Scottish Muscle Network

Scottish guideline for management of Mitochondrial Disorders

Patients with mitochondrial disease typically have a multisystem disorder. This guideline suggests minimum surveillance to be performed at review appointments, and referral triggers.

Neurology
At each review, enquiry should be made regarding the possible development of ataxia, deafness, seizures, muscle weakness or pain, migraine, hallucinations, autonomic dysfunction, psychosis, dementia, weight loss, and dysphagia. Patients with any suspicion of these or other neurological symptoms should be referred to a neurologist.

Drugs
Presence of mitochondrial disease should be considered when prescribing. The following agents should be considered in this context and likely avoided: metformin, sodium valproate, aminoglycosides, zidovudine. Monitor CK if starting statins. There is limited evidence supporting the prescription of Coenzyme Q and riboflavin and this should only be prescribed by a neurologist.

Endocrine
Annual HbA1c. If > 6.5% refer to Diabetologist.

Cardiology
ECG annually
Echo annually (three yearly if normal after 3 years *)
Patients with abnormal ECG or echo or concern over cardiac symptoms, should be referred to a cardiologist, preferably with an interest in inherited cardiac conditions.

Visual
At each review, enquiry should be made regarding visual acuity. If vision is deteriorating, patients should be referred to ophthalmology, in addition to conventional diabetic retinopathy screening.

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Respiratory
Patients with limb weakness should be referred to a respiratory physician, ideally with an interest in muscle disorders, at the point of diagnosis.
At each review, enquiry should be made of appropriate features of aspiration, recurrent infection and sleep disordered breathing. Any symptoms should prompt referral to a respiratory physician, ideally with an interest in muscle disorders.

Nephropathy
Annual U&E's, eGFR

Gastrointestinal
At each review, BMI should be assessed. Enquiry should be made about nutrition and bowel habit. The importance of regular meals and adequate hydration should be emphasised. There should be a low threshold for referral to dietetics. Enquire about bowel habits and any events that may be suggestive of intestinal pseudo-obstruction or malabsorption. It is important to recognise pseudo-obstruction since the management is conservative rather than surgical. Identifying patients at risk and informing patients and professionals within the respective GI teams is paramount.

Pre-pregnancy
All women considering a pregnancy should be referred for pre-conception counselling, ideally to a diabetic-obstetric clinic for review of anti-epileptic and other medication, discussion of high dose folic acid and with addition of diabetes counselling for women with pre-existing diabetes.

Pregnancy
All women should be offered high dose folic acid (5mg/day) in pre and early pregnancy and referred to an obstetric clinic, ideally a diabetic-obstetric clinic, where consideration should be given to cardiac and respiratory screening as well as anaesthetic review.
Women not known to have diabetes should be offered screening in pregnancy for pre-existing diabetes and gestational diabetes. The screening programme for women with a "risk factor" in SIGN 116 should be offered for those with high risk mutations (mt.3243 A>G, mt.14709T>C, single mtDNA mutations).

Genetics
All patients should be referred to the Genetic clinic at the point of diagnosis for genetic counselling and to allow cascade testing of relatives. Clinicians managing diagnosed patients should ensure that a Genetic referral has been offered.

For further information see www.mitochondrialncg.nhs.uk

*As per NCG recommendation

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.