



Deborah Raymond is a genetic counselor with expertise in movement disorders.

# Test Taking Tips

## *Genetic Counselors Inform Patients & Families about Testing for Dystonia*

“Every family has its own relationship with dystonia,” says Deborah Raymond, MS, CGC, Genetic Counselor at Mount Sinai Beth Israel. “Genetic counseling is an opportunity for families to learn about dystonia, how it’s inherited, and discuss options for testing—including how testing might or might not benefit the family.”

The genetics of dystonia are complex. A lot has changed in the 20+ years that Raymond has worked in movement disorders: multiple dystonia genes have been discovered, new testing technologies are available, companies have begun offering direct-to-consumer testing without requiring a physician’s prescription, and the potential for confusion among patients and families is greater than ever.

### Decisions, Decisions

A genetic counselor can help dystonia patients and their family members determine whether they are candidates for genetic testing, whether they wish to be tested, and what method of testing might be appropriate. A genetic counselor also helps families understand results from testing.

A gene is a segment of genetic material responsible for a specific life function. A genome is the complete genetic make-up of a living thing. The technologies available for diagnostic genetic testing allow for single-gene testing, testing for mutations in multiple genes at once, and targeting sections of a patient’s genome to identify faulty genes.

“Testing is obviously evolving a lot, and there are a number of genes that we usually focus on first when we see a dystonia patient,” explains Raymond. Examples of dystonia-causing genes include DYT1/TOR1A, DYT6/THAP1, DYT11/SGCE, and many others.

Recommendations for genetic testing depend on a number of factors: the clinician’s findings from the neurological exam, patient history, family history, and sometimes ethnicity. “All these factors can play into the most likely gene that is involved and where testing should start,” says Raymond. “For example, if somebody has childhood onset dystonia and has Ashkenazi Jewish background, there’s more than a 90% chance that it’s DYT1 dystonia, especially if symptoms started in a leg. Now, there is some newly available testing that can be helpful in cases where the person has a constellation of symptoms that includes dystonia, but you can’t quite fit them into any of the known classifications. In that situation it may be appropriate to use a type of testing where we can look at many genes at once.”

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### Issues to Consider Before Testing

Part of the genetic counselor's role is to help patients and families think through all the implications of testing, some of which may not be obvious. For example:

- **Genetic testing may influence treatment options.** Research suggests that patients who are positive for certain dystonia genes, including DYT1/TOR1 and the newly discovered DYT28/KMT2B, seem to respond especially well to deep brain stimulation surgery.
- **There are different rules for testing adults and children.** "A lot of thought has been given to testing as it applies to children," says Raymond. "If a child has symptoms, the genetic information could be helpful medically. However, there is consensus that it's not recommended for parents to have an asymptomatic child tested, because that child should be able to make that choice for themselves when they are of age."
- **Genetic testing can be a psychologically and emotionally sensitive issue.** Genetic counselors are not licensed clinical psychologists, but they are trained in counseling. Raymond explains: "People have their own values and contexts and relationships that influence how they see the risk. For example, there can be anxieties or feelings of guilt. All these aspects are important to discuss and think about before testing and sometimes after."
- **There are laws that protect against genetic discrimination by health insurance providers, but these laws are not absolute and do not apply to disability or life insurance.** Issues of insurance and privacy are complex and can have far-reaching consequences that are important to anticipate as much as possible.
- **Genetic testing may not provide all the answers.** Raymond explains: "There are certain people for whom finding a dystonia gene with the testing available is unlikely, so genetic counseling may be more about clarifying what testing will tell us, what it won't tell us, and exploring the inheritance pattern in the family for clues to help explain the risk. But if you have questions, by all means, it's appropriate to have counseling."

### Direct-to-Consumer Genetic Testing

Up until recently, genetic testing has been available only through physicians and genetic counselors. Direct-to-consumer genetic testing is advertised directly to patients by private companies. Patients provide their genetic information via a saliva sample directly to the company without necessarily involving a doctor or insurance provider in the process.

Multiple medical and scientific organizations and regulating bodies have expressed concerns about taking genetic testing out of a healthcare setting. Some of these concerns involve patient education, marketing, lack of regulation, and privacy.

"Direct-to-consumer testing is definitely an option that is available in some states, but for all of the reasons that we are discussing, the concern is that people aren't really able to anticipate all the aspects when they send in their spit sample because they don't get counseling before the testing," Raymond explains. "Anybody who is considering direct-to-consumer testing should speak with a genetic counselor, or their physician, before they make the decision to test so they can think through the complexities ahead of time."

### Working with a Genetic Counselor

If you have questions about the genetics of dystonia, consider talking to your doctor about seeing a genetic counselor, ideally one with a background that includes neurological disorders. Genetic counselors can be identified by referrals from your doctor, through your health insurance provider, or professional genetic counseling organizations such as the National Society of Genetic Counselors.

*Deborah Raymond, MS, CGC, is a certified genetic counselor and clinical researcher in the Mirken Department of Neurology Center for Movement Disorders at Mount Sinai Beth Israel Medical Center where she counsels patients and families about genetic movement disorders and options for genetic testing, and is a senior coordinator of research studies aimed at understanding the genetic causes of dystonia and Parkinson's disease.*

This article is the first of a two-part series. Look for an update on the genetics of dystonia in the next *Dystonia Dialogue*.

## Common Misconceptions about Dystonia Genetics

- **There is one genetic test for dystonia.** This is not accurate. There are several types of genetic testing available for dystonia, and several dystonia genes that can be detected.
- **If you are negative for the DYT1 gene, your dystonia is not genetic.** This is not necessarily true. There are multiple genes for dystonia. A dystonia patient can test negative for all known genes, and the dystonia may still be genetic—the gene has not yet been discovered.
- **If you have a dystonia gene, you are guaranteed to get dystonia.** This is not true. Most people who inherit a dystonia gene never develop symptoms. However, their biological children may be at risk of inheriting the gene and developing symptoms.

## Types of Testing

### Single Gene Sequencing

Dystonia can be caused by a single faulty gene. Testing can be focused to look at one specific gene.

### Genetic Panels

Testing can look for multiple dystonia-causing genes at once.

### Whole Exome Sequencing

Testing can look for changes in the ~20,000 genes involved in the body's functions and identify those genes that may be contributing to dystonia symptoms. However, this type of testing cannot pick up all types of mutations and may generate findings that are inconclusive.

### Whole Genome Sequencing

Testing can look for changes in the entire genetic code including parts that do not contain genes but may be important for gene function. This method is used primarily for research purposes and does not yet have a clear clinical application for dystonia.

# Dystonia 101

Dystonia can be a confusing disorder to understand. Here are the basics:

- Dystonia is a neurological disorder. It affects the brain's ability to control voluntary muscle movement.
- Dystonia does not affect vital organs such as the heart.
- There are many forms of dystonia. It can affect a single body area or multiple muscle groups.
- Dystonia may occur with other movement symptoms such as tremor, myoclonus, or parkinsonism.
- Inherited dystonias are those with a known genetic origin, for example mutations in the DYT-designated genes such as DYT1, DYT5, or DYT11.
- Dystonia may result from birth injury, drug exposure, brain injury, infection, and other secondary causes.
- Treatment options include oral medications, botulinum neurotoxin injections, deep brain stimulation, and less invasive methods such as physical or occupational therapy.
- Individuals with dystonia may experience depression and anxiety disorders, including social phobia.
- Stress does not cause dystonia, but symptoms may worsen in stressful situations.

For more information, visit [dystonia-foundation.org](http://dystonia-foundation.org)



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