

SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA: REPORT OF A CASE AND REVIEW OF THE LITERATURE

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ABSTRACT

Spondyloepiphyseal dysplasia congenita (SEDC) is a disorder of type II collagen synthesis that primarily affects the spine and proximal epiphyseal centers. The abnormalities are present at birth and may include short stature, flattened facies, kyphoscoliosis, lumbar hyperlordosis, coxa vara and genu valgum. The defects may complicate into gait abnormality, early degenerative changes, joint fusion, osteopenia and neurological compromise. Early diagnosis of SEDC may prevent unnecessary diagnostic testing for other causes of short stature and/or osteoarthritis and guide towards timely protective measures. We report here, a 4-years-old child, who presented with SEDC and was treated with analgesics and counselling of parents for prognosis, precautions, potential complications, treatment options for the future and inheritance of the disease.

Key Words: Spondyloepiphyseal Dysplasia, Scoliosis, Short stature

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INTRODUCTION

The spondyloepiphyseal dysplasia (SED) is a type of skeletal dysplasia that mainly involves the spine and proximal epiphyseal centers¹. SED may present as the congenita variant or the tarda variant, with considerable genetic heterogeneity². In the congenita variant, there is abnormal synthesis of type-II collagen and abnormalities are present at birth and may include short trunk, dwarfism with short limbs (proximal and middle), disproportionately larger hands and feet and flat face; while in the tarda variant, there is normal appearance at birth and short trunk becomes apparent around five to six years of age^{1,2}. In distinction to the congenita variant, the face and extremities are normal in the tarda variant². Early diagnosis of SED may prevent unnecessary diagnostic testing for other causes of short stature and/or osteoarthritis. We present here, report of a case of SED congenita (SEDC) who presented with signs and symptoms of short stature, spinal deformity and leg pains.

CASE REPORT

A four-years-old child presented with progressive back deformity and pain in legs. He had normal hearing and vision. There was no history of such complaints in his family. On examination, he had mild bowing of legs, marked lumbar lordosis, thoracic scoliosis, pectus carinatum and flat feet (Figure 1). His height was between

3rd and 15th centile according to the world health organization height-for-age chart for boys. He had a waddling gait. The oral and ocular examination was normal and there was no joint laxity.

The radiographs of the spine and pelvis demonstrated flattening of dorsal vertebral bodies (platyspondyly), thoracolumbar scoliosis, spina bifida at LV5 and SV1 levels and dysplastic bilateral proximal femoral epiphysis and metaphysis (Figure 2). The radiographs of the wrist and forearm revealed dysplastic bilateral radial epiphysis, dysplastic bilateral distal radial and ulnar metaphysis; and inconspicuous ossification centers of carpal and metacarpal bones. Acetabulae showed mild dysplasia more on the left side. The ilium, ischium and pubis were, however, unremarkable (Figure 3). The ultrasonography of the abdomen did not show hepatomegaly, splenomegaly or renal malformation.

All essential laboratory investigations for the child were carried out. The complete blood count, thyroid function tests, serum calcium, phosphate and alkaline phosphatase levels were within normal limits. Based on clinical evaluation and radiological investigations, he was diagnosed to have SEDC. The child was advised oral paracetamol (120 mg/5ml) 10 ml twice daily. His parents were counselled about the prognosis, potential complications, treatment options for the future, and inheritance of the disease. They were warned

Figure 1: Pectus carinatum, short neck, increased lumbar lordosis and scoliosis



Figure 2: X-rays of the dorsal-lumbar-sacral spine demonstrating dorsal vertebral platyspondyly, thoraco-lumbar scoliosis, spina bifida at LV5 & SV1 levels and dysplastic bilateral proximal femoral epiphysis & metaphysis. Acetabulae showing mild dysplasia more on the left side

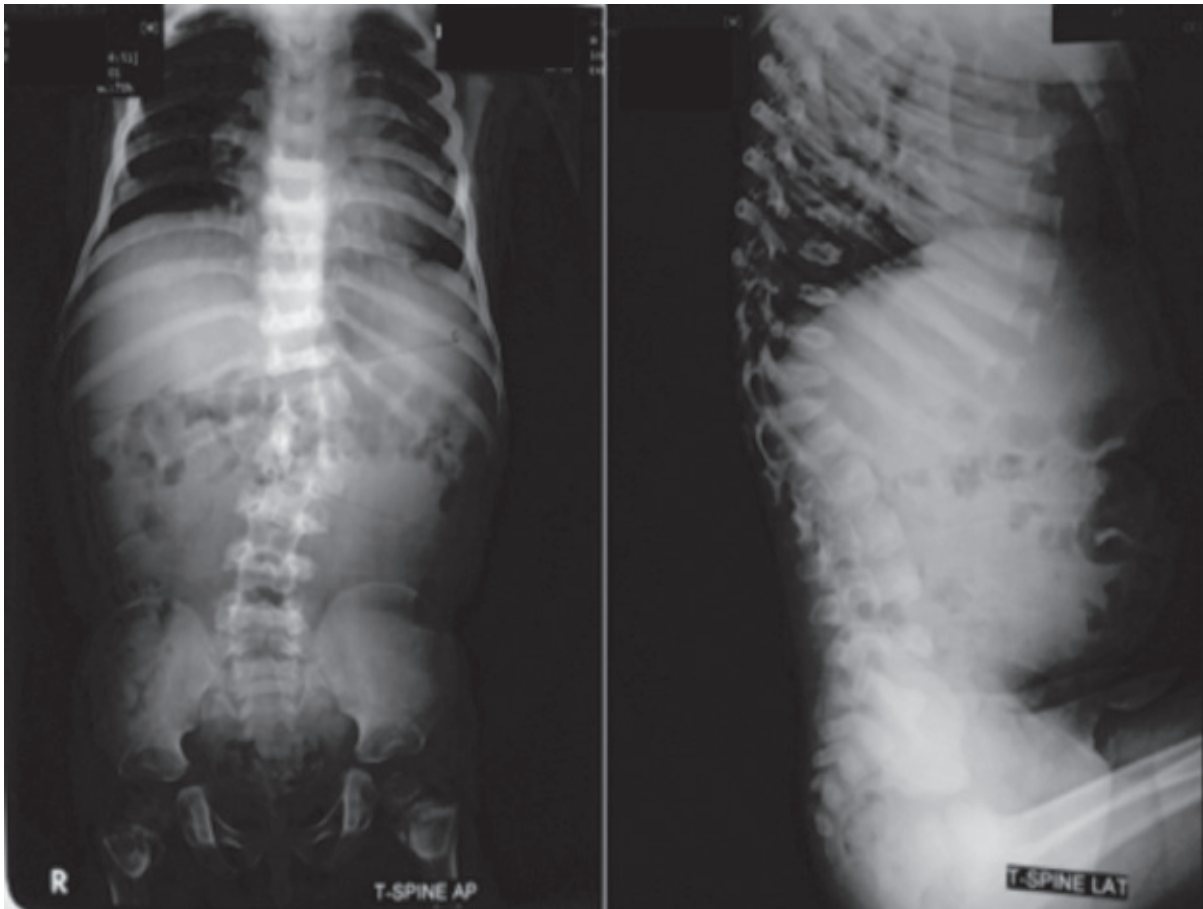
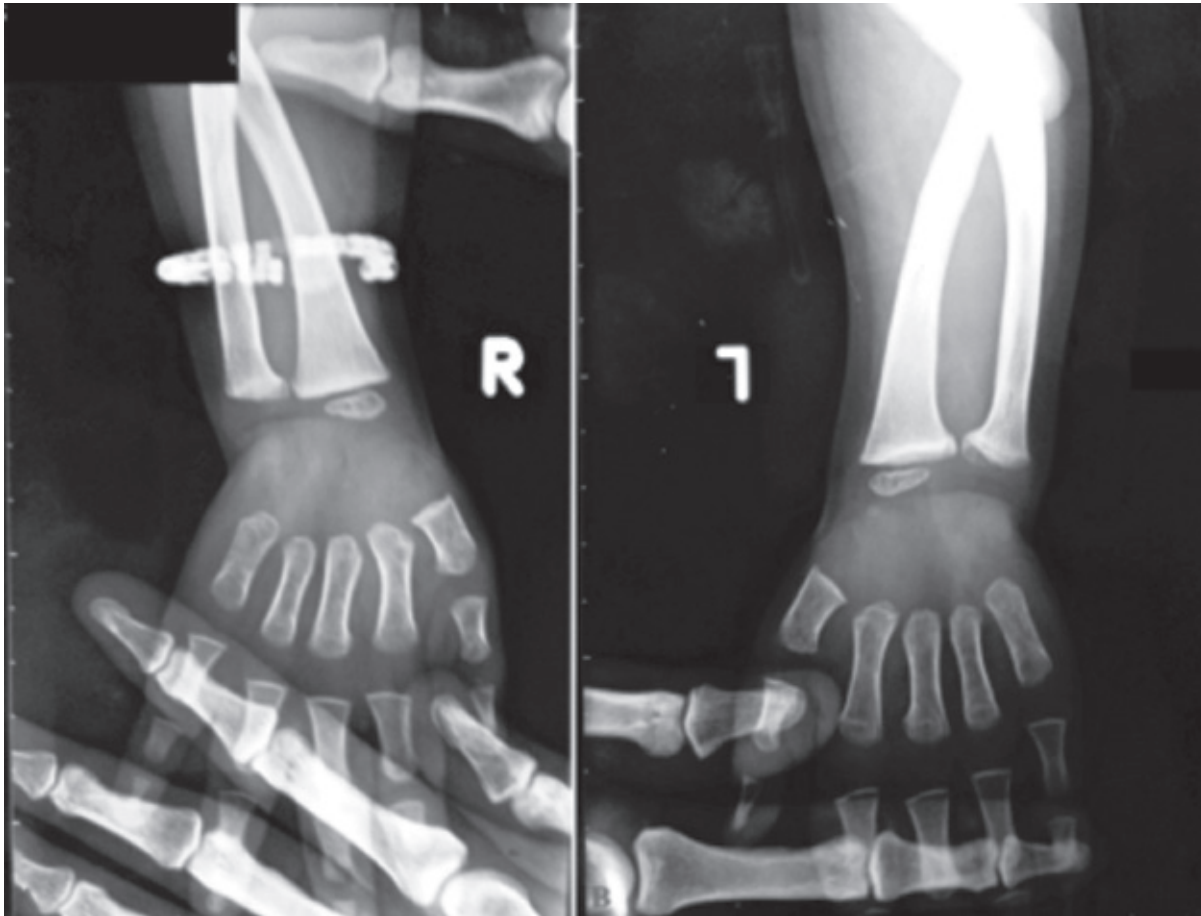


Figure 3: X-rays of both wrists demonstrating dysplastic bilateral distal radial and ulnar metaphysis & radial epiphysis and inconspicuous ossification centers of carpal and metacarpal bones



against activities posing undue stress on the spine and weight-bearing joints. An annual follow-up for assessment of joint pain and scoliosis was also advised.

DISCUSSION

The term SED is used to embrace a group of conditions characterized by platyspondyly and dysplasia of other bones. The degree of spinal and tubular bone involvement and the amount of dwarfism vary between the different groups³. SEDC is a rare disorder of bone growth that results in dwarfism, characteristic skeletal abnormalities and occasionally, problems with vision and hearing. The name of the condition indicates that it affects bones of the spine (spondylo-) and the ends of bones (epiphyses), and that it is present from birth (congenital). SEDC is autosomal dominant or sporadic (most common) and is a subtype of collagenopathy, types II and XI^{2,3}. Features of SEDC include disproportionate dwarfism with spine and hips more involved than extremities, waddling gait and muscular weakness, flat facies, short neck, deafness and cleft palate. Axial skele-

ton shows ovoid vertebral bodies, severe platyspondyly, hypoplasia of odontoid process (which results in atlantoaxial instability that can cause cervical myelopathy), progressive kyphoscoliosis (short trunk) involving thoracic and lumbar spine, narrowing of disk spaces (resulting in short trunk), broad iliac bases, deficient ossification of pubis and flat acetabular roof. The chest shows bell-shaped thorax and pectus carinatum. Extremities appear slightly shortened, with severe coxa vara, genu valgum, multiple accessory epiphyses in hands and feet, and talipes equinovarus. Retinal detachment and myopia (50%) are common associations. Secondary arthritis in weight-bearing joints is a disabling complication⁴.

We searched similar cases of SEDC with physical and radiological description available freely on the internet and identified twenty-four cases (Table 1)⁵⁻¹⁷. There were 10 (41.7%) males and 14 (58.3%) females with a mean age of 19 ± 18 years. The family history was positive in 6 (37.5%) and negative in 10 (62.5%) cases while in eight cases no relevant description was available. All had a short stature. Disproportionately small trunk with rel-

atively larger hands and feet were identified in fifteen (62.5%) cases. Spinal hyperlordosis and/or scoliosis was described in 23 (95.8%) individuals. Four (16.7%) individuals had conductive deafness, four (16.7%) had myopia, two (8.3%) had cleft palate and renal impairment each and one (4.2%) had retinal detachment, cataract and anisohyperopia each. Facial abnormalities (microcephaly, mid facial hypoplasia, flat face and hypertelorism) were spotted in five (20.8%) cases. Knee (genu valgum and knock knees) and chest (broad and short thorax and pectus carinatum) abnormalities were found also in five (20.8%) cases each. One individual had pes planus and one had pes cavus. Finger and phalangeal abnormalities (clinodactyly, bipartite phalanges and phalangeal hypoplasia) were noticed in four cases. Gait abnormalities were observed in 14 (58.3%) individuals. One individual developed tetraparesis and one died of renal failure.

On radiological evaluation, platyspondyly was detected in twenty (83.3%) individuals. Pelvic abnormalities included hypoplasia and delayed ossification of ilium, ischium & pubis and sloping acetabular roofs. Hypoplastic and deformed femoral heads, coxa vara, short and wide femoral necks and short long bones were found in radiographs of pelvis and long bones in most individuals. Our case had a short stature, lumbar hyperlordosis, thoracic scoliosis, pectus carinatum and pes planus. The radiographs detected platyspondyly, thoracolumbar scoliosis, dysplastic femoral epiphysis and metaphysis, mild acetabular dysplasia, and dysplastic radial epiphysis and radial & ulnar metaphysis. The ilium, ischium and pubis were, however, unremarkable.

SEDC needs to be differentiated from achondroplasia, hypothyroidism; and other dysplasias with universal platyspondyly in young children including metatropic dysplasia, thanatophoric dysplasia, achondrogenesis and hypophosphatasia. Typical findings of achondroplasia are short rectangular vertebral bodies with relatively wide disk spaces and narrowing of the distal lumbar spinal canal. In hypothyroidism, marked vertebral body flattening with a beak-like projection from the anterior inferior aspect of lumbar vertebrae are present in neonates and there is also a delay in skeletal age².

The goals of treatment in SEDC are avoidance of activities/postures that may compromise atlantoaxial stability (e.g. extreme neck flexion and extension) or place undue stress on the spine and weight bearing joints. Annual follow-up for assessment of joint pain and scoliosis and radiological evaluation of the cervical spine prior to general anesthesia for clinically significant odontoid hypoplasia may be the appropriate choice. Standard pain management protocols as with other musculoskeletal problems such as physical modalities, exercises, analgesics, injection procedures, etc. may be followed for joint pains. In advanced stages, joint replacement for

hip, knee and shoulder and surgical correction of scoliosis or kyphosis may be considered. Genetic counseling of the family for the inheritance pattern of the disease must be carried out.

CONCLUSION

Our case of SEDC presented with short stature, spinal deformity and leg pains and was found to have platyspondyly, kyphoscoliosis, pectus carinatum, pes planus and delayed epiphyseal ossification. He was managed with analgesics and parental counselling about prognosis, potential complications, precautionary measures, future treatment options and disease inheritance.

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