NHS Highly Specialised Services for Rare Mitochondrial Disorders

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 2nd 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

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