At a glance guidelines:

Constipation in Children with Mitochondrial Disease

For full guideline visit:

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

First published October 2017
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. Constipation is a common feature in childhood mitochondrial disease and can result in significant morbidity. We recommend referral to a specialist mitochondrial centre in all cases [www.mitochondrialdisease.nhs.uk](http://www.mitochondrialdisease.nhs.uk)

Prevention is better than cure, but specific measures may be required when complications develop. We therefore advise / recommend the following:

1. Specialist opinion should be sought from a Paediatric Gastroenterologist, in conjunction with a Mitochondrial Specialist, if symptoms are recurrent or difficult to treat

2. **Diet & lifestyle**
   - Educate children and families about the importance of good bowel hygiene
   - Aim for passage of a formed soft stool at least once daily
   - Ensure an adequate daily fluid intake, ideally water, appropriate for age, body-weight, and any associated conditions (e.g. renal or cardiac)
   - Bulk-forming drugs or diet may worsen constipation in mitochondrial disease; low fibre diets should be considered
   - Immobility can contribute to constipation therefore regular physical activity (or changes in posture and positioning for those with severe physical disability) is recommended
   - Numerous medications have constipation as an associated side-effect; risk/benefit and alternatives should be considered

3. **Investigations**
   - Screening for hypothyroidism should occur in all children with mitochondrial disease and constipation
   - Screening for coeliac disease should be considered
   - Other contributory causes, such as electrolyte imbalances, should be considered
• Constipation does not need to be routinely investigated with abdominal X-rays, ultrasound, transit studies or endoscopy
• AXR may be helpful in children with mitochondrial disease and constipation displaying gastroenterological symptoms and prevent unnecessary surgical intervention in the future

4. **Laxative maintenance therapy**
• Maintenance therapy should only be commenced once faecal impaction has been excluded
• Paediatric polyethylene glycol 3350+electrolytes maintenance should be started according to age and if previous disimpaction has occurred; usually daily dosage is ½ - 4 sachets
• The dose of polyethylene glycol 3350+electrolytes should be adjusted according to response, aiming for the daily passage of soft formed stools
• If polyethylene glycol 3350+electrolytes is not tolerated then a stimulant laxative such as sodium picosulphate or senna should be commenced; docusate should be added if stools are hard
• Children should be reassessed to ensure impaction has not (re)occurred
• Most children with constipation due to mitochondrial disease will require laxative therapy throughout childhood and likely into adult life
• Medication should not be abruptly stopped
• Opinion from a Paediatric Gastroenterologist should be sought in cases of intractable constipation despite optimum diet, lifestyle and laxatives

5. **Faecal impaction**
• Faecal impaction (severe constipation) may present as ‘overflow diarrhoea’ and needs urgent management
• **Disimpaction regime**
  o Child aged <1yr: Paediatric polyethylene glycol 3350+electrolytes ½ sachet/day increasing to 1 sachet/day
  o Child aged 1-5yrs: Paediatric polyethylene glycol 3350+electrolytes 2 sachets/day increasing in steps of 2 sachets every 2 days up to a maximum of 8 sachets daily
Child aged 5-12yrs: Paediatric polyethylene glycol 3350+electrolytes 4 sachets/day increasing in steps of 2 sachets/day up to a maximum of 12 sachets daily

The aim is to pass solid stools initially progressing to loose then watery stools; once watery stools have been passed, maintenance therapy can commence

6. **Surgical management**
   - Surgical intervention is rarely required
   - **Rectal biopsy**
     - Not required unless clinical features of Hirschsprung’s disease are present, or required for investigation of another condition
   - **Manual evacuation of the bowel under general anaesthesia**
     - Should not be performed unless optimum treatment with enteral laxative medication (+/- rectal medication) where appropriate has failed
     - Discussion with the Paediatric Anaesthetist and consideration as to appropriate anaesthesia will be required if manual evacuation is planned
     - Close monitoring of serum glucose and electrolytes will be required
     - See Perioperative Care in Children with Mitochondrial Disease guideline if surgery planned
   - **Antegrade colonic enema procedure**
     - Not generally advised in children with metabolic disorders due to a lack of evidence
     - Children with intractable constipation despite optimum management should be referred to Paediatric Gastroenterologist before considering an ACE procedure
     - If an ACE procedure is planned, appropriate support and follow-up must be in place from those with expertise in managing children who have had an ACE procedure

7. **Complications**
• Urinary retention, or incomplete urinary voiding, can occur in children with severe constipation and should be sought in the history and on clinical examination (palpable bladder)
• Consider performing an abdominal USS if concerns regarding retention
• Some children may develop urinary tract infections as a consequence of chronic constipation and should be treated promptly
• Urinary microscopy and culutre should be performed in constipated children with mitochondrial disease presenting unwell, febrile, vomiting or with abdominal pain
• Upper or lower GI obstruction can occur and needs urgent management

8. **If a child presents acutely with signs of mechanical obstruction:**
   a. Keep the child nil by mouth and commence IV fluids (0.9% saline + 5% glucose)
   b. Consider passing a nasogastric tube +/- flatus tube
   c. Perform a plain abdominal X-ray
   d. Serum glucose, electrolytes and lactate should be checked regularly
   e. Consider TPN if prolonged fasting is likely
   f. Faceal impaction should be treated accordingly
   g. Early advice from a Paediatric Gastroenterologist / Paediatric surgeon is advised
References


NHS Choices Constipation in young children


Gordon M, MacDonald JK, Parker CE, Akobeng AK and Thomas AG. Osmotic and stimulant laxatives for the management of childhood constipation. Cochrane Database of Systematic Reviews 2016, Issue 8. Art No.: CD009118
