

FIRST PERSON //

## Striking Unlucky

■ BY LAUREN WARE

44



**D**on't worry too much about these tests," the pediatric gastroenterologist reassured us. "If he has any of these conditions, it would be like getting struck by lightning twice." We all laughed nervously.

My husband and I sat quietly as she checked off boxes on the form for blood work. Our 15-month-old son, Jake, bounced around the office, crinkling the paper on the examination table, wiping his hands on the floor.

A year earlier, Jake had stopped growing and began suffering from severe digestive problems. I had watched helplessly as he became lethargic and seemed to be in constant pain. When his pediatrician told us that Jake's sweat test for cystic fibrosis was positive, I was devastated. Matt and I walked out to the car in slow motion. I buckled Jake into his seat, sat down in the front passenger seat and started bawling. It was like winning the lottery in reverse: I never thought something that horrible would happen to a child of mine.

After several months on a digestive enzyme replacement, Jake was not rallying the way his medical team had expected. Hovering over me like a brewing storm was the feeling that something was terribly wrong.

Then lightning struck again. The pediatric gastroenterologist told us that Jake has Alpha-1 antitrypsin deficiency, a rare lung and liver disease caused by an autosomal recessive mutation like CF. She explained that there is no evidence the two mutations are inherited together, that it was sheer chance Matt and I were carriers for both disorders. One in 30 people in the United States walks around silently carrying one of the mutations that cause CF, and one in 25 people carries the mutation that causes Alpha-1. The likelihood that a child would have CF *and* Alpha-1? One in 7.5 million. Yet because we are his parents, Jake's chances were narrowed to 1 in 16.

The absurdity of the situation left me numb. Alpha-1 typically causes liver disease and lung damage, which people with CF already face. We found specialists experienced with both disorders, but they didn't know how the Alpha-1 would interact with the CF, except to say: "We can't imagine it will be good."

But whenever I felt my heart pierced by the tragedy, I only had to look at Jake to be renewed. He was—and is—an extraordinarily happy, peaceful little guy. When he stayed happy-go-lucky but didn't start talking by two years old, our extended circle of family and friends brushed it off. Look at all he's been through, they said. He just needs time to catch up. But by the time Jake turned three, that familiar feeling of dread returned. Then the third lightning strike came: autism.

Maybe having been through it twice already helped me accept this new development. Or perhaps I knew autism was just a name for who Jake had been all along. Happy, but on his own terms. Blissful, but in his own universe. He wanders among the flowers and grasses, not worried about another lightning strike. Maybe the bright side of his random bad luck is that he doesn't understand it, doesn't question why it happened. And maybe he's here to teach me about randomness in my life. ■

→ Post-Op originates at the other end of the stethoscope, presenting essays and commentary from patients, consumers and other medical outsiders. *Proto* invites your contributions; please send ideas to [protoeditor@mg.harvard.edu](mailto:protoeditor@mg.harvard.edu).

AD