LESSON OF THE MONTH

An unusual cause of atlanto-axial subluxation

Philip Windrum, Gary D Wright, Michael B Finch

Case report

A 55 year old woman of short stature (fig 1) presented with a four week history of paraesthesiae in the fingers of both hands. This began suddenly and she subsequently developed a progressive weakness in all four limbs until she was unable to walk or stand unaided despite being previously active. For the two weeks before presentation she had noticed an occipital headache that was exacerbated by neck movements. There was no recent trauma to account for the development of these symptoms. In the past she had been diagnosed as having Schmid chondrometaphyseal dysplasia (CMD) in childhood, had undergone bilateral total hip replacements for coxa vara and required a caesarean section for the birth of her son. There was no known family history of dwarfism. On examination she was of short stature (height 4' 3") with normal facies and trunk length but short limbs. She had a thoracic scoliosis convex to the left. Neck flex-

ion produced Lhermitte's phenomenon. Power was reduced in all muscle groups to grade 3/5 and tone was increased symmetrically. All reflexes were brisk with ankle clonus present and the plantar responses were extensor. Sensory examination was normal. There was no evidence of a peripheral arthritis. Initial investigations including biochemical screen, full blood picture, erythrocyte sedimentation rate, C reactive protein, thyroid function, rheumatoid factor and autoimmune screen were normal. Radiographs of the cervical spine showed marked atlanto-axial subluxation (AAS) with exaggerated cervical kyphosis (fig 2) while radiography of the spine and peripheral joints confirmed the underlying diagnosis of Schmid CMD. Magnetic resonance imaging was performed showing partial absence of the odontoid peg with anterior subluxation of C1 on C2 and marked secondary cord compression (fig 3). She was transferred to the neurosurgical unit where trans-oral odontoidectomy and posterior cervical fusion of C1 and C2 was performed. Postoperative recovery was slow though at a recent outpatient review her symptoms had resolved and her mobility had returned to normal.

Discussion

The disorders of skeletal development include a vast array of differing conditions although attempts have been made to clarify this problem. The "Defects of growth of the tubular bones and/or the spine manifesting in childhood"¹ include the CMDs. Here there is a growth disturbance at the metaphyses of tubular bones. The epiphyses and diaphyses are less commonly involved and the spine can also be affected. The CMDs can be further subdivided to include the Schmid, McKusick and Jansen types.

Schmid first reported a mild variant of CMD in 1949.² The condition has a prevalence of 3–4 per million.³ Patients appear normal at birth and present after infancy with short stature.⁴ Symptoms resulting from lower limb involvement include pain, bony tenderness, a waddling gait, increased lumbar lordosis and bowing of the legs and may lead to confusion with rickets.⁵ Further confusion may result from neurological complications such as spinal cord compression in CMD and the proximal myopathy seen occasionally in rickets leading to lower limb weakness. Careful neurological examination should enable a distinction to be

Figure 1 Patient with Schmid chondrometaphyseal dysplasia.

Rheumatology
Department, Royal
Victoria Hospital,
Belfast BT9 7JB
P Windrum
G D Wright
M B Finch

Correspondence to:
Dr M B Finch.

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Figure 2 Cervical spine radiograph showing atlanto-axial subluxation.

Figure 3 Magnetic resonance imaging of the spine confirming spinal cord compression at C1/C2 level.

made. In contrast with CMD however, rickets can affect the complete skeleton, has the characteristic radiographic changes of looser zones and may show the typical biochemical changes of hypophosphataemia and an increased alkaline phosphatase level. Hypocalcaemia and hypocalciuria may also be noted in rickets. The

confusion has led to some patients with CMD receiving vitamin D treatment, which, needless to say, has no effect.

Unlike the more severe but less common McKusick and Jansen CMDs, patients with Schmid CMD such as this woman have a normal facies. Growth retardation and spinal involvement is classically said to be less marked with an average height of 59 inches attained. A wide spectrum of disease severity does exist however, which may account for our patient's AAS and more severe growth retardation.

The pattern of symptoms and signs heralding subacute AAS in this patient was classic with cervical pain associated with shooting pains on neck flexion, weakness and gait disturbance noted. She did not report any sphincter disturbance. The typical spastic weakness with hyperreflexia was evident and a lack of sensory symptoms affecting proprioception and vibration sense would indicate that there had been sparing of the posterior columns.

The condition is usually inherited as an autosomal dominant trait although late onset and autosomal recessive forms have also been described.⁶⁻⁷ This may account for our patient's lack of positive family history. It is caused by defects of the collagen X gene.⁸⁻¹² Collagen X is normally expressed by chondrocytes in the developing growth plates. The defect gives rise to characteristic radiological appearances with broadened and shortened tubular bones and widening of the metaphyses. This is most obvious at the knees, resulting in genu varum or valgum, or hips leading to coxa vara.¹³

In a review of 80 patients with short stature all were found to have some form of spinal involvement with odontoid hypoplasia leading to AAS occurring in 6%.¹⁴ AAS itself was more likely to occur in multiple epiphyseal dysplasia, achondroplasia and the mucopolysaccharidoses. Cases have also been associated with Down's syndrome.¹⁵ AAS is more commonly seen in rheumatoid disease and the seronegative spondyloarthropathies.¹⁶⁻¹⁸ In such conditions the mechanism of AAS differs from the congenital cases and results from erosion of the odontoid process and weakening of the transverse ligament of the atlas, which allows the odontoid process to move posteriorly from the atlas on neck flexion. Although commonly seen on radiographs in up to 25% of cases of rheumatoid disease it less commonly leads to spinal cord compression.

Spinal involvement is usually mild in Schmid CMD with sclerosis or irregularity of the vertebral end plates most typically seen. Although rare, odontoid hypoplasia and AAS have been described in Schmid CMD. Therefore, all patients of short stature with neurological symptoms or signs should be investigated with radiography, computed tomography or magnetic resonance imaging for AAS and secondary cord compression.¹⁹ Some authors also suggest routine investigation in the absence of symptoms and even prophylactic cervical fusion at an early age in high risk patients such as those with the more severe CMDs.²⁰ Patients with short stature require specific precautions

for surgery including the cervical fusion required in our patient to prevent her imminent quadraplegia. Neck flexion or extension should be avoided, which can be achieved by cervical immobilisation in a collar. The patient can also be intubated for surgery using a nasotracheal fibre endoscope.²¹ In the event of emergency surgery the anaesthetist must presume that the patient with short stature has an unstable cervical spine. At present the optimal surgical management of AAS is either posterior atlanto-axial fusion or fusion of the posterior axial arch to the occiput.

The lesson

- AAS must be suspected in patients with abnormalities of skeletal development presenting with limb weakness or paraesthesiae.
- While AAS is rare in the milder Schmid CMD, it may still occur.
- Care should be taken in all patients of short stature requiring intubation for general anaesthesia.
- AAS may result from conditions other than rheumatoid disease and the seronegative spondyloarthropathies.

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