

# Myotonic Dystrophy

## At A Glance

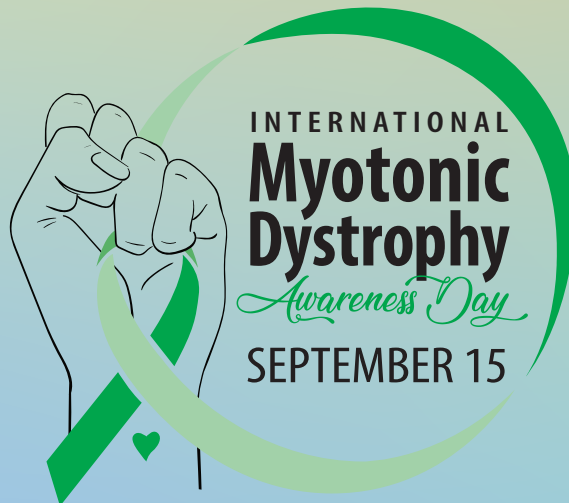
Myotonic dystrophy is a rare, multi-systemic, inherited disease that affects an estimated 1 in 2,100 people, or over 3.6 million individuals across the world.<sup>1</sup>

Myotonic dystrophy is the most common form of adult muscular dystrophy and considered the most variable of all known conditions.<sup>2</sup>

Myotonic dystrophy is commonly referred to as DM, an abbreviation of the Latin name used by doctors and researchers worldwide: dystrophia myotonica. Other names for DM include myotonic muscular dystrophy (MMD), Steinert's Disease for DM1, and Proximal myotonic myopathy (PROMM) for DM2.<sup>3-31</sup>

Mutations prevent genes from carrying out their functions properly, which can impact multiple body systems. Myotonic dystrophy type 1 is caused by a mutation in the DMPK gene, while myotonic dystrophy type 2 is caused by a mutation in the CNBP gene.<sup>3-31</sup>

Myotonic dystrophy symptoms usually become more severe with each generation, yet there is currently no cure and there are no approved treatments.<sup>74-89</sup>



Millions of people are living with DM globally, yet millions of people do not know they have the disease and are in need of care.<sup>1</sup>

Myotonic dystrophy is inherited - people living with myotonic dystrophy have a 50% chance of passing on the mutated gene to their children.<sup>8</sup>

People living with myotonic dystrophy experience varied and complex symptoms, from skeletal muscle problems<sup>91-112</sup>, to heart<sup>113-126</sup>, breathing<sup>127-150</sup>, digestive<sup>151-166</sup>, hormonal<sup>198-219</sup>, speech and swallowing<sup>246</sup>, diabetic<sup>247</sup>, immune<sup>220-237</sup>, excessive daytime sleepiness<sup>248</sup>, early cataracts and vision challenges<sup>238-245</sup>, and cognitive difficulties.<sup>167-197</sup>

Myotonic dystrophy doesn't always look the same. The different body systems affected, the severity of symptoms, and the age of onset of those symptoms vary greatly between individuals, even in the same family.<sup>249-250</sup>

Over 35 biopharmaceutical companies are leading promising research which may result in new treatments for myotonic dystrophy, and, one day, a cure.<sup>251</sup>

Delays in diagnosing myotonic dystrophy are common. Despite the availability of simple genetic tests, a lack of familiarity with the disease on the part of healthcare providers can allow misdiagnoses to persist for decades.<sup>251</sup>

Learn more at:

[www.myotonic.org/myotonic-dystrophy-glance](http://www.myotonic.org/myotonic-dystrophy-glance)