At a glance guidelines:

Screening & Surveillance in Children with Mitochondrial Disease

For full guideline visit:

http://www.mitochondrialdisease.nhs.uk/professional-area/care-guidelines

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Mitochondrial diseases are a diverse group of conditions presenting in many different ways and at varying ages. The genetics, clinical features, progression and prognosis are equally variable. We recommend referral to a specialist mitochondrial centre in all cases www.mitochondrialdisease.nhs.uk

Prevention is better than cure, but specific measures may be required when complications develop. We therefore advise / recommend the following surveillance in all children with confirmed, or suspected, mitochondrial disease:

**Growth:**

- All children should have their height (or length) and weight measured following initial diagnosis and at 6 monthly intervals thereafter
- Height and weight should be plotted on a WHO-UK growth chart and centiles recorded
- Growth faltering should be addressed early
- Children exhibiting growth faltering should have their height and weight recorded every 3 months, with any interventions made noted on the growth chart
- Referral to a paediatric dietician is indicated for all children below 2\(^{nd}\) centile for weight / height or low BMI

**Blood tests:**

- All children should have blood tests performed following initial diagnosis (if not done as part of their work-up) and at intervals thereafter (timing dependent on clinical status, symptoms or indicated earlier by other professionals involved in the child’s care)
• Blood tests include: FBC, U&Es, LFTs, bone profile, CK, lactate, FGF-21, HbA1c, random glucose
• Additional blood tests may be required depending upon the child’s clinical status and associated conditions
• Blood taking should be performed by a Paediatric phlebotomist, Paediatrician, or nurse experienced in paediatric phlebotomy
• Cryogesic spray or local anaesthetic cream (e.g. Ametop) should be offered to all children having blood tests
• Support with a play specialist should be considered before and during the procedure

Cardiac:

• All children should have an ECG and transthoracic ECHO following initial diagnosis (if not done as part of their work-up), unless already under a Paediatric Cardiologist
• The ECG and ECHO should be repeated annually for 3 years after diagnosis. After that time, if the ECHO remains normal then the interval can be extended to 2-3 yearly. For most patients ECG should be performed at the same time interval as ECHO but in those patients with a higher risk of rhythm disturbance such as Wolff-Parkinson White (m.3243A>G and m.8344A>G) or cardiac conduction block (large-scale single deletion of mtDNA) annual ECG is recommended. Timing for surveillance should be agreed with local Paediatric Cardiology services.
• If the child harbours a cardiac-specific mutation (e.g. m.4300A>G, ACAD9, TAZ) then the interval for cardiac screening should be reduced to annually

Audiology:
• All children should have audiological testing following initial diagnosis (if not done as part of their work-up), unless already under an audiologist

• Audiology should be performed at 5 yearly intervals, unless the child is symptomatic

• Audiology should be repeated if the child suffers from recurrent or persistent tinnitus, or hearing impairment is suspected by a parent or carer

• Children known to harbour the m.1555A>G mutation should avoid aminoglycoside antibiotics (e.g. gentamicin) and should have audiology performed if they have been exposed to these antibiotics

**Ophthalmology:**

• All children should have their visual acuity tested and fundoscopy performed (with a direct ophthalmoscope) following initial diagnosis (if not done as part of their work-up)

• Visual acuity and fundoscopy should be performed annually, unless there is a history of visual deterioration as reported by the child, or suspected by a parent or carer

• Children should be assessed by a Paediatric Orthoptist if aged <5 years, there is restriction in eye movements or squint evident

• All children with ptosis, limitation of eye movements or significant visual impairment should be assessed by an Ophthalmologist, ideally with experience in mitochondrial diseases

**Respiratory:**

• Spirometry should be performed in all children with suspected respiratory impairment, clinical myopathy, or history of aspiration
• Symptoms of nocturnal hypoventilation or obstructive sleep apnoea should be actively sought
• Referral to a Paediatric Respiratory physician for further assessment is advised
• Annual influenza and pneumococcal vaccines are advised

**Other:**

• Routine electroencephalogram (EEG) are not advised unless seizures have been reported
• Routine nerve conduction studies are not advised unless there is a history of consistent with peripheral neuropathy, or reduced/absent reflexes on clinical examination
• Routine MRI of brain, spine or muscles is not indicated and should be requested only upon the basis of clinical findings
• Speech and language therapy assessment should be requested if there is a history of dysphagia, choking on food or aspiration, dysphasia, dysarthria, dysphonia or speech fatigue
References


Nesbitt V, Pitceathly RDS, Turnbull DM et al. The UK MRC Mitochondrial Disease Patient Cohort Study: clinical phenotypes associated with the m.3243A>G mutation – implications for diagnosis and management. J Neurol Neurosurg Psychiatry 2012; 00: 1-3


Pfeffer G, Majamaa K, Turnbull DM, Thorburn D and Chinnery PF. Treatment for mitochondrial disorders. Cochrane Database of Systematic Reviews 2012 (4)