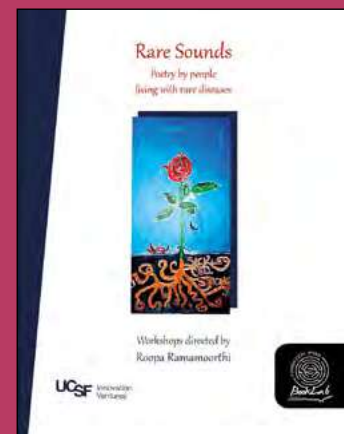
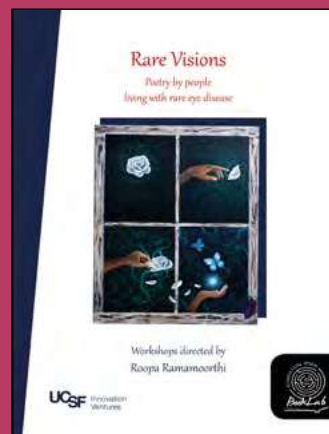
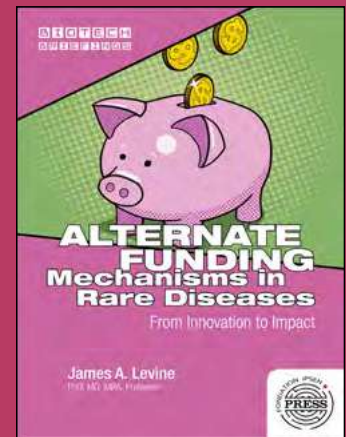
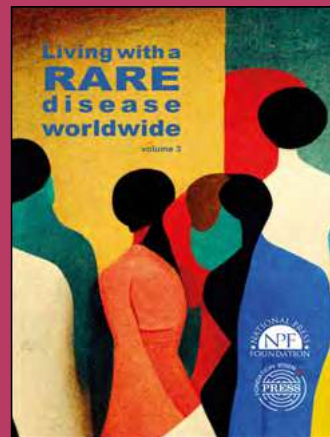
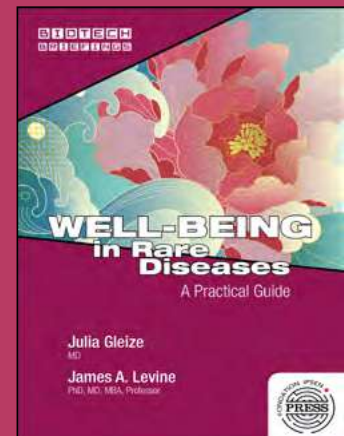
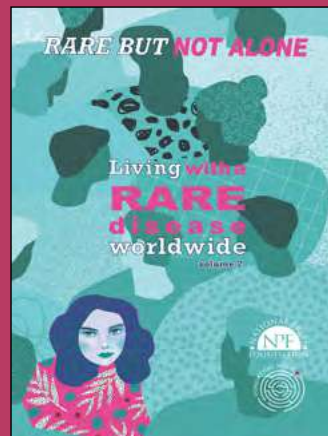
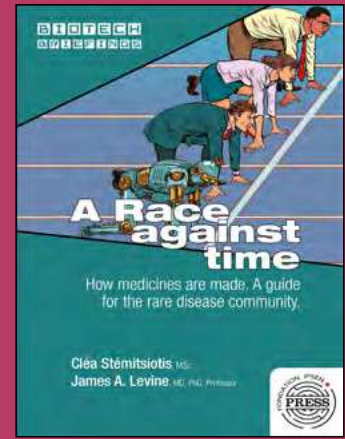
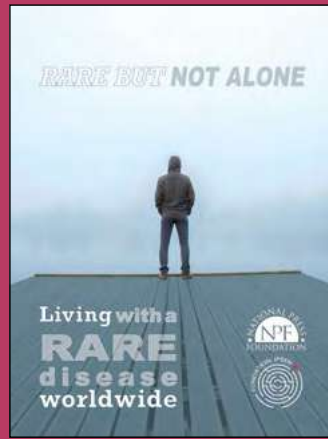




Living with a **RARE** disease worldwide

volume 4





Rare Disease Reporting: Family to Family, Continent to Continent

Recent history-making biotechnological innovations in the rare disease realm herald the dawn of a hopeful new era.

The research that produced a successful personalized medicine treatment for Baby K.J. Muldoon in 2025, led by researchers at Children's Hospital of Philadelphia, made headlines around the world – and for good reason.

Few would argue against communicating technology's ability to produce specialized cures for the complex, often deadly rare diseases affecting more than 300 million people around the world. But the expertise needed to provide vital context about those biotech advances is still elusive for many international journalists.

For the fifth time in 2025, the National Press Foundation continued its mission to fuel better reporting on the research, advocacy and physical challenges of living with rare diseases by convening the Rare Diseases Reporting Fellowship via Zoom from Nov. 17-21, 2025.

The program, hosted by NPF through funding from Fondation Ipsen, convened one of the largest training cohorts ever curated by NPF. The 146 international applicants for the program hailed from six continents. More than 60 journalists from 29 countries participated in the week-long virtual training, hearing from experts including:

- Rebecca Ahrens-Nicklas and Kiran Musunuru, the history-making Children's Hospital of Philadelphia researchers;
- Fernando Goldzstein, the Brazilian businessman who founded the Medulloblastoma Initiative to save his son's life;
- Ida Mirkovic-Knaus and Yamina Husseini, the Croatian and French youth advocates for people living with rare diseases and their families;
- Philip Langat, the Kenyan agricultural scientist who lost five relatives to motor neuron disease and who's determined to help other families on the African continent.

NPF's core training strategy around covering rare diseases centers on the urgent need to curate information relevant to a varied international audience. While many U. S. and European journalists have access to the research institutions and established advocacy organizations that can support their reporting and story development, many journalists in the Global South face a range of challenges. In countries like Nigeria, Colombia and Malaysia, people living with rare diseases and their families often endure intense isolation, silenced by religious and cultural taboos, outright disinformation or lack of medical resources. Journalists who may encounter rare disease patients are often hampered by lack of medical and/or research facilities that can provide contextual support. As a result, the potential to lessen stigma and inform the public becomes even more elusive.

NPF's successful training format reached a new peak during the 2025 program, with journalists giving extremely high ratings for sessions about access to genetic testing, the use of

AI in the rare disease realm and testimony from the first person to be successfully cured of sickle cell disease using CRISPR therapy, Victoria Gray. In her second conversation with NPF fellows, Gray detailed enduring a lifetime of suffering due to sickle cell disease before receiving the historic gene-editing therapy in 2019. Said Evans Jona of The Post in Zimbabwe, "She put a human face to what advanced medical innovation can achieve when patients are placed at the center of care. Listening to her describe life before and after treatment made it painfully clear how far behind our own systems still are. Patients in Zimbabwe are still fighting for basic pain management, reliable testing, and consistent clinical follow-ups."

That critique perfectly illustrates the enormous opportunity that the Rare Diseases Reporting Fellowship offers to journalists worldwide. Just as technology is potentially easing some of the barriers of access to information, research and treatment for people living with rare diseases, the ability to convene a cohort of journalists for exclusive briefings yields tremendous dividends, as this fourth volume of projects produced by fellowship participants illustrates.

Said Colombian freelance journalist Carlos Cordero, who produced a comprehensive feature about Huntington's Disease post fellowship,

"Ahrens-Nicklas's presentation on genetic diagnostics and the clinical management of hereditary disorders was extremely relevant. Understanding how inherited conditions are detected, monitored, and communicated to families gave me essential context for reporting on communities where Huntington's has been present for generations."

NPF is tremendously grateful for the opportunity to continue offering this vital expertise to journalists worldwide. The 2026 program will leverage this solid track record to advance the goal of educating communities around the world about rare diseases.

Rachel Jones

Director of Journalism Initiatives, National Press Foundation
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Because we can't wait longer

One of the great challenges faced by people living with rare diseases is that the public knows so little of their struggles, challenges, and the need for unrelenting resilience in a world that doesn't understand. Coupled to the 300 million people living with rare diseases are hundreds of millions of caregivers who sacrifice time, work and money in the name of love. Working with the National Press Foundation, top world journalists highlight in these writings the plights of patients living with rare diseases and those who care for them. The stories are informative, dramatic, and heart-rending. I thank most the patients living with rare diseases and the people who love them for sharing their stories with these gifted journalists.

James A. Levine, MD, PhD, Professor
President, Fondation Ipsen

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Alex

ABABIO

in Ghanaian Watch | Ghana



Mr **Alex Ababio** is a Managing Editor of Ghanaian Watch with diverse media experience, specializing in health , science, environment and human rights.

He reports on how emerging challenges-including public health crises shape lives in Ghana .

He is also a fellow of the Internews Health Journalists Network, the International AIDS Society ‘s HIV Vaccine Research Advocacy Fellowship, and the Thomson Reuters General News Reporting fellowship.

He co-founded the Society of Journalists, Ghana to empower journalists with investigative

storytelling skills and expertise development.

In 2015, he co-founded FOKLEX Media Production to honor Ashanti region journalists, contributing to community development.

As an MPT Internews Health Journalists Network fellow from June to August 2023, Mr. Ababio authored a feature article titled “Transcending Taboos and Illegality: Pioneering Inclusivity in Ghana’s HIV Solutions.

In 2015, he joined the Ashanti Regional National Media Commission team, researching political shows to improve the political atmosphere.

3 Million Ghanaians, No Clear Plan: How the Diagnostic Gap Is Leaving Children with Rare Diseases to Die Early



Born With Symptoms, Still Undiagnosed

The afternoon heat in New Tafo Krofrom, a crowded inner-city community in Kumasi, was intense. The air barely moved, and sweat gathered easily on the skin. Inside a dim, poorly ventilated room, Rita Fosua sat on the edge of a thin, worn mattress.

The roof leaks when it rains. The floor is cracked and full of holes. Insects crawl along the walls. This is where she lives with her four children.

In her arms lies Kwaku — one and a half years old — his head bent backward at an unnatural angle, his tiny ribs visible beneath fragile skin. He has never sat on his own. He cannot look straight; his eyes drift sideways. Since birth, he has never suckled his mother's breast.

"My son was born at Old Tafo Government Hospital," Rita Fosua recalls, her voice steady but tired. "Right from birth he did not cry. His eyes were closed and he couldn't open them. The doctors put him in a machine for a long time. They could not figure out what was wrong."

From old Tafo Government Hospital to South Suntreso Government Hospital. From there to Komfo Anokye Teaching Hospital. At each stop, the same question echoed: What is wrong with this child?

No Doctor Could Name His Illness :

At Komfo Anokye, Kwaku finally opened his eyes — but he still could not fix his gaze. Doctors issued internal referrals. They asked for physiotherapy. They ordered checks on his eyes, ears and cognitive function. Rita and her husband managed two physiotherapy visits at Manhyia Government Hospital in Kumasi. Each visit cost GH¢100.

And That's When the Family Hit Empty Pockets

The father, who survives on irregular menial jobs, has spent more than GH¢10,000 since Kwaku was born. The family now buys canned food because Kwaku cannot breastfeed. Some days, they cannot

afford even that.

“We don’t know how much they will charge to check his mind, eyes and ears,” Rita says. “We have no money again.”

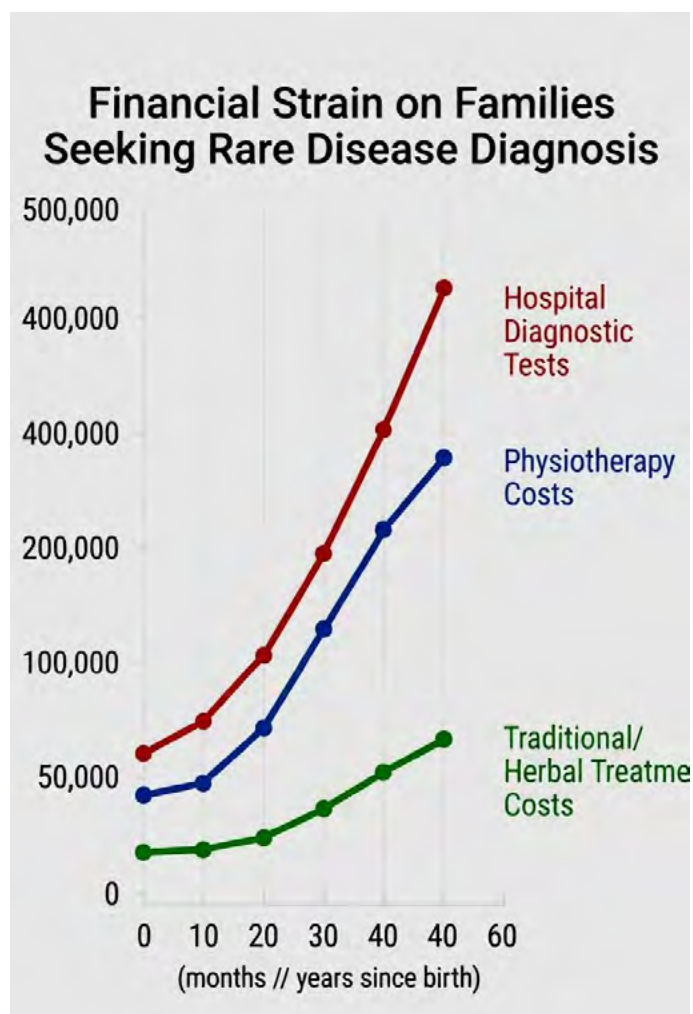
They turned briefly to herbal medicine when orthodox treatment brought no answers. It was even more expensive. They stopped.

Now Kwaku lies quietly in the dark room that smells of dampness and exhaustion — a child without a diagnosis, a family without hope.

Kwaku’s Story Reflects the Plight of Millions of Unseen Ghanaians.

In Ghana, millions of children like Kwaku are trapped in a diagnostic desert — rooms full of uncertainty, tests that don’t exist locally, and doctors struggling with conditions they can’t name.

American geneticist and bioethicist Dr. Charmaine Royal emphasizes : The global imbalance in genetic data not only undermines equity in diagnosis but also limits the development of treatments for underrepresented populations,” highlighting why the lack of local data has devastating consequences.



A Hidden Population, Hurting: Three Million Forgotten Ghanaians Living with Rare Disease

In Ghana, an estimated three million people — roughly 10% of the Ghana’s 35.5 million population — are living with rare diseases

According to Professor Jerry John Kponyo, Scientific Director and Project Lead of the Responsible Artificial Intelligence Lab (RAIL) at the Kwame Nkrumah University of Science and Technology Engineering Education Project,



The Scientific Director and Project Lead of RAIL at Kwame Nkrumah University of Science and Technology Engineering Education Project
Professor Jerry John Kponyo

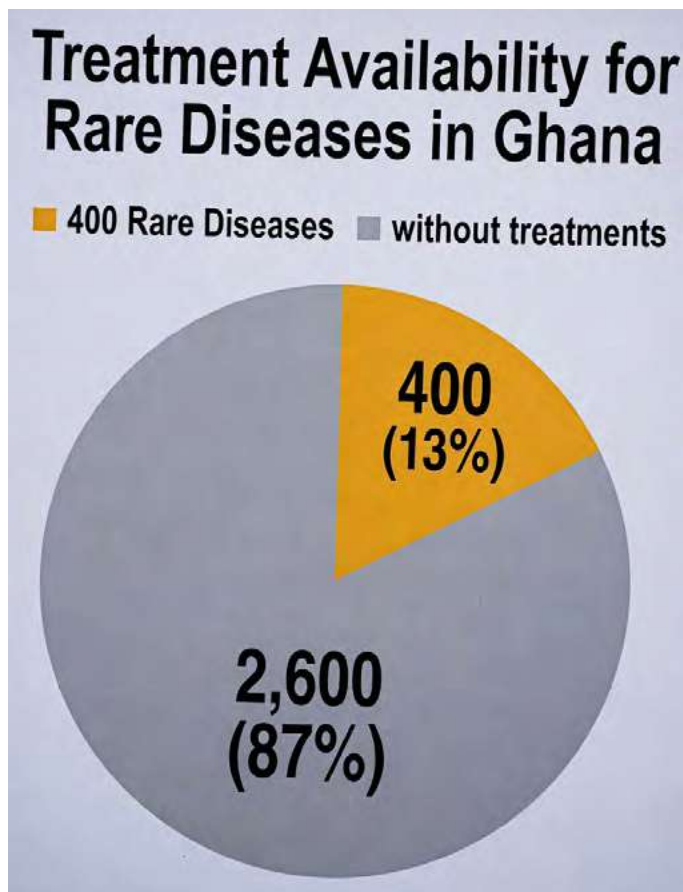
Globally, more than 350 million people live with rare diseases, according to the France-based Fondation Ipsen. The U.S.-based National Organization for Rare Disorders estimates that 30 million Americans — also about 10% of the population — are affected. Around 95% of rare diseases have no approved treatment.

Yet in Ghana, the challenge is compounded by a severe diagnostic gap.

Professor Kponyo explains that between 6,000 and 8,000 rare diseases have been identified globally. Only around 400 have treatment plans — and many of those depend on precise genetic understanding.

“One thing about rare conditions,” he says, “is that they are often caused by a single gene. If you can identify that gene, you can tailor treatment.”

But identifying that gene requires genetic testing — a service that can cost up to \$10,000 per family. How many Ghanaian families have \$10,000 sitting idle? Certainly not Rita.



No Name, No Hope: The Diagnostic Gap :

The Executive Director of Rare Diseases Ghana Initiative, Samuel Adjei Wiafi, does not mince words.

“The biggest issue is that there isn’t a policy. There’s no clear national plan for rare disease patients,” he says. “The diagnostic infrastructure is woefully inadequate.”



Through a philanthropic diagnostic access program, his organization has supported nearly 400 families with genetic testing — at roughly \$10,000 per test. Multiply that.

That is nearly \$4 million raised largely through philanthropy, not government systems.

“It’s not sustainable,” Wiafi admits. “If a patient is suspected anywhere in the country, they should be able to get testing done affordably, ideally covered by insurance.”

But Ghana’s National Health Insurance Scheme (NHIS) reimburses based on coding systems. Many rare diseases lack proper coding integration. Even though the global ICD-11 classification includes many rare diseases, it remains unclear whether full integration into Ghana’s reimbursement structure has occurred.

NHIS Ashanti Regional Director Muniru Mohammed Muntaka argues that about 95% of diseases and medications are covered. He points to the Ghana Medical Trust Fund — popularly known as Mahama Care — as an initiative to support conditions not covered.

“The scheme must be sustainable,” he explains. “If we bring in all cancers and rare diseases at once, it could collapse the scheme.”



The Ashanti Regional Director of the National Health Insurance Scheme (NHIS), Mr. Muniru Mohammed Muntaka

He emphasizes actuarial projections and financial stability. The government, he says, is building capacity gradually.

For families like Rita’s, gradual feels like never.

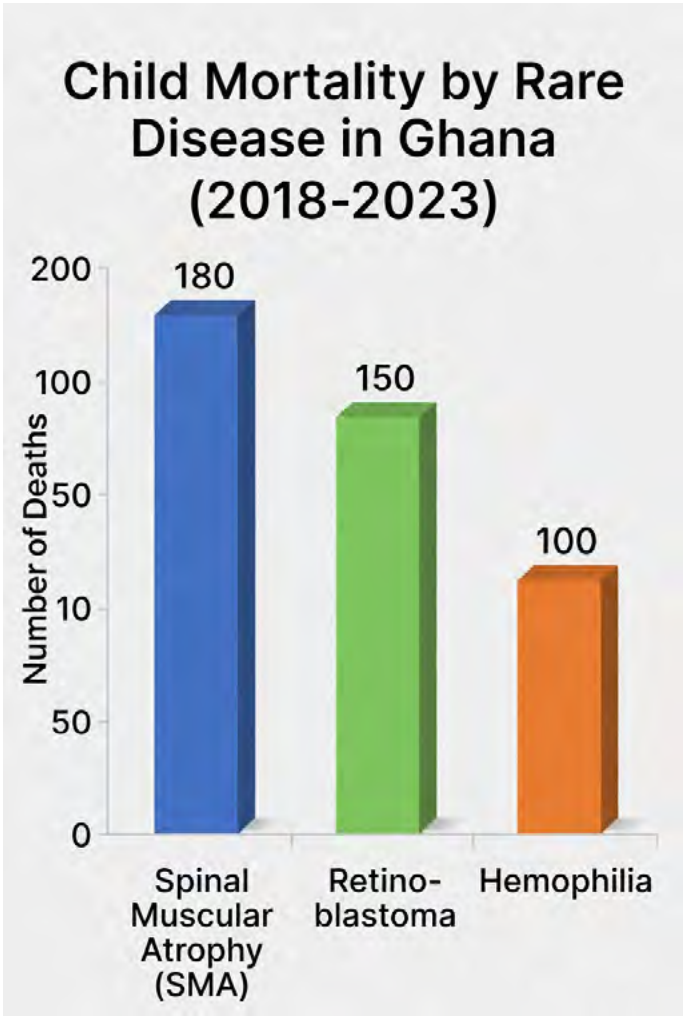
Children Dying Before Diagnosis

Spinal Muscular Atrophy (SMA) offers a window into the crisis.

Between 2018 and 2023, 17 SMA cases were confirmed across five centers in Ghana, according to hospital records cited by Dr. Charles Kumi Hammond,

Associate Professor, Department of Child Health, KNUST & Consultant Pediatric Neurologist, Komfo Anokye Teaching Hospital KATH, Kumasi

Nine of the 17 children — 53% — died. Four had originally been referred as cerebral palsy. Four were labeled broadly as neuromuscular disorders. Five families paid for overseas genetic testing. Twelve relied on special programs. Globally, SMA incidence ranges from 1 in 6,000 to 1 in 10,000 births. In the United States, prevalence is about 1 in 10,000 individuals. In Ghana, prevalence is unknown — largely because newborn screening does not exist. Management here remains supportive: physiotherapy, respiratory care, genetic counseling. The life-saving gene therapies available elsewhere are not accessible. One surviving Ghanaian child with SMA type I received newborn screening overseas and gene therapy. That child lives. Others did not.



The Eye Cancer That Steals Sight

Dr. Samuel Tenagyei, a leading optometrist in Kumasi, speaks about retinoblastoma — a rare childhood eye cancer affecting children under five. “The symptoms are classical,” he says. “It is not difficult to diagnose. But it is difficult to manage.” Prognosis is often poor. Saving vision — sometimes even life — becomes uncertain. He highlights systemic weaknesses: limited state-of-the-art equipment, inadequate genetic testing, insufficient data systems. “Most of the time it goes undiagnosed or is labeled cause unknown,” he says.



Dr. Samuel Tenagyei, a leading optometrist

He describes the psychological trauma of families shuttling from one hospital to another, enduring trial-and-error treatments. “Imagine the anxiety, the fear, the loss of hope,” he says. “And of course, in our setting, we quickly resort to spiritual explanations.” In the absence of diagnosis, superstition fills the vacuum.

Stigma, Silence, and the Weight of Spiritual Blame :

Elizabeth Emmanuella Nimon, National Secretary of the Ghana Society of the Physically Disabled, describes a 12-year-old boy in Bekwai. The child suffers mysterious symptoms — mouth sores that migrate, sensations of intestines “folding,” constant hospital visits without answers. “Every time they go, they are told a different thing,” she says. “They are not able to get specific treatment.”



Elizabeth Emmanuella Nimon, National Secretary of the Ghana Society of the Physically Disabled

Families are accused of witchcraft. Children are whispered about. Mothers are blamed. Neurofibromatosis patients with facial tumors struggle to attend school or church without ridicule. Down syndrome children with congenital heart defects require surgeries families cannot afford.

Wiafi puts it bluntly: “The moment they have a child with a rare disease, they start moving toward the poverty line.”

Kwaku’s family is already there.

Who Profits, Who Suffers: Following the Money
Rare disease care is expensive everywhere. But in high-income countries, insurance systems, social safety nets and research funding cushion the blow. In Ghana, families pay out-of-pocket.

Physiotherapy: GH¢100 per session.

Genetic testing: up to \$10,000.

Cochlear implants: tens of thousands of cedis.

Congenital heart surgeries: unaffordable for most. NHIS may cover consultations and some labs, but advanced diagnostics, genetic tests, assistive devices and orphan drugs often fall outside reimbursement structures.

Childhood cancers were recently considered for inclusion. That is progress. But thousands of other rare conditions remain uncovered.

The financial burden pushes families downward — sometimes permanently.

A Global Mandate — Will Ghana Act?

On 24 May 2025, the World Health Assembly unanimously adopted a Resolution on Rare Diseases. It recognizes rare diseases as a global health priority and calls for improved diagnosis, treatment, care and research.

Ghana co-sponsored the resolution.

The resolution requests a 10-year Global Action Plan.

The world has spoken.
Will Ghana listen to itself?

Artificial Intelligence and a Flicker of Hope
At KNUST’s Responsible AI Lab, researchers are building tools to analyze genomic data at scale. Their model compares patient genetic information with a global pool of 29 million shared datasets.

In one case, a woman lost four babies shortly after each birth. Doctors treated recurrent convulsions without identifying the root cause. The KNUST Engineering Education Project -RAIL Genetic analysis revealed a condition triggered by breastfeeding. When she avoided breastfeeding with her next child, the baby survived.

From Numbers to Hope: How Data Saved a Life

But hope is emerging. Pilot programs using genomic data have already changed outcomes for some children, turning numbers into lifesaving interventions.

“Professor Shahida Moosa, a leading genomic clinician in South Africa, says building diagnostic capacity tailored to African genomes is essential — not optional — if millions are to be diagnosed accurately and early,” signaling that with proper investment, the story of Kwaku could be replicated for thousands more.

The Scientific Director and Project Lead of RAIL Professor Kponyo believes Ghana can lead Africa by integrating AI-driven genomic tools into public health systems.

But algorithms cannot function without data. Data cannot exist without testing. Testing cannot happen without funding and policy.

“At the Responsible Artificial Intelligence Lab, our main goal is to ensure that the innovative solutions we develop actually reach and benefit people. There’s no point in creating a solution if it’s never used. That’s why, with our Care for Rare Disease program, we aim to partner with the Ministry of Health and the Ghana Health Service to address challenges related to rare diseases.”, Professor Kponyo explained

Technology alone cannot fix systemic neglect. What Must Change

Experts agree on several urgent priorities:
 A comprehensive national rare disease policy.
 Integration of rare diseases into NHIS coding and reimbursement.
 Investment in genetic testing and newborn screening.
 Expansion of specialist services — physiotherapy, speech therapy, occupational therapy.

Inclusion of orphan drugs and assistive devices on the essential medicines list.
 Incentives for research and data management.
 Public awareness campaigns to reduce stigma.

Rare Diseases Ghana Initiative is engaging policymakers. Advocacy forums have been held with the Ministry of Health.
 But policy drafts do not breathe for children like Kwaku.

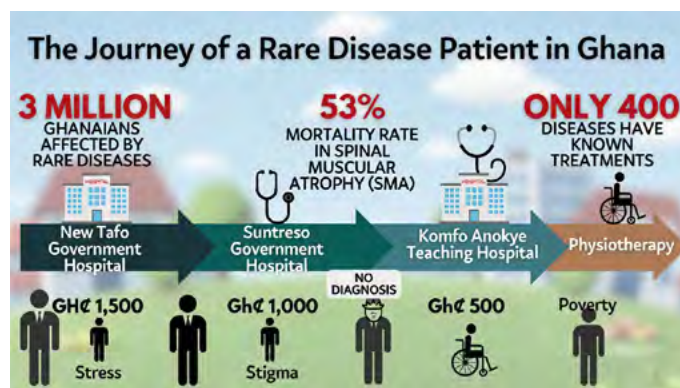
Trapped in the Dark: Life in a Forgotten Room :

As evening falls in New Tafo Krofrom, Rita adjusts Kwaku's head gently so he can breathe comfortably. She does not know the name of her son's condition. She does not know whether he will sit, speak, or

survive.

She only knows that she has walked from hospital to hospital, from hope to hope, spending money she did not have.

Three million Ghanaians live in similar uncertainty — invisible in statistics, absent in policy, waiting in dark rooms across the country.



Rare but deadly.

The question is no longer whether Ghana can afford to prioritize rare diseases.

It is whether Ghana can afford not to.

Because every day without diagnosis is not just a policy delay.

It is a childhood slipping away. ■



Rashad

AHAMAD

in New Age | Bangladesh



Rashad Ahamad is an award-winning investigative journalist based in Dhaka, Bangladesh, with over 15 years of experience reporting on public health, healthcare, health education, labor rights, and human impact stories.

He has covered major health crises, including the Covid-19 pandemic, tropical diseases, antimicrobial resistance, and vaccine hesitancy in remote communities.

Rashad's reporting focuses on how health inequities and systemic neglect affect vulnerable populations. His

work has appeared in leading national outlets such as New Age and The Daily Star, as well as in international media including The New York Times, Deutsche Welle, Al Jazeera, NZZ, Le Monde and NPR.

He has received national and international awards and fellowships for his reporting. As President of the Marine Journalists' Network, Rashad advocates for press freedom, ethical, data-driven, and compassionate storytelling that gives voice to the unheard and brings rare and neglected issues to public attention.

Thalassaemia spreads as the prevention plan stalls



Thalassemia affected child treated by blood transfusion at the country's largest child health institute, Bangladesh Shishu Hospital and Institute in the capital's Sher-E-Bangla Nagar.
| New Age photo

Thalassaemia, a genetic blood disorder that is considered relatively rare worldwide, is gradually spreading in the country because of limited screening, weak prevention measures and costly treatment.

Although the government adopted a National Thalassaemia Prevention Action Plan in 2018 to eliminate the disease by 2028, public health experts say that the implementation of the plan has stalled.

The deadline for the national prevention plan has now, however, been extended to 2030.

According to the Bangladesh Bureau of Statistics, 11.4 per cent of the population were thalassaemia carriers in 2024 though the rate was 4 per cent in 2000.

With a population of 174.7 million, this suggests that the country has nearly 20 million carriers. Globally, about 1.5 per cent of people are carriers, with significantly higher rates in South Asia.

The Bangladesh Thalassaemia Foundation estimates that 60,000 to 70,000 people, mostly under 35, are living with the disease in Bangladesh. Each year, 6,000 to 8,000 babies are born with thalassaemia.

Professor Dr Syeda Masooma Rahman, vice-chair of the foundation, said that the government had yet

to establish a functional patient registry or introduce mandatory premarital and prenatal screening.

‘Not only the government, but international organisations and donors have overlooked this serious but preventable health issue,’ she said.

She noted that most district hospitals lack electrophoresis testing facilities, which are essential for diagnosing thalassaemia and other blood disorders, as well as adequate treatment services.

She also criticised insufficient public awareness campaigns about the disease.

State minister for health and family welfare MA Muhit acknowledged that thalassaemia had not been prioritised amid the country’s heavy burden of other diseases such as tuberculosis, malaria, diabetes and hypertension.

‘Treatment of all diseases is a struggle here. We will work to overcome the situation,’ he said, stressing that prevention through public awareness was more effective than treatment.

In 2023, the High Court directed the authorities to form an expert committee to draft a policy to curb the spread of thalassaemia.

However, Directorate General of Health Services disease control director Professor Dr Md Halimur Rashid said that activities under the non-communicable disease control programme, which included thalassaemia, were suspended after its operational plan was scrapped in 2024.

State minister Muhit said that the authorities were uncertain when actions against non-communicable diseases could resume.

Data from the Bangladesh Shishu Hospital and Institute show that E-beta thalassaemia accounts for 60 per cent of cases in the country, while beta thalassaemia represents the remaining 40 per cent.

Thalassaemia is an inherited disorder that impairs the body’s ability to produce normal haemoglobin, leading to chronic anaemia, fatigue and severe complications affecting the heart, brain and kidneys.

Patients often require lifelong blood transfusions, which can cause iron overload if not managed with iron chelation therapy.

At the Bangladesh Thalassaemia Foundation's charity hospital, about 8,440 patients from across the country are receiving treatment, reflecting the limited availability of services elsewhere. The country has only five specialised thalassaemia centres.

More than 60 per cent of thalassaemia patients are under 21, while only 2.4 per cent are over 60. The average life expectancy in Bangladesh is 72.

Doctors say many patients die prematurely due to complications linked to inadequate treatment and iron overload.

Professor Dr Md Belayet Hossain of paediatric haematology and oncology at Bangladesh Shishu Hospital and Institute said that treatment remained largely unavailable and unaffordable, contributing to early deaths. There is no official data on thalassaemia-related mortality.

He also warned that blood transfusion safety remains a concern in some facilities.

Currently, red blood cell transfusion combined with iron chelation is the mainstay of treatment. Advanced options such as bone marrow transplantation, stem cell transplantation and gene therapy remain financially out of reach for most families.

Monthly treatment costs for the disease range from Tk 20,000 to Tk 30,000, which is nothing but a crushing burden for many households, said Bangladesh Thalassaemia Patients and Parents Welfare Association's founding president Kamrun Nahar, who is also a thalassaemia patient.

Iron chelation tablets cost around Tk 250 each, she said, urging price reductions.

'We are neglected. Nobody cares how we are dying. Nobody even counts our deaths as thalassaemia deaths,' she said, breaking down while recalling friends who had died.

Physicians explain that if both parents are carriers, each pregnancy carries a 25 per cent risk of

producing a child with thalassaemia, a 50 per cent chance the child will be a carrier, and a 25 per cent chance of being unaffected. If only one parent is a carrier, the child will not develop the disease but may inherit carrier status.

The experts strongly recommend premarital screening and prenatal testing before 12 weeks of pregnancy to prevent new cases.

However, systematic screening remains absent, leaving most carriers unaware of their status until complications emerge or a child is born with the condition.

Dr Mohammad Abdullah Al Zubayer Khan, head of haematology at the National Institute of Laboratory Medicine and Referral Centre, said that the absence of a national registry was a major barrier to control efforts.

'There are no pragmatic measures on the ground from the government to eradicate thalassaemia,' he said, adding that only charities and a few non-governmental organisations were working on a limited scale.

In a recent survey, he said, he found that carrier rates reached up to 40 per cent in some areas, particularly among ethnic communities in the hill tracts and northern districts. The survey report is yet to be published.

'We need to investigate why prevalence is higher there,' he said.

The human cost is evident. Rahima Begum and her husband Hafiz Uddin, a rickshaw puller, had three children with thalassaemia and only one of them, Sumon Mia, survives. Their daughter Juni Akter died aged 10 in 2010, and their son Sohagh Mia died three years later.

'If we had known about thalassaemia, we would not have brought children into the world with this disease,' Rahima said, recalling how unqualified practitioners repeatedly misdiagnosed her children's illness as jaundice.

Without urgent investment in nationwide screening, public awareness and affordable treatment, thalassaemia will continue to expand silently, doctors warned. ■



Jennifer

AMBOLLEY

in The Chronicle Newspaper | Ghana



Jennifer Ambolley is an award-winning journalist with over four years of experience. She is passionate about using storytelling to spotlight underreported issues, influence public policy, and drive sustainable development.

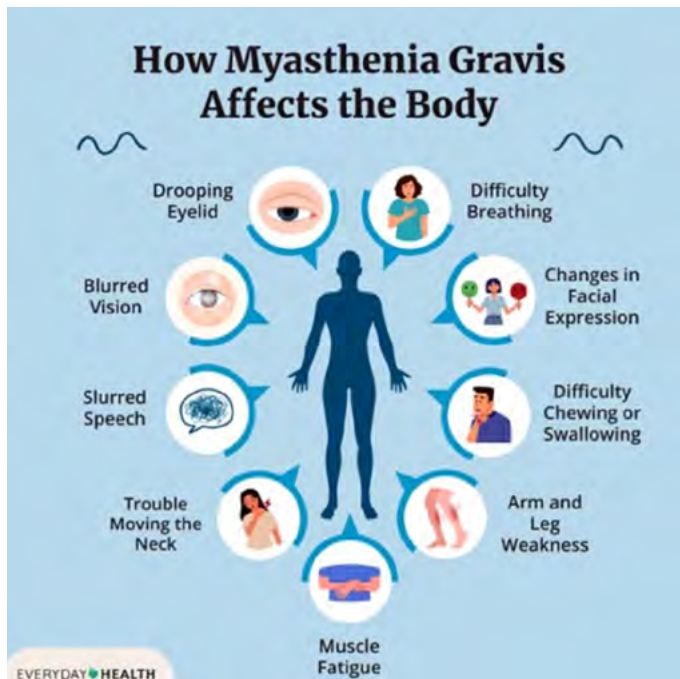
Jennifer currently works with The Chronicle Newspaper in Ghana, where she produces stories on public health, environmental justice, and technology.

Her exceptional contributions have earned her recognition, including two First Prizes in the Merck Foundation Media Recognition Awards (“Diabetes & Hypertension” – 2023, West Africa; “More Than a Mother” – 2024, West Africa) and a nomination for African

Print Journalist of the Year at the Ladies in Media Awards.

She is also a multi-fellowship recipient, including the Her Press Fellowship 2025, Digital Public Infrastructure (DPI) Journalism Fellowship 2024, Oxford Climate Journalism Network Fellowship (Cohort 7), African Investigative Journalism under 30 Fellowship 2024, CPHIA Journalism Fellowship 2023, and the Clean Air Reporting Fellowship 2024, among others. Through her work, Jennifer continues to shape narratives around digital rights, public health, environmental advocacy, and governance across Africa.

Feature: Diagnosis in the Dark: A Mother's Search for Hope



Myasthenia Gravis Symptoms

When COVID-19 arrived in Ghana in early 2020, it came wrapped in fear and uncertainty. Streets emptied. Schools shut their doors. Hospitals became places people avoided unless absolutely necessary. Every cough raised suspicion, every fever felt dangerous. Families stayed indoors, waiting for normal life to return.

But for Charity Juliet Asamani and her family, COVID did not simply bring lockdowns and masks. It arrived bearing a cruel, almost ironic burden, one that would reshape their lives long after the pandemic faded from public consciousness.

Charity Juliet Asamani is the mother of Freda (not her real name), her nine-year-old daughter, a once extremely active child now living with Myasthenia Gravis, a rare autoimmune neuromuscular disease.

Freda was just three years old when the first signs appeared, during the height of the COVID lockdown, marking the beginning of a six-year journey through misdiagnosis, uncertainty and a health system unprepared for the rare.

When the Eye Would Not Open

It began quietly.

Freda had gone out to ride her bicycle. When she returned, Juliet noticed what looked like a small pimple near her left eye. By evening, the eyelid appeared damaged, and the lower lashes were missing. By the next day, the eye would not open at all.

Alarmed, Juliet rushed her daughter to a clinic. The response was unexpected.

“They asked me why I was only coming now,” she recalls. “I said the thing just happened. I don’t even know what is wrong.”

What followed was a maze of tests and referrals eye examinations, scans, and MRIs most of which came back normal. Yet Freda’s eye remained shut. More confusing still was the pattern Juliet began to notice: every morning, Freda’s eye would open normally. As the day progressed, it slowly drooped shut again.

That daily cycle, strength at rest, weakness with activity would later become the key to unlocking the diagnosis.

After several referrals, Freda was sent to see a Neurologist. There, a simple test changed everything. An ice pack was placed over her eye. When it was removed, the eyelid lifted.

The diagnosis was Myasthenia Gravis.



Drooping eyelid in Myasthenia Gravis (MG)

A Rare Name, an Uncertain Path

Before the diagnosis, some doctors suggested surgery to correct Freda's drooping eyelid. Others hinted that her eye might eventually need to be removed. Juliet refused.

"I didn't even sit down," she says. "I left and went to another facility."

Initially, the weakness appeared limited to one eye. Plans were even made for minor surgery. Then, two weeks later, the other eye began to droop.

Over time, the weakness spread from the eyes to the neck, and then to the limbs. Today, Freda struggles especially with her lower body. Walking for long distances exhausts her. The child Juliet describes as "very, very, very active" now lives within the limits set by fatigue.

Medication helps but only to a point.

"They told me it's not something that can be cured. She will be on medication for life," Juliet says.

The dosage has increased steadily since 2019. Yet the weakness continues to descend.

From the COVID lockdown to today, Juliet's life has been defined by hospital visits: teaching hospital corridors, eye clinics, paediatric neurology units follow-ups, referrals and second opinions.

"Sometimes we were admitted and not even given medication," she says. "They don't really know which medicines to give her and which to avoid."

Even with a diagnosis, uncertainty remains.

Freda takes medication four times a day, supplemented with other drugs and multivitamins. One bottle costs thousands of cedis and does not last long. Juliet must wake her at night, administer the first dose before school, send medication along, and give the final dose when Freda returns home.

"She's getting weaker and weaker each day," she admits. "I think it's the medicine, but what choice do we have?"

Freda's schooling has also been disrupted. Some days, she cannot attend at all. On others, she goes

but struggles through exhaustion invisible to most.

Juliet has explored treatment options abroad, contacting doctors online and searching for help beyond Ghana's borders. The responses often require tests, funds, or connections she does not have.

A System Unprepared for the Rare

Freda's story reflects a broader national gap in the care of rare conditions.

Myasthenia Gravis (MG) is a rare autoimmune neuromuscular disorder in which the immune system disrupts communication between nerves and muscles, leading to fluctuating muscle weakness.

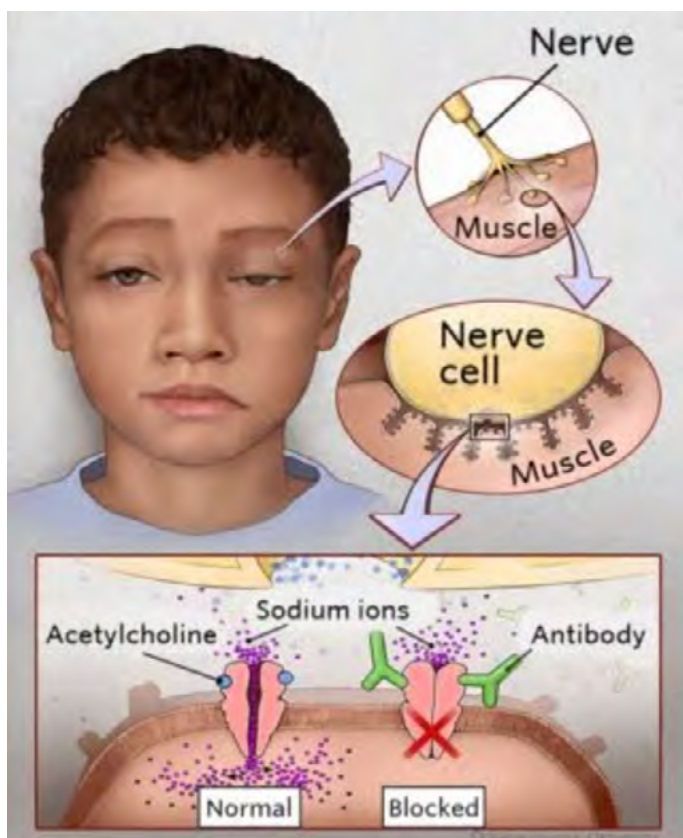
Globally, an estimated one million people live with the condition, though experts say the true number is likely higher particularly in low- and middle-income countries where diagnosis is often delayed or missed.

Common symptoms include drooping eyelids, blurred or double vision, difficulty chewing or swallowing, slurred speech, neck weakness, and fatigue that worsens with activity and improves with rest. In children, these signs are often mistaken for eye problems, infections, or developmental issues, delaying proper diagnosis.

This pattern explains why Freda's eyelid would open in the morning and gradually droop as the day progressed. While MG is considered manageable with early diagnosis and consistent treatment, it is rarely suspected in children in Ghana, where specialized testing and long-term care options remain limited.

Ghana has no national rare disease policy, no centralized data system, limited genetic testing capacity, and few specialists trained to recognize rare conditions early especially in children. As a result, patients drift from facility to facility, accumulating misdiagnoses, financial strain, and emotional exhaustion.

Early diagnosis, critical for preventing complications and reducing long-term costs, is often delayed by years, leaving families to navigate a health system that was never designed with rarity in mind.



The uncertainty has affected Freda's daily life.

"She is easily fatigued and weak, and the family is always afraid she might fall or hurt herself," Mr Waife said, noting that her condition limits her ability to participate in normal social activities.

Care coordination, he added, has largely fallen on Freda's mother, who is also grieving the recent loss of her husband. "The mother has to do everything navigate the system, talk to doctors, and manage the condition on her own," he said.

According to Mr Waife, Freda's experience reflects a wider systemic failure.

"Without a national rare disease policy, families like Freda's are left on their own. We are not training experts, investing in diagnostics, or creating clear care pathways," he said.

He argued that Ghana must prioritize awareness, early diagnosis, regional centres of care, and sustained research investment to prevent families from carrying such an overwhelming burden alone.



Rare Disease

What Health Care Providers Say

Samuel Agyei Waife, a clinical psychologist and rare disease advocate, first met Freda and her family through a mental health referral not as part of her medical care. The family had been advised to seek psychological support as the emotional strain of prolonged uncertainty and repeated hospital visits became overwhelming.

Freda has Myasthenia Gravis (MG), a rare neuromuscular condition with only a few documented cases in Ghana. Diagnosing the condition, Mr Waife explained, is especially difficult.

"Unlike some rare diseases where you can run a genetic test and get a clear answer, this one is different. It requires largely clinical assessment and specialized biomarker tests, and most of these tests are very expensive," he said.

For Freda's family, this meant moving from one hospital to another, consulting different specialists and undergoing multiple tests without clear answers.

"They have seen different hospitals, different teams, and done many tests. They have a whole file of investigations, yet care is still not coordinated," he said.

What Experts Say

Dr Nicholas Ekow Thomford, Senior Lecturer at the University of Cape Coast and Head and Principal Investigator of the Pharmacogenomics and Genomic Medicine Group and Laboratory, says Ghana's rare disease burden is far heavier than official attention suggests.

"There are a lot of rare diseases in Ghana, but there is very limited rare disease research in the country," Dr Thomford explains. "Generally, anything rare is overlooked. And in a country with limited resources, there is added incentives to focus on diseases policymakers believe affect large numbers of people."

Most rare diseases, he notes, occur in roughly one out of every 2,000 people. On paper, that low prevalence often pushes them out of policy conversations. In reality, when taken together, rare diseases affect thousands of families many of whom like Freda's, spend years searching for answers.

Dr Thomford describes Ghana's lack of a national rare disease policy as a critical failure point.

"The absence of a policy confirms the level of importance placed on rare diseases," he says. "It makes research almost non-existent and more expensive, local diagnosis complicated or unavailable, and significantly lowers the odds for patient survival and good outcomes."

For families, this policy vacuum translates into long diagnostic odysseys multiple referrals, inconclusive tests, and clinicians forced to rely on guesswork rather than clear diagnostic pathways. Freda's early experience, where surgery was proposed before the underlying neurological cause was understood, reflects this systemic gap.

At the research and diagnostic level, Dr Thomford points to major deficiencies that fuel delayed or incorrect diagnoses, especially in children. These include limited capacity for functional genetic variant identification and poor access to orphan drugs medicines developed specifically for rare conditions.

"Most rare diseases have a genetic basis," he explains. "Because they are rare, they require advanced methods to identify the causative

functional genes. These methods demand funding, expertise, and sophisticated equipment which are limited in Ghana."

Genetic testing and proper data collection, he adds, could dramatically shorten the diagnostic journey for patients like Freda.

"Phenotypic and genotype data help in the easy identification and management of rare disease symptoms that clinicians have already encountered," he says. "But the biggest barriers remain funding, access to advanced equipment, and trained personnel."

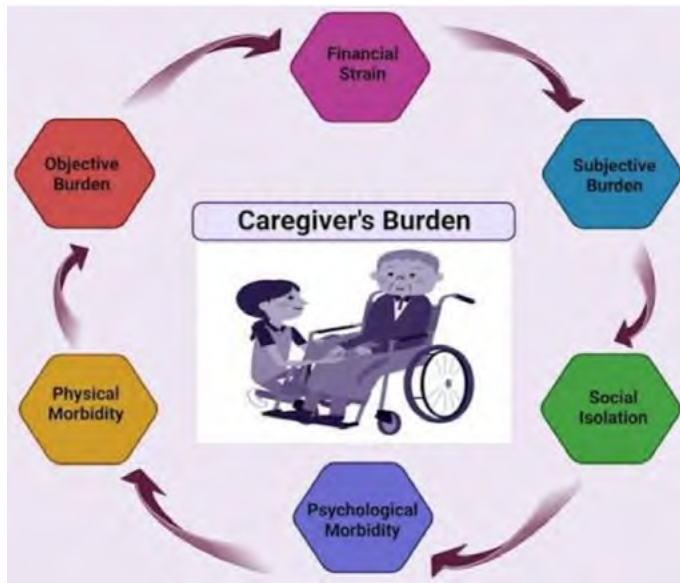
Despite these constraints, Dr Thomford believes progress is possible if rare diseases are deliberately brought into national health planning.

"If we engage researchers and clinicians to develop practical policies, we can improve clinical care and long-term outcomes for rare disease patients," he says.

For Juliet, such changes feel distant but deeply necessary. Each hospital visit, each new prescription, and each school day Freda struggles through underscores the cost of delay not just in cedis, but in childhoods reshaped by fatigue and uncertainty.

Until rare diseases are recognized, studied, and planned for, many children will continue to be diagnosed in the dark, their lives shaped not only by biology, but by the absence of policy, preparedness, and timely care. ■

Burden of Care: The Psychological Toll on Rare Disease Caregivers



Caregiving often begins with quiet concern a parent notices subtle changes, unexplained symptoms, or developmental delays. What follows for many families in Ghana is not immediate clarity, but a prolonged search for answers that stretches across hospitals, specialists, and years.

For caregivers of people living with rare diseases, care becomes constant and consuming. Emotional distress, financial strain, and social isolation accumulate over time.

Yet while attention is often focused on patients, caregivers parents, siblings, and extended family members carry a psychological burden that remains largely invisible.

Alongside them are healthcare professionals who witness this struggle closely. Pediatricians encounter caregivers at the very start of the journey, while clinical psychologists support families as uncertainty deepens. Public health experts warn that without systems to support caregivers and providers alike, the cost of care becomes unsustainable.

The Long Road to a Diagnosis

Dr. Richard Bright Danyoh, a pediatrician at the Korle Bu Teaching Hospital, says delayed diagnosis is one of the most emotionally taxing aspects of rare disease care—for families and clinicians alike.

“Parents are in limbo for a very long time,” he explains. “They begin to lose interest in the health space because they are not given answers. Sometimes even health workers themselves are not too conversant with what may be wrong.”

This uncertainty stretches over months and sometimes years. As investigations stall and symptoms progress, complications set in. In some cases, Dr. Danyoh says, the window for effective medical intervention closes entirely.

“These cruel delays can lead to complications where only rehabilitation becomes an option,” he says. “As a pediatrician, there is an emotional weight that comes with knowing you are unable to offer meaningful help to the child.”

For caregivers, repeated hospital visits become cycles of hope and disappointment. Anxiety deepens as children deteriorate without explanation. Pediatricians often find themselves managing not only the child’s physical symptoms, but also the mounting distress of parents who feel abandoned by the system.

The emotional toll does not end there. When outcomes are poor, the strain intensifies.

“Sometimes, when death occurs, it reduces morale,” Dr. Danyoh admits. “You feel the weight of not having been able to change the course of things.”

Working in a health system without clear rare disease protocols further compounds this strain. Decision-making becomes uncertain, referrals are unclear, and investigations lack structured pathways.

“Support for care and a clear track for investigation is paramount,” he says. “Guidelines can only be produced if we talk about and diagnose these conditions. Until there is a concerted effort with a dedicated team working on rare diseases, policies will be difficult to formulate.”

Despite their low prevalence, Dr. Danyoh emphasizes that rare diseases carry immense significance for the families affected.

“The numbers may be few,” he says, “but they are significant for individual families. And pediatricians must be central to any national effort, along with other health workers.”

Without institutional support, pediatricians are left vulnerable to compassion fatigue and moral distress knowing what should be done, but lacking the tools to do it.

What would make the biggest difference, he says, is not abstract reform but practical change.

“Having support for diagnostics and treatment in Ghana would go a long way,” Dr. Danyohnotes. “We also need to elevate awareness across all levels of healthcare.”



Caregiving in a System Not Built for Rare Diseases

According to Samuel Agyei Waife, a clinical psychologist who works with persons living with rare diseases, Ghana’s healthcare system is ill-equipped to meet the complex realities caregivers face.

“Many rare diseases are challenging. It’s not normal,” he explains. “Our healthcare system is not set up to meet those needs.”

By the time caregivers reach psychological services, many are already experiencing depression, anxiety, trauma, hopelessness, worthlessness, and chronic stress. These struggles are shaped by unanswered questions why the condition occurred, what treatments exist, and what the future holds.

For caregivers, uncertainty becomes a form of chronic psychological exposure.

The Emotional, Social, and Financial Weight of Care

Caregiving extends far beyond hospital visits. Many caregivers abandon employment or reduce work hours. Social lives shrink. Families reorganise daily routines around appointments, medication schedules, and supervision.

“They are concerned about everything,” Waife says, “from treatment and finances to stigma, future childbirth, and whether they can live a normal life.”

Financial strain intensifies emotional distress. Rare disease care often requires multiple specialists physiotherapists, ophthalmologists, dermatologists, nephrologists—most of whom are not covered by insurance.

Over time, caregivers experience burnout not because they are weak, but because care is unrelenting.

When Professionals Become Caregivers Too

While families are primary caregivers, psychologists often become secondary ones. Supporting rare disease families demands extensive time researching unfamiliar conditions, navigating unclear diagnostic pathways, and advocating within fragmented systems.

This gap between ethical responsibility and systemic reality creates moral distress, where professionals are unable to act according to their training. It becomes increasingly difficult to support caregiver wellbeing when core medical needs remain unmet.

Vicarious Trauma in Rare Disease Care

Rare diseases are often chronic and progressive, requiring long-term engagement. Over time, psychologists develop deep relationships with patients and caregivers.

“For rare disease patients, you become part and parcel of their life,” Waife says. “They become your family.”

Witnessing deterioration sometimes rapid and irreversible leaves lasting psychological scars. Waife describes this as vicarious trauma, a

hidden occupational risk in rare disease care.

“You are watching them suffer, and you can’t do much,” he says. “It’s traumatizing.”

Why Policy Often Ignores the Human Cost

From a global public health perspective, Dr. Tomia Austin, rare disease educator and public health expert, says this psychological toll is not unique to Ghana but is magnified in countries without national rare disease policies.

“Globally, policies focus on diagnosis, treatment access, and research infrastructure,” she explains. “The emotional labour of providers is often overlooked.”

In higher-resource settings, some protections exist. The European Union encourages National Rare Disease Plans, with countries like France implementing coordinated care networks and multidisciplinary teams. These systems reduce isolation for caregivers and clinicians alike.

However, even in these settings, provider wellbeing is often treated as an afterthought. “There’s still an assumption that clinicians will simply cope,” Dr. Austin says. “Asking for help is still seen as weakness.”

What Best Practice Looks Like Elsewhere

International models show that caregiver and provider wellbeing can be protected when treated as a quality-of-care issue.

In the United Kingdom, structured clinical supervision and reflective practice are embedded in the National Health Service, giving professionals protected time to process emotional strain. Australia’s National Strategic Action Plan for Rare Diseases includes workforce wellbeing strategies such as peer support systems and psychosocial training.

Trauma-informed care used widely in Canada, the United States, and the UK emphasizes provider safety, shared decision-making, and organizational transparency. These frameworks help prevent professionals from internalizing families’ trauma.

“These models recognize that sustainable care depends on protected caregivers and supported providers,” Dr. Austin notes.

Policy Silence and Moral Distress in Ghana

In Ghana, the absence of a national rare disease policy compounds stress for caregivers and clinicians alike.

Dr. Austin describes this as a textbook case of moral distress. Professionals may know what best-practice care looks like but lack the tools to deliver it. Psychologists become emotional anchors for families while witnessing repeated suffering without systemic change.

What Ghana Could Realistically Do

According to Dr. Austin, meaningful change does not require massive investment. Peer supervision networks used in the UK could be implemented virtually. Regional rare disease focal points, as used in Australia, could improve coordination. Simplified referral pathways would reduce uncertainty for caregivers and professionals alike.

Most importantly, a national policy must explicitly protect provider mental health.

“Burnout and secondary trauma should be formally recognized,” Dr. Austin says. “Caseload standards, supervision, debriefing, and counseling access must be part of the policy.”

Who Cares for the Caregivers?

Caregivers remain the invisible backbone of rare disease care in Ghana emotionally exhausted, financially strained, and often unsupported. Pediatricians see their distress first. Psychologists carry it longest. Public health experts warn that without systemic reform, both families and providers will continue to suffer in silence.

Until caregiver wellbeing is recognized as a public health priority and until rare diseases are addressed through a coherent national policy the burden of care will remain personal, uneven, and heavy. ■



Paravez

BABUL

in The Daily Observer | Bangladesh



Parvez Babul is an award-winning health, environment, climate change journalist, and columnist gained twenty years of professional working experience in large and respected English newspapers in Bangladesh and abroad. He is a public Radio storyteller on health, climate change, environment, women and gender issues too. He has completed his higher study (MS) in Environmental Science. One of his articles/ chapters, titled: Women Climate Change and Food Security in Bangladesh has been incorporated in the text curriculum at the Connecticut University of USA.

He has been awarded international

Media fellowships from different organizations include the Canada-based IDRC / International Development Research Center.

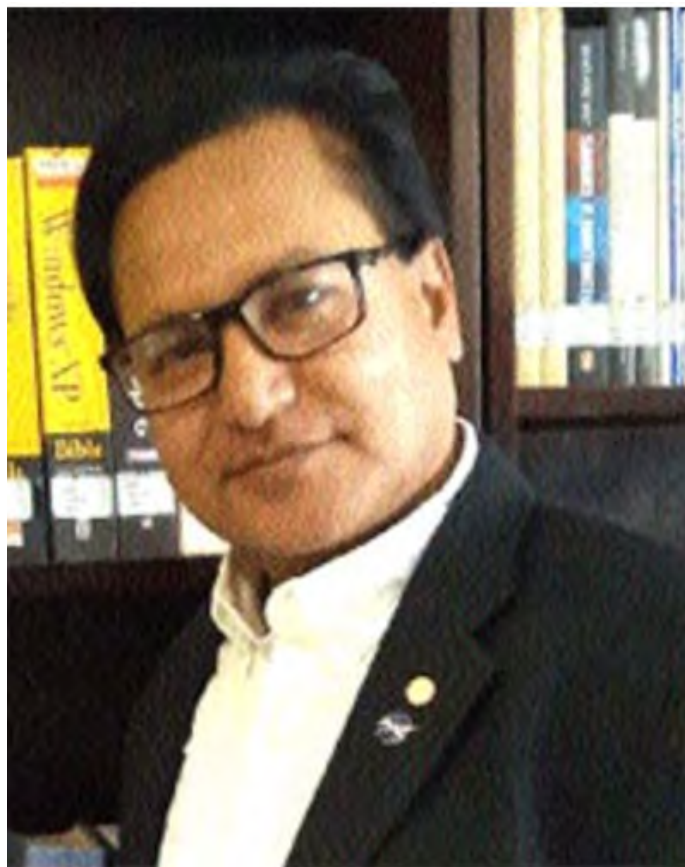
He took part and covered COP 22, of UNFCCC, Marrakesh, Morocco, 2016.

Singapore-based Asia-Europe foundation/ ASEF.

International Centre for Integrated Mountain Development / ICIMOD, Nepal.

Global Alliance for Incinerator Alternatives / GAIA, Asia Pacific, among others.

Thalassemia disease getting worse in Bangladesh



In 2021, there were approximately nine million cases of Thalassemia among children worldwide. The condition is a major global health burden, with about 300,000 to 400,000 children born annually with severe hemoglobin disorders, including Thalassemia. The majority of these affected children are born in low- and middle-income countries including Bangladesh.

Pediatric Thalassemia in Bangladesh is one of the rare diseases of children among others. It claims thousands of lives of children annually.

Status of Thalassemia in Bangladesh: Thalassemia is a major, growing public health concern in Bangladesh, with approximately 11.4 -12 percent of the population 17-22 million carrying the thalassemia gene. Roughly 6,000 to 13,000 children are born with thalassemia major annually, with over 60,000 patients currently suffering from the disease, requiring regular, costly, and often inaccessible blood transfusions.

Studies estimate that 6-12 percent of the population are carriers, with some studies showing rates as high as 10.9-13.3 percent. It is the most common hereditary blood disorder in the country, and it is considered a significant, growing health issue. Studies have shown higher rates of beta-thalassemia traits in specific areas, such as the Barisal division 8.1 percent, followed by Rajshahi 5.5 percent and Sylhet 5.2 percent.

Children are often diagnosed around 18 months of age with cases are common among school-aged children, average age around 16. Due to a lack of premarital screening and awareness, an estimated 6,000 to 13,000 children are born with the Thalassemia annually.

Regular, life-long blood transfusions and iron chelation therapy is crucial but often poorly managed due to high cost. Families often bear 74 percent of treatment costs out-of-pocket, leading to financial distress. Over 80 percent of parents struggle to find regular blood donors, and many receive no support from organized blood banks.

Researches show that children face limitations in daily activities and schooling, with many experiencing social stigma. Despite 7,000 children being born with thalassemia annually in Bangladesh, the country lacks a national prevention program.

While most parents are aware of the disease, fewer than 10 percent of mothers received prenatal diagnosis in previous pregnancies. And 40 percent of parents face social stigma. Health professionals strongly recommend mandatory premarital screening, awareness campaigns, and increasing access to prenatal diagnosis.

Rare diseases and high-mortality conditions in Bangladesh claim the lives of thousands of children annually due to poverty, limited diagnostic facilities, lack of specialized treatment.

In Bangladesh, most of the children with Thalassemia were deprived of regular blood

transfusion and medical follow-up at the district level during the COVID-19 restrictions. Over 80 percent children with Thalassemia had no access to healthcare services at all.

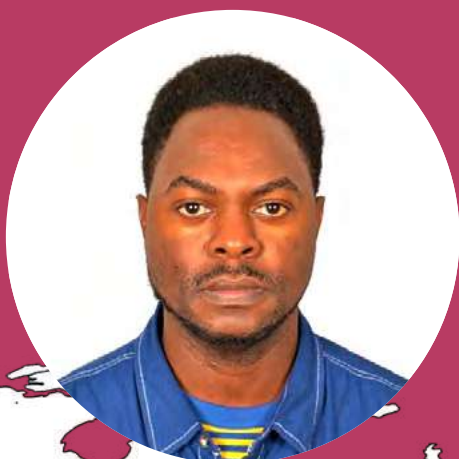


In Bangladesh, more than 70 percent of people live in rural areas. And Rural populations face significant, widespread disparities in healthcare access compared to urban counterparts. As a result, over 56 percent of global rural residents lacking essential health services. Consequently, rural residents experience poorer health outcomes, including higher rates of chronic illness and non-communicable diseases (NCDs) such as diabetes, heart disease, and increased mortality.

The COVID-19 pandemic has exacerbated this situation further due to an acute shortage of blood supply and inadequate medical care. So considering the vulnerability, Dr. Shamima Sultana of Dhaka Medical College Hospital said, “Emergency response with appropriate mitigation measures must be a priority for ensuring adequate blood supply and proper treatment during any pandemic both in rural and urban areas.”

Every year Thalassemia Day is observed globally on May 08 through World Health Organization (WHO). The global stakeholders come together to help create a world where people with Thalassemia are empowered, heard and prioritized. As the United Nations Sustainable Development Goals (SDGs) aim: No one is left behind.

It must be ensured that the challenges, needs, and rights of those living with Thalassemia are not only acknowledged but placed at the heart of healthcare policies and practices. ■



Davis

BUYONDO

in New Vision | Uganda



Davis Buyondo is an award-winning multimedia science journalist based in Uganda and working with New Vision, a government newspaper. He specialises in a spectrum of science disciplines, including Environmental sustainability, gender and social sciences, public health and biomedical research, Gender dynamics and emerging innovation systems. His journalistic works have been featured in several local and international media outlets, including New Vision Printing and Publishing Company Ltd, Uganda Radio Network, Insight Post Uganda, Africa21, China Global South Project and InfoNile. Buyondo has covered international high-level

events, including Science Forum South Africa (2017), COP29, Africa Climate Summit 2023, Kam-pala Declaration 2023, where 48 African countries signed the Kampala Ministerial Declaration on Migration, Environment, and Climate Change (KDMECC), among others. He has received several prestigious awards, including the 2023 Multimedia Award, organised by the US Embassy in Uganda and Ultimate Media Consultant (UMC), and the Best Nile Basin Media Awards 2023. He is currently mentoring emerging journalists interested in science reporting to ensure all unreported or underreported issues are given attention to transform society.

Data gaps leave Uganda's motor neuron disease patients undiagnosed

MND is a rare progressive disease that affects the nerves controlling voluntary movement leading to weakness and stiffness in the muscles of both arms and legs. This also leads to paralysis, mobility challenges, and speech difficulties. However, Amyotrophic Lateral Sclerosis (ALS) is the most commonly diagnosed form.



For Joseph Sentamu, a resident of Lutungu village in Lwanda-sub-county, Rakai district, nearly three decades passed without knowing what he was suffering from due to delayed detection and wrong diagnosis. (Credit: Davis Buyondo)

In Uganda and other developing countries, many patients with motor neuron disease (MND) remain largely invisible and wrongly diagnosed. They struggle to afford or access specialised drugs/treatment that could enable them live longer.

In the absence of national data systems with inadequate care, MND and other rare diseases/conditions are often left out of the healthcare planning, making it harder for many patients, especially in rural communities, to survive.

For Joseph Sentamu, a resident of Lutungu village in Lwanda-sub-county, Rakai district, nearly three decades passed without knowing what he was suffering from due to delayed detection and wrong diagnosis.

The 39-year-old single father first showed signs of

the strange illness at only five-years-old, stumbling frequently, losing grip on things and dropping objects.

However, the doctors at Rakai government hospital repeatedly misdiagnosed him, suspecting sickle cell disease until the disease (MND) had already crippled and left him bedridden for 16 years.

He depends entirely on his children for almost everything, including feeding, bathing, dressing, making his bed and pushing his wheelchair.

“Sometimes I miss medication and physiotherapy due to a lack of money. For now, I am really stuck,” he says quietly.

His experience reflects the wider struggles faced by patients with rare neurological disorders across Uganda. These include delayed diagnosis, incorrect diagnosis and the high cost of the only known treatment. This has made the management of rare conditions quite harder especially in rural communities.

MND, unseen rare condition

MND is a rare progressive disease that affects the nerves controlling voluntary movement leading to weakness and stiffness in the muscles of both arms and legs. This also leads to paralysis, mobility challenges, and speech difficulties. However, Amyotrophic Lateral Sclerosis (ALS) is the most commonly diagnosed form.

Doctors don't know exactly what is causing MND. However, it is believed that MND can develop due to a combination of environmental, lifestyle and genetic factors.

According to the World Health Organisation (WHO), a rare disease is often described as a specific health condition affecting one in 2,000 individuals or fewer in the general population.

Currently, there are more than 7,000 known rare

diseases worldwide affecting more than 300 million people, with 70% of these conditions starting in childhood. While most rare diseases are measured by prevalence, some are better described more precisely by incidence.

Health experts add that the number of MND patients in Uganda is unknown, and a few known patients are often misdiagnosed or underdiagnosed, not necessarily because it is rare but because it is difficult to detect and poorly documented.

As a result, it is frequently mistaken for other conditions, delaying proper treatment. In addition, the country does not have a national registry for rare neurological conditions.



Joseph Ssentamu was crippled by motor neuron disease (MND). (Credit: Davis Buyondo)

Years of misdiagnosis

For Ssentamu, the disease was already progressing quietly until it manifested when he was a child. He recalls that his struggles began after a severe malaria episode that left him in a coma for two weeks.

Shortly after, the weakness in his arms, legs and feet. “I would struggle with everyday tasks that others found easy,” he recounts. Little did he know that these were early signs of a rare condition that would later be diagnosed as MND.

Ssentamu has been visiting Rakai Government Hospital, where the doctors could not detect or treat the condition all this time. Over the years, he would only receive painkillers and immune booster, which could not do much about the condition.

It wasn’t until 2018, when he was referred to Masaka Regional Referral Hospital for further attention. The doctors diagnosed MND. By then, the disease had taken much of his strength. “They said that my condition might not have deteriorated severely if it had been detected,” he recounts.

By his early 20s, he could no longer sit or stand unaided. He dropped out of school in Senior Five (S.5) in 2009. “My dreams of becoming a lawyer and a successful businessman were shuttered,” he says.

Cost of living with MND

Currently, there is no cure for MND. But the treatment focuses more on symptom control, slowing progression and palliative care for those in late stages of the condition, like Ssentamu.

The only drug known for extending survival by slowing progression is Riluzole. But in Uganda, it is not only scarce but also too expensive for underprivileged families.

At least each month, Ssentamu needs above Sh1,000,000 (approx. USD300) for his medication, more than what many households earn.

“Each tablet costs sh18,000. I have to take one in the morning and another in the evening,” he says, adding that the condition is becoming too expensive to manage alongside his family.

New Vision has established that Riluzole is not stocked in Ssentamu’s nearby health centres or even at the Regional Referral Hospital in Masaka district. His nearest and most reliable access point is Kampala, more than 165km away.

“I can only afford drugs (Riluzole) through social mobilisation,” he says. “When I get money, I stock medicine for three months to avoid frequent travels,” he adds.

Ssentamu further says he also attends physiotherapy to help reduce pain and stiffness but he has to cater for his helper in all these visits. “As a result, everything from transport, lodging and food doubles,” he recounts.

With medication, Ssentamu’s pain from stiffness of the muscles and pain or discomfort significantly

reduces. But when he misses a dose, his condition worsens. “My body aches, the weakness spreads faster, and speaking becomes harder,” he explains.

Clare Naluze, a pharmacist in Masaka, who works closely with patients with neurological conditions, confirmed that the drugs that treat rare diseases are not always stocked because they are imported in small quantities, unlike those treating common diseases and conditions.

“These drugs are not procured in large quantities, yet there is no government subsidy to help lower the cost,” she says.

However, Uganda lacks a national health insurance policy that could help make treatment affordable and improve access to healthcare, leaving most patients to cover medical bills out of pocket.

Impact on family, Children

Ssentamu cannot work, yet he has a family, including orphaned dependents to look after. He relies on handouts from well-wishers. Still, his eldest son was forced to leave school to take care of him and his siblings.

“My children are psychologically stressed because they have never seen me walking,” he says. They are also stigmatised as the community members believe that he was bewitched and the condition is a curse punishing the family.

For Ssentamu, he has lost his privacy since it is his children who bathe and dress him. “To avoid being overwhelmed, they take turns. I know I am a burden to them, but they do not show it,” he explains.

Experts speak

Experts have explained that MND can affect multiple parts of the nervous system and becomes common with age, rising around 40 and peaking in the 80s. They say that the risk factors include cigarette smoking and pesticides being spread on crops.

According to Dr. Martin Kaddumukasa, a Consultant Neurologist and Physician at Mulago National Referral Hospital, there is no data for MDN patients in Uganda and there is no specific treatment currently available.

MND Diagnosing Challenges

Dr. Kaddumukasa explains that MND is difficult to diagnose because it does not follow a clear and straightforward pattern. It also takes a long time before the symptoms appear.

Usually, he adds, patients often start with weakness in the hands, legs and feet, and muscle wasting (shrinking), signs common in malaria and many other diseases or conditions. “This makes it hard to screen for MND among thousands of other possible diseases,” he says.

Kaddumukasa says that diagnosing MND requires specialised expertise and not all doctors are trained to do it. While some patients receive care, the treatment is so complex and comprehensive.

It involves neurologists who treat the brain and nerves, physiotherapists when weak muscles need exercises, speech therapists as the muscles that lift the tongue become weak, respiratory physicians because the diaphragm also becomes weak, dieticians, nurses and social workers. “Palliative care is a good.

“To reduce the chances of developing MND, we must avoid smoking, prevent exposure to pesticides by wearing a mask, limit exposure to metals such as Lead and excessive copper, manganese and others,” he advises.

While there is no cure, he says, palliative care would play a great role in easing symptoms, improving the quality of life, providing emotional and psychological support, supporting family and caregivers, and other symptoms, improving comfort and dignity.

Dr. Iddi Matovu is a public health specialist at Kitovu Mobile, a not-for-profit, faith-based organisation under the Diocese of Masaka, where he works with people with rare diseases in need of palliative care.

He explains that electrolytes like calcium, magnesium, sodium, and potassium help muscles to move and relax normally. Without these minerals, he adds, the muscles can become stiff and weak. “In some serious cases, they may shrink or even become paralysed,” he says.

A system not built for shortage

Uganda's health system has made significant gains in infectious diseases and maternal health, but rare diseases remain largely invisible in policy planning.

“Currently, we (Uganda) don't have a formal classification of rare diseases,” says Dr. Diana Atwine, the Permanent Secretary of the Ministry of Health (MoH).

This means that rare neurological conditions are not clearly identified, recorded, or given priority within the country's healthcare system. As a result, diseases such as MND are often grouped with other illnesses, making it difficult to measure how many people have them to support planning for specific services for them.

Health experts further explain that since rare disease patients do not show up in statistics, they are not planned for. This gap makes it harder to carry out early diagnosis, secure funds, procure specialised drugs and policy development.

Therefore, without official classification, these rare conditions remain largely invisible during national healthcare planning, leading to delayed diagnosis, delayed treatment, misdiagnosis and limited access to specialised care.

Palliative Care in Uganda

The government does not provide home-based care services for people with rare diseases, especially those with progressive neurological conditions like MND. A few palliative care services that exist are disproportionately distributed and mostly focus on cancer and HIV patients.

Palliative Care Association of Uganda (PCAU) supports the work of MOH that aligns with its strategic objectives. PCAU works with the ministry to formulate policies and guidelines to enhance the scale-up of palliative care service provision and integration in the health care system.

In partnership with the Ministry of Health, PCAU conducts periodic and on-schedule visits to districts for mentorship and support supervision for palliative care providers.

The visits held jointly with the Ministry of Health

are intended to address the knowledge, skills, and attitude needs for continued quality improvement of palliative care services.

“I have not been able to access these services. I wish there could be an opportunity,” he states.

New drug brings hope

A groundbreaking drug, Tofersen is giving renewed hope to people living with MND, not only in the United Kingdom but around the world. The drug, developed by Biogen, has been shown to slow and, in some cases, even stop the worsening of MND in people with Superoxide Dismutase 1 (SOD1).

Patients like Ssentamu remain optimistic that the ongoing research is helping patients to get closer to the life-extending treatment and nearing a cure. “Accessibility and affordability remain the biggest challenges, especially for patients in developing countries like Uganda,” he says.

Global contrasts

In developed countries, people with MND normally access specialised clinics under the supervision of teams of medical experts and physiotherapists in addition to specialised medication. However, in Uganda, and many parts of Sub-Saharan Africa, very little research has been done on MND.

The patients here are rarely part of international medical trials. Therefore, doctors often use treatment guidelines from other countries yet they may not fit local conditions.

Recent global research is giving much hope for a better understanding and treatment of MND. However, scientists are working on new tests that may help to detect the condition/disease much earlier. One study found that a simple blood test could identify people with MND with about 97% accuracy.

This raises hope for quicker and less invasive diagnosis in the future.

In addition, researchers are also studying blood tests that look for proteins and other signs that change years before symptoms start, which could help detect MND much earlier. ■



Robi
CHACHA
in K24 TV | Kenya



My name is **Robi Chacha**, a dynamic journalist, TV host, and digital content creator passionate about amplifying community voices and driving conversations that inspire social change. As the host of a mid-morning television show and a growing YouTuber and podcaster,

I cover impactful stories on health, gender, and social justice, spotlighting communities often left unheard. Beyond the screen, Robi is a media trainer and communication consultant who empowers professionals to engage the public with clarity, confidence, and purpose.

Raising A Child With Turner Syndrome | Beyond The Diagnosis | Seven Years Of Grace



Video [here](#)

Turner Syndrome | Rare Genetic Condition



Video [here](#)

Alagille Syndrome | My Son Is Living With This Rare Disease



Video [here](#)

Narcolepsy : Living With A Sleep Disorder | Rare Disease Day



Video [here](#)



Charlie CORDERO

Freelance | Colombia



Charlie Cordero (Barranquilla, 1990) is a Colombian documentary photographer based in the Caribbean and a member of the Rejo Colectivo. His work spans personal projects, assignments for NGOs, and collaborations with international media. Themes such as social justice, climate change, and Caribbean identity recur throughout his visual storytelling.

He has contributed to publications including The New York Times, The Guardian, Geo, The Washington Post, Vogue, El País, and National Geographic. Cordero has been a fellow of the Gabriel García

Márquez Foundation (2020), National Geographic (2021), VII Academy (2022), PhotoVogue (2024), and Vital Impacts (2025). His work has been exhibited internationally, including Sin Fronteras (PhotoEspaña, Madrid, 2015), Coca – La Guerra Perdida (Centro de la Imagen, Mexico, 2021), Inside the Curve (National Geographic, 2022), Square Mile (VII Foundation, 2024), and Photoville (New York, 2024).

He is currently developing a long-term project about Santa Cruz del Islote, the world's most densely populated island, and its struggle against climate change.

The children of Huntington's: the orphan disease of the Caribbean

In Yeimy Lozano's life, there are no clear distinctions between love and pain. She is 45 years old and her body is covered in wounds. Sometimes, when she speaks, she pauses to show one: "Look, Eduardo did this to me last night," and she points to a bruise on her leg or a scratch on her chest.

These are the marks left by her children, Eduardo and Yeideline. He is 30 years old and she is 27. They live with their mother in the San Isidro neighborhood of Barranquilla, where they migrated from Venezuela nine years ago. Yeimy doesn't blame them for the beatings: "They are just children, my poor little ones."

Yeideline and Eduardo Lozano are Huntington's patients, a neurodegenerative disease that impairs the brain's ability to control movement, including speech and eating. It also alters behavior, with daily

outbursts of anger that Yeideline tries to control, sometimes at the expense of her own health.

Huntington's disease is considered a rare, inherited, and currently incurable condition that affects a very small percentage of the population in most countries. It arrived in South America about 200 years ago, brought by Europeans, and spread through large families in the Caribbean regions of Colombia and Venezuela. These two countries are home to the two largest populations in the world living with the disease.

The most distinctive feature of Huntington's disease is involuntary movements. Eduardo and Yeideline's legs and arms never stop moving. They swing in erratic, unpredictable directions, in a constant back and forth that makes it difficult for them to walk or hold objects with their hands.



Yeimy Lozano and her son Eduardo Lozano. Eduardo inherited the disease from his father, Eduardo Goteras. Photo: Charlie Cordero.

In Venezuela, the disease was known as St. Vitus' Dance. During Yeimy's childhood in Maracaibo, there were rumors of an illness that was passed down from parents to children, and that over generations had settled in a fishing village near the city called San Luis, a neighborhood in the municipality of San Francisco, in the state of Zulia.

The people of San Luis were feared and discriminated against. They were popularly known as "Sanviteros," and many tried to hide their origins. It wasn't difficult: Huntington's disease doesn't manifest immediately. If one parent has it, their children have a 50% chance of inheriting it, but in most cases, symptoms begin after age 20.

The disease waits, dormant, like a coin tossed in the air, its secret verdict already decided at birth. It was written in the genes of Eduardo Goteras, the father of Yeimy's children, that he would develop Huntington's disease around the age of 27, and so it happened. But when they met in the 1990s, he was 20 years old and lived with the hope that he hadn't inherited it.

Yeimy was 16. She studied in the El Manzanillo area of San Francisco. After school, she would often go for ice cream at a shop owned by Eduardo's paternal grandmother in the neighborhood. They met and fell in love. He didn't tell her that he was from San Luis, nor that by then his mother was already showing the first signs of Huntington's disease.

Yeimy and Eduardo's first child, whom they named after his father, was seven months old when she began to suspect something was wrong. "Eduardo, why does your mother move like that?" she asked him once. He replied that it was Parkinson's, without making a big deal of it, and Yeimy felt sorry for her mother-in-law who had gotten sick so young.

But the fears didn't go away. "You're not from one of those families with St. Louis disease, are you?" Yeimy asked a few days later, and Eduardo denied it again, to no avail. The more time they spent together, the more clues appeared about his family's secret. Other uncles and cousins with unusual behavior, other questions evaded.

Then came the confession, then the panic. Yeimy wanted to have surgery to prevent having more children, but when she went to the doctor, she received the news that she was pregnant with

Yeideline. She was 17 years old and had two children, each with a 50% chance of developing Huntington's disease.



Yeideline Lozano, daughter of Yeimy Lozano. She developed the disease three years ago and has been cared for by her mother ever since. Photo: Charlie Cordero.

"You cheated on me. If you had any conscience, you would have thought about your children," Yeimy told Eduardo when he tried to leave her. She went to live in another house in Maracaibo with her children. He stalked her. He snuck into her room, tried to burn her with boiling water, shot her in the foot, and punched her, leaving her with a deformity in her mouth. She still bears the scars, alongside the recent wounds from the fits of rage that her illness triggers in her children.

Eduardo, the father, died of Huntington's disease years later. His children, Eduardo and Yeideline Lozano, spent their childhood waiting, not knowing if they had inherited the disease. Yeimy watched them grow up haunted by a fear that never truly left, a silent companion. The fear was with her in the bedroom when she prepared her children for bed, it accompanied her in the morning when she took them to school, and it remained there in the afternoon when the children returned home.

When she felt they were both old enough, she told them the truth: that one day, one of them might get sick like their father. Yeideline took it with humor: "When I regain my mobility, I'm going to put a radio on my shoulder so they think I'm dancing," she replied.

It was harder for Eduardo to accept. He hadn't even turned 10 and he already had to deal with a destiny over which he had no control. He hugged his mother and made only one request: "Never leave me alone."

“One does not travel alone, one travels with one’s genes”

More than a millennium ago, on Christmas Day in 1021, the parish priest of the Saxon village of Kölbigk cursed 18 people who were “dancing obscenely” in front of the church door. This is the first documented record in medieval chronicles of the “dancing movements,” or chorea, that we now associate with Huntington’s disease.

In her article, “[Huntington’s Disease: A Journey Through History](#),” physician Esther Cubo records this episode and notes that similar incidents occurred in the following centuries. Hundreds of Huntington’s patients were accused of witchcraft or demonic possession. During this time, the belief also spread that people could be cured by praying to a 4th-century Sicilian saint, Saint Vitus, whose name eventually became the popular name for the disease.



Rosa Salgado and her husband, Luis Martinez. Rosa was diagnosed with Huntington’s disease a decade ago. They live in Algarrobo, Magdalena, one of the towns with the highest concentration of cases in Colombia. Photo: Charlie Cordero.

The cause of Huntington’s disease remained a mystery for centuries. The first step toward understanding the disease was the detailed description made by the American physician George Huntington in 1872. One hundred years later, Nancy Wexler’s work in Venezuela finally unlocked the secret of the genetic mutation.

The research focused there due to the extraordinarily high concentration of patients in the Maracaibo area. While the rest of the world has an average of 2.7 Huntington’s disease patients per 100,000 inhabitants, in that Venezuelan region, [one in ten people](#) has the disease.

[Wexler’s team](#) found the specific gene that causes Huntington’s disease, located on chromosome 4. By studying it, they discovered that patients have excessive repetitions of nitrogenous bases in that gene. It’s an alteration in the genetic code, like an incorrect instruction within the cells.

“Its effect is a degenerative spiral that progressively atrophies areas of the brain responsible for controlling movement, behavior, and cognitive abilities, leading to dementia,” explains Sonia Moreno, a researcher with the Neuroscience Group at the University of Antioquia. Ultimately, it causes

death approximately 15 to 20 years after the onset of symptoms.

That “error in the system” is what Huntington’s patients inherit from their parents; the mutation that traveled for centuries along the paths of geography and genetics: transmitted through generations, transported on ships that crossed the Atlantic from Europe, until concentrating in the Caribbean region of Colombia and Venezuela.

“You don’t travel alone; you travel with your genes and the diseases you’re going to have,” says Sonia Moreno. There are indications that the cases in Colombia and Venezuela stem from what is known as a founder effect: a patient with the disease who has many children who inherit it and spread it throughout entire populations.

In Venezuela, the founder is believed to be a Spaniard named **Antonio Justo Doria**, who arrived in the Maracaibo region in the 18th century. In Colombia, **documents** point to a Frenchman, Luc Etcheberry, who settled in Juan de Acosta, Atlántico, around 1842, where he started a lineage that today extends throughout the Caribbean.

It was a lineage that spread without people even being aware that they carried the disease. In Algarrobo, Magdalena, one of the hotspots of cases in Colombia, most inhabitants had never heard of Huntington’s disease until just over a decade ago. Each family had its own explanation for the disease that reappeared in every generation.



Inés Carmona, 54, lives in Algarrobo, Magdalena. Her father had Huntington’s disease, and some of her siblings and nieces and nephews inherited it. She has not had any symptoms. Photo: Charlie Cordero.

Inés Carmona, 54, remembers her father, José Cardeno de Ávila, ill and almost paralyzed by

Huntington’s disease, appearing every now and then with a new sibling. She spent her childhood on a path outside Algarrobo, watching her family grow as her father’s health deteriorated.

Without knowing the words to name it, Inés sensed how with each birth the roulette wheel of genetics began to spin: “My mom told him she didn’t want any more children, but my dad was a sexist. I was a child and I thought to myself, I didn’t tell anyone, that we were all going to get sick from then on.”

| *“Why are you moving like that?”*

It was 12:30 in the afternoon when Eduardo Lozano, Yeimy’s eldest son, felt himself losing control of his body. He was 14 years old. He collapsed in the middle of the street, next to his classmates from the seventh grade of the Vicente Lecuna High School in Maracaibo. Eduardo tried to get up, but fell again. His classmates, frightened, kept asking him the question he didn’t want to answer: “Why are you moving like that?”

He never went back to school. He spent the next year hiding in his house. “I don’t want anyone to see me. I don’t want to, I don’t want to,” he kept repeating to his mother. There, protected by the walls, Eduardo learned to walk for the second time. He adapted to the arrhythmic and incessant movements, to the arms and legs that had stopped obeying him.



Eduardo Lozano and his mother. During the early years of his Huntington’s disease, Eduardo was still able to perform some daily activities, but his condition has worsened over the past year. Photo: Charlie Cordero

A few years earlier, he had wanted to be a professional baseball player. “He was a tremendous pitcher,” Yeimy says proudly. She remembers

that some scouts from the United States saw him and spoke with her about Eduardo playing in that country: “But I told them about his father, and they were discouraged. If I had kept quiet, maybe Eduardo would have had a little more fun.”

Because of the age at which the symptoms began, Eduardo’s illness is classified as juvenile Huntington’s disease. These are the rarest cases, appearing before the age of twenty. The more repetitions there are in the specific gene that causes Huntington’s disease, the faster the symptoms appear.

“In that sense, the genetic inheritance from the father is more problematic than that from the mother: the length of the repetitions can be twice as long if it is transmitted through the male line,” explains Sonia Moreno, from the Neuroscience Group of the University of Antioquia.

Yeidelina, Yeimy’s second daughter, showed no symptoms during her adolescence. She cared for her brother, Eduardo, who already had Huntington’s disease, while her mother worked during the day. At 15, Yeidelina became pregnant. The father was one of her neighbors and childhood friends, Eugenio, and they had a daughter whom they named Eugenelys.

“Thank you, Father,” Yeimy thought a few months after giving birth, when she confirmed that Yeidelina did not exhibit the symptoms of Huntington’s disease. One of the most widespread beliefs among the families of patients is that, for women, pregnancy triggers the disease.

According to Sonia Moreno, there is no evidence that giving birth influences the onset of symptoms, but she believes that “it may be part of the beliefs that become popular to explain something as complex as a genetic disease. In Antioquia, in the case of Alzheimer’s, many people believed that the disease was caused by touching a tree in the center of town.”

It’s difficult to go through life without explanations that offer comfort. Yeimy has her own to process why Yeidelina’s illness awoke at 24: “I told her not to have any more children, to stop smoking, not to go live in Ecuador. She didn’t listen to me.”

Yeimy and her family had migrated to Colombia in 2017, forced out by Venezuela’s social and economic crisis. Yeimy could no longer afford medicine and food for her children. They settled in Barranquilla, but Yeidelina never liked the city. She separated from Eugenio and decided to travel to Guayaquil with her new partner, leaving Eugenelys with her grandmother, who has raised her ever since.



Yeidelina Lozano at her home in Barranquilla. Photo: Charlie Cordero.

After some time in Ecuador, Yeidelina began to experience symptoms of Huntington’s disease. At first, she tried to hide the signs. She stopped turning on her camera when she spoke to her mother on the phone so that her mother wouldn’t notice the involuntary movements.

Yeimy had to decipher what her daughter wasn’t telling her. She did it through the photos she posted on social media. In those still images, she saw the signs of the illness. “You could see it in her eyes,” she recalls.

Those who have lived with Huntington’s disease agree that it first manifests in the eyes. The disease affects the entire motor system, linked to movement, including the facial muscles. Most patients have a particular, dreamy expression; a calm way of looking that contrasts with the constant restlessness of their bodies.

After speaking with some acquaintances, Yeimy confirmed her suspicions: her daughter had the disease. She also learned that Yeidelina’s partner had abandoned her in Ecuador as soon as she began showing symptoms. “He kicked her out, telling her he was ashamed to be with her. I went to Ecuador to find my girl,” she says.

He took a bus to the border and returned to Barranquilla with her. Since then, they have all lived together in a house in the San Isidro neighborhood, along with Yeimy's Colombian friend, Susan. She is the guarantee that they can keep the house rented. As in other Colombian cities, many landlords in Barranquilla refuse to accept Venezuelan tenants.

But the Yeideline who returned to Colombia was not the same. She was angry. Eugenelys, who barely had any memories of her mother, found herself facing a distant woman, prone to sudden fits of rage, tormented by involuntary movements that continued to gain control over her body.

"They fight like two sisters, over anything: because Eugenelys doesn't like her smoking, or because Yeideline gets jealous if I buy something for the girl," says Yeimy.

Sometimes, Yeideline tries unsuccessfully to reclaim her authority. "Listen to me, I'm your mom," she says, struggling to speak because of the movements caused by Huntington's disease. Eugenelys always responds the same way: "My mom is Yeimy."



Eugenelys, daughter of Yeideline Lozano. She has been living with her grandmother, Yeimy Lozano, in Barranquilla since she was 2 years old. Photo: Charlie Cordero.

In many cases, Huntington's disease causes a rift within families. The illness disrupts traditional roles for parents, children, and siblings, which can lead to emotional isolation and increased conflict between them, as [several](#) neuropsychological studies have shown.

For Yeimy, her children's illness has not only plunged them back into a stagnant childhood, but has also forced her to bear their resentment. Upon

returning from Ecuador, she endured her daughter's rage for several months, simmering like a bitter brew. The worst part wasn't the pushing or the scratching, it was the words. At that time, Yeideline could still articulate them clearly.

"It's your fault my husband left me. It's your fault I'm like this. It's your fault. You brought me into this world."

"She turned out just like me, the same color as me."

Maripaz is nine years old and is playing jump rope with her cousins on a street in Algarrobo, Magdalena. Her mother, Diana, watches her from the front yard and calls out: "Maripaz, come here." The girl takes one last leap to dodge the rope and runs over. She has her mother's tan skin and her father's curly hair.

"What is the name of your grandmother Rosa's illness?" Diana asks softly.

"I don't remember," she replies.

"Remember," the mother insists. "His name is Huntington." The girl looks at her, not knowing what to say.

From afar, a child's voice calls Maripaz back to the game. Her mother signals to her, and she runs off. The children raise the rope higher and higher, but Maripaz's jumps are precise and controlled; she has no trouble reaching it.

When she was younger, Maripaz used to complain to her mother about her skin color. "She would say to me, 'Mommy, why didn't you make me dark-skinned like my dad?' And I would answer that she turned out just like me, with my skin color. But she insisted that no, she wanted his."

Parents choose their children's names. Everything else is beyond their control: skin color, eyes, voice, the talent for jumping rope, genetic code. No one chooses what they inherit. Genetic inheritance is always an involuntary movement.

"What will become of my life?"



Diana Martínez shows some family photos at her mother's house, Rosa Salgado. Photo: Charlie Cordero

Yeimy sells aphrodisiac juices made from borojó and chontaduro fruit outside her home in Barranquilla. Every day she brings out a cart full of bottles with logos shaped like a bull, a lightning bolt, a fully charged battery, and colorful signs promising sexual or physical feats.

It's noon on Tuesday, and she's sold 9,000 pesos worth of goods all morning. "It doesn't matter. Right now, I'll ask Susan to watch the kids for me, and I'm going downtown to sell trash bags," she says.

Each month, she must come up with 900,000 pesos to pay the rent and expenses for the house she shares with her friend. In addition, she has to find money for Eduardo's diapers, Eugenelys's school fees, Huntington's medication, and co-payments for doctor's appointments, all while caring for her two sick children and her 12-year-old granddaughter.

Huntington's disease is **considered a high-cost** illness, not only because of the treatment of involuntary movements, but also because it requires care from a wide range of areas: neurology, psychiatry, nutrition, nursing, physical therapy, among others.

Expenses often exceed the income of most affected families. The most comprehensive characterization of Huntington's disease cases in Colombia comes from the international Factor H Foundation, which has been supporting families in three countries with outbreaks of the disease—Venezuela, Colombia, and Peru—since 2012.

It is difficult to estimate an exact number of people with Huntington's disease. Most patients never receive a genetic test that would include them in official statistics. In total, Factor H has identified 75 patients with a medical diagnosis in Colombia and 501 people "at risk." But many of these latter cases are part of an underreporting of the disease. Eduardo and Yeideline, for example, never had a laboratory test.

Data from Factor H also indicates that 84% of people affected by Huntington's disease in Colombia live in poverty or extreme poverty. This is another consequence of the disease. "Because of the ages at which it appears, and the limited time caregivers have to work, the disease ends up condemning families to progressive economic decline," says Brayan Hernández, legal advisor for projects at Factor H.



Yeideline Lozano at her home in Barranquilla. Her mother, Yeimy Lozano, sells energy drinks and aphrodisiacs. Photo: Charlie Cordero

Someone knocks on Yeimy's door; it's a customer. She gets up and, while preparing an energy juice, talks about her exhaustion. "I have no one. I have to solve everything on my own. It's been like this for 15 years. Sometimes I just want to sleep. Because I'm tired. I have to eat in secret. The illness makes Eduardo anxious if I eat in front of him."

Her granddaughter, Eugenelys, appears in the doorway. The little girl makes excuses to approach, curious about strangers. Her grandmother's face lights up. In the last hour, she has mentioned at least four times that Eugenelys graduated with honors from fifth grade last year.

"She is my friend, she is my sister, she is everything to me," says Yeimy.

Her cell phone is full of photos and videos of her granddaughter: Eugenelys in her school carnival queen costume; Eugenelys in her graduation gown

and with her diploma; Eugenelys during a dance performance, her steps precise and agile; Eugenelys during a biology presentation about the nervous system.

"She built the model of the brain herself, with all the insides, right, my love?" Yeimy says. She asks the girl to repeat the presentation. She hesitates at first, but her grandmother's confidence encourages her.

He speaks in an expository tone: "This is the brain, and it has different parts. This is the happy part, and this is the boring part. When you're bored, your brain tells you: get active, you have to do something. Dance, draw, whatever. And then the brain becomes happy."

Eugenelys doesn't like to talk much about Huntington's disease. Ever since she can remember, she has seen her uncle, and later her mother, as possible reflections of a future she prefers not to speak of aloud. Only once, when she came home from school and saw Yeimy bent over in pain from a shove by Eduardo, did she say between sobs, "Mommy, I don't want that to happen to me. Look what they did to you. What will become of my life?"

"Think about it, that's not going to happen to you. You're a girl who goes to church. Think about it, that's not going to happen to you," Yeimy told her granddaughter that night. For her, the threshold after which Eugenelys will be safe is 15 years old. She trusts that if she reaches that age without symptoms, she won't develop the disease. She knows that hope is the only comfort when the coin of genetics, invisible and resounding, hangs in the air. ■



Márcia

DEMENTSHUK

in Agência Pública | Brazil



Graduated in Social Communication - Journalism from the Pontifical Catholic University (RS), specialized in Communication and Marketing from the Higher School of Advertising and Marketing-RS, **Márcia Dementshuk** holds a professional master's degree in Journalism from the Federal University of Paraíba. About 20 years of experience as a journalist. Worked for the print newspapers Diário de Pernambuco, O Norte, and Correio da Paraíba. Dedicated for 10 years in the fields of science, technology, and the environment. She is the author of the

book “Birds Fly in a Flock” (about the early days of the Internet in Brazil). Is currently a freelance journalist. Produces and hosts the weekly column/podcast “Horizons of Innovation”. Collaborates with Eco Nordeste Content Agency and is a member of the Pública's Fellows. Winner of the 2014 Petrobras Journalism Award in the Social and Environmental Responsibility Reporting category - National. Second place in the 2021 Semear International Journalism Award, Print category.

Rare Disease Patients in Brazil May Total 10 Million

There is a guiding principle in the realm of rare diseases: “Rare diseases are rare, but patients with rare diseases are very numerous.” Even if each specific condition affects only a few individuals, their collective impact is vast. Up to 10 million Brazilians may face a rare disease at some point in their lives, a figure driven by the existence of between 6,000 and 10,000 different types of these conditions.

The France-based Fondation Ipsen considers the more than 350 million rare disease patients worldwide to be “invisible.” Identifying these individuals within healthcare systems remains a significant challenge, and approximately 95% of them have no available treatment. In the United States, the National Organization for Rare Disorders (NORD) estimates the patient community at 30 million people — roughly 10% of the population — and notes that the healthcare system is not currently designed to provide adequate assistance. This reality is mirrored across Africa, Asia, Oceania, and Europe.

Advocating for this cause, the European Organisation for Rare Diseases (Eurordis), which represents over 1,000 patient organizations across 77 countries, proposed in 2008 that February 29 (during leap years) or February 28 be designated as “Rare Disease Day.” The suggestion gained global traction and is now observed worldwide.

In Brazil, medical and patient associations are preparing awareness campaigns. In João Pessoa, the Liga Acadêmica de Genética Médica da Universidade Federal da Paraíba will host a roundtable discussion titled “From Invisible to Recognized” on February 27 at 2:30 PM at the Centro de Ciências Médicas. Additionally, the Associação Paraibana de Doenças Raras (Aspador) is promoting the “Raros Run 2026,” starting from the Busto de Tamandaré on Sunday, March 1, at 6:00 AM. Registration is available at www.race83.com.br.

Specialized Center in João Pessoa is the First of its Kind in the Northeast

In Brazil, a disease is classified as rare when it affects up to 65 people per 100,000 individuals — a benchmark established by the Ministério da Saúde. The public agency recognizes the existence of approximately 5,000 different types in the country. However, determining the exact number of people suffering from a rare disease in Brazil is a complex arithmetic challenge, as data within the Sistema Único de Saúde (SUS) is not yet properly categorized to identify these individuals.

Natan Monsores, General Coordinator of the Coordenação-Geral de Doenças Raras (CGRAR/MS), noted that to measure this population, the agency has adopted projections from Orphanet, an international data platform. “Between 2.5% and 5% of the Brazilian population will suffer from a rare disease at some point in their lives,” he stated. These figures represent between 5 and 10 million Brazilians.

To reach this population, the SUS established a specific policy in 2014: the Política Nacional de Atenção Integral às Pessoas com Doenças Raras (PNAIPDR), enacted through Ministério da Saúde Ordinance No. 199/2014. This regulatory milestone paved the way for the accreditation of specialized reference centers, research funding, the integration of new medical technologies and treatments, the availability of genetic testing, and the creation of mechanisms to identify patients within the system.

As a result of these efforts, the City of de João Pessoa inaugurated the Centro de Referência Multiprofissional em Doenças Raras in 2022. The facility is the first in the Northeast and the second in the country to offer this level of specialization.

Saionara Araújo, the center’s General Coordinator, explained its operations: “It functions as an outpatient clinic tailored to the needs of rare disease patients. We have 13 specialists on staff. While Brazil has fewer than 500 geneticists, we have two here. We offer space for physical and psychological rehabilitation, social assistance, and even a play area

designed for children.” Most of the center’s patients are pediatric.

As of December 15, 2025, 1,199 people had undergone screening; of these, 231 cases are currently under investigation, with a total of 216 different types of diseases recorded, according to the center’s Epidemiological Profile. Other accredited services in Brazil follow this model or are based in hospitals where more complex treatments are administered.

Diagnosis Takes Over Four Years on Average

One of the regular patients at the Centro de Referência em Doenças Raras de João Pessoa is 12-year-old Luís Eduardo de Souza. His mother, Vanderléia Rocha de Souza, shared that he has Duchenne Muscular Dystrophy, a condition that causes the necrosis of muscle fibers. When her son began showing symptoms — such as imbalance and lack of muscle strength — at age six, Vanderléia embarked on a “diagnostic odyssey,” a grueling journey through clinics and laboratories to understand what was happening.

A pediatrician eventually suspected the condition and advised her to seek a specialist. “It took me more than three years to get a pre-diagnosis; I searched the internet and only found despair. Finally, I found a group of Duchenne mothers in Paraíba. They reassured me, comforted me, and guided me toward the right doctors.” According to studies by Rede Raras — a Brazilian research initiative coordinated by geneticist Têmis Félix to gather epidemiological data — the average time to obtain a diagnosis is four and a half years.

Vanderléia noted that there are 20 families affected by Duchenne in Paraíba, and she also participates in a national group of nearly 300 families. While Luís Eduardo knows there is currently no cure, he undergoes weekly physical therapy and consults with a psychologist and nutritionist at the Reference Center. He also has semi-annual follow-up appointments at the Hospital Universitário Lauro Wanderley with neurologists, pulmonologists, and other specialists as needed.

In João Pessoa, caregivers and families who dedicate themselves 24 hours a day to those in need also rely on the Associação Paraibana de Doenças Raras (Aspador). On average, the association serves 900 people per month.

SUS Negotiates Multi-Million Dollar Deal for Gene Therapy for SMA Patients

The implementation of the Rare Disease Policy gained significant momentum in 2023 with the creation of the Coordenação-Geral de Doenças Raras (CGRAR/MS). According to Natan Monsores, the number of annual patient consultations jumped from 30,000 to 70,000, and the network of reference services more than doubled, growing from 16 to 37 across 16 Brazilian states.

Management is focused on initiatives such as improving neonatal screening to detect rare conditions early in childhood. Differential payment for consultations encourages investigative and diagnostic work: while a standard SUS consultation pays approximately R\$ 20, a Rare Disease consultation pays R\$ 800. Each patient can be billed four times a year, which also helps fund high-cost complementary exams. Total annual investments have reached R\$ 4 billion.

The integration of new technologies has been a major milestone. The SUS already offers and is currently refining “Exome Sequencing,” a high-complexity genetic test. Since September 2025, it has also provided gene therapy for Spinal Muscular Atrophy (SMA). While the treatment costs approximately R\$ 12 million, the SUS closed a risk-sharing deal with the supplier for R\$ 7 million. Under this agreement, payments are made in installments based on patient outcomes over five years.

Since the program began, 15 children have received the single-dose therapy and are currently being monitored. Brazil is the sixth country in the world to offer this treatment through its public healthcare system. ■

Pain and Hope: A Firsthand Account of Rare Disease Policy within the SUS

In late May 2022, at the age of 50, my night's sleep was suddenly interrupted by excruciating, paralyzing pain. What began as extreme fatigue evolved into a sensation of “thousands of tiny shards” piercing my feet, violent cramps in my legs and feet, and joint pain. The first suspicion, chikungunya, triggered a harrowing pilgrimage through clinics and laboratories — a journey marked by misdiagnoses and the despair of uncertainty. Nearly three years later, the answer arrived, shrouded in mystery: “immune-mediated axonal polyneuropathy,” a rare disease.

This personal experience serves as the starting point for an investigation into a paradoxical universe: a small number of people are grouped into thousands of types of rare diseases, resulting in millions of patients. My journey — from an anonymous citizen to a participant in a specific Ministério da Saúde policy — mirrors that of millions of Brazilians navigating the ‘diagnostic odyssey,’ seeking refuge for their rare conditions within the Sistema Único de Saúde (SUS).

WHY THIS MATTERS

There is still a significant lack of data regarding who people living with rare diseases are, which leads patients to spend years trying to obtain a diagnosis.

On the other hand, in Brazil, the SUS has been the primary path for both advancing research and enabling people to access diagnoses and treatments, reinforcing the importance of public policies for rare diseases.

Up to 10 Million Brazilians May Have Rare Diseases at Some Point in Their Lives

A disease is considered rare in Brazil when it affects up to 65 people per 100,000 individuals. This is the reference value used by the Ministério da Saúde. The public agency estimates the existence of about 5,000 different types in Brazil. However, this number is considered uncertain among scientists, who mention the existence of between 6,000 and

10,000 types. The World Health Organization (WHO) uses the International Classification of Diseases 11 (CID 11, not yet adopted in Brazil), which includes 5,500 rare diseases.

Determining how many people suffer from a rare disease in Brazil is a complex arithmetic operation because information is not properly categorized to identify these individuals. Within the Sistema Único de Saúde, the SUS, “current information systems do not record the number of visits for people with a specific condition,” justifies the Coordenação-Geral de Doenças Raras do Ministério da Saúde (CGRAR) in response to a freedom of information request. This gap hinders the planning of public policies. Data from SUS services — from the Sistema de Informações Hospitalares (SIH/SUS) and the Sistema de Informação Ambulatorial (SIA/SUS) — are records of consultations, hospitalizations, and procedures performed. I certainly appear in this database, but in a fragmented way — a consultation here, an exam there — without any indication in the system that my case is rare.

Intrigued by the issue, I took the topic directly to the General Coordinator of CGRAR, Natan Monsores. He revealed that, faced with the need to know this population and unable to rely on SUS projections, he preferred to adopt the results of discussions raised by Orphanet, an international data platform: “Between 2.5% and 5% of the Brazilian population will suffer from a rare disease at some point in their lives.” These numbers represent, in reality, about 5 to 10 million Brazilians. This scale underpins the Orphanet slogan: “Rare diseases are rare, but patients with rare diseases are very numerous.”

Brazil responded to this challenge by establishing the Política Nacional de Atenção Integral às Pessoas com Doenças Raras (PNAIPDR) through Ordinance 199 of 2014. This regulatory framework paved the way for the accreditation of reference centers for specialized care, research funding, the incorporation of new treatments with the availability of genetic testing, and even the creation of means to identify patients in the system. But its implementation was — and is — no bed of roses.

Rare Diseases: Building Policy within the SUS

The SUS can be used by any Brazilian citizen, and even foreigners visiting the country, for medical consultations, exams, vaccines, treatments, surgeries, and hospitalizations, all free of charge. The Ministério da Saúde reported that 84% of the Brazilian population does not have private health insurance. These are people who, therefore, depend directly on the SUS.

Although the SUS has progressed toward including a greater number of services and assistance, gaps remained regarding rare disease cases. In well-articulated words, with a friendly tone and a smiling expression, Natan Monsores explained: “Our healthcare system prepared for two things: infectious diseases, which decimated our population for decades, and chronic diseases (cardiac, respiratory, and others). However, there was always that small percentage of children at the mercy of this entire system, the patients that I captured inside the reference hospitals in Brasília. It was leukemia, it was a syndromic condition, it was the patient with a suspected disease that no one knew what it was...” These were experiences Monsores lived through as a public health physician.

With the advancement of science, high-cost genetic tests such as the “exome” (which identifies genetic alterations associated with rare diseases) and high-complexity treatments became possible in Brazil, although costs were high. Aware of these “innovations,” patients began to claim their right to exams and treatments through legal channels. During this period, families began to unite, associations were born, and patients on the fringes of the system grew stronger to assert their rights.

Monsores continues the narrative: “In the years 2012 and 2013,” health professionals (a group I was part of), representatives of organized civil society, and medical societies, such as the Sociedade Brasileira de Genética Médica, drafted the final text of what would become the Ordinance establishing the Política Nacional de Atenção Integral às Pessoas com Doenças Raras.” In 2014, the Ordinance was numbered 199 and signed by Health Minister Alexandre Padilha. It establishes guidelines for “Comprehensive Care for People with Rare Diseases” within the scope of the SUS and signals financial funding incentives.

The policy was managed by the Coordenação-Geral de Média e Alta Complexidade do MS. Much of the initial work was administrative, involving the accreditation of rare disease reference services. The Instituto Fernandes Figueira in Rio de Janeiro, the Hospital de Clínicas de Porto Alegre (HCPA), both in 2016, and the Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo (HC FMRP-USP) in 2019, were some of the pioneering services to serve the population. However, there was still no dedicated coordination for the policy. Ahead lay a long road to make what was on paper a reality.

Rare Disease Policy More Than Doubled SUS Consultations

The creation of the Coordenação-Geral de Doenças Raras (CGRAR) in 2023 marked a turning point. The number of annual rare patient consultations jumped from 30,000 to 70,000, and the network of reference services more than doubled in two years of work, increasing from 16 to 37, present in 16 of the 26 states and the Distrito Federal. Management is focused on actions such as improvements in neonatal screening, aiming to detect rare disease conditions early in childhood. Differential payment for consultations encourages investigative and diagnostic work: a regular SUS consultation pays about R\$ 20. A Rare Disease consultation pays R\$ 800. Each patient can repeat this billing four times a year, which also aims to fund high-cost complementary exams.

The incorporation of new technologies has been one of the main achievements. Jonas Saute, PhD in Medical Sciences, a member of the Academia Brasileira de Ciências, and a neurologist and geneticist at the Hospital de Clínicas de Porto Alegre (HCPA) — one of the pioneering centers — describes the current moment as a “therapeutic revolution.” He narrates the complex journey to implement “Exome Sequencing” in the SUS, a high-complexity genetic test that became available through the network in 2020.

“We had the possibility of doing it here, but our equipment was not adequate, and we lacked the expertise to analyze this type of exam to generate a report. In 2021, we created a project with the hospital’s research directorate. The hospital invested in forming a group with a molecular biologist, a medical geneticist, and a bioinformatician so we could create the conditions to learn how to analyze

and, with that, offer the service clinically.”

“In 2023, we began performing exome exams clinically (for people with rare diseases). Since then, we have performed about 200 exams through the SUS. With Ordinance 199, we achieved a certain fiscal balance, so to speak, allowing us to perform the exam without generating a loss.”

To circumvent the shortage of specialists capable of interpreting these exams, the Ministério da Saúde is implementing an analytical center with bioinformaticians and geneticists that will support reference services nationwide, issuing reports that will be translated into clinical diagnoses by the attending physician.

Another emblematic case of this new phase of the Rare Disease policy is the incorporation of gene therapy for Spinal Muscular Atrophy (SMA), a severe degenerative disease. The treatment, one of the most expensive in the world, was integrated into the SUS through an unprecedented risk-sharing agreement with the pharmaceutical company. Brazil became the sixth country in the world to offer this therapy in its public system.

“Before treatments existed, half of the children who died of respiratory failure by eight months or one year of age probably had SMA, but the presence of the disease was not identified,” Saute informs gravely.

The discovery of an effective treatment for SMA is considered recent. “Here at HCPA, we performed three different treatments that were approved for use in Brazil by Anvisa in 2017; all three were incorporated into the SUS, including gene therapy,” Saute revealed.

This therapy then cost R\$ 12 million. Unfeasible, even for the SUS, although it has already authorized 150 court-ordered requests, according to the MS. The Coordenação-Geral de Doenças Raras prepared a service structure with specialists qualified to monitor and evaluate the drug’s efficacy, a set of guiding documents, and closed a risk-sharing negotiation with the drug supplier for R\$ 7 million, for which installments will be paid according to the results observed in patients over five years.

These arrangements were aligned during 2025, and as of September, gene therapy for SMA was

implemented within the SUS. Since then, 15 children have received the therapy (it is only one dose) under this model and are being monitored.

The financial machinery behind this policy is a work in progress. According to Monsores, maintaining each of the 37 accredited reference services costs around R\$ 700,000 per year. That is R\$ 25.9 million for these centers. Adding to this the neonatal screening programs, assistance, research promotion, the supply of high-cost medications, exams, and litigation — which costs about R\$ 3.97 billion — the total bill, in 2025 alone, reached R\$ 4 billion.

Who Are the Patients and Where Are They?

One of the biggest obstacles to implementing the policy is the absence of consolidated epidemiological data, as seen at the beginning of this report. To overcome this barrier, the Ministério da Saúde funds one of the most important scientific projects in this area: the Rede Nacional de Doenças Raras (Raras). Coordinated by medical geneticist Têmis Maria Félix, PhD in Child and Adolescent Health, the network is dedicated to building a national epidemiological overview.

The physician, a member of the Sociedade Brasileira para o Progresso da Ciência, reported that the first retrospective survey, published in a scientific article in 2024 with data from 12,530 patients, brought crucial information to light: “71.9% of rare diseases are of genetic origin, and the SUS was the main funder of both diagnoses (84.2%) and treatments (86.7%). Regarding skin color: 47.5% of patients are mixed-race (mestiços) and 45.9% are white. Only 5% are Black, and less than 1% are Asian (amarelos) or Indigenous. The average time for these people to obtain a diagnosis was 5.4 years.”

The ultimate goal is to integrate this data into a unified electronic medical record with SUS data, developing algorithms capable of identifying patients with a history of symptoms suggestive of a rare disease. “We expect the pilots of these projects to start happening this year,” says Monsores, with the hope that this technology can shorten the painful diagnostic odyssey experienced by so many patients.

My Adventure in the Diagnostic Odyssey

While rare disease public policy was consolidating

in research environments, reference centers, and Ministério da Saúde offices, I was traversing an unusual and unknown terrain. I was bedridden, with paralyzed arms and legs, and without knowing it, I would benefit from this policy.

The first stage of my journey to find out what was happening to me lasted about a month. I started at a Unidade de Pronto Atendimento (UPA), a SUS health service focused on emergencies, in João Pessoa, where I live. The initial and mistaken diagnosis was chikungunya. I saw a doctor at a private clinic who wrongly diagnosed “fatty liver” and prescribed unnecessary medications; I made a strategic stop at a rheumatologist who discovered the nervous origin of the problem, until finally finding a neurologist who began the correct investigation.

Neurologist Bruna Nadielly began seeing me in her private practice. I could not bear the pain and could not wait for a SUS consultation. But she is also a doctor at the Hospital Universitário Lauro Wanderley (HULW), a SUS unit in João Pessoa. So, three months later, through the Posto de Saúde da Família (PSF), a municipal SUS unit, I managed to get an appointment with her at the hospital. She has accompanied me on a second leg of the diagnostic pilgrimage since June 2022. She conducted a series of exams through which various diseases were excluded: cancers, leprosy (hanseníase), Guillain-Barré Syndrome, thyroid problems, diabetes... With the exception of leprosy, I paid for all the exams, relying on financial help from people close to me at the church I attend and from family members.

The results, always negative, directed the clinical evaluation toward an autoimmune factor as the cause. It is as if a certain defense substance in my cells identified the proteins in the neurons as “enemies.” My defense cells attack the neurons in the peripheral nervous system — in the legs and arms — by mistake. This causes inflammation and even the death of neurons. This is “immune-mediated axonal polyneuropathy,” which, after three years, is considered chronic.

The story summarized here hides the pain and suffering I went through during this entire period. If it were not for my family, the support of the people from the church, and the confidence in the work of a prepared doctor, the trauma of this odyssey would be multiplied by ten.

All the treatments I have had so far, infusions of immunoglobulin, methylprednisolone, and cyclophosphamide, were through the SUS. Exhausting these possibilities, the medication I could use as an immunosuppressive agent is high-cost. I had to go to court to obtain it, as it is not listed as a medication offered by the SUS for the disease I have. Fortunately, the Court understood the need, and I’ve been receiving the medication for the infusions free of charge. They are done every six months.

With treatment, I regained part of my mobility through physical therapy. But I face permanent damage daily. The simplest tasks are complex for me and performed with pain: peeling a vegetable to make lunch; walking to the bakery; being here at this moment, sitting, typing.

At the Forefront of Care: Reference Centers

Opened in April 2022, the Centro de Referência em Doenças Raras de João Pessoa functions as a multidisciplinary outpatient clinic, offering consultations with 13 specialists (including two geneticists, a scarce resource in the country), rehabilitation, psychological support, and social assistance. By December 2025, the center had already served 1,199 people with rare diseases, with 231 cases still under investigation, covering 216 different types of diseases, according to the institution’s Epidemiological Profile. Most patients are children, reflecting the predominantly pediatric nature of rare diseases. Today, I am being treated by doctors at this center, in addition to the HULW.

It is in this environment that I met Luís Eduardo de Souza, 12, who has Duchenne Muscular Dystrophy. His mother, Vanderléia Rocha de Souza, found support in the center and in support groups to care for her son. “It took me three years to get the pre-diagnosis; I went to the internet and only found misfortune. Finally, I found a group of Duchenne mothers from Paraíba. They reassured me, comforted me, and guided me regarding the doctors,” she said.

“It’s not a favor anyone is doing. The SUS exists, and it is very good. When it doesn’t work, it’s because the front line wasn’t qualified,” says Saionara Araújo, director of the center.

Hope in Genetics

With about 80% of rare diseases having a genetic origin, scientific research is the primary source of hope. Centers of excellence such as the Centro de Estudos do Genoma Humano e Células-Tronco (Genoma USP), coordinated by renowned geneticist Mayana Zatz, are at the forefront of identifying disease-causing genes and developing new therapies.

The center, which has its roots in the pioneering work of Professor Oswaldo Frota-Pessoa in the 1960s, has already served about 100,000 people and has established itself as the largest center for serving people with genetic diseases in Latin America. The center's work is not limited to basic research: it also performs genetic counseling, a fundamental procedure for families to understand the risk of disease recurrence. The research is funded by the Fundação de Amparo à Pesquisa do Estado de São Paulo (Fapesp).

Mayana Zatz, of the Academia Brasileira de Ciências, began her research with Duchenne Muscular Dystrophy, the disease that affects Luís Eduardo. Over decades, the research group she coordinates has studied more than 30,000 families with neuromuscular diseases and identified more than 10 new genes responsible for these pathologies, which gives a chance for gene therapy treatment.

Dr. Jonas Saute explains the logic behind gene therapy: “If I know the cause, if I know the mechanism that leads to the disease (...) How do I treat it? I replace that protein. As complex as it seems, it's simpler. Most genetic diseases are monogenic, meaning they involve only one gene and one protein. This characteristic facilitates the development of specific treatments, unlike multifactorial diseases, where the cause is a summation of probabilities.”

However, the path from research to treatment is long and costly. Roberto Giugliani, a recognized name among geneticists for promoting the development of the fields of Medical Genetics and Rare Diseases in Latin America, has coordinated clinical trials for more than 20 years in Porto Alegre. In 2023, he founded the Casa dos Raros, an example of an institution dedicated to both assistance and research.

He clarifies that the average development time for a therapy is about nine years, passing through preparatory phases, study conduct, and clinical trials. These trials, funded by private agents, face ethical submission deadlines in Brazil that can reach 290 days. Although it is forbidden to pay research participants, there is an important secondary benefit: early access to medications that often represent the first treatment available for the disease.

When Urgency Is a Matter of Life or Death: Frederico Could Not Wait

The story of Frederico Goldsztein illustrates how urgency and immediate investment can accelerate research. In 2015, at age nine, he was diagnosed with medulloblastoma, a rare brain tumor that affects about 15,000 children worldwide each year. The standard treatment until then had been the same since the 1980s — highly toxic and with a relapse rate of 30%, which is almost always fatal.

His father, the businessman from Rio Grande do Sul, Fernando Goldsztein, did not accept the ‘death sentence’ after his son’s relapse. Without time to wait for a solution through the SUS, he sought clinical trials in the United States and, realizing that even there there was no specific research for medulloblastoma, proposed a donation to accelerate the studies. In 2021, he created the Iniciativa Medulloblastoma, an international philanthropic organization that raises private funds to finance high-level research. The consortium brings together 16 laboratories in 12 institutions in the USA, Canada, and Germany.

The results were impressive: in 2024, just three years after the initiative’s creation, two clinical trials for new treatments were approved by the FDA (Food and Drug Administration), the American regulatory agency. “It’s not a question of ‘if’ we will find it, but ‘when’ we will find the cure,” Fernando says with conviction. Frederico, who will turn 20 in 2026, lives with a quality of life never before seen in similar cases.

Although the initiative’s focus is global, Fernando promises to give back to Brazil: “when the clinical trials advance, the country will be the first to receive the new technology,” he stated. In addition, the initiative has already established agreements with Brazilian hospitals to share knowledge through webinars with local doctors.

Persistent Challenges and the Road Ahead

Despite the advances, significant challenges remain. The shortage of geneticists — fewer than 500 in the entire country — limits the capacity for diagnosis and follow-up. The geographical distribution of reference services is unequal, concentrating in the South and Southeast regions. Litigation remains an important avenue for access to non-incorporated treatments, generating unpredictable costs for the system.

The issue of high-cost medications is particularly complex. Less than 10% of rare diseases have specific drug treatments, and those available often cost millions of dollars. The risk-sharing model adopted for SMA may be a solution, but its replication depends on the willingness of pharmaceutical companies to negotiate.

Monsores recognizes the complexity: “Are we going to solve everything at once? No. But we are now starting to have mechanisms to try to mitigate these long itineraries.” The expectation is that with the implementation of electronic medical records, the formation of the specialist database, and the expansion of the service network, the SUS can offer increasingly agile and effective care.

Têmis Félix reinforces: “We know that most rare diseases have no treatment at all. There is a lot that can be done in terms of physical therapy and rehabilitation, but most do not have drug treatment.” In this context, genetic counseling, psychological and social support, and the building of support networks are as important as the search for a cure. ■



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China's 7,000-year-old sunmao woodwork leads to world's most efficient DNA editing tool

Quest to beat the US saw a Chinese team turn to ancient traditions, leading to a new technique 10 times more effective than other methods



The new gene-editing strategy is more precise at insertion and replacement in rice. Photo: Shutterstock

How to beat the United States in gene editing? A research team from Peking University and the Chinese Academy of Sciences had struggled with this question for three years, with no results.

Then they decided to draw inspiration from an ancient woodwork tradition and cut DNA like wood. It led to a gene-editing tool with unprecedented efficiency.

In the Chinese mortise-and-tenon joint technique known as sunmao, no nails or glue are used. Instead, a protruding “tenon” on one piece fits securely into a matching “mortise” on another, creating a solid interlocking structure.

One of the earliest examples can be traced to China's Hemudu culture in Zhejiang province around 7,000 years ago.

The new editing strategy achieves precise insertion and replacement efficiencies of up to 59.47 per cent in rice, surpassing those of existing mainstream tools.

At certain genomic loci, which refer to the fixed physical locations of genes or other DNA sequences on a chromosome, its editing efficiency is 10 times higher than that of traditional Cas9-based systems.

CRISPR-Cas9, the most popular tool in use, was pioneered by researchers affiliated with Harvard and MIT. Its core patents remain solid until 2038.

“Gene editing tools are like lithography machines; if they are ‘choked’, nothing gets done,” Zhang Huawei, a researcher at the Institute of Advanced Agricultural Sciences of Peking University, said in a January 6, interview with China Science Daily.

“Just as we need our own lithography machines, we need independently developed tools for gene editing.”

In December 2019, he established his own laboratory at the university with a goal to create a framework for precise gene editing with independent intellectual property rights for China.

In crop breeding, precise and scarless DNA fragment insertion or replacement represents a pivotal technology for overcoming bottlenecks in varietal improvement and improving national food security.

However, for the first three years, the experiments yielded no usable data. Then the ancient mortise-and-tenon structure sparked a new idea.

The techniques of Chinese wooden architecture, including mortise-and-tenon joints, have been inscribed on Unesco's Representative List of the Intangible Cultural Heritage of Humanity.

Zhang realised that the mortise-and-tenon principle closely resembled the gene-editing framework they sought: achieving precise integration by constructing complementary “tenons” and “mortises” at the DNA level.

The team engineered a special double-strand break structure – forming a “mortise” – at target sites in the rice genome, featuring a defined overhang. They then synthesised a double-stranded DNA donor that served as the “tenon”, with ends complementary to the “mortise”. ■

Through precise pairing and ligation, their “mortise-tenon connection system” (MT system) enables directional insertion and replacement of DNA fragments.

At a single target locus, the MT system’s forward insertion efficiency reached 46.55 per cent and 48.12 per cent, with no reverse insertion. In contrast, the traditional Cas9 system paired with standard donors achieved only 1 per cent efficiency.

At optimal sites, the MT system’s forward insertion efficiency hit 83.54 per cent, with a precise insertion rate of 59.47 per cent – more than 10 times that of the Cas9 system.

Beyond higher efficiency, the MT system also enables “scarless editing”. Previous systems often relied on recombinases, which leave short recognition sequences at insertion sites – akin to surgical scars.

“Our mortise-tenon system doesn’t require such sequences; after editing, it leaves no extra bases behind,” Zhang added.



Mangaiyarkarasi

J

in The Hindu Businessline | India



Mangaiyarkarasi is a journalist with The Hindu BusinessLine in Chennai, India. She has worked across newsrooms and classrooms, focusing on issues at the intersection of science, medicine, health, pharma and nutrition. Her stories explore how health policies, access to medical care, and awareness shape public health.

Her foundation in health journalism was laid nearly a decade ago with The Diet Food, a Tamil-language health magazine, where she wrote

extensively and also authored four books in Tamil that makes health and nutrition more accessible to the common man. Before joining the newsroom, she was an Assistant Professor teaching Journalism and Mass Communication at SRM Institute of Science and Technology. She brings together editorial rigour and balance to her reporting and strongly believes in storytelling as a tool to create awareness and bring policy changes.

The silent epidemic. ‘Rare diseases aren’t rare, they’re orphaned’

Despite India’s national policy on rare disease treatments, access is marred by delays and exorbitant costs.



Ahead of Budget 2026, patient groups call for centralised procurement, new-born screening and cap-less funding | Photo Credit: istock.com

Eight-year-old Krish Dev is recovering from a surgery performed to support his neck, as his body stopped growing even as his head continued to do so. Witty and sharp as a tack, Krish longs to play with other kids, but cannot as the bones in his legs have protruded outwards, making it difficult for him to even walk, says his mother, Abirami.

Krish has mucopolysaccharidosis type IV (MPS IV), a rare genetic disorder that progressively affects bones and joints, leading to a short stature, spinal cord compression and breathing problems. Abirami has come to terms with her son’s diagnosis, but not with the wait for care, as hospital visits begin with hope, only to end with no clear treatment pathway.

Last year, the Union Budget put a spotlight on rare diseases when it exempted certain drugs from basic customs duty to reduce treatment costs. As Budget 2026 approaches, affordable treatments and care still remain elusive for many patients, even as the disease progresses on schedule.

The National Policy for Rare Diseases (NPRD), notified in 2021, covers 63 conditions under three groups and offers financial assistance through designated centres. But advocates for patient groups say the current model — including a ₹50-lakh

ceiling for many cases — is inadequate in meeting the cost of lifelong therapies and recurring support care, pushing patients’ families into debt and appeals for crowdfunding.

Abirami says it took nearly four years for her family to even understand what was happening to Krish. Doctors at a government hospital in Salem treated him for vitamin deficiency for years, she says, before eventually prescribing genetic testing at a private clinic. “We finally got to know that Krish has a rare disease that cannot be cured and he will have to live with it throughout his life,” Abirami says.

She was later told that the medicines for Krish’s condition are not easily available in India, and are prohibitively expensive. The estimate still haunts her: one dose a week, each potentially costing up to ₹2 lakh.

For now, Krish’s life revolves around his surgeries and recovery. Abirami says his head has grown normally for his age, while this body has not kept pace making it difficult to bear the weight of the head. He underwent surgery to insert support structures for this neck. Six months later, she says, he needed another operation — this time for his legs, after bone complications made walking difficult.

Different disease, same corridor

For Rajesh, a high school teacher in Villupuram, the disease in question may be different but the uncertainty is familiar. His wife lives with paroxysmal nocturnal haemoglobinuria (PNH), a rare blood disorder requiring frequent blood transfusions.

He says she passed blood in her urine for a long time, even as the couple dealt with repeated miscarriages. It took about a year, he says, for doctors to identify what was wrong.

His wife needs a blood transfusion every month, which costs between ₹40,000 and ₹60,000, including the travel to Chennai for the treatment.

Each transfusion involves three or four units. However, within weeks, he says, the symptoms return — fatigue, headaches, nausea, breathlessness, dizziness, and blood in the urine. Since her blood group is quite rare — B-negative — it's not easy to find either, he says.

medicines that can reduce the frequency of transfusions, cost up to ₹3-4 lakh a vial. Rajesh says his wife took the drug once during pregnancy, after borrowing heavily to pay for it, but still suffered miscarriage.

“If it's a one-time drug, we can pull up the money somehow,” he says. “But if they have to take it every three or six months, it's unaffordable for me and my family.”

Funds in cold storage

Meanwhile, funds given to designated centres remain severely under-utilised even as thousands of patients wait for the ₹50 lakh support, says Manjit Singh, President, Lysosomal Storage Disorders Support Society (LSDSS).

In response to an RTI filed by Manjit Singh, accessed by *businessline*, the Health Ministry said that about ₹30.79 crore had been utilised under the NPRD in FY25-26. However, for the same period, ₹299.59 crore was allocated under the NPRD, according to a written reply to a Lok Sabha question.

Singh, himself a parent of a child with a rare disease, says the funding cap fails to acknowledge how rare disease treatment works in real life. “Fifty lakh rupees is peanuts,” he says.

Patients' groups have repeatedly urged the ministry and judiciary to remove the cap and allow continued support on a case-by-case basis. “Patients who improve on getting the treatment should be given continued treatment, not a one-time grant,” he says, as any break can undo the gains from the treatment, and families are left stranded.

For some like Krish, the support never came. He has been registered with the Institute of Child Health, E (an NPRD designated centre) in Egmore, Chennai, for three years, but has not heard from them, even after this condition started progressing, his mother alleges.

In an order dated October 4, 2024, the Delhi High Court directed the health ministry to release funds for patients who had exhausted the ₹50-lakh cap, and create a pooled national fund for rare diseases with an allocation of ₹974 crore across FY2024-25 and FY2025-26. “This was not implemented and over 50 patients have died in the past two years. These were preventable deaths. Their treatments were halted when the ₹50 lakh ceiling was exhausted, despite the court's clear intent to eliminate such limits and ensure continuity of care,” Singh says.

The government has appealed against the Delhi HC order in the Supreme Court, with the hearing scheduled for March 2026, without interim directions on financing.

Singh alleges that the cap discouraged some designated centres from initiating treatment. For many diseases, he says, enzyme replacement therapy (ERT) may be the only option, which can push up the cost to ₹1 crore. “They were afraid that if treatment starts and then stops at ₹50 lakh, families will go to court to ask for continuation,” he says.

Lack of transparency

“There is no clarity on how this operates. If a patient goes to a centre, we don't know when they will get the treatment, how long they have to wait, whether they will get the treatment or not. Only through an RTI will we come to know. The patient has to wait till the call comes,” says Prasanna Shirol, co-founder of the Organisation for Rare Diseases India (ORDI), calling for clear timelines and communication to families.

Shirol argues that fragmented procurement is a bottleneck. When each centre has to procure drugs independently, delays become inevitable, negotiations are weaker, and costs remain high. “If 100 per cent of the funding is coming from the Centre (government), then why should each centre do its own chaos?” he asks. A centralised procurement mechanism, he says, would allow faster delivery across states.

The states, meanwhile, are attempting to create their own models. Kerala has devised a programme to provide free treatment for certain rare diseases in children under 18. The Kerala United Against Rare Diseases (KARE) initiative — soft-launched in 2022

— provides risdiplam (procured from Swiss pharma company Roche, at a negotiated but undisclosed price) free of cost to children with spinal muscular atrophy (SMA), and has —expanded the coverage to other rare conditions, including lysosomal storage disorders (LSDs), Gaucher and Pompe diseases, as well as growth hormone deficiency in paediatric patients requiring long-term hormone therapy.

In August 2025, Goa introduced a value-based pricing policy for innovative lifesaving therapies to make high-cost treatments for rare genetic conditions more accessible. It partnered with organisations like AstraZeneca and Qure.ai to integrate AI-driven diagnostics for early detection and treatment.

Ask from the Budget

Shirol calls for a national newborn screening policy, together with a genetic counselling facility, saying this could pre-empt the progression of many diseases. “This is where I need the Budget,” he says. “One, for treatment costs of existing rare diseases in children. But for the long term, I need the Budget for improving the infrastructure, encouraging research

and development, drug development.” He points to the US’ Orphan Drug Policy (1983) to show how tax incentives can encourage companies to make these medicines.

Even if treatment is unavailable, Shirol argues, supportive care should not be neglected. Of the nearly 7,000 identified rare diseases worldwide, he says, only a small fraction have definitive therapies, and insurance cover is almost non-existent. For the rest, families rely on supportive care like rehabilitation, physiotherapy, speech and hearing therapy, mental health counselling — services that already exist in India but are not systematically integrated into rare disease care. Caregivers, he adds, are invisible in policy. “The mother spends her lifetime taking care of the child,” says Shirol, who turned to advocacy after losing his 24-year-old to pompe disease

Singh, too, believes the biggest missing piece is not policy design but follow-through. “The government is not sensitive at all to these diseases,” he says. “Rare diseases are not rare, they have just become orphaned.” ■



Evans JONA

in Post On Sunday | Zimbabwe



Evans Jona is a qualified journalist and communications specialist with more than eight years of experience in radio broadcasting, digital media, health and development communication. A graduate of the National University of Science and Technology, he has worked with leading Zimbabwean commercial radio stations such as Diamond FM and Skyz Metro FM, producing and reporting powerful human-interest stories. Since October 2024, Evans has been with Post On Sunday, an online

news publication, where he reports on health and community development issues. His health reporting has included collaborations with the Rare Diseases and Disabilities Foundation Africa (RaDDA), the AIDS Healthcare Foundation (AHF), and the Zimbabwe AIDS Network (ZAN). Dedicated to people-centred storytelling, Evans strives to use journalism as a tool to raise awareness, promote social change, and highlight the resilience of communities across Zimbabwe.

A Life of Pain, A Heart of Hope: Molyn Chima's Fight Against Sickle Cell in Zimbabwe



HARARE — It all began with a sharp backache when she was just nine years old. “It was consistent, and it felt like broken pieces were being rubbed on my back,” recalls Molyn Chima, founder of the Sickle Cell Anaemia Trust Zimbabwe. “I was a happy, playful child, completely innocent, and I didn’t know that my life had changed forever.”

As the pain worsened, Molyn struggled to breathe, felt weak, and could not keep any food down. “All I could do was cry,” she remembers. Her parents rushed her from clinic to clinic, desperate for answers, but doctors could not provide a diagnosis. Soon, her life, and the lives of those around her, were turned upside down.

“I had to be carried on my mother’s back at nine years old,” she says. “It was difficult for the family. My younger sibling had to mature before her time, helping take care of me. I doubt if they ever slept because every time I turned, one of them would be watching me and asking if I was okay.”

Eventually, the diagnosis came. Sickle cell disease, a genetic condition, had run through her family for generations. “It did not come as a shock because we had seen it in other members of the extended family,” she explains. “A relative recognized the signs from our past experiences. It was a relief to know what I had, but we also braced ourselves for the unknown.”

Sickle cell disease is characterized by severe pain, anaemia, and general body weakness. For Molyn,

the disease is not just physical, it carries a heavy emotional and social burden.

“People often doubt the frequency of painful crises because the disease is unpredictable,” she says. “A sickle cell crisis can last a day, weeks, or even months, depending on the cause and access to care. Severe pain is the hallmark, but because of this, some patients are wrongly accused of drug-seeking. That delays proper management and makes life harder.”

Living with the condition is like carrying an invisible disability. “Most people cannot see it, but it weighs heavily on the mind,” she says. “Discrimination happens in workplaces and in communities. A crisis can happen at any time, and some people doubt, judge, or ignore you when you need help the most.”



Molyn’s experiences inspired her to take action. She founded the Sickle Cell Anaemia Trust Zimbabwe, a support group that brings patients together to share their stories, counsel one another, and offer comfort in knowing someone understands exactly what they are going through.

“The support group has been life-changing,” she says. “Hearing the lived experiences of fellow warriors helps me understand my condition better. We give each other hope and strength. Just knowing someone knows what I am feeling is always comforting.”

The financial cost of living with sickle cell is significant. “It’s expensive,” she says. “We need blood transfusions from time to time, frequent hospital admissions, and chronic medications. Hydroxyurea, which is vital for many of us, is difficult to get locally, and sometimes we have to source it from other countries. Medical aid is costly and not user-friendly for people with rare conditions like ours.”

While medical advances, including genetic treatments, offer hope, access remains a challenge. “I personally abstain from bone marrow transplants for now,” she says. “I lost a sister after she underwent one, and the doctors experimented on her for reasons I still do not fully understand. But I remain positive. As treatment technology improves, maybe one day I will reconsider. My biggest concern is affordability, how can we access such treatments when we struggle to get Hydroxyurea?”

Molyn is passionate about the need for better health policies for rare diseases. “Everyone should know about these conditions, and hospitals must treat crises as medical emergencies to prevent complications like organ damage,” she says. She advocates for newborn screening, genetic counselling, and the decentralization of specialized care so patients do not have to travel long distances when they are unwell.

“Specialized personnel, from doctors to nurses and village health workers, are essential,” she says. “It is important that people understand what they are treating. Early detection helps prevent complications like stroke in babies and reduces mortality. Families should know their genotype to make informed choices and prevent future cases.”

Through it all, prayer has been her anchor. “There are times when even loved ones get frustrated and

hopeless,” she says. “Sometimes it is I, the patient, who reassures them that all will be well. Prayer keeps me grounded.”

Earlier this year, Molyn faced one of the toughest battles of her life. Her right leg became unbearably painful. “I sometimes slept for only ten minutes at a time,” she recalls. “I even called my orthopedic surgeon asking for an amputation, but he refused. He assured me it would pass. I spent weeks in a wheelchair, but God was there. I saw Him in my doctor, my parents, my daughter, and the provisions that came through. Thank you, Dr. Mwoyofiri, for following up, for not judging, and for refusing to remove my leg.”

She also expresses heartfelt gratitude to her family and community. “I saw God in my parents, my daughter, and all those who stood by me. Prayer, support, and love carried me through some of the darkest moments.”

For newly diagnosed patients, Molyn has words of encouragement drawn from experience. “Hold on to your faith first. Be strong, patient, and protect your mind and body to avoid unnecessary crises. Understand that some people will not understand you, identify those who do and stick with them because they will be your pillars. Get a doctor who listens without judgment, and if possible, build a friendship with your healthcare team. Never be shy to take your medication. Almost everyone is taking medicine; the only difference is why we take it. Be comfortable with your treatment.”

Through the Sickle Cell Anaemia Trust Zimbabwe, Molyn continues to advocate for awareness, provide support, and challenge misconceptions about the disease. Her story is a testament to resilience, faith, and the power of community support in the face of chronic illness.

“Living with sickle cell is not easy,” she says, “but with faith, knowledge, and support, you can survive, thrive, and help others do the same.” ■

A Rare Condition, a Childhood of Silence, and Tinotenda Mudarikwa's Fight for Dignity



BULAWAYO – When Tinotenda Mudarikwa speaks about childhood, his voice is steady, but the memories he carries are anything but. He was born with anorectal malformation, a rare congenital condition commonly referred to as imperforate anus, a defect in which a baby is born without a normal anal opening. Surgeons had no choice but to bring part of his intestine out through his abdomen, attaching a colostomy bag to collect waste. From that moment, life followed a path few could see, and even fewer understood.

“There is no control,” Mudarikwa says quietly. “It just comes out.”

That single medical reality shaped an entire childhood. Growing up with incontinence meant living in constant fear, fear of smells, of stains, of laughter. Well into his teenage years, Mudarikwa struggled to control when he defecated, a condition that isolated him from peers and pushed him into silence. At Lady Tate Primary School in Kadoma, the classroom was not always a place of learning. Sometimes, it was a place of shame. He remembers one day vividly: a teacher wrote a letter home saying her pupil had diarrhoea. It was break time, and he was asked to go home.

Children looked away. Some laughed. Some chuckled. Others simply wrinkled their noses. “That’s when you know you are different,” he says. “That’s when you start pulling away from people.”

According to global medical data cited by the World Health Organization (WHO), anorectal malformations occur in about one in every 5,000 live births. Using Zimbabwe National Statistics Agency (ZimStat) birth estimates of more than 350,000 births annually, paediatric specialists say at least 60 to 70 Zimbabwean children are likely born with the condition each year, most into families and communities with little understanding of what it means.

But numbers do not capture humiliation. They do not capture being labelled “smelly”. Or being avoided in class. Or being told, as Mudarikwa was, that you are a curse.

“In the community, I was told I was bewitched,” he says. “That I was cursed.”



Those words dug deep. Over time, he began to believe them. Zimbabwe’s cultural landscape, like much of Africa, often interprets rare medical conditions through superstition. For children like Mudarikwa, that interpretation can be more

damaging than the condition itself. According to UNICEF Zimbabwe, children with disabilities are twice as likely to experience bullying, exclusion and emotional distress compared to other children.

By 2004, the emotional weight became unbearable. Mudarikwa suffered a severe mental health breakdown, a psychotic episode that forced his mother to make a painful decision: to seek psychiatric help for her son. “She accepted that I was mentally ill,” he says. “And she took me to a psychiatric hospital.” For mental health experts, the link is clear. The WHO Mental Health Atlas (2023) notes that people living with chronic congenital conditions face higher risks of untreated depression and anxiety, particularly in low-income countries where psychosocial support is limited.

Yet, in Zimbabwe, disability conversations remain focused on what can be seen, wheelchairs, crutches, canes. “Disability is not always visible,” Mudarikwa insists. “Some of us look okay. But we are fighting battles you cannot see.”

Globally, rare diseases affect more than 300 million people, according to Rare Diseases International, with over 70 percent of rare conditions presenting in childhood. Despite this, many African countries, Zimbabwe included, have no dedicated rare disease policy. Zimbabwe’s National Disability Policy of 2021 recognises invisible disabilities and commits the State to inclusive healthcare and education. The country is also a signatory to the United Nations Convention on the Rights of Persons with Disabilities. But advocates argue that implementation remains weak, especially for children with rare conditions requiring long-term medical, psychological and educational support.

“Rare diseases fall between the cracks,” says a policy analyst Takudzwa Madondo. “They are not counted, not tracked, and therefore not prioritised.”

For Mudarikwa, silence was no longer an option.

In 2018, he registered the Rare Diseases and Disabilities Africa Foundation, born not from theory or policy papers, but from lived pain. The organisation advocates for awareness, inclusion, data collection and dignity for people living with rare and invisible conditions.

He worries deeply about children growing up today, not just in classrooms, but online.

“What happens to a child with a colostomy bag in a world of social media?” he asks. “In a world of cyberbullying?”

Schools, he argues, are ill-prepared. Many lack private sanitation facilities, counselling services, or basic training for teachers to understand medical incontinence. The result is exclusion, sometimes silent, sometimes cruel.

Mudarikwa believes change must start with mindset. In kombis, in buses, in queues, in schools. “When you hold your nose at someone, when you mock them, you don’t know half of what they are carrying,” he says.

He pauses, then adds: “Those moments stay with a person for life.” Today, Mudarikwa speaks not just for himself, but for children who cannot yet explain why they smell, why they struggle, why they are different. Children who are being born every year with conditions medicine can manage, but society still rejects.

“I believe our mindset can change,” he says. “And when it does, we will stand and speak for every child born like me. For every child living with incontinence. For every child growing up in shame for something they did not choose.”

He was born without an ending, without a normal anatomical closure. But through his voice, he is opening something far more powerful: a conversation Africa can no longer afford to ignore. ■

Born Invisible: A Rare Condition, a Childhood of Silence, and Tinotenda Mudarikwa's Fight for Dignity



Video [here](#)

Living With an Invisible Battle: Bulawayo Woman's Eight-Year Fight With Systemic Lupus Exposes Zimbabwe's Rare-Disease Gaps



BULAWAYO – When Colette Sophie Morris speaks about her life, she often pauses, as if replaying years of confusion, misdiagnoses and the slow unraveling of a mystery that nearly cost her life. Today, at 43, she lives with systemic lupus erythematosus (SLE), a chronic autoimmune disease that attacks healthy tissues and organs. She remembers her diagnosis clearly: “I was thirty-five when they finally told me what was wrong. That was on the 13th of November 2017.”

For years before that moment, she had been cycling through illnesses that seemed unrelated. “Doctors later told me I had suffered from SLE long before I even knew it,” she said. Her description of the disease is both simple and heartbreaking: “Your immune system turns against your own body. It uses abnormal antibodies to attack healthy cells. It can damage anything – your organs, your skin, your blood. Everything.”

Some of her symptoms are visible; others are not. Excessive sweating, for example, has become a part of her daily life. “If the sweating is too much, it means I am not well,” she explained. “I have to carry extra clothes on a warm day because I become soaking wet.” Fatigue often keeps her in bed for long stretches, and her fingers frequently crack and develop infections, making basic tasks like laundry a battle. Her face, hands and feet sometimes swell without warning. On some mornings, she wakes up partially blind. “But as the day goes on, I start seeing normally again,” she said, as if describing something ordinary.

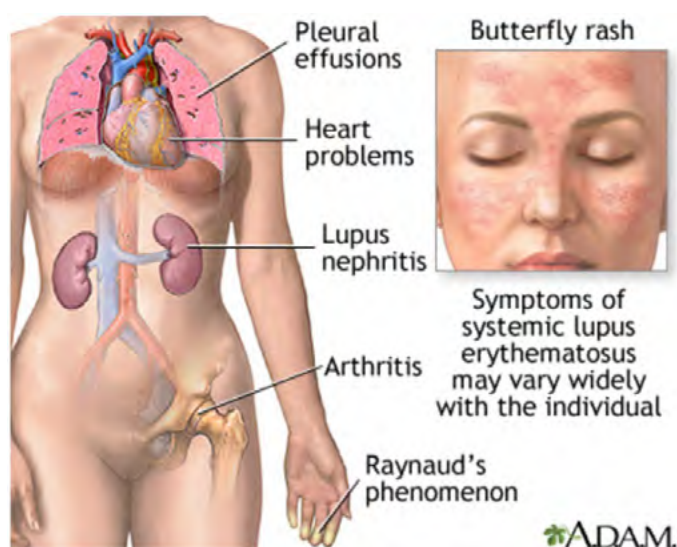
Her journey to diagnosis spans nearly three decades. She traces the beginning of her ordeal to 1996 when she began collapsing and experiencing fits. “It was a long journey, full of confusion,” she said quietly. “I felt I was sick for too long. At one point I even asked to be started on HIV treatment because I thought maybe everyone was missing something.”

Everything changed in November 2017 when her body suddenly went into severe distress. “My face swelled beneath the eyes and around the cheekbones. My hands and feet swelled. I had maroon marks on my palms and the soles of my feet,” she recalled. Then came symptoms that terrified even the doctors: bloody tears, blood vomit, painful ulcers in the mouth and ears, a butterfly-shaped rash across her nose, and seizures that would not stop. She had been discharged from hospital just two weeks earlier. When she returned, doctors ordered a series of tests. “That is when they said I fit the criteria of SLE,” she said. Her family was called in and informed she had a rare condition. One doctor warned the family to prepare for the worst. “They told us I might live for only six months.”

Before that diagnosis, she endured years of misdiagnoses. She was repeatedly admitted for epilepsy, then treated for diabetes even though she wasn’t diabetic. “One doctor asked me if I had diabetes. I said no, but I had been treated for it,” she said, shaking her head. She later learned she was

pre-diabetic. She was also treated for meningitis many times. “They gave me injections even before test results came out,” she said. “Then the results would be negative.” Her confusion, headaches and psychotic episodes were misunderstood. “They said I was going mad. They used the word ‘hysteria,’ and every time I heard it, I cried.”

Reactions at home were mixed. Her immediate family accepted the diagnosis, but relatives outside the home doubted her. “Some thought I was pretending, hiding HIV, or just being lazy because I spent so much time in bed and had lost a lot of weight.”



Accessing healthcare after diagnosis brought a new wave of challenges. She had no money for medication or frequent hospital visits. “Even when I had flare-ups, I wouldn’t go because I couldn’t afford it,” she said. Eventually she was referred to United Bulawayo Hospitals, where specialists could monitor her. But information remained scarce. “They didn’t explain much. They showed me the butterfly rash in the mirror, said it meant I had SLE, and then told me to Google what SLE was.”

Medication, for years, was beyond her reach. “My family helped for some time then stopped. Friends helped here and there,” she recalled. Donors through a church network assisted her for three years before the support ended. Tests were expensive and often impossible to afford. Follow-up appointments were equally difficult. Many times she walked long distances to hospitals while hungry or extremely fatigued. “Sometimes I ended up admitted because I was too weak,” she said. Today, she still lives without two major medications used to manage lupus. “I only take hypertension medicine, epilepsy

medicine, sleeping tablets and depression medicine.”

Her condition has reshaped every part of her life. She left marriage twice and now lives with her parents and children. She rarely socialises. “I am mostly at home sleeping,” she said. Yet she managed to return to school at 39 and earned a distinction in Biology at the United College of Education despite frequently getting sick. “I lagged behind my classmates. If I had a choice, I would have given up long before halfway. But they refused to let me give up,” she said. Although she finished training, she is still waiting for deployment as a teacher, something she believes is delayed because of her health. “It is stressful seeing my colleagues working while I wait.”

Everyday tasks remain unpredictable. Headaches strike without warning. Tooth infections have led to many removals. She collapses sometimes, suffers chest pain that affects her breathing, and cannot be exposed to much sunlight. Insomnia leaves her exhausted for days. Her memory fails her. She moves slowly. But she has learned a simple rule: “I have learned not to get angry because anger makes inflammation worse.”

Emotional support did not come easily at first. “I used to cry and frustrate everyone around me,” she said. Over time, she built ways to cope. Her mother-in-law became a refuge whenever confusion or illness overwhelmed her. Sometimes she sought treatment without telling her family, just to avoid worrying them. She finds healing in music, prayer, and spending time at the Zimbabwe Red Cross Society. She ministers in children’s church, a role she says brought unexpected strength. “Interacting with children healed me faster than sitting in the main church,” she said. Her family calls her umafa evuke, the one who dies and rises again. “When I recover, you can’t even tell I was ever sick.”

Stigma, however, remains a constant shadow. “People don’t understand what you’re going through,” she said. She rarely discloses her condition unless she believes it will help someone else. She believes communities must learn that rare conditions are not witchcraft and not a curse. “Families should not point fingers looking for someone to blame,” she said. She also warns lupus patients against traditional medicines. “I was told that boosting the immune system can make me worse.”

Morris is now connected to support structures like RADDa and an epilepsy group, which opened her eyes to others living similar lives. But she believes Zimbabwe still lacks emotional, financial and medical support for rare-disease patients. “The voices are there but not loud enough,” she said. If she ever had the chance to address policymakers, she knows exactly what she would ask for: “A national foundation for persons with rare conditions. A place where all the services they need are under one roof.”

She believes rare-disease awareness in Zimbabwe has improved since 2017, especially through social media. She now feels more understood by healthcare workers. “These days they know me. Even at the local clinic, they call me by my name,” she said warmly. The improvements she hopes for are practical: jobs for adults who can still work, free schooling for children, government-funded medication, food vouchers, support groups, and family education. “Love within families facing this ordeal is important,” she said. “When there is love, healing becomes faster.”

Despite everything, she considers herself resilient. “This journey taught me to be tough. The tests

I went through were more painful than what an ordinary person could handle,” she said. She knows her triggers now. She pushes herself not to remain bedridden for long. She listens to church audios, writes down scriptures, and drinks water when headaches begin. Her greatest victory remains her education. “Getting a distinction in Biology made me believe that anything is possible, even with a rare condition.”

She thinks about the future of rare-disease care in Zimbabwe often. “I hope that medicine will be available. That everyone will know about these conditions. That a foundation will be opened,” she said, laughing softly as if catching herself dreaming. “Maybe by then I won’t be alive to see it.”

Her message to anyone newly diagnosed is grounded in faith and compassion. “I would pray with them, explain what I know, help them understand their condition,” she said. And then she mentions the scripture she holds closest: “In Isaiah 41:10, God says, ‘Fear not, for I am with you.’ That is what kept me going.” ■

From Library Discovery to Global Stage, The Young Zimbabwean Challenging Rare-Disease Neglect



GWERU – When Vincent Mugwati stumbled upon a laptop screen in the Midlands State University medical library one evening in 2023, he never imagined it would spark a journey that would take him from local classrooms to global advocacy forums. The screen displayed a simple advertisement for a rare-disease advocacy website, a term he had never encountered before in textbooks or lectures.

“I had never even heard the term ‘rare diseases’ before that night,” Mugwati recalls. “I spent the entire night researching. I needed to understand what it was and whether people in Zimbabwe were affected. What I found shocked me.”

That night ignited a determination that would see the third-year medical student quickly emerge as one of Zimbabwe’s youngest voices in rare-disease advocacy. Within months, Mugwati joined the Rare Diseases International Youth Leadership Programme, one of only 13 young global advocates

trained to amplify the voices of patients and influence policy. “Being part of RDI allowed me to participate in global forums like the World Health Assembly,” he says. “It showed me that Zimbabwe can learn from international best practices, but only if we first know who our patients are.”

Mugwati’s clinical rotations exposed him to the stark realities for patients in Zimbabwe. Rare diseases are largely absent from medical curricula, leaving future doctors ill-prepared to diagnose life-threatening conditions. A case that remains etched in his memory is that of a patient with Stevens-Johnson syndrome. “The doctors took a long time to diagnose it,” he says. “The patient went into a coma, and even the clinicians seemed uncertain if they would survive. How many people have died without anyone recognizing a rare condition?”

Compounding the problem, public misconceptions often delay care. In many communities, unexplained symptoms are attributed to witchcraft, intensifying trauma for patients and families. “Many patients I’ve met still struggle with the psychological impact of being misunderstood when they were younger,” Mugwati says. “That can last a lifetime.”

Structural gaps in Zimbabwe’s healthcare system make timely care even more elusive. The country has no genetic clinics, no clinical geneticists, and no genetic counselors. Medicines are often unavailable locally or prohibitively expensive, and referral pathways are fragmented. “Families sometimes have to source life-saving medication from outside Zimbabwe at their own expense,” Mugwati says. “It’s heartbreaking.”

For Mugwati, data is the first step toward reform. “We need a national rare-disease registry,” he asserts. “Without it, policymakers can’t make informed decisions. Budgets are drawn blindly, and patients suffer.”

Despite these challenges, Mugwati sees hope in Zimbabwe’s younger medical professionals. Modern

curricula now introduce modules in precision medicine, genomics, and biomedical sciences. “Young doctors can adopt what I call a ‘rare-way mindset,’” he explains. “When common diagnoses fail, they should ask: could this be something rare? That mindset alone could save countless lives.”



Emerging technologies, from genomic screening to telemedicine and AI-assisted diagnostics, also offer promise, but only if the healthcare system is prepared. “Early detection could transform outcomes,” Mugwati says. “But strong policies, data privacy regulations, and trained medical personnel are essential. Technology alone isn’t enough; it must be combined with education and patient-centered care.”

His vision is ambitious but clear: a national rare-disease ecosystem with a fully functional registry, specialized clinics, genetic services, integrated care pathways, and a patient-centered approach. “I want doctors who can diagnose rare diseases early, a health system that treats patients with dignity, and policymakers who plan based on evidence, not guesswork,” he says.

Mugwati’s work resonates strongly with the mission of the National Press Foundation (NPF),

a U.S.-based nonprofit that equips journalists and advocates to shine a light on underreported health issues. NPF emphasizes storytelling that drives policy, community engagement, and evidence-based reform. While Mugwati is not a participant in NPF programs, his advocacy mirrors the foundation’s principles: using data, patient narratives, and grassroots campaigns to promote systemic change.

During the 2025 NPF Reporting on Rare Diseases Fellowship, journalists engaged with young advocates worldwide who shared experiences remarkably aligned with Mugwati’s observations. Presenters included Dimitrios Athanasiou, Rare Disease Patient Advocate; Phillip Langat, Founder of the Motor Neuron Disease Association of Kenya; Fernando Goldzstein, Founder of the Medulloblastoma Initiative; and Sarita Edwards, Founder of E.WE Foundation and rare-disease parent advocate. They highlighted delays in diagnosis, systemic barriers, and the role of youth and caregivers in shaping rare-disease advocacy across Africa, Europe, and the Americas. Their stories emphasized the power of data-driven reporting, digital engagement, and innovative solutions to improve patient outcomes.

Mugwati’s initiatives, digital awareness campaigns, school outreach, advocacy for registries, and pushing for policy inclusion, echo these lessons. “Every time we educate a family, every time we help a patient access care, we’re contributing to a system that is accountable and humane,” he says. “That is exactly what NPF encourages in journalists and advocates: using information to empower and protect communities.”

By bridging local realities with global insights, Mugwati’s work demonstrates how young advocates can build resilient, patient-centered health systems even in resource-limited settings. His story highlights the universal need for recognition, early diagnosis, and equitable care for rare-disease patients, a mission that transcends borders and resonates with advocates worldwide.

“Advocacy is not easy,” he admits. “Sometimes it feels like no one is listening. But the resilience I’ve seen among people living with rare diseases in Zimbabwe keeps me going. Even if we reach one person at a time, that person becomes a lifeline for someone else. That is how change begins.” ■

A Zimbabwean Mother's Harrowing Battle With Myasthenia Gravis, a Rare Invisible Disease



JOHANNESBURG / MUTARE — When the first symptoms surfaced in April 2013, 28-year-old Letwin Nyakudya tried to shrug them off. Her speech began to slur, her eyelids felt unusually heavy and her body seemed to fatigue faster than her mind could keep up. “People looked at me and saw a normal woman,” she recalls. “But inside, my body was shutting down.” She never imagined that she was at the beginning of a five-year medical odyssey that would alter every part of her life, her autonomy, her relationships and even her sense of self.

As the months rolled on, her body continued to behave in strange, frightening ways. She started tripping over nothing, as though her legs had forgotten their instructions. Food sat in her mouth longer than it should, and swallowing, a simple act most people perform without thinking, became an exhausting task. Some mornings she woke up ready for work, only to fall back into bed moments later, held down by a crushing fatigue she could not explain.

Clinics in Zimbabwe gave her answers that never fit. She was told it was high blood pressure. She was told it was tonsillitis. She was told it was stress. Each visit ended with a new prescription but no relief. “I took all the medications they gave me, but I kept getting worse,” she says. As her health declined, rumours filled the vacuum created by the absence of a diagnosis. Some relatives whispered that she had been bewitched. Others believed her husband was mistreating her because she was losing weight so rapidly. “When people don’t see the illness on your body, they create stories,” she says quietly. “And those hurt more than the symptoms.”

Her independence, once taken for granted, slowly disappeared. Doing laundry became a struggle. Opening a peg felt like lifting a brick. Eventually, even bathing herself became impossible. Her young daughter, only seven at the time, began helping her dress, wash and move around the house. “You don’t want your child to wash you,” Letwin says. “But she did. She had to.”

Her condition worsened sharply. One night, a simple sardine lodged in her throat, leaving her gasping for breath. “I thought: this is it. This is how I die,” she remembers. Nurses again insisted it was tonsils and discharged her. By then, her right eyelid had drooped almost completely shut, and she was seeing double. When she asked a doctor what would happen if her other eye closed, she was told not to worry. “But how do you not worry,” she asks, “when you’re going blind in one eye?”

By 2017, after four years of being shuffled between hospitals without answers, a speech therapist finally urged her to push back. “She told me, ‘Next time, refuse the medication. Demand answers.’” Taking that advice changed everything. Letwin returned to the hospital and refused more prescriptions, forcing a reassessment and eventually a referral to a major facility.

In 2018, desperate for answers, she travelled to Johannesburg. There, on 17 October, a neurologist simply looked at her and named the condition that had stolen half a decade of her life: Myasthenia

Gravis, a rare autoimmune neuromuscular disorder that disrupts communication between nerves and muscles. “I finally had a name,” she says. “I wasn’t crazy. I wasn’t cursed. I wasn’t imagining things. I was sick.” Within two weeks of beginning the correct treatment, her strength slowly returned.



But the diagnosis marked only the beginning of a different kind of struggle. The medication she needed could cost up to R8,000 per month. The nearest hospital equipped to help her was nearly 90 kilometres away. And the drugs, while effective, came with debilitating side effects, severe acidity, painful cramps, weight gain and unpredictable daily weakness. “Some days my legs give in; some days my arms,” she says. “You learn to live hour by hour.”

Despite the challenges, she refused to surrender her aspirations. At the peak of her illness, she applied for a teaching job, completed a cake-decorating course and later earned a postgraduate qualification.

“You can die while you are still alive,” she says. “Or you can keep going.”

Public understanding of her condition has been limited. Because Myasthenia Gravis is largely invisible, Letwin is often judged harshly. At work, some colleagues assume she is exaggerating. Relatives, lacking accurate information about rare diseases, occasionally revert to cultural interpretations. The result has been emotional isolation and a shrinking social world. “I don’t have the energy to explain myself to everyone,” she says.

Fortunately, her three children have become her greatest source of support. “God knew what He was doing when He gave me those three,” she says. “They are my caregivers, my strength.”

Her long journey through misdiagnosis, stigma and delayed recognition shines a harsh light on the realities of rare-disease care in Zimbabwe and much of Africa. There is little public awareness, minimal training for frontline health workers, weak referral pathways and no national policy addressing rare conditions. “When HIV started, there were posters, songs, campaigns everywhere,” she says. “For rare diseases, nothing. People are suffering without knowing what they have.”

Her faith deepened during the most difficult days. “When you try everything and nothing works, you know only God remains,” she says. She also remembers meeting a woman named Elaine through an online support group, another Myasthenia Gravis patient taking the same medications. Elaine later died. “It taught me that medicine alone is not enough,” she reflects. “You need God to go through each and every day.”

Today, Letwin shares her story to educate others, not to seek pity. “Don’t feel sorry for me,” she insists. “Just learn. So you can help the next person.” She is writing a book about her experience and dreams of establishing a national support network for rare-disease patients in Zimbabwe.

Her advice to newly diagnosed patients is grounded in lived truth: accept the new reality, rest when the body demands it, protect your mental health and surround yourself with positivity. “You don’t need to prove anything to anyone,” she says. “Listen to your body. And never, ever give up.”

Her personal struggle echoes a wider global pattern. At the 2025 National Press Foundation Rare Disease Reporting Fellowship, a week-long programme of advanced training, expert briefings and in-depth Q&A sessions, advocates from around the world described similar experiences of misdiagnosis, stigma and disbelief. Rare-disease activist Yamina Hsaini, founder of “Yamina’s Life,” told journalists how she endured years of dismissal from doctors who insisted her symptoms were psychological. “All the doctors did was give me medication that didn’t work,” she said. “They kept saying it was all in my head.” Her testimony mirrors Letwin’s experience

and that of countless others navigating health systems unprepared to recognise rare and complex conditions.

What Letwin endured in silence for five years is a reality for millions across Africa and beyond. Her voice, steady, brave and unshaken, breaks through that silence. And as advocates like Hsaini continue to push for global recognition, their stories form a united call: for healthcare systems to see the unseen, name the unnamed and finally act on the suffering that has been ignored for far too long. ■

How One Man with Cowden Syndrome is Redefining Advocacy and Awareness



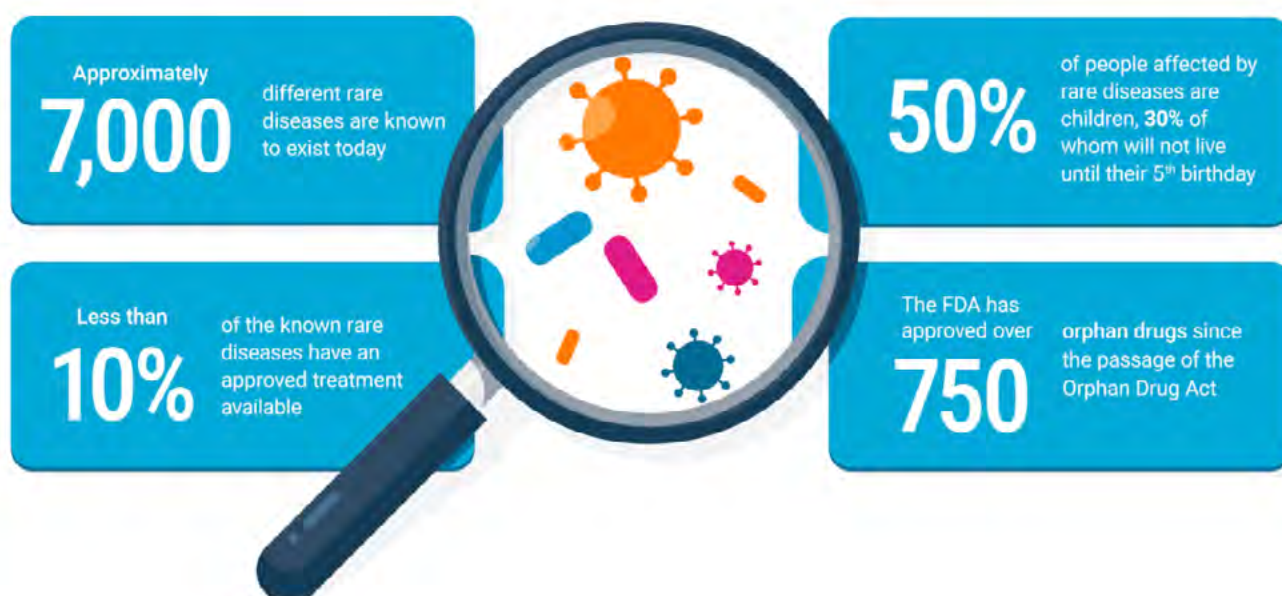
LONDON/MUTARE — At 43, Dave Hogan received a letter from his late mother that would change the course of his life. His mother had been diagnosed with Cowden syndrome, a rare condition affecting one in 200,000 people worldwide. A year before her death in 2015, she urged him to get tested. “I felt fine at the time, but after she passed, I realized I needed to know,” he recalls. Within three months of seeing a genetics counselor and doctor, he was diagnosed.

Cowden syndrome carries elevated risks for cancers, benign growths, developmental delays, and in many cases, autistic traits. “Some people seem not too badly affected, while others suffer severely,” Hogan explains. “A few of us can maintain full-time work; some can’t. It’s a spectrum.”

His diagnosis brought clarity but also new challenges. The medical system, while supportive, could not fully address the complexity of his condition. “Doctors don’t know enough about rare diseases, and that’s not necessarily their fault. Patients often know more than the doctors because we learn from support groups and patient communities in the UK, US, and beyond,” Hogan says.

Daily life with Cowden syndrome is a balancing act. Hogan experiences persistent leg pain, ongoing health monitoring, and social challenges. “Self-awareness has been key. It’s taught me my strengths and limitations and how to live with a rare condition,” he reflects. Stigma and misunderstanding are constant companions. “People often don’t understand or want to ask questions. Isolation is intense, especially right after diagnosis,” he says.

RARE DISEASES



Hogan has channelled his experience into advocacy. He has run Facebook support groups, participated in family councils, and attended charity events. “Mental health must be treated with the same priority as physical health,” he emphasizes. He also highlights the financial and personal costs of advocacy: “Many patient advocates fund their own participation in events or take unpaid leave for clinical trials. Rare-disease advocacy isn’t easy, and it can be exhausting.”

He calls for healthcare systems to better integrate patient voices. “Doctors need to involve patients in workshops, town halls, and decision-making spaces. Patients should be at the table, not fighting to be heard,” Hogan insists. He stresses the importance of joined-up care: rare-disease patients often juggle multiple conditions, requiring communication across specialists.

Despite these challenges, Hogan’s outlook is grounded in hope. “If you can accept having a rare condition, that’s half the battle,” he says. “You need time to process emotions, understand your condition, and learn how to live with it.” His advocacy underscores the value of community, conversation, and shared experience.

Hogan’s work aligns with the vision of the National Press Foundation (NPF), which seeks to inform, connect, and inspire journalists covering complex health issues globally. While Hogan was not part of the NPF fellowship, his advocacy mirrors the goals of the programme: to amplify rare-disease voices, highlight gaps in care, and drive public understanding.

Through his work, Hogan embodies the resilience, insight, and persistence that NPF encourages in journalists and patient advocates alike. His story sheds light on the unseen and unrecognized struggles of rare-disease patients worldwide. “Resilience is continuous,” he says. “Some days it’s exhausting, some days it’s empowering. But speaking up, connecting with others, and demanding better care, those are the victories. That is how change begins.”

Hogan’s voice, steady and unshaken, joins a growing chorus of global advocates urging healthcare systems to see the unseen, name the unnamed, and act decisively on the suffering that has too long been ignored.

For any questions you might have. ■

Ida Mirković Knaus Urges Stronger Advocacy & Policy For Rare Diseases



MUTARE, ZIMBABWE — Expert Associate and Secretary of Rare Diseases Croatia and Vice President of the Croatian Neurofibromatosis Association Advocate Ida Mirković Knaus, warns that fragmented advocacy and weak national frameworks leave rare disease patients underserved.

She was speaking to fellows on Day 4 of the 2025 National Press Foundation (NPF) Rare Disease Reporting Fellowship, held online from Nov. 17–21, where 62 journalists from around the world participated in a week-long series of advanced trainings, briefings, and interactive Q&A sessions. This year’s program carried the theme: “Reimagining the Future: Communicating What’s Possible in Rare Disease Innovation.”

“Without national rare disease strategies, everything becomes piecemeal,” Mirković Knaus told participants. “We need coordinated plans, registries, clear referral pathways, and sustained commitment. Advocacy must be organised, strategic, and aimed at policy change.” She stressed that rare disease advocacy cannot rely solely on passion. “Political will is essential. Families deserve visibility, structured care, and systems that function reliably. Patient advocacy groups must work strategically to

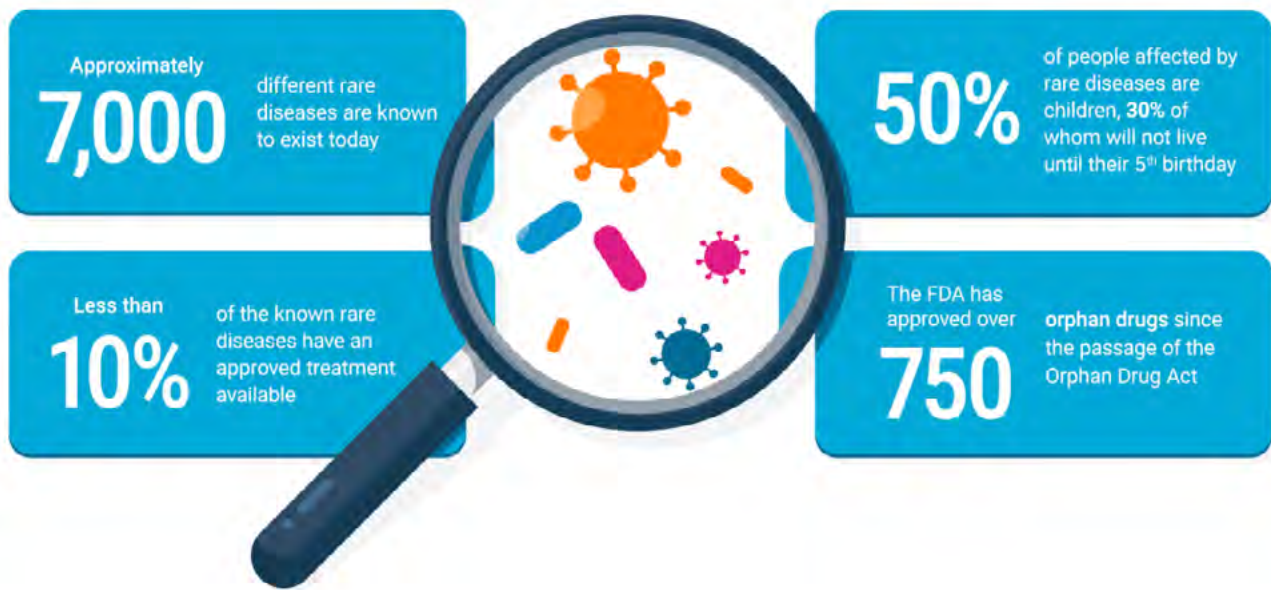
influence policy, gather data, and secure resources.” Although Mirković Knaus’s examples came from developed countries, Zimbabwean health-science journalist Luthando Mapepa told Post on Sunday that the challenges she described closely mirror conditions in Africa, particularly in Zimbabwe.



“What Ida described, fragmented advocacy, lack of registries, and absence of national strategies, is exactly what we see in most African countries,” Mapepa said. “Rare disease patients are often left to navigate complex systems alone. Families go without support, medical guidance, or clear treatment pathways.”

Mapepa shared a case from Chimanimani, Manicaland Province, involving a 23-year-old male patient who developed paralysis in his left limbs in 2015. The young man underwent extensive testing, including HIV, heart function, blood pressure, and diabetes screenings, all of which returned normal results. A CT scan in Harare, however, revealed an infarct in the right side of his brain. Doctors determined that his blood was abnormally thick and prone to clotting, though the underlying cause remains undiagnosed to this day. He was prescribed warfarin, a blood-thinning medication, with weekly INR monitoring.

RARE DISEASES



“The treatment carried significant risks,” Mapepa said. “Any injury, accident, or dental procedure could have caused excessive bleeding, even death. Unfortunately, financial difficulties later forced him to stop the medication, leaving him at continued risk.” Through consistent physiotherapy, the patient regained mobility in his legs, but he cannot use his left arm, highlighting both the long-term physical consequences of delayed and partial treatment and the gaps in Zimbabwe’s healthcare support system.

Mapepa said the Chimanimani case illustrates the urgent need for systemic changes. “We cannot leave patients to struggle on their own. The government and health sector must develop a national rare disease strategy, including a patient registry to track conditions. Diagnostic tools, such as CT scans and genetic testing, must be accessible and affordable beyond the capital. Rural clinics should have clear referral pathways to tertiary hospitals to ensure timely care. Financial barriers must also be addressed, so patients do not have to choose between essential medication and survival. Healthcare workers, communities, and patients’ families need

training and awareness programs to identify rare diseases early and support patients throughout their treatment journey.”

Mirković Knaus reinforced the importance of structured advocacy. “The most successful rare disease ecosystems, particularly in Europe, are built on decades of consistent policy development, patient-led advocacy, and systemic collaboration,” she said. “Change happens only when advocacy is strategic, coordinated, and backed by committed governments.”

Her presentation was part of the NPF fellowship’s week-long online program, which convened experts in health systems, genetics, patient advocacy, and emerging technologies to help journalists worldwide strengthen reporting on rare diseases.

Evans Jona, the author of this story, is a proud fellow of the 2025–2026 National Press Foundation Rare Disease Reporting Fellowship, joining a global cohort of journalists dedicated to telling these vital stories with both scientific rigor and humanity. ■

Zimbabwe's Rare Disease Patients Remain Invisible As Advocates Demand National Registry



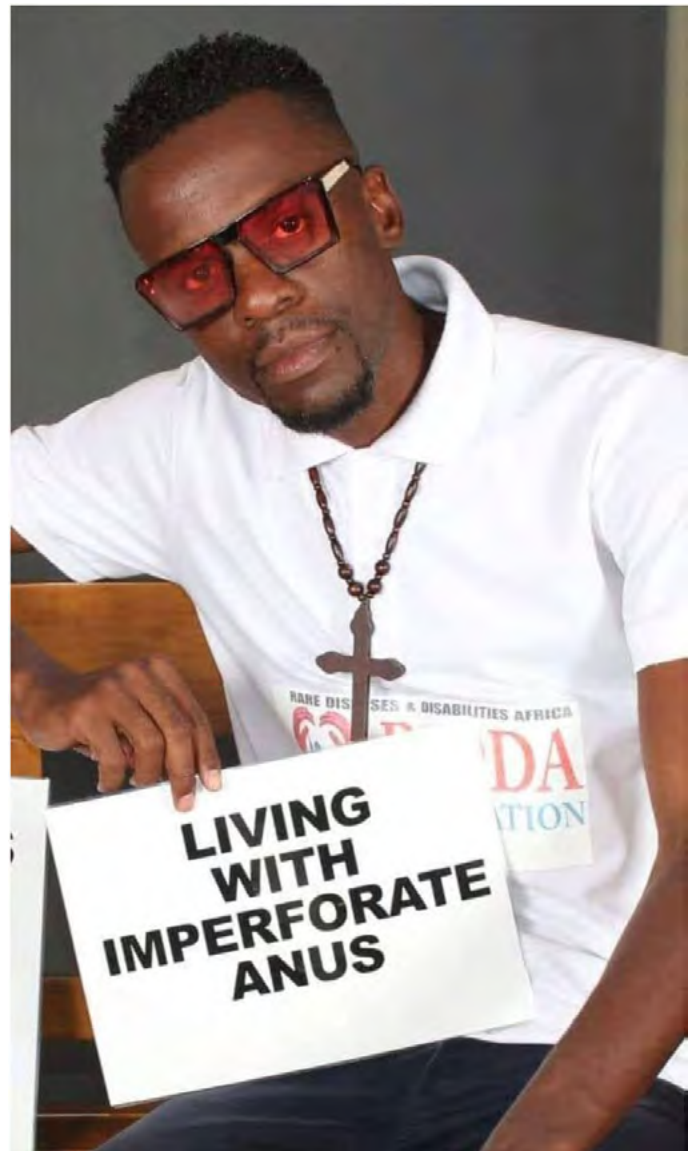
MUTARE — Zimbabwe's rare-disease patients remain largely invisible, uncounted and underserved due to the country's failure to establish a coordinated national registry or policy framework. This invisibility continues to expose families to misdiagnosis, stigma and catastrophic financial strain while global awareness of rare diseases grows rapidly.

According to the World Health Organization (WHO), more than 300 million people around the world live with rare conditions, yet many African countries, including Zimbabwe, have no national systems to identify or support them. WHO has acknowledged that this lack of documentation leads to delays in diagnosis, limited treatment options, and severe economic hardship for affected families.

In an exclusive interview with Post On Sunday, Rare Diseases and Disabilities Foundation Africa (RaDDA) founder Tinotenda Mudarikwa said the government "cannot plan for what it does not count," noting that many Zimbabwean rare-disease patients "do not exist on paper" despite living with life-threatening conditions. He emphasised that without accurate data, policymakers remain blind to the realities faced by families in every

province, leaving them to navigate complex and misunderstood conditions alone.

RaDDA has also been appealing for support for affected families who cannot afford assistive devices, medications or basic necessities. NewsDay Zimbabwe has previously reported on cases where individuals living with rare diseases rely entirely on donations to obtain wheelchairs, crutches, white canes or even food. Mudarikwa said these challenges reflect the urgent need for a national system that recognises and responds to the burden of rare diseases.



At the 2025 National Press Foundation (NPF) Rare Disease Reporting Fellowship, a week-long series

of advanced trainings, briefings and Q&A sessions with international experts and advocates, global youth voices underscored how misdiagnosis and stigma can devastate lives. Rare-disease activist and advocate Yamina Hsaini, founder of “Yamina’s Life,” shared her painful journey of misdiagnosis, recounting how doctors dismissed her symptoms as psychological.

“All the doctors did was give me medication that didn’t work. They kept saying it was all in my head,” she told journalists. Her story mirrors what many Zimbabwean patients experience when they encounter clinicians unfamiliar with rare or complex conditions.

Hsaini explained that social media became a turning point in her journey, saying, “Social media didn’t just give me a voice, it helped many people get the right diagnosis on time.” She urged journalists to embrace their responsibility in shaping public understanding, adding, “As journalists, you have the power to make the invisible visible. Your reporting can truly change lives.” Her message has been welcomed by Zimbabwean advocates, who say that responsible storytelling remains one of the few tools available to bring attention to neglected conditions.

The National Press Foundation notes that the world is entering a transformative moment for rare-disease reporting and scientific innovation. The organisation highlights that for the 300 million people living with rare diseases globally, this period marks an unprecedented era of new tools, treatments and understanding.

However, Zimbabwe risks being left behind if it continues to operate without proper records, diagnostic pathways and policy structures.

Isolated research in Zimbabwe offers glimpses into the hidden burden. A landmark study published

in the Journal of Medical Genetics found that oculocutaneous albinism affects approximately one in every 4,728 schoolchildren, signalling significant dermatological and visual-health needs. In 2024, a sickle-cell newborn screening pilot tested 550 infants, the country’s first attempt at early detection of genetic blood disorders, yet no national screening programme exists. International analyses, including a 2023 review in PLOS Digital Health, highlight that rare diseases remain largely unconsidered in African health-data systems, leaving millions untracked.

Families living with rare diseases in Zimbabwe often carry the burden alone. According to Gender Links Southern Africa, many patients experience discrimination, social exclusion and extreme emotional strain, made worse by cultural misconceptions that link unexplained illnesses to curses or witchcraft. Parents frequently travel long distances in search of answers, depleting their savings while enduring repeated misdiagnoses and limited access to specialised care.

In Zimbabwe, advocates like Mudarikwa argue that a national rare-disease registry would transform the landscape by identifying affected individuals, informing resource allocation, guiding procurement of essential medicines and devices, and supporting health-worker training. They warn that without such a system, Zimbabwe will continue to overlook some of its most vulnerable citizens.

As the global rare-disease community moves forward with new technologies, treatments and advocacy models, Zimbabwe’s rare-disease patients remain on the margins of visibility. Their stories, advocates say, must be heard.

As Mudarikwa stated, “If we cannot count the patients, we cannot care for them.” ■

National Press Foundation Fellows Learn from James Levine: Telling Rare Disease Stories with Heart and Science

Millions of people worldwide live with rare diseases, conditions so uncommon that many remain undiagnosed, misunderstood, and invisible to the public. Yet during the 2025–2026 National Press Foundation (NPF) Rare Disease Reporting Fellowship, journalists from across the globe gained firsthand insight into the complex scientific and human landscapes of rare diseases, guided by James Levine, President of Fondation Ipsen.

“This is a massive unmet medical need,” Levine told fellows, emphasizing the dual responsibility of science and storytelling. Fondation Ipsen, a nonprofit organization, supports education, advocacy, and research for rare diseases, reaching millions through publications, digital platforms, and collaborations with over 240 organizations worldwide.

The NPF Rare Disease Reporting Fellowship, now in its latest 2025–2026 cohort, selects accomplished journalists and emerging reporters to deepen their understanding of rare disease science, advocacy, and policy. The program provides fellows with unparalleled access to leading experts, patient communities, and real-world case studies, equipping them to tell stories that can shape public awareness, policy, and global funding priorities.

Levine’s session, *Reimagining the Future: Communicating What’s Possible in Rare Disease Innovation*, outlined the critical role journalists play in bridging science and society. Rare disease stories are often buried under technical jargon, delayed diagnoses, and fragmented healthcare systems.

“The oxygen of science is suffocated in jargon,” Levine said. “Your journalistic capacity is the oxygen we need in the rare disease community.”

Journalists were urged to report with precision and heart, balancing complex scientific advances, gene

therapies, with the human stories of patients and caregivers. Levine highlighted the courage and resilience of people living with rare diseases, from Claude, who ran 55 kilometers for his birthday despite his condition, to Maxine, an author with autism, and Sophia, a student in Cape Town whose only educational resource is a rare disease advocacy magazine reaching 300,000 children.

Levine addressed common myths and misconceptions: rare diseases are not always congenital, not always visible, and even when treatments do not exist, hope remains through supportive therapies and emerging innovations. Journalists were provided with a practical framework for impactful storytelling, emphasizing the importance of working closely with rare disease associations and using trusted scientific sources such as PubMed.

Fellows also explored challenging ethical questions raised by patients themselves, including how to share personal stories without fear of stigma, and how to balance hope with the risk of false expectations in regions with low scientific literacy. Levine emphasized that each story matters, not only for raising awareness but for shaping policy, funding, and access to therapies worldwide.

“This fellowship equips journalists to be the bridge between science and society,” Levine said. “Report with your heart, report with your mind, and help us mobilize impact.”

For the journalists at the National Press Foundation fellowship, rare diseases are more than medical conditions, they are human stories waiting to be told, understood, and acted upon. Through careful, compassionate reporting, these fellows now have the tools to make the invisible visible, bringing hope and understanding to millions. ■



Stephen KENECHI

in TheCable | Nigeria



Stephen Kenechi is a senior multimedia journalist with over five years of experience in high-impact report-ing focusing on the intersection of public policy, socio-economic vulnerabilities, and humanitarian crises. As a Desk Head at TheCable, he led

projects that drove tangible public accountability. His work is marked for its ethical rigour and human-centred narrative. Stephen is passionate about deploying in-depth report-ing to address the global health equity crisis as it affects Africa.

Nigeria struggles with essential care for bleeding disorders as global medicine moves to gene therapy



Inside the living room of a home in Nigeria's up-north city of Kaduna, a newlywed couple looked at their third-born son and named him Onwuzueike, which translates in the Igbo language to "death must cease".

It was less a nomenclature and more a desperate petition.

They had already buried two sons before their tenth birthdays, one lost to a harmless bike accident and another to what could have been a routine eye surgery.

Both were victims of an unceasing bleed that family rumours had instead attributed to a "spiritual curse". But for the third son, Victor Ugochukwu, the curse would later find a name: Haemophilia A.

He was born into a body with a "Factor 8" protein level of 0.01%, a misfortune that meant a fall at age 5 wouldn't just result in a bruise, and then a year-long bedriddenness and two horrific encounters with an unknowing bone-setter who cracked apart his blood-stiffened knees without anaesthetic.

"I might as well have died that day," said Ugo, now 31, sighing and shuddering at the memory of being subdued by hefty men, the swollen legs bent so far backwards that his heels touched his thighs.

What the bone-setter did not know was that Ugochukwu's knee joint was not stiffened from structural damage but was internally drowning in blood, a pool no amount of traditional manipulation could drain.

When bodily trauma causes tissue rupture, a clotting mechanism naturally plugs the wound to stop any bleeding in minutes. This "coagulation" is an autonomic response for most people, but not patients with haemophilia, an inherited disorder in which wounds don't clot properly, leading to prolonged bleeding.

The prevalence of haemophilia is estimated at 1 in 10,000 live births, affecting up to **465,000 individuals** globally, according to data from the World Federation of Haemophilia.

Haemophilia A, which occurs when a clotting

protein called “Factor 8” is dangerously below 50% in the blood, is the most common form of the disease. It impacts about 1 in 5,000 to 10,000 male births.



Victor Onwuzueike Ugochukwu tutoring students

The condition affects **between 11,000 and 20,000** Nigerians, with over 90% still undiagnosed and some 946 patients already identified, according to state sources and data from independent non-profit research.

TURNING TO TRADITIONAL MEDICINE

In Nigeria’s south-eastern state of Enugu, Josephine, 38, is a mother of three boys with Haemophilia A.

The first she heard of the disorder was after her first son, Chidebere, was cut, and a doctor informed her of the condition. The doctor himself had doubted that an African could really present with what he had deemed to be “white people’s sickness”.

Eight days after a local midwife helped bring the boy to life, the circumcision bleed would not stop.

“The child was in diapers. The whole diaper was soaked with blood. He was pale. His eyes were wide, darting left and right. We knew something was wrong,” Josephine narrated.

Transfusing the baby with “factor-rich” blood helped clot the bleed, but this was just the beginning.

In the sixth week, Josephine would accidentally nick the baby’s flesh while trying to trim his fingernails, and the tiny cut would again not stop bleeding.

Panic set in when the toddler got knocked down a

flight of stairs in pre-school, falling on his knee.

A warm massage only doubled the swell by morning. And while a doctor confirmed the knee wasn’t broken, he lacked the words to explain why it was “ballooning”.

Desperate for answers the clinic couldn’t provide, Josephine turned to a local bone-setter whose attempt to stretch out the knee only “worsened” the internal bleeding.



Empty vial of factor 8

It took the bone-setter’s own bafflement and a fortuitous referral to a teaching hospital to finally replace traditional ointments with the science of haematology.

When Chidiebere was injected with a vial of donated Factor VIII protein by the non-profit Haemophilia Foundation of Nigeria, the agony that had defied every herb and bandage vanished in mere minutes.

Haemophilia is a **sex-linked disorder** carried on the “X” chromosome. Because males have only one X chromosome (XY), inheriting a single defective gene from their mother causes the disease.

Women have two X chromosomes (XX). A healthy gene on one compensates for a defective one on the other, only making the woman a carrier. She may show no symptoms, but still has a 50% chance of passing the hemophilic gene to her sons and a 50% chance of also making her daughters carriers.

According to **medical research**, about one-third of babies diagnosed with haemophilia have no pre-existing family history of the disorder,

signalling **spontaneous mutations** as culprits.

The WFH database shows Nigeria had 758 Haemophilia A patients as of 2024, 33% of whom were between the ages of 5 and 13. Some 30% are in adulthood, aged between 19 and 44.

Josephine's own parents had three male children, none of whom were hemophilic. Her sister had four boys, none of whom were also hemophilic.

MEDICATION PRICED BEYOND REACH

The treatment options for haemophilia in Nigeria are primarily **clotting factor** concentrates, which are administered either on-demand during crisis or at intervals as low-dose "prophylaxis" to prevent bleeds.

There are also newer but expensive non-factor therapies like the monoclonal antibody **Emicizumab**, whose longer-acting effect, clinicians say, could give haemophilia patients a totally normal quality of life.

But access is severely challenged by a lack of government subsidisation, forcing patients to perennially rely on donations by international non-profits due to the prohibitive cost of commercial purchase.

Market research says a 30mg vial of Emicizumab costs up to \$3,500, about N5.25 million at N1500/USD. Some patients, haematologists confirm, require up to seven vials a month, depending on body mass.

A 500 IU vial of the "Factor 8" concentrate could cost up to \$1,500 at commercial price. A standard dose to stop a medium-severity bleed for an average adult in on-demand treatment can be up to 1,500 IUs.

Austin Usifo, the head of marketing at Biofem Group, said the pharmaceutical company opted out of importing haemophilia medication due to the lack of substantial commercial demand for the products.

Unsubsidised commercial access is so prohibitively expensive that families are forced to choose between financial ruin, waiting on donations, and a patient's severe disability or death.

Despite the universal coverage mandate of Nigeria's National Health Insurance Authority Act, the **novel legislation** excludes high-cost treatments for rare conditions like haemophilia.

Factor VIII and IX coagulants **are listed** among essential medicines to which health institutions must ensure access, but they are excluded from the _____, meaning insurance does not cover them.

In 2022, **Nigeria captured** bleeding disorders in its national non-communicable diseases strategic plan. But an implementation guideline is required before state funds can be set aside for factor procurement.

LIVING DANGEROUSLY ON DONATIONS

Sanusi Omobolaji, 32, had just returned to his home in Ogun state from a long-dreaded trip to Akure, the Ondo state capital. In under 24 hours, the adrenaline of travel had given way to the throbbing heat of a spontaneous ankle bleed.

He was stranded; the empty Factor 8 vial on the creative designer's work station had been his last. And the nearest replenishment was only in a 4-hour traffic-fraught trip to Lagos; he could no longer manage.



Creative Designer Sanusi Omobolaji in Ogun

Omobolaji lives in a clinical blind spot. In the last 12 months, he has not attended his quarterly haemophilia clinic at the Lagos University Teaching Hospital, where he could have received a take-home packet.

He moves with a cane, favouring a right knee that has bowed under the weight of decades of internal

haemorrhaging, admittedly unable to walk for up to ten minutes without his body demanding a rest.

“All my joints, both major and minor, are affected by deformity,” he noted with a sobering honesty he rarely affords himself. “Whether sleeping, lying down, walking, or sitting, I’m constantly in pain.”

For Omobolaji, care is not merely about the existence of medicine, but also the ability to survive the terrain required to reach it. He was considering the difficult interstate trip to the Lagos University Teaching Hospital (LUTH). Unknown to him, at that time, even the hospital’s model haemophilia treatment centre at Idi-Araba, Lagos, was out of stock.



TheCable, Index	
Haemophilia Treatment Centres in Nigeria	
South-West	
Lagos	Lagos University Teaching Hospital
Oyo	University College Hospital Ibadan
Oyo	Lautech Teaching Hospital
Ondo	State Specialist Hospital
Osun	OAU Teaching Hospital
South-East	
Enugu	University of Nigeria Teaching Hospital
South-South	
Rivers	University of Port Harcourt Teaching Hospital
Edo	University of Benin Teaching Hospital
North-West	
Kaduna	Barau Dikko Teaching Hospital
Kaduna	Ahmadu Bello University Teaching Hospital
Kano	Aminu Kano Teaching Hospital
Sokoto	Usmanu Danfodiyo University Teaching Hospital
Katsina	Federal Medical Center
North-Central	
Kwara	University of Ilorin Teaching Hospital
Abuja	National Hospital
Kogi	Federal Medical Centre
Nasarawa	Federal Medical Centre
North-East	
Gombe	Gombe State University Teaching Hospital
Bauchi	Federal Medical Centre

Source: HFN

LUTH is one of the 17 hospitals designated by the Haemophilia Foundation of Nigeria as an HTC. LUTH’s haemophilia clinic, as of January 2026, reported 136 registered patients, with about 70 in active care, meaning they must undertake two to three clinical contacts in any year for checkups and education.

HTCs receive a quarterly shipment of WFH products through the HFN, alongside training, to provide comprehensive care for patients, ideally “supposed” to include orthopaedic, physiotherapy, mental health counselling, and social support.

But donations from WFH’s humanitarian **aid programme** have been the sole source of life-sustaining factor concentrates available to these patients and all others among the 946 registered with Nigeria’s 17 HTCs, an anomaly even the global non-profit itself admitted it has been fighting to correct and discourage.

A donation cycle begins with securing a NAFDAC nod to import, followed by a standoff with Customs at an airport cargo terminal.

Once liberated from the tarmac, the temperature-sensitive vials are hauled inland on a cold truck to the Haemophilia Foundation of Nigeria’s headquarters in Kaduna, from where they are distributed to HTCs.

And not even the coveted donor products are spared misfortune.

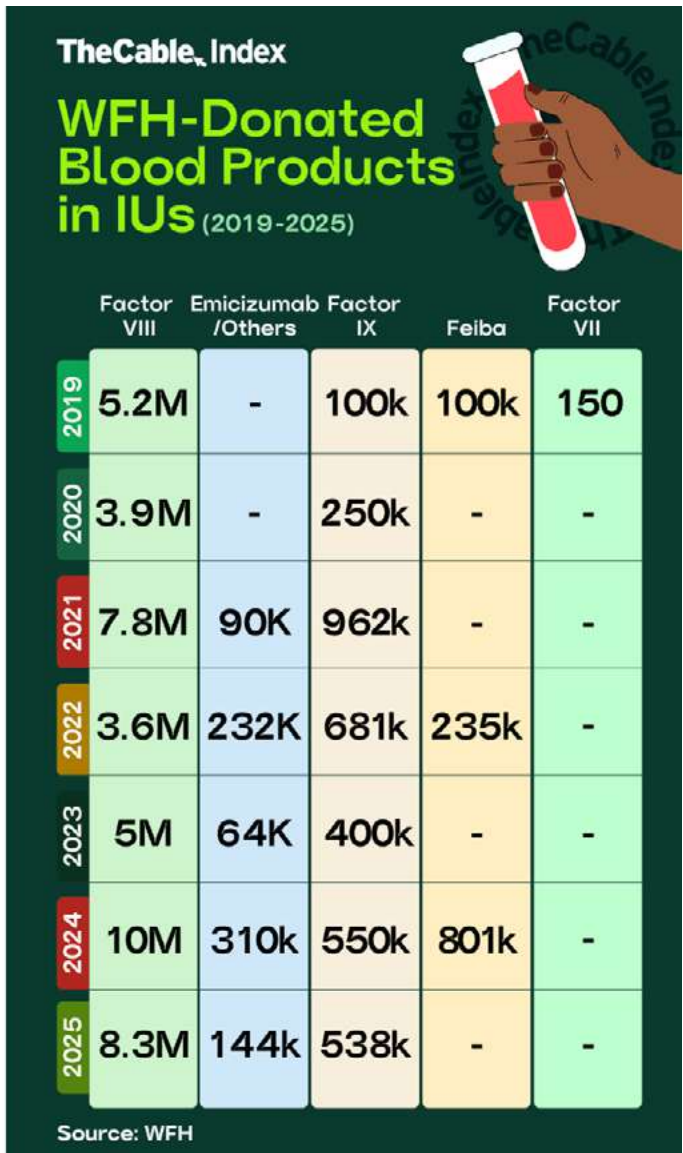
Benjamin Clousier, communications manager at the WFH, said the shipment approval timeframe with Nigeria’s health authorities has significantly increased to three months, stalled by demands for documentation “unrelated” to core clearance priorities like the medicine’s safety and efficacy.

This drag, he noted, has forced the UN-affiliated non-profit to slash its shipping frequency from quarterly to twice a year.

“Several patients are on prophylaxis. If, for any reason, the donations were delayed, these Nigerians will have to stop treatment, which will expose them to life-threatening bleeds,” he stated in an email.

The resultant defaults in quarterly shipments, the director of LUTH’s HTC, Titi Adeyemo, said, force the treatment centre to reserve prophylactic care for

only children under age 12.



It means the adult patients do not have access to preventive prophylaxis.

“They’re treating bleeds when the standard of care should be prevention,” the doctor said. “More bleeds mean more joint disease. Eventually, the joints fail, and you start using a walking aid or wheelchair.”

DIAGNOSTIC CAPACITY STILL A LUXURY

For Iyabo Akinleye, the true cost of Nigeria’s healthcare deficit can be measured in the several years of clinical guesswork she had to endure.

As a kid, she bled a lot, enough so that her parents panicked. After immunisation shots in infancy, she bled. When she was teething at 6 months old, she bled. During tooth-shedding at 7, she also bled. Every growth milestone meant a frantic race to the

hospital ward.

When a seven-year-old Iyabo was put to bed at her family’s Kaduna home after a chloroquine injection for malaria, her mother expected the restorative sleep of a recovering child.

Instead, she woke to find her daughter in a pool of red. What she had mistaken for a child wetting the bed turned out to be blood slowly draining out from a needle site that a simple tuft of cotton wool should normally have sealed.

Iyabo was thereafter subjected to a strict list of Dos and Don’ts, as her worried parents sought answers.

Puberty would usher in unusually heavy, long-lasting menstrual periods that had her skipping school.

In search of a diagnosis, she was at the Obafemi Awolowo University Teaching Hospital in Osun, the University of Ilorin Teaching Hospital in Kwara, and the Ahmadu Bello University Teaching Hospital in Kaduna.



Iyabo Akinleye

“I told them I had a bleeding problem at the time. They didn’t know what was wrong,” Iyabo recounted.

Nighttime offered no reprieve, as Iyabo would wake several times to expel blood that had pooled in her throat while she slept. A doctor had hinted at haemophilia. But it was with repeated therapeutic trials that her condition would be pronounced with fairly reasonable certainty as “Von Willebrand’s Disease”.

Research says VWD is a more common bleeding disorder **affecting** 1 in 100 people. In Nigeria, however, there were only 18 **confirmed cases** as of 2025, a reality experts attribute to a lack of diagnostic capacity.

VWD patients either lack or are low in a clotting protein called the von Willebrand factor, or it doesn't work properly. For a person to make a clot, VWF binds to factor 8, another clotting protein, and platelets in blood vessel walls. In a normal person, this process forms a platelet plug, enabling a bleed to cease.

Iyabo was subjected to a panel of basic blood tests that established she had 7 IU/dL of the VWF, where normal levels should be between 50 and 200. None of the hospitals she visited could conduct the gene tests required to establish why her VWF levels were low and, therefore, determine her type of VWD.

Titi Adeyemo, LUTH's HTC director, said Nigeria's tertiary hospitals and labs lack the infrastructure to reliably test for bleeding disorders, with even privately-run clinics resorting to sending samples **abroad**.

"Hence, we rely on therapeutic trials. There are different bleeding disorders and different treatment options. If we give them one treatment and it works, we can focus on it. This is how we've been going."



Titi Adeyemo, LUTH HTC director

A coagulometer is a machine needed to measure blood clotting and diagnose bleeding disorders like haemophilia. But it was only in 2025 that LUTH acquired one through a donation from the Danish non-profit **Novo Nordisk**, allowing it to conduct factor assessment and name a condition, but not much else.

Nigeria's maternal mortality is high, with UN data showing that one in 100 women dies in labour or in the following days, and postpartum haemorrhage is **identified** as one of the culprits.

Iyabo, now 39 with two dissuasive childbirth experiences that had her family on edge, thinks bleeding disorders could be a factor. Her fear is that Nigeria may never know for sure without data and the proper diagnostic infrastructure.

NIGERIA SIDELINES GENE THERAPY AS PRICE TAG STALLS ACCESS

Haemophilia is due to a missing or defective gene that prevents the body from making a clotting protein.

Beyond "miracle" injections like Emicizumab that rebalance the system, WFH data shows **gene therapy** offers a longer-lasting treatment enabling the body to naturally manufacture the missing clotting factor.

As the delivery medium or "vector", scientists use "**harmless**" Adeno-associated viruses, infused with a working copy of the missing clotting gene.

The modified virus is delivered to a patient via a single intravenous injection, travelling through the blood to the liver cells, which are the body's natural factory for clotting factors. The gene, once added, remains in the cells while the vector leaves the body through urine, faeces, blood, saliva, and semen.

Factor levels, successful clinical trials show, begin to rise after three to four weeks of gene therapy.

Australia's **Mark Lee** is one of the most famous success stories for haemophilia treatment with gene therapy. After receiving the injection in a trial, his clotting factor levels were reported to have moved into the normal range. He went from needing factor infusions thrice a week to living without any bleeds.

In the US, three landmark gene therapies have been approved for haemophilia care, including **Hemgenix** and **Beqvez** for Hemophilia B, along with **Roctavian** for Hemophilia A.

The therapies, priced between \$2.9 million and \$3.5 million per dose, are transforming haemophilia care.



In the UK, trials at the University College London and St. Jude Children’s Research Hospital **reported** that haemophilia patients have maintained stable clotting levels for over 13 years after a single dose.

Due to variations in outcome, clinicians say the medical community is focused on long-term monitoring to see if the clotting factor levels produced by these “one-time” shots would stabilise over the decades.

Interestingly, artificial intelligence is emerging for personalised treatment and usage in conjunction with point-of-care ultrasound to quickly detect early joint damage for inherited disorders like haemophilia.

But Megan Adediran, founder and executive director at the HFN, said the prohibitive costs, gaps in infrastructure, and competing health priorities hinder Nigeria’s deployment of these innovations.

Emmanuel Kolawole, researcher at the University of Connecticut, said demand for pricey gene therapy is low even in high-income markets, citing the pharmaceutical firm Pfizer’s **discontinuation** of global development for Bequez.

Afees John Olanrewaju, a medical scientist at Nigeria’s Babcock University, worries there is limited effort by big pharma to facilitate licensing, regional manufacturing, and competitive pricing in the global south.

A geneticist, Christian Happi, in a lab on the red earth of Osun state, pioneered a landmark, albeit donation-funded, public health programme using CRISPR to surveil for deadly diseases.

Yet, Nigeria has not notably begun exploring this gene editing tool in actual treatment, beyond fielding patients for international clinical trials.

At LUTH, Titi Adeyemo watched these gaps consume a patient who, after a lifetime of undiagnosed agony, presented with a leg so ravaged by gangrene that only amputation could save him. Insisting he would rather “die whole”, Michael, 22, signed himself out into the silence of a certain death.

“I felt very hurt,” the surgeon admitted, wincing from behind her office desk. “You wonder if you could have done more.”

Access to even the most essential haemophilia care in Nigeria remains poor, devastating the education, sex life, and mental health of many, alongside the already strained finances of their families. ■



Australia’s Mark Lee

A study of Haemophilia A patients three years after Roctavian treatment **reported** that average Factor VIII levels were 42% after 1 year, dropped to 23% in 2 years, and dipped further to 15% in year 3.



Anna LARSSON

in Swedish National Radio | Sweden



Anna Larsson has studied journalism at the University of Gothenburg, and she also studied medicine at Karolinska Institutet in Stockholm to get a better background for medical journalism.

She was employed by Swedish National Radio, where she still works as a medical reporter. Her

duties are mainly to cover news in the medical field and health care. She has published hundreds

of radio documentaries, reportages on air and articles at the website of Swedish Radio.

She is rewarded for her work several times, recently by the Foundation for children's cancer in Sweden for excellence in

reporting on difficult subjects in cancer treatment and care.

In February 2022 she defended her thesis at Karolinska institutet, on the subject

‘Medicine and the Media: to communicate medical news to those who need it most’.

Anna Larsson lives in Stockholm.

Adam lider av sällsynt sjukdom – ny medicin ger hopp



Audio [here](#)

Rare diseases: Long wait in Sweden for new medications for Adam and other boys

Audio [here](#)

Sällsynt diagnos: Vidar fick vänta i två år på rätt behandling



Audio [here](#)



Leonard MASAULI

in Malawi News Agency | Malawi



Leonard Masauli – Investigative Journalist, Malawi

Leonard Masauli is an enterprising and passionate investigative journalist from Malawi, dedicated to un-covering stories that inspire change and accountability. With over five years of experience at the Malawi News Agency (MANA) under the Ministry of Information and Digitalization.

He holds Bachelor of Arts in Journalism and Mass Communication from Amity University in India and both his education and professional

experience have given him a global outlook and deep insight into local ground realities.

Leonard’s journalism spans a variety of science, Health, climate change, development, and social justice issues that are typically underreported in the mainstream media. His work has been published both locally and internationally with reputable outlets such as The Guardian, FairPlanet, and El País, amplifying Mala-wian voices and views to be heard abroad.

Genotype testing before marriage could curb sickle cell burden

As Malawi grapples with limited specialist care and low awareness of genetic disorders, patients living with sickle cell disease continue to endure pain, stigma and high treatment costs, raising questions about whether premarital genotype testing could help protect future generations.

Sarah Charity Kumwenda, 30, from Luwinga Township in Mzuzu, knows the burden all too well.

She and her sister are living with sickle cell disease, while their first-born brother died from the condition after inheriting the gene from their parents.

Growing up, Sarah says she faced stigma and discrimination both from teachers and fellow learners.

“Sometimes my eyes turned yellow and that attracted insults. I was forced to change schools just to continue learning in peace,” she recalls.



Despite the challenges, she completed secondary school at Our Lady of Wisdom and was selected to pursue Computer Science at Chancellor College, a constituent college of the University of Malawi.

However, in 2017 she fell seriously ill and stayed away from campus for two years, eventually failing to continue with her studies.

After recovering, she moved to South Africa where she enrolled at Damelin University in 2023 to study a Diploma in Information and Communications Technology.

Sarah says managing sickle cell disease is expensive and emotionally draining. Doctors have advised that a bone marrow transplant in India, costing over US\$25,000, could cure her condition — an amount far beyond her family’s reach.

Efforts to raise funds through online campaigns have yielded minimal support.

Their mother, Rank Nkosi Kumwenda, a single parent of six children, says caring for three children born with sickle cell has been financially and emotionally exhausting.

“When they fall sick at the same time, it is frightening. Sometimes hospitals run out of medication, and buying it privately costs about K80,000 for three months,” she says.

She recently graduated with a Master’s degree in Education from Mzuzu University, hoping to inspire her children to persevere.

Sarah now runs an online advocacy platform called “Sarah the Sickle Cell Warrior” to raise awareness and calls on government to train more specialists and expand access to genotype testing.

According to a Ministry of Health press release issued during World Sickle Cell Day commemorations on June 19, 2025, about 0.1 percent of Malawi’s population lives with sickle cell disease, while 9.1 percent carry the trait.

Globally, about 7.7 million people live with the disease and an estimated 300,000 children are born with the condition each year, with over 90 percent of cases occurring in Africa. ■

Beyond the ‘bad spell’: the lonely fight for Malawi’s children with rare diseases

Machinga, February 26, Mana: In many communities across Malawi, rare diseases such as spina bifida, hydrocephalus, and sickle cell disease are widely perceived as bad spells or divine punishment.

Such beliefs fuel stigma, isolate families, and delay timely care-seeking for children born with life-altering conditions.

Estimates from Child Help Malawi suggest that more than 20,000 Malawians live with spina bifida and hydrocephalus, but only about 3,000 access hospital-based care, leaving the majority without consistent medical support.

For Rose Amos from Bangwe Township in Blantyre, the struggle is deeply personal. She is raising her eight-year-old son, who was born with both spina bifida and hydrocephalus.

According to Stanford Medicine Children’s Health in America, Spina bifida is a birth defect in which part of the spinal column fails to form properly, leaving spinal nerves exposed.

On the other hand, Hydrocephalus involves the accumulation of excess cerebrospinal fluid in the brain due to disrupted formation, flow, or absorption.

According to Amos, the diagnosis puts her in fear and isolation with people in the community believing that the disease is a result of witchcraft or punishment for past wrongs.

“I could not take it at first and I believed that it was a bad spell cast on me. It took me time to understand what is happening. Even my relatives left me alone; but with time, they accepted the situation,” she says.

Amos further laments that discrimination is high in schools as some refuse to enroll children with such conditions.

“I would like to call on government to recognize children with rare diseases in the same way it supports persons with disabilities, ensuring equal

access to healthcare, education, and medication,” Amos says.

She adds that accessing care remains an uphill battle as hospitals often lack proper medication and equipment for surgeries related to her son’s condition.

But life begins to change for Amos after she connecting with Child Help Malawi, a Blantyre-based organisation that provides a lifeline by offering primary healthcare, specifically tailored for spina bifida and hydrocephalus.

Beyond medical treatment, they empower Amos through caregiver training and provision of the essential equipment her son needs to lead a healthier, more dignified life.

“They teach us how to care for our children properly and encourage us to keep going. Without the organization’s efforts, it would be an impossible situation,” she explains.

The organization’s Executive Director, Blessings Chapweteka, says government’s efforts on rare diseases are falling behind as compared to priorities given to common conditions.

He says, currently, it is only non-governmental organizations taking lead to fight rare conditions, thereby creating an imbalance because NGOs drive the agenda without clear national policies on management and awareness.

He explains that many communities continue to view Spina bifida and hydrocephalus as curses linked to witchcraft or divine punishment which, at times, fuels stigma and discrimination.

“To counter this, we work with communities and support government hospitals with surgical materials such as shunts. We also provide continence management training for children who cannot control urine or stool because an issue of surgery alone does not fix.

“We do this so that the children can stay dry, attend school, work, and interact with others,” he says.

Beyond treatment, the organization provides a number of interventions such as distributing folic acid supplements to mothers to help prevent Spina bifida; training healthcare workers; conducting advocacy and community sensitization: offering social support, and carrying out clinical outreach and home visits.

Chapweteka stresses that prevention and long-term care require serious investment, saying it costs up to 791,200 US dollars—about (K137 billion) to care for one person over a lifetime.

He calls for intensified community awareness, expanded training for healthcare workers, and national management guidelines that go beyond surgery to include continence care, physiotherapy, and rehabilitation.

He warns that NGO-led interventions are not sustainable because NGOs can pull out at any time and that Government must treat this as a ticking time bomb.

Health expert Maziko Matemba agrees, noting that Malawi faces deep structural challenges in managing rare diseases.

He says the country’s per capita health expenditure stands at about 40 US dollars—far below the World Health Organization recommended 86 US dollars for low-income countries.

“We need serious health financing reforms. Malawi also needs specialized hospital facilities to manage these conditions if we are to achieve universal health coverage by 2030, a commitment the country made at the United Nations General Assembly in 2019,” Matemba says.



The Ministry of Health had not yet responded on the matter as this story went to press.

In Malawi, more than 60 percent of health financing comes from external sources.

For Rose Amos, the goal of Universal Health Coverage by 2030 is not a distant policy figure—it is the lifeblood of her eight-year-old son.

As she continues to navigate a landscape of scarce medication and crowded hospital corridors, she

represents a growing voice demanding that the government step out from the shadow of NGO-led aid.

Until rare diseases are integrated into the heart of Malawi's national health agenda, families like hers will continue to fight their battles in the dark, waiting for a dawn that only systemic change can bring. ■



Ferdinand MBONIHANKUYE

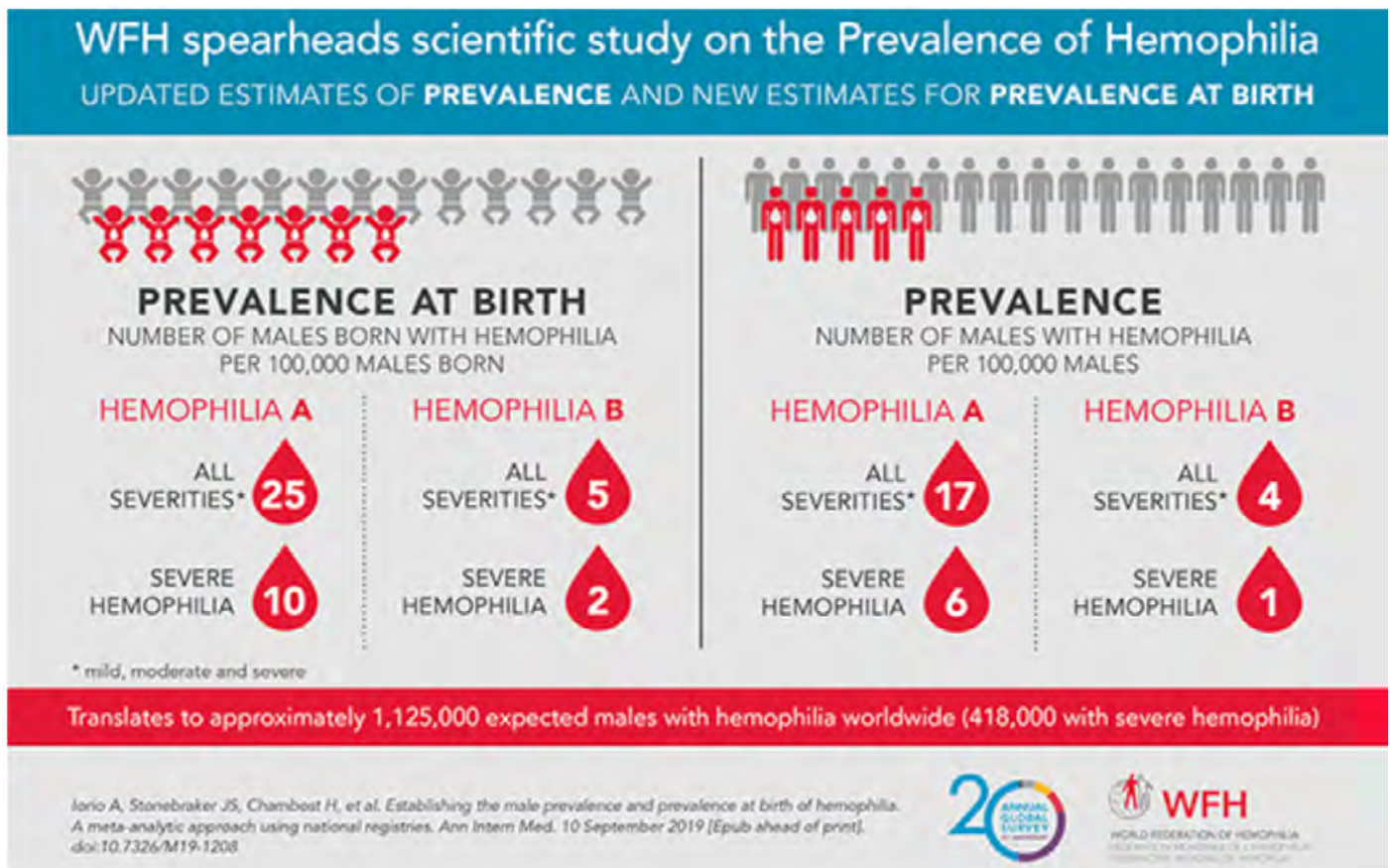
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Ferdinand Mbonihankuye is a Burundian investigative journalist specialising in data, science, the environment and science journalism. He is a member of the Network of Science Journalists in French-speaking Africa (RJSAF), co-founder of Ibihe.org, and correspondent for SciDev.Net. With seven years' experience in the media, he specialises in investigative journalism, with a particular focus on environmental crime, water and environmental health. He is the recipient of renowned fellowships, including iwmf.org (Global Health Reporting Initiative), CPHIA 2023, infonile.org, Rainforest Journalism

[Fund/pulitzercenter.org](https://fund/pulitzercenter.org), Aga Khan University, and the Climate Finance Reporting Academy. He is active as a reporter for several media outlets, including [TheNiles.org](https://theniles.org), [Dialogue.earth](https://dialogue.earth), mongabay.com, trtafrika.com, [Global Voice](https://globalvoice.org), and africanliberty.org, and has also been a blogger on platforms such as ijambo.org, [mondoblog](https://mondoblog.org), yaga-burundi.com, [cde greatlakes.org](https://cdegreatlakes.org), and insideburundi.org. His passion for science journalism enabled him to secure financial support from the African Development Bank to take part in the African Climate Summit 2023.

Hemophilia: a little-known but very real and costly disease



Infographic showing global estimates of the prevalence of hemophilia A and B at birth and among the male population, highlighting the extent of this hereditary disease according to a scientific study coordinated by the World Federation of Hemophilia (Photo credit: WFH)

In East Africa, including Burundi, hemophilia kills not only as a disease itself, but also through the lack of diagnosis, the shortage of medication, and dependence on international donations. As climate disruptions weaken the cold chain essential for treatments, this rare condition becomes a stark reminder of regional health vulnerabilities. For families, every unexpected bleeding episode can turn into a tragedy, exposing children and adults to serious complications, lasting disabilities, and profound economic hardship.

“Hemophilia is an inherited blood clotting disorder caused by a deficiency in clotting factors,” explains Dr. Stella Rwezaula, consultant hematologist at Muhimbili National Hospital and lecturer at Muhimbili University of Health Sciences and Allied Sciences (MUHAS) in Dar es Salaam, Tanzania. She is also the President of the Tanzania Hemophilia Society. This statement was made during a

telephone interview about the medical and structural challenges posed by this rare disease in the region.

“The main symptom is prolonged bleeding that doesn’t stop without treatment,” she continues. For families, it often begins with a period of confusion: a child who bleeds longer than expected, a teenager whose knees swell painfully after minor bumps, or a patient whose bleeding recurs several times after a tooth extraction.

A disease that is neither African nor European

In East Africa, hemophilia is sometimes described as a “foreign” or imported disease. This idea is medically unfounded. *“Hemophilia affects all countries in the same way. It doesn’t choose Africa or Europe,”* explains Dr. Rwezaula.

According to the World Federation of Hemophilia (WFH), approximately one in 10,000 people worldwide lives with the disease. Hemophilia A, the most common form, affects one in 5,000 male births, while form B affects approximately one in 25,000 to 30,000. These averages allow us to estimate the theoretical number of cases in the region, which is often much higher than the recorded figures.

In Tanzania, with over 800,000 births per year according to the [World Bank](#), approximately 160 new cases of hemophilia could appear annually. Kenya, the most populous country in the region with over 1.3 million births per year, would have more than 250 theoretical new cases.

In neighboring countries, the orders of magnitude are more modest but significant: Rwanda (approximately 350,000 births/year) and Burundi (nearly 300,000) could each register 60 to 70 hemophiliac boys each year — even if most are never diagnosed.

In reality, national registries only list a few dozen. The [2023 World Report](#) from the World Federation of Hemophilia mentions fewer than 200 identified patients in Tanzania, fewer than 100 in Kenya, and barely a few confirmed cases in Rwanda or Burundi.

This discrepancy is alarming to doctors. *“Hemophilia is very much present here, but invisible in the statistics,”* laments Dr. Rwezaula. A lack of specialized laboratories, the cost of screening, and a lack of awareness about the disease all contribute to this invisibility. As long as diagnosis remains a luxury, hemophilia will continue to be perceived as a distant disease—when in fact it is very much present here.

A genetic disease, but not exclusively male.

Hemophilia is often described as a “male disease.” The explanation is genetic. *“Hemophilia primarily affects men because it is carried on the X chromosome,”* explains Dr. Rwezaula. Men only have one X chromosome: if it carries the mutated gene, the disease will manifest in its full form, due to the lack of a spare X chromosome.

Women typically have one affected X chromosome and one functional X chromosome. In most cases, they are asymptomatic carriers. However, they are not completely protected. *“In rare cases, women*

can also have hemophilia if both X chromosomes are mutated, or if the second X chromosome is inactive. This phenomenon is known as lyonization,” explains Dr. Rwezaula.



Dr. Stella Rwezaula, consulting hematologist at Muhimbili Hospital, Tanzania

Considering the disease as “impossible” in women has consequences: patients with hemorrhagic symptoms may be ignored, misdiagnosed, or deprived of specialized care.

A disease that destroys in silence

The medical mechanism of hemophilia is simple to explain, but it is devastating in practice. When the body lacks clotting factors, bleeding does not normally stop after an injury, surgery, or even a routine procedure.

“Circumcision can endanger the life of a boy with hemophilia,” says Dr. Rwezaula. She explains that in mild cases, bleeding occurs mainly after trauma or surgery. In severe cases, it can appear spontaneously. *“In cases of severe hemophilia, patients can bleed even without injury,”* she adds.

Even more concerning, the bleeding isn’t always visible. *“They might wake up with bleeding in their joints or muscles,”* she explains. Internal bleeding can be fatal, particularly intracranial hemorrhage. The gastrointestinal tract and urinary system can

also be affected, in which case patients vomit blood or have blood in their stool or urine.

Hemophilia is therefore not just a childhood disease. It is a chronic disease that can progressively damage the body, especially when repeated joint bleeding causes chronic pain and deformities.

In Burundi, a lack of awareness about hemophilia has already claimed victims. Gilbert Niyonkuru, president of the Burundian Society of People with Hemophilia (Sobuh), speaks about it with restrained gravity. His commitment stems from a personal tragedy.

“I went to almost every hospital in Burundi. No healthcare professional could tell me what illness my child was suffering from,” he says.

For months, his son presented with unexplained symptoms. Consultations multiplied, without clear answers. Diagnoses were inconclusive, treatments changed, but the child’s condition did not improve. Faced with this impasse, the father decided to cross the border.

He went to King Faisal Hospital in Kigali. There, doctors found a large hematoma on his back, the result of a lumbar puncture performed before cranial surgery. A clotting disorder was suspected.

“The doctors sent me to the University Hospital Center of Kigali (CHUK) to confirm or refute the hypothesis,” he emphasizes.

At this center, the diagnosis was made: hemophilia type A. Treatment was then initiated. Thanks to the support of the Rwandan Hemophilia Association and the University Hospital of Kinshasa (CHUK), the child began receiving factor VIII, essential for blood clotting. However, the relief remained fragile.

“I had to transport my son to Kigali every time he had a shock. Whether it was morning or evening, rain or shine, I had to cross the border,” Niyonkuru explains.

After lengthy negotiations, Rwandan doctors exceptionally agreed to entrust him with a few vials of clotting factors, which were then stored at the Muyinga hospital. A precarious solution, dependent on goodwill and available supplies.

Then came the Covid-19 pandemic. Borders closed. Travel became impossible. *“The doctors wouldn’t let me return to Burundi with medication,”* he says. His son died at the age of four.



Gilbert Niyonkuru, president of SOBHU (Burundian Society for People with Hemophilia). Photo credit: Ferdinand Mbonihankuye.

In 2022, devastated by this loss, Gilbert Niyonkuru founded the Burundian Society of People Affected by Hemophilia, Sobuh, officially approved by the Burundian Ministry in charge of the Interior.

Between medical ignorance and mystical accusations

“Without treatment, a patient can bleed to death,” warns Dr. Rwezaula. When a child continues to bleed after a minor injury, many families in Burundi and East Africa don’t consider a rare genetic disease. Instead, they suspect witchcraft and a curse.

Several hundred kilometers from Muyinga, in the Jabe neighborhood of Bujumbura, Blandine Kabanegeye doesn’t contradict this observation. She relives the early years of her son’s illness. Sitting in her living room, she recounts a gradual descent into incomprehension.

“My son was less than six years old when I noticed the first signs. He had bruises all over his body. His knees were swelling,” she recalls.

At the hospital, doctors initially suspected blood infections. Medication was prescribed. Dietary advice was given. Nothing changed. Three months later, the symptoms returned, more severe than before.



Blandine Kabanegye, mother of a child with hemophilia: “The family devotes almost all of its resources to medical care.” Credit: Ferdinand Mbonihankuye.

A doctor then suggested surgery to remove the fluid that had accumulated in the knee. But in the operating room, the surgeons discovered not fluid, but blood. The wound healed poorly. The bleeding continued. The child’s condition deteriorated. *“The doctor didn’t really know what was wrong with my son,”* she said.

Around her, the explanations take a different turn. Some speak of a curse. Others talk of ancestral wrath. Her community accuses her.

“We tried everything. Incantations, herbal baths, amulets. Nothing worked,” she confides.

Years passed. The family’s savings dwindled. Finally, the mother found more specialized medical care at the Central Polyclinic of Bujumbura, and then at King Khaled Hospital. Extensive examinations were carried out.

The diagnosis was made: hemophilia. But the delay in treatment left lasting effects. Due to repeated bleeding in his joints, the child struggles to walk. He had to repeat his third year of primary school, unable to attend school regularly.

As with the Niyonkuru family, Blandine Kabanegye’s story reveals the flaws in a system where hemophilia remains poorly understood, often confused with other pathologies, and sometimes interpreted through the prism of beliefs.

“There are no specialized laboratories in the country. Basic coagulation tests are insufficient to confirm hemophilia and determine its type. There

are no specialist doctors or laboratories equipped to differentiate between hemophilia A and B, nor are there any medications. The risks are numerous: loss of patients or permanent joint disability. Of the more than 2,000 patients that the association should be registering based on prevalence at [birth](#), we only have 19, of which 18 are alive and one has died,” explains Mr. Niyonkuru.

A financial black hole for families

For Burundian families, hemophilia is not just a medical ordeal. It is an economic catastrophe. A single injection of clotting factor can cost several hundred dollars, in a country where the average monthly income is projected to be around forty dollars in 2025, according to the [World Bank](#).

Ms. Kabanegye describes an exhausting daily life, punctuated by trips to the hospital up to three times a week. *“We were only eating one meal a day,”* she says sadly. Her son, forced to repeat his third year of school, can’t even go home alone during a seizure. *“His classmates carry him on their backs all the way home when he has a seizure,”* she explains.

Beyond the medical expenses that devour family savings, the disease deprives parents of any possibility of working. Maintaining income-generating activities is impossible when the child requires constant care. *“The Burundian health system is powerless against hemophilia. There is no national registry, no follow-up care is provided. Patients have to obtain treatments themselves, often from abroad. The state focuses its efforts on malaria, HIV, tuberculosis, and malnutrition,”* laments Gilbert Niyonkuru.

Sobuh has 23 members, mostly men, many of whom are children, in a country where the prevalence of hemophilia A is estimated at 1 case per 5,000. Clotting factor concentrates, donated by the World Federation of Hemophilia, are free but scarce. Additional costs—transportation, customs clearance, consultations, injections—combined with widespread poverty, hinder access to medication in Burundi. Without trained doctors or adequate equipment, late diagnoses worsen joint and bleeding complications, and perpetuate social exclusion and early infant mortality, Mr. Niyonkuru stated.

Population size matters

While the disease affects approximately one in 10,000 people in Burundi, every country in East Africa is expected to have hundreds of patients, whether diagnosed or not. *“The number of patients depends on the size of the country’s population,”* Dr. Rwezaula points out.

The diagnosis requires laboratory capacity concentrated in the capital and referral centers. *“It requires coagulation tests, and many rural hospitals don’t have them,”* she points out. *“Most of the trained specialists are concentrated in national and tertiary hospitals,”* she adds.

This results in a pyramid-shaped system: a well-equipped apex in the capital, and a very broad base of under-resourced facilities in rural districts. For a disease whose early diagnosis can prevent disability and death, this configuration is catastrophic. Burundi is rarely mentioned in global discussions on hemophilia, but it is likely not exempt from international assessments.

The treatment exists, but it remains out of reach.

Modern hemophilia care is among the great achievements of medicine. In high-income countries, patients live long lives with few complications, thanks to prophylaxis and regular access to factor concentrates.

In East Africa, these same medications exist, but they are rarely accessible. *“Most African countries don’t buy factor concentrates because they are too expensive,”* says Dr. Rwezaula. *“The appropriate treatment is to replace the missing clotting factor. When factors are unavailable, we use blood products such as fresh frozen plasma. Cryoprecipitate can also be used, particularly for hemophilia A,”* she explains. However, this is a compromise. Blood products contain clotting factors in lower concentrations, which necessitates large volumes and repeated infusions. *“They also carry risks of transfusion-transmitted infections, including hepatitis and HIV,”* she adds.

Dr. Rwezaula also mentions the arrival of long-acting treatments and factor-free therapies such as Emicizumab, injected subcutaneously and capable of preventing bleeding for several weeks, and gene

therapy. However, she acknowledges that access to these remains limited in Africa.

Atmospheric changes affect storage

In debates about climate change and health, hemophilia is rarely mentioned. Yet it should be. The disease remains genetic; climate is not the cause. However, rising temperatures and energy instability threaten the systems that keep patients alive. This genetic disease requires treatment dependent on a stable cold chain and continuous access to electricity—two resources that the climate crisis is making increasingly precarious.

The central issue is storage. *“Factor concentrates must be kept between 2 and 8 degrees Celsius,”* explains Dr. Rwezaula. This requires functioning refrigerators, a reliable power supply, and an unbroken cold chain from international transport to the patient. In many parts of East Africa, these conditions cannot be guaranteed.

Heat waves, rising temperatures, and unreliable electricity supplies are further complicating storage. Some rural clinics lack refrigeration, hospitals experience frequent power outages, and patients live far from the centers that store the concentrates. *“The distance between hospitals is a major risk, as hemorrhages are emergencies,”* emphasizes Dr. Rwezaula.

To address this, his team trains families in traditional cooling techniques using water and local materials, inspired by methods applied to other heat-sensitive biological products. However, the very fact that families have to improvise reveals the vulnerability of care to climate shocks and energy insecurity.

Organizations such as the [World Federation of Hemophilia](#) and [Health Care Without Harm](#) are calling for these challenges to be integrated into climate policies. A study published in *The Lancet Planetary Health* highlights that global warming is already threatening blood and blood product supply chains, making the development of climate-resilient healthcare systems urgent.

Up to \$100,000 per patient per year

“Hemophilia is rare, but it is extremely expensive,” Dr. Rwezaula continued. Global estimates put the cost of treating a patient at around

\$100,000 per year in some settings. While this cost varies by country and care model, the message is clear: treatment remains out of reach for the vast majority of families.

She observes that *“families cannot afford to pay. Governments also struggle to cope.”* This is why donor support has become essential for hemophilia treatment in Africa. But this model is unsustainable. *“We cannot rely on donors forever,”* she insists.

Dr. Rwezaula’s warning takes on a more urgent tone when global politics are involved. International aid to health systems is evolving. In some contexts, funding is decreasing, programs are being restructured, and priorities are changing.

For countries that rely on donations, the consequences are dire. *“If donations decrease, the consequences for patients could be catastrophic,”* she says. When factor concentrates stop arriving, patients face not just discomfort, but death. Hospitals revert to plasma transfusions, with a higher risk of infection. Families sell their possessions, children drop out of school, and joint hemorrhages lead to lasting disabilities.

At Muhimbili National Hospital, Dr. Rwezaula describes the clinical burden associated with hemophilia. *“The appropriate treatment involves replacing the missing clotting factor,”* she reiterates. But, here again, *“most African countries don’t buy factor concentrates because they are too expensive.”* She adds that *“many countries depend on donations,”* particularly those from the World Federation of Hemophilia.

Gilbert Niyonkuru summarizes the consequences for families. *“Hemophilia creates great psychological distress and financially exhausts families, because*

patients must receive two to three injections per week without receiving free care.” He calls for political action. *“The government should provide the necessary medications to treat patients who remain in critical condition due to a lack of treatment.”*

Sobuh, for its part, operates primarily through contributions from poor families. It organizes awareness sessions in schools, health centers, and through community radio stations. However, reaching the entire country remains impossible with such limited resources.

Group purchases

Dr. Rwezaula’s recommendations are unequivocal. Governments must first begin purchasing factor concentrates, even if only gradually. *“Regional group purchasing in East Africa could reduce costs,”* she states. Decentralizing diagnosis is also essential. Coagulation tests should not remain limited to national hospitals, and healthcare professionals in all districts must be trained to suspect and diagnose hemophilia. Finally, stigma must be combated through public education. *“Media awareness campaigns, as well as training for patients and the wider community, are crucial to reducing stigma,”* she believes.

In Burundi – as has also been established in Tanzania, Kenya and Uganda – the priorities are clear: develop a national strategy on rare diseases, establish free care for hemophilia patients, create well-equipped reference centers, establish a national patient registry and conduct large-scale awareness campaigns.

“The government should provide the necessary medicines to care for patients who remain in critical condition due to a lack of treatment,” calls Gilbert Niyonkuru. ■



Linord MOUDOU

in VIP News Network | United States



Linord Moudou is a seasoned multimedia journalist. She is the Founder and Managing Editor of the VIP News Network online media start-up and hosts the VIP with Linord webcast.

Before that, Moudou worked with Voice of America in Washington, D.C. for nearly 20 years as a bilingual (French-English) news anchor, health editor, writer, reporter, producer, project manager, and trainer for journalists in Africa.

Moudou has conducted interviews with leaders, newsmakers, and medical experts, including Microsoft Co-Founder Bill Gates, Dr. Benett Omalu,

who discovered CTE in American Football, Gro Harlem Brundtland, Former Norwegian Prime Minister, and WHO Director-General Tedros Adhanom Ghebreyesus. She has moderated numerous high-level events, such as the Social Good Summit in New York and the Global Fund Replenishment Summit in Lyon, France.

Linord Moudou studied Journalism and Media Productions at George Mason University. She holds an MBA in Organizational Management and an Honorary Doctorate in Broadcasting Leadership.

Innovating Rare Disease Trials: Insights from Dr. Harsha Rajasimha



Video [here](#)

Rare but Not Invisible: Ellis–Van Creveld and the Challenges in Rare Disease Care

When a rare genetic diagnosis lands in a family’s lap, biology is only part of the story. In this feature, one family’s experience with Ellis–Van Creveld syndrome shows how genes, health systems, and geography together shape survival.



Erica Shoemate at home, reflecting on her family’s journey with Ellis–Van Creveld syndrome.

“Terminate, heal, and try again, in that order.”

That was the recommendation.

At just over 20 weeks pregnant, Erica Shoemate sat in an exam room as doctors told her and her husband their daughter had a lethal genetic condition. The message was blunt: end the pregnancy, recover, move forward.

“There was no behavioral health support in that moment,” Shoemate recalls. “You’re being told your child is not compatible with life, and you’re expected to process that clinically.”

The suspected diagnosis was Ellis–Van Creveld syndrome (EVC), a rare inherited disorder that affects skeletal development and often the heart. In most parts of the world, EVC is estimated to occur in roughly 1 in 60,000 to 200,000 births, according to the NIH’s Genetic and Rare Diseases Information Center. Orphanet, the European rare disease database, notes that only a few hundred cases have been formally documented in the medical literature, many clustered in specific communities due to founder effects.

Shoemate and her husband chose not to terminate the pregnancy.

“You don’t know what you’re going to do until you’re in that situation,” she says. “It’s not black or white. It’s very gray.”

Their daughter survived.

But survival depended on reaching the right specialists.

When Survival Depends on Specialization

Ellis–Van Creveld syndrome is inherited in an autosomal recessive pattern: both parents must carry the altered gene, and the condition appears when a child inherits two copies. Children with EVC typically present with short stature, extra digits, dental abnormalities and nail differences. The most serious complications often involve the heart.

“The most challenging aspect of EVC is the heart condition,” says Dr. Laura Poskitt, medical director of the Clinic for Special Children in Pennsylvania.

She estimates that between 60 to 80 percent of children with EVC are born with significant congenital heart disease, often atrioventricular canal defects in which the walls separating the heart chambers are incomplete and the valves leak. Some defects are manageable. Others require complex surgical repair early in life.

Historically, surgeons moved quickly. Babies with large cardiac defects underwent openheart surgery within weeks of birth. For children with EVC, whose rib cages and lungs are smaller than average, that approach often led to devastating complications.

Over time, clinicians caring for Amish children with EVC altered the timeline. Instead of rushing to surgery, they stabilized infants at home with tracheostomies, ventilators and feeding tubes, allowing the chest and lungs to grow. Heart surgery was delayed until closer to nine to twelve months of age. Survival improved.

“Someone with EVC who has a normal heart has a full life expectancy,” Poskitt says. “Cognitively, these children are typically normal.”

That clarity rests on experience.

In Pennsylvania, EVC is more common among the Old Order Amish because of a genetic founder effect.



Amish communities in Pennsylvania have been the focus of research on Ellis–Van Creveld syndrome, which is linked to a genetic founder effect.

Roughly one in eight Amish individuals carries an EVC mutation, a rate far higher than in the general population. Poskitt explains that the mutation in this community can be traced back to a single immigrant, Samuel King, who arrived in Pennsylvania in the eighteenth century.

That concentration allowed earlier recognition and refined care protocols. Outside that context, familiarity with the condition is limited.

When knowledge is limited, conversations about prognosis can become overly rigid.

When Data Does Not Reflect the Patient

“We’re not Amish. We’re not Dutch,” Shoemate says. “So, what does that mean for us?”

Most published descriptions of EVC come from Amish cohorts. On paper, her daughter’s suspected condition resembled a different, uniformly lethal shortrib polydactyly syndrome.

Shoemate’s instincts pushed her to question assumptions.

“You can either skew the data or ignore the data,” she says, “and you get two completely different outcomes.”

Her daughter’s care changed only after referral to a national skeletal dysplasia center operating with true interdisciplinary coordination. There, specialists recognized the nuances between lethal and non-lethal skeletal dysplasia, understood the bell-shaped chest and long, slender lungs that characterize many EVC patients, and drew on protocols developed with Amish children in nearby Pennsylvania.

“It wasn’t just multidisciplinary,” she explains. “It was interdisciplinary. That changed everything.”

Instead of separate specialists working in parallel, her daughter’s cardiologist, pulmonologist, orthopedist, and other clinicians met around a single plan. Care became coordinated rather than fragmented. The difference showed up not in theory, but in oxygen saturations, hospitalizations avoided and a little girl who now attends school, jokes with her friends and prepares for orthopedic surgery while living what her mother calls an “ordinaryextraordinary” life.

The broader rare disease landscape reflects similar gaps.

“Individually rare, collectively common,” says Dr. Harsha Rajasimha, founder and executive chairperson of the IndoUS Organization for Rare Diseases.

Approximately 11,000 rare diseases are recognized in the medical literature. By U.S. definition, each affects fewer than 200,000 Americans, yet collectively they impact more than 30 million people in the United States. Globally, rare diseases are estimated to affect between 300 and 400 million people.



Abstract DNA illustration representing the genetic basis of many rare diseases.

“Only five percent of these rare diseases have an approved treatment as of now,” Rajasimha says.

The remaining 95 percent have no approved therapies.

Diagnosis itself can take years.

“On average, it takes five to seven years to get a proper diagnosis, even in a developed country like the United States,” he explains.

Representation in research compounds the challenge.

“Historically, 80 percent of clinical trial participants have been Caucasian male adults with affluent socioeconomic backgrounds,” Rajasimha says.

Federal analyses and independent reviews have repeatedly documented underrepresentation of women and racial and ethnic minorities in clinical trials, particularly in earlyphase research.

The economic burden is equally sobering. A 2019 analysis by the EveryLife Foundation estimated that rare diseases collectively cost the United States nearly one trillion dollars annually, including direct medical expenses and indirect costs such as lost productivity and unpaid caregiving.

The African Gap: First, Recognition

While Ellis–Van Creveld syndrome is frequently associated with Amish founder populations in Pennsylvania, a molecularly confirmed case has been documented in Tanzania in peerreviewed medical literature.

Yet documentation does not guarantee recognition.

Prof. Leon Mutesa directs the Center for Human Genetics in Rwanda.

“It’s also present in African populations,” he says of Ellis–VanCreveldtype conditions. “Sometimes misdiagnosed or underdiagnosed because of a lack of access to diagnostics. Most of the professionals don’t recognize these disorders.”

In the United States, families may struggle to access specialized centers. In parts of Africa, the challenge begins earlier.

“We only have one genetic center in the country,” says Dr. Aime Lumaka, professor of genetics and pediatrics at the University of Kinshasa in the Democratic Republic of Congo.

“We only have four clinical geneticists correctly trained. We have zero genetic counselors for the entire country of about 110 million inhabitants.”

“The burden of not knowing is really destroying families,” he adds. And conditions may sometimes be attributed to spiritual causes.



Dr. Aime Lumaka consults with a mother and child at the genetics clinic in Kinshasa. Photo courtesy Dr. Lumaka.

This is a reality across many African settings where diagnostic infrastructure remains limited. A child born with shortened limbs and extra digits in a rural clinic may never receive molecular confirmation of EVC or a related skeletal dysplasia. Without routine echocardiographic screening, congenital heart defects may remain undetected until they become life-threatening.

In Pennsylvania, surviving EVC may depend on the timing of surgery. In parts of Africa, survival may depend first on access to diagnosis.

The difference is structural.

Trauma Beyond the Operating Room

Shoemate’s daughter now attends school. She continues to see a cardiologist and prepares for orthopedic surgery to correct knockkneed alignment in one leg. Her life is active.

But trauma lingers.

“During winter months, if I hear a cough, my body reacts,” Shoemate says. “It’s very somatic.”

She speaks openly about postpartum anxiety and PTSD.

“Sometimes you need prayer, therapy and prescription,” she says.

Rare disease care often measures success by surgical survival. Families measure it differently.

“As a rare dad,” Rajasimha says, reflecting on losing a child to Edwards syndrome, “I realized that science and technology are critical, but patient advocacy is very powerful.”

A Crisis of Systems

Ellis–Van Creveld syndrome is rare.

The vulnerabilities it reveals are not.

Across continents, families living with rare diseases encounter delayed diagnoses, uneven access to expertise and limited representation in research.

“Rare does not mean unimportant,” Lumaka says.

Genetics is not the crisis. Systems are.

To describe EVC as rare is accurate. To allow rarity to become invisibility is a failure of structure, not science.

For families living with rare diseases worldwide, visibility is not symbolic. It is survival. ■



Rakiya Abubakar MUHAMMAD

in RM Times | Nigeria



Rakiya Abubakar Muhammad is an independent journalist whose work has appeared in multiple outlets, including Daily Trust, RM Times, The Guardian, Liberalist Magazine, and Factcheck News.

She served as Bureau Chief at the Daily Trust Newspapers until August 2020.

Rakiya claimed the 2024 African Regional Antimicrobial Resistance

Champion Journalist Award (Print).

In 2023, she was a finalist for both the Climate Now Award and the Nigeria Media Merit Award.

In 2022, she won the Pamela Howard Forum on Global Crisis Story Contest and the Nestle Nigeria Media Awards, and has clinched the Daily Trust Best Feature Stories Awards on multiple occasions.

Enhancing Journalistic Coverage of Rare Diseases



Grounding hopefulness in evidence, grounding hype in data, and being fundamentally honest to the reading public plea is a plea from the rare disease community through science.

This was a key takeaway from the presentation by renowned physician and innovator, Professor James Levine at the 2025 National Press Foundation(NPF) Rare Diseases Reporting Fellowship.

Professor Levine, President of Fondation Ipsen, an international science foundation focused on rare disease innovation, addressed 62 journalist fellows from 29 countries across five continents.

“It’s too easy to say that there are new gene therapies, hope is on the horizon,” he pointed out.

“But can you imagine what that’s like for a family who’s been looking after a child, whether that’s in a wealthy country or a less wealthy country, and they read that there’s sort of hope where in fact hope with respect to an immediate cure does not exist.”

Accurate, well contextualised journalism, he said, is essential to scientific advancement: it is the “oxygen of science” that can breathe hope in the futures of people living

Uncommon Disorders: Communicating the Scale

Levine described Rare Diseases as the world’s largest invisible medical community, affecting

350 million lives across 7000 conditions -yet 95% remain untreated.

The word “rare” is almost a paradox in that it suggests these conditions must be uncommon, the healthcare innovator observed.

“But when you combine all these rare diseases together, you’re talking about an agency of more than 300 million people living with rare diseases, and you’re talking about the size of the United States,” he highlighted.

“And because of this misconception, you can understand why this minimizes the urgency in the political attention.”

He also pointed out: New therapies are on the way and they’re coming fast but because sometimes many of the rare diseases by definition affect only a few people, some people argue that there’s not enough of an incentive to develop therapies for them.

Levine counters: “If you look at the data, approximately \$50 billion of business was done with mergers and acquisitions in the rare disease space in 2023.”

“There’s a lot of activity in this space and there are reasons for this. Some of it is related to the ability to deliver therapies, but also part of it is related to the spillover economics.”

He added: “So you can develop a therapy, for example, for an ultra-rare obesity condition, but the spillover can be into more commonplace disorders.”

“And so, there are significant opportunities. So because their disease is rare, that doesn’t mean people aren’t interested in finding cures. It’s just not true.”

From Victims to Visionaries: Breaking Pity Narrative

Levine called for a paradigm shift: change the story from one of victimhood to one of resilience and agency:: Break the pity narrative, from victims to visionaries.

“The community is not looking to be saved. They’re looking to be supported, the people are looking for diagnoses and people are looking for cures, but they’re not looking to be saved,” he said.

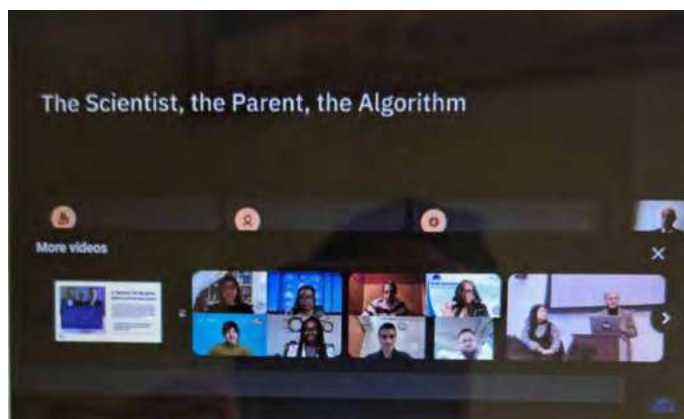
“There’s a very different communication between agency and tragedy. And the way the language is written in whichever language you are writing in is extremely important with respect to how members of the public who are reading your articles perceive the shapes of possibility.”

He spotlighted resilience through inspiring stories—like that of a Fondation Ipsen author living with a severe neurological disorder yet thriving as a journalist.

“She’s met three popes. She’s written 18 books, which are romance novels and has been in a wheelchair her whole life,” he stated.

“Can you imagine what it takes to do that? People with rare diseases are not looking to be pitied. They’re looking to be elevated. They show agency over adversity. They have so much to teach us.”

After more than three decades in medicine, Levine has often witnessed families eager to share their viewpoints.



“The people living with rare diseases are more than willing to share not only their own data or even the data of their children, but even genetic data because

the solutions come from sharing this data,” he observed.

“As journalists, you’re going to find a remarkable willingness to share lifetime stories and personal stories as it ultimately is going to raise awareness of the actual condition and accelerate cure.”

Language of Hope: Focusing on Progress

He encouraged journalists to honour each step forward, fostering an optimistic spirit that shines a light on advancement.

“Focus on progress—not perfection,” he said, stressing the ‘grammar of hope’.

He highlighted a shift in language and sees journalists as guides, helping society embrace this fresh outlook.

“Medicine is becoming creative engineering; the lexicon shifts from cure to construction, patient to partner,” he stated.

“Journalists can teach society this new grammar. Every story you tell expands the public vocabulary of progress.”

To guide storytelling, Levine introduced his DACMAR framework—six vital questions:

Disruption: What’s changing?

Adoption: Who’s using it?

Collaboration: Who’s partnering?

Management: How is it governed?

Adaptability: Can it scale?

Resources: What’s the cost?

He shared invaluable tips for interviews:

For scientists, ask: “What keeps you awake at night?”—uncovering doubts, fears, triumphs, and the emotional core behind discoveries. “Ask about failures, not just successes; explore the personal cost of persistence; find the moment of uncertainty before breakthrough; connect the science to the scientist’s story.”

For patients, ask: “What do you want the world to know?”—empowering them to tell their stories on their terms. Move from extraction to collaboration, let patients co-author their story, he added.

The researcher emphasised the journalist’s role in weaving together three distinct voices: The Scientist, representing rigorous research and clinical expertise;

the Parent, offering lived experience and advocacy; and the Algorithm, providing data analysis and pattern recognition.

“Stay inspired, stay honest,” Levine advised.

“Exaggeration erodes credibility. Your reporting must be rooted in evidence—realistic, transparent, and hopeful.” ■

Gene Editing: Illuminating Landmark Leap in Rare Disease Treatment



In 2025, doctors corrected a life-threatening genetic disorder in an infant using gene therapy, marking a medical breakthrough.

From birth, baby KJ Muldoon faced CPS1 deficiency, a rare liver disorder caused by a lack of the enzyme carbamoyl phosphate synthetase 1 (CPS1). This enzyme is essential for removing ammonia, a waste product from protein breakdown. Without CPS1, ammonia levels in the body can become dangerous, requiring immediate intervention.

The scientists, Rebecca Ahrens-Nicklas and Kiran Musunuru, with the Innovative Genomics Institute, pioneered the CRISPR gene-editing therapy—a method for making precise changes to DNA—that enabled targeted correction of KJ’s DNA.

The landmark achievement was featured at the 2025 Rare Disease Reporting Fellowship, where experts detailed KJ’s case and discussed the broader

significance of the therapeutic advance.

Addressing journalists from 29 countries, the doctors presented KJ’s therapy as a milestone offering new hope for the treatment of rare diseases.

CPS1 Deficiency: Unravelling a Rare Genetic Puzzle

Kiran Musunuru, Professor of Translational Medicine at the University of Pennsylvania, explained that KJ’s case was especially dire because his liver lacked an essential enzyme, which is necessary for protein processing.

He added that inheriting two faulty gene copies, as in KJ’s diagnoses, results in no enzyme activity, leading to ammonia buildup and severe disease.

“You get one from mom, you get one from dad. If you get a broken one from mom, but a healthy one from dad, you’re going to make half the protein,”

Musunuru stated.

If you make 50% of the enzyme, that is enough to clear ammonia and remain healthy, the gene editor explained.

The only way for someone like AJ to get this disease is by inheriting two defective copies—one from each parent, he pointed.

“Now you have 0% working protein, and that’s when you get into trouble because then you can’t do anything with the ammonia,” he added.

“It builds up in the body and is very toxic and causes a very bad disease.”

CRISPR: From Ancient Defender to Modern Marvel

Musunuru highlighted that CRISPR, a natural system bacteria use to cut viral DNA, now allows scientists to precisely edit human genes.

He described how bacteria evolved CRISPR to fight viral threats, drawing a parallel to how humans combat pathogens.

“Just like we have to fight off invaders—microorganisms, pathogens that are trying to invade our body and grow in our body and reproduce and so forth—it turns out the microscopic bacteria deal with the same sort of threats just on a different level,” he highlighted.

“In their world, viruses are the big threat. And so, they have evolved ways to help them. If they see a virus for the first time, they can’t do much about it. They either survive it or not.”

Musunuru expounded: “If they survive it, then they take little snippets of DNA sequence from that virus and incorporate it into their own genome. And they have a system called CRISPR that allows them to use those little snippets and monitor for any repeat infection by that virus,”

“And if that same virus shows up and has a matching DNA sequence to the snippet that was stored away from the past encounter, the bacterium can use CRISPR to actually search out and destroy that invader. And so that’s how CRISPR developed as an immune system.”

The gene editor elaborated on how scientists harness this system, comparing the human genome to a textbook. Think of each chromosome as a chapter, and each gene as a paragraph. If there’s a typo—like a misspelling—CRISPR can precisely correct that single letter, restoring proper function.

“And in K J’s case, he had one of these misspellings, actually two misspellings in both copies of a particular gene.”

He noted that advances in CRISPR technology have led to more precise DNA editing.

Base editing, or Version 2.0 of CRISPR, he explained, enables scientists to correct genetic mutations by changing a single DNA letter rather than cutting the entire strand.

“Out of the 6 billion letters in that entire textbook, we can find just the one letter that needs to be changed and corrected; we have that degree of precision,” he asserted.

“That’s what we ultimately used in KJ’s case.”

Engaging Parents: Transforming Hope into True Partnership

Rebecca Ahrens-Nicklas, Assistant Professor of Paediatrics at the Children’s Hospital of Philadelphia and the University of Pennsylvania, described how KJ’s parents actively participated by asking questions and becoming true research partners.

“It was pretty amazing. They asked a lot of good questions about what this meant for their child, but very quickly pivoted to start asking, what does it mean for the greater urea cycle disorder community and the greater rare disease community?”

“In that first conversation, they agreed that we should keep going with the scientific research, and from that moment on, we’ve been partners in all of this. So, I would give them regular science updates and research updates.”

The paediatrician emphasised the importance of clear communication and educating families about the disorder and therapies, describing this as especially rewarding.

She said, “The part of my job I love most is helping families understand their diagnosis and available therapies.”

Treatment Timeline: Mapping the Journey

Ahrens-Nicklas provided a concise overview of KJ’s treatment from the first dose to discharge, highlighting the remarkable advances in the care pathway.

“We provided the first dose right before he was seven months old, and within a week or two, we could increase the amount of protein that he took in his diet,” she detailed.

“Kids with this disease, one way you help treat them is by restricting how much protein they take. And very quickly, within about a week, we were able to get him up to the recommended daily allowance for protein for a baby his age. And this is really important because protein is necessary for babies to grow.”

The paediatric geneticist added that they had hints that something might be helping him very quickly, and that he subsequently received two doses over the next couple of months.

Baby KJ was discharged home at about 300 days old, while the medical experts continued to follow him closely.

“Throughout this process, we continue to see signs that he has benefited from the therapy,” Ahrens-Nicklas observed.

How Enduring is Gene Therapy? Unpacking the Longevity

The researchers said early signs are promising. “We anticipate durable genomic change, and time will tell in humans,” Ahrens-Nicklas remarked.

“But in animal models, we see that that correction is there, right? It changed the genome; it changed the patient’s DNA.”

Musunuru also expressed confidence that the genetic edit will remain stable and durable as the child grows.

“I would say pretty confident. It’s hard at this point to be a hundred per cent confident since we’re in very early days,” he stated.

“There are, at this point, a few hundred patients who have received this kind of gene editing therapy. But what we’re seeing in the adults who received the treatment 2, 3, 4 years ago now is that the editing appears to be entirely stable.”

The professor gave a little context: “Some of the adult patients who have received therapies have received them for heart disease. As a cardiologist, I’ve obviously been very interested in what’s been going on in that respect.”

Musunuru, who participated in helping to develop some of the therapies for heart disease, asserted: “What we see is that the effect, the therapeutic effect, for example, reduction of cholesterol levels or reduction of other proteins in the blood, they immediately go down after the patients get the treatment.”

He added that once they’re down, levels remain low for a very long time.

“As I said, 2, 3, 4 years now, and there’s no hint that they’re coming back up. It really does look like one-and-done. Once you have the effect at the DNA level, even though the liver is still growing or at least turning over some cells, those cells eventually die off,” he accentuated.

“We can’t yet say that it’ll be good for the lifetime, but I think there is a very good chance that it will be good for the lifetime. And we’re expecting that in adults. And we’re also expecting it in children and infants like KJ, but we’ll have to see to be sure.”

Opening Doors: Expanding Patient Access

Ahrens-Nicklas assured that efforts are underway to broaden access through clinical trials, so more patients can benefit from this historic therapy.

“You need formal trials where you essentially assess multiple versions of this drug; essentially the same drug that’s just tuned to an individual’s genetic variant,” she illuminated.

“Whether it’s patient A, patient B, or patient C, they might have different genetic misspellings, but the

safety profile of the drug is actually pretty similar across those versions of the drug.”

The physician disclosed that some of the diseases for which people have inquired about the drugs for their children or their patients require correcting organs other than the liver.

Currently, the delivery system works only for liver disorders. Hundreds of inquiries have shown a huge unmet need, she noted.

“And so, we cannot do what’s called this expanded access one-off compassionate use approach for each of those children because the resources we would put into that would not be generating the data we need to get a drug approved down the road,” said Ahrens-Nicklas.

“The formal clinical trial we hope to launch later this winter will allow us to both help children through that trial, but most importantly, generate the data we need to try to get approvals such that there can be a sustainable model for all of these children that are trying to reach out.”

She believes that, eventually, it may be cheaper than procedures such as liver transplants, and that funding and insurance reimbursement are critical for widespread adoption.

“But we need to be able to show the data that is a benefit, or that is a worthwhile use of health insurance resources, public health resources,” she pointed out.

“And so, we are helping families to the best of our ability by scaling through a clinical trial to provide more access to more families and then ultimately, hopefully get an approved therapy which will ultimately allow us to scale.”

Global Gaps: Bridging Disparities with One-Time Remedy

Ahrens-Nicklas observed that access to care for patients with rare diseases differs from country to country.

She cited KJ’s case, noting that without gene editing, he would have required a special metabolic formula, dialysis and later liver transplant.

“I will tell you that metabolic formula, which is life-changing, these medical foods for kids with metabolic diseases are not available in many, many, many, many places in the world,” she highlighted.

“To the point where those patients that have the resources to travel, let’s say to the US or maybe to Europe for their care, will take back suitcases of cans of formula so that their child could have the lifesaving food that they need where they are.”

The physician added, “Liver transplantation is similarly not available everywhere. And so, if we could develop a process by which one could deploy a one-and-done therapy.”

“It would keep a patient safe without having to undergo a liver transplant, and the ongoing management of liver transplant or needing medical foods across their lives that aren’t available in their home country, it would actually increase access in some ways.”

Ahrens-Nicklas revealed how they envision expanding access worldwide. “One of the key components of this is the technology itself. As I said before, the synthesis and the manufacturing of this type of drug product is actually something that’s simpler in some ways than some other cell and gene therapies,” she said.

“It uses techniques that have already been developed for a variety of other uses and deployed throughout the world, including developing nations.”

“So, the same sort of technology to make that soap bubble, to put the cargo inside that soap bubble, that’s appropriate, has been used to manufacture things such as vaccines. And those manufacturing sites are being set up around the world, outside of just the United States or over Europe.”

Ahrens-Nicklas argued that repurposing local manufacturing sites for drug development, or enabling patients to travel to a centre before returning home, could better meet the needs of patients with rare diseases worldwide.

“So, I worry about access all the time. I worry about access within the United States. I worried about access around the world,” she revealed.

“But I think by developing a cost-effective one-and-done therapy, you actually might increase access as compared to the current standard of care right now,

which I have to tell you is incredibly difficult when you start thinking about access for patients around the world.” ■



Nokukhanya MUSI - AIMIENOHO INHLASE

in Centre of Investigative Journalism | Eswatini



Nokukhanya Musi Aimienuho is an experienced Print, Radio & TV Media Practitioner based in Manzini, Kingdom of eSwatini. A widely published journalist, her articles have appeared in various online publications and websites across the African continent and the United States. She has been worked with the VOA Africa and Central Asia Division as well as TrT World. She has also written for a number

of international media outlets such as the Earth Island Journal, the Christian Science Monitor, BlueDotLiving to name a few. As a freelance journalist, Nokukhanya covers a diverse range of topics, including climate change, health, wildlife conservation, politics, and human rights issues. Her commitment to these important subjects reflects her passion for raising awareness and driving meaningful conversations.

Eswatini Rural Boy's Struggle: Bladder Exstrophy and Dignity



Born into a web of challenges, a young boy with bladder exstrophy fights for his chance at an everyday life in the impoverished rural community of Shewula in Eswatini. His birth marked not just the arrival of a new life but the beginning of a daunting battle against societal stigma, inadequate healthcare, and the relentless grip of poverty.

For 10-year-old Phiwoyinkosi Masimula, each day is a testament to resilience as he navigates the complexities of his condition while clinging to the hope that a brighter future awaits.

In the early morning light of a cool June day in 2017, the quiet hills of Shewula cradled the birth of this baby boy. Its arrival was met not with the usual cries of a newborn but with a startled, persistent wailing that echoed through the modest homestead.

The infant's tiny body bore an unmistakable anomaly: his bladder lay exposed on the lower abdomen, a rare condition known as bladder exstrophy. Bladder exstrophy is a rare congenital defect affecting approximately 1 in 30,000 to 50,000 births, where the anterior bladder wall fails to fuse, leaving the bladder exposed.

It often includes other complications and may require staged surgical repair.

The sight shocked his grandmother, Gogo Busisiwe Nhlabatsi, who had never witnessed such a birth in her village. A mix of fear and concern washed over her as she held the delicate infant, realising the challenges that lie ahead.

At 62 years old, Gogo walks with the aid of two canes and relies on Eswatini's elderly grant—recently increased from E 500 to E 600 per month (about US \$30).

“Although I am old and have health issues, I made a decision that I wouldn't stop caring for him. The grant I receive barely covers our daily needs, but we make do”, she says.

The boy's mother, Nokuthula Mnisi, left him with Gogo as a toddler and visits occasionally; day-to-day care falls to his grandmother.

Phiwoyinkosi wears disposable nappies (Pampers). In Eswatini, a pack costs roughly E180–E550* (US\$ 10–\$30). The used nappies are buried in a shallow

pit dug behind the homestead.

From the moment the condition was noticed, Gogo embarked on a relentless journey through the health system.

“We ran from clinic to clinic, and each time they said they’d figure it out, but the answers never came,” she recalls.

Financial exhaustion quickly depleted the modest elderly grant. A South African doctor offered tentative hope, but the family was told to wait in Eswatini.

In 2024, the Luke Commission (TLC) was approached; they promised specialist care abroad—a promise they are still waiting to see fulfilled.



The endless wild goose chase for help was frustrating and drained Gogo of the little she had left. To date, Phiwoyinkosi continues to wear diapers and has not yet undergone corrective surgery.

In 2025, at the age of eight, Phiwoyinkosi enrolled at Emajebeni Primary School in Shewula. The headmaster initially refused admission, citing concerns about the child’s age and need for diapers.

Gogo persisted, emphasising that “he may wear a diaper, but his mind is sharp.”

The school eventually relented; he completed Grade 1 and was promoted to Grade 2.

However, the denial of entry delayed his educational development and set him back compared to his peers.

In the face of these challenges, Gogo remains a

pillar of strength for Phiwoyinkosi. She makes sure he feels loved and cared for, despite the struggles they face. Together, they navigate the trials of poverty while nurturing the boy’s bright spirit and determination to learn. Gogo’s unwavering love and commitment serve as a beacon of hope in a world that often feels unforgiving.

The realities of families managing rare conditions are further illuminated by insights from Fikile Lukhele, a caseworker at the Deputy Prime Minister’s office.

Her experiences reveal critical gaps in the support system for these families. She explains that rare disease cases often come to their attention through community observation rather than direct contact.

“Unfortunately, we have never been contacted directly by families seeking assistance for rare conditions; we usually find such cases while working in the communities,” she shares.

This lack of direct communication highlights the barriers families face when seeking help, often leading to delays in care and support.

“We’ve discovered during field visits to communities that many people suffer from different ailments and conditions that significantly impact their health and social lives, as well as their financial situations. Counselling is something they need,” Fikile says.

She recounts a recent case involving a child with a rare condition, which she cannot discuss in detail due to ongoing support efforts. However, she describes a successful collaboration.

“After many challenges, we managed to get assistance from organisations in America for this child, who is now being cared for by a family,” she adds.

A significant challenge Fikile faces is the perception of her role.

“As junior officers, we encounter difficulties in getting people to listen to us. They often only respond to our superiors, which can hinder timely intervention,” she explains.

This systemic issue limits frontline workers’ effectiveness in advocating for vulnerable families.

Lukhele emphasises the need for the Ministry of Health to play a more proactive role, particularly during immunisation efforts.

“I often wonder what happens during immunisations at clinics. They should identify cases early and provide assistance, rather than us discovering these issues only when children are older,” she says.

Lukhele further emphasises the need for increased budget allocation for social work services.

“If the Ministry of Health could invest more in community sensitisation and social support, it could significantly impact families accessing care sooner,” she concludes.

While programs exist that could provide some support, significant gaps remain in specialist care and resources.

As Phiwoyinkosi continues his journey, there is little hope on the horizon. Echo VanderWal, Executive Director of The Luke Commission, admits:

“There are currently no known government-led programmes or clinical guidelines specific to rare diseases in Eswatini,” she explains. “However, we recognise the importance of raising awareness and improving diagnostic capabilities. Many rare conditions likely go undiagnosed, as families often navigate care through existing community structures.”

Echo notes that The Luke Commission is committed to leveraging every available resource for patients like Phiwoyinkosi.

“We have diagnosed and clinically managed several rare conditions, including Duchenne muscular dystrophy and malignant hyperthermia. Our approach emphasises accurate diagnosis confirmed with laboratory testing, along with family education and linkage to specialised care whenever feasible,” she says.

Moreover, she expresses a desire for expanded research and diagnostic efforts on rare diseases and welcomes initiatives that develop policy frameworks focused on rare conditions.

“Increased collaboration—both within the country and through partnerships on regional and global

scales—could significantly change the landscape for families facing these challenges,” she explains.

The Ministry of Health has initiated several publicly funded programmes that, while primarily targeting more prevalent conditions, also address rare diseases.

Drawn for comment, Dr Adman Shabangu, Senior Medical Officer at the Ministry, explains that there are existing programmes, such as the Neglected Tropical Disease (NTD) initiative, that oversee conditions such as leprosy, which the country is targeting for elimination, and schistosomiasis, which primarily affects children.

“We have integrated health services to assist in screening, diagnosing, and managing these conditions in schools and health facilities. Funding for these initiatives comes from the national budget, with additional support from UN agencies like WHO and UNICEF,” he further says.

Dr Shabangu also highlights the role of the National Cancer Program, which assists children with childhood cancers by providing funding for treatment and operations.

“There are stakeholders involved in activities related to prevention and linkage to care,” he notes.

While these programmes show promise, the landscape of scientific research on rare diseases remains challenging.

“Unfortunately, we are not yet seeing a significant number of researchers venturing into this field. Attracting funding for rare conditions remains a primary challenge,” Dr Shabangu reveals.

Looking ahead, Dr Shabangu is optimistic about future initiatives, adding that the Ministry of Health plans to further invest in primary healthcare and encourage pregnant women to seek genetic counselling and early screenings.

“We are also focusing on radiological and laboratory diagnostic improvements. However, gaps still exist in the availability of specialists to manage these rare conditions effectively,” Dr Shabangu adds. ■

Breaking Cycle: Success Story Exposes Rare Disease Care Gaps



Eswatini's healthcare landscape is marked by significant challenges in addressing rare diseases, with many conditions progressing to disabilities due to research gaps, a lack of knowledge on how to diagnose, and a lack of support for affected individuals.

The country faces critical gaps in clinical research capacity, funding, and data-sharing, leaving rare disease communities underserved and often isolated. Amahle Simelane, a 10-year-old, from Siphofaneni, is one such story. For a decade, Amahle lived with the debilitating effects of Blount disease, a rare paediatric orthopaedic disorder that causes bowlegs and mobility issues.

Blount disease is extremely rare, with a prevalence of less than 1% in the general population, and even lower in Eswatini. Without access to corrective treatment, Amahle's condition would have led to permanent disability, limiting her future opportunities and quality of life.

In a groundbreaking collaboration, the Eswatini government partnered with Taiwan to arrange life-changing surgery for Amahle at Mbabane Government Hospital.

The successful operation, facilitated by Deputy Prime Minister Thulisile Dladla and Taiwan's Ambassador to Eswatini Jeremy Liang, has given Amahle a new lease on life. She is expected to regain normal mobility and requires eight weeks of bone healing and five months for full recovery.

However, Amahle's story is not the only one. Philile Ndlela's 21-year-old daughter Miselo, born with cerebral palsy, illustrates the harsh realities faced by families living with rare diseases in Eswatini.

Cerebral palsy is a rare neurological disorder that affects movement, muscle tone, and coordination, with an estimated prevalence of 2-3 per 1,000 births in Eswatini.

Philile's daughter was born with cerebral palsy due to a blood clot in her head, which occurred when she was just three months old.

“She had two operations in South Africa to remove the clot, and the doctors told us she would never be normal again,” Philile recalls.

Despite the challenges, Philile has dedicated her life to caring for her daughter.

Philile’s story is one of resilience and determination.

“I wake up and give her a bath and feed her,” she says, her voice filled with love and exhaustion.

“She’s heavy to lift, and it’s challenging to give her enough attention since I work.”



Philile Ndlela and her 21-year-old daughter Miselo

Philile’s experience underscores the need for improved support systems for families living with rare diseases in Eswatini.

The government’s meagre E450 (approximately \$24.50 USD) monthly grant barely covers her basic needs, and more needs to be done to address the systemic gaps in healthcare and social support.

To understand the systemic context surrounding Amahle and Miselo’s case, Inhlase spoke with Simay Neves, Acting Director at the Deputy Prime Minister’s Office (DPMO).

Offering valuable insights into Eswatini’s approach to rare disease management, Simay says, “An overview of the current landscape reveals several critical gaps in research, diagnosis, and support for rare conditions, many of which remain undetected and ultimately contribute to disability,” Simay explains.

“A further challenge within the country is the limited clinical research capacity, which remains thinly spread and insufficient to address the needs of rare disease communities adequately,” she further says.

When asked about post-surgical care and long-term follow-up, Simay redirects the inquiry to the Ministry of Health, emphasising the DPMO’s role in referring cases for expert evaluation and intervention.



Amahle (blurred), Thuli Dladla, Jeremy Liang

Simay points out that the lack of a dedicated Disability Information System and a rare disease registry means that data is currently captured through broader national mechanisms.

“The establishment of a comprehensive Disability Information System has been included in the Deputy Prime Minister’s Office five-year plan, signalling a commitment to strengthening data collection and management in the future,” she notes.

In linking families to support services, Simay referenced the 2024 Handbook on Disability Mainstreaming and Disability-Inclusive Budgeting, which outlines referral pathways through community-based organisations, social workers, and partnerships with organisations such as the Federation of the Disabled in Eswatini (FODSWA).

Amahle’s mother, Jabulile Ndzimandze, expressed gratitude for her daughter’s operation, saying, “I’m so relieved to see Amahle recovering and looking forward to a brighter future.”

Amahle's story highlights the urgent need for improved rare disease management in Eswatini, from early detection to treatment and long-term support.

As Simay Neves emphasises, addressing these gaps will require sustained investment, coordinated data infrastructure, and inclusive approaches that prioritise patient perspectives. ■

Rare Disease, Rare Strength: Cross-Border Collaboration Transforms Lives



In a world where rare diseases often mean isolation, two neighbouring countries are breaking down barriers and building bridges.

For Itumeleng Morole, a 36-year-old South African cancer survivor, a stage-4 anal cancer diagnosis in 2021 was just the beginning of her remarkable journey.

The permanent colostomy that saved her life also brought new challenges – stigma, shame, and a sense of disconnection.

But Itumeleng refused to let her rare disease define her.

Instead, she founded the Ubunye Ostomates Foundation, a beacon of hope for ostomates in South Africa and Eswatini.

Itumeleng's journey is not just about personal

triumph, but about a shared struggle and a shared hope that transcends borders.

Her foundation is working tirelessly to create a community of support, awareness, and empowerment for ostomates in both countries.

Itumeleng starts her day with a routine check of her stoma.

“The first thing I do is check my stoma, clean it or change it,” she says.

But it is not just about the physical aspect; it is about the emotional and psychological toll of living with a rare disease.

“There are days where I am grateful, but sometimes it's painful. I will have all sorts of questions with no one to answer because only God can,” she shares.

Despite the challenges, Itumeleng and others living with rare diseases have learnt to adapt, to navigate the complexities of living with a stoma.

Her foundation is working with ostomates in Eswatini, providing support and a sense of community.

“I have learnt that Eswatini and South Africa are facing the same challenges. The real challenge is little to no ostomy supply in both countries, causing health risks for ostomates,” she says.

The Ubunye Ostomates Foundation is eager to collaborate with NGOs, hospitals, and communities in Eswatini to break down the stigma surrounding ostomies. By creating awareness and promoting education, they hope to empower ostomates to live openly and without shame.

“Most ostomates in both countries are in hiding because they never knew about anyone living with an ostomy, so it is still very foreign to them,” Itumeleng explains.

Itumeleng’s journey and the work of the Ubunye Ostomates Foundation is a reminder that even in the darkest moments, there is hope.

And it is not just about surviving; it is about thriving.

“I am alive, healthy, I get to do what I love and spend time with the people I love. It is a new chapter, one that is full of purpose and meaning,” she further says.

As the Eswatini Cancer Network (ECAN) continues to champion cancer awareness and support, its Program Manager Dumsile Ngwenya emphasizes the power of community in transforming the lives of cancer survivors.

“Cancer survivors in Eswatini face significant challenges, but through community support, we can address these issues and empower survivors to thrive,” she says.

Ngwenya highlights ECAN’s collaborative efforts with the Ministry of Health and other partners to provide holistic care, including medical, psychosocial, and practical support.

“Through ECAN, we facilitate quarterly regional survivor support group meetings and an annual Survivors’ Day, creating a sense of belonging and solidarity among survivors,” she states.

Ngwenya notes that the benefits of community support are clear.

“Survivors often trust advice from fellow survivors, which improves health-seeking behaviour, treatment adherence, and overall resilience. By sharing experiences and supporting one another, survivors can rebuild confidence and advocate for their needs,” she points out.

However, Ngwenya further admits that cancer survivors in Eswatini still face significant challenges, including late diagnosis, interrupted treatment, and stigma, adding that community support helps address these issues through peer support, practical assistance, and emotional care.

“Community support plays a vital role in improving the mental and emotional well-being of cancer survivors. Opportunities to speak openly with others who understand the cancer journey help reduce isolation, anxiety, and depression,” she adds.

ECAN has seen success with initiatives like the annual Cancer Walk – Brave the Breast (BTB) Challenge, which promotes awareness and raises funds for cancer support services.

“The walk brings together survivors and the general public, providing an important platform for cancer education, promoting physical activity, and encouraging cancer screening,” she explains.

ECAN promotes awareness and reduces stigma through community outreach, media engagement, and survivor testimonies.

“By sharing accurate information and real survivor stories, we challenge myths and normalize conversations around cancer and ostomy,” Ngwenya says.

Individuals can support cancer survivors and community-building efforts by volunteering, contributing skills, and participating in awareness activities. ECAN also welcomes financial contributions and partnerships.

“Every day acts of solidarity, such as offering practical assistance, sharing information, or speaking openly about cancer prevention and survivorship, also help build a stronger, more compassionate community,” she says.

Looking ahead, Ngwenya sees opportunities to expand survivor-led networks, strengthen regional partnerships, and integrate community support into the broader health system. ■

Eswatini's New Research Review Board to Tackle Rare Diseases



Eswatini, a small landlocked country in Southern Africa, is making strides in strengthening its healthcare system. With a population of approximately 1.2 million people, the country faces various health challenges, including infectious diseases like HIV and tuberculosis, as well as non-communicable diseases such as diabetes and hypertension.

Amidst these challenges, rare diseases often slip through the cracks, affecting individuals and families in profound ways.

Enter Dr. Sabelo Vusi Dlamini, a seasoned public health expert and parasitologist, who has taken the helm of the Eswatini Health and Human Research Review Board (EHHRRB) as its new chair.

Dr. Dlamini brings a wealth of experience and qualifications to his new role, including a Doctor of Public Health (DrPH), Master of Science in Public Health (MScPH), Bachelor of Science (BSc), and certification as a Diabetes Educator (CDE).

As a Senior Lecturer at the Faculty of Health Sciences, University of Eswatini, Dr. Dlamini is well-positioned to drive change in the country's health research landscape.

In an exclusive interview, Dr. Dlamini shares the importance of creating awareness in tackling rare diseases in Eswatini.

“Unfortunately, many rare diseases are neglected in the country, suggesting that we need to create awareness through the Health Promotion department. This could also improve awareness among attending healthcare workers (diagnosing doctors and nurses) so that they think of the possibility of a patient developing a rare disease when first reporting for diagnoses. Health promotion could also empower parents of affected children to identify rare conditions early and to seek corrective measures,” he explains.

He believes that creating awareness and gathering data on rare diseases are crucial steps towards

improving diagnosis and treatment.

“We do need data on rare diseases which we can use to develop patient handling processes (diagnosis and management). Many rare diseases are neglected in the country, suggesting that we need to create awareness through the Health Promotion department,” Dr. Dlamini says.

He further notes that rare diseases affect a few people and most of them are non-communicable. Their impact on communities or nationally is small but they have severe impact to the affected individual and close family members that have to take care of the affected individual.

When asked about the challenges in diagnosing rare diseases, Dr. Dlamini notes the healthcare professional such as the doctor, nurse, or other does not immediately think of the rare disease but focuses on the most common cause of the symptoms and treats according to that.

“This delays appropriate diagnosis of the rare disease,” he says.

Dr. Dlamini further emphasizes the need for improved laboratory services, saying, “Lack of adequate laboratory services at some facilities limit progress to conclusive diagnosis, causing delay in confirmation of the rare disease.”

Explaining the role of the EHHRRB, Dr. Dlamini says, “The Research Review Board has very little to do with healthcare practice but looks at the safety of research processes and does not conduct research itself.”

He stresses the importance of research in understanding rare diseases, stating, “Research is supposed to determine the cause(s) of, for example, Blount disease in specific individuals whether it’s environmental, genetic or overload of bones from obesity.”

Dr. Dlamini is further committed to prioritizing rare diseases.

The EHHRRB, under Dr. Dlamini’s leadership, aims to strengthen the country’s research ecosystem and promote ethical research practices.

Dr. Rejoice Nkambule, Deputy Director of Public Health in Eswatini’s Ministry of Health, echoes this sentiment, saying, “The creation of the Board demonstrates Eswatini’s commitment to upholding international ethical standards, safeguarding rights and welfare of participants, and ensuring accountability and transparency in health and human research.”

She explains that the EHHRRB will work closely with researchers, policymakers, and stakeholders to ensure that research is conducted with integrity and that findings are translated into policy and practice.

“The Board will also provide guidance on ethical issues, review research proposals, and monitor research activities to ensure compliance with national and international standards. We are committed to strengthening the research ecosystem in Eswatini and promoting a culture of research excellence and ethics,” she further states.

Both Dr. Dlamini and Dr. Nkambule agree that strengthening Eswatini’s research ecosystem is crucial for addressing rare diseases and promoting ethical research practices.

By creating awareness, gathering data, and ensuring research is conducted with integrity, Eswatini can better support individuals affected by rare diseases and contribute to the country’s overall health development. ■



Sharon

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Sharon Muzaki is an environmental and science journalist reporting on the intersection of science, climate change, and environmental issues. She holds a degree in Journalism and Communication from Makerere University (2019) and has received extensive training in storytelling, data journalism, mobile reporting, agroecology, climate journalism, and multimedia reporting. Sharon is the inaugural fellow of the Advancing Science Journalism in Africa and the Middle East Fellowship under the

Knight Science Journalism Program at the Massachusetts Institute of Technology, established in honour of Mohammed Yahia and funded by Springer Nature. As a KSJ Research Fellow, she completed an intensive program of seminars, study, and fieldwork across Cambridge and Boston. She currently writes for Nature Africa, focusing on high-quality science journalism and raising awareness of scientific innovation in Africa and beyond.

Can gene therapy transform sickle cell survival in Africa?



At the PEN-Plus clinic in Nhamatanda, Mozambique, Lindolfo dos Santos provides patients and family members with nutritional guidelines for managing sickle cell disease. Credit: Ivan Simone Congolo / World Health Organization

Gene-editing breakthroughs are bringing curative therapies closer to reality. The challenge now is ensuring African health systems are ready to deliver them.

Long neglected in global research and funding priorities, sickle cell disease is now at the centre of a wave of gene-based innovation. Science is proving that a cure is possible. Less certain is whether those advances can reach the patients who need them most.

An inherited blood disorder that causes severe anaemia, organ damage and recurrent pain crises, sickle cell disease requires lifelong management.

Around 515,000 babies were born with sickle cell disease worldwide in 2021, nearly 80% of them in sub-Saharan Africa, according to estimates from

the Global Burden of Disease Study¹ published in *The Lancet Haematology* in 2023. Without early diagnosis and treatment, 50–90% of affected children in parts of sub-Saharan Africa may die before age five.²

In laboratories in Europe, the United Kingdom, and the United States, often led by scientists of African descent, work is accelerating.

Feyisayo Eweje, a bio-engineer at Harvard University, is developing non-viral delivery systems designed to improve precision and reduce immune reactions in gene-editing therapies. These innovations aim to make curative treatments safer and potentially more accessible for patients in Africa.

“For sickle cell, safety is critical,” he says. “Many patients can already manage symptoms with

therapies such as hydroxyurea, so any curative intervention must demonstrate a very strong safety profile.” His team focuses on improving targeting accuracy while minimising unintended genetic changes.

Much of this research is being developed in laboratories abroad, and translating these advances into treatments for African patients will require strong local clinical and regulatory systems. “How this research reaches patients in Africa will require collaboration across academia, industry, and public health systems,” Eweje adds.

Building systems for cure delivery

Scientific breakthroughs alone will not determine who benefits.

“These therapies require healthcare infrastructure, trained personnel, and long-term follow-up,” says Gene Bukhman, a cardiologist and global health researcher at Harvard Medical School. “Without strengthening district hospitals and referral networks, most patients will not benefit from these advances.”

On the continent, efforts to build that foundation are already under way.

Julie Makani, a physician-scientist at Muhimbili University of Health and Allied Sciences, in Tanzania, has spent years developing locally led research and care infrastructure for sickle cell disease. Through the [SickleInAfrica network](#), she and her collaborators have recruited more than 40,000 people with the condition across multiple countries, creating one of the largest coordinated sickle cell research platforms globally.³

“People with sickle cell disease are often treated as research subjects rather than partners,” Makani says. “From early on, we encouraged active participation

and engagement within local health systems.” Most participants in the network receive care close to home, something she sees as essential as advanced therapies emerge. “If we can establish bone marrow transplant services and in vivo gene therapy delivery in Africa, patients will not have to travel abroad for treatment.”

Strengthening routine care

While curative therapies continue to evolve, strengthening routine care remains critical. Initiatives such as [PEN-Plus](#) expand services for severe chronic diseases, including sickle cell disease, type 1 diabetes, and congenital heart disease, to district and primary hospitals where care is often more accessible.

By providing interventions such as hydroxyurea therapy and improving referral systems, the programme saves lives, while preparing patients and facilities for more complex treatments in the future.

Neil Gupta, senior director of policy for the [NCIDI Poverty Network](#), explains that PEN-Plus decentralises services to rural and district hospitals, trains nurses and community health workers, and provides diagnostic tools. “By bundling care for multiple chronic conditions, we reach more children, reduce costs, and attract broader funding,” he says.

Countries including Kenya and Ethiopia have launched national PEN-Plus plans, and Uganda is expanding newborn screening.

Looking ahead

For Makani, building a research network across multiple African countries has required navigating regulatory bottlenecks, harmonising data collection, and earning trust in diverse settings. But the experience demonstrates what African-led science can achieve. ■



Safina

NABI

in Independent | India



Safina Nabi is a multimedia journalist and a Pulitzer Center grantee from South Asia. She writes about gender, health, human rights, social justice, development, and the environment.

India's Silent Village Shows What Happens When Genetic Clusters Go Unaddressed

DHADKAI, India—In a remote mountain hamlet nestled in the rugged beauty of Jammu and Kashmir, Mohammad Aftab begins his mornings tending to cattle—work that sustains his family, but does not reflect his true calling. Aftab, who is about 30 years old, is a trained mason, widely regarded as one of the area's most skilled. Yet he rarely works beyond Dhadkai's narrow paths—all because of a hearing and speech impairment that he's lived with since childhood.

“He was like this from the beginning,” says Aftab's nephew, Mohammad Ali, who explains that he cannot work outside the village because of the communication barrier. “That's why, despite being really good at this job, he hardly gets to put it to use,” Ali says.

But in Dhadkai, Aftab is far from alone. Often called the “Silent Village of India,” Dhadkai is known not only for its breathtaking landscape of steep hills and dense forests, but also for an unusually high number of residents who cannot hear or speak. Out of about 2,000 people who live in the village, more than 90 are estimated to live with hearing and speech impairments—an extraordinary concentration in a small, close-knit community. In Aftab's immediate family alone, all four of his siblings share Aftab's condition, even though their parents have no hearing or speech impairments.



A view of the village of Dhadkai, in the Doda district of Jammu and Kashmir. February 23, 2026. Safina Nabi

For global health experts, the village represents more than a local curiosity. It raises urgent questions about genetics, early screening, intergenerational health patterns, and the gaps in rural disability care that allow conditions such as hearing impairment to persist largely unaddressed.

Most of Dhadkai's residents belong to the [Gujjar community](#), one of the largest tribal groups in the region. Traditionally pastoralists, the Gujjars rear cattle and buffalo and often migrate seasonally, moving to higher mountain pastures in summer and descending to the plains in winter. According to India's [2011 Census](#), Gujjars, along with the closely related Bakarwal community, make up nearly [12%](#) of Jammu and Kashmir's population, with Gujjars forming the largest Scheduled Tribe in the territory. They are not a marginal sliver of society; they are a significant part of its rural and tribal backbone.

For years, the hearing condition was attributed to fate, environment, or lack of medical care. But a 2017 [genetic study](#) published by the Journal of Indian Speech-Language & Hearing Association offers a clearer explanation.

One of the lead researchers, Sunil Raina, MD, has been working on hearing loss in the community since 2009, when he worked as an assistant professor at a medical college in Jammu. “A few patients from the village had approached us [for treatment], and there were reports of an unusually high number of people with hearing impairment. A small group of us, led by my colleague Dr. Sushil Razdan, spent a few days in the village collecting blood samples,” says Raina.

The team hypothesized that they might find a specific genetic defect responsible, but when they tested a small, randomly selected group of people, they found multiple genes that could be responsible in some of the patients. The team had published the preliminary findings—the [first scientific paper on hearing loss in Dhadkai](#)—in 2012. A few years later, the Indian Council of Medical Research approached the team of doctors to conduct a full-scale investigation.

“Between roughly 2012 and 2018, we collected around 600 blood samples from the village and carried out detailed genetic analysis,” says Raina, who was leading the study. “Again, we were surprised. It was not just one gene defect; there were multiple mutations involved. That made the genetic picture far more complex than we had initially assumed.”

Researchers analyzed DNA from affected families and identified a harmful change in the OTOF (otoferlin) gene. This gene plays a crucial role in how the inner ear transmits sound signals to the brain. When a child inherits two copies of this gene that carry the mutation—one from each parent—a child is born with hearing loss.

This pattern is known as autosomal recessive inheritance, meaning parents can carry the altered gene without having any hearing problems themselves. If both parents carry the same mutation, there is a chance their child may inherit two copies and be affected.

Raina says that the solution might take time, but at this point, the best long-term option would be gene therapy, working directly at the level of the gene to correct the defect. But gene therapy is not yet widely available in India, and when it is, it is only for life-threatening diseases. Raina suggested a more immediate and practical approach would be an intervention at the premarital level.

India has nearly 4,600 to 5,000 distinct populations, many of which have practiced endogamy, marrying within the same community, for thousands of years. “If we can identify carriers through genetic screening before marriage, families can make informed choices. If one partner carries the mutation and the other does not, the likelihood of having an affected child drops significantly. Prevention, in many ways, may be the most realistic starting point,” Raina says.

As Kumaraswamy Thangaraj, a senior scientist at the Centre for Cellular and Molecular Biology in Hyderabad, explains, “Over time, endogamy allows certain recessive genetic mutations to become more common within specific groups. People may appear completely healthy but still carry the mutation and unknowingly pass it to their children. In smaller or more isolated communities, that can lead to a higher number of people developing the same condition, such as congenital hearing impairment.”

What makes Dhadkai unusual is the sheer number of affected individuals.

“The most practical intervention in such communities is not advanced treatment but prevention,” Thangaraj says. “Through genetic counseling, carrier screening, and prenatal diagnosis, families can understand the risks before a child is born. Over time, these measures can significantly reduce how frequently certain mutations appear in a population.”

But it also raises pressing public health questions. The village has limited access to routine newborn screening, genetic counseling, or early hearing intervention services—support systems that, in many countries, help families make informed decisions and provide children with assistive technologies or language support within the first months of life.

In the silence of Dhadkai’s mountain valleys, the issue is not only one of biology but also of access and awareness, and whether rural health systems are equipped to respond when science finally provides answers.

A few houses away from Aftab’s home lives 35-year-old Shabir Ahmad, who, like many others in the village, has been deaf and unable to speak since childhood.



Shabir Ahmed, photographed on a balcony with the mountains in the backdrop. February 23, 2026. Safina Nabi

Shabir is married to a woman who does not have a hearing impairment. Together, they have two children, both of whom can hear and speak,

and are described by their teachers as bright and active in school. Their presence complicates the assumption that every family in the village will inevitably pass on the condition.

But while the children are thriving academically, the family's economic reality is fragile. Shabir and his wife struggle to afford school expenses; the children's grandfather has stepped in to cover their educational costs, ensuring they remain in school.

In a village where disability, poverty, seasonal mobility, and limited access to specialized health care intersect, families like Shabir's illustrate both the genetic complexity of the condition and the social consequences that follow. Hearing loss is not only a medical issue—it shapes livelihoods, marriage prospects, and intergenerational opportunity.

“Gene therapy is scientifically promising, but it remains extremely expensive and difficult to deploy at scale, even in developed countries,” Thangaraj says. “India does have a [National Policy for Rare](#)

[Diseases](#) [which recognizes the need for better diagnosis and financial support for certain genetic conditions], but implementation is complicated, and hearing loss often falls outside the policy's definition of a rare disease.”

Hearing loss, unless linked to a high-profile rare disease, often receives limited targeted attention—especially in remote tribal belts, according to Thangaraj. In geographically isolated communities like Dhadkai, where marriages often take place within extended kinship networks, certain genetic traits can quietly become concentrated over generations. Without early diagnosis and counseling, such patterns may continue.

Dhadkai is not just a “silent village.” It is a reminder that when large communities like the Gujjars remain at the margins of structured health systems, genetic patterns can become entrenched before systems respond. Science has offered clarity. What remains uncertain is whether policy and public health will move quickly enough to meet the needs of people living with its consequences. ■



Lilys

NJERU

in Nation Media Group | Kenya



Lilys Njeru is a multimedia award winning health and science journalist with the nation media group. She is the current acting content lead- health at the eastern africa's largest media house.

Nick Wambugu, known for documentary ‘The People Shall’, battles rare blood disorder



Nick Wambugu, an award-winning filmmaker, creative visionary and storyteller, during an interview. Wilfred Nyangaresi | Nation Media Group

As the year draws to a close, few would look back on 2025 and call it an ordinary year for filmmaker and storyteller Nicholas Nick Wambugu. A lot has happened. The year brought acclaim for his ‘People Shall’ documentary, recognition as one of the ‘Business Daily’s’ Top 40 under 40, and an arrest over claims linking him to the BBC expose ‘Blood Parliament’, which both he and the broadcaster deny.

It also ends with a diagnosis that has reshaped everything- hypocellular MDS.

According to Dr Rohini Radia, a haematologist, MDS stands for Myelodysplasia. “Myelo is blood cells, and dysplasia is where they are disorganised. The common type of Myelodysplasia is hypocellular, where there are a lot of cells, but essentially, the bone marrow has become non-functional. It is disorganised and produces too few blood-forming cells.”

“Myelodysplasia is commonly a disease of much older patients,” she says. “There are no obvious known causes. Hypocellular myelodysplasia is rare, and it accounts for about 10 per cent of patients with myelodysplasia. For hypocellular myelodysplasia,

the important thing is to distinguish it from aplastic anaemia, which is also present in younger patients. Aplastic anaemia is where the bone marrow is also empty and non-functional, but it’s not associated with a pre-leukemic phase, and the treatment is different. So, the treatment for aplastic anaemia is immune suppression or a bone marrow transplant. But with hypocellular myelodysplasia, you tend to treat it with a bone marrow transplant or chemotherapy if the patient is not fit enough,” she offers.

It began quietly. The 34-year-old storyteller started feeling unusually tired. He did not immediately read the fatigue as sickness.

“That was around October. The ‘People Shall’ documentary was premiering at the NBO film festival. At the time, I was high on adrenaline and working long hours. I told myself, we work hard right now and rest after the festival,” he offers.

There was also a persistent whooshing sound. One that he remembers too well, but his film, one that depicts the 2024 anti-finance bill protest, was his priority. So, this too he ignored.

“When the festival ended, I decided to rest at my sister’s place,” he says.

We meet there weeks later. Nick sits across from this reporter, the conversation repeatedly punctuated by coughs. We slow the interview, waiting each time for him to recover.

“I was resting but feeling even more fatigued, and the whooshing sounds. It was then that my sister suggested that I see a doctor. The specialist said my condition was serious and required urgent admission to the high dependency unit for close monitoring. I was admitted for five days. On the fourth day, they did a bone marrow biopsy. Three weeks later, I learned that I had hypocellular MDS. I had never heard of it before,” he says.

In his telling, the doctors did not point to a specific cause but suggested it could be genetic, possibly triggered by other factors.

“The condition impacts all three main blood cells,” Dr Rohini says. “Low red cells cause fatigue, low white cells increase infection risk—including neutropenic sepsis—and low platelets make you prone to bleeding easily.

For the filmmaker, it all came down to this: staying away from others. His work, his stories, and his career—all of it is temporarily on pause. Blood Parliament arrests

Nick was among four filmmakers roughed up by police at their Karen studio on May 2, linking them to the BBC documentary. The documentary alleged that the Kenya Defence Forces and the National Police Service were involved in some of the killings during the Gen-Z-led protests of June 25, 2024. The case is still a matter in court.

“I tell stories of the mavericks, the misfits, the people who never get the spotlight but form the backbone of who we are. I cannot do that now, and it feels weird being on this other side, where I am being interviewed and talking about a condition I knew nothing about a few months ago. Besides the shock and the stress that came with the arrest, I have enjoyed great health in the recent past,” he remarks.

Now, the condition demands at least Sh100,000 every week from his pocket to manage it. “It is an expensive affair. I miss the camera, I miss the long nights edits, but the one thing that’s on my mind right now is, ‘how do I raise the Sh9 million for the transplant?’

According to Dr Rohini, the treatment depends on how severe the low blood counts are and the risk of developing or progressing to acute leukaemia from myelodysplasia.

Ultimately, the only curative option is a bone marrow transplant. As there are no known certain triggers, it’s not considered preventable.

“For patients with hypocellular myelodysplasia who have very low blood counts, there is an urgent need for bone marrow transplant because of the risk of having a number of blood transfusions, and the risk of life-threatening bleeding as they have low platelets.”

Friends, colleagues, and well-wishers have raised Sh3 million to support his treatment. “The worry is that whatever resources we raise go into the management of the condition, and that’s why I urgently need a bone marrow transplant. The longer I stay here, besides the cost, I risk contracting other diseases as I am now vulnerable to opportunistic infections,” Nick offers. ■

Fibroids and fertility: The choices Kenyan women are forced to make



HIFU JC 300 machine at Nairobi HIFU Treatment Centre. It is used as a non-invasive treatment option for uterine fibroids through focused ultrasound waves to heat and destroy fibroid tissues, achieving one-time ablation like surgery. Photo I Pool

What you need to know:

- Ada Zillah lived with fibroids for nearly four years, managing heavy bleeding, pain and fertility fears with painkillers while avoiding surgery.
- Her story highlights how fear, misinformation and limited treatment options leave many women trapped between suffering and risky choices.

In July this year, Oscar-winning actress Lupita Nyong'o posted a message on Instagram that stunned her millions of followers. For more than a decade, she revealed, she had been battling a condition in silence.

"I no longer want to remain silent," she declared. "That's why I want to share my personal story for the first time... so we can invest in this research and find early detection and treatment and solutions."

The condition was uterine fibroids. Since her disclosure, other celebrities have come forward to share their own struggles. But for every famous woman who speaks out, millions of others continue to suffer without a platform, without a diagnosis, and often without hope.

Ada Zillah, now in her 30s, battled with uterine fibroids for close to four years fearing that surgery would result in loss of her fertility. Mid this year,

at Nairobi HIFU Treatment Centre, she underwent high-intensity focused ultrasound treatment, a non-invasive, and precise medical technology that targets the lesions.



Ada Zillah, now in her 30s, battled with uterine fibroids for close to four years, fearing that surgery would result in loss of her fertility. Mid this year, at Nairobi HIFU Treatment Centre, she underwent high-intensity focused ultrasound treatment, a non-invasive, and precise medical technology that targets the lesions. Photo I Pool

Ada Zillah knows this silence intimately. For nearly four years, fibroids dictated the rhythm of her life—what she wore, where she could travel, how she planned her days. Like Lupita, she carried her burden quietly. Unlike Lupita, she had no idea that a stranger's instinct would eventually lead her to a treatment that changed everything.

The hidden epidemic

Data from the International Federation of Gynaecology and Obstetrics shows that up to 70 per cent of women are affected by fibroids by the time they reach menopause. Other estimates suggest the figure may be as high as 80 per cent by age 50.

Yet public awareness remains strikingly low.

"I like that we can discuss issues like postpartum depression openly. Look at cervical and breast cancer. There are a lot of awareness campaigns around them. But not all women's health issues get that spotlight. Take fibroid diagnosis for instance. While many people seem to know October is breast cancer awareness month, very few know that July is dedicated to fibroids," says Sharon Mukami, a post-fibroid patient.

Dr Mugambi Jackline, a certified obstetrician and gynaecologist and skilled in high-intensity focused ultrasound technology for non-invasive treatments (HIFU), states that many women carry their health burdens silently, adding that Black women are disproportionately affected by uterine fibroids compared to other races due to factors such as genetics, environmental exposures, and delayed diagnosis and care.

Dr Naushad Karim, Consultant Interventional Radiologist at Aga Khan University Hospital, explains that fibroids are “non-cancerous growths that develop from the muscle layer of the uterus and can appear in different locations, inside the uterine wall, bulging into the cavity, or projecting outward into the pelvis. Though their exact cause remains uncertain, hormones such as oestrogen and progesterone play a major role in their development, and genetics often influence whether a woman is predisposed to them.”

Further, fibroids respond to reproductive hormones, particularly oestrogen and progesterone. They usually do not appear before puberty, tend to shrink after menopause, and can grow quickly during pregnancy.

When it began

Ada first noticed something was wrong in 2021. She works in project management, and she was on a field assignment with colleagues when her body began sending signals she could not ignore.

“The first time I experienced it was in 2021,” she narrates. “I work in project management, and my colleagues and I were out for a field assignment. One week after my monthly period, which used to last at most three days, I realised that there was still some discharge. In large quantities. Two weeks, three weeks. It dawned on me that I was literally in a menstrual pad for the entire month. I was also cramping in between. Not extreme, but uncomfortable.”

A gynaecologist confirmed that she had six fibroids. The largest measured approximately 2.7 centimetres.

“So, I told myself, that’s tiny. What’s the rush to have them removed?”

Her indifference, she later reveals, was anchored in statistics.

“I know dozens of women with uterine fibroids. At the top of your mind, you probably know tens of them who are just managing them. When a problem is very common, one can be nonchalant.”

When the body rebels

Her nonchalance did not serve her well.

“But, I think I spoke too soon because after that, I started experiencing an increase in flow and a lot of blood clotting. And when I say clotting, I mean serious clotting.”

That was in 2022, a year after the diagnosis. She managed the pain through different types of painkillers and menstrual products.

“By 2023, the symptoms intensified. I could vomit from period pain. When it started happening every other month, I knew, I just knew that something had changed,” she offers.

Upon monitoring the fibroids, the examining doctor found that one of them had grown to about 3.6 centimetres. The treatment option was surgery.

“I didn’t want it.”

Those are words that Margaret Kamau, a counselling psychologist and mental health advocate, has heard many times.

“There are cases of shame, there are those who have concerns around their identities and are often wondering if they will ever give birth. And, will the pain ever go away?”

“In Kenya and around the world, women have long endured the discomfort, sometimes believing that hysterectomy, the surgical removal of the uterus, is their only option,” offers Dr Karim.

The fertility dilemma

For Ada, the resistance to surgery was rooted in a deeply personal fear.

“I don’t have a child yet. I wanted the reassurance that someday, I could have a child if I wanted. Because of the many women I know who at one point in their lives, have been diagnosed with fibroids, I also know quite a number who have had to go through a hysterectomy or a surgical removal of the uterus. So, instead, I normalised taking painkillers. But I also noted that I had become lethargic. I wanted to preserve my fertility.”

“That’s why I didn’t want to do anything that might interfere with my uterus. Doctors told me that because of where the fibroid was, I had one inside the uterus, even if I tried to conceive, the risk of miscarriage would be very high. They advised me to be extremely careful and to make a very considered decision.”

She found herself trapped.

“But living with the fibroid was also dangerous. And from what I had seen, removing it surgically felt just as risky. So, I found myself stuck, managing the pain with painkillers, trying to cope, because either option felt dangerous.”

A stranger’s intervention

Ada had been sick for two years, but she had not planned on seeking treatment that day. In fact, the decision was not even hers.

She was in the back of a taxi when the driver caught her eye in the rear view mirror. Noticing her exhaustion, he bypassed her destination and insisted on driving her to the nearest hospital, without extra charge.

He confided in her that he was a widower, having lost his wife to anaemia.

“I know women go through a lot. You got in and slept. That is worrying.”

“It’s just fatigue,” she protested.

But he insisted.

This time, she had a low haemoglobin level, and she was put on blood boosters. Every month, she made a trip to a health facility to make sure that she was on the right track. Still, surgery was not a route she was prepared to take.

A life on hold

“Between 2021 and 2024, I had visited at least four specialists. One time, I went to one who cautioned me about carrying a pregnancy because of the low blood levels. By then, my menstrual cycle had doubled. I was now having six days of heavy flow, which means that I had to change the menstrual products more frequently. Bloating became part of my life,” she says.

Margaret Kamau emphasises the importance of addressing both the physical and emotional dimensions of chronic conditions like fibroids.

“When handling women with this chronic condition, one of the things I do is psychoeducation; encouraging women to understand as much as possible about fibroids: what they are, the symptoms, and how they present. This helps reduce uncertainty and stress because when you understand what’s happening, it becomes easier to cope. I also focus on the emotional toll. I equip them with skills for emotional regulation, particularly to manage anxiety linked to worry and shame, and the self-questioning that often comes with the condition.”

Over time, Ada says, it felt as if her life was centred around the fibroids. “When I was travelling for work, I had to request that I be assigned a female driver during my menstrual period because of the many stops we’d need to take. This, coupled with blood boosters and painkillers, was weighing me down. I saw yet again another specialist who gave me a variety of treatment options.”

The search for alternatives

“When it comes to treatment, there are different approaches depending on the size of the fibroids, their location, and the symptoms a woman experiences,” explains Dr Karim.

“The first is watchful waiting, suitable for women nearing menopause or those with mild symptoms, since fibroids often shrink naturally when hormone levels drop. The second approach is medical therapy, which uses hormonal medication to reduce bleeding or temporarily shrink fibroids, though symptoms often return once treatment stops.”

“The third is surgery, which can be either a myomectomy (removing only the fibroids and

preserving the uterus) or a hysterectomy (removing the uterus completely). The fourth is uterine fibroid embolization, a minimally invasive procedure that works by cutting off the blood supply to the fibroids, causing them to shrink and die off. Gradually.”

A new technology

“In June, my doctor introduced me to a new treatment option known as High-Intensity Focused Ultrasound (HIFU). It was the first time hearing about it,” Ada offers.

For years, Dr Mugambi, working at the Nairobi HIFU Treatment Centre, has seen women at her clinic suffering from uterine fibroids.

“HIFU is a relatively new clinical technology that uses non-invasive ablative technology where we use ultrasound sound waves to burn off the fibroid. The low-intensity sound that comes from the ultrasound is directed through the abdomen to where the fibroid is, and where the ultrasound waves converge, they convert that energy into thermal energy that kills off the fibroids. With time, they start shrinking. We use the same diagnostic ultrasound machine to monitor the real-time progress of the treatment,” she explains.

Evidence from multiple studies suggests HIFU is a viable non-invasive option for some women with fibroids. Minor side effects such as fatigue, mild fever, or light bleeding may occur for a few days, but they typically resolve quickly, Dr Mugambi offers.

Crucially for women like Ada, the procedure offers hope of preserving fertility.

“HIFU is a fertility-sparing procedure because the procedure does not damage the endometrium; we don’t damage the integrity of the uterus because we don’t cut it open. We have had patients who were supposed to undergo IVF and are advised to first have the fibroids removed, especially if they are small, so that implantation can be done,” she explains.

The results

Dr Mugambi shares that fibroids are expected to shrink over time, with a follow-up being

recommended at three months, six months, and one year. For Ada, the results exceeded expectations.

“In my case, the doctors noted that the fibroid had shrunk by about 90–95 per cent just three months post-procedure, far faster than I had expected. By the time I was going for the procedure, the largest fibroid was about 6.5 centimetres. My symptoms, including heavy bleeding, clotting, and pain, have almost completely resolved. For the first time in years, my periods are lighter, less painful, and easier to manage,” offers Ada.

Today, sitting comfortably on a sofa with a pillow gently placed on her lap, Ada looks at the wall clock and stops mid-conversation. We watch the second hand sweep past the 12. “If this were a year ago,” she notes, “I would have probably excused myself a couple of times already.”

The question of access

However, while it is a viable choice for women desiring to preserve fertility, its cost may limit access.

“I paid around Sh500,000 for the procedure. My medical insurance covered part of the bill, but I didn’t receive any support under the Social Health Insurance Fund (SHIF) pool. When I inquired, I was informed that the Social Health Authority (SHA) is not currently endorsing HIFU for fibroid treatment, pending further assessment of its technical and clinical considerations,” she offers.

For a condition that affects up to 80 per cent of women by age 50, Ada, who is now recovering, laments that awareness, research, and support for women is still low. “As I was doing my research for the HIFU treatment, I realised that many women are going through reproductive health issues. And I remember that by the time I was going for the procedure, I had a distended abdomen, which made some people think I was pregnant.”

She pauses.

“We have been told that period pain is normal, and that’s why some people don’t get checked. But it’s not normal, so it’s important to get checked.” ■



Precious NWONU

Freelance | *Nigeria*



I'm a freelance journalist who has reported different news stories on education, health and social issues in the society. I have written stories that have gotten the attention of public leaders.

I'm passionate about using journalism as a tool to amplify voices that are often unheard in the society.

Economic Burden of Sickle Cell Disease in Nigeria: When Treatment Costs More Than Survival

In Nigeria, sickle cell disease (SCD) affects an estimated 2–3% of the population, with roughly 150,000 babies born with the condition each year. Beyond the medical challenges, the financial toll on families is staggering, deeply entwined with the country's harsh economic realities. Routine management including regular consultations, blood tests, hydroxyurea, and folic acid medications can cost tens of thousands of naira each month. In severe cases, hospital admissions and emergency care can push monthly expenses into the hundreds of thousands, sometimes reaching up to ₦1,000,000 for a single patient.

For many families, treatment is paid entirely out of pocket, as health insurance rarely covers the full spectrum of care. In an economy marked by inflation and limited social safety nets, this creates a severe financial strain. A study in southeastern Nigeria found that the median monthly economic burden for a patient was about ₦76,711 (≈ US\$385), with even higher catastrophic costs hitting the poorest households hardest. The data underscores a grim reality: medical care for SCD can eclipse basic living expenses like food, housing, and education.

The financial pressures extend beyond hospitals and clinics into the everyday lives of patients and caregivers. Surveys indicate that over 70% of families caring for children with SCD report lost income, as time spent on caregiving interferes with employment. Many families borrow money, sell assets, or make painful sacrifices just to meet the cost of care, highlighting the intersection of health, poverty, and inequality. Women most often mothers bear the brunt of caregiving responsibilities, juggling unpaid care with dwindling job opportunities, sometimes even sacrificing their careers entirely. In a country already grappling with inflation-driven price increases and job insecurity, SCD creates a cycle in which health hardship and poverty reinforce each other, making the disease not only a medical but also a profound economic and social challenge.



When Treatment Costs More Than Survival: Patients Speak

To understand the human side of these statistics, several sickle cell patients shared their experiences through in-depth interviews. They described the routine care required, the financial sacrifices, and the toll on work, school, and family life.

Alabi Motola, a young adult student living with SCD, spoke candidly about the daily routines and challenges that define his life:

“I have been keeping up with a lot of routine since medical tests confirmed my genotype group. I normally visit hospital at least every two months whether I’m sick or not. I have been doing this because doctors said the illness is complicated, and I need on-time assessment to avoid complications. I use Folic Acid and Mayanil tablet every day, in a bid to keep my cell strong. I take too much water according to doctors’ advice, and I ensure I don’t expose myself to heat or cold.”

“All the above are my routine whether I’m well or unwell. But if I’m sick, those periods are the trying time. I visit hospital almost every day due to the kind of pain I normally go through those times. I normally received treatments at Usmanu Danfodiyo University Teaching Hospital. The section that attend to me is a special department (MCOP) and they opened once in a week between 8am to 11am.”

Motola also highlighted the financial challenges of living with SCD:

“There’re a lot of financial responsibility that comes with managing my health conditions. If we look at it, I will normally end up spending more than people that are not facing similar health challenges like me. For the daily drugs, I normally buy the two packs that contain 100 tablets each at the rate of 3500. They last me for three months. I take sachets water, at least half bag in a day, and a bag is sold at 350 to 500 depending on my location.”

“Whenever I’m sick, I spend a lot of money on transport, more than 2000 per day many times because the hospital is far distance to where I’m. Although, I don’t pay much for the hospital bill, I spend a lot of money on drugs. Sometimes, I spend 5000 on a single pack drug of drugs that doesn’t contain more than 10, and which may not last me than a week. I buy more of those kind of drugs like four or five depending on my condition and doctors’ prescription.”

“As an adult, I normally settle nearly all my financial responsibilities from my pocket. Although, I’m a student, I have skills that generate income for me. Nevertheless, there are some times when I go broke. Majority of the time when I get sick, the illness always meet money with me. Sometime when the expenses outsource my budgets, I seek assistance from my parents, siblings and at times, my friends support me.”

Motola also spoke about the emotional and family impact of his illness:

“There’s a lot responsibility that comes with it honestly. I have gave up a lot of things I love doing because my health didn’t support it. Whenever I fall sick, I hardly move around or do anything because the pain always comes heavy. So, within those intervals I’m always cut from my work, I miss classes, and some other things too. About two to three years ago when I’m working in a bakery, that

I don’t even know I’m living with such condition, whenever I miss works because of my illness, the company will deduct each day I miss from my payment at the end of the month. But now, I work remotely, and don’t really work under pressure. Whenever I’m sick, I notified my superior who always understand my condition and bare with me until I feel better.”

“There are people whom their support have become a weapon in navigating the health challenge. Apart from caring and standing by me, they also use their money to support me. Honestly, taking care of me do affects them because attending to me consume some of their time. When I was a kid, the sickness has once drained my family physically, emotionally, and financially. That year, I was critically ill more than ever before. My parents, siblings and some relatives stay in the hospital since morning we’ve left home for my treatment until the late in the night. That day, all of them were afraid I may not survive because all my hands and eyes were white and I felt a lot of pain I couldn’t withstand that day, I later ended up receiving blood. The money, stress, and other treatments after we left hospital really affected my whole family.”

“Also, the last time I was badly ill was around October last year. I was in school not at home. I was admitted for two days before I insisted I should be discharged. My friends that stood by me were unable to proceed with their business. They missed classes because one of them need to stay with me at the hospital and the others stay from morning till the evening whenever they bring food.”



*Usman Dan Fodio Teaching Hospital, Sokoto State
Picture by: Precious Nwonu*

“Nigeria is just a country where government’s assistant is nearly inaccessible. Relying on subsidized treatment is not something achievable with the whole situation of the country. So, one needs to strive to take care of things independently, if not the consequences may be daring.”

“Even in this hospital where you pay for everything, many will nearly die on queue before it reach your turn to get medical attention. The last time I got sick badly, my condition worsened around 11pm. I had to hold up with pain with a lot of cry throughout the night. When it was dawn, we managed to get a bike that transported me to the hospital. We get to the hospital around 6:30am, and the worst happened. I was writing with pain on the floor, but I wasn’t attended to because the hospital was short of staff as at that time. Doctors were not around, and there was no bed space. I was attended to by 10am, after a doctor arrived and got a space after a patient was discharged. This part is so saddening. This is the fate of thousands of patients in Nigeria hospital today. Imagine you are on the verge of death, and you are helpless despite being in the hospital. We just hope the system gets improved.”

Another SCD patient, Hafsat Abdul, shared her approach to managing care:

“Follow ups with hematologist, routine drugs folic acid, hydroxyurea and paludrine. Also home pain management plans or visiting the hospital during pain crises. I manage cost through health insurance. In the event of an unexpected health emergency, my family and I rely primarily on health insurance to cover most treatment expenses or my sister who happens to be a doctor manage to take care of me and we also try to make financial plans no matter how little. Living with sickle cell disease has affected my ability to work and study consistently due to unpredictable pain crises, fatigue, and frequent medical needs, the challenges can lead to missed days, reduced hours, or the need for flexible schedules, making it harder to maintain a steady source of income. They play a vital role in supporting me by helping with my medical appointments, pain management plans and daily activities during illness and sometimes it requires them to take time off work or adjust their schedules which can affect their income and daily activities. Careful financial planning with increased support from family to manage both routine and emergency care.”

Privilege Meets Challenge

Bilal Sani Umar, while acknowledging the expense of care, benefits from financial stability:

“Managing sickle cell disease is expensive. Alhamdulillah, I come from a financially stable background, which has ensured that my medications are always available. During emergencies, my parents promptly rush me to the hospital and settle all medical bills without hesitation. I have been truly privileged educationally. From primary school through my master’s degree, I never missed an examination despite missing many school days due to sickle cell-related illness. Alhamdulillah for that strength. I have always encouraged myself with the belief that ‘I am the sickler; sickle cell disease will not define me.’ My family remains my greatest source of support. They have played an indispensable role throughout this journey, standing by me through every challenge. I sincerely pray that Allah rewards them abundantly and grants them Jannatul Firdaus. However, access to healthcare in Nigeria remains a major challenge. Frequent strikes by healthcare workers make timely medical care difficult—not only for sickle cell patients but for many Nigerians who rely on government-owned hospitals. In emergency situations, I am sometimes forced to seek care from private health facilities, which are extremely expensive.”

NGO Perspective: Ibrahim Khalil Abubakar Bagudo Foundation

Mariya Abubakar, the executive director of the Non-Governmental Organization for Sickle Cell patients, Ibrahim Khalil Abubakar Bagudo Foundation (IKAB Foundation) based in Sokoto, shared detailed insights in an exclusive interview about the challenges sickle cell patients face, particularly the financial strain in Nigeria’s current economic climate. When asked to describe the regular medical care someone living with sickle cell disease typically needs, and the kinds of costs families have to plan for, she responded:

“I have had collaborative sessions with the Kebbi state Sickle Cell association. So to describe the regular Medicare health care, I’ll tell you it’s a routine, because firstly, there is daily medications, medications you take daily for your health, like B-Complex, like Folic acid, like Hydroxy like DF. These are mostly medications most sickle cell

patients take on a daily. And then there are abrupt medications for when you have medical flare ups, when you have a very critical flare up, when you have sudden crisis. You know, sickle cell patients' crisis is literally just what happens when your blood flow is affected by your sickle cell. So, like all those crises they have, they are just called sequesteria crisis."

"There's also overlapping conditions that come with sickle cell anemia, like sickle cell complications. Some could have cardiovascular, some could have gastro, some could have neurological complications that would overlap with the supposed condition, and then this would also incur more financial cost – which are different kinds of costs families have to plan for. So I would argue that you can't even plan directly for it, because while you can plan for the monthly, you don't know what will happen in the month. You don't know."

"I love to emphasize that not every sickle cell patient has mental health challenges, but there are people that suffer from mental health challenges depression, anxiety due to discrimination or constant stigmatization. These are also kinds of costs, because you may have to see a psychiatrist or psychologist, or pay for therapy. And then also there's the cost of comfort. Some people with sickle cell anemia cannot withstand any unnecessary stress. They can't use public transport. They can't stay where there's no light, so their house has to be fully ventilated, always, always has to have light. So there are also families that have to deal with the cost of fuel, or the cost for solar or inverter so that your place is always air-conditioned. There are extra costs, like having massage devices, like having some medical equipment at home that you can just use. It could be a pulse oximeter, a glucometer, because, like I said, there are overlapping conditions that could come with sickle cell anemia."

"So like these are, they are different kinds of costs that families really, really have to plan for with sickle cell anemia. And it's not direct; it depends from one patient to another. One could have these complications, one could not have these complications – some could have to have very, very, highly specialized treatment. And then there's also the one for food. Like my brother, he had certain foods. He cannot eat certain meals; he can only take certain meals. So there's also that mental cost, or physical cost, of having to cook different

food from the family. There's this stress of having two meals every day, so like you're cooking six meals in the morning, for him, for the patient, and for others. And then there's also the financial cost of food. Most of the time, the food, especially for those with dietary restrictions, for those with anemia, are very expensive. So there's also that cost. You can see that there are different tiers, different levels, to the different kinds of cost families usually have to – which are all part of medical care, because medical care is not just about the drugs they take. It's not just about the hospital. It's about holistic care of their mental, physical, psychological, and nutritious state. So all of this tends to affect the kind of what families have to plan for."

"So, like I said, it depends on families literally. There are families like middle class, and then there are families that cannot afford the finances to pay for the medical bills. Most families can't afford. Some go to family, relatives, uncles, aunts. Some have to sell jewelries, family belongings. There's literally this patient, she's deceased now; she died in November, but her family is still yet to repay the loans they took from different people to pay off their medical debts for her health condition. She was a sickle cell patient with a plethora of healthcare conditions."

"There's also drugs that go to NGOs like ours. We fundraise for different patients to afford healthcare bills, and we had a programme that we are trying to fully elaborate this year that covers basic healthcare for people with sickle cell anemia. Literally, we pay medical bills for people with sickle cell anemia. So there are also those that go to NGOs that kind of help pay vendors, taking loans, and then they're selling off family belongings or personal belongings so that they can kind of still pay for it."

"So I don't know if you know, but according to the committee for people living with disability, DCRPD, chronic illness is a form of disability. We call it the invincible disability. These are disabling chronic health conditions that affect day-to-day life. Now, one of these is sickle cell anemia. Well, not everybody falls under it, but most fall under it. Why? Firstly, there's the fact that it's unpredictable. You can be healthy now, and the next moment you're writhing in pain. You are rushing to the hospital. You are rushing to the emergency room. It's literally unpredictable to your energy levels. It's not the same as other people. You have low energy levels. You suffer from chronic exertion, chronic

pain, and chronic fatigue. Now, it's not the normal kind of, 'Oh, I'm tired, I want to rest.' No, this is a chronic kind of fatigue that comes and it's just untreatable. It's there in your body."

"And I love to emphasize this: what people don't understand is that even when these sickle cell patients look healthy, they aren't. They are never 100% healthy. It's just that, 'Oh, the pain today is bearable.' It's not weakening me, it's not the kind of glue-to-the-bed, levitating pain. So there's that, like, keeping in mind that these are people who suffer from chronic fatigue. There are days you wake up and you just can't show up at work because some people have sensory overload. People with chronic illness cannot just deal with sounds or light, and it affects them badly. They cannot show up. They fall sick on exam days, or on the day they have a presentation at work, and that's why I most times say that hiring people with chronic illness disability is harder than hiring people with physical disability."

"Because with physical disability, sometimes, once we have a ramp, you can manage. I'm not downplaying the struggles, but once you have a ramp and these things, it works. But with invisible disability, it's unpredictable. So yes, it affects people in so many ways, with chronic fatigue, chronic exertion. For some, like my late brother, if the day is sunny, once he goes out, he's going to fall sick the sun affects his body a lot. There are people who cannot work long hours; they have to have breaks in between, to stand up, stretch, relax, to not think about work. Most workplaces are not flexible enough."

"All of this makes them high-risk employees. It makes it harder to employ them. And then, when they are employed, people think, 'Oh no, they're just seeking special treatment. How are you always sick? You're just pretending.' Yes, we know you're sick, but you can chest it now. You can be strong today."

"I did a project where I interviewed people with sickle cell anemia, and one of them told me she has a business. There are days that she doesn't open her business because she wakes up very sick. She cannot do anything and shuts her business on those days. That means whenever she is sick, her income goes a little low. She's not able to show up to work. Some employers deduct pay for missed days, especially casual workers. I know someone who is a nurse; she finished nursing, but she does not work now because

she cannot afford it. It's too strenuous. When she goes to work, she gets back pain, hand pain, leg pain, affecting her ability to work and maintain a stable income."

"This is emotional. There's a psychological and financial aspect. When you talk to people with sickle cell anemia, you realize that a lot of their parents have strange relationships. Not a lot, but quite a few. Almost like the mother is the primary caregiver, always in the hospital, always doing the blood tests, scans, investigations. She is particular about what the child eats and does not eat. The father is like the financial backbone. But he gets tired, saying, 'You're always spending my money.' The mother is like, 'I am always in the hospital, always giving care. You're not even here.' I've seen a lot of sickle cell patients who cannot use their hands or legs during a crisis. Most times, it's the mother carrying them on her back."

"It's always the female primary caregiver feeding, bathing, even washing when the child defecates, even when grown. This puts a strain on parents' relationships. There's also the academic part. My mother had to give up her PhD in the early years of my brother's illness because she couldn't cope with the hospital and her studies. She returned almost seven to eight years later, which delayed her career progression. Your life achievements and ambitions take a backseat because your child is in pain. And there's guilt. You feel it's your fault somehow. Sickle cell anemia is preventable if genotype compatibility is checked, but for my parents, my mum didn't have genotype testing before marriage. It was only later she discovered she was AS, not AA, but by then, the deed was done."

"Let me tell you one thing, I'm not going to sugarcoat taking care of a sickle cell patient in Nigeria is a lot. From food to medications to hospital visits it's hundreds of thousands, sometimes even millions of naira. If you have other kids, you have to care for them too. Your personal self suffers; you can't show up for yourself or others. Sickle cell patients get sick multiple times a month; sometimes admitted twice or thrice. If you work in a government or private institution, you're the primary caregiver you have to take leaves. You can't just quit your job, one income isn't enough. You need physiotherapy, mental health support, and a safe, nurturing environment. Caring for a sickle cell patient is multifaceted and extremely demanding."

“When my brother was starting, he had to take medical drugs daily, and it will cost a lot. His drugs are going to cost like I’m talking about as way back as say, 2006 2007 2008 My brother died in 2019, but up until his death, his drugs would cost like thirteen – seventeen thousand naira, depending on the severity of the kind of drugs you only get. Now you’re spending nothing less than 30. You’re spending 25 you’re spending 30. You’re spending 35k on a monthly routine drugs – this is not something you take just because you’re something you need daily, daily. You have to take it 30 days in a way, in a month, seven days a week. So how many households can afford that? What’s the minimum wage? 70k how many households in Nigeria and above minimum wage? You got siblings who have who their school fees have to be paid. You’ve got many mouths to feed. You’ve got family responsibilities. You’ve got a lot of things. How many households can absolutely afford for their child to take routine, daily drugs? Now, the cost of this is that you see families to cut costs if you take this drug today, then you don’t take tomorrow, and that means you are reducing the dosage your body needs to function properly. That means it’s going to affect the quality of life you’re living, because you’re not taking your drugs as prescribed as you’re supposed to, because you’re trying to cut costs – which is a very big economic challenge. You see a sickle cell patient have severe health condition, a flare up, a crisis, and he’s at home. He’s wincing in pain. He’s crying in pain. Parents cannot afford to take him to the hospital, because when they take him to the hospital in a day, 50,000 naira is gone. 70,000 naira is gone, 100,000 naira is gone – this is at a local, manageable level.

“What does that mean? It means that you watch your child in pain, you watch your family in pain. You watch your loved one in pain, and you’re able to do nothing because you can absolutely not afford it. So your child is wincing and writhing in pain, and you’re not doing anything about it. There’s a lot of people who have to do this hip surgery. You know there’s, there’s this hip surgery.

“There’s this health condition whereby the hip surgery, you have to hip surgery, because their hip bone kind of shift from its position and my brother had to do it. And even as of then, it runs into hundreds of thousands. We’re talking currently about millions of Naira, not 1 million, about two plus million naira, while in some hospital is three to four million naira to do this.

“And what it means is that a lot of people cannot afford it, hat’s why you see some sickle cell patient, one leg of theirs is, higher up than one leg. I don’t know how to experience it. You can see, how do you walk? You can see their legs are not balanced. It’s the it’s that condition that causes it for them, and you cannot afford that. Well, it also means that it has you have to cut costs. You have other like I said earlier. You have other mouths to feed. So how do you do it? Because when you are when you have to feed so many people, that means other people would not get as much as this person. And then there’s also subtle resentment. it’s something that we don’t talk enough about, but it’s something that in my years of advocating for sickle cell patient, talking to people with sickle cell patients in their families, there’s this subtle resentment with siblings. Like, yes, you know, he said, But why? Why? Why do you have to put your life on hold because your sibling as of, why are your demands and your needs and your wants put last because your brother, your sister has sickle cell condition, but that’s what has to be done. He has to survive, but I hear you. You empathize with him, you sympathize with him, you understand. But it’s also kind of hurts you, and it’s breeds subtle resettlement and everything, and that’s one of the effects of the economic challenges family affected by sickle cell disease face that is in a more severe level than before.”



Mariya Abubakar, Executive Director, Ibrahim Khalil Abubakar Bagudo Foundation (IKAB)

Call to Action: Turning Awareness Into Impact

The stories of families struggling with sickle cell disease in Nigeria make one thing painfully clear: without systemic support, the economic burden of this condition will continue to trap patients and caregivers in cycles of hardship. Research shows that the median monthly cost of care for a sickle cell patient in Nigeria can be around ₦76,711 (about US \$385), with outpatient costs making up the bulk of that burden. Many households face catastrophic spending, especially the poorest families who spend disproportionate shares of their income on care. A report from Nairametrics also highlights that average monthly treatment costs including routine medications, consultations, and hospital stays can range from ₦70,000 up to ₦1,000,000 or more in severe cases.

These financial realities demand urgent, coordinated action. Policymakers must strengthen public healthcare infrastructure, including expanding comprehensive health insurance coverage that truly covers routine treatments, emergency care,

and essential medications like hydroxyurea and folic acid. There is also a need to roll out universal newborn screening and genotype education programs nationwide to prevent future cases and catch complications early.

Beyond government action, community support and NGO involvement are vital. Increased funding for sickle cell advocacy groups and support networks can help fill gaps in care and raise awareness about the disease's genetic nature and preventability. Employers and educational institutions should adopt policies that accommodate the unpredictable health needs of people living with SCD, such as flexible work hours and protected time off for medical care.

Finally, as health professionals, advocates, and everyday Nigerians, we all have a role to play in challenging stigma, increasing public understanding of sickle cell disease, and pushing for solutions commensurate with the scale of the problem. Without a concerted and sustained commitment, the silent struggle of millions will continue to unfold in hospitals, homes, and communities unmet and unheard. ■

Rare Disease Day 2026: Moving Forward, Looking Ahead



Why Nigeria Must Not Be Left Behind

Every year on 28 February, the world comes together to recognise Rare Disease Day, a global health awareness event dedicated to the hundreds of millions of people living with rare conditions that too often go unseen and unsupported. Established in 2008 by EURORDIS – Rare Diseases Europe, this day has grown into an international movement spanning over one hundred countries and countless organisations, clinicians, researchers, caregivers, and, most importantly, patients. The date, the last day of February symbolizes rarity itself, reminding the world that though each rare disease may affect only a few individuals, collectively they represent one of the largest underserved health populations on the planet. In 2026, the theme "Moving Forward. Looking Ahead. An Event for Patients" places

the lived experiences of patients at the centre of the global conversation, urging health systems, policymakers, researchers, and communities to think not only about discovery, but about meaningful progress that improves people's lives. For Nigeria, this day offers a powerful opportunity to reflect on how rare diseases shape human experience and reveal gaps in health access, even as it connects the Nigerian voice to a global movement.

Rare Diseases: Common in Impact, Invisible in Conversation

Though the term "rare disease" might suggest exceptional or isolated cases, the reality is deeply human and widespread. There are thousands of identified rare diseases, most of them chronic, progressive, and life-long, covering a spectrum of conditions that include metabolic disorders, genetic syndromes, immunodeficiencies, neurological conditions, and many more. When rare diseases are viewed collectively, they impact hundreds of millions of people worldwide, from cities in Europe and North America to villages in Asia, Latin America, and Africa. The vast majority of these diseases have limited or no approved treatments, meaning that patients and their families often navigate a landscape of uncertainty and unmet medical need. Many rare diseases begin early in life, a fact that adds urgency and complexity to the global effort to improve diagnosis, care, and research. While scientists around the world work to unlock the mysteries of these conditions, the everyday reality for patients from Lagos to London to Lima involves managing symptoms, seeking answers, and building resilience in the face of systemic silence.

Global Challenges, Shared Human Experiences

Across the globe, people living with rare diseases encounter similar barriers: long diagnostic odysseys, limited access to specialists, isolation, and stigma. In wealthier nations where genetic testing and specialised clinics exist, patients may still wait years sometimes more than a decade before receiving an

accurate diagnosis. In lower- and middle-income countries, including many parts of Africa, Asia, and Latin America, these challenges are compounded by gaps in health infrastructure, education, and public awareness. In countries like Brazil, community organisations have pushed for rare disease registries and newborn screening programs to identify conditions early, enabling earlier interventions and support structures. In Canada and Australia, advocacy networks have brought rare disease considerations into national health policy, emphasising patient engagement in research and decision-making. In Japan and South Korea, advances in genomic medicine have accelerated diagnosis for some rare conditions, though access is still unequal across regions and populations. These diverse global experiences underline a shared truth: rare diseases are not only clinical phenomena, but social, economic, and cultural realities that affect families everywhere.

The Role of International Organisations and Data in Shaping Global Action

Central to the global understanding of rare diseases are international organisations that champion research, policy, and patient rights. Groups such as the International Rare Diseases Research Consortium (IRDiRC) bring together scientists, clinicians, and funders with the goal of accelerating diagnosis and therapy development. The World Health Organization (WHO) includes rare diseases in its broader Universal Health Coverage agenda, urging countries to integrate rare disease care in national health planning and to develop data systems that capture the true burden of these conditions. The Global Genes Project and Rare Diseases International work to amplify patient voices across continents, promote global equity in research and treatment access, and provide educational resources for caregivers and healthcare professionals alike. These organisations collect and synthesise data from across the world, offering evidence that rare diseases are a significant global public health challenge, not a collection of isolated curiosities.

Lived Realities: Lessons From Diverse Countries

In the United States and parts of Europe, rare disease communities often have access to well-established patient groups, diagnostic pipelines, and, in some

cases, approved therapies through orphan drug legislation designed to incentivize pharmaceutical research. Even so, patients still face high costs, fragmented care, and psychosocial burdens that national systems struggle to address. In parts of Africa, where specialised services are limited and genetic testing infrastructure is sparse, families frequently rely on local advocacy networks and non-profit organisations to connect with information and support. In parts of Asia, traditional medical systems coexist with modern healthcare, prompting unique challenges in distinguishing rare disease presentations from cultural interpretations of illness. In Latin America, patient advocacy groups have successfully lobbied for inclusion of rare diseases in national health strategies, resulting in expanded newborn screening and better access to multidisciplinary care. These regional nuances reveal that while rare diseases are a global phenomenon, the paths toward awareness, diagnosis, and care are shaped by historical, economic, and cultural contexts.

Nigeria's Case: A Story of Everyday Realities

In Nigeria, the conversation around rare diseases often unfolds not in national health plans or registries, but in the quiet stories of families who navigate uncertainty without clear signposts. A child with unexplained symptoms, an adult whose condition defies simple diagnosis, a caregiver who becomes an expert out of necessity — these are the human faces of rare diseases in communities where the term itself might not be widely recognised. Cultural beliefs about illness, limited access to specialists, and the absence of formal genetic services can mean that many rare conditions go undiagnosed or misunderstood. The Nigerian experience does not need national prevalence numbers to be real; it is embedded in the lived struggle for answers, dignity, and compassionate care. On Rare Disease Day, these stories deserve space, not just as anecdotes, but as part of a collective call for recognition, awareness, and action.

Stigma, Culture, and the Burden of Being Different

Across every continent, rare disease patients face not only medical challenges but social ones. Visible

symptoms physical differences, developmental delays, or complex health needs can attract misunderstanding, fear, or exclusion. In many communities, people living with rare conditions may be labelled as “different,” “cursed,” or “burdened,” simply because their conditions do not fit common illness narratives. This stigma affects mental health, social opportunities, education, and economic inclusion. In countries with strong community networks and social safety systems, peer support groups often provide solace and shared understanding. In places where stigma is more pronounced and resources are limited, families may internalise shame and isolation. Rare Disease Day offers an opportunity to replace fear with understanding, to challenge myths with evidence, and to build empathy across societies.

Moving Forward, Looking Ahead: A Global Call to Action

The theme of Rare Disease Day 2026, “Moving Forward. Looking Ahead. An Event for Patients” is a powerful reminder that progress must be rooted in the real lives of people affected by rare diseases. Globally, this means investing in data systems that capture rare disease prevalence, expanding access to genetic testing, and supporting research that prioritises patients’ needs. It means shaping national health policies that recognise rare diseases as part of the broader health landscape, not as outliers or afterthoughts. It means empowering patient associations, strengthening multidisciplinary care teams, and ensuring that no one lives in the shadows of their condition. For Nigeria and countries around the world, this shared global effort is not about statistics alone it is about expanding compassion, improving quality of life, and honouring the human dignity of every individual whose journey has been shaped by a rare disease. ■

More Than Pain:

Social Burden of Neuroma-Fibrosis

The first thing many people notice is not the pain. It is the way others stare.

In a crowded clinic waiting room in Benin City, a young woman sits quietly with a scarf wrapped around her arm. Beneath the fabric is a swelling that has grown slowly over the years, twisting the shape of her forearm into something she no longer recognizes as part of herself. She has learned to keep it covered, not because it hurts all the time, but because of the way people recoil. Some whisper.

Some move seats. A few ask if it is contagious. She has heard the question so many times that she now answers before it is asked: “No, it is not an infection. It is not a curse. It is not something you can catch.” What she is living with is a condition many Nigerians have never heard of, yet quietly exists across communities neuroma-fibrosis.



An image of someone dealing with Neurofibromatosis
Credit: Getty Image

Dr. Loretta Oduware Ogboro-Okor, an Obstetrician and Gynaecology Surgeon who has encountered patients with these lesions through multidisciplinary referrals, explains that neuroma-fibrosis is an umbrella description for conditions involving abnormal growth of nerve tissue combined with fibrous tissue proliferation. In simpler terms, it is when nerves and scar-like tissue grow in a way they should not, leading to swellings, nerve compression, chronic pain, and often visible deformities of soft tissue. It can manifest as neurofibromatosis,

a genetic disorder that causes tumors to grow along nerves, or as traumatic neuroma, which develops after surgery, injury, or amputation when regenerating nerve fibers become trapped in scar tissue. Though medically benign in many cases, the social, psychological, and functional consequences are anything but mild.

The most common form is Neurofibromatosis Type 1 (NF1), also known as von Recklinghausen disease. International data from institutions such as the National Institutes of Health and the World Health Organization show that NF1 affects about 1 in every 3,000 people worldwide, making it one of the most common genetic neurological disorders. In Nigeria, precise prevalence data is scarce due to underdiagnosis and poor reporting systems, but teaching hospitals such as the University College Hospital Ibadan, Lagos University Teaching Hospital, and University of Benin Teaching Hospital have documented multiple cases over the years, often discovered only when tumors have grown large and disfiguring. NF1 typically presents with multiple cutaneous or subcutaneous nodules called neurofibromas and characteristic skin spots known as café-au-lait spots.

However, these spots pose a unique diagnostic challenge in Nigeria and across Africa. On darker skin, the light brown patches described in textbooks are not easily visible. Some researchers have described them in African patients as “café-sans-lait” or blackish-brown spots that blend into the skin tone. Because they are not obvious, early diagnosis in infants and children is frequently missed. By the time tumors begin to grow along nerves, the opportunity for early monitoring has already passed.

Another severe form is the plexiform neurofibroma. These are congenital or early developmental lesions that grow along multiple nerve branches, often described by doctors as feeling like a “bag of worms.” They can burrow deep into tissues, involving limbs, the face, or the trunk, causing massive disfigurement. International studies show

that between 5–10% of these plexiform tumors can transform into malignant peripheral nerve sheath tumors (MPNSTs), an aggressive cancer. In Nigeria, many patients present only when the tumor has already reached a size that interferes with movement, vision, or breathing, and in some cases, has undergone malignant change. Traumatic neuroma tells a different story.

It is not genetic. It is the body's attempt to heal. After surgery, injury, or limb amputation, the cut nerve tries to regenerate. When the growing nerve fibers are blocked by dense scar tissue (fibrosis), they coil into a painful knot. Patients describe the pain as electric, burning, or stabbing, triggered even by light touch. This is commonly seen in people who have had amputations due to road traffic accidents, diabetic complications, or severe infections situations not uncommon in Nigeria. What appears as a small lump at the end of a stump can be a source of relentless, life-altering pain.

Localized intraneural neurofibromas, though solitary, can cause fusiform enlargement of a nerve trunk, compressing nearby structures and producing numbness, weakness, or persistent discomfort. Without imaging like MRI or CT scans, which are not easily accessible in many parts of the country, these are often mistaken for other soft tissue tumors.

Dr. Ogboro-Okor notes that the challenges of managing neuroma-fibrosis in Nigeria go far beyond the medical textbook. Patients frequently present late, sometimes after living with the condition for decades. By then, tumors are massive, deeply seated, and intertwined with muscles, blood vessels, and vital structures. Surgical removal becomes risky, complex, and expensive. Diagnostic limitations also play a major role.

Advanced imaging required to map these tumors is unavailable in many centers, and histological diagnosis is often limited to basic staining methods without the immunohistochemical studies needed for precise classification.

There is also a scarcity of multidisciplinary teams. Ideal management requires neurosurgeons, plastic surgeons, neurologists, geneticists, pain specialists, and rehabilitation experts working together. In reality, many patients are managed in isolated departments with limited collaboration. The cost of surgery, especially reconstructive procedures after

tumor removal, is beyond the reach of most patients who pay out of pocket.

Follow-up, which is crucial because of the risk of malignant transformation, is inconsistent due to financial hardship and poor health-seeking behavior. Beyond the hospital walls lies another painful layer culture and perception. In many communities, people with visible neurofibromas are treated as outcasts.

There is an unspoken discomfort with bodies that look different. Some believe the condition is spiritual. Others think it is infectious. Children are teased. Adults are avoided. Marriages are affected. Employment opportunities shrink.

Patients speak more about stigma than about pain. They hide their limbs in wrappers, wear long sleeves in heat, and avoid social gatherings. As one patient told a clinician, “The tumor does not hurt me as much as the way people look at me.”

Management often begins with surgery. Symptomatic neurofibromas and traumatic neuromas may be excised. For large plexiform tumors, total removal is rarely possible, so surgeons perform debulking procedures to reduce size and relieve pressure. Pain management is essential, especially for traumatic neuromas, using medications or surgical resection of the affected nerve segment. Rehabilitation follows, helping patients regain function and adapt to physical changes.

Yet, treatment is only part of the story. Awareness is equally critical. Early recognition of café-au-lait spots in children, even when faint on dark skin, can prompt monitoring. Public education can reduce stigma by reinforcing a simple truth: neuroma-fibrosis is not contagious. It is not a curse. It is a medical condition involving nerve tissue and fibrous growth.

In clinics across Nigeria, people continue to arrive quietly with scarves, long sleeves, and years of silence. Their condition may be rare, but their experience of stigma, delayed care, and resilience is common. Neuroma-fibrosis is a story of nerves and scars, but also a story of society, perception, and the urgent need for earlier diagnosis, better resources, and kinder understanding for those whose bodies tell a different story. ■

Where Public Support Falls Short, Families, Foundations Step In for Children With Down Syndrome

Ruth Ugwuegbulam's journey as a mother began more than two decades ago, the day her son, Shadrach, was born. The pregnancy had been uneventful, the delivery smooth. At first, everything seemed ordinary.

"Shadrach was born 21 years ago, just like every other child," she said, remembering those first fragile days. He had a healthy weight, and she went home from the hospital relieved and exhausted, thinking life was simply beginning as expected.

The first indication that something was different came several months later when Shadrach struggled to control his head at about four months old. Concerned, her brother-in-law, a consultant in trauma and orthopaedics at the University of Benin, recommended further medical examinations. During visits to the University of Benin Teaching Hospital, doctors carried out several tests and asked detailed questions about the baby's birth and early health.

"That was the first time in the University of Benin Teaching Hospital that I heard the word Down Syndrome," she recalled.



Down Syndrome, a genetic condition caused by an extra copy of chromosome 21, affects approximately **1 in every 700** live births globally. In Nigeria, estimates suggest that around 25,000 to 30,000 children are born with Down Syndrome

every year, though official data remains limited and many cases go undiagnosed. Children with the condition experience developmental delays, congenital heart defects, and increased vulnerability to infections.

Following the diagnosis, Shadrach was referred for occupational therapy at Yaba Psychiatric Hospital. The therapy was designed to help him achieve developmental milestones such as walking, standing and other motor skills that had been delayed.

As he grew older, new complications surfaced. At one point, doctors discovered a cataract in his left eye. He was referred to the Eye Clinic at the Lagos University Teaching Hospital, where he eventually underwent surgery to replace the damaged lens with an artificial one.

But the hospital visits themselves became a test of endurance.

Simple procedures were not always simple. During eye examinations, for instance, Shadrach could not read the letters or numbers on the screen. Doctors had to find other ways to assess him, adjusting their methods in a system that was not built with children like him in mind.

Even arriving at the hospital before dawn did not mean they would be attended to early. Ruth would get there as early as 6 a.m., only to wait for hours. Care often depended on a specific staff member familiar with his case file. If that person was unavailable, they waited.

"It was frustrating," she said.

But the hospital visits were only part of the strain.

At home and within the extended family, Ruth faced a different kind of challenge. Shadrach's delayed development drew attention. Some relatives and neighbours mocked him.

“When he was crawling, he moved on his stomach on the floor,” she said. “They called him a ‘water snake.’”

He was slow to speak, and people made assumptions. Some thought he could not hear. Others believed he could not understand.

At the time, Ruth was largely raising him on her own. Her husband was abroad.

“The father was away, so I was the one on the ground,” she said.

She said it took time to come to terms with the diagnosis and the reactions around her. Over time, she stopped trying to explain her son to everyone.

“It took me time,” she said. “But I told myself that whatever people call my child does not matter. What matters is that I have accepted him.”

Years later, the family had to adjust again when Shedrach began to lose his vision. At home, they started teaching him how to move around safely, memorising the layout of rooms, doorways and stairs.

“These are some of the daily challenges we face,” she said.

Ruth’s experience is not isolated.

Muyiwa Majekodunmi, a jazz musician, has spent more than three decades raising his son, Olumakinwa, known as BJ, who was born with Down syndrome in 1988.



Jazz Musician Muyiwa Ma; jekkodunmi with his son Olumakinwa Majekodunmi. Photo Credit: Femi Odugbemi

“My son was born when there was no internet and very little awareness,” he said.

He described those early years as uncertain. Information was limited, and many schools were not prepared to admit children with developmental disabilities. Families, he said, often had to figure things out on their own.

Despite that, his son found a path through music. BJ is now a percussionist and has performed publicly in Nigeria. His father said music helped him develop confidence and communication.

Like many families, the Majekodunmis also had to navigate the cost of care. Therapy, schooling and medical needs required steady spending over the years.

He said one concern has never gone away.

“You worry about what will happen to them if you die before them,” he said.

Majekodunmi recalled instances where other parents objected to their children sharing classrooms with his son. Even so, he said the experience has reshaped how he measures progress.

“When you see your child grow and express their talents despite the odds,” he said, “you realise every little achievement matters.”

For Iduma Egwu, those milestones have looked different, but no less significant.

His daughter struggled with early development, including holding her head upright. For months, the family moved between hospitals without clear answers. It was only after a visit to a teaching hospital in Lagos that doctors confirmed the diagnosis.

“Down Syndrome has different levels,” he said. “Some children have more delays than others.”

Over time, she learned basic routines such as washing her clothes and managing aspects of her personal hygiene. But she still depends on the family for many daily tasks.

Schooling has been one of the most difficult decisions.

Egwu said they first enrolled her in a special needs school in Ikeja. After relocating, they tried a mainstream school, but she struggled to keep up.

“She was mostly just playing,” he said.

The family later moved her to a specialised school run by the Down Syndrome Foundation Nigeria, where she could learn alongside children with similar needs.

The cost, he said, is substantial. Each term, the family spends close to half a million naira on school fees and related expenses.

Like many parents, he is also planning for the future.

“I have an insurance where I put my daughter as my benefactor,” he said, explaining that it is meant to provide for her if anything happens to him.

For Honourable Olayinka Abayomi Ijal, the experience has taken a slightly different shape.

His 26-year-old son, Moyosoreoluwa, can manage many daily activities independently, but still needs occasional guidance.

“He can function properly without being supervised,” he said, “but sometimes needs a little more guidance.”

Finding the right school was one of the family’s biggest challenges. Many institutions were not equipped to support children with developmental disabilities, leaving families to rely on private options or specialised centres.

He said misconceptions about the condition remain common, with some people assuming children with Down syndrome cannot learn or live independently.

In another family, care has stretched across generations.

Olubunmi Omokorede Sosina said responsibility for her sister, who is 42 and lives with Down Syndrome, shifted after their mother’s death.

“When my mother was alive, she handled most of it,” she said. “After her death, it became something all of us had to take on.”

Now, the siblings share caregiving responsibilities, helping with daily routines and monitoring her well-being.

Sosina said they have not faced severe discrimination, but misunderstandings still arise. When they do, the family tries to explain.

“We tell them that Down Syndrome is not contagious,” she said.

In some cases, families have found more structured support.

Down Syndrome Foundation Nigeria: Providing Care Amid Persistent Stigma

One of the organisations filling that gap is the Down Syndrome Foundation Nigeria, established in 2001. The foundation works with children and adults living with Down syndrome, offering education, therapy and support for families navigating the condition.

For parents like Ruth and Egwu, the foundation has provided an alternative to a system where support is often inconsistent. Children are grouped with others facing similar challenges, and the focus is on routine, social interaction and basic life skills.

Innocent Okuru, the foundation’s Deputy Head of Education and Project Manager, said many families arrive at the centre after struggling to find answers elsewhere.



Down Syndrome Foundation Nigeria (DSFN) Inter-house sport activities. Photo Credit: DSFN

“From my experience, one of the major challenges is stigmatisation and non-acceptance,” he said.

He said the foundation’s first point of intervention is often counselling, especially for parents encountering the diagnosis for the first time. From there, children are introduced to structured learning and therapy, depending on their needs.

Medical conditions are also common. Okuru said some children require specialised care, including corrective surgery for heart defects. Through a program known as the Save a Life Project, the foundation helps connect families to treatment, sometimes outside the country.

But access remains uneven.

Okuru said many families cannot afford the cost of care, particularly when it involves travel or long-term therapy. Even within Nigeria, he said, services are limited and not always tailored to children with developmental disabilities.

“There are still gaps,” he said. “Awareness is growing, but it is still a work in progress.”

He also pointed to challenges within the healthcare system itself. Some medical professionals, he said, have limited experience working with people with Down Syndrome, which can affect communication and care during hospital visits.

“Some health professionals do not have the patience to listen,” he said.

The foundation’s work shows that where support exists, it is often driven by private organisations or families themselves, rather than a coordinated public system.

Medical Insights: Down Syndrome and Early Intervention

Professor Bede Ibe, an emeritus professor of paediatrics, said early diagnosis is possible in some cases, even before birth. With experienced radiologists and the right equipment, signs can be

detected during pregnancy. Further tests, including analysis of amniotic fluid, can confirm the condition.

But those options are not widely available.

In many cases, he said, diagnosis comes later, after parents have already noticed delays. That pattern is reflected in the experiences of families like Ruth and Egwu, who moved between hospitals before getting clear answers.

From a clinical standpoint, delayed development is expected.

“Development is usually slow,” he said, explaining that milestones such as sitting or walking often come later than they would for other children.

He also pointed to other conditions that often appear alongside Down syndrome, particularly congenital heart defects. These can affect growth and require specialised care, sometimes beyond what is available locally.

Where support exists, timing matters.

Early intervention, including physiotherapy and speech support, can improve long-term outcomes. But access is uneven, and many families start late or cannot sustain the cost of care.

That gap shows up not just in hospitals, but in everyday life. Parents become the primary support system, managing therapy, schooling and medical care over many years.

“There are no strong social welfare structures,” he said.

The result is a system where outcomes often depend on what families can afford or find on their own. Organisations like the Down Syndrome Foundation Nigeria provide some structure, but they operate within the same limits of cost, awareness and reach. ■



Angeline OCHIENG

in Nation Media Group | Kenya



Angeline Ochieng is a print journalist with over four years' experience working for Nation Media Group (NMG) based in Kisumu, Kenya. Her journalism focuses on health, gender and human interest stories. Driven by a commitment to give a voice to the voiceless and act as a public watchdog, she is passionate about Solution Journalism (SOJO) that inspires hope and fosters positive

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System and governance failure: Why Kenyan mothers are still dying in childbirth



A mother is diagnosed with PPH if she loses over 500ml of blood after vaginal birth or 1,000ml of blood during a caesarean section.

What you need to know:

- With Kenya's maternal mortality rate currently standing at 355 deaths per 100,000 live births, health experts are united in their verdict, this is unacceptably high for a nation that possesses both the infrastructure and the expertise to do far better.

Across the country, mothers are dying; not because their doctors lack knowledge or their nurses lack training, but because the systems meant to save them are failing. Health workers are too few. Referrals take too long. Blood is unavailable when it matters most. And when a mother's life hangs in the balance, the right decisions are often not made fast enough.

These were the hard truths laid bare this week at the Kenya Obstetrical and Gynaecological Society (KOGS) annual conference in Mombasa, where the country's top doctors, gynaecologists, and health leaders gathered to confront a crisis that is both persistent and preventable.

What emerged from the discussions was sobering: the problems driving maternal and newborn deaths in Kenya are not new. They have been documented, discussed, and decried for years. Yet year after year,

they remain the leading causes of death; pointing not to a failure of clinical knowledge, but to a deeper, systemic failure that continues to cost lives.

Health Director General Dr Patrick Amoth, a keynote speaker at the conference, did not mince words about where the failures begin. A mother's chance of surviving childbirth, he reminded delegates, is shaped long before she enters a delivery room; by decisions made from the moment she conceives, throughout her pregnancy, and right up to the final moments before delivery.

"Lack of knowledge, poor financing, delays by a mother to access a health facility, and delays by a health worker to attend to a client, all have a heavy impact on maternal health outcomes," Dr Amoth said.

The message was stark: when a mother delays seeking care, or when she arrives at a facility only to wait too long to be seen, the consequences can be fatal.

With Kenya's maternal mortality rate currently standing at 355 deaths per 100,000 live births, health experts are united in their verdict, this is unacceptably high for a nation that possesses both the infrastructure and the expertise to do far better.

The national government has pledged to work more closely with county governments to close the governance and accountability gaps that continue to cost mothers their lives. But pledges, as many in the room knew, are not enough.

What is needed is action: faster referrals, better-stocked facilities, more health workers, and a system that responds decisively when every second counts. According to the Ministry of Health, postpartum haemorrhage (excessive bleeding after childbirth) remains one of the leading causes of maternal deaths in Kenya.

Dr Amoth noted that while Kenyans continue to donate blood in significant numbers across the country, weak infrastructure often makes it nearly impossible to get the life-saving commodity to mothers who need it most. The gaps, he explained, span the entire supply chain; from screening and transportation to timely delivery at the bedside.

“For a very long time, Samburu County did not have a blood transfusion centre, forcing mothers and health workers to travel all the way to Nakuru—a distance of more than 200 kilometres—to access blood,” Dr Amoth said. “Imagine an expert having to travel from Maralal to Nyahururu just to collect blood and rush back to save a mother’s life.”

Despite these logistical hurdles, he pointed to progress in public willingness to donate. “Last year, we collected 101,000 pints of blood, surpassing our target of 80,000. This shows that Kenyans still have the zeal to donate. We are now focused on strengthening the infrastructure to ensure a smooth flow of services.”

Also present at the event was the chairperson of the County Executive Committee Members (CECMs) for Health, Dr Gregory Ganda, who said that every maternal death often points at a system failure.

According to Dr Ganda, preventable maternal and neonatal deaths is always a governance failure, before it becomes a clinical failure.

“Somewhere in Kenya, a woman is going to die today while trying to give birth and sadly, one of us in this room could have saved her. The question is not the skill, but whether our system allows the skill to reach her on time,” said Dr Ganda.

Dr Ganda, also the Kisumu County CECM for Health, said while a woman may have made it to the hospital for a skilled delivery, a lot of factors may affect the pregnancy outcomes.

While there might be a health worker to attend to the client, lack of knowledge of what to do to stop bleeding due to a lack of mentorship still stands as a barrier.

“The system of blood donation, referral, lack of equipment and staffing may fail the mother. Imagine an instance where an expert wants to conduct an operation but the drips are not working, a procedure is being delayed because of a specialist who rarely reports to work and a failed oversight system that never holds the involved staff accountable,” said Dr Ganda.

He added: “Over time, all of us in this room have learnt that when a medic delivers a healthy baby, he or she is not the only hero. So much goes on, starting from the lowest level of the nurse who did the antenatal care clinic,”

Dr Ganda, however, lauded the role of inter-county Maternal and Perinatal Deaths Surveillance and Response (MPDSR), a peer-to-peer learning and accountable system connecting frontline health workers in all the 47 counties, the national referral and county and national leadership in reducing the preventable deaths.

With the help of the system, he said, 37 counties are currently running weekly reviews of maternal child health, sharing dashboards for comparisons, shared mentorship engagements and hosting of national webinars based on crosscutting themes.

While sharing a presentation on the impacts of the intercounty MPDSR, Dr Grace Wanjiku, a consultant obstetrician and gynaecologist in Murang’a, said the county’s fifth mortality rate currently stands at 133 per 100,000 live births.

In 2025, she said, out of the 18,000 deliveries in the county, only 26 maternal mortalities were reported.

“Reliable commodity availability is one of the key reasons behind the positive outcomes,” said Dr Wanjiku.

Dr Ganda argued that as much as the health services are devolved, patients and referral pathways are not, thus the reason behind women and newborns traversing counties in search of care.

He, however, cautioned that MPDSR remains incomplete unless it examines the full patient journey across the counties and facilities involved in care.

“We want to learn from each other through the system so that the mistakes made in one county are not replicated,” he said.

Dr Amoth said in order to improve maternal and child outcomes, the national government is going to work closely with the county government in capacity building and training of health workers, commodity supply, equipment and workforce availability to ensure a smooth flow of services.

The government, he said, is also working to address governance and accountability gaps through setting up and strengthening maternal and perinatal death surveillance responses.

“We have also decided to come up with a rapid result initiative to address the increasing burden of maternal and newborn mortality while prioritising the 26 counties which contribute more than 60 per cent of maternal and newborn deaths across the country,” he said.

Dr Amoth further disclosed that, with support from partners, the government is revamping the regional blood transfusion centre in Garissa to serve the neighbouring counties of Tana River and Wajir, areas where geographical isolation has long delayed access to emergency blood supplies.

He also highlighted a critical shift in how health data is collected and used. In the past, the government relied on the demographic health survey—conducted every five years—to inform decision-making, a lag that often derailed timely interventions in maternal and child health. The introduction of a digital health system in facilities is now changing that.

“One of the challenges with waiting five years for data is recall bias: experts often struggle to remember the exact causes of death,” Dr Amoth explained.

“As we speak, level four hospitals and above are already transmitting real-time data, thanks to the new system. We are now working to digitise level two and three facilities.”

During the conference, the government also launched the first Basic Obstetric Protocol 2026, a landmark resource designed to offer evidence-based, practical guidance to healthcare professionals at every level of service delivery.

According to KOGS President Dr Kireki Omanwa, the protocols will address seven priority conditions in maternal health: obstructed labour, sepsis, abortion care, anaemia, antepartum and postpartum haemorrhage, and hypertensive disorders.

“These pocket-friendly booklets will be distributed to experts across the country in a concerted effort to reduce maternal and child deaths,” Dr Omanwa said. ■

Blamed for genes: How a sickle cell diagnosis drove a young mother out of marriage



Every year, roughly 14,000 people in Kenya are born with sickle cell disease—a hereditary blood disorder that reshapes soft, flexible red blood cells into rigid, crescent-like forms.

Mercy* steadies her one-year-old son as he stirs on a mat where he was having his afternoon nap. She moves through familiar motions; water for a bath, clothes to wash—the endless rhythm of a Mother’s Day.

But there is a fracture running through these ordinary moments. For the past seven months, she has done this alone in her brother’s three-roomed house in Kisumu County, far away from the home she shared with her husband of over eight years.

Mercy does not speak about that home easily. And when she does, tears well up in her eyes. “I left my matrimonial home earlier this year following continuous emotional abuse by my husband,” she says, her eyes betraying her pain.

The abuse did not begin because of infidelity, money or the ordinary conflicts that afflict fracture marriages. It began with a diagnosis.

In October 2024, Mercy’s newborn son was diagnosed with sickle cell disease, an inherited

blood disorder caused by a mutation in the gene that produces abnormal sickle-shaped red blood cells. These cells block blood flow and cause severe health complications.

The diagnosis should have brought the couple together. However, it tore them apart. “My husband said he has never heard of the disease in his lineage, and, therefore, I am responsible for our baby’s condition,” Mercy recalls.

“He also refused to pay the medical bills and opted to visit a herbalist in search of a cure.”

The misunderstanding on how inherited diseases work has cost Mercy her marriage, livelihood, and nearly everything that she had built. Eight years ago, in 2017, when Mercy moved in with her spouse, they were hopeful of a good future together. A few months into the marriage, she conceived their first child. However, after birth, the child suffered ill health from the outset. Hospital visits became routine.

In 2019, a paediatrician advised the couple to get the child tested. The test results revealed that he had sickle cell disease. Their firstborn died of the illness.

"None of us thought about getting screened for the blood disorder," Mercy says. "Neither of us had also come across the disease in our families before."

That couple would never know if their child might have lived had they understood the disease sooner. What they knew was loss, and they moved on. Six years later, Mercy conceived again.

This time, in October 2024, after delivering the baby at Kisumu County Referral Hospital, she followed a health worker's advice and had her newborn screened. Three months later, the hospital called her to pick the results.

"I was scared of breaking the news to my husband. I requested the health worker inform him on the phone," she says, adding that her husband did not answer his phone.

That evening, as they prepared for bed, Mercy finally told her husband about the results. She remembers him blaming her. Mercy says that he was emphatic that no one in his family had ever suffered from sickle cell disease. He says that I am the one who brought the disease, she says. Read: Beyond survival: Ensuring adolescents with sickle cell disease can thrive

What followed was months of escalating cruelty. Mercy says he called her names. She says that he even threatened her when she asked for help to buy their son's medication. When she asked him to go for screening to understand how their son had inherited the disease, he refused.

Instead, he blamed her daily for bringing what he called a "killer disease" to the family. He swore he would never have another child with her.

Mercy had a choice: stay and endure, or leave and try to survive alone. By May 2025, the emotional abuse had become unbearable. She walked out.

Approximately 6,000 infants are born with sickle cell disease every year in Kenya. According to Ministry of Health statistics, 80 per cent of them die before their fifth birthday. Kisumu, one of the country's high-burden areas for the disease,

records 3,000 children born with the blood disorder annually. These are not rare cases. Yet the violence that follows a diagnosis is rarely discussed.

Ms Lily Webala, the founder of Zuwi Afya, a community-based youth-led health organisation, says sickle cell cases are common.

"Cases of women exposed to gender-based violence after giving birth to children with genetically inherited diseases, especially sickle cell, remain common," she says.

Her organisation supports sickle cell disease warriors across the country who face physical or emotional violence from partners who blame them for their children's illness. In some cases, the violence comes not just from husbands but from in-laws who put pressure on the men to remarry and bear "healthy children".

"We have encountered one case of a man who was pushed out of his home by his wife who believes he is the cause of the illness," Ms Webala notes.

But women bear the heaviest burden of the blame. And many, like

Mercy, find themselves leaving their marriages. Others stay but they become the sole caregivers. They carry the emotional and financial burden alone. Some women end up enduring violence in silence, with the hope that things will change.

Ms Webala says that when women finally understand the biology behind the disease that sickle cell is inherited from both parents, not one-the blame dissolves. However, that requires access to information. Last month, the organisation launched a campaign to create awareness about sickle cell disease. People were educated on how the disease is inherited and how it is managed. The campaign includes free screening services for couples and children, as well as counselling for parents who are carriers and those whose children have been diagnosed.

"Our major message, as we mark the 16 Days of Activism against gender-based violence is to make the masses understand that the disease is not a fault of one of the parents, but it is inherited from both," Ms Webala says.

For Mercy, that understanding came too late to save her marriage. After she left, she lost her teaching job at a private school. She was told that she had become unreliable. Before the separation, Mercy was frequently absent from work because she needed to take her son to hospital.

"Before I was sacked, I was not able to report to work regularly because I had to take care of my ailing son," she says. "I am his sole caregiver. As time went by, my employer relieved me of my duties, saying that I should go and take care of my child."

The hospital visits have consumed her savings. The Social Health Authority covers some of her son's medication, but many drugs are rarely available in public health facilities. "A number of drugs are rarely available in the public facilities, forcing me to pay out of my pockets," Mercy says.

Seven months into her separation, Mercy now depends on her family and friends for support to take care of her son. ■

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Li Wei SOON

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Li Wei Soon is a bilingual journalist at the Malaysian National News Agency (BERNAMA), covering climate change, biodiversity, planetary health, human rights, and culture. Her recent work includes reporting on how climate change influences rat populations and raises Leptospirosis risks, as well as how extreme heat affects the physical and mental health of low-cost housing residents. In June 2022, she produced

an investigative report on illegal logging in Kelantan, highlighting its devastating impact on biodiversity and Indigenous communities. The project, supported by Cent-GPS and the U.S. Embassy in Kuala Lumpur, gained wide attention after her exclusive story on BERNAMA was picked up by other media. Li Wei writes in English, Bahasa Malaysia, and Simplified Chinese.

New Policy On Rare Diseases Brings Hope To Patients, Carers

Nine years ago, Sivasangaran Kumaran and his wife Prema Latha Letchimun were overjoyed to welcome their second child, Swathi Nisha Nair, who was delivered naturally.

Nothing seemed unusual until six months later, when everything changed.

The parents noticed that Swathi had developed a prolonged bout of pneumonia, prompting doctors to perform an X-ray.

The results were alarming: her heart was enlarged and an echocardiogram showed that it was functioning at only 25 percent capacity.

Soon after, Swathi was diagnosed with Pompe disease, a rare genetic disorder that weakens the heart and muscles. The disease, affecting only one in 40,000 people worldwide, is linked to a deficiency of an enzyme called acid alpha-glucosidase (GAA), which breaks down glycogen, a complex sugar.



Swathi was diagnosed with Pompe disease, a rare genetic disorder that weakens the heart and muscles, during 6 months old (Pix credit to: Sivasangaran Kumaran)

“There are two main forms of Pompe disease – infantile-onset and late-onset. Swathi has the infantile variant, the more severe form that affects the heart and can be fatal within the first year or two of life if untreated,” Sivasangaran, 46, who works for a multinational company, told Bernama recently.

FUNDING

He was told that the only available treatment for Swathi is enzyme replacement therapy (ERT), one of the world’s most expensive life-saving treatments. He also found out that his daughter’s treatment would cost about RM500,000 per year.

However, at that time, the annual government allocation for rare disease treatments at Hospital Kuala Lumpur (HKL) – which acts as the national referral centre for genetic, including rare, diseases – had already been exhausted, forcing the family to pay a huge sum out of pocket to begin treatment immediately.

Sivasangaran also approached the Malaysia Lysosomal Disease Association to help fund Swathi's treatment temporarily and he repaid the NGO in instalments over the following months. The medication was just enough to keep his daughter alive until government funding resumed the following year.

“We were very lucky that the ERT worked for Swathi. Not all children respond to the drug (used in ERT), and some families lose their children within one or two years despite treatment.

“On top of this, she gets physiotherapy, occupational therapy, aqua therapy and speech therapy a few times a week at Seremban to help her reach developmental milestones,” he said.

“We have to get up at 4 am once a week to travel to HKL so Swathi can undergo ERT... my wife had to give up her job as a medical assistant in 2017 to take care of our daughter.”

Though Swathi is still non-verbal, her heart and liver functions have improved. She is also now able to communicate through gestures and alternative communication tools.

Swathi is now nine years old and is at Level 3 of the Special Education Integration Programme, where students with special needs are placed in special classes within mainstream primary and secondary schools.

According to her father, Swathi can understand letters, numbers, colours and shapes. She can read and write, not perfectly, but clearly.

“Although her mobility is limited, she is very smart... she can walk independently for short distances with specialised shoes, but a wheelchair may eventually be necessary,” he said, adding his elder son Harrish, 17, acts as a supportive “third parent” to Swathi.

In light of World Rare Disease Day on Feb 28, Sivasangaran hopes that Swathi's story can inform Malaysians about the existence of Pompe disease and rare diseases as a whole.



Swathi is now nine years old and is at Level 3 of the Special Education Integration Programme, where students with special needs are placed in special classes within mainstream primary and secondary schools. (Pix credit to Sivasangaran Kumaran)

POMPE DISEASE DIAGNOSIS

As of 2025, a total of 28 patients with Pompe disease have been referred to HKL. A majority of them were already in the advanced stage of the disease and were too late for life-saving medical intervention. Currently, only seven patients are still surviving.

Commenting on the disorder, HKL clinical genetics consultant and head of the genetics department Dr Ngu Lock Hock said Pompe disease is a rare,

inherited neuromuscular disorder caused by a deficiency of the GAA enzyme, which leads to a harmful buildup of glycogen in muscle cells, causing progressive muscle weakness and damage.

He said this rare disease is unique among neuromuscular disorders because it is both a lysosomal storage disease and a metabolic myopathy, primarily affecting the skeletal, heart and respiratory muscles.

“Pompe disease is difficult to diagnose early because of its non-specific early symptoms, which can easily be mistaken for more common, less severe or infectious conditions,” he said, adding that muscle weakness or breathing difficulties in Pompe disease are frequently misdiagnosed because they overlap with more common neuromuscular or respiratory conditions.



Hospital Kuala Lumpur's clinical genetics consultant and head of the genetics department, Dr Ngu Lock Hock

“Because Pompe disease is rare, physicians often explore more common diseases first, leading to a diagnostic delay that can span over 10 years in late-onset cases,” he told Bernama.

Elaborating on the two forms of Pompe disease, Dr Ngu said infantile-onset Pompe disease (IOPD) is the most severe and usually presents within the first two to four months of life, with the babies appearing ‘floppy’ due to poor muscle tone and lack of head control.

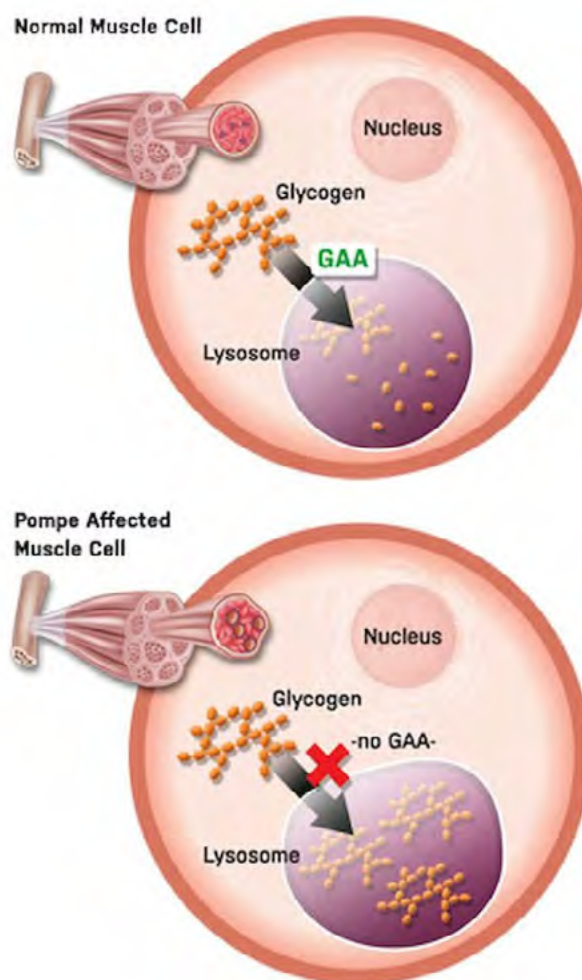
“They often experience profound muscle weakness, feeding difficulties and rapid enlargement of the heart, which can lead to heart failure if untreated,” he said.

He added that respiratory distress and recurrent infections are also common in IOPD, and some infants may have an enlarged liver or tongue.

“In contrast, late-onset Pompe disease (LOPD) can present from late childhood to adulthood, and symptoms are often more gradual and may initially be overlooked.

“Patients usually develop progressive muscle weakness, especially in the legs, hips and trunk, leading to difficulties climbing stairs, rising from a seated position or walking,” he said.

Breakdown of glycogen to glucose in the lysosome is impaired in Pompe disease.



Graphic shows how the breakdown of glycogen in the Lysosome is impaired in individuals with Pompe disease (Pix courtesy of Dr Ngu Lock Hock)

TREATMENT AND CHALLENGES

Dr Ngu said currently, ERT is the available medical treatment for Pompe disease that supplies, or “replaces”, the missing or deficient enzyme in patients. Alglucosidase was the first marketed ERT product for this rare condition. The treatment has now transitioned toward a newer, more effective next-generation ERT product called avalglucosidase.

alfa, which was approved by Malaysia in May 2024 for long-term use. The drug is given bi-weekly, via intravenous infusions.

“Due to the rarity of the disease, experts and treatment centres are limited to major public hospitals such as HKL and Universiti Malaya Medical Centre (UMMC),” he said.

This year, the government allocated RM42 million to HKL and a few other state hospitals for the treatment of rare diseases, including RM30 million for ERT.

Commenting on the shortage of rare disease specialists in Malaysia, Dr Ngu said it is a critical issue, with only about 15 clinical geneticists and a handful of genetic counsellors serving a population with an estimated 10,000 to over a million people affected by rare diseases.

“This limited expertise, concentrated mainly in the Klang Valley, causes significant gaps in the healthcare system, leading to long diagnostic delays (meaning patients often face a ‘diagnostic odyssey’, spending years seeking an accurate diagnosis) and inequitable access to treatment,” he said.

Dr Ngu added that since more than 80 percent of rare diseases, including Pompe, are genetic in origin, clinical geneticists and genetic counsellors play a crucial role in explaining inherited conditions and advising families on reproductive options and family planning.

“Incorporating genomic sequencing into newborn blood screening could help detect Pompe and other rare diseases, as well as hundreds of additional childhood conditions at a much earlier stage, enabling earlier diagnosis and timely treatment,” he said.

UMMC consultant clinical geneticist Prof Dr Thong Meow Keong said expanding newborn screening to include inherited metabolic conditions and other genetic disorders could significantly reduce long-term morbidity and economic burden.

“Early detection of treatable metabolic and genetic disorders improves survival and reduces downstream healthcare costs,” he said.



Prof Dr Thong Meow Keong

He added that awareness and proper counselling play a critical role in improving uptake of genetic services.

“Once awareness is established and genetic counselling is provided, the uptake usually improves. This is why public education, particularly among high-risk groups, is essential,” said Dr Thong, who is also Malaysian Rare Disorders Society (MRDS) advisor.

POLICY MATTERS

Meanwhile, commenting on the National Policy for Rare Diseases (NPRD) launched by the Ministry of Health on Feb 4, Dr Thong praised the initiative and said the government must now invest in specialised training pathways, fellowships and regional rotations to expand expertise in clinical genetics, metabolic medicine, dietetics and genetic counselling.

“Without the right talent pool, policy ambitions cannot be translated into clinical reality,” he said.

The NPRD is a reform initiative aimed at strengthening the country’s healthcare ecosystem in addressing rare diseases in a more structured and sustainable manner. Health Minister Datuk Seri Dr Dzulkefly Ahmad said the policy reflects the government’s commitment to ensuring that rare diseases are no longer regarded as a marginal issue, but are instead elevated as a mainstream agenda.

Dr Thong said sustaining progress in Malaysia’s rare disease landscape requires structural reforms across the five key domains outlined in the NPRD.

“To ensure long-term impact, we need legislative and policy consolidation such as a dedicated Rare Disease Act, similar to those implemented in Japan

and Taiwan, that would provide stronger rights-based access to treatment, continuity in financing and clearer multi-agency accountability,” he said.



The National Policy for Rare Diseases (NPRD) launched by Health Minister Datuk Seri Dr Dzulkefly Ahmad (middle) on Feb 4

A comprehensive national funding mechanism is equally critical with a ring-fenced rare disease fund, supported through blended financing models such as government allocations, social insurance and public-private partnerships, he said, adding that this would help ensure ethical, equitable and sustainable access to high-cost therapies.

“In fact, it is important to build a robust data infrastructure as a unified national rare disease registry integrated with genomic sequencing platforms to generate high-quality epidemiological data, which is essential for precision medicine and evidence-based policymaking,” he added.

RAY OF HOPE

For Sivasangaran and other parents and caregivers of children with rare diseases who had long been fighting for patients’ rights, the NPRD is like a ray of hope, whether in accessing treatment, education, employment or recognition in society.

“Many of us caregivers are forced to bear the extremely high medical costs, and a significant number of families affected by rare diseases fall within the B40 income category.

“However, we are thankful that this policy has finally been introduced to ensure long-term governance, sustainable funding and cross-ministry coordination.

“We hope that under this new policy, more hospitals and research institutions will be involved in the research and development of rare diseases, so patients can receive treatment closer to home without having to travel far,” he said.



The Family potret of Swathi Nisha Nair with her father Sivasangaran Kumaran (right), mother Prema Latha Letchimun (middle) and eldest brother Harrish (left) (Pix credit to Sivasangaran Kumaran)

Meanwhile, MRDS president Nadiah Hanim Abdul Latif said from a grassroots perspective, the NPRD must look beyond medical treatment in addressing rare diseases in Malaysia.

“I definitely echo the importance of (genetic and newborn) screening. We have seen how life-changing it can be, especially for families with hereditary rare diseases. It allows families to make informed decisions about whether they wish to have another child

“However, we must also not forget that 30 per cent of rare diseases happen/ manifest in adolescence or adulthood. So, timely and accurate diagnosis across the life course is key,” she said.

She also stressed that prioritising interventions requires focusing resources where impact can be maximised.

“We need to strengthen diagnostic capacity, expand targeted screening, negotiate for affordable medicines, and most importantly, provide social support to prevent catastrophic health spending.

“The care pathway after diagnosis is equally important — we must invest in how patients are supported once they enter the system,” she said.



MRDS President, Nadiyah Hanim Abdul Latif (pix credit to Motherhood Malaysia)

She added that ensuring rare disease patients live with dignity should not rest solely with the Ministry of Health, but must also involve other agencies.

“Inclusive education, for example, falls under the Ministry of Education. Youth with rare diseases who are capable of pursuing higher education must not be denied that opportunity, and we have recently seen reports of persons with disabilities being blocked from educational pathways, which is deeply concerning.

“Workplace discrimination and economic vulnerability should be addressed. Under the principle of equal work for equal pay, individuals with visible rare conditions or those who require regular hospital visits, such as for infusions, should not be the first to lose their jobs during voluntary or mutual separation schemes. Work must come with dignity,” she said. ■



Simon

SPICHAK

in Being Patient | Canada



Simon Spichak is a journalist specializing in science, health, and technology. He regularly covers Alzheimer's and dementia for Being Patient. His work has appeared in the New York Times, MIT Technology Review, The Scientific Observer, and more. Simon holds a Master's in neuroscience from University College

Cork, where he researched interactions between gut bacteria and the brain. Beyond journalism, he co-founded Resolve, a virtual therapy clinic providing affordable mental health support in Ontario. In his free time, Simon enjoys bouldering, coffee, and travel.

The Race to Treat Genetic FTD: New Therapies Target the GRN Mutation



Mutations in the GRN gene are the most common genetic cause of frontotemporal dementia. Experimental pills and gene therapies are vying to be the first to prevent and slow the disease.

Heidi Hall suspected something was off when her mother, a nurse, began having language difficulties. She could no longer spell her own last name and had difficulty finding the right words to make a restaurant reservation over the phone.

Her grandmother, at the same time, was experiencing similar problems. A year later, they learned the diagnosis for both of them: **frontotemporal dementia (FTD)** caused by a mutation in the GRN gene.

Hall described the disease as being in a “prison in their own bodies,” where both her mother and grandmother could see and hear the world but couldn’t express their thoughts.

FTD, also sometimes called frontotemporal degeneration, is an umbrella term for several rare

forms of dementia affecting 60,000 Americans. Unlike Alzheimer’s, where the first symptoms are often memory-related, people with FTD may experience changes in language abilities, personality, thinking, and behavior.

Around 20 percent of FTD cases are caused by mutations in a single gene, with a **half of those cases** caused by the GRN gene. Inheriting one copy of that mutated gene guarantees the disease. By age 60, people with the gene have a **60 percent chance** of showing symptoms. By 75, that goes up to 95 percent.

The gene mutation lowers levels of a protein called progranulin, which is key for clearing cellular waste, including misfolded proteins like **TDP-43**. When aggregates of abnormal TDP-43 accumulate, neurodegeneration starts.

“This long-term reduction of progranulin levels is exacerbating a lot of that failure of proper clearance of those damaged proteins,” Penny Dacks, chief science officer of the Association for Frontotemporal Degeneration, told Being Patient.

Many drug companies are working to develop treatments for people living with the GRN gene to slow or prevent the disease altogether.

What makes GRN an appealing target?

With so many forms of FTD, characterized by a variety of aggregated proteins, including TDP-43, tau, and FUS, and with a lack of reliable biomarkers, researchers have set their sites on targeting GRN.

In some genetic diseases, a mutation might cause the formation of toxic or misfolded proteins. Treating them might require fixing these misfolded proteins or preventing them from wreaking havoc. People with the GRN mutation have, on average, 50 percent less progranulin.

“That leads to a really straightforward therapeutic intervention to increase progranulin levels,” Laura Mitic, president of the Bluefield Project, a non-profit supporting the development of GRN-targeted therapies for FTD. Genetic testing also allows researchers to easily spot people with the GRN gene that are likely to benefit from raising progranulin levels.

The other advantage of GRN is cross-correction. Scientists don’t need to repair every single cell so that it produces progranulin, said Mitic. The progranulin from one cell could be released and spread to other parts of the brain as well.

To make it easier for researchers to find families like hers that carry the gene for trials, Wanda Smith formed CureGRN in March 2025. “We had this richness of investigative clinical trials, and I knew families were out there,” Smith told Being Patient. “They just weren’t connected.”

Raising progranulin levels to treat FTD

Companies are taking different approaches to treating people with the GRN gene.

Several companies are developing therapies that boost the levels of progranulin. Denali Therapeutics and Takeda are in the [early stages](#) of testing their synthetic version of progranulin, provided intravenously, that crosses into the brain. [Passage Bio](#), [Aviadobio](#), and [Eli Lilly](#) are testing a gene therapy approach that delivers a healthy version of the GRN gene into the brain via a virus.

Other companies, like Versper Bio, are developing drugs that prevent progranulin from breaking down. It will test its experimental pill in a [Phase 2](#) trial after showing promising results early in trials.

“I’m really excited that they are all trying different ways to boost the program and levels,” Rachael Honeyfield, who is asymptomatic but carries the GRN gene, told Being Patient.

It isn’t clear which approach will succeed. Early on, many of these drugs are effective in increasing progranulin levels in the brain and cerebrospinal fluid. Larger Phase 3 trials must show that they also slow disease progression. It also isn’t clear what type of treatment — gene therapies, monoclonal antibodies, or pills — will be first on the scene.

“There’s some concern that if you’re ramping up progranulin too much in the periphery, you might end up having long-term side effects, and you won’t be getting enough diverted into the brain to have the benefit that you’re seeking there,” Dacks said.

Does progranulin play a role in other dementias too?

Finding treatments that target GRN could also help treat other diseases.

“Some of those companies are also now looking at how the progranulin molecular pathways also relate to Alzheimer’s, Parkinson’s, and a lot of age-related neurodegenerative diseases,” Dacks said.

Alector and Glaxo-Kline Smith’s monoclonal antibody latozinemab, which binds to a protein called the sortilin receptor preventing progranulin degradation, [failed to slow](#) FTD in its Phase 3 trial. But because of the molecular overlap between the diseases, it is now being [tested in a Phase 2](#) trial for Alzheimer’s, with results expected by the end of the year.

Hall took part in the latozinemab trial as an asymptomatic participant, saying that doing so gave her “that little bit of peace of mind that I’m taking this drug and I’m not going to get what my mom and my grandmother got.”

Though she was disappointed when she learned the trial didn’t succeed, she said she hopes a treatment will be developed in her lifetime. ■

Frontotemporal Dementia Is Often Misdiagnosed — New Tests Could Help



Many with FTD receive a wrong initial diagnosis. Emerging biomarkers and genetic testing could help.

For generations, Wanda Smith’s family faced a range of neurological disease diagnoses — including senility with depression, Alzheimer’s, and **Lewy body dementia**. They were all wrong.

Doctors hadn’t realized the family members all had a genetic form of **frontotemporal dementia (FTD)**, also called frontotemporal degeneration, caused by mutations in the GRN gene. She later founded CureGRN to connect families with researchers and trials. Smith’s family wasn’t alone in getting the wrong initial diagnosis. **One in three people** eventually diagnosed with FTD are first misdiagnosed with psychiatric conditions like depression, anxiety, or psychosis.

“We have a lot of people who are being told there’s nothing wrong with you, it must be a midlife crisis,” Penny Dacks, chief science officer of the

Association for Frontotemporal Degeneration (AFTD), told Being Patient.

Affecting as many as **60,000 Americans**, FTD is an umbrella term for several rare dementias that often strike people in their 50s or 60s.

Finding biomarkers for FTD

Since there aren’t any tests that confirm that FTD causes a person’s symptoms, many people don’t get diagnosed quickly. The variety of symptoms, too, makes it more challenging. Some people develop the behavioral variant (bvFTD) resulting in personality and behavior changes while others develop primary progressive aphasia, losing their language and speech abilities. Other forms affect movement abilities first.

Even among those with the GRN gene, there’s a range of symptom presentations. Most people with this gene develop either behavioural variant FTD or primary progressive aphasia, though some show other symptoms. Even with identical GRN

mutations, the symptoms and age of onset can vary substantially.

Sandra Ahten, a semi-retired business owner living in Urbana, Illinois, cared for her mother, whose FTD progressed during the early years of the COVID-19 pandemic. When Ahten started having concerns about her mother's symptoms, which resembled stroke more than dementia, some of the doctors they went early on didn't make the right diagnosis, despite a family history of FTD.

As one of the doctors fumbled around on the computer, Ahten recalled her mother joking, "They might need to see a doctor more than I do."

It took four years for her mother, initially told she had Alzheimer's, to get the right diagnosis. A brain scan that showed the frontal and temporal lobes of her brain were shrinking finally helped doctors figure out it was FTD.

Researchers and companies are searching for a biomarker that could confirm a diagnosis through brain imaging, skin biopsies, saliva, blood, or cerebrospinal fluid. Dacks believes that when it's developed, it could screen patients, helping doctors identify who needs to be referred out to the limited number of U.S. specialists who can make the final diagnosis.



Sandra Ahten in San Francisco

Since **20 percent of cases** are caused by mutations in a specific gene, major organizations like the AFTD recommend that people with a family history

receive a genetic test.

Preparing for genetic testing

A genetic test revealed that a mutation in the GRN gene caused Ahten's mother to develop FTD. Ahten decided she wanted to know whether she carried the gene too. Before getting tested, Ahten purchased **long-term care insurance**, which she might not be able to get if she knew she had the gene. She tested positive and is currently asymptomatic.

Genetic counselling is recommended before and after testing. A counsellor helps people understand any drawbacks and risks and explains the results of the test. The **Progranulin Information Navigator** was created by the Bluefield Project, a non-profit helping accelerate new treatments for the genetic form of FTD caused by a GRN mutation, to help individuals and their families by providing free genetic counselling and testing.

Heidi Hall, another asymptomatic carrier of the GRN gene, told Being Patient that anyone considering genetic testing should prepare for the emotional and practical aftermath. She urges finding a therapist, connecting with a support group, and bracing for family tensions. Getting tested caused a temporary rift with her siblings, she said, who decided not to find out whether they carried the gene as well.

One man whose wife carries the GRN gene told Being Patient that one of their children got tested while family planning. With some forms of in-vitro fertilization, it may be possible to select for embryos that don't carry the mutation.

The positives of an early diagnosis

Soon after Ahten's mother received the correct diagnosis, her condition progressed. She passed away the following year during the early stages of the COVID-19 pandemic. As a caregiver for her mother, Ahten said she had a "long goodbye" and closure rather than a sudden loss.

But if the diagnosis had come sooner, "she could have gotten into a trial" that might slow the disease, Ahten told Being Patient. Ahten herself is taking part in the ALLFTD (Advancing Research and Treatment for Frontotemporal Lobar Degeneration),

a longitudinal study following people at risk of developing FTD across their lifespan.

“I use that study to reduce stress,” said Ahten. Rather than ruminating over every minor lapse in memory, she gets extensive testing once a year in Philadelphia, monitoring her brain health as part of the study. “That’s not your job to have your brain monitor your own brain,” she said “That’s the doctor’s job.”

Knowing she carries the GRN gene was also a “wake-up call” for Ahten, and it’s not necessarily an entirely negative experience. She’s planning a large trip later this year, and took up biking and breathwork. “What do I have to do? Go to Morocco. Go to Philadelphia once a year,” she said. “You get a long goodbye, starting when I’m completely healthy.” ■

Heidi Hall Is Defying the GRN Gene That Caused Dementia in Her Family



Heidi Hall as a child with her mother and grandmother.

After testing positive, Hall navigated family tensions while turning to ultramarathons, therapy, and clinical trials to stay hopeful about frontotemporal dementia.

Despite her best efforts growing up, Heidi Hall, the youngest of five siblings, could never seem to outsmart her mother, Mary Margaret. The day after secretly hosting a house party, she remembers her mother slyly asking: “What did you do with all the beer cans?”

Mary Margaret was the “glue” for her family and community, Hall said. A school nurse with a signature 1980s perm, she was the president of the Parent Teacher Advisory Committee, helped with her husband’s business, and managed wedding dress shopping, cross-country moves, and all appointments for her kids.

But when she was about 21, Hall noticed the glue dissolving. Her mother, 56, had trouble spelling her last name, recalling words in conversation, and couldn’t remember how long she’d been married during her anniversary. “One time I asked her to schedule a doctor’s appointment for me,” Hall told *Being Patient*. “She called me up crying that she couldn’t.”

Unbeknownst to Hall, her 79-year-old grandmother, Margaret Mary (the inverse name of her mom) was experiencing similar symptoms at the same time.

Dementia runs in the Hall family

Tracing the line back to her great-great grandmother in Slovakia, dementia has always run through Hall’s family, though no one was sure what caused it. After hearing about the Hall family’s history and the language difficulties during a medical appointment, University of Pennsylvania neurologist Dr. Murray Grossman recommended genetic testing.

“When she was trying to make dinner reservations, she was having problems finding the right words,” Hall recalled about her mother. “She was quickly losing the ability to speak the words she wanted to say.”

By the summer of 2008, the testing showed that both Hall’s mother and grandmother carried the GRN mutation, which slashed the levels of the protein progranulin. Without it, waste clearance in the brain is impaired, leading to a build up of **TDP-43 proteins**, damaging brain cells and ultimately causing **frontotemporal dementia (FTD)**. Their form, primary progressive aphasia, is the same type that actor **Bruce Willis** has.

Hall described the disease as locking them inside a “prison in their own bodies,” where her mother and grandmother could see and hear the world but couldn’t express their thoughts.

Graduating from college, Hall had to grow up fast. “My dad was so focused on my mom that it left me very much on my own,” she said. “It changed me, having to learn how to be very independent.”

Her thoughts about her early 20s are filled with regrets. “I wanted to forget that my mom had dementia,” said Hall. “I would just get frustrated because she was declining so quickly.”

In 2013, Hall’s mother and grandmother both passed away, donating their brains to research. This left Hall and her four older siblings, who all had a 50/50 chance of inheriting the gene, with a daunting question: Did they want to know if they carried it?

Learning her genetic status

In 2010, the Hall siblings took part in an observational study for people at risk of FTD, but none of them chose to learn whether they carried the gene. The study ended due to a lack of funding.

“I don’t want to know the results for something that can’t be prevented or there’s no clinical trials and nothing in the pipeline,” said Hall.

Hall’s winding career path eventually brought her to a contractor role within the National Institutes of Health (NIH) working with clinical research. “I

always feel like I was brought to NIH to redirect me into getting genetically tested,” she said.

Scrolling through a trials database one day, she discovered new studies at the University of Pennsylvania. In 2018, at age 31, she joined **ALLFTD**, a longitudinal study that tracked people at risk of FTD over their lifetime.

This time, she chose to learn her genetic status and found out she carried the GRN gene.

Hope in the Aftermath

After testing, Hall started making changes. She adopted a puppy, cut toxic relationships from her life, got a therapist, and started training to get back into ultramarathon running.

“I had to deal with my anxiety and my anger,” she said, “and focus on being even more healthy.” Her siblings, who opted not to join trials or learn their genetic status, distanced themselves at first. “The sister who had told me she would be there for me, ended up not being able to fully be there,” said Hall. Another sibling who heard she got tested cut off contact for eight months.

“You can’t be angry at those family members,” she said. With time, her siblings reconciled.

Hall urges anyone considering genetic testing for FTD to prepare for the emotional and practical aftermath by securing life insurance, finding a therapist, and connecting with a support group.



Heidi Hall running in a marathon.



Heidi Hall and her dog Sky. She adopted the dog after finding out she had the GRN gene.

The drug that could prevent her disease

In 2019, the University of Pennsylvania got in touch with Hall to tell her about a new Phase 3 clinical trial. Alector was testing a drug called latozinemab

in people with the GRN gene, who either had FTD or were still asymptomatic.

Latozinemab is a monoclonal antibody like Leqembi or Kisunla. But instead of sticking to beta-amyloid, latozinemab binds the sortilin receptor, preventing the destruction of progranulin.

She joined the trial, saying, “I couldn’t save my mom, but I can possibly save other family members.”

Unfortunately, latozinemab failed to slow the disease in Phase 3 trials and was discontinued in 2025. It’s now being tested as a potential treatment for Alzheimer’s instead.

When the study began, Hall said she felt the lack of support from her siblings, but when it stopped, “they all called me that day to check up on me, because they knew they had to be there for me.”

Despite setbacks, Hall remains hopeful. Several new therapies targeting GRN — including gene therapies — are now in Phase 2 trials and could prevent or delay FTD in her lifetime. ■



Robert

WANJALA

in Journalism Hub East Africa | Kenya



With over a decade of experience in investigative journalism, I have dedicated my career to amplifying marginalized voices and uncovering systemic injustices through evidence-based reporting. My work explores the intersections of social justice, human rights, governance, and corruption, translating complex data and lived

experiences into compelling public interest stories. Guided by a belief that journalism should inform, empower, and hold power to account, I strive to produce stories that not only expose wrongdoing but also inspire reform and resilience within communities often left unheard.

From Survival to Strength: New Technology Offers Lifeline to Sickle Cell Patients in Western Kenya



JOOTRH Victoria Annex. Sickle cell patients can now access apheresis treatment at JOOTRH following the installation of the Spectra Optia machine. Photo courtesy JOOTRH

Spectra Optia system at Kisumu hospital transforms sickle cell care, replacing conventional transfusions with life-saving automated red cell exchange

Kisumu, February 10 – When the chest pain struck, Antony Lubuka Olila thought his time on earth had come to an end. “I felt a sharp, intense pain I couldn’t explain,” he recalls. “It was the worst chest pain I had ever experienced. Doctors identified it as acute chest syndrome, one of the most dangerous complications of sickle cell disease. It was tough.”

Between 2016 and 2019, the Busia-based teacher at Bugengi Primary Special School, which serves learners with special needs, endured repeated episodes of the life-threatening condition. At 54, Lubuka has lived with the genetic blood disorder his entire life. He has buried siblings who had it.

He has survived blood transfusions, crippling pain crises and decades of stigma. Yet he still walks into a classroom each day to teach and impart life skills to special needs learners.

“I have been a teacher for 30 years,” he says. “Sickle cell disease is not a death sentence.”

For Lubuka, a husband and father of two, the disease has never been an abstract statistic. It has been a lifelong companion – sometimes silent, other times brutal. “Pain is almost a constant companion,” he says calmly. “Sometimes you can go for months, even a year, without a major crisis. But when it comes, it hits hard.”

Listening to his body has become as critical as any prescription. In 2016, he made a personal choice to stop taking sugar and heavily spiced foods. “I stopped using sugar, and I started feeling some relief from the constant pains,” he says. “I don’t know if sugar caused the chest pain, but nowadays I don’t feel it.”

He is cautious not to generalise. “I don’t say sugar is bad because I have no medical evidence. But in my experience, it affected me.” He switched to simple, mostly boiled foods. “Spicy foods upset my stomach and cause discomfort, so I stick to natural foods.”

His story mirrors that of thousands of families across Western Kenya, where sickle cell disease remains one of the most under-recognised public health challenges.

According to the World Health Organization (WHO), about 14,000 children are born with sickle cell disease (SCD) each year in Kenya. Across Africa, an estimated 240,000 children are born with the condition annually. Globally, nearly 100 million people live with SCD, with over 70 per cent of affected births in Sub-Saharan Africa. In the region, 50 to 90 per cent of affected children die before their fifth birthday, many without ever being diagnosed.

Many face preventable complications such as stroke, severe anaemia and acute chest syndrome simply because specialised care has been out of reach.

Behind those statistics are lives shaped by pain, resilience, and increasingly, hope. For patients like Lubuka, awareness remains a major gap. He is not yet aware of new treatment technologies, which reflects the broader need for increased public awareness. “We need support, care and information,” he says. “If people knew more about treatments, more lives could be saved.”



Nurses observe the newly installed Spectra Optia Apheresis System at JOOTRH, enabling timely and reliable treatment for sickle cell patients. Photo courtesy JOOTRH

A new technology improves survival prospects

In Kisumu, that awareness gap is being addressed through medical innovation. At the Victoria Annex

of Jaramogi Oginga Odinga Teaching and Referral Hospital (JOOTRH), clinicians have permanently installed the Spectra Optia Apheresis System, developed by Terumo Blood and Cell Technologies. The machine shifts care from conventional transfusions to automated red cell exchange, selectively removing sickled red blood cells and replacing them with healthy donor cells.

Using centrifugation, the system separates blood components, removes the abnormal cells, and transfuses healthy donor red cells back into the patient. This reduces haemoglobin S levels from nearly 100 per cent to below 30 per cent, improves blood flow and oxygen delivery, and significantly lowers the risk of complications such as stroke and acute chest syndrome.

Sickle cell disease is caused by a mutation in the beta-globin gene. Instead of round, flexible red blood cells, patients produce rigid, crescent-shaped cells that block blood flow, deprive tissues of oxygen and trigger severe pain. Over time, organs including the lungs and brain are damaged. Acute chest syndrome – the complication Lubuka endured – remains leading causes of death. Stroke is another life-threatening complication, particularly in children.

Traditional management relies on supportive care, hydroxyurea to increase protective fetal haemoglobin, and manual blood transfusions. But chronic transfusions can lead to iron overload, damaging the liver and heart. Medical experts say automated red cell exchange reduces that risk more precisely.

Kevin Otieno of Conrad Science, which collaborated in installing the system, says its capabilities extend beyond sickle cell. “We have introduced one of its key capabilities: red cell exchange for sickle cell patients. Beyond that, we will expand its use to therapeutic plasma exchange, platelet collection for oncology patients, stem cell collection and bone marrow processing. Over time, we can even begin autologous transplants and specialised depletion procedures for patients with complex haematological needs.”

Dr. Thadeous Masawa, a consultant pathologist, says the treatment is structured, not curative. “Red blood cells live about 120 days, so patients need repeat procedures every three to four months. The goal is to

reduce crises and improve quality of life.”

Dr Brenda Misore, a paediatric haematologist and oncologist, highlights its importance for children. “Many complications seen in adulthood begin in childhood. The goal of red cell exchange transfusion is to prevent these complications early, allowing children to grow, develop and live healthy lives.”

Previously, patients travelled to Nairobi or relied on visiting private facilities, paying up to Sh120, 000 per session. “Permanent local availability reduces delays, travel costs and overall expenses,” she says.



JOOTRH Chief Executive Officer Dr Joshua Clinton Okise checks on a sickle cell patient. Photo Courtesy JOOTRH

The silent burden of stigma and data gaps

Lubuka was diagnosed at about two years old. In a family of nine children, four were born with sickle cell disease. Today, he is the only one alive. “My sister died in 2016 after a stroke. One brother died at 10 after severe malaria. Another died in a road accident,” he says.

At 46, Lubuka began taking hydroxyurea. “Hydroxyurea has strengthened me and reduced the frequency of crises.”

He has also endured critical transfusions. At 24, shortly after starting his teaching career, a severe malaria-induced anaemic crisis left him hospitalised for nearly a month with a haemoglobin level of 2.8 g/dL.

“That was my first worst crisis. It almost knocked me off,” he recalls. Yet he returned to the classroom. “Even now, I am still serving,” he says with a smile. “I have educated Kenyans. We can be beneficial to society.”

His voice sharpens when he speaks about stigma. “Many people believe that people living with sickle cell disease cannot live for long. Another thing they believe is that people living with sickle cell disease are weak and they cannot do anything. People with sickle cell disease are just human beings like any other person.”

Beyond stigma, he says, the country is still grappling with a data gap. “We don’t have a national register to know how many cases we have,” he notes, highlighting how weak surveillance continues to hinder planning and response.

Globally, early screening has transformed outcomes, cutting childhood mortality from over 50 per cent to below 5 per cent in high-resource settings. SCD not only causes severe medical complications and chronic pain but also imposes heavy emotional and economic strain on families.

Locally, however, progress is emerging. Dr Steven Ojuma, Director of Victoria Annex Hospital, says the impact is visible. “Sickle cell disease touches almost every family here. This is the first public facility in Kenya with this technology permanently installed.” In Kisumu, about one in five children is born with the sickle cell trait, and among 500 children followed from birth over four years, only 11 died – a 2.2 per cent mortality rate compared to historical figures of up to 80 per cent, says Dr. Ojuma.

Western Kenya, in a malaria-endemic zone around Lake Victoria, bears a disproportionate burden. Kisumu County is among the hardest-hit areas. About 1,500 children are born with sickle cell disease annually. Approximately 30 per cent of the population carries the sickle cell trait, meaning two carriers have a 25 per cent chance with each pregnancy of having a child with the disease.

According to the Ministry of Health (MoH), about 248,718 Kenyans are living with SCD, with an estimated 400 deaths annually. The disease is most prevalent in 18 out of 47 Counties, particularly in Western, Nyanza and Coastal regions. Recent data from Kisumu and Kisii counties indicate prevalence rates of between 1.5 and 4 per cent, with migration and intermarriage contributing to its wider spread. Although a centralised national registry is still in development, regional registries at JOOTRH and KEMRI continue to support data collection.

Dr. Patrick Amoth, Director General for Health in the MoH says it has developed a strong policy framework to guide prevention, diagnosis, management and patient support, including the establishment of a multi-sectoral Hematological and Rare Diseases Technical Working Group bringing together key stakeholders.

Dr. Amoth says SCD financing is structured through the Primary Healthcare Fund, which covers infant screening and essential preventive care such as folate, penicillin and malaria prophylaxis, pain management and immunisation boosters. Under the Social Health Insurance Fund (SHA), patients can access outpatient care capped at Sh2,000 per visit (up to four visits annually), as well as inpatient services, medical imaging, and pharmacy and specialised laboratory services limited to Sh5,000 per quarter per person.

However, sickle cell programming in Kenya remains hampered by low public awareness and stigma, limited newborn screening, and diagnostic services confined to five counties – Kisumu, Uasin Gishu, Busia, Nairobi and Kilifi (KEMRI), the ministry notes. Shortages of trained providers, frequent stock-outs of essential medicines, weak surveillance systems pending a national registry, and high out-of-pocket costs further restrict access to care.



Medical experts from across East Africa gathered in Nairobi in October 2024 to discuss hemophilia, sickle cell, and other blood disorders. Photo courtesy Sickle Cell Federation of Kenya

Beyond Kisumu

About 100 kilometres away in Bungoma County, Dr Dickens Lubanga estimates nearly 1,000 children are living with sickle cell anaemia. “Access to hydroxyurea and specialist care remains limited,

especially in rural areas. SHA coverage for outpatient medication is inadequate. This machine, alongside newborn screening and outreach, can reduce these barriers,” he says. Without routine screening and care, children risk dying from preventable infections, severe anaemia, or acute chest syndrome before their fifth birthday, adds Dr Lubanga.

While Kisumu County has made strides through specialised centres, partnerships for affordable medication, and expanded newborn screening, however, the burden of sickle cell disease remains heavy. Patients from Bungoma, Vihiga, and Busia counties are expected to benefit from the regional availability of automated exchange transfusion. For children at high risk of stroke, regular red cell exchange can cut incidence by more than 90 per cent. While it does not replace hydroxyurea, it offers a vital option for severe cases.

The technology is supported by a financing and sustainability model designed to keep the service accessible. JOOTRH Chief Executive Officer Dr Joshua Clinton Okise says the installation was made possible through a public–private partnership. The hospital has secured five years of technical support, ongoing staff training, and funding for consumables and maintenance. If demand rises, a second machine will be added to expand capacity, says Dr. Okise

The resilience behind the medicine

While clinicians celebrate innovation, survival for patients like Lubuka is built on daily discipline as well. “Doctors told me to reduce overtraining and rest more,” he says. “But as an African man with family obligations, I have to keep working hard.”

His resilience is also rooted in self-advocacy – monitoring his diet, tracking symptoms, and adjusting daily habits. “What has given me strength,” he says, “is acceptance. Accepting my condition early in life and choosing to live positively with it.”

“I feel like a fighter,” he says with conviction. The school day is over. He swings his backpack over his shoulder as he stands ready to lock his office door, and as we walk towards the gate, he adds, “I have faced many battles, and the journey has not been easy.” He had asked not to be photographed.

Now a grandfather, he sees himself as proof that survival beyond childhood is possible. “I am 54 years old and still going strong. With proper medication, regular clinic visits and self-care, people living with sickle cell disease can grow, thrive and give back to the community.”

He is not asking for sympathy. He is asking for increased awareness. He spends every moment of his free time leading advocacy in schools and his community to reduce stigma and correct myths about sickle cell disease. ■

About Fondation Ipsen

There are 7,000 rare diseases affecting 300 million people worldwide. 75% of patients are children. 1 in 2 patients do not have an accurate diagnosis. A quarter of patients wait 4 years to get a diagnosis. Our program brings together world experts to improve this dire situation.

About The National Press Foundation

The National Press Foundation is a 501(c)(3) whose mission is to “make good journalists better.” We educate journalists on the complex issues of the day and train them to use the latest reporting tools and techniques. The foundation recognizes and encourages excellence in journalism through its awards and fellowships.

Since 1976, the foundation has provided in-person professional development opportunities to thousands of editors, producers and reporters, helping them better understand and explain the effects of public policy on readers and viewers. All NPF programming is free and on the record.

National Press Foundation is funded by journalism organizations, foundations, corporations and individual benefactors. We are grateful to our funders who are listed here.

Prior to the novel coronavirus, NPF programs were held in the nation’s capital, around the United States and overseas. During the global pandemic, we are offering all-virtual training, and continuing to bring journalists together with leading authorities to discuss significant issues ranging from health and economics to politics and policy. NPF produces digital curriculum from these fellowships and briefings that are posted to our website, allowing journalists across the world to access the best expertise and enhance their reporting.

Journalists are currently under fire, overworked, underpaid, and too often threatened with violence. The landscape for media continues to deteriorate with widespread layoffs, newsroom closures, mistrust, and disinformation. Against this grim background, the National Press Foundation’s mission—making good journalists better—has become more necessary than ever.

About the collaboration

The National Press Foundation and Fondation Ipsen plan to select and train a delegation of the world’s leading journalists to focus on Rare diseases. The goal of this meeting is to connect leading Science journalists to patients, scientists, caregivers and patient groups.

The National Press Foundation and Fondation Ipsen will select a group of international journalists to participate in a four-day training program to offer them the opportunity to interact with scientific experts and learn better ways to communicate information on rare diseases to the general public (detection, diagnosis, molecular biology, symptoms, etc.). The convening will reveal newly published scientific information and facilitate interaction between journalists, scientists and patients.



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One of the great challenges faced by people living with rare diseases is that the public knows so little of their struggles, challenges, and the need for unrelenting resilience in a world that doesn't understand. Coupled to the 300 million people living with rare diseases are hundreds of millions of caregivers who sacrifice time, work and money in the name of love.

Working with the National Press Foundation, top world journalists highlight in these writings the plights of patients living with rare diseases and those who care for them. The stories are informative, dramatic, and heart-rending. I thank most the patients living with rare diseases and the people who love them for sharing their stories with these gifted journalists.

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