Scottish Muscle Network

Myotonic Dystrophy type 1 “at a glance” sheet

Also known as “dystrophia myotonica” or “Steinert disease”

NOTE

This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined based on 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.

Diagnosis / Genetics

- autosomal dominant inheritance
- diagnosis must have molecular confirmation
- in successive generations of an affected family, symptoms typically start earlier and are more severe than in the previous generation (“anticipation”)
- **all** affected individuals, particularly females, are at risk of having babies with a severe, congenital-onset form
- genetic counselling is advised for all family members

Clinical features and outlook

A multisystem disorder with variable age of onset, severity of symptoms and prognosis. Muscle weakness may result in difficulties with practical tasks and mobility problems. Additional associations include diabetes mellitus, cataract formation, gut motility disorders, sleep-disordered breathing, daytime somnolence, and variable cognitive changes which may include apathy or avoidant-type personalities. **Affected individuals are at a substantially increased risk of life-threatening events including cardiac arrhythmias, respiratory issues and anaesthetic complications.** Involvement from various AHPs is important for the management of myotonic dystrophy type 1 including physiotherapy, OT, orthotics and speech and language therapy.

Anaesthetic risks

Anaesthesia and sedation present a **major risk** and should be performed with full cardiac and respiratory review. Given the risk of anaesthesia, elective procedures should be undertaken with identified HDU and ITU bed availability. Regional or local anaesthesia may be preferable. **Opiates** and **sedatives** should be avoided where possible and if administered the patient should be carefully monitored. Patients should be encouraged to carry an alert card, bracelet or device.

Cardiac manifestations

Cardiac arrhythmias are common, and pacing is often required. There is also an increased risk of dilated cardiomyopathy. An echocardiogram at diagnosis, and yearly ECG as a minimum is recommended with early referral to cardiology if abnormal. Palpitations or syncope should be promptly investigated.

Respiratory manifestations

Respiratory failure is common. Dysphagia can predispose to aspiration pneumonia. Regular symptom review for respiratory muscle weakness or evidence of sleep-disordered breathing is important and the use of nocturnal non-invasive ventilation may be indicated.

Cognitive manifestations

Some patients may have learning difficulties, behavioural problems or autistic traits. It would be important to address them accordingly.

Further guidance for management of patients with myotonic dystrophy, including anaesthetic guidelines can be viewed at

www.nn.nhs.scot/smn/

Sources of additional information:

www.musculardystrophyuk.org
https://neuromuscular.wustl.edu/