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## Braving the Pain: Youngster fights rare disease with courage

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Staff Photo: Jason Braverman

Kim Avery tries to comfort her 12-year-old son, Tyler, who deals with excruciating pain during a recent visit to Emory Johns Creek Hospital in Duluth. Tyler initially went to his pediatrician's office when he was not feeling well but got so bad the doctor called for an ambulance. He was transported to Johns Creek before being taken to Children's Healthcare of Atlanta where he was admitted for three days. Tyler has been in the hospital more than 100 days in 12 months.

### Finding the problem

A number of mitochondrial disease cases are present in infants, many of whom can die within hours and days. Most

The sign, one of several on the hospital's walls, likely goes unnoticed by many parents on trips to Children's Healthcare of Atlanta.

It shows a pain scale, rating a child's discomfort with numbers, smiles and frowns. Fortunate parents don't have to use the scale, and many don't have to see their children reach the higher levels of the chart.

Others aren't so lucky.

Tyler Avery's pain scale is always in effect. On good days, the 12-year-old rates a 2 or 3. On his worst, it's a 10, the most excruciating pain imaginable.

That kind of agony has become part of the Sugar Hill resident's life while dealing with a rare mitochondrial disease, one that has put him in the hospital for more than 100 days in the past 12 months.

It was at its worst for two emotional months late last year, when Tyler split time between CHOA and the Mayo Clinic in Rochester, Minn.

"For two months, (the pain) never got below a 6 and it reached 10 on a daily basis," said John Avery, Tyler's father.

John and his wife Kim have endured every parent's nightmare on a frequent basis over the past several years, first while searching for a diagnosis of their son's condition and then while managing a complex disease. They ran the gamut of emotions from concern to frustration to fear, never more frightful than during one of Tyler's episodes, which began occurring as often as every few weeks.

Each episode is the most gut-wrenching scene imaginable, with Tyler screaming, groaning and fearing for his life. He can only lie flat, even a pillow or thin sheet causes intense pain. For 24 to 48 hours he doesn't even have the strength to squeeze his parents' hands.

There's also another cruel kicker - pain medicines don't alleviate the symptoms. His body metabolizes them so quickly they only give minutes of slight relief and then can't be administered again for hours. There's also a fine line - too much pain medicine causes him to code.

"It feels like he's been in a bad car accident, there's a car on top of him and he's begging us to move the car," Kim Avery said of her son, the second-oldest of her four children. "And there's nothing we can do. All we can tell him is 'It's going to be OK.' And he's looking at us like, 'No, it's not going to be OK. I'm not going to make it through. Knock me out. Do something.'"

cases occur in children, but others can develop the symptoms into their adult life.

The disease is caused by the failure of mitochondria, specialized compartments in every cell of the human body with the exception of red blood cells. Mitochondria are counted on for producing more than 90 percent of the energy needed to sustain life and growth, so cells begin to be damaged and eventually affect a large number of major organs. In the long term, conditions like organ failure and heart disease can be a serious side effect.

Mitochondrial disease is also very hard to diagnose, something the Averys know as well as anyone. It took countless appointments and hospital trips to make headway with Tyler's pain, which began with migraines, muscle pain and weakness when he was 7. The migraines are so harsh that they occasionally paralyze one side of his body.

The reoccurring problems were followed by repeated tests, including four painful (and as it turns out unnecessary) spinal taps to rule out meningitis. There were MRIs. Tests for leukemia. Every type of scan imaginable.

Unfortunately many hospitals offered a less than ideal answer for the Averys, telling them their child was just sensitive, or that he was suffering growing pains.

"It's incredibly frustrating when you feel there's something mysterious your child has," Kim Avery said. "We could tell that this was not normal for an 11-year-old to sleep all the time. ... They did all the basic tests, there are 20-something scans they do, and once they reach that, there's just like, 'We can't find it. It must be in his head.' We got his records later and it said things like 'parent driven' and 'psych component,' because they couldn't figure it out. When the doctors would leave, Tyler would say, 'Why, mom, why? Why are they saying that?'"

Eventually the Averys got a beacon of hope with the help of local pediatrician Vidya Krishnamurthy, who works in clinical genetics. She took one simple step by writing a care letter for the Averys that they could take to the emergency room, explaining that the way to treat Tyler was to administer fluids as quickly as possible.

That alone was a major step.

But the family also got an answer when a muscle biopsy - the best way to discover mitochondrial disease - revealed that he had point mutation in his DNA and Complex I and Complex IV mitochondrial disease.

"My doctor is my hero," said the soft-spoken Tyler, who recently spent his 12th birthday in the hospital. "She was the first one that understood my situation."

A normal kid

What makes Tyler's case so difficult is that it comes in episodes, unlike some children whose cases are so severe that they can't eat, walk or talk. He not only functions, he functions exceptionally well.

Some people who know Tyler don't even know he has such a painful disease.

He swims with Wild Timber Extreme, his neighborhood team, and qualified for the county championships last year. He has played basketball and soccer. He spent most of this fall playing baseball - his father John is a former Atlanta Braves minor-leaguer - and even pitched a late-season game.

He also loves to golf and fish, proudly discussing the 7-pound bass he caught while fishing with his grandfather this summer. He enjoyed a Boy Scout trip over the summer. He plays the drums and the guitar.

"He looks so healthy and so normal," Kim Avery said. "People say it's so strange how he looks so normal, how can he be so sick? Because they don't understand the episodes."

Kim is a home-school teacher, a blessing because it allows Tyler to stay on track in school despite his long periods of illness. He fits in classes during the summer and on weekends when it's necessary, and tests very highly to the amazement of his doctors.

He's already doing pre-algebra and teaching his two younger siblings, even though he has to take the occasional breaks when his hands hurt from writing.

His maturity makes him very aware of his condition.

"He's a bright kid, very articulate," said Fran Kendall, an assistant professor in the Emory University School of Medicine's Department of Human Genetics and one of Tyler's doctors. "He has a good sense of what he's going through and he's able to explain to other people what he's going through.

"Kids that go through this with courage and understanding, you have to respect and admire them for going through this in a positive manner."

Tyler has even maintained his dry sense of humor.

"One doctor told him when he grew up he should be a geneticist," Kim Avery said. "When (the doctor) left, Tyler said, 'So the new plan is for me to grow up, become a geneticist and save myself. That doesn't sound too promising mom.'"

#### Long-term treatment

Public awareness of mitochondrial disease isn't high, despite its relatively frequent occurrence.

The United Mitochondrial Disease Foundation estimates that every 30 minutes a child is born who will develop a mitochondrial disease by the age of 10. It also presents the statistics in another way - each year 1,000 to 4,000 children in the United States are born with a mitochondrial disease.

"It's not that uncommon," Emory's Kendall said. "It's interesting that people know nothing of it. For us doctors in this field and certainly for parents, it's something we struggle against. We get very little air time and certainly very little funding."

Since his two-month ordeal late last year, Tyler has traveled the country to see specialists and attend symposiums, enduring long car and plane trips that wear out his aching back. The uniqueness of his disorder led to his being accepted into the exclusive National Institutes of Health's Human Genome Research Institute in Bethesda, Md.

Only the rarest of cases are accepted to the institute, which will use a large team of experts to attack Tyler's problems. At the very least, the Averys are hopeful that the program will offer new ways to treat their son, to relieve his pain.

But they feel Tyler is up to the challenge.

"It's amazing how much he's matured," John Avery said of his son. "I don't discount any of my kids, everybody has a different personality. But I don't know another kid, even my other three kids, who could get through this. Ty is the one who can get through. He has the biggest heart and he just cares so much for other people.

"He doesn't think, 'I might get sick soon.' He doesn't think about next week. He just thinks about today, which is neat."

That kind of attitude, his family says, keeps him positive through his treatments. A port in his chest allows him to receive IV fluids at home, which has helped limit his emergency room visits this fall.

He takes a steady round of medicine, an astonishing 22 pills and one liquid a day - most are vitamins.

"The liquid vitamin is the worst," Tyler said. "You've got to take it with Coke or something strong."

The Averys know their son is plenty strong, too.

He faces his daily pain without whining. He manages not to get discouraged when planned vacations or activities get postponed, when he has to miss sporting events. He doesn't sulk, instead using the days when he has energy for his favorite activities.

Those traits have impressed many in Tyler's life, including his doctors.

"Tyler's a very good boy," Krishnamurthy said. "He's tough for what he's gone through. You and I, with all these treatments, we wouldn't be good patients. I'd be an angry patient. But Tyler, he's a trooper. He's holding up really well. He's a very good boy."

And one who knows the extent of pain far too well.