NICHQ National Institute for Children's Health Quality

Insights

The Impact of Rare Disease

Stories shared from patients and families affected by rare diseases

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In the U.S., a rare disease is defined as a condition that affects fewer than 200,000 people. Millions of Americans, many of them children, are living with a rare disease. To help show the impact and prevalence of rare diseases, we invited our network to share their stories. Read their submissions below and share your own experiences with us on social media and tag @NICHQ.

Interested in sharing your story? We're continuing to accept submissions for NICHQ's 2021 Rare Disease Day campaign.

Acquired Aplastic Anemia, Emily, West Virginia

> Acquired Aplastic Anemia, is "a rare and serious blood disorder due to failure of the bone marrow to produce blood cells." Learn more here.

My oldest son was diagnosed with what we initially thought was leukemia, but quickly discovered was Aplastic Anemia. His health deteriorated: he was immunocompromised and had chronic nosebleeds and no energy. We were in and out of the hospital with fevers and infections. He was transfused to increase his cell lines. He was the oldest of four siblings (three biological). Both of his brothers and both my husband and I had testing to see if anyone was a bone marrow match. His brother, only 13 months younger than him, was a perfect match. Children's Hospital of Colorado completed a bone marrow transplant with great success. My son has fully recovered but the fear and anxiety of being so close to death follows him every day, as it does for all of us. He struggles with the emotional fallout from the trauma of both the symptoms he experienced, as well as the treatment. His outcomes were really a best-case scenario.

Myelodysplastic Syndromes, Bambie, Washington

> Myelodysplastic Syndromes a rare group of blood disorders that occur as a result of disordered development of blood cells within the bone marrow." Learn more here.

As a public health nurse for both Early Head Start and Washington's state preschool program, I interact with families daily who carry the burden of living with or having a child with a chronic illness. The weight of living with a chronic illness takes its toll on a person in more ways than I have the vocabulary to describe. But on a positive note, living with my own chronic illness, [Myelodysplastic Syndrome], allows me to empathize and advocate for the families I serve. I sometimes share my story with my families so they know I can relate and that I have a heart (and ears!) open and ready to simply listen to them and understand their intimate struggles. Sometimes we cry together; sometimes we just have to laugh so we won't cry. It's a very humbling and personal experience for me and I try to use it for good.

RYR-1 Congenital Fiber-type Disproportion (CFTD), Nikki, Ohio

> RYR-1-related diseases are conditions caused by changes (mutations) in the RYR-1 gene, which are typically associated with core myopathies**.** Congenital Fiber-type Disproportion is "a rare genetic muscle disease that is usually apparent at birth. It belongs to a group of muscle conditions called the congenital myopathies that tend to affect people in a similar pattern." Learn more here.

A genetic condition that causes weak muscles and disability, RYR1 CFTD has stolen a lot from my family. My sister, Chavon, born when I was 8, entered the world with a weak cry and challenges with eating, breathing and moving. She would grow up to become my best friend, and she persevered in spite of every challenge. Dealing with significant motor impairment was difficult throughout her life. She finished college, started a doctoral program in bioengineering, and traveled the country, but in 2012, when she was 27, she succumbed to the condition.

In 2011, my son Richie was born, weak and with a cry like Chavon's. We discovered Richie's correct genetic diagnosis after years of testing—Chavon had been misdiagnosed with muscular dystrophy as a child. For Richie, respiratory function was a major health concern. The tracheostomy, back braces, ventilator and feeding tube are among the many technologies that have supported his life and improved his quality of life over the years. There is no cure for RYR1 CFTD, but we hold hope for the future. Through research, we can better provide diagnoses and treatment that can lead to better lives, even for the rarest conditions.

Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT), Ruth, Illinois

> Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) is "a genetic disorder that causes an abnormally fast and irregular heart rhythm in response to physical activity or emotional stress." Learn more here.

At 10 years old, our son was swimming at camp when the lifeguard noticed he was no longer moving in the water. He was rescued successfully but could only say he felt cold and his vision went black. Six months later, he was running at the bus stop when he fell to the sidewalk. Again, same story but this time he added that he felt like his fingers were tingling. In both scenarios, all medical tests were negative.

Premature Ventricular Contractions (PVCs):
Premature ventricular contractions (PVCs) are extra heartbeats that begin in one of your heart's two lower pumping chambers (ventricles). Learn more here.

He was sent for a cardiac workup. During a stress test, he started showing multifocal PVCs, and started complaining of tingling in his fingers and that ‘he was going to go down.’ Many more cardiac exams followed. His cardiologist mentioned CPVT but said it was very uncommon. He put him on a beta blocker. The third incident occurred when he was playing baseball. He had just run to first base when he went down. Luckily, he was wearing an event monitor which captured the abnormal rhythm. It was then that the cardiologist said, "I can't cure it, but we need to prevent these episodes from causing a cardiac arrest which we cannot bring him back from."

At 11 years old, our son received an implantable cardioverter defibrillator (ICD) [an implanted device that monitors heart rate]. He has had two replacements since then. He has successfully graduated college and is working in a job he loves. Through genetic testing it was determined that he had the RYR2 form and did not inherit this from his parents. Any children he has will need to be tested for the gene shortly after birth.

Colpocephaly, Tess, Connecticut

> Colpocephaly is "a congenital brain abnormality in which the occipital horns – the posterior or rear portion of the lateral ventricles (cavities) of the brain – are larger than normal because white matter in the posterior cerebrum has failed to develop or thicken.” Learn more here.

My brother Greg was diagnosed with colpocephaly as an adult. He led a good life with a successful career, was a member of MENSA and held a pilot's license. Even with the diagnosis, he held out the hope that he was "recovering from colpocephaly" but with the normal loss of neurons from aging, he lost many physical capabilities, and succumbed last year, just after his 59th birthday. My wish is for innovative treatments, perhaps stem cell transplants, for patients like Greg to continue to live happy and productive lives.

Congenital Cytomegalovirus (CMV), Amanda, Iowa

> Congenital Cytomegalovirus (CMV) a viral infection that, when found in newborns, “ranges in severity from being without symptoms to being a severe disease affecting the liver, spleen and central nervous system, with possible developmental disabilities.” Learn more here.

We had a typical, healthy, planned pregnancy until 20 weeks gestation. At that time, we were informed that our baby's brain was not developing typically. An amniocentesis, [a prenatal test to screen for developmental abnormalities], revealed that our baby was infected with Cytomegalovirus. We had never heard of CMV before, and certainly had no idea how devastating it can be to a developing baby. We found out that experimental treatment was available, and we had four Cytogam [a medication] infusions during the pregnancy.

Our baby, Pippa, was born at 34 weeks gestation. Her symptoms at birth included intracranial calcifications and thrombocytopenia [a condition causing low blood platelet count]. She did well and came home from the hospital after three weeks. Pippa also received six months of oral antiviral therapy at home.

At around 6 months old, Pippa started to fall behind on the typical "milestones" for infants. An MRI revealed that the CMV infection had done more damage to Pippa's developing brain than we had originally thought. She was diagnosed with generalized polymicrogyria [a condition characterized by abnormal development of the brain before birth]. Today Pippa's diagnoses include developmental delay, intellectual disability, swallowing disorder, speech delay, epilepsy, and hearing loss.

Our lives have completely changed because of Pippa's diagnosis. We built a more accessible home for her and moved school districts. We have meetings with the school regarding her individual education plan. She goes to physical therapy, speech therapy, feeding therapy, and occupational therapy. Pippa sees a neurologist, audiologist, and ophthalmologist regularly. From the activities we do together to the dinners we make, every aspect of our family life has been impacted by Pippa's diagnosis. Congenital CMV has also turned our family into advocates. We want people to know how it can be prevented and how it can be treated. We helped get a law passed in our state that helps CMV get diagnosed early so they can have the best possible outcome.

Congenital CMV has caused our family an enormous amount of stress and pain, but with that has also come a sense of belonging and purpose. We are so lucky to know Pippa; she has brought an immeasurable about of joy to our lives, and the lives of everyone she meets.

Our sincerest gratitude to all those who shared their stories with us. Raising awareness about rare diseases can help build national momentum to improve care and outcomes.