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CASE REPORT

Kozlowski type spondylometaphyseal dysplasia: a case report with literature review

Mehmet Selim Nural, H. Barış Diren, Özgür Sakarya, Türkay Yalın, Ayhan Dağdemir

ABSTRACT
Spondylometaphyseal dysplasia is a type of bone dysplasia characterized by vertebral and metaphyseal changes of varying severity. Diagnosis of the disease is difficult because the severity of bone involvement differs and symptoms change according to the age of the patient. In this study, radiographic findings of a 16 month-old male patient diagnosed as Kozlowski type spondylometaphyseal dysplasia is reported.

Key words: • *dysplasia* • *bone dysplasia* • *osteochondrodysplasia*

Spondylometaphyseal dysplasia (SMD) is an extremely rare bone dysplasia that was defined by Kozlowski for the first time in 1967 (1). Its various subtypes were defined in terms of radiological findings and genetic transmission. Prompt and correct diagnosis is very important in order to ensure correct treatment and to provide the necessary consultation services to families. Herein, we present a case of SMD that was diagnosed with radiological and clinic findings, and typed by literature review.

Case report

A 16-month-old male presented with bronchopneumonia and was evaluated for the presence of a metabolic disorder or skeletal dysplasia because costal ends appeared enlarged on chest radiogram. The patient was the only child of a first-degree relative mother and father. Neither his parents nor other family members had any anomalies such as gait disorder, short stature, or kyphoscoliosis. He was a full-term baby, delivered by normal, spontaneous vaginal route, with a birth weight of 3000 gr. He had a bilateral inguinal hernia operation 3 months after birth and was treated several times for lung infections; additionally, his ability to walk had been delayed and upon presentation he could not walk without assistance. His blood calcium, phosphorus, and alkaline phosphatase levels were normal. There were no amino acids or mucopolysaccharides in his urine analysis. He was 64 cm in length (< 3rd percentile), his bilateral total arm length was 65 cm (3rd-10th percentile), his pubis-calcaneal length (lower segment length) was 26.5 cm, and his upper/lower segment ratio was 1.39 (normal: 1.52). On anterior-posterior chest radiogram, widening and scalloping of anterior costal ends were present (Figure 1). On vertebral radiograms, there was increment of intervertebral disc space, significant decrease of vertebral corpus height, and sharpening of anterior aspects of the vertebrae; posterior elements were normal (Figure 2). On anterior-posterior pelvic radiogram, the acetabular roof was smooth and pubic distance was increased secondary to delay of ossification. The femoral neck was short and blunt, femoral metaphyses were significantly widened, and the proximal femoral epiphysis was undeveloped. On extremity radiograms, the tibia and fibula were short and thick, and the proximal and distal metaphyses were widened bilaterally (Figure 3). The humerus, radius and ulna were short and thick, the proximal and distal metaphyses were significantly widened, metaphyses of short tubular bones of hands were scalloped, especially distal metaphyses of the metacarpi, and epiphyses were observed as buried. Additionally, pseudo-epiphyses were present in metacarpal bones (Figure 4).

Discussion

SMD is a skeletal dysplasia characterized by involvement of metaphyses of vertebrae and tubular bones, but various types are defined

From the Departments of Radiology (M.S.N. ✉ *msnural@omu.edu.tr*, H.B.D., T.Y.) and Pediatrics (Ö.S., A.D.), Ondokuz Mayıs University, School of Medicine, Samsun, Turkey.

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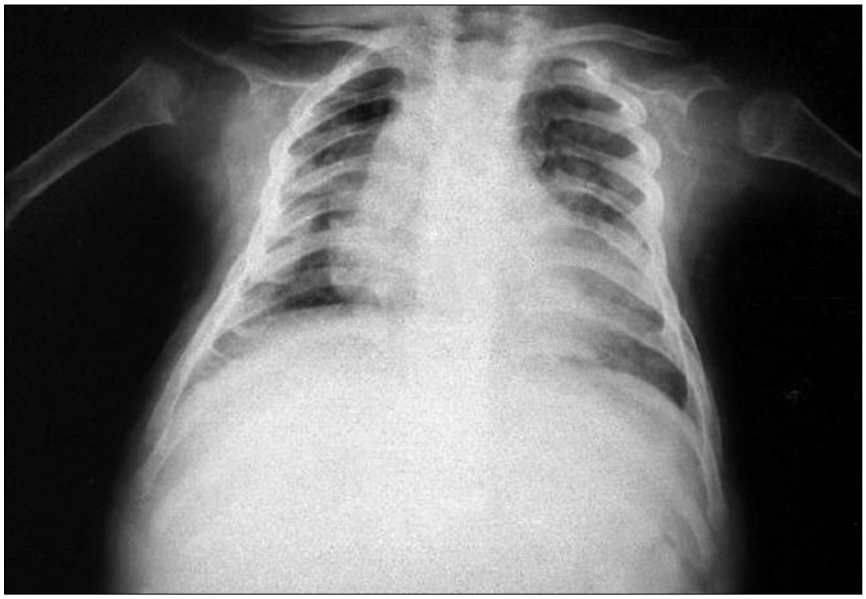

Figure 1. Widening of anterior costal ends and scalloping are seen on posteroanterior chest radiogram.

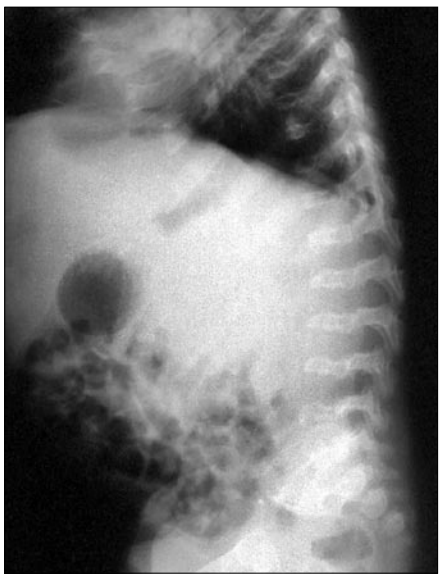

Figure 2. Sharpening of vertebral bodies anteriorly and flattening of bodies are seen on thoracolumbar vertebral graphy.

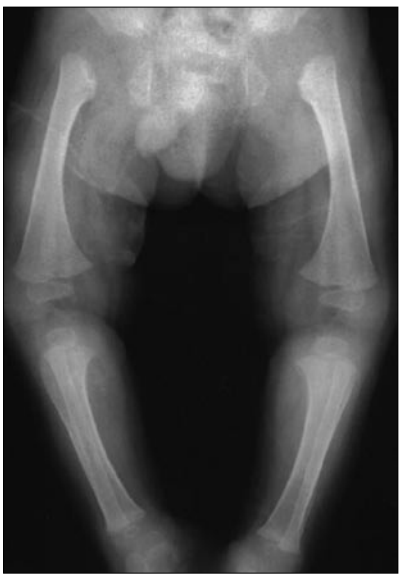

Figure 3. Shortening and widening of long tubular bones, significant widening of proximal and distal metaphyses, widening and blunting of femoral necks, and undeveloped proximal femoral epiphysis are seen on anteroposterior lower extremity radiogram. Retardation of ischiopubic bone ossification and widening of symphysis-pubis distance are seen.

according to severity of the condition and affected segments, its clinical findings increase with age. Kozlowski type is the most commonly seen SMD with autosomal dominant transmission, but because of mutations, X related recessive transmission is also possible (1, 2). Patients are normal at birth; in early infancy growth retardation, truncal shortness, joint movement restriction, genu valgum, and mild scoliosis can be seen. In adulthood, significant dwarfism develops. Radiographic findings are, widening, scalloping, and irregularity of the metaphyses of tubular bones, shortness of the femoral neck, progressing coxa vara, severe and diffuse platyspondyly of vertebral bodies, delay in ossification of carpal bones, and delay in bone age (2, 3). In addition to these classical findings, Bagga et al. defined hypocalciuric hypercalcemia in a Kozlowski type SMD case (4).

Several subtypes of SMD were defined according to severity of the findings, properties of the involved bones, and transmission pattern. Kozlowski made the first classification in 1982 and categorized SMD into 7 types (Table 1) (5). The classical form with usual findings and autosomal dominant transmission is type 1. Type 1 is also referred to as Kozlowski type SMD in the literature (5, 6).

Maroteaux and Sprange divided SMDs into 3 main groups regarding femoral neck involvement, in 1991. In this classification, subgroups were also defined according to vertebral involvement (Table 2) (7).

Apart from the classification models presented above, additional SMD types were also reported. Some are referred to by the names of those who defined the condition, such as Sutcliffe type (8), Borochowitz type (9), and Jansen type (10); others are referred to by the country in which it was defined, such as Algeria type (11), East Africa type (3), and Japan type (12); some are named after the most significantly affected body part as axial type (13).

Kozlowski, in a case study of two brothers in 2003, described different findings resembling SMD, such as compressed nose root, forehead protuberance, forearm deformity, and hyperlordosis. This case was reported to be different from previously reported SMDs and may be a unique SMD (14). Ehara et al. reported 3 cases (2 of whom were brothers) with platyspondyly, small thorax and scalloping of the anterior aspects of ribs, irregularity of the proximal femoral metaphysis, and lentiform changes in iliac wings, with retinitis pigmentosa and optic atrophy. It was noted that this was a new SMD not previously described, and was called axial SMD (13). However, based on Maroteaux classification, this type was type A4.

The findings in our case were severe generalized platyspondyly, scalloping of anterior costal ends, shortness of femoral necks, widening and irregu-

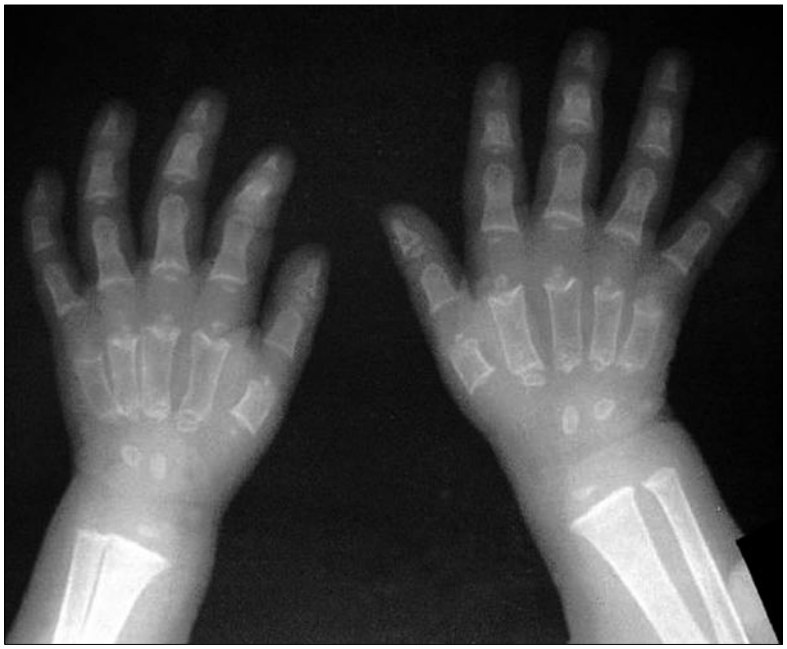

Figure 4. Widening of distal metaphyses of the radius and ulna, shortening and widening of short tubular bones of the hand, scalloping of metaphyses especially in distal ends, buried epiphyses and metacarpal pseudoepiphyses are seen on anteroposterior hand radiogram.

Table 1. Spondylometaphyseal displasia classification of Kozlowski, 1982 (5)

Spondylometaphyseal displasia	Severity of radiological finding		Genetic transmission
	Vertebrae	Metaphyses	
Type I	++++	++++	Autosomal dominant
Type II	++++	++	Autosomal recessive
Type III	+++	++	Autosomal recessive
Type IV	+	+	Autosomal dominant
Type V	++++	++++	Autosomal recessive
Type VI	++	++	Autosomal recessive
Type VII	++++	+++	Autosomal recessive

larity of proximal femoral metaphyses, growth retardation of proximal femoral epiphyses, widened metaphyses representing severe involvement of short tubular bones of the hand and scalloping of metacarpal metaphyses, shortness of long tubular bones, and metaphyseal widening. The most characteristic features of Kozlowski type SMD, platyspondyly, bone development retardation, and metaphyseal scalloping were observed in our case. However, since our patient was 16 months old and proximal femoral epiphyses were underdeveloped, coxa vara deformity could not be evaluated. According to these findings, our case was considered type 1.

Kozlowski type SMD should be differentiated from Morquio disease. In the past, Kozlowski type SMDs were reported as Morquio disease (2); but Morquio disease can be easily diagnosed by the presence of corneal opacities and keratin sulphate in urine.

Dyggve-Melchior-Clausen syndrome (Smith-McCart dysplasia), which does not include mental retardation, should be included in the differential diagnosis. Lentiform appearance of iliac crests with a double hump view of vertebral disc surfaces and widespread platyspondyly, barrel chest, and ossification deficiency of the inferior part of the scapula are characteristic radiographic features (15, 16), which help differentiation. Sponastrime dysplasia, which affects both vertebrae and metaphyses, should also be included in the differential diagnosis; however, findings like metaphyseal linings, forehead protuberance, saddle nose, spinal changes (17) and mild systemic osteoporosis differ from those observed in our case.

Table 2. Dysplasia classification of Maroteaux and Spranger, 1991

Severe coxa vara	Mild vertebral and metaphyseal changes (type A1 (Sutcliffe))
	Mild vertebral changes, more severe metaphyseal changes (Type A2 (Schmidt, Langer)
	Round vertebral bodies (type A3)
	Anteriorly protruded flat vertebral bodies (type A4)
Average degree changes in femoral neck	Mild irregularities of vertebral corpus, shortening of tubular bones of the hand, and metaphyseal irregularity (type B1)
	Widespread platispondyly, brachyolmy-like (type B2)
Mild metaphyseal changes in femoral neck	Irregularly contoured square-shaped vertebral corpus (type C1)
	Average flattening and long vertebral bodies on lateral film (type C2)
	Trapezoid appearance of vertebral body (type C3)

In conclusion, SMD is a rarely seen disease in which, radiological evaluation is of extreme importance in diagnosis and classification. Early and correct diagnosis is important, especially in terms of genetic consultation for the family. Various conflicts in typing and defining subgroups highlight the necessity for additional studies about SMD.

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