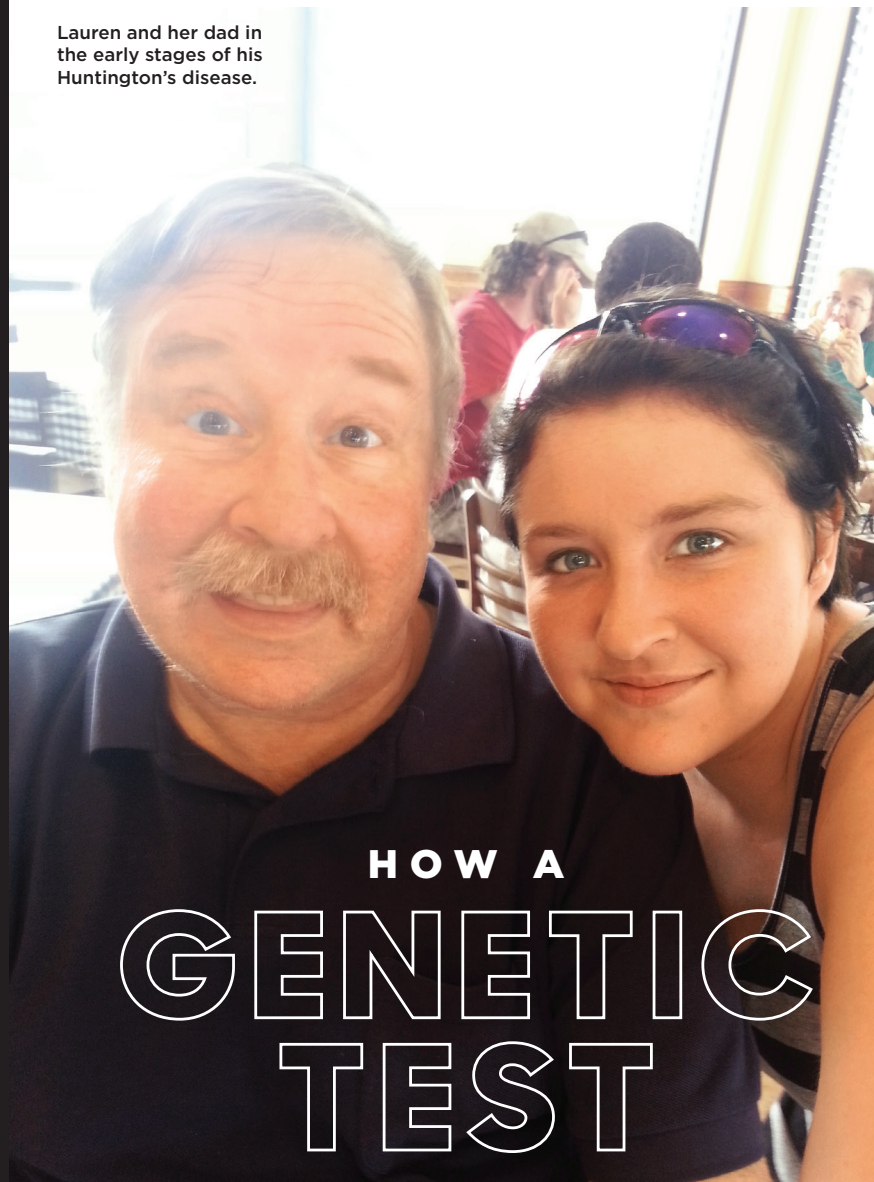


Lauren and her dad in the early stages of his Huntington's disease.



HOW A GENETIC TEST

CHANGED MY LIFE



As a teen, **Lauren Holder** learned that a cruel and fatal disease was stalking her family—and that she might have inherited the gene for it from her dad. Now 30, she tells *GH* about her journey to discover the truth

as told to **VIRGINIA SOLE-SMITH**

When I was 5, my grandpa Rose took my dad and me out on his boat off the coast of Florida, where we live. It was a hot, bright day, and as the boat skimmed over the water, I asked my dad to put up the sunshade so we could cool off. It was a small request, but Grandpa Rose exploded in rage: “No! We’re not doing that!” Then he began to curse. I was shocked, but my dad wasn’t. As I would soon find out, violent outbursts were the norm for Grandpa Rose and had been for some time. His reputation as a cruel abuser was no secret in our family.

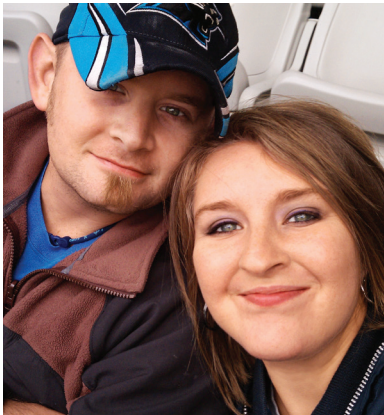
And then, 10 years later, Grandpa Rose became someone else entirely to me. That was the day my aunt told me he had Huntington’s disease (HD).

FEAR SETS IN

Until I was 15, I had never heard of HD, a neurodegenerative disorder that is like Alzheimer’s disease, Parkinson’s disease, bipolar disorder and Tourette’s syndrome rolled into one. But my aunt Amy told me what it was and explained that all of Grandpa Rose’s behavior—his temper, slurred speech (which we had wrongly thought indicated a drinking problem) and recent difficulty swallowing—had been symptoms. The disease would worsen, Amy said; near the end, he would be trapped inside a stiff body, unable to eat, talk or move. The scariest part: HD is inherited. There was a 50/50 chance my father had it, and if he did, I faced the same odds.

Upon hearing the diagnosis, my grandpa, in total denial, jumped in his car, drove across the country and never came back. My family was actually relieved—his being so far away made it easier to avoid thinking about the situation. My dad didn’t seem to take the news seriously. “Just chain me up in the backyard with a drink, and I’ll be fine!” he joked about the possibility of developing HD, but inside, he must have been terrified. My grandpa was the meanest person I knew. Now there was a chance that my dad could turn into that kind of monster...and that I could, too.

Then, around the time I turned 18, I was chatting with my dad in our kitchen when he suddenly started screaming in anger and squeezing my arm hard enough to leave bruises. We didn’t talk about it, but I know we were worried about the same thing: He might be getting HD, too. →



MAKING IT COUNT Lauren at a football game with husband Josh in 2014 (*left*) and reconnecting with her grandpa Rose before he died in 2007.



This is it. It was also difficult to deal with my parents: Though my dad’s angry outbursts continued, neither of them had accepted that HD was the cause. Talking to my mom about it was like talking to the air.

Still, I was determined that my test results should help someone. I knew my grandpa Rose, then 73, was in a nursing home in Seattle, so I flew out to see him. At first glance, I couldn’t tell if he was alive; he’d lost so much weight, and his body had become stiff. That night, I cried myself to sleep.

But the next day I forced myself to visit again, bringing family photos. Though he hadn’t been out of bed in six months, I asked him, “Do you want to get up and sit with me?” It took a few minutes for him to get a word out, but when he did, he said yes, and so we sat and looked through the pictures. When it was time to leave, I said, “I love you.” For the first time in my life, Grandpa Rose told me, “I love you, too.”

When I got home, I sent my dad pictures of the trip, and days later he and his sisters flew to see their father. Grandpa Rose died just months later. I’m forever grateful that we all made peace and said goodbye.

READY TO LIVE

It’s been 10 years since I got my results, and I’m still presymptomatic. My doctors think that will change when I’m in my mid-40s.

There have been times when I’ve tried to push Josh away, to protect him. But he always says, “We’re in this together.” Still, I know he wishes we could have kids as badly as I do. But I don’t want to pass this gene to a child, nor would I want a child to have to take care of me at a young age.

But whenever I think *Poor me*, I remember my dad. Three years ago, he said, “Lauren, I looked up Huntington’s disease. I think I have it.” I realized that it had taken me facing my truth for my dad to accept his. Soon afterward, he joined me in volunteering with the Huntington’s Disease Society of America.

At 57, my dad has little short-term memory; by noon, he can’t tell you what he ate for breakfast. He walks slowly, has poor balance, suffers from mood swings and has other health problems, like complications from diabetes, that disorient him. But there are good moments, too, when I know he’s still Dad. If my mom gets flustered, he’ll tease her by saying, “Oh, no—you caught my Huntington’s!” And we laugh, because no one is living in denial anymore. We’re all determined to get as much as we can out of life. We are living with Huntington’s disease, but that doesn’t mean we can’t live.

THE 411 ON HD

HUNTINGTON’S DISEASE is an incurable inherited brain disorder that results in the progressive loss of mental faculties and physical control. Signs usually appear between ages 30 and 50 and worsen slowly over the next 10 to 25 years.

WHO’S AT RISK?

You can develop the disease only if one of your parents has the mutated HD gene and you inherit it. It’s estimated that one in 10,000 Americans has HD, and more than 250,000 others are at risk of having inherited the gene.

IS THERE HOPE?

Research has yet to find a means of slowing the deadly progression of HD, but last year scientists launched the first clinical trials to study the safety of a drug designed to silence the gene. For more information, visit hdsa.org.

LEARNING MY FATE

As soon as Grandpa Rose was diagnosed, when I was still in high school, I flung myself into researching HD. One of the first things I learned was that the same blood test that had found HD in him could tell me whether I would get it. When my dad began showing symptoms a few years later, he wasn’t interested in the test for himself, but I decided to get tested as soon as possible.

The genetic counselor I found wasn’t as supportive as I had hoped. “If you have the genetic mutation for Huntington’s, there’s nothing we can do,” she told me. “There is no medical benefit [to knowing].” I understood, but I also knew that without the test result, I would obsess over whether, when and how I might get sick. Testing would put the biggest piece of the puzzle in place.

After the blood draw, the wait for the results felt endless. I kept asking myself the same questions: What would a positive result mean for me and my dad? Would I hurt those I loved like Grandpa Rose had? Would my husband, Josh, whom I’d wed at 19, leave me?

When the day finally arrived, the counselor didn’t make us wait. As we walked into her office, she said it: “You tested positive. You are going to get Huntington’s disease.”

A NEW REALITY

When I heard the news, I burst into tears—of sadness, but also of relief, because I wasn’t wondering anymore. But as we began to talk, fear settled in. It’s surreal to have someone tell you how you’re going to die.

Adjusting to my new reality over the next few days, weeks and months wasn’t easy. I developed insomnia, had anxiety attacks and got very depressed. Any time I got mad or acted clumsy, I would freak out and think,

GENETIC TESTING

Depending on your family history, your DNA could tell you *a lot* about your health. Intrigued? Start here *by* VIRGINIA SOLE-SMITH

GENETIC TESTING isn’t only relevant to people who have a disease like Huntington’s in their families. “I think in the not-too-distant future, a person’s genome sequence will be a part of his or her health story. We’ll refer to it whenever a health question presents itself,” says Lisa Edelmann, Ph.D., an associate professor of genetics and genomic sciences at Mount Sinai’s Icahn School of Medicine in New York City.



Thanks to major strides in genetic technology, scientists have tied more than 2,000 genes to specific diseases, and there are over 900 tests available to patients. But the science isn’t perfect yet. Today, testing walks a fine line between lifesaving and life-complicating: Clear results could bring relief or help you make a change, but inconclusive ones may take an emotional toll or require tough choices. Below, find out what testing can and can’t answer.

AM I LIKELY TO GET A SERIOUS ILLNESS?

FIND OUT WITH:
PREDICTIVE SCREENING

These tests can be done on adults of any age to assess whether they have a predisposition to certain conditions, including breast cancer, heart disease and Huntington’s disease.

BENEFITS: Positive results often motivate people to take preventive measures like exercising, eating better, undergoing screenings such as breast MRIs and more. With unavoidable conditions like HD, testing can help you spot early symptoms and reexamine your life goals.

TESTER BEWARE: Inconclusive results—that is, you show some susceptibility to a condition, but a geneticist can’t calculate your exact risk—are common. And even definitive testing can’t tell you when a condition will set in or how severe it will be, says Joy Larsen Haidle, of the National Society of Genetic Counselors. Before testing, “Ask yourself, *Will I use this information, or will it only make me anxious?*”

WILL I PASS A “BAD” GENE TO MY BABY?

FIND OUT WITH:
CARRIER SCREENING

Either before a couple conceives or during early pregnancy, this testing can be done to calculate a mother and father’s odds of passing disorders like sickle cell anemia and cystic fibrosis to their offspring.

BENEFITS: If you’re considering becoming pregnant, you’ll likely feel reassured: Only about 1% of couples who go through carrier screening will turn out to be at risk of passing on a disease. Couples in this group can consider preimplantation genetic diagnosis (PGD), which is done in conjunction with in vitro fertilization to weed out embryos with a condition. A prenatal diagnosis can also help doctors plan treatments, like surgery for a newborn with a genetic disorder that causes congenital heart defects.

TESTER BEWARE: Depending on the results, couples who wait until pregnancy for testing may have to make difficult decisions. Plus, “a negative result doesn’t mean you have zero risk,” says Edelmann. “Everyone has some degree of ‘residual risk.’”

IS THERE MORE TO THESE SYMPTOMS THAN IT SEEMS?

FIND OUT WITH:
DIAGNOSTIC TESTING

This is used when a patient displays mysterious symptoms or a child has severe developmental delays.

BENEFITS: “These patients and their families have often been on diagnostic odysseys trying to understand what’s wrong,” says Haidle. Finding the genetic cause of their condition can end that quest and tell parents how likely they are to have another child with the same problem.

TESTER BEWARE: As with predictive screening, inconclusive results can be common. If initial tests come back negative or inconclusive, your genetic counselor may suggest exome sequencing—a technique for uncovering all of the protein-coding genes in a genome. “Since we’re looking so broadly, you need to be prepared for what else we might find,” says Edelmann. She’s had patients do it to help with an undiagnosed childhood disease only to learn about an increased risk of cancer.

WHO KNEW?



YOUR GENES COULD HELP YOU FIND A PAINKILLER. A new type of test called a pharmacogenetics (PGx) screen uses DNA to determine whether a patient is a good match for certain medications (including codeine, oxycodone and Plavix, a medication that prevents blood clots). In the next few years, this test should help doctors pinpoint the ideal usage and dosage for around 200 drugs.

THE GENETIC-TESTING PROCESS

WHAT TO EXPECT

STEP 1: First, you'll meet with a genetic counselor (click on Genetic Counselor Search at nsgc.org to find one) to discuss which tests might be a fit.

STEP 2: Your blood or saliva will be drawn; results take one to several weeks.

STEP 3: You'll likely get results in person.

Bring a spouse or friend for support and as a second set of ears.

STEP 4: You'll process the news. Depending on the conditions tested for and the complexity of the results, you may feel relieved, shocked, scared or something else entirely. Your counselor can refer you to support groups or a therapist as needed.



From Our
Health Lab

TRIED + TESTED

DNA KITS

Online services decode your genes to reveal your ancestry. The GH Institute vetted three

Not interested in health information? There are personal genomics companies that investigate ancestry. All you have to do to get results is send in a sample of saliva. Whether you're adopted or merely have a murky sense of your roots, the results can be eye-opening.

**GH'S WINNER: 23ANDME**

The GH Institute parsed more than a dozen options and had one local tester—Jennifer, 30—try the three most robust services. All three kits gave an accurate (and similar) picture of her origins and

ethnicity, the results corresponding with her extensive family knowledge. But 23andMe presented the information in the most digestible format (via an interactive color-coded map). The other services were harder to navigate and, at times, glitchy. Another bonus: 23andMe has more than 1 million active users, which increases your odds of finding a long-lost relative. It's also the only kit that provides health information: Thanks to recent FDA approval, 23andMe now delivers carrier screening for 36 gene variants along with a detailed results explanation that could be similar to what a genetic counselor would provide. Established clients who used the service before this new feature can receive carrier results at no extra charge. **\$199, 23andme.com**

IMPORTANT SAFETY INFORMATION (Continued)

Do not take BOTOX® (onabotulinumtoxinA) if you: are allergic to any of the ingredients in BOTOX® (see Medication Guide for ingredients); had an allergic reaction to any other botulinum toxin product such as *Myobloc®* (rimabotulinumtoxinB), *Dysport®* (abobotulinumtoxinA), or *Xeomin®* (incobotulinumtoxinA); have a skin infection at the planned injection site.

The dose of BOTOX® is not the same as, or comparable to, another botulinum toxin product.

Serious and/or immediate allergic reactions have been reported. These reactions include itching, rash, red itchy welts, wheezing, asthma symptoms, or dizziness or feeling faint. Tell your doctor or get medical help right away if you experience any such symptoms; further injection of BOTOX® should be discontinued.

Tell your doctor about all your muscle or nerve conditions such as amyotrophic lateral sclerosis (ALS or Lou Gehrig's disease), myasthenia gravis, or Lambert-Eaton syndrome, as you may be at increased risk of serious side effects including severe dysphagia (difficulty swallowing) and respiratory compromise (difficulty breathing) from typical doses of BOTOX®.

Tell your doctor about all your medical conditions, including if you: have or have had bleeding problems; have plans to have surgery; had surgery on your face; weakness of forehead muscles, such as trouble raising your eyebrows; drooping eyelids; any other abnormal facial change; are pregnant or plan to become pregnant (it is not known if BOTOX® can harm your unborn baby); are breastfeeding or plan to breastfeed (it is not known if BOTOX® passes into breast milk).

Tell your doctor about all the medicines you take, including prescription and non-prescription medicines, vitamins, and herbal products. Using BOTOX® with certain other medicines may cause serious side effects. **Do not start any new medicines until you have told your doctor that you have received BOTOX® in the past.**

Especially tell your doctor if you: have received any other botulinum toxin product in the last 4 months; have received injections of botulinum toxin such as *Myobloc®*, *Dysport®*, or *Xeomin®* in the past (be sure your doctor knows exactly which product you received); have recently received an antibiotic by injection; take muscle relaxants; take an allergy or cold medicine; take a sleep medicine; take anti-platelets (aspirin-like products) or anti-coagulants (blood thinners).

Other side effects of BOTOX® include: dry mouth, discomfort or pain at the injection site, tiredness, headache, neck pain, and eye problems: double vision, blurred vision, decreased eyesight, drooping eyelids, swelling of your eyelids, and dry eyes.

For more information refer to the Medication Guide or talk with your doctor.

You are encouraged to report negative side effects of prescription drugs to the FDA. Visit www.fda.gov/medwatch or call 1-800-FDA-1088.

Please refer to full Medication Guide including Boxed Warning on the following pages.

BOTOX®
onabotulinumtoxinA injection