

Improving health outcomes in Rett syndrome  
**Nutritional and Digestive Health**

An information booklet for families and carers



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### Family Stories:

We asked family carers to contribute their personal stories on how nutritional and digestive issues had impacted on the life of their daughter with Rett syndrome. Twenty-two families from countries around the world offered to share their personal experiences of nutritional and digestive issues with other families living with Rett syndrome.

A special thanks to all the families who contributed their stories and photos. We also thank members of the Australian Rett Syndrome Study Community Reference Group for reviewing the booklet.

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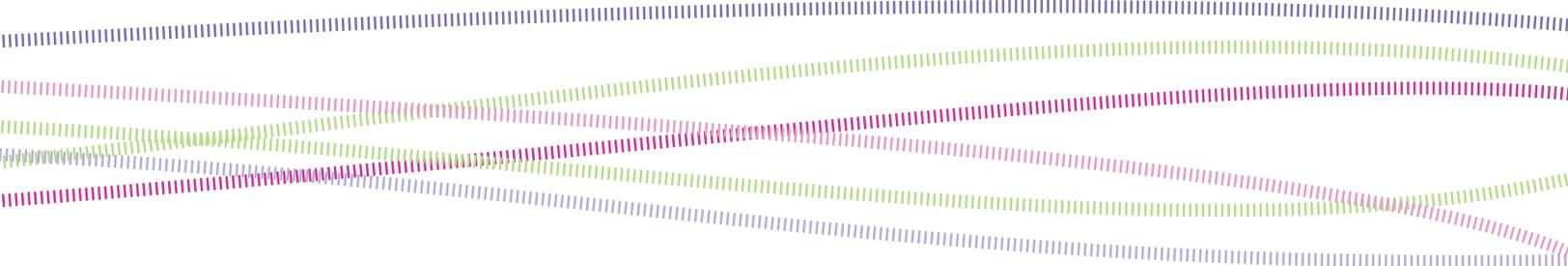
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## About Rett syndrome ...

Rett syndrome occurs almost exclusively in females with a prevalence of approximately 1:9000 female births.

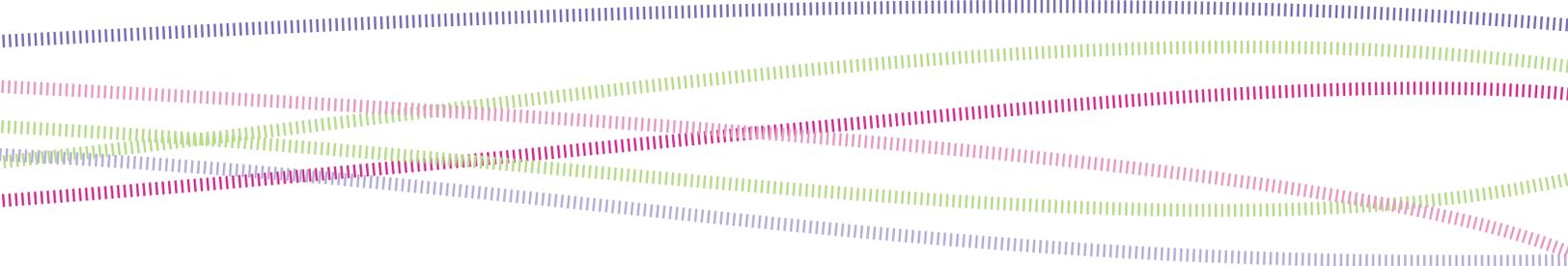
Most girls and women with Rett syndrome have a mutation in the methyl-CpG-binding protein 2 (*MECP2*) gene.

Because the *MECP2* gene affects how nerve cells work, a mutation of this gene can affect the normal functioning of many body systems including the nervous, respiratory, musculoskeletal and digestive systems.

Some of the features of Rett syndrome can limit the body's capacity to take in, absorb, and use the essential nutrients needed for optimal health and functioning.

This booklet provides advice on how nutritional and digestive health can be monitored, managed and improved in girls and women with Rett syndrome.





## This booklet ...

This booklet has been produced by the Rett syndrome study team at the Telethon Institute for Child Health Research, Perth, Western Australia. The aim is to provide a ready reference for families and carers on the best ways of improving the nutritional and digestive health of girls and women with Rett syndrome.

The topics covered in the booklet are drawn from questions families and carers said they wanted answered.

After reviewing the research literature, we sought input from a panel of international experts who provided feedback on how nutrition and digestive health should be assessed and improved in girls and women with Rett syndrome.

This booklet on Nutritional and Digestive Health in Rett syndrome is divided into two parts – the first part deals with nutritional health.

The second part deals with digestive health focusing on how best to manage some common gastro-intestinal problems.

We have sought to make it easier for you to identify which parts of the text will be most useful to you by highlighting key points and data in text boxes and by including summary tables throughout the booklet.

We also asked families and carers to share their personal stories of how they had dealt with any nutritional issues, feeding difficulties and/or gastro-intestinal problems affecting their daughter. Parent comments on what has and what has not worked for them are featured throughout the booklet.

We hope we have presented the information in a way that will be easy to understand and apply to people's everyday lives, as well as empowering families and carers in interactions with health and medical practitioners.

This booklet is part of a series of publications by the Child Disability Group of the Telethon Institute for Child Health Research summarising the latest research on the assessment and treatment of health conditions in Rett syndrome.

*We have referred to the girl or woman with Rett syndrome as your daughter. In some circumstances, this will not be your daughter, but will be the person for whom you will provide care.*

## Introduction

Nutrients are the essential ingredients that our body needs to absorb from the food we eat and from the fluid we drink to enable us to be healthy.

Before the nutrients can be absorbed and used by the body, food needs to be broken down into smaller components so that the nutrients can be distributed throughout the body and absorbed by the cells.

When the food first enters the mouth, it is chewed, coated with saliva, swallowed and moves down the oesophagus into the stomach.

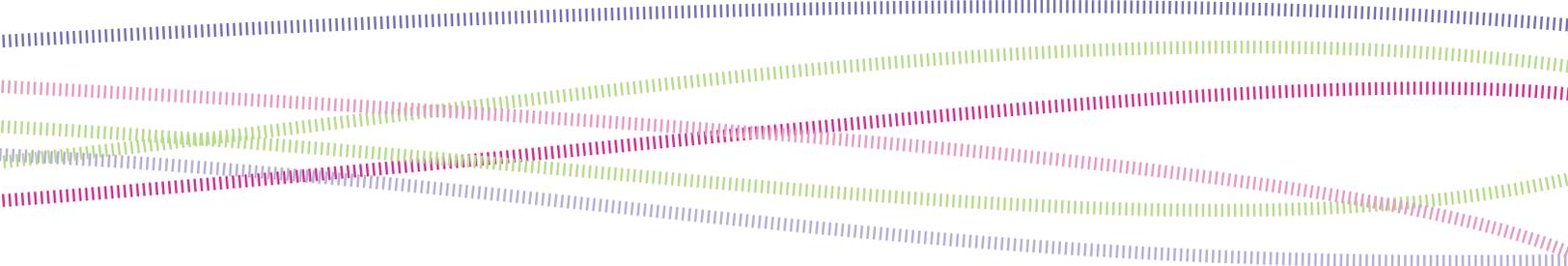
From the stomach, food is chemically digested and then moves into the small intestine from where the smaller molecules are absorbed into the blood and carried to cells throughout the body. Leftover waste products are processed and then eliminated through the large intestine.

### Knowledge is power ...

You will be in a better position to identify and monitor problems that may be affecting your daughter's health if you have a basic understanding of how the human body normally works, and what it needs to work best.

You will also be in a better position to advocate on management options that may be best for your daughter.





## Key messages

Good nutrition is important for everyone's health. For girls and women with Rett syndrome it is especially important to:

- build strength and stamina for everyday living
- strengthen the immune system
- limit the negative impacts of some of the other health conditions associated with Rett syndrome.

Many girls and women with Rett syndrome experience problems with feeding, swallowing and digestion. This can make it more difficult for them to get the nutrition they need. If your daughter has problems with feeding or with her digestive system – you are not alone.

Brush up on your basic knowledge of human biology – why nutrition is important and how the digestive system is supposed to work. It will help you to identify and monitor problems so you can advocate from an informed position on what treatment options may work best.

Learn the language of nutrition and digestion – communication with health professionals is easier if you know the basic medical terms they are likely to use. We have included a glossary of terms on pp 41-43.

Make sure your daughter has a regular medical assessment to check for any nutritional and/or digestive problems so that the most appropriate corrective action can be taken. A thorough nutritional and digestive health assessment is recommended once every six months for children under 12 years and at least once a year for teenagers and adults.

Not all gastrointestinal problems can be attributed to Rett syndrome. When making a diagnosis other possibilities may also need to be considered.

You know your daughter best – monitor any feeding difficulties she may be experiencing (including how long it takes to feed her, her preferences for different foods and textures and how they seem to affect her digestive processes). Keep a detailed diary. Share your observations with your doctor.

If your daughter seems to be having difficulties with her feeding, or with her digestive processes, as a first step, try making gradual changes to her diet and/or her feeding routines. Be prepared to experiment with different foods and textures. Changes in posture while feeding may also make feeding easier. Monitor the effects. Keep a record of what you try and what effect it has.

If your daughter is underweight, try being creative in the kitchen to increase her calorie intake. We have included some simple ways to add calorie-rich foods to her diet (see page 15).

If simple changes to diets and eating routines don't work, there are medications that may assist. Doctors follow protocols to find the best medication and dosage depending on the type of problem your daughter may be experiencing. Medications should only be used after consultation and careful monitoring by your medical practitioner.

If changes to diet, feeding routines or medications don't work, there may be surgical options available that may improve your daughter's nutritional and digestive health.

## PART 1:

## Nutritional health

In this part of the booklet we provide information on:

- why nutritional health is important (and what's required to achieve good nutritional health)
- the essential nutrients the human body needs
- nutritional issues commonly encountered in Rett syndrome
- recommendations for nutritional health in Rett syndrome
- options for improving feeding and nutritional health in Rett syndrome.

### 1.1 What do we mean by “nutritional health”?

Nutritional health requires the:

- adequate intake of nutrients (from a variety of foods and fluids)
- effective processing and absorption of nutrients (as food and fluids are digested)
- effective use of nutrients (as nutrients are metabolised to provide energy and build and repair body tissues).

#### Good nutrition is important for everyone's health ...

For girls and women with Rett syndrome good nutrition is especially important to:

- build strength and stamina for everyday living
- build and repair body tissues (including building stronger bones and muscle tissue)
- strengthen the immune system

As well as to:

- limit any negative impacts of some of the other health conditions typically associated with Rett syndrome.



The amount of energy available in food is expressed as kilocalories or kilojoules/gram. The higher the number of kilocalories or kilojoules/gram, the greater the energy-producing value of the food. Energy not expended in growth, development and activity is normally stored as body fat.

### Your daughter needs a variety of foods from different food groups.

Your daughter needs:

- Macronutrients (needed in larger quantities).

There are three sources of energy producing macronutrients:

- carbohydrates (mainly found in grains and some vegetables)
  - proteins (mainly from animal products but also in legumes, some nuts and vegetables)
  - fats (mainly from animal products but also in some vegetables or manufactured artificially).
- Micronutrients (needed in smaller quantities).

Vitamins and minerals are “micronutrients” because only a small amount is required of each one to do the job required of it. A deficiency can lead to health problems.

**Maintaining a healthy body weight requires a balance between the intake of energy-producing foods, and the expenditure of energy used in maintaining bodily functions and during physical exertion.**

Girls with Rett syndrome are often short in height for their age but their weight should be in proportion to their height. Body mass index (BMI) is useful to assess this. If there is insufficient food intake over a significant period of time, growth may slow below potential but these girls are also likely to be thin. A small proportion is overweight.

**An adequate daily intake of fluids is essential to maintaining good nutritional health.**

Many families report that they are worried about the adequacy of the fluid intake of their daughter. Girls and women with Rett syndrome are also likely to have a higher rate of fluid loss due to related conditions such as drooling or hyperventilation. An increased rate of fluid loss makes it even more important to ensure adequate fluid intake.

### HOWEVER ...

As many parents and carers will know ... getting their daughters to eat and drink as much as they need is not always easy.



## 1.2 Why do some girls and women with Rett syndrome have feeding difficulties?

Eating is a complicated process requiring the ability to co-ordinate different muscle groups in the mouth, jaw, throat, oesophagus and diaphragm in the correct sequence.

Difficulties in muscular coordination commonly associated with Rett syndrome mean that girls and women with the syndrome may also experience difficulties in feeding, chewing and swallowing.

These, and other Rett syndrome features that may contribute to feeding difficulties are listed in the table below.

### Factors that may contribute to feeding difficulties in Rett syndrome

|  |   |
|--|---|
| <b><u>Appetite</u></b> may be affected by medication   | Some medications, especially those used to control epilepsy, can affect appetite – increasing or decreasing the desire to eat.  |
| <b><u>Oral sensitivity</u></b> may affect responses to the taste, texture or temperature of foods or fluids          | Some may respond negatively to the taste, texture or even the temperature of some foods and fluids.   |
| <b><u>Chewing</u></b> requires the coordination of muscles in the tongue, jaw and cheeks                             | Many find it difficult to move food around the mouth with their tongue to help with chewing and swallowing.<br>Repetitive grinding of the teeth (bruxism) is also common in Rett syndrome. Teeth grinding can wear down the teeth as well as cause other dental problems which may affect the ability to chew and soften food ready for swallowing.   |
| <b><u>Swallowing</u></b> requires the coordination of muscles in the lips, tongue and throat in the correct sequence | The throat and pharynx below it provide a passage for the intake of air into the lungs, as well as regulating the movement of food and fluids down the digestive tract. When the food passage is open, the air passage is normally closed.<br>Some girls and women with Rett syndrome experience difficulty swallowing causing them to cough, gag or choke while eating or drinking and increasing the likelihood that food, fluids or air may go down the wrong passage.<br>Abnormal breathing patterns can also interfere with the swallowing process further increasing the risk of food, fluids and air going down the wrong way. |
| <b><u>Posture</u></b> can affect the alignment of the digestive tract  | Poor posture especially slouching over while eating can squeeze and cramp the digestive organs.<br>The presence of a scoliosis may make it difficult to maintain an upright posture while eating and drinking.  |
| <b><u>Self-feeding</u></b> requires finely tuned coordination between the eye, hand and mouth                        | The ability to feed ourselves enables us to control what, how much and the pace at which we eat or drink.<br>Many are unable to independently feed themselves and rely on others to provide them with adequate food and fluid.  |

## Feeding difficulties can reduce the intake of essential nutrients

**An adequate and balanced intake of food across the main food groups is required to ensure healthy nutrition.**

Many girls and women with Rett syndrome do not get enough of the nutrients they need for health. This is despite most parents reporting their daughter has a good appetite and eats about the right amount of food or even more than expected.

*“When Sarah was about 10, eating had become so slow that she wasn’t getting enough nutrients and she began to spiral downwards. She was too tired to eat and because she wasn’t eating enough, she had no energy.”*

## Feeding difficulties can increase the time taken for eating and drinking

**An extended feeding time can be stressful for the person being fed and for the person feeding them.**

Many girls and women with Rett syndrome depend on others to assist with feeding and take longer to feed than would normally be expected.

The time taken to feed is often used as a measure of difficulty in feeding.

*“Kate was eating regular food until she lost her skills. From that point on, she only ate pureed foods. But it still took 45 minutes for her to eat 1/2 cup. It was also difficult to get enough fluids into her and she was usually constipated.”*

## Difficulties with swallowing can increase the risk of choking

**Coughing, gagging or choking during eating or drinking may indicate problems with swallowing, as well as increase the risk of aspiration (inhalation) of food or fluids into the lungs.**

Approximately 25% of girls and women with Rett syndrome regularly experience coughing, gagging or choking when eating or drinking. Around 40% have difficulty swallowing.

*“Angie finger fed until chewing and swallowing became inadequate and she began to choke. This was about 10 years ago. We turned to pureed casseroles and soups and soft foods for about seven years.”*



## 1.3 How do I know if my daughter is getting enough nutrition?

To monitor the adequacy of your daughter's nutritional intake, it's very important to have your daughter's nutrition and growth assessed by a qualified health care practitioner on a regular basis.

Nutritional assessments are recommended:

- every six months for children under 12 years of age
- at least once a year for teenagers and adults and throughout adulthood.

### 1.3.1 What is a nutritional assessment?

A standard nutritional assessment will usually include:

- [measurement](#) of weight, height and body mass index (BMI) to monitor growth
- [oral health check](#) of teeth, tongue and gums to determine whether any oral health problems may be interfering with eating and drinking and/or whether a referral to a dentist for treatment is required
- [assessment of daily diet](#) to determine what and how much food and fluid is being consumed
- [assessment of feeding abilities and difficulties](#) including:
  - how long it takes to feed
  - chewing and swallowing
  - self-feeding abilities
  - whether different feeding techniques have been trialled and what effect they had (such as changes to posture, use of special equipment or utensils, modifying the consistency of foods etc).

- [blood tests](#) to measure the chemical composition of the blood indicating how well the organs are working and whether essential nutrients are available in sufficient quantities to enable the body to grow, build and repair tissues and to function most effectively.

**At least part of the growth restrictions that occur in Rett syndrome are likely to be as a result of genetic rather than nutritional factors.**

Special growth charts have been developed that take account of the smaller stature of girls and women with Rett syndrome. These charts provide a basis for comparing growth patterns with others with Rett syndrome.

<http://www.rettsyndrome.org/understanding-rett-syndrome/living-with-rett-syndrome/symptom-management/health-matters/growth-references>



## 1.3.2 Additional tests

### Blood tests

Your doctor may conduct blood tests if your daughter:

- is at risk of being deficient in specific micronutrients - particularly if she has a low BMI, or is taking anti-epileptic medications which affect the absorption of some micronutrients
- has symptoms or signs suggesting that nutrients are not being properly absorbed in the digestive system (such as poor weight gain, weight loss or anaemia).

### Chewing and swallowing ability

If your daughter has difficulty eating and drinking, your doctor may refer her to an allied health care professional so that a more thorough assessment can be conducted of her feeding, chewing and swallowing abilities.

### Videofluoroscopy

Swallowing may be investigated with a special x-ray procedure called a *videofluoroscopy* or *Modified Barium Swallow Study*. Your daughter will be given foods and fluids of different textures and consistencies mixed with barium (a white powdery substance that shows when x-rays are taken). By taking a rapid, moving series of x-ray images, the contrast of the barium makes it possible to observe the swallowing mechanism and the functioning of other organs in the upper part of the digestive tract. Recording the images on video allows for closer examination and analysis of the results after the test has been conducted.

Videofluoroscopy can be especially useful in seeing whether food and fluids are being aspirated into the airway and lungs rather than passing down the oesophagus into the digestive tract.

Videofluoroscopy can also be useful in assessing whether modification to the texture of foods may help improve swallowing ability.

## 1.3.3 Your contribution to the nutritional assessment

While doctors and other health care professionals have the knowledge and expertise to undertake tests, make diagnoses, and advise on treatment options, they do not know your daughter as well as you do.

For a nutritional assessment, your daughter's doctor or dietician will also need information from you about what, and how much, your daughter eats and drinks on a daily basis and how she typically reacts to the different foods and fluids she consumes.

Your experiences, observations and understanding of your daughter's dietary preferences, feeding abilities, and feeding difficulties are vitally important to ensuring that the nutritional assessment and treatments are informed by real-life experience.

### **You know your daughter best ...**

You are the best person to monitor what your daughter is eating and drinking on a daily basis as well as how different foods or fluids affect her.

Keep a detailed diary of her preferences for different types of foods or different textures of foods. What does she seem to like or not like?

Record how she responds to the consistency of food and fluids as well as how they seem to affect her.

Record any feeding difficulties she may be experiencing, including when and under what circumstances she seems to experience more or less difficulty.

Share your observations with your daughter's doctor or health professional.

## 1.4 How can I improve my daughter's nutritional health?

To improve your daughter's nutritional health you may consider:

- altering her diet
- trying strategies to reduce feeding difficulties
- enteral feeding options (See section 1.6 page 17).

Seek advice from your doctor or a dietician about a reasonable target weight.

### 1.4.1 Changes to the diet

The amount of energy (number of calories) your daughter needs to maintain a healthy weight is determined by comparing her usual intake with a series of growth measures.

If your daughter is underweight for her height and age, her intake of calories should be increased until she reaches a satisfactory weight.

You can increase the number of calories in your daughter's diet by:

- supplementing her diet with [high calorie nutritious foods](#) (breads, cereals, pasta, dried fruits, dairy foods, legumes, olives, avocados or by adding healthy fats and oils). If foods need to be puréed, blend family food but add milk if more liquid is needed. Cheese, yoghurt, oil or margarine can also be added
- offering milk desserts or yoghurts, or milk drinks after meals or as snacks. Age appropriate high calorie nutritional supplements can be useful. These should only be commenced on the advice of an experienced dietician.

*"We give her honey greek yoghurt with high calorie count by mouth. It is pleasant for her to eat and slides down well. Texture seems to be important to determine what "sticks" in her throat and what slides through."*

*"Maree eats orally at lunch time and has Jevity Hi Cal at other times. She usually has the meal from the previous day for lunch. We have found that this hasn't kept her iron levels up so has had to have a natural iron supplement to get the levels up again."*

An intake of energy-producing nutrients that is too high for the amount of energy expended may lead to your daughter putting on too much weight.

A small proportion of girls or women with Rett syndrome are overweight for their height and age.

*"Now that Sarah is in full-time care, we have to be careful that carers are not continually feeding her and giving her the wrong foods. Sarah always wants to be in the kitchen looking for food, stealing fruit from the fruit bowl, taking food from the bench. Carers think she is hungry because she goes to the kitchen. We have had to encourage carers to give her lots of fruit and veg, less meat, not biscuits, chips etc and to have coke zero. She also has lite milk."*

*"Meg is quite solid for a Retts girl. She constantly wants food - sometimes she is hungry although at other times we think this is due to pain in her tummy, boredom or frustration. When finger feeding, Meg will put too much in her mouth and will eat far too quickly. To manage this, we practice counting to 10 slowly with Meg's each bite. Meg responds well to us counting, and more frequently now days she will slow her eating down when I tell her to "Count to 10". "*



## For girls and women with Rett syndrome who are underweight

### Some tips and recipe suggestions for increasing calorie intake

#### TIPS FOR INCREASING CALORIES

One of the easiest ways to increase the calorific value of the diet is to add high energy foods to the meals and snacks your daughter is already eating.

Examples of high energy and high nutrient foods that can be added to meals and snacks include:

- **Cream or full cream milk (fluid or powder) or non-dairy alternatives** – added to milkshakes, smoothies, porridge, cereal, sauces, scrambled eggs, omelettes, soups, pancakes, purées, custards and desserts
- **Cheese** – added to scrambled eggs, omelettes, soups, purées, sauces, baked beans, pasta dishes, toast
- **Yoghurt (dairy or non-dairy)** – added to smoothies, cereals, desserts
- **Avocado** – added to bread, toast, crackers
- **Meat, legumes (eg baked beans)** – added to casseroles, soups
- **Oils and butter** – added to bread, pasta, and in cooking
- **Offering a dessert after the main meal**

**High energy snacks** can include:

- sandwich with plenty of butter and margarine and soft filling
- nut spreads on bread / toast / crumpets
- muffins with dried fruit and nuts
- yoghurt or custard (full cream)
- milk drink

#### EASY RECIPES

##### **Quiche (all in one)**

4 eggs  
1 small onion, chopped  
1 cup cheese, grated  
1 ½ cups milk  
½ cup self-raising flour  
Beat eggs and then add all other ingredients and mix.  
Add chosen flavours\*.  
Cook in pie dish or shallow casserole in moderate to hot oven 190°C for 40 minutes or when skewer comes out clean.  
\*You can add ingredients of your choice such as salmon, asparagus, spinach, ham, creamed corn, grated vegetables etc.

##### **Passionfruit Shape**

1 packet lemon jelly crystals  
1 egg  
1 cup sugar  
1 cup milk  
6 passionfruit  
Dissolve the jelly in 1 cup hot water.  
Beat together the egg, sugar and milk.  
Mix with jelly and beat well.  
Add passionfruit pulp (with or without seeds).  
Pour into individual bowls or serving bowl and chill.

*Recipes with permission from 'By Special Request: Recipes from Granny's Kitchen' by V. Reith (2006). Published by Penfolk Publishing, Victoria: Australia.*

## 1.4.2 Reducing feeding difficulties

You may be able to reduce your daughter's feeding difficulties by:

- **offering frequent small feeds** - to help her practise her feeding skills and reduce the effort required for each meal
- **supporting her jaw** and ensuring it is not over-extended - to make it easier to chew
- **modifying food textures** and the consistency of fluids - to help with swallowing and to reduce the effort required to chew and move food in the mouth, particularly if she tires quickly
- **offering foods she especially likes** - to increase her motivation to eat
- **using special equipment or utensils** such as modified plates, spoons, cups and feeding bottles - to increase her capacity to feed herself
- **providing verbal and physical prompts** - to increase active participation in eating and make mealtime a more enjoyable social experience
- **monitoring breathing patterns** and offering food or fluids only when her breathing rhythm is steady - to reduce the risk of aspiration
- **adjusting her physical position** or supporting her posture - to ease the passage of food down her digestive tract.

You might also consider getting help from an allied health professional to improve your daughter's feeding skills.

Some girls and women lose their feeding skills after an acute illness or surgery. If this is the case, your daughter may benefit from more intense therapy to help her regain her skills and normal feeding patterns.

If your daughter is living in out-of-home care, all care staff involved in feeding her should be provided with instructions by an experienced speech therapist, dietician or nurse on the best feeding techniques for her. These feeding instructions should be based on the results of your daughter's growth and nutrition assessment.



## 1.5 Enteral feeding options

“Enteral feeding” is a term used to describe the delivery of nutrients via a tube inserted directly into the stomach, or into the duodenum or jejunum in the small intestine.

If changes to the diet or feeding practices do not improve your daughter’s nutritional intake, enteral feeding may help.

There are three main types of enteral feeding:

- **a naso-gastric tube** where a feeding tube is inserted down the nose into the stomach (usually suitable for use on a short-term basis only)
- **a gastrostomy tube** (sometimes termed a G-tube) where a feeding tube is implanted into the stomach either endoscopically (via the mouth and called a PEG [Percutaneous Endoscopic Gastrostomy]) or surgically
- **a gastro-jejunostomy or PEG-J tube** where a feeding tube is placed into the jejunum in the small intestine through a gastrostomy or surgically. This is used less commonly.

Your doctor may recommend that your daughter would benefit from enteral feeding if she:

- fails to put on weight despite an increase in her calorie intake
- finds it difficult to coordinate the muscles in her mouth and throat (and especially if restrictions in her swallowing ability increase the risk of aspiration into the airway and lungs)
- experiences a lot of discomfort and becomes distressed when feeding or drinking
- takes an unusually long time to feed.

Enteral feeding can:

- provide assurance that your daughter is getting the essential nutrients she needs
- reduce feeding hassles caused by resistance to eating or by excessively long feeding times.

*“Lily is so thin and I am desperately worried. I have succumbed to the acceptance that she will need a feeding tube to help her gain some weight and maintain it in times of ill health. This has been a difficult thing for me as I am sure it has been for all those who have gone through the same with your angels. There have been many years of anticipation and consideration, and feelings of dread and guilt that I could not nourish my own child.”*

*“I also put off agreeing to a PEG for years, but have not regretted having it put in. It is not that you cannot nourish your child, she just needs to get it in a different way.”*

*“Jess had been thin for years but after a rough two winters of chest infections and a particularly bad bout of pneumonia, she had to have a G-tube placed as an emergency procedure. I wish we had opted for the surgery prior to this, when she was well. I think in my mind I was using the excuse of Rett syndrome for her being so skinny. I would say to people ‘girls with Rett are often small for their age’. But looking back at photos I can now see Jess wasn’t just small for her age, she was too thin.”*

**Many parent carers who initially seemed reluctant to go down the tube feeding route have subsequently reported how much easier life has become - both for their daughter, and for them - after she moved onto enteral feeding.**

In the Australian Rett Syndrome Database, about one in five girls or women with Rett syndrome are tube fed, either for some or all of the time.

## Advantages and disadvantages of different enteral feeding options

| Option   | Advantages   | Disadvantages   |
|--|--|---|
| <b><u>Naso-gastric tube</u></b> - inserted down the nasal passage into the stomach                             | <ul style="list-style-type: none"> <li>• provides immediate increase in nutrient intake:               <ul style="list-style-type: none"> <li>◦ in cases of acute illness or where the feeding difficulty is likely to be temporary</li> <li>◦ to correct existing malnutrition</li> <li>◦ to test whether a more permanent gastrostomy tube may improve nutrition.</li> </ul> </li> </ul> | <ul style="list-style-type: none"> <li>• usually short-term use only</li> <li>• may cause discomfort</li> <li>• possibility of tube becoming clogged or dislodged or migrating</li> <li>• skin irritation where tube is taped</li> <li>• risk of reflux.</li> </ul>   |
| <b><u>Gastrostomy</u></b> - inserted through the abdomen into the stomach                                      | <ul style="list-style-type: none"> <li>• allows oral intake without discomfort and amount can be varied depending on how much is eaten at meals</li> <li>• reduces feeding stresses (fewer feeding times of shorter duration) and risk of chest infection due to aspiration</li> <li>• ease in administering medications.</li> </ul>   | <ul style="list-style-type: none"> <li>• possibility of the tube leaking, becoming dislodged or migrating</li> <li>• possibility of surgical complications during insertion</li> <li>• red irritated skin and/or hypergranulation at gastrostomy site.</li> <li>• risk of reflux.</li> </ul>  |
| <b><u>Gastro-jejunosomy (PEG-J)</u></b> - inserted through the abdomen into the jejunum in the small intestine | <ul style="list-style-type: none"> <li>• similar benefits to gastrostomy</li> <li>• recommended in cases of gastro-oesophageal reflux that are difficult to manage, where gastrostomy feeds are not tolerated, and/or where there is a high risk of aspiration.</li> </ul>   | <ul style="list-style-type: none"> <li>• need for continuous rather than bolus feeds</li> <li>• possibility of tube leaking, becoming dislodged or migrating, irritating skin.</li> <li>• may decrease transit time of liquids leading to poor absorption and risk of undigested nutrients entering the small intestine if flow is too fast</li> <li>• possibility of surgical complications during insertion.</li> </ul> |

### Tube feeding can provide long term assurance of adequate nutrient intake

*“Seven years ago, when Sarah was about 10, we started having weight issues again. I was watching my daughter fade away before my very eyes! Now Sarah is 17 and I am forever grateful to have the PEG. It means I know she is getting sufficient nutrients and fluids as well as making giving medicines easy. When she was little she would often spit out her anticonvulsant meds and you never knew if she had the full amount or sufficient cover. Now you measure the amount and that’s what goes in unless you spill it!”*

### Tube feeding can reduce feeding stresses

*“Switching to J tube feedings versus g tube feedings sounds daunting, but can make our girls so much more comfortable. It’s not much of a big deal (at least if your child is non-mobile) to simply hang the bag on the chair all day and stay hooked up. Actually I found it easier. With the g-tube, I would occasionally forget a feeding on time, which then messed up the rest of the day’s schedule. Our pump (Zevex Infinity) can run all day, and into a second day, without needing to be recharged, so we simply plug it in overnight.”*

### Feeding through a naso-gastric tube can improve short-term nutritional intake

*“Sarah got so weak we had to give her a naso-gastric tube and any further surgery had to wait till she was physically a bit stronger. I think we used the tube for six months, using Jevity with fibre, (which she is still on) which clogs the tube very easily. We had many hospital visits to get new NGs because the last one was unworkable, but this was still so much better than seeing my girl so sick. When Sarah was well enough and she had regained sufficient weight, the operation was planned to re-do the gastrostomy.”*

### Tube feeding can reduce the risk of aspiration

*“I stopped feeding Sarah by mouth with the last gastrostomy, as it just wasn’t worth the risk of aspiration and possibility of pneumonia. Occasionally she can have a taste of something, like a nibble of birthday cake, but this is not often and only when she is well and there hasn’t been any hint of coughing!”*

### “Just a different way of nourishing your child”

*“When Jess first had the tube placed, an older nurse at the hospital mentioned how, when she started nursing, they didn’t have formula and used to blend meals or make smoothies to go into the tube. Once I looked into it, I found that lots of people still do this. Initially I only had a hand held stick blender and would make smoothies with sustagen, yoghurt, milk, apple purée, avocado, cooked pumpkin and sweet potato – things that didn’t need too much blending. Then some friends and workmates got together and held a fundraising night to buy us a Thermomix. This has been great and changed Jess’s diet. She now eats a huge range of healthy foods and gets much more variety than she ever did when she ate everything orally. We can blend up raw fresh fruit and veggies and nuts smooth enough to go in via the tube. Yesterday for lunch Jess had celery, spinach, apple, pear, carrot and pumpkin seeds.*

*Jess still eats – probably about 50% orally and 50% via the tube, but if she has a cold or is overly tired, I can still give her something to eat – quickly and easily without any pressure on either of us! Now Jess can enjoy eating. I love that in hot weather I can quickly give her some water and we can be on our way. Having a feeding tube means you can still nourish your child – just done in a different way!”*



## The importance of regular monitoring ...

While there are many benefits associated with tube feeding, there are also some potential risks and complications.

For girls and women who are on enteral feeding support it is especially important to have regular medical check-ups to ensure enough nutrients are being supplied and that the tube continues to be positioned correctly and working effectively.

Your dietician will be able to advise on the most cost-effective way to obtain feeds.

*“Since Sarah’s spinal surgery in 2010, the angle of the PEG has changed slightly and there is some leakage from the gastrostomy site. Gauze pads don’t really help as they just stick to the area and hurt to get back off. We have been using a ‘stoma’ powder which helps to limit any leaking. Sometimes the area looks sore but this may be because her wheelchair belt or pants have been rubbing on the area. The soreness doesn’t seem to continue over night (at this stage anyway).”*



## PART 2:

## Digestive health

This part of the booklet focuses on what happens to nutrients as they are processed and absorbed and as waste products are eliminated by the digestive system.

In this section we provide information on:

- how the digestive system works (including the role of different digestive organs)
- how conditions associated with Rett syndrome can affect digestive health
- the assessment, symptoms and management of three of the main gastro-intestinal problems that affect women and girls with Rett syndrome
  - reflux
  - constipation
  - abdominal bloating.

Although this booklet is primarily concerned with the conditions listed above, it is acknowledged that girls and women with Rett syndrome may also be prone to problems with the gallbladder (such as gallstones) which may cause pain as well as lead to other complications.

### 2.1 How does the digestive system work?

The digestive system is under the control of the *autonomic nervous system* which is responsible for those bodily functions that occur without us having to consciously think before performing them.

As well as digestion, the autonomic nervous system controls involuntary body processes such as circulation, respiration, perspiration, salivation, dilation of the pupils, etc.

Within the autonomic nervous system, the *enteric nervous system* controls the mechanical and chemical processes required to break down food and fluids into their smaller molecular components so that nutrients can be absorbed and used by the body.

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

### 2.1.1 The digestive tract

The digestive tract is a long, twisting, hollow tube that connects organs and stretches from the mouth to the anus.

Muscles lining the digestive tract propel food and fluids from the mouth, down the oesophagus, into the stomach and from there into the small intestine and finally into the large intestine (or colon).

Specialised circular muscles (sphincter muscles) along the digestive tract seal off the different digestive organs so that each part of the process can be performed in its correct sequence without mechanical or chemical interference from other parts.

Tiny glands are embedded in the mucous membrane (mucosa) of the digestive tract. These glands produce juices that chemically break down food molecules so their nutrients can be absorbed.

Each organ in the digestive tract performs a distinct function which depends on the successful execution of the function preceding it. Problems encountered at any level of the process may have repercussions further down the line as the digestive process continues.

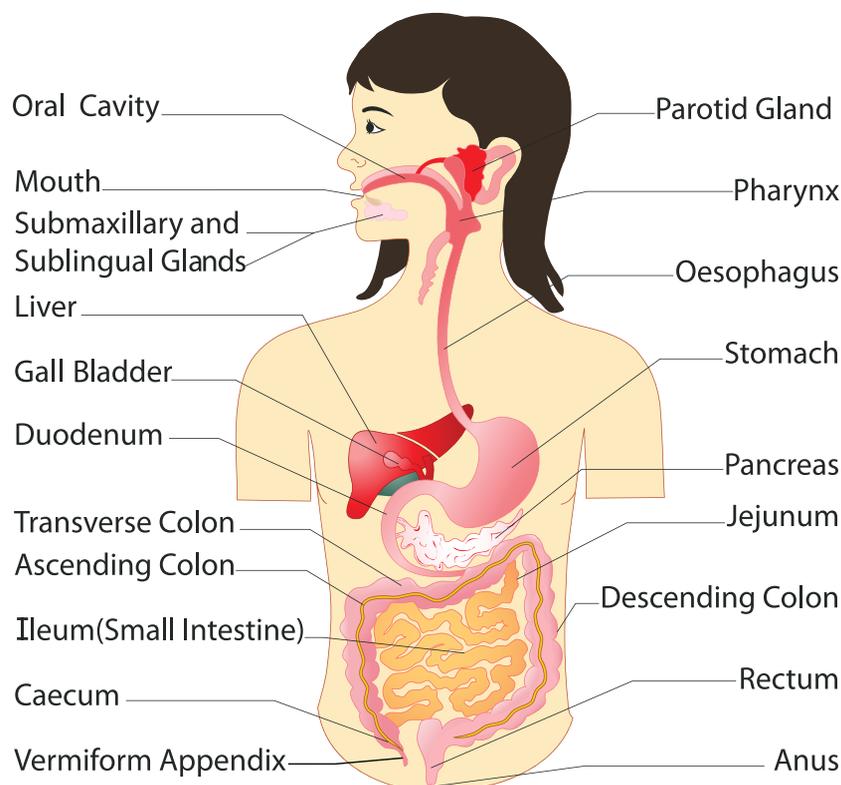
### 2.1.2 Mechanical and chemical processes of digestion

The mechanical stage of digestion enables food to be mashed, mixed and moved through the digestive system via a synchronised sequence of muscular contractions called peristalsis.

Digestive chemicals (enzymes) secreted by the mouth, stomach and small intestine break down larger food molecules into their component parts ready for absorption.

The movement of food and fluids through the digestive organs and the production of digestive juices within them are stimulated and regulated by hormones released into the blood stream.

A network of nerves controls the direction and force of muscular contractions in the different digestive organs and the release of digestive juices into them.



## Stages of digestion

| Digestive organ  | Process  | What can go wrong   |
|--|--|---|
| <p><b>Mouth</b> - provides a point of entry for food and fluids and breaks down food through chewing</p>         | <ul style="list-style-type: none"> <li>• the tongue moves food around for chewing and positions it ready for swallowing</li> <li>• saliva moistens and softens food and begins the chemical breakdown of carbohydrates</li> <li>• muscles at the back of the mouth contract to swallow the food.</li> </ul>  | <ul style="list-style-type: none"> <li>• poor oral health or teeth grinding (bruxism) can interfere with chewing.</li> <li>• poor muscular coordination can restrict:               <ul style="list-style-type: none"> <li>◦ tongue movements</li> <li>◦ effective chewing</li> <li>◦ contraction of muscles in the correct sequence.</li> </ul> </li> </ul>  |
| <p><b>Throat</b> - transports food from back of the mouth into the oesophagus</p>                                | <ul style="list-style-type: none"> <li>• the epiglottis closes the windpipe (<i>trachea</i>) to prevent food entering the lungs</li> <li>• a muscle at the top of the oesophagus (<i>the upper oesophageal sphincter</i>) opens to allow food into the oesophagus.</li> </ul>  | <ul style="list-style-type: none"> <li>• inadequate muscular contractions and/or poor muscular coordination can:               <ul style="list-style-type: none"> <li>◦ interfere with swallowing</li> <li>◦ cause food and fluids to enter the windpipe rather than the oesophagus.</li> </ul> </li> </ul>   |
| <p><b>Oesophagus</b> - propels food from throat to stomach</p>   | <ul style="list-style-type: none"> <li>• wave-like muscular contractions (<i>peristalsis</i>) propel food and fluids toward the stomach</li> <li>• a muscle at the bottom of the oesophagus (<i>the lower oesophageal sphincter</i>) opens to allow food and fluids to enter the stomach.</li> </ul>   | <ul style="list-style-type: none"> <li>• inadequate muscular contractions and/or poor muscular coordination can:               <ul style="list-style-type: none"> <li>◦ restrict the smooth passage of food or fluids down the oesophagus</li> <li>◦ inhibit appropriate opening and closure of the lower oesophageal sphincter allowing stomach acids to pass back into the oesophagus.</li> </ul> </li> </ul>   |
| <p><b>Stomach</b> - breaks down larger food molecules into their smaller molecular components</p>                | <ul style="list-style-type: none"> <li>• muscular contractions continue to break down food mechanically</li> <li>• food and liquids are churned and mixed with stomach acids, digestive juices and protein-digesting enzymes</li> <li>• a muscle at the bottom of the stomach (<i>the pyloric sphincter</i>) opens to allow the contents of the stomach to empty slowly into the small intestine.</li> </ul> | <ul style="list-style-type: none"> <li>• inadequate muscular contractions and/or poor muscular coordination can lead to:               <ul style="list-style-type: none"> <li>◦ food remaining in the stomach for longer than normal or failing to empty from the stomach at all (delayed gastric emptying or <i>gastroparesis</i>)</li> <li>◦ undigested food entering the small intestine too quickly (<i>gastric dumping syndrome</i>).</li> </ul> </li> </ul> |
| <p><b>Liver</b> – filters blood from the digestive system, processes food nutrients and regulates metabolism</p> | <ul style="list-style-type: none"> <li>• drugs are filtered and neutralised</li> <li>• bile is produced to break down dietary fats and enable absorption of fat soluble vitamins</li> <li>• carbohydrates are converted to glucose for energy</li> <li>• ammonia from protein metabolism is converted into urea for excretion in urine.</li> </ul>   | <ul style="list-style-type: none"> <li>• liver can become enlarged, scarred or damaged by drugs, being overweight, diabetes or through infection by viruses leading to impaired liver function.</li> </ul>  |

## Stages of digestion (continued)

| Digestive organ  | Process   | What can go wrong  |
|--|---|--|
| <p><b>Gallbladder</b> – stores bile produced by the liver before release into small intestine</p>                              | <ul style="list-style-type: none"> <li>water is extracted from the bile previously produced by the liver</li> <li>concentrated bile liquid is squeezed into the small intestine when fatty foods are present.</li> </ul>  | <ul style="list-style-type: none"> <li>formation of gallstones through crystallisation of excess cholesterol in the bile and/or the failure of gallbladder to fully empty.</li> </ul>  |
| <p><b>Pancreas</b> – releases digestive juices into small intestine to break down food molecules</p>                           | <ul style="list-style-type: none"> <li>digestive juices and enzymes are released into the first part of the small intestine via a small duct</li> <li>release of sodium bicarbonate helps to neutralise stomach acids</li> <li>(also produces insulin which helps to control blood sugar levels).</li> </ul>  | <ul style="list-style-type: none"> <li>can become inflamed (sometimes as a result of gallstones).</li> </ul>   |
| <p><b>Small intestine</b> - continues the chemical breakdown of food molecules and absorbs nutrients into the blood stream</p> | <ul style="list-style-type: none"> <li>digestive juices released by the pancreas, liver and the walls of the intestine continue to break down carbohydrate, fat and protein</li> <li>nutrients are absorbed into the bloodstream through tiny finger-like projections lining the walls of the small intestine (villi and microvilli)</li> <li>a sphincter muscle at the base of the small intestine (the ileocaecal valve) opens to allow fluids, undigested food and bacteria to pass into the large intestine.</li> </ul>           | <ul style="list-style-type: none"> <li>inadequate muscular contractions and/or poor muscular coordination can: <ul style="list-style-type: none"> <li>restrict the passage of food or fluids through the small intestine leading to nausea, vomiting, bloating, pain or altered bowel movements</li> <li>contribute to a build-up of bacteria in the upper part of the small intestine inhibiting absorption of nutrients and releasing excess gas.</li> </ul> </li> </ul> |
| <p><b>Large intestine (colon)</b> - processes and stores wastes ready for elimination from the body</p>                        | <ul style="list-style-type: none"> <li>muscular contractions move fluid, bacteria and undigested food back and forth to allow sufficient time for most of the fluid to be reabsorbed</li> <li>the remaining fluid and waste products are compacted into a stool</li> <li>muscle contractions push the stool towards the rectum</li> <li>sensory nerves detect the filling of the rectum and trigger reflexes enabling the anal sphincter muscle to relax and the stool to be passed from the body during a bowel movement.</li> </ul> | <ul style="list-style-type: none"> <li>inadequate muscular contractions and/or poor muscular coordination can: <ul style="list-style-type: none"> <li>slow the movement of waste through the colon leading to too much fluid being absorbed and excessive drying of the stool leading to constipation</li> <li>inhibit relaxation of anal and rectal muscles necessary to allow the stool to pass.</li> </ul> </li> </ul>  |

## 2.2 How does Rett syndrome affect the digestive system?

In this part of the booklet, we provide information on the symptoms, assessment and treatment for three of the most common gastro-intestinal problems affecting girls and women with Rett syndrome:

- Gastro-Oesophageal Reflux Disease or Gastro-Esophageal Reflux Disease – GORD or GERD)
- constipation
- abdominal bloating.

These gastro-intestinal problems can cause considerable pain and discomfort as well as:

- compromising the healthy enjoyment of food
- limiting the intake of nutrients needed for optimal growth and bodily functioning.

Due to limited communication skills in those with Rett syndrome, it can be difficult making an accurate diagnosis.



**Most girls and women with Rett syndrome experience one of more gastro-intestinal problems.**

- The majority have some **gastro-intestinal dysmotility**
- Around 80% have **constipation**
- About 40% have gastro-oesophageal **reflux**
- Approximately half experience **abdominal bloating**

*“We could write an entire book on Kara’s GI problems. She has reflux issues, many gas/air issues, delayed gastric motility, etc, etc ...”*

### The good news is...

Effective treatments for gastro-intestinal problems are available and they’re the same used for anyone else who experiences these problems.

Just because a person has Rett syndrome, it doesn’t mean they have to put up with gastro-intestinal pain and discomfort.

### Start with the simple...

Some digestive problems can be easily remedied with changes to diet, positioning or the feeding environment. If simple strategies don’t work on their own, drug treatments may help. When all else fails, surgery may help to alleviate persistent problems that have not responded to other, less invasive treatments.

### It’s important to remember that...

Not all digestive problems can be attributed to Rett syndrome. If symptoms persist after treatment, it may indicate that something else is amiss. That’s why it’s important for your daughter’s doctor to do a thorough assessment and relevant investigations to rule out other possibilities.

## Many girls and women with Rett syndrome have multiple digestive problems

*“Sarah’s had lots of GI issues thru the years ... She screamed, cried, gagged, threw up and was miserable from constipation. She was losing her ability to swallow...never chewed a lot as early as when she was 3.5 years old.*

*Looking back so many symptoms were ignored by the medical world until we as parents found doctors who truly understood our pleas.*

*We tried everything including many visits to the feeding clinic at children’s hospital who felt the problem was me personally! Can you imagine! They felt I was spending too many hours...up to 15 hours a day...feeding her. I was watching Sarah wither away”.*



## Not all digestive problems can be attributed to Rett syndrome

*“Because she was diagnosed with Rett syndrome, no one looked further as to why my daughter has such horrible reflux and feeding problems.*

*When I questioned the extent of her reflux and discomfort I was always told that it was just Rett. Though I have been a good advocate for her in other areas, this seemed typical for Rett, so I didn’t push. If they had discovered she had intestinal malrotation when she was much younger it would have saved a lot of problems later on.*

*Instead, it was not discovered until she was almost 13 years old, after she had already moved to tube feeds, and a gj tube had failed.*

*Just because our children have Rett syndrome does not mean there can’t be other “problems” as well, push to look for the possibility, not everything is Rett related.”*



## 2.3 Reflux

### 2.3.1 What is reflux and what causes it?

In everyday use, the word “reflux” means “to flow back” which is a good description for what happens when reflux occurs in the digestive system.

When the muscle at the bottom end of the oesophagus (*the lower oesophageal sphincter*) does not close properly after food has passed through to the stomach, the contents of the stomach are able to flow backwards and up into the oesophagus.

The action of stomach acids in the oesophagus can be painful. Reflux may also cause nausea or vomiting. In extreme cases it can damage the lining of the oesophagus.

Acid may also flow back into the oesophagus when the stomach does not empty properly. This condition is called *delayed gastric emptying* (or *gastroparesis*).

**Reflux is more common in girls and women with Rett syndrome than in the general population.**

Approximately 40% of girls or women with Rett syndrome have problems with reflux. This is between 2-4 times more than occurs in the general population (in Western countries).

Reflux is more common in girls and women with Rett syndrome where mobility is also restricted – the greater the physical disability, the more likely the person will also have problems with reflux... especially if they spend a lot of time lying down.

Scoliosis also increases the likelihood of reflux.

*“At age 17, Rachel had a test for acid in the oesophagus (a pH probe study) that revealed she was refluxing terribly: 500 times in 48 hours and 15 of those times lasted longer than five minutes.”*

### 2.3.2 How do I know if my daughter has reflux?

Because most girls and women with Rett syndrome are not able to communicate their symptoms, it may be more difficult to work out why they seem to be experiencing pain after eating.

Your daughter may have reflux if she:

- loses weight, despite maintaining a good appetite for food
- has anaemia (due to iron deficiency) – suggesting reflux esophagitis
- has sour breath, burps or belching
- regularly vomits or brings back her food – (sometimes with blood)
- refuses food, or chews repeatedly on food with a reluctance to swallow (rumination)
- has recurrent pneumonia, coughs after feeds or poor respiratory health generally
- displays behaviour problems during or immediately after eating (including self-harm, screaming and/or restlessness).

#### Regular vomiting may be a sign of reflux

*“Sarah’s reflux was so bad that if I went grocery shopping with her in the trolley, I’d have to call for a mop and bucket to clean up the mess. By the time she was three, I had to take multiple changes of clothes wherever we went.*

*It wasn’t long before her weight was going backwards as well as it being socially difficult. (You can imagine she never smelled nice and clean for a cuddle, let alone the car seats, prams and carpet at home!)”*

*“At 11 months old, Liora began vomiting. The problem became chronic and it took us almost a year to discover the source.*

*We noticed as a result of the swallowing study, that Liora’s stomach was not emptying. So, all the food that I was feeding her was sitting in her stomach.”*

#### Increasing respiratory problems may be a sign of reflux

*“When she was five, she started hyperventilating a lot. She was very agitated and her eating was getting worse and worse.*

*After taking reflux medication for one month, she got much better. After a few months, she stopped hyperventilating completely. She also stopped drooling. She became her old smiley self.*

*Feeding her became easier. She started putting some weight on again.”*

#### Worsening behaviour problems may be a sign of reflux

*“When Rachel was around 6 and was irritable, we realized it was due to reflux.”*



### 2.3.3 Testing for reflux

Your daughter's doctor may order some tests to determine whether reflux is occurring.

#### Tests for Reflux

24-hour oesophageal pH monitoring measures the acid/alkaline (pH) balance in the oesophagus over a 24 hour period to see whether and how much stomach acid is in the oesophagus and whether the symptoms are consistent with the reflux

Multichannel intraluminal impedance monitoring may be relevant when the symptoms of reflux occur despite no or very low levels of acid in the oesophagus or when symptoms persist despite the use of acid suppressing medications

Upper gastro-intestinal endoscopy involves the insertion of a thin, flexible viewing instrument (called an endoscope) through the mouth into the oesophagus enabling the doctor to look for inflammation, bleeding, ulcers or other problems in the lining of the oesophagus. The endoscope can also be used to view the inside of the stomach and the first part of the small intestine. In children, this is done under general anaesthesia

Radionuclide scintigraphy enables two-dimensional images to be captured of the inner workings of the body's organs based on the gamma rays emitted from previously ingested radioactive chemical tracers. Radionuclide scintigraphy may help to test whether reflux is contributing to aspiration into the lungs.

### 2.3.4 What can be done to relieve my daughter's reflux?

Treatments for reflux can range from making simple changes to the diet or feeding practices, through to drug treatments, or surgery as a last resort.

#### Changing feeding strategies

Simple changes to diet and feeding strategies may be all that is required to control your daughter's reflux.

- **Diet** - try eliminating foods that may be associated with reflux (see box below). Eliminate one at a time and monitor whether there is any beneficial effect
- **Size and frequency of feeds** - try giving your daughter smaller and more frequent feeds to increase the speed of digestion and reduce the stomach acid available to flow back into the oesophagus
- **Thickened feeds** - talk to your doctor of health professional about thickening your daughter's feeds with commercial or food based thickeners
- **Posture while eating** - the digestive organs work best when they are not cramped. Gravity can also help the movement of food, fluids and digestive juices in the right direction. Try to ensure your daughter is in an upright position while she is eating. You may even consider feeding her while she is standing. If your daughter needs to eat lying down, make sure you elevate the head of the bed
- **The feeding environment** - stress can interfere with the normal digestive process. Try creating a calm environment for eating. You may even consider playing soothing music in the background – calming for your daughter and possibly for you as well.

#### Foods associated with reflux

Some foods are more likely to cause reflux in susceptible people including: citrus fruits, chocolate, caffeinated drinks, fatty fried foods, garlic, onions, mint flavourings, spicy foods, tomato-based foods, fizzy drinks.

## Drug treatments for reflux

If simple strategies don't work on their own, drug treatments may be used to supplement these strategies.

There are two main categories of drugs used to treat reflux, drugs that:

- decrease the amount of acid produced by the stomach
- increase the frequency or strength of contractions of muscles in the digestive system helping to move food through the stomach more quickly (especially in cases of delayed gastric emptying).

While these drugs may help relieve reflux, they may also have other side effects. That is why any drug needs to be introduced after careful consideration and its effects monitored carefully.

Drug treatments should only be administered on prescription from your doctor at the lowest dose for control of symptoms.



## Drug treatments for reflux

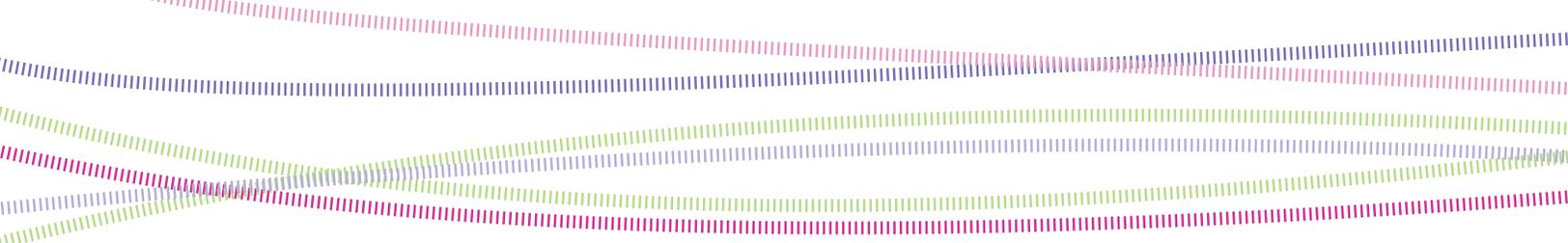
| Action of Drugs  | Type of drug   | Possible side effects   |
|--|--|---|
| <u>Drugs that decrease the amount of acid produced by the stomach</u>                                      | <b>Proton pump inhibitors (PPIs)</b> such as Lansoprazole, Omeprazole, Pantoprazole are the first choice and can be used over the long term.   | <ul style="list-style-type: none"> <li>• can be associated with diarrhoea, constipation or pain</li> <li>• high dose or long-term use can increase the risk of infection.</li> </ul>  |
|  | <b>H2-blockers or H2-receptor antagonist</b> also reduce acid secretion but are considered to be less effective than PPIs.   | <ul style="list-style-type: none"> <li>• minimal side effects have been reported but can be associated with gastrointestinal disturbances such as diarrhoea</li> <li>• development of intolerance and less effective with long term use.</li> </ul> |
| <u>Drugs that increase the frequency or strength of muscular contractions</u><br><b>(Prokinetic drugs)</b> | <b>Domperidone</b> medications such as Motilium, Motinorm, Costi and Nomit increase gastrointestinal motility where there is prolonged gastric emptying time. They may also increase tolerance to feeds. | <ul style="list-style-type: none"> <li>• can be associated with dystonia or abnormal muscle contraction around the head and neck</li> <li>• may increase the level of the hormone prolactin.</li> </ul>   |
|  | <b>Erythromycin</b> often prescribed as an antibiotic but at low doses can also be used to increase muscle contractions.   | <ul style="list-style-type: none"> <li>• can be associated with diarrhoea, nausea, abdominal pain or vomiting.</li> </ul>   |
|  | <b>Bethanechol</b> can also increase gastrointestinal motility.  | <ul style="list-style-type: none"> <li>• contraindicated if asthma or gastric ulcers.</li> </ul>  |



**Prokinetic drugs** such as Domperidone and Erythromycin can increase the heart's QT interval upsetting the timing of the heartbeat and triggering dangerous heart rhythms. Long QT interval is more common in Rett syndrome.

## Erythromycin can help in cases of delayed gastric emptying

*“Liora’s stomach was not pumping and not emptying. We consulted with a pediatric motility specialist in the US and he suggested four options. The first was erythromycin. Liora took this for a short while and within three weeks, the vomiting had stopped, her stomach learned to pump again and the vomiting went away. We have not seen this problem since. The doctor who suggested Liora try an antibiotic is a shining star in my eyes. He solved the problem in a quick and easy way. The stomach is now doing what a stomach should do.”*



## Surgical management of reflux

If severe reflux persists despite attempts to treat it with drugs, a surgical procedure called a *fundoplication* may need to be carried out.

This involves the upper part of the stomach (the fundus) being wrapped around the lower end of the oesophagus and stitched into place to tighten the lower oesophageal sphincter and prevent stomach contents flowing back into the oesophagus.

As well as helping to reduce reflux, fundoplication may also help to improve weight gain. However, it may also contribute to abdominal bloating, retching or swallowing difficulties.

A fundoplication can be performed by *laparoscopic* or *keyhole surgery* which reduces recovery time and requires only a short time in hospital.

Regular monitoring is required after a fundoplication to check that it is still operating effectively and/or whether any complications have resulted from the procedure.

*“Alana had a fundoplication at the very young age of three. The decision wasn’t taken lightly but a decision we knew had to go ahead.*

*Alana had numerous investigations for profuse vomiting and dehydration, and a cause was never found. She had difficulty keeping any meals down and nutrition obviously became an issue. I was told that when Alana had the fundoplication she would be having what they referred to as a full wrap and this would mean she could not vomit. Alana therefore needed a peg put in at the same time so gas could be released. This was also of great concern to me at this time. Just all the unknown really. I was concerned about Alana needing to vomit and the feeling for her not being able to. There were lots of things to weigh up.*

*I was also told Alana may not eat orally again but I had some kind of instinct that these things wouldn’t happen to Alana. After the surgery, I was determined not to wait too long to start her on small amounts of soft foods. I was very excited when she ate her 1st custard and it was all good from there with improvements every day. Alana has never had an issue with vomiting since and if uncomfortable I can just attach an extension tube to the peg and release gas this way. Unbelievably, the vomiting urge had disappeared for Alana. The fundoplication was the answer and a fantastic decision as we couldn’t go on the way we were.*

*I remember getting Alana dressed ready to go out before her fundoplication took place and before we would reach our destination, I would be pulling over on the side of road changing Alana. It was very stressful and horrible for Alana.*

*We have never had any issues with the fundoplication and it has been an excellent decision.”*

## 2.4 Constipation

In the last stage of digestion, left-over waste products are processed in the large intestine and eliminated from the body via the rectum and anus.

This process can occur more or less frequently for different people - from several times a day to only once every few days – which may be quite normal and healthy depending on the individual.

### 2.4.1 What is constipation and what causes it?

A person is considered to be constipated if their stools are dry, hard and difficult to pass. In Rett syndrome, reduced intestinal motility contributes to constipation.

The most common dietary and lifestyle factors that contribute to constipation are:

- not enough fibre in the diet
- not enough fluid in the diet
- not enough physical activity
- disruptions to the daily routine (or not enough predictability in the daily routine).

### 2.4.2 How do I know if my daughter has constipation?

Monitoring the frequency and consistency of your daughter's bowel actions provides the best guide as to whether she may be suffering from constipation.

Constipation can cause considerable discomfort and pain.

Your daughter may not be able to express her discomfort or explain where it is occurring in which case you need to be alert to other signs.

Your daughter may be suffering from constipation if she:

- grimaces or cries when passing a stool
- loses her appetite
- withdraws from interaction with others
- is more irritable or her behaviour changes especially if behaviour problems increase as the interval between the passage of a stool increases.

*“Sarah would roll around our living room screaming when constipation was so very bad ... brought me to tears...”*

Your daughter's doctor may also be able to diagnose constipation by conducting a physical examination of her abdomen and or sometimes by requesting an X-ray.

### 2.4.3 What can be done to relieve constipation?

Making changes to diet and lifestyle may be effective in reducing constipation or the severity of symptoms.

If changes to diet and lifestyle are not sufficient to relieve constipation on their own, drug treatments may help.

Surgery is an option of last resort which may be necessary if your daughter has an obstruction in her bowel.

## Managing constipation

### Diet and lifestyle factors

**Not enough fibre in the diet** - Dietary fibre helps form stools that are easier to pass.

Insoluble fibre (found in wholemeal/wholegrain flour, bread and cereals, as well as nuts and vegetables) draws back some of the water that is being absorbed by the large intestine. This gives the stool its volume and weight which in turn stimulates the muscle contractions that move the stool through the colon and into the rectum.

Soluble fibre (found in oats, peas, beans, lentils and many fruits) absorbs water as it moves through the large intestine turning into a soft gel that is incorporated into the faeces and makes them softer.

**Inadequate intake of fluids** - While most of the fluid that enters the large intestine is reabsorbed back into the body, about 10% is used in the formation of stools that:

- have enough bulk to stimulate muscle contractions
- are soft enough to pass easily.

If fluid intake is too low, stools are likely to be harder and drier. Urine will also be a darker colour and the number of wet nappies or diapers reduced.

**Physical inactivity** - Physical activity increases blood flow and helps to tone and strengthen the body's muscles - including the muscles involved in moving food through the digestive tract. Inactivity, on the other hand, increases the likelihood of constipation.

**Changes to routine** - The natural rhythms of the body work best when we also maintain a predictable routine in our lifestyle and habits, including our toileting habits.

Changes to routine, or unpredictable routines, may disrupt the body's normal rhythms.

### Management

**Increase fibre intake** - The recommended level of fibre in the diet is about 14-25 grams per day depending on age. Eating foods such as whole grain cereals and breads, vegetables, legumes, and fruit especially rhubarb, pears and dried fruit including prunes or drinking prune juice is an easy way to boost the intake of fibre in the diet. Foods that are high in fibre have a natural laxative effect.

Some nutritional supplements (such as Benefibre, psyllium marketed as Metamucil or Konsyl) are also high in fibre. An adequate intake of fluid is especially important when psyllium or other high fibre preparations are taken.

**Increase fluid intake** - The recommended intake of fluids for girls and women with Rett syndrome is generally the same as for the rest of the population – about 4-8 cups per day depending on age.

However, in cases of hyperventilation or drooling more fluid may be required to compensate for the fluid lost through excessive breathing or because saliva is dribbled out of the mouth rather than being swallowed.

**Increase physical activity** - Maintaining mobility and/or physical activity can also ease the symptoms of constipation.

**Establish a regular toileting routine** - You can help program your daughter's body clock for the elimination of wastes by establishing a regular toileting routine. The best time for toileting is within 30 minutes of finishing the meal to take advantage of the reflex which increases the movement of faeces in response to the stomach muscles stretching after a meal (called the *gastrocolic* reflex).

## Changes to diet and maintaining a regular daily routine can help relieve constipation

*“Veronica has been on a high fibre diet for more than 20 years. She is almost 34 years old now and I find that if I follow a strict routine and ask other people who care for her to do the same, she will be quite regular. Whereas if we start relaxing the rules with less fibre, less fruit and veggies or less fluids, after a few days she starts to get constipated. After her lunch at 12 noon, I take her to the toilet and allow her to sit for 5-10 minutes. For Veronica, this is her most frequent time for ‘success’. So, it is written into the routine for her day program, wherever she is and whoever she is with.*

*If she misses a day without using her bowels, I need to give her extra opportunities on the toilet, extra fluids, maybe more exercise, until success.*

*I should add I try not to stress her about using her bowels. I simply take her to the toilet and allow her to sit. If she doesn’t go after lunch, she will be taken to the toilet mid-afternoon, after her drink. If she still hasn’t gone, I take her again after dinner.”*



## Veronica’s high fibre diet:

### **Breakfast @ 7.45-8 am:**

*High fibre cereal (eg oatmeal) with ½ teaspoon psyllium soaked in soymilk; or two weetbix with chopped fruit and soymilk. Fruit may include chopped strawberries, kiwi fruit, pear, chopped dates and apple. We never give her more than ½ a banana because a full one seems to cause constipation. Plus two cups of drink – ½ water and ½ juice with no added sugar (eg apple, blackcurrant, pineapple or a combination). We never give her orange juice as it seems to make her itchy.*

### **Lunch @ 12 noon:**

*Two thick slices of wholemeal bread with tuna, egg, tahini and hommus; or baked beans; or kidney beans. Fruit – usually a pear or occasionally apple or mandarin or other fresh fruit in season. Pear seems to work best. Another two cups of drink same as breakfast.*

### **Mid-afternoon:**

*1 cup lite soy milk.*

### **Dinner @ 5.30 – 6.30 pm:**

*Meat or fish and veggies (including potato, pumpkin, sweet potato or peas). She likes frozen mixed veggies too. Unfortunately no broccoli – she hates it. She also likes savoury mince or casserole with veggies and mince and rice (never white rice alone, always ½ brown, ½ white). Plus 1½ cups of drink.*

*Veronica doesn’t have sweets (except the occasional ½ piece of cake with no icing) or dried fruit (maybe three dates) because sugar makes her itchy and sometimes gives her thrush.*

## Other treatments for constipation

If your daughter is still suffering from constipation, it may be necessary to use medications or other treatments.

Options in the table below are listed in order of preference. Less invasive options are always recommended as the first course of action.

Always consult your doctor before administering any drug treatment.



### Other treatments for constipation

| Treatment  | Possible side effects   |
|--|---|
| <b>High fibre preparations</b> – <i>psyllium (Metamucil, Konsyl) or other high fibre preparations such as Benefibre can be used to increase bulk of the stool</i>  | <ul style="list-style-type: none"> <li>• be careful to ensure that there is also sufficient fluid intake.</li> </ul>  |
| <b>Osmotic laxatives</b> - <i>increase the amount of water in the intestines helping to produce softer, easier to pass stools. Lactulose, Docusate, Polyethylene glycol (Movicol or Miralax) will soften the stool</i> | <ul style="list-style-type: none"> <li>• can contribute to feelings of nausea, bloating, cramping, flatulence or diarrhoea.</li> </ul>                                |
| <b>Lubricant laxatives</b> – <i>include paraffin oil and Parachoc, and coat the stool to make it easier to pass</i>  | <ul style="list-style-type: none"> <li>• avoid oil based laxatives if there is a possibility of aspiration</li> <li>• can lead to oil in the stools .</li> </ul>      |
| <b>Suppository</b> - <i>include glycerine suppositories to stimulate muscle contractions to empty the bowel</i>  | <ul style="list-style-type: none"> <li>• suitable for occasional use only</li> <li>• can be associated with rectal irritation, abdominal pain or cramping.</li> </ul> |
| <b>Enema</b> – <i>flushes liquids into the rectum and colon causing powerful muscle contractions.</i>  | <ul style="list-style-type: none"> <li>• may result in uncomfortable bloating or cramping.</li> </ul>   |

## Many parents report that the use of laxatives has been effective in managing their daughter's constipation

*"Sarah also receives an adult sachet of Movicol every day to help keep things moving. This is very important and really does make a difference. How do I know, I ran out of a script once and since I was seeing the doctor in a fortnight's time thought I would wait till then. Bad move, from now on, if I run out it's worth paying the full amount than letting her get completely blocked!"*

*"She has a sachet of Movicol in the afternoon and another the following morning if she has had no bowel motion the previous day and 45mls of Parachoc after the Movicol in the afternoon... If there is no bowel movement in 48 hours she has half a Piccolax."*

*"For the constipation we added a calcium magnesium supplement which initially worked wonders - immediately got her back on track (although she seems to not be benefitting as much from that right now). Her liquid intake is not great so I have not tried Miralax as she doesn't have enough liquid intake to benefit from that. We have used Pedilax laxatives as needed - probably about 10 times in the last 18 months."*

## Maintaining fluid intake is especially important when taking laxatives

Maintaining an adequate fluid intake is important for all girls and women with Rett syndrome, especially if laxatives are being taken to regulate bowel movements. Fluids are needed to soften bowel movements and make them easier to pass.

Fluid can come from drinks and fluids used in preparing foods (eg milk on cereal) as well as foods with a high fluid content (eg soups, jelly).

Individual fluid requirements vary depending on body size and fluid losses. However if your daughter is having regular wet nappies or diapers during the day then it is likely that she is receiving enough fluid.

*"All I can say is with the constipation medications we have found you need to have a girl that takes in lots of liquids or they don't work (unless they are like Movicol which are taken with plenty of water)."*

## Surgical management of constipation

If your daughter develops symptoms indicating she may have an obstruction in her bowel and this cannot be resolved through conventional medical treatment, she may require a general anaesthetic to enable the obstruction to be removed manually.

## 2.5 Abdominal bloating

The term, “abdominal bloating”, is used to describe swelling of the abdominal area between the chest and the hips. It can be accompanied by a feeling of tightness and fullness. It may also be associated with discomfort and pain.

Abdominal bloating is reported to affect up to one third of the general population, but like other gastro-intestinal problems, it occurs more frequently in Rett syndrome.

### 2.5.1 What causes abdominal bloating?

In Rett syndrome, the most likely cause of abdominal bloating is the intake of excess air due to the tendency to hyperventilate, breath hold and swallow air.

It may occur on its own but it is more commonly associated with other gastro-intestinal problems such as reflux, delayed gastric emptying, and/or constipation.

*“Esther started swallowing air. She burps and passes the air a lot and I am massaging her stomach as often as I can to help it. It’s worse when she sits for a while. That is why she is usually standing or walking.”*



Pain, anxiety and excitement can also exacerbate abdominal bloating which often decreases at night as hyperventilation is extremely rare in sleep.

*“One thing with Sarah that does bother me is the bloating of her stomach. She does this when holding her breath (which is on a regular basis). When she bloats her back arches forward and makes her stomach look very large and it feels very hard.”*

### 2.5.2 How do I know if my daughter has abdominal bloating?

Your daughter may have abdominal bloating if:

- she seems to be experiencing pain in her abdomen in association with distension in the abdominal area.

Clinical tests for abdominal bloating may include:

- feeling the abdomen with gentle pressure to see whether and how much resistance there is to the pressure exerted
- monitoring breathing patterns to determine whether there is breath holding or air swallowing.

Your doctor will also check carefully for other gastro-intestinal problems such as reflux, delayed gastric emptying and constipation.

If your daughter is experiencing very severe pain in her abdominal region and/or has a very tense abdomen, her doctor may recommend that she needs more extensive tests to determine whether other conditions may be contributing to these symptoms (including whether she has other problems in the gastro-intestinal tract).

### 2.5.3 What can be done to relieve abdominal bloating?

Effective management of constipation can alleviate symptoms of abdominal bloating.

Drug treatments may be tried to reduce bloating but they have limited effectiveness.

#### Drug treatments

If bloating is due to excess gas, drug treatments such as **Simethicone** may help to reduce the surface tension of gas bubbles and reduce bloating.

If anxiety is contributing to the bloating, **serotonin reuptake inhibitors** or other drugs may help.

However, as with all drug treatments, these drugs have side effects which may cause other problems.

Drug treatments need to be taken only on a doctor's advice and their effects carefully monitored.

#### Surgical treatments

If bloating is very severe and causing a lot of discomfort, a gastrostomy procedure may be used to release the build-up of air. This may make a difference, but only allows air to be removed from the stomach not further down the gut. Ask your doctor or health professional to show you how to vent air through the gastrostomy.

### A gastrostomy can release the build-up of air

*"Maree has been admitted to hospital many times because of wind. On her latest admission, the Xray showed she had a lot of gas. The radiologist commented, "No wonder she was vomiting with that amount of wind."*

*She seems better if we give her probiotics. 10mls of alovera juice each day also seems to reduce her abdominal pain.*

*When Maree has a distended stomach due to a lot of gas, a catheter bag is fitted to her peg so as to let the air out and to decompress her stomach.*

*I am going to suggest that her house staff use the catheter bag at night if Maree isn't sleeping and is air swallowing a lot even though her stomach may not be distended and also give her some Motilium. Hopefully this will stop the accumulation of gas as it is always between 5 to 7am when she has ended up vomiting."*

#### AN IMPORTANT NOTE ABOUT ...the possibility of gallbladder dysfunction

Gallbladder dysfunction (including the presence of gallstones in the gallbladder) can occur more frequently in girls and women with Rett syndrome than in the general population.

Signs of gallbladder dysfunction may include abdominal pain, vomiting and fever.

If more common causes of abdominal pain and discomfort have been excluded, the possibility of gallbladder dysfunction should be considered.

Problems with the functioning of the gallbladder can be tested by an ultrasound or by a special nuclear scanning test called a HIDA scan.

If the gallbladder is not functioning properly, it may need to be removed via a surgical procedure called a cholecystectomy.

Reassuringly, the body can function quite well without a gallbladder.

## Final words from families

*"Luckily Sarah has not had too many problems in this area. She has always been a great eater and generally doesn't have a problem swallowing. She has always been able to eat most foods (generally eating what the family eats and tends not to like mushy food).*

*She sometimes has diarrhoea and is sometimes constipated, but generally regular and normal. When Sarah was young she was always very skinny even though she ate very well. We were always trying to fatten her up feeding her lots.*

*Now she is 23 we have to have her on a diet because she still continues to love food and she will eat whatever is offered to her... and more... and is putting on the weight.*

*She is also not as active as she was, so maintaining her weight is important. She has managed to maintain her weight at 60kg.*

*Generally Sarah is very well, rarely sick, still walking pretty good and generally happy."*



*"We've been lucky with Hannah and have been able to manage her nutrition and digestive health without any particular issues.*

*She eats without problems and is a good healthy weight for her size. To manage constipation, she has a Dulcolax suppository most days followed by a bowel motion. Sometimes if she swallows air, she has a chewable Rennie (an antacid) which helps with expelling any wind causing her pain.*

*As I said, we feel we are lucky."*

*"Deciding to get the gtube was the hardest part. Once we did that, the rest was easy. Within a week, we felt like we had been in the routine forever. Within a month, I wished I had done it a year ago."*

*"Selena had her gtube done when she was only three after her reflux had become so bad she was down to 18lbs and in a state of chronic dehydration. Although I fought it at the time, it has proven to be one of the best medical decisions made for her. She gained weight right away and has maintained a good weight ever since. She is never dehydrated because even if she doesn't feel well she can get all the calories and fluids she needs. She never 'wears her medications' or refuses to take them because everything just goes right in the gtube! Once she healed, life became so easy."*

*"After years of treatment, these days Sarah is one of the happiest people I know. She never cries, whines or whimpers. Her GI system is under control. She takes 1600 ml of Peptamin jr daily along with 400 ml Pedialyte and another 1000 ml water. She's done growing and is perfect at 4'8" and 90 pounds. She has not cried once in nearly ten years!"*

# Glossary

|                                  |   |
|----------------------------------|---|
| <b>Abdominal bloating</b>        | distension of the abdominal region often caused by excess gas   |
| <b>Aerophagia</b>                | excessive air swallowing  |
| <b>Anaemia</b>                   | low levels of haemoglobin in the blood  |
| <b>Anal sphincter</b>            | the muscle that regulates the opening and closing of the anus through which digestive wastes are eliminated   |
| <b>Anus</b>                      | the external opening at the bottom of the digestive tract   |
| <b>Aspiration</b>                | food or fluids inhaled into the respiratory tract or lungs instead of the digestive tract (sometimes referred to as pulmonary aspiration)                             |
| <b>Autonomic nervous system</b>  | controls body functions that occur unconsciously (eg digestion, circulation, respiration etc)   |
| <b>Bile</b>                      | liquid produced by the liver and stored by the gallbladder for release into the small intestine to break fats down into fatty acids                                   |
| <b>Bolus</b>                     | a “ball” of food after it has been chewed and swallowed   |
| <b>Bruxism</b>                   | excessive grinding of the teeth and/or clenching of the jaw   |
| <b>Calorie</b>                   | a measure of the energy-producing potential of food   |
| <b>Cholecystectomy</b>           | surgical removal of the gallbladder   |
| <b>Colon</b>                     | the last part of the digestive tract in which water is reabsorbed back into the body and wastes are processed ready for elimination – also called the large intestine |
| <b>Constipation</b>              | infrequent or difficult passage of stools   |
| <b>Domperidone</b>               | a drug used to treat reflux which increases the frequency or strength of muscular contractions in the digestive system  |
| <b>Duodenum</b>                  | the first part of the small intestine responsible for breakdown of food and regulating the rate at which the stomach empties  |
| <b>Dysmotility</b>               | slow or restricted movement of food or fluids in the digestive tract due to poor muscle action – may also be described as gastro-intestinal dysmotility               |
| <b>Dysphagia</b>                 | difficulty in swallowing  |
| <b>Endoscopy</b>                 | thin flexible viewing instrument inserted down the oesophagus to observe the health and workings of the upper gastro-intestinal system                                |
| <b>Enema</b>                     | flushing of liquids into the rectum and colon to stimulate muscle contractions for treatment of constipation  |
| <b>Enteral Nutrition Support</b> | the delivery of nutrients via a tube inserted directly into the stomach, duodenum or jejunum  |
| <b>Enteric nervous system</b>    | controls mechanical and chemical processes in the digestive system – part of the autonomic nervous system   |
| <b>Enzymes</b>                   | complex proteins that produce a chemical change in the body such as breaking down food molecules  |
| <b>Erythromycin</b>              | mainly used as an antibiotic but in low doses can also be used to stimulate muscle contractions in the digestive system   |
| <b>Epiglottis</b>                | a flap of cartilage at the back of the throat which covers the windpipe during swallowing to prevent food and fluids passing into the lungs                           |
| <b>Faeces</b>                    | waste products of digestion, including bacteria – also called a stool   |
| <b>Fundoplication</b>            | a surgical procedure in which the upper part of the stomach (fundus) is wrapped around the lower end of the oesophagus and stitched into place                        |
| <b>Fundus of the stomach</b>     | the upper part of the stomach   |
| <b>Gallbladder</b>               | small organ responsible for storing bile produced by the liver ready for release into the small intestine to help digest fats   |
| <b>Gallstones</b>                | small stones in the gallbladder which may result from the crystallisation of excess cholesterol in bile or from the failure of the gallbladder to fully empty         |

|   |   |
|---|---|
| <b>Gastric dumping syndrome</b>                       | undigested food is emptied too quickly from the stomach   |
| <b>Gastrocolic reflex</b>                             | a reflex action that increases the muscle contraction in the colon in response to the stomach muscles stretching after a meal   |
| <b>Gastro-jejunosomy</b>                              | implantation of a feeding tube via a gastrostomy with the tip in the jejunum in the small intestine – also referred to as PEG-J tube  |
| <b>Gastro-oesophageal reflux disease</b>              | the back flow of stomach acids into the oesophagus with associated complications – also referred to as GORD or GERD depending on whether English or American spelling is used   |
| <b>Gastroparesis</b>                                  | delayed emptying of the stomach which can contribute to reflux  |
| <b>Gastrostomy</b>                                    | implantation of a feeding tube into the stomach with an opening onto the abdomen through which feeds are delivered – also referred to as G tube   |
| <b>H2-blockers</b>                                    | category of drugs that reduce acid secretion by the stomach used to treat reflux  |
| <b>Hormones</b>                                       | chemicals released by a cell, gland or organ in the body to stimulate a response in other cells   |
| <b>Ileocecal valve</b>                                | a muscle separating the small intestine from the large intestine which prevents the flow back of digestive materials  |
| <b>Ileum</b>  | the last section of the small intestine   |
| <b>Intestinal malrotation</b>                         | abnormal alignment of parts of the small and large intestine which can lead to obstruction of the small intestine and its blood supply  |
| <b>Jejunum</b>  | the middle section of the small intestine   |
| <b>Joule</b>  | the metric unit of energy used that shows energy content of foods   |
| <b>Kilocalorie</b>                                    | a measure of the energy-producing value of food – 1 kilocalorie = 1000 calories   |
| <b>Kilojoule</b>                                      | 1000 joules (4.184 kilojoules = 4,184 joules = 1 Calorie = 1 kilocalorie = 1,000 calories)  |
| <b>Laparoscopic surgery</b>                           | an operation performed via a flexible fibre-optic instrument (laparoscope) inserted through a small incision in the abdominal wall with magnification of the surgical site via TV monitors – also referred to as minimally invasive surgery or keyhole surgery      |
| <b>Large intestine</b>                                | the last part of the digestive tract in which water is reabsorbed back into the body and wastes are processed ready for elimination – also called the colon   |
| <b>Larynx</b>   | situated in the neck, the larynx is involved in breathing and production of sound. It protects against the aspiration of food or fluids into the lungs – commonly called the voice box  |
| <b>Laxative</b>                                       | taken orally, help to produce softer, easier to pass stools   |
| <b>Liver</b>  | large organ located on the upper right side of the abdomen with multiple functions including filtering blood from the digestive system, removing toxins from the body, producing bile to digest fats, processing nutrients, and helping to regulate body metabolism |
| <b>Lower oesophageal sphincter</b>                    | the muscle at the bottom end of the oesophagus helps to prevent back flow of stomach acids  |
| <b>Milk of magnesia</b>                               | a drug used to treat constipation through the absorption of water into the stool making it easier to pass   |
| <b>Multichannel intraluminal impedance monitoring</b> | a test for reflux   |
| <b>Naso-gastric tube</b>                              | a feeding tube inserted down the nose into the stomach  |
| <b>Nissen fundoplication</b>                          | a surgical procedure in which the upper part of the stomach (fundus) is wrapped around the lower end of the oesophagus and stitched into place  |
| <b>Oesophageal pH monitoring (pH probe study)</b>     | a test for reflux in which the acid in the oesophagus is monitored usually over a 24 hour period  |
| <b>Oesophagus</b>                                     | the part of the digestive tract that connects the throat to the stomach   |

|                                      |  |
|--------------------------------------|--|
| <b>Oropharangeal</b>                 | relating to the mouth or throat  |
| <b>Palpation</b>                     | feeling and exerting slight pressure on the outside of the body – used to test for abdominal bloating  |
| <b>Pancreas</b>                      | a small organ located behind the stomach releases enzymes and digestive juices into the small intestine to help digest food (particularly proteins), also produces insulin which helps to control blood sugar levels |
| <b>Para-oesophageal hernia</b>       | when the stomach bulges up through the diaphragm into the chest  |
| <b>Peristalsis</b>                   | wave-like muscular contractions that propel food and fluids along the digestive tract  |
| <b>Pharynx</b>                       | the part of the throat situated below the nasal cavity, behind the mouth and above the oesophagus – plays a critical role in swallowing  |
| <b>Prokinetic</b>                    | A type of drug that increases the frequency and strength of contractions in the digestive tract – used to treat reflux and delayed gastric emptying  |
| <b>Proton pump inhibitors</b>        | a type of drug that reduces the amount of acid produced by the stomach – used to treat reflux  |
| <b>Pyloric sphincter</b>             | a muscle at the bottom of the stomach that opens to allow the contents of the stomach to empty slowly into the small intestine   |
| <b>QT interval</b>                   | a measure of the timing and rhythm of the heartbeat from the electrocardiograph (ECG or EKG ) test.  |
| <b>Radionuclide scintigraphy</b>     | captures images of the inner workings of the body's organs based on recording gamma rays from previously ingested radioactive chemical tracers – used to test for aspiration   |
| <b>Rectum</b>                        | the last part of the large intestine which temporarily stores digestive wastes before they are eliminated from the body  |
| <b>Reflux</b>                        | the back flow of stomach contents into the oesophagus  |
| <b>Rumination</b>                    | Frequent regurgitation of previously ingested food into the mouth  |
| <b>Simethicone</b>                   | a drug that reduces the surface tension of gas bubbles in the digestive system – used to treat abdominal bloating  |
| <b>Serotonin reuptake inhibitors</b> | medicines used to treat anxiety disorders – may help to reduce abdominal bloating where anxiety is considered to be a contributing factor  |
| <b>SOMA</b>                          | acronym for Schedule for Oral Motor Assessment – a specialised assessment tool for feeding, chewing and swallowing abilities   |
| <b>Small intestine</b>               | the part of the digestive tract that follows the stomach in which most of the digestion and absorption of nutrients takes place – also called the small bowel  |
| <b>Sphincter muscle</b>              | circular muscle that closes or opens to either contain the contents of an organ or bodily passage or release its contents  |
| <b>Stoma</b>                         | a surgically created opening in the body   |
| <b>Stool</b>                         | waste products of digestion, including bacteria – also called a faeces or feces  |
| <b>Suppository</b>                   | a medication inserted into the rectum to stimulate muscle contractions – used to treat constipation  |
| <b>Trachea</b>                       | a tube structure that connects the pharynx and larynx to the lungs allowing air into the lungs   |
| <b>Upper oesophageal sphincter</b>   | a circular muscle at the top of the oesophagus that opens during swallowing to enable food and fluids to pass into the esophagus   |
| <b>Videofluoroscopy</b>              | a special x-ray procedure used to test for swallowing ability and/or difficulties  |
| <b>Villi</b>                         | small, finger-like projections that increase the surface area and absorptive capacity of the walls of the intestine.   |
| <b>Volvulus</b>                      | twisting of the intestine which may cause a blockage and/or cut off blood flow causing damage to the intestine.  |

## Resources and references

In today's information technology age, there's an abundance of information posted on the internet. Some of this information is supported by the evidence, some of it is unreliable, and some of it is actually harmful. When searching the internet for information, we recommend you favour websites that have been endorsed by reputable research institutions, by government health authorities or by relevant professional bodies. We have listed a few sites below that fit these criteria. The sites we have listed are mainly sourced from Australia, the UK and the US. There are many more in countries around the world committed to promoting nutritional and digestive health for those with rare disorders.

### Nutritional health

**Nutrition Australia** (non-government, not for profit organisation)

with expert and community membership committed to promoting health and wellbeing in the community.

<http://www.nutritionaustralia.org/>

**US Department of Agriculture – Choose my plate**

<http://www.choosemyplate.gov/food-groups/>

<http://www.choosemyplate.gov/healthy-eating-tips.html>

**British Nutrition Foundation (UK)**

<http://www.nutrition.org.uk/>

<http://www.nutrition.org.uk/healthyliving>

### Increasing energy intake

<http://kidshealth.schn.health.nsw.gov.au/fact-sheets/high-energy-eating-infants-toddlers>

<http://www.medicalhomeportal.org/issue/power-packing>

### Digestive health

**National Digestive Diseases Information Clearinghouse** is an evidence-based site that provides a user-friendly summary of how the digestive system works and common gastro-intestinal disorders. Here's the link to a listing of different digestive diseases including a section on how the digestive system works

<http://digestive.niddk.nih.gov/ddiseases/a-z.aspx>

### Rett syndrome websites

**Rett syndrome research in Australia**

Aussie Rett <http://www.aussierett.org.au>

InterRett <http://www.interrett.org.au>

**International Rett Syndrome Foundation IRSF**

<http://www.rettysyndrome.org/>

The Rett syndrome specific growth charts are available on the IRSF website.

<http://www.rettysyndrome.org/understanding-rett-syndrome/living-with-rett-syndrome/symptom-management/health-matters/growth-references>

**Rett UK**

<http://www.rettuk.org/rettuk-public/rettuk.html>

### Main references used to inform this booklet

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## Developing the content of this booklet

In developing the evidence-based content for this booklet, we have adopted a staged methodology:

- conduct of a comprehensive literature search to identify relevant research and statements on growth and nutrition, and gastrointestinal problems in Rett syndrome
- consultation with parents and caregivers on their experiences of poor growth, calorie intake, feeding difficulties and gastrointestinal problems
- preparation of a draft checklist for clinicians on assessment and clinical management (based on statements from the literature search, parent input, and questions requiring further clarification)
- appointment of an international panel of expert clinicians from different specialities who reviewed successive drafts of the recommendations on assessment and management until agreement was reached on their content.

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

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**AussieRett:**  
Australian Rett  
Syndrome Study

**InterRett:** International  
Rett Syndrome Database  
International Rett Syndrome Foundation



The **Royal Children's  
Hospital** Melbourne



**Murdoch Childrens  
Research Institute**  
*Healthier Kids. Healthier Future.*



Government of **Western Australia**  
Department of **Health**  
Child and Adolescent Health Service

the  
**children's  
hospital** at Westmead

