Managing poor growth and nutrition in Rett syndrome

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What is meant by poor growth?

Good nutrition is important to build stamina, strengthen the immune system and maintain a healthy lifestyle. Nutritional health requires adequate nutrient intake, effective nutrient processing and absorption (digestion), and effective nutrient use (metabolism). Food intake should provide the body with adequate quantities of macronutrients (carbohydrates, proteins, and fats) and micronutrients (vitamins and minerals).

Why does poor growth occur in Rett syndrome?

Poor growth is one of the supportive diagnostic criteria of Rett syndrome (RTT), usually observed as slowing of the rate of head growth, height and weight for age. Difficulties with feeding, swallowing and digestion can make it difficult to provide the body with the necessary nutrition. Mealtimes can be long and stressful. Many families express concern that their child does not drink sufficient fluids, especially when excessive fluid loss may result from conditions such as drooling or hyperventilation. Factors that might affect growth include the following:

- Appetite may be affected by medications used to control other conditions (increased or decreased).
- Oral sensitivity may affect responses to taste, texture or temperature of foods and liquids.
- Chewing requires muscle coordination and some individuals find it difficult to move food with the tongue. Repetitive grinding (bruxism) is common and worn-down teeth are less capable of softening food prior to swallowing.
- Difficulties in swallowing may cause gagging or coughing while eating or drinking and make it more likely for food/liquid to pass into the lungs and for air to pass into the digestive tract. This can increase the risk of chest infections. Altered breathing patterns such as breath holding or hyperventilation also interfere with this process.
- Poor posture can squeeze and cramp the digestive organs. For example, scoliosis may make upright posture difficult.
- Many individuals with RTT are unable to feed themselves and rely on others to provide food/drink.

How common is poor growth in Rett syndrome?

Many individuals with Rett syndrome are underweight for their height and age although a small proportion of are overweight for their height and age. Difficulties with feeding and nutrition occur commonly. Approximately 25% regularly experience coughing when eating or drinking and in Australia about 20% of individuals with Rett syndrome receive some or all nutrients through a tube into the stomach (gastrostomy).

How can families manage this at home?

To improve the nutritional health of those with Rett syndrome, families can consider dietary adjustments or support strategies to reduce feeding difficulties. For example, offering frequent small feeds can enable practice of feeding skills yet make feeding more manageable.

Providing jaw support may facilitate chewing. Modifying food textures can assist with swallowing and reduce chewing effort. Offering favourite foods may increase appetite and motivation to eat. Using special utensils (eg. feeding bottles) may assist capacity for independent feeding. Providing physical and verbal prompts can increase mealtime participation, making it a social and enjoyable experience. To reduce the risk of aspiration, families should monitor breathing patterns and offer food when breathing is steady. Maintaining upright posture and providing support if necessary can ease the passage of food down the digestive tract.

Are there established treatment/management options?

There are a variety of treatment and management options depending on the needs of the individual. Based on assessment findings treatment can be advised through several pathways: increased calorie intake, alterations of diet and feeding strategies, and enteral feeding options.

What are the initial treatment options?

In individuals with low Body Mass Index (BMI) or who are taking some anti-epileptic medications, a doctor may request a blood test to look at micronutrient levels. Eating and drinking difficulties may be assessed by an allied health care professional for feeding/chewing/swallowing ability.

An X-ray procedure called a video fluoroscopy is used to observe the swallowing mechanism and functioning of other organs in the upper part of the digestive tract. This is useful in identifying aspiration and in assessing whether modification to food texture may improve swallowing.

A doctor or dietician can provide advice about a reasonable target weight for those with RTT and growth charts for RTT are now available. If underweight, calorie intake should be increased to help them meet this target. This can be done by supplementing the diet with high calorie nutritious foods (breads, cereals, pasta, dried fruits, dairy foods, legumes, olives, avocados etc.). Age appropriate high calorie nutritional supplements may also be useful but only on the advice of a dietician. Changing diet and feeding practices may also be of assistance (see family management at home section).

If changes to diet and feeding practices do not improve nutritional intake, enteral feeding options may be explored. These deliver nutrients via a tube inserted directly into the stomach, the duodenum or the jejunum. Enteral feeding options may be beneficial to girls who fail to put on weight despite increases in calorie intake, who find it difficult to coordinate the necessary muscles when swallowing, who find feeding/drinking uncomfortable or distressing, or who take unusually long to feed. It can also reduce feeding time while ensuring essential nutrient intake.

There are three main types of enteral feeding. A naso-gastric tube can be inserted down the nose and into the stomach but this is only for short term use. A gastrostomy tube, or G tube, is inserted into the stomach either percutaneously (a Percutaneous Endoscopic Gastrostomy, PEG) or surgically. A gastro-jejunostomy, or PEG-J tube is a feeding tube placed into the jejunum of the small intestine through a gastrostomy or surgically. This latter option is less commonly used.

What follow-up is needed?

Assessment of nutritional and digestive health is recommended once every 6 months for children under 12 years and at least once a year for teenagers and adults. This should include measurement of weight, height and BMI, assessment of oral health, and review of daily diet,

feeding abilities and difficulties. This includes assessing how long it takes to eat, chew and swallow, self-feeding abilities, the effect of new feeding strategies, and blood tests to measure chemical components of blood and observe organ function. As families are most familiar with their daughter, their input provides great insight for doctors and dieticians when prescribing optimal feeding plans and treatments.

When enteral options are being used, monitoring is necessary to observe for complications such as skin irritations at the insertion site, tube leakages, movement of the gastrostomy tube or side effects such as reflux. Any person receiving at least 50% of daily nutritional needs via enteral support should have regular blood tests to check for blood chemistry and micronutrients. Emotional and practical support may be needed by families adjusting to their daughter (or son) receiving nutrition via enteral means.

References

Leonard H, Ravikumara, R, Baikie G, Naseem N, Ellaway C, Percy A, Abraham S, Geerts S, Lane J, Jones M, Bathgate K, Downs J. Assessment and management of nutrition and growth in Rett syndrome. Journal of Pediatric Gastroenterology & Nutrition. 2013;57(4):451-460.
See also guidelines for gastrointestinal issues at https://rett.telethonkids.org.au/resources/guidelines-and-reports/