

Newcastle Mitochondrial Disease Guidelines

At a glance guidelines:

Gastrointestinal Involvement in Adult Mitochondrial Disease

For full guideline visit:

<http://www.newcastle-mitochondria.com/service/patient-care-guidelines/>

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There are many different forms of mitochondrial disease, varying greatly in their genetic basis, clinical presentation, progression and prognosis. Gastrointestinal involvement is easily overlooked but can result in major morbidity and mortality. This is most commonly seen in disease due to the m.3243A>G mutation, but occasionally in other forms of disease including MNGIE. Both upper GI and lower GI presentations occur. We recommend referral to a specialist mitochondrial centre (www.newcastle-mitochondria.com).

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

1. Early satiety, anorexia, weight loss, constipation and symptoms mimicking irritable bowel syndrome may be due to mitochondrial disease
2. Upper and lower GI symptoms should be screened for regularly and treated promptly. Aim for normal bowel motions once per day. This is especially important if any form of elective surgery is planned.
3. Low residue diet (bulk forming drugs and/or diet may worsen constipation in mitochondrial disease)
4. Be aware that faecal impaction may present as 'overflow diarrhoea' and needs exclusion
5. High dose macrogols (osmotic laxatives) or bowel cleansing solutions may be required for stubborn faecal impaction.
6. Specialist opinion should be sought if symptoms are recurrent or difficult to treat.

7. Acute Presentations – upper GI (mimicking mechanical obstruction): Prompt recognition of clinical symptoms and radiological findings is crucial so that drainage of the stomach and small bowel content can be achieved with the insertion of *wide-bore* nasogastric (NG) tube ('Drip and suck'). Prominence of vomiting, particularly projectile vomiting or vomiting of bilious or faecal matter in the setting of a distended abdomen following oral intake should alert the clinician to this as a possible serious acute complication of CIPO. This can be particularly dangerous in patients unable to adequately protect their own airway – such as those with encephalopathy, seizures, or bulbar dysfunction. Patients should be kept nil by mouth and enteral feeding is contra-indicated until the pseudo-obstruction has resolved. IV fluids (with avoidance of Ringers lactate solution) should be commenced to provide adequate fluid intake. Total parenteral nutrition (TPN) should be considered early if prolonged fasting is likely.

8. Acute Presentations – Lower GI (mimicking mechanical obstruction): Surgical intervention should be a last resort and is associated with high mortality rates. Prior discussion with a mitochondrial specialist is strongly advised. Distended bowel loops without other features of an 'acute abdomen' usually respond to conservative measures (see full guideline). Baseline lactate levels may be raised in m.3243A>G patients and should be interpreted with caution. Be aware that upper **and** lower GI dysmotility usually coexist.

Appendix A:

Management of chronic upper and lower gastrointestinal symptoms in mitochondrial disease

Clinical Assessment

Upper GI symptoms

- Bloating and abdominal pain
- Gastric reflux
- Decreased appetite
- Early satiety and/or post-prandial nausea/vomiting

Consider Gastroparesis

- Review diet/medications
- Bloods & AXR
- Refer to gastroenterologist for consideration of scintigraphy and/or radio-opaque markers

Treatment options

- Implement low fibre diet
- Consider prokinetic agent (eg. erythromycin)
- Parenteral or Enteral feeding (preferably jejunostomy) may be considered if refractory symptoms/weight loss

Lower GI symptoms

- Bloating and abdominal pain
- Difficult/infrequent defaecation
- Abnormal Bristol Stool Chart
- Clinical evidence of faecal loading

Consider Constipation

- Review diet/medications
- Bloods & AXR

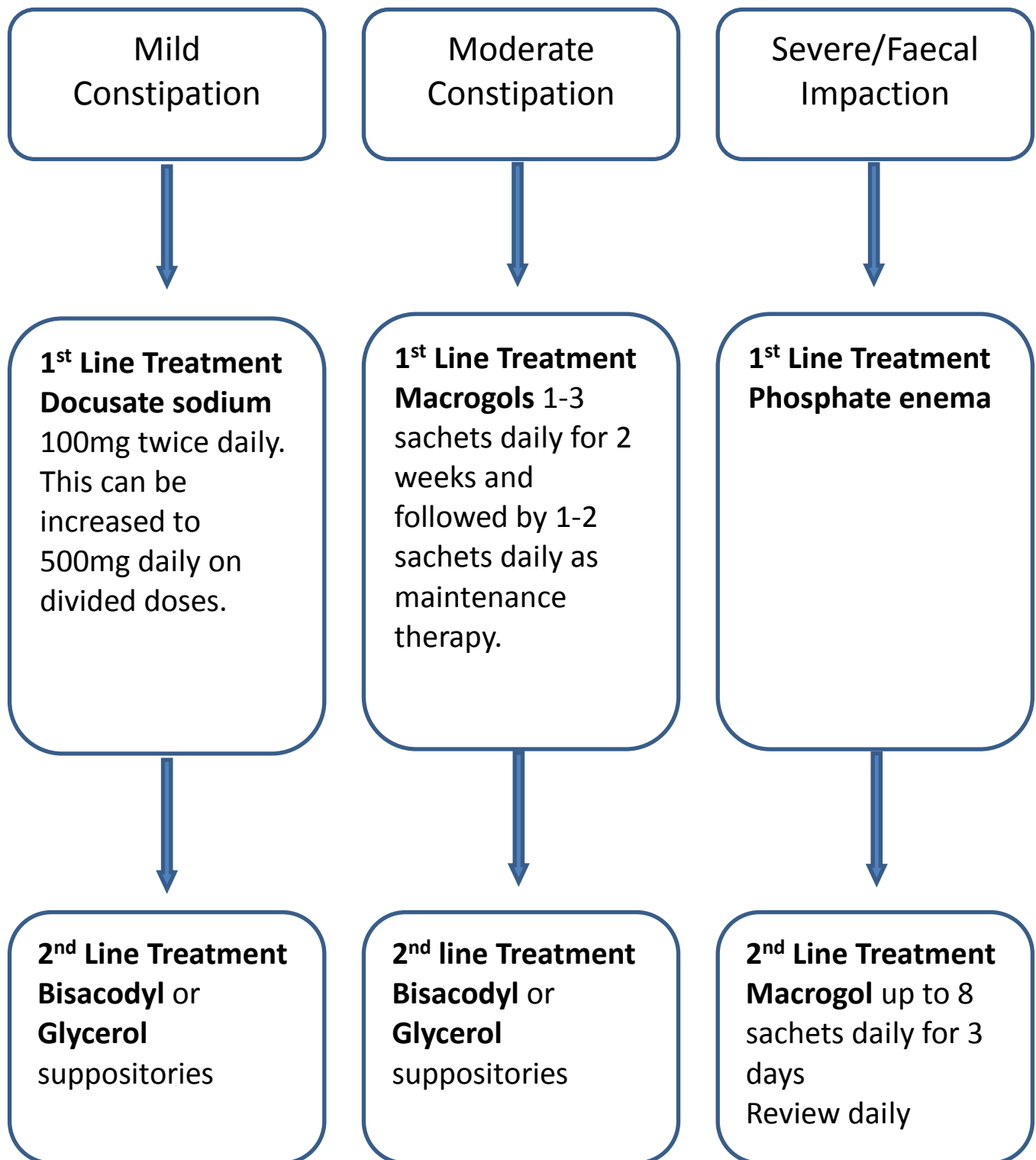
Treatment options

- Implement low fibre diet
- See Appendix B for treatment of:
 - Mild constipation
 - Moderate constipation
 - Severe constipation/Faecal impaction/ loading

Treatment Failures

- Refer to Mitochondrial Centre
- Refer to gastroenterologist
- Consider colonic irrigation
- Consider other newer agents – e.g. prucalopride, lubiprostone

Appendix B: Treatment of Constipation



NOTE: 1. Adopt low-fibre diet. Avoid bulk-forming laxatives e.g. Fybogel
2. All medications mentioned above should only be administered as prescribed.