

Myotonic Dystrophy Foundation Advocacy Priorities 2021 Rare Disease Day

Myotonic dystrophy is a multi-systemic inherited disease that affects at least 1 in 2,100 people or over 150,000 individuals in the United States. The disease is caused by a mutation in a gene required for normal muscle function which prevents the gene from carrying out its function properly. The genetic mutation is an autosomal dominant mutation, where one copy of the altered gene is sufficient to cause the disorder. As a result, affected individuals have a 50% chance of passing on the mutated gene to their children. A child is equally likely to have inherited the mutated gene from either parent. If both parents do not have the disease, their children cannot inherit it. Children with congenital myotonic dystrophy almost always inherit the disease from an affected mother.

Individuals affected by the genetic mutation may have skeletal muscle problems, heart function abnormalities, breathing difficulties, cataracts, issues with speech and swallowing (dysarthria and dysphagia), cognitive impairment, excessive daytime sleepiness, or diabetic symptoms. Any single individual is unlikely to have all or even most of these symptoms. Myotonic dystrophy is a highly variable and complicated disorder. The systems affected, the severity of symptoms, and the age of onset of those symptoms vary greatly between individuals, even in the same family. In general, the younger an individual is when symptoms first appear, the more severe symptoms are likely to be. A complete diagnostic evaluation, which includes family history, physical examination, and medical tests, is typically required for a presumptive diagnosis of myotonic dystrophy. The presence of the disorder can then be confirmed by genetic testing. Prenatal testing, where the DNA of the fetus is checked for the presence of the myotonic dystrophy mutation, is also available. The disease symptoms grow more significant and disabling with each new generation.

Delays in diagnosing myotonic dystrophy are common. This is usually because of the lack of familiarity with the disease on the part of healthcare providers and that more common diseases with symptoms that mimic myotonic dystrophy must typically first be ruled out before this disorder is considered. There are currently no Food and Drug Administration (FDA) approved treatments for myotonic dystrophy, however, over 40 biopharmaceutical companies are working on treatments for the disease, with many leading promising clinical trials.

Call to Action

During Rare Disease Day, Myotonic Dystrophy Foundation advocates from across the country seek to create awareness of this rare genetic disorder and urges our Representatives and Senators to take the following actions:

- Increase fiscal year 2022 National Institutes of Health (NIH) funding for myotonic dystrophy research that will improve health outcomes, reduce disability, and increase life expectancy for individuals living with the disease,
- Maintain myotonic dystrophy as an eligible condition for research funding under the Department of Defense Peer Reviewed Medical Research Program (PRMRP) in the fiscal year 2022 Senate Defense Appropriations bill.
- Finally, we are anticipating Congress will consider legislation recognizing Myotonic Dystrophy Awareness Day this year, and we hope we can count on your support.