

Spine and Spinal Cord Problems in Bone Dysplasias

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The spine (or spinal column) has four major functions. First, it is the primary **supporting structure** of our body. While most animals are principally supported by their four legs (with the spinal column holding the internal organs that hang down from it) humans, having assumed an upright pattern of walking, are far more dependent on the strength and straightness of the spine for support. Secondly, the bones which make up the spinal column, called the *vertebrae*, provide **protection for the spinal cord**; the spinal cord carries the nerve tracts which transmit signals from our brain to the rest of our body and are critical, for example, in the control of our muscles, in sensation (pain, touch) and so forth. The vertebrae enclose the spinal cord in a bony, protective case. That case is jointed, however, and that is important in terms of the third major function of the spinal column. That it is made up of lots of different vertebrae rather than a single, solid piece of bone, means that a person's trunk has **mobility** — can bend forward and back and side to side and can twist. Finally, the backbones grow and much of a person's **growth** is dependent on the increasing size of those vertebrae.

So, if a person has a disorder of bone growth, it may affect any or all of the functions of the spine. (Likewise, necessary medical treatment for one of these functional problems can result in problems in other areas of function of the

spinal column.)

A diagram of the human spinal column is shown in *Figure 1*. Of the various levels of the column, most often problems, arise in individuals with bone dysplasias (various kinds of dwarfing conditions) either in the *cervical* (neck) or the *lumbar* (waist) regions. I have chosen to use an example or two of possible problems which may develop in each of those regions related to the functions of the spine which we've already talked about.

Stenosis. Stenosis means narrowing. Because the spinal cord is completely

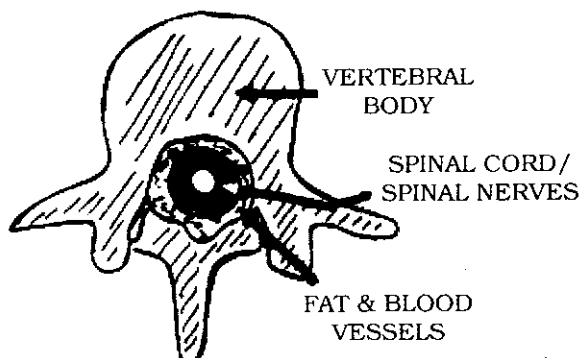


Figure 2

surrounded by bone (see *Figure 2*) it shouldn't be surprising that abnormalities of bone growth can result in pressure on the spinal cord or the nerves coming from it. That is, the cord is of normal size but its "container" — the vertebrae — grow more slowly. When we talk about spinal stenosis generally what is implied is that such bony narrowing is sufficiently severe that it is or will in the future cause medically important problems.

Example 1: Cervical cord stenosis in infants with achondroplasia. A few years ago, with the help of many colleagues, I recognized that a small number of infants with achondroplasia appeared to have risk of sudden and unexpected breathing problems that could be life-threatening. Ultimately it became evident that those risks are related to abnormal stenosis at the foramen magnum — the opening in the skull between the base of the brain and the spinal cord. Such stenosis apparently can sometimes damage the breathing control centers in the base of the brain. Babies can't tell us if they are having symptoms of cervical spine stenosis. Evaluations done here and elsewhere suggest that infants at special risk

special evaluations. For this reason I recommend that *every* baby with achondroplasia be seen in a clinic specializing in bone dysplasias and have an evaluation including careful neurologic examination, overnight sleep study (called a polysomnogram), neuroimaging (using CT scan or MRI scan to look at the region of the foramen magnum) and so forth. *Figure 3* shows an MRI scan of a baby with achondroplasia; the arrow points to the area which is of concern where, in some infants, compression of the brainstem and upper cervical cord may be present.

Certain precautions to protect the neck — neck support with movement, use of a rear facing car seat, avoidance of swing-o-matics, johnie-jump-ups and umbrella strollers — may decrease risk. If a baby is clearly developing indicators of cord compression then a neurosurgical procedure called suboccipital decompression can be done to remove a piece of bone in the region and decrease those risks. No one yet knows how many babies are at risk for this complication. Certainly it is a small number. Probably considerably less than 10% of all infants with achondroplasia have sufficient indications to undergo such decompression.

Example 2: Lumbosacral stenosis in adults with achondroplasia. All adults with achondroplasia have narrowing of the spinal canal in the lumbar and sacral region (low back). Many develop temporary symptoms, such as leg pain associated with exercise, which seems to be because of temporary pressure on the nerves from the spinal cord or blood vessels which become engorged with blood during physical activity. This "spinal claudication" goes away with rest. That kind of symptom does need to be evaluated but usually is not an emergency. In contrast, if bone is pressing on those same nerves then permanent damage could result. So, if an individual with this or other bone dysplasias is experiencing constant leg pains, 'shooting' pains down the legs, evidence for weakness or asymmetry of strength, persisting numbness or tingling or pins and needles sensations or problems with bowel or bladder control, then it's possible that lumbosacral

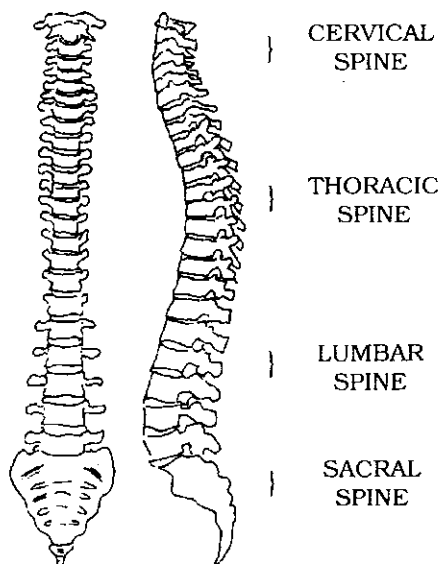


Figure 3



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spinal stenosis is present and should be evaluated quickly. If severe and uncontrolled pain or evidence of bony compression is present, then treatment is through removal of the compressing bone from the back part of the vertebrae, called lumbar laminectomy.

Curvature. Spine curvature in people with bone dysplasias may be due to *scoliosis* or *kyphosis* or a combination of the two. Scoliosis is side to side curvature while kyphosis refers to an abnormal backwards curve in the spine. (*Lordosis* is an abnormal forward curvature or 'sway-back' which usually isn't a major medical concern.) When mild, such curvatures may not have much importance. But if they progress, then mechanical, neurologic and heart and lung complications can be exceedingly serious. Early evaluation and monitoring is crucial in making sure that such curves are appropriately treated.

Example 1: Kyphoscoliosis in spondyloepiphyseal dysplasia (SED). Most forms of SED carry a markedly increased risk of spinal curvature, usually in the thoracic and lumbar region. Unlike the scoliosis in many individuals with average stature, the curves in people with SED more often progress, and progress faster. Without treatment kyphoscoliosis can eventually result in problems of balance, in risks for nerve related complica-

Children and adolescents with these disorders (and others) need to be watched for development of such curvature. And, very often, bracing and/or surgery will be needed to prevent the serious secondary effects.

Example 2: Cervical kyphosis in diastrophic dysplasia. Infants with diastrophic dysplasia seem to have a special and marked risk to develop backwards curvature of the vertebrae of the neck. This kyphosis is sometimes harmless, but if sufficiently severe it can cause nerve related pressure problems and nerve and muscle problems of sufficient severity to require surgery. So far there seems to be no clear cut way of preventing this kyphosis, nor has there been any way to predict which individuals have high risk beforehand.

Example 3: Thoraco-Lumbar kyphosis in achondroplasia. Most babies with achondroplasia develop a 'hump' in their lower back (sometimes referred to as a gibbus). For most of these that early kyphosis will go away by itself without any symptoms resulting. An x-ray of early thoraco lumbar kyphosis in a baby with achondroplasia is shown in *Figure 4*. In few percent of individuals with achondroplasia, however, that kyphosis will become stiff and angulated. And in those individuals, nerve damage can arise later on in life that, if not cared for, can result in paralysis. Rather than having to treat the kyphosis at that stage, what we would

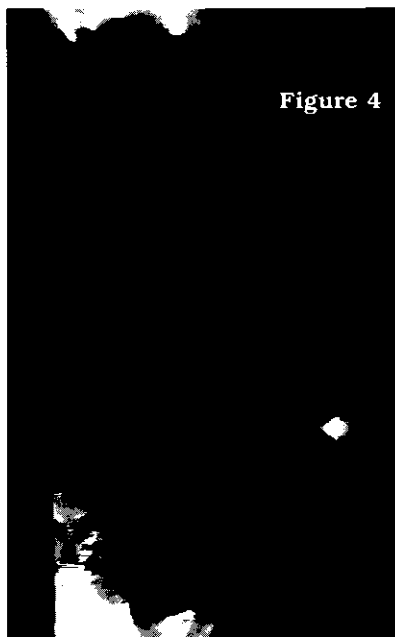


Figure 4

UW'S WAISMAN CLINIC HELPS FAMILIES DEAL WITH DWARFISM

University of Wisconsin-Madison
News Service

MADISON—Lucas Case looks almost like any other bright-eyed 16-month-old boy. But as bubbly and as cute as he is, there is something just slightly different about him. A closer look tells you that Lucas's head is slightly too big for the rest of his body, and that his arms and legs aren't quite as long as they should be.

Lucas, whose parents regularly bring him to the Genetics Bone Dysplasia Clinic at the University of Wisconsin-Madison's Waisman Center, has achondroplasia. It's the most common of the 150 to 200 specific disorders that constitute dwarfism: "bone growth failure," in the words of Dr. Richard Pauli, director of the clinic and associate professor in the departments of pediatrics and genetics at UW-Madison. Dwarfs, he said, will typically

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like to be able to do is prevent those severe kinds. Evaluations done here (and similar assessments carried out at other centers) suggest that some simple interventions may be effective in preventing those complications. It seems that if one can prevent gravity from exerting abnormal pressures on the bones of this region then much less frequently will the severe curves develop. How can those effects of gravity be minimized? It seems that prohibiting any unsupported sitting in the first year of life and always providing good back support markedly decreases the risk of serious and rigid curves developing. About 85% of all babies develop some kyphosis. With careful prohibition of such unsupported sitting all but about 15% of those will take care of themselves. In our clinic the remainder are braced for a time and in that group none have gone on to have worrisome curves.

Instability. The spine bones are bound to each other by connective tissues. Usually those connective tissues allow just enough movement for bending, stretching and twisting but are tight enough that slipping between the vertebrae doesn't occur. In quite a few bone dysplasias the connective tissues are also affected and, therefore, in quite some

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number of these there is a risk of abnormal slippage between the vertebrae. This is particularly so in the neck and can result in either acute or chronic nerve damage. In addition, sometimes the neck bones themselves are abnormally formed and more easily allow such slipping to occur with neck movement.

Example. Cervical spine instability in Morquio syndrome. Individuals with Morquio syndrome almost always have some abnormal movement between the bones of the neck. Similarly, people with metatrophic dysplasia, various SEDs and other bone dysplasias have such risks. A physician with special expertise in bone dysplasias knows which have this risk and can assess whether a particular person needs to be evaluated more thoroughly. All individuals with any of these disorders need to have x-rays of the cervical spine taken looking for this kind of instability. (In fact,



Dr. Richard Pauli

the DAAA requires such x-rays before anyone with these kinds of causes of their short stature can participate in sports.) If sufficiently severe, surgical fusion of the bones of the neck can be carried out.

As for any other part of the body, the specific kinds of spine problems for which a person may have risk is diagnosis specific. Only with a specific diagnosis can we use the experience with other individuals with exactly the same diagnosis to predict, evaluate and prevent. It's through the continued cooperation of LPs that such experience has and, hopefully, will continue to be accumulated in order to help others in the future.

Editor's Note: Rich Pauli is Professor of Pediatrics and Medical Genetics at the University of Wisconsin-Madison where, among other duties, he is Director of the Bone Dysplasia Clinic. He has been a member of LPA, and of the Medical Advisory

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grow to be about 4 feet tall.

The Madison center is one of only about 15 in the country that deal specifically with dwarfism, a condition that affects one of every 10,000 babies born in the United States.

Physicians can miss the disorder, said Pauli. The signs can be subtle, and X-rays are required to make a diagnosis with certainty.

Pauli said it's important for parents of achondroplastic children to get medical advice and counseling as soon as possible after a child is born with the disorder.

Achondroplastic infants run the risk of sudden death because a malformed skull can cut off breathing by constricting the brain stem, which controls respiration. Surgery can relieve the problem, Pauli said.

A specialized clinic such as the one at the Waisman Center, which gets about 150 visits annually, can sort out other risks to achondroplastic infants and help parents prevent them by providing "anticipatory care."

For example, children with achondroplasia are prone to middle ear disease in infancy, which can cause hearing problems and interfere with language development. Antibiotics or, if need be, surgery, can correct it.

Back curvature is another potential hazard, and one that can lead to paralysis. Simply teaching children to sit properly can mitigate that problem.

"We can't make the children 'not dwarfs', but we can help them become as healthy as possible," said Pauli.

Pauli, who is a medical adviser to Little People of America, a social and support organization for dwarfs, said children with the disease almost always develop normal intellects, but special attention must be paid to their physical development.

Lucas's parents, Dennis and Eraina Case, brought him to the clinic, which sees patients from six Midwest states, when he was two weeks old. Their pediatrician diagnosed Lucas's disorder at birth, but it was a nurse at the plant where Dennis was working who told the couple about the Waisman clinic. The family has made eight visits from their Rockford, Illinois home.

Eraina Case said Pauli and the clinic staff have been able to patiently and clearly answer questions about their son's disorder and to explain what they could expect from Lucas.

"I never had so much education so quickly," she said.

During a recent visit, Pauli told Lucas's parents that pressure on the child's eardrum should be checked, and would be discussed the next time they came. It is not currently a major concern.

"Lucas is hearing everything that he needs to," Pauli said. "And overall, he's doing beautifully."

As he did during previous visits, Pauli told the Cases that achondroplasia is caused by a genetic mutation in the egg or sperm, that no cause of the mutation has been found, and that having one child with achondroplasia doesn't necessarily mean they will have another.

"Your risks are just the same as anyone else's," Pauli told them. "The only person who has an increased risk is Lucas. His risk of having a child with the disease is 50 percent." (There is a 50% of his passing on sperm that have the dwarf gene; his chance of having a dwarf child depends on who the mother is.)

Pauli said providing such information to the families along with medical recommendations is a key function of the clinic. He wants parents to know as much as they possibly can about the disease that afflicts their children.

"The clinic's aim is to empower the family to become expert in the disorder, to act as expert-advocates for their children, not just as parents," he said. You can contact Dr. Richard Pauli at (608) 263-6874.