CAMPTOMELIC DYSPLASIA : A case with bifid scrotum and hypospadias
Kamptomelik displazi: Bifid skrotum ve hipospadias'lı bir vaka

M Adnan Öztürk[1], Ercan Genç[2], Mustafa Öztürk[3], Bahri Elmas, Şükrü Doğan

Summary: *Camptomelic dysplasia syndrome is part of a large spectrum of bone dysplasias. It is a distinct entity that should not be confused with other conditions associated with congenital bowing of the long bones and is characterized by camptomelia, pretibial skin dimples, flattened facies, posterior cleft palate and hypoplastic scapulae. It is a rare congenital disorder. We report a case of camptomelic dysplasia associated with bifid scrotum and hypospadias. Our patient was seven days old and had all the characteristic findings for this syndrome. There were typical facial features, skeletal and roentgenographic findings.*

Key Words: *Dysplasia, Hypospadias, Scrotum*

Özet: *Kamptomelik displazi sendromu, kemik displazilerinden olup, farklı bir antite olarak uzun kemiklerin doğumsal kavisleşmesi ile ilişkili diğer durumlarla karıştırılmamalıdır. Bu sendrom, alt ekstremitelerde uzun kemiklerin kavis yapması, pretibial ciltte gamze, basık yüz görünümü, arka damak yarığı ve küçük skapula ile karakterize, nadir doğumsal bir hastalıktır. Biz bifid skrotum ve hipospadias' lı kamptomelik displazi vakası rapor ettik. Hastamız yedi günlüktü ve bu sendromun tüm karakteristik bulgularına sahipti. Tipik yüz görünümü, iskelet ve röngen bulguları vardı.*

Anahtar Kelimeler: *Displazi, Hipospadias, Skrotum*

Camptomelic dysplasia is a rare skeletal disorder and a form of congenital short limbed dwarfism, that is usually lethal. It is characterized by camptomelia (bowing of the long bones of the lower extremities), pretibial skin dimples, in association with a posterior cleft palate, flattened facies and hypoplastic scapulae (1).The inheritance pattern of this syndrome has not yet been definetely established. An autosomal recessive pattern is postulated (2). In this paper, a case of camptomelic dysplasia associated with bifid scrotum is reported.

Case Report

A newborn baby was born to a gravida-1 mother with head presentation following full term pregnancy. On the seventh day , this baby had respiratory distress and feeding difficulties. Birth measurements were as follows;weight 1950g(<3 P), length 42 cm (< 3 P), head circumference 33.5 cm (10- 50 P). The general condition of the child was moderately poor and hypotonic. He displayed normocephaly and disproportionately short trunk and lower limbs. The anterior and posterior fontanel widths were measured as 4x3 cm and 0.5x0.5 cm 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. His ears were abnormally low-set (Figure 1). Small mouth and long philtrum, micrognathia, retrognathia and posterior cleft palate were other abnormalities. The thoracic cage was small bell shaped and kyposcoliotic. Lower limbs showed anterior bowing of the tibiae and characteristic skin dimples (Figure 2). The femora were also mildly angulated and there were talipes equinovarus, brachydactyly and clinodactyly of the fifth fingers. In addition there were bifid scrotum and hypospadias (Figure 3). Testicles were in the upper scrotum.

Erciyes Üniversitesi Tıp Fakültesi 38039 KAYSERİ
Çocuk Sağlığı ve Hastalıkları. Doç.Dr.[1], Araş.Gör.Dr.[2]. Radyoloji. Y.Doç.Dr.[3].

Geliş tarihi: 30 Ekim 1996

The complete blood count, blood chemistry, urine analyses, cranial computerized tomography, echocardiography and abdominal ultrasonography findings were normal. Chromosomal pattern was 46 XY. Roentgenogram of the total body is shown (Figure 4). Our patient died at the twenty fifth day of life due to respiratory failure. Necropsy findings were hypoxic edema of brain, pulmonary hemorrhage and occasional hyalinized appearance due to adult type respiratory distress syndrome, bilateral normal testicles in the upper scrotum, hypoplastic inferior scapulae. No abnormality was noted related to internal genitalia.

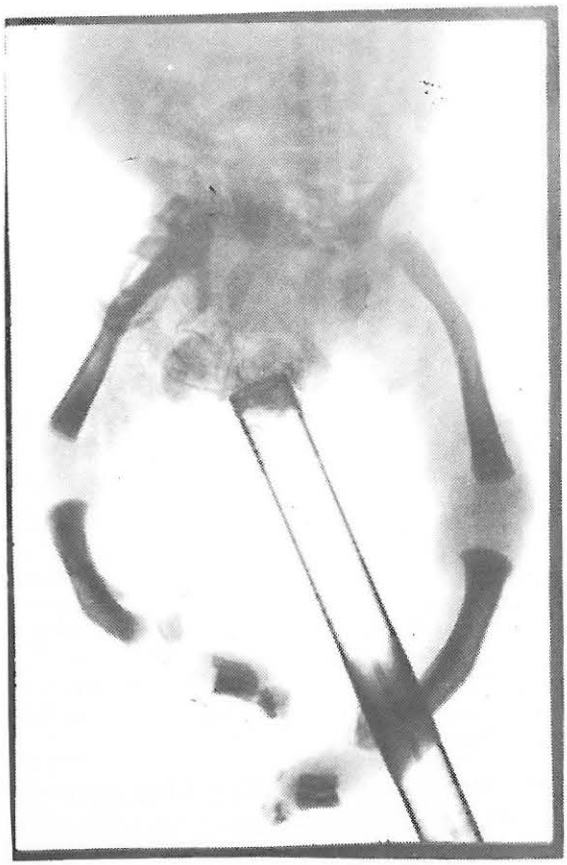

Figure 2. Low limbs roentgenogram demostrates bowed femora and tibiae

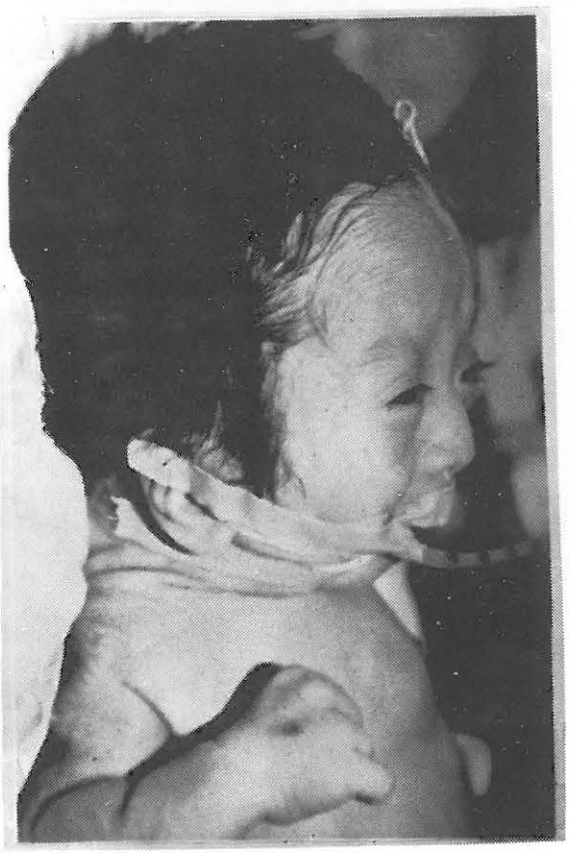

Figure 1. Characteristic facial features in camptomelic dysplasia, including flattened facies and nasal bridge, high forehead, low set ear and small mouth

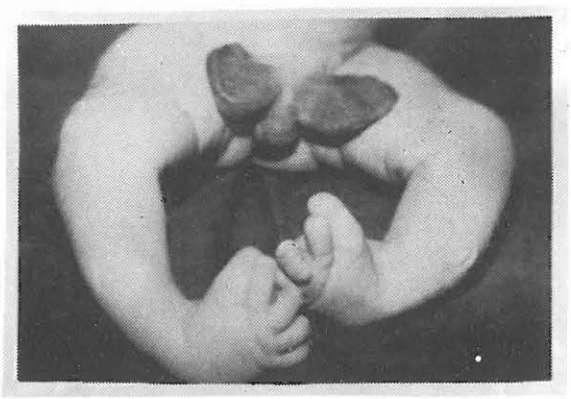

Figure 3. Angulated tibiae with pretibial skin dimples, talipes equinovarus and bifid scrotum, hypospadias

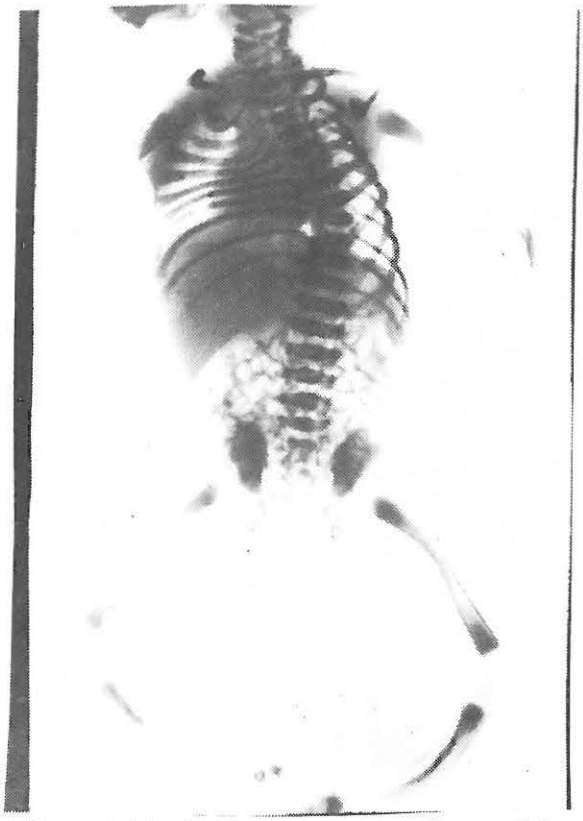

Figure 4. Total body roentgenogram; Short and flat vertebrae, small bell-shaped chest, 11 pairs of ribs with scoliosis and bowed femora and tibiae

Discussion

Camptomelic dysplasia was first fully and originally described by Maroteaux et al (3) in 1971. Infants affected with camptomelic dysplasia have extreme hypotonia at birth, low or normal birth weight, low birth length (35 to 49 cm), macrocephaly or normocephaly and disproportionately short trunks and lower limbs. Characteristic facial features include flat appearing small face with high forehead, large anterior fontanel, low and flattened nasal bridge. The palpebral fissures were narrow, giving the appearence of hypertelorism. The mouth is small, with long philtrum, micrognathia and retrognathia. A cleft of the soft palate is present in two thirds of patients with camptomelic dysplasia . The ears are abnormally malformed and/ or low set (1-3). The skeletal findings are the most characteristic and prominent features. The lower limbs show prenatal anterior bowing of the tibiae and characteristic skin dimples (1-3). The pretibial dimples appear to result from the loss of subcutaneus tissue secondary to marked strecthing of the skin overlying the apexes of the bony curvatures during fetal life. The femora are also mildly angulated and talipes equinovarus and dislocation of the hips are usually present. Short fibula, scoliosis, kyphoscoliosis, brachydactyly and clinodactyly are common (1-4).

Our case did not have dislocation of the hip. Roentgenographic findings included curved femora and tibiae, short and somewhat flat vertebrae (particularly cervical), hypoplastic scapulae, small bell- shaped chest often with slender and eleven pairs of the ribs and a poorly mineralized sternum. Roentgenograms of the spine show kyphoscoliosis, abnormal cervical vertebra, hypoplastic pedicles of the thoracic pedicles. Roentgenograms of the pelvis may show small iliac wings with relatively wide pelvic outlet (1-5). Tracheobronchial findings include incomplete cartilaginous development with tracheobronchiomalacia. Larynx may be small. Central nervous system abnormalities include a large brain with hydrocephaly and absence or hypoplasia of the olfactory bulbs and tracts. Less common findings include marked hypoplasia of the corpus callosum and anomalies of the caudate nucleus, thalamus and cerebral pedincles. Defects of the respiratory apparatus contribute significantly to the early death of affected infants. In addition to the small bell-shaped thoracic cage, which creates a restrictive respiratory effect, there are significant upper airway and pulmonary abnormalities. Genitourinary abnormalities in this syndrome include hydronephrosis, hydroureter, renal hypoplasia and renal cysts, ambigious genitalia (1-5). A sex reversal phenomenon has been described in patients with male 46 XY karyotypes and female phenotypes which may explain the apparent sex predilection. It has autosomal recessive inheritance (6). Genetic counseling can then be offered for further pregnancies. Prenatal diagnosis, using ultrasonography, is possible at as early as 18 weeks gestation (7). Differential diagnosis should be made among osteogenesis imperfecta, hypophosphatasia, diastrophic dwarfism and Larsen's syndrome (2,4,7). Only supportive medical care can be offered to the newborn.

REFERENCES

1. *Smith DW. Camptomelic dysplasia syndrome. In: Smith DW (ed).Recognizable Patterns of Human Malformation. (3 rd ed), WB Saunders Philadelphia, 1982; pp 246-247*
2. *Argaman Z, Hammerman CA, Kaplan M, et al. Picture of the month. Am J Dis Child 1993; 147: 205-206*
3. *Maroteaux, Spranger JW, Opitz JM, et al. Le Syndrome Campomelique. Presse Med 1971; 22: 1157-1162*
4. *Hall BD, Spranger JW. Camptomelic dysplasia. Further elucidation of a distinct entity. Am J Dis Child 1980; 134: 285-289.*
5. *Austin GE, Gold RH, Mirra JM, et al. Long-limbed camptomelic dwarfism: A radiologic and pathologic study. Am J Dis Child 1980; 134: 1035-1037.*
6. *Mansour S, Hall CM, Pembrey ME. A clinical and genetic study of camptomelic dysplasia. J Med Gen 1995; 32: 415-420.*
7. *Sanders RC, Greyson-Fleg RT, Hogge WA. Osteogenesis imperfecta and camptomelic dysplasia: Difficulties in prenatal diagnosis. J Ultrasound Med 1994; 13:691-700.*