Case Report

Total hip replacement for a case of spondyloepiphyseal dysplasia tarda with premature hip arthritis: two case reports

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ABSTRACT

Spondyloepiphyseal dysplasia (SED) tarda is an extremely uncommon form of skeletal dysplasia, characterized by congenital dwarfism with a short trunk and epiphyseal dysplasia in the long bones and vertebral bodies. It is an X-linked recessive condition not evident at birth, the usual age of presentation is after the first decade of life. These patients commonly present with short trunk and arm spans significantly greater than height and premature hip arthritis. In this article, we present two short-statured male patients of age 28 years with early hip arthritis diagnosed to be SED Tarda with clinical examination and radiological findings, who underwent primary hybrid total hip replacement of dysplastic hip with excellent post-operative functional outcome.

Key words: Short stature, Premature osteoarthritis, Platyspondyly, Case series, Total hip replacement

Spondyloepiphyseal dysplasia tarda (SEDL) is an X-linked primary skeletal dysplasia that predominantly affects the spinal vertebral bodies and epiphyses during skeletal growth [1]. The condition is not evident at birth, and the usual age of presentation is after the first decade of life. It is a rare condition with a prevalence of 1 in 2 lakh live births [2]. Disproportionate (short-trunked) short stature in a male, with or without back pain, is the common presenting feature. Other characteristic features include a broad chest with sternal protrusion and limitation of joint motion at the hip and elbow. The radiographic manifestations of SEDL are diagnostic, clearly distinguishing it from other dysplastic conditions. It includes generalized platyspondyly, narrowing of intervertebral disc spaces, and pathognomonic superior and inferior "humps" involving the posterior two-thirds of the flattened vertebral bodies. The major potential medical complication of the disorder is premature arthritis, predominantly affecting the spine and hip joints. Hip joint disease may be severe, necessitating replacement in early adult life. Preoperative planning is crucial in these cases, demanding custom-made implants to accommodate narrow femoral canals. Early diagnosis of the condition facilitates genetic testing and counselling, thus reducing the incidence of the disease.

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CASE SERIES

Case 1

A 28-year-old man, a driver by occupation, presented with a complaint of left hip pain for 7 months associated with limping. He had no trauma or any other history suggestive of inflammatory pathology. He was short statured, which he noticed first at the age of 13 years in school. He had an elder brother who was also short-statured and an elder sister of normal height.

The patient was conscious, afebrile, and oriented to time, place, and person. The patient was moderately built and well nourished, 140 cm in height, with an UL: LL segment ratio of 1.2, indicating disproportionate dwarfism with a short trunk and barrel-shaped chest. Hip examination showed adduction deformity of 10° with restricted internal rotation on the left side. There was a true shortening of 1 cm confined to the supratrochanteric region of the femur, confirmed by the Bryant's triangle. The patient also had barrel shaped chest with pectus carinatum and reduced chest expansion (1 cm) (Fig. 1a). Radiographic examination revealed multiple epiphyseal abnormalities, platyspondyly with characteristic superior and inferior "humping" seen on the lateral view, with Hip X-ray showing evidence of premature osteoarthritis (Fig. 1b).

We planned for a total hip replacement on the left side and periodic observation of the right hip for the development of

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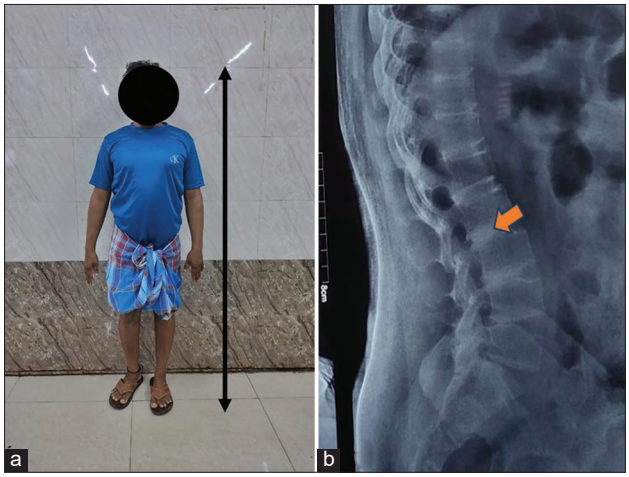

Figure 1: (a) Image shows the short stature of the patient (case 1); (b) X-ray lateral view of lumbar spine (case 1) showing platyspondyly with superior and inferior bony "humps" affecting the middle and posterior thirds of the vertebrae

arthritic changes. Pre-operative evaluation with a computed tomography (CT) scan showed an acetabular cup size of 40 mm (Fig. 2). Under spinal anesthesia, the patient was placed in the right lateral position, and through a lateral (Hardinge) approach, a skin incision was made 5 cm proximal and 8 cm distal to greater trochanter. The gluteus maximus and tensor fascia lata were cut, and the trochanteric bursa was incised. A conjoint flap of gluteus medius and vastus lateralis was elevated, and capsulotomy was done. *In situ*, a neck cut was made 1 cm proximal to the lesser trochanter, and the head was extracted out with a corkscrew. Serial acetabular reaming done and 40 mm size cup fixed with 6.5 mm screws with liner lip covering the posterosuperior aspect. The femoral canal was found to be narrow, and we encountered difficulty in broaching the femur. Prophylactic cerclage was applied with stainless steel (SS) wire (Fig. 3). After cementation, a femoral stem size of 00 (MERIL) with a head size of 28 mm was fixed. The head reduced back into the acetabulum, and stability was checked in all planes. The wound closed with a functioning drain, and the patient was placed in limb abduction. The post-operative protocol involved hip abduction and quadriceps strengthening exercises, and toe touch weight bearing with a walker was started immediately. Partial and full weight bearing was advised after 3 weeks and 6 weeks, respectively (Fig. 2d).

Case 2

A 28-year-old man, a daily wage worker by occupation, presented with right hip pain for 6 months associated with limping. He had no trauma or any other history suggestive of inflammatory or infective pathology. On examination, he was conscious, afebrile, and well-oriented. No signs of nutritional deficiency. Short statured (145 cm) with UL: LL segment ratio of 1.1, indicating disproportionate dwarfism with a short trunk. Hip examination showed flexion up to 45° (further movements restricted) and adduction deformity of 15° on the right side with restricted internal rotation on both sides (Fig. 4a). There was no true shortening. Radiographic examination

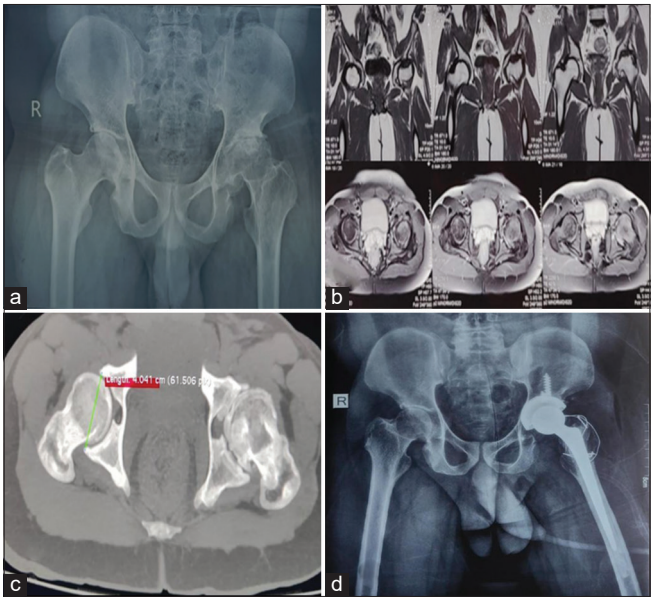

Figure 2: (a) Loss of femoral sphericity with reduced superolateral joint space, sclerotic changes in acetabulum and broken shentons line (left side); (b) Magnetic resonance imaging of hip showing cortical irregularity with subchondral collapse at superolateral aspect of left femoral head; (c) computed tomography axial cut of bilateral hip (case 1 showing acetabular cup size of 40.4 mm on right side (unaffected); (d) Post-operative X-ray anterior-posterior view of bilateral hip showing acetabular cup with screws and cemented femoral stem with stainless steel wire cerclage (Case 1)

revealed platyspondyly with characteristic superior and inferior "humping" seen on the lateral view, with hip X-ray showing evidence of premature osteoarthritis (Fig. 4b).

He underwent primary total hip replacement on the right side through a lateral approach, as previous case. Pre-operative evaluation with a CT scan showed an acetabular cup size of 48 mm (Fig. 5a and b). Intraoperatively, a 44 mm acetabular cup was found to be stable, and we proceeded with that. A cemented femoral stem of size 7 was used (the smallest uncemented stem available was 7 size, which was bigger for his very narrow femoral canal. The post-operative protocol was the same as that followed for the previous case (Fig. 5c).

DISCUSSION

Spondyloepiphyseal dysplasia (SED) is a general term that describes the radiographic abnormalities seen in several skeletal dysplasias, including pseudoachondroplasia. The "congenita" form can be evident at birth, whereas the "tarda" form is usually seen in early adulthood. SEDL commonly refers to the X-linked recessive form of the disorder, although rare autosomal dominant and autosomal recessive "tarda" forms have been described. It is not population specific as cases had been reported from Korea [3], China, Japan [4,5], and other parts of the world. A pedigree analysis done on a Chinese family showed 6 affected males in the past 5 generations, confirming the X-linked inheritance of the condition [6].

Craniofacial appearance, vision, hearing, and intelligence are unaffected in SEDL, and there are no consistently associated extraskeletal anomalies. Barrel shaped chest due to a short trunk

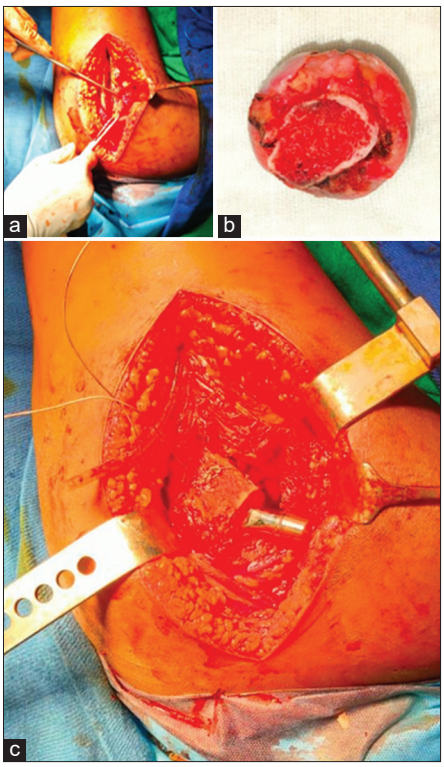

Figure 3: Intraoperative image showing (a) Conjoint flap of gluteus medius and vastus lateralis (Case 1); (b) Extracted femur head with arthritic changes, (c) Femoral stem with SS wire cerclage at lesser trochanter level

Figure 4: (a) Comparison of the stature of the patient was done by standing him beside a normal mal; (b) X-ray Lateral view of Lumbar spine showing platyspondyly with superior and inferior bony "humps" (case 2)

has been reported [7]. The radiographic manifestations of SEDL include generalised platyspondyly, narrowing of intervertebral disc spaces, and pathognomonic superior and inferior "humps" involving the posterior two-thirds of the flattened vertebral bodies. These changes are best appreciated in late childhood and adolescence and may become superimposed by secondary arthritic changes in later decades. The condition can be differentiated from other forms of SED by their age of onset (SED Congenita). Morquio syndrome is often confused with SEDT The differences between them are shown in Table 1.

Table 1: Difference between Morquio syndrome and SEDT

Features	SEDL	Morquio
Mental retardation	-	+
Clouding of the cornea	-	+
Vertebral body	Platyspondyly with Heeped up vertebra	Central beaking
Pelvis	Iliac flaring	Iliac flaring
Hip	Coxa vara and premature arthritis	Coxa valga

SEDL: Spondyloepiphyseal dysplasia tarda

Trafficking protein complex subunit 2 (*TRAPPC2*) gene, which encodes a protein named SEDLIN is the causal gene for this disease. The protein is involved in intracellular transport mechanisms [8]. SEDLIN [4] binds to other components of the TRAPP complex and plays a critical role in the traffic of vesicles between the endoplasmic reticulum and the Golgi apparatus [9]. When performed, molecular genetic testing of all mothers of affected sons determined that regardless of family history, all were carriers of a pathogenic variant in *TRAPPC2*. Carrier females are at a 50% risk of transmitting the pathogenic variant in each pregnancy: males who inherit the pathogenic variant will be affected; females who inherit the pathogenic variant will be carriers and will not be affected. Carrier testing of at-risk female relatives and prenatal testing for pregnancies at increased risk are possible if the pathogenic variant in the family has been identified. Reduced expression of the *TRAPPC2* gene both at the mRNA level and protein level, and its impact on the expression of protein COL2A1 was studied by Lou *et al*. [10] in 2023 and highlighted the importance of molecular testing in the diagnosis of this rare disease in clinics.

There is no specific treatment apart from the management of the complications of the condition. The most common of these is the management of hip dysplasia, which may require hip replacement. Advice should be given regarding the prevention of premature arthritis by maintaining a healthy weight for height and regular, low-impact exercise such as swimming and cycling. Data regarding the use of the growth hormone in this condition and its effect on final adult height are not available. Significant short stature is usual in this condition, and appropriate ongoing psychosocial support of the patient and the family is important. Genetic counselling [11] should be provided to discuss X-linked inheritance.

Orthopedic surgery is directed at treating precocious hip arthritis. Valgus osteotomy of the proximal femur with acetabular augmentation when needed has been proposed for younger patients with SEDL, but the influence of osteotomy on the long-term outcome of these hips has not been established. Degenerative arthritis is treated by total hip arthroplasty in early adulthood. Pre-operative templating and custom components may be needed because of the narrow canal width of the femur [12]. Spinal surgeries are rarely required, although odontoid hypoplasia with instability demands stabilization.

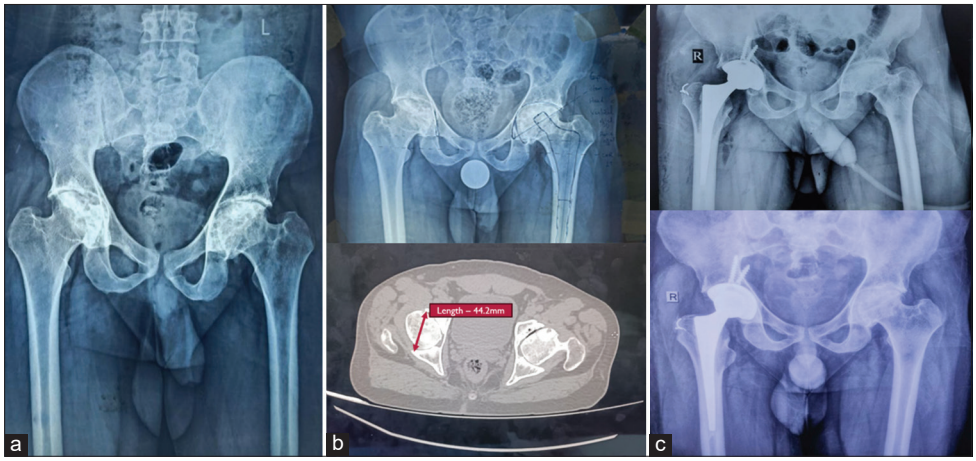

Figure 5: (a) Pre-operative hip X-ray with bilateral hip arthritis; (b) Pre-operative templating (on normal side shows Acetabular cup of size 44mm and stem size 7, which is confirmed with computed tomography scan (on affected side); (c) A Immediate post-operative X-ray B 2-month follow-up X-ray shows good cement bone integration with no signs of osteolysis (Case 2)

CONCLUSION

SEDL is an X-linked recessive genetic disorder causing back pain and early osteoarthritis, which may require replacement surgeries. Good pre-operative planning and custom implants may be required to prevent intraoperative complications and improve functional outcomes. Early diagnosis of the condition helps in prenatal testing and genetic counseling, thus reducing the incidence of the disease.

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