

Genetic Testing

We are uncovering the genetic basis of many neurological disorders.

I vividly remember the first patient for whom I ordered a genetic test for Huntington's disease. As you will learn later in this issue, Huntington's is a genetic disease that carries a 50:50 chance of inheriting it from a parent who has the disorder.

My patient was a man in his late fifties. He had all the symptoms. About 10 years earlier he began experiencing uncontrollable movements of his arms and legs that forced him to quit his job driving a taxi. Not long after that, his memory started to fail. He became easily confused and quick to anger, and even hit his wife and children on a number of occasions. The movements of his arms and legs became nearly continuous—except while asleep—burning up so many calories that he could barely eat enough to maintain his weight.

The family had seen several doctors but were not comfortable with the Alzheimer's disease diagnosis they were given. I recognized the disease immediately as Huntington's. I explained this to his family, including information about the genetic aspects. I asked about whether anyone else in the family had any symptoms similar to what my patient had developed, but they had no information about illnesses in his family.

I told them that we now had a new blood test that could tell for sure whether or not my diagnosis of Huntington's disease was correct. I also explained that knowing this for certain was important, because if their father had Huntington's, it meant that each of his children would have a 50 percent chance of also developing the disease.

The oldest child in the family was 28 years old. She had a child who was 3, and she was planning to have another one soon. The other siblings were in their early twenties and had not yet started their families. Everyone agreed that their father should have the test. It was positive, confirming the diagnosis of Huntington's disease.

When they came back to see me to get the test results, they wanted to know whether or not the test could tell them if they were going to get the disease. Yes, I told them, a positive test

result would mean that they were going to get Huntington's.

After several weeks of soul searching and lengthy discussions with me and a genetic counselor, the oldest daughter asked to have the test done. She wanted to know before having another child. We had her meet with a psychologist to ensure she really wanted the test—and was prepared to deal with the results. I don't know the outcome of the test, but I do know she had another baby, so I suspect that the result was negative. Her siblings, however, chose not to take the test. I thoroughly respect both decisions.

In this issue of *Neurology Now* we feature Dame Julie Andrews, an incredible performer and an advocate for new therapies and a cure for genetic diseases like Huntington's disease. In our special report on neurogenetic advances, we learn about the efforts of dedicated Huntington's researchers like Nancy Wexler, whose own mother died from the disease, and we report on recent advances in autism, Parkinson's, Alzheimer's, and other neurological diseases with a genetic basis.

As we uncover the genetic basis of these diseases, we have the potential to learn facts that will help us find ways to treat and even cure them. We also have

the potential to make early diagnoses—which, if therapy is available, can slow down or even prevent disease. That, of course, is our hope. Sometimes, before we have the knowledge to treat or cure, the information can at least help prevent the disease in the next generation.

The choice to take a test that will diagnose an as-yet incurable disease that may not become symptomatic for years is a complicated and agonizingly difficult one. I cannot say what I would have done if I had been in the place of my patient's daughter. What would you do?



But the choice to get tested is complicated.

Robin L. Brey, M.D.
Editor-in-Chief