

## FIVE THINGS TO KNOW ABOUT ...

**Myotonic dystrophy type 1**

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**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.<sup>1</sup> Myotonic dystrophy type 1 is especially prevalent in certain regions of Quebec.<sup>2</sup> 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.

**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.<sup>4</sup> Strength and ability to feed and breathe improve in those who live beyond the first year.

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**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](http://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,<sup>3</sup> first-degree relatives and women of childbearing age who are at risk of carrying an affected child should be referred for genetic counselling.

**Cardiac arrhythmia 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.<sup>5</sup> Sudden cardiac death may be predicted by severe abnormalities detected through ECG monitoring and a diagnosis of symptomatic atrial tachyarrhythmias.<sup>5</sup>

**References**

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**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).<sup>6</sup> In a small RCT, methylphenidate was found to decrease excessive somnolence seen in this disorder.<sup>7</sup> There are some promising disease-modifying therapies entering clinical trials.

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