Shining a spotlight on Duchenne Muscular Dystrophy

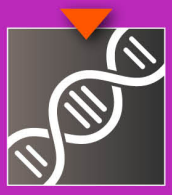
DMD is one of the most common genetic disorders[1]

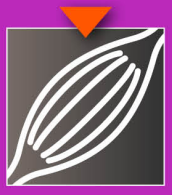
- DMD is an inherited genetic disorder caused by variants in the gene that makes dystrophin[1]
- It mostly affects males, with disease onset typically at 3–5 years of age[1]

Diagnosing DMD[1]

A physical examination and evaluation of family history are conducted first. If DMD is suspected, laboratory testing can be used to confirm a diagnosis:


Enzyme tests for blood CK levels


Genetic testing for dystrophin variants


Muscle biopsy to assess atrophy

DMD is progressive (symptoms worsen over time)[1]


Abnormal movements


Respiratory, orthopaedic and cardiac complications


Loss of independent movement


Respiratory and cardiac failure

Early diagnosis and novel treatment options can improve outcomes for patients with DMD[3]

CK, creatine kinase; DMD, Duchenne muscular dystrophy.

1. National Organisation for Rare Disorders. Available at www.rarediseases.org/rare-diseases/duchenne-muscular-dystrophy/. Accessed September 2021.
2. World Duchenne day. Available at www.worldduchenneday.org. Accessed September 2021.
3. van Ruiten HJA, et al. *Arch Dis Child.* 2014;99(12):1074–1077.

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