Managing scoliosis in Rett syndrome

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What is meant by scoliosis?

Scoliosis is a sideways curvature of the spine. The spine can also twist and some rotation may occur. The earliest signs of scoliosis include a sidewards lean during sitting, standing and/or walking. At first, a scoliosis is flexible but with time it may become stiff and fixed. The degree and severity of spinal curvature is reported as the Cobb angle. *See figure.*

Why does scoliosis occur in Rett syndrome?

Scoliosis develops because of altered muscle strength and tone. An unbalanced spine may make it harder to maintain sitting and standing and can make it harder to walk. Scoliosis can increase susceptibility to respiratory infection or cause pain.

How common is scoliosis in Rett syndrome?

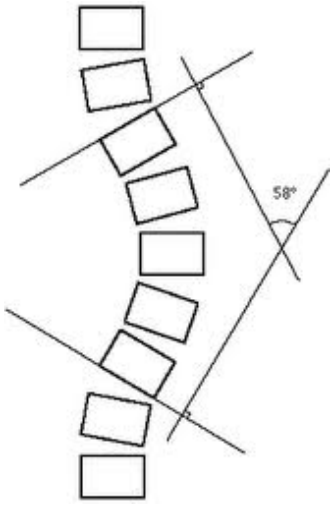
Although scoliosis does not affect every girl with Rett syndrome, it is the most common orthopaedic condition in Rett syndrome. Approximately one quarter of girls will develop scoliosis by the age of 6 and three quarters by 15 years, with an average age of onset of 11 years.

The type of mutation can influence the development of scoliosis. For example, individuals with p.Arg255* or large deletion mutations may develop scoliosis when younger and it may progress more rapidly. Girls who haven't learnt to walk are twice as likely to develop scoliosis. Those who can walk, either independently or with assistance, may develop a less severe scoliosis, if at all. An individual with no scoliosis or a small curve <25^0 and who is able walk independently at age 10 years is very unlikely to develop a severe scoliosis.

Are there established treatment/management options?

Management for scoliosis should begin prior to a diagnosis. General daily activities, in combination with physiotherapy, occupational therapy, hydrotherapy, and/or hippotherapy should aim to develop and maintain walking for as long as possible, to strengthen back muscles, and to promote correct posture while sitting and sleeping.

Because scoliosis may occur suddenly and progress rapidly, each visit to a doctor should include a physical examination of the spine, ideally every six months. More frequent monitoring may be required in children who have never learned to walk and have low muscle tone, are experiencing a growth spurt, have developed scoliosis at a young age or already have a very severe scoliosis. Physical examination includes assessment of growth (height/weight), spinal posture, muscle tone, and motor skills such as sitting, standing and walking.



Diagnosis is made by a doctor following a spinal assessment and x-ray. Your daughter would usually then be referred to an orthopaedic surgeon to assess and monitor for any progression. Follow up x-rays are taken every six or 12 months depending on how the scoliosis is progressing until skeletal maturity, then approximately every 12 months thereafter

until the Cobb angle stops changing. Based on this monitoring, the optimal treatment pathway can be determined: physical activity, spinal bracing or spinal surgery. The main aim in managing scoliosis is to prevent further curve progression and maintain maximum function.

What are the treatment options?

There are three main treatment pathways:

1. **Physical therapy and activity**
   This is important for improving and maintaining physical abilities, muscle strength and joint flexibility. Walking, whether assisted or independently, should be encouraged as much as possible, aiming to achieve two hours each day. If walking is not an option, daily use of a standing frame is a good alternative.

   Daily stretching, prescribed by a physiotherapist, can help to maintain range of movement of muscles and joints. A physiotherapist or occupational therapist can also advise on sitting postures to benefit the spine. Exercise and activity is important for all individuals with Rett syndrome.

2. **Spinal bracing**
   A spinal brace may be recommended to help with sitting balance and delay the need for surgery. Surgery carries greater operative risks for very severe curves and for younger patients. In this way, it is hoped that a brace might limit the progression of scoliosis and so delay the need for surgery until the child is older. However, to date no evidence that the brace can affect the progression of a scoliosis curve has been able to be demonstrated in the medical literature.

   Some girls can experience discomfort due to pressure sores or skin irritation and, if not fitting correctly, a brace can restrict breathing or exacerbate gastro-oesophageal reflux. If a brace is prescribed, the team of orthopaedic surgeons, physiotherapists, and orthotists should work together to ensure that it is both comfortable and helping to correct the scoliosis.

3. **Spinal surgery**
   The aim of spinal surgery is to correct the curve and prevent further development of scoliosis, through achieving a balanced and fused spine. Surgery is considered in girls, preferably over 10 years of age, with a Cobb angle over 40-50 degrees. Ideally, surgery should occur before the scoliosis becomes very severe. The decision of whether or not to proceed with surgery is done on a case-by-case basis and careful discussion between the family and surgeon is vitally important.

The girls must be as strong as possible prior to surgery to maximise recovery. This is helped by having a thorough preoperative assessment conducted in the weeks prior to surgery. From this any necessary adjustments can be made, for example providing extra nutritional supplementation.

Following surgery, monitoring and administration of pain medications often occurs in the intensive care unit (ICU) although some girls do not need ICU after their surgery. Assisted ventilation may be required immediately post-operation. When in hospital, family support is vital to ascertain their daughter's comfort.

Mobility must be encouraged as soon as possible to improve breathing, muscle strength and function as well as general comfort. A typical mobility program includes: log rolling to move around in bed, sitting on the edge of the bed (where possible) one day after surgery, bed-chair transfer (where possible) two days following surgery, and walking (where possible) three days

following surgery.

Hospitalisation and surgery can be very stressful for families who may need to plan to take time off work or organise care for other children during this time.

Depending on the individual, some changes may be necessary following surgery. Equipment may be required (eg slings to assist with transfers) and doses of some regular medications may need adjustment. In general, spinal surgery has been observed to improve general health, comfort, upright stability and sometimes mobility.

Following spinal surgery, follow-up appointments in general begin 6 weeks following surgery, then every 2-3 months for the first year. As per the individual, the surgeon may continue checking the spine annually.

References

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