Picture story

Spondylo-metaphyseal dysplasia in a 2-year-old Sri Lankan girl

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Introduction

Spondylo-metaphyseal dysplasia is a heterogenous group of rare genetic disorders characterized by malformations of the axial and extra-axial skeleton resulting in vertebral and metaphyseal abnormalities[1]. The reported prevalence is less than one in one million worldwide[2]. The disease-causing mutations are mapped to the collagen type II alpha 1 chain (*COL2A1*) gene and transient receptor potential cation channel subfamily V member 4 (*TRPV4*) gene. However, in many patients, the mutations are not identified[3,4]. Due to the subtleness of the clinical features, the diagnosis of spondylo-metaphyseal dysplasia is only made during childhood when clinical features are apparent[5]. We report a 2-year-old girl with spondylo-metaphyseal dysplasia who had short stature since birth.

Case report

A two-year and seven-month-old Sri Lankan girl has been followed up for extreme short stature since birth. She was born at term to healthy non-consanguineous parents with a birth weight of 1990g (less than -3SD). Antenatal ultrasonography at 28 weeks of gestation revealed a short femur length; however, the antenatal period was uncomplicated. Her length at birth was 40cm (less than -3SD) and the head circumference at birth was 33cm (median to -1SD). She has been short throughout; however, her motor and cognitive development have been age-appropriate. There was no family history of short stature, skeletal or extra-skeletal abnormalities or abnormal body posture in parents or relatives. She was the only child in the family. On examination, her height was 59cm (below -3SD) (Figure 1).

Father's and mother's heights were 166cm and 146cm respectively. The child's height was well below the mid-parental height range, and the upper-to-lower segment ratio was 1.6:1. Her neck was short, and the chest was visibly small with pectus carinatum deformity (Figure 2). Limb examination showed bowlegs with varus deformity and short stubby hands and feet bilaterally.

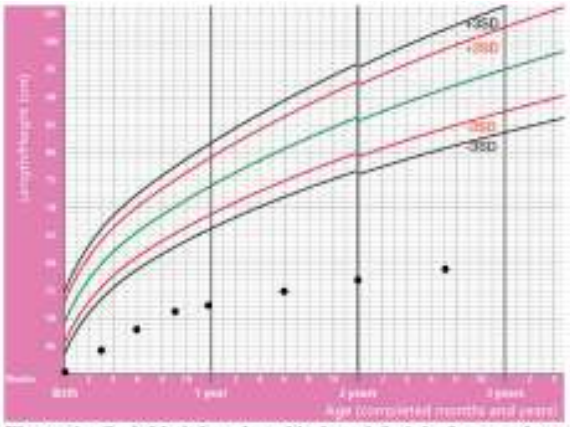

Figure 1: ***Serial heights plotted in length/height for age chart***

Figure 2: ***showing extreme short stature, bowlegs and pectus carinatum***

She did not have kyphoscoliosis, organomegaly, corneal clouding or joint contractures. Her cardiovascular, abdominal and nervous systems were clinically normal. Haematological and biochemical investigations, including

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full blood count, electrolytes, renal and liver function tests, calcium, inorganic phosphate and alkaline phosphatase were normal. The skeletal survey revealed generalized platyspondyly involving vertebral bodies (Figure 3), metaphyseal changes of long bones (Figure 4 and Figure 5) and short square iliac wings. The bone age was between 6-12 months and the urinary mucopolysaccharide screen was negative.

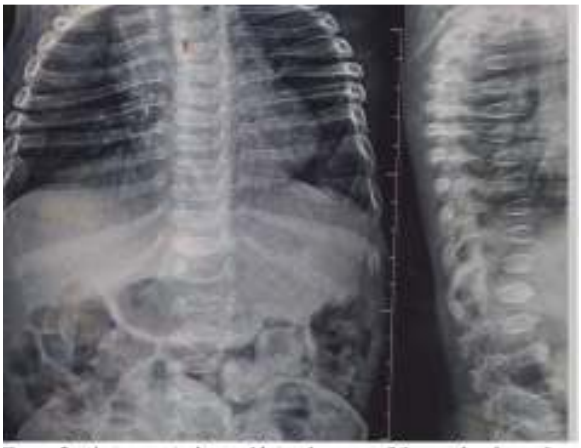

Figure 3: *Anteroposterior and lateral x-rays of thoracolumbar spine showing diffuse platyspondyly, anteriorly rounded/wedged vertebral bodies and increased intervertebral disc spaces*

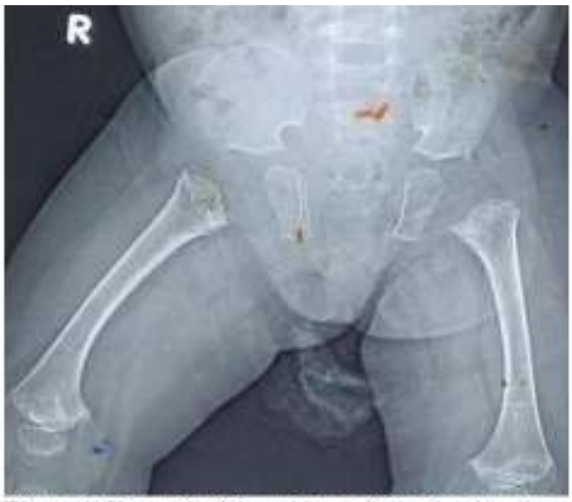

Figure 4: *X-ray of pelvis and lower limbs showing short flared iliac wings, irregular hypoplastic acetabular roof and widening, sclerosis and irregularity of metaphysis*

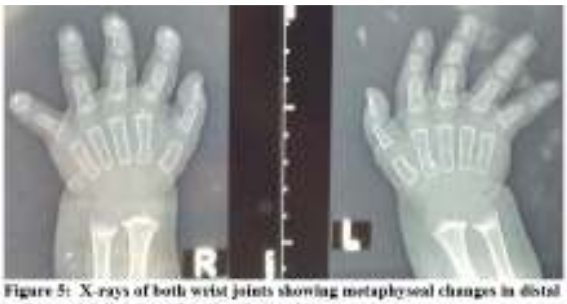

Figure 5: X-rays of both wrist joints showing metaphyseal changes in distal radius and delayed ossification of carpal bones

Spondylo-metaphyseal dysplasia was diagnosed based on the disproportionate short stature, small thoracic cavity, pectus carinatum and radiological features of generalized platyspondyly and metaphyseal changes. Molecular genetic testing was not performed due to financial constraints. Parents were counselled and regular paediatric clinic follow-up was arranged for height monitoring.

Discussion

Spondylo-metaphyseal dysplasia is characterized by metaphyseal dysplasia, generalized platyspondyly and radiographic changes in the spine, pelvis and proximal femur[1,5]. The child has classic features of spondylo-metaphyseal dysplasia that include short stature, short neck, pectus carinatum, varus deformity of limbs, and waddling gait[6]. Dorsal kyphoscoliosis, also a classic feature, is expected to develop during adolescence. Diagnostic radiographic findings in our case were generalized platyspondyly with anteriorly rounded/wedged vertebral bodies, widening, sclerosis and irregularity of metaphysis and carpal ossification delay[6,7].

Kozlowski classified spondylo-metaphyseal dysplasia into seven types based on severity, individual bones involved and transmission pattern[1]. Clinical features of our patient resemble spondylo-metaphyseal dysplasia type 1 - Kozlowski type; however, definitive diagnosis can only be made following molecular genetic studies. The differential diagnoses of type 1 include Kniest dysplasia, Jansen type metaphyseal chondrodysplasia, spondyloepiphyseal dysplasia, metatropic dysplasia and Morquio disease. All these diseases exhibit vertebral and metaphyseal abnormalities; however, they also have other features or organ anomalies that were not present in our patient[8,9]. Morquio disease was excluded in our patient by a negative urine test for mucopolysaccharidosis.

Spondylo-metaphyseal dysplasia does not have a cure. The treatment is targeted at symptomatic therapy, support, and counselling. Potential late complications include kyphoscoliosis and cervical spine instability due to odontoid hypoplasia[6]. Screening x-rays of the cervical and thoracolumbar spine and neck are recommended 2-3 yearly to monitor for cervical spine instability, kyphosis and scoliosis[2]. Affected individuals generally survive into adulthood and have a near-normal life expectancy[5].

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