# Scottish Muscle Network

# Duchenne Muscular Dystrophy (DMD) “at a glance” sheet

**Diagnosis/Genetics**

- X-Linked Recessive
- Dystrophin, an essential protein in the muscle cell membrane, is usually absent
- Diagnosis must have molecular confirmation
- Genetic counselling mandatory for affected individual and female relatives at risk of being carriers

**Clinical features and outlook**

DMD affects males, and causes progressive muscle weakness. Loss of ambulation is inevitable in the teens or twenties, with timing partly defendant upon management. DMD is a life limiting condition, although with appropriate cardiac and respiratory support young men may live to the fifth decade**.**

**Associated features and anaesthetic risk**

Anaesthesia presents a significant risk and cardio-respiratory review must occur prior to surgery when possible. There is additional risk of malignant hyperpyrexia like reactions. Sedative analgesia should be prescribed with careful monitoring. Anaesthesia should be undertaken by experienced teams.

**Cardiac manifestations**

DMD is associated with cardiomyopathy. Early treatment for deteriorating cardiac function is recommended. Formal cardiac surveillance is mandatory.

**Respiratory manifestations**

Respiratory decompensation will always occur and all patients should be under review by an informed respiratory physician. Cough can become weaker and equipment such as a cough assist machine can help to clear excessive mucus production.

**Gastrointestinal**

In some, swallowing function can become weaker so that patients find it difficult to complete meals or eat solid foot and keep up with their daily nutritional intake. In those who lose a significant amount of weight, this can cause significant problems with fatigue. We refer some patients for a gastrostomy to prevent further weight loss and to ensure that patients keep up with their daily recommended calorie intake.

**Treatments**

Steroid therapy is recommended for all although there can be side effects including osteoporosis and fractures. Monitoring of bone health is carried out by the endocrine team. All patients should be under the care of a specialist team so that the role of disease modifying drugs can be discussed. Spinal surgery should be considered for progressive scoliosis on loss of ambulation. A range of specialist equipment is available to provide postural support and assist independent living.

**All patients should carry an Alert bracelet or similar. Patients who are receiving steroids should also carry a steroid card.**

**Additional information**

www.smn.scot.nhs.uk
www.musculardystrophyuk.org
https://dmdcareuk.org/
https://dmdhub.org/
https://treat-nmd.org
www.neuromuscular.wustl.edu/index.html

*NOTE*

*This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined on the basis of all clinical data available for an individual case and are subject to change as scientific knowledge and technology advance and patterns of care evolve. Adherence to guideline recommendations will not ensure a successful outcome in every case, nor should they be construed as including all proper methods of care or excluding other acceptable methods of care aimed at the same results. The ultimate judgement must be made by the appropriate healthcare professional(s) responsible for clinical decisions regarding a particular clinical procedure or treatment plan. This judgement should only be arrived at following discussion of the options with the patient, covering the diagnostic and treatment choices available. It is advised, however, that significant departures from the national guideline or any local guidelines derived from it should be fully documented in the patient's case notes at the time the relevant decision is taken.*