SPONDİLOEPİFİZYAL DİSPLAZYA KONJENİTA
(Olgu Sunumu)

SPONDELOEPHYSEAL DYSPLASIA CONGENITA
(Case Report)

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Özet

Spondiloepifizyal displazya konjenitalı, karakteristik olarak normal eller ve ayaklar, paytak yürüyüş, pektus karinatus, kısa boyun ve yassı yüzü 7 yaşında bir erkek vaka sunuldu. Radyolojik bulgular epifizler ve metafizlerde nokta kalsifikasyonlar, sklerotik ve radyolusent bölgelerle metafizin genişlemesiyle metafizyal disorganizasyon, generalize osteopeni ve geri kalmış kemik yaşını içerir. Karakteristik olarak femur ve tibia kışırdı ve distal femurda metafizyal düzünsizlikler görülü. MRI da kemik taramasında ulna ve radiusta, el bileğinde, kalça ve eklemlerde kronolojik yaşa göre beklenenden daha az epifizyal aktivite görülmekteydi.

Anahtar kelimeler: Spondiloepifizyal displazya konjenitala, Kemik sintigrafisi, Manyetik rezonans görüntülemesi

Summary

A case of spondyloepiphyseal dysplasia congenita is presented here in a seven-year-old male with lumbar lordosis, flat face, short neck, pectus carinatus, waddling gait and characteristically normal hands and feel. Radiological findings included retarded bone age, generalized osteopenia and metaphyseal disorganization with widening of metaphysis with sclerotic and radiolucent areas and stippled calcifications in epiphysis and metaphysis. Characteristically femur and tibia were short with metaphyseal irregularities appearing in the distal femur. Magnetic Resonance Imaging (MRI) revealed hyper intense metaphysis and epiphysis of ulna and radius and in bone scanning, elbows, hips and ankles revealed less epiphyseal activity than expected for the chronological age.

Key words: Spondyloepiphyseal dysplasia congenital, Bone scintigraphy, Magnetic resonans Imaging
Skeletal dysplasias are a heterogeneous group of disorders, many of which have been well defined by clinical, radiological and morphological criteria, but the underlying biochemical defect is unknown in most of them (1). Spondyloepiphyseal dysplasia congenital (SEDC) is characterized by short-trunk dwarfism identifiable at birth, and generalized platyspondyly and dysplasia of the proximal epiphyses. Ocular complications, including myopia, retinal detachment, cataracts, and glaucoma are known to be associated with this disorder (2). Here we report a case with SEDC and would like to draw attention to the findings of skeletal scintigraphy and MRI.

Case Report

A seven-year-old boy was admitted to the Department of Pediatrics of Atatürk University Medical School for growth retardation and chest deformity. The family did not know the birth weight and length. There was no similar case in the family but a cousin. He had no history of a previous serious illness. His physical measurements were: height 97 cm (<3%), upper segment 47 cm, lower segment 40 cm, weight 13 kg (<3%), head circumference 50 cm (between 2% and 50%). His face was flat and neck was short. Pectus carinatus, lumbar lordosis, waddling gait were noticed, his hands and feet were normal in length and shape (Figure 1 a- b). Neither retinal detachment nor myopia were detected. Blood cell count, urinalysis, serum calcium, phosphorus and alkaline phosphatase values and growth hormone, T3, T4 and TSH were within normal levels. Radiological findings included retarded bone age and generalized osteopenia and lumbar lordosis. Thorax was broad and short, intercostal spaces were narrow. Radiographs of hands showed minimal widening of metaphysis with sclerotic and radiolucent areas. The space between metaphysis and epiphysis was also wide (Figure 2a).
Figure 3. Magnetic Resonance Imaging (MRI) of Hand and Arm. Note Hyper Intense Metaphysis and Epiphysis of Ulna and Radius, and the Hypo Intense Space Between Them

Figure 4. Bone Scintigraphy Shows Expected Epiphyseal Activity on Shoulders, Wrists and Knees, and Inadequate Activity on Elbows and Ankles

Stippled calcifications in epiphysis and metaphysis of humerus were noticed. Pelvis was small, iliac wings were low and broad, femur and tibia were short with metaphyseal irregularities appearing in the distal femora and femur had no epiphysis; acetabular roofs were more horizontal than normal and femoral heads looked granular. Irregular and flattened aspect of the epiphyseal centers is more evident in the femoral capital epiphysis. Metaphyseal ossification lines were irregular and convex (Figure 2b). Varus deformity of the femoral neck was noted. MRI of the hand revealed hyper intense metaphysis and epiphysis of ulna and radius. A hypo intense space was noticed between them (Figure 3). In bone scanning with Tc-99-MDP(methylene di-phosphate), increased uptake on shoulders, wrists and knees was interpreted normal due to high epiphyseal activities in these areas compatible to patient's age. Elbows, hips and ankles revealed less epiphyseal activity than expected for the chronological age (Figure 4).

Discussion

Spondyloepiphyseal dysplasia congenital (SEDC) is a well recognized skeletal disorder with estimated incidence of approximately 1 in 300000 (3). Although SEDC may be suspected at birth, no history about birth was obtained. Inheritance is by an autosomal dominant trait (4), having a cousin with short stature and disproportional shape was compatible with autosomal inheritance. SEDC has been reported to result from the deletion, the duplication of part of the COL2A1 gene, and from single base substitutions (5,6). Murray et al (1) reported that some of SEDCs are the result of mutations in Type II collagen that occur between the carboxyl terminus and CNBr peptide II. Differential diagnosis is primarily concerned with other short-trunk dwarf conditions which roentgenographically show platyspondyly or dwarf conditions causing kyphoscoliosis such as achondroplasia, metatrophic dwarfism, Morquio's disease, spondyloepiphyseal dysplasia tarda and diastrophic dwarfism (4-7). Although, no kyphoscoliosis was noticed in our case, lumbar lordosis, flat face and short neck, pectus carinatus, waddling gait and normal hands and feet are characteristically signs of SEDC (8). SEDC may be easily confused with Morquio's disease (4). It is differentiated from Morquio's disease by its earlier manifestations, characteristic roentgenographic features, its inheritance as a dominant trait, its lack of corneal clouding, and keratosulphaturia (7). In Morquio's disease there is a wide flare of the ilia and acetabular hypoplasia with an iliac angle surpassing 180° in some cases, ossification proceeds normally and the well developed femoral neck is in valgus position (7). Sponyloepiphyseal dysplasia tarda appears at puberty and manifests a peculiar hump-like deformity of the lumbar centra with a deep narrow pelvic configuration which differentiates it from the congenital form (4). Diastrophic dwarfism can be differentiated by the presence of clubfeet and the absence of severely retarded skeletal maturation and progressive severe kyphoscoliosis and peculiar "battle-ax" pelvic configuration, with marked iliac flaring of metatrophic dwarfism distinguish it from SEDC (4). Retinal detachment is one of the most serious complications of SEDC, but the incidence remains a controversial topic (2). Although in different series, different results were reported, we did not detect retinal detachment or myopia in our patient. The only MRI finding of our case was the hyperintensity of metaphysis and epiphysis of ulna and radius. In our case x-ray findings revealed retarded bone age, generalised osteopenia and metaphyseal disorganization, and widening of metaphysis with
sclerotic and radiolucent areas, stippled calcifications in epiphysis and metaphysis were reported. Especially, alterations in acetabular roofs and fossae were important. Additionally bone scanning revealed less epiphyseal activity in elbows, ankles and hips probably due to inadequate synthesis of new bone and cartilage. Although the findings will be enlightening some aspects of the SEDC, the family refused the bone biopsy. We reported this case especially in order to emphasise bone scanning and MRI findings. We believe further studies of bone metabolism will explain the exact defect in this disease.

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References


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