

CASE REPORT

Achondroplasia and lumbar spinal stenosis: A Case report and review of literature

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ABSTRACT

Achondroplasia is the most common non-lethal skeletal dysplasia. Its incidence is between 1 in 10,000 to 1 in 30,000; with half of the patients showing various neurological manifestations. We are presenting an interesting case of a 26 year old lady with short stature, who presented with acute pain in the front of the thigh for three months. Plain x-rays showed platyspondylosis with developmental anomalies of the posterior neural arch/dysplasia. MRI dorsolumbar spine showed congenital stenosis in this area. The stenosis was more pronounced at L2-3 .She was treated with L2-3 laminectomy and discectomy. On follow-up, she was symptom free with no residual neurological damage.

Keywords: Achondroplasia; Backache; Lumbar spinal stenosis

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INTRODUCTION

Achondroplasia is the most common non-lethal skeletal dysplasia.¹ It is the most common type of rhizomelic dwarfism and is caused by a mutation of fibroblast growth factor receptor-3.^{2, 3} The primary defect found in patients with achondroplasia is abnormal endochondral ossification.⁴ Half of the patients show various neurological complications. The most serious complication of achondroplasia is respiratory impairment, apnea and sudden infant death, resulting from compression of the medulla oblongata.⁵ Spine surgery, requires evaluation of both spinal stenosis and instability. These patients are best evaluated by a multidisciplinary team.⁶ Fusion procedures are recommended in patients with a large decompression, overlying a thoracolumbar kyphosis to avoid progressive postoperative deformity.⁴ The tendency toward paralysis and paraplegia is much higher in this set of patients. Special attention must be given to technical details when operating on achondroplastic dwarfs. Early and most extensive surgery is recommended.⁷

CASE REPORT

A 26 year old lady who was bed ridden, presented with acute pain in front of the thigh bilaterally for three months. The pain in front of the thigh was so severe that the patient was unable to sleep at night. There was no history of backache or trauma.

Patient was initially seen by the medical department and Doppler USG of the lower limb was advised to rule out a vascular pathology. On examination power was reduced at the hip flexors to 4/5. Rest of the examination was unremarkable. Clinical vascular examination of the lower limb was also normal. No past medical and surgical history of significance was present.

All routine investigations were within normal limits. A plain x-ray showed platyspondylosis with developmental anomalies of the posterior neural arch/dysplasia. MRI of the dorsolumbar spine showed congenital stenosis from L1 to L5, more pronounced at L2-3 (Fig: I, II and III). L2-3 laminectomy and discectomy was done. The patient became symptom free just after surgery and returned to his routine work.



Fig I: MRI dorsolumbar spine T2 weighted image sagittal view

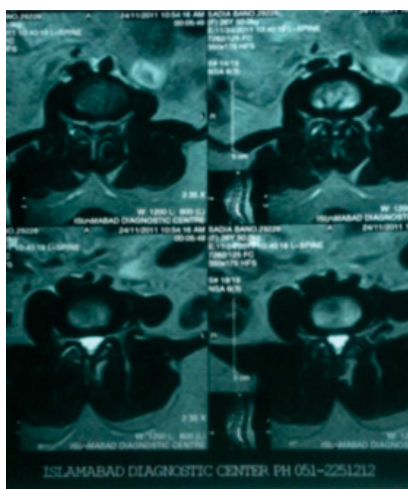


Fig II: MRI Dorsolumbar spine T2 weighted image axial view

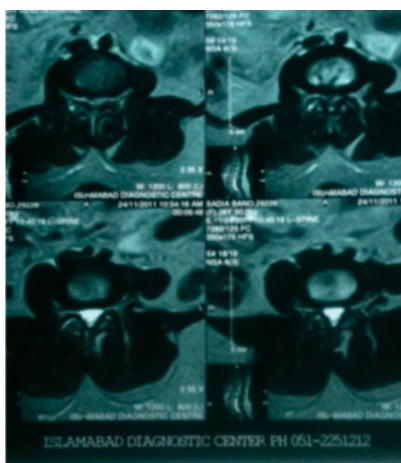


Fig III: MRI dorsolumbar spine myelogram

DISCUSSION

The skeletal dysplasias are a heterogeneous group of disorders characterized by intrinsic abnormalities in the growth and/or remodeling of cartilage and bone. These dysplasias affect the skull, spine, and extremities in varying degrees.⁸ The abnormal spinal development of the achondroplastic dwarf can result in neurologic damage due primarily to the following two syndromes: lumbar spinal canal stenosis and thoracolumbar kyphosis.³

In the general category of congenital spinal stenosis, there is a group of patients with particular features; patients suffering from a hereditary or systemic disease with congenital narrowing of the spinal canal. Evaluation of their neurological symptoms is influenced by the peculiarities of the underlying disease.⁷ They frequently cause a disproportionately short stature, the standing height falls below the third percentile for age.

Fibroblast growth factors are structurally related proteins associated with cell growth, migration, wound healing, and angiogenesis. At the cellular level, their function is mediated by transmembrane tyrosine kinase receptors, known as fibroblast growth factor receptors (FGFR). Mutation in *FGFR3* is responsible for achondroplasia, hypochondroplasia, and thanatophoric dysplasia. The primary function of *FGFR3* is to limit osteogenesis. Mutation causes enhancement in its function of limiting endochondral ossification. Mutation in *FGFR3* in achondroplasia is due to transition of guanine to adenine (G to A) at nucleotide 1138 of complementary DNA.⁹

Wide, multilevel laminectomies extending to the pedicles and lateral recesses with foraminotomies may be necessary. Extradural removal of herniated disc material is performed as necessary.¹⁰ The length of decompression usually extends from the lower thoracic spine to the sacrum to prevent recurrence. Maintaining the integrity of facet joints is necessary to prevent postlaminectomy instability. If instability does occur, anterior fusion may be necessary.

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Professor Koji Sumikawa joins APICARE

We are pleased to announce that Professor Koji Sumikawa has joined the Editorial Board of APICARE. Koji Sumikawa is Professor of Anesthesiology at Nagasaki University School of Medicine, Nagasaki (Japan).

He was born on August 3, 1947. He qualified his MD in 1972, and later earned Ph.D. in 1978 from Osaka University Medical School. He is Japanese Board certified in Anesthesiology, Pain Clinic and Critical Care Medicine. He is Vice President, Board of Directors of Japanese Society of Anesthesiology (JSA), Chairman of Committee on Ethics in JSA and member, Board of Directors of Japanese Society of Cardiovascular Anesthesia. He has also served as President of Japanese Society of Cardiovascular Anesthesia, Japanese Society of Circulation Control in Medicine, Japanese Society of Neuroanesthesia and Japanese Society of Reanimatology. He is a member of ASA, ASRA, IARS and IASP.