



Testing, Testing

Are “over-the-counter” genetic tests ready for prime time?

Many human characteristics—from height to eye color to a tendency to develop certain diseases—run in families. I’m left-handed, a trait that came from my mother’s side. I have a friend who runs ultra-marathons (100 miles); his son, who shares his lanky build, is an exceptional long-distance runner as well. Our genes are a powerful form of “inheritance” that helps shape who we are. I choose the words “helps shape who we are” intentionally. More about that in a minute.

The Human Genome Project, a 13-year, global collaboration among scientists, was completed in 2003. In the midst of this effort, the United Nations adopted a Universal Declaration of the Human Genome and Human Rights that says, “The human genome underlies the fundamental unity of all members of the human family, as well as the recognition of their inherent dignity and diversity.”

This declaration is especially poignant when you realize that human beings share genes that are much more similar than different. Small differences, sometimes magnified by environmental effects on gene function, are responsible for human diversity.

All of the approximately 25,000 genes in human deoxyribonucleic acid (DNA) have been identified.

And the three billion chemical “base pairs” that form the basic DNA building blocks have been sequenced. Scientists are now working to determine which types or combinations of genes lead to certain diseases and why certain characteristics are passed down.

With 25,000 genes, figuring out which genes are involved in different diseases and traits is complicated. Plus, genes have alternative forms that function differently depending on the sequence of base pairs used to make them. And, depending on its alternative form, one gene can influence (or be influenced by) other genes or environmental factors such as toxins or medications. This means that knowing the names or even all of the alternative forms of all of your genes will not necessarily allow you to understand everything about your health risks.

In this issue of *Neurology Now* we take on the topic of genetic testing. (See “Risky Business,” page 18.) While great strides have been made in understanding the role genes play in disease, we have a long way to go. For one thing, scientists don’t always agree on which genes or combinations of genes put a person at risk for a certain disease. Having said that, there are some important reasons to do

genetic testing in individual circumstances. Some of these reasons include:

- ▶ Finding possible genetic diseases in unborn babies
- ▶ Finding out if people carry a gene for a disease and might pass it on to their children
- ▶ Screening embryos for disease
- ▶ Testing for genetic diseases in adults before they cause symptoms
- ▶ Confirming a diagnosis in a person who has disease symptoms

In some cases, a disease can be prevented, treated, or treated *differently* if a particular form of a gene is found. For example, we know that people with certain genes have a greater sensitivity to the blood-thinning drug warfarin. Knowing which form of the gene a person has can allow their physician to select the correct dose to prevent bleeding complications.

Knowing a person’s genetic make-up can help physicians customize prevention and treatment. Experts believe that so-called “personalized medicine” might be the norm in as few as five to 10 years. My advice now is that you talk with your doctor about whether or not genetic testing can help you. The genetic screening available “over the counter” is not, in my opinion, ready for prime time.

My very best,

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



Knowing a person’s genes can help physicians customize treatment.