



Module 2

How is myotonic dystrophy type 1 (DM1) diagnosed?

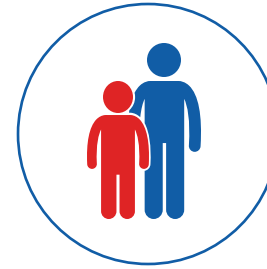


Module summary



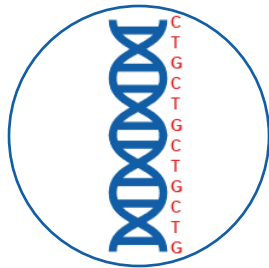
DM1 is associated with a diagnostic delay of 6–7 years^{1,2}

Delays are due to the diversity in age of onset, the wide range of body systems involved, and the variability in symptom severity^{1–3}



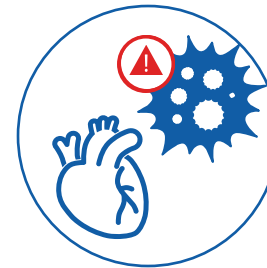
A diagnosis of DM1 should be suspected in individuals with a family history of DM1 and/or associated features^{4–6}

Features vary between individuals and by age, and include muscular and non-muscular manifestations^{4–6}



Genetic testing detects abnormal CTG repeat expansion and confirms a DM1 diagnosis^{4,7,8}

After diagnosis, the risk to family members should be explained, and a genetic study should be proposed to those aged > 18 years⁸



Early diagnosis of DM1 is important to introduce appropriate management of disease-associated risk factors¹

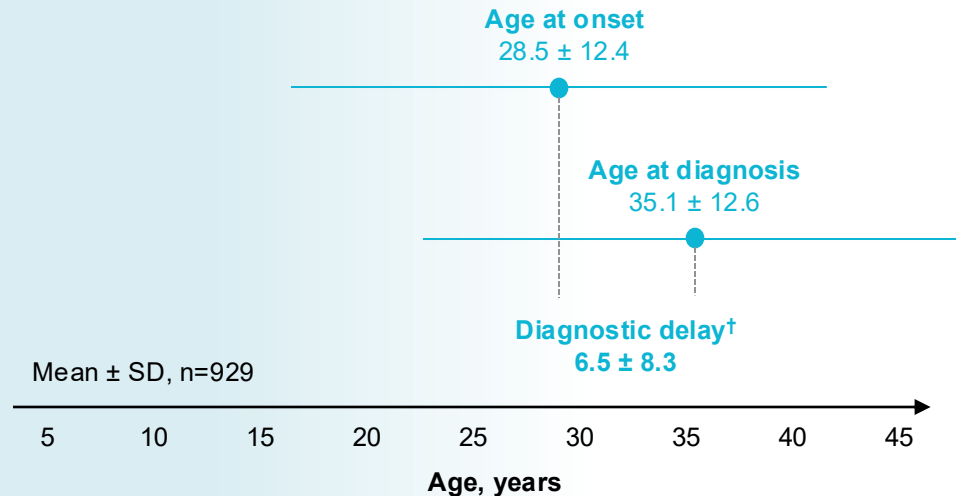
Individuals with DM1 have a higher risk of cardiac abnormalities, cancer, and certain surgical complications than those without DM1^{9–16}

CTG, cytosine-thymine-guanine; DM1, myotonic dystrophy type 1.

1. Hilbert JE, et al. *J Neurol*. 2013;260:2497–2504; 2. Hageman KA, et al. *Muscle Nerve*. 2019;59:457–464; 3. Hamel JI, et al. *Muscle Nerve*. 2022;66:508–512; 4. Johnson NE, et al. *Neurol Clin Pract*. 2019;9:443–454; 5. Ho G, et al. *World J Clin Pediatr*. 2015;4:66–80; 6. Bird TD. Myotonic Dystrophy Type 1. 1999 Sep 17 [Updated 2021 Mar 25]. In: Adam MP, Everman DB, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2022; 7. Chakraborty S, et al. *Curr Protoc Hum Genet*. 2016;91:9.29.1–19; 8. Gutierrez Gutierrez G, et al. *Neurologia (Engl Ed)*. 2020;35:185–206; 9. McNally EM, et al. *J Am Heart Assoc*. 2020;9:e014006; 10. McNally EM, et al. *Heart*. 2011;1094–1100; 11. Petri H, et al. *Int J Cardiol*. 2014;174:31–36; 12. Alsaggaf R, et al. *Muscle Nerve*. 2018;57:316–320; 13. Galdalla SM, et al. *JAMA*. 2011;306:2480–2486; 14. Zampetti A, et al. *J Am Acad Dermatol*. 2015;72:85–91; 15. Win AK, et al. *Mayo Clin Proc*. 2012;87:130–135; 16. Kim CS, et al. *Sci Rep*. 2021;11:8.

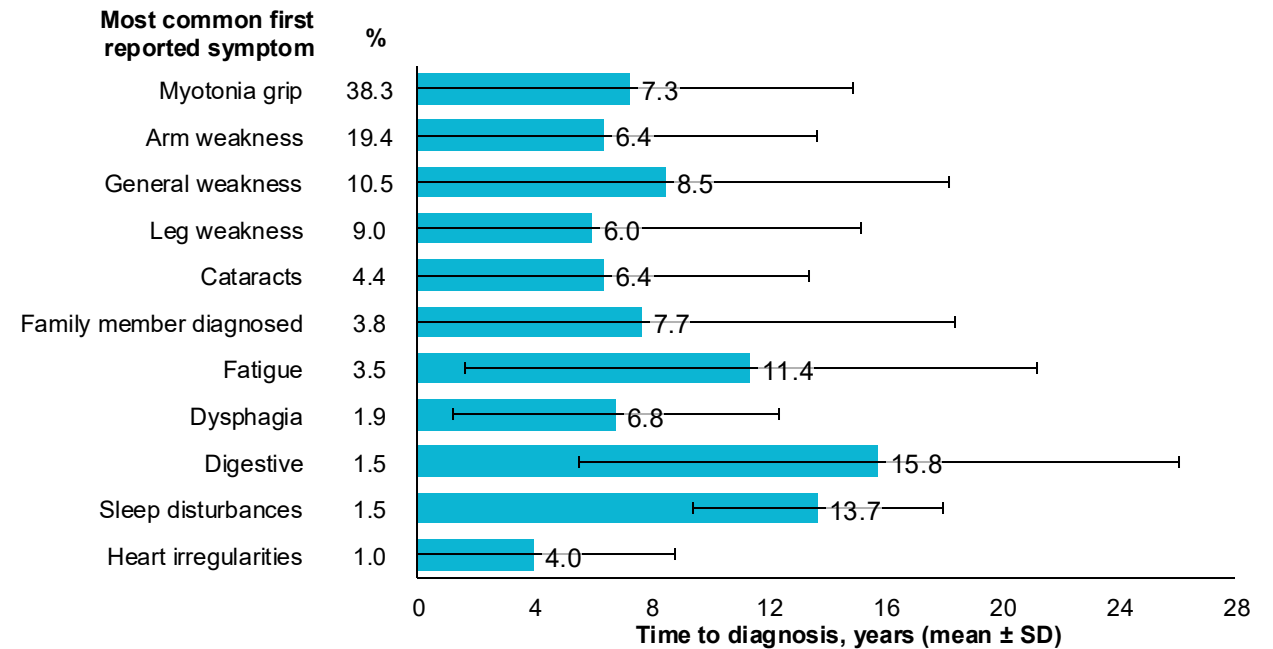
How long does it typically take for DM1 to be diagnosed?

Onset and diagnostic delay^{1*}



- Individuals with DM1 experience significant delays in diagnosis due to the diversity in age of onset, the wide range of body systems involved, the wide variability in severity of the signs and symptoms of the disease, and the number of different practitioners consulted^{1–3}

Time to diagnosis by most common first reported symptom^{3†}



- Myotonia (49.2%) and overall weakness (43.3%) are the most common first reported symptoms leading to a DM1 diagnosis³
- Individuals with symptoms such as fatigue, sleep disturbances, and digestive issues often experienced the longest diagnostic delay³

DM1 is associated with a diagnostic delay of 6–7 years^{2,3}

*Data from the US National Registry for Myotonic Dystrophy.

[†]This study analyzed de-identified data from Registry members classified with genetically proven or clinically defined DM1.

DM1, myotonic dystrophy type 1; SD, standard deviation.

1. Hamel JI, et al. *Muscle Nerve*. 2022;66:508–512; 2. Hageman KA, et al. *Muscle Nerve*. 2019;59:457–464; 3. Hilbert JE, et al. *J Neurol*. 2013;260:2497–2504.

What signs or symptoms may lead to a suspected DM1 diagnosis?

Neonates



- Family history of DM1¹
- Hypotonia¹⁻³
- Facial muscle weakness³
- General weakness^{1,3}
- Positional malformations (e.g. clubfoot)^{1,3}
- Respiratory distress¹⁻³
- Feeding problems^{1,2}
- Cognitive impairment²
- Motor and developmental delay²

Children/Juveniles



- Family history of DM1¹
- Eyelid ptosis and/or oral motor weakness¹
- Distal weakness¹
- General muscle weakness²
- Myotonia and muscle atrophy^{1,2}
- Scoliosis¹
- Visual impairment²
- Sleep apnea²
- GI issues¹
- EDS and fatigue^{1,2}
- Autism or social communication difficulties¹
- Conduction disturbances^{1,2}
- Testicular atrophy²
- Learning disabilities (e.g. dyslexia) and developmental/speech delays^{1,2}
- ADHD, anxiety, and behavioral problems¹

Adults



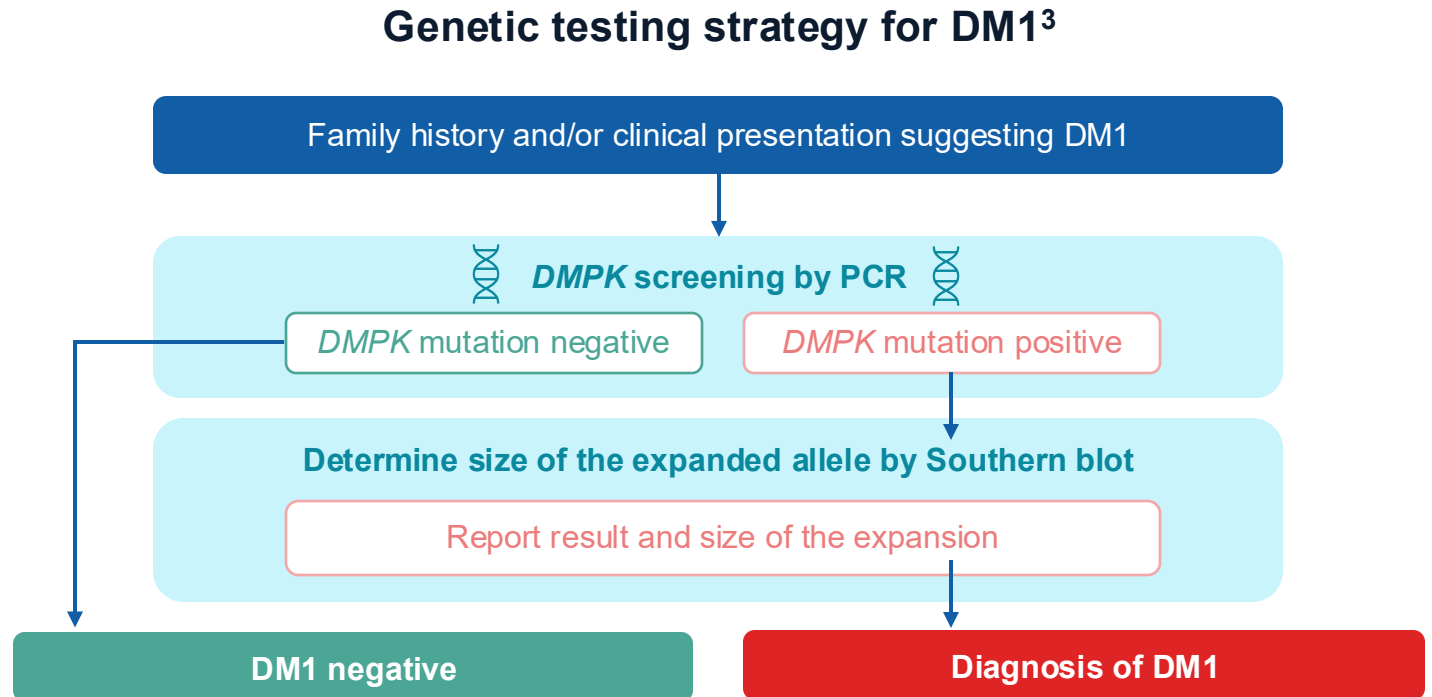
- Family history of DM1¹
- Muscle weakness³
- Myotonia^{2,3}
- Posterior subcapsular cataracts^{2,3}
- Conduction defects²
- Insulin resistance²
- Respiratory failure²

Parents of a child with DM1 should be assessed for the possibility that they, too, have DM1¹

A diagnosis of DM1 should be suspected in individuals with a family history of DM1 and/or associated features¹⁻³

How is a diagnosis of DM1 confirmed?

- Once DM1 is suspected based on family history and/or clinical presentation, molecular testing using a blood sample is used to confirm the diagnosis^{1,2}
- Muscle biopsies are not required to confirm the diagnosis of DM1²
- After diagnosis, the risk to family members should be explained, even in the absence of symptoms, and a genetic study should be proposed to those aged >18 years²
- Asymptomatic individuals found to have CTG repeat expansion should be considered at risk for developing DM1, although they may never develop symptoms⁴



Genetic testing detects abnormal CTG repeat expansion and confirms DM1 diagnosis¹⁻³

Why is early diagnosis of DM1 important?

Early diagnosis can help ensure that individuals have the opportunity to be managed by a multidisciplinary team so that known DM1 risk factors are recognized and addressed in a timely manner¹



High incidence of cardiac abnormalities and risk for arrhythmias, cardiomyopathy, conduction disturbance, and sudden death^{2–4}

Regular screening by a cardiologist familiar with neuromuscular disease is recommended²



Higher risk of cancer, compared with the normative population, regardless of lifestyle factors^{5–8}

Population-based cancer screening guidelines should be incorporated into clinical care⁵



Increased risk of surgical complications when under general anesthesia and cardiac complications after surgery⁹

Anesthesia in individuals with DM1 needs to be carefully planned¹⁰

Early diagnosis of DM1 is important to introduce appropriate management of disease-associated risk factors¹¹

DM1, myotonic dystrophy type 1.

1. Wenninger S, et al. *Front Neurol*. 2018;9:303; 2. McNally EM, et al. *J Am Heart Assoc*. 2020;9:e014006; 3. McNally EM, et al. *Heart*. 2011;1094–1100; 4. Petri H, et al. *Int J Cardiol*. 2014;174:31–36; 5. Alsaggaf R, et al. *Muscle Nerve*. 2018;57:316–320; 6. Galdalla SM, et al. *JAMA*. 2011;306:2480–2486; 7. Zampetti A, et al. *J Am Acad Dermatol*. 2015;72:85–91; 8. Win AK, et al. *Mayo Clin Proc*. 2012;87:130–135; 9. Kim CS, et al. *Sci Rep*. 2021;11:8; 10. Myotonic Dystrophy Foundation. Practical suggestions for the anesthetic management of a myotonic dystrophy patient. Accessed February 12, 2025. https://www.myotonic.org/sites/default/files/pages/files/MDF_PracticalSuggestionsDM1_Anesthesia2_17_21.pdf; 11. Hilbert JE, et al. *J Neurol*. 2013;260:2497–2504.