

CDKL5 and Scoliosis

There is not a lot known about scoliosis in CDKL5. Therefore, these are general comments to reassure and make you aware of the possibility of scoliosis being a feature of CDKL5.

Scoliosis is a curvature of the spine. There are many known causes of this. Neuromuscular diseases and certain syndromes can cause or be associated with curvature of the spine. It is not usually painful. Scoliosis that appears after the age of 8 does not usually interfere with the development and function of heart, lung or other internal organs. Scoliosis in children under the age of 6 may sometimes restrict lung development. Scoliosis seems to be less common in children that have the ability to walk or spend some time standing.

In CDKL5, the frequency of scoliosis is not known and there are no scientific studies yet available on this specific topic. In Rett syndrome about 70% develop scoliosis and about half of those might have surgery for the scoliosis. CDKL5 families should therefore be aware of the possibility of scoliosis developing so that they can be involved in shared decision making with health care professionals.

Monitoring and intervention regarding diagnosis

Physical assessment of the spine should be conducted at the time of diagnosis and at clinical reviews at between 6 and 12 month intervals depending of rate of growth and size of any scoliosis. If the curve is mild it may not require health care professional review.

Therapy

Therapy should aim to:

- Develop, maintain and promote walking or standing for as long as practical
- Maintain flexibility of the spine

Implement a postural management scheme that includes appropriate support for correct sitting posture. This may require specialist wheelchair assessment and provision

Monitoring following a diagnosis of scoliosis

Referral should be made to a specialist in managing scoliosis. Physical examination of the spine should occur frequently throughout growth, but particularly if there is an early age of onset of curve, a large curve, low muscle tone, and a child that never managed to walk.

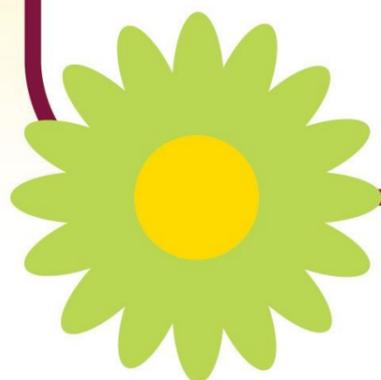
Imaging

The curve may be monitored by sitting or standing whole spine x-rays. The frequency of x-rays depends on the need for clinical monitoring. In children there is a desire to reduce radiation exposure.

Therapy and conservative management

Involvement of physiotherapists and occupational therapists should occur. There is no evidence that physiotherapy will prevent progression of an established scoliosis. The aim is to prolong ambulation and maintain musculoskeletal wellbeing. For those that cannot walk the use of standing frames should be encouraged. Symmetrical seating is of value for the child's comfort and function. Supported seating will optimise posture.

Spinal bracing has not been shown to be useful. If it is used at all it should only be in the skeletally immature child. Pressure sores, respiratory impairment, skin irritation, and exacerbation of gastro-oesophageal reflux are some of the potential problems with bracing.



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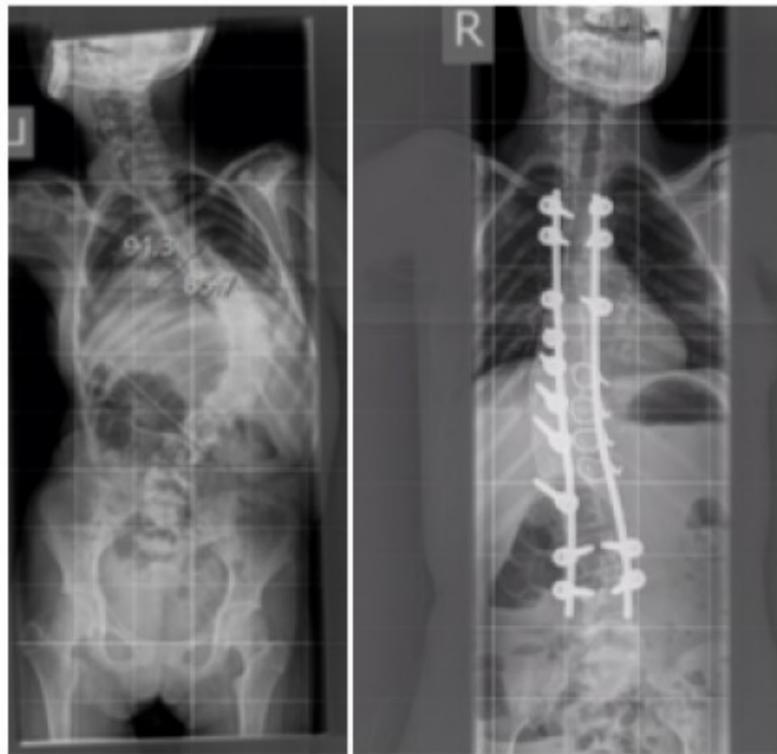


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Surgery

Surgery if considered should be of functional benefit to the child. There should be shared decision making between the patient and carers and the surgical team, with careful consideration of the risks and benefits.



Scoliosis surgery should only occur in specialist centres due to the high risk of anaesthetic and post-operative complications. Nutritional status should be optimal before surgery.

In the majority of cases a posterior only spinal fusion would be used. Metal screws and rods are used to correct the curve and provide stability until the bone fusion is solid. The goal of surgery should be good sitting balance with level hips, improved well-being and function of the child and improved carer well-being.

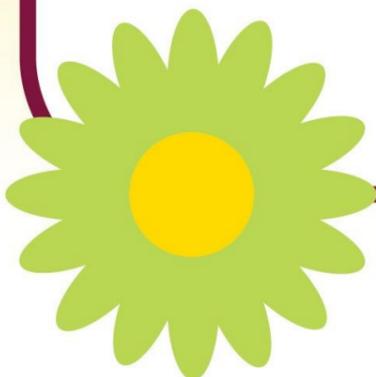
Links

Specific information on scoliosis in CDKL5 is not yet available. These sites provide useful general information regarding neuromuscular scoliosis and some of the syndromes that are associated with scoliosis.

Scoliosis Association U.K. <http://www.sauk.org.uk/>

Scoliosis Research Society <http://www.srs.org/>

This article has been kindly written for CDKL5 UK by Dr Philip Sell, Consultant Orthopaedic and Spinal surgeon, University Hospitals of Leicester and Queens Medical Centre Nottingham (UK).



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