ELLIS -- VAN CREVELD SYNDROME
(A case report)

A. MAHAKRISHNAN,* S. VELU † AND P. I. PANDIAN *

Summary

A 23 years old male with typical features of Ellis-Van Creveld Syndrome is presented for its rarity. This is the second living case being reported from India.

Ellis-Van Creveld syndrome is characterised by ectodermal defects, dwarfism with characteristic foreshortening, and polydactylism involving the ulnar side of hands. 60% of cases may show cardiovascular abnormalities. 115 cases were reported in world literature upto 1976[1]. Sohi, et al reported the first living case from India[2]. We are presenting a case showing all the somatic features of the syndrome. No cardiac abnormalities were present in this case.

Case:

23 years old male born of non consangunous marriage was seen in the Department of Dermatology, Tirunelveli Medical College Hospital, Tirunelveli for defective finger and toe nails. On examination he showed disproportionate dwarfism with extreme shortening of distal segments of the limbs (Fig. 1). Skull and trunk were normal. Polydactyly on the ulnar side of both hands (Fig. 2) and lateral side of both feet were present. There was a severe degree of genu valgum on both sides with inability to hold the feet together (Fig. 3). The hands were as broad as their length.

Fig. 1 Shows dwarfism with normal trunk and skull extremities show foreshortening.

Patient was not able to make a firm fist. The finger and toe nails were thin and brittle. Scalp, axillary and pubic hairs were normal in structure and quantity. There were number of frenulae binding down the upper lip to the gum. The teeth showed wide spacing, poor development of enamel, absence of lower

* Department of Dermatology
† Department of Radiology
Tirunelveli Medical College Hospital

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Fig. 2 Short hands with polydactyly

incisors, and several broken teeth (Fig 4). Systemic examination revealed no abnormalities. Family history revealed that 5 elder siblings of the patient had died in their childhood. The exact mode and cause of their death could not be elicited.

Investigations :

Routine urine and haematological examinations were normal. ECG–Normal-VDRL-Non-reactive. Radiological survey of the skeletal system showed the following : Skull and chest-Normal. Short fibulae and exostosis on the medial side of upper end of both tibiae (Fig. 5), polydactylism on the ulnar side of both hands, fusion of hamate and capitate on both sides, partial synmetacarpalism (Fig. 6) of the left hand.

Discussion :

Ellis and Van Creveld described a disorder of chondro-ectodermal dysplasia in 1940. The main clinical features of the disorder are ectodermal defect, polydactylysm and dwarfism with characteristic foreshortening of the limbs with normal trunk and skull. The ectodermal defects are present as nail dysplasia. Nails may be small, flutted, brittle or abnormally pigmented. The teeth may be small, defective, wide spaced and peg shaped. The upper lip may be bound down to the gum by number of frenulae. The hair is usually normal but alopecia may be a rare feature.

The dwarfism is characterised by normal head and trunk with short limbs. The short limbs themselves show a disproportionate distal shortening. The hand may be as broad as their length. Polydactyly affects the ulnar side of the hands. In 10% of the patients extradigit may also affect the feet. Radiological survey of the skeletal system will show shortening of the limbs polydactylysm, synmetacarpalism, polymetacarpalism as well as fusion of capitate and hamate. In the lower limbs the fibulae show greatest shortening, the tibia shows exostosis on the medial aspects of proximal ends, defective ossification of the lateral ends leading to extreme genuvalgum. The pelvis may be small, short with squared iliac rings. The sacro sciatic notch may be short. The skull and spines are spared. Cleft lip, hare lip, hypoplasia of mandible, strabismus, coloboma of the iris, longitudinal pattern of dermatoglyphics, hypospadiasis and undescended testis are other rare features of the syndrome. 60% of the cases may show cardiac abnormalities presenting with single artium, A.S.D., V. S. D. and transposition of great vessels[3,4,5].

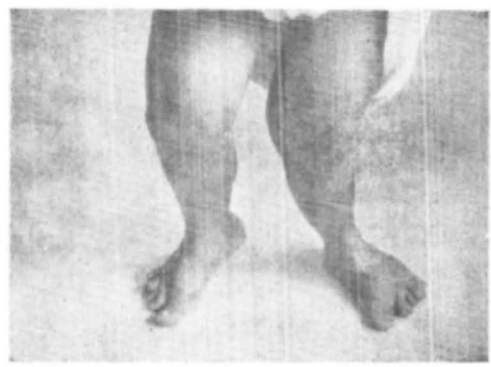

Fig. 3 Shows extreme genu valgum

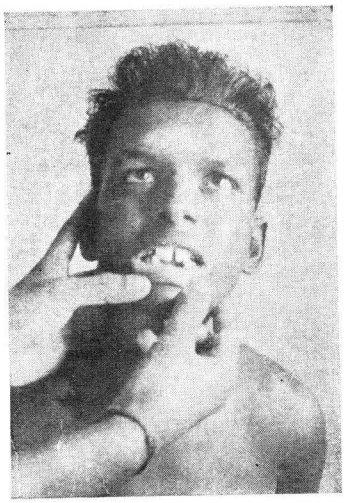

Fig. 4 Shows absence of lower incisors wide space in betweeu the upper incisors

This condition differs from achondroplasia wherein hydrocephalus frontal bossing, affection of vertebrae and proportionate shortening of the limbs are noted features. Cartilage hair-hypoplasia (Mckusick's syndrome), which may also show a distal shortening of the limbs will always present with sparse and thin hair. The fibulae will

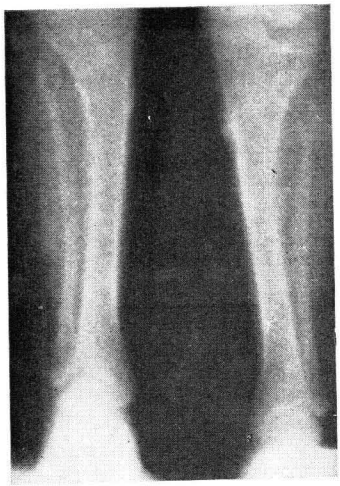

Fig. 5 X-Ray of the legs showing short fibulae exostosis upper end of the both tibiae

be long giving a characteristic ankle deformity. The loose joints of the feet leads to flat foot[4].

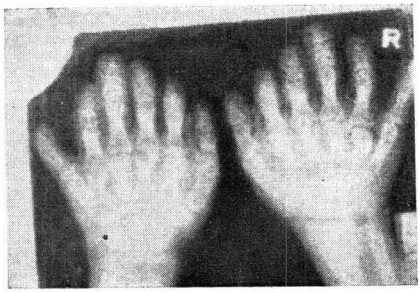

Fig. 6 X-ray of the hands showing polydactyly, fusion of hamate and capitate on both sides, the left hand shows partial synmetacarpalism

Jeune's osteodystrophy is characterised by narrow thorax with hypoplastic short ribs usually associated with renal disease unlike Ellis–Van Creveld syndrome where congenital heart disease is a usual association[4].

The cause of Ellis-Van Creveld Syndrome is not known. Study among the old order of Amish by Mckusick have suggested an autosomal recessive inheritance. The effect of the genes seems to have an inhibitory action of the normal development at the end of second and beginning of the third foetal month as evidenced by

(i) disturbance of proximo distal elongation of the limbs

(ii) persistent absence of the sulcus labio gingivalis and

(iii) frequent failure of the heart septum to close completely[4].

Conclusion :

We present a patient with typical skeletal and ectodermal features of Ellis-Van Creveld Syndrome who is leading a normal life. The family history of early death of the siblings suggest that the cause of death in them may be due

to cardiovascular abnormalities which is a noted feature in this syndrome. This is the second report of a living case of Ellis-Van Creveld Syndrome from India.

Acknowledgement

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ABSTRACT

Comparison of the Tensile Properties of Human Scalp Hair With And Without Natural Pigment. A. B. Gupta, K. G. Ghosh and B. Haldar (Physical and Earth Sciences Division, Indian Statistical Inst., Calcutta) Indian J Dermatol, 1979 ; 24 : No. 4.

The tensile properties - ultimate tensile strength (UTS) and ultimate percentage elongation (UPE) of human hairs (240 strands) with and without natural pigment, which were extracted from the occipital region of the scalp of 12 Bengali-speaking Hindu subjects of both sexes in the age range 15–72 years have been examined. The tensile properties of hair with natural pigment (Black : Type I) have been compared with those with no natural pigment (Grey : Type II) for each individual and the difference has been tested statistically to assess its significance. It is observed that there is no significant difference ($P > 0.05$) in both the UTS and UPE-values between the given two types of scalp hairs of an individual. From the point of view of forensic scientists in particular and the human biologists in general, the finding of the present explorative study is expected to be of immediate importance.