Newcastle Mitochondrial Centre

At a glance guidelines:

Ocular Involvement in Adult Mitochondrial Disease

For full guideline visit
http://www.mitochondrialncg.nhs.uk/newcastle_guidelines.html

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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 for appropriate counselling and guidance (www.mitochondrialncg.nhs.uk or see appendix for international centres).
Most have the potential for ocular involvement. This includes ptosis, diplopia, cataracts and retinal or optic nerve disease. We therefore recommend the following:

1. All patients should have testing of their Snellen visual acuities at diagnosis and at subsequent visits.
2. Fundoscopy with a direct ophthalmoscope should be performed at diagnosis and at subsequent visits.
3. Visual field testing should be assessed in the clinic by confrontation where there is a history of seizures, encephalopathy, or stroke-like episodes.
4. Carriers of Leber's Hereditary Optic Neuropathy (LHON) should be advised not to smoke and to avoid excessive alcohol consumption.
5. Patients with mitochondrial disease should be referred for a comprehensive neuro-ophthalmological assessment if they report:

(i) significant visual loss at first presentation or progressive visual failure during follow-up.
(ii) diplopia (± limitation of eye movements)
(iii) functionally disabling ptosis (e.g. severe enough to occlude the patient's visual axis) or ptosis causing a significant aesthetic impact.