Spinal stenosis is the leading cause of disability among individuals with achondroplasia and other skeletal dysplasias. This condition involves the narrowing of the spinal canal, leading to compression of the spinal cord and nerve roots. Such compression can impair vital functions, including breathing, bowel and bladder control, and limb movement, often resulting in significant pain and disability.

In achondroplasia, spinal stenosis is prevalent due to the abnormal formation of vertebrae, which narrows the spinal canal and leaves less room for the spinal cord and nerve roots. Additional factors like excessive lumbar lordosis and kyphosis can exacerbate this compression. Both children and adults are affected, with infants at risk for foramen magnum stenosis and adults commonly experiencing thoracic and lumbar stenosis starting in their twenties, though it can occur as early as adolescence.

The most vulnerable areas of the spine in achondroplasia are the foramen magnum, where the brainstem and spinal cord pass through, and the thoracolumbar region. Compression at the foramen magnum can lead to serious neurological complications, including sleep apnea, disrupted breathing patterns, hydrocephalus, myelopathy, and even sudden infant death. Lumbar and thoracic stenosis often present with lower back pain, numbness, tingling, and weakness in the limbs. In severe cases, loss of bowel or bladder control may occur, necessitating immediate medical attention.

Preventative measures are crucial. Regular monitoring by physicians familiar with skeletal dysplasia is essential to detect early signs of stenosis. Diagnostic tools like physical evaluations, X-rays, CT scans, and MRIs are important, and sleep studies may be recommended for infants. Educating parents and caregivers on safe handling and positioning of infants can reduce the risk of cervical compression. For older children and adults, routine doctor visits and imaging studies help in early detection and management.

Treatment varies based on individual cases. For cervical spinal cord compression, some children may require cervicomedullary decompression surgery, which involves removing a portion of the skull base and possibly parts of the upper spine to relieve pressure. Thoracic or lumbar stenosis may necessitate surgical procedures like decompressive laminectomy or spinal fusion, especially in severe cases associated with spinal deformities such as kyphosis. Spinal fusion, involving rods and screws to stabilize and straighten the spine, is often the most effective treatment to prevent instability and severe kyphosis.

Recovery from spinal fusion surgery can take from 2 to 6 weeks for resuming simple activities, with complete recovery potentially taking up to a year. Physical therapy plays a vital role in rehabilitation, helping patients regain strength, improve mobility, and learn safe movement techniques. Activities like walking and pool therapy are beneficial once the surgical incision has healed. Patients should avoid heavy lifting and excessive bending or twisting during the healing process. Additional therapies, including massage and acupuncture, may aid in recovery. In cases where extensive fusion limits arm mobility, patients might consider arm lengthening procedures to assist with daily activities.

Ongoing dedication to rehabilitation and regular follow-up appointments with healthcare providers are essential for monitoring the spine and ensuring the success of the treatment. With appropriate care and intervention, individuals with achondroplasia can achieve significant improvements in quality of life, moving beyond pain and disability to lead fulfilling lives.