CHONDRO-ECTODERMAL DYSPLASIA (ELLIS-VAN-CREVELD SYNDROME)

HELEN SENDER, M.B., B.CH., D.M.R.D., D.P.H., *Department of Radiology, Johannesburg General Hospital, Johannesburg*

It is the purpose of this paper to discuss briefly the diagnosis, the variability of the manifestations, and the hereditary factors in the syndrome of chondro-ectodermal dysplasia (Ellis-van-Creveld syndrome).[1,2]

The clinical diagnosis can be made on inspection of the patient even immediately after birth. There is shortening of both upper and lower extremities, especially below the knees and elbows, with short stubby hands, polydactyly, and sometimes syndactyly. There is hypoplasia of the nails and sometimes of the hair. The teeth, if erupted, are usually dysplastic.

THE RADIOLOGICAL APPEARANCE

This is diagnostic.

Hands

These show multiple deformities which include supernumerary phalanges and metacarpals, fusion of capitate and hamate bones, delayed maturation of primary phalangeal ossification centres, and accelerated maturation of secondary phalangeal epiphyseal ossification centres.

All the metacarpals are short. In the rudimentary 6th finger there is no centre for the distal phalanx. An example of affected hands is shown in Fig. 1.

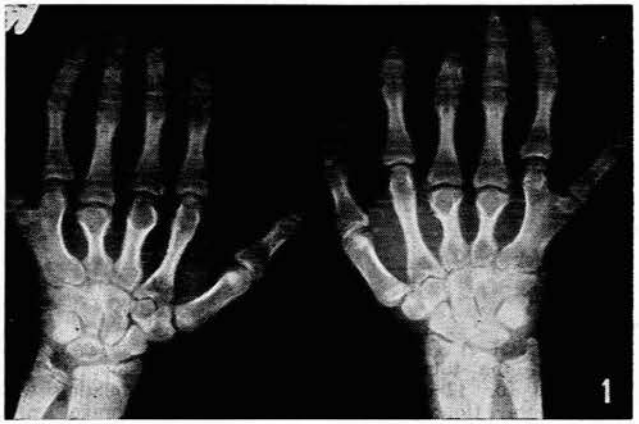

Fig. 1. Polydactyly with short metacarpals.

Lower Extremity

The tibia and fibula are both shortened. The proximal end of the tibial shaft is widened and its cephalic edge is pointed, with a lateral slope and a shorter medial slope. The latter is capped by a small thin triangular ossification centre in the epiphyseal cartilage. A small exostosis projects medially and caudally from the medial cortical wall of the tibia. The patella is large and well mineralized. Most of these features are well shown in Fig. 2.

Pelvis

The sacrum is small. The necks of both femora are bent upwards and outwards into extreme reverse coxa valga deformities. The femoral heads are flattened medially and their lateral segments project laterally well beyond the confines of the hypoplastic acetabular cavities (Fig. 3).

OTHER ANOMALIES

The Cutaneous Dysplasia

This has been severe and consistent in all the reported cases. Teeth in both the upper and lower jaws in all cases

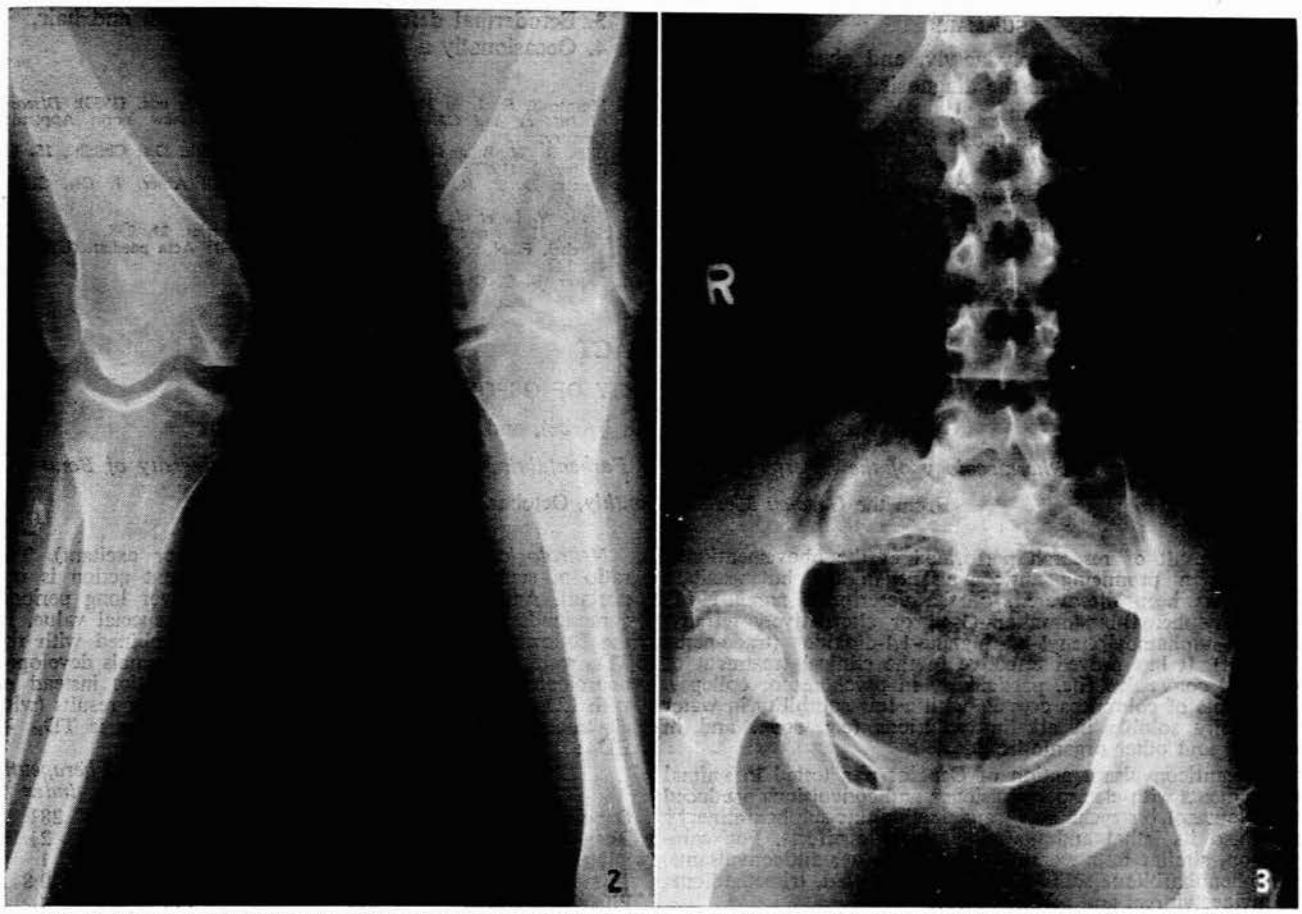

Fig. 2. Femora and tibiae showing: classic genu valgum deformity, short medial slope and long lateral slope of widened proximal end of tibial shaft, normal patellae, and bilateral tibial exostoses directed medially and caudally.
Fig. 3. Bilateral coxa valga deformity.

have been defective, and deformed and retarded in their development. Every nail of every finger and toe of every patient has been too small, often deformed and often excessively friable. The nails on rudimentary sixth digits are usually absent.

Cardiac Involvement

Congenital cardiac disease occurred in 3 of the 5 reported cases of John Caffey.[3]

DISCUSSION

All the tubular bones in the extremities are shortened and relatively thick. These bones are short absolutely in proportion to the trunk.

Dwarfism is due almost exclusively to shortening of the legs. It is noteworthy that the fibulae, which are relatively the shortest bones in the legs of patients with chondro-ectodermal dysplasia, are relatively the longest bones in the legs of achondroplastic subjects.

The tibial deformity is a pathognomonic lesion of the Ellis-van-Creveld syndrome. The proximal widened ends of the tibial shafts are sharpened into low peaks with relatively long lateral slopes and short medial slopes. The latter are capped by thin hypoplastic ossification centres in the proximal epiphyseal cartilages. This accounts for the classical genu valgum deformity. Patellae are usually normal.

The hands of all patients are practically identical. Each hand has a rudimentary 6th digit attached to its ulnar side. Each of the hands has six metacarpals.

Diagnosis

In the diagnosis of this condition one relies heavily on the X-ray changes. Caffey[3] has stressed the pathognomonic pattern seen in cases of chondro-ectodermal dysplasia, i.e. progressively increased shortening from the trunk to the distal phalanges. The root bones of the extremities are relatively the longest, in contrast to classical achondroplasia in which the reverse is true.

In every case there has been a secondary characteristic feature,[4-7] e.g. the widened ends of the tibial shafts which are irregular, and the ossification centres in the proximal epiphyseal cartilages which are hypoplastic.

Inheritance

The mechanism for inheritance of these congenital anomalies is a subject needing further elucidation.[8] The statistical probability of a sibling having the complete tetrad of defects is rather remote. A single recessive defect appears in the siblings of affected persons in only 25% of cases.

SUMMARY

Ectodermal dysplasia, polydactyly, and chondro-dysplasia must be present to make the diagnosis. There may or may not be congenital heart disease.

An autosomal recessive mode of inheritance is strongly suggested.

The syndrome of Ellis-van-Creveld consists of:

1. Osseous deformities with exostoses and peripheral chondro-dysplasia.
2. Polydactyly.
3. Ectodermal defect affecting nails, teeth and hair.
4. Occasionally a cardiac defect.

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