NHS Highly Specialised Services for Rare Mitochondrial Disorders

At a glance guidelines:

Constipation 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. 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

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*
    - Child aged <1yr: Paediatric polyethylene glycol 3350+electrolytes ½ sachet/day increasing to 1 sachet/day
    - 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

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