CHONDRO-ECTO DERMAL DYSPLASIA (ELLIS-VAN CREVELD SYNDROME) (Case reports with review of literature)

B. K. SOHI,* A. S. SOHI † AND M. K. GHOSH ‡

Summary

Two sisters presenting features of Chondro-Ectodermal Dysplasia (Ellis-van Creveld syndrome) are described and literature reviewed. They were youngest of six sibs, elder four being normal. Their mother showed feature of ectodermal dysplasia in the form of nail and teeth defects. These are the first living cases being reported from India.

Chondro-ectodermal dysplasia was first described by Ellis and Van Creveld in 1940[1]. These cases have involvement of ectodermal derivatives such as nails and teeth in addition to chondrodystrophy. There may be other congenital anomalies as well. Mode of transmission is recessive. (Metrokos and Fraser, 1954)[2]. Lynch et al[3] reviewed 103 cases upto 1968 while Madhavan et al[4] (1976) mentioned that 115 cases were registered in world literature and also described a neonate with this malady. We are describing this syndrome in two sisters. They were youngest of six sibs. Four elder siblings were normal and their mother had features of ectodermal dysplasia. These are the first living cases being reported from India.

* Department of Paediatrics, Command Hospital (SC) Pune.
† Department of Dermatology, Command Hospital (SC) Pune. (At present – Military Hospital, Jullundur Cantt),
‡ Department of Radiology, Command Hospital (SC) Pune.
Request for reprints to :
Lt. Col. A. S. Sohi, AMC,
Dept of Dermatology, Military Hospital,
Jullundur Cantt.
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Case Reports

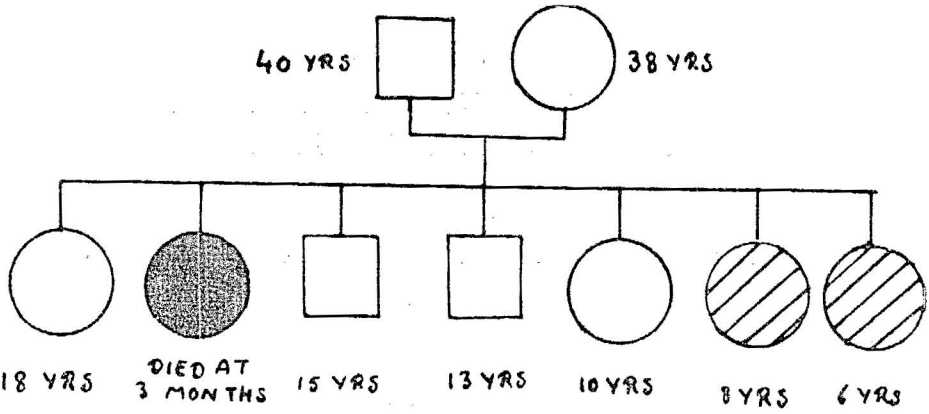
Case No. 1

K, eight years old female child was brought with history of marked deformities of upper and lower limbs for 5 years, which were incapacitating her for locomotion. She was born normally at full term and milestones were normal. From the age of 3 years she started getting progressive deformities of bones of both upper and lower limbs.

Her family tree is depicted below.

Father was short statured and thin built (Height 156 cm). Mother was of same height as father but edentulous, having lost her teeth in childhood. Her nails especially of fingers, did not require trimming for many months at a stretch indicating thereby very slow growth. She had no other skeletal or other congenital defects. Patient's youngest sister was also affected but other sibs were normal. (Fig 1).

On examination, the child was intelligent, co-operative and cheerful. Her height was 81 cms, weight 10 kg and head circumference 48 cms. Her hairs



were thin and sparse. Except the lower two incisors which were peg shaped all other teeth were missing giving the child, an 'old woman look'! Upper gingvolabial fold was obliterated but lower one was normal. Nails were small, thin, ridged and hardly ever required trimming. Upper limbs were short and forearms were bowed having convexity posteriorly (Fig 2). Humerii were bowed with thickened lower ends. Chest was cylindrical, narrow and flaring at base and sternum was bulging forwards. Lower limbs were short and deformed with shortening and bowing of thigh and leg bones. There was no other congenital anomaly nor was any physical abnormality detected in any other system clinically. Radiologically both femora were short, thick and curved with marked oseteoporosis of femora and pelvic bones (Fig 3). Tibiae and fibulae were short, curved in lower halves, with widening of lower ends, separation of epiphysis and marked osteoporosis. Tarsal and metatarsal bones also showed marked osteoporosis. Spine showed verterba plana, six lumbar vertebrae, beaking of L 4 and osteoporosis (Fig 4). In upper limbs humerii showed fragmentation of heads, thin pencil like lining on inner sides with fractures and callus formation (Fig 5, marked with arrows). Radii and ulnae were shortened, thickened and bent, though curvature was much less compared to that of lower limbs. Lower ends of these bones were widened and osteoporotic with separation of epiphysis. There was fusion of carpal bones. Metacarpal and phalangeal bones showed osteoporosis, thinning of cortex and separation of epiphysis (Fig 6). X-Ray of the jaws showed 5 teeth in upper jaw and 8 in lower jaw. Mandible was thin with marked osteoporosis (Fig 7). Maxillae were hypoplastic and frontal sinuses were absent. On 3-5-76 she had a fall from a height of about 45 cms and sustained fracture of right femur, which was fixed by nailing and healed well.

Lab investigations were as follows:

Blood-HB 13 gms%, TLC-8,500/cmm, P-50%, L-40%, M-2%, E-8%, Blood for STS non-reactive Blood urea-38 mgm%, Blood creatinine 1.0 mgm. Liver function tests showed: Serum bilirubin-1 mgm%, total proteins, 6.9g%, albumin-4.5 g%, globulin-2.4 g%, A/G ratio 1.8:1, serum calcium-4.9 mEq/L organic phosphate 1.9 mEq/L, alkaline phosphatase 8.0 KAU. Urine calcium 2.4 mgm/100 ml, inorganic phosphate-80 mgm/100 ml, aminoacid nitrogen 45 mg/100 ml. ECG was normal. Routine urine and stool examination were normal.

Case No. 2

Younger sister of case No. 1 aged six years gave history of inability to walk and progressive deformities of both

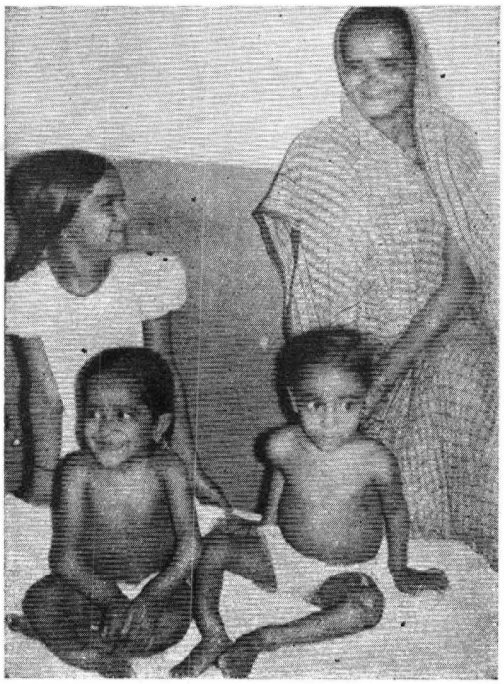


Fig. 1 Two affected sisters, case I (Right) and case II (Left). Sitting. Mother and one normal sister standing at back.

Fig. 2 Case I, showing deformities of limbs chest and teeth.

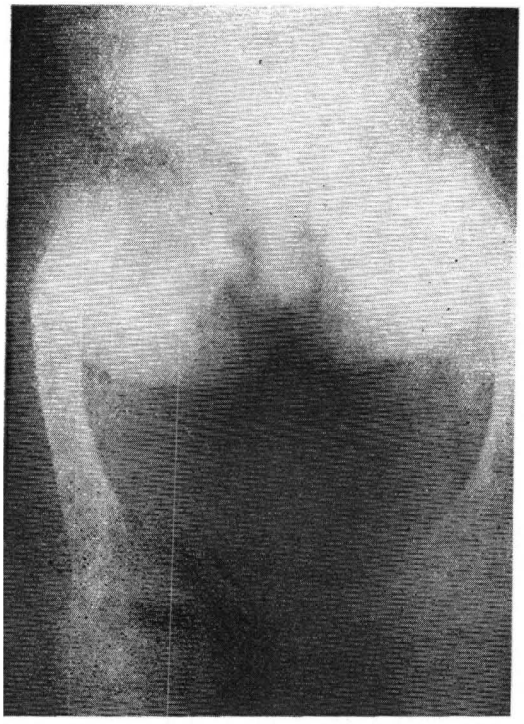


Fig. 3 X-ray of pelvis and femora. Showing marked osteoporosis.

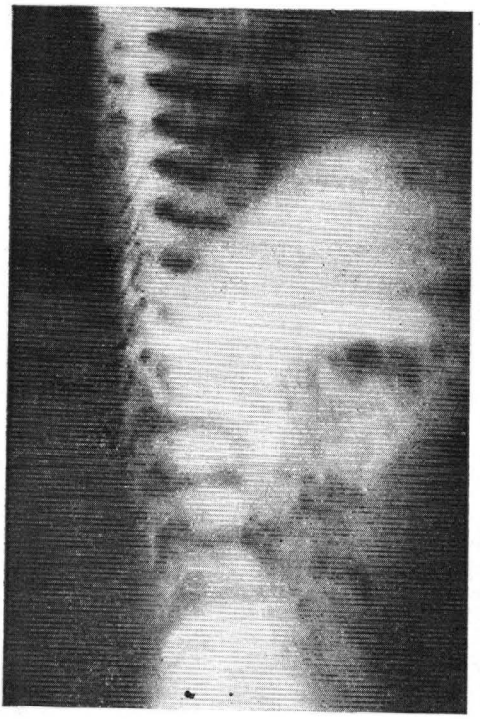


Fig. 4 X-Ray spine lateral view - showing vertebra plana, 6 lumbar vertebrae, beaking of L4 and osteoporosis.

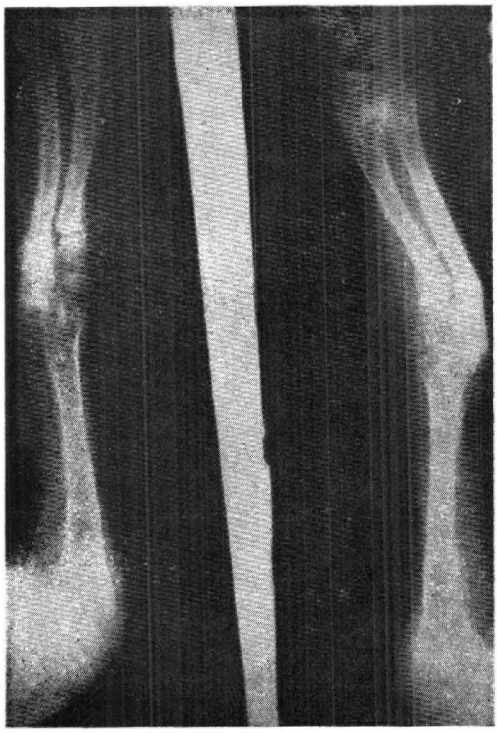


Fig. 5 X-ray of humerii radii and ulnae-showing fracture and callus formation. (arrows)

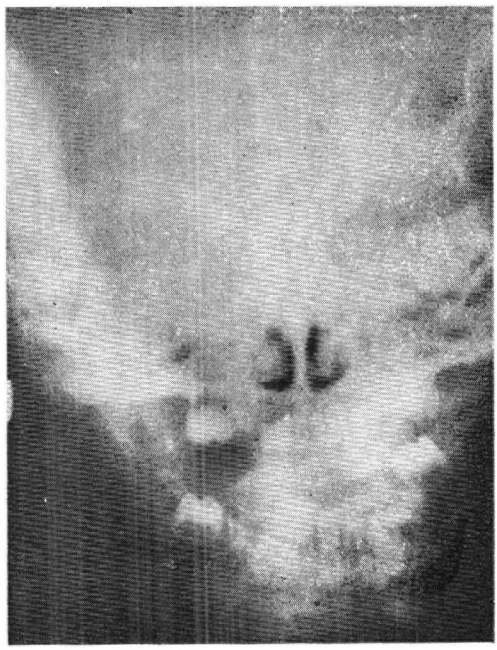


Fig. 7 X-Ray both jaws and skull - showing 5 teeth in the upper jaw and 8 in lower jaw.

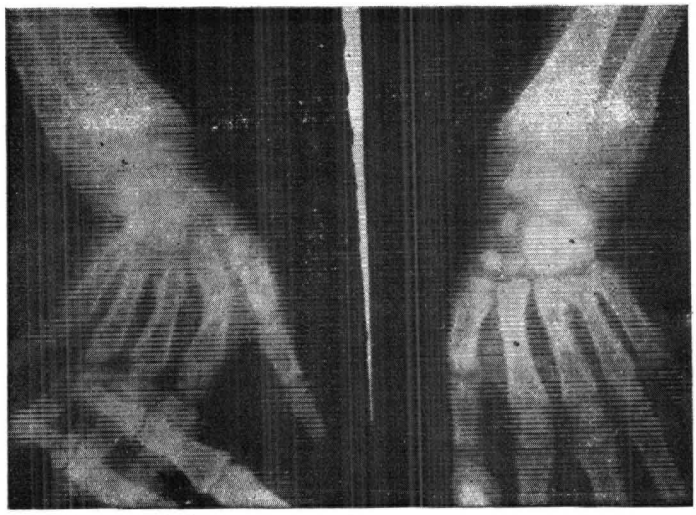


Fig. 6 X-Ray wrists and hands-showing osteoporosis, thinning of cortex and separation of epiphysis.

upper and lower limbs for three and a half years. She was born normally at full term. Mile stones were normal. She was an intelligent and cheerful girl of height 79 cms, weight 9 kg and head circumference 47 cms. There was frontal bossing and facial configuration resembled that of the elder affected sister. Hairs were thin and sparse. Nails looked small and thin. Patient did not remember having ever trimmed them. Only few teeth were present in both upper and lower jaws, and those present were irregular and deformed (Fig 8). Both upper and lower limbs were shortened and deformed with bowing, but deformities were less marked than those of her elder sister. No other congenital deformity or physical defect was detected in other systems. Radiological findings resembled those of the elder affected sister.

Lab investigation were as follows : Blood Haemoglobin 12.5 g% TLC-6, 600/cmm, P-61%, L-35%, M-2%, E-2%, Blood urea 38 mgm%, Blood creatinine-1.0 mgm, serum sodium 140mEq/L potassium - 5.3 MEq/L, chloride - 97 mEq/L, CO^2 combining power-23, serum calcium-4.5 mEq/L, inorganic phosphate-2 mEq/L, alkaline phosphastase-8.0 KAU. ECG and X-Ray chest normal.

Discussion

Though this syndrome is named after Ellis and Van-Creveld in 1940, one of the three cases included in their report was first described in 1933 by Mc Intosh. This patient lacked congenital heart lesion and hairs were unaffected. Ellis and Andrew (1962)[5] reported two cases and discussed in detail chondro-ectodermal dysplasia. Upto that time 40 cases had been reported. As the name implies, this syndrome affects both mesodermal and ectodermal tissues. Polydactyly more or less a consistent feature in earlier reported cases was absent in our patients.

This condition has been described in sibs previously, and isolated feature of syndrome has occurred in forebears, and sibs. In the present report, mother had hair and teeth defects. The syndrome has been described in Negro, Arab, Jewish, Turkish, Mediterranean, North European and American children, but only in one neonate from India who died within a few minutes after birth[4]. The present reports are those of the first living cases in India.

The shortening of limbs typically affects the distal segment to a greater degree than the proximal. Humerii and femora may be bowed, thickened and expanded distally. The paired bones commonly show unequal shortening. There is enlargement of the head of ulna and distal end of radius. Proximal end of tibial shaft is widened and pointed, epiphyseal ossification centre is hypoplastic. Caffey[6] regarded the appearance of tibia as diagnostic. All the features mentioned so far with exception of bowing of humerii were seen in our cases.

The phalanges are markedly shortened and the centre of ossification for terminal phalanges may be absent. Frequently there is fusion of two or more carpal or tarsal bones. In no instance has the skull or facies shown the appearance characteristic of classical achondroplasia. The nails and teeth are almost invariably affected as in our cases. In some cases hairs are sparse or fine and our cases also showed these features.

Skin was not involved and sweating was normal in both the sisters as had been reported in earlier cases. The nails are small, dystrophic, scale like and they may be absent on one or more digits. In our cases nails were small with thin nail plate and very slow rate of growth. The teeth are markedly

irregular (as in case No. 2) and individual teeth may be pointed, dystrophic and absent (as in case No. 1 and the mother). Premature eruption of malformed teeth may be noted at birth or soon afterwards. There may be fusion of maxillary gum pad with upper lip obliterating the normal gingvo labial sulcus, as was seen in our first case.

Ellis detected congenital heart disease in 22 out of 38 cases but in our cases heart was normal. Various congenital anomalies associated with this syndrome like malformation of genitalia, undescended testes, cleft palate, cleft lip, strabismus, coloboma of iris and hepato-splenomegaly have been described.[1] However there were no congenital anomalies in our cases, nor was intelligence affected.

Jansen's disease[7] is a condition affecting metaphysis, characterized by spreading, cupping and defective irregular mineralization of the metaphyses of tubular long bones and to some degree affects the edges of flat bones. Blood chemistry and renal functions are normal. Striking features of this metaphyseal dysostosis are smooth edges of the epiphyseal ossification centre in contrast to the rough edge of its contiguous metaphysis. Mild types are much more common simulating radiographic changes of refractory rickets, and hypophosphatasia; differentiating point being normal blood chemistry. Radiological appearances of this condition, resembles to some extent those of our cases but our cases had clear cut association of ectodermal dysplasia which is not described in Jansen's disease.

Prognosis with regard to life expectancy of these cases depends upon those of associated congenital anomalies, like heart disease. Pelvic deformities are liable to cause obstructed labour in females and limb deformities limit locomotion. Rarefaction of bones make them prone to fracture with slight trauma as happened in our case No. 1. There is no specific treatment.

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