Myotonic Dystrophy Facts

What is myotonic dystrophy?

- Described as "the most variable of all diseases found in medicine," myotonic dystrophy (DM) is a multi-systemic genetic disorder that is the most common form of adult-onset muscular dystrophy.
- DM is the only form of muscular dystrophy that has impacts on cognition and brain function, in addition to impacts on the heart, lung, muscles, gastrointestinal system, and many other parts of the body.
- There are two forms of DM: DM1 and DM2. Both are passed from parent to child by autosomal dominant mutations of a particular gene. Because this gene is not located on the X or Y chromosome, it can be passed to male and female children with equal frequency.

What makes myotonic dystrophy a rare disease?

- In the United States, a disease is considered rare if it affects fewer than 200,000 Americans. DM is classified as a rare disease as it affects somewhere between 1:3,000 and 1:5,000 people worldwide, or an estimated 100,000 Americans.
- Many rare diseases, like DM, are genetic. DM impacts entire families and becomes more severe with each successive generation. The symptoms and disease progression can vary widely, even among members of the same family.
- Some rare diseases are apparent at birth, and approximately two-thirds of those living with rare diseases are children. The congenital and juvenile-onset forms of DM are particularly devastating as they often present life-threatening issues from birth or a very young age.

What causes myotonic dystrophy?

- Individuals with DM have a particular gene that conveys faulty instructions, resulting in the symptoms of DM.
- DM1 and DM2 are caused by mutations in different genes. While they show similar symptoms, they have fundamentally different origins.
- Scientists are currently looking into the possibility that there may be additional forms of DM caused by genetic mutations at different sites.

Widely varying symptoms often lead to a long diagnostic odyssey.

- Since DM symptoms often mimic more common diseases, extensive testing is usually required to rule these out.
- Delays in diagnosis are common, and it typically takes over seven years to diagnose DM1 and more than 11 years to diagnose DM2.

Like many other rare diseases, there are currently no treatments or a cure for myotonic dystrophy.

- Researchers are close to finding the answers to restore cell function in the body systems that are attacked by DM. It presents an enormous research opportunity that extends far beyond this disease alone.
- DM is considered a breakthrough disease in terms of its ability to advance science around triplet repeat, RNA diseases, and other neuromuscular disorders.
- Scientists have reversed the condition in laboratory mice, and human clinical trials are scheduled for 2014.

P.O. Box. 29543 | San Francisco, CA 94129
www.myotonic.org | 86-myotonic or 415.800.7777 | info@myotonic.org