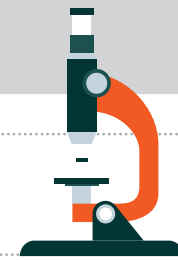


# HOW GENETIC NEUROMUSCULAR DISEASE HAPPENS



**DNA is a complex molecule that contains our genetic code.** A gene variant is a change in the DNA code that alters how cells grow or work. A variant can cause a genetic condition.

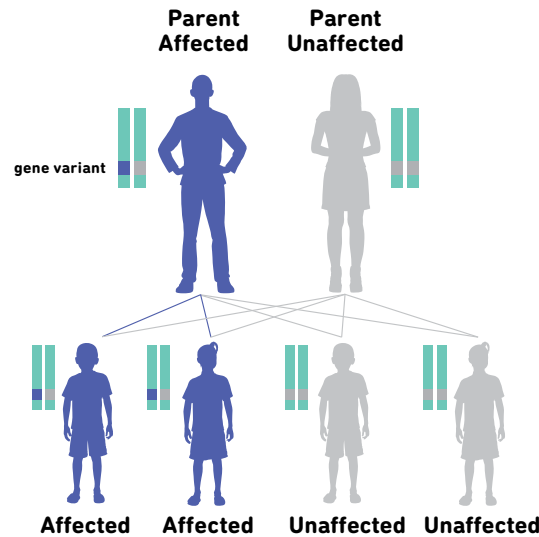
There are two ways for an individual to acquire a disease-causing gene variant.

- ▶ **Spontaneous:** Changes in a gene can result from random errors in natural biological processes and cause disease in someone who has no family history.
- ▶ **Inherited:** Gene variants can be passed down from parent to child.

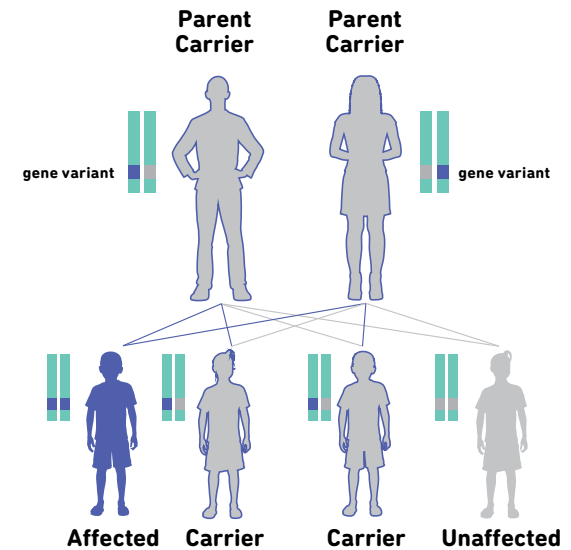
## Inheritance patterns

Every person has two copies of each gene — one from their mother and one from their father. There are four ways that an inherited genetic condition can be passed along in a family.

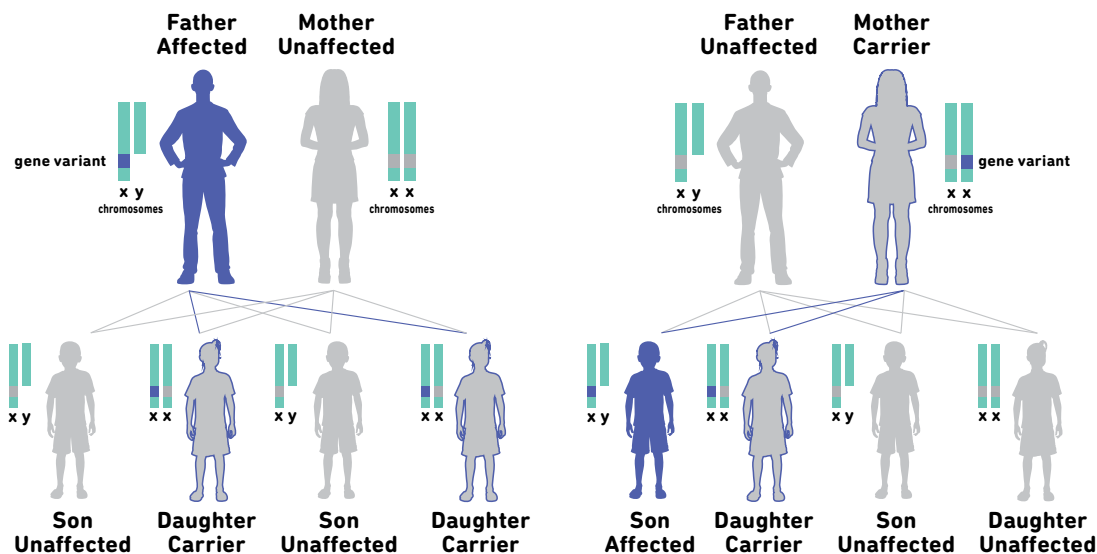
### AUTOSOMAL DOMINANT INHERITANCE



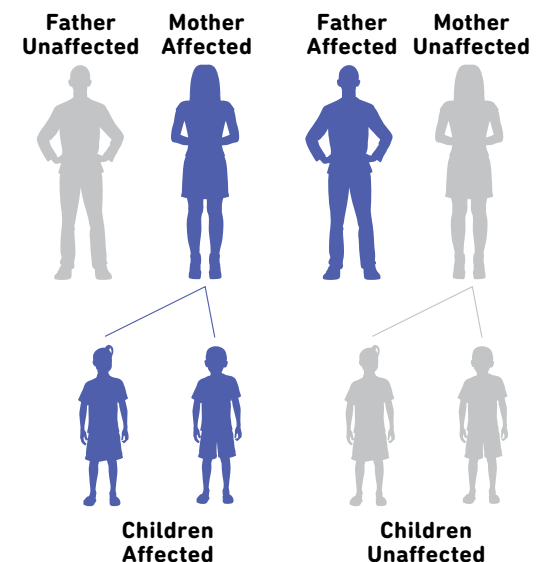
### AUTOSOMAL RECESSIVE INHERITANCE



### X-LINKED RECESSIVE INHERITANCE



### MITOCHONDRIAL INHERITANCE



● This inheritance pattern is not sex-specific. ● This inheritance pattern is sex-specific.

**A diagnosis confirmed by genetic testing** can provide an answer for why an individual is experiencing symptoms, as well as the exact cause of their condition.

Genetic testing can:

- ▶ Enable proper diagnosis, especially when no one else in the family displays symptoms
- ▶ Guide management of a condition that is known to have many different causes and/or different treatments for the same symptoms
- ▶ Help determine the inheritance pattern based on who has the condition in the family and tell you who else in the family is at risk for the same condition
- ▶ Determine eligibility to participate in research studies, which could provide symptom relief or lead to a cure for the condition

Whether to undergo genetic testing is a choice that an individual and their family must make together. It is important to hear the pros and cons of genetic testing for your condition or the condition in your family from a genetic counselor so you can make an informed decision.

**Read more at [mda.org/education](https://mda.org/education):**

- ▶ **Genetics and Neuromuscular Disease**
- ▶ **Basics of Genetic Testing**

**MDA's Resource Center may be able to help you find resources for genetic testing. Call us at 833-ASK-MDA1.**

This resource was developed with the expertise and knowledge of Tiffany Grider, MS, LGC, from the University of Iowa Hospitals and Clinics.

66-0051 June 2021