

DEVELOPING THIS CHECKLIST

These recommendations have been developed according to the process recommended by the National Health and Medical Research Council of Australia.

A staged method was employed:

- comprehensive literature search to identify relevant information
- consultation with parents and caregivers on their experiences of poor growth, calorie intake and feeding difficulties
- preparation of a draft set of recommendations
- appointment of an international and multidisciplinary panel of expert clinicians who reviewed successive drafts of the recommendations until agreement was reached.

Expert Panel

Members of the expert panel who reviewed the earlier drafts are listed below and we thank them for their contribution.

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References

This checklist is based on the following:

Baikie G, Ravikumara M, Downs J, Naseem N, Wong K, Percy A, Lane J, Weiss B, Ellaway C, Bathgate K, Leonard H. Guidance in the management of gastroesophageal reflux, constipation and abdominal bloating in Rett syndrome. 10.1097/MPG.0000000000000200.

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Gastro-intestinal disorders in Rett syndrome Checklist for clinicians on assessment and management

Rett syndrome is a rare neuro-developmental disorder that occurs almost exclusively in females. The prevalence rate is approximately 1:9000 of the female population.

Rett syndrome is caused by mutations in the methyl-CpG-binding protein 2 (*MECP2*) gene which affects the nerves in the central nervous system. This affects many systems of the body:

- the **nervous system** (cognitive impairment, communication difficulties, and epilepsy)
- the **respiratory system** (hyperventilation and breath holding)
- the **musculo-skeletal system** (altered muscle tone, contractures, low bone density and scoliosis)
- the **gastrointestinal system** (gastro-oesophageal reflux, gastrointestinal dysmotility, abdominal distension, constipation and oromotor dysfunction).

The multiple impacts of Rett syndrome have major health and lifestyle implications, both for the girls and women affected and for their family carers.

This leaflet is one of a series of publications developed by the Telethon Institute for Child Health Research in Western Australia to support better clinical assessment and management of health conditions commonly associated with Rett syndrome.

The content reflects issues cited by parent carers as important to them in responding to their daughter's gastro-intestinal problems.

Other publications currently available in this series focus on clinical assessment and management of:

- *scoliosis in Rett syndrome*
- *nutrition and growth in Rett syndrome.*

A more comprehensive booklet on nutritional and digestive health in Rett syndrome has also been developed for families and carers and is available from rett.childhealthresearch.org.au

Many females with Rett syndrome experience gastro-intestinal problems that may inhibit the healthy enjoyment of food and/or limit nutritional intake. This brochure provides an overview of recommended practice in the clinical assessment and management of three of the most common gastro-intestinal problems associated with Rett syndrome:

- gastro-oesophageal reflux disease (GERD)
- constipation
- abdominal bloating.

REFLUX

Those with Rett syndrome are vulnerable to GERD, especially if motor function is poor. Reflux is also more likely if the person has scoliosis (commonly associated with Rett syndrome) or spends a lot of time in the supine position.

Assessment

The following signs may indicate gastroesophageal reflux:

- regurgitation, sour smelling burps or vomiting
- dental erosion
- unexplained weight loss
- iron deficiency anaemia
- food refusal and/or rumination
- recurrent lower respiratory tract infections
- behaviour problems (including agitation, self harm, screaming, restlessness for no apparent reason).

Investigations

Options to test for reflux include:

- 24 hr oesophageal pH monitoring (with preference for conducting a multiple channel intraluminal impedance study in combination with pH testing)
- upper GI endoscopy to assess whether there is reflux oesophagitis and/or gastritis
- radionucleotide scintigraphy to test for aspiration due to reflux.

NOTE:

A barium study may not be useful in the assessment of reflux.

Management

A trial of conservative strategies is recommended before pharmacological and surgical strategies are considered.

Feeding, postural and behavioural strategies

Options may include:

- providing small, frequent feeds and/or thickened feeds
- creating a calm environment for eating (including the possibility of playing soothing music)
- eliminating selected foods from the diet commonly associated with GERD, and monitoring any impacts
- encouraging an upright position while eating (including the possibility of standing)
- elevating the head of the bed if the person is recumbent.

Pharmacological management of reflux

If more conservative management options are ineffective, pharmacological interventions may be trialled to supplement these strategies.

Proton pump inhibitors (PPIs, e.g. Lansoprazole, Omeprazole, Pantoprazole) are recommended as the drugs of choice.

It is recommended that PPIs initially be trialled once/day increasing to twice daily if symptoms of GERD persist.

Dosage should be maintained at the lowest level possible for effective symptom control.

PPIs can be used over the long term once they have been shown to be clinically effective. However, they may also be associated with diarrhoea, constipation or pain. If these side effects are experienced, an alternative PPI may be trialled, or the dosage of the existing drug reduced.

Other pharmacological options include:

- **H2-blockers or H2-receptor antagonist** reduce acid secretion but are less effectiveness than PPIs

Prokinetics should be used with extreme caution or avoided in girls with Rett syndrome because of their effect in prolonging QT interval. 20% of girls have a prolonged QT as part of Rett syndrome.

- **Domperidone** has a prokinetic action increasing gastrointestinal motility where there is prolonged gastric emptying time and may increase tolerance to feeds.
- **Erythromycin** at sub-antimicrobial doses also has a prokinetic action and may shorten gastric emptying.

Surgical management of reflux

Surgical fundoplication is indicated if reflux is severe despite PPI therapy. Some treatment centres perform a fundoplication at the same time as a gastrostomy.

Nissen fundoplication has also been shown to improve weight gain in some children with Rett syndrome.

A laparoscopic Nissen's fundoplication is recommended if no contraindications exist as feeding goals are achieved more quickly and the duration of the hospital stay is shorter than with an open Nissen's fundoplication.

The effectiveness of the fundoplication needs to be monitored carefully as symptoms may recur. Following a fundoplication girls and women with Rett syndrome may experience abdominal bloating, retching, dysphagia and/or dumping syndrome.

NOTE:

There is no role for a prophylactic surgical anti-reflux therapy.

CONSTIPATION

Constipation is common in Rett syndrome. Females with Rett syndrome have limited communication skills and may not be able to express their discomfort due to constipation.

Assessment

Seek advice from the caregiver on the frequency and consistency of bowel actions.

Signs of constipation may include:

- grimacing or crying with passage of stool
- decreased appetite
- irritability or behaviour changes

Testing for constipation

Physical examination of the abdomen is recommended. An x-ray of the abdomen is not required in a routine investigation for the diagnosis of constipation.

Management

Nutritional and behavioural strategies

Consistent use of strategies designed for each individual is helpful in preventing and managing constipation with particular attention to adequate intake of fluids and fibre, and maintaining mobility and physical activity.

Fluid intake

Fluid requirement for girls with Rett syndrome is generally the same as for the general population but is increased in the presence of hyperventilation and/or drooling. Ensuring adequate fluid intake is an important management strategy.

Fibre intake

Intake of fibre consistent with a healthy diet is recommended from wholegrain foods, fruit and vegetables.

Supplement the diet if necessary eg prune juice or soluble fibre eg Benefibre.

Physical activity

Inactivity can increase the likelihood of constipation. Maintaining mobility and/or physical activity is recommended in the management of constipation.

Toileting routine

Toilet timing to take advantage of the gastro-colic reflex should be encouraged. Where possible females should be encouraged to sit on the toilet within 30 minutes of meals

Enteral feeding can also be supplemented with fibre.

Pharmacological management of constipation

If nutritional and behavioural strategies are not effective alone, pharmacological interventions should be used. The options below are listed in order of preference. More potent options are required if conservative approaches have been trialled and found to be ineffective. Consistent use is important once an effective dose has been found.

- Psyllium (Metamucil, Konsyl) or other high fibre preparations such as Benefibre can be used to increase the bulk of the stool (sufficient fluid intake is essential when psyllium or other high fibre preparations are used)
- Oral osmotic laxative treatments include Lactulose, Polyethylene glycol (Movicol or Miralax) and Milk of Magnesia and will soften the stool
- Lubricant laxatives include paraffin oil and Parachoc and coat the stool to make it pass more easily through the colon. Possible side effects relate to where there may be a risk of aspiration and oil in stools.
- Suppositories are indicated if dietary management and oral laxatives are inadequate
- An enema may be indicated if other strategies have proven ineffective.

NOTE:

- Soapsuds enemas and water enemas are not suitable because of the risk of serious complications There is currently no evidence for the effectiveness of probiotics in the management of constipation.

Surgical management of constipation

If obstructive symptoms develop that are not responsive to medical treatment, manual disimpaction under general anaesthesia may be necessary.

ABDOMINAL BLOATING

Girls and women with Rett syndrome are prone to hyperventilation, breath holding and air swallowing that may contribute to abdominal bloating. Anxiety, pain and stress may exacerbate breathing dysfunction and abdominal bloating.

Assessment

Abdominal bloating may be indicated by:

- caregiver report of abdominal distension and pain
- history of change in abdominal girth
- lesser bloating on waking increasing during the day.

Investigations

Options include:

- observation of breath holding
- abdominal examination and palpation
- video fluoroscopy during feeding to monitor air swallowing.

NOTE:

If bloating is associated with severe pain, tense abdomen and/or signs of shock, urgent medical assessment is required to exclude the rare complication of gastric perforation.

Management

Manage constipation as may exacerbate abdominal bloating.

The action of simethicone in reducing the surface tension of gas bubbles may be effective in reducing abdominal bloating.

Where anxiety is associated with abdominal bloating, serotonin reuptake inhibitors may be considered. However, the potential risk of a prolonged QT interval should be taken into account before prescribing these drugs.

In cases of extremely severe abdominal bloating, a gastrostomy procedure may be considered. The gastrostomy can be used to release the build up of excess air.