

Dyggve–Melchior–Clausen Syndrome

V GUPTA, A KOHLI AND V DEWAN

From Department of Pediatrics and Neonatology, Dr Ram Manohar Lohia Hospital, New Delhi, India.

Correspondence to:

Dr Vijay Gupta,

C/o Chaudhary Traders, Dal Bazaar,

Lashkar, Gwalior 474 009, MP, India.

yzaygupta@gmail.com

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Dyggve–Melchior–Clausen syndrome is a rare autosomal-recessive disorder, characterized by progressive spondylo-epi-metaphyseal dysplasia associated with mental retardation. The clinical and radiological findings resembles Morquio disease at the onset of condition, which may hinder its diagnosis. Two siblings with characteristic clinical (progressive postnatal dwarfism and mental retardation) and radiological features (irregular lace-like appearance of the iliac crests) are reported.

Key words: *Dyggve–Melchior–Clausen syndrome, Spondylo-epi-metaphyseal dysplasia.*

Dygve-Melchior-Clausen syndrome (DMCS) is a rare autosomal recessive disorder produced by mutations in the Dymeclin gene (mapped in the 18q12-21.1 chromosomal region). The patients have short trunk dwarfism, striking barrel-shape chest, sternal protrusion, kyphoscoliosis, microcephaly and various distal deformities, including genu valgum or varum, and minimal decrease in joint mobility with variable degrees of mental retardation(1,2). The most notable radiographic findings are a lacy iliac crest apophysis, hip dysplasia, platyspondyly, double vertebral hump, and odontoid hypoplasia with atlanto-axial instability. The diagnosis may be confirmed histologically, but no biochemical defect has been defined as yet(2-8). Only 58 cases have been reported in the English literature worldwide(4), mainly from Lebanon, Greenland and Morocco. For the first time, two cases are being reported from India.

CASE REPORT

Case I

A 12-year-old boy was admitted with complaints of not gaining height, with delay in developmental milestone. The patient underwent detailed clinical

examination followed by radiographic examination, which revealed skeletal dysplasia. He was the fourth child in birth order with parents having consanguinity. Birth history was normal with average size at birth. Family history revealed except the younger sibling having similar complaints. Height was 98 cm (<-3SD), weight was 12.3 kg (<-3SD percentile), pubic bone-heel distance (lower segment) was 53 cm, an upper segment length was 45 cm and an upper to lower segment ratio was 0.85 (normal ~ 0.9). Head circumference was 44.5 cms (<-3SD) (**Fig. 1**). Mental retardation was present (IQ = 41) with no other neurological signs. Waddling gait was present. His skeletal age was 6 years. An increased anteroposterior chest diameter and kyphoscoliosis was present, and proximal portions of the upper and lower limbs were short. Urine amino acids were normal and there was no urinary excretion of mucopolysaccharides. Radiological findings are detailed in **Fig. 2**.

Case II

The younger 4-year-old sib of Case I presented with similar complaints of not gaining height with delay in developmental milestones. He was 8th in birth order with normal birth history. His height was 84 cm



FIG. 1. Siblings with Dyggve-Melchior-Clausen Syndrome.

(<-3SD), weight 9.7 kg (<-3SD), pubic bone-heel distance (lower segment) was 41 cm, an upper segment was 43 cm and an upper to lower segment ratio was 0.85 (normal ~ 1.2). Head circumference was 43 cms (<-3SD). Mental retardation was present (DQ/ IQ = 30) with no other neurological signs. Waddling gait was present. His skeletal age was 4

years. An increased anteroposterior chest diameter and kyphoscoliosis was present and proximal portions of the upper and lower limbs were short. Urine amino acids were normal and there was no urinary excretion of mucopolysaccharides.

Radiological findings in both cases were similar (**Fig. 2.**)

DISCUSSION

Dyggve–Melchior–Clausen syndrome (DMC, MIM 223800) needs to be differentiated from its rare variant Smith-McCort dysplasia, mucopolysaccharidosis, spondyloepiphyseal dysplasia tarda and spondylometaphyseal dysplasia (SMD).

Both cases presented here had typical clinical and radiological features of DMC. Clinical and radiological presentation of DMC mimics storage disorder, in particular Morquio's disease (MPS IV, MIM 253010), but the presence of mental retardation and absence of corneal clouding, hyperextensibility of joints, deafness, valvular disease or mucopolysacchariduria in DMC serves to differentiate the two conditions(1).

Smith-McCort dysplasia is a close variant of DMC with all features resembling the later including the radiological findings except that there is no evidence of mental retardation in the former.

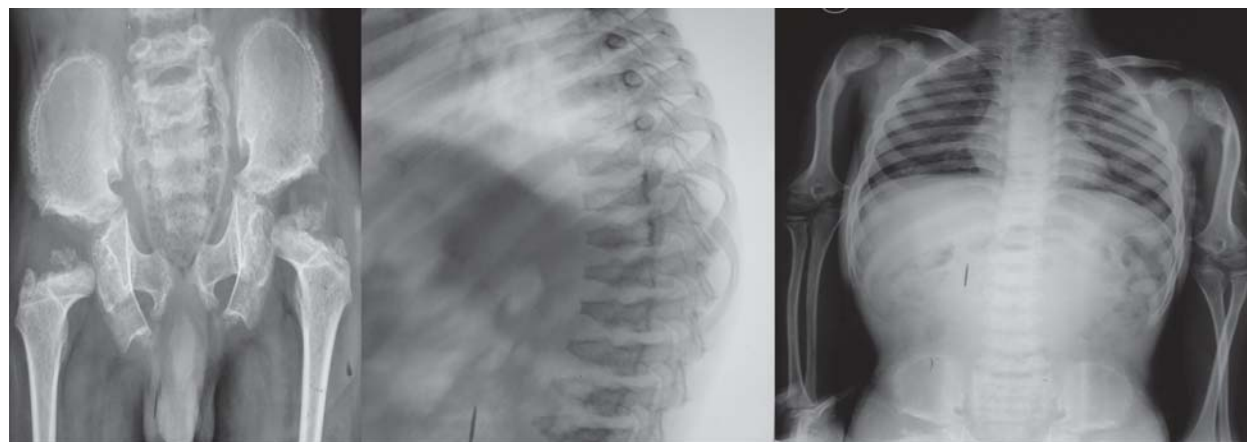


FIG. 2 Iliac bones are hypoplastic with characteristic irregular lace-like appearance of the iliac crests, Irregular ossification was present on the acetabuli and the acetabular roofs are flat. Femoral necks are short and both epiphyses and metaphyses are irregular causing lateral displacement of the femoral heads. Characteristic doublehumped appearance of vertebral bodies. Epiphyseal and metaphyseal dysplasia is present at both ends of humerus with destruction of shoulder joint.

Spondyloepiphyseal dysplasia tarda and spondylometaphyseal dysplasias can be differentiated from DMC based on their differentiating clinical and radiological features(9,10).

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