

Fighting a rare disease: inside Neurofibromatosis

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“Incurable disease.” Those two words devastated Emily Boyas, LZHS reading specialist, when her daughter, Madelynn, was first diagnosed with a life-changing disease at a year old.

Madelynn suffers from Neurofibromatosis (NF), an incurable disease that comes in various forms and attacks the nervous system, according to Boyas. Luckily, Madelynn has the less intense of the two types, NF1.



“Neurofibromatosis can cause blindness, stunted growth, a lot of pain, and scoliosis,” Boyas said. “There are brown spots, called ‘café au lait spots,’ which Madelynn has all over her face, body, and even her brain.”

Madelynn, who is five years old, sees a developmental therapist, a physical therapist, and a speech therapist for her disabilities. Once a year, she also sees a neurologist for the tumor on her brain. She has recently started seeing the NF clinic at University of Illinois Chicago for a growth on her face.

“She was born with the NF,” Boyas said. Although she was born with the disease, Madelynn was not diagnosed until a year later. “We do not know if it is inherited or a mutation, but neither my husband or I show symptoms of the disease.”

Boyas said Madelynn’s diagnosis had more of an effect on the family when she was first diagnosed because of their emotional hardships and because they constantly had therapists in the house.

“While we’re able to cope with the learning disabilities, the development delays, and the physical delays, not knowing what’s going to happen is the hardest thing for our family. The disease is incurable and the tumor in Madelynn’s brain is not removable, so we just watch it and hope it doesn’t grow,” Boyas said. “She is now five, so she is becoming aware of the spots on her body. She is starting to ask why she has these spots and why me and my husband don’t, so it’s difficult for us to explain to her that she’s just special.”

Boyas knew something was wrong with Madelynn when she was born because of the 'café au lait spots' all over her body, a clear NF indicator. Even though these spots are an indicator of NF, they do not cause any medical concern, according to The NF Center at Washington University of Medicine at St. Louis. When Madelynn was about eight weeks old, her doctor sent them to a neurologist.

"I had no hope. I was devastated because everyone wants their first child to be healthy," Boyas said. "I felt guilty that I had done something wrong or hadn't eaten the right foods [when I was pregnant] and I blamed myself. We decided to follow what the doctor said and have a positive outlook. Madelynn has challenges, but not as bad as some."

Even though Madelynn has disabilities that make it harder for her to perform simple, everyday tasks like getting dressed, she is still a happy kid who works through her challenges says Boyas.

To help cope with Madelynn's disease, Boyas and her husband participate in a walk every year in order to raise money for a cure for NF through the organization NF Midwest.

"The walk is in Naperville every year and some families raise a ton of money. It brings awareness to the disease that most people don't know about," Boyas said. "Families from all around get together, and it helps to see that you're not alone."

Boyas also tries to raise money for NF research by getting involved in fundraisers, like Scentsy. Scentsy specializes in flameless candle products, according to the Scentsy website. Boyas sells these products on her own Scentsy page as a consultant.

Along with Scentsy, Boyas sells Mary Kay products and used to sell candy bars to benefit NF research. Her most successful attempt to raise money for NF research so far has been through a fundraising website called *First Giving*.

"We have raised over \$1000 on Madelynn's *First Giving* page," Boyas said. "[People can help raise money] by sharing the First Giving page on their Facebooks, or liking the awareness page on Facebook to spread the word about Madelynn."

Even through her disease, Boyas says Madelynn is still a happy and crazy five-year-old girl.

"She does deal with focus issues because of her disease, but she's happy and she is not defiant," Boyas said. "She loves everything about life and she's always smiling. She gets frustrated sometimes when things are hard for her, but for the most part she's just a happy kid."