# The Role of Genetic Counseling in Informing Families About Genetic Testing Results

Genetic testing can provide important information for making an accurate neuromuscular disease diagnosis. But for patients whose results identify a pathogenic variant or mutation, sharing that information isn't an exact science.

Kelly Minks, MS, CGC, a genetic counselor at the University of Rochester Medical Center, is experienced with helping individuals and families with neuromuscular diseases interpret their genetic test results and decide when, how, and with whom to share that information.

According to Kelly, there are several good reasons to share genetic test results with family members. "They might have health implications for family members who may be at risk of developing diseases," she says. "They may want the information so they can look into the availability of disease-specific therapy, screening for related medical conditions, and the option to participate in research studies and clinical trials, along with family planning purposes."

## Addressing family dynamics

For patients, one of the hardest parts of genetic testing can be sharing the results. "We know family relationships can be complicated, and not everyone is comfortable sharing information," Kelly says. While acknowledging the tricky nature of family dynamics, she tells patients that sharing genetic test results is important, so everyone in a family who may be affected can make their own decisions about genetic testing, screening, or treatments.

Genetic testing results can be complicated and often difficult to explain to relatives. "It's important to get across for predictive or presymptomatic testing, for example, that it's a misconception that genetic testing will always tell you if you will develop a disease or not," she says. "We know for some conditions that this is not the case, so it's important to present the information correctly."

Kelly notes a genetic counselor can help patients with these difficult conversations by:

- Brainstorming ways to discuss information and help practice those conversations
- Writing a letter explaining the genetic test results and risks, which a patient can give to their relatives instead of or in addition to a conversation
- Providing local genetic counseling resources for at-risk relatives to discuss their own personal risk and options for genetic testing

## Who needs to know?

Kelly recommends that patients share their genetic test results with all at-risk relatives. This may include first-degree relatives — parents, siblings, and children — and others, depending on the inheritance pattern of the disease. A genetic counselor can help a patient identify who is at risk in their family.

"It's important for the ones who it's being shared with to get a copy of the test results, so a genetic counselor or geneticist knows the specific gene and variant causing disease in the family," Kelly says. "It allows for more specific and accurate testing for relatives."

When discussing a child's genetic test results, sharing can be a different challenge. "There is concern about a child's ability to understand information and potential for psychological harm," she says. "There are many resources for sharing genetic information and history of family disease with children, such as publications, in an appropriate way. A genetic counselor can help with navigating these difficult conversations as well."

Kelly recommends the following resources for talking to children about risk or a diagnosis:

- New Diagnosis and Early Care Guide (from Parent Project Muscular Dystrophy)
- Discussing MD with Children — for Parents and Caregivers (from Muscular Dystrophy Australia)
- Books About Duchenne for Children and Parents (from Cure Duchenne)

## Who should get tested?

Genetic testing is an option for any at-risk relative of a person with an identified pathogenic variant or mutation.

“The decision to have genetic testing is a personal one, and at-risk relatives should meet with a genetic counselor to discuss the benefits and limits of testing,” Kelly says. “Some genetic diseases also have associated medical risks that have available screening, such as cardiac disease risk. Genetic testing would allow for earlier screening for those individuals.”

Limitations to genetic testing include reduced penetrance in certain diseases, meaning not everyone with the mutation or the pathogenic variant will develop the disease. In these cases, being identified as having the mutation could cause unnecessary psychological distress.

## Addressing testing concerns

Some people may be reluctant to get genetic testing even when presented with the benefits and limitations.

“One common reason for hesitation is potential costs for genetic testing,” Kelly says. “But counselors can help them navigate the billing options. Negative emotions can also be triggered, like anger and guilt, and it may cause some hesitancy to pursue genetic testing. A person may also be concerned about social and family repercussions — they may worry about how other family members will feel if the test results reveal information about them.”

## Family planning

Another factor to consider in disclosing genetic testing results is that it could give relatives the option to consider family planning. For this option, it’s best to have genetic testing results before pregnancy.

“I recommend a relative considering pregnancy meets with a reproductive genetic counselor to learn about potential risks to their own health, risks for pregnancy, and the available genetic testing options, which include preimplantation genetic diagnosis or prenatal testing,” she says. “It’s also important to know that prenatal genetic testing and preimplantation genetic diagnosis are only an available option if there is a known pathogenic variant or mutation in the family.”

Discussing health issues that affect oneself or a loved one is never easy. Genetic counselors can make sure patients consider all the factors when deciding if or how to share their genetic test results with their families.

## Resources

MDA Community Education offers virtual learning programs, webinars, print-ready educational materials, and more to inform the neuromuscular disease community. Consider sharing these resources on topics including genetics and genetic testing with your patients. Find them at mda.org/care/community-ed.