FIVE THINGS TO KNOW ABOUT ...

Myotonic dystrophy type 1

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1 Myotonic dystrophy type 1 is a common form of muscular dystrophy

The prevalence of myotonic dystrophy type 1 ranges between 0.5 and 18.1 per 100 000 population, making it the most common muscular dystrophy, ahead of Duchenne and facioscapulohumeral muscular dystrophies.[1] Myotonic dystrophy type 1 is especially prevalent in certain regions of Quebec.[2] It is an autosomal dominant nucleotide repeat disorder. The mutant messenger RNA with expanded repeats has a toxic gain of function, resulting in widespread splicing dysregulation.

2 Myotonic dystrophy type 1 is multisystemic

Myotonic dystrophy type 1 can present at any age but is typically diagnosed in adults. Although this disorder can present in many different ways (Appendix 1, available at www.cmaj.ca/lookup/suppl/doi:10.1503/cmaj.151384/-/DC1), facial and distal muscle weakness, along with grip myotonia, are key findings on examination. Given the risk of sudden death (described below), dominant inheritance and high penetrance,[3] first-degree relatives and women of childbearing age who are at risk of carrying an affected child should be referred for genetic counselling.

3 Myotonic dystrophy type 1 shows genetic anticipation

Genetic anticipation, the earlier and more severe presentation in offspring, relates to the unstable trinucleotide repeat mutation expanding in subsequent generations. Congenital myotonic dystrophy type 1 is the most severe manifestation of this disorder. Congenitally affected neonates with this manifestation are weak and hypotonic, and often require ventilation and feeding support. Mortality is about 25% in the first year.[4] Strength and ability to feed and breathe improve in those who live beyond the first year.

4 Cardiac arrythmia is a frequent cause of death

Unlike other dystrophies, arrhythmias and other conduction abnormalities are the primary cardiac manifestation in myotonic dystrophy type 1 and require regular electrocardiographic (ECG) monitoring and referral to cardiology.[5] Sudden cardiac death may be predicted by severe abnormalities detected through ECG monitoring and a diagnosis of symptomatic atrial tachyarrhythmias.[5]

5 Treatments are symptomatic

No treatment to modify disease currently exists. However, mexiletine is effective in reducing myotonia based on evidence from randomized controlled trials (RCTs).[6] In a small RCT, methylphenidate was found to decrease excessive somnolence seen in this disorder.[7] There are some promising disease-modifying therapies entering clinical trials.

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