Severe spinal stenosis in an adult achondroplastic dwarf – case report

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Abstract

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. Achondroplasia is estimated to occur in 1 in 10,000–30,000 live births4,7. The disease is autosomal dominant, but 80% of patients have new mutations. It is commonly associated with several neurological conditions such as hydrocephalus, cervicomedullary compression, cervical or thoracic cord compression, and lumbar spinal compression due to bone stenosis along the neuraxis. We report a case with severe spinal stenosis at the lumbar and thoracic levels, with minimal involvement of the cervical spine with late neurological onset in an adult patient with achondroplasia. Neurological and radiological findings and surgical procedures are discussed. The patient was admitted with profound spastic lower paraparesis and urinary incontinence. In the first operation we performed lumbar decompression and the patient improved and on the fifth day she was able to take a short walk. 3 months after the first surgery we intervened on the thoracic spine with a multi-level decompression which allowed for further neurological improvement, continued in a specialized medical facility. The case stands out as the clinical picture was dominated by the lumbar stenosis (although both lumbar and thoracic stenosis were severe at the time of presentation) with a late onset and sparing of the cervical spine.

Keywords: achondroplasia; spinal stenosis; surgical decompression

Introduction

Achondroplasia is the most common form of congenital bony dysplasia human and is one of a diseases group caused by mutations in the FGFR3 gene (1,8). It is characterized by abnormal formation of enchondral bone that leads to the stenosis of the craniospinal axis (6). The classic features of achondroplasia include a long, narrow trunk and short limbs. The head is generally large, with frontal prominence, and the face is hypoplastic. Hypotonia is a common feature in infancy and is a factor in motor developmental delay. Thoracolumbar kyphotic deformity is common. The spinal X-rays demonstrate a narrowing of the interpedicular distances in the lumbar spine (8). Head and spine MRI is recommended in achondroplasia to document the extent of spinal stenosis, cranio-spinal abnormalities (such as stenosis of foramen magnum) and the presence of hydrocephalus). The surgery of choice is currently considered to be the decompression of the spine as early as

possible to avoid significant damage to the spinal cord (8). It is worth noting that the earliest report of neurological complications associated with achondroplasia was provided by Walter Dandy in 1921 (2). We present the case of an adult achondroplastic dwarf with thoraco-lumbar spinal stenosis with relative sparing of the cervical spine with a late onset of neurological symptoms and with good response to the decompressive procedures.

Case report

A 40 year old female presented for severe hypotonic paraparesis (4/5), ongoing for two years with worsening in the last two months prior to presentation, urinary incontinence, paresthesias with hypoaesthesia in the lower limbs, and sciato-cruralgia. The upper limbs were spared of neurological deficit, motor or sensory. She presented a typical picture of achondroplasic dwarf, with a large head, short arms and legs. She exhibited hypotonic paraparesis, with Babinski sign bilaterally and ankle clonus positive bilaterally. An MRI study of the brain and entire spine was performed (Figure 1 A - C). The brain MRI showed no sign of hydrocephalus. Spine MRI showed instead a severe spinal canal stenosis at the level of lumbar spine (the minimal diameter was 4mm), especially at L2-L5 levels as well as a equally severe stenosis at the level of thoracic spine, maximal at D7-D10 levels (with a minimal diameter of 6 mm). The cervical spine showed signs of stenosis but they were minimal. Correlated with clinical status of the patient we decided a two staged decompressive surgery of the spine, starting with the lumbar stenosis. A laminectomy was performed from L2 through L4 with recalibration of the lumbar

canal through resection of the interior aspects of the articular processes. Bilateral foraminotomies were performed at all levels of the decompression.

A
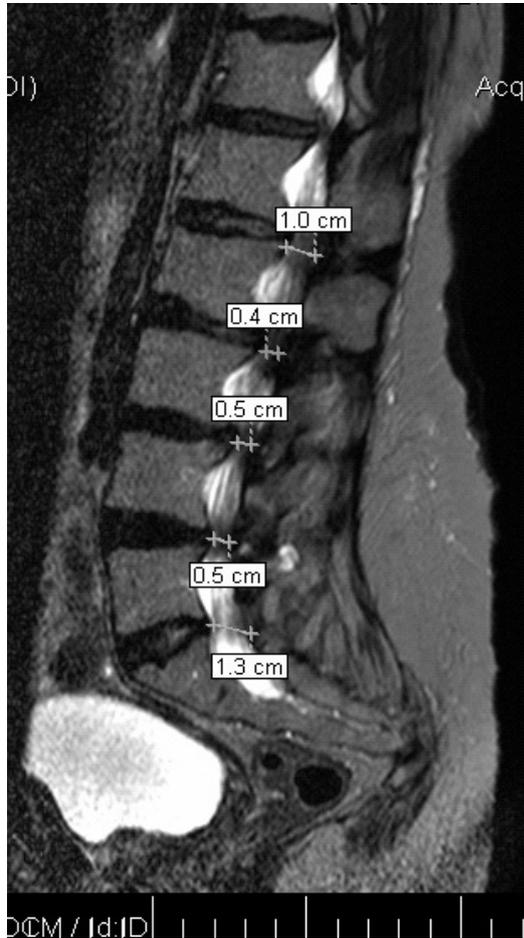
Figure 1 A. T2 weighted MR images (left – sagittal, right – axial) of the thoracic spine showing the the minimal stenosis at level of D9.
B. T2 weighted MR image of the lumbar spine with measurements of the stenosis at all levels.
C. T2 weighted MR imageof the cervical spine showing the relative sparing of the cervical and cranio-cervical junction elements.

Postoperatively, on the 5th day she was able to walk assissted. Although initially she was reserved to undergo a surgical operation on the spine, the clinical improvement after the first operation result was so significant that she decided to come back for a second surgery, on the thoracic spine, fearing a future worsening of the paraparesis. The second operation took place four months later when a decompression at D7-D10 levels was performed with further improvement in the clinical status and she entered a recovery program in a specialized facility. No complications related to surgery or anesthesia were documented during any of the interventions.

Discussion

Stenosis of the achondroplastic vertebral canal is progressive and is the substrate of neurological deficits such as intermittent claudication, nerve root compression, paraparesis, paraplegia and quadriplegia, depending on the level of stenosis. Hydrocephalus is frequently seen in these patients yet it is frequently asymptomatic and stops progressing (5). The achondroplasia presents a difficult management problem in spinal surgery and the preoperative duration of signs and symptoms and the age of the patient may equally determine the outcome of the operative treatment. Generalized stenosis is considered to be a poor prognostic factor

(due mostly to the cervical stenosis and its possible complications) (6). Prospective studies will be necessary to document that prophylactic decompression is indicated in those patients before the onset of a neurological deficit. The third and fourth decades of life are the most common age for development of thoracolumbar spinal stenosis syndrome in achondroplasia. The mean age on admission with spinal stenosis is 31 (22–37) years (4). Multi-level laminectomy is indicated if the clinical features mainly result from a narrow canal. The indication, timing, and procedure for surgical treatment of spinal canal stenosis with thoracolumbar kyphosis in achondroplasia have not been well established. Our case confirms the fact that a decompressive laminectomy even without instrumentation is a useful procedure in treating the severe spinal stenosis at both lumbar and thoracic level with a good functional outcome and an effective relief of neurological symptoms (as long as a critical clinical exam assigns the neurological status to stenosis and not to other neuraxis pathology).

Conclusions

Our patient presented in her fifth decade of life with a clinical picture dominated by the lumbar stenosis (although both lumbar and thoracic stenosis were severe at the time of presentation). Her cervical spine and cranio-cervical junction were spared from osseous stenosis typically associated with achondroplasia. The initial lumbar decompression through multileveled laminectomy provided significant relief of the neurological function. A second operation, multileveled laminectomy at the thoracic level, was performed due to clear imagistic aspects and patient's understading of possible future development, with further improvement in neurological outcome.

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