Patient Guide to Achondroplasia

Kyphosis

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What is achondroplasia?
Achondroplasia is a genetic condition. This means that a gene that growth in the body does not work properly. In this particular condition, a protein in the body called the "Fibroblast Growth Factor Receptor" begins to function abnormally. This results in abnormal cartilage where cartilage cells do not grow as much. Bones grow from cartilage and people with achondroplasia have shorter bones, abnormally shaped bones, and shorter stature.

What is the normal anatomy of the spine?
The spinal cord is a cord of nerve tissue that runs from the brain to the lower back. It sits within a protective bony canal called the spinal canal. The spines of the bone called vertebra form the spinal canal and protect the spine.

Each level of the spine has a different name and natural curvature. The neck spine is called the cervical spine. The upper back spine is called thoracic spine. The lower back is called the lumbar spine. The sacral spine is the area near the hips.
Different parts of the spine have different normal curvatures in the front-to-back plane. The neck and lower back have a normal curvature called lordosis. The upper back and hip spine have a normal curvature called kyphosis.

What is kyphosis?
Kyphosis is a normal curvature of the upper back or thoracic spine. Normal people have kyphosis. Looking at a person in profile will reveal a rounding or curvature in the upper back with the apex of the curve pointing backwards. Physicians can take X-rays and measure in degrees the amount of kyphosis a person possesses. Normal thoracic kyphosis is usually 20-40 degrees.

When does kyphosis become a problem?
Too much kyphosis is a problem. When the spine begins to bend forward too much, the spinal cord can become compressed. This impairs normal spine function such as bowel or bladder control, leg and feet movement. It also can worsen any spinal stenosis, narrowing of the spinal canal, present or other curvatures lower down on the spine.

Where in the spine does problematic kyphosis occur?
The extreme kyphosis tends to occur in the middle of the spine between the upper and lower backs. This is called the thoracolumbar junction. It involves the twelfth thoracic or first lumbar vertebra.

When does this problem usually occur in achondroplasia?
Kyphosis is usually present in infants with achondroplasia. It is thought to occur in infants with achondroplasia because they often have low muscle strength, large heads, flexible ligaments resulting in the inability to keep their spines straight. However, most infants improve when they are 2 or 3 years old. At that time they develop more muscle strength and tend to walk more. 10% of infants will unfortunately continue to keep the excessive kyphosis.

What are the signs and symptoms of kyphosis?
Kyphosis is usually noted as the child begins to sit. Symptoms of spinal cord compression can also be present. This can include numbness, tingling, or weakness in the lower legs. Loss of bowel or bladder control are other signs of spinal cord compression.

Should I see a doctor regularly?
Yes. It is important to have regular visits with a doctor to watch for and monitor kyphosis. Kyphosis endangers the spinal cord and normal spine function. The physician will ask questions, perform physical examinations to look for spinal cord
compression and order tests such as X-rays to measure the amount of kyphosis at each visit and look at the shape of the vertebra. The physician may also order other tests such as a CT or MRI to evaluate the spine.

What is the treatment of kyphosis?
The treatment for kyphosis depends on the severity of the curvature and the age at which it happens. Generally, treatment can mean observation, sitting modification, bracing, casting, or surgery. Usually people with achondroplasia require treatment at 3 phases of life.

In infancy, treatment is aimed to keep the kyphosis that is usually present from worsening. Typically, sitting modifications are used. In only 10% of the time does a child need to wear a brace all the time while awake. Sometimes a body cast must be used instead if a brace is not adequate.

In childhood, the goal is to correct those 10% of children that do not correct by themselves. This may require surgery. The surgery is usually prophylactic posterior fusion. This surgery helps the bones of the spine in the back grow or fuse together. The surgery will straighten the spine initially and keep it straight for the long-term. It is often performed so that patients do not develop other problems down the road.

In adulthood, the goal is to treat the kyphosis that is worsening spinal stenosis. Again, posterior fusion attempts to straighten the spine and allow the bones of the spine to grow together.

How will I do in the long run with kyphosis?
With careful treatment and proper follow-up, people with achondroplasia and treated kyphosis do very well. They can often return to active, productive lives with few restrictions and problems.

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