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Rare Case of a Dyggve-Melchior-Clausen Syndrome Revealed by a Spinal Cord Compression

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Abstract **Case Report**

Dyggve-Melchior-Clausen syndrome (DMSC) is an rare autosomal recessive skeletal disorder that belongs to the category of spondyloepiphyseal dysplasia. The present work reports a case of DMCS in a nine- year-old kid who was admitted for spinal cord compression accompanied by mental retardation and dysmorphic syndrome. The patient had shown a C1-C2 dislocation with compression of the bulbo-medullary junction, platyspondyly with double ondulation of all the vertebral endplates, widely open acetabulums and epi- metaphyseal dysplasia of the femoral heads. ***Discussion***: The syndrome is clinically characterized by a staturo- ponderal delay of varying severity. Radiological characteristics consist of a scalloped appearance of the iliac wings, spondyloepimetaphyseal dysplasia and platyspondyly. The diagnosis of Dyggve-Melchior-Clausen syndrome can be suspected after a clinical evaluation, a detailed patient history, and identification of characteristic clinical and radiological signs.

Keywords: Dyggve-Melchior-Clausen, spondyloepiphyseal dysplasia, spinal cord compression.

INTRODUCTION

Dyggve-Melchior-Clausen syndrome (DMCS) is a constitutional bone disorder, with an autosomal recessive transmission, and belongs to the category of spondyloepimetaphyseal dysplasia (SEMD). It is characterized by a short trunk, harmonious dwarfism, mental retardation and microcephaly. Radiological features show scalloped iliac wings, epiphyseal-metaphyseal dysplasia and platyspondyly [1].

CASE REPORT

We report the case of a 9-year-old boy who was admitted with an evolving spinal cord compression for one week, clinically manifested by genital-sphincter disorders and heaviness in all four limbs. He was born at term through vaginal delivery, to second-degree consanguineous parents of Moroccan origin, in apparent good health and without any notion of a family disorder. On clinical examination, he was found to have a dysmorphic syndrome consisting of harmonious dwarfism, a short trunk and neck, abdominal distension, effacement of the lumbar kyphosis and a deformed, shield-shaped thorax. Imaging assessment showed C1-C2 dislocation with compression and myelopathy of the bulbo-medullary junction (Fig.1), odontoidum, hypoplasia of the odontoid process (Fig.2), platyspondyly with double ondulation of the vertebral plates (Fig.3), flattening and irregularity of the acetabular cups, widening of the pubic symphysis and upper femoral epiphyseal dysplasia (Fig.4). An

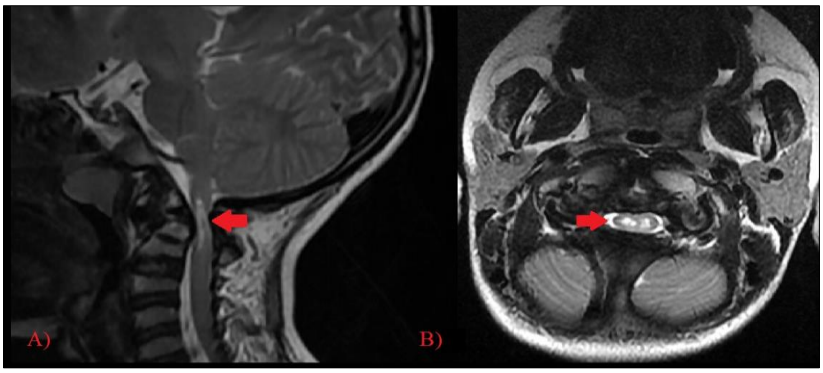

Figure 1: Cervical spine MRI in T2 weighted images sagittal (a) and axial (b) reconstructions revealing an atlantoaxial subluxation with compression and myelopathy of the bulbo-medullary junction (arrow)

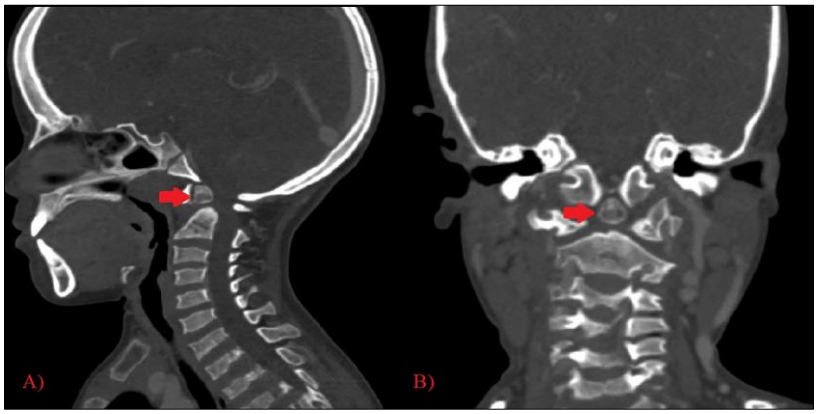

Figure 2: Cervical spine CT scan in bone window with multiplanary reconstructions in sagital (a) ans coronal (b) reconstructions showing an hypoplasia of the odontoid process (arrows)

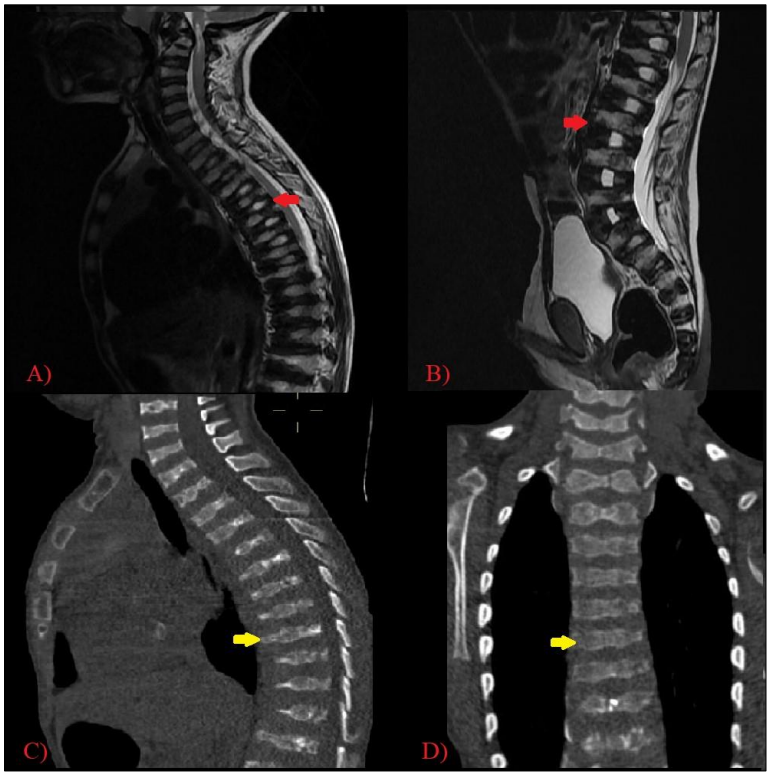

Figure 3: Spine MRI in T2 weighted images sagittal (a, b) reconstructions and bone window CT scan in sagittal (c) and coronal (d) reconstructions revealing a platyspondyly (red arrows) with double ondulation of the vertebral plates (yellow arrows)

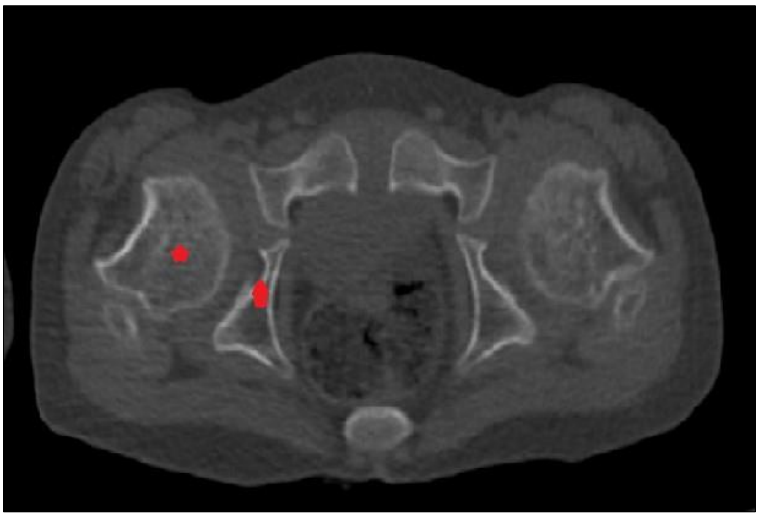

Figure 4: Pelvic CT scan in axial reconstruction showing widely opened acetabular cups (arrow) and femoral epiphyseal dysplasia (star)

DISCUSSION

Dyggve-Melchior-Clausen syndrome is a rare primary bone disease of genetic origin, belonging to the SEMD category. autosomal recessive transmission, with the gene located on chromosome 18q21.1 [2].

The syndrome is clinically characterized by a progressive and severe statural deficit affecting the trunk, postnatal microcephaly with facial dysmorphia, moderate to severe intellectual disability, rhizomelic shortening of the limbs, a shield-shaped thorax and coarse facial features. And coarse facial features [1, 2].

Radiologically, the syndrome is defined by a pathognomonic semiology featuring epiphyseal-metaphyseal dysplasia, which may be associated with hypoplasia of the odontoid process, and platyspondyly with double ondulation of the vertebral bodies. The iliac wings are narrow, with a thickened, scalloped appearance, the femoral epiphyses are subluxated, the metaphyses tend to become horizontal and the acetabuli are widely open. The thorax is cylindrical and broad. X-rays of the hands show a stocky appearance with no deformity of the metacarpal extremities [1].

This pathology frequently leads to orthopedic complications, including scoliosis, thoracic kyphosis and lumbar lordosis, spinal cord compression secondary to atlantoaxial instability, hip dislocation and knee deformity [1].

The differential diagnosis is mainly with Smith-McCort syndrome and mucopolysaccharidosis type IV, which are clinically similar but the first has no associated mental retardation, and the later presents specific radiological and enzymatic findings [2].

CMDS requires multidisciplinary management. The progressive nature of the symptoms calls for prolonged medical follow-up and possibly genetic counseling [1].

CONCLUSION

DMC syndrome is an inherited bone disorder belonging to the spondyloepimetaphyseal dysplasias, accompanied by mental retardation and dysmorphic syndrome. The diagnosis may be suspected after a thorough clinical evaluation, detailed patient history and identification of characteristic clinical signs. Imaging confirms skeletal abnormalities compatible with DMC syndrome, and identifies atlantoaxial instability, which can be life-threatening due to spinal cord compression [1, 2].

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