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X-RAY FINDINGS OF A RARE CASE OF SPONDYLO-EPIPHYSEAL DYSPLASIA TARDA

Radiology

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ABSTRACT

Spondyloepiphyseal dysplasia tarda (SEDT) is a rare skeletal dysplasia that involves abnormal bone development, resulting in disproportionate short stature and various skeletal deformities. This condition typically arises in late childhood or early adulthood and often presents with musculoskeletal symptoms that necessitate further investigation. Patients with SEDT commonly experience issues such as back pain, joint stiffness, and reduced mobility. Radiological assessments reveal specific features associated with the condition, including platyspondyly (flattened vertebrae), shortened long bones, and irregular epiphyseal formation. Given the variability in its presentation, SEDT can be easily underdiagnosed or misdiagnosed, highlighting the need for a thorough understanding of both clinical and radiological characteristics for timely diagnosis and effective management. The genetic factors associated with SEDT are usually linked to an X-linked recessive inheritance pattern, which provides important insights for affected families and emphasizes the importance of genetic counseling. Enhancing awareness of this condition among healthcare providers, especially radiologists, is crucial for ensuring prompt recognition and appropriate treatment, thereby improving patient outcomes and overall quality of life.

KEYWORDS

epiphyseal irregularities, platyspondyly, short stature, skeletal dysplasia, sedt

INTRODUCTION

Spondylo-epiphyseal dysplasia tarda (SEDT) is an inherited disorder causing short stature. It occurs only in males with radiological findings seen at adolescence or adult age. The diagnosis can be made on plain radiographs [1].

Case Presentation

An 18-year-old male presented to the department with bilateral hip pain and back pain. Patient was normal until 16 years of age after which he started developing curvature of spine and gradually increasing bilateral hip pain which had become severe in last 6 months. The patient and his parents were aware about his short stature. There was no history of consanguinity.The patient stated that he had 2 siblings, one brother and one sister, and they were of normal height.

Physical examination showed 18 year old male who had a shortened neck, shortened trunk with increased curvature of spine and shortened proximal extremities. His height was 5 feet, 2 inches. The limitation of motion was found on examination of bilateral hip. On muscle biopsy, no muscle wasting was noted.

X-Ray:

- On plain radiograph (lateral view), the DL spine showed kyphosis with mild platyspondyly and prominent sternum.
- A radiograph of the lumbar spine (AP view) showed flattened vertebral bodies and hump like dense bone centrally placed on the superior and inferior plates.
- A radiograph of the pelvis showed squaring of iliac bones and flattening of the femoral head.
- A radiograph of the elbow joint showed bilateral mal-alignment proximal radioulnar joint and left fused radioulnar joint at an early age.

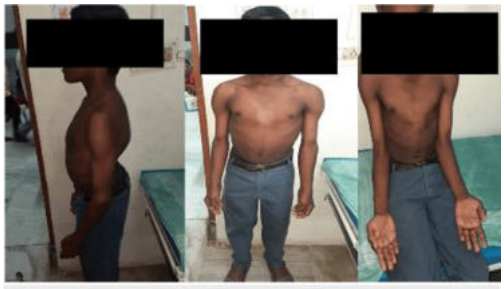

FIGURE 1: External appearance of the patient showing increased curvature of spine, shortened neck and trunk and shortened proximal limbs with normal hands

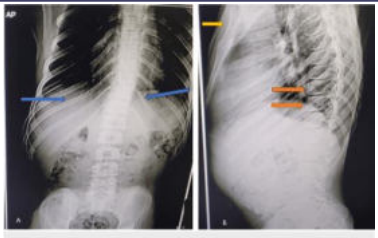

FIGURE 2: A) X-ray lumbar spine (AP view) showing long slender ribs (blue arrow) on both sides of thorax. B)Radiograph of DL spine (lateral view) showing Platyspondyly (orange arrows) and prominent sternum (yellow arrow).

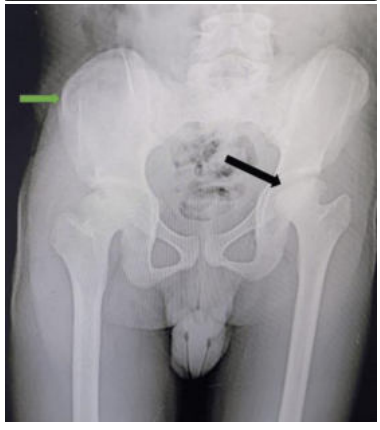

Figure 3: A radiograph of the pelvis showing squaring of iliac bones (green arrow) and flattening of the femoral head (black arrow).

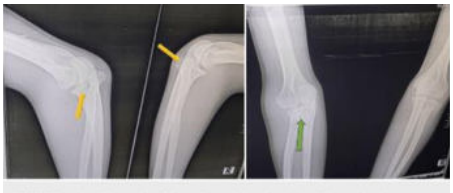

FIGURE 4: A radiograph of the elbow joint showing bilateral mal-alignment proximal radioulnar joint (yellow arrow) and left fused radioulnar joint (green arrow) at an early age.

DISCUSSION

Spondylo-epihyseal dysplasia tarda is a hereditary chondroplasia

which is X-linked recessive trait. It appears in adolescence or adult life as deformities of epiphysis of long bones, flattening of articular surfaces of small and large joints causing joint restriction and flattening of vertebral bodies causing short stature. Spondylo-epiphyseal dysplasia tarda can be diagnosed on plain radiograph itself. The incidence of Spondylo-epiphyseal dysplasia is estimated to occur in approximately 1 in 100,000 live births.

The present case had classical features like shortened neck, shortened trunk with increased curvature of spine and flattened vertebral bodies (Platyspondyly), flattening of articular surfaces of small and large joints, Squaring of iliac bones with flattened femoral head and waddling gait with normal IQ and no muscle pathology. Thus leading to a diagnosis of SEDT.

CONCLUSIONS

Patients with SEDT are males and are affected with a X-linked recessive transmission. No muscle pathology or metabolic defects have been demonstrated on muscle biopsy. Only bony defects occur which are seen in the form of short neck, short stature and short extremities. The radiographic appearance of the lumbar vertebral bodies in the adult is characteristic of the condition. The patients have normal IQ and can live a normal life. Physiotherapy is advised in most cases. The families of such patients should be given moral support and counselling for accepting their kin and relatives with such conditions.

Additional Information

Disclosures

Human subjects: Consent was obtained or waived by all participants in this study.
Conflicts of interest: In compliance with the ICMJE uniform disclosure form, all authors declare the following:
Payment/services info: All authors have declared that no financial support was received from any organization for the submitted work.
Financial relationships: All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work.
Other relationships: All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

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1. Leonard O. Langer, Jr.: Spondyloepiphysial Dysplasia Tarda. RSNA. 1964, https://doi.org/10.1148/82.5.833