Scottish Muscle Network

Manifesting carriers of Duchenne and Becker muscular dystrophy “at a glance” sheet

Diagnosis/Genetics

- X-Linked Recessive
- Dystrophin, an essential protein in the muscle cell membrane, is altered in quality or quantity
- Diagnosis must have molecular confirmation
- Genetic counselling mandatory- some female relatives may be carriers and some male relatives (including seemingly asymptomatic ones) may be affected

Clinical features and outlook

Duchenne and Becker muscular dystrophy affects males causing progressive muscle weakness. The majority of female carriers do not have any signs of the condition at all but a proportion of (usually <20%) manifesting carriers can have muscle problems than can range from mild to occasionally as severe as males with Duchenne muscular dystrophy.

Associated features and anaesthetic risk

Anaesthesia may present a risk especially if the patient is severely affected. There is additional risk of ’Malignant Hyperpyrexia-like’ reactions. Anaesthesia should be undertaken by experienced teams.

Cardiac manifestations

Cardiomyopathy may be the only clinical feature in a manifesting female carrier and affected males. Early treatment for deteriorating cardiac function is recommended. Echocardiogram should be performed at diagnosis and, if normal, every five years thereafter.

Pregnancy

Specialist cardiac evaluation should be considered in all carriers prior to conceiving a pregnancy or as soon as the pregnancy is recognized. This is considered especially important for manifesting carriers. Those with cardiomyopathy should be treated as high risk and monitored by a high risk obstetrician and a cardiologist.

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.