Abstract:

Three and a half year old boy with a rare Smith-McCort dysplasia is presented. Dyggve-Melchior-Clausen syndrome without mental retardation has clinical and radiographic findings similar to those of Smith-McCort dysplasia. Both of these syndromes are rare autosomal recessive disorders affecting skeletal development. The radiographic appearance of generalized platyspondyly with double-humped end-plates and the lace-like appearance of iliac crests are pathognomonic of these syndromes.

Keyword: skeletal deformity, mental retardation, short stature

Dyggve-Melchior-Clausen syndrome (DMC) and Smith-McCort dysplasia (SMC) are rare autosomal recessive osteochondrodysplasias. DMC was first described by Dyggve, et al in 1962 and SMC was originally described by Smith and McCort in 1958 as skeletal dysplasias. Since both of the genes responsible for these disorders are localized on the same chromosome, they are thought of as allelic disorders (1). DMC with mental retardation and SMC without mental retardation have similar clinical and radiographic findings.

Case Report

Three and a half year old male child, first born of third degree consanguinous parents admitted for fever, cough and breathlessness of 3 days duration. Patient on detailed clinical and radiological examination revealed skeletal dysplasia. He was born at term of vaginal delivery with birth weight of 2800g. Child attained age appropriate milestones and no other significant illness except for the skeletal deformities. There is no history of abnormality in walking, kyphoscoliosis or short stature in mother, father, sibling or relatives. On examination he was short, short neck, pectus carinatum, kyphosis, widened wrists and knock knees. His weight was 7 kilograms against the expected of 15 kilograms (less than third percentile as per...
WHO normogram), height was 72 centimeters against the expected of 98 centimeters (less than third percentile as per WHO normogram), pubic bone-heal distance (length of the lower segment) of 35 centimeters and upper segment length of 37 centimeters and an upper to lower segment ratio of 1.05:1 (normal is 1.3:1). Head circumference was 46 centimetres (between -2 SD and -3SD as per WHO standards). Blood investigations showed a normal blood count, normal haemoglobin and normal blood urea, serum creatinine and electrolytes level, serum calcium was 10mg/dl, serum phosphorus was 3.8 mg/dl, Serum Alkaline Phosphatase was 24 KA units (normal upto 22KA units). Urine for Metabolic Screening was negative and X-ray chest showed bronchopneumonia and ultrasound abdomen was normal, echocardiogram and ophthalmic evaluation were normal. Bone age was appropriate. Skeletal survey revealed characteristic double humped vertebrae and lace like appearance of iliac crests. IQ assessment was done which was normal.

Discussion

DMC and SMC are autosomal recessive allelic osteochondrodysplasias caused by mutations on the same genes. Both phenotypes have been mapped to chromosome 18q21.1 and mutations in the DYM (dymeclin) gene (2). DMC and SMC are both disorders of bone and cartilage that are characterized by a short trunk and extremities and a barrel shaped chest, the radiographic appearances of both disorders are similar (3). Platspondyly with double-humped endplates, anterior beaking and scoliosis are seen in all vertebrae. A barrel-shaped chest, significant anterior convexity of the sternum and widened costochondral junctions may also be present in radiological examinations. Small iliac bones, irregular lace-like appearance of iliac crests, widened sacroiliac joints, decreased sacrosciatic notches, widened pubic rami and ischiopubic synchondrosis, a flat acetabular roof with irregular ossification, lateral displacement of the femoral heads, and a widened symphysis pubis may be present. A small scapula, concave inferior scapular angle with irregular ossification, flat glenoid fossa, and a wide and spreading acromion may also be seen. Shortness in varying degrees in long bones, irregular metaphyses, multicentric ossification and deformities on humeral and femoral epiphyses, and flat epiphyses may be present as well. Small carpal, metacarpal, and metatarsal bones in the hands and feet, cone-shaped epiphyses, and accessory epiphyses are also encountered (1, 3). Double-humped vertebrae and an irregular lace-like appearance of iliac crests are pathognomonic for DMC and SMC (3). In this case the characteristic double-humped vertebrae and an irregular lace-like appearance of iliac crests pathognomonic for DMC and SMC were present. There was scoliosis, some degree of shortening of long bones and concave inferior scapular angle with irregular ossification. In the differential diagnosis achondroplasia must be considered in which vertebrae are short and flat, pedicles are short, and the spinal canal is narrow. Anterior wedge-shaping may be present in one or more vertebrae. In this case, the double-humped end-plates due to wedge shaped ossification defect were not compatible with the vertebral changes seen in achondroplasia. Mucopolysaccharidosis Type IVA is one of the most important diseases that must be distinguished from DMC and SMD with radiographic findings (4). Severe
Platyspondyly, vertebral irregularities, hypoplastic odontoid process, atlantoaxial subluxation, pectus carinatum of the thorax, increased anteroposterior diameter of the chest, narrowed iliac crests, an oblique acetabular roof, coxa valga and femoral epiphyseal changes, as well as epiphyseal and metaphyseal irregularities of long bones, especially in the end-stage of the disease, are seen in mucopolysaccharidosis and are useful and important for making a differential diagnosis. A lace-like appearance of iliac crests, concavity of the edge of the scapula due to defective ossification, no excretion of mucopolysaccharides in urine screen, a horizontal acetabular roof, and double-humped end-plates of vertebrae are important signs supporting the diagnosis of DMC or SMD. Metatrophic dysplasia (MD) may mimic the signs of DMC and SMD. Kyphosis or scoliosis, small and spherical sacrosciatic notches, a flat and irregular acetabular roof, and odontoid hypoplasia are the signs of MD. But MD causes general shortness in all long and short tubular bones. Furthermore, a lace-like appearance of iliac crests is a significant sign for making a differential diagnosis between MD and SMD or DMC. Metatrophic dysplasia (MD) may mimic the signs of DMC and SMD. Spondyloepimetaphyseal dysplasia (SEMD) and spondylometaphyseal dysplasia (SMDI) may also mimic the radiological findings present in this patient. Both of these disorders primarily affect the vertebrae. Irregularities on end-plates of vertebrae and platyspondyly may be present in both disorders. SEMD may mimic DMC or SMD, especially with features such as platyspondyly type tarda, hill-shaped irregularities in the central and posterior aspects of the superior and inferior end-plates of vertebrae, a hypoplastic cone-shaped odontoid process, mild or severe epiphyseal dysplasia, a small pelvic bone, and a short femoral neck. Irregular lace-like appearance and a double humped appearance of the end-plates of vertebrae due to wedge-shaped defect, as seen in vertebrae radiographs, are important signs for distinguishing DMC or SMD from SEMD-type tarda or other SEMDs.

Since the child had characteristic double humped vertebrae and an irregular lace-like appearance of iliac crests without mental retardation was diagnosed as Smith-McCort dysplasia

References


5 Dyggve-Melchior-Clausen syndrome without Mental retardation (Smith-McCort dysplasia); Diagn Intervent Radiol 2005; 11:163-165
Three and a half year old child with Smith–McCort syndrome with short stature, short neck, pectus carinatum, kyphosis, widened wrists and knock knees. The characteristic double humped vertebrae in spine skiagram in Smith–Mccort syndrome.

Picture 1: Three and half year old child with Smith–Mccort syndrome with short stature, short neck, pectus carinatum, kyphosis, widened wrists and knock knees.

Picture 2: The characteristic double humped vertebrae in spine skiagram in Smith–Mccort syndrome.

Picture 3: The characteristic lace like appearance of iliac crest in x ray pelvis in Smith–Mccort syndrome.