Clinical Research

Surgical treatment for lumbar canal stenosis in adult achondroplasia

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ABSTRACT

Introduction: Achondroplasia is the most common form of human short-limbed dwarfism caused by mutations in the FGFR3 gene, with an incidence of 1 in 30,000 live births annually in global.Achondroplasia is characterized by foramen magnum stenosis, thoracolumbar kyphosis, spinal stenosis, shortened pedicles, genu varum, and short stature.

Methods:We report a female, 51 years old, with lumbar canal stenosis due to degenerative disc disease and achondroplasia. We decided to perform decompression (laminectomy) and posterior stabilization. Post-operatively, we observed the general condition of the patient, neurovascular complication, Visual Analogue Scale (VAS), Oswestry Disability Index (ODI), implant position and spinal structure condition.

Results: 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 L5 and S1 dermatome region of the right leg was improved, as well as the radiological evaluation.

Conclusion: Decompression followed by posterior stabilization in a one-stepsurgery is an effective surgical modality to treat lumbar canal stenosis in achondroplastic patients. However, pedicle screw instrumentation in patients with achondroplasia and short stature can be challenging. Safe insertion requires technical experience and knowledge of the achondroplastic pedicle morphometry, which differs markedly from that of the normal spine.

ABSTRAK

***Pendahuluan** : Akondroplasia merupakan bentuk yang paling sering ditemui dari dwarfisme yang disebabkan oleh mutasigen FGFR3, dengan kejadiansatu dari 30.000 kelahiran hidup di seluruh dunia. Akondroplasia ditandai dengan stenosis foramen magnum, kifosis torakolumbal, stenosis spinal, memendeknya pedikel, genu varum, dan perawakan pendek.*

***Metode**: Kami melaporkan seorang perempuan berusia 51 tahun dengan stenosis kanalis lumbal akibat penyakit sendi degeneratif dan akondroplasia. Kami memutuskan untuk melakukan dekompresi (laminektomi) dan stabilisasi posterior. Setelah tindakan operasi, kami melakukan pengamatan kondisi umum pasien, komplikasi neurovaskular, Visual Analogue Scale (VAS), Oswestry Disability Index (ODI), posisi implan dan stuktur tulang belakang.*

***Hasil:** Tidak ditemukan adanya komplikasi neurovaskular pasca bedah. Keluhan leg pain berkurang dengan skor VAS dari 5 menjadi 2. Skor ODI juga menurun, sebelumnya 18 dari 45 (40%) menjadi 8 dari 45 (17.7%). Fungsi sensorik pada dermatom level L5 dan S1 pada tungkai mengalami perbaikan begitu juga dengan evaluasi radiologis.*

***Kesimpulan:** Prosedur dekompresi dan stabilisasi posterior dalam satu tahap dapat menjadi modalitas bedah yang efektif untuk mengatasi lumbar canal stenosis pada pasien akondroplaksia. Meskipun demikian, instrumentasi menggunakan pedicle screw pada pasien dengan akondroplaksia dan berperawakan pendek merupakan suatu tantangan tersendiri. Prosedur insersi pedicle screw pada pasien akondroplaksia membutuhkan pengalaman teknis dan pengetahuan yang baik terhadap bentuk anatomis dari pedikel tulang belakang yang jelas berbeda dengan bentuk tulang belakang pada pasien normal.*

Keywords: achondroplasia, lumbar canal stenosis, degenerative disc disease, decompression, laminectomy, posterior stabilization

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INTRODUCTION

Achondroplasia is the most common form of human short-limbed dwarfism and is one of a spectrum of diseases caused by mutations in the FGFR3 gene.[1,2] Globally, achondroplasia is the most common skeletal dysplasia, with an incidence of 1 in 30,000 live births annually.[2] Achondroplasia is characterized by foramen magnum stenosis, thoracolumbar kyphosis, spinal stenosis, genu varum, and short stature. Disordered endochondral bone formation producescharacteristic limb and spinal deformities. These spinal abnormalities, including thoracolumbar kyphosis, spinal stenosis, shortened pedicles, and underdeveloped sacrum, have been recognized in achondroplasia.[2,3]The achondroplastic spinal canal is one-third to one-half the size of a normal spine secondary to decreasing interpedicular distance caudally, thoracic kyphosis with lumbar lordosis, anterior vertebral body wedging, and shortened, thickened pedicles and laminae. These features also contribute to narrowed foramens.[3,4]

There are several characteristics of the achondroplastic spine that make the spinal canal narrow throughout and increase the chance ofcompression in the spinalcord or the cauda equina tooccur. The pedicles are short in these patients, particularly in the thoracolumbar region, and there is a reduction of interpedicular spacing of the lumbar vertebrae, which in contrast to regular spinal anatomy dimensions is progressively smaller in the caudal direction. In addition to these reduced interpedicular distances, pedicular thickening has been described in these patients, which adds to the narrowing of the spinal canal. [5]

Stenosis of the achondroplastic vertebral canal is progressive and is the substrate of neurological deficits.[4] Worsening stenosis of the canal and foramens eventually can lead to spinal cord and root compression, resulting in sensory dysfunction, intermittent claudication, paraparesis, paraplegia and quadriplegia, bladder dysfunction, and in severe cases, fecal incontinence, which depends on the level of stenosis.[4,6] Symptomatic lumbar stenosis can be observed in 20% to 30% of the achondroplastic adult patients, and surgical treatment is required in nearly 10% of the patients. Multilevel spinal laminectomy is widely accepted as the treatment of choice. More extensive decompression to sacrum has also been recommended.[6]But recently, Thomeer *et al,* showed that L2 and/or L3 levels are involved in 97% of a series of 36 cases underwent interapophyseolaminar decompression.[7] Xin Qi *et al* suggested that laminectomy at these levels could provide enough decompression for most patients.[7,8]

Several measurement tools and questionnaires were provided to evaluate the spinal disorders pre- and post-operatively. The Oswestry Disability Index (ODI) has become one of the principal condition-specific outcome measures used in the management of spinal disorders.[9,10]

CASE ILLUSTRATION

A 51 year old patient came to our hospital with chief complain of leg pain in the last2 years that was getting worsefor the past 2 months. This condition started 2 years ago, the pain sharp in quality, and especially felt when the patient was standing, sitting, or walking for long time.She also sometimes felt moderate tingling and numbness in the regio of calf when she felt too tired. She hadno trouble in walking except after walking for a long distance (more than 1 mile) that she had to stop due to pain on her leg. She felt relieved when she was inresting or in sitting position. There was no history of trauma.

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 hyperlordotic deformity of the lumbar spine (Figure 1). There was local tenderness inthe 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 L5 and S1 dermatome of the right leg, there was hypoesthesiainthose regions. We assessed this patient with Moderate Disability (40%) based on Owestry Disability Index (ODI Score 18 out of 45).

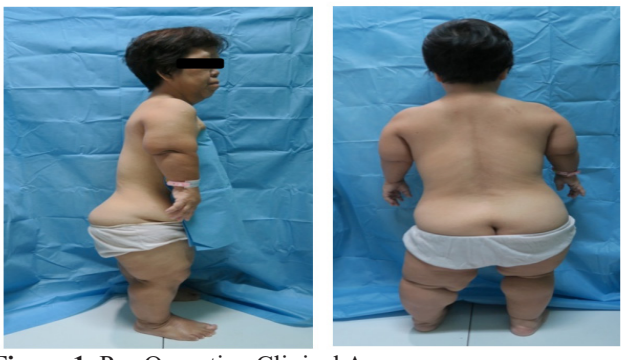

Figure 1. Pre-Operative Clinical Appearance

In radiological analysis, based on the initial anteroposterior and lateral view of the lumbosacral vertebrae plain radiograph, we found lordotic of L5 to S1 spine, ostheophite, sclerotic endplate of L5 and S1 of the vertebral body, and disc space narrowing at level L4-L5 and L5-S1 (Figure 2). Based on the MRI of lumbosacral vertebrae there are degenerative disc disease from level L2-3 to L4-5, bulging discs at L1-2 to L5-S1 that compress the spinal canal and bilateral radix at the level L2-3, L3-4, and L4-5, and facet joint degeneration at the levelL4-5 and L5-S1 (Figure 3).

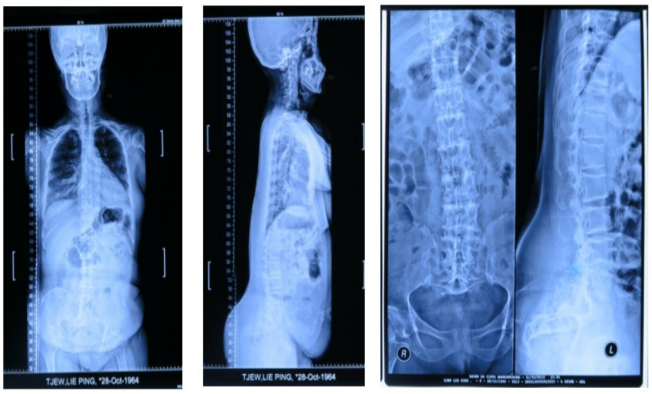

Figure 2. Pre-operative Radiology Examination

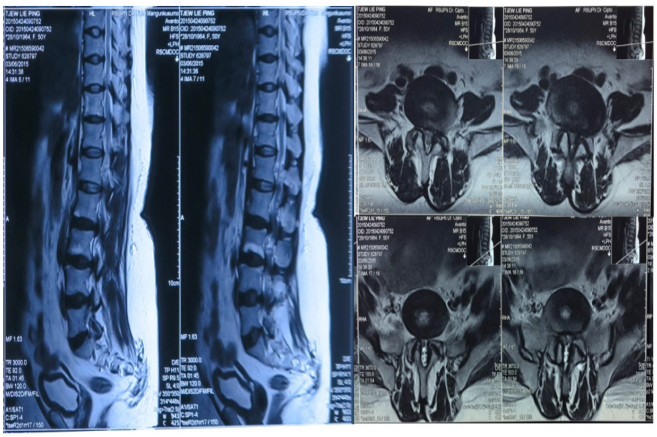

Figure 3. Pre-Operative MRI Examination

The patient was diagnosed with leg pain due to lumbar canal stenosis of L2-3, L3-4, and L4-5 due to degenerative disc disease. We decided to perform decompression and posterior stabilization to this patient. The incision was made by posterior approach in the midline along the spinous process at the level L2 to S1. We identified the spinous process and the facet joint structure, and then performed spinous process detachment by conductingosteotomy on both lateral sides. We continuedto laminectomy at the level L2-L3, L3-L4, and L4-L5 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 perfomed posterior stabilization by putting cortical polyaxial lumbar pedicle screws size 5.5 – 35 mm and rods on bilateral sides of L2, L3, L4, L5. Then we put crosslink rod between the L3 and L4 level. The screw placement was confirmed by using the image intensifier. The bleeding was controlled and we put drain before the closure (Figure 4).

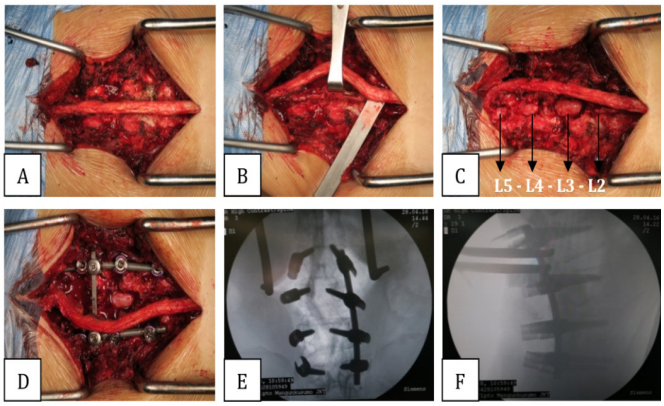

Figure 4. Intra-Operative Procedures. A: Exposed; B: Detachment of Spinous Process; C: Decompression (Laminectomy and Flavectomy); D: Posterior Stabilization; E & F: Image intensifier confirmation

RESULTS

There was no neurovascular complication following the surgery. Mean VAS score of the leg pain improved from 5 to 2. Owestry 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 L5 and S1 dermatome region of the right leg was improved, there was no longer hypoesthesiainthose regions. By checking the post-op X-ray of the lumbosacral spine AP and lateral view, we found that the implant remained in a good position and the spinal structure was also in a good condition (Figure 5).

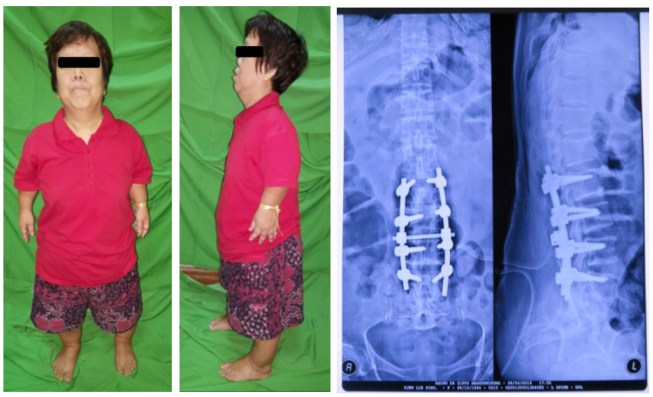

Figure 5. Post-Operative Clinical Appearance and X-Ray

DISCUSSION

Despite its rarity, achondroplasia is well documented in the literature. Recognisable at birth, it is the most common form of congenital bony dysplasia.The abnormal formation of enchondral bone leads to stenosis of the craniospinal axis.[11]

The patient presented in this case report had a chief complaint of progressive leg pain that became worse with activitiessuch as standing, sitting for a long time or walking for a long distance (more than 1 mile) and relieved after taking a rest. The patient also felt moderate tingling and numbness in the regio of calf. There was hypoesthesiainL5 and S1 dermatome of the right leg. The signs of spinal compression are clearly identified inthis patient by theclinical manifestations and radiology examination. Based on the radiographic findings there werehyperlordotic lumbar, degenerative disc disease of the lumbar spine showed as disc space narrowing, bulging discs that compressed the lumbar spinal canal and bilateral radix of the lumbar spine, and there wasalso facet joint degeneration.

In achondroplastic patient, the spinal X-rays demonstrate thenarrowing of the interpedicular distances in the lumbar spine. A small spinal canal is present from birth but the signs and symptoms of spinal cord compression are noted in middle age or later.[11,12] There are several pathognomonic findings in theVertebral MRI examination in achondroplastic patients. Based on S.-T. Jeong *et al,* studies, the normal spinal canal,as measured by its interpedicular distance, widens from L1 to L5, but in patients with achondroplasia this distance progressively decreases. Most of the achondroplastic patients havestenosis at the level of the intervertebral disc, suggesting that the stenosis isdegenerative. There are also protrusion of the intervertebral disc and hyperthrophy of the ligamentum flavum. Most of the patients also havea combination of stenosis of the central canal and the lateral recess, none hassignificant compression of the dural sac at the level of vertebral body. The most common level affected areL1 and L2. Achondroplasia patients with narrow bony canal at the higher spinal levels (L1 to L3) are more likely to develop symptoms of spinal stenosis. Degenerative disc changes which occur in the intrinsically narrow canal are probably the reason why narrowing occurs more at the higher lumbar levels.[12] Modi HN *et al,* in their study, postulated that symptomatic lumbar stenosis in achondroplasia is primarily a central stenosis secondary to degenerative spondylosis rather than nerve root compression attributable to foraminal stenosis because (1) the areas of the foraminal canal and nerve root in the foramina of patients with achondroplasia are smaller than those without achondroplasia and (2) nerve root occupancy in the foraminal canal of patients with achondroplasia is not more than that of those without achondroplasia.[13]

We ruled out the diagnosis of the patient in this case report as leg pain due to lumbar canal stenosis caused by degenerative disc disease. We performed decompression and posterior stabilization to overcome the posterior instability of the lumbar spine after decompression, as definitive surgical treatment ofthis patient.

Post-operatively, we found no neurovascular complication and there wereconsecutive improvements of VAS pain score and ODI score as well as the sensory function. Overall, surgical procedure performed tothis patient resulted in a good anatomical and clinical outcome.

In general population, short-term (≤5 years) results of the surgery tend to be promising, with many studies reporting statistically significant and often dramatic improvements in symptoms and functions.Long-term benefit is more likely with earlier surgery, patients with achondroplasia who develop symptoms of stenosis should be encouraged to seek medical advice sooner rather than later.[6,14]

However, this case report isfar from perfect. The measured outcomes areonly coming fromclinical 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 theresults for both short-term and long-term outcome

CONCLUSION

Decompression followed by posterior stabilization in a one-stepsurgery isan 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.

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