**CASE REPORT**

**Imaging features of a rare case of Scapuloiliac dysostosis (Kosenow**

**syndrome) in an ambulatory child.**

**Introduction**

Scapuloiliac dysostosis, also known as pelvis-shoulder dysplasia and Kosenow syndrome, is a rare skeletal dysplasia described first by Kosenow et al in 1970(1). The main components of the syndrome are varying degrees of bilateral iliac and scapular hypoplasia. Various other skeletal abnormalities including exaggerated lumbar lordosis, clavicle, vertebral, rib and long bone anomalies and extra skeletal abnormalities, most commonly those of eye and ear, have also been reported(2). We describe the plain radiography, CT and MRI findings of an interesting case of the syndrome.

**Case report**

A 4 year old child, the second born of non consanguineous marriage, presented with abnormal gait and had a normal neurological examination except for waddling gait. The elder sibling was normal

Plain radiograph of the pelvis showed complete bilateral absence of ilium with normal well formed ischium, the superior and inferior pubic rami as well as the femoral heads bilaterally. Frontal chest radiograph also revealed severe hypoplasia of the body of scapulae bilaterally while the acromion, coracoid and glenoid were well formed. The shoulder joint was normally aligned.

CT scan of the pelvis confirmed the radiographic findings. Band like soft tissue density areas were seen in the normal location of the ilium and the femoral heads were seen lying within the soft tissues. Sacral spina bifida was also noted.

MRI was performed which showed absence of osseous or cartilaginous tissue in the normal location of ilium. Instead there was a soft tissue structure, hypointense in all sequences suggestive of fibrous tissue, which along with the surrounding skeletal muscles seemed to be supporting the well formed femoral heads.

**Discussion**

Scapuloiliac dysostosis (pelvis-shoulder dysplasia, Kosenow syndrome) is an uncommon hereditary skeletal dysplasia. It is classified as autosomal dominant due to one of the original families, though both dominant and recessive inheritances are described (3). The chief components of the syndrome are varying degrees of bilateral hypoplasia of the ilium and scapulae, usually severe. The sacrum, ischium and pubis are normal while the acetabulum is markedly dysplastic with severe hip instability. The acromion and coracoid process of the scapula are usually preserved despite severe hypoplasia of the body. Milder phenotypical variants may have involvement limited to the pelvis. Several other skeletal and extra skeletal anomalies have also been described. These include hypoplastic or abnormally elongated clavicles, lumbar hyperlordosis, rounded appearance of the vertebral bodies, rib anomalies and overconstriction of the femora and tibiae (2, 3).The common soft tissue associations involve the eye and ear such as microophthalmia, coloboma of the lids, low set ears and deafness.(4).

A unique feature of this case is the well formed femoral heads and ambulation with a fully functional, though abnormal gait, despite the complete bilateral iliac agenesis. To the authors’ knowledge, only one case of this syndrome with complete iliac agenesis and complete ambulation has been reported where as the others had severe hip instability and were bedridden (5). The support for the femoral head, in the absence of ilium, seems to be provided by the soft tissues such as the surrounding muscles and the fibrous tissue replacing the ilium, as shown by MRI. This seems to be the first report of the MRI findings of this syndrome.

This case further supports the theory that, the dysplastic changes in the femoral head in congenital dislocation of hip is a result of its abnormal contact with the ilium or false acetabulum and that the femoral head will grow normally even if dislocated, in the absence of this abnormal contact(5).

This case report is unique in terms of the rarity as well as the complete multi modality radiological evaluation of the syndrome. In addition, the surprising degree of ambulation and well formed femoral heads despite complete iliac agenesis provides insights into the development of hip and femoral head.

**References**

1. Kosenow W, Niederle J, Sinios A. 1970. Becken-schulter dysplasie. Fortschr Geb Rontgenstr Nuklearmed 113:36–48.
2. Taybi H, Lachman R.1996. Radiology of syndromes, metabolic disorders, and skeletal dysplasias, 4th ed. St. Louis: Mosby-Year Book Inc. p 444, 773, 789
3. Elliot AM, Roeder ER, Witt DR, et al. Scapuloiliac dysostosis (Kosenow syndrome, pelvis-shoulder dysplasia) spectrum: Three additional cases. Am JMed Genet 2000;95:496–506.
4. Blane CE, Holt JF, Vine AK. Scapuloiliac dysostosis.Br J Radiol 1984;57: 526–8.
5. Mac-Thiong JM, Leduc S and Labelle H. Complete bilateral agenesis of the ilium in a 7-year-old ambulatory girl.Spine 2005;30:E420–E423

**Legend for images** :

**Figure 1** – Frontal radiograph of Chest and Abdomen shows bilateral absence of ilium and severe hypoplasia of the body of scapulae. The coracoid and acromion are normal bilaterally.

**Figure 2** – Axial CT section of pelvis in soft tissue window settings show bilateral absence of bony ilia replaced by band shaped soft tissue density structures.

**Figure 3** – Axial CT in bone window showing the well formed femoral heads, supported by soft tissues. The ischium and pubis are normal. Sacral spina bifida is also seen.

**Figure 4** - MRI of pelvis, axial T1 image show bilateral iliac agenesis with hypointense band like fibrous tissue replacing ilia.

**Figure 5** – Coronal T1 pelvic MRI reveals iliac agenesis with the femoral heads normal and supported by surrounding muscles and fibrous tissue.