

A Fatal Turkish Case of Campomelic Dysplasia

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Campomelic dysplasia (CD) is a rare form of congenital short-limbed dwarfism, classically characterised by campomelia (bowing of the long bones of the lower extremities) in association with a posterior cleft palate, flattened facies and hypoplastic scapulae. It was first fully and originally described by Spranger et al and Maroteaux et al in 1971.¹⁻³ CD is caused by mutations in the SRY related gene SOX9, mapped to 17q24.3-q25.1. A small proportion of cases are associated with structural rearrangements involving 17q and it has been proposed that this subgroup have a milder phenotype and better prognosis compared to those with mutations in the SOX9 gene.⁴ Herein, a newborn infant with CD is presented because of rare presentation.

A newborn girl was born to a gravida 4 mother with face presentation following full-term pregnancy. At birth, the 1st min and 5th min Apgar scores were 4 and 7, respectively. The parents were not relative. The first infant of the family was stillborn and did not have any anomalies. The second child, a 6-year-old, was normal. The third pregnancy ended with curettage. On physical examination, the general condition of the baby was moderately poor and she had moderate hypotonia. The birth measurements were as follows: weight was 2400 g (10th-25th percentile), length was 39 cm (10th percentile) and head circumference was 36 cm (90th percentile). She displayed macrocephaly and disproportionately short trunk and lower limbs. Her cry was hoarse. There were large ecchymotic plaques on her face secondary to face presentation delivery. The anterior and posterior fontanelles were 4 x 5 cm and 0.5 x 0.5 cm in diameter, respectively. Facial features included flattened and high forehead. The nasal bridge was also flattened and the palpebral fissures were narrow giving the appearance of hypertelorism. Besides a small mouth and long philtrum, micrognathia and retrognathia were also noted. No cleft in the soft palate was present. Her ears were abnormally low set. The thoracic cage was small and bell-shaped, and there was kyphoscoliosis. The lower limbs showed anterior bowing of both tibia and characteristic skin dimples. The femurs were also mildly angulated and there were talipes equinovarus, brachydactyly and clinodactyly of the 5th finger. Laboratory investigations of the blood and urine findings were normal. Radiograph of thorax showed a small bell-shaped chest, and 11 pairs of ribs with scoliosis and hypoplastic scapulae. The pelvis and lower limbs X-ray demonstrated bowed femur and

tibia (Fig. 1). The patient was hospitalised since birth. Aside from supportive therapy, nasal oxygen was given because she had respiratory distress secondary to retrognathia, small bell-shaped thoracic cage and kyphoscoliosis. Her general condition did not improve and she died on the 7th day of life.

Infants affected with CD have extreme hypotonia at birth, low birth weight, short birth length (35 to 49 cm), macrocephaly (average occipitofrontal circumference is 37 cm) and disproportionately short trunks and lower limbs.³ Characteristic facial features include flat appearing small face with high forehead, large anterior fontanel, low and flattened nasal bridge, the palpebral fissures are narrow, giving the appearance of hypertelorism. The mouth is small; long philtrum, micrognathia and retrognathia are present. The ears are abnormally malformed and/or low set, and most affected individuals are deaf.¹⁻⁵ The skeletal features are the most characteristic and prominent. The lower limbs show prenatal anterior bowing of the tibia and characteristic skin dimples. The femurs are also mildly angulated, and talipes equinovarus and dislocation of the hips are usually present. Short fibulae, scoliosis, kyphoscoliosis, brachydactyly and clinodactyly are common. In addition, flat vertebrae particularly cervical, hypoplastic bladeless scapular, small bell-shaped chest often with slender and 11 pairs of the ribs and a poorly mineralised sternum may also be observed.^{1,2,5} Our patient's clinical and radiographic findings were consistent with CD. After the critical first year, quality of life tends to improve. Most infants are mentally retarded. The oldest reported survivor was a 17-year-old who had an IQ of 45.^{1,5} Our patient died on the 7th day of life despite high-quality supportive management.

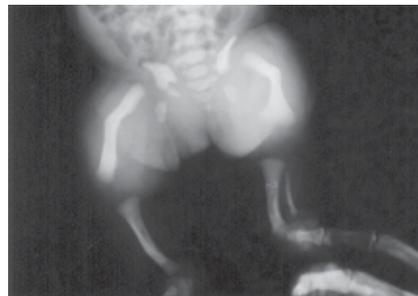


Fig. 1. Lower limb X-ray demonstrates bowed femur and tibia.

REFERENCES

1. Argaman Z, Hammerman CA, Kaplan M, Schimmel M, Rabinovich R, Tunnessen WW Jr. Picture of the month. Campomelic dysplasia. *Am J Dis Child* 1993;147:205-6.
2. Hall BD, Spranger JW. Compomelic dysplasia. Further elucidation of a distinct entity. *Am J Dis Child* 1980;134:285-9.
3. Horton WA, Hecht JT. The skeletal dysplasia. In: Behrman RE, Kliegman RM, Jenson HB, editors. *Textbook of Pediatrics*. 16th ed. Philadelphia: WB Saunders, 2000:2113-32.
4. Savarirayan R, Bankier A. Acampomelic campomelic dysplasia with *de novo* 5q;17q reciprocal translocation and severe phenotype. *J Med Genet* 1998;35:597-9.
5. Jones KL. *Smith's Recognizable Patterns of Human Malformation*. 5th ed. Philadelphia: WB Saunders, 1997:344-5.