Management of gastrointestinal disorders in Rett syndrome

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What is meant by gastrointestinal disorders?

The three most common gastro-intestinal problems affecting girls and women with Rett syndrome are: constipation, Gastro-Oesophageal Reflux Disease or Gastro-Esophageal Reflux Disease (GORD/GERD or reflux) and abdominal bloating.
Constipation occurs where stools are dry, hard and difficult to pass. Constipation can cause considerable discomfort and pain. Reflux occurs when the muscle at the lower end of the oesophagus does not close properly after food has passed through to the stomach. As a result, stomach contents can pass back up the oesophagus which can be painful, and in extreme cases, damage to the oesophagus lining. Acid may also flow back into the oesophagus when the stomach doesn't empty properly. This is known as delayed gastric emptying. Abdominal bloating describes the swelling of the abdomen, accompanied by feelings of tightness, fullness, discomfort and pain.

Why do gastrointestinal disorders occur in Rett syndrome?

As the digestive system operates alongside other systems in the body (the nervous, circulatory, muscular-skeletal, respiratory and endocrine systems), disruptions to any of these systems can affect the healthy operation of the digestive system.

- Reduced intestinal motility, physical activity and some side effects from various medications may cause individuals with Rett syndrome to suffer constipation.
- Reflux is likely to occur in individuals with restricted mobility and/or scoliosis.
- The tendency to hyperventilate, breath hold and swallow air can cause abdominal bloating.

Pain, anxiety and excitement may exacerbate these issues and increase discomfort. This can compromise the healthy enjoyment of food and limit the intake of nutrients, leading to poor growth and bodily functioning.

How common are gastrointestinal disorders in Rett syndrome?

Most individuals with Rett syndrome experience one or more gastro-intestinal problems. Constipation affects around 80% of individuals with Rett syndrome, approximately 40% have reflux and approximately 40% experience abdominal bloating.

How can families manage this at home?

Simple changes to diet and feeding strategies may be sufficient to effectively manage reflux, constipation and abdominal bloating.

For the management of reflux: Changing the diet can be effective, eliminating foods that may trigger reflux (ex. citrus, chocolate, caffeinated drinks, fizzy drinks). Families should eliminate suspected foods one by one and monitor for any improvement. Providing smaller more frequent feeds may increase digestion speed and prevent backflow of stomach acid. Thickening feeds using commercial or food-based thickeners (upon consultation with a doctor or dietician) may prove effective. Posture while eating is also important. An upright position ensures digestive organs are not cramped and gravity can help the correct movement of food, fluids and digestive juices. If feeding needs to occur lying down, then families should elevate the head of the bed. Finally, the feeding environment should cultivate a positive eating experience. Stress can interfere with the normal digestive process so creating a calming and enjoyable environment can be helpful.

For the treatment of constipation: Fibre, fluids, physical activity and routine are all important for family management. The recommended level of daily dietary fibre is ~14-25g depending on age. High fibre foods (eg. whole grain cereals and breads, vegetables, legumes, fruit, and dried fruit) have a natural laxative effect and these should be included in the diet. Maintaining adequate fluids is very important. In cases of frequent hyperventilation or drooling or where laxatives are being taken, fluid intake should be higher to compensate for fluid loss. Maintaining mobility and engaging in some regular physical activity can ease constipation symptoms. Finally, establishing a regular toileting routine sets the body clock for waste elimination. The best toileting time is within 30 minutes of finishing a meal, when faecal movement is increased in response to stomach muscles stretching. Lifestyle changes to manage constipation can also relieve abdominal bloating.

Are there established treatment/management options?

The least invasive option is always considered as the first course of action.

Symptoms of constipation include at least 2 of the following: straining during at least 25% of defecations, lumpy or hard stools in at least 25% of defecations, sensation of incomplete evacuation for at least 25% of defecations, sensation of anorectal obstruction/blockage for at least 25% of defecations, manual manoeuvres to facilitate at least 25% of defecations, and/or fewer than 3 defecations per week.

There are many symptoms that might indicate reflux. These might include: weight loss despite maintaining good appetite (when accompanied by other symptoms), sour breath, burps or belching, regular vomiting, refusing food or repeated chewing with a reluctance to swallow (rumination), coughing after feeding, poor general respiratory health, behavioural problems during or immediately after eating, dental erosion. Diagnosis depends on the overall presentation of these symptoms.

Symptoms of abdominal bloating include: abdominal pain associated with distension in the abdominal area. Physical examination of the abdomen may be necessary and monitoring breathing patterns to determine whether breath holding or air swallowing are contributing factors.

A doctor may request additional tests to confirm a diagnosis of reflux including pH monitoring, or an upper gastro-intestinal endoscopy. For constipation, additional monitoring of bowel movements and abdominal x-rays may be requested.

What are the clinical treatment options?

Some digestive problems can be easily remedied with changes to diet, positioning or the feeding environment (see family management at home section). If simple strategies do not work, then pharmaceutical treatments may help. Medical assessment is necessary to determine if these are indicated.

For constipation, high fibre preparations such as psyllium and Benefibre, so long as fluid intake is sufficient, increase the bulk of the stool. Osmotic laxatives such as Lactulose, Docusate or Polyethylene glycol can be used to produce softer, easier to pass stools. Lubricant laxatives are oil based and coat the stool to make it easier to pass. Suppositories may be helpful. For more severe constipation, an enema can be used to flush liquid into the rectum and colon, causing strong muscle contractions.

For reflux, there are two main categories of drugs. Drugs in the first category decrease the amount of acid produced by the stomach. They include proton pump inhibitors and H2-blockers or H2-receptor antagonists. Drugs in the second category increase the frequency or strength of muscle contractions in the digestive system. These include prokinetics such as domperidone, low dose erythromycin and bethanechol.

If abdominal bloating is due to excess gas, drugs such as Simethicone may help to alleviate symptoms by reducing the surface tension of gas bubbles. If anxiety is a contributor, serotonin reuptake inhibitors may help. However, drug treatments typically have limited effectiveness for abdominal bloating in Rett syndrome.

Sometimes surgery is indicated. For the treatment of reflux, a fundoplication can be useful. The upper part of the stomach is wrapped around the lower end of the oesophagus and stitched into place, preventing backflow of stomach contents. If constipation is severe and the bowel becomes obstructed, the stool can be removed under general anaesthetic. Finally, if abdominal bloating is very severe and causing lots of discomfort, a gastrostomy may release the build-up of air in the stomach.

What follow-up is needed?

A doctor should prescribe the lowest dose for the control of symptoms and any effects should be monitored. If symptoms persist after treatment, or if there is severe abdominal pain and tension, then the digestive problem may not be attributed to RTT and may be indicative of other complications. A thorough assessment by a doctor can diagnose/rule out other possibilities.

References

Baikie G, Madhur R, Downs J, Nasseem N, Wong K, Percy A, Lane J, Weiss B, Ellaway C, Bathgate K, Leonard H. Gastrointestinal dysmotility in Rett syndrome, Journal of Pediatric Gastroenterology and Nutrition. 2014;58(2):237-44.

Mackay J, Downs J, Wong K, Heyworth J, Epstein A, Leonard H. Autonomic breathing abnormalities in Rett syndrome: Caregiver perspectives in an international database study. Journal of Neurodevelopmental Disorders. 2017;9:15.

See also guidelines for gastrointestinal issues at https://rett.telethonkids.org.au/resources/guidelines-and-reports/ .