Robert Stone's Story – Genomics Used for A Diagnosis After 14 Years

Fast Crowdfunding and Community Building Leads to a Breakthrough

Robert was born in 1997 and was a typical, happy baby until just after he turned one. Then over the course of three days, he had brief episodes of slowed or interrupted movements. This culminated in a dramatic medical breakdown.

At first, "It was a few seconds on vacation in our hotel, when he fell backwards and just froze. Then he got up and looked fine. What did I just see?" Jeneva Stone, Robert's mother, asked herself. "You can't press rewind on your life." The reasonable explanation was he was tired, off schedule.

Then, "It was so sudden and so terrible and completely out of the blue," Jeneva describes the day when he had his medical crash. He lost use and control of his arms and legs, his mobility and his ability to communicate.

Jeneva took Robert to numerous doctors for testing and the months turned into years without a diagnosis. All diagnostic testing strangely came back normal. "At first you feel like you won the lottery when you get negative test results for some life-threatening disease, but after over a decade, you are devastated when test results have no diagnosis," Jeneva said.

Fifteen years later, Robert, now 16, is in a wheelchair and has a feeding tube. Although difficult, he tries to communicate using his limited mobility. He lifts his right hand for "yes" and his left hand for "no," or by facial expressions. Robert's parents always believed there was some rare genetic disease causing Robert's condition.

"After 14 years with no diagnosis, last year we pursued gene sequencing through Rare Genomics Institute and finally found an answer."

- Jeneva Stone

Jeneva connected with Rare Genomics Institute through another mother who has a child with an undiagnosed medical condition. "I saw it as a last-ditch effort to figure out what was wrong," Jeneva says. The Stones brought Robert and their binder filled with over 100 diagnostic tests to RGI. After years of testing with no answers and many medical bills, the Stones decided to try raising money for Robert's genomic sequencing through RGI's crowdfunding campaign.

Crowdfunding, as the name suggests, is asking a crowd of people to donate to a cause. RGI helps families create a patient profile, which allows parents to share their child's photograph and medical story on the RGI website. Friends, family, or anyone can donate directly to the child's research site to raise the funds needed for sequencing. See the Parent's Toolkit.

Roger Stone moves his son Robert from his wheelchair to the family couch. Robert suffers from Dystonia 16 - which only 8 other people in the world have. His family spent 14 years trying to get him diagnosed and finally got a diagnosis in 2012.

"We empowered an entire community," Dr. Lin of Rare Genomics Institute said, and within six weeks, Robert's campaign reached its fundraising goal of $7,500 for sequencing the parents and Robert. The average donation was less than $50. About one-third of the money came from complete strangers and the rest from a community effort using Facebook to tell family and friends about the RGI crowdfunding site for Robert.

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Robert began genomic sequencing, and within 9 months, the researchers found what was causing Robert's condition. Robert and his parents finally had a diagnosis. He has a syndrome called Dystonia 16. Robert is only the ninth person ever reported to have this syndrome. "It was a one in three billion lightning strike," explained doctors to the Stone family. This would never have been diagnosed without exome sequencing.

The only other people with this illness are in Brazil and one in Germany. He has an extremely rare combination of two mutations, one on each copy of a gene called PRKRA. The diagnosis gave the family answers and a sense of peace. They found the answer for Robert. They didn't give up, and they were right. They now know what they always suspected, that Robert has normal intelligence. They always believed he did. He does understand them.

With this diagnosis, they can use medications that can be targeted towards his illness that work better. They are using a repurposed drug called Sinemet, for Parkinson's disease to initiate movement. The knowledge of what caused their son's illness has given the Stones new information to understand and improve his quality of life. "We'll do whatever we can to connect with people who are doing research in this area," Jeneva said. They are now hoping that this discovery will lead to new treatments for their son and others with dystonias and movement disorders. "We always knew the science would catch up to Robert," Jeneva said. "We didn't give up, we found out what was wrong with him."

Takeaways:

1. Crowdfunding enabled a medical breakthrough. A fourteen-year medical mystery for Robert Stone and his family was solved from many $25 or $50 donations from friends and family. Now they can focus on targeted treatments for their son.

2. Exome sequencing found a disease so rare that it would never have been solved without sequencing. He is the ninth person ever diagnosed with Dystonia 16. The only other reported cases in all medical literature are in Brazil and Germany.

3. Many times genomic testing will reveal a more important result. An exact diagnosis tells you what it is, and what it is not. Some thought that a child with severe physical disabilities automatically had severe mental disabilities. Some assumed Robert could not understand or communicate. Exome sequencing determined he had a movement disorder. He does understand and can communicate. The Stones now know with certainty what they always suspected, that Robert has normal intelligence.

4. Document your child to track patterns. “You can’t rewind your life,” Jeneva said. If you think something is off or not quite right, write down a description of what you observed, including the date of when it happened.

5. Construct a binder that contains all your relevant and significant medical records, including MRI disks. The binder that Jeneva Stone kept for her son Robert was so well done that it is famous among doctors. You should get a copy of every lab report on lab stationary. Not every lab runs tests the exact same way. This will give you the most accurate information and smallest chance to have to repeat the same test. Get test results and any other medical reports. See the Parent’s Toolkit section for a detailed breakdown and photos of what was in her binder and how she organized it. Doctors have said having this information and organizing it can save 3-6 months of time to diagnosis.

6. Building a community around your child adds great value. Jeneva Stone, Robert's mother, is a writer. She has built of community of support around her and her son. She has written in prominent journals on disability and parenting. She has been a strong advocate for her son.

7. What happened to Robert and finding this diagnosis can be used to help others suffering from rare diseases. Research can be done on Robert's Dystonia 16, as well as other dystonias and movement disorders.

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