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RECOGNIZING SPONDYLOMETAPHYSEAL DYSPLASIA: AVOIDING MEDICOLEGAL MISINTERPRETATION IN PEDIATRIC RADIOLOGY

Radio-Diagnosis

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ABSTRACT

Spondylometaphyseal dysplasia (SMD), Kozlowski type, is a rare skeletal disorder characterized by platyspondyly, metaphyseal flaring, and occasional corner fractures, often mimicking nonaccidental trauma. Misdiagnosis can lead to unwarranted legal consequences. In a five-year-old boy with short stature and delayed ambulation, radiographs revealed flattened vertebral bodies with anterior beaking, metaphyseal widening and cupping at the distal femur, proximal tibia, and distal radius/ulna, and a distal femoral corner fracture. A structured radiologic approach, combined with clinical insight, confirmed SMD, preventing misinterpretation as child abuse. This case emphasizes the importance of skeletal surveys, differential diagnosis, and multidisciplinary consultation in pediatric radiology for accurate diagnosis and management, supported by recent advances in imaging and genetics.

KEYWORDS

Spondylometaphyseal Dysplasia, Pediatric Radiology, Metaphyseal Corner Fractures, Nonaccidental Trauma, Skeletal Survey, Platyspondyly

INTRODUCTION

Skeletal dysplasias, encompassing over 450 genetic disorders, affect bone and cartilage development with a prevalence of approximately 1 in 5,000 births [1]. Spondylometaphyseal dysplasias (SMDs) are a rare subset involving the spine and metaphyseal regions of long bones, with the Kozlowski type linked to pathogenic variants in TRPV4 or COL2A1 genes, critical for chondrocyte function and skeletal growth [2]. SMD manifests in early childhood with disproportionate short stature, delayed motor milestones, and radiologic findings like platyspondyly and metaphyseal flaring [3]. Recent advances in prenatal imaging have improved early detection of such disorders [4].

Diagnosing SMD is challenging due to its rarity (incidence ~1 in 100,000) and overlap with conditions like rickets, mucopolysaccharidoses, metaphyseal chondrodysplasias, and achondroplasia [5]. Notably, metaphyseal corner fractures in SMD can resemble nonaccidental trauma, posing significant medicolegal risks [6]. Misdiagnosis may trigger unnecessary child protection investigations, causing family distress. This report illustrates the radiologic hallmarks of SMD, Kozlowski type, strategies to differentiate it from nonaccidental trauma, and the critical role of skeletal surveys, supported by comprehensive genetic and imaging studies [7].

Case Presentation

A five-year-old boy presented with short stature and difficulty walking. He began ambulating at 2.5 years and struggled with running and climbing stairs. There was no history of trauma, fever, joint pain, or family history of skeletal disorders, and birth history was unremarkable, with no prenatal indications of severe skeletal dysplasia [8]. Physical examination revealed disproportionate short stature (height <3rd percentile) with rhizomelic limb shortening (upper segment:lower segment ratio 1.3, reference ~1.1). The trunk appeared relatively long, with no joint swelling, scoliosis, cardiac involvement, or neurologic deficits, distinguishing this case from more severe forms like Sedaghatian SMD and some TRPV4-related cases with neurological overlap [9]. Laboratory tests, including serum calcium, phosphate, and alkaline phosphatase, were normal, ruling out metabolic bone diseases like rickets [10]. Informed consent was obtained from the guardians, and the case was exempt from institutional review board review.

Imaging Findings

A skeletal survey was conducted to assess the growth pattern in a five-year-old boy with suspected spondylometaphyseal dysplasia (SMD), as recommended for evaluating skeletal dysplasias [11]. The survey revealed key radiologic findings, detailed in Table 1 and illustrated in Figures 1–6. Lateral thoracolumbar radiographs demonstrated generalized platyspondyly, characterized by flattened vertebral bodies with reduced height and anterior beaking at T12–L2, without scoliosis or segmentation anomalies, distinguishing this presentation from SMD corner fracture type (Figure 1). Anteroposterior radiographs of the lower extremities showed pronounced metaphyseal widening, flaring, and cupping, particularly at the distal femur and proximal tibia, with irregular, trumpet-shaped metaphyseal margins consistent with SMD Kozlowski type (Figures 2, 5, and 6). A subtle metaphyseal corner fracture was identified at the distal femur, lacking soft tissue swelling or trauma history, indicative of skeletal fragility rather than nonaccidental injury (Figure 3) [12]. Wrist radiographs, both anteroposterior and lateral, revealed mild metaphyseal widening and irregularity at the distal radius and ulna, with trumpet-shaped margins and normal carpal bones, showing no delayed ossification or abnormal morphology (Figure 4). Additionally, hand radiographs indicated mild metacarpal shortening, with normal epiphyses, pelvis, and ribs, aligning with established descriptions of skeletal dysplasias (Figure 5) [13].

Table 1: Radiologic Findings in SMD

Region	Findings
Spine	Generalized platyspondyly with flattened vertebral bodies; anterior beaking at lower thoracic/upper lumbar levels; preserved disc spaces.
Long Bones	Metaphyseal widening, irregularity, flaring, and cupping at distal femur, proximal tibia, and distal radius/ulna; trumpet-shaped metaphyseal margins.
Fracture	Subtle metaphyseal corner fracture at distal femur, without soft tissue swelling.
Wrist	Mild metaphyseal widening and irregularity at distal radius and ulna; normal carpal bones.
Other	Mild metacarpal shortening; normal epiphyses, pelvis, and ribs.

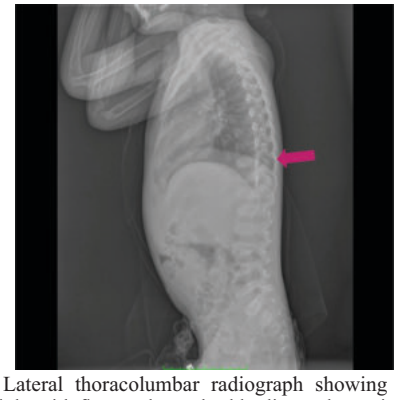

Figure 1 Lateral thoracolumbar radiograph showing generalized platyspondyly with flattened vertebral bodies and anterior beaking at T12–L2 (arrow indicates anterior beaking).

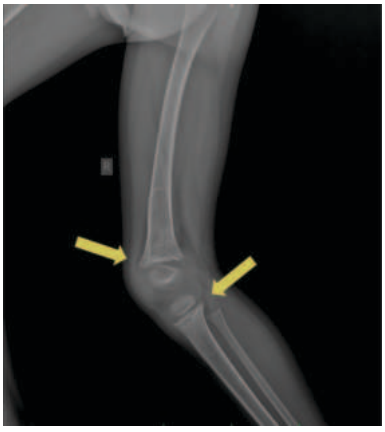

Figure 2 Anteroposterior radiograph of the lower extremities demonstrating metaphyseal widening, flaring, and cupping at the distal femur and proximal tibia (arrows highlight trumpet-shaped metaphyseal margins).

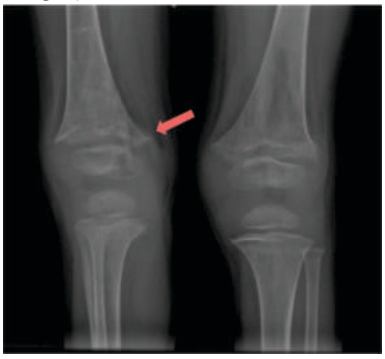

Figure 3 Anteroposterior radiograph of the distal femur revealing a subtle metaphyseal corner fracture without soft tissue swelling (arrow points to the corner fracture).

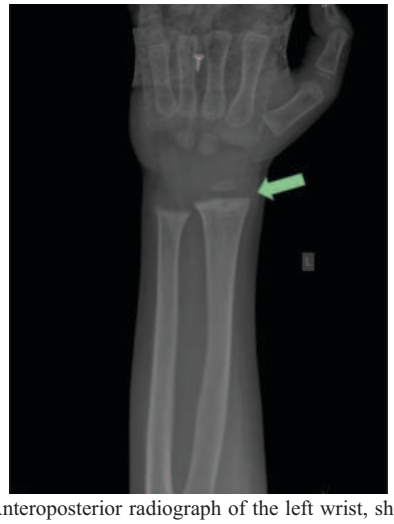

Figure 4 Anteroposterior radiograph of the left wrist, showing mild metaphyseal widening and irregularity at the distal radius and ulna (arrows indicate trumpet-shaped metaphyseal margins)

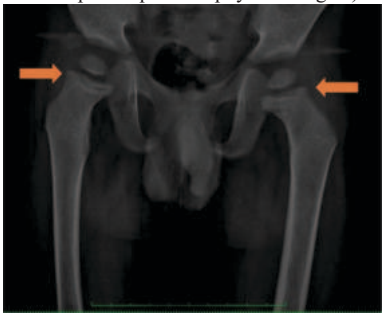

Figure 5 Anteroposterior radiograph of the pelvis and proximal femora demonstrating normal epiphyses and pelvis with subtle metaphyseal flaring at the proximal femora (arrows highlight flaring).

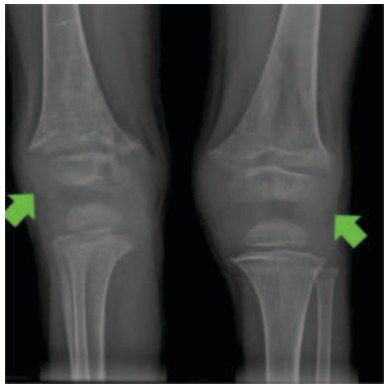

Figure 6 Anteroposterior radiograph of the lower extremities showing metaphyseal widening, irregularity, and cupping at the distal femora and proximal tibiae (arrows indicate trumpet-shaped metaphyseal margins).

Diagnosis

The combination of disproportionate short stature, delayed motor milestones, normal biochemical parameters, and radiologic findings (platyspondyly, metaphyseal flaring, and distal femoral corner fracture) supported a diagnosis of SMD, Kozlowski type [14]. Genetic testing for TRPV4 and COL2A1 variants was recommended to confirm the diagnosis, as these mutations are well-documented in SMD, but was not pursued due to financial constraints, unlike cases where prenatal detection might prompt early genetic evaluation. Differential diagnoses, such as achondroplasia, Sedaghatian SMD, or other SMD variants, were ruled out based on normal epiphyses, absence of cardiac involvement, and lack of other characteristic features [15]. The patient was referred to pediatric orthopedics and genetics for multidisciplinary management.

DISCUSSION

SMD, Kozlowski type, is an autosomal dominant disorder caused by TRPV4 or COL2A1 mutations, disrupting chondrocyte regulation and skeletal development [16]. Clinically, it presents with disproportionate short stature and gait difficulties, with normal cognitive development. Radiographically, platyspondyly, metaphyseal flaring, and corner fractures are hallmark features, as seen in this case (Figures 1–6). The differential diagnosis includes metaphyseal chondrodysplasia (Schmid type), Morquio syndrome, rickets, spondyloepiphyseal dysplasia, achondroplasia, and Sedaghatian SMD, each distinguished by specific clinical and radiologic features [17]. Management requires a multidisciplinary approach, including orthopedics, physical therapy, and genetic counseling [18]. Metaphyseal corner fractures, often associated with nonaccidental trauma, must be carefully evaluated with clinical history and skeletal surveys to avoid misdiagnosis, as emphasized by the American Academy of Pediatrics [19]. Skeletal surveys remain the primary imaging modality, with whole-body MRI reserved for complex cases due to cost and availability.

CONCLUSION

This case highlights the importance of a structured radiologic approach in diagnosing SMD, Kozlowski type. Key features—platyspondyly, metaphyseal flaring, and corner fractures—must be interpreted in clinical context to avoid misdiagnosis as nonaccidental trauma. Radiologists should advocate for standardized skeletal survey protocols and multidisciplinary collaboration, leveraging recent imaging and genetic advances. Future research should address barriers to genetic testing and the role of advanced imaging in skeletal dysplasias.

Teaching Points

- Recognize platyspondyly, metaphyseal flaring (Figures 2, 5, and 6), and wrist metaphyseal irregularities (Figure 4) as hallmarks of SMD to differentiate it from rickets or mucopolysaccharidoses.
- Evaluate metaphyseal corner fractures with clinical history and skeletal survey findings to distinguish skeletal dysplasia from nonaccidental trauma (Figure 3).
- Use skeletal surveys as the primary imaging modality for suspected skeletal dysplasias, reserving MRI for complex cases.
- Collaborate with geneticists, orthopedists, and pediatricians for holistic care.

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