Spinal Stenosis with Kyphotic Deformity in Achondroplasia Patient: A Case Report

Abstract: Achondroplasia refers to the absence of cartilage development, and was first described in 1878. Achondroplasia is the most prevalent type of human short-limbed dwarfism caused by FGFR3 gene abnormalities, affecting one in every 30,000 live births worldwide. This case report describes a case of a 21 years old male patient with Achondroplasia with lumbar spinal stenosis of vertebral Th12-L1, L1-L2, L2-L3 and kyphotic deformity. Patient had a surgical treatment with decompression laminectomy and posterior stabilization fusion. There was no neurovascular complication following the surgery. Mean VAS score of the leg pain improved from 5 to 2. ODI scores also improved consecutively from 18 out of 45 (40%) to 8 out of 45 (17.7%). The sensory function at the Th12 and L3 dermatome region of the leg was improved, as well as the radiological evaluation.

Keywords: Achondroplasia, LSS, Kyphotic Deformity, Laminectomy.

INTRODUCTION

Achondroplasia refers to the absence of cartilage development, and was first described in 1878 (Agrawal, S.N. 2020; Liao, J. C. et al., 2006; & Akbar, A., & Tobing, S. D. A. 2018). Achondroplasia is the most prevalent type of human short-limbed dwarfism caused by FGFR3 gene abnormalities, affecting one in every 30,000 live births worldwide. Achondroplasia causes limb and spine development problems due to an endochondral ossification deficiency. Short stature, spinal stenosis, shorter pedicles, genu varum, foramen magnum stenosis, and thoracolumbar kyphosis are all symptoms of achondroplasia. Because of reduced interpedicular space caudally, thoracic kyphosis with lumbar lordosis, anterior vertebral body wedging, and shorter, thicker pedicles and laminae, the achondroplastic spinal canal is one-third to one-half the size of a normal spine. These characteristics also lead to foramen narrowing (Pauli, R.M. 2019).

MATERIAL AND METHOD

We reported a male, 21 years old, with lumbar canal stenosis due to achondroplasia and kyphotic deformity. We decided to perform decompression (laminectomy) on VTh12 – VL2 and posterior stabilization (PSF). We assessed the patient's overall condition, neurovascular complications, Visual Analogue Scale (VAS), Oswestry Disability Index (ODI), implant location, and spinal structure status after surgery.

Case Study

A 21 year old patient came to our hospital with chief complain of leg weakness for the last 2 years. Pain was felt albeit a little, and was a no disturbance for daily activities. His back was known that it was bending and the patient had already gone to physio therapy for 19 times in two different hospitals.
Patient has no history of trauma and there was no family history of the same complaint. In general physical examination, the body stature was only 115 cm and the body weight was 39 kg. Based on physical examination of the spine, there was kyphotic deformity of the lumbar spine (Figure 1). There was local tenderness in the lumbar region with Visual Analog Scale 3. No step off sign was found on the palpation. The flexion and extension range of motion of the lumbar spine was limited due to pain. The motoric strength of both lower limbs were good as well as the sensory function except for the L2 and L3 dermatome of the leg, there was hypoesthesia in those regions. We assessed this patient with Moderate Disability (40%) based on Owestry Disability Index (ODI Score 18 out of 45).

Based on the first anteroposterior and lateral view of the lumbosacral vertebrae plain radiograph, we discovered kyphotic deformity of the Th12–L3 spine with 86° angle and disc space narrowing at level Th12–L1 (Figure 2).

Based on the MRI of lumbosacral vertebrae there are reduced space of vertebrae Th12-L1, L1-L2, L2-L3, bulging discs at Th12-L1 to L2-L3 that compress the spinal canal and bilateral radix at the level L2-3, L3-4, and L4-5, and facet joint degeneration at the level L2-3 and L5-S1 (Figure 3).
Fig 3. Pre-operative MRI, T1WI appears hypointense to hyperintense at T2WI, and T2 STIR with increased contrast in the medulla, cortex, and periosteum.

The patient was diagnosed with leg paresthesia caused by lumbar spine stenosis of vertebrae Th12-L1, L1-L2, and L2-L3. We chose to treat this patient with decompression and posterior stabilizing fusion. The incision was performed by a posterior approach in the midline along the spinous process from Th12 to L3. We identified the spinous process and the facet joint structure, and then performed spinous process detachment by conducting osteotomy on both lateral sides. We continued to laminectomy at the level vertebrae Th12-L1, L1-L2, L2-L3 as well as flavectomy at the same levels. We continued until the duramater was exposed and it was identified as being in good condition.

After that, we performed posterior stabilization by putting cortical polyaxial lumbar pedicle screws size 5.5 mm – 40 mm and rods on bilateral sides of vertebrae Th12-L1, L1-L2, L2-L3. Then we put crosslink rod between the L1 and L2 level. The bleeding was controlled and we put drain before the closure (Figure 4).

Fig 4. Intra-operative Procedures

RESULTS
Patient showed with favourable outcome at 6 months follow up, there was no new lesion and patient was able to walk normally. There was no pain, and there was no other signs or symptoms of recurrence was reported. There was no neurovascular complication following the surgery. Mean VAS score of the leg pain improved from 5 to 2. Oswestry Disability Index (ODI scores) also improved consecutively from 18 out of 45 (40%) to 8 out of 45 (17.7%). The sensory function at the L2 and L3 dermatome region of the leg was improved, there was no longer hypoesthesia in those regions. We discovered that the implant stayed in a decent position and that the spinal structure was also in good condition by inspecting the post-op X-ray of the lumbosacral spine AP and lateral view. (Figure 4)
DISCUSSION

Despite its rarity, achondroplasia has been widely reported in the literature. It is the most prevalent kind of congenital bone dysplasia and is visible from birth. The improper development of enchondral bone causes craniospinal axis stenosis. The incidence of this disorder is around 1 in 25,000 children birth in a year (Agrawal, S.N. 2020; & Akbar, A., & Tobing, S. D. A. 2018).

The etiology of this entity is yet unknown, however it was genetically linked. All cases of achondroplasia are caused by autosomal dominant mutations. These mutations are completely penetrant and exhibit relatively little expression variability. Because of its dominant inheritance pattern, an individual with achondroplasia (and whose partner is of average height) has a 50% chance that each of their kids would be similarly afflicted. However, the majority of cases of achondroplasia — approximately 80 percent — are caused by new, spontaneous mutations. As a result, around 80% of afflicted infants are born to two unaffected, average-sized parents (Ravichander, B., & Kundu, S. 2017; & Hoover-Fong, J. E. et al., 2021).

Because to decreased interpedicular space caudally, thoracic kyphosis with lumbar lordosis, anterior vertebral body wedging, and shorter, thicker pedicles and laminae, the achondroplastic spinal canal is one-third to one-half the size of a normal spine. These characteristics also lead to foramen narrowing. In addition to these baseline anomalies, as these individuals age, they are susceptible to the stenotic processes that afflict all populations: disc herniation, degenerative spondylosis, and arthrosis (Carlisle, E. S. et al., 2011).

Frontal bossing, rhizomelic shortening of the limbs, pelvic tilt, and fixed flexion deformity of the hip joints are all skeletal anomalies of this disease. As the canal and foramen stenosis worsens, it can cause spinal cord and root compression, resulting in sensory dysfunction, radicular pain, neurogenic claudication, bladder malfunction, and in extreme cases, fecal incontinence (Aziz, A. et al., 2019).

According to one research, 19 of 193 people with achondroplasia (10% ) were diagnosed with spinal stenosis before the age of 40, while another 27 of 193 (14%) were identified afterwards (Coi, A. et al., 2019). In one research, 139 of 193 individuals (72 percent) had neurologic symptoms by the age of 50 (although were not always diagnosed with spinal stenosis). Only a small percentage of people with achondroplasia never have any spinal problems (Adamova, B. M. et al., 2013).

Several techniques and early assessments have been proposed and used. It is worth noting that none of these have been shown to be useful in the future, but are largely based on subjective reasons. Such prospective studies of which evaluation method is most beneficial are critically required yet extremely difficult to produce. Many facilities have decided to skip CT and go straight to MRI. Better visibility of neural tissue is on the way, but because to the length of the operation, sedation or general anesthesia is typically required. Multi-position MRI is frequently chosen. Flow investigations may also be useful in evaluating if surgical intervention is necessary (Panda, A. et al., 2014).

To enhance height, surgical limb lengthening and growth hormone therapy have been performed; however, both of these therapeutic methods are controversial. In such instances, growth hormone treatment has been utilized to promote growth. Surgical managements such as limb lengthening operations, lumbar laminectomy, and spinal fusion have all been
shown to be helpful. Neurologic improvement has usually been observed following craniomedullary compression operations. The prognosis of achondroplasia is determined on the severity of the disease. Approximately 5% of babies with an exceptionally severe type of achondroplasia die during the first year of life, despite the fact that the majority of people with the illness live a normal life (Vleggeert-Lankamp, C., & Peul, W. 2012; & Adamova, B. M. et al., 2013).

In achondroplasia, the indication, timing, and method for surgical treatment of spinal canal stenosis with thoracolumbar kyphosis are not well established. For spinal stenosis above three disc levels with kyphosis more than 40°, anterior decompression with anterior interbody fusion and posterior decompression with posterolateral fusion are recommended. Although posterior instrumentation can help to support the spine and improve posterolateral fusion after laminectomy, the hook system is not recommended due to the small canal in achondroplasia. It has been documented that Harrington compression instrumentation caused intraoperative neurological impairment in an achondroplasia patient. This surgical complication can be prevented by using pedicle screw instrumentation in achondroplasia (Akbar, A., & Tobing, S. D. A. 2018; & Vleggeert-Lankamp, C., & Peul, W. 2012).

However, this case report needs some improvement. The measured outcomes are only coming from clinical evaluation, scoring measurement and radiological findings. Furthermore, with more samples of patients, longer time of follow-up, functional aspects and daily activities considerations of the patients, we could provide more evidence and recommendations about the treatment and the results for both short-term and long-term outcome of the treatment that has been done.

**CONCLUSION**

A 21 years old male with Achondroplasia with lumbar spinal stenosis of vertebrae Th12-L1, L1-L2, L2-L3 and kyphotic deformity had a surgical treatment of decompression (laminectomy) and posterior stabilization surgery. Six months following surgery, patient showed with good outcome, he was able to walk without gait disturbance and there was no new lesion reported. Decompression followed by posterior stabilization in a one-step surgery is an effective surgical modality to treat lumbar canal stenosis in achondroplastic patients. However, pedicle screw instrumentation in patients with achondroplasia and other patients with short stature can be challenging. Safe insertion requires technical experience and knowledge of the achondroplastic pedicle morphometry, which differs markedly from that of normal spine. Post-operative outcome can be achieved by evaluating the neurovascular complication, VAS score, ODI score, and the general condition of the patient.

**REFERENCES**