DYSTONIA MEDICAL RESEARCH FOUNDATION

What is Myoclonus-Dystonia?

Myoclonus-dystonia (M-D) is a rare and complex neurological movement disorder that affects individuals and families around the world. Treatments are available and researchers are actively pursuing improved therapies and ultimately a cure. Resources are available to help support affected individuals and families.

Causes of Myoclonus-Dystonia

M-D often affects several members and generations of a family, demonstrating a clear genetic component. M-D can also occur without a family history. About 30-40% of individuals and families impacted by M-D have mutations in the *SGCE* gene (also referred to as *DYT11*).

Individuals who develop M-D 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.

Children who inherit an *SGCE* mutation from their father will almost always (90%) develop symptoms. Only 5% of children who inherit the mutation from their mother will develop symptoms.

About 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).

The myoclonus jerks typical of M-D most often affect the neck, trunk, and upper limbs. Approximately 50% of affected individuals have dystonia affecting the neck and/or hand. Dystonia or myoclonus in some individuals may mimic tremor.

M-D typically includes more than movement symptoms. Non-movement features may include obsessive compulsive disorder, depression, anxiety, alcohol abuse, and panic attacks. M-D does not affect cognition, intelligence, or shorten lifespan.

Symptoms can vary significantly among individuals, including within the same family. Individuals with M-D report fluctuation of symptom severity, episodic escalation of movement symptoms (with or without trigger event), respiratory effects, and voice impairment.

Symptoms typically begin in childhood or early adolescence but may also first appear in adulthood.

Many individuals experience a dramatic decrease of the myoclonus jerking symptoms, and sometimes improvement of dystonia, upon ingesting alcohol.

How Common is Myoclonus-Dystonia?

M-D is rare, but little is known about how prevalent it is within the United States or elsewhere.

M-D affects males and females equally, and across nationalities and ethnicities.

See back for more information.

What Research is Being Done?

Research on M-D is being conducted at centers around the world and is focused on developing a more thorough understanding of the disorder while seeking breakthroughs in genetics and therapeutics.

The Myoclonus-Dystonia Research Program, a partnership of the Brown Family Foundation and the Dystonia Medical Research Foundation (DMRF) is a focused effort specifically on M-D. The Myoclonus Dystonia Research Program has funded research projects led by leading researchers and workshops that bring together renowned experts to review what is known about M-D and assess next steps for the field.

Efforts are underway to better understand how mutations in the *SGCE* gene lead to symptoms, and to clarify the function of the protein encoded by this gene, epsilon-sarcoglycan. Researchers know that, when mutated, this protein is implicated in the development of M-D, but do not yet understand what purpose this protein serves in the body in its normal state.

Researchers are actively trying to better understand the genetics of M-D by identifying possible new mutations or additional genes. Studies are also ongoing to look for links between the *SGCE* gene and epsilon-sarcoglycan protein and the genes and proteins known to play a role in additional movement disorders. Understanding the genetics of M-D is directly linked to developing therapies, because understanding the genetic basis of the disorder provides targets for new treatment strategies.

Treatment

At this time, there is not yet a cure for M-D, but treatments are available to help minimize symptoms. Treatment must be highly customized to the individual. Each component of the M-D—the myoclonus, the dystonia, and the mood component—requires attention for complete care and the most positive results.

Therapies to address the motor symptoms of M-D typically include oral medications and botulinum neurotoxin injections. Deep brain stimulation is a neurosurgical procedure that is showing promise as an effective therapy for M-D. Complementary therapies such as physical or occupational therapy may support medical treatments.

If obsessive compulsive disorder, depression, anxiety, personality disorders, alcohol abuse, and/or panic attacks are present, these symptoms must also be addressed. Although sometimes difficult to talk about, individuals with M-D who experience signs of these complications are encouraged to bring them to the attention of their treating physician. If family members or loved ones observe signs of such complications, it may be helpful to share their concerns with the individuals and/or with healthcare providers.

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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

Find the DMRF on Facebook, Twitter, and YouTube.

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