The Washington Post
Democracy Dies in Darkness

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Their children have a rare condition. They didn't know its name – until now.

Five families with children who have the newly discovered rare condition recently gathered in a D.C. park. Scientists believe 100,000 more people have it.

Today at 6:00 a.m. EDT

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Five-year-old Adeline Strohbeck lugged a box of White Cheddar Cheetos Puffs across the grass and watched as the other children and their parents started to arrive. Like a small mayor with a large white ribbon in her hair, she walked up to each and tried to engage them.

She didn't use words. Most of the other children didn't either.

At one point, Adeline plucked a tiny flowering weed and tried to hand it to Eleanor, a 3-year-old girl.

The adults standing by waited to see what the two would do next.

To someone watching from the outside, the interactions of five children in a D.C. park on a warm weekend afternoon would not have seemed extraordinary. But to those children's parents, every move offered insights — into what they could do now, into what they might be able to do someday.

The families recently learned they share a profound connection. They had each watched their children fail to hit the milestones others did with seeming ease. They were late to crawl and walk. Some have trouble eating and don't speak. A few have seizures. "Global developmental delays," the doctors call it when such lags in language, motor skills and cognition happen at the same time.

Doctors couldn't tell them why their children were experiencing what they were experiencing, and with no diagnosis, they had no clear prognosis for their kids, or themselves. It was an excruciating mystery that hummed through years of specialist visits and brain scans that yielded no clear answers.

Until last year.

Thanks to a global partnership among genetics researchers and the relentless organizing of parents, the five D.C.-area families who gathered in the park — and many more who aren't yet aware of the breakthrough — now have a name for their children's condition. The rare genetic anomaly that causes it was discovered just last year, and families that once felt alone with their questions are now finding answers and one another.

Eleanor clapped when she saw the flower that day. But instead of taking hold of it, she let more than a minute pass, seeming unable to grasp it. Three-year-old Rae lay beside the two, warming herself in the sun.
Her mom, Leila Levi, marveled at seeing the children together.
“You just hope,” she said, “that the world is kind to them.”

The discovery
A little over a year before, a leading expert on rare diseases at the University of Oxford, Nicky Whiffin, was looking at the data in front of her and thought there was a mistake.
One of her students, Yuyang Chen, had been searching through a vast British database, known as the 100,000 Genomes Project, as part of her team’s effort to ferret out answers for people with undiagnosed conditions. For years, Whiffin’s strategy had been to scour the less-studied regions of the genome. Out of nearly 9,000 patients with unexplained brain development disorders, Chen found that 46 of them had an identical change in their DNA, a single added letter in their 6-billion-part genetic code.
“That looks to me like it’s an error in the data,” Whiffin told him. In the world of rare conditions, that was a stunningly high number of cases. Something must be wrong with the sequencing technology or their analysis tools, she thought. So they set about trying to knock down their discovery.
“And we just couldn’t disprove it,” Whiffin said.
She reached out to collaborators around the world looking for confirmation, including a scientist in Boston who is part of the GREGoR Consortium, a national network of researchers working to diagnose rare disease cases. That scientist, Anne O’Donnell-Luria, sent a request to a geneticist at Children’s National Hospital in D.C. for more information about patients with the same genetic change, or similar ones in nearly the same place.
Seth Berger, a medical geneticist at Children’s National, looked at about 300 patients. A big part of his job is helping to discover new diseases. He had never seen anything like this.
“Typically when we find a new syndrome, I’ll find one in this region. And then we’ll find one in like Texas, and one in Florida, and maybe a couple in Europe somewhere. And you maybe find five people around the world,” Berger said.
Four families were found in the Washington area alone.
Whiffin published an early version of the findings in April 2024, and a final version in the journal Nature that July. The math was astounding — the implications for families even more so. She estimates that 0.4 percent of babies born with severe neurodevelopmental disorders have this newly identified genetic condition.
“Huge,” Berger said of the discovery.
“Kind of madness,” Whiffin said.
It’s called ReNU syndrome, a nod to the gene at issue, RNU4-2. By Whiffin’s calculations, 100,000 people around the globe are living with it.
Life without answers
When Kathy Yang got the call last year from Children’s National finally putting a name and scientific explanation to what was happening with her daughter Eleanor, she began to cry.
From the moment Eleanor was born, Yang knew something was different. The doctors told her she had nothing to worry about. But she had two other kids, and deep down, she knew.
Within months, doctors confirmed what she sensed. Eleanor was diagnosed with hypotonia, which is also called “floppy infant syndrome” and is characterized by low muscle tone. She had microcephaly, meaning her head was smaller than was typical of children her age. Yang went

into an Instagram spiral, trying to understand her daughter through photos of others with disabilities. “Is that Eleanor? Is that Eleanor?” she wondered.

During evenings left alone with her thoughts, she feared it was her fault.

“I was in my head, like, ‘Is it because I ate something wrong? Is it because I had that one sip of wine?’ So it haunts you,” Yang said.

What was facing Eleanor was not caused by, or inherited from, her parents. ReNU stems from what scientists call “de novo” genetic variants, or changes that are “newly arisen in the child,” Whiffin said.

When Yang finally learned the cause of her daughter’s condition, she cried. “I could take away some of the guilt,” she recalled.

There was much she didn’t know about raising a child with a developmental disability, and there are many questions that remain, like what will Eleanor’s life be as an adult? But, she said, she knows this: “She’s a joy to have in our family.”

Eleanor’s dad, Henry Liu, was struck watching his daughter with the other children affected by ReNU. “They’re all like twins,” he said. “They’re very good-natured. They’re happy kids.”

For eight years, Lindsay Pearse and her husband, Grant, who live in Warrenton, Virginia, tried to figure out what was happening with their son, Lars. They were in and out of the hospital, seeing specialist after specialist. They addressed Lars’s “global delays” with physical and speech therapy, a feeding tube and leg braces. But they, like the others, had not been able to dig out the cause of their child’s challenges. They thought they might never know.

Then they got a call early last year. Lars — like Adeline, Eleanor and Rae — had been enrolled in a research project with Berger at Children’s National. Their DNA had provided vital pieces of evidence in Whiffin’s global study.

For families, it’s been like they’ve somehow been given access to a time machine. Beyond the kinship and camaraderie of finding families who have gone through what they have, they now have a view into the opportunities and dangers coming years or decades down the line.

Lars had his first seizure when he was 2. They were sporadic at first, but in the last year they’ve ramped up, which had worried his parents. Now, the family is part of a community that includes parents of teenagers and young adults with ReNU syndrome who saw the same pattern in their kids and shared their experiences. From them, the family also learned that weak bones could emerge as a problem, better preparing them to try to address it.

The rush of practical information has pared back a sense of helplessness — and injected new energy and hope for many.

Pearse has used the fear and frustration of her family’s experience to fuel efforts to organize, helping found a patient advocacy group last fall, ReNU Syndrome United. The group, at its first conference in July, is developing a global road map, ranging from fundraising to coordination with researchers and companies, for finding therapeutics that could help people with ReNU.

“What we’ve learned from the scientific community is it’s very targetable,” Pearse said, adding that their sights are on a “bullish” five-year timeline to try to find treatments.

Many questions remain about what parts of the condition might be preventable or reversible. For example, a gene therapy breakthrough might address Lars’s troubles expressing himself through language, which she finds heartbreaking. That would be a relief as a parent, she said, and yet, “I also very much personally believe that Lars is perfect as he is.”

Whiffin said with new genetic therapies, including tools to edit DNA and target crucial proteins, ReNU could end up being “one of the fastest discoveries of a disorder to getting a treatment.” But there are many caveats, leaving her with “tempered hope.”

Researchers must learn more about the basic biology of ReNU, which affects a complex process within cells called splicing, to know if a therapy will be safe and effective, Whiffin said.

Since ReNU is a disorder of development, a child might need treatment very early on to make a difference, she said. Yet sometimes that's not needed. ReNU is drawing crucial interest from drug companies, as well as other researchers, she said, because so many people are affected. Turns out, their island wasn't deserted after all, Pearse said. Vast numbers of families are still out there. "We'd love to find them," she said.

Finding one another

Four of the families had met before the recent park gathering. They called themselves Team Real — for Rae, Eleanor, Addie and Lars.

A fifth Washington-area child, Sam, joined them for the first time that Sunday. His family just learned his diagnosis in April, thanks to a network of eagle-eyed moms — including Rae's — and a rapid-fire series of connections.

"Our sweet girl was diagnosed with a newly discovered genetic mutation last year," Rae's mother, Leila Levi, of Bethesda, Maryland, wrote in a Facebook post on April 3. She's normally guarded about exposing her family's business on social media but wanted to share information about ReNU and a fundraising page for the D.C. families.

Levi's law school classmate saw the post, thought the symptoms sounded familiar and told her friend, Sam's mom, Susannah Rosenblatt, who contacted Berger. He asked for Sam's raw data from previous genetic testing and easily pinpointed the added letter that Whiffin's team had identified.

When Sam, 5, rolled up to the park in jeans and a blue fleece, Adeline took an interest. She's social, which her parents credit partly to her twin brother Oliver, who has pushed her from the start. At school, she tries to take care of other kids, and is particularly drawn to those with special needs. At home, she sets out lunch boxes for her dolls, mothering them.

She'd never met Sam before. That day, she reached for a colorful pop-it toy he was holding. Then she tried to borrow a tablet Sam uses to communicate, since he makes sounds and gestures but does not speak.

"Addie, see that's his," her mom said.

"Does she like your toy? So you need to hug mommy about it?" his mom said.

"He's really good with the talker," Eleanor's mom said. "That's amazing." He had pushed a button generating the message: "My name is Sam."

The gathering sometimes appeared too much for Sam, who as his mom held him, began tugging at her hair, a coping mechanism she hopes to help him grow out of — and a scene familiar to some of the other parents there. That's something Lars does too sometimes, when he can't communicate what he wants or needs, his mother explained.

"Nobody was bothered," Rosenblatt said later, describing her relief. "There was no awkwardness or judgment if your child's acting in an unexpected way, because they can totally understand and relate."