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Patient Guide to Achondroplasia Spinal Stenosis

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What is achondroplasia?

Achondroplasia is a genetic condition. This means that a gene that directs 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 so 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 bottom of the back. It sits within a protective bony canal called the spinal canal. The bones of the spine called vertebra form the spinal canal and protect the spine.

Depending on the level of the spine, the spine has different names. The neck spine is called the cervical spine. The upper back spine is called the thoracic spine. The lower back spine is called the lumbar spine. The spine near the hips is called the sacral spine.

The spinal cord has many different functions. It helps control breathing, bowel and bladder function and helps control movement of the hands and feet.

What is spinal stenosis?

Spinal stenosis is a condition where the spine bones or vertebra that surround the spinal cord do not provide enough room. Stenosis means narrowing of the spinal canal. The spinal cord can become compressed by the vertebra and result in loss of normal spine function.

Why is it so common with achondroplasia?

Spinal stenosis is the most common serious problem that people with achondroplasia may develop. One of the bones that grows abnormally in achondroplasia is the spine bone or vertebra. The vertebra form in such a way that the spinal canal has less room. This leads to compression of the spinal cord.

What part of the spinal cord is most commonly affected or compressed?

Most of the stenosis of narrowing of the spine occurs in the lumbar area of the spine. This part of the spine occurs in the lower back. The stenosis of achondroplasia usually increases in severity the further down the spine. Unstable neck bones are not usually a problem in achondroplasia.

What are the signs and symptoms of spinal stenosis?

The most common symptom of spinal stenosis is pain. This pain is often called neurogenic claudication and has some unique features. The pain occurs in the lower back, buttocks or thighs. It often occurs after a set amount of walking or activity and is often better when bending forward at the waist or resting. Numbness, tingling, or weakness in the lower legs may also be present. Loss of control of bowel or bladder function is another sign of spinal cord compression.

When does spinal stenosis typically occur?

Achondroplasia spinal stenosis usually occurs in a person's twenties. However, it can occur earlier and has been noted to occur as early as 11 years of age.

Should I see a doctor regularly?

Yes. It is important to have regular visits with a doctor to make sure that spinal stenosis does not develop. The physician will ask questions, perform physical examinations to look for spinal cord compression and if necessary order tests such as X-rays, CT, and MRI to evaluate the spine.

What is the treatment of spinal stenosis?

Spinal stenosis eventually requires surgery. Without surgery,

the spinal cord will continue to be compressed and result in loss of various spine functions. This could mean continued pain, loss of bowel and bladder control, loss of leg and foot control. The surgery is usually decompression of the spinal canal. This means that specific parts of the spine bones are removed carefully to relieve the pressure on the spinal cord.

How will I do in the long run with spinal stenosis?

With surgery and proper follow-up, people with treated spinal stenosis do very well. They usually report relief of pain, possibly renewed control of any weakness and continue to lead productive and active lives.

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