

Achondroplasia. MR imaging findings in the lumbar spine

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Section: Musculoskeletal system

Area of Interest: Musculoskeletal spine

Procedure: Diagnostic procedure

Imaging Technique: MR

Special Focus: Congenital Case Type: Clinical Cases

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Patient: 27 years, male

Clinical History:

Man undergoing lumbar spine MRI due to symptoms suggestive of spinal canal stenosis. The patient has short stature.

Imaging Findings:

On the sagittal images there was posterior scalloping of the vertebral bodies. The spinal canal was compromised at the level of the intervertebral disks, with compression of the neural elements (figure 1). On the axial images the pedicles were shortened, there was also evidence of both central and lateral canal stenosis (figure 2).

Discussion:

Achondroplasia is an autosomal dominant dwarfing condition [1, 2] with an incidence between 1/13, 500[3] to 1/25, 000 live births [4].

Achondroplasia is due to a nucleotide substitution in FGFR3 gene [5] and over 75% of them are newly paternally derived [6].

It is characterized by rhizomelic limb shortening, macrocephaly, midface hypoplasia with frontal bossing, bowed legs [2], short broad hands with trident finger configuration [1].

Children present with recurrent otitis media [2], delay in mental and motor development, hypotonia, feeding, sleep disorders, and compressive spinal syndromes [7, 8]. More severe early complications include hydrocephalus, sleep apnoea and sudden death related to foramen magnum stenosis [9].

There is considerable debility and reduction in the quality of life[3], increased overall mortality from birth to 34 years[10], mainly accidental, neurological, and heart disease related[4]. The average life expectancy is decreased by 10 years in achondroplastic patients [4].

Achondroplasia is associated with developmental stenosis of the cervical and lumbar canals because of a vertebral development defect, with premature fusion of the posterior elements [11, 12]. This is due to an enchondral ossification disorder resulting in early fusion between pedicles and vertebral bodies at the neurocentral synchondrosis[7, 8]. Normally the spinal canal, as measured by its interpedicular distance, widens from L1 to L5, but in patients with achondroplasia this distance progressively decreases [3]. Thus adults and some teenagers have complications [2] related to lumbosacral spinal stenosis [7, 8] that include pain, muscle weakness, and paralysis. In such cases imaging confirms shortened pedicles and decreased interpedicular distance resulting in a narrowed spinal canal [3].

Jeong et al found that all achondroplastic patients in the symptomatic group had stenosis at the level of the

intervertebral disc, suggesting that the stenosis was degenerative [3]. Therefore, a developmentally narrow canal, due to short pedicles on the one hand and premature or accelerated degenerative changes on the other hand seem to be significant for developing symptomatic stenosis of the lumbar canal[3].

Jeong et al also suggested that achondroplastic patients with a narrowed spinal canal at the L1-L3 levels are more likely to present with spinal stenosis symptoms due to concomitant disc degeneration[3]. There is need of larger studies for developing guidelines concerning the quantification, diagnosis and treatment of lumbar spinal stenosis in achondroplastic patients [3].

Differential Diagnosis List: Achondroplasia, increased intracranial pressure, dural ectasia

Final Diagnosis: Achondroplasia

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Figure 1

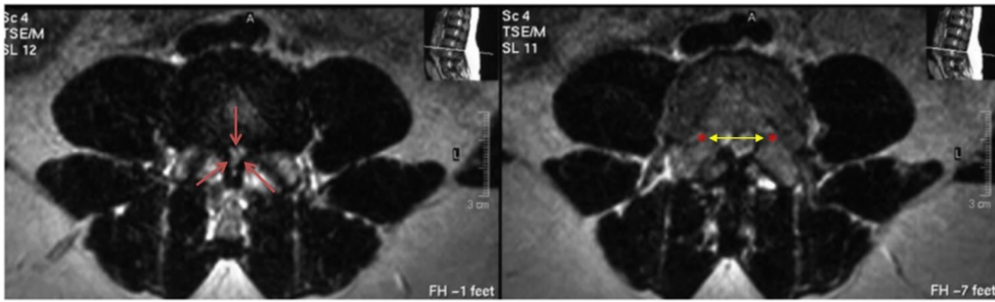
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Description: There is scalloping of the posterior aspect of the vertebral bodies. The spinal canal is stenosed at the intervertebral disk level. The upper lumbar intervertebral disks show more degenerative changes. **Origin:** E. Solomou, Department of Radiology, University Hospital of Patras, Greece

Figure 2

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Description: Cranial to caudal images: There is marked canal stenosis at the disk level (orange arrows). The pedicles of the L4 vertebra are shortened (red asterisk). The interpedicular distance is also short (yellow arrow). **Origin:** E. Solomou, Department of Radiology, University Hospital of Patras, Greece