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INSIGHTS

Family reflections: Wolfram syndrome-Lauren's story

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My name is Pat Gibilisco, and my husband and I have two daughters. Lauren is 32 years old with Wolfram syndrome (WS) and our other daughter Megan is 30 years old and a carrier of WS. We live in Nebraska. Lauren was diagnosed 20 years ago at age 12 with WS. At that time there were no foundations representing this disease nor was there any research taking place, I referred to that time period as "The Dark Ages". Rob Birkinshaw, another parent who had children with WS and I decided to start the very first WS Family Support Group in 1990. In the beginning, it was just four families; two in the United States, two in England and one in Australia. Our WS support group has since grown over the years to more than 160 families.

Wolfram syndrome is a deadly and multifaceted disease. WS presents many manifestations, and unfortunately, my daughter, Lauren, suffers from most of them; diabetes mellitus, diabetes insipidus, extreme hearing loss, optic atrophy, choking issues, seizures, total loss of taste and smell, ataxia, heat intolerance, short term memory loss and problems with her bladder and bowels. Our family has seen an army of specialists trying to treat all these symptoms. Throughout all of Lauren's struggles, the most amazing thing is I have never heard one complaint out of her mouth. She just takes on every new issue with determination. This is a common factor with all Wolfram children. They are very bright, kind, and considerate, they are warriors.

These WS patients look completely normal on the outside, so people do not realize how sick they truly are. Other than using a cane for Lauren's blindness many people are not aware of what my daughter goes through each and every day. It was difficult during Lauren's school years because Lauren appeared normal like her other classmates, unfortunately, she was picked on and some of her classmates were very cruel, she was an easy target. As a parent with a special needs child, you are a constant advocate for them. I had to make sure that Lauren was receiving the accommodations that she needed in order to ensure she was receiving the education that she deserved.

Wolfram Syndrome has affected our whole family. If we planned to leave the house, this took time and consideration. Factors that impacted our ability to leave the house were, temperature, location of nearest bathrooms, Lauren's ability to hear and navigate in her surroundings. There were many places and things we as a family could not see or do.

A glimmer of hope was presented by a doctor named Alan Permutt. Dr. Permutt announced he was starting a WS research clinic with mice. After five years of mice research, Dr. Permutt was ready to start human studies. Lauren was among the first patients to be invited to Washington University School of Medicine in St. Louis where Dr. Alan Permutt and Dr. Tamara Hershey set up a clinic for Wolfram syndrome patients. Unfortunately, Dr. Permutt passed away in 2012 but before his death, he made sure that his research would continue. Dr. Tamara Hershey has been conducting the Wolfram syndrome research in clinics for the past nine years, she has written several research papers, and through grants, she has received funding for these important clinics. Dr. Bess Marshall, a pediatric endocrinologist at Children's Hospital St. Louis has been a great help for all the WS families. She helps us navigate medical issues and also writes letters explaining patients' needs in regards to insurance, school accommodations or physicians. Our WS family is fortunate to also have Dr. Fumihiko Urano, one of the world's top researchers on Wolfram syndrome, he just finished the first safety trial on a drug that could help slow the progression of this dreadful disease. All of these doctors and researchers have been there to help us in our quest for a cure.

My daughter's struggles of living a life totally blind and having limited hearing leave her literally in the dark quite often. These patients suffer from severe bladder issues, most of these children have to catheterize in order to drain their bladder. Lauren is a little different in that, her bladder constantly spasms and leaves her in a constant state of feeling the need to void. She uses a neuro-stimulator and Botox treatments to control these constant urges. These symptoms are the worst for Lauren, but other symptoms might affect other families differently.

The problem with Wolfram syndrome is that it is an Ultra Rare Disease and trying to get funding to continue research has been a constant battle. We tried for many years to get this disease recognized and receive the help but we were not successful, it seemed as if no one cared. We finally received some help when a woman named Stephanie Gebel joined our group. Stephanie's five-year-old daughter was diagnosed with WS and she too learned that very little was taken place in regards to WS research and funding. In 2012, Stephanie started the first non-profit in the United States and has raised over a million dollars to help fund research and clinical trials. It is unfortunate that our WS family has to rely solely on ourselves to raise money in hopes of a cure.

Our hope is that researchers will look at drug repurposing and accelerate the pace which therapies developed for diabetes and other neurodegenerative diseases can be tested in the lab and brought to clinical trials. I would like researchers to apply powerful new technologies like CRISPR, single-cell sequencing technology for achieving a cure for Wolfram syndrome. We need a cure desperately. Our biggest issue is financial support. It is the single limiting factor in our ability to get the necessary drug therapy. Because Wolfram syndrome is a rare disease it is often overlooked by big pharma companies. It is so painful to see your child lose their vision, lose their hearing, have trouble walking because of balance, lose the thrill of knowing what you are eating and smelling, not being able to be out in the summer because of the

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heat, suffer from seizures and have the embarrassment of dealing with bladder and bowel issues. Our children are deteriorating because we cannot find someone who will financially support our research. It truly is heartbreaking. I would like to see the worldwide scientific community come together with their expertize to bear on behalf of WS. We have been able to eliminate many of the obstacles that once stood in the way of research such as lack of tissue, disease models, and coordination among researchers. Now we need a cure. Medium age of death for someone with Wolfram syndrome is 40 years. Time is running out for Lauren and many of the other Wolfram syndrome patients.

The vital work of the Snow Foundation is made possible by generous individuals, corporate partners, and foundations who believe strongly in supporting breakthroughs in scientific research that not only impact WS patients but millions of people suffering from diabetes and other neurological diseases as well. For more information on Wolfram syndrome or to donate towards our cause, please visit thesnowfoundation.org.