Picture Stories

A case of campomelic dysplasia causing a pathological fracture

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(Key words: Campomelic dysplasia; pathological fracture)

Case report

A 67 day old baby boy, presenting with hypotonia and respiratory distress, had abnormal mobility of the left lower leg for past15 days following gentle manipulation of leg while dressing the baby at home. On x- ray he had a fracture of the left tibia with normal callus formation (Figure 1).

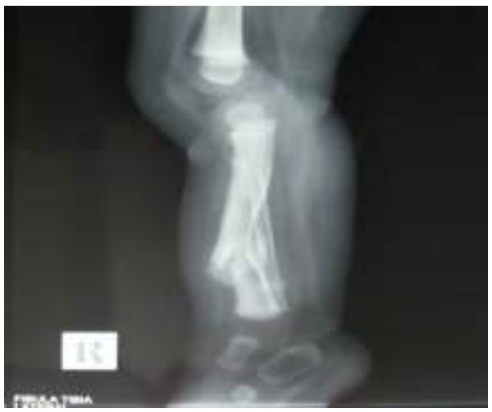

Figure 1: ***Fracture of left tibia***

This 4 kg baby boy was delivered at 37 weeks from a second gravida mother by caesarean section and developed respiratory distress just after birth. He had macrocephaly, large forehead, low set ears, flat nasal bridge, cleft palate, small jaw, short bowed upper extremities, small deformed thoracic cavity, short stature (42 cm, <3rd percentile), a webbed short neck and normal male genitalia (Figure 2).

Radiographs showed bowed humerus, radius and ulna of both upper limbs and a deformed chest cavity with 11 pairs of ribs (Figures 3 & 4). Mother was not exposed to medications or radiation during pregnancy and there was no family history of skeletal abnormalities or congenital malformations. Baby had a male karyotype 46, XY. A diagnosis of campomelic dysplasia was made on the basis of clinical and radiological findings.

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Figure 2: ***showing bowed upper extremity, small jaw and low set ears***

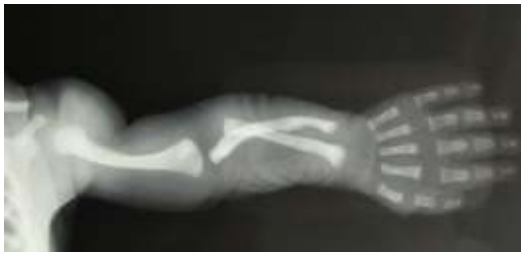

Figure 3: ***Bowed humerus, radius & ulna***

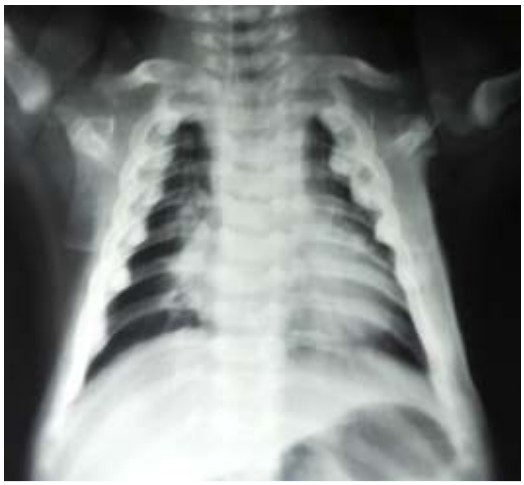

Figure 4: ***Deformed chest cavity***

Discussion

Campomelic dysplasia (CMD) is a rare autosomal dominant osteochondrodysplasia with or without sex reversal and with skeletal and non-skeletal defects, caused by de novo heterozygous mutations in the SOX9 gene (SRY-related HMG box gene family) on chromosome 17q[1,2]. The abnormal curvature of long bones, the main characteristic of CMD, is absent in 10% of patients, and the disease is then called acampomelic campomelic dysplasia[3]. Muscular hypotonia, craniofacial dysmorphism, cleft palate, brachydactyly, malformations of thoracic spine, and gonadal dysgenesis with female external genitalia and müllerian duct derivatives in the presence of a male karyotype are the common features of CMD[4]. Pathological fracture is not uncommon in this disease[4]. Respiratory distress, caused by small thoracic cage and narrow airways from defective tracheobronchial cartilages, is the main cause of death, which occurs mostly in the neonatal period.

References

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