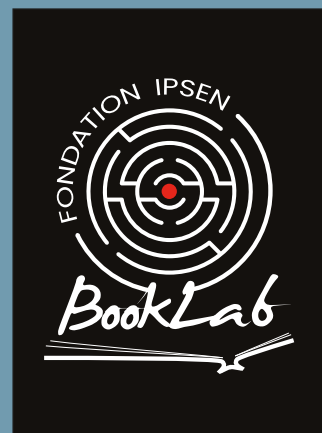
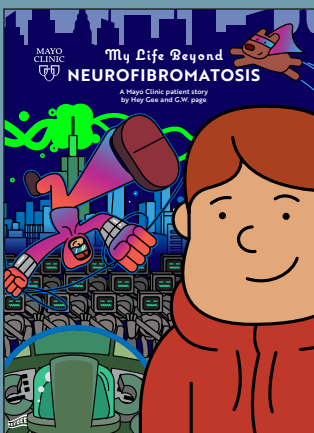
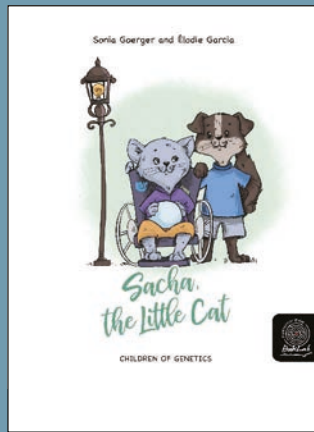
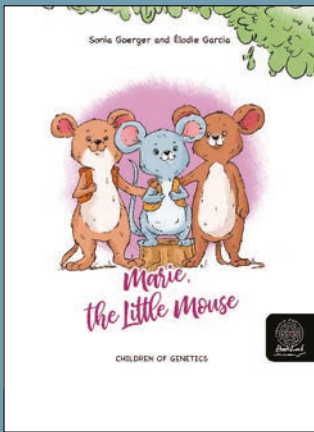
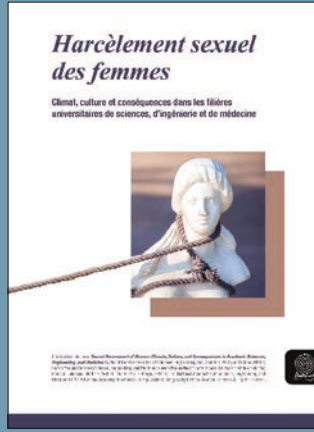


RARE BUT **NOT ALONE**



Living with a
RARE
disease
worldwide





Expanding and Informing Journalistic Coverage of Rare Diseases

In 2021, with newsrooms around the world riveted by the impact of COVID-19, the world's healthcare journalists found it daunting to pitch to their editors any stories that were not related to the virus. Yet at the same time, health reporters and editors were thirsty for compelling stories about topics other than COVID-19 — especially ones that gave hope to pandemic-weary audiences.

Covering rare diseases is a particular challenge for journalists because the word “rare” itself implies the topic is not of particular interest to mass-market audiences. On the contrary, the number of rare disease patients and those who love and care of them may easily number half a billion people – and the advances being made in diagnosis and treatment are of interest to man more.

Against this backdrop, the National Press Foundation with the support of the Ipsen Foundation tackled this challenge by organizing a three-day online conference to inform journalists about recent scientific progress in understanding, diagnosing and treating and writing about rare diseases with nuance and sensitivity.

Twenty fellows were then selected and offered the opportunity to travel wherever they needed to do the stories they deemed most important to their audiences.

This volume brings together some of their extraordinary reporting. Their work includes newspaper, magazine and science journal articles, video, online stories, radio broadcasts and moving photographs. The fellows' work comes from the United States, Central and South America, Europe, Africa and Asia.

For Radio Sweden, Anna Larsson reported that new genomic techniques have given 1,000 to 2,000 Swedes new genetic diagnoses and brought clarity to their mysterious symptoms. U.S. Freelance biomedical journalists Bob Rohr wrote about advances in phenylketonuria and Meeri Kim's story about acute myeloid leukemia offered hope in the form of information about emerging research.

This volume also includes stories on rare diseases from across Africa. In Kenya, Mactilda Mbeny wrote about microcephaly, debunking the widespread belief that the condition is caused by witchcraft. Ridwan Karim Dini-Osman produced a six-minute radio feature about the availability of rare disease treatment in Ghana for the PRX program “The World,” which has a worldwide

reach. And Clement Edward Msiska filed a poignant account of a family with a rare form of hemophilia for Malawi's Capital One Radio network.

We hope that this selection of articles will give offer patients and their families solace in knowing that readers and listeners around the world care about their fate. And we offer all resources and videos from the training free to download from the National Press Foundation website, in hopes they will inspire other journalists around the world to cover this vital topic.

National Press Foundation thanks the Ipsen Foundation, in particular Dr. James Levine and Yannick Tanguy, for their unfailing support.

Sonni Efron

CEO, National Press Foundation, Washington D.C.

Because we can't wait longer

The Fondation Ipsen is committed to improve the condition of people living with a rare disease and to raise public awareness about their daily lives.

Because living with a rare disease can be difficult or even very difficult.

Because a rare disease is not always easy to diagnose, whether because of a lack of knowledge or a lack of resources. As you will see in this book, not all countries have the same resources to support patients, unfortunately.

Because seeing your health decrease is a terrible ordeal, especially if there is no treatment. Because living with a difference is not easy, especially in communities where norms are suffocating and draconian.

Because living sick without doctors being able to determine a diagnosis is a leap into the unknown.

It is for all these reasons that this book exists.

It is for all these reasons that we have selected all these talented journalists: to bring you testimonies from all over the planet to enable you to understand, if not to join in, the daily fight of 300 million people who live with a rare disease.

You can now prove by your commitment that these persons are not alone.

Yannick Tanguy, PhD

Fondation Ipsen, Paris.

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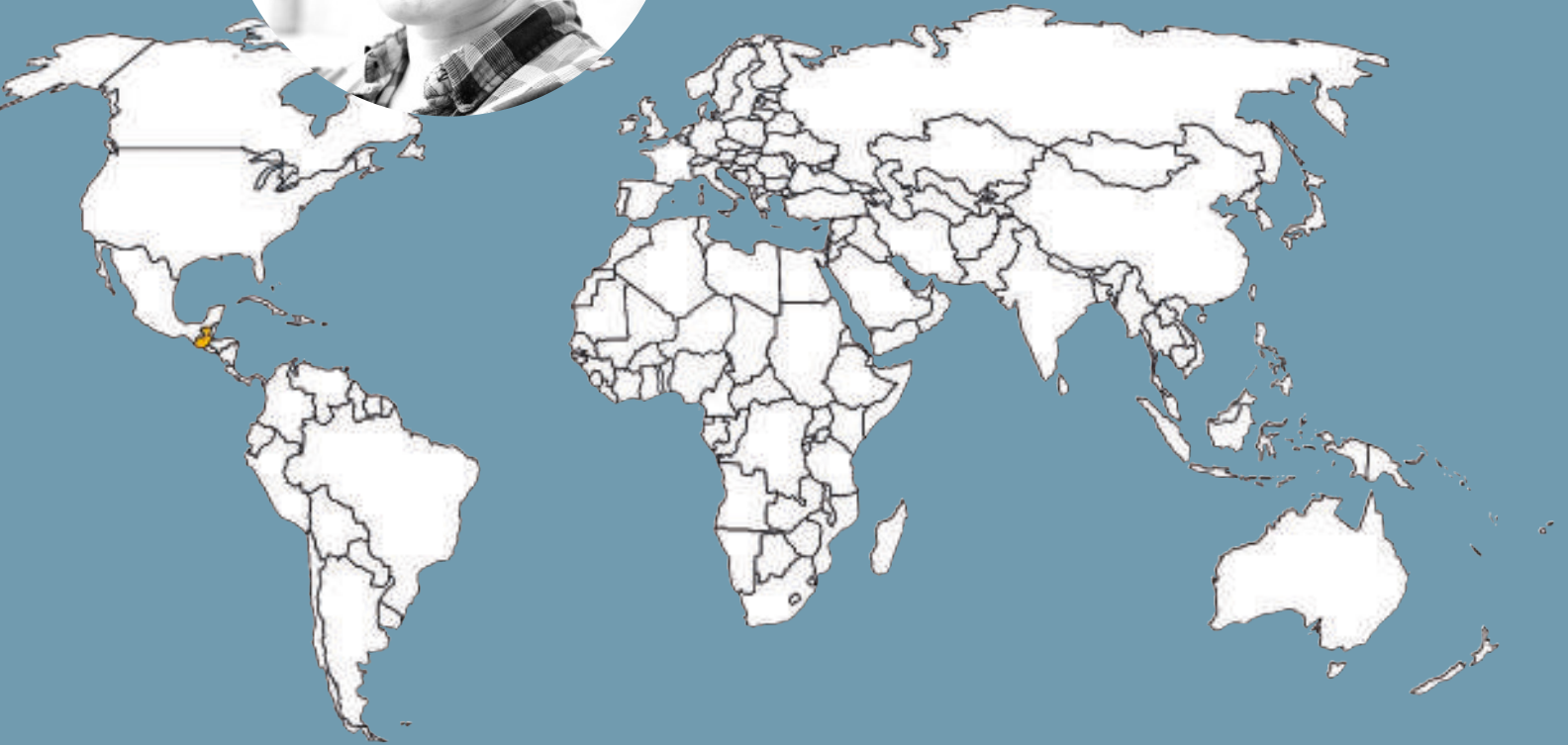
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Sandra CUFFE

Guatemala



Sandra Cuffe is a freelance journalist based in Guatemala, where she covers politics, human rights, migration, and social movements. Her work has been published by Al Jazeera, the Los Angeles Times, The Intercept, the Guardian, and many

other publications. She has lived in Central America for 14 of the past 20 years and has reported from 12 countries in the Americas.

Rare disease patients push for access to treatment in Panama

Panama has Central America's only law for rare disease health care, but advocates say it is not being fully implemented.

Reporting for this story was supported by a grant from the National Press Foundation.

Panama City, Panama - Maria has lost track of how many times she has seen a healthcare worker over the past three years. But despite innumerable medical checks, she still does not have any clear answers as to what is ailing her.

"I have been sick for so long. I want to know why," said Maria, who asked that her last name not be used to avoid possible repercussions at her Panama City workplace. "One doctor said maybe I have some kind of rare condition, but he did not say which."

Now in her early 20s, Maria said she has been living with chronic fatigue, muscle aches and other symptoms for several years - and that they have been gradually getting worse.

"I like to watch the birds," she told Al Jazeera from a bench in the city centre as songbirds flitted about in the shade of a nearby tree. "They help take my mind off everything."

Push for answers

If Maria is in fact living with a rare disease, she may have more waiting ahead as a proper diagnosis can often take several years in Europe and longer in Panama. Even then, if Maria is diagnosed with one of the five percent of rare diseases that has an approved therapy, she might not have access to it if Panama does not purchase it for the public health system.

But a movement has been mobilising in the Central American nation to change that.

Patient- and parent-led Panamanian associations are joining forces to lead a push for concrete action to ensure early diagnosis, medical care and access to treatment for people living with rare diseases, and they officially formed an umbrella group - the Panama Rare Disease Network Federation, RedER - late last year to have their voices heard.

"We are stronger when we are united," said Alaisa Arauz, director of the Panamanian Hemophilia Foundation, one of 10 organisational members of RedER, which also

incorporates individuals with diseases so rare there are not enough patients in the country for a group.

Rare diseases are only rare when considered individually. Globally, an estimated 300 million people live with one or more of more than 7,000 known rare diseases. Most rare diseases are genetic in origin, many are life-threatening, and 95 percent have no approved therapy.

Hemophilia has an approved therapy in Panama, but it took 14 years of non-stop, patient-led advocacy in the country in the 1990s and early 2000s to pressure the government into acquiring enough Factor VIII concentrates to cover patients' needs. Factor VIII is the missing clotting factor in Hemophilia A patients' blood.

"It was a huge achievement," Arauz told Al Jazeera. "It was years of struggle. With every government that took office, we were there."

Arauz has two sons who have been diagnosed with hemophilia, and she lives with Von Willebrand disease, another genetic bleeding disorder. But it was decades before she was diagnosed.

"It was [bleeding episodes] my whole life," said Arauz, who also acts as the delegate for rare disease patients on an intersectoral, government-created committee tasked with ensuring access to treatment for such patients. "Imagine if I had had an early diagnosis. I would not have suffered for so long."

Rare diseases law

Formally known as the Intersectoral Committee of Prevention, Diagnosis, Comprehensive Attention and Research for the Treatment of Rare, Uncommon and Orphan Diseases, the committee was born out of a 2014 Panamanian law that deals specifically with rare diseases - the only such legislation in Central America.

In Panama, as in the European Union, rare diseases are defined as those that affect fewer than 1 in 2,000 people.

The law aims to ensure social protections, diagnosis and treatment access for people with rare, uncommon and orphan diseases. But while the legislation has been on the books for more than seven years, it has yet to be fully implemented.

"It is a very good law, a very complete law, but as we have called it, a 'decorative' law," said Arauz.

The law, for example, instructs the Ministry of Health to set up a national programme for purchasing medications for the treatment of rare diseases, "to enable immediate and equitable access for all patients". But that key aspect has not yet been fulfilled.

One of the committee's responsibilities is establishing cooperation agreements with governmental and private sector bodies to fund treatments for rare disease patients. But the committee was not established until 2019 and has made little headway since. Along with Arauz, it includes six representatives from different government entities, as well as a pharmaceutical industry delegate.

The obstacle, according to Arauz and other rare disease patient association leaders, is that the Ministry of Health, which presides over the committee, has switched its representative several times and those representatives also have other health ministry duties and are not solely dedicated to rare diseases.

The Ministry of Health was unavailable for comment before time of publication, and the institution has made very few public statements on the issue of rare diseases over the past two years.

In a statement from October 2020, the ministry noted some advances, including the identification of more rare diseases in Panama and a committee tasked with developing disease-specific management and treatment guides.

'A full life'

But patient advocates say the longer it takes to implement the law, the longer people go without care.

"The law is not being fulfilled and patients are not being treated," said Enma Pinzon, president of the Rheumatoid Arthritis Foundation of Panama and a longtime patient movement leader also involved with the National Federation of Associations of Patients with Critical, Chronic and Degenerative Diseases.

Pinzon told Al Jazeera that when rare diseases do get public attention in Panama, it is through charity and aid campaigns to improve the living conditions of people diagnosed with diseases for which there are no treatments. That is important, but the focus ends up overshadowing the fact that there are some rare diseases with approved therapies and patients that need them, she said.

The focus of RedER will be advocacy to push the government to implement the law and other measures, added Pizon, stressing that the goal is to ensure everyone that needs medical care can get it.

"I have a chronic disease," she said. "Thirty years ago, I had the luck of receiving medication. I had the luck of receiving an early diagnosis and here are the results. I have had a full life and that is what I am seeking for everyone with illnesses in Panama."

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Ridwan Karim DINI-OSMAN

Ghana



Ridwan Karim Dini-Osman is a multiple award-winning development journalist and news anchor based in Ghana.

Dini-Osman is a Pulitzer Centre Grantee and recipient of the 2018 Lorenzo Natali Media Prize, a prestigious global award run by the European Commission.

In 2020, he was awarded best African TV Journalist in Environmental and Change Reporting by the Pan-African Climate Justice Alliance (PACJA).

Ridwan Karim Dini-Osman is also a 2021 fellow of the University of Rhode Island's Metcalf Institute Annual Science Immersion Programme for Journalists and won the 2021 International Center for Journalists' Global Health Crisis Award for Covid-19 reporting.

He holds a Bachelor's Degree in Communication Studies from the Ghana Institute of Journalism.

By definition, a rare disease affects a very small number of people, but add up the thousands of rare diseases out there and you're looking at an impact on some 400 million people worldwide. The vast majority of those rare diseases have no treatment. Drug companies see little financial incentive to develop them. In West Africa, raising children with rare diseases can bring traumatic hardships. Young patients with limited access to healthcare are suffering unnecessarily.

Reporter Ridwan Karim Dini-Osman has our story from Accra, Ghana.

In the sprawling settlement of Dome, a suburb of Accra, I meet this family of five humming along to a local gospel tune in their small apartment. When Samuel Donkor tides the note with his beloved wife in the year 2000, he very much looked forward to the joys of fatherhood. But that dream was shattered when his first child arrived with a rare disease. Without knowing what it was, he sought answers in many places.

"Going to Hospital. Buying medicine from roadside. Buying medicine from drugstores."

To make matters worse, Donkor's second and third children also arrived with the same condition. The 52-year-old says he constantly endures stigma from neighbors.

"One day I have a problem with a certain lady and the lady told me that if I have medicine I should cure my children there, the sickness. I sit down for some time. Why God? why?"

The first among Donkor's children, Jonathan dropped out of school owing to mobility challenges. The 19-year-old just like his siblings, has deformed bones, short limbs, small hands with clubbed fingers, dwarfism and a large upper face. His brain is unaffected by the disease.

"What do you want to become in future?"

I wanted to become a doctor or banker in the past. But now when I get money out to support those who are like me. I want to help them. Now that they can get their freedom."

But he is trapped in a small chair with his ankles and knees constantly hurting. His mother, Grace Adabelle, says the children condition has taken a toll on every aspect of family life.

"When I look at my sister's children and then I look at my own, the pain in my heart is just too much to bear due to their condition. I can't even engage in any meaningful economic activity because caring for them is a full-time job."

For 17 years, this couple did not know the exact condition of their three children until a not-for-profit organization, Rare Disease Ghana Initiative intervened. Samuel Agyei Wiafe started the NGO in 2017.

"I had opportunity to meet a family with an unknown syndrome. Looking at how the health system was set up, it was very challenging to support this family and so that drew me into the rare disease space to advocate for the family."

His organization is the only one of its kind in West Africa. There is no recognized definition for rare disease in many African countries and very little data on prevalence, treatment options and support structures. This is compounded by a lack of capacity to recognize and test for rare diseases. It turned out Donkor's children have been living with a rare genetic disease called mucopolysaccharidosis type 4. There is no cure, except an enzyme replacement therapy. But it is very expensive, meaning families like the Donkors cannot afford it. Durhane Wong-Rieger, chair of Rare Diseases International based in Toronto, Canada.

"Around the world, probably less than 1/10 of 1% are getting access."

But the possibilities for diagnosis and treatment for millions of rare diseases patients around the world are advancing rapidly.

“Huge amount of resources innovation put into developing these therapies. And guess what? Nobody is getting them. So this is one of the challenges that we have to address.”

Doctor Ann Perisa is director of the Office of Rare Diseases Research at the National Institutes of Health.

“We're in a time of just unprecedented scientific opportunity, and what we're really facing now are more operational logistical barriers. We really do need to get away from this one disease at a time strategy for any disease.”

To fix this, the international community of rare disease patients and their families want to see the United Nations adopt a resolution in support of people who live with rare conditions. They believe this resolution would encourage governments around the world to update their policies regarding rare diseases.

In Accra, caregivers like Samuel Donkor have added their voice to this call.

“Not my children alone. Everywhere the whole world, we have to help children living with a rare disease.”

But because the world is already dealing with so many health crises, it is unclear whether the UN will act.

For the world, I am Ridwan Karim Dini-Osman in Accra, Ghana.



Kelechukwu IRUOMA

Nigeria



Kelechukwu Iruoma is a multi-award-winning Nigerian freelance journalist with five years of experience in investigative, development and solutions journalism. His works on global health, environment, education, social justice, politic, and business have been published by Al Jazeera, Thomson Reuters Foundation, NPR, The Fish Site, African Business Magazine, TRT World, Global Sisters Report, among many others. Kelechukwu is a committed journalist who speaks for the voiceless.

Kelechukwu has won local and international awards for his work on environmental crime, corruption, and social injustice. Last year, he emerged the overall winner in the Business Category of the West Africa Media Excellence Conference and Awards (WAMECA). He was recently announced as the runner-up for the Society of Environmental Journalist Reporting Awards on Environment.

INVESTIGATIONS

FEATURES: Rise in Monkeypox infection troubling Nigeria's rural population



Published 1 week ago on December 24, 2021
By Ripples Nigeria



*In this features, KELECHUKWU IRUOMA goes into the rural communit
Bayelsa State, south-south Nigeria to uncover the plight of residents
with monkey pox disease, and the challenge of authorities to deal v*

On a sunny afternoon in April 2021, Desmond Ere, 45, had just had his bath when his began to itch.

Ere, who owns an online fashion store, thought his itchy skin would stop. But it didn't

“At first, I thought it was a waterborne disease because I started experiencing skin ch the form of rashes,” he said.

Whenever he poured warm water on his body, he would be relieved for some minutes itching would begin again.

When he could no longer bear the discomfort, he approached a chemist in Yenagoa, t of Bayelsa State in the Niger Delta, where he lives. He was told he had contracted Chi

“They gave me some drugs for Chicken Pox and said I would be given an injection for days,” he said. “Three days after I received the injection, there was no improvement,”



Ere managed to complete the dose of the injection. Then the symptoms became severe and his entire body broke out in a rash. His head and face were swollen. He had to shave his hair.

Some of his family and friends told him the illness was spiritual. They suggested he seek traditional medicine to treat himself. They contributed money and got a traditional medicine which cost N25,000, and asked Ere to take it. But he refused and said he would rather go to hospital.

In late April, Ere met a doctor who instructed him to go to the Community Medicine and Public Health Department of the Federal Medical Centre, Yenagoa, where he would meet Doctor Stella Rotifa, a public health physician, who would help find out what the problem was.

At this stage, Ere became afraid of what was happening to him. He could not stand and walk properly. He had to use a walking stick. Each time he had to use the bathroom, he was restless.

When he got to the hospital, Rotifa observed his body and asked him some questions which she took his blood samples and carried out a clinical laboratory test to find out what was wrong with him.

When the result came out, it was discovered that he was positive for Monkeypox.

“It was at this point that Rotifa started giving me the right drugs to take,” he said. “She gave me drugs that would serve me for one week because I was coming for treatment and going home.”

Monkeypox situation in Nigeria

Monkeypox is a viral infectious disease that jumps from a non-human animal to humans. It primarily occurs in rainforest areas of Central and West Africa. Typically, it comes with fever, and swollen lymph nodes. While it is mostly transmitted to people from wild animals, human-to-human transmission also occurs.

The virus is transmitted from one person to another by contact with lesions, body fluids, respiratory droplets, and contaminated materials such as bedding.

Data from the Nigeria Centre for Disease Control (NCDC) website revealed that between 1970 and 1978, ten human Monkeypox infections were reported in Nigeria. While three were laboratory-confirmed – two in 1971 and one in 1978.

However, between September 2017 and June 2021, Nigeria has had a surge in the number of suspected Monkeypox cases – 466 out of which 205 were confirmed. Out of the confirmed cases, there were 88 in 2017, 49 in 2018, 47 in 2019, 8 in 2020, and 13 in 2021.

There have been eight deaths from the disease since September 2017.

The number for 2021 is likely to be an under-representation because many people are likely to have been avoiding healthcare facilities for fear of contracting COVID-19 disease.

A rare disease

A report from the Centre for Disease Control says Monkeypox is a rare zoonotic infection having been identified first in humans in the Democratic Republic of Congo back in 1970 during a period of effort to eliminate smallpox.

This was after the virus was first discovered in 1958 when two outbreaks of a pox-like disease occurred in colonies of monkeys kept for research, hence the name ‘monkeypox.’

The first time that a case was reported in Nigeria was in 1978, when a 4-year-old child in the southeastern part of the country fell ill with the disease, according to the NCDC.

It only came up again thirty-nine years later on September 22, 2017, when a suspected case was reported to the Nigeria Centre for Disease Control (NCDC).

It can easily be confused with other rash illnesses such as smallpox, chickenpox, measles, bacterial skin infections, scabies, syphilis, and medication-associated allergies which are difficult to diagnose easily.

Failure to detect and report on time will result in the further spread of the disease, which can cause death in at least one in 10 of those infected. The disease can spread quickly.

READ ALSO: Indian COVID-19 strain now in Nigeria, Disease Center warns

Enugu State Epidemiologist, Dr. Chinyere Ezeudu, said that transmission of monkeypox occurs when a person comes in contact with the virus from an animal, human, or material contaminated with the virus.

She explained that human-to-human transmission results from close contact with respiratory tract secretions, skin lesions of an infected person, or objects recently contaminated by patient fluids or lesion materials.

“Household members of a person infected with the virus also have a very high risk of infection through droplet respiratory particles during prolonged face-to-face contact,” she said.

She advised that people who are caring for patients should wear gloves and face mask and avoid any direct or droplet contact, adding that the symptoms of the virus include fever, headache, muscle aches, and swollen lymph nodes.

To treat the virus, smallpox vaccination should be administered within two weeks of exposure to monkeypox.

Studies suggest that people vaccinated against smallpox have about 85% chance of being protected from monkeypox and that is because smallpox and monkeypox are closely related.

Every Tuesday of the month, Ere would go to the FMC for drugs and gradually, he started healing.

“I took it for the entire one month and I also had other antibiotics I was taking too,” Ere said.

He finally regained himself in early June when his drugs finished.

“Rotifa told me there was no need to keep coming.”

Out of pocket spending

For the entire one-month period he took his medication at FMC, Ere spent N10,000 daily because he had a drug he was taking every 2 hours which cost N2,500 per dose.

While it was easy for Ere to access healthcare due to the financial support from his family and friends, many symptomatic patients in Nigeria cannot afford to pay their bills.

“They fail to come for diagnosis and treatments,” said Oyaba Diemebonso, disease surveillance and notification officer in charge of Yenagoa LGA.

According to Diemebonso, treatment for Monkeypox, like other infectious diseases, which should be free is not in Bayelsa State and this is because the state government has not taken up that responsibility of paying bills for patients.

When the monkeypox virus broke out in 2017, the state government provided free treatment and that encouraged more positive patients to go for treatment. But now, people have

present themselves, even when they are symptomatic.

“There ought to be a designated treatment center where patients will not have to pay treatment, unlike the one we have at the Niger Delta University Teaching Hospital, O where patients are made to pay,” Diemebonso said.

Difficulty in accessing treatment

He explained that the funding for Monkeypox ought to be tripartite, coming from the state, and local governments. While the federal government is doing its best in terms making funds available, the state and local governments are not doing enough.

Diemebonso and other disease surveillance and notification officers in the state have trying to improve surveillance and response as well as raise awareness about the mor virus and how to access treatment across Bayelsa State communities. With little fund support, there is only little they can achieve.

“How can we possibly do our surveillance across communities, send messages and do when we are not provided with enough financial support?” Diemebonso asked.

He said that a letter signed by all DSNOs, pointing out their challenges and demands written to the commissioner for health in the state. But nothing has been done about

He also said that the deputy governor had instructed Local government chairmen in t to give DSNOs N20,00 monthly as a stipend. But since March when the instruction wa some LGAs have refused to do that.

Challenges in tracing, diagnosis and treatment

Dr. Ezeudu said apart from lack of adequate funding, another major challenge associat Monkeypox is that of stigmatization, which often makes it difficult for victims to pres themselves at the hospital.

“Victims of Monkeypox often have big rashes all over their body, hence they resort to management and that increases the risk of further spread of the disease which is high infectious, “she said.

She however suggested that the best way to deal with the challenge of stigmatization for improvement in the sensitization and education of people on the need to always present themselves for treatment.

She explained that another challenge lies with the fact that although laboratories exist in the country, not all of them have the capacity for Monkeypox diagnosis which is spec

“Most often, we take our samples to the National Reference Laboratory in Abuja where they can be diagnosed and that takes time because we must transport the samples and wait for the results to be checked and produced,” she said.

She said that people must avoid contact with wild animals, especially monkeys and other animals known to be sources of monkeypox virus include apes and a variety of rodents (including mice, squirrels, and prairie dogs) and rabbits as well as avoid eating any meat from such animals.

She further said that there was a need for the government to improve the capacity of laboratories across the country to aid diagnosis to save time.

On the challenge of funding, she said that the various governments both at the state and federal level might appear not to be providing enough funding for the treatment of Monkeypox cases because they have been engaged more in combating the COVID-19 pandemic and Ebola Virus Disease which have been more ravaging.

Diemebonso said despite the challenges, DNSOs across the country are working hard to ensure that responses are given to all epidemic-prone diseases.

“In Bayelsa state, we have the manpower to handle monkeypox. All that remains is sensitization,” he said.

Ere is well now and wants to help in creating awareness about Monkeypox and the need for people to always present themselves at the hospital when they begin to notice symptoms.

“Whenever I see anyone with the symptoms, I will be willing to take them to the hospital,” he said. “There is nothing to be afraid of because the virus is treatable, and we need to let people know about this,” he said.

This story was produced as part of the National Press Foundation’s Covering Rare Diseases Fellowship



Meeri N. KIM

USA



Meeri N. Kim, Ph.D. is a freelance science and health journalist based in Pasadena, CA. She contributes regularly to a number of news publications including The Washington Post, Inside Science, CURE Magazine, and Optics & Photonics News. Her work has also appeared in VICE's Tonic, HuffPo, The Philadelphia Inquirer, and Slate.com. Meeri enjoys writing about the intersection of technology and medicine. She has written about the use of virtual reality for pain

management, people with diabetes hacking their own insulin pumps, and how virtual assistants can help the elderly feel less lonely.

Meeri received her Ph.D. in physics with a dissertation focus on biomedical optics and neurology from the University of Pennsylvania in 2013. In her free time, she enjoys hiking with her dog Zeus, exploring the city on her bike, and eating dessert.

With CRISPR gene editing, unique treatments begin to take off for rare diseases

Meeri Kim

Paddy Doherty remembers his father as a proud, hard-working family man who stayed physically fit for most of his life. A career in construction and various home improvement projects kept him active until his 60s, when Doherty first caught glimpses of a worrying decline in his dad's health.

"I noticed him getting breathless on walks. He'd stop for a while and maybe make an excuse for stopping, saying, 'Oh, isn't that a lovely tree' or whatever," said Doherty, who lives in Ireland. Doctors chalked it up to angina, or chest pain caused by reduced blood flow to the heart, symptomatic of an underlying heart problem.

But two years later, when Doherty's father died of a sudden heart attack, the true cause was discovered: a rare disease called [transthyretin \(ATTR\) amyloidosis](#), characterized by a misfolded protein that builds up in the heart and interferes with normal function.

"Patients left untreated with this type of amyloidosis develop heart failure, low blood pressure, horrible bowel disturbance, and eventually become incontinent of urine and feces," said Julian Gillmore, nephrologist and head of the National Amyloidosis Centre at University College London. "It's a truly awful, gradually progressive disease that is ultimately fatal."

In February last year, Doherty — now about age 65 — began to experience the same early breathing symptoms his father had had. As an avid hiker who has trekked the Himalayas, he was surprised to find himself getting winded on local hill walks. Testing confirmed that Doherty had a hereditary form of ATTR amyloidosis.

But there was one bit of good news: If Doherty had been diagnosed even a year earlier, no treatment options would have been available to him — an all-too-common situation for over 30 million U.S. patients with rare diseases. But Gillmore, Doherty's doctor, offered him the chance to participate in an early stage clinical trial using [CRISPR, a groundbreaking genome editing therapy](#) with the potential to cure his ATTR amyloidosis in a single dose.

Groundbreaking therapy

"I had no side effects and left the facility after two days," Doherty said. "The walk that I felt breathless on, which is a steep kind of mountain walk through a forest, I'm doing that every Sunday now." CRISPR-Cas9, whose creators were awarded the [2020 Nobel Prize in chemistry](#) a mere eight years after its discovery, allows researchers to alter the DNA of living things at will. It works like genetic scissors that can insert, repair or edit individual genes to rewrite the code of life. The system itself consists of two molecules — a protein known as Cas9 that works like scissors and a guide RNA that takes Cas9 to the right place in the genome — that can be inserted into cells or the bloodstream.

"Broadly, there are more than 7,000 rare diseases impacting over 30 million Americans. Ten or 15 years ago, the technologies to effect change [in terms of treatment] were just not available," said Rich Horgan, founder and president of [Cure Rare Disease](#), a Boston-based nonprofit group.

“But the onset of CRISPR has shown that gene editing has incredible promise, and I think we’re just scratching the surface of what CRISPR will unlock for society.”

Studies to treat a variety of rare conditions in human patients with CRISPR are ongoing, but initial results appear promising.

Promising results

In June, Vertex Pharmaceuticals and CRISPR Therapeutics reported the [latest data](#) from small clinical trials testing CRISPR therapy for two rare inherited blood disorders: [sickle cell disease](#) and [beta-thalassemia](#). Sickle cell disease, which disproportionately affects Black Americans, is caused by genetic mutations that turn red blood cells from flexible discs into rigid sickles that can block blood flow and cause sudden painful crises. Patients with beta-thalassemia have reduced production of hemoglobin, and patients often require regular blood transfusions.

For both diseases, CTX001 involves removing blood stem cells from patients’ bone marrow, turning on a gene that produces usable hemoglobin and infusing the cells back in. After the therapy, the researchers saw marked improvements in both disease groups for anywhere from three to 26 months of follow-up. All 15 patients with beta-thalassemia, who were previously dependent on blood transfusions, no longer needed them. Similarly, the seven patients with sickle cell disease had no sudden painful crises because of blocked blood flow since the treatment.

“We don’t have the final results of this clinical trial, but so far, it seems to be quite successful,” said Mario Amendola, group leader at Généthon, a French nonprofit research institute dedicated to the development of gene therapy treatments for rare diseases. He was not involved in the study. “The idea is that they use CRISPR-Cas9 not to correct the malfunctioning gene, but to [switch on] a gene that can functionally replace the missing one.”

In the case of the [clinical trial on patients with ATTR amyloidosis](#) Gillmore and his colleagues aimed to not only edit the malfunctioning gene itself but also demonstrate for the first time that direct infusion of CRISPR molecules into the bloodstream is safe and effective.

The hereditary form of ATTR amyloidosis affects [roughly 50,000 people worldwide](#) — with a large cluster of patients like Doherty with roots in Donegal County, Ireland. A mutation in the gene that provides instructions for producing the transthyretin protein causes it to misfold and deposit in the body’s tissue and organs, disrupting normal function.

Because circulating transthyretin is made almost entirely in the liver — and everything that enters the bloodstream is carried to the liver to metabolize — the researchers realized they could simply inject patients with the CRISPR-based therapy.

The therapy, called NTLA-2001 and developed by Intellia Therapeutics, appeared to knock out the mutated gene as intended. Only six patients were tested in total, but the three who received the higher of two doses — including Doherty — saw their transthyretin levels drop by an average of 87 percent after 28 days.

The results remain preliminary, and several more patients will need to be tested before the trial is complete.

“It’s important to emphasize that this is very early data, but at the moment, things look incredibly promising,” Gillmore said. “What we will hopefully see over the next five to 10 years is the ability to deliver a therapeutic to not just the liver, but whatever cell you want.”

Patients with other kinds of rare diseases would need a CRISPR-based drug that distributes itself throughout the entire body.

For example, Horgan founded Cure Rare Disease to develop just this kind of treatment for his younger brother, Terry, who has [Duchenne muscular dystrophy](#), a progressive, muscle-wasting disease. At 26, Terry requires round-the-clock care from his parents to do basic physical tasks such as showering and using the bathroom. His condition stems from a mutation in a gene that provides instructions for making dystrophin, a protein primarily in muscles used for movement and cardiac muscle.

Although FDA-approved therapies exist for some patients with Duchenne muscular dystrophy, Terry's particular mutation makes him ineligible for those drugs. Few individuals with the disease live past their 30s.

"I wondered if there was anything in clinical development to treat this mutation, and the answer was no, which really scared me," Horgan said.

Recent research that created a tailor-made drug within less than a year for a 6-year-old girl with a rare, fatal neurodegenerative disorder inspired him to attempt the same feat for Duchenne. He rallied a group of gene therapy scientists to work on a CRISPR-based therapy for Terry's specific mutation.

The hope is that CRISPR will then boost production of dystrophin in his muscles. "We'll dose Terry in late winter or early spring," Horgan said. "We really look forward to that being our first proof-of-concept for subsequent patients that we'll dose over the coming years."

Many other strategies for tackling rare diseases with CRISPR are in even earlier stages of research. For instance, Amendola and colleagues are taking somewhat of an opposite approach as Horgan's team. Instead of creating a drug based on a single patient's disease, they are developing a modular treatment that could theoretically treat many different conditions.

7,000 rare diseases

"There are about 7,000 rare diseases, and for each disease, there can be different mutations," Amendola said. "So if you want to correct every mutation, it will probably take forever. Our idea is to find some kind of strategy which could be adapted — not for all of them, because that's impossible, and they are too different — but at least for a family of diseases."

"We would like to use this as a plug-and-play platform for many different diseases, so that we don't have to re-optimize the whole system every time," he said. "We just need to change it a small bit, which hopefully can speed up the drug development process."

But taking the CRISPR route for rare diseases has its own set of [risks and challenges](#). One possible issue includes minimizing off-target effects that occur when the technology acts on unintended parts of the genome. Also, researchers need to make sure the CRISPR molecules stay in the part of the body that needs treatment. While longer follow-up is still required, subjects in the clinical trials mentioned have not experienced any unexpected harmful side effects so far.

Lastly, the costs of CRISPR-based therapies remain exorbitant at the moment, with price tags exceeding \$1 million per treatment.

"For us to be able to continue gene therapy and all that it's unlocking, it'll be very important for there to be good conversations around reimbursement of these drugs — not just for customized therapeutics, but for gene therapies as a whole," Horgan said. "It's very expensive to make them

right now.”

Doherty said he hopes his family members and fellow Donegal residents will be able to benefit from CRISPR as much as he has. Fortunately, testing shows his two daughters did not inherit ATTR amyloidosis. And along with his father, Paddy’s uncle and cousin both died of the disease.

“When the trial is over, I hope that CRISPR is available and affordable for all amyloidosis patients,” Doherty said. “If a pharmaceutical company can mass-produce something like that and sell it at a good price, it would be a godsend.”

Better Days Ahead for Patients With AML

October 20, 2021

[Meeri Kim](#)

CURE, Hematology Special Issue (October) 2021, Volume 1, Issue 1



An abundance of clinical trials is giving patients with acute myeloid leukemia (AML) hope for a better future.

As an almost 30-year veteran of the Secret Service, Jim Helminski took pride in maintaining his physical health – both for his own sake and to be better fit to protect others. His demanding career placed the safety of Presidents Bill Clinton and George W. Bush and then-Vice President Joe Biden in his hands.

In late 2015, he retired as deputy assistant director to live a more tranquil life on Orcas Island, Washington, with his wife, Teresa Patrick, a former Department of Justice attorney. Not one to sit still, Helminski started a security consulting business on the side while running, weightlifting and practicing karate. He even earned his private pilot's license and began flying a vintage Cessna.



During a routine yearly checkup in 2019, his primary care doctor remarked that Helminski was in better shape at 61 than most 30-year-olds who came into the office. But later, that changed.

"I went for a jog one day that week after my exam, and my cell phone rings. It was my doctor, and he said, 'Jim, there's something wrong with your blood. Your white blood cell count is dangerously low,'" Helminski, now 63, recalls. "I had no symptoms. I felt nothing."

The direness of the situation hit him when he arrived at a Seattle-based health facility for a consultation with a hematologist that ended with a bone marrow biopsy. A few days later, he received a diagnosis of myelodysplastic syndrome (MDS), a type of cancer in which immature blood cells in the bone marrow do not mature or become healthy blood

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AML, which starts in the bone marrow, usually moves quickly into the blood. From there it can spread to other parts of the body, including the lymph nodes, liver, spleen, brain and spinal cord. Typically, AML develops from the malignant transformation of cells that would turn into white blood cells, but it also may start in very immature forms of red blood cells or cells that make platelets. Approximately 19,940 new cases of AML were diagnosed last year in the U.S., with most occurring in adults.

"It was a huge shock," Helminski says. "I scheduled an appointment at the (regional) cancer center to see one of their top leukemia doctors, and it became the lowest point in my life."



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JIM HELMINSKI
received a diagnosis of myelodysplastic syndrome, which quickly progressed into AML, after he retired following almost 30 years with the Secret Service.

Jim Helminski received a diagnosis of myelodysplastic syndrome, which quickly progressed into AML, after he retired following almost 30 years with the Secret Service.

The reviewing oncologist advised him that standard treatment gave him only a 10% to 20% chance of remission and provided little guidance about which therapy to pursue. Instead, he simply recommended that, rather than attempting standard chemotherapy, Helminski and his wife look online to find a list of clinical trials for AML and pick one.

Understandably, they left the appointment dissatisfied and began looking elsewhere for guidance. Meanwhile, Helminski broke the bad news to family and friends, including his flight instructor, whose daughter happened to be an oncologist at the University of California, San Diego. She shared his patient profile with a colleague at Johns Hopkins Medicine in Baltimore, who in turn recommended that Helminski consider a clinical trial helmed by her friend, Dr. Courtney DiNardo, an associate professor of leukemia in the division of cancer medicine at The

an oral medication approved for adults 75 years and older or adults who cannot tolerate chemotherapy; and azacitidine. After an extensive new-patient visit, he was accepted into the clinical trial as one of 48 participants.

Similarly, Irma Smith saw her doctor for isolated pain in her toe and received a diagnosis of AML in 2016. The 75-year-old lived in Fort Wayne, Indiana, with no major health problems and had worked as a real estate agent for the past 29 years.

"I didn't have a clue. I felt great. I went into shock when I heard the diagnosis because I thought, 'How can somebody feel so good and then get hit with AML?'" Smith, now 80, says. "The doctor gave me two weeks to two months to live."

Smith and her daughter decided to seek a second opinion from Dr. Hamid Sayar, a professor of clinical medicine at Indiana University School of Medicine in Indianapolis. Instead of painting a bleak picture, he went straight to work by putting Smith on induction chemotherapy followed by consolidation chemotherapy. After successfully achieving remission in early 2017, Smith was entered into a clinical trial for oral azacitidine as maintenance therapy to prevent relapse.

In September 2020, oral azacitidine was approved by the Food and Drug Administration (FDA) for patients aged 55 years and older with AML who achieve remission after chemotherapy and are not able to complete intensive curative therapy with a stem cell transplant.

PAVING THE WAY

After decades of stagnation, progress in AML treatments has experienced a resurgence in recent years due to rapid advances in genetics, understanding of molecular mechanisms and development of novel therapeutics. Since 2017, nine new drug approvals by the FDA have significantly changed the treatment landscape of the disease. As a next step, clinical trials such as DiNardo's aim to find which combinations of therapies will offer patients the best outcomes.

"AML is still, unfortunately, a very life-threatening cancer. Cancer is clever — it's going to figure out a resistance mechanism to evade a single agent," DiNardo says. "Putting agents together, if you don't have overlapping toxicity, is just a smarter way of giving cancer therapy. So we're trying to move these drug combinations into the frontline setting where they have the best chance of eradicating all disease and preventing relapses."

Other studies focus on improving treatment for elderly individuals with AML, given that the average age at receiving a diagnosis is 68.

Researchers are also testing new targeted therapies. immunotherapies

“AML is more a disease of older populations. Historically, one challenge in the treatment of older adults has been exposing them to intense therapies, which we can do for the younger patients,” Sayar explains. “But treatment of AML at any age, at any phase of the disease, is a challenge. There is an unmet need at every aspect of treatment.”

REFINING AND PERSONALIZING THERAPY

The approval of more therapies has certainly helped many patients, and researchers such as DiNardo are looking to optimize their administration even more by finding the most effective combinations and timings. The clinical trial that Helminski participated in brought together three approved agents for the first time in the frontline setting for patients who have AML with an IDH1 mutation.

Helminski underwent a 28-day isolation in his hospital room at MD Anderson while receiving the three medications due to his immunocompromised state from the leukemia and the treatment. It kept him as infection-free as possible until his immune system recovered. He remained as active as he could – using an exercise bike daily, strength training with resistance bands and meditating with the help of a smartphone app – but did experience some setbacks, such as pneumonia and minor liver inflammation attributed to an antifungal drug.

On the 28th day, the results of Helminski’s follow-up bone marrow biopsy showed that he went from 40% leukemic myeloblasts – immature blood cells that serve as a marker of AML progression – to just 1%.



Irma Smith was treated with induction chemotherapy followed by consolidation chemotherapy.

“He’s a great example of someone who went into a really deep remission. He’s been leukemia-free and doing great for over a year now,” DiNardo says. “We have all of these new approvals now, and they were approved in the single-agent setting. But that’s probably not the best way to actually use them in the real world.”

administered a placebo as part of the control group instead of the maintenance therapy. She is currently being treated indefinitely with Venclexta (venetoclax) and decitabine, a chemotherapy drug, every six weeks – which temporarily takes a toll on her body.

“I don’t want to go out or see anybody. I’m pretty miserable, but I know it’s not going to last,” Smith says. She still enjoys maintaining her house, going on long walks with her dog and spending quality time with family: “The side effects stop in a week and a half, and the rest of the time, I’m just fine. I have a lot of energy.”

Other studies aim to help patients like Smith who relapse after undergoing therapy. Sometimes another round of chemotherapy can put the leukemia into remission again, but it is not likely to be long-lasting. A stem cell transplant or newer targeted therapy for a specific genetic mutation could be better options, but with much more toxicities, and patients must be eligible for these therapies.

“Unfortunately, AML still is a disease where the leukemia does recur or come back, so a lot of research is focused on treating patients who have relapsed after upfront therapy,” Dr. Sangmin Lee, an assistant professor of medicine at Weill Cornell Medicine in New York City, says.

“Several ongoing clinical trials are geared toward relapsed and refractory settings, such as those investigating cell-based therapies, targeted therapies and drugs that overcome resistance.”

For example, a number of clinical trials are exploring the use of chimeric antigen receptor (CAR)-T cell therapy, a novel treatment that involves engineering a patient’s own immune cells, for AML. CAR-T cell therapy has shown promise in other blood cancers. Research is still in the early stages for AML, with initial studies looking at the safety and feasibility of the therapy in adults and children.

Overall, experts agree that several avenues are being explored by researchers to help patients with AML, who are recommended to look into clinical trials as soon as they receive their diagnosis. Helminski, for example, emphasized that he would not have been eligible for DiNardo’s study if he had undergone standard therapy first.

“There is a lot of research trying to see if novel therapies provide benefit, so patients should be on the lookout for clinical trials – both in an upfront setting and also in the relapsed or refractory setting – when they (receive a diagnosis of) leukemia,” Lee says.



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After going into remission in 2017, SMITH enrolled in a clinical trial for oral azacitidine as maintenance therapy to prevent disease recurrence.

After going into remission in 2017, Smith enrolled in a clinical trial for oral azacitidine as maintenance therapy to prevent disease recurrence.

Since the trio of medications Helminski received worked well enough to induce a deep remission, he was able to undergo a curative stem cell transplant in February 2020 from an unrelated donor.

After the stem cell transplant, he gradually regained his strength over the course of 100 days. Today, at more than 500 days post-transplant, Helminski shows no signs of measurable residual disease. He is back to having a full life on Orcas Island with his wife, exercising regularly, flying his airplane and building furniture.

“I initially believed that I was terminally ill, and there was no hope for me. And there’s always the monster of a possibility of a recurrence,” Helminski says. “But I take life one day at a time, and I’m very appreciative of every day that I have.”



Anna LARSSON

Sweden



Anna Larsson has studied journalism at the University of Gothenburg. She also studied medicine at Karolinska Institutet in Stockholm for three years. She is employed as medical reporter at Swedish National Radio. Her duties are mainly to cover news in the field of medicine and health care. She has published radio documentaries, reportages and articles on the website of Swedish Radio. 1999 came her first book about caring for elderly relatives (partly documentary) - 'Med varmt hjärta och vässade klor'. 2009 she

was rewarded the Söderbergiska journalistpriset for integrity, knowledge and high quality in journalism. She also got awarded for reportage on children's cancer 2018. She is a pHD student at Karolinska institutet with the subject 'Medicine and the Media', now preparing for disputation. 2020-2021 most of her working time at the newsdesk has been dedicated to the effects and future of the corona pandemic.

Anna Larsson lives in Stockholm.

Rare diseases- publication

Morning show (P1Morgon) 29 nov 2021, two audio pieces:

<https://sverigesradio.se/artikel/sallsynta-sjukdomar-genanalys-kan-ge-nya-diagnoser-at-fler>

Many people with very rare diseases have missed both diagnose and treatment for a long time.

The new genomic techniques have now made it possible to make diagnose on close to 2000 children and adults in Sweden, who to date have not had any explanation of their symptoms.

Celeste, 6 years, is one of those children and her mother Jenny-Li Örsell tells about the daughter's first years in life, and how important it is to get a diagnose.

On the same link, a bit further down: Listen also to Oskar Ahlberg, board member of the patient organization Riksförbundet sällsynta diagnoser, an umbrella organization for different patient associations. One of his children have the diagnose MPS, which consists of a group of different diagnoses of serious diseases.

News (Morgonekot) 29 nov 2021:

<https://sverigesradio.se/artikel/genanalys-kan-ge-nya-diagnoser-at-fler>

New genomic technique have given 1000-2000 persons in Sweden a new genetic diagnosis and brought an explanation to their unclear symptoms.

The project Genomic Medicine Sweden is an example of how to use precision medicine to clarify diagnoses and give tailor-made treatment to the patients.

The technique is however uneven distributed in Sweden and hitherto the most genomic analyses are still being made in Stockholm. The patient organization Riksförbundet Sällsynta diagnoser claim a more equal application of diagnosis and treatment in the country.

News (Dagens Eko) 5 dec 2021:

<https://sverigesradio.se/artikel/samhallet-vill-dela-kostnader-for-dyr-behandling>

New and very expensive drugs against rare diseases are underway in the pipelines of the pharmaceutical industries, but since the official prices are around 100 000 dollars or more, they are unlikely to reach everyone in need in our country.

The advisory board for new drug therapies (which are negotiating prices with the industry) have started discussions about new ways of financing the new treatments.

Sharing cost responsibility, meaning that full price only is payed if the patient achieves good effect of the therapies are discussed but will take time to reach.

Summary

For people suffering from unclear symptoms and disabilities, a new and explaining diagnose could make a big difference. The new genomics has during the last years brought diagnoses to close to 2 000 persons in Sweden, which makes new therapies getting a bit closer.

One of the patients is Celeste, six years, who after a long wait got a very rare diagnose. Her mother Jenny-Li recalls the first months of her life.

--She was always such a happy baby with a bubbly laughter.

But the development of Celeste was slower than expected. She spoke her first word 'Mama' twice, but then she remained silent.

--I really thought that this was the only word she needed, at least in my opinion, Jenny-Li remembered and smiles.

Still today, by the age of six 'Mama' is the only word Celeste can pronounce.

Her communication now is based on sign language and she manages around 200 signs, Jenny-Li explains.

She also uses a sort of I-pad with speech synthesis, which works very well. The I-pad 'talks' on command, and can pronounce the sentences that Celeste chooses, for example:

"-My name is Celeste. Do you want to play?"

But it has been a difficult time for her parents. The child care center ignored their worries and it took several years before they got the diagnose – a combination of letters: SATB2, showing which gene in the DNA that is affected. Globally only 500 persons shares the same mutation. So Celeste is a very rare person. Her problem consists of intellectual disability, speech problems, weak skeleton and not fully developed fine motor skills.

It took several genome analyses of both Celeste and her parents to finally get her diagnose through the GMS project – Genomic Medicine Sweden – which started five years ago. The goal is to develop the concept of precision medicine and to use the new knowledge for tailor-made therapies to individual patients. Professor Anna Lindstrand at Karolinska Institutet works with rare diseases at GMS, and she has big hopes for the future.

--Today there are few cases where you have an individual therapy, around five percent of all rare diagnoses. But many new treatments are underway so in the future you may find the cause of the disease and an effective treatment. Maybe not curable, but something that facilitates life a lot.

So what happens in society when a lot more patients get diagnoses?

Oskar Ahlberg has a son with a rare diagnose called MPS and he is also member of the board of the patient organization Riksförbundet Sällsynta diagnoser. He says that he recognizes the story of Celeste, and all the questions and worries that many parents experiences.

How did you react?

--I recognized this story very well – Before you are worried and then you get a diagnose and you are suddenly more afraid of being excluded from many situations in society and possibly looked upon as very strange. But when the first shock is over there are possibilities. There might be treatments, assistance, support and aid. There are possibilities to get a better life for your child.

--Many tells us that now that I have got the diagnose it is positive but it is also still difficult to explain to authorities what kind of disease this is.

--There are also problems with upcoming new drugs that are so costly that it will be almost impossible to pay for.

-- I really think it is a big help to get a diagnose, you can ask for aid and support. But still many persons have their unclear symptoms but no diagnose and we must not forget them.

Who will pay for the new treatments that lies in the pipelines of big pharma?

Gene therapies, immunotherapies and newly developed drugs – many ways of treating rare diseases are on their way to the market. Some of them can eventually cure a number of those diseases, but there are no guaranties for an effect that is good enough. There is a great need for new ways to finance new drugs. One solution could be that only pay the full price if the patient really gets better.

--If society should subsidize new and expensive therapies, when you do not know that they will work in the long term, then it will be necessary to share the economic risks, says Gerd Lärfars, who is president of the advisory board on drugs of the county councils in Sweden – NT-rådet (=Rådet för nya terapier).

-We want to make sure that if we are going to subsidize a therapy, we will have to know that it gives the expected effect, she says. And I am quite convinced that we will reach an agreement on this.

One of the problems with the area of drug development and rare diseases is that there are seldom enough number of patients to do a randomized controlled study, to get a scientific proof of the effect. So therefore lots of price negotiations are going on between health authorities and manufacturers. One example is a gene therapy for a spinal muscle atrophy in children, which might be the most expensive treatment hitherto, around 2 000 000 dollars officially. Untreated the disease often leads to death in early childhood. 2020 it got approval from the European Medical Agency, and is now used in United Kingdom and Norway. Negotiations were still going on in Sweden in November 2021.

Anna Larsson

Swedish National Radio

News desk



Mactilda MBENYWE

Kenya



Mactilda Mbenywe is a Health and Science Journalist at The Standard Group a Mainstream Media House in Kenya, for the past four years, Mactilda has transformed Health Reporting in Kenya. Apart from her passion for health, she has been a key pillar in reporting Gender issues, especially issues touching on the vulnerable groups in society.

Mactilda has also worked at Nation Media Group another Mainstream Media house in East Africa.

She is a member of The Association of Media Women in Kenya (AMWIK) and The Media for Environment, Science, Health and Agriculture (MESHA) she holds Honors in Journalism Studies from Maseno University and is currently a Masters Degree Student in Communication Studies at the same university.

My son sneezed, and just like that, broke his right hand

Mactilda Mbenywe

Seven-year-old George Ouma groans in pain as he tries to lift his heavily bandaged left leg.

He sustained a fracture in the arm and leg two weeks ago, confining him to a bed as he recovers.

“He has had over 30 fractures, with an equal number of operations in which surgeons have tried to fix his broken bones while avoiding implants due to his tender age,” explains his mother, Diana Akumu, as she stares at his frame which is smaller compared to his peers.

As a child, Ouma was healthy, albeit dull and slow, until trouble started when he turned two.

“One day he was sitting and stretched his leg to stand up and just like that, he broke a hip bone,” recalls Akumu, who took the boy to the Kenyatta National Hospital (KNH) where doctors found that the bone had snapped.

A few tests later, the doctors discovered that Ouma suffered from Brittle Bone Disease, also known as Osteogenesis Imperfecta (OI). The condition makes bones weak and susceptible to breaking. As a result, Ouma has had spent much of his time in hospital.

Three months after the first fracture he suffered yet another in the upper limb, again after stretching it.

“I feel so bad for him especially because of the accompanying pain. One time Ouma broke his right hand just by sneezing,” says his mother.

Vincent Owaa, an orthopedic surgeon at Jaramogi Oginga Odinga Teaching and Referral Hospital (JOOTRH), says that OI is a rare genetic disease which occurs due to lack of high-quality collagen, the protein which forms the framework for the bones.

He explains that without collagen, bones, including teeth and cartilage, can lack flexibility and strength, making them susceptible to fractures.

The disorder, which is incurable, disrupts the body’s ability to form strong connective tissue and to grow new bone tissue.

OI is a lifelong condition, and those affected by it experience physical pain due to frequent fractures.

“The bone weakness sometimes worsens and as the bones weaken, they become progressively crooked, causing deformed limbs and difficulties in walking or activity,” Owaa explains.

For young Ouma, his hands and legs have visible deformities. He cannot stand or do much on his own.

“He needs someone round the clock to support him with everything. I live in constant anxiety at the thought of any slight move leading to a fracture in one of his bones,” laments Akumu. “It breaks my heart seeing him in pain. It means he cannot play or make friends like boys of his age.”

There are drugs that strengthen the bones, but nothing can completely cure the condition. Those affected are advised to adopt a healthy lifestyle to maintain bone health.

“It is important for patients to get adequate calcium intake and enough exercise to maintain healthy body weight. Being overweight can worsen the condition,” says Owaa.

Surgeries are often required to put metal implants in the bones or spine to prevent fractures. Being a genetic disorder means taking down a family’s history when treating children in addition to the physical examination.

X-Rays allow a doctor to see current and past broken bones besides identifying defective bones.

The AIC Kijabe hospital receives an average of 10 to 15 patients with Brittle Bone Disease monthly and most are aged between one year and 18 years, says Dr Federico Sibona, a pediatric orthopedic surgeon at the hospital where some patients report up to 40 fractures a year.

“There is no cure for brittle bone disease,” says Dr Sibona, “but treatment can relieve symptoms, prevent breakage of bones, and maximise movement. Severe forms of the disease can affect the shape of the rib cage and spine, which can lead to life-threatening breathing problems.”

Health experts warn that the cost of treatment can be exorbitant with Owaa saying “a patient needs an average of Sh500,000 to restore a hip bone when fractured.”

Dr Sibona, on his part, says some patients need blood transfusion “and a single surgery on a broken bone costs about Sh90,000 to Sh130,000.”

However, there is new drug and telescopic nail (standard treatment for stabilising long bones and correcting deformities in Brittle Bone Disease). Both “will reduce pain with a reduced number of surgeries after frequent breaking of bones,” reassures Dr Sibona.

Bernard Ndemo, an occupational therapist at Kisumu County Hospital, says that lab tests are used to analyse the structure of children’s collagen and “the management course for OI in children is determined by their age, overall health and medical history.”

Ndemo explains that the ultimate goal of treatment is to prevent deformities and fractures and to allow the child to function as independently as possible but “there may be use of wheelchairs braces, and other custom-made equipment which could maximise the individual’s functional ability.”

For Akumu and Ouma, pain and anxiety are not the only thing that worries them as “there is a lot of stigma, and many stay away from us.”

But for now, she takes her son’s challenge a day at a time, hoping that medical interventions and constant prayers will cure her only child.

“I have hope that one day he will get help and attend school like other children.”

Wiping open defecation cut cholera, created jobs

 standardmedia.co.ke/health/health-science/article/2001431070/wiping-open-defecation-cut-cholera-created-jobs



A SATO toilet bowl in Bar Agulu village, Siaya County. [Denish Ochieng, Standard]

Mary Achieng walks shoulder-high at her home in Baragulu village, Siaya County. Her family of seven has not suffered any sanitation-related illnesses in five months thanks to a pan placed on her pit latrine.

The Safe Toilets (SATO) are mechanical with air-tight seals that close off latrines and thus keeping away insects which spread diseases after coming into contact with human waste.

Use of SATO pans helped reduce open defecation and has enhanced sanitation levels in Siaya, Homa Bay and counties in Nyanza where previously “we had frequent diarrhea but since all has been well,” says Achieng.

Besides sanitation, the toilets have come in handy for James Odhiambo who was disabled during the 2007 post- election violence from which he has undergone several surgeries on his hip joint, back and he also experiences numbness below the knee of his right leg.

A SATO stool has made life easier as going to the toilet is no longer a problem as “it is clean and serves me well.”

The sanitation project has been a joint effort between UNICEF and the county governments with Siaya achieving Open Defecation Free status in 2018 and has since launched market-based sanitation activities to upgrade sanitation which includes construction of low-cost improved sanitation systems installed in markets, schools, public toilets and churches.

Siaya County has installed 24,115 latrines –enabling more than 120,000 people to access basic sanitation courtesy of UNICEF accelerating the elimination of Open Defecation and facilitating access to basic sanitation in communities and schools.



Use of SATO pans helped reduce open defecation and has enhanced sanitation levels in Nyanza. [Denish Ochieng, Standard]

The sanitation project sees vulnerable households access financing to upgrade their basic sanitation while providing employment and entrepreneurship to members of the community. For instance, some 130 artisans were equipped with skills to build low –cost toilets.

Siaya County had been grappling with at least two incidences of cholera outbreak leading to the loss of lives since 2016, the year 558 cases of cholera left 15 dead with the most affected sub-counties being Alego Usonga, Ugunja and Ugenya.

There were 1,117 cases of diarrhea in December last year down from 2,177 that November 2020.

Now, however, diarrhea cases have been declining since June last year and no single case has been reported this year, according to data from the Siaya Department of Health and Sanitation.

UNICEF is also working to improve sanitation in Homa Bay with the project covering the sub-counties of Rangwe (359 villages), Homa Bay Township (266) and Kasipul (293) and 30 schools with the aim of being Open Defecation Free.

In Kisumu, about 60 percent of villages are open defecation-free after the county began implementing community-led total sanitation strategy which has seen latrine coverage stand at 96.3 percent up from 80.1 percent since 2014, according to Kenya National Bureau of Statistics 2019 Census Report.

The strategy has since stopped recurrent cholera outbreaks in both urban and rural areas via improved clean water supply and safely managed sanitation services.

Only 3.6 percent of households in Kisumu are practicing open defecation and still “85 percent of Open Defecation takes place in 15 counties,” including Homa Bay said Unicef Kisumu chief field office, Wangui Karanja but she hopes the project aims “to eliminate Open Defecation in 15 counties by 2025.”

The county and Unicef introduced Market-based Sanitation (MBS) and Open Defecation that stood at 38 percent in 2015 had been reduced to less than 10 percent, according to Homa Bay County director and Water Sanitation and Hygiene (WASH) Partner Coordinator, Mark Aguanda.

“Three sub-counties Ndiwa, Homa Bay and Kasipul, have become Open Defecation Free zones,” Aguanda said adding that a drive had been launched for building modern latrines with the focus being on schools.

Kenyans abuse antibiotics, painkillers in wet evenings

 standardmedia.co.ke/health/health-science/article/2001429592/kenyans-abuse-antibiotics-painkillers-in-wet-evenings



There is widespread misuse of antibiotics, a trend that has resulted in bacteria that is resistant to medication, a study has shown.

Antibiotics, the first-line miracle drugs, enjoy increasing availability. About 90 per cent of Kenyan households use antibiotics annually, an alarming rate of consumption, according to a study conducted between November and December 2020.

The cross-sectional survey titled, Antimicrobial Resistance Knowledge Among Kenyans, revealed that antibiotics were the most sold product in 58 per cent of Agrovets, and 43 per cent of pharmacies, where sale of antibiotics was only rivaled by painkillers, cough medicine and antiparasitic drugs.



Watch Video At: <https://youtu.be/ykxeKHoIRyM>

Antibiotic sales contributed to 38.8 per cent of total revenues in pharmacies and 30 per cent in Agrovets.

More than two-thirds of human drug stores sold antibiotics without a prescription, compared to veterinary drug stores.

Reasons for increased sales included the perception that antibiotics could treat all diseases, including non-bacterial infections like cold and flu. But of more interest is that Kenyans also reported failure of completing their antibiotic prescriptions and doses, besides sharing and stockpiling of remaining medications.

The misuse and resulting resistance has been a growing concern not just in the country, but globally, according to a report released by World Animal Protection ahead of the World Antimicrobial Awareness Week.

The study titled, Evidence of Superficial Knowledge Regarding Antibiotics, and published in 2017, reveals that the mutant bacteria commingle with other pathogens in sewage canals, hospital wards and livestock pens, and can share their genetic resistance traits, making other micro-organisms impervious to antibiotics.

Farmers were the most frequent consumers of antibiotics, with dairy farmers at 49.6 per cent and poultry ones at 38.9 per cent. Dairy and poultry rearing are associated with intensive farming systems in which farmers overuse and misuse antibiotics to mask poor welfare practices, despite relative knowledge of consequences.

“Inappropriate practices included rampant sale of antibiotics without prescriptions, self-medication practice, failure to complete course of antibiotic therapy, sharing of antibiotics, and stockpiling of remaining antimicrobials were most common,” the study read in part.

The report also notes that veterinary and human pharmacists reported increased antibiotic sales in wet seasons and lowest in the dry seasons, highlighting a seasonal pattern of sales of antibiotics and “majority of antibiotic sales happened in the evening peak hours.”

Dr Victor Yamo, Farming Campaigns Manager, World Animal Protection, said: “Antibiotics were the commonly sold product in human and veterinary stores. This contrasts with the low sales of vaccines and other biosecurity measures,” meaning there were significant gaps in knowledge and practice that needed attention like shifting farmers from low welfare practices in which antibiotics are misused to cover them up to adopting higher animal welfare standards.

Inside Nyanza's fight against sickle cell

standardmedia.co.ke/health/health-science/article/2001428501/inside-nyanzas-fight-against-sickle-cell

[Home](#) / [Health & Science](#)



Lab technologists during routine clinical research at Kombewa Clinical Research centre in Seme Kisumu county. [Collins Oduor, Standard]

Nyanza has one of the highest burdens of sickle cell disease, but patients will soon access specialised care and treatment from the region's largest referral hospital.

Data from the Kisumu County Department of Health shows that about 1,500 children are born with sickle cell disease annually, meaning three to four out of 100 newborn babies have sickle cell disease – with 50 to 90 per cent of them not celebrating their fifth birthday.

But why the high sickle cell burden in Western Kenya?



Watch Video At: <https://youtu.be/2--4BKAg6kQ>

Dr George Rae, the CEO of Jaramogi Oginga Odinga Teaching and Referral Hospital (JOOTRH), says sickle cell disease is a painful, major blood disorder, which in Africa was historically common among Nilotic communities living near rivers and water bodies, where malaria was common.

“Sickle cell was body adaptation to fight malaria in endemic zones, genetically people are born with it,” explains Dr Rae. Dr Bernard Awuonda, a pediatrician at JOOTRH, adds that Nyanza’s high burden of malaria led to sickle cell gradually evolving as a protective mechanism against malaria. However, the sickled red blood cells do not survive for long compared to normal red blood cells.

Rae cited malaria and carbon monoxide (use of charcoal) as the common triggers that precipitate a crisis among sickle cell patients, including stroke, destroying red blood cells used for transporting oxygen and hence low blood levels and a need for transfusion.

Dr Lutz Hegemann, Novartis Group Head of Corporate Affairs and Global Health, says they will consider a combination of approaches to better health delivery, including partnerships with the Kisumu County government.

Patients like Miriam Adhiambo’s four-year-old son, who had been admitted for seven days at the paediatric ward, will be the biggest beneficiary.

“My son was diagnosed with the disease when he was one and a half years old, since then life has been unbearable as cost of treatment and care has been a tall order for my family,” says the 28-year-old.

George Moturi, the Franchise Head – Oncology – at Novartis, acknowledges that sickle cell is endemic in the lake region where there is a prevalence of about 4.5 per cent.

“So JOOTRH is a centre of excellence that we would like to partner with to assess the plight of the patients and families affected by sickle cell disease. We have come together with JOOTRH to improve aspects to do with training and capacity building so that patients coming to the hospital can be taken care of well.”

The JOOTRH pediatric ward admits about 30 patients with different types of sickle cell crisis every week but the cost burden will be eased with the Novartis partnership.

According to Dr Rae, this will not only reduce dependency on supplies from the Kenya Medical Supplies Authority (Kemsa), but also ensure specific drugs are accessed at half price from Novartis, including those for sickle cell, leukemia, multiple sclerosis, malaria, cancer and epilepsy.

He says beyond the partnership, Novartis will carry out researches, which JOOTRH will take part in “towards advancing our pharmaceutical products to improve the treatment of sickle cell disease”, which will also be made easier with the installation of a multimillion-shilling machine to detect and manage sickle cell.

Novartis Country President Racey Muchilwa says, “Screening will help in genetic counselling for parents who could potentially be having sickle cell trait as well as prepare them to take care of their children.”

Through a partnership with American Society of Haematology (ASH), JOOTRH will not only get expertise, training and research to address the burden of sickle cell, but also equipment, including a special apheresis machine, which will diagnose and replace defective red blood cells with normal ones in a therapeutic process.

Rae says the hospital currently does tests for sickle cell using mechanisms that also diagnose other ailments and which thus “do not present accurately the volume of the cells that have been sickled”.

He says many sickled cells pass out as normal and healthy and that is the major reason “we have opted to import a machine, which can detect sickle cells in anybody whether one is a carrier or has the disease.”

Rae says with advanced treatment, interventions can be done to ensure children transition and live productive adult lives.

Screening

“We plan to screen 38,000 newborn babies for sickle cell annually for the next five years,” says Dr Awuonda. “This will help reduce mortality among children born in the region as it’s the epicentre of sickle cell disease in Kenya.”

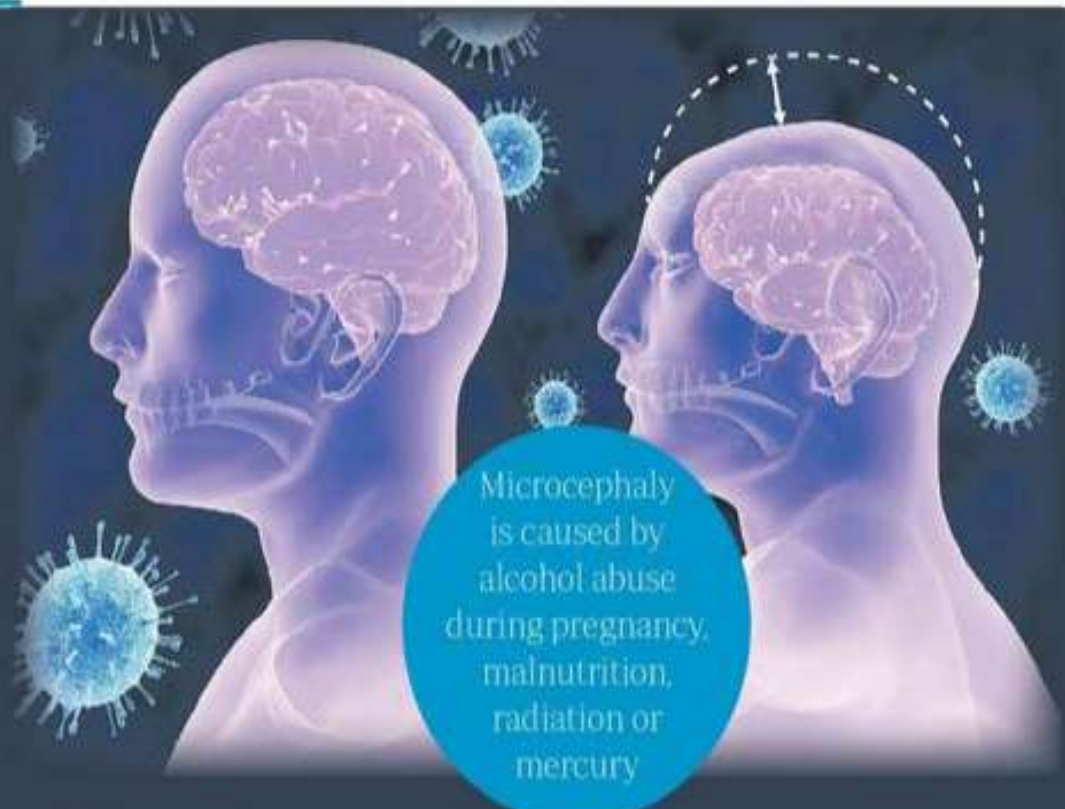
Many patients report to hospital when it’s late and bones destroyed, making rehabilitation a challenge, but Dr Rae adds that “ultimately the centre will be equipped to offer bone marrow transplant services, which will also be useful for haemato-oncological conditions like leukaemia.”

Rae also notes that Norvatis is keen on supporting JOOTRH carry out kidney transplants, hence the establishment of a kidney transplant unit. The partnership with Norvatis will give JOORTH a footing in the region's healthcare sector.

“Such a partnership should enhance evidence-based, people-centred, focused leadership that is result-oriented and promotes cohesion among patients and healthcare providers,” said Kisumu Governor Anyang Nyong'o.

Statistics show that about 30 out of 100 people in Kisumu County are carriers of sickle cell gene. Data from JOOTRH indicates that between 20 and 30 per cent of patients have sickle cell traits, with annual new cases at three per cent.

FLESH & BLOOD



Microcephaly is caused by alcohol abuse during pregnancy, malnutrition, radiation or mercury

Children with extra small head not a curse, witchcraft

By Mactida Mbenywe
Health reporter at Standard Media House

After Dorothy Acheng gave birth to her daughter 15 years ago, she woke up and found nurses staring at her with unusual curiosity.

She had not experienced any complications during pregnancy or when giving birth to Zaida, her second born, but Dorothy got anxious when a neurologist started examining the baby before asking: "Did you know your child developed Microcephaly?" Confused Acheng, replied, "What is Microcephaly?"

More tests revealed that Zaida's head was significantly smaller than expected as her brain had not fully developed.

Dr Lee Ogutha, a consultant neurosurgeon and a lecturer at Maseno University, explains that microcephaly is a rare condition in which a baby's head is smaller than average for that age.

Dr Lee Ogutha says the size of a baby's head is measured and compared to a Head Circumference Chart for boys and a Chart for girls.

A baby's head is measured every month and it's supposed to grow at a certain rate based on the Standard Chart. But the heads of

head is within the normal range, whether too large or too small."

The condition has no cure and in Kenya, there is no documented data on it. However, in the USA it occurs in about two to 12 per 10,000 live births.

Dr Ogutha has attended to three cases of microcephaly at Pediatric Neurosurgery Centre in Nairobi in his career as a neurosurgeon and notes that "some children who have this condition end up with birth defects and congenital effects. They might die before we get to see them in hospital."

Dr Ogutha explains that microcephaly is caused by congenital malformations which can be hereditary like Down syndrome—in which a baby has three copies of chromosomes instead of the usual two copies in all cells, affecting the brain.

Microcephaly can also develop via mother to child infections while the baby is in the uterus. Toxins like mercury, alcohol abuse during pregnancy, malnutrition and exposure to radiation can also cause microcephaly.

In the case of Dorothy, medical records indicate she lacked proper vitamins and nutrients besides alcohol consumption during pregnancy and the reason Zaida suffered from the condition.

Dr Ogutha also adds that microcephaly can



Dorothy Acheng (left) with her daughter Zaida, a microcephaly patient, at their home in Manyatta slums, Kisumu County. [Denish Ochieng, Standard]

The condition has no cure
There is no documented data on microcephaly in Kenya
Microcephaly occurs in about 2 to 12 per 10,000 live births in the USA

whose spread in Brazil in early 2015, also saw a peak in the number. Microcephaly cases from between 3.6 million and 54 million in two years to 2017.

The bombing of Nagasaki and Hiroshima, Japan during World War II led to many being exposed to radiation which also saw increased number of babies born with micro-

cephaly have other kinds of destructive injuries that affect blood vessels such that fetus develop stroke even before and after they are born causing a disruption in the development of the brain.

Whole about 10-12 percent of these babies have normal intelligence, the majority suffer intellectual disability and learning difficulties, speech delays and seizures which are commonly accompanied with abnormal muscle function ending up with trouble walking and balancing.

For Zaida, Dorothy says "she depends on me on everything, she cannot control call of nature, she is now a teenager with normal menses but I have to take care of her."

the condition and the environment in which the child grows, as those whose damage to the brain is not severe can live a long life in a supportive environment compared to those who are not.

Dr Ogutha says since there is no cure, all an expectant mother can do is have good maternal nutrition and "minimize risk by avoiding alcohol during pregnancy and situations that can cause infections to occur during pregnancy."

Support for children with microcephaly include physiotherapy, speech therapy and occupation therapy to help them adapt to activities they can do by maximizing their ability and cost depends on type and class of hospital, but those with NHH cover and on UHC scheme can afford basic treatment in public hospitals.

But for Zaida her mother could not afford treatment and "for the last five years she has not received medical care save for malaria and other common illnesses," says Dorothy.

Another neurologist, Dr Symon Karuki of KEMBO-Welcome Trust observes that stigma in some communities that microcephaly is caused by curses or witchcraft, was parents hiding their children, yet the condition "is a result of accidents that happen or due to congenital malfunctions the public and parents must understand this, it shouldn't be feared nor parents blamed."

In the case of Dorothy, Zaida's father "left us and we had to continue, he blamed me to



Clement MSISKA

Malawi



Clement Msiska is a Social and Governance Journalist working with Capital Radio Malawi since 2016.

He holds a Bachelors Degree in Journalism and Mass communications obtained at Pentecostal Life University (PLU) in 2021 and a Diploma in Journalism obtained at the Malawi Institute of Journalism (MIJ) in 2013.

Before Joining Capital Radio, Clement worked as a communications assistant (internship) for Youth for Development one of the local organisation working towards the

advancement of young people.

Clement also worked as an intern for Matindi Radio for a period of six months, during his time at Matidi Radio, he was assigned to report on governance issues.

Over the years, he has won two awards, one in health (Tuberculosis) reporting courtesy of the World Bank in Malawi (in 2018) and another one on the promotion of human rights courtesy of the United Nations Office on Drugs and Crime UNODC in Malawi (2019)

For social issues Capitol Hill is being challenged to step up its efforts towards eradicating rare diseases other than focusing its attention on a few elements that are common in Malawi and neighboring countries. Malawi has instances of rare tropical diseases like leprosy and ancylostomiasis. As well as non-tropical diseases including hemophilia, a deadly hereditary disease that affects people from a tender age.

From Lilongwe, Malawi, Clement Msiska reports.

On a normal day, that is the jovial sound of children as they play different kind of games. But, for five-year-old Charles, not his real name, such luxury does not come easy. As most times, he is indoors, not by choice, but due to mobility challenges caused by massive hemophilia attack affecting his limbs and also having a breathing disorder. Charles is the last-born son among the three children in the family of Davy and Agnes Paolo, who are living some kilometers outside the capital city Lilongwe.

While the majority of hemophilia cases are passed on from mother side to the child, some cases like that of little Charles are not inherited and there is no family history of the disease due to the genes that are on the X chromosome.

According to his mother Agnes Paolo, they realized that something was wrong with their son when he was just three months old. Agnes is at pains, as she believes the future of her five-year-old son, who has already been forced to withdraw from school, is in tatters.

“The problem started when he was about seven months. I enrolled him at a nearby school, but he would only go for two days because some parts of his body would get swollen and was bleeding. As a result, he wouldn't try it.”

His father, Dave Paolo, a tent and a truck driver, reveals how he and his wife wasted thousands of quarters by visiting places in different parts of the country, in search of solutions to their science problem, but not to avail. At one of the traditional healers' shrines, Mr. and Mrs Powell were told that their son's condition was as a result of witchcraft.

“While we were still thinking about the next move, we thought of visiting a traditional healer. We were told that our son was cured and that he would never get sick again after he was given some helps. But they never will.”

The witch doctor prescribed various concoction for the boy and incisions all over his body, but weeks later his condition moved from bad to worse. On their visit to the hospital, doctor suspected the boy could have been suffering from hemophilia, but we're not certain due to lack of testing coagulation. The couple were referred to the Society of Hemophilia and Allied Disorder unit within the hospital, where Charles was put on factor 8 treatment. While things seem lousy, another challenge arose. The factor 8 treatment requires that the child be injected every day, which meant to the couple should fork out 14,000 Kwacha every week for transportation to and from the hospital, which they can't afford. Chairperson for the Society, who is also a hematologist, Dr. Mombay remains that the unit survives on donations from the World Federation for Hemophilia, WFH, supplies them with 100,000 of Factor 9 units and 300,000 Factor 8 units every three months to be given to patients.

“We've never had any firm commitment for the government to it that they give permission, like when they established the service. The government, you can't just go there and they establish a service. You explain what you want to do and he said to go ahead. But getting the government to commit to buying the 8 factors for these people has been a problem.”

However, Mombay placed down possibilities of negative effects of treating patients based on signs and symptoms due to lack of diagnosis.

“The actual factors that are sought are factor 8 and factor 9. And we don't give them only....Clinical suspicion are those that we are suspecting clinically. We give blood transfusion. With all the factors, we got dressed the issue.”

Meanwhile, chairperson for the Parliamentary Committee on Health, Matthew Zgwai observed that most rare diseases in Malawi are neglected due to the fact that the government is not willing to spend money on expensive medicine and equipment on diseases that affect a handful of people.

“That’s unfortunate. If we look at hemophilia, It’s a difficult situation, but how big is it for when we look at it from the position of Malawi? You might find that it’s a very small area, and then, if we have limited resources. Can we be looking at that sometimes right now?”

20 years ago, Malawi and other African countries agreed to adopt a policy that will ensure nations allocate 15% of their national budgets towards health sector to address the never ending challenges. Up-to-date, Malawi is still crawling towards the same but limited resources with many competing healthcare needs make this rare disease face common problems such as lack of access to correct diagnosis, lack of equipment and lack of quality information on the same. Currently, neighbouring Mozambique is the nearest country where one can get a full diagnosis as estimates indicate that Malawi has about 40 hemophilia and bleeding disorder patients, the majority of whom are children. But the questions are: what happens if the World Federation against hemophilia runs out of funds? And what will be the future of those beneficiaries, including young Charles, who at the age of five has already dropped out of school and is deprived of social interaction due to his fragile condition?

Reporting for capital firm in Lilongwe. I am Clement Msiska.



William NEWTON

USA



William Newton is a healthcare reporter for GlobalData Healthcare and Clinical Trials Arena covering drug development in rare diseases, central nervous system diseases, ophthalmology, and head and neck cancers. His work focuses on the implication of clinical trial results on drug advancement, approval and uptake, and insurance coverage. Previously, he worked at the healthcare information firm Close Concerns, where he covered breaking news in diabetes therapeutics and technology for the company's

industry-facing publication. He graduated Williams College in 2020 and worked as a News Editor, Executive Editor, and Managing Editor of the Williams Record. Outside of health journalism, William enjoys ultimate frisbee, basketball, and crosswords.

ANALYSIS

Platform trials: an opportunity for rare dystrophies or gene therapies?

By William Newton | 06 Dec 2021 (Last Updated December 8th, 2021 14:48)

Platform trials offer a streamlined approach to rare disease research, but regulatory and design obstacles loom large.

Designing clinical trials for rare diseases can prove challenging with limited resources and high operation costs. But three nontraditional trial designs—platform trials, basket trials, and umbrella trials—could revolutionise the space. In this series, Clinical Trials Arena will explore each design and its potential to shape the future of rare disease research.

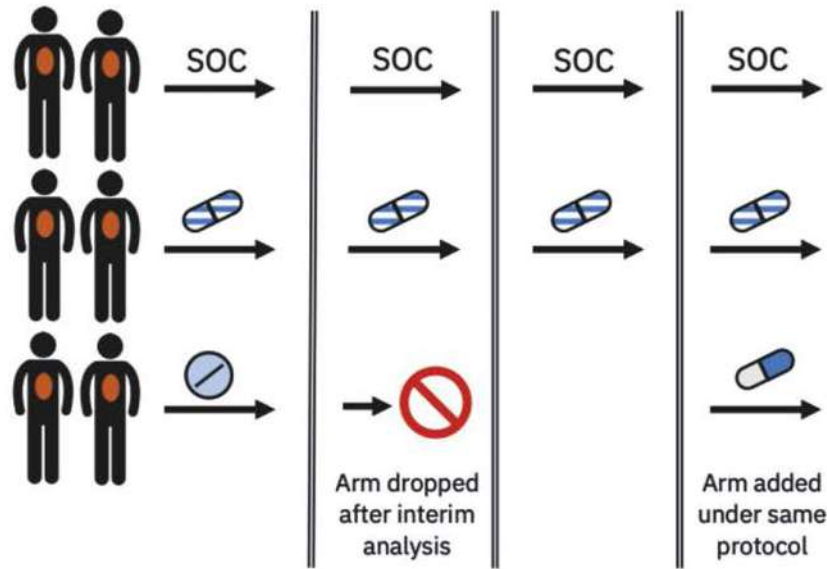
Platform trials can streamline rare disease research as they evaluate multiple treatments and diseases under a single, durable master protocol. This trial design has the flexibility to add and drop treatment arms as data develops and new drug candidates emerge.

By focusing on disease groups with similar phenotypes and manufacturing needs, rare disease researchers can utilise platform trials to increase efficiency and reduce operation costs. A successful platform trial requires choosing compatible diseases, negotiating with regulators and selecting appropriate endpoints and sites.



Platform trial

Single master protocol; can add and drop treatments and diseases over time



CLINICAL TRIALS ARENA

Can platform trials streamline dystrophies and gene manufacturing?

Rare muscular dystrophies have similar disease profiles and an overlapping pool of experts, making this field a strong candidate for a platform trial design, explains NIH Director of Rare Disease Research Anne Pariser.

Most dystrophies have different underlying causes and thus require different drugs. But monitoring, equipment, and endpoints could be similar, Pariser notes. As a result, a durable master protocol could live on even if the trial

Platform trials are also attractive for rare disease gene therapies, where different gene constructs can rotate in and out of viral particles, Pariser adds. There are opportunities to streamline across gene therapy manufacturing, preclinical work, assay development, and clinical monitoring expertise, she notes. Streamlining these processes could add efficiencies, resulting in significant cost savings, she told this news service.

Contending with regulatory concerns

From a regulatory perspective, master protocol designs typically don't directly lead to approvals, a biostatistician notes. Instead, they are often intended to detect an efficacy signal that forms the basis for a subsequent registrational trial, he says.

Poll |

What are your thoughts on the cost of decentralized/virtual clinical trials vs. traditional trials?

- Decentralized/virtual clinical trials are more expensive than the traditional trials
- Decentralized/virtual clinical trials are less expensive than the traditional trials
- Cost of decentralized/virtual clinical trials is same as traditional trials

Next

While the FDA will initially recommend a traditional, randomised, placebo-controlled trial investigating a single treatment, the agency can be flexible with rare diseases, a former FDA orphan drug official says. For example, if a rare disease only affects a few hundred patients, the FDA would be willing to consider a nontraditional trial that can still demonstrate a drug's safety and efficacy, adds the former official, who now works as a regulatory consultant. To maximise chances, companies should come to FDA trial design meetings prepared with extensive animal data in all the indications and treatments they plan to pursue, she says.

Pariser adds that in rare diseases, the regulatory burden of evidence can be lower. "With rare diseases, it's not

Platform trials's location and endpoints are key

Since rare disease experts are usually clustered in specific centers and paediatric hospitals, these sites could be well-suited trial locations. If a center specialises in multiple, similar rare diseases, it could already have the basic infrastructure to pursue a platform trial, Pariser says.

Ideally, efficacy endpoints of a platform trial design would include a short-term biomarker, making it easier to drop arms that underperform in interim analyses, Pariser says. But selecting an endpoint can be challenging in rare diseases, which often do not progress as fast or as predictably as cancer, she adds.

In cancer, there are multiple standardised endpoints such as progression-free survival (PFS) and overall response rate (ORR). In contrast, rare diseases often lack similar universal endpoints. "If we could better understand rare diseases, then that would make it a whole lot easier to design good quality clinical trials," Pariser says.

Platform trials in action

In addition to numerous platform trials in oncology and Covid-19, there are several important platform designs in rare diseases in early stages. For instance, the NIH is breaking into the rare disease space with the Platform Vector Gene Therapy (PaVe-GT) trial.

Collagen Q deficiency—and two metabolic diseases—propionic acidemia and isolated methylmalonic acidemia.

PaVe-GT will develop and test AAV-9, with a different gene for each indication, using a single master protocol. Though not yet at the stage of clinical testing, Pariser says her team has seen signs of early progress: “On a small scale, we’re already seeing some efficiencies and really seamless information sharing between our investigators and our team.”

Outside of rare diseases, the Phase II/III STAMPEDE trial ([NCT00268476](#)) is an active platform trial with an estimated enrollment of more than 12,000 patients with prostate cancer. There are currently eleven treatment arms, three of which are open for recruitment: standard of care, metformin, and transdermal oestradiol. Overall survival is the primary endpoint, with secondary endpoints including adverse events and cost effectiveness measured by EuroQol.



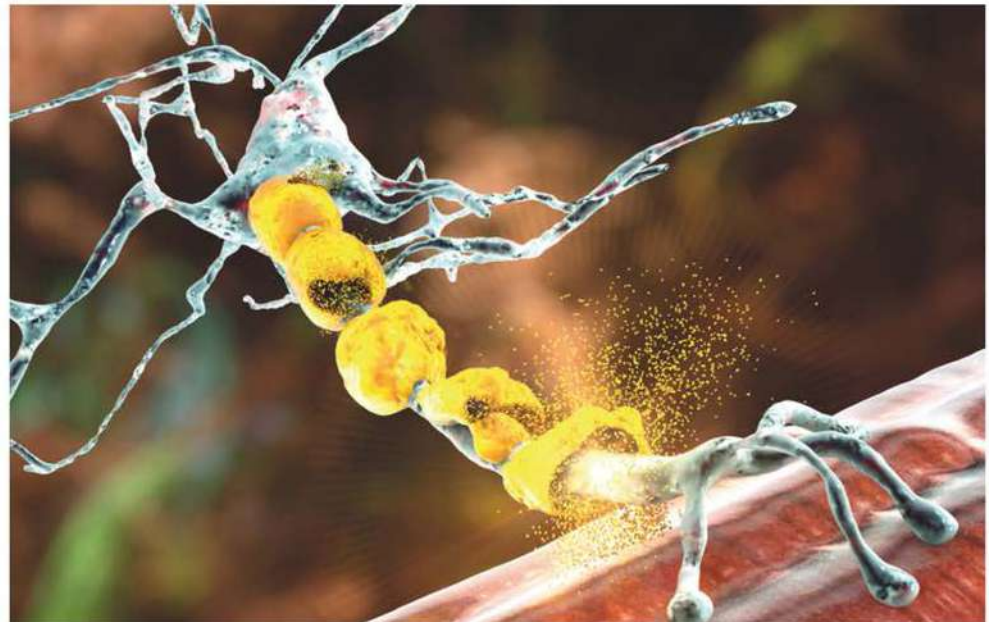
ANALYSIS

Basket and umbrella trials: untapped opportunities in rare disease

By William Newton | 21 Dec 2021 (Last Updated December 21st, 2021 12:30)

Basket trials have more traction than umbrella trials, but both are a drop in the bucket in total rare disease studies.

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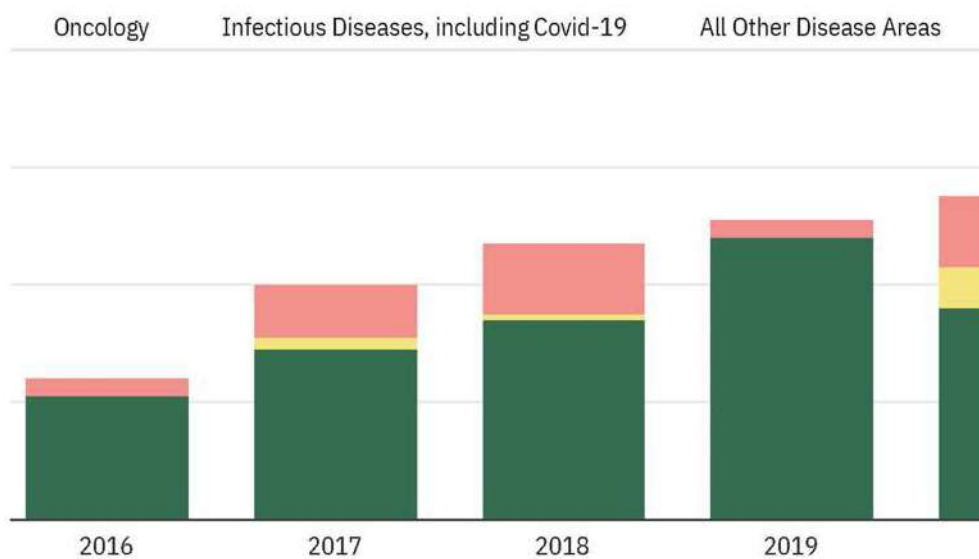


Designing clinical trials for rare diseases can prove challenging with limited resources and high operation costs. But three nontraditional trial designs—platform trials, basket trials, and umbrella trials—could revolutionise the space. In this series, Clinical Trials Arena will explore each design and its potential to shape the future of rare disease research.

Basket and umbrella trial designs could open new doors in therapeutic rare disease research, but they remain largely underutilised outside of cancer. Even though basket trials have gained some momentum in rare diseases, umbrella trial use remain relatively stagnant.

Outside of cancer and Covid-19, basket and umbrella trials still underutilised

Number of clinical studies described as basket or umbrella trials for any disease, sorted by therapy area



Source: GlobalData

CLINICAL TRIALS AR

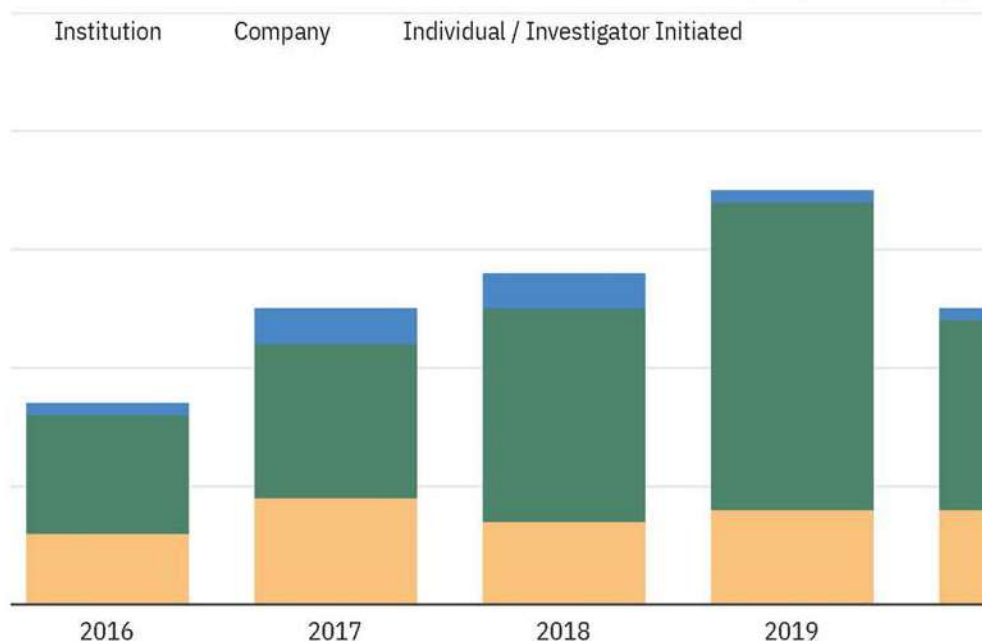
Growth in use of basket and umbrella trials has primarily been

“basket” or “umbrella” in their trial title or description, sorted by initiation year over the past six years, shows that these designs have remained underused in indications outside of cancer. A slight uptick of these nontraditional trials in infectious disease treatments over the past two years is likely a consequence of Covid-19 investigations.

Pharmaceutical companies are leading the recent surge in basket and umbrella trials for rare diseases, according to the GlobalData database. In 2016, companies began an estimated ten such trials while institutions started to run around six. Fast forward to 2021, companies initiated an estimated 40 basket or umbrella trials while institutions started approximately eight studies.

Recent growth in basket and umbrella trials driven by pharmaceutical companies

Number of clinical studies described as basket or umbrella trials for rare diseases, sorted by sponsor type



Umbrella trials lag behind basket approach

Basket trials can investigate multiple rare diseases simultaneously with a single treatment intervention. Meanwhile, umbrella trials can look at multiple treatments for one disease, saving on recruitment and protocol design costs.

Poll |

If you are using the decentralized/virtual clinical trial model or at least part of the trial management is virtual, was it:

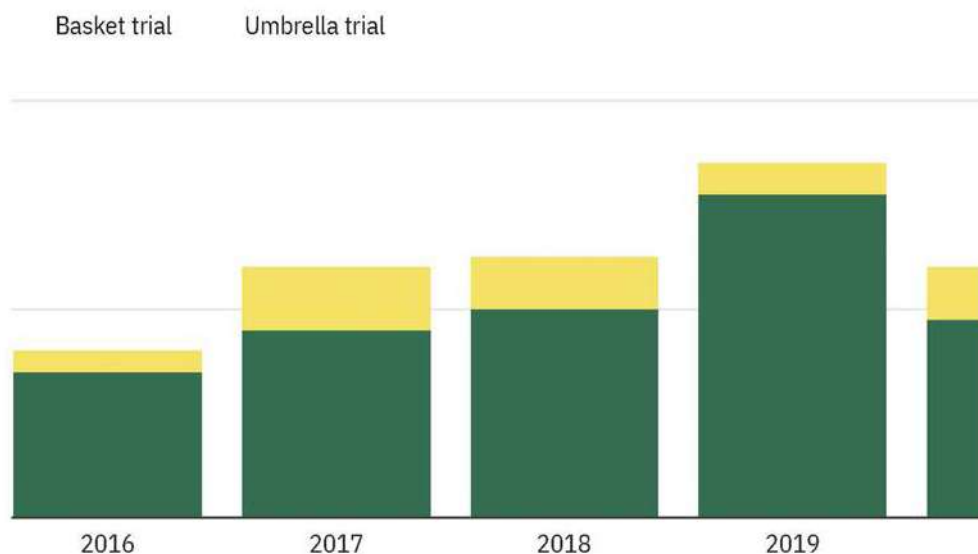
- Before the COVID-19 pandemic
- Because of the COVID-19 pandemic
- Plan to initiate this model in the future

Next

But thus far, there have been far fewer clinical studies for rare disease treatments described as “umbrella” than as “basket” trials. Nevertheless, both nontraditional designs still make up only a very small fraction of the total number of rare disease investigations. In 2021, there were an estimated 42 basket trials and seven umbrella trials initiated, but more than 5,000 rare disease trials started in the same year.

Basket trials outpace umbrella trials in prevalence and growth

Number of clinical studies in rare diseases described as basket or umbrella trials



Source: GlobalData

CLINICAL TRIALS AR

Because umbrella trials require multiple treatments for one disease, they often require various companies and institutions to provide therapies. This requires more risk and complicated collaboration, which could include different stakeholders that normally don't work together, explains NIH Director of Rare Disease Research Anne Pariser. As a result, it would likely take multiple umbrella trials in rare diseases before significant efficiencies appear, she notes.

Like platform trials, umbrella trials would work well for gene therapies in rare diseases areas such as neuromuscular junction disorders, Pariser says. A single capsid could be used as a delivery mechanism for multiple genes targeting a single

Basket trials target shared mutations

Rare diseases with shared underlying genetic mutations could benefit from basket trials, including when these clusters of diseases have different clinical manifestations, Pariser explains. Rare demyelinating diseases, which often have similar underlying causes, would also make sense for this design, notes a former FDA official who now works in regulatory consulting. Examples of rare demyelinating diseases include acute disseminated encephalomyelitis, neuromyelitis optica spectrum disorders, as well as multiple sclerosis and its rarer variants.

Rare metabolic disorders often have causes within the same pathway, making them strong candidates for a basket trial design, Pariser says. Rare metabolic disorders include Fabry disease, Crigler Najjar syndrome, and Gaucher disease. The purpose of the basket designation is to save on administrative costs, such as avoiding the need to rerecruit clinical sites, a biostatistician adds.

For a nontraditional trial design to lead to an approval, the individual components of the basket trial would need conventional trial designs, the biostatistician adds. As in the case with platform trials, drug companies should propose basket or umbrella trial designs to the FDA with extensive animal studies, natural history data, and examples of previous, similarly structured trials, the former FDA official notes. There is always room for the FDA to be flexible, but due diligence in compiling non-clinical supporting research is essential, she says.



Marielba NUNEZ

Venezuela



Marielba Nunez is a freelance Venezuelan journalist, based in Caracas, specialized in health and science. Her recent work has been focused on portraying the complex humanitarian emergency in her country. She has worked as reporter and editor in Venezuelan print and digital media, mainly in the newspaper *El Nacional*, where she coordinated the sections of science, society and in-depth journalism. She has also collaborated with national and international media outlet as *Armando.Info*, *Scientific*

American in Spanish and *Scidev*. She holds a bachelor's degree in Social Communication from the *Universidad Central de Venezuela*, a master's degree in Science Communication from *Universitat Pompeu Fabra*, Barcelona, Spain, and was selected as a Knight-Wallace fellow at the *University of Michigan* 2020. She wrote chapters for the books *Así nos tocó vivir. Historias que están detrás de la pobreza* (UCAB, 2006) and *Unbias the news. Why diversity matters for journalism* (Hostwriter, 2019).

Long forgotten is the community that helped understand Huntington's disease

elnacional.com/venezuela/en-el-olvido-esta-la-comunidad-que-ayudo-a-entender-la-enfermedad-de-huntington

December 31, 2021



Yoelvis Foto is distressed by the discrimination Huntington patients face. Photo: Lisbeth Torres

At the age of 25, Yoelvis Soto, a resident of the San Luis neighborhood, in the San Francisco de Maracaibo municipality, began to manifest the symptoms of Huntington's disease, a hereditary condition that he shares with many of his neighbors in that small town of the Zulia state. He had training as a paramedic and worked in the shrimp farms that operate in the area, activities that he would soon have to abandon.



Today, at 32 years old, the involuntary and jerky movements that characterize the disease completely prevent him from working. From his time in the world of work, he did not have any pension left that could help him support his 7 and 10-year-old children. The smallest has begun to show symptoms of early development of the disease and walks, his relatives describe, on the tips of his feet. She often misses school because her mother is afraid she may fall and hurt herself. Not only because of the difficulties to maintain balance, typical of the disease, but because other children bully and push him. “Some days, she comes home with scrapes,” describes her aunt, Evelyn Soto.

Yoelvis is mortified precisely by the rejection and contempt shown by others towards those like him who suffer from the disease for which his father died at 50 years of age. "In other municipalities and here there is not much support," he says. The stigma makes daily life even more difficult for the inhabitants of San Luis, an area punished by extreme poverty, the lack of access to drinking water and electricity, and where the public health clinic that should serve the community has been closed since about two years ago, when the covid-19 pandemic began.

Although the health crisis is widespread throughout the state of Zulia, in San Luis it is even more evident, precisely because of the high presence of Huntington's disease. Along with Barranquitas, another population on the western shore of Lake Maracaibo, has the highest

prevalences of this condition worldwide: while, in general, it is reported to affect 2.7 people per 100,000 inhabitants, here it is estimated that there are 700 cases per 100,000 inhabitants.

Origin of the Huntington

In addition to uncontrollable movements, Huntington's disease is accompanied by other problems, such as language and behavior disorders, including mood swings, depression, and aggressiveness, as well as headaches, insomnia, and progressive cognitive decline. The origin is a genetic mutation that causes an abnormality in the production of a protein known as huntingtin, which in this case becomes a toxic substance for neurons.

Advertisements

In early December, Norma Gotera, who had recently seen one of her three daughters die, Rudy Soto, 44, a Huntington patient, was making efforts to get medical care for another of her daughters, Alexandra Soto, 46, also diagnosed with the disease and affected by a picture of fever and seizures. With sacrifices he managed to collect 16 dollars that he needed to pay in a private laboratory for the blood tests that were requested to determine what was wrong with Alexandra. To this he must have added the effort involved in transferring his daughter, very weak and almost prostrate, to the clinic, located several streets from his house, to be examined.

In the house that has belonged to Nelly Guerra for 38 years, where a community center in Barrio Adentro runs that she is in charge of coordinating, the situation is not encouraging either. He shows the shelves where medicines used to be kept, completely empty. "We have four years that do not send anything from the ministry (of Health)." The aid that he manages to distribute does not come from the State, but from private national and international foundations that send him bags of food to distribute in the neighborhood, as well as some medicines and other items, such as diapers, which quickly run out.

But if anything speaks eloquently of the oblivion in which this community survives, it is a red brick building at the entrance to the neighborhood. On its façade there is still a sign that reads Huntington's Korea Home House "Love and Faith". A care center for people with this condition operated there between 2000 and 2013. Some 65 patients could come to reside there when they got worse. Today, the doors to the center are closed and the place has been vandalized.



The Casa Hogar Amor y Fe, founded with the support of the scientists who conducted the genetic studies on Huntington's disease, is closed and has been vandalized. Photo: Lisbeth Torres

The memory of a contribution

Few remember the relationship of the community of San Luis, today sunk in abandonment, with a historical scientific advance. Thanks to the fact that its inhabitants agreed to have samples taken to participate in international scientific studies, it was possible to identify the gene linked to Huntington's disease, a milestone that was published in 1993 in the journal *Cell*.

There, reference is made to the “Venezuelan pedigree”, the genealogical tree built with information on more than 18,000 people that the American researcher Nancy Wexler managed to put together thanks to her constant visits, during the eighties and nineties, to the areas affected by Huntington's disease in Maracaibo.

The roots of this story can be traced a few decades earlier, when the Venezuelan doctor Américo Negrette, who was a professor at the Faculty of Medicine of the University of Zulia, described in a first publication of 1955 his observations on several dozen cases of what then It used to be called Mal de San Vito.

The work of Negrette and her disciples was what made the international scientific community turn its eyes to the “sanviteros” after a few years, as they still identify with each other as the inhabitants of this small fishing village.

An article published in 2004 in the *Proceedings of the National Academy of Sciences* of the United States, relates that Wexler and his collaborators developed for their research “a protocol of neurological, cognitive and psychiatric evaluations to specifically satisfy the cultural and educational requirements of this community, mainly without education”. That allowed them to feed, for 23 years, a database with the information of 2,547 people who were examined annually for these studies.

Only the older ones now seem to have a memory of the visits of those international investigation teams. Norma Gotera remembers that at those times they used to receive donations of clothes and medicines. The Hereditary Disease Foundation to which Wexler belongs played a role in the opening and maintenance of the Casa Hogar Amor y Fe.

A letter published by Wexler in *The Lancet*, in 2013, speaks of the financial difficulties to maintain the institution: to pay its director, the doctor Margot de Young, two nurses and 30 employees they needed 750,000 dollars annually and in total they required one million dollars annually to keep running. In an interview with *The New York Times* in 2020, Wexler pointed out that the house had been closed by decision of the Venezuelan government.

The entities that could provide support to Huntington patients, but which are currently closed, are joined by the Institute for Genetic Research of the University of Zulia, affected by the acute lack of resources suffered by the university, says geneticist Lennie Pineda, Retired researcher from that organization.

Pineda prepared a few years ago a review from the bioethical point of view of the studies carried out by international researchers among Huntington de Maracaibo patients, which questioned the way in which the samples were obtained, due, among other reasons, to the social vulnerability of the disease. studied population that, in the long run, did not have access to the results of the genetic analyzes.

He adds that the contribution of those who volunteered to participate in the studies has not ceased, although the great promise of a cure has not yet materialized. “This genetic material has allowed the discovery of other genes. The samples of these people are kept in repositories of multiple laboratories around the world ”.



The clinic that should provide services in San Luis has been closed for two years. Photo: Lisbeth Torres

Debt to remedy

For Marina González, from the NGO Hábitat LUZ, which has developed cooperation projects in the area, it is not enough to remember the debts that the international scientific community has with the San Luis community, we must also highlight those that the state has with it Venezuelan. “We must not forget that this is a disease that attacks people in their productive ages, so their families are extremely vulnerable. Hence, we must combat exclusion and give them tools to strengthen their education and economy”.

The poverty in which the inhabitants of San Luis live becomes a factor that increases the risk of earlier development of Huntington's disease, as has been described by scientific research to which the inhabitants of this area themselves contributed.

Dozens of children with Huntington's family histories do not have access right now to the genetic counseling they need or the conditions that could make a difference in their lives. Jhonny and María Soto form one of those families that every day begs for the disease to pass them by. His mother died from that cause and she, who comes from Barranquitas, says that one of her aunts suffered from it. "But my mother, thank God, he didn't get hurt by that," he says.

The psychiatrist Rey Varela, who voluntarily visits the area every fifteen days, and who was linked to the community through his work with the NGOs Paluz and Factor H, emphasizes that treatments should be individualized and not standardized. In each case, it should be evaluated whether antidepressants, antipsychotics, or other drugs, such as tetrabenazine, are needed to control exaggerated movements.

The accompaniment of family members is also essential: they are the ones who assume the tasks of care, including feeding, which must be specially prepared to avoid drowning when patients cannot swallow. However, the main concern remains poverty. "If a person does not have to eat three times a day, he will not have to take medicine because the first thing he has to do is satisfy the need for hunger."

By **Marielba Núñez**



Esther OLUKA

Uganda



My name is **Esther Oluka**, a journalist working with the Daily Monitor, a print Newspaper in Kampala, Uganda. I ventured into Journalism, professionally, in 2012 soon after graduating with a Bachelors degree in arts in Mass Communication at Uganda Christian University (UCU), a learning institution in Uganda.

One of the areas I love writing about is health because it's relatable and by

writing stories on the subject, they make an impact on people's lives in one way or another.

I am delighted to be part of this fellowship program covering Rare Diseases. I hope to learn a lot from other colleagues during the program as well as understand more about the subject.

Rarely diagnosed diseases: The silent killers in Uganda

Diagnosing and treating rare diseases has become a huge burden because of different challenges the country faces. In this first of our four part series on covering rare diseases, **Esther Oluka** explains the reasons why they are the new silent killers.

John Paul Ofwono rose to fame during the 1990s as the tallest man in the country.

He stood at about 7 feet 6.5 inches (about 230cm) tall. His incredible height was, however, a disorder.

Some media reports claim that Ofwono was suffering from over secretion of growth hormone (from the pituitary gland) which in the end caused enlargement of bones in his feet, hands and face.

The medical term for this condition is acromegaly which manifests in adults.

Before succumbing to diabetes in 2003 at the age of 43, there had been ongoing discussions to take him abroad for specialised medical care. However, Ofwono is reported to have declined the treatment fearing the outcome.

Due to the rarity of his condition in the country, some experts in the medical profession, who spoke to *Daily Monitor*, reason that it is why help came a little too late for Ofwono. The disorder had taken a toll on his body.

"With rare conditions, patients might take years before knowing what they are really suffering from or they may never discover," Dr Alex Kakoraki of Murchison Bay Hospital in Luzira, says.

Perhaps, the challenges in managing rare diseases in the country is partly why there are no readily available statistics on these disorders.

Some medical professionals were able to give statistics from only their respective fields.

For instance, Dr Peter Wasswa, a paediatric hematologist at Mulago hospital, said about 300 patients are registered with Hemophilia Foundation of Uganda



The late John Paul Ofwono poses with a health worker in Tororo District. PHOTO/ FILE

Haemophilia Foundation of Uganda (HFU), a non-government organisation which helps create awareness on hemophilia, a bleeding disorder among patients.

"Yet one would expect to have about 2,000 patients with the condition in a

population of about 42 million Ugandans," Dr Wasswa says.

There are many cases that are undiagnosed or undetected and Dr Wasswa believes that it is because of limited access to the right treatment.

Dr Richard Kwizera, a medical mycologist at the Infectious Diseases Institute, College of Health Sciences, Makerere University, says Chronic Pulmonary Aspergillosis (CPA) is another often rare and misdiagnosed condition.

"It is a slowly progressive and destructive parenchymal lung disease which affects both immunocompetent and immunocompromised patients with previous or underlying lung disease," he says.

Dr Kwizera says the condition may be misdiagnosed as any other lung disease including tuberculosis. In the end, patients may miss out on getting the right treatment.

The condition affects three million people worldwide.

The other challenges with handling rare diseases, Dr Kwizera says, is that



Witchcraft. Patients may ignore the condition or sometimes conclude that it is witchcraft. They will then go out seeking for alternative medicine and only when things get worse do they then start seeking better treatment, when the situation has already spiraled out of control," Dr Richard Kwizera, a medical mycologist at the Infectious Diseases Institute, College of Health Sciences, Makerere University

some of these conditions do not manifest as chronic in nature and are not that painful.

"And for such reasons, patients may ignore the condition or sometimes conclude that it is witchcraft. They will then go out seeking for alternative medicine and only when things get worse do they then start seeking better treatment, when the situation has already spiraled out of control," he says.

He added that in some circumstances, "a section of patients easily give up on life after discovering they are suffering from a mysterious condition."

One patient battling an unusual skin condition told *Daily Monitor* on condition of anonymity for fear of being stigmatised, that she has given up on seeking the cure.

"Where haven't I gone in the past five years? I have been to almost every specialist but they cannot quite figure out what is wrong with my skin," she says.

Due to her condition, some individuals don't want to be near her, a move the

ISSUE

Summary of some of rarely diagnosed diseases in the country

- Acromegaly; over secretion of the growth hormone (from the pituitary gland) which in the end causes enlargement of the bones in the feet, hands and face.

- Gigantism; a rare condition caused by excessive amounts of growth hormone during childhood.

- Autoimmune disease; a condition in which your immune system accidentally attacks your body instead of protecting it.

- Sleeping Sickness, or human African trypanosomiasis; one of the most important but equally most neglected tropical infections caused by related parasite strains transmitted by a tsetse fly. The disease can lead to disruption in sleeping patterns, confusion, among other signs.

- Giant Cell tumor of the bone; a rare, aggressive non-cancerous tumor which develops near a joint at the end of the bone.

- Chronic Pulmonary Aspergillosis (CPA); a slowly progressive and destructive parenchymal lung disease which affects both immunocompetent and immunocompromised patients with a previous or underlying lung disease.

woman says is mentally tormenting her.

Dr Alex Bakenga, a clinical medical officer at the Uganda Cancer Institute (UCI), said one of the biggest challenges when handling any patient with a rare condition is that a lot of time and resources need to be injected into investigating a respective health issue.

"This is all because a medical expert is trying to be sure with what they are dealing with before commencing treatment," Dr Bakenga says.

But even with thorough attention that might be given in conducting an investigation on a rare disease, it can be misdiagnosed, as the disease continues spreading fast in the body.

In September, while giving an online presentation on the theme: Rare Diseases: A Global Challenge, Samuel Wiafe, a clinical psychologist in Ghana, stated that poorer (African) countries are more focused on treating major infectious diseases than rare ones.

"We have been focused on dealing with cholera, tuberculosis, HIV, malaria, and many infectious diseases," Wiafe said.

To be able to get decision makers to pay close attention to the concept of rare diseases, Wiafe said "Remains a huge challenge largely because of limited funding to respective health sectors and in so doing, governments will have no option but to prioritise and in so doing, will opt to neglect dealing with rare diseases."

The reporting of this story was supported with a grant offer from National Press Foundation (NPF) to exclusively cover Rare Diseases in the country.

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Dr Richard Kwizera, a medical mycologist, on duty at the Infectious Diseases Institute, Makerere University. PHOTO/ COURTESY

17,000 university students risk missing study loans

The Higher Education Students Financing Board has not received applications from students in those universities.

BY DAMALI MUKHAYE

At least 17,000 students from Makerere, Gulu and Soroti universities slated to be admitted for the 2021/2022 academic year intake are at the verge of missing out on the government study loans after the institutions failed to admit private students within the deadline.

The application for the study loans that kicked off last month ends today, something that will see the students locked out.

For one to qualify for the study loan, he or she must be admitted to the approved universities and other tertiary institutions and must be pursuing Science, Technology, Engineering and Mathematics (STEM) programmes or those approved under affirmative action.

The Higher Education Students Financing Board (HESFB), which is mandated to award the study loans, has not received



Makerere University students on campus. The university management wants the deadline application for the study loans extended. PHOTO/COURTESY

any application from students admitted at the three universities.

Sources in the HESFB yesterday told Daily Monitor that the three universities have not yet released the lists of privately sponsored students, and if the board does not extend the application deadline, their students will be locked out.

"The fate of students in those universities lies in the hands of the HESFB. You find that first year students from some universities such as Kabale have started

11,187

Loan beneficiaries since 2014

studying and the board is supposed to pay their tuition," a source said.

"But there are other universities that have not yet admitted students so the board is at the crossroads of how to serve

all the students," the source added.

Makerere admits majority of the students in the country with last year's intake totaling to 14,500, while Gulu and Soroti universities admit between 1,000 and 1,600.

The HESFB spokesperson, Mr Bob Nuwagira, confirmed that they have not yet received any application from students in the universities in question, but added that the board is handling the issue.

"The deadline ends today and the

board will communicate the way forward on whether to extend the deadline to cater for the affected students or to go with those who have applied," Mr Nuwagira said.

The Deputy Vice Chancellor of Makerere University, Prof Umar Kakumba, said they have not been able to complete the admission exercise due to the high number of students they handle.

He, however, said they are in talks with the HESFB to extend the deadline because the students are not the ones at fault.

"You know we admit students who come directly from Senior Six, diploma students, law pre-entry students and those on mature entry. Now sorting all these is not an easy task," Prof Kakumba said.

"We are, however, at 90 percent of admitting our students and hopefully that by the end of this week, the lists will be out. We are hopeful that the deadline is extended to also cater for our students," he added.

The Vice Chancellor of Gulu University, Prof, said the academic registrar was handling the admissions.

The government last month asked eligible needy students to apply for study loans for the academic year 2021/2022.

The government is slated to give study loans to 1,600 needy students who want to pursue degree and diploma programmes.

The loans cover tuition, functional, and any other fees required by the learning institutions. They also cover aids and appliances for people with disabilities. Online applications officially kicked off yesterday with suitable candidates expected to demonstrate a financial need.

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Nangulu's long battle with bleeding disorder

BY ESTHER OLUKA

KAMPALA. In this second part series of covering rare diseases in the country, we look at Mr Peter Nangulu's story of coping with hemophilia, a rare condition that affects the clotting of blood.

When one meets Mr Peter Nangulu for the first time, it is hard to imagine that he is battling any serious disease. Physically, he seems fine, yet all is not well with him.

Mr Nangulu, 26, has been battling haemophilia, a rare bleeding disorder, since childhood.

The disorder affects the clotting of blood in the body and also makes body joints swollen.

Mr Nangulu, who is a youth coordinator with Haemophilia Foundation of Uganda (HFU), says no one in his family understood what was happening with his body whenever he sustained either a bruise or cut that could not stop bleeding.

"My parents were quite confused with what was happening with me. They tried searching for answers from doctors and other family members, but no one had clear responses for them," he says.

One of his sisters also has a son with the same disorder.

Mr Nangulu recalls that the first time he noticed something strange with his body was when a sharp-stick pierced

his left thigh when he fell down as child while playing.

"This accident made me bleed for weeks until I had to be taken to hospital to get urgent medical attention," he says.

Then in 2007, while playing with friends, a game that involved running and jumping over desks in class, Mr Nangulu fell hard and hit both knees on the floor.

"My left knee got very swollen to the extent that I failed to wear school trousers," he recalls.

Mr Nangulu was in Primary Six at that time at Buwesa Primary School

in Butaleja District. Due to the extent of his injuries, he was taken to hospital where he was admitted for three months.

After leaving the facility, he started using crutches and earned the nickname "delicate" from his fellow pupils because of how fragile his body was.

Mr Nangulu continued using crutches during his secondary school life at Kiyeyi High School in Tororo District where he completed Senior Four in 2012.

He later sat Senior Six in 2015 at Town View College, in Tororo before joining Young Men's Christian Association

(YMCA) Comprehensive Institute, where he obtained a Diploma in Computer Science in 2020.

"I got into another serious (bleeding) accident last year when I bit myself while eating," he says.

It took some weeks for the bleeding to stop.

The diagnosis

Since no one knew what Mr Nangulu was suffering from, his treatment was based on assumptions.

His swollen joints worsened his condition, affecting his ability to walk and stretching his hands.

DOCTOR'S TAKE ON THE RARE BLEEDING CONDITION

Dr Peter Wasswa, a paediatric hematologist at Mulago National Referral Hospital, says: "Haemophilia is a type of hereditary bleeding disorder due to deficient production of one of the clotting factors specifically Factor VIII (type A haemophilia) and Factor IX (type B haemophilia). About 85 percent of patients have type A haemophilia, while the rest have haemophilia B.

Typically, the condition affects boys because the deficient gene lies on the X chromosome. Males have got an X and Y chromosome unlike females who have got two

X chromosomes. So, if a male has got one X chromosome and it is deficient, then, they will likely get hemophilia. Meanwhile, for girls, they have an X chromosome but they don't manifest symptoms because their other chromosome (Y) is enough to let them produce either Factor VIII or IX.

The girls can only become carriers of the disease and may pass on the deficient gene to their sons, leading to disease. Carriers do not have symptoms but can produce boys with the disease.

It is estimated that about one in 5,000 new born males have he-

mophilia. But you would expect that in a population of about 42 million Ugandans, you would expect to have about 2,000 patients with the condition, however, about 300 patients are registered with Haemophilia Foundation of Uganda, and so obviously many cases are undiagnosed or undetected.

The common signs and symptoms include excessive bleeding or it may occur for such long periods of time and swollen painful joints.

Treatment is clotting factor replacement prescribed by only trained medical personnel."

"The doctors treated me based on what they suspected was wrong with me. That is the burden of having a rare condition. Some sections of people began saying the condition was as a result of witchcraft," Mr Nangulu says.

It was not until 2016 when he was diagnosed with the rare condition following an assessment by a specialist who handles haemophilia disorders.

It was discovered that he had haemophilia, type A which is caused by missing factor VIII, a clotting protein in the blood.

Mr Nangulu says there has been a great improvement in his life after the diagnosis because he now knows how to deal with the condition, including getting the right medication.

He stays away from doing things that easily make him bleed or get injuries.

His advise

If one ever notices anything suspicious with their body, Mr Nangulu advises them not to keep ignoring the problem.

"Search for answers by continuously asking doctors or doing your own research even on the Internet. Taking such initiatives is what will save your life early," he says.

The reporting of this story is supported by National Press Foundation (NPF).

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Government has set January 15, 2022 as the new deadline.

BY DAMALI MUKHAYE

More than 20,000 students from various government universities are relieved after government bowed to pressure and considered extension of the deadline for study loans.

The move is meant to cater for Makerere, Lira, Soroti, Gulu and Kyambogo universities that have not admitted students yet.

Addressing journalists at the Uganda Media Centre yesterday, the Executive Director of the Higher Education Students Financing Board (HESFB), Mr Michael Wanyama, said students from various universities and other tertiary institutions that have not yet admitted learners were in panic after failing to meet the initial deadline of yesterday.

Mr Wanyama revealed that a number of students and managers of universities had petitioned the board to extend the deadline.

Thus, the board has extended the application for study loans from today to January 15, 2022.

"While the board had set the deadline for application for midnight November

Govt extends application deadline for study loans

30, it was established that some universities had not completed the admission exercise while others, most especially the tertiary institutions that admit students on diploma programmes, have not yet started the admission process," Mr Wanyama said.

He asked the affected universities to admit students within the extended period warning that there will be no more extension.

However, the extension caters for only students from the five universities, people living with disabilities as well as students from Karamoja sub-region.

Meanwhile, Mr Wanyama said the board will proceed to process and issue out study loans to all applicants who have applied by the deadline timeline.

Mr Wanyama revealed that the list of beneficiaries in the first slot will be released in December while those in institutions that delayed or admitted students will be out in February 2022.

Government reopened all institutions of higher learning (universities and Oth-

ISSUE

While the board had set the deadline for application for midnight November 30, it was established that some universities had not completed the admission exercise while others, most especially the tertiary institutions that admit students on diploma programmes have not yet started the admission process," Mr Michael Wanyama, ED of Higher Education Students Financing Board

er tertiary institutions) on November 1 but only continuing students who had joined these institutions commenced

their studies. First year students who sat for the 2019 Uganda Advanced Certificate of Education did not report for studies since most universities had not completed the admission exercise.

Whereas these categories of students have started their studies in some institutions, the majority are slated to start their studies next year, once the current first year students have progressed to second year.

Officials from various universities that had not concluded the exercise have indicated that by end of December, all their students will have received admission letters.

Prof Eli Katunguka, the vice chancellor of Kyambogo University, explained that they were delayed because they were switching from the Academic Information Management System (AIMS) to a new scheme dubbed ACMIS (Academic Certified Management Information System).

"The transition took a lot of time but I can assure our students that we have cap-

tured all the information and by December 15, we should have finished the verification process and should have the admission lists," Prof Katunguka said.

The government is slated to give study loans to 1,600 needy students who want to pursue degree and diploma programmes.

The loans cover tuition, functional, and any other fees required by the learning institutions. They also cover aids and appliances for people with disabilities. Online applications officially kicked off yesterday with suitable candidates expected to demonstrate a financial need.

Since 2014, the board has awarded loans to 11,187 beneficiaries (9,632 undergraduate degrees and 1,555 undergraduate diplomas) to study 130 degree programmes and 76 diploma courses.

At least 3,000 loan beneficiaries have completed their studies and are slated to start servicing their loans.

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In this third part (of four) series of covering rare diseases in the country, Ms Daniella Akellot opens up about having classical Ehlers-Danlos Syndromes (EDS), a genetic connective tissue disorder with symptoms including stretchy skin, joint hypermobility and fragile skin easily prone towards getting bruises.

She talks to Esther Oluka about the condition and why it took her 28 years to get diagnosed.

While growing up, Ms Daniella Akellot, did not understand what was going on with her body.

She noticed that her skin was stretchy, the wounds took long to heal and once they did, wide scars were left on her body.

Also, her joints were quite flexible and easily got dislocated.

"I remained quiet about my condition. I did not tell anyone, even in my family," she says.

Akellot adds: "I kept the condition a secret out of fear of being judged, left out and laughed at."

According to her, the dislocations of joints, for instance, happened from time to time.

"There were times I would either be getting up from a chair or walking and then hear a popping sound in my hip, a sign that something had snapped," she recounts.

Normally, the dislocations did not require her to seek medical attention as she always knew how to properly position them back into place.

"There was one night I dislocated my shoulder and was able to fix it back by myself," she says.

Because the wounds took long periods to heal and left behind huge scars, this sometimes would attract unwanted stares or prompt people to ask what was wrong with her skin.

The older she became, the more the symptoms grew worse and as such, she restrained herself from engaging in activities that easily brought harm on her body.

For instance she stopped playing sports, doing heavy household chores

It took me 28 years to be diagnosed - Akellot



Ms Daniella Akellot who has classical Ehlers-Danlos Syndromes (EDS), a genetic connective tissue disorder with symptoms including stretchy skin and joint hypermobility. PHOTO/COURTESY

and carrying heavy items.

"Some people then began calling me lazy and weak, not knowing this was a way of protecting myself," she reveals.

Over time, Ms Akellot was forced to research about her condition so as to find better ways of coping with it.

The diagnosis

While reading a medical-related book in 2016, Ms Akellot came across a picture of a young boy who exhibited similar symptoms.

She then approached one of the orthopedic surgeons at Comprehensive Rehabilitation Services in Uganda (CoRSU), a non-profit organisation with a

core mandate of rehabilitating patients with orthopedic occasions.

"After a thorough medical examination, the orthopedic surgeon eventually diagnosed my condition as Classical Ehlers-Danlos Syndrome (EDS). Imagine finding out after 28 years!" she exclaimed.

Confused and stunned by the revelation, Ms Akellot inquired about the condition and fortunately, the orthopedic surgeon offered the needed answers.

Classical EDS exhibits symptoms such as joint hypermobility, joints that easily dislocate, fragile and stretchy skin as well as wounds that take long to heal and leave wide scars.

Knowing about the condition has enabled her to manage it better by mostly refraining from activities that manifest the symptoms.

Ms Akellot says the biggest problem with having a rare condition is that it is difficult to get both a diagnosis and support from members of the community since the patient numbers are very few.

She advises those suffering from rare conditions to research about the symptoms before consulting a specialist.

The reporting of this story was supported with a grant offer from National Press Foundation (NPF) to exclusively cover rare diseases in the country.

WHAT IS EHLERS-DANLOS SYNDROME (EDS)?

Dr Paul Muwa, an orthopedic surgeon at CORSU, says; "EDS is a spectrum of rare inherited diseases that affect connective tissue. Some of the varieties include;

- Vascular EDS, considered the most serious type, affects internal organs and blood vessels by making them more prone to tear, in the long-run, causing bleeding. Patients with this type are disposed to issues such as bowel tearing and womb tearing.

- Hypermobile EDS where patients present symptoms such as joint hypermobility, easy dislocation of joints, extreme tiredness and skin that easily bruises.

- Classical EDS whose symptoms include joint hypermobility, joints that easily dislocate, fragile and stretchy skin as well as wounds that take long to heal and leave wide scars, among other symptoms.

- Kyphoscoliotic EDS where patients mostly present symptoms such as curvature of the spine, joint hypermobility and unstable joints that easily dislocate. Treatment involves managing symptoms (depending on the type). For instance, if a patient dislocated their joint, the problem is resolved by medically (fixing) it back.

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Buganda Kingdom mourns Kabaka's caretaker mother

Nagawa Siwoza, 76, died on Tuesday after suffering a stroke for years.

BY JAMES KABENGWA

Cohorts of Kabaka Ronald Muwenda Mutebi have said the death of his caretaker mother Margaret Nagawa Siwoza, has left him devastated.

Siwoza, 76, who was a resident of Katuuso-Buziga in Makindye Division, died on Tuesday night.

During a press briefing at Bulange Mengo, Buganda Katikkiro (prime minister) Charles Peter Mayiga said he had instituted a special committee to organise the burial of Siwoza.

The committee, which includes Mr Patrick Mugumbule, the speaker of Buganda Lukiiko (parliament), and Buganda Kingdom minister of culture Kyewalabye Male, will be headed by the second deputy Katikkiro Robert Wagwa Nsibirwa. Other members are Kabaka's uncle Ssabaganzi Emmanuel Ssekitooleko, and Mr Daniel Muyanja, a son of Siwoza.

"The committee will arrange the burial following all the cultural requirements," Mr Mayiga said.

Siwoza's friends, owing to her love for God, dedicated Biblical verses that they said were befitting to the woman of God.

These include Philippians 3:20, which reads: "But our citizenship is in heaven. And we eagerly await a Saviour from there, the Lord Jesus Christ".

Another Revelation 21:4, reading: "He will wipe away every tear from their eyes, and death shall be no more, neither shall there be mourning nor crying nor pain any more, for the former things have passed away."

Siwoza became Kabaka's caretaker mother through hereditary lineage of the cultural title of Namasole, an offi-



Kabaka Mutebi's caretaker mother Margaret Nagawa Siwoza died aged 76. PHOTO/COURTESY

cial mother of a reigning Kabaka.

Kabaka Mutebi was born to the late Sarah Lule Kisosonkole and the first Presi-

dent of Uganda Kabaka Edward Mutesa II.

When Kisosonkole died in 1974, her



Loss. "She has been receiving medicare from Mulago National Referral Hospital and in Nairobi. She has, however, not made it. The committee instituted will announce the burial date in the due course," Buganda premier Peter Mayiga

roles were taken over by Rebecca Zirimbuga Musoke, after the coronation of Ssabataka Mutebi as Kabaka but when she died in 2013, Siwoza took over her roles.

Mr Mayiga said Siwoza has suffered a stroke for one year.

"She has been receiving medicare from Mulago National Referral Hospital and in Nairobi, Kenya. She has, however, not made it. The committee instituted will announce the burial date in the due course," he said.

In August 2020, Kabaka Mutebi and Kenyan President Uhuru Kenyatta and former prime minister of Kenya Raila Odinga, visited Siwoza who was then hospitalised at Agha Khan Hospital.

One of her sons, Mr Daniel Muyanja, told *Daily Monitor* in a telephone interview that his mother worked in Nairobi before she returned to Uganda, to form a secretarial company, HEMU enterprises.

"When she retired, she became the Namasole. We will miss an outgoing, hardworking, loving, friendly mother, who guided us through the challenges of this world," Mr Muyanja said. Her vigil was held at Ring Road in Buziga last night.

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Owiny-Dollo lauds Comboni missionaries

BY STEPHEN OTAGE

KAMPALA. Chief Justice Alfonso Owiny-Dollo has lauded the Comboni Missionaries for setting up schools in Acholi Sub-region.

While launching the autobiography of Rev Fr John Scalabrini on Sunday last week at Bishop Cipriano Kihangire Senior Secondary School, Justice Owiny-Dollo, said northern Uganda is what it is today because of the missionaries.

"Today, 60 per cent of prominent Acholi leaders were educated by Comboni Missionaries and 40 per cent by the Church Missionary Society," he said.

Justice Owiny-Dollo said Fr Scalabrini came to Uganda in 1962 and became an icon of the Catholic Church as he built schools and hospitals.

"The school where we are today has educated so many children from the Acholi region and it was named after the first black African Catholic Archbishop Cipriano Kihangire. I believe this school was supposed to be located somewhere in the Acholi Sub-region," he said.

Justice Remmy Kasule, the board chair of Emmau's Foundation, a non-profit organisation, said Fr Scalabrini, who died in 2016, was twice exiled by the Idi Amin government and military commission that overthrew the latter. The respective governments feared that Fr Scalabrini's power could be used to their detriment, he said.

Justice Kasule revealed that when Fr Scalabrini returned to Uganda in 1980, he revived commercial agriculture in Gulu.

"He also organised the laity in the northern district and raised funding for schools which caused discomfort in the sitting government. It thought he was a change agent and consequently limited his movements and operations to within Kampala Archdiocese. This eventually led to the birth of Bishop Cipriano Kihangire Senior Secondary School in Luzira in 1993. At the time, Nakawa Division did not have a high quality secondary school," he added.

Justice Owiny-Dollo also launched *The Faces in Their Eyes: A Memoir of a Spirited Love*, written by Poppilla Sue Agalo over a 10-year period to celebrate the life and times of Fr Scalabrini.



Fr John Scalabrini

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BY ESTHER OLUKA

KAMPALA. A section of medical experts and patients has urged the government to invest in diagnosing and treating rare diseases.

The appeal follows a story *Daily Monitor* published on Monday about how rarely-diagnosed diseases are the new silent killers in Uganda.

Dr Paul Muwa, an orthopaedic surgeon at Comprehensive Rehabilitation Services in Uganda (CoRSU), a not-for-profit organisation with a core mandate of rehabilitating patients with orthopaedic conditions, says it is imperative that funding in the area improves.

"That money would cater for many great projects, including pushing for awareness among masses, buying specialised equipment and training medical personnel to dig deep into diagnosing these ailments," Dr Muwa says.

Dr Richard Kwizera, a medical mycol-

Build capacity to tackle rare diseases - experts

ogist at the Infectious Diseases Institute, College of Health Sciences, Makerere University, says: "The reason why patients with rare conditions continue to suffer is because of limited medical equipment in managing their cases."

He added: "You find a patient going to a health facility to seek medical attention only to be turned away because of lack of equipment. In the end, such a patient will resign to fate by returning home and suffering silently with their condition."

Mr Peter Nangulu, who suffers from haemophilia, a disorder that affects the blood's ability to clot, says absence of statistics around the condition shows that patients like him are rarely catered for.

Ms Daniella Akello, who suffers from classical Ehlers-Danlos syndromes

(EDS), a genetic connective tissue disorder, says it is about time the government stepped up.

"These patients get isolated at times

WHO

According to the World Health Organisation (WHO), rare diseases present fundamentally different challenges from those of more common ailments.

WHO, therefore, urges that more investments be dedicated to build infrastructure and international networks such as biobanks, registries and networks of expertise.

because of their respective rare conditions," she says, adding: "If the government invests time and energy in sensitising the masses, the stigma will reduce."

Dr Alex Bakenga, a clinical medical officer at the Uganda Cancer Institute (UCI), says investing in medical personnel is a great strategy of motivating them to handle rare diseases.

During a recent online training for journalists on rare conditions, Dr Durhane Wong-Rieger, the chair for Rare Diseases International and President for Canadian Organisation for Rare Disorders, urged the governments from the global south to copy the best practices of their peers from the global north.

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USA



Erin Prater is a veteran communications professional with more than 20 years of experience in writing, editing, content planning, social media/website management and page/graphic design for newspapers, magazines and publishing houses. She currently serves as the assistant city editor at *The Gazette* in Colorado Springs, Colo., where she also covers education, health and the military. Aside from *The Gazette*, her work has been published on Military.com and in *Stars and Stripes*, among other publications. She has earned a graduate certificate in health

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Diagnostic odyssey: The lonely road walked by thousands of Coloradans with disorders so rare even their doctors hadn't heard of them

[denverpost.com/premium/diagnostic-odyssey-the-lonely-road-walked-by-thousands-of-coloradans-with-disorders-so-rare-even/article_d994be10-64f2-11ec-b5e6-539b30f3b61a.html](https://www.denverpost.com/premium/diagnostic-odyssey-the-lonely-road-walked-by-thousands-of-coloradans-with-disorders-so-rare-even/article_d994be10-64f2-11ec-b5e6-539b30f3b61a.html)

Erin Prater

December 31, 2021



When Keegan Joines was born as a low-birthweight baby, his parents saw the rough start as a mere bump in the road.

“He plumped up quickly and everything seemed fine,” said his mom, Susan Joines, an elementary school assistant and pediatric nurse by trade who lives in Castle Rock.

But by the time he was a year and a half, the Joineses were noticing developmental delays, including in walking and speech. A year later he would be diagnosed with Type 1 diabetes, an autoimmune disorder that destroys the body’s ability to create insulin, a vital hormone that allows the body to use glucose for energy.

“We noticed more global abnormalities and we always just kept thinking, ‘Something is related here. These are not all separate instances occurring.’”

Little did they know that, half a decade later, Keegan would be diagnosed with a rare genetic disorder — *KCNJ11*, which affects the pancreas and the brain, resulting in developmental delay and juvenile-onset diabetes; the diagnosis was one of only 30 identified cases in the world at the time similar to his.

Even with the diagnosis, the search for answers continues, as the Joineses await therapies that could unlock his potential, allowing him to develop beyond the kindergarten-to-first grade level he functions at as a 10-year-old — or could have no effect at all.

“Unfortunately we don’t know what his future is — and you never can, even with a well-researched disorder,” his mom said. “Of course, the sky’s the limit for these kids. But, without having much research, we just don’t know what to expect.

“There’s just not enough kids like him to know.”

‘Diagnostic odyssey’

Alone, rare disease can be isolating. The U.S. National Institutes of Health defines a rare disorder as one that affects fewer than 200,000 people nationwide — a definition created by Congress in the 1983 Orphan Drug Act, which established financial incentives for drug companies to develop medications for such conditions.

Collectively, however, rare is common, with approximately 7,000 known diseases affecting an estimated 25 million-30 million Americans — nearly 10% of the population, according to the National Human Genome Research Institute. Worldwide, 263 million to 446 million are affected by rare disorders at any point in time — between 3.5% and 6% of the global population, according to a 2019 article in the *European Journal of Human Genetics*.

With nearly 1 in 10 Americans — and Coloradans — affected by rare disorders, we likely encounter them daily — at schools, at grocery shops, at places of worship, at workplaces. Some prefer to keep quiet, realizing the likelihood of being misunderstood is much greater

than that of finding common ground. Others become vocal advocates, on a quest to raise awareness of a disease very few — if any — others have been diagnosed with. Others yet are oblivious to their disorders, on a quest for an answer to their health maladies that may never materialize.

The quest for a diagnosis the Joineses were on seemed foreign, rare, esoteric. But patients with rare disorders, on average, spent six to eight years — and often untold thousands of dollars — searching for an answer. While waiting, there's the uncertain no man's land of "undiagnosed," a label that can call into question one's symptoms and even one's sanity.

And when a diagnosis is finally received, it's not always the right one.

"It's hard to diagnose people (with rare disorders) — it takes a really long time," said Dr. Anne Pariser, director of the Office of Rare Diseases Research at the National Institutes of Health's National Center for Advancing Translational Sciences.

"This happens so often in rare diseases. We call it the diagnostic odyssey."

Beyond frustration, accompanying the diagnostic odyssey are consequences that have the potential to take a toll on health and finances.

"Being undiagnosed carries a substantial monetary and also human cost," Pariser said. "People are treated for the wrong disease. They don't receive therapies that may be available, or there aren't specific ways that we can intervene to lessen suffering."

Pariser cited a 2019 study by the EveryLife Foundation for Rare Diseases that estimated the economic cost of nearly 400 rare diseases in the U.S. that year at nearly \$1 trillion, surpassing the estimated economic burdens for diabetes, heart disease and cancer — among the costliest common chronic diseases.

There's a mental cost, as well. Without a diagnosis, Susan Joines spent years attributing Keegan's issues to pregnancy complications.

She blamed herself.

"I had to go on beta blockers just for myself to survive because I wasn't profusing to him well," she said. "He just wasn't thriving super well in my body. As we started seeing the gap widening between him and his peers, I was just like, 'I should have done better.' That mom guilt just never goes away, because it always feels like you could do more. Even with your neurotypical kids, you always feel like you're not doing things well enough.

"It's especially hard with a kiddo with extra needs because there's always so much you feel like you should or could be doing."

Complicating each patient’s search for answers is the reality that every rare-disease patient is unique. A patient’s symptoms can be caused by a single gene or chromosomal abnormality; multiple genetic errors; nongenetic factors; or a combination thereof. Even those considered to have the same disorder can have similar but distinct genetic errors that result in different presentations and health outcomes.

When “we think about rare disease, each patient is essentially unique in their characteristics — and that makes studying them, diagnosing them and understanding the public health impacts of rare disease patients very, very challenging,” said Melissa Haendel, chief research informatics officer at the University of Colorado Anschutz Medical Campus and director of the National Center for Data to Health.

Case in point: You’ll find differing estimates of the number of rare diseases, depending on the source and the country the data originates in — more than 7,000, according to the National Institutes of Health; between 5,000 and 8,000, according to the World Health Organization; more than 6,000, according to Rare Diseases Europe.

“Why do we care about how many rare diseases there are? Despite having 10% of the population potentially having a rare disease, the inability to count them really underlies an inability to identify them in the first place,” Haendel said.

“It’s not the count that we care about. The fact that we can’t count them is an indication of our inability to understand and define them, to diagnose them, to treat them.”

Zebras, not horses

For Elliott Wellnitz, 3, of Colorado Springs, the diagnostic odyssey was blessedly short — seven or eight months, as his mom, Christine, recalls.

A few things were “off” during the pregnancy — Christine had only one artery in her umbilical cord instead of two, and Elliott was born prematurely — but “we didn’t know at the time we were going to have a special-needs kid,” she recalls.

But soon medical providers began to point out other anomalies — a widely spaced big toe, port wine stains under his lips, an abnormally large head.

A basic genetic test showed no abnormalities, nor did a more sophisticated test.

The Wellnitzs were sent home with a tank of oxygen, a myriad of specialist appointments and no answers.

Several months later they learned he had Megalencephaly-capillary malformation syndrome — an exceedingly rare genetic syndrome involving developmental delay, intellectual disability, poor muscle tone, parts of the body that are larger than usual and epilepsy. The disorder places patients at risk of fluid buildup in the brain and of cancerous tumors.

For Christine, the diagnosis has meant wearing more hats than parents already wear — those of honorary therapist and educator, and that of an actual nurse.

“When we got out of the hospital, no one pointed me in the right direction,” said Christine, a hospice nurse whose husband stays home and cares for their son. “I kind of had to figure it all out on my own. To me, that’s the most frustrating aspect of this journey.”

Susan Joines ran a group for special needs families where she lived last, working as a parent liaison to the early childhood education system. She found that many parents of children with rare disorders are “desperate for answers and support and help.”

“I feel like very rarely do we get all three of those — or even two out of three of those — from providers.”

She’s encountered a wide variety of personality types in doctors over countless visits, from the kind who say, “Wait, give it time, they’ll be fine, and then they’re 16 and end up with an autism diagnosis” — to the ones who say, “Here you go, here’s your child’s list of problems, and we’ll see you later, completing writing a kid off.”

The former have served as roadblocks, the latter sources of immense hurt and devastation.

A psychologist once administered an IQ test on Keegan without Susan's permission or understanding, then delivered dismal news.

"Essentially they just said he was in the bottom 15th percentile, so probably under 40," she said of his score. "Just basically, 'Here is his IQ, and most likely he'll never live alone, he'll never be able to have a job or live independently,' and they just kind of left it there. We were like, 'Should we follow up with you?' They were like, 'No, this is it.'"

"I feel like they gave him a life sentence."

With myriad rare disorders and such fuzzy definitions of many, it’s often a struggle to find a provider equipped to diagnose, no less treat, a rare disorder.

The problem at least partially originates in medical schools, where doctors in training are taught that “when you hear hoofbeats, think horses, not zebras — common things occur commonly. Don’t give somebody a rare diagnosis if they probably have a common problem,” Pariser said.

Providers need to be trained to look for “zebra triggers,” she said, invoking a symbol of the rare disease community: zebras, which each have a unique stripe pattern, as humans do fingerprints.

“We want (doctors) to think of zebras, and we want them to think of zebras when they start seeing certain clusters of things: young age, high (medical system) utilization, multiple consults, having to travel great distances. Also, some of these what we call ‘basket (medical)

codes,' like 'developmental delay' or 'motor delay.'”

“Rare diseases — we have many, many diseases that affect small numbers of patients each and very few treatments. But when you consider this collectively, it really is a large public health problem.”

‘How long is my child going to live?’

Even with a diagnosis, the future is uncertain for Keegan.

Now 10, he performs in school three to four years behind grade level, “even with extensive developmental services,” Susan said.

“We’ve exhausted just about everything we think we can think to do for him. He struggles greatly with academics. He still can’t write his name. He still isn’t reading well,” she said, adding that, regardless, Keegan is a joyful, humorous child who loves life, his family, school and friends.

“To get him to recognize the word ‘the’ — that’s a (school) goal for him, to have an 85% success rate on just recognizing that word. He’s in a severe special-needs program. We aren’t seeing progress like we hope. Just from the little bit we do know, early to late elementary school is typically, developmentally, where they see him maxing out.”

For Christine Wellnitz, receiving a diagnosis was comforting — but it didn’t come with a road map of what to expect.

“It was definitely a huge relief when there was actually a name for what he had,” she said. “I cried. But there wasn’t a whole lot of information, even on life expectancy.

“How long is my child going to live?”

A Facebook group Christine joined, for those affected and their families, has a couple of older patients in their 30s and 40s, “and that makes me happy.”

“But beyond that, we don’t really know, and that’s pretty scary. I always tell my husband, ‘Right now we’re doing pretty good, and things are going pretty well, but I never hold my breath,’ because every time I think that, something else pops up, or we need to see another specialist.”

Elliott’s pediatrician is supportive but doesn’t have much to offer in the way of specialized knowledge.

“Our pediatric doctor basically just goes with whatever I want,” Christine said. “If I call and say, ‘I think I need this,’ she usually does it. I think part of it is because I’m a nurse. I love our pediatric doctor, but I feel like there should be more specialists in town.

"I really have no idea exactly where my child is" when it comes to development. "I just know that he's somewhat delayed."

When it comes to enrolling her child in school and ensuring he receives proper education and support, "I don't even know what the next steps are. I hear the school districts aren't great when it comes to working with special needs children, and that just puts fear in my heart.

"I haven't heard of one good district in this town, unfortunately."

For Susan Joines, looking to the future is equally tough.

"My husband and I frequently think we might be forever-nesters," she said. "There's such a grieving process — we will never put a cap on him, of course, but it is a process of this potential grief of maybe not achieving the life we hope for him. He may never drive. He may never graduate. He may never have a job. He may never get married. We may never see grandkids from him."

"Maybe we will," she added hopefully.

Susan leans on her personal faith in the fact that God made her son for a purpose and has plans for him.

"But it's definitely not without grief," she said. "It's the heaviest and hardest thing we deal with on a regular basis."

Commentary | And so, we sail: Of diagnoses and demons on this diagnostic odyssey

Erin Prater erin.prater@gazette.com
Dec 31, 2021



Cyrus waves to a window-washer dressed in a Batman costume after his cecostomy surgery, which allows the flushing colon, at Children's Hospital in Aurora in the spring of 2019.

Erin Prater, The Gazette

When I first read the phrase, I thought he was waxing poetic, striving to be both a doctor and a wordsmith — a Renaissance man, of sorts.

“Cyrus’ hypotonia and congenital malformations have led his mother along a diagnostic odyssey for possible underlying genetic causes,” wrote a young doctor at a hospital in Utah, where we’d flown to see a pioneer in genetics.



Diagnostic odyssey.

The phrase was perfect — satisfyingly so.

That’s what this is, I thought. There was a name for this “fresh hell” I’d experienced for the past five years — this string of doctor appointment, doctor appointment, hospital visit, doctor appointment. For this journey to five states, seeking answers to what was “wrong” with my son, who I would later learn has genetic “errors” so rare that it is much easier for doctors to dismiss them than to admit they don’t have the time, interest or expertise to put a name to them.

There was a name for this living out of bags — this grabbing the one we’d half-unpacked from the latest out-of-state medical trip, on my way out the door to the ER. For this grabbing the same bag the next week for a several-day stint at the Ronald McDonald House in Denver during a “tour de force” of appointments.

There was a name for this constant state of confusion — for this being told in the NICU that my son looked “Downsy,” to being told that nothing was “wrong,” to collecting a laundry list of subdiagnoses like tethered spinal cord and macrocephaly, to being diagnosed with a rare form of primordial dwarfism, to being told that diagnosis wrong, to being sent back to the no man’s land of the undiagnosed.

12:26



Others, too, were on this journey of rare disease — each in his or her own boat, wading in isolation, tossing up a flare when an occasional boat passed in the distance.

Having nothing to offer his fellow traveler save an affirmation of his circumstances and threadbare sanity.

You, too? Fair winds, following seas.

The isolation patients with rare disorders feel defies the numbers. There are hundreds of millions of families worldwide being tossed around by the choppy waves of rare disorders. Between 5,000 and 8,000 conditions have been identified, according to the

World Health Organization — all "serious chronic diseases" with the potential to be life-threatening. Approximately one in 15 people worldwide are affected by a rare disease, or will be during their lifetime.

But no two rare patients are the same. Symptoms and presentations vary. Some patients have multiple conditions. And when you're one of only a handful of individuals worldwide identified with a particular issue, it can be difficult to connect with those like you — even in this digital age.

Happily, in Colorado Springs there are groups to support families touched by Down syndrome, cerebral palsy, autism.

But for FG syndrome, for Mitochondrial complex I deficiency, for patients who have both? It is, in all likelihood, a club of one — even on a global scale.

I revel in the fact that there is no one in the world quite like my son. I love my quirky, my contrary. He truly broke the mold — scratch that, he obliterated it.

But there's a price to pay for such uniqueness. No one else truly knows what I'm going through when test results return that can't rule out yet another ultra-rare disorder. When doctors spar over whether or not my son has "mental retardation" or simply global developmental delay and global learning disabilities — as if intellectual disability is that much of a gray area.

In this world, everything is.

Day in, day out, I sail this sometimes stormy sea of gray, flecked with the colorful confetti of my son's personality that he tosses as he zips about.

Cyrus is the sun, warming me — and all those nearby — with his persistent, easy, forgiving love and gooberish smile.

I'm guided by the north star of my "mama gut," following its commands even when they seem more like whims — even when they seem quieter than usual, less authoritative and more confused.

I have no map.

12:26



I have only one constant companion in this small boat, ours alone: my co-captain, my son. Someday he'll need to take over the wheel. Perhaps I can navigate the worst of the waters for him so by the time he reaches adulthood, smoother seas abound.

Perhaps, if I try hard enough, I can steer us to shore before he must take over. Then he can rejoin the rest of the world on land, molded by his journey into an empathetic, compassionate soul, but not daily afflicted by it. Not scarred, but sculpted — like sea glass freshly washed up on a beach, glimmering in the sun.

We have hope of better days ahead.

And so, we sail.

Godspeed.

Erin Prater

Night Metro Editor

Night metro editor, The Gazette



Bob ROEHR

USA



Bob Roehr is a freelance biomedical journalist based in Washington, DC who has written extensively for The BMJ, Leaps Magazine, New Scientist, Scientific American and other publications. Beginning in 1993 and continuing for fifteen years he was Washington correspondent for a dozen gay newspapers, the lead ones being The Bay Area Reporter (San

Francisco) and Windy City Times (Chicago). An historic focus of his reporting has been the biological, environmental, and social factors of HIV and infectious diseases; more recently it has expanded to include the human microbiome and an ecosystem approach to health.

Beyond Henrietta Lacks: How the Law Has Denied Every American Ownership Rights to Their Own Cells

Bob Roehr

Bioethics | Feature Story

A 2017 portrait of Henrietta Lacks.

Collection of the Smithsonian National Portrait Gallery and National Museum of African American History and Culture, Gift from Kadir Nelson and the JKBN Group LLC.

The common perception is that Henrietta Lacks was a victim of poverty and racism when in 1951 doctors took samples of her cervical cancer without her knowledge or permission and turned them into the world's first immortalized cell line, which they called HeLa. The cell line became a workhorse of biomedical research and facilitated the creation of medical treatments and cures worth untold billions of dollars. Neither Lacks nor her family ever received a penny of those riches.

But racism and poverty is not to blame for Lacks' exploitation—the reality is even worse. In

The case that established this legal precedent is *Moore v. Regents of the University of California*.

John Moore was diagnosed with hairy-cell leukemia in 1976 and his spleen was removed as part of standard treatment at the UCLA Medical Center. On initial examination his physician, David W. Golde, had discovered some unusual qualities to Moore's cells and made plans prior to the surgery to have the tissue saved for research rather than discarded as waste. That research began almost immediately.

"On both sides of the case, legal experts and cultural observers cautioned that ownership of a human body was the first step on the slippery slope to 'bioslavery.'"

Even after Moore moved to Seattle, Golde kept bringing him back to Los Angeles to collect additional samples of blood and tissue, saying it was part of his treatment. When Moore asked if the work could be done in Seattle, he was told no. Golde's charade even went so far as claiming to find a low-income subsidy to pay for Moore's flights and put him up in a ritzy hotel to get him to return to Los Angeles, while paying for those out of his own pocket.

Moore became suspicious when he was asked to sign new consent forms giving up all rights to his biological samples and he hired an attorney to look into the matter. It turned out that Golde had been lying to his patient all along; he had been collecting samples unnecessary to Moore's treatment and had turned them into a cell line that he and UCLA had patented and already collected millions of dollars in compensation. The market for the cell lines was estimated at \$3 billion by 1990.

Moore felt he had been taken advantage of and filed suit to claim a share of the money that had been made off of his body. "On both sides of the case, legal experts and cultural observers cautioned that ownership of a human body was the first step on the slippery slope to 'bioslavery,'" wrote Priscilla Wald, a professor at Duke University whose career has focused on issues of medicine and culture. "Moore could be viewed as asking to commodify his own body part or be seen as the victim of the theft of his most private and inalienable information."

The case bounced around different levels of the court system with conflicting verdicts for nearly six years until the California Supreme Court ruled on July 9, 1990 that Moore had no legal rights to cells and tissue once they were removed from his body.

The court made a utilitarian argument that the cells had no value until scientists manipulated "In effect, what Moore is asking us to do is impose a tort duty on scientists to investigate the consensual pedigree of each human cell sample used in research," the majority wrote. In other words, researchers don't need to ask any questions about the materials they are using.

One member of the court did not see it that way. In his [dissent](#), Stanley Mosk raised the specter of slavery that "arises wherever scientists or industrialists claim, as defendants have here, the right to appropriate and exploit a patient's tissue for their sole economic benefit—the right, in other words, to freely mine or harvest valuable physical properties of the patient's body. ... This is particularly true when, as here, the parties are not in equal bargaining positions."

Mosk also cited the appeals court decision that the majority overturned: "If this science has become for profit, then we fail to see any justification for excluding the patient from participation in those profits."

But the majority bought the arguments that Golde, UCLA, and the nascent biotechnology industry in California had made in amici briefs filed throughout the legal proceedings. The road was now cleared for them to develop products worth billions without having to worry about or share with the persons who provided the raw materials upon which their research was based.

Critical Views

Biomedical research requires a continuous and ever-growing supply of human materials for the foundation of its ongoing work. If an increasing number of patients come to feel as John Moore did, that the system is ripping them off, then they become much less likely to consent to use of their materials in future research.

Some legal and ethical scholars say that donors should be able to limit the types of research allowed for their tissues and researchers should be monitored to assure compliance with those agreements. For example, today it is commonplace for companies to certify that their clothing is not made by child labor, their coffee is grown under fair trade conditions, that food labeled kosher is properly handled. Should we ask any less of our pharmaceuticals than that the donors whose cells made such products possible have been treated honestly and fairly, and share in the financial bounty that comes from such drugs?

Protection of individual rights is a hallmark of the American legal system, says Lisa Ikemoto, a *Judgment* series, where authors may only use legal precedent in effect at the time of the original decision.

"Why is the law willing to confer property rights upon some while denying the same rights to others?" asks Radhika Rao, a professor at the University of California, Hastings College of the Law. "The researchers who invest intellectual capital and the companies and universities that invest financial capital are permitted to reap profits from human research, so why not those who provide the human capital in the form of their own bodies?" It might be seen as a kind of sweat equity where cash strapped patients make a valuable in kind contribution to the enterprise.

The Moore court also made a big deal about inhibiting the free exchange of samples between scientists. That has become much less the situation over the more than three decades since the decision was handed down. Ironically, this decision, as well as other laws and regulations, have since strengthened the power of patents in biomedicine and by doing so have increased secrecy and limited sharing.

"Although the research community theoretically endorses the sharing of research, in reality sharing is commonly compromised by the aggressive pursuit and defense of patents and by the use of licensing fees that hinder collaboration and development," Robert D. Truog, Harvard Medical School ethicist and colleagues wrote in 2012 in the journal *Science*. "We believe that measures are required to ensure that patients not bear all of the altruistic burden of promoting medical research."

Additionally, the increased complexity of research and the need for exacting standardization of materials has given rise to an industry that supplies certified chemical reagents, cell lines, and whole animals bred to have specific genetic traits to meet research needs. This has been more efficient for research and has helped to ensure that results from one lab can be reproduced in another.

The Court's rationale of fostering collaboration and free exchange of materials between researchers also has been undercut by the changing structure of that research. Big pharma has shrunk the size of its own research labs and over the last decade has worked out cooperative agreements with major research universities where the companies contribute to the research budget and in return have first dibs on any findings (and sometimes a share of patent rights) that come out of those university labs. It has had a chilling effect on the exchange of materials between universities.

But that was the dawn of a new technological age and standards have changed. Now cell lines are kept in state-of-the-art sub zero storage units, tagged with the source, type of tissue, date gathered and often other information. Adding a few more data fields and contacting the donor if and when appropriate does not seem likely to disrupt the research process, as the court asserted.

Forging the Future

"U.S. universities are awarded almost 3,000 patents each year. They earn more than \$2 billion each year from patent royalties. Sharing a modest portion of these profits is a novel method for creating a greater sense of fairness in research relationships that we think is worth exploring," wrote Mark Yarborough, a bioethicist at the University of California Davis Medical School, and colleagues. That was penned nearly a decade ago and those numbers have only grown.

The Michigan BioTrust for Health might serve as a useful model in tackling some of these issues. Dried blood spots have been collected from all newborns for half a century to be tested for certain genetic diseases, but controversy arose when the huge archive of dried spots was used for other research projects. As a result, the state created a nonprofit organization to in essence become a biobank and manage access to these spots only for specific purposes, and also to share any revenue that might arise from that research.

"If there can be no property in a whole living person, does it stand to reason that there can be no property in any part of a living person? If there were, can it be said that this could equate to some sort of 'biological slavery'?" Irish ethicist Asim A. Sheikh wrote several years ago. "Any amount of effort spent pondering the issue of 'ownership' in human biological materials with existing law leaves more questions than answers."

Perhaps the biggest question will arise when -- not if but when -- it becomes possible to clone a human being. Would a human clone be a legal person or the property of those who created it? Current legal precedent points to it being the latter.

Today, October 4, is the 70th anniversary of Henrietta Lacks' death from cancer. Over those decades her immortalized cells have helped make possible miraculous advances in medicine and have had a role in generating billions of dollars in profits. Surviving family members have spoken many times about seeking a share of those profits in the name of social justice; they intend to file lawsuits today. Such cases will succeed or fail on their own merits. But

Bob Roehr

Bob Roehr is a biomedical journalist based in Washington, DC. Over the last twenty-five years he has written extensively for The BMJ, Scientific American, PNAS, Proto, and myriad other publications. He is primarily interested in HIV, infectious disease, immunology, and how growing knowledge of the microbiome is changing our understanding of health and disease. He is working on a book about the ways the body can at least partially control HIV and how that has influenced (or not) the search for a treatment and cure.

People With This Rare Disease Can Barely Eat Protein. Biotechnology May Change That.

Bob Roehr

Health | Feature Story

The Brown family at the Grand Tetons (2019). Clockwise from left, Christine, Kevin, Keagan, Connor, and Kellen.
Courtesy Brown family

Imagine that the protein in bread, eggs, steak, even beans is not the foundation for a healthy diet, but a poison to your brain. That is the reality for people living with Phenylketonuria, or PKU. This cluster of rare genetic variations affects the ability to digest phenylalanine (Phe), one of the chemical building blocks of protein. The toxins can build up in the brain causing severe mental retardation.

Can a probiotic help digest the troublesome proteins before they can enter the bloodstream and travel to the brain? A Boston area biotech start up, Synlogic, believes it can. Their starting point is an E. coli bacterium that has been used as a probiotic for more than a century. The

But Christine Brown knew none of this when the hospital called saying that standard newborn screening of her son Connor had come back positive for PKU. It was urgent that they visit a special metabolic clinic the next day, which was about a three-hour drive away.

"I was told not to go on the Internet," Christine recalls, "So when somebody tells you not to go on the Internet, what do you do? Even back in 2005, right." What she saw were the worst examples of retardation, which was a common outcome from PKU before newborn screening became routine. "We were just in complete shell shock, our whole world just kind of shattered and went into a tail spin."

"I remember feeding him the night before our clinic visit and almost feeling like I was feeding him poison because I knew that breast milk must have protein in it," she says.

"Some of my first memories are of asking, 'Mommy, can I eat this? There were yes foods and no foods.'"

Over the next few days the dedicated staff of the metabolic clinic at the Waisman Center at the University of Wisconsin Madison began to walk she and husband Kevin back from that nightmare. They learned that a simple blood test to screen newborns had been developed in the early 1960s to detect PKU and that the condition could be managed with stringent food restrictions and vigilant monitoring of Phe levels.

Everything in Your Mouth Counts

PKU can be successfully managed with a severely restricted diet. That simple statement is factually true, but practically impossible to follow, as it requires slashing protein consumption by about 90 percent. To compensate for the missing protein, several times a day PKU patients take a medical formula – commonly referred to simply as formula – containing forms of proteins that are digestible to their bodies. Several manufacturers now add vitamins and minerals and offer a variety of formats and tastes to make it more consumer friendly, but that wasn't always the case.

“When I was a kid, it tasted horrible, was the consistency of house paint. I didn't think about it, I just drank it. I didn't like it but you get used to it after a while,” recalls Jeff Wolf, the twang of Appalachia still strong in his voice. Now age 50, he grew up in Ashland, Kentucky and was part of the first wave of persons with PKU who were identified at birth as newborn screening was rolled out across the US. He says the options of taste and consistency have

more than the body can handle and toxic levels of Phe begin to accumulate in the brain.

“Some of my first memories are of asking, ‘Mommy, can I eat this? There were yes foods and no foods,’” recalls Les Clark. He has never eaten a hamburger, steak, or ribs, practically a sacrilege for someone raised in Stanton, a small town in northeastern Nebraska, a state where the number of cattle and hogs are several-fold those of people.

His grandmother learned how to make low protein bread, but it looked and tasted different. His mom struggled making birthday cakes. “I learned some bad words at a very young age” as mom struggled applying icing that would pull the cake apart or a slice would collapse into a heap of crumbs, Les recalls.



Les Clark
Courtesy Clark

Controlling the diet “is not so bad when you are a baby” because that's all you know, says Jerry Vockley, Director of the Center for Rare Disease Therapy at Children's Hospital of Pittsburgh. “But after a while, as you get older and you start tasting other things and you say, Well, gee, this stuff tastes way better than what you're giving me. What's the deal? It becomes harder to maintain the diet.”

First is the lure of forbidden foods as children venture into the community away from the watchful eyes of parents. The support system weakens further when they leave home for college or work. “Pizza was mighty tasty,” Wolf' says of his first slice.

Vockley estimates that about 90 percent of adults with PKU are off of treatment. Moving might mean finding a new metabolic clinic that treats PKU. A lapse in insurance coverage can be a factor. Finally there is plain fatigue from multiple daily dosing of barely tolerable formula, monitoring protein intake, and simply being different in terms of food restrictions. Most people want to fit in and not be defined by their medical condition.

Jeff Wolf was one of those who dropped out in his twenties and thirties. He stopped going to clinic, monitoring his Phe levels, and counting protein. But the earlier experience of living with PKU never completely left the back of his mind; he listened to his body whenever eating too much protein left him with the “fuzzy brain” of a protein hangover. About a decade ago he reconnected with a metabolic clinic, began taking formula and watching his protein intake. He still may go over his allotment for a single day but he tries to compensate on subsequent days so that his Phe levels come back into balance.



Jeff Wolf on a boat.

Courtesy Wolf

Treatments

Most rare diseases have no treatment. There are two drugs for PKU that provide some benefit to some portion of patients but those drugs often have their own burdens.

KUVAN® (sapropterin dihydrochloride) is a pill or powder that helps correct a protein folding error so that food proteins can be digested. It is approved for most types of PKU in adults and children one month and older, and often is used along with a protein-restricted diet.

“The problem is that it doesn’t work for every [patient’s genetic] mutation, and there are hundreds of mutations that have been identified with PKU. Two to three percent of patients will have a very dramatic response and if you’re one of those small number of patients, it’s great,” says Vockley. “If you have one of the other mutation, chances are pretty good you still are going to end up on a restricted diet.”

PALYNZIQ® (pegvaliase-pqpz) “has the potential to lower the Phe to normal levels, it’s a real breakthrough in the field,” says Vockley. “But is a very hard drug to use. Most folks have to take either one or two 2ml injections a day of something that is basically a gel, and some individuals have to take three.”

Many PKUers have reactions at the site of the injection and some develop anaphylaxis, a severe potentially life-threatening allergic reaction that can happen within seconds and can occur at any time, even after long term use. Many patients using Palynziq carry an EpiPen, a self-injection device containing a form of adrenaline that can reverse some of the symptoms of anaphylaxis.

Then there is the cost. With the Kuvan dosing for an adult, “you’re talking between \$100,000 and \$200,000 a year. And Palynziq is three times that,” says Vockley. Insurance coverage through a private plan or a state program is essential. Some state programs provide generous coverage while others are skimpy. Most large insurance company plans cover the drugs, sometimes with significant copays, but companies that are self-insured are under no legal obligation to provide coverage.

Les Clark found that out the hard way when the company he worked for was sold. The new owner was self-insured and declined to continue covering his drugs. Almost immediately he was out of pocket an additional \$1500 a month for formula, and that was with a substantial discount through the manufacturer’s patient support program. He says, “If you don’t have an insurance policy that will cover the formula, it’s completely unaffordable.” He quickly began to
It’s easy to see why PKUers are eager for advances that will make managing their condition more effect, easier, and perhaps more affordable. Synlogic’s efforts have drawn their attention and raised hopes.

Just before Thanksgiving Jerry Vockley presented the latest data to a metabolism conference meeting in Australia. There were only 8 patients in this group of a phase 2 trial using the original version of the company’s lead E. coli product, SYN1618, but they were intensely studied. Each was given the probiotic and then a challenge meal. Vockley saw a 40% reduction in Phe absorption and later a 20% reduction in mean fasting Phe levels in the blood. The product was easy to use and tolerate.

The company also presented early results for SYN1934, a follow on version that further genetically tweaked the E. coli to roughly double the capacity to chop up the target proteins. Synlogic is recruiting patients for studies to determine the best dosing, which they are planning for next year.

“It’s an exciting approach,” says Lex Cowser, Director of Research Development at the [National PKU Alliance](#), a nonprofit that supports the patient, family, and research communities involved with PKU. “Every patient is different, every patient has a different tolerance for the type of therapy that they are willing to pursue,” and if it pans out, it will be a welcome addition, either alone or in combination with other approaches, to living with PKU.

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Anna Sawerthal is an editor at the Austrian daily newspaper „Der Standard“, at the international desk. Her main focus of interest is Asian affairs. She received her MA degree in Tibetan and Buddhist Studies from the University of Vienna and studied

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Thin air: rare diseases overshadowed by the corona pandemic

ST derstandard.at/story/2000132218266/duenne-luft-seltene-krankheiten-im-schatten-der-corona-pandemie

Global health

Even in normal times, patients with rare diseases are not very well heard. During a pandemic in Nepal, patients with pulmonary hypertension are dependent on NGOs

Anna Sawerthal from Kathmandu

December 30, 2021, 6 a.m.

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In the wake of the Covid wave last spring, the medical facilities in Nepal were well over their limit.

Ram is lying on the couch with a breathing tube hanging from his nose through which he supplies oxygen to his body. The 42-year-old looks into his cell phone camera; he is almost 24 hours by car from Kathmandu, in a city in southern Nepal. Talking is sometimes difficult for him. Again and again he has to take a break and take a deep breath.



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It all started 13 years ago, he says. Suddenly he got a fever. He had to cough a lot back then, and on the third day there was blood. In the local hospital, the doctors first diagnosed tuberculosis - a disease that is common in Nepal. After further testing, they eventually found a heart problem and sent him to Kathmandu. He only got his diagnosis from a specialist in the capital: pulmonary hypertension, i.e. pulmonary hypertension (PH).

The disease is considered a "rare disease". According to the European definition, such a disease affects less than 1 in 2,000 people. According to US standards, it occurs less than 200,000 times in the country. In developing countries, however, the extent of these diseases has usually not been investigated.

Long and expensive journey

Mahindra was also diagnosed with PH in adolescence. Today he is almost 50, an operation as a child would have completely eliminated the problems. But when he went to a clinic in India as a teenager, it was too late for that. "In a country like Nepal - back then there were no heart surgeons," he says. On some days he can only walk a few hundred meters at a time, and he quickly becomes out of breath. The 47-year-old should actually have a check-up twice a year. But the journey to Kathmandu is long and expensive.

Lungenhochdruck liegt eine Verengung der Lungengefäße zugrunde. Das Blut muss also mit erhöhtem Druck gepumpt werden. Das kann sich langfristig wiederum auf das Herz auswirken, das überbeansprucht wird. Im schlimmsten Fall kann es zu Herzversagen kommen. PH kann entweder durch Erkrankung der Lungenarterien an sich entstehen, oder er tritt als Begleiterscheinung anderer Erkrankungen auf – was häufiger der Fall ist.

Die Corona-Pandemie hat ein neues Schlaglicht auf Lungenerkrankungen weltweit geworfen. Aber schon vor der Pandemie hatte die Weltgesundheitsorganisation WHO gewarnt, dass Lungenerkrankungen in den nächsten Jahren steigen werden.

Große Höhe als Risikofaktor

Im Westen findet PH daher immer mehr Beachtung. Manche werfen sogar die Frage auf, ob man überhaupt noch von einer seltenen Krankheit sprechen sollte. Doch in wirtschaftsschwachen Ländern wie Nepal fehlt es oft an Möglichkeiten zur Diagnose, zur Behandlung und an Geld.

Dabei herrschen gerade in Nepal Faktoren vor, die das Risiko, an PH zu erkranken, drastisch erhöhen. Dazu zählen die enorme Luftverschmutzung in der Hauptstadt oder Abgase durch das Kochen am offenen Feuer. Ein entscheidender Faktor ist aber vor allem: die Höhe. Im Himalaja-Land Nepal leben mehr als die Hälfte der knapp 30 Millionen Einwohner in den Bergen. Während sich manche Höhenbewohner über Generationen an die veränderten Bedingungen angepasst haben, zeigen Studien, dass der längere Aufenthalt in großer Höhe zu chronischem PH führen kann.

Um solche Fälle aufzudecken, hat Rozam Khatiwada schon mehrere Gesundheitscamps in ländlichen Gegenden in Nepal durchgeführt – nicht nur in den Bergen, aber auch. Im Solu-Khumbu-Gebiet etwa, der Heimat der Sherpas am Fuße des Mount Everest, hat der Arzt aus Kathmandu mit seinem Team rund 1.000 Schüler und Schülerinnen untersucht. Rund 30 zeigten bei den Grunduntersuchungen auffällige Werte und wurden daher zur weiteren Abklärung in die Hauptstadt gebracht.

Eine der Patientinnen war eine elfjährige Vollwaise, die in sehr schlechtem Zustand war, erinnert sich der Arzt. In Kathmandu hatten sich die Ärzte rasch zu einer Operation entschieden. Nur sieben Tage später hatte er sie kaum wieder erkannt. "Es ist eine echte Erfolgsstory, es war unglaublich! Wenn wir sie nicht operiert hätten, hätte sie vielleicht noch drei oder vier Jahre gehabt. Jetzt geht es ihr aber gut."

Abgeschnitten vom Gesundheitswesen

Organisiert werden die Camps von der NGO [PHA Nepal](#). Die Organisation will das Bewusstsein um die Krankheit vor allem in abgelegenen Gebieten erhöhen, wo die Menschen vom Gesundheitswesen des Landes größtenteils abgeschnitten sind. Rozam und seine Kollegen wollen dabei vor allem Kinder und Jugendliche unter 14 Jahren, die noch keine Symptome zeigen, finden, um sie rechtzeitig behandeln zu können.

Finanziert wird die Organisation hauptsächlich von Spendern aus den USA, erzählt Rozam. Der US-Nepalese Choodamani Kanal hat die Organisation gegründet, nachdem er selbst an PH erkrankt war. Neben den Camps stellt PHA Nepal Geld für OPs, Transport und Nachversorgung zur Verfügung. Denn in Nepal existiert kaum medizinische Grundversorgung. Auch Ram ist bei PHA Nepal tätig. Eigentlich hatte er Umwelttechnik studiert, doch aufgrund seiner Erkrankung kann er nicht arbeiten und lebt in seinem Elternhaus. Die Medikamente, die er nehmen muss, sind teuer. Ohne Hilfe von außen würde das Geld oft nicht reichen.

Rare Infrastruktur

PH-Patientinnen wie Sajana haben mehr Glück: Als Krankenschwester in einem großen Spital in der Nähe von Katmandu bekam sie rasch die richtige Diagnose und auch die richtige Behandlung, deren Kosten außerdem vom Spital übernommen wurden. Das Dulikhel Hospital gehört etwa zu jenen Einrichtungen, die das volle Programm an Technik bieten können: Echokardiogramme, Computertomografien, Bluttests oder auch Katheter.

Die Covid-Pandemie hat allerdings gezeigt, wie schnell das Gesundheitssystem in Nepal ans Limit gerät. Rajendra Kaju, Leiter des Spitals, bekommt jetzt noch Gänsehaut, wenn er an die zweite, verheerende Welle im vergangenen Mai denkt. Ihnen ist schlicht der Sauerstoff für die vielen Intensivpatienten ausgegangen. Und doch hat die Pandemie auch ein neues Bewusstsein für Lungenkrankheiten geschaffen. Der Spezialist für Höhenkrankheit Buddha Basnyat nannte die neuen Forschungsoperationen, die durch die Pandemie entstehen, jüngst [in einem Podcast des "British Medical Journal"](#) die "gute Seite" in all der Düsternis.

However, access to this infrastructure is still only possible for a small part of the population. Rozam Khatiwada is already planning the next health camp. In the best case, the doctor says, he would like to go out once a month. He wants to uncover as many cases as possible - and collect as much patient data as possible to put pressure on the government. And ideally he would like to buy a portable echo device. Then important examinations could be carried out on site - ideally in order to be able to give the all-clear right away. (Anna Sawerthal from Kathmandu, December 30th, 2021)

The research was funded by the National Press Foundation and the Ipsen Foundation. All patient names have been changed.



Limpho SELLO

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Limpho Sello is full-time journalist attached to Lesotho Times and Sunday Express newspapers based in Maseru Lesotho.

She started her career eight years ago and has keen interest on health related issues. She holds an Associate Degree in Journalism and Media.

The award-winning journalist is working hard to grow her brand name both nationally and internationally.

Limpho has written general news, politics, health, social and human rights issues until she moved and found passion in health and social issues. She is a recognised senior Health and Social issues Journalist

in the country. She has received recognition from the Ministry of health where she scooped her first award as a best health reporter in 2016; appreciation award from the Lesotho Nurses Association in April 2021 and overall winner of 2021 LNIG/MISA Lesotho Women in Media Awards.

Limpho has been fortunate to become part of the 52nd Training Course for Young African Journalists in Cairo, Egypt in 2018. She participated at the Breaking Down Borders Africa Youth summit in Johannesburg, South Africa in 2019, and the 3rd Southern Africa Youth Forum (SAYoF) 2021 in August 2021, as a panellist.

Govt, QMMH Accused Of Denying Minor Life-Saving Operation



By Lesotho Times

On Dec 13, 2021

FEATURES & ANALYSIS LOCAL NEWS NEWS



Queen 'Mamohato Memorial Hospital (QMMH)

Limpho Sello

A HA-LEQELE, Maseru family has accused the government and Queen 'Mamohato Memorial Hospital (QMMH) authorities of denying their six-year-old daughter an opportunity to go for crucial surgery to remove a brain tumor in Cape Town.

The child, Lerato Moshoeshe*, was diagnosed with a brain tumor shortly after birth at QMMH.

Two years ago, specialists in Bloemfontein attended to Lerato and referred her to the Red Cross Children's Hospital in Cape Town for surgery. They said the child's life was under threat and the only thing that could save her was surgery at the Cape Town facility.

However, she has still not been operated on. Her parents are accusing the government and QMMH authorities of throwing spanners in the works by refusing to write the necessary referral letter for the minor whose identity has been withheld due to her age.

Narrating her ordeal in a recent interview with the *Sunday Express*, the Lerato's mother, Lisebo Moshoeshoe*, said she was born in December 2015 with tuberous sclerosis syndrome – a rare multisystem genetic disease that causes the growth of tumors.

The syndrome causes overgrowth of tissues in different parts of the body. In the case of Lerato, the overgrowth is on the left side of her brain.

“The scans showed that she has a rare condition called tuberous sclerosis,” Ms Moshoeshoe said.

“This is a rare disease that causes growths in the brain and other organs. The condition is not cancerous but it can be fatal if it gets severe. With my daughter, the growths are on the left side of the brain and that is why her right side is very weak. When the tumors are on the left, the right side of the body is the one that is affected.

“We were referred to a Bloemfontein doctor who in turn referred Lerato to Cape Town for surgery after she had started experiencing seizures. The doctor wrote in Lerato's medical booklet, saying he had failed to control the seizures and that the only option to save her life was a brain surgery by specialists at the Red Cross Children's Hospital.

“Unfortunately, we hit a rock at QMMH (then run by the Tšepong Consortium on behalf of the government). They told us that their agreement with the government did not allow them to fund the operation in Cape Town,” Ms Moshoeshoe said.

She said one her friends advised them to seek help from Rare Diseases South Africa – a non-profit organisation which is dedicated to excellent health care for people with rare diseases.

She said QMMH would not give them the referral letter even after being assured that the South African organisation would help facilitate Lerato's admission in Cape Town for the surgery.

Lerato's father, Lerotholi Moshoeshoe*, also expressed his anger and frustrations at the authorities' refusal to help them.

“Lerato's health continues to deteriorate. She is currently on four different medications to reduce frequency of seizures. Ours continues to a struggle due to delaying tactics by the authorities.

“At one-point Tšepong said the referral letter must written by the management at the Bloemfontein hospital that referred Lerato to Cape Town. Out of desperation, we went to the extent of writing to the Director General of Health Services, Nyane Letsie, begging for the referral letter. We even attached copies of Lerato's medical records but to no avail,” Mr Moshoeshoe said.

The *Sunday Express* has made several unsuccessful attempts to obtain comment from Dr Letsie. Most of the times, her mobile phone rings unanswered and on the rare occasions it is answered, she says she will be unable to talk because she is in meetings.

This publication has similarly got no joy from the health ministry's principal secretary, Khothatso Tšooana, who also said he was in a meeting and referred all questions to Dr Letsie.

QMMH Deputy Director General for Clinical Services, Lipontšo Makakole, said they were only allowed to refer patients to hospitals in Bloemfontein, hence they could not refer Lerato to Cape Town.

“You are talking about a patient who has been referred to hospitals that are not approved by the government. There is no way we can write referrals to hospitals that are not approved by the government,” Dr Makakole said, adding Lerato’s case had to be addressed by the health ministry itself.

Valuable time wasted.

Valuable time has been wasted. Lerato’s condition has been deteriorating over the past two years that both the Ministry of Health and QMMH have failed to assist the Moshoeshoe family with the much-needed referral letter.

While the family has been shunted from one office to another by the authorities, Lerato’s tumor has been growing bigger.

“The sad thing is that Lerato was a lot better than she is now when we were told to go to Cape Town. She was still a normal child but her condition has worsened since 2020. At times I look at her and think that she is now mentally disturbed.

“She has even developed other diseases. At Bloemfontein, they have also recommended speech therapy which she was attending at Ladybrand Hospital. She has had to stop going for sessions after the therapist moved to Bloemfontein,” Ms Moshoeshoe said.

She was full of praises for her husband who she says has been fully supportive to the extent of preparing different meals for Lerato whenever she refuses to eat what has been placed before her.

“He gives everything when it comes to caring for his daughter. He attends to her more than anyone else. When she refuses to eat, *Ntate* Moshoeshoe goes out of his way to prepare something else for her.

“If Lerato refuses to eat pap and milk, he will prepare noodles. If she still refuses, he will prepare spaghetti and try everything until she agrees to eat. He even has time to sit down with her and our son to watch cartoons. He now knows all the cartoons now,” Ms Moshoeshoe said.

Mr Moshoeshoe said it was important for the family to stick together in such trying times and he was prepared to do anything to assist his wife and daughter.

“I accompany my wife whenever Lerato has to go for check-up. Sometimes I even go with Lerato on my own.

“Even when my masculine side makes me feel otherwise, I fight the feeling and remind myself that we are in this together as a family,” Mr Moshoeshoe said.

Lerato could die

But all the maternal and paternal love will not suffice. It is no substitute for the medical attention that Lerato needs.

With a shaky voice and tears streaming down her cheeks, Ms Moshoeshoe fears that Lerato will soon die if she does not get the life-saving brain surgery as per the medical advice of the Bloemfontein specialists.

“Last year we were told that with every seizure, there is some brain damage that occurs. I get mad knowing that my daughter has been denied a simple referral letter that would enable her to have the surgery she deserves. What has made us angrier is the unsympathetic attitude of a QMMH staffer who said the money required for Lerato’s treatment could be used to help many other children instead of assisting just a single child.

“It would have been better if they had given us the referral letter and left us to struggle on our own afterwards. Things might have been different but as it is, QMMH has failed us by denying us the assistance our baby needs.

“Right now, I’m not coping at all. Every time I hear about epileptic children who have died, I just cry the whole night because I have an epileptic child.

“All we need is for our daughter to be given the referral letter so she can go to Cape Town for surgery. Even her Bloemfontein doctor pitied us, saying ‘if you were South Africans, I would send you to Cape Town right away before this thing becomes cancerous.

“We are appealing to the authorities to urgently help us with the referral letter to Cape Town. We are prepared to take her anywhere for assistance. They can even refer to India and she will go there as long she can get help to enable her to live a normal life,” Ms Moshoeshoe said.

***These are not the family’s real names. Pseudonyms have been used to protect the minor child’s identity.**



Stephen TSOROTI

Zimbabwe



Stephen Tsoroti is a journalist from Zimbabwe. He is a versatile writer who reports on a number of beats that including health, environment, politics, human rights, arts and culture, and developmental issues. Some of his articles have resulted in policy shifts at government level and sparked national debate in Zimbabwe. Since working as a full time and as a freelance journalist, Stephen's work has been published in his home country and abroad. He has won several journalism awards and fellowships that includes; Journalist of the Year Award (Zimbabwe

Biodiversity Award 2000), Best Idea Awards (The British Council Arts Competition Award 2000), Journalist of the year Award (Environment Africa Award 2015), World Bank Investigative Journalism fellowship Arusha Tanzania (2004), National Press Foundation: Journalist to Journalist health fellowship (2015), Money Trail Illicit Financial Flows Investigation Project Grant recipient- Journalismfund.eu (2019), Thomson Reuters Foundation fellow,- COVID-19 Crisis Reporting Hub – sub-Saharan Africa (2020).

Tales of the covid-19 burden on other diseases

zambianeye.com/tales-of-the-covid-19-burden-on-other-diseases

December 9, 2021 | Filed under: [Breaking News, Health & Nutrition](#) | Posted by: [Editor 2](#)
652 Views



...the impact of covid-19 on rare diseases...

Stephen Tsoroti

In March 2020, Kamwala Township south of Lusaka, Zambia's capital city wore an ethereal frown. The deafening yells and shouts from street vendors were not heard. The often crowded streets were deserted, a creepy air patched every house. It was as if an apparition had visited and declared death upon it.

It was no longer a rumour.

The virus was here and was killing people. For now, the only remedy was to stay indoors," described Mainess Chileshe. For me, it was the stark reality that our visit to the University Teaching Hospital (UTH) will be restricted, and even if I go, the chances of getting the regular supply of moisturizers and ointment needed for my child will not be there.

Since then, the situation has not changed much.

“We are still struggling to get the basic treatment for our child,” says Chileshe.

At the dawn of COVID-19 in Zambia in 2020, Chileshe's daughter Peggy 6 years old, had been diagnosed with ichthyosis vulgaris an inherited skin disorder in which dead skin cells accumulate in thick, dry scales on the skin surface.

The scales also called fish scales disease or fish skin disease can be present at birth but first appear during early childhood. Though not contagious. It is a life condition, and a genetic disorder which occurs 1 in 10,000 people.

Doctors at UTH prescribed hydrocortisone ointments and recommended that the family buy creams to keep her skin moisturized at all times.

For Young Kumbirai Nhondo* in Zimbabwe, COVID-19 was a body blow. he could not survive the waiting. After years of mis-diagnosis physicians found the cause to be Mucopolysaccharidosis (MPS) type 1, -is a rare disease in which the body does not have enough of an enzyme needed to breakdown long chains of sugar molecules and remedied a drug.

The problem was that the medicines were not available in the country and the family decided to source the drugs from USA. With virtually the entire world under lock down Nhondo's medicines came months late after he had succumbed to the disease.

Since the COVID-19 days, people living with Rare Disease (RD) in southern Africa have been disillusioned. Their stories-punctuated by sad encounters of not having access to healthcare services, clinical trials being halted, patients visit to health care facilities or trial sites not possible due to risk of COVID-19 infection, health care providers busy caring for covid-19 patients and unable to carry out day to day clinical trials duty, many laboratories for scientific have been closed due to COVID-19 public health concerns.

Trudy Nyakabangwe head of the Rare Diseases Foundation in Zimbabwe portrays situation as dire as the pandemic has stretched families seeking treatment. She says through her organizations work, families have been struggling to get diagnostic tests, get treatment and access care services as COVID-19 restrictions take center stage.

It's heart rending, we have to find fast solutions to arrest the situation.

Nyakabangwe feels ways and means should be found quickly to simplify the tortuous regulatory process that pharmaceutical wholesalers, NGOs and health institutions must undergo to bring drugs to market to help southern Africa consumers of life saving drugs,” says Nyakabangwe.

Pediatric and Consultant at Parirenyatwa Hospitals Pediatric unit in Harare Loyce Hlatwayo depicts COVID-19 as a barrier to diagnosis, treatment and drug supply. But sees the lack of statistics around RD as an impediment to drug supply issues.

We have to count all the cases for us to get a total picture of rare conditions in the country and region.

Hlatwayo contends that whereas the country and region has done much in communicable disease, adding that the availability of specialized diagnostic equipment and registry can produce different results.

“It is ineffective to deal with the bane of RD and the drug supply side without addressing the records and registry challenges,” challenges Hlatwayo.

Haem-oncologist Hlatwayo indicates that rigorous effort needs to be concentrated on finding the numbers, pushing for proper records and putting up a registry to inform stakeholders to ease the drug supplies side as well as showing the trends of rare conditions in the region.

Melissa Haendel university of Colorado Anschutz in the USA is of the same mind, knowing how many RD are, is critical in order to know their treatment regime. If rare diseases are not counted rare disease patients will not count.

“To know how many rare diseases there are, clinical systems require a common set of definitions to best aid diagnosis and care. We need publicly available information about individuals with rare diseases because better counting of rare diseases will lead to better patient outcomes,” says Haendel.

Generally, life threatening, chronic and incapacitating RD have not been captured well in national records of southern African countries, leading to health experts, researchers and care givers concluding that lack of data is a barrier to the procurement of RD drugs.

On the other hand, drug manufacturing and pharmaceutical companies worldwide have favored volumes and profit in the supply of drugs, but the same companies claim that drug development also requires patient recruitment in clinical trials, regulatory requirements, profitability and sustainability.

Unlike countries in the west where robust generic industry exists, only one country in Africa, South Africa, has made strides in becoming a high tech hub of pharmaceutical distributor and manufacturer of generic drugs.

But South Africas pharm market, and local production scene is dominated by generics mostly focused on the countrys more prominent killers HIV/ AIDS, tuberculosis TB and hepatitis A and B, as well as common medications for influenza, pain and other everyday ailments.

That scenario leaves Zimbabwe and its neighbors under severe pressure to import from elsewhere, where the process becomes expensive and cumbersome for people with RD.

Battered economies and deteriorating health facilities and service infrastructure, also worsens the southern African countries abilities to procure drugs for its people.

Local health experts say the will to combat RD will also hinge on the governments prioritizing non-communicable diseases.

Which means countries like Zimbabwe, with enabling local drug regulatory bodies such as the Medicine Control Authority of Zimbabwe (MCAZ), can do much to advocate and recommend funding for research or procurement of life-saving drugs.

Perhaps coming late though, the MCAZ intends to discuss with various patients interfacing bodies to understand the needs of members of community that need guaranteed access to life-saving medicines. “We are aware of the supply challenges that have come as a result of COVID-19 pandemic,” says MCAZ Projects and Public relations officer Shingia Gwatidzo.

“It makes sense at the moment to advocate for more drugs importation, but in future we need more recognition at home and internationally that patients with rare conditions needs critical attention, maintains Nyakabangwe. If we have a goal to attain universal health, then all people need to included.”

According to the International Rare Diseases Research Consortium Assembly, 500 rare disease drugs have reached the market, and there are 700-800 treatments in development. Worldwide, 300 million people worldwide are known to have a rare disease.

Anne Pariser Managing Director office of Rare Disease Research National Centre for advancing translational sciences NH, warns — each year 200 to 250 new rare diseases are being discovered, of which 50 percent of them first manifest in children. How much in Africa? – is a big guess.

Meanwhile, Chileshe pins her hopes on improved supply of medicines, hoping COVID-19 behaves well.

*correct names of source changed to protect their privacy



David WAHLBERG

USA



David Wahlberg has been the health reporter at the Wisconsin State Journal in Madison since 2005. He previously covered health and other topics at the Atlanta Journal-Constitution, Ann Arbor (Mich.) News, San Bernardino (Calif.) Sun and Wausau (Wis.) Daily Herald.

At the State Journal, his projects on patient safety, rural health care, doctor discipline, organ transplants and cancer research have won national awards.

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https://madison.com/wsj/news/local/health-med-fit/newborn-screening-not-equal-among-states-leaving-some-babies-untreated/article_e42c56e2-7416-56c8-96be-ea8620abf70c.html

TOPICAL ALERT

NO TIME TO LOSE | FINDING RARE DISEASES IN INFANTS

Newborn screening not equal among states, leaving some babies untreated

From the No time to lose: Finding rare diseases in infants series

David Wahlberg | Wisconsin State Journal
Dec 20, 2021



Kyle and Jenna Heckendorf, of Spring Green, with children Carter, 6, and Ava, 2, look at photos of Bryce, the couple's first child, who died at 18 months old from a rare disorder called Krabbe disease. Ten states test newborns for Krabbe, allowing parents to seek early treatment. Wisconsin has twice rejected screening for Krabbe. "I wouldn't want any other family to have to go through that, because it was devastating," Jenna Heckendorf said.

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David Wahlberg | Wisconsin State Journal

With blue eyes and a tuft of blond hair, Jenna and Kyle Heckendorf's first child, Bryce, seemed healthy, smiling and giggling during a family vacation at a lake resort.

Jenna and Kyle Heckendorf lost their son, Bryce, to Krabbe disease seven years ago when he was 18 months old

But shortly after he turned 5 months old, he started eating less. His arms and legs became rigid. When placed on his stomach, he would wail.

He was diagnosed with Krabbe disease, an inherited disorder that causes muscle weakness and other problems, **typically becoming fatal by age 2.**

“They handed us tissues and said, ‘We’ll help you keep him comfortable as he passes away,’” said Jenna Heckendorf, a teacher from Spring Green, whose son died at 18 months old seven years ago after being on a feeding tube and an oxygen machine.



At Kyle and Jenna Heckendorf's home in Spring Green, family photos include Bryce, their oldest child, who died in 2014 from a rare condition not included in Wisconsin's newborn screening program.

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Ten states test infants for Krabbe

(pronounced crab-AY), using a few drops of blood collected from their heels a day or two after birth for routine newborn screening. Families whose babies have the condition can try stem cell transplants, which studies show can lead to longer and more normal lives if done within 30 days, before symptoms appear.

Wisconsin has twice rejected adding Krabbe to its newborn screening

program. State officials say it's not clear how babies would be sent out of state to the few centers that do the risky transplants for infants with the condition. A **bill before the state Legislature** would circumvent Wisconsin's scientific committee process for evaluating conditions and require the state to test babies for Krabbe.

“We wish we would have had that option,” Heckendorf said of a transplant. “It gives you hope ... versus being told there's nothing you can do.”

Nationwide, newborn screening has tested babies for rare diseases that are unnoticeable at birth but treatable **since the mid-1960s**, with some states checking for only a few disorders in the initial decades and others detecting many. Despite a federal effort to make testing more consistent in 2010, states still choose which conditions to add, and considerable variation remains.

Connecticut **screens for 73 diseases**, while **Hawaii looks for 28**, according to the federal Health Resources and Services Administration, or HRSA. **California and Tennessee test for 67 disorders**, while **Louisiana does 30** and Alabama, Arizona and Arkansas **do 31**, the agency says. Wisconsin's **panel includes 47 disorders**.



In a storage chest in their bedroom, Kyle and Jenna Heckendorf of Spring Green keep belongings of their late son Bryce, including his favorite book, "Commotion in the Ocean." They have donated copies of the book to UW Health's American Family Children's Hospital, where Bryce received much of his care. With them is daughter Ava, 2.

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Debates over cost, the prevalence and severity of an illness, the effectiveness of treatments and false positive results account for much of the disparity, said Natasha Bonhomme, director of **Baby's First Test**, a newborn screening education center in Washington, D.C. Some states require legislative approval to add conditions, and others rely on doctors, with varying levels of public input.

States name and count some conditions differently, so the magnitude of differences is hard to pin down, Bonhomme said.

“If you see one state is screening for 30-some conditions and another state is screening for 60-some conditions, the variation may not actually be a difference of 30 but there is going to be a difference,” Bonhomme said.



Bonhomme



Many Wisconsin hospitals send some newborn screening samples late

Federal recommendations

In 2010, the federal government adopted a **Recommended Uniform Screening Panel**, or RUSP. It currently lists 35 conditions states should screen for and 26 other disorders that can be picked up when testing for the core group.

“We continue to push for states to definitely provide all 35 of the core screenings,” said Stacey Brayboy, a senior vice president at the March of Dimes, which has **long promoted newborn screening**.

Some disorders on the list are relatively well known, such as cystic fibrosis and sickle cell disease, but most are unfamiliar and have complicated names. With most, each parent is an unaffected carrier — usually, unknowingly — and each of their children has a 25% chance of getting the disease.



Like many parents, Jenna and Kyle Heckendorf, shown with daughter Ava, 2, didn't pay much attention to newborn screening — tests for rare diseases using a few drops of blood taken a day or two after birth. “We knew newborn screening existed, but we didn't look into what was screened for,” Jenna said.

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The prevalence of the conditions ranges from roughly 1 in 3,000 babies to 1 in 300,000. Combined, when hearing loss is included, about 1 in 300 newborns, or **nearly 13,000 U.S. babies a year**, has one of the disorders.

The diseases typically can be treated with special diets, therapies such as enzyme infusions or procedures such as stem cell transplants. In most cases, doctors say early treatment is essential to prevent death or disability.

Missing early treatment

Wisconsin screens for 32 of the 35 core conditions. The state doesn't test for **Pompe disease, which disables the heart and skeletal muscles**, and was **added to the RUSP in 2015**. It doesn't screen for **mucopolysaccharidosis type 1, or MPS 1**, a disease that affects many parts of the body, or **X-linked adrenoleukodystrophy**, also called X-ALD, which gained attention in the 1992 film "Lorenzo's Oil." Both were **added to the RUSP in 2016**.

Twenty-seven states screen for Pompe, 26 test for MPS 1 and 22 look for X-ALD, **according to NewSTEPS**, a resource center run by the Association of Public Health Laboratories.

The Wisconsin Department of Health Services **approved adding Pompe (pom-PAY) in May 2020**, and the testing is scheduled to begin Jan. 10. Committees are looking at adding X-ALD and have discussed MPS 1, but for now aren't pursuing it.



Atlas Faucher, 5, of Oshkosh, with nurse Cassidy Pethke, was born with a rare condition called Pompe disease. Wisconsin doesn't test newborns for Pompe as 27 states do, and Atlas wasn't diagnosed until he was nearly 4 months old, preventing early treatment that could have prevented many disabilities. The state in May 2020 approved screening for Pompe, which is scheduled to begin Jan. 10.

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In a pilot project, Wisconsin **screened babies for Pompe from July 2017 to March 2019**. No cases of the severe infantile form of the disease were found, but **13 babies were identified as having a less-serious, later-onset form**. The state has more than 60,000 births a year.

In June 2016, Genevieve and Austin Faucher of Oshkosh welcomed their first child, Atlas. He seemed fragile and floppy, which doctors attributed to being born five weeks early. When the boy developed a persistent cough, his mother demanded a chest X-ray.

The scan found a significantly enlarged heart. Atlas, by then nearly 4 months old, was taken by helicopter to Children’s Wisconsin hospital near Milwaukee, where six days later tests revealed he had infantile Pompe. He started on an enzyme therapy that can slow the worsening of symptoms, but it doesn’t reverse deterioration that already occurred.



Born with Pompe disease, an inherited disorder that impacts muscles and organs, especially the heart, Atlas Faucher of Oshkosh gets physical therapy, occupational therapy, speech therapy and breathing treatments, which are mostly covered by Medicaid.

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“That’s months of (disease) progression, of damage that he can’t get back,” Genevieve Faucher said.

Atlas, 5, is fed through a tube and uses a wheelchair. He speaks in muffled words that can be hard to understand. Seven nurses rotate caring for him on weekdays, and his working parents handle weeknights and weekends.

Since he is susceptible to complications from infections, Atlas attends kindergarten online. He receives physical therapy, occupational therapy, speech therapy and breathing treatments, which are mostly covered by Medicaid. A fan of the Disney movies “Aladdin” and “Cinderella,” he likes to spin his wheelchair on the family’s hardwood floors to the soundtrack of the 2020 film “Zombies 2.”



Atlas Faucher, 5, of Oshkosh, attends kindergarten online at home, where he has an adaptive bathroom. His Pompe disease creates challenges, but “Atlas is a very happy kiddo and, all things considering, healthy,” said his mother, Genevieve Faucher.

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With a 3-year-old sister and an 18-month-old brother who don’t have Pompe, comparisons are hard to accept. When his brother took his first steps, “Atlas got so mad, he started yelling at him and said, ‘You’re not allowed to walk,’” their mother said.

Atlas' future, and longevity, are "a big old question mark," Faucher said.

If Pompe had been part of newborn screening when Atlas was born, he could have started treatment earlier and almost certainly would have fewer disabilities, she said. For babies born with the condition today, "it could literally save people like Atlas' lives," she said.



Genevieve Faucher's son Atlas, 5, must be fed through a tube and use a wheelchair. He speaks in muffled words that can be hard to understand.

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In Wisconsin, experts appointed by the state health department vote on adding conditions to newborn screening before **consideration by a larger committee** that meets twice a year.

Another panel, the Secretary's Advisory Committee on Newborn Screening, **considers nine criteria**, including how accurate tests are and whether treatment is readily available, before making its recommendation to the

health department. The department's rulemaking process to add a condition can take 30 months, spokesperson Jennifer Miller said.



Physical therapy for Atlas Faucher, 5, of Oshkosh, includes playing a matching game with his nurse Cassidy Pethke. His muscles are weakened from a rare disorder called Pompe disease. "If he doesn't stretch for a couple of days, his muscles get very tight and painful," his mother said.

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Newborn screening is considered mandatory in all states, though in Wisconsin and many states **parents can opt out for religious beliefs or personal convictions.** About 99% of babies in the state are screened, said Dr. Mei Baker, co-director of newborn screening at the Wisconsin State Laboratory of Hygiene.

Positive results typically require follow-up testing to confirm a diagnosis, Baker said. False positives can alarm parents, and treatments need to be effective enough to justify early identification of the disease, she said.

“If you list a condition, you also attach an obligation,” Baker said.



Baker

Krabbe screening

New York was the **first state to start screening for Krabbe, in 2006,** after former Buffalo Bills quarterback Jim Kelly’s son, Hunter, **died from the condition the previous year.** Missouri added Krabbe to newborn screening in 2012, followed by Kentucky and Ohio in 2016, Tennessee and Illinois in 2017, New Jersey in 2019, Indiana in 2020 and Pennsylvania and Georgia this year, according to the nonprofit Hunter’s Hope, started by Kelly.

When Wisconsin **first refused to add Krabbe in 2016,** state experts said there were too many false positives, and stem cell transplants weren’t clearly effective. The federal committee overseeing the RUSP **voted against adding Krabbe seven years earlier for similar reasons.**

committee, which said only a few centers do transplants for babies with Krabbe, notably Duke University in North Carolina.



Kevin and Judy Cushman, of Wisconsin Rapids, lost their son Collin in January 2019 at age 8 to a rare disorder called Krabbe disease. The Cushmans met with state Sen. Patrick Testin, R-Stevens Point, who introduced a bill to require Wisconsin to screen babies for Krabbe. The state has twice rejected such testing. Babies with the condition can receive stem cell transplants. "It gives parents a choice," Kevin Cushman said.

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This February, health department Secretary Karen Timberlake ruled against adding Krabbe, saying follow-up procedures and referral centers need to be identified. In July, Hunter's Hope re-nominated Krabbe at the federal level. The review process could take at least a year.

DHS letter denying newborn screening for Krabbe disease

Dr. Joanne Kurtzberg, who has done 57 stem cell transplants for Krabbe at Duke since 1996, **published a paper on the earliest patients in 2018**. The report said 79% were alive 10 years after the procedures and those transplanted before 30 days moved, spoke and fed better than those transplanted later.



Kurtzberg

hospital.

In an interview, Kurtzberg said about a third of the patients end up needing a wheelchair, another third walk with difficulty and another third are minimally impaired, including a 17-year-old who takes dance classes. “If you met her on the street, you would never know,” she said.

Early this year, Dr. Kristin Page, who trained under Kurtzberg, moved to the Medical College of Wisconsin near Milwaukee. Page has had “preliminary conversations” about starting stem cell transplants for Krabbe, said Evan Solocheck, a spokesperson for the affiliated Children’s Wisconsin

A difficult life

Toys, blankets and pillows, including one stuffed with their son’s hair, are strewn across a hospital bed in Judy and Kevin Cushman’s living room in Wisconsin Rapids.

It has been nearly two years since Collin Cushman died from Krabbe at age 8, but the couple can’t bring themselves to get rid of the bed or his breathing machines stored beneath it. For most of his life, Collin couldn’t walk, talk, see or eat. He lived on the bed.



Judy and Kevin Cushman, of Wisconsin Rapids, store son Collin's breathing machines under the hospital bed he stayed on for much of his eight years living with Krabbe disease. Children with the condition typically die by age 2, doctors say. The Cushmans believe their close attention to Collin's treatments gave him a longer life than most.

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“It was months and months before I would spend much time in this room,” Kevin Cushman said, his voice quivering.

Judy studied a photo of Collin waving as she held him on a slide when he was 9 months old. “This was one of his last waves,” she said.

Soon after, Collin became irritable, had trouble sitting up and clenched his fists. Four months of testing led to the diagnosis of Krabbe.

“You go into shock, denial, anger, all those stages of grief,” Kevin said.

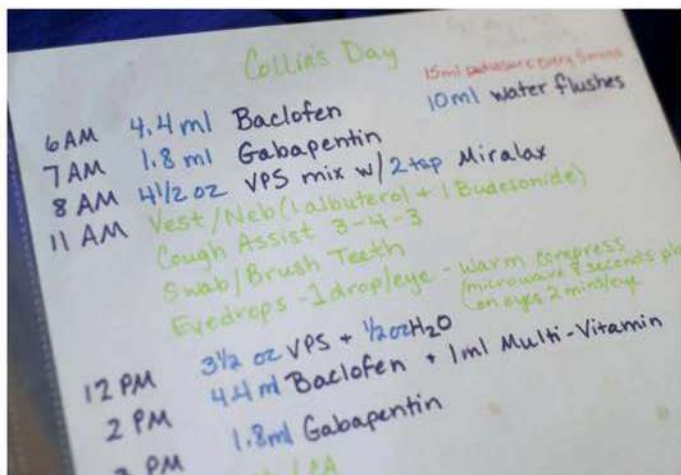


After their son Collin became stiff and irritable at age 9 months, it took four months of testing to diagnose him with Krabbe disease, which took his life at age 8, Judy and Kevin Cushman said. They're trying to get Wisconsin to test babies for the condition as part of newborn screening.

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The couple persisted as parents, including with a healthy daughter born four years after Collin. They traded shifts watching over their son and worked with his phalanx of nurses. All the while, they wondered how life might have been different if Collin had been tested for Krabbe at birth, in time for a transplant.

“To have a child who could have sleepovers, who could go to sleepovers, who could live somewhat of a normal life, would be huge,” Kevin said.



Kevin and Judy Cushman said caring for Collin, their son who died at age 8 from a rare disorder, was around-the-clock. “Our typical day was where we would get three hours of good sleep,” Judy Cushman said.

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deviating from the state’s reliance on scientific committees. The process aims to “be sure that there will be an overall benefit to families and society, and to avoid negative or unintended impacts,” he said.

Living with Krabbe

On a recent afternoon, Jeremy Thoms shifted his body from his wheelchair to an easy chair between his parents’ recliners at their home in Eau Claire. He began to sing, joining in a Josh Groban recording on YouTube.

“You raise me up, so I can stand on mountains,” Jeremy bellowed off-key, while imparting the inspiration of the song. “You raise me up, to walk on stormy seas.”

They approached state Sen. Patrick Testin, R-Stevens Point, who in March introduced the **bill to require Wisconsin to screen for Krabbe**. Testin amended the bill to make the state start Krabbe testing **when it begins screening for Pompe**, which involves a similar test. That way, it would bring little or no additional cost, state officials said.

Dr. Robert Steiner, newborn screening consultant for the state health department, expressed concern about



Randy Thoms helps his son, Jeremy, up a wheelchair ramp to their house in Eau Claire after getting him off the school bus. Jeremy was tested for Krabbe disease shortly after birth, unlike most babies in Wisconsin, because his older brother died from the disease. "He's alive today because he got treated," Randy said.

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Lego sets, with Harry Potter, Batman and dinosaur themes, covered much of the floor. At night, Jeremy lines up the dinosaurs so they stare at his father, a running joke.

Jeremy, 21, is in his sixth year of high school by choice. He is slow to speak but can converse. Why Batman over Superman? "Batman doesn't have super powers," he said. "He's a hero no matter what."

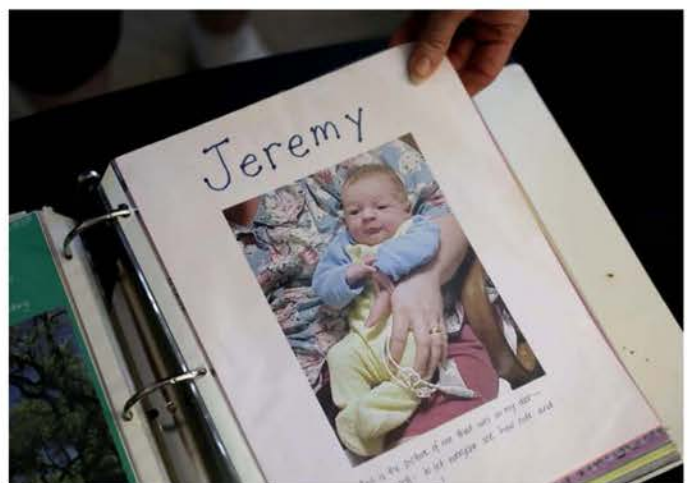


Jeremy Thoms, 21, likes to joke with his parents, Tanys and Randy Thoms, and play-fight with his plastic swords. He was born with Krabbe disease, a rare inherited condition that is typically fatal by age 2 if untreated. Jeremy, who has disabilities, got a stem cell transplant for the condition as a baby.

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Jeremy was diagnosed with Krabbe when he was 8 days old. Kurtzberg gave him a transplant at 28 days. Tanys and Randy Thoms had Jeremy tested because their first son, Alex, died from Krabbe at 13 months. Their middle son, Adam, 33, is not affected.

Jeremy walked on his own until he was 5, when he started using a walker. He has relied on a wheelchair since he was 10. A few years ago, doctors put metal rods in his back to address a related spine condition that can cause fatal breathing problems.



Randy works as Jeremy's caregiver, helping him shower, get on the toilet and change clothes. Tanys works from home as an administrative assistant for Mayo Clinic Health System.

Jeremy Thoms got a stem cell transplant for Krabbe disease from Dr. Joanne Kurtzberg at Duke University in 2000, less than a month after he was born.

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Tanys and Randy Thoms help son Jeremy use a power lift chair to get into their van in October to go out to dinner to celebrate Jeremy's 21st birthday.

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"I wouldn't trade my time with Jeremy for anything," Randy said. "He means so much to us."

Wanting a choice

In Spring Green, the Heckendorfs keep a plastic tote with photo albums and scrapbooks of their son Bryce's 18 months of life. Photos show him lying beside a stuffed toy firefly, propped up on a tractor at the farm where Jenna grew up and strapped into a standing device to help his bones grow.

One shows the book, "Commotion in the Ocean," Bryce's favorite before he became ill.



Jenna and Kyle Heckendorf have nearly a dozen photo albums and scrapbooks showing their son Bryce's life before he died at 18 months old from Krabbe disease. "I put every photo I have of Bryce in an album," Jenna said.

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"We'd get to the shark page, and he'd just light up," said Kyle Heckendorf, who teaches math at River Valley Middle School, where Jenna leads student support sessions in math and reading.

The photos don't show the days when Bryce was in so much pain from his disease that he was inconsolable. "We'd take turns, around the clock, holding him 24 hours a day," Jenna said. "It was horrible."

Like many babies with untreated Krabbe, Bryce lost his sight and hearing. His parents will never know how much of life he absorbed. “To the day he died, we talked to him and read to him,” Jenna said.



Kyle Heckendorf plays with children Carter, 6, and Ava, 2, at their home in Spring Green. Jenna Heckendorf said the family tries to keep the memory of son Bryce, who died in 2014, alive while enjoying their other two kids. “We talk about Bryce a lot,” she said. “You move forward but you don’t move on.”

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The couple have two healthy children, Carter, 6, and Ava, 2. They wish they also still had Bryce, who would be 8 today.

“After witnessing what Bryce went through, we would give anything to have been given a somewhat normal life,” Jenna said. “We didn’t have a choice.”

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https://madison.com/wsj/news/local/health-med-fit/many-wisconsin-hospitals-send-some-newborn-screening-samples-late/article_cb41d9df-c492-58a4-835b-6a169c16a500.html

TOPICAL TOP STORY

Many Wisconsin hospitals send some newborn screening samples late

David Wahlberg | Wisconsin State Journal
Dec 18, 2021



Colton Hidde, 9, left, and brother Christian, 4, right, were born with a rare disease called argininosuccinic aciduria, or ASA. Brother Caleb, 6, middle, doesn't have the condition. Treatment for Colton was delayed after birth because of a late newborn screening test, Karen and Michael Hidde alleged in a lawsuit that was settled. Christian was treated sooner. "Colton will never be verbal, where Christian has the potential to be verbal," Karen Hidde said.

AMBER ARNOLD, STATE JOURNAL
David Wahlberg | Wisconsin State Journal

Colton Hidde can put his pants on, but he hasn't figured out shirts. When eating, he uses an adaptive high chair. The 9-year-old speaks only a few words, including "ish" for "fish."

A delayed newborn screening test in 2012 left the boy brain-damaged before doctors could diagnose and treat him for **argininosuccinic aciduria, or ASA**, a rare genetic disorder in which ammonia builds up in the body, according to a lawsuit filed by his parents.

STATE OF WISCONSIN CIRCUIT COURT COUNTY OF OUTAGAMIE
BRANCH 7

**COLTON HIDDE, a minor, by his
Guardian Ad Litem, TARA R. DEVINE,
KAREN HIDDE, Individually, and
MICHAEL HIDDE, Individually,**

Plaintiffs,

**UNITED HEALTHCARE SERVICES, INC.,
a Foreign Corporation,
WISCONSIN DEPARTMENT OF HEALTH SERVICES,
a Governmental entity,
NETWORK HEALTH INC., a Wisconsin Corporation, and
BLUE CROSS BLUE SHIELD OF WISCONSIN,
a Wisconsin Corporation,**

Involuntary Plaintiffs,

vs.

Case No.: 15-CV-78

**THEDACARE MEDICAL CENTER – NEW LONDON, INC.,
a Wisconsin Corporation, Doing Business as
NEW LONDON FAMILY MEDICAL CENTER,
MMIC INSURANCE, INC.,
MICHAEL L. FETTEROLF, M.D., and
WISCONSIN INJURED PATIENTS AND FAMILIES
COMPENSATION FUND, a Statutory Corporation,**

Case Code: 30104
Medical Malpractice

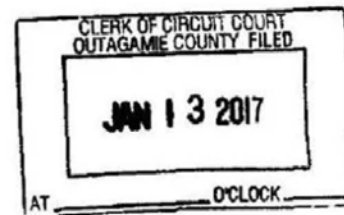
Defendants.

PLAINTIFFS' AMENDED COMPLAINT AT LAW

COUNT ONE

(COLTON HIDDE v. ThedaCare, MMIC, and WI Injured Patients and Families Fund)
(Medical Negligence)

NOW COME the Plaintiffs, COLTON HIDDE, a minor, by his parents and Next Friends, KAREN and MICHAEL HIDDE, KAREN HIDDE, Individually, and MICHAEL HIDDE, Individually, by and through their attorneys, SALVI, SCHOSTOK & PRITCHARD, P.C., and



Colton's younger brother, 4-year-old Christian, was also born with ASA but treated right away. He is also developmentally delayed but uses a fork and spoon, unlike Colton, and already speaks many more words.

"There's no way of knowing what Colton would be like if (the newborn screening test) would have been on time, but there's no doubting he wouldn't have been as sick as he was because he would have had normal treatment from the beginning," said his mother, Karen Hidde, of New London.



Colton Hidde, 9, who has a rare genetic disorder that causes ammonia to build up in the body, speaks only a few partial words. He is learning to use a device to communicate.

AMBER ARNOLD, STATE JOURNAL

Hospitals are supposed to **collect blood samples from babies 24 hours to 48 hours after birth** and send them to the Wisconsin State Laboratory of Hygiene in Madison **within 24 hours after collection**, according to the lab. But some samples don't make it to the lab on time, which can prevent early treatment that could save or dramatically improve newborns' lives.

"Recently we've noticed a trend towards submitters collecting and submitting samples outside of this recommended time window," said an Oct. 15 letter to providers from the lab and the state Department of Health Services. "As a reminder, delayed submissions can have negative consequences."

The lab, allowing for some flexibility, **reports how many samples are received within three days of collection**. In April to June, Mayo Clinic Health System-Northland in Barron, SSM Health St. Agnes Hospital in Fond du Lac, Ascension

Columbia St. Mary's Hospital Ozaukee in Mequon, Richland Hospital in Richland Center and Tomah Health sent more than 5% of samples more than three days late, according to the lab. Many other hospitals in the state sent some specimens late.



Michael Hidde said liver transplants have reduced ammonia levels from a rare disease in his sons Christian, 4, and Colton, 9, but led to other challenges such as the need to take anti-rejection drugs for life. "The liver transplant took them from being a metabolic child to being a transplant child," he said. "It was like a double-edged sword."

AMBER ARNOLD, STATE JOURNAL

The Mequon and Richland Center hospitals continued to send more than 5% of samples more than three days late in July to September, when Westfields Hospital in New Richmond joined the list. At Aspirus Medford Hospital, more than 5% of samples were more than three days late the first and third quarters of the year.

The Mequon hospital, part of Ascension Wisconsin, sent 26 samples, or more than 12%, more than three days late during the second and third quarters, the worst level at any hospital in the state this year.

When courier and related problems are included, the vast majority of Wisconsin hospitals had some delayed samples in January to September. Ascension Columbia St. Mary's Women's Medical Center in Milwaukee topped the list with 119 delayed samples, including 17 the lab considered to be the hospital's fault. Ascension St. Elizabeth Hospital in Appleton had 110, including 22 that were its fault, and Froedtert Menomonee Falls Hospital had 88, including nine that were its fault.

In Madison, SSM Health St. Mary's Hospital had one delayed sample and UnityPoint Health-Meriter had six, none considered their fault.

The state lab and the **Wisconsin Hospital Association** started making the data **publicly available** in 2014 after a **Milwaukee Journal Sentinel** investigation, triggered by Colton Hidde's story, uncovered flaws in the nation's newborn screening practices. Colton's sample, taken at what is now ThedaCare Medical Center of New London, didn't arrive at the state lab until five days after it was collected, the Journal Sentinel reported. By then, he had already been taken by helicopter to Children's Wisconsin hospital near Milwaukee, arriving in a coma.



A delay in Colton Hidde's newborn screening test cost crucial days in starting treatment for a rare genetic disorder in which ammonia builds up in the body.

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Lawsuit settled

Karen and Michael Hidde in 2018 settled their lawsuit against the New London hospital, which was filed three years earlier. They have a third son, Caleb, 6, who doesn't have ASA.

Colton has a feeding tube, through which he gets medications, but he mostly eats by mouth unless he's ill. He is learning to use a communication device to say his name and some words. He is not expected to become verbal, his mother said.

Colton and Christian have received liver transplants, which helps prevent ammonia buildup but requires them to take anti-rejection drugs that weaken their immune systems.

Karen Hidde said she wishes Colton's screening had been processed more quickly so he could have been treated right away, but she is glad the family's ordeal helped spur public reporting of test transfer times.

"Now that they have to post it and let people know, I think it holds hospitals more accountable," she said.

Wisconsin State Laboratory of Hygiene newborn screening specimen collection
Jan-March 2021 [\(full tables online\)](#)

Submitter Number	Submitter Name	Submitter City	Total Specimens	Specimens Received <=3d	% Specimens Received <=3 d	Specimen Delayed
800106	MINISTRY DOOR CO MEDICAL CENTER	STURGEON BAY	32	32	100	
800107	HOWARD YOUNG MEDICAL CENTER	WOODRUFF	0	0		
800110	MEMORIAL HOSPITAL OF LAFAYETTE COUNTY	DARLINGTON	0	0		
800113	SAUK PRAIRIE HOSPITAL	PRAIRIE DU SAC	77	77	100.00	3
800115	ASCENSION CSM HOSPITAL OZAUKEE	MEQUON	80	80	100.00	7
800117	THEDACARE MEDICAL CENTER-WAUPACA	WAUPACA	25	25	100.00	3

From December 19th through December 26th we will be granting free access as a gift to our readers presented by SSM Health

https://madison.com/wsj/news/local/health-med-fit/wisconsin-wants-to-hold-onto-newborn-screening-specimens-longer/article_d7974692-1b13-5c73-b7fd-06d8f9725f51.html

TOPICAL

WISCONSIN | BABIES' BLOODSPOTS

Wisconsin wants to hold onto newborn screening specimens longer

David Wahlberg | Wisconsin State Journal
Dec 18, 2021



Drops of blood from a baby are put on a filter paper card at UnityPoint Health-Meriter in Madison as part of routine newborn screening in this photo from 2013.

AMBER ARNOLD, STATE JOURNAL ARCHIVES

Minnesota had to destroy more than 1 million newborn screening blood samples after families sued, saying the state was keeping the dried bloodspots for research without their permission.

After a similar lawsuit in Texas, that state got rid of more than 5 million bloodspot cards. In Michigan, a judge ruled in July that the state's retention of specimens before 2010, when **the state started seeking consent** for storing them, **violated the constitutional rights of parents of two children born in 2008.**

Wisconsin has long **stored babies' blood samples for a year before destroying them**, while keeping samples with positive results indefinitely. Now, scientists who lead the state's newborn screening program are looking at holding onto samples for 10 years. That could improve infant testing, enable biomedical research and assist doctors and families in identifying causes of diseases children can develop later in life, they say.

“Retaining for a period of time is really justifiable,” said Dr. Mei Baker, co-director of newborn screening at the Wisconsin State Laboratory of Hygiene.



A few drops of blood are taken from the heel of Greyson Banes for newborn screening at UnityPoint Health-Meriter as his mother, Michol, looks on, in this photo from 2013.

AMBER ARNOLD, STATE JOURNAL ARCHIVES

Baker and others who sit on the state’s umbrella committee for newborn screening **voted Dec. 3 to keep samples for 10 years**, a move that still needs to be approved by the state Department of Health Services. The committee also voted to develop a policy on how to handle research proposals and inform the public, including about how people could opt out of storing samples for research.

Newborn screening is mandatory in all states, though in Wisconsin and many states parents can opt out for religious reasons or personal convictions. Five or six drops of blood are collected from the pricked heels of babies and placed on filter paper cards. After screening tests are completed, some states store residual samples for a few months, others keep them for decades and several store them indefinitely, typically without explicit parental permission, according to a University of Utah **study published in 2019 in the journal Genetics in Medicine**.

Programs **use some of the samples to improve infant testing**, but scientists also use de-identified specimens to study genetic disorders and other childhood diseases, the Utah researchers found. Some bloodspot cards are used for forensic purposes, such as to identify human remains.

A push to make states obtain consent specifically for retaining samples has been met with concerns that too many parents will say no, making the remaining bloodspots less representative of overall populations. However, consent “may improve trust in newborn screening programs,” the Utah researchers said.

Destroying samples

Minnesota in 2014 destroyed 1.1 million bloodspot cards after **settling a lawsuit filed in 2009 by 21 families**. Under updated laws, samples since August 2014 have been **stored indefinitely unless parents opt out**.



Atlas Faucher, 5, who has significant muscle weakness from a rare disorder called Pompe disease, gets physical therapy at his home in Oshkosh, where he attends kindergarten online.

AMBER ARNOLD, STATE JOURNAL

Texas in 2009 **agreed to destroy 5.3 million bloodspot cards** stored since 2002 following a lawsuit by the Texas Civil Rights Project. The state had given 800 de-identified samples to a U.S. Armed Forces lab to help build a DNA database to identify missing people and solve cold cases, **the Texas Tribune reported in 2010.**

Today, Texas **keeps bloodspot cards for two years** for use by the state health department **unless parents agree to store them for up to 25 years**, which can include research outside of the department.

In Michigan, **bloodspot cards since 2010 are stored for 100 years**, but aren't used for research unless parents consent, according to the state. A trial is scheduled for April on how widely samples should be stored and used for research, and whether proper consent is being obtained.

“Do they really need to keep everybody’s sample to do that?” asked Philip Ellison, an attorney from Hemlock, Michigan, who **sued in 2018 on behalf of four parents of nine children.**

What’s allowable

Wisconsin doesn't seek consent for keeping bloodspot cards, but parents can ask the state lab not to retain them, Baker said. Positive samples are kept indefinitely or until they're used up, and de-identified for use by the lab to improve testing, she said.



The front closet of Genevieve and Austin Faucher's house in Oshkosh is filled with medical supplies for their son Atlas, 5, who has a rare disorder called Pompe disease.

AMBER ARNOLD, STATE JOURNAL

Baker said that if bloodspot cards were kept longer, they could help families of children who develop symptoms of certain diseases long after birth. Hearing loss can stem from cytomegalovirus, or CMV, which can be acquired in the womb or after birth; a stored sample can help discern the origin.

A small minority of children with cystic fibrosis aren't caught by newborn screening, Baker said. If children later develop symptoms of the lung and digestive disorder, a stored sample could determine if their test was a known type of false negative or an error.

From December 19th through December 26th we will be granting free access as a gift to our readers presented by SSM Health

https://madison.com/wsj/news/local/health-med-fit/wisconsin-newborn-screening-list-shorter-than-in-many-states/article_3d1788d8-3d75-5a5d-954f-1dda11dc5d93.html

TOPICAL

Wisconsin's newborn screening list shorter than in many states

David Wahlberg | Wisconsin State Journal
Dec 18, 2021



Chemists Mary Carlstedt, Michael Cogley and Marcy Rowe open packages containing newborn screening blood sample cards for testing at UW-Madison's Wisconsin State Laboratory of Hygiene in this photo from 2013.

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David Wahlberg | Wisconsin State Journal

Connecticut tests babies for 73 disorders shortly after birth, the most of any state, while Wisconsin screens for 47, fewer than surrounding states, **according to the federal Health Resources and Services Administration**, or HRSA.

But the numbers can be deceiving, said Dr. Mei Baker, co-director of newborn screening at the Wisconsin State Laboratory of Hygiene. She is also a member of HRSA's Advisory Committee on Heritable Disorders in Newborns and Children.

"We never get into a counting game," Baker said. "You really cannot judge a program by how many conditions they list."

Connecticut says it screens for five hemoglobin traits that make people carriers for sickle cell disease from birth, though carriers don't have the blood disease. Wisconsin also detects and reports the carriers but doesn't list them on its newborn screening panel, Baker said.

Connecticut also lists two conditions related to phenylketonuria, or PKU, a protein disorder that can cause intellectual disability. Wisconsin, which lists PKU, can also pick up the related conditions but doesn't list them, Baker said.

Similarly, Connecticut, Michigan and Minnesota list T cell conditions related to severe combined immune deficiency, or SCID. Wisconsin lists SCID and also detects the T cell conditions but doesn't list them.

Additional examples involve disorders Wisconsin picks up incidentally when screening for targeted conditions, Baker said. While some states list those, Wisconsin doesn't unless they **meet the state's newborn screening criteria**, she said.

In 2011, Wisconsin removed three enzyme conditions from its panel because they were found to be benign. One **had been found primarily in the state's Hmong community**. Connecticut, Michigan, Minnesota and Illinois list some or all of them. Wisconsin screens for two hemoglobin disorders not listed by many states: beta thalassemia major and hemoglobin e-beta thalassemia. They are more prominent among southeast Asians, Baker said.

Mateo Medina has never walked or talked. The 10-year-old breathes and feeds through tubes.

His brother, Javier, 5, marches around their house in Fond du Lac and talks up a storm, but he can't run or jump and his speech is garbled. He can eat soft foods.

Their sister, Amelia, 3, sprints, chats and eats like most kids her age. An occasional morning tremor is the only sign that she, too, was born with the most serious form of **spinal muscular atrophy, or SMA**. The genetic disorder, which progressively weakens muscles, typically has been fatal by age 2.

New treatments are changing the face of SMA, and the Medinas are Exhibit A. Mateo was born five years before approval of the first treatment, which Javier received in a clinical trial. Amelia got a newer gene therapy — considered most effective if given within a few weeks after birth, before symptoms appear — when she was 11 days old. For families without a history of the disease, the main way to catch it early is newborn screening. While Wisconsin and most states have added SMA to their infant testing programs, **12 states haven't**, including Alabama, after **a federal committee recommended it in 2018**.

“A large part of me feels like the state of Alabama failed my son,” said Lauren Hendrix, of Troy, Alabama, whose son Graham died in January. He was diagnosed with SMA nearly six weeks after birth after developing symptoms and received the gene therapy, called Zolgensma, two weeks later.



“Had he gotten Zolgensma a month sooner, because (SMA) had been tested for in the newborn screening, I fully believe my son would still be alive today,” Hendrix said.

Dr. Mary Schroth, chief medical officer of Cure SMA, based in Elk Grove Village, Illinois, has been **urging all states to add SMA to newborn screening**. Other states that haven't are Alaska, Arizona, Hawaii, Idaho,

Louisiana, Nevada, New Jersey, New Mexico, Oregon, South Carolina and Virginia. Some plan to start next year.



Amy Medina, with children Mateo, 10, foreground, Amelia, 3, and Javier, 5, said she and husband Adan worked full-time jobs, with opposite shifts, until their daughter was born. “We didn’t see much of each other,” she said. Adan started staying home with the kids until recently, when he went back to work and she started staying home.

AMBER ARNOLD, STATE JOURNAL

“An infant being diagnosed with SMA used to be essentially a death sentence, and it’s not anymore,” said Schroth, who previously was a pediatric lung specialist at UW Health. But, “the sooner a patient with SMA is identified and treated, the better the outcomes.”

New treatments

Like most disorders detected in newborn screening through a few drops of blood collected from a baby’s heel a day or two after birth, SMA is rare, occurring in about 1 in 11,000 births. If both parents are carriers, each of their children has a 25% chance of having SMA.

The condition is caused by a mutation in a gene that normally makes a protein needed by nerve cells that control muscles. Without the protein, the nerve cells die and people lose the ability to move.

A drug called Spinraza, approved in 2016 for SMA patients of all ages, helps a related gene make more of the protein. Given by spinal injection every four months for life, the drug costs \$750,000 the first year and \$375,000 each subsequent year.



Javier Medina gets breathing treatments to remove secretions from his airway and mucus from his lungs twice a day. His brother, Mateo, gets the treatments three times a day.

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Zolgensma, approved in 2019 for SMA patients younger than 2 years, is a one-time infusion. It uses a harmless virus to replace the mutated gene with a normal one, restoring regular protein production. The gene therapy costs \$2.1 million, believed to be the most expensive one-time treatment on the market.

A third treatment, Evrysdi, was approved last year for SMA patients 2 months and older. It's a daily oral liquid that helps make and

maintain more of the protein, costing up to \$340,000 a year.

Spinraza has helped most patients treated early maintain the ability to swallow and eat by mouth, **its maker, Biogen, reported in June.** With Zolgensma, most children with severe SMA sat, stood and walked alone at age-appropriate times, **according to clinical trial data by Novartis.** Most infants who started Evrysdi after developing symptoms were free of ventilators at their first birthday, **Genentech said in July.**

With its hefty, ongoing price tag, Spinraza “exceeds common thresholds for cost-effectiveness,” **according to the Boston-based Institute for Clinical and Economic Review, or ICER,** which studies the cost and benefits of medical treatments.

Zolgensma’s price is “right at the **upper bounds of what could be considered cost-effective**,” said David Whitrap, a spokesperson for ICER, which hasn’t reviewed Evrysdi.



Javier Medina, left, plays on his tablet as his brother Mateo lies on his bed at their home in Fond du Lac. Sometimes Javier or his sister Amelia will grab Mateo's hand and help him play a game, their father said. “They include him as much as they know how to include them,” Adan Medina said.

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Among about 80 patients with SMA at UW Health, including adults with a later-onset form, 17 take Spinraza, a dozen have received Zolgensma and about 50 are on Evrysdi, said Dr. Jennifer Kwon, a UW pediatric neurologist. Insurance coverage is sometimes a challenge for Spinraza and Evrysdi, but most plans promptly cover Zolgensma, especially for infants, Kwon said.

Since Wisconsin **added SMA to newborn screening in October 2019**, nine babies **have been found to have the condition through the testing**. Eight have received Zolgensma and one got Spinraza.

One Wisconsin baby who had symptoms of SMA before getting Zolgensma shortly after birth has had physical delays, but the others who got the gene therapy “are doing remarkably well,” Kwon said.



Dr. Jennifer Kwon, a pediatric neurologist at UW Health, tests Piper Droessler's strength in this photo from January 2020, as her parents Caiti and Ben Droessler look on.

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What their lives might be like years from now “is a question we can’t answer,” she said. “While it seems like it’s continuing to be effective, we just don’t have longer-term data.”

Results vary by state

Seated in her high chair, Piper Droessler bounced up and down, kicked up her feet and ate strawberry-banana yogurt with a spoon — until she decided to scoop it up with her fist and smear it across her face.



Piper Droessler eats with her family at their home near Platteville a few weeks before turning 2 on Nov. 25. Piper was the first baby to test positive for spinal muscular atrophy through newborn screening in Wisconsin, which started the testing in October 2019. She got a new gene therapy for the condition shortly after birth and shows no signs of SMA.

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A few weeks before her second birthday last month, the toddler chased her older brother and sister around their home near Platteville, occasionally falling on the floor before springing back up and racing again.

Piper was the **first child in Wisconsin to test positive for SMA through newborn screening** and the first identified through screening to get Zolgensma, 23 days after she was born. Today, she shows no signs of the disease.



Caiti Droessler said she's grateful Wisconsin started screening babies for spinal muscular atrophy six weeks before daughter Piper was born. After Piper tested positive, Caiti and husband Ben learned they are carriers, meaning each of their children has a 25% chance of having the disease. Ellianna, 8, and Finley, 5, don't have it, but "if one of them would have had it, it would have been a whole different story," Caiti said.

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"You wouldn't even know," said her father, Ben Droessler.

Caiti Droessler, Piper's mother, interacts online with parents whose children with SMA have died or use feeding tubes. "I don't know their struggle at all," she said.

In Alabama, after Lauren Hendrix's son died, she **started a petition to get SMA added** to newborn screening. The state plans to start screening for SMA by late January, said Arrol Sheehan, spokesperson for the Alabama Department of Public Health.



Graham Hendrix

LAUREN HENDRIX

Graham Hendrix looked normal at birth in October 2020, but a few weeks later he stopped moving his arms and his head, his mother said. After being diagnosed with SMA, he got Zolgensma at Children’s of Alabama hospital in Birmingham. The treatment seemed to help, but he stopped breathing in January and died the next day.

“It blows my mind that we don’t test for (SMA) in our state, but we have a team for it at the children’s hospital,” said Hendrix, who is expecting a daughter in April.



Wisconsin first state to screen newborns for rare immune disorder

Haley Comer of Middleton, Idaho, said she and medics had to resuscitate her son, Ryder, a month after he was born. Tests showed he had SMA, and he got Zolgensma two weeks later.

Today, at 10 months, Ryder is moving his arms and sucking on his fingers again, but he doesn't sit or hold his head like most children his age, his mother said. Idaho plans to start screening for SMA in February, said Greg Stahl, spokesperson for the Idaho Department of Health and Welfare.



Ben Droessler, with daughters Ellianna, 8, and Piper, who turned 2 on Nov. 25, said Piper shows no signs of the spinal muscular atrophy she was born with but treated for shortly after birth.

AMBER ARNOLD, STATE JOURNAL

“It’s very frustrating” Idaho hasn’t started the testing yet, Comer said. “Getting treatment sooner gives them such a better chance.”

Audrey Wiley's daughter Adelynn was born in Storm Lake, Iowa, on June 25, 2020. Six days later, **Iowa started screening newborns for SMA.**

Adelynn, diagnosed with the disease at 3½ months, got Zolgensma a month later. She started moving more but stopped breathing in January and died.

“It’s as bad of luck as it gets,” Wiley said. “Obviously, she was not treated soon enough.”

Two sons with SMA

Like most parents affected by SMA, Amy and Adan Medina had never heard of the disease before their son Mateo was diagnosed with it a month after he was born in 2011.

He has used a feeding tube since he was 3 months old and a breathing tube, attached through a hole in the front of his neck, since 7 months. He wears diapers. He can’t move, except for his eyes, and, very subtly, his feet and left index finger and thumb. “You have to really be watching to see it,” Amy Medina said.



Adan Medina cleans around his 10-year-old son Mateo's breathing tube before getting him dressed. Nurses provide some care for Mateo, but Adan and his wife Amy also spend a lot of time caring for him.

AMBER ARNOLD, STATE JOURNAL

Three times a day, his parents or a nurse use machines to clear secretions from his airway and mucus from his lungs.

Mateo spends his days in bed or on a wheelchair-stroller, extended upright to support his spine and prevent the buildup of secretions. He goes to public school, where he has been trying to use a communication device activated by eye movement to converse with fellow fourth-graders.

When the Medinas got pregnant again, they knew they had a 25% chance of having another child with SMA. But, “one in four is also a 75% chance you’re not going to have an affected baby,” Amy said.

More than five months into the pregnancy, amniocentesis confirmed the baby would have SMA. The news was no easier to accept the second time. “It was still heartbreaking,” Adan said.

After Javier was born, the family drove 14 hours to Johns Hopkins University in Baltimore, where he got his first dose of Spinraza in a clinical trial at 12 days. They made 18 trips to Baltimore, most by airplane, before Javier started getting the injections last year in Chicago.



Photos in the hallway of Amy and Adan Medina's home in Fond du Lac show them with their sons, Mateo and Javier, who were born with spinal muscular atrophy. Photos of Javier, right, show him grinning before age 3, when his facial muscles weakened so much he became unable to smile.

AMBER ARNOLD, STATE JOURNAL

Mateo also got Spinraza, starting at age 6, before switching last year to Evrysdi.

Javier eats — mostly oatmeal, mashed potatoes, cookies and Cheetos — but gets much of his nourishment through a feeding tube at night. He also receives supplemental oxygen while sleeping, especially when he’s sick.



Javier Medina, 5, and his 3-year-old sister, Amelia, get going soon after waking up on a recent morning at their home in Fond du Lac.

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In addition to getting treatments to remove secretions and mucus twice a day, the kindergartner receives physical therapy, occupational therapy and speech therapy.

Family photos show him grinning until he was 3, when his facial muscles weakened. “He lost his smile,” his mother said. “He says it’s broken.”

A third child

When the Medinas got pregnant a third time, they figured the odds must be in their favor. But they learned, again through amniocentesis, that their third child would also have SMA.

“I cried, loud, uncontrollably and a lot for a period of time,” **Amy wrote on the family’s CaringBridge page.**

Amelia was born in March 2018, when another clinical trial was available, for Zolgensma. After getting the gene therapy at UW’s American Family Children’s Hospital, she is doing well today.

Outside the family’s house, a big red van sits in the driveway, equipped to carry the five Medinas and all of their gear. On a recent morning, Amelia ran along the sidewalk. Javier swung his arms as if he was running, but his feet stayed on the ground. Mateo watched them from his horizontal perch.



Amy Medina, shown with 3-year-old daughter, Amelia, said the only sign Amelia shows of having a spinal muscular atrophy is an occasional tremor in the morning.

AMBER ARNOLD, STATE JOURNAL

Amelia pushed her scooter, adorned with a unicorn on the front. Javier rode his three-wheel, lime green adaptive bicycle, sporting a hotdog costume he likes to wear around the house.

“They’re two peas in a pod, but they fight like crazy too,” Amy said.

With three children who represent the spectrum of SMA today, the Medinas see the importance of newborn screening.

“Catching it early makes a world of difference,” Amy said. “You shouldn’t have to wait until your child is showing symptoms to figure out what’s happening.”



Javier Medina, 5, wakes up at his home in Fond du Lac. He started treatment for spinal muscular atrophy less than two weeks after he was born, but he still has some symptoms of the muscle weakening disease. At night, he receives supplemental oxygen and nourishment through a feeding tube.

AMBER ARNOLD, STATE JOURNAL

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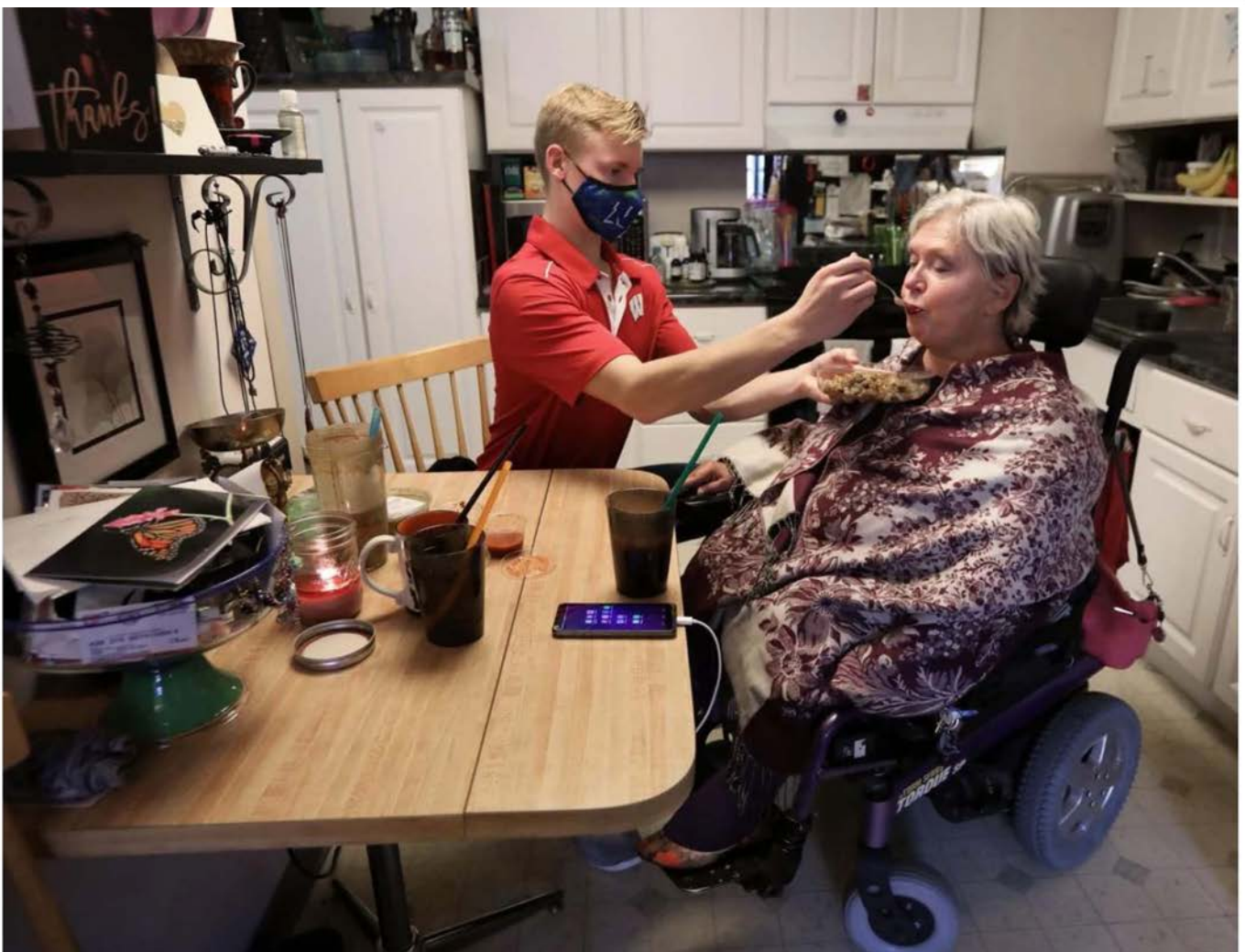
https://madison.com/wsj/news/local/health-med-fit/in-adults-new-treatment-for-muscle-disease-can-have-mixed-results/article_c3f59118-d438-5ca2-b208-523b9f790d43.html

TOPICAL

ADULTS WITH SPINAL MUSCULAR ATROPHY

In adults, new treatment for muscle disease can have mixed results

David Wahlberg | Wisconsin State Journal
Dec 20, 2021



Personal care worker Andrew Wolfram helps Karen Foxgrover eat lunch at her Downtown Madison apartment. Foxgrover, who has a later-onset form of spinal muscular atrophy, has been taking a new drug for the condition for about five months.

AMBER ARNOLD, STATE JOURNAL

David Wahlberg | Wisconsin State Journal

Karen Foxgrover relies on personal care workers to help her eat, go to the bathroom and get dressed. She hoped a new medication for spinal muscular atrophy would give her enough strength to live more independently.

But after swallowing the fruity liquid, called Evrysdi, daily for five months, she's not sure if it has helped with the muscle-weakening condition. She struggles to find enough workers to help her get by day to day.

"I don't feel like I'm getting any weaker," the 64-year-old from Madison said. "But I don't feel like I'm getting significantly stronger. ... I don't think I've noticed any difference."

Evrysdi, approved last year for SMA patients 2 months and older, is one of two treatments available for adults with SMA. The other, Spinraza, a spinal injection given three times a year, was approved five years ago.



Karen Foxgrover, who has spinal muscular atrophy, receives her daily dose of the oral liquid medication Evrysdi, which she said tastes like fruit punch. She hoped the treatment would give her more muscle strength, but it's hard to tell if it has helped or not, she said.

AMBER ARNOLD, STATE JOURNAL

There are **four main types of SMA**. Type 1, the most common and most severe, is usually diagnosed within the first few months of life and is typically fatal by age 2 if not treated. People with type 2, diagnosed between 6 months and 2 years, and type 3, diagnosed as late as the teen years, generally can move or walk in childhood but eventually need wheelchairs. Type 4, or adult-onset SMA, is very rare.

Tyler Engel, 32, a Madison man who has type 2 SMA, has been taking Evrysdi for a year. His strength has remained about the same, but he doesn't get tired as quickly as he previously did doing activities like wheelchair soccer, he said.

He hopes the drug will allow him to maintain the strength he still has. "It's life-changing knowing this is the worst-case scenario that I think I'm going to be in," Engel said.

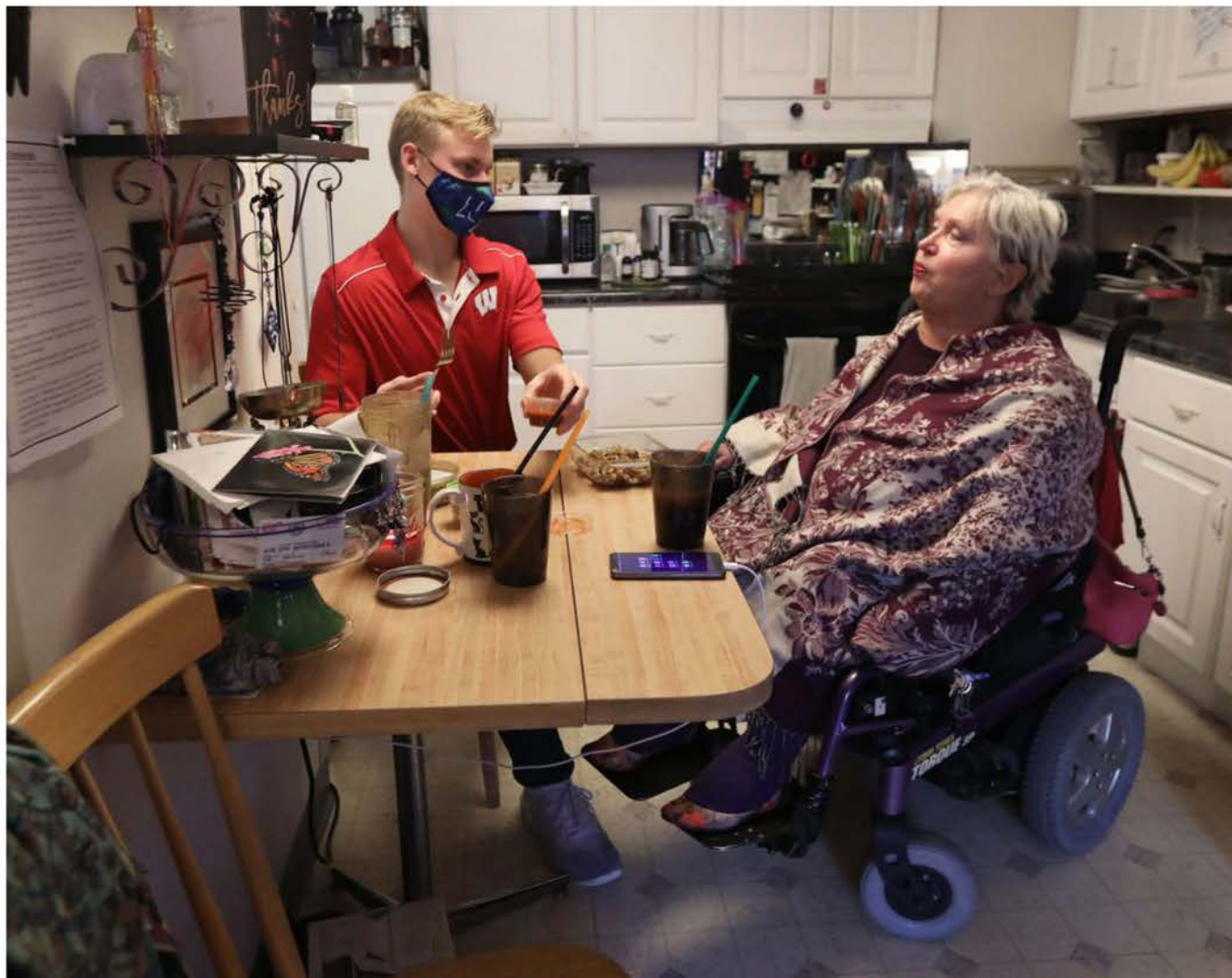
Foxgrover, who has type 3 SMA, moved to Madison in 1982 after graduating from UW-Whitewater with a business degree. She was born and raised in Appleton.

She walked with difficulty as a young child, on the sides of her feet, quickly growing tired from the exertion, she said. By fifth grade, she started using a wheelchair and attending a school for children with disabilities.

Her oldest brother had what was likely SMA and died in his teens. Their other six siblings don't have it, in line with the 25% chance that parents who are carriers will pass it on to each of their children.



Engel



Andrew Wolfram, a pre-medicine student at UW-Madison, is one of several personal care workers who help Karen Foxgrover eat, go to the bathroom and get dressed. Foxgrover said it's a never-ending challenge to find enough workers to assist her.

AMBER ARNOLD, STATE JOURNAL

When Foxgrover moved into her Capitol Centre apartment in Downtown Madison nearly 40 years ago, she was still strong enough to feed herself and transfer herself from her wheelchair to the toilet and the shower.

The disease eventually left her unable to use her arms and reliant on regular help.

“It’s just so obvious how much I’ve lost,” she said. With Evrysdi, “I was hoping I could get some muscles back ... but I think I’m a little too old for it.”

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https://madison.com/wsj/news/local/health-med-fit/wisconsin-first-state-to-screen-newborns-for-rare-immune-disorder/article_70ec1602-575c-56a4-a73d-97560b66f54e.html

TOPICAL

Wisconsin first state to screen newborns for rare immune disorder

David Wahlberg | Wisconsin State Journal
Dec 20, 2021



Claire Hood, 8, reading in a swing in her backyard in West Bend. Born with a severe immunodeficiency, she received a stem cell transplant at 4 months old.

AMBER ARNOLD, STATE JOURNAL

David Wahlberg | Wisconsin State Journal

Less than a week after Amanda and Brandon Hood’s daughter Claire was born, they got a call from their doctor. Claire, who seemed healthy, had **severe combined immunodeficiency disorder, or SCID**, sometimes called “bubble boy disease.”

Amanda Hood, a nurse, was familiar with immune deficiencies but not SCID, which affects **about 1 in 45,000 babies**. “We were very surprised,” she said. “I thought maybe it was a lab error.”

Further tests confirmed SCID, marked by having few or no T cells to fight infections. Without treatment, the condition is usually fatal by age 2.

Doctors at UW Health’s American Family Children’s Hospital introduced T cells in Claire’s body by giving her a stem cell transplant when she was 4 months old, before showing any symptoms. Now 8 and in third grade, she is doing well, playing with her 3-year-old brother on their backyard swing set in West Bend and writing a short story tentatively titled “Fire Mountain.”

“What would have happened if we didn’t have newborn screening then?” Amanda asked.

Most states didn’t test babies for SCID in 2013, when Claire was born. Only 16 did, including Wisconsin, which **five years earlier became the first state to add the condition to newborn screening**. SCID was the **first disorder included in the federal Recommended Uniform Screening Panel in 2010**. By 2018, all states were testing infants for it.

Many Americans are familiar with SCID from the 1976 film, “**The Boy in the Plastic Bubble**,” starring John Travolta, who played a child with the disease based on two real patients.



Brandon Hood, a sergeant with the Washington County Sheriff's Department, plays with children Claire, 8, and Evan, 3, in their backyard in West Bend.

AMBER ARNOLD, STATE JOURNAL

word 'cure,'" Baker said.

On Jan. 1, 2008, Wisconsin started a pilot project to test babies for the disorder, using funding from Children's Wisconsin hospital and the New York-based Jeffrey Modell Foundation, named after a boy who died from a related condition at 15.

New molecular technology presented an opportunity for testing, and stem cell transplants were shown to be beneficial, said Dr. Mei Baker, co-director of newborn screening at the Wisconsin State Laboratory of Hygiene.

"With the ... transplant, it's the first disorder where we ever can use the



Amanda Hood and daughter Claire, 8, look at photos of Claire's early life. Born with severe combined immunodeficiency, or SCID, Claire received a stem cell transplant at 4 months old to give her a new immune system. It took a while for the transplant to take hold, so she had to live in isolation until she was nearly 3.

AMBER ARNOLD, STATE JOURNAL

Once she was able to socialize, Claire was shy during play dates because she was not used to interacting, her mother said. But that soon changed.

In 4K, Claire was exposed to chicken pox, for which she has not been vaccinated because the live vaccine can still be dangerous for her. She received preventive antibodies and didn't get sick.

In school today, she wears a face mask to help prevent COVID-19, as do a few classmates even though West Bend schools don't require them. Amanda stopped working as a nurse because she didn't want to bring infections home. Brandon is a sergeant with the Washington County Sheriff's Department.

Other than washing their hands frequently and trying to avoid sick people, the family lives like any other.



A photo shows Claire Hood on her "4th Life Day," the anniversary of when she received a stem cell transplant for severe combined immunodeficiency, or SCID, a disorder sometimes called "bubble boy disease."

AMBER ARNOLD, STATE JOURNAL

"She's like a normal kid, where she gets sick and she gets better," Amanda said.



Regents boost salary ranges

Move aims to give UW System more negotiating power as high-profile searches for new leaders get underway

LOCAL&STATE. A3

EYING PRIZE

Packers appear to have easiest schedule among NFC bye contenders

SPORTS. B1



Hostages managed to flee

Missionaries held in Haiti found freedom last week by making a daring overnight escape

NATION&WORLD. A10

Biggest stories of 2021

This full-page photo gallery takes a look at some of the top headlines from a busy year

NATION. A11

PARTLY SUNNY 34 • 14 FORECAST, A14 | TUESDAY, DECEMBER 21, 2021 | MADISON.COM

COVID-19 | THE HEALTH CARE SYSTEM

Close to breaking point

Officials urge small holiday gatherings as surge continues

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As a COVID-19 surge leads some Wisconsin hospitals to cancel procedures and turn away pa-

tients, health officials on Monday urged residents to get vaccinated, wear masks indoors and keep holiday gatherings small to prevent cases of the new omicron variant from overwhelming already strained facilities.

Dane County extended its mask mandate until February, with the county's weekly COVID-19 case rate nearly three times that from

early November and coronavirus-related hospitalizations at the highest level this year.

"Our health care organizations have been pushed to the breaking point, and it is quite possible that omicron will push us beyond the breaking point," said Dr. William Melms, chief medical officer of the Marshfield Clinic Health System, which has discontinued elective

surgeries requiring inpatient beds and is turning away 100 referrals a week. "Something's got to give."

With 96% of intensive care beds and 98% of intermediate care beds already in use, and hospitals and nursing homes struggling to

find nurses and other staff to care for patients, state Department of Health Services Secretary Karen Timberlake pleaded for unvaccinated people to get shots and for

Please see SURGE, Page A5

■ **Inside:** Justices to rule on challenge to Dane County's mask mandate. A3

■ **Nation:** Omicron now the dominant strain across the United States. A9

NO TIME TO LOSE | FINDING RARE DISEASES IN INFANTS



AMBER ARNOLD PHOTOS, STATE JOURNAL

Theo Gutzdorf, with parents Rikki and Josh Gutzdorf at their home in Watertown, was born with numerous medical problems for which routine tests couldn't identify a cause. Through genome sequencing, he was diagnosed with the rare disorder Stuve-Wiedemann syndrome, which helped doctors know how to treat him. Josh custom-made Theo a walker before the 3-year-old started walking on his own.

Next step, sequencing

Mapping genome can help when screening doesn't have answer

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Rikki and Josh Gutzdorf knew from prenatal testing that their child would have abnormally shaped bones.

After son Theo was born, other problems emerged: He could barely breathe and had a dangerously high fever. Tests, including a spinal tap, couldn't identify a cause.

Doctors at Children's Wisconsin hospital near Milwaukee turned to genome sequencing, mapping the protein-making parts of the baby's DNA. The analysis showed Theo had Stuve-Wiedemann syndrome, a rare genetic disorder marked by curved leg bones, respiratory distress, poor regulation of body temperature, swallowing problems and, often, death within a year.

The finding helped doctors decide to adjust the boy's oxygen level, start him on a



Rikki Gutzdorf gives Theo, 3, medication through his feeding tube. "He's a little person with a lot of orthopedic structure issues," Rikki said.

Please see DNA, Page A6

FAR EAST SIDE

Jail calls support homicide charge

Decomposed body found in September

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A Madison man has been charged with first-degree reckless homicide after police in September found the decomposed body of his former girlfriend at the Far East Side home where they had lived, then listened to jail calls between him and his current girlfriend as the two settled on a story to tell police, according to a criminal complaint.

Gregg G. Raether, 56, told police he had nothing to do with the death of a 55-year-old woman identified in the complaint only by her initials but by the Dane County Medical Examiner's office on Monday as Patricia A. McCollough, of Madison.

The complaint, filed late Friday, says police believe McCollough died on or about July 22. Police went to her home on Sept. 16 after a caller who identified herself as McCollough's daughter asked police to check on her mother's welfare since she had been unable to reach her by phone.

After looking through windows and seeing that the house was in disarray and getting no answer at the door, police entered the home and found McCollough's body in a bedroom, buried under a pile of dresser drawers, shelving, televisions and bedding material.

Please see HOMICIDE, Page A5



Raether



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COMICS	B3	OPINION	A11	STOCKS	A9
METRO/REGIONAL	A10	SCOREBOARD	B5	TABLETS	B10
OPINION/ADFS	A4	SPORTS	B1	TELEVISION	B12





AMBER ARNOLD PHOTOS, STATE JOURNAL

Rikki Gutzdorf regularly takes Theo, who has Stuve-Wiedemann syndrome, to therapy appointments and does stretches at home. The rare disorder can be fatal by age 1, but with an early diagnosis and regular care, Rikki said her son can thrive. "We want to make sure we're giving Theo the best possible life," she said.

DNA

From A1

medication to lower his temperature and give him a feeding tube. Since the disorder also involves a lack of the reflex to blink, they initiated eye drops. Theo spent his first 100 days in the neonatal intensive care unit, but he hasn't been in the hospital since and turned 3 in September.

"It was very much life or death," said Rikki Gutzdorf, of Watertown. "We firmly believe that without the genetic testing, we wouldn't have had a diagnosis and known how to treat it, and possibly would not have our son with us today."

Doctors are increasingly sequencing babies' genes in addition to newborn screening, especially in critically ill children whose symptoms aren't explained by the routine screening a day or two after birth that mostly involves metabolic tests.

Charting the A, C, G and T bases in the DNA molecules of sick infants can yield answers — sometimes in three days or less, according to a California study — that can improve treatment and save lives. The technology, which often involves enzymes made in Madison by the company Illumina, offers the potential to sequence all babies — not just for childhood disorders but, as a Boston study showed, for risk of adult-onset diseases such as some types of cancer.

The prospect of universal newborn sequencing, and looking for adult-onset conditions, raises questions about personal autonomy, according to a federally funded ethics group. In 2018, the group said the evidence "does not support genome-wide sequencing of all babies at birth."

Still, "at some point, that's going to be a reality," said Dr. Donald Basel, medical director of genetics at Children's Wisconsin. "We're all going to have knowledge of our genomic data."

For now, Basel said, genome sequencing at Children's Wisconsin is done on about 200 patients a year, primarily children in the neonatal or pediatric intensive care units. The testing leads to a diagnosis about 43% of the time and often leads to better care, he said.

"We've definitely made some significant changes in treatment based on the genomic data we've gotten back," Basel said.

At UW Health, doctors in April started sending blood samples from perplexing NICU patients to Rady Children's Hospital in San Diego, which led an effort to do rapid whole genome sequencing on sick infants. In July, UW opened an Undiagnosed Genetic Disease Clinic to identify rare diseases in children and adults, with the somewhat slower sequencing involved taking place on campus.



Amanda Maegli, research specialist at the DNA sequencing lab at the UW-Madison Biotechnology Center, loads a cartridge with DNA samples into a DNA sequencer.



LEFT: Rikki Gutzdorf was a preschool teacher specializing in special education before Theo was born with Stuve-Wiedemann syndrome. Her background, she said, "was the universe's way of preparing us for Theo."

RIGHT: Josh Gutzdorf said he's grateful for every day with Theo, who spent his first 100 days in the neonatal intensive care unit. "We didn't know if he was going to make it past one year of age," Josh said.



Each of more than 7,000 known rare genetic diseases affect relatively few people, but collectively the conditions are believed to be present in about 8% of the population, or roughly 450,000 Wisconsin residents, said Dr. Stephen Meyn, director of the UW Center for Human Genomics and Precision Medicine, which includes the new clinic.

Until recently, it took three to five years to diagnose one of the conditions, and doctors were successful less than 10% of the time, Meyn said. Now, with genome sequencing, the speed is quicker and the success rate is closer to 40%, he said.

"We are, as geneticists, thrilled with those numbers," Meyn said. "But that means most patients walk out of the clinic without a diagnosis. We have a lot of work to do still."

Rapid sequencing

A federal program that explored newborn genome sequencing in recent years included projects by researchers in San Diego and Boston.

Dr. Stephen Kingsmore, CEO of the Rady Children's Institute for

Genomic Medicine, led Project Baby Bear, which used rapid whole genome sequencing to analyze the genetic code of 184 infants at five NICUs in California covered by the state's Medicaid program.

The sequencing produced a diagnosis for 74 babies, or 40%, in a median time of three days, the researchers reported in June in the American Journal of Human Genetics. The testing, which cost \$1.7 million, led to about \$2.5 million in savings in treatment, the researchers said. California has approved \$3 million annually to expand the program statewide.

In one case, a scan showed problems in the brain of an inconsolable 5-week-old boy, whose older sister died at 11 months after seizures. Genome sequencing on the boy, completed in a record 13 hours in October 2020, identified a severe vitamin B12 disorder called THMD2. Soon the boy was started on a vitamin treatment and recovering from the condition that likely killed his sister, Kingsmore and colleagues reported in June in the New England Journal of Medicine.

"This case illustrates the potential for decreased suffering and improved outcomes through the implementation of rapid genome sequencing," they wrote.

The BabySeq Project, at Boston Children's Hospital, involved sequencing of nearly 1,000 genes on

127 healthy babies and 32 NICU patients. Some 8% of healthy babies and 9.4% overall had mutations for childhood genetic disorders, which were not predicted by clinical or family history, researchers reported in 2019 in the American Journal of Human Genetics.

Parents of more than half of the sequenced babies agreed to look for actionable adult-onset conditions. Three babies had such genetic risk factors, two for breast cancer and one for colon and endometrial cancer.

The information gleaned from the sequencing didn't cause undue distress for the parents, including the 15 families whose babies had an unexpected risk gene for childhood disease, according to survey results reported in August in JAMA Pediatrics. Now, BabySeq2 aims to expand the research to more diverse families in Boston, New York City and Birmingham, Alabama.

Wisconsin doctors seek answers

In Michigan, Dr. Caleb Bupp heads up Project Baby Deer — which, like its California precursor, is named after the state animal. Eight hospitals in the state have sent samples from more than 80 children to Rady in San Diego for rapid genome sequencing,

About this series

Sunday: Wisconsin doesn't screen newborns for some disorders, which can lead to disability and death.

Monday: Testing and treatment have greatly improved the outlook for people with a rare muscle disease.

Today: Doctors are increasingly turning to DNA sequencing to explain conditions missed by screening.

Series online

Point your smartphone's camera here to view this series online, including additional photos and video.



with analysis of the first 30 cases showing 40% led to diagnoses and 45% resulted in changes in treatment, said Bupp, chief of medical genetics at Spectrum Health Helen DeVos Children's Hospital in Grand Rapids.

Michigan's Medicaid program in September started covering genome sequencing for children up to a year old. Michigan's project also involves private insurers, including the state's largest carrier, Blue Cross and Blue Shield, for whom children with unexplained symptoms up to age 18 can be sequenced.

The testing helps parents by providing answers and doctors by providing direction, Bupp said. "It's also good for hospitals and payers, because if we know why we're treating folks, we give them better care and the cost of that care is lower," he said.

UW's American Family Children's Hospital is among 10 hospitals in the Sanford Children's Genomic Medicine Consortium — led by Sanford Health, based in Sioux Falls, South Dakota — that are sending samples to Rady for rapid genome sequencing, Meyn said.

Since April, sequencing has been performed on four NICU patients from UW, with three leading to diagnoses within five days, Meyn said. He said "the results led to major changes in medical management for all three of the diagnosed patients."

UW doesn't have enough cases to justify running costly sequencing equipment around the clock on-site for the rapid results needed to help deteriorating newborns, Meyn said.

Please see DNA, Page A7



AMBER ARNOLD, STATE JOURNAL

Theo Gutzdorf, who has a type of Stuve-Wiedemann syndrome that involves dwarfism, or short stature, uses a stool to navigate his play kitchen.

DNA

From A6

With older children or adults who have spent years trying to pinpoint the cause of their conditions, speed is not as vital, he said. That's why genome sequencing for patients at the new Undiagnosed Genetic Disease Clinic is done at UW-Madison's



Bupp

Biotechnology Center, which can run so-called "long-read" sequencing that can provide even more answers. UW plans to study up to 500 patients at the clinic over five years, with the goal of diagnosing rare disorders and discovering new disease genes.

Children's Wisconsin has been doing genome sequencing on some patients for more than a decade, Basel said. In 2009, the hospital received national attention for reading the genetic script of 4-year-old Nic Volker of Monona to diagnose his rare condition and treat him with a cord blood transplant.

Most sequencing at the hospital yields results in two or three weeks, but the timing can be sped up to about five days for urgent cases, Basel said. In those

situations, scientists using the sequencing equipment for research are asked to temporarily give it up.

"They basically have to put everything they're doing on hold so we can run our sample," he said. "We flood the plate with just our sample for a quick turnaround."

Adapting to a rare disease

At the Gutzdorfs' house in Watertown, Theo climbed a step to arrange pots and pans in his play kitchen on a recent afternoon before scuttling across the floor to pet Phil, the family's Lab mix.

Theo started walking on his own a year ago, a few months after he turned 2. Earlier, he used a walker Josh Gutzdorf fashioned out of PVC pipe. Medical walkers, even pediatric ones, were too big.

Theo is a little person, or dwarf, one aspect of his Stuve-Wiedemann syndrome. With his leg bones not only short but curved, doctors plan to operate on his hip to help him walk better. After surgery, he will need to be in a partial body cast for six weeks. His parents scheduled the procedure for January, when it's cold.

"Theo's body temperature can still be hard to regulate, even

with the medication he takes.

"It's a lot easier to warm him up than it is to cool him down," Josh said. When it's hot, "we can only be outside for 15 minutes, maybe, before he starts showing a lot of distress."

Theo is largely nonverbal but recently learned to say a few words, including "bye." His parents are trying out communication devices and teaching him some American Sign Language. Signing is a challenge, though, because his condition makes it difficult for him to open his fingers. At rest, his hands form fists.

He goes to speech therapy, physical therapy and occupational therapy. Though he eats mostly by mouth, he still has a feeding tube, used for medication, hydration and, when he's ill, nutrition, Rikki Gutzdorf said. His parents give him eye drops several times a day to prevent scarring of his cornea and hope he'll learn to blink when he gets older.

Stuve-Wiedemann is exceedingly rare, especially in the United States, so Theo's parents aren't sure what to expect for his future. They credit genome sequencing for helping him so far.

Without it, "we wouldn't be in the place we are today," Rikki said. "I wish for the families that don't have a diagnosis or a (disease) name."

Illumina makes reagents, tools for DNA sequencing

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At the west end of University Research Park on Madison's West Side, workers brew a potion with a powerful purpose.

Their flasks and tanks resemble those at breweries, but these workers aren't fermenting yeast to make beer. They're using harmless strains of E.coli bacteria to grow enzymes for genome sequencing.

San Diego-based Illumina has a major share of the global DNA sequencing market, and the company's manufacturing center in Madison makes enzymes — proteins that speed up chemical reactions — to run its sequencing machines. With 180 employees, the hub also produces what is called "library prep," or kits to prepare blood or saliva samples for sequencing.

"We're producing all the reagents and tools for everything the company does," said Bill Chcewicz, general manager at Illumina's Madison campus.

In 2011, Illumina bought Epicentre Biotechnologies, a Madison company founded in 1987. Epicentre developed a technology, called Nextera, to simplify and speed up preparation of samples for DNA sequencing, which is now part of Illumina's products.

In a building that opened in 2019 on Genomic Drive, workers make about 40 different enzymes. They induce E.coli cells to produce the desired proteins and purify the mixtures by passing

them through centrifuges, filters and columns.

In the fermentation room, equipped with a 1,000-liter tank, a caramel-colored liquid containing glucose and other ingredients feeds the E.coli cells. "The more cells you have that are expressing enzyme, the more enzyme you have when you go to break the cells open and purify them," said Carolyn Pettersson, associate director for manufacturing operations.

Genome sequencing conducted through efforts such as Project Baby Bear in California and BabySeq in Boston has used Illumina sequencers, said Julia Ortega, the company's director of scientific research. The DNA sequencing lab at UW-Madison's Biotechnology Center also has Illumina machines.

A study led by Illumina involved 354 infants with suspected genetic conditions at five neonatal intensive care units in five states. Half of the patients had whole genome sequencing within 15 days of admission and the other half got it within 60 days.

In both groups, the sequencing doubled the portion of babies who received precise diagnoses that altered their care, the researchers reported in September in JAMA Pediatrics.

"It really shows the power of the genome to provide a single test that can replace multiple tests that might be done and hopefully diagnose children earlier," Ortega said.



AMBER ARNOLD, STATE JOURNAL

Will Wayland, a manufacturing specialist at Illumina, checks valves on a fermentation tank. The company's facility in Madison makes enzymes for use globally in its DNA sequencing machines.

ECONOMY

Inflation puts squeeze on holiday budgets

ANNE D'INNOCENZO
Associated Press

NEW YORK — Enmarilis Velazquez is paying higher prices on everything from food to clothing.

Her monthly grocery bill has ballooned from \$650 in almost \$850 in recent months. To save money, she looks for less expensive cuts of meat and has switched to a cheaper detergent. She also clips coupons and shops for her kids' clothing at thrift stores instead of Children's Place.

For the holidays, she's scaling back on gifts. She plans to spend \$600 on her three young children instead of \$1,000, and she won't be buying any gifts for relatives.

"It's stressful," said the 33-year-old stay-at-home mother from Boardman, Ohio, whose husband earns \$30,000 a year making pallets for stores. "You want to give it all to your kids, even though (Christmas) is about family. They still expect things. It is hard that you can't give them what they ask for."

Retailers may be forecasting record-breaking sales for the holiday shopping season, but low-income customers are struggling as they bear the brunt of the highest inflation in 39 years.

The government's report last week that consumer prices jumped 6.8% over the past year showed that some of the largest cost spikes have been for such necessities as food, energy, housing, autos and clothing.

Overall, rising prices are changing shopping habits for many Americans. For some, they're a mere inconvenience, pushing them to delay building a deck on their house amid higher lumber prices. But for lower-income households with little or no cash cushions, they're making harder choices such as whether they can put food on the table or if they'll have to drastically scale back on holiday presents for their children — or forgo them completely.



DARRON CUMMINGS, ASSOCIATED PRESS

Customers wait in line to check out during a Black Friday sale Nov. 26 at Macy's in Indianapolis. Retailers overall are expecting record-breaking sales for the holiday shopping season, but low-income customers are struggling as they bear the brunt of the highest inflation in 39 years.

"Inflation is devastating the pocketbooks of low-income households," said C. Britt Beemer, chairman of the American Research Group, estimating that low-income households are cutting back their holiday buying by 20% from a year ago. "They are going to have to decide what they're going to buy and what they're going to eat."

Even some retailers that built their businesses around the allure of ultra-low prices have begun boosting them. Dollar Tree — the last true dollar store — is increasing its prices to \$1.25 for a majority of its products because of higher costs of goods and freight, Velazquez says that 25 cents extra per item adds up, and the increase will force her to scale back on impulse buying there.

Growth forecast

Despite the inflation pressures — as well as supply chain disruptions and the new COVID-19 omicron variant — the National Retail Federation says this year's holiday shopping season appears to be on track to exceed its sales growth forecast of between 8.5% and 10.5%.

According to a poll by The Associated Press-NORC Center for Public Affairs Research, about three-quarters of Americans say they will be giving gifts to friends and family to celebrate the winter holidays this year. But the rising costs have not gone unnoticed. About 6 in 10 Americans say holiday gift prices are higher than usual, while only 2 in 10 say they are not. Roughly 2 in 10 say they did not purchase gifts recently.

Overall, 4 in 10 Americans say it has been harder to afford the things they want to give as gifts this year. Roughly half say it's neither easier nor harder, while few say it has been easier.

But people in lower-income groups are feeling the cost pressures most acutely.

Forty-five percent of Americans in households earning less than \$50,000 annually and 40% in households earning between \$50,000 and \$100,000 say it has been harder to afford gifts this year, compared with 30% in higher-income households.

"It was hard enough a year ago, five years ago, for lower-income families to find extra money to buy gifts. But it is that much harder now," said Ted Rossman, senior industry ana-

lyst at CreditCards.com, whose survey in October found a significant number of low-income people were completely opting out of holiday gifting this year amid higher prices on essentials.

Financial stress

Such financial stress is being felt at the food pantries such as the one at Shiloh Church in Oakland, California. In the past three months, Shiloh has seen a spike in the number of people, particularly those with jobs, coming in to pick up a weekly box of essentials or shop at its market for free produce and other food, according to Jason Bautista, who runs the food pantry.

That prompted Bautista to bring in more holiday toys for the annual giveaway set for this Saturday. It will have about 2,000 toys to donate to families this weekend compared with about 1,500 a year ago.

"Families that would normally go to Safeway can't afford to with their fixed incomes," Bautista said. "Their dollar is not stretching."

Miriam Canales, 34, of Oakland, has been going weekly to Shiloh for free food since the beginning of the pandemic. Her husband lost his job as a chef at a restaurant that permanently closed in the spring of 2020. He got another job at a different restaurant a few months ago, but he's only working on average six hours a week.

She said higher food prices have added financial stress, and she will not be buying gifts for her children, ages 13 and 6. Instead, she plans to pick up toys on Saturday at Shiloh Church.

But Canales says she feels grateful this holiday season because of her husband's job as well as her daughter's recovery from brain radiation that landed her in the hospital with epilepsy a year ago. Now she's healthy again.

"I feel blessed," Canales said.

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.

Webinars:



Podcasts:



Books:



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In an era of global social media, rampant disinformation, and distrust of experts, the role of well-educated science and medical journalist is more critical than ever.

Today, journalists do not merely inform the public of new scientific advances. They must provide critical context and model a dispassionate attitude and a long-range outlook that can boost public confidence. At best, they awaken compassion.

Sonni Efron

CEO, National Press Foundation, Washington D.C.

The plight of patients with rare diseases is a critical unmet need of patients in healthcare. The statistics are frightening; there are 7000 rare diseases in the world that affect 350,000,000 people. One in eleven Americans has a rare disease. Three-quarters of patients with rare diseases are children and only half of patients receive an accurate diagnosis. The average delay for a patient to receive a diagnosis with a rare disease is 1 1/2 years. It is deeply concerning that one in four patients with a rare disease waits four years for an accurate diagnosis. There is an urgent need to communicate knowledge and expertise in the field of rare disease detection.

James A. Levine

MD, PhD, Professor, Fondation Ipsen, Paris

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