My husband and I were thrilled when our son, Andy, was born in June 1989. Our delight soon turned to concern, though, when we noticed that something was different. Andy couldn't nurse despite repeated attempts. He cried nonstop, and he had poor muscle tone. We consulted several doctors, but none could identify any underlying health problems. When Andy was 2, our second child, Laura, was born. As she reached all the expected milestones of infancy, Andy's differences were even more pronounced.

In addition to his physical problems, Andy exhibited extreme anxiety. He would panic when my husband or I put him down and walked out of his view for even a few seconds. I
remember him at age 3 screaming, flapping his hands, and stomping his feet. He seemed unable to tolerate being in his own skin.

Convinced that something was seriously wrong, we met with more doctors, including a developmental pediatrician who conducted genetic testing and delivered a diagnosis: Andy had fragile X syndrome, a genetic condition characterized by intellectual disability and learning and behavioral problems, and sometimes by seizures. This rare disease is caused by mutations in the FMR1 gene on the X chromosome that interfere with production of a protein involved in normal brain development.

Now 31, Andy can dress and feed himself and understands most of what we say to him, but he has trouble communicating and can string together only about seven or eight words. He's also very shy. We usually do not have guests at our house, but if he really likes someone, he will spend all his time with him or her.

He still lives at home with us in Newburyport, MA, where he attended a day program before the pandemic. Called Shared Living Collaborative, it has a farm where Andy would take care of horses, donkeys, and other animals. For a while he enjoyed riding horses, but in the past few years he's preferred caring for and training them. And he's particularly attached to one of the donkeys.

About four years after Andy was born, we met another parent who had a child with fragile X syndrome, and together we established the FRAXA Research Foundation. We began by reaching out to the scientific community to develop an advisory board, which now has 16 members, including two Nobel laureates. When we started, there was no such thing as social media, so we reached people through word of mouth and by attending meetings for families affected by the disease. Since 1994 we have raised $27 million and funded biomedical research grants, chosen through a peer-review process led by our scientific director.

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We haven’t found a cure yet, but we’ve moved closer to understanding fragile X syndrome and identifying treatment targets. We hope this will help our son and other families dealing with this life-changing condition. —As told to Paul Wynn