Original Article

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Spondyloepiphyseal Dysplasia: A Rare Entity

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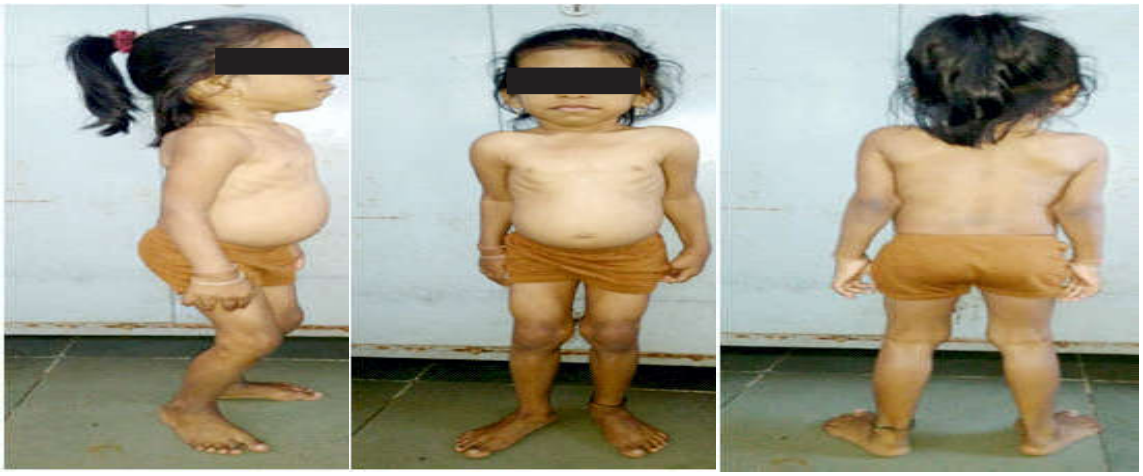
Abstract

Spondyloepiphyseal dysplasia congenita is an autosomal dominant inherentdisorder characterized with abnormal growth of epiphyseal center,spine and disproportionate dwarfism. Here we report a child presents with short stature evaluated for short stature and abnormal gait turned out as skeletal dysplasia.

Keywords: Spondyloepiphyseal Dysplasia; Skeletal dysplasia.

Case Summary

A 4 year old female child brought with complaints of short stature, developmental delay, difficulty in walking and weakness of upper limb since age of 1.5 years. There was difficulty in walking in form of externally rotated limb, abnormal gait, difficulty in squatting position and sitting with folding limbs. Child had upper limb weakness in the form of raising hands above shoulder level with difficulty in shoulder joint rotation. There was no history of repeated respiratory tract infection or any other chronic medical illness. On birth history child was full term cesarean section delivery in view of oligohydroamnios with low birth weight, there was no history of any other perinatal event. On general examination child has height/ age of 2 years, weight /age of 2 years, Ht was less than 3rd centile with ratio of US/LS was 0.9. Child had dysmorphic features like microcephaly, short neck with mid facial hypoplasia. There was no



Fg. 1:

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associated winging of scapula, had swelling at multiple joints. The hands and feet were normal in length.Bilateral shoulder abduction was 60%, with restricted internal and external rotation with restricted bilateral hip abduction, right lower limb showed coxa vara deformity. Lumbar lordosis was present;there were no features of rickets or widening of carrying angle. On initial investigations Ca/Po4, VitD3, ALP level were normal. X-ray pelvis showed bilateral coxa vara. On evaluation ofX ray long bones suggestive of dysplastic femur neck with shortening and widening of humerus with incomplete cervical rib with generalized osteopenia, Suggestive of spondyloepiphyseal dysplasia. Bone age was normal with normal karyotype and thyroid function test. EMG/NCV studies were normal.

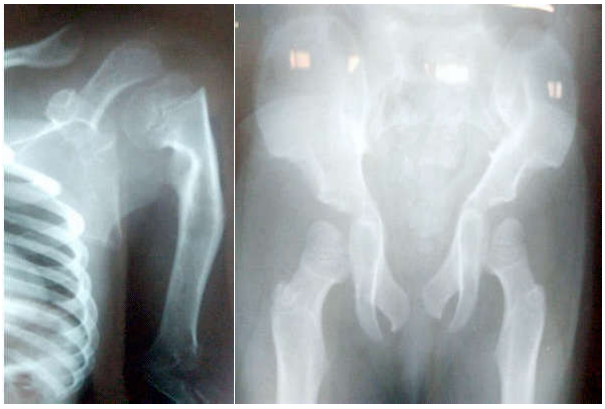

Fig. 2 a: Shortening of epiphysis with widening of metaphysis
b: Increase neck shaft angle s/o coxa vara deformity

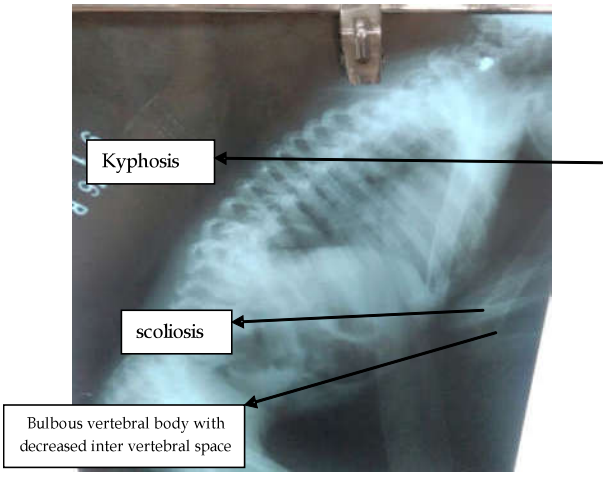

Fig. 3:

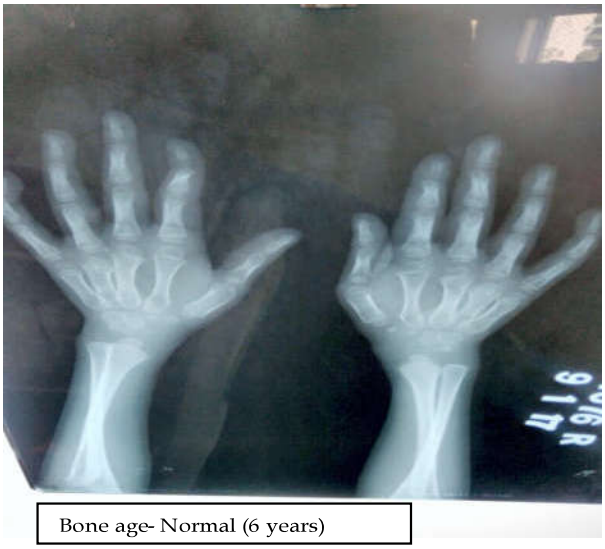

Fig. 4:

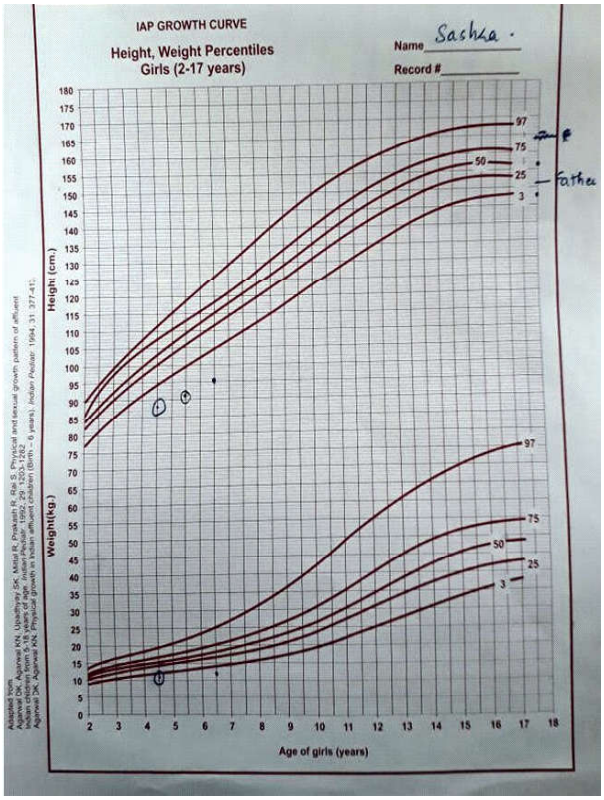

Fig. 5: Fathers ht: 165cm,Mothers ht: 160cm, MPH: 156 cm

- Pathological short stature
- Short stature since beginning
- Growth velocity: 4 cm/year

Discussion

Spondyloepiphyseal dysplasia is a bone growth disorder that results in short stature, skeletal deformity and problems with hearing and vision. This condition affects bones of the spine and ends of long bones. Thesechildren have short trunk and neck and shortened limbs. They have hands and feet of average sized adult height of 3-4 feet, when they grow adult.They also have spine deformity with platyspondyly, coxa varaand club foot deformity. Arthritis develops earlier. There are two major types

SED congenital and SED tarda. The clinical and radiographic differences among the various spondylodysplasias are frequently age related. Gait problems are often attributed to hip and knee deformities. Mortality occurs because of cervical myelopathy or cord compression. The gene for SED congenital has been mapped to the long arm of chromosome 12. Most cases results from random mutations.

Conclusion

Spondyloepiphyseal dysplasia congenita is a rare entity. A detail skeletal survey and endocrine evaluation needs in a case of short stature. Early detection of SED congenital is necessary as growth can be achieved by implementation of growth hormone therapy.

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Conflict of Interest: None

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