DYSTONIA MEDICAL RESEARCH FOUNDATION

Genetics of Myoclonus-Dystonia

Myoclonus-dystonia (M-D) is a movement disorder characterized by a combination of rapid, brief muscle contractions (myoclonus) and/or sustained twisting and repetitive movements that result in abnormal postures (dystonia). Non-movement features may include obsessive compulsive disorder, depression, anxiety, personality disorders, alcohol abuse, and panic attacks. Symptoms typically begin in the first or second decades of life, and rarely in adulthood. M-D may affect multiple members in a family or occur without a family history.

A genetic counselor can help individuals learn more about the genetics of M-D in their unique families and what kinds of genetic testing may be appropriate.

Genes Associated with M-D

M-D often affects several members and generations in a family, demonstrating a clear genetic component, but M-D can also occur without a family history. About 30-40% of individuals with M-D have mutations in the *SGCE* gene (also referred to as *DYT11*). Although mutations in two other genes, *DRD2* and *DYT1*, have been associated with M-D, the significance is unknown at this time.

How is M-D Inherited?

Individuals who develop M-D and test positive for a *SGCE* mutation may have inherited the disorder from a parent or have a brand new mutation. M-D is dominantly inherited, which means that only one parent needs to have a mutation for a child to develop M-D.

Each child of an individual with M-D has a 50% chance of inheriting a mutation. However not every child who inherits the mutated gene will develop symptoms.

Children who inherit an *SGCE* mutation from their father are very likely (90%) to develop symptoms, while only 5% of children who inherit the mutation from their mother will develop symptoms. It is possible to be a carrier of the mutation without having M-D symptoms. Carriers can also pass along the mutation to their biological children.

Who is Eligible for Genetic Testing?

Although genetic testing is available as part of the diagnostic process, an individual can have M-D without testing positive for a genetic mutation. A qualified genetic counselor can help individuals and families understand what testing may be appropriate for them. Generally speaking, testing may be appropriate for:

- Adults and children who exhibit clinical symptoms of M-D.
- Adult biological relatives of individuals who have been genetically confirmed as having M-D.

Genetic testing is generally not recommended for children who do not exhibit symptoms, even if a biological relative tests positive for a disease-causing mutation.

Options exist for carriers of known M-D mutations who wish to decrease the likelihood of passing the mutation onto their children. These options include prenatal testing and pre-implantation genetic diagnosis (PGD). PGD uses *in vitro* fertilization and sophisticated gene testing techniques prior to pregnancy to dramatically reduce the chance of a child inheriting a disease-causing mutation.

See back for more information.

When a Child has M-D: Emotional Impact on Parents

The diagnosis of a disorder like M-D in a family can cause a range of reactions and emotions. The inherited nature of M-D can add another dimension to the impact on both the individuals affected and the family as a whole.

Parents often feel a powerful sense of guilt and grief at having contributed to M-D in their children. Especially because M-D is rare and can be debilitating, this sense of guilt may be multiplied. This is a common experience of parents with children of genetic disorders.

Although parents are not at fault when a child develops M-D, they may grieve the life they had envisioned or wished for their child. While a genetic counselor can help families understand the inheritance patterns of M-D, parents who experience grief or guilt are encouraged to seek the assistance of a professional mental health therapist or counselor to work though these feelings.

Parents of children with M-D are also encouraged to reach out to fellow parents, consider joining the DMRF's Support4Parents of Children with Dystonia Facebook group, and actively help their children navigate life with M-D.

Locating a Genetic Counselor

A genetic counselor is a medical professional who specializes in genetic diseases and how diseases affect families. A consultation with a genetic counselor can help identify what testing, prevention strategies, or research trials may be appropriate for a particular individual or family impacted by M-D. Individuals may choose to locate a genetic counselor by contacting their movement disorder specialist or health insurance provider. Professional organizations can also provide contact information for genetic counselors.

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About the DMRF

The Dystonia Medical Research Foundation (DMRF) is a 501(c)(3) non-profit organization dedicated to advancing research for more dystonia treatments and ultimately a cure, promoting awareness, and supporting the well-being of affected individuals and families.

http://www.dystonia-foundation.org

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