

Newcastle Mitochondrial Centre

At a glance guidelines:

Respiratory Involvement in Adult Mitochondrial Disease

For full guideline visit:

<http://www.newcastle-mitochondria.com/service/patient-care-guidelines/>

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There are many different forms of mitochondrial disease, varying greatly in their genetic basis, clinical presentation, progression and prognosis. We recommend referral to a specialist mitochondrial centre (www.mitochondrialncg.nhs.uk or see appendix for international centres).

Many have the potential for respiratory muscle involvement and this may develop in the absence of symptoms. We therefore recommend the following:

1. All patients should be offered a Forced Vital Capacity (FVC) and Forced Expiratory Volume (FEV1) measurements (in both the erect and supine position) following initial diagnosis.
2. The FVC and FEV1 should be repeated annually in patients with known respiratory impairment or a clinically detectable myopathy. This interval should only be extended after discussion with a specialist.
3. Symptoms of nocturnal hypoventilation or obstructive sleep apnoea should be actively sought.
4. Contributory factors such as aspiration of food should be considered.
5. All patients with respiratory impairment should have other causes excluded and referral to a specialist in respiratory medicine arranged.
6. All patients due to undergo significant surgery or a general anaesthetic should have their diagnosis highlighted to the anaesthetist. Recent respiratory investigations (as above) should be available for review.
7. Annual immunisations for influenza are indicated.