Daniel Barvin always wanted to be a parent. "It was just something that was in me," says Daniel, 35, of Houston, Texas. "My wife and I met when we were quite young, and five years in I was like, 'OK, let's start having kids.' We weren't even married yet, but I just knew I wanted to be a father."

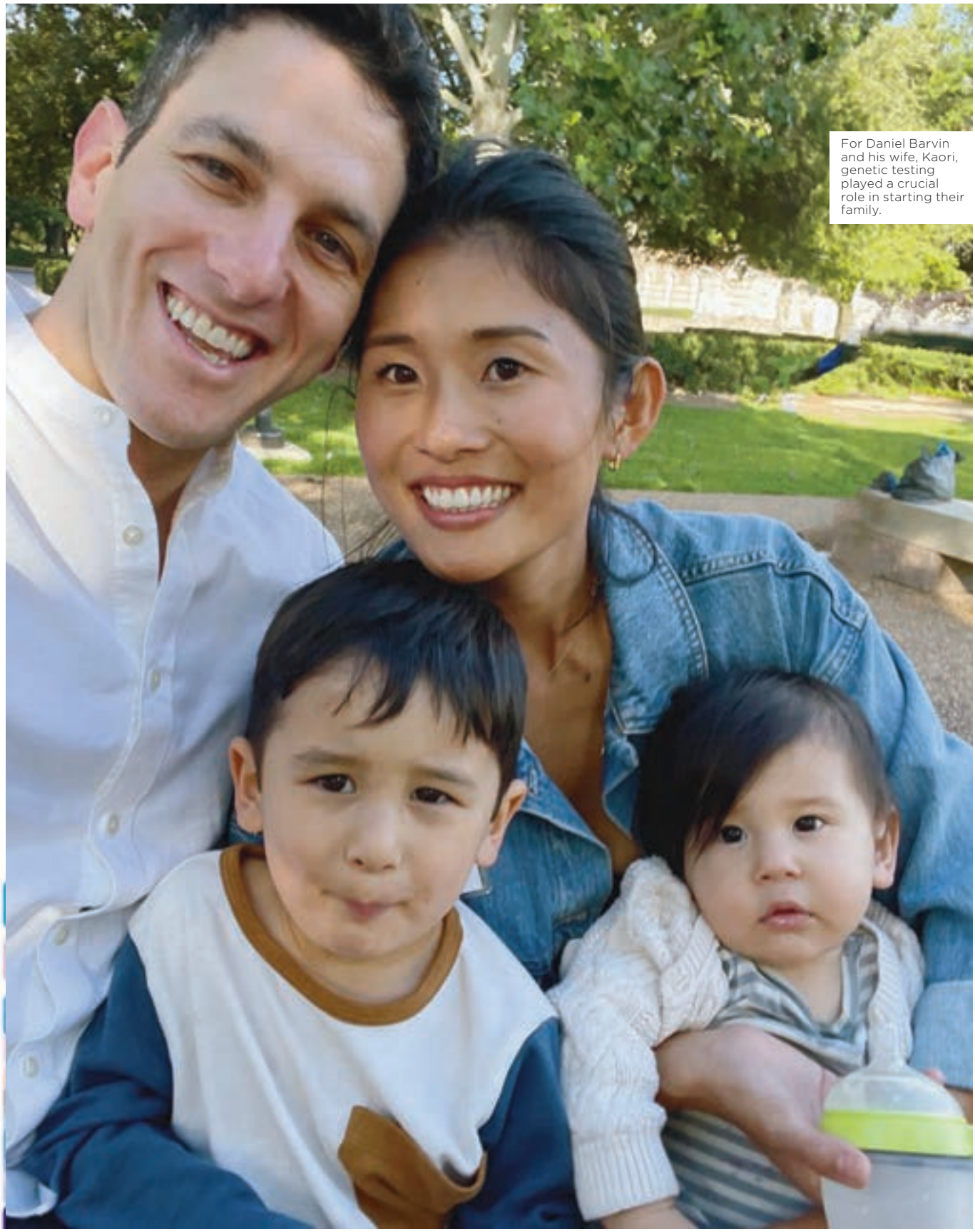
But fatherhood wasn't the only thing that was "in" Daniel. He also carries the *C9orf72* gene mutation that can lead to familial amyotrophic lateral sclerosis (ALS), frontotemporal dementia (FTD), or both.

A FAMILY AFFAIR

Genetic testing can help current and aspiring parents

BY MATT ALDERTON

Image: iStock.com/natrot



For Daniel Barvin and his wife, Kaori, genetic testing played a crucial role in starting their family.

FREQUENTLY ASKED QUESTIONS

My doctor gave me a diagnosis. Why should I get a genetic test?
In many cases, genetic testing can confirm and improve the accuracy of a clinical diagnosis by pinpointing an individual's disease-causing mutation. This can lead to eligibility for certain therapies or medications and clinical trials, as well as a better understanding of how the disease will progress. This can also facilitate accurate genetic testing for affected or unaffected members.

Can't I order a genetic testing kit on the internet?
There are many DNA testing kits on the market that claim to reveal your ancestry or provide health insights. But over-the-counter genetic testing kits are not medical tests, can't match the specificity of the tests ordered in a clinical setting, and don't offer the expert interpretation and counseling your care team delivers with test results.

I already had a genetic test. Do I need another?
Genetic testing is advancing quickly. If your previous genetic test was negative or inconclusive, keep asking about new testing. Anyone considering a gene therapy or participating in a clinical trial also should ask if their testing needs to be repeated.

Is genetic testing covered by health insurance?
Many health insurance plans will cover genetic testing and carrier screening when it is ordered by a physician. Check with your plan for coverage and reimbursement details.

If I have a diagnosis, should my family be tested?
Talk with your genetic counselor about who in your family should be tested based on the type of disease and who else is showing symptoms. Relatives may be interested in genetic testing to find out if they are at risk of developing symptoms later in life or if they're carriers. Carriers have a chance of having children with the same genetic disease.

In people with FTD, damage to the brain's temporal and frontal lobes can cause dramatic changes in language, personality, and behavior. Approximately 15% of people with FTD also develop ALS.

Although doctors didn't recognize it at the time, Daniel's grandfather died of FTD when Daniel was 8 years old. Later, his father developed early-onset dementia and an uncle and an aunt died of ALS.

"It was brutal," recalls Daniel, who says doctors never raised the specter of heredity. "This was before genetic testing was prevalent, so every doctor told us these illnesses were sporadic and we had nothing to worry about."

When Daniel's father died following a dementia episode in 2016, his family ordered an autopsy and discovered he had the *C9orf72* gene mutation. Suddenly, Daniel's tragic family history made a lot more sense. From that clarity, however, came an unforeseen crisis of conscience: Knowing the gene was in his DNA, Daniel asked himself whether he could still become a father — and whether he should.

He isn't alone. Daniel is among millions of aspiring and current parents whose genes could predispose their offspring to hereditary neuromuscular conditions. However, unlike the generation before them, they don't have to wonder helplessly about the future. Thanks to genetic testing and family planning, they can understand risks, prepare for them, and take action if they choose to.

What is genetic testing?

Genes tell your cells how to grow and work. When you have genetic testing, medical professionals examine your genes for changes — called mutations or variants — that may cause diseases.

"Persons with a wide variety of symptoms could have those symptoms as the result of an underlying genetic change, and identifying and documenting that change increasingly is the way that we make a diagnosis," explains Jennifer Roggenbuck, a licensed genetic counselor at

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The Ohio State University College of Medicine, where she specializes in neuromuscular diseases.

Although the underlying science is similar, the clinical genetic tests one gets in a medical setting are different from the commercial genetic tests one might order from the internet to research their ancestry or their predisposition to baldness. "Those tests use a different technology," continues Jennifer, who says commercial tests examine fewer gene variants with less rigor. "With medical genetic testing, there's an element of clinical expertise to ordering the genetic test and interpreting the results that's completely different than what you might get from direct-to-consumer genetic testing."

And yet, medical and commercial tests have one thing in common: They're non-invasive. "Sometimes we need blood samples and sometimes we need saliva samples. Often, we're just using a very simple cheek swab," says Kristin Engelstad, a certified genetic counselor in neurology at Columbia University.

Proactive parenthood

Genetic testing can be diagnostic or predictive — it can help you understand the cause of symptoms you have or your risk for developing a genetic disease when you don't have symptoms. In both cases, the results of a genetic test may qualify you for life-enhancing and life-saving therapies, medications, and clinical trials.

That's what initially motivated Daniel to seek genetic testing in 2018: He wanted to know if he carried the same genetic mutation for ALS and FTD that had affected his relatives, and what he could do about it if he did.

"I found out that there is a 95% chance that I will develop the disease," Daniel says. "I had prepared myself for that news, and I approached it with a mindset of action. The generation before me had no idea this was coming. They were blindsided by the disease, but I have the ability to prepare for it."

The ability to prepare was especially important to him as he pondered parenthood. "When my wife and I eventually met with a genetic counselor, the key takeaway was that we could do family planning, which we had never heard about," continues Daniel, who learned that he has a 50% chance of passing down the *C9orf72* gene mutation to a child. "That became a torch that I carried forward. This had decimated my father's generation, and who knows what it will do to mine. But I decided to do everything I could to save the next generation."

"The generation before me had no idea this was coming. They were blindsided by the disease, but I have the ability to prepare for it."
— Daniel Barvin

Any prospective parent — regardless of family history — may want to consider carrier screening, according to Ellen Moran, a certified genetic counselor at the Center for Children at NYU Langone's Hassenfeld Children's Hospital in New York. Comprehensive carrier screening encompasses broad genetic screening for autosomal recessive disorders like spinal muscular atrophy (SMA), X-linked disorders like Duchenne muscular dystrophy (DMD), and autosomal dominant disorders like forms of limb-girdle muscular dystrophy (LGMD). With carrier screening, future parents may identify potential risk factors.

Armed with this information, couples can decide whether to pursue family planning options, which may include preimplantation genetic testing (PGT) with in vitro fertilization (IVF) or using donor eggs or sperm.

With PGT and IVF, eggs are retrieved from the mother and fertilized in a laboratory with sperm from the father. Fertilized eggs can be tested for dominant and recessive genetic mutations, and healthy embryos can be implanted in the mother.

When using donor eggs and sperm, Kristin emphasizes the importance of working with reproductive centers that routinely conduct carrier screening. Genetic counselors might be able to refer couples to reproductive centers that understand and appreciate genetics.

"There are a lot of options. It just depends on what people are comfortable with, what their insurance will cover, and what they can afford out of pocket," Kristin says.

Daniel and his wife opted for PGT and IVF and now have two children, a 1-year-old daughter and a 3-year-old son — neither of whom carry the *C9orf72* gene mutation.

"I'm in a much better position than the previous generation to take steps that protect my children's future," says Daniel, whose personal experience inspired him to start a career at Coya Therapeutics, a biotechnology company developing therapeutics for ALS, and to establish a nonprofit called End The Legacy: Genetic ALS and FTD. "My kids don't have this gene, but I'm going to be incredibly proud to tell them the story of their ancestors and the proactive stance we've taken against ALS and FTD."

AFFORDABLE GENETIC TESTING

As technology advances, genetic testing is becoming more affordable. In addition, some sponsored programs offer genetic testing or carrier screening for specific neuromuscular conditions at no cost. Generally, your doctor will need to verify your eligibility and order the test.

To learn about sponsored genetic testing programs and other resources, contact MDA's Resource Center at **833-ASK-MDA1** or **ResourceCenter@mdaUSA.org**.

Genetic testing for children

In cases where there is a known risk of genetic disease after conception, parents have options to be prepared to welcome their baby.

"We can check the genetic status of the fetus with a variety of tests," notes Jennifer, who says the most common tests are chorionic villus sampling, which involves genetic testing of the placenta beginning at 12 weeks of pregnancy, and amniocentesis, which involves genetic testing of the amniotic fluid around the fetus starting at 15 weeks of pregnancy.

Parents who learn of a neuromuscular disease in their family after having children should consult with a genetic counselor to find out if their children should be tested.

For children with no symptoms, genetic testing typically can wait. "We do not recommend genetic evaluation for children who are currently healthy," says Jennifer, who points out that genetic testing in childhood can have personal and

ONLINE EXCLUSIVE

KNOW BEFORE YOU TEST

Find out what you need to know about your rights regarding your genetic status and how it could impact you in "Can Genetic Testing Make You Uninsurable?" at **MDAQuest.org/insurable**.

financial implications in adulthood when individuals apply for insurance, start relationships, etc. "We want to respect the child's autonomy. So, unless there's a medical reason to do genetic testing early, we usually say the child can make that decision when they're 18 or older," she says.

Families of children showing symptoms may want to pursue testing. "Genetic testing could help you get an accurate diagnosis," says Ellen, who explains a genetic diagnosis is necessary to provide accurate genetic counseling to the family and determine eligibility for clinical trials and treatments, including disease-modifying therapies that tend to be more effective when they're given early. "The sooner you start, the better the prognosis is for treatment."

When it comes to getting a diagnosis, parents are their children's best advocates. "If you have a child with a mysterious medical condition that your physician doesn't understand — particularly one that's lasted a long time and was present from an early age — it might be worth asking, 'Could my child benefit from genetic testing?'" Jennifer says. "Don't wait for your doctor or pediatrician to bring it up because it may not be at the forefront of his or her mind."

Where to start

Individuals seeking genetic testing for family planning should see a genetic counselor. Often, the first step is getting a referral from your primary care physician or neurologist. Alternatively, you may be able to make an appointment directly with a genetic counselor. (Use the National Society of Genetic Counselors "Find a Genetic Counselor" tool at **FindaGeneticCounselor.nsgc.org**).

"Ideally, you want to see a genetic counselor who specializes in neuromuscular disorders," advises Jennifer, who says demand for specialists outstrips supply. "Fortunately, many genetic counselors offer telemedicine appointments as long as you're in the same state."

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It's helpful to understand what a genetic counselor does, according to Jennifer, who says there's more to genetic testing than the actual test. Genetic counselors also help patients determine what kind of genetic test to get, what the implications of testing might be, and what actions they can take in response to their test results.

"Genetic counselors can help you review the medical landscape surrounding genetic testing, as well as the psychological, emotional, and family issues that come into play with these types of decisions," Jennifer explains. "We're patient-focused, so we'll never tell you what to do. Our role is to provide risk assessment, education, and support to individuals as they determine what steps are right for them at a particular time in their lives."

Whether your motivation is your own health or that of your future children, genetic testing can be fraught but fruitful.

"It can be a scary process, but we try to impress upon families that they're not in this alone," Ellen concludes. "There's a whole team of people there to work with you as you go through this." Q

Matt Alderton is a Chicago-based freelance writer who frequently covers health topics.