Chicago

A few Faces of Wilson's Disease in Chicago

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Wilson's Disease is a rare disorder that affects approximately 1 in 30,000 people. No two cases look the same. The purpose of this presentation is to spread awareness of Wilson's Disease by sharing a few stories.

Steve and Julie Goostree

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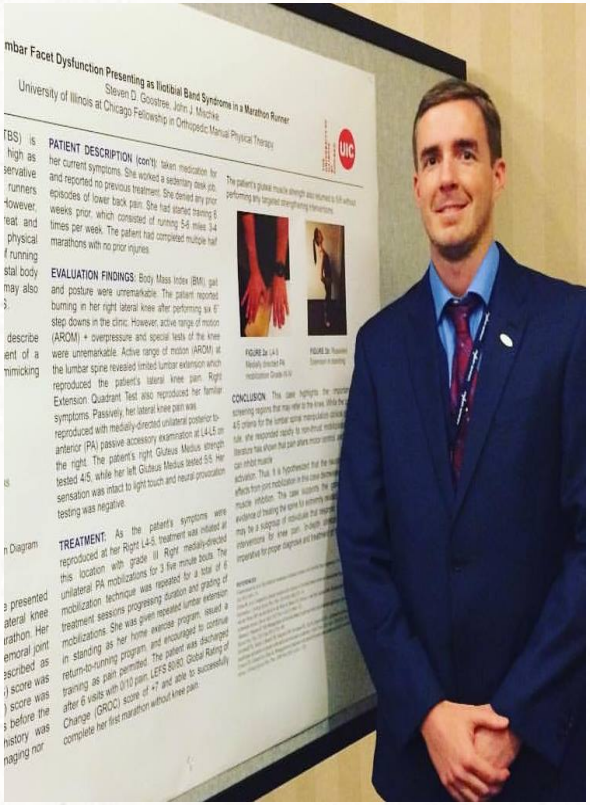
Steve

30 years old

Diagnosed at age 17



Julie

28 years old

Diagnosed at age 14

Steve and Julie Goostree

In the summer of 2002, as 14-year old Julie was getting ready to enter high school, her mom took her to the doctor for a sports physical. Julie had just switched from a pediatrician to an internist, so he did more thorough blood tests than the pediatrician would have. When her results came back, her liver enzymes were skyrocketing. After much confusion and a lot of testing, Julie was diagnosed with Wilson's Disease. After both of her brothers were tested, it turned out her brother Steve, who was 17 at the time, also had Wilson's Disease. Mansoor Khan from Alexian Brothers and Fred Askari from University of Michigan were both extremely helpful to Julie, Steve, and the entire family. Julie is currently a teacher and Steve is a physical therapist. They are both extremely grateful to have found out about their Wilson's Disease when they did, and diligently take their medicine. They are extremely appreciative of their parents who have provided endless amounts of support and guidance. The entire family hopes to spread awareness about Wilson's Disease.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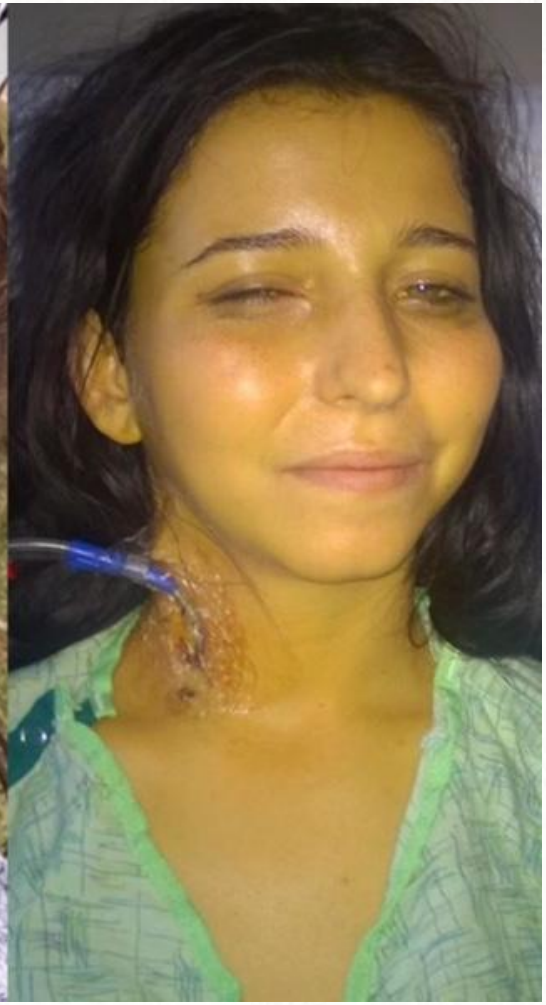

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On March 30, 2015, I was a seemingly normal, healthy, 18 year old enjoying the last semester of high school. I was a varsity soccer player and deciding between spending my college years at Cal Poly in California or at Clemson University in South Carolina the following fall. My parents took me to the doctor because my skin and eyes were yellowing dramatically. A blood test revealed that my liver enzyme, as well as most of my other blood work, was extremely elevated. I was immediately rushed to Northwestern Memorial Hospital by ambulance and admitted straight onto the transplant floor. Countless doctors began to stream in from hepatology, neurology, gastroenterology, ophthalmology and many other departments.

It was in the early hours of April 1st that we initially heard about Wilson's Disease. The doctors explained the condition and its rarity. They said if left undiagnosed and untreated Wilson's will result in a liver transplant. The doctors were fairly certain that I was suffering from end stage Wilson's Disease and registered me on the national transplant list. My team of doctors assured us that once they confirmed that my ailment was Wilson's, I would rise to the top of the list as a status one patient. Many tests were done in search for this diagnosis. One test was done to analyze my urine, which was now as dark as coffee. The result was the first confirmation that I in fact did have Wilson's Disease. On April 3, 2015 I was listed as status one.

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During my time in the ICU waiting for a lifesaving liver transplant, I underwent many procedures to prolong my life and keep me stable. One of which is called plasmapheresis. This was done by attaching two needles to the catheter tube that was inserted in my heart. Simply, my blood was being taken out of my body through one of the catheter tubes. It was then run through the apheresis machine. Once in the machine, my blood cells were separated from the plasma and the copper was stripped out by spinning the blood at high speeds. After, my blood cells were mixed with replacement plasma and the new mixed blood was then returned into my body through the other catheter tube. This procedure took two hours each time. My parents and I would sit and watch the large bag fill with another dark brown fluid that is supposed to be white. Beyond the plasmapheresis, I was also given medicine in hopes of eliminating the ammonia from my system before it started to negatively affect my brain. All of these were temporary treatments to extend my life. Nearly one week after my original acute liver failure and diagnosis, on April 5, 2015 I received a lifesaving liver from a deceased donor.

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Today, in hindsight, I think about other symptoms I had throughout my childhood that could and should have pointed to Wilson's Disease. I was tired and moody. I constantly wanted to chew on ice. The size of my legs were getting bigger each year even though I was an avid athlete. In fact, we even went so far to blame the enlargement on my years of soccer. We thought it must be muscle development from all time I spent playing, but actually my legs had been filling with more fluid every year due to my failing liver. The larger missed symptom was when I was 16, two years prior to my liver failure. I had been suffering from extreme fatigue and could not chew enough ice. As a result, I had blood work done to rule out anemia. The bloodwork did not show signs of anemia, but did show high liver enzymes! Tragically, this sign was misdiagnosed and I was sent to an oncologist instead of a gastroenterologist. I was instead diagnosed with mononucleosis and my Wilson's Disease continued to go undetected.

My hope and purpose in sharing my story is to spread awareness about Wilson's Disease. Through education, and stories like mine, there is a chance that someone who is suffering from Wilson's will be diagnosed early enough to treat the condition efficiently and effectively before it can to do great harm to anyone else.


Dana Schaumann

Dana Schaumann

Dana just turned 13 and was diagnosed with Wilson's disease in September of 2015.Dana said that she first started to realize something was wrong with her last spring when she started to drool on her homework. Playing the trumpet became very difficult for her, but she just thought she wasn't as good as the other trumpet players at her school. She didn't know at the time that her mouth muscles were weakening and that was the real reason why it became so hard to play. Her mom even signed her up for private lessons through the summer, perplexed as to why she had gone from being a good player to having such difficulty getting a clear sound out of the instrument.

About five months after a gradual onset of symptoms (drooling, slurred speech, anxiety, muscle rigidity in her hands, arms, and legs), she had an MRI that revealed abnormalities in her basal ganglia. A local pediatric neurologist wasn't exactly sure what tests to order from there, but upon mentioning that Dana's aunt has Wilson's disease, the neurologist thought that should be the first testing to happen. When Dana received her diagnosis, she was relieved to know that it wasn't a nameless, mystery illness no one would ever figure out, and she was glad that at least she could still eat Skittles.

Dana's biggest sadness has been having to give up chocolate, and she wishes the empty-stomach window didn't have to be so long for taking her medicine. Her friends have been very supportive and help her with fine-motor tasks as needed. There are some kids who look at her differently who don't know she has a medical problem, but she hasn't felt the need to share her medical difficulties with everyone.She has worked with a speech therapist at school, and her speech is a lot better than it was last summer. After seven months on a waiting list, she has finally been able to start occupational therapy for her hands.

A week after she was diagnosed, they held the first Wilson's Walk in Chicago, and her family signed up to go. While talking to all the people there, Julie and Steve's dad Doug convinced the family to take her to the University of Michigan. Dr. Askari there has been a tremendous help with her treatment.

To learn more about Wilson's Disease, please visit http://www.wilsonsdisease.org/about-wilsondisease.php

THANK YOU for taking the time to read our stories! Please help spread the word about Wilson's Disease, and if you are not already walking in the BIG WOW, please consider signing up for this October 1st event!