

# SCOLIOSIS IN RETT SYNDROME

A collaboration between parents, clinicians and researchers





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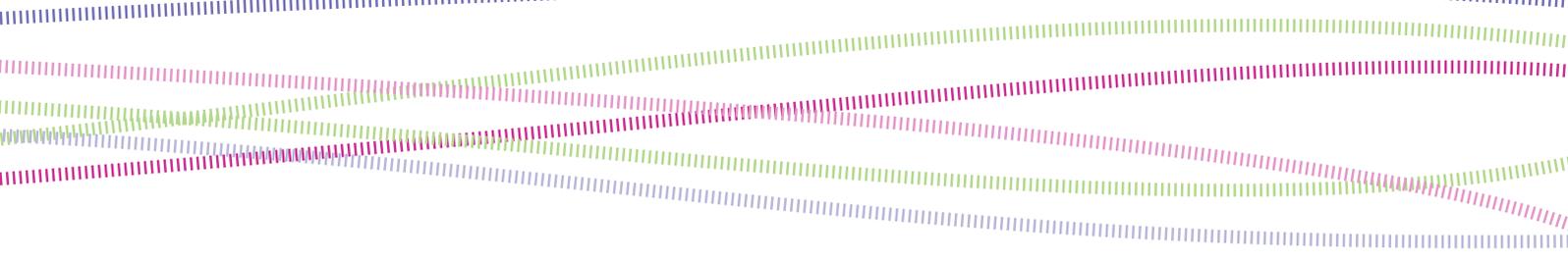
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### Parents have also provided input to the development of this booklet.

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## Table of contents

### **Purpose**

The purpose of this booklet .....	4
-----------------------------------	---

### **Background**

What is Rett syndrome? .....	5
What is scoliosis? .....	5
How common is scoliosis in Rett syndrome? .....	5
Does my daughter have scoliosis? .....	6

### **Diagnosis and non-surgical management of scoliosis**

Monitoring and treatment before a diagnosis of scoliosis .....	8
Monitoring after a diagnosis of scoliosis .....	8
Medical care for the spine with scoliosis .....	9
Physiotherapy and exercise .....	10
Spinal bracing for scoliosis .....	12

### **Surgical treatment of scoliosis**

Indications and goals of surgery .....	14
Before scoliosis surgery .....	14
The operation itself .....	15
After scoliosis surgery .....	15
Going home after surgery .....	17
Surgery: Not always the best option for those with very severe scoliosis .....	19

<b>Final words from families</b> .....	20
--	----

<b>Additional resources for families and clinicians</b> .....	21
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<b>Glossary of terms</b> .....	22
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<b>Contact details</b> .....	23
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## The purpose of this booklet

Rett syndrome is a rare neurodevelopmental disorder affecting approximately 1 in 8,500 live born females. Given the rarity of this condition, most doctors have limited experience treating patients with this disorder. A common complication of Rett syndrome is the development of **scoliosis** (curvature of the spine).

There is limited information in the medical literature regarding the management of scoliosis. We have therefore developed some guidelines to provide information for parents and carers and to be helpful to doctors caring for girls and women with Rett syndrome. The main aim of managing scoliosis is to maximise function whilst preventing the progression of the spinal curve. These guidelines are not a formula for every girl with Rett syndrome who develops scoliosis but rather, they highlight the key features of accepted current best practice. They are based on the available medical literature and on expert clinical opinions from various fields.

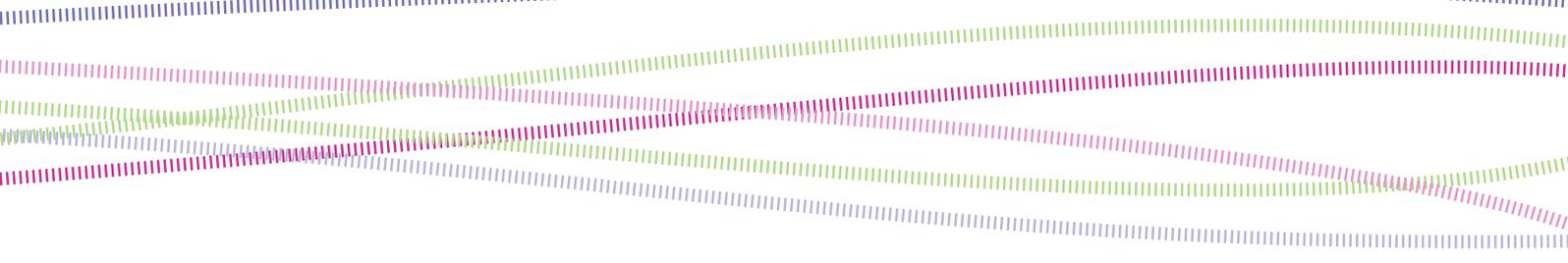
This booklet was designed to outline some recommended management guidelines for scoliosis in Rett syndrome and provide the relevant background information for parents and clinicians. There is a glossary of terms that you may find helpful on page 22 and the words contained in the glossary are printed in **bold** the first time they are used in the text.

In addition, we have gathered first-hand accounts from parents regarding their daughter's scoliosis. These parents have a wealth of knowledge, understanding and experiences of the different aspects of management of scoliosis in Rett syndrome. Parents form an important partnership with their daughter's doctors, providing perspective and goals that ultimately guide the treatment.

## JOURNAL REFERENCE FOR THE SCOLIOSIS GUIDELINES

Downs J , Bergman A, Carter P, Anderson A, Palmer GM, Roye DP, van Bosse H, Bebbington A, Larsson E-L, Smith BG, Baikie G, Fyfe S, Leonard H (2009). Guidelines for management of scoliosis in Rett syndrome patients based on expert consensus and clinical evidence, SPINE, 1;34(17):E607-17.





## What is Rett syndrome?

Rett syndrome is a neurodevelopmental disorder which mainly affects females and results in physical and intellectual disability. It is usually associated with a fault (mutation) in the *MECP2* gene.

The girl with Rett syndrome usually has a normal period of development during infancy. This is followed by a period of rapid developmental regression when some skills are lost. These include loss of communication skills and purposeful use of hands. Walking and other gross motor skills are affected and stereotyped hand movements appear.

The preschool to primary school years are characterised by some stabilisation in developmental skills, and whilst variable, there can be some improvement in gross motor skills. A scoliosis may develop during this period. Over time, walking skills may be lost.

Rett syndrome can also be associated with:

- altered breathing patterns (such as breath holding),
- poor growth,
- altered muscle tone,
- epilepsy,
- **osteoporosis**,
- feeding difficulties,
- constipation, and
- altered sleep patterns.

## What is scoliosis?

The human spine is made up of bony vertebrae aligned on top of each other in a vertical column. Scoliosis occurs when there is a side to side deviation in the alignment of the vertebrae and a curve develops. There can also be some associated twisting of the spine along its long axis as well as chest wall deformity.

In Rett syndrome, scoliosis usually develops because of altered neurological function and this is called a “neuromuscular scoliosis”. The exact reason for the development of scoliosis in Rett syndrome is not entirely clear, but it is thought to be related to muscle weakness or tightness, altered muscle tone and limited mobility. However, “idiopathic scoliosis” is the term used when a scoliosis develops in otherwise normal healthy people for no apparent reason.

Scoliosis can stiffen the spine as it becomes more severe, and may make it harder for the girl or woman with Rett syndrome to balance whilst sitting, standing or walking. This leads to decreased function and quality of life. In addition, large curves can alter the position of vital organs in the chest and abdomen.

## How common is scoliosis in Rett syndrome?

The chance of scoliosis developing in Rett syndrome increases with age. Approximately one quarter of girls develop scoliosis by six years of age and three quarters by 13 years of age. Girls with Rett syndrome who never learn to walk have been shown to be twice as likely to develop scoliosis as girls who have learned to walk.

## Does my daughter have scoliosis?

The earliest sign of a scoliosis developing is often leaning to one side during sitting, standing or walking. The curve may initially vary in appearance in different body positions. With time it may become fixed and rigid. If you notice your daughter leaning to one side or you are concerned that her back is not straight you should arrange for your daughter to see her doctor. The doctor will carefully inspect the spine and perform physical examination manoeuvres, such as the forward bending test to assess for spinal curvature (see picture).

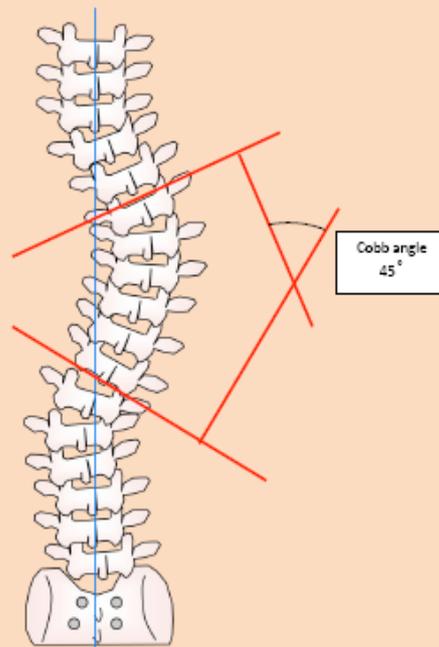
If the spine appears straight and fully mobile on clinical examination, an X-ray may not be necessary. If the doctor finds that a scoliosis is present, he/she will arrange a spinal X-ray. The X-ray will confirm the presence of a scoliosis, define its contour and the size of the curve.

The size of the curve is reported as an angle from the X-ray using the Cobb method, which is the standardized method for measuring spinal curves world-wide. The **Cobb angle** is determined by drawing lines from the vertebrae at the upper and lower ends of the curve. The Cobb angle is the angle made by intercepting the perpendiculars from these two reference lines (see diagram).

To find the Cobb Angle, draw lines perpendicular to those lines. The perpendicular lines intersect at an angle. The degree measurement of that angle is the magnitude of curvature (Cobb Angle).

To find the Cobb Angle:

1. Locate the most tilted vertebrae at the top (proximal end vertebra) and draw a parallel line to the vertebral endplate.
2. Locate the most tilted vertebrae at the bottom (distal end vertebra) and draw a parallel line to the vertebral endplate.
3. Erect intersecting perpendicular lines from these.
4. The angle formed is Cobb's angle.



Diab, M. "Cobb Angles." Diagram. Vitale et al. Assessment and Decision Making in Adolescent Idiopathic Scoliosis: Use of the Lenke Classification System, 2009.



# Diagnosis and non-surgical management of scoliosis



## Monitoring and treatment before a diagnosis of scoliosis

Scoliosis occurs frequently in Rett syndrome and therefore each visit to your daughter's doctor should include a physical examination of the spine. We suggest that the back be assessed by the doctor for scoliosis approximately every six months.

**Physiotherapy, occupational therapy, hydrotherapy, hippotherapy** and other **activities of daily living** should aim to:

- develop, maintain, and promote walking for as long as possible,
- strengthen the back muscles, and
- promote correct posture while sitting and sleeping.

Therefore, daily activities should be practised to develop and maintain strength and function. Regular assessment is key.

## Monitoring after a diagnosis of scoliosis

If your daughter does develop a scoliosis, this should be monitored and a referral to an **orthopaedic surgeon** should be made. An orthopaedic surgeon will assess the severity of the scoliosis and regularly monitor its progression. He/she will also take into account your daughter's previous medical history, her general health, growth and development.

The physical assessment will include measurement of your daughter's height and weight, examination of the spine, assessment of posture, muscle tone, and sitting, standing and walking skills.

The orthopaedic surgeon or your daughter's general doctor will usually assess the spine every

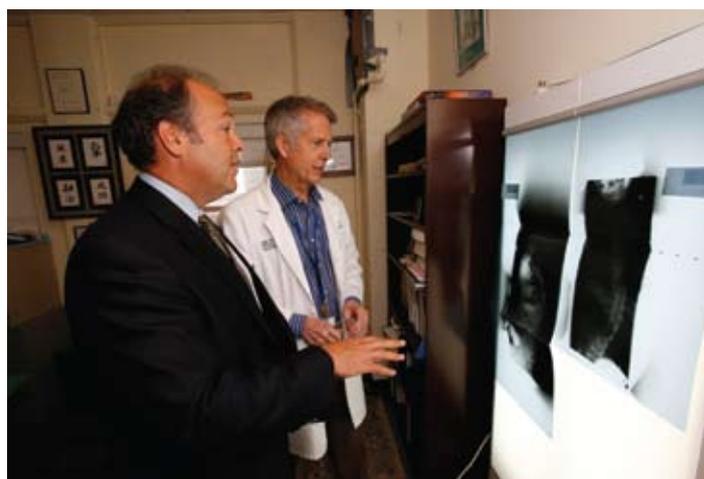
six months or so. It may be recommended that your daughter be assessed more frequently if:

- she has never learned to walk,
- she has very low muscle tone,
- she is having a growth spurt,
- the scoliosis developed at a very young age, and/or
- the scoliosis is severe.

X-rays of the spine will be repeated at various intervals and these will help the surgeon assess the rate of progression of the scoliosis.

### When should X-rays be taken?

- An X-ray of the spine will be taken on the first visit if a curve is noted.
- X-rays should be taken approximately every six months before skeletal maturity if the Cobb angle is  $>25$  degrees.
- X-rays should be taken approximately every 12 months after skeletal maturity until the Cobb angle stops changing.



## Medical care for the spine with scoliosis

General nutrition and medical care as your daughter grows and develops will have many physical benefits including benefits for the growing spine. For example, girls with Rett syndrome are very prone to developing osteoporosis. Osteoporosis can be minimised by eating a diet high in calcium (commonly found in dairy products such as milk and cheese), getting as much exercise as possible and participating in activities in the sunlight to promote production of **vitamin D** in the skin.

However, sun exposure needs to be balanced against the need to protect skin from the harmful effects of ultraviolet light. Therefore, it is necessary to consider the time of the day and the length of time your daughter is out in the sun. Talk to your doctor about monitoring your daughter's calcium intake, monitoring vitamin D levels and about safe sunlight exposure in your area. Some girls may require vitamin D supplements, particularly if they live in less sunny areas.



## Physiotherapy and exercise

There is agreement that physiotherapy has an important role to play in preserving and maybe even improving physical abilities, muscle strength and joint flexibility. We recommend that:

1. Your daughter should attempt to walk for as long and far as possible. Aim to increase the distance that your daughter can walk and/or the length of time on her feet, with or without assistance, depending on her ability (at least two hours per day when possible).
2. If your daughter cannot walk, use a standing frame for at least 30 minutes a day.
3. Practice daily stretches to maintain the range of movement of the muscles and joints. A physiotherapist may suggest types of stretches that may be used and for how long each day these stretches should be practised.
4. Talk with your physiotherapist and occupational therapist about methods of sitting that could be beneficial to your daughter's spine. This could be practising sitting on a stool or making sure that her wheelchair has adequate supports to keep her spine evenly balanced.
5. However, even with your best efforts not all girls will be able to continue to walk as they grow and mature.

Many of these activities are illustrated in the pictures on this page and also on pages 7 and 9.



Here are some descriptions of the types of physical activities in which girls with Rett syndrome participate.

*“Amy, at 16 years old, has had mild scoliosis and kyphosis for many years now and it has progressed very little in that time. I believe that this is due to the emphasis we have put on including physical activity in Amy’s daily life. For example, she does supported standing and walking for a few hours each day, active sitting when not in her wheelchair, minimal time spent in her wheelchair (she has a postural seating system on her wheelchair for these times), physio stretches weekly, swimming with exercises in a heated pool at least weekly in spring/autumn, more often in summer and holidays .... All these activities I think have helped strengthen her trunk muscles and will hopefully prevent the scoliosis worsening and therefore avoid the need for bracing and/or surgery in the future.”*

*“Our daughter Hannah is almost 10 years old. She has had scoliosis since about 6 years, and it has progressed gradually. In the last couple of years her scoliosis has increased and currently measures 31 degrees. We are attempting to ensure she gets 1 - 2 hours of walking and standing in a day (she has a standing frame at home that we love). Hannah does therapeutic horseback riding and she rides an adapted bicycle. We do a stretching session at home daily. Currently she has a program of weekly walking on a treadmill through our local hospital PT department (with a support system to hold up her weight). She’s now up to 32 minutes of walking at one time, at a speed of about .8 or .9 miles per hour.”*

We are not sure of the exact benefits of physical activities for minimising scoliosis. However, families generally agree that physical activity is beneficial to their daughter.

*“ [Physiotherapy] kept her walking longer, it kept her moving and standing longer, it strengthened her considerably and increased her endurance.”*

*“It is impossible to know if the therapy slowed down the scoliosis because Ashley’s scoliosis continued to worsen in spite of intense physical therapy. We had therapy for many years. We have no idea if the spine would have got worse faster without the therapy.”*



## Spinal bracing for scoliosis

There are many different views as to how useful bracing is for the management of scoliosis. It is generally believed that spinal bracing will not alter the progression of the curve. However, there is agreement that bracing helps sitting balance. Careful and close follow-up with an **orthotist** is needed whilst the **spinal brace** is being worn to deal with growth, adjustment of the brace for activities of daily living, and any complications caused by the brace (eg, skin irritation). Frequent brace adjustments may be needed.

When a child requires surgery at a very young age the risks of surgical complications may be increased. In some cases, the use of a well-fitted spinal brace in a young child may help delay the need for scoliosis surgery.

*“When we first put on the brace, I thought that she would not tolerate it but instead we saw a look like a sigh of relief, so I would assume it alleviated some pain and pressure.”*

*“We were able to postpone surgery until she reached her teens.”*

*“Jenn had **hypotonia** and it was a major effort to hold herself up. With the brace she didn't have to worry about it, it held her up! That gave her the energy to do other things, to interact more with people.”*

Wearing a brace can be difficult and complications can include pressure sores, difficulty breathing, general discomfort, skin irritation and exacerbation of **gastro-oesophageal reflux**. Wearing a brace can make it difficult to practise mobility skills and physical activity and may lead to loss of trunk strength and flexibility.

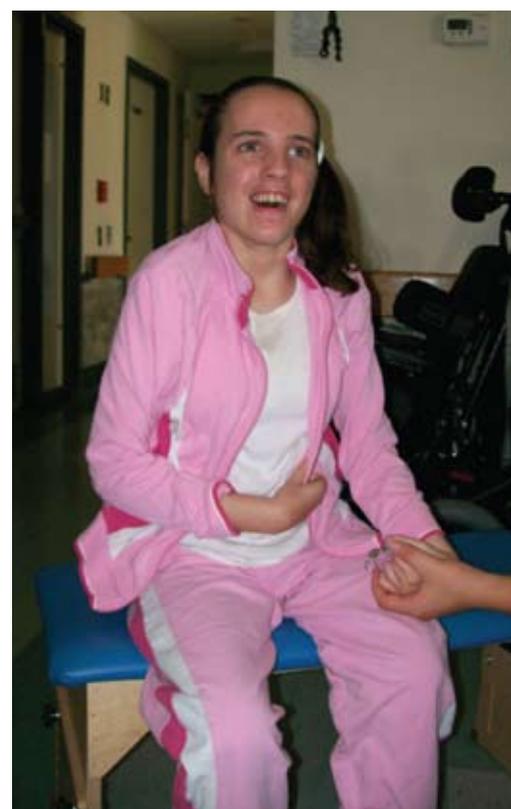
*“We had problems with the G-tube leaking due to pressure on her stomach which caused skin breakdown around the tube insertion site. It also decreased her independence as she was unable to sit up or roll when on the floor due to the bulkiness of the brace.”*

*“The brace made it difficult to care for her physically. It was harder to transfer her, difficult to change her diapers, and hot in the summer.”*

Parents should work in conjunction with orthopaedic surgeons, physiotherapists and orthotists to determine if bracing is appropriate for their child.



# Surgical treatment of scoliosis



## Indications and goals of surgery

If the scoliosis does continue to progress and your daughter's Cobb angle becomes greater than 40-50 degrees, your surgeon will consider and may advise that she have **spinal fusion** surgery to correct and prevent further progression of the scoliosis. Correction of curves that are larger may carry higher operative risks. The need for surgery is considered on a case-by-case basis.

Surgeons usually try to wait until after the child is 10 years of age before performing spinal fusion. They may often recommend bracing, physiotherapy and physical activity to delay surgery in order to allow for as much growth as possible. The aims of surgery are to achieve a balanced, fused spine so that in upright positions such as sitting or standing, the shoulders and hips are more level. This improvement in balance will increase her comfort level while sitting and will help with transfers during everyday activities.

*"It was a very difficult decision for us to put Louise through major high-risk surgery - a spinal fusion and insertion of rods. We were anxious about the anaesthetic, risk of infection and slow recovery. However, after I had attended a parent information session where there was a presentation from another well-respected orthopaedic surgeon, it was easier to make the decision. This surgeon talked about the 'window of opportunity' whereby if your child met all the criteria he listed, it was the optimum time for surgery."*

Spinal fusion is a major operation and is usually performed in a specialist centre due to the risk of complications. This can be a very stressful time for families and it is important to discuss the reasons for surgery, the expected medical and surgical plan in detail, and the surgical outcomes with your child's doctors.

## Before scoliosis surgery

Your daughter's body needs to be as strong as possible before surgery to decrease peri-operative risks and improve recovery time. Looking after the health of your daughter's bones as described in *Medical care for the spine with scoliosis (page 9)* will be helpful. Leading up to the surgery:

1. The therapist will assess your daughter's day to day skills such as sitting, standing and walking.
2. If your daughter's weight is very low for her height, nutritional supplementation may be necessary for a period of time before surgery. Your doctor may recommend short-term insertion of a feeding tube into your daughter's stomach or small intestine to provide extra calories.
3. Your daughter will have routine blood tests prior to surgery to detect and correct any abnormalities.
4. The anaesthetist will assess your daughter's fitness for anaesthesia.
5. Additional medical assessments may include, amongst others, checking the oxygen saturation levels of the blood by a probe attached to the finger. An electrocardiogram (ECG) may be performed to check the heart rate and rhythm and measure the QTc value.
6. Your daughter may be referred to other specialists to ensure that any other underlying medical problems such as epilepsy or gastro-oesophageal reflux are well managed.
7. Now is also an important time for families to talk further with doctors and other staff about what to expect during the hospital stay.

*"There had been a thorough pre-operative assessment done weeks before, involving an ECG (which diagnosed mild prolonged QT Syndrome - a surprise to us), blood tests, review with a respiratory physician and x-rays. She was also monitored by an oxygen saturation monitor at home overnight."*

## The operation itself

There are a number of different techniques to reduce the curvature and stabilize the spine. The extensive preoperative work-up described above will play a large role in informing surgical decisions. The most common spinal fusion procedure performed for girls with scoliosis and Rett syndrome uses posterior instrumentation.

No two cases are the same, and you will play a role in preoperative discussions. It is important to understand the surgical options and the preferred plan for your daughter. You will discuss the details of your daughter's posterior spinal fusion, such as the incision(s), the type of rods to be used, and the levels of the spine that will be included in the fusion. In some cases the fusion extends to the bones of the pelvis for additional stability. The curve is corrected by an amount your surgeon deems safe, and metal rods are inserted in order to stabilize the spine in its new position.

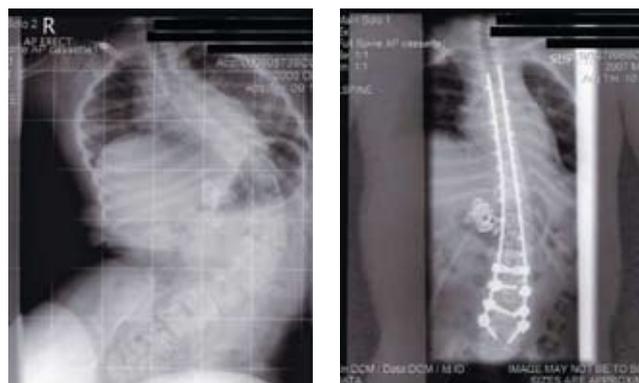
In some cases, an anterior approach to the spine will be used in addition to the posterior approach described above. An anterior approach allows for different types of instrumentation to be used, and allows for the surgeon to stabilize the anterior aspect of the spine in such a way that is not possible using the posterior approach alone. The risks and benefits of the anterior approach will be discussed in detail during preoperative visits.

Scoliosis surgery such as the approaches described above may take three hours or more, often a stressful waiting time for parents. In addition to the careful preoperative planning described above, many parents must plan time away from their jobs and care for other children.

*"Brooke had scoliosis surgery when she was 13 years old. The time leading up to the operation is very emotional to say the least.... Brooke on the day was fine - guilt from me was horrible ..... I felt so guilty but had to be strong - no sleep, worry, guilt, other kids - not to mention our jobs."*

## After scoliosis surgery

Following spinal fusion, your daughter will be admitted to an intensive care unit (ICU) for constant monitoring and care. A specialist paediatric pain team will work with you and your daughter to achieve comfortable pain relief. Pain medications and surgery can lead to constipation. Your input will be vital in providing feedback to the pain specialists, as you know your daughter best. Assisted ventilation may be required in the immediate post operative period. Careful monitoring of the health of the lungs, suctioning of the mouth and chest physiotherapy will help to prevent lung complications such as infection.



*"Recovery was much more rapid than expected. She was extubated just under 24 hours post surgery, spent one day in intensive care and a total of 7 days in the hospital. Pain management, constant suctioning of secretions to avoid lung complications and fighting constant constipation have been her biggest challenges."*

*"It was a difficult time for everyone as the operation was 10 hours long. She spent 3 days in ICU and was ventilated as she stopped breathing during surgery. The healing process was very long and tiring. She spent 4 weeks in hospital and 3 months at home recovering.... Her spine was corrected to 40 degree. She is much better as a result of the surgery, although she has stopped weight bearing."*

*“The surgery went well. The scariest time for us as parents was after the surgery was completed. They took her out of PICU after 14 hours. Samantha slept literally almost 24 hours, waking for just a few minutes. After not waking up for four days, the doctor tried to wean her off the morphine and onto Tylenol w/codeine. Her sleeping improved by maybe being awake 2 hours a day. On day six, the doctor withdrew her meds from her except for Tylenol (no codeine). She woke up almost immediately. I don’t know if all girls with Rett syndrome react to pain meds like this or just our Sami.”*

*“Tracy had full posterior spinal fusion. She had just turned 7 a month prior and I was told she was the youngest and smallest to have this done at the time. She got through the surgery with no complications, she was in ICU for 3 days and had received 2 bags of blood on the second day and one on the third day then all turned around and she was back to Tracy wanting to watch cartoons. She was now at 34 degrees. She did so well I was able to bring her home within a week. She recovered well and sooner than expected; I had to drive her back to her 3 week check which is a 5 hour drive and she didn’t complain, then 5 hours back and she did then complain. She was able to sit again (didn’t walk before surgery from different reasons). She was sleeping better once healed.”*

Mobility will be encouraged in the post operative period. Early mobility will improve your daughter’s breathing, muscle strength and function, and will help your daughter feel more comfortable. A typical pattern of mobilization after spinal surgery includes

- immediate log rolling to move around in bed,
- sitting on the edge of the bed one day after surgery,
- transferring to a chair from her bed two days after surgery,
- walking (if possible) three days after surgery, and
- over the course of the hospital stay, you should be taught how to lift and transfer your daughter.

*“She had a full length fusion with rods when she was 15 ½ years old. The day after the surgery she was able to roll from side to side and three days later she was weight-bearing. Pain management by epidural was excellent and by the time she went home a week later she was off all pain medications. Her seizures actually stopped during the first 10 days recovery time. She seemed to have grown 4 inches over night. That made us realise how bent over she had been. Her appetite increased and she smiled a lot more often.”*



## Going home after surgery

By the time the girls are ready for discharge from the hospital, they should be able to sit in their wheelchairs or take a few steps if they were able to walk preoperatively. Surgery and peri-operative medications may affect the way your daughter processes the medications she was taking prior to surgery. Medication doses may need to be adjusted.

The time to full recovery can be quite variable depending on the level of disability and the length of surgery. Wound healing is variable, but by two weeks, most wounds should be healed. If your daughter is having trouble with wound healing you may need to see a doctor or specialist more frequently for wound care.

Depending on the length of the fusion there may be some restrictions on how the child can be lifted or moved. Slings may be advisable for fusions that extend to the pelvis. Most children will tire rather quickly in this initial period but the establishment of normal daily routines should be encouraged with the expectation that endurance levels will improve. Your doctor and therapist will talk with you about the return to normal activity.

*“Once we had her home with suitable equipment (you need to plan for this well beforehand – electric hi-lo bed, customised shower commode), she recovered well and was back at school in six weeks. Additional post-operative care (for 3 weeks) was provided through Home & Community Care to give me a break, and the local council made sure the care workers were trained by the nurse in how to assist her (doing log rolls, keeping her spine straight). Additional respite was a must so that I didn’t go too stir-crazy. I also have a very understanding employer which helped enormously. The six weeks went very slowly as she was not able to do any of her normal activities such as standing, sitting in chairs other*

*than her wheelchair etc. We were provided with an excellent sling by the hospital and made sure we had a waterproof one as well so we could leave her on a sling all the time in her wheelchair or shower chair. We were also initially very nervous moving her around to shower and dress her, worrying something was going to snap or fall out. But it didn’t - and six months later, she was able to go back to her usual activities. Two years down the track, she stands in a tilt table, walks on a treadmill, uses a Meywalker, rides a bike and lies in a gel chair. In fact she is more mobile now than ever, due to an excellent intensive physiotherapy program at school.”*

*“Recovery was very hard and long. As she would do no weight-bearing and she is a big girl, caregiving was very difficult, at least for the first three months. It took nine months before Sami would be able to take really good steps in her gait trainer, which she could do really well before the curve got bad. It was about a year and a half before her full stamina returned. Another difficult thing was that she would not hold her head up. It laid on her chin. The doctor was right (again!) about it being caused by the neck muscles being pulled during surgery. Our worry was that she would never recover from it. It did take lots of physical therapy, but she did build those muscles back up and holds her head up well. Her spine is wonderfully straight now. Two and a half years out, she cannot stand as well as before the curve got to 30ish, but better than prior to the surgery. The same goes for walking in the gait trainer. Better than before the scoliosis surgery, where it had dropped to almost nothing, but not nearly as good as before scoliosis had set in.”*

Surgery for scoliosis has many advantages for girls with Rett syndrome. After recovery, families often report satisfaction with the outcomes for their daughter. Activities of daily living are mostly maintained and in some cases, improve following scoliosis surgery. Sitting posture improves and mobility is usually maintained.

*“Despite the drama of the post-op recovery, neither we nor Ella regret the decision to proceed with surgery. In fact, have asked Ella on a number of occasions (when she’s particularly bright and alert) her opinion, and she has always confirmed she is pleased to have had her back “fixed”. This remains very much the case. We agonised for some time over whether to proceed down the surgery road, but have no regrets with the result. She sits so straight and tall, and, most of the time, appears to have much better stamina. Notably, her increased comfort and more natural position enabled her to better engage people. Her general health has generally been excellent and, amazingly, despite other complications, she has remained cold/virus free for the 18 month period since her surgery....”*

Some families however have reported that sitting on the floor, embracing and moving their daughter are more difficult after surgery.

*“I believe this surgery saved her. I do fear though at times, since she is not mobile and must be transferred everywhere, that those two stainless steel rods must be watched..... everyone must be aware of her surgery. i.e., she no longer bends, anywhere. She is now 16 and a half years old.”*

*“Not being able to hold her in our lap any more as she can’t be twisted in any way. We would like to figure out how to snuggle with her as mother and daughter miss this very much.”*

*“Loss of some mobility though she never walked before or after the surgery. Not able to sit alone on the floor after surgery as she used to.”*

Post-operative appointments with your orthopaedic surgeon are needed, initially at around six weeks after the surgery and then approximately every two to three months over the first year. Some surgeons will want to check the spine annually thereafter. Feedback from families is important if there are any concerns.



## Surgery: Not always the best option for those with very severe scoliosis

Surgery may not be appropriate for some girls and women with Rett syndrome who have a very severe scoliosis. The risks of surgery may outweigh the potential benefits. In such cases, the medical management and therapy strategies described above remain the mainstay of treatment. It is important to

- keep the bones healthy as described in *Medical care for the spine with scoliosis (page 9)*,
- promote as much time in upright postures of standing and walking as possible, and
- provide supported seating to achieve a balanced sitting posture.

Monitoring and treatment of pressure sores is also necessary. Antibiotics may be required more frequently to minimize the possibility of developing a serious chest infection with restricted chest wall movement. Encouraging cough and upright positioning at times of respiratory illness are also helpful.

*"In 2003 she had gone 15 degrees since surgery in 1996 so she was now at 49 degrees. From 2003 to 2007 she had a slow rotation with her internal organs moving along with the rotation with no complications except for a little pressure on the left lung ... At this time she was experiencing other health issues so her pediatric orthopedic surgeon said to leave well enough alone as she may not be strong enough for a full double surgery. She was at 64 degrees in 2006 and took her big and last growth spurt and was at 68 degrees in 2007. Tracy has had hydrotherapy for about 9 years now so her doctors feel it is helping her as her last measurement was at 67 degrees so now that she is not growing they feel the therapist can now keep the muscles loosened better and for longer periods."*



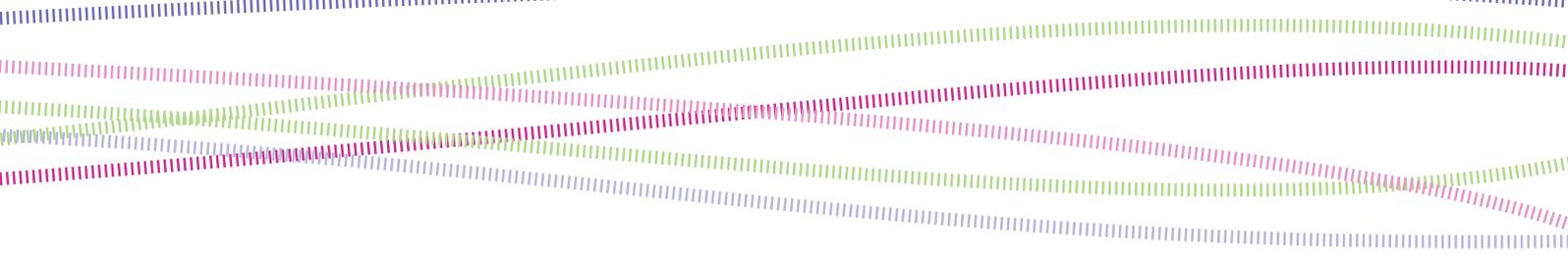
## Final words from families

*“Scoliosis seems to be a huge cause of caretaker stress .... and can affect function and cause pain for our girls. To be able to hear the stories from other parents and to learn “best practices” in the management and treatment of scoliosis ..... will be invaluable to every clinician and parent. While no two cases are exactly alike, the experiences before and after surgery will help parents feel more comfortable as they make decisions for their child.”*

*“Andrea began seeing an orthopaedic surgeon in relation to scoliosis at about 11 or 12. The scoliosis had a severe impact on her quality of life and caused her much pain. Initially, she became unsteady on her feet but as the scoliosis progressed her breathing became compromised, her skin colour grey and she became more and more bent. Her health had deteriorated to such an extent that I genuinely feared for her life. At around the time of Andrea’s 15th birthday, the specialist raised the issue of whether we should go ahead with surgery, given her general deterioration. I had a very distressing 3 days before I spoke to the neurologist who assured me that the issues were related to the scoliosis. Even after surgery was recommended, we had to wait more than 3 months for the surgery. This was a time of agony for all of us – literally for Andrea and emotionally for the rest of the family. But this changed after the surgery. Even when she was recovering, Andrea was in a different place. Her colour returned to normal and the quality of the pain was very different and it disappeared as she recovered. It was amazing to see her with a straight back again. After a few weeks, she was walking, at first with a brace. All but a few vertebrae were fused, so Andrea has virtually no flexibility in her spine but she is still walking 12 years later.”*

*“Since Abby was three, Nuria and her team of healthcare givers had been keeping an eye on the curvature of her spine. When Abby turned 8, they witnessed a rapid shift of her spine’s curve from 21 degrees to 45 degrees in six months. In February she brought her daughter and their X-ray films to meet Dr. Roye for the first time. At that point, Abby couldn’t sit up – bending off to the right – and showed a look of pain on her face. “He explained exactly what we would be facing,” explained Nuria, “and he told me the decision to have surgery was about the quality of her life. Only two weeks after her spinal surgery, Abby had her turn around. There were no more spasms, and at 12 days after surgery she was sitting perfectly straight – with no look of pain. She was brighter. Happier.” Nuria acknowledges her difficulty in making the decision for surgery for her daughter. But today she knows she made the right choice. Abby is able to enjoy her days – out of pain. She loves the water when they take her to the pool; she loves to be moving as they take her for long walks; and she loves watching her videos.”*





## Additional resources for families and clinicians

### For Families

International Rett Syndrome Foundation  
[www.rettsyndrome.org](http://www.rettsyndrome.org)

Rett Syndrome Association of Australia  
[rettaust@bigpond.com](mailto:rettaust@bigpond.com)

Rett Syndrome Association of the United Kingdom  
[www.rettsyndrome.org.uk](http://www.rettsyndrome.org.uk)

Rettnet e-mail list  
[www.rettsyndrome.org](http://www.rettsyndrome.org) - then click "online communities", then click "Rettnet"

ANZAC Rett  
[au.groups.yahoo.com/group/ANZACRett](http://au.groups.yahoo.com/group/ANZACRett)

Australian Rett Syndrome Study (Aussie Rett)  
[aussierett.ichr.uwa.edu.au](http://aussierett.ichr.uwa.edu.au)

International Rett Syndrome Database (InterRett)  
[interrett.ichr.uwa.edu.au](http://interrett.ichr.uwa.edu.au)

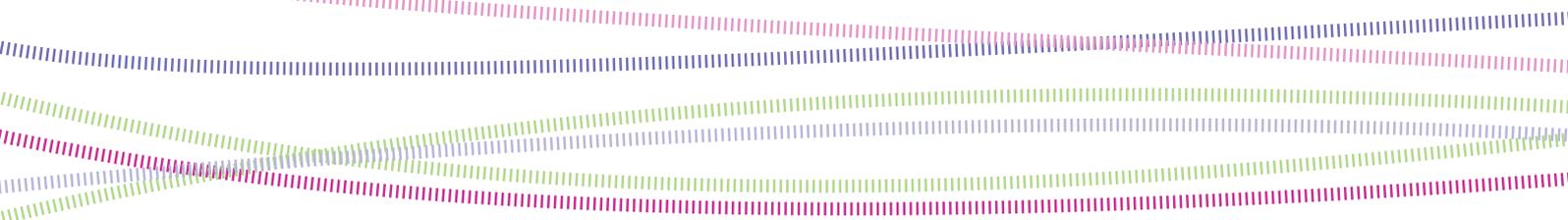
Division of Pediatric Orthopaedics of Morgan Stanley Children's Hospital of New York - Presbyterian  
[childrensorthopaedics.com](http://childrensorthopaedics.com)

### For Clinicians

Downs J , Bergman A, Carter P, Anderson A, Palmer GM, Roye DP, van Bosse H, Bebbington A, Larsson E-L, Smith BG, Baikie G, Fyfe S, Leonard H (2009). Guidelines for management of scoliosis in Rett syndrome patients based on expert consensus and clinical evidence, *SPINE*, 1;34(17):E607-17.

### Suggested Citation

Downs J, Roye DP Jr, Torode IP, Ellaway C, Baikie G, Larsson E-L, McKenzie A and Leonard H on behalf of the Australian Rett Syndrome Study, Scoliosis in Rett syndrome: A collaboration between parents, clinicians and researchers, Perth, Telethon Institute for Child Health Research, 2009.



## Glossary of Terms

**Activities of daily living:** Usual activities such as mobility, dressing, toileting and communication that are necessary for function in daily life.

**Cobb angle:** Used to assess the magnitude of spinal curvature; made by drawing a perpendicular to a line drawn across the superior endplate of the upper-end (most tilted) vertebra and the inferior endplate of the lower-end vertebra; the angle formed by the intersection of the two perpendicular lines is the Cobb angle. For more detail see *Does my daughter have scoliosis?* (page 6).

**Gastro-oesophageal reflux:** regurgitation of the stomach contents into the esophagus or trachea, e.g. heartburn.

**Hippotherapy:** Therapeutic use of activity in conjunction with the 3-dimensional movement of a horse to improve motor skills and functional activities.

**Hydrotherapy:** Therapeutic use of activity in an aquatic environment to improve motor skills and functional activities.

**Hypotonia:** A reduction of muscle tone.

**Occupational therapy:** use of self-care, work, and recreational activities to increase independent function, promote development, and to prevent or delay disability.

**Orthopaedic surgeon:** Physician concerned with the preservation and restoration, of the musculoskeletal system, extremities, and spine by medical and surgical methods.

**Orthotist:** Professional who makes splints and braces to improve the position of joints of the body.

**Osteoporosis:** Reduction in the quantity of bone characterized by decreased bone mass and abnormal skeletal microarchitecture, leading to increased susceptibility to fractures.

**Physiotherapy:** Promotion of health and rehabilitation with treatment by physical therapeutic measures as opposed to medical, surgical, or radiologic measures.

**Scoliosis:** Sideways curvature of the spine.

**Spinal brace:** An orthotic device that supports the spine in correct positioning in order to prevent and/or delay curve progression.

**Spinal fusion:** An operation to fix two or more vertebrae in alignment. The procedure can be performed on the posterior (back) portion of vertebrae or the anterior (front) portion of vertebrae.

**Vitamin D:** Vitamin that promotes the proper use of calcium and phosphorus, thereby producing skeletal growth, along with proper bone and tooth formation.

Definitions adapted from Stedman's Medical Dictionary - 28th Ed., 2006.

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**We would like to  
extend our appreciation  
to all the families who  
shared their stories  
and photos for this  
booklet.**





**Aussie Rett:**  
Australian Rett  
Syndrome Study

**InterRett:** International  
Rett Syndrome Database



RETT SYNDROME  
ASSOCIATION UK

**Australian Rett Syndrome  
Association Inc.**