DUCHENNE MUSCULAR DYSTROPHY[1]

Duchenne is a lethal, X-linked recessive neuromuscular disorder caused by mutations in the dystrophin gene

Dystrophin – a cytoskeletal protein that ensures the strength, stability and functionality of myofibres

Absent or insufficient functional dystrophin

Progressive muscular damage and degeneration

Muscular weakness

Motor delays

Loss of ambulation

Respiratory impairment

Cardiomyopathy

DIAGNOSING DUCHENNE[1]

Unexplained increase in transaminases

Family history of Duchenne

Avoid diagnostic delay with prompt referral to a neuromuscular specialist

STEP 1 — *Testing for serum creatine kinase (CK)*

STEP 2 — *If CK elevated, testing for dystrophin gene deletion/duplication*

STEP 3 — *Genetic sequencing**

Confirmed Duchenne diagnosis

No family history, but...

Motor symptoms:

- Not walking by 16–18 months
- Abnormal gait and toe walking
- Gross motor delay or loss of motor skills
- Difficulty climbing or running
- Frequent falling or clumsiness
- Muscle pain, cramping or low muscle tone
- Decreased head control
- Gowers' sign

Other symptoms:

- Behavioural problems
- Cognitive delay
- Speech delay or articulation difficulties
- Learning and attentional issues
- Failure to thrive or poor weight gain

*If genetic testing does not confirm a clinical diagnosis of DMD, then a muscle biopsy should be performed

The information contained within this summary of the 2018 International Care Considerations for Duchenne muscular dystrophy is not exhaustive; please refer to references 1–3 for full information.

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THERAPEUTIC INTERVENTIONS IN DUCHENNE

Physiotherapy and treatment with glucocorticoids are the mainstays of treatment for Duchenne.[1]

PHYSIOTHERAPY[1]

The maintenance of passive ranges of movement and muscle extensibility can help:

- *Optimise movement*
- *Maintain ambulation*
- *Prevent contractures and deformities*
- *Optimise respiratory function*
- *Maintain skin integrity*

GLUCOCORTICOIDS

Benefits of long-term glucocorticoids may include:

Loss of ambulation at a later stage

Preserved upper limb function

Preserved respiratory function

Avoidance of scoliosis surgery

There is currently no consensus about which glucocorticoids are best and at which doses[1]

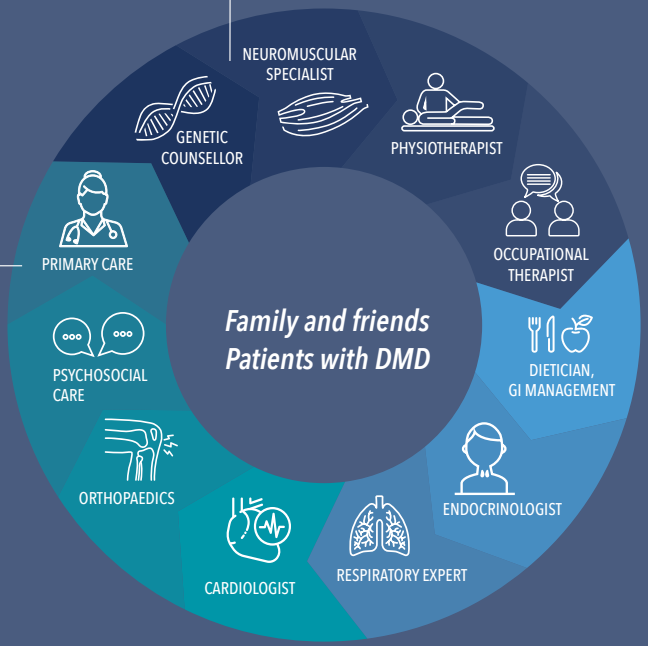
Adapted from reference 1.

Side effects of glucocorticoids can include weight gain and obesity, acne and warts, cushingoid features, growth retardation and delayed puberty, cataracts, immune/adrenal suppression, glucose intolerance, hypertension, adverse behavioural changes, gastro-oesophageal reflux, peptic ulcer, gastritis, osteoporosis and myoglobinuria[4,5]

DUCHENNE REQUIRES A MULTIDISCIPLINARY APPROACH

will serve as the lead clinician

is key to diagnosis, and will coordinate care with appropriate specialists



Adapted from references 1–3.

With multidisciplinary care the survival of patients with Duchenne has improved,[1] with patients now living into their 30s and beyond[3]

Adapted from reference 1.

References: **1.** Birnkrant DJ, *et al. Lancet Neurol.* 2018;17:251–267. **2.** Birnkrant DJ, *et al. Lancet Neurol.* 2018;17:347–361. **3.** Birnkrant DJ, *et al. Lancet Neurol.* 2018;17:445–455. **4.** Nascimento Osorio A, *et al. Neurologia.* 2018; DOI: 10.1016/j.nrl.2018.01.001 [Epub ahead of print]. **5.** Bushby K *et al. Lancet Neurol* 2010;9:77–93.

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