MYOTONIC DYSTROPHY FOUNDATION

Care and a Cure

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 affects somewhere between 1:3000 and 1:8000 people worldwide.
- While the adult-onset form of DM is the most common form of muscular dystrophy, there are also very severe forms that present at birth or during childhood that can cause permanent developmental delays and severe, often life-threatening impacts to many organs and systems of the body.
- 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 types 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 causes myotonic dystrophy?

- Individuals with DM have an abnormally expanded stretch of DNA, which is caused by a high amount of triplet RNA repeats. Due to a phenomenon known as "anticipation," these repeats increase with each successive generation, causing symptoms to present earlier and become more severe.
- DM1 and DM2 are caused by mutations in different genes. The genes for DM1 are found on chromosome 19, while the genes for DM2 are found on chromosome 3. While they both show similar symptoms, the two types of DM are distinct and 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.
- As a genetic disease, DM impacts entire families. However, even among members of the same family, the symptoms and disease progression can vary widely, which further complicates diagnosis.
- Delays in diagnosis are common: it typically takes over seven years to diagnose DM1 and more than 11 years to diagnose DM2.

There is currently no 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, toxic RNA diseases, and other neuromuscular disorders.
- Scientists have reversed the condition in laboratory mice, and the first human clinical trials were launched in 2014.

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