



Are spine problems common in skeletal dysplasia conditions?

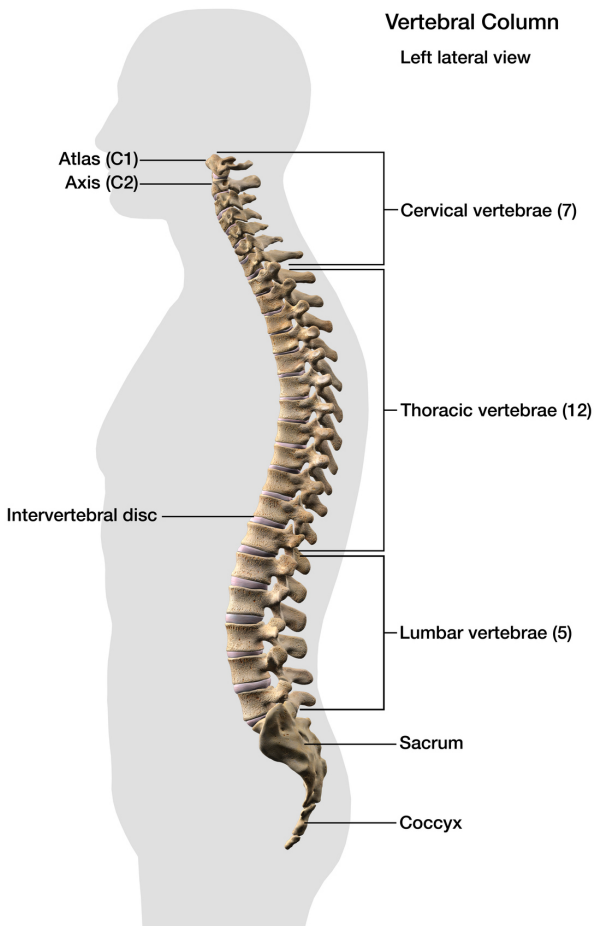
Yes, they are. Different problems and combinations of problems can occur, depending on the type of condition a person experiences (see Table 1 below for details). These problems occur because the spinal building blocks (vertebrae) might be a different shape or weak and easily compressed, or the hip joints can be stiff, changing the shape of the spine.

Does having a curve in the spine matter?

The job of the spine is to keep the body upright and to protect the spinal cord. Deformity in the spine can cause pain and compress the spinal cord. Regular monitoring of the shape of the spine and medical checks for any signs that things are getting worse is important if you have skeletal dysplasia.

Are some spine problems more common in certain conditions?

Sometimes, the appearance of the spine on X-ray can tell your doctor exactly what kind of skeletal dysplasia condition is there. For example, flattening of the vertebrae is more common in collagen-II



disorders, whereas sway back lordosis is a feature of achondroplasia. The shape of the spine may change as time goes by – we see a kink in the spine in infants with achondroplasia, but with the right spine care, this will go away by the time they can walk. *See Table 1 for more details.*

What is “cervical spine instability”?

The spine is divided up into different sections and the vertebrae from top to bottom are called cervical, thoracic, lumbar and sacral, and they are numbered.

At the very top, the cervical vertebrae 1 and 2 (C1, C2) connect and hold the skull in place on the spine – for this reason C1 is also called the Atlas.

When C1 and C2 are not properly linked because the C2 peg that should hook into the ring of C1 is underdeveloped or hypoplastic, the upper spine is not stable and can move, pressing dangerously on the spinal cord. This is “cervical spine instability.” Surgery to fix the upper spine is needed. *See Table 1 for more details.*



Are there activities that should be avoided for someone who may have cervical spine instability?

Until instability has been ruled out, anything that jolts the neck should be avoided, like trampolining, horse-riding and contact sports.

What is spinal stenosis?

This is a certain kind of narrowing in the spine, not because of deformity, but because the bone and ligaments around the spinal cord thicken and fill up the space, pressing against it, causing pain and neurological problems, like pins and needles, weakness, pain and sometimes incontinence. People with this might squat down to relieve the symptoms they experience. *See Table 1 for more details.*

Can spine problems get worse if they are not treated?

With the right care, some spine deformities improve as time goes by. For example, kyphosis in achondroplasia will improve if the spine is properly supported and kept straight early on. Other spine problems can worsen and start to press on the cord. At their most severe, problems can change the shape of the chest so much, the person can experience serious breathing issues. In a number of conditions, it is important to keep monitoring the spine and tracking the progress of any deformity as it worsens.

If it gets worse, what happens next?

Your doctor will do some more tests to see if there is any pressing on the cord. This involves asking questions about numbness or pain in the arms and legs, doing an MRI of the spine to look at the cord and sometimes some electrical tests of the nerves in the arms and legs. Repeat checks are needed to track the progress and when things become more severe, sometimes spine bracing can stop things from becoming worse. Your doctor may also suggest spine surgery to relieve the pressure on the cord. It is also important to keep healthy and not become overweight. Physical therapy is also recommended.

Is surgery dangerous?

No surgery is without risks and spine surgery is no exception. It is important that it is carried out in a specialist center by experts.



Table 1. Spinal deformities experienced by condition and recommendations of the SDMC team for treatment. You should be looked after by experienced specialists from multiple fields and seen at least once a year in childhood and whenever required in adulthood.

Spinal deformity	Description	Condition	Monitoring and treatment	Severe complications to prevent
Kyphosis	Forward kink or curve near the top of the spine	Achondroplasia, storage disorders, metatropic dysplasia; osteogenesis imperfecta	Physical examination, plain X-rays, whole spine MRI if worsening. Spinal bracing or casting, corrective surgery if severe	Spinal cord compression, respiratory difficulties
Scoliosis	Curvature of the spine to the left and right	Type II collagen disorders, conditions with flattened or wedged vertebrae, brittle bones	Physical examination and plain X-rays, whole spine MRI if worsening. Spinal bracing, surgery if severe	Pain, chest deformity, mobility issues, respiratory difficulties
Lordosis	Sway in the lower back	Achondroplasia	Monitor hip joints by clinical examination and plain X-rays	Lower back pain, reduced movement of hips
Cervical spine instability	Weakness in the connection between the top parts of the spine, called C1 and C2	All skeletal dysplasia conditions, especially collagen II disorders, metatropic dysplasia, Morquio syndrome	X-rays cervical spine in flexion-extension (child must do this themselves – not forced) +/- CT & MRI	Pithing of the top of the spinal cord
Spinal stenosis	Narrowing in the spine due to bone and ligaments around the spinal cord thickening and filling up the space, causing pain and neurological problems	Achondroplasia, acrodysostosis	Physical examination and medical assessment (limb pain, numbness, weakness)	Irreversible compression of the spinal cord and paralysis