



Genetic Testing: A Critical Step in Helping You Obtain a Definitive Diagnosis

A definitive diagnosis is the first step to effectively managing your neuromuscular disease. It can help ensure you receive the most appropriate treatment and supportive care, achieve the best outcomes, and gain access to clinical trials or specific treatment regimens.

What is genetic testing?

- Every person has genetic information, called DNA, within his or her cells.
- DNA is inherited from your parents and contains coded information within genes.
- Your genes determine how your cells are built and, ultimately, the way your body looks and functions.
- Sometimes a mutation, or a change in your DNA, occurs that makes up a specific gene.
- One mutation can completely change the way a gene is expressed.
- Some types of mutations can be harmless and undetectable, while others affect the way your body may appear or function. And some mutations can be the underlying cause of your neuromuscular disease.



What happens once I have genetic testing done?

Once your doctor suspects that you may have a neuromuscular disease, he or she may recommend that you have a DNA test of your saliva or blood to confirm the diagnosis.

The results of your genetic testing will help your doctor find common mutations associated with your disease that will inform which types of treatments and supportive care might be best to help manage your disease.

The results can also help identify disease-specific registries and clinical trials in which you may be eligible for participation.



TO LEARN MORE ABOUT THE IMPORTANCE OF GENETIC TESTING, VISIT MDA.ORG



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