Multiple disc herniations in spondyloepiphyseal dysplasia tarda

A case report

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Summary. *Spondyloepiphyseal dysplasia (SED) tarda is a group of inherited dysplasias in which the spine and the epiphyses of long bones are affected from late childhood. A 19-year-old male was diagnosed as SED tarda. He had a thoracic and then lumbar disc herniations which were separated by a 4-year interval. Surgical excision was performed for each disc herniation. This is the first case report of multiple disc herniations in SED.*

Résumé. *La dysplasie spondylo-épiphysaire tardive appartient à un groupe de dysplasies héréditaires dans lesquelles la colonne vertébrale ainsi que les épiphyses des os longs sont touchées tardivement dans l'enfance. Un garçon âgé de 19 ans a été diagnostiqué avec cette maladie. Il a eu deux hernies discales thoracique et lombaire à quatre ans d'intervalle. Une excision chirurgicale a été pratiquée pour chaque hernie discale. C'est le premier cas rapporté de dysplasie spondylo-épiphysaire tardive avec multiple hernies discales.*

Introduction

Spondyloepiphyseal dysplasia (SED) tarda is characterized by shortness of stature due to generalized vertebral flattening or platyspondyly. Nilsonne [11] was the first to describe this group of skeletal dysplasia which was later denoted as SED tarda by Maroteaux et al. [10]. Although Maroteaux et al. described this disorder as an X-linked recessive, both autosomal

dominant and autosomal recessive forms have been described in later reports [2, 15, 17, 22]. Since severe degenerative changes of the spine are features of SED tarda, it is probable that this condition may have a predisposition to intervertebral disc herniation [7, 8]. We report a case of thoracic and lumbar disc herniation in SED tarda.

Case report

The first admission

A 19-year-old male, who worked as a carpenter, was admitted with complaints of spastic paraplegia and dysuria. His growth was normal until the age of 4–5 years, when his short stature became conspicuous. On admission, the patient was 139 cm high and weighed 54 kg. His arm span measured 158 cm. He had a barrel-like chest, a short trunk, and disproportionately long limbs. He was of normal intelligence with a normal face structure. There was no history of consanguinity. The family history was unremarkable except that his father had a similar type of skeletal dysplasia (father's height: 140 cm).

A neurological examination revealed spastic paraplegia with loss of sensation below the T 4 level, and bladder-bowel dysfunction. He did not have bladder sensation and was totally unable to control it. Laboratory studies were normal. Radiologically, the skull and hands were normal except for clinodactyly of the fifth fingers. Radiographs of the spine showed platyspondyly, end-plate irregularity, and disc space narrowing, especially in the thoracic spine (Fig. 1a, *arrows*). There was mild acetabular dysplasia and coxa vara of the left hip joint (Fig. 2). A myelogram and a postmyelogram CT showed spinal cord compression at the T 2–3 level with a calcified extradural mass (Fig. 3, *arrowhead*).

A T 2–3 hemilaminectomy was performed, which was extended laterally to the right. The pedicles and the posterolateral aspects of the vertebral bodies of the T 2 and T 3 were removed. The disc protrusion at the T 2–3 level was excised piece by piece, and showed the degenerative cartilage histologically. No instrumentation was performed. Sensation and strength in the lower extremities improved, and bladder-bowel

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Fig. 1. Lateral radiographs of thoracic **a** and lumbar **b** vertebrae, showing platyspondyly (arrows), end-plate irregularity, and no posterior hump

Fig. 2. An antero-posterior radiograph of the pelvis. There is mild acetabular dysplasia and coxa vara of the left hip joint

Fig. 3. A myelogram **a** and a postmyelogram CT **b-d** showing spinal cord compression at the T 2-3 level **c** with a calcified extradural mass (**c**, arrowheads)

Fig. 4. MRI showing herniation of nucleus pulposus and severe compression of the dural tube at the L 2-3 level (arrow)

dysfunction was resolved. After a course of physiotherapy, he walked without assistance. In the postoperative course, the progress of kyphosis was not detected.

The second admission

He returned to his heavy labor as a carpenter after the first operation. However, after 4 years, he developed acute pain in the

low lumbar area and in the lateral area of his left thigh; he could not walk due to pain immediately prior to the second admission. On neurological examination, the Lasègue test proved positive in both legs, although there was no muscle weakness, sensory disturbance, or bladder-bowel dysfunction. A radiological examination, including a myelogram and a postmyelogram CT, revealed spinal cord compression at the L 2-3 level. An MRI revealed severe compression of the dural tube at the L 2-3 level (Fig. 4, arrow).

A partial laminectomy was performed at the L 2–3 level, extending laterally to the left. The herniated disc was excised. Immediately after the operation, his pain decreased markedly, followed by full recovery within 4 weeks.

Discussion

The patient had the tarda form of SED. His symptoms became apparent when in childhood. He was a short-trunk dwarf with a normal face, generalised platyspondyly (Fig. 1) and dysplasia of the proximal epiphysis (Fig. 2) [8]. The patient was similar to his father and therefore the inheritance pattern appeared to be autosomal dominant. No posterior hump was noted in the spine which is again consistent with the autosomal dominant pattern of inheritance (Fig. 1b). A posterior hump has been reported to be characteristics of the X-linked recessive type [8, 15].

The association of herniation of an intervertebral disc in SED tarda is rare [7]. It may perhaps be due to an abnormality of the nucleus pulposus. There is evidence of molecular abnormality of the type II collagen in the cartilage and nucleus pulposus in both SED congenita [1, 4–6, 9, 18–20] and tarda [3, 12–14, 16, 21]. Another possibility is a structural defect in the annulus which consists mainly of type I collagen, but abnormality of type I collagen has not been noted in SED. However, degenerative changes in the spine, particularly in the thoracic vertebrae, are a feature of this condition [8]. These changes, together with the patient's occupation as a carpenter may have led to the disc herniations.

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