Scottish Muscle Network

Mitochondrial disease “at a glance” sheet – for adults

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

- mutations are within mitochondrial DNA, less common nuclear DNA mutations also occur
- inheritance maternal, autosomal dominant or recessive or sporadic
- formal genetic counselling mandatory

Clinical features and outlook

A multisystem disorder with variable features. Commonly patients have a myopathy and complain of exercise intolerance. They are at risk of developing migraine, epilepsy, lactic acidosis (aggravated by infection), cardiac manifestations (arrhythmia or cardiomyopathy or cardiac failure), respiratory muscle weakness, deafness, and visual problems (including ptosis, progressive external ophthalmoplegia and blindness). Other features in adults include neuropathy and ataxia, gastrointestinal dysmotility, malabsorption and low BMI. Rarely patients present with bouts of pseudo intestinal obstruction. They may develop various endocrine problems with diabetes being the commonest but also thyroid disease, reduced fertility, premature menopause and hypopituitarism. Bone density may be low, and they may be at risk of fractures. There may be renal and liver involvement. Some are at high risk of developing refractory epilepsy and stroke-like events. These are not true strokes but are episodes due to refractory focal epileptic seizures. They may be difficult to recognize since they do not present in a conventional way. Psychiatric manifestations, learning and behavioural difficulties may be present and cognitive disturbance are not infrequent.

Associated features and anaesthetic risks

Anaesthesia presents a major risk and should be performed with full cardiac and respiratory review. There is a risk of Malignant Hyperthermia-like reactions and adverse reactions to Propofol. Sodium Valproate should be avoided where possible and is absolutely contraindicated specifically in POLG-associated mitochondrial disorders.

Cardiac manifestations

Cardiac follow up is important with yearly ECG. Cardiac disease can occur even when skeletal muscle weakness is minimal. Echocardiography is important to monitor for cardiomyopathy. Yearly ECHO initially is recommended but if stable then 3-yearly would suffice. Some patients may also be at risk of autonomic failure.

Respiratory manifestations

Respiratory failure may occur, and patients should be referred accordingly.

Patients should carry an alert bracelet or similar.

Sources of additional information:

www.nn.nhs.scot/smn/
www.musculardystrophyuk.org
https://neuromuscular.wustl.edu/
www.thelilyfoundation.org.uk
Care Guidelines - Rare Mitochondrial Disorders Service (mitochondrialdisease.nhs.uk)
General information about mitochondrial disease – Wellcome Trust Centre for Mitochondrial Research (newcastle-mitochondria.com)