CHONDRO-ECTODERMAL DYSPLASIA *

Ellis-Van Creveld Syndrome : A Case Report

S. Bilir, M. D.,** N. Bilâloğlu, M. D.***
and K. Soydan, M. D.****

A new type of chondrodysplasia was first described by Ellis and van Creveld [1] in 1940 and later called the Ellis-van Creveld syndrome (E-vC). The disorder, which consists of chondrodystrophy, ectodermal dysplasia affecting the hair, nails and teeth, polydactyly and congenital heart disease, is rare. Several authors collected case reports and added their own.[2-5] Until 1964, the number of the reported cases was 52 according to McKusick.[5] This syndrome is seen in many different countries, including Turkey.

Our purpose in presenting another case is to draw greater attention to this type of disorder. In reviewing the Turkish medical literature, we found no reports on the E-vC syndrome, but, in our opinion, this is not because of a lack of cases in this country. The majority of patients with E-vC syndrome are either stillborn or born with a heart involvement which causes death either in the newborn period or in early infancy. However, patients with E-vC syndrome, but with no heart involvement, do not go to a doctor because of extra digits, hair or teeth troubles, or, if they do, the doctor does not pay enough attention to these symptoms. Since endogamy occurs frequently in Turkey, especially in villages and small towns, one would expect more cases of E-vC in the families who present such anomalies.

Case Report

A one and a half-year-old female infant was admitted to the hospital on April 20, 1967, with a history of constipation since birth. She had been given many laxatives, but without result, and her last bowel movement was fourteen days prior to admission.

The patient's parents, who came from Southeast Turkey, were first cousins. They were normal and healthy, showing no abnor-

* From Sami Ulus Children's Hospital, Ankara.
** Assistant Professor, Sami Ulus Children's Hospital.
*** Associate in Pediatrics, Sami Ulus Children's Hospital.
**** Assistant in Pediatrics, Sami Ulus Children's Hospital.

mality nor smallness of stature; however, the paternal grandfather had six digits on each hand. Other than this, family history was negative.

At the time of delivery the mother was 21, the father 25 years old. Pregnancy and delivery were normal and the baby full term. This was the second child of the family, the first having died of unknown causes at two months old. At six months the patient had head control, at seven months she could sit, but when admitted she could not yet walk and could say only a few words.

Physical examination on admission : Weight 8.2 kg (normal 11 kg),* length 68 cm (normal 80 cm), stem length 43 cm (normal 50.4 cm), length of upper extremity 22 cm (normal 31 cm), brachium 9 cm (normal 12 cm), head circumference 42 cm (normal 47 cm), chest circumference 45 cm (normal 48 cm), abdominal circumference 49 cm (normal 45 cm). The abdomen was distended, the anterior fontanel was losed and the hair smooth and fine. The patient had only five teeth, four of them upper and lower first molars, which erupted only two months prior to admission, and the fifth an upper lateral incisor, which erupted at the age of 12 months. The teeth were dysplastic, cylindriform and spaced irregularly. The lower incisors had not erupted, and the gingival mucosa had several irregular grooves. The buds of the lower incisors had not developed.

Figure 1 shows the general appearance of the child. The ears, nose and throat were normal, but the thorax was shortened. Physical examination of the lungs revealed negative results. Heart sounds and contours were normal, and no murmurs were heard. The liver and spleen were not palpable. Rectal examination showed that the anal sphincter was extremely tight, and an enormously large part of the rectum (about 1.5 to 2 cm from the anal ring) was filled with compact feces. Examination of the central nervous system gave a negative result, and the child did not show any sign of mental retardation. On the ulnar side of both hands there were six digits, all of which were short and stubby (Figure 2 - A) and both the fingernails and toenails were found to be hypoplastic and friable. A pes equinovarus deformity was observed on the right foot and a syndactyly between the third and fourth toes of the left foot (Figure 2 - B). The genitals were normal.

Laboratory examination : Blood picture, bone marrow and urine were normal : total serum protein 6.25 gm/100 ml, albumin

* Normal figures given according to the 50th percentile.

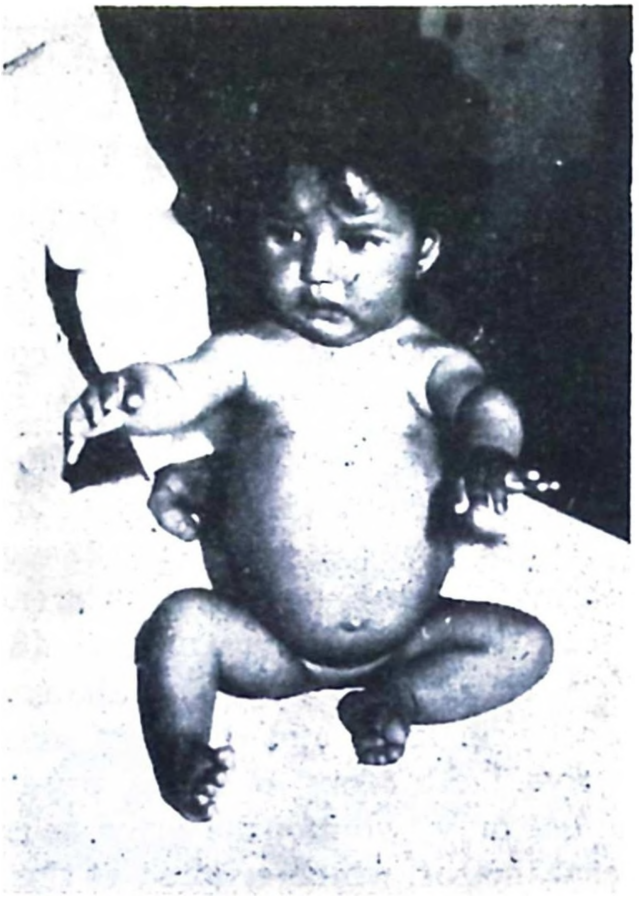
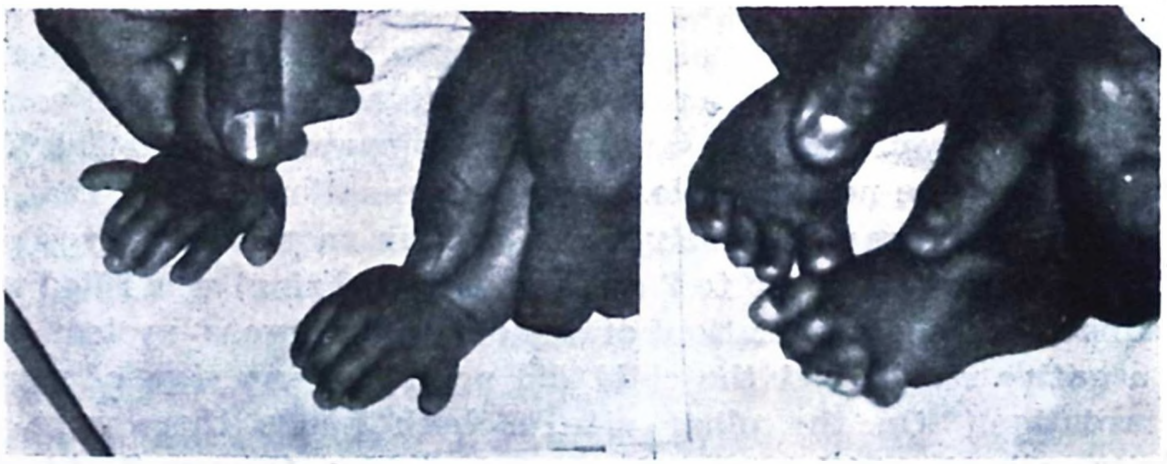


Figure 1. General appearance of child showing shortening of extremities and polydactyly.



Figures 2-A and 2-B. Polydactyly, Pes equinovarus, syndactyly.

4 gm/100 ml, globulin 2.25 gm/100 ml, blood sugar 100 mg/100 ml, serum calcium 9.5 mg/100 ml, serum phosphate 4.5 mg/100 ml, alkaline phosphatase one Bodansky unit, serum cholesterol 170 mg/100 ml. Tuberculin test was negative. Complement fixation reaction from both parents and child was negative. Chromosome studies were normal. The biopsy specimen taken from the rectal mucosa

showed the presence of parasympathetic ganglion cells in the myenteric plexus.

X-ray Studies : Roentgenogram of chest and heart was negative. Skull x-rays revealed normal bones and the size of the sella was normal. Posteroanterior view of the face showed no buds for the lower incisors (Figure 3). The long bones showed characteristic chondro-ectodermal dysplasia, an obvious shortening of the bones, more pronounced distally (Figure 4) with a definite widening of the head of the ulna and of the distal end of the radius. The hands revealed a shortening of the metacarpals and phalanges with fusions between the third and fourth metacarpals of both hands (synmetacarpalism). The spine was normal.

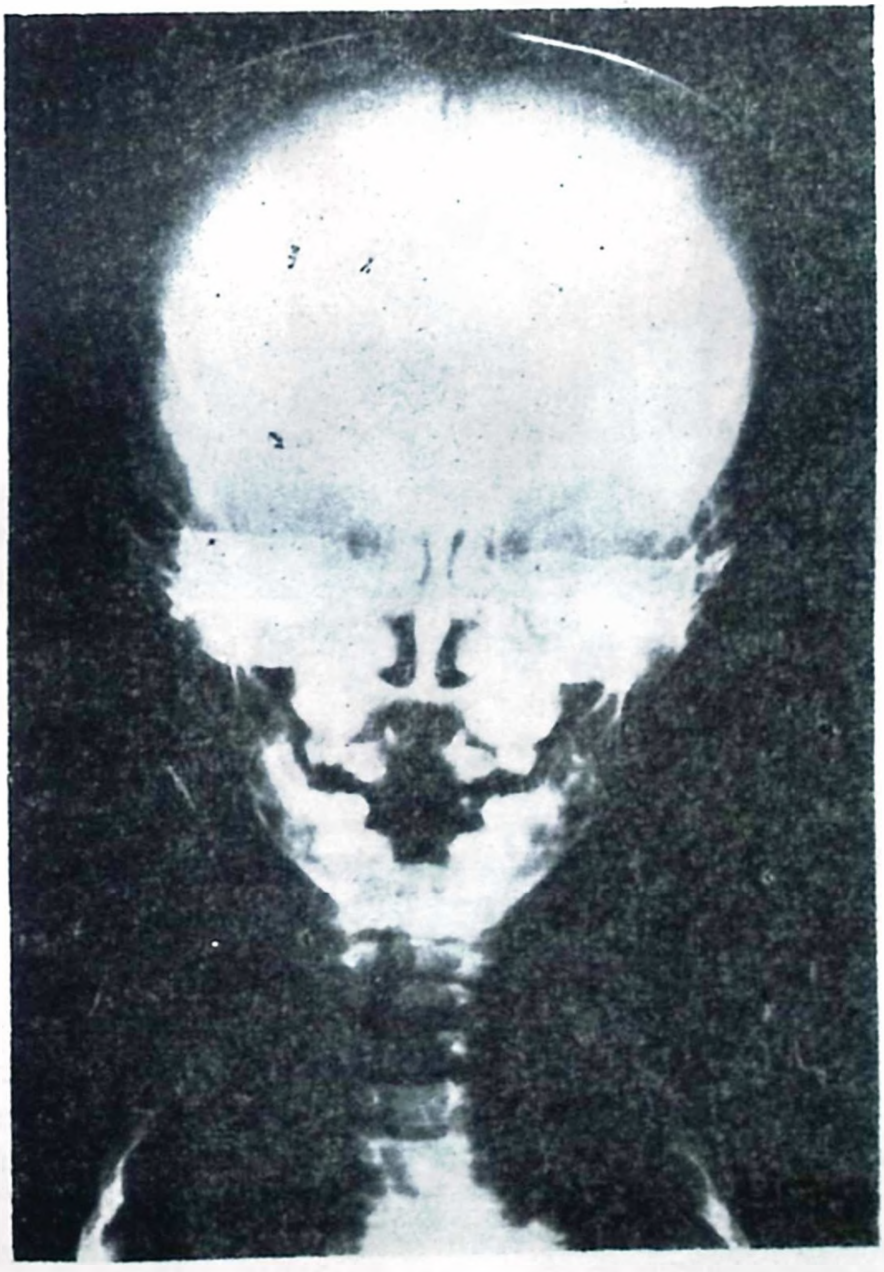


Figure 3. Buds of lower and upper incisors have not developed.

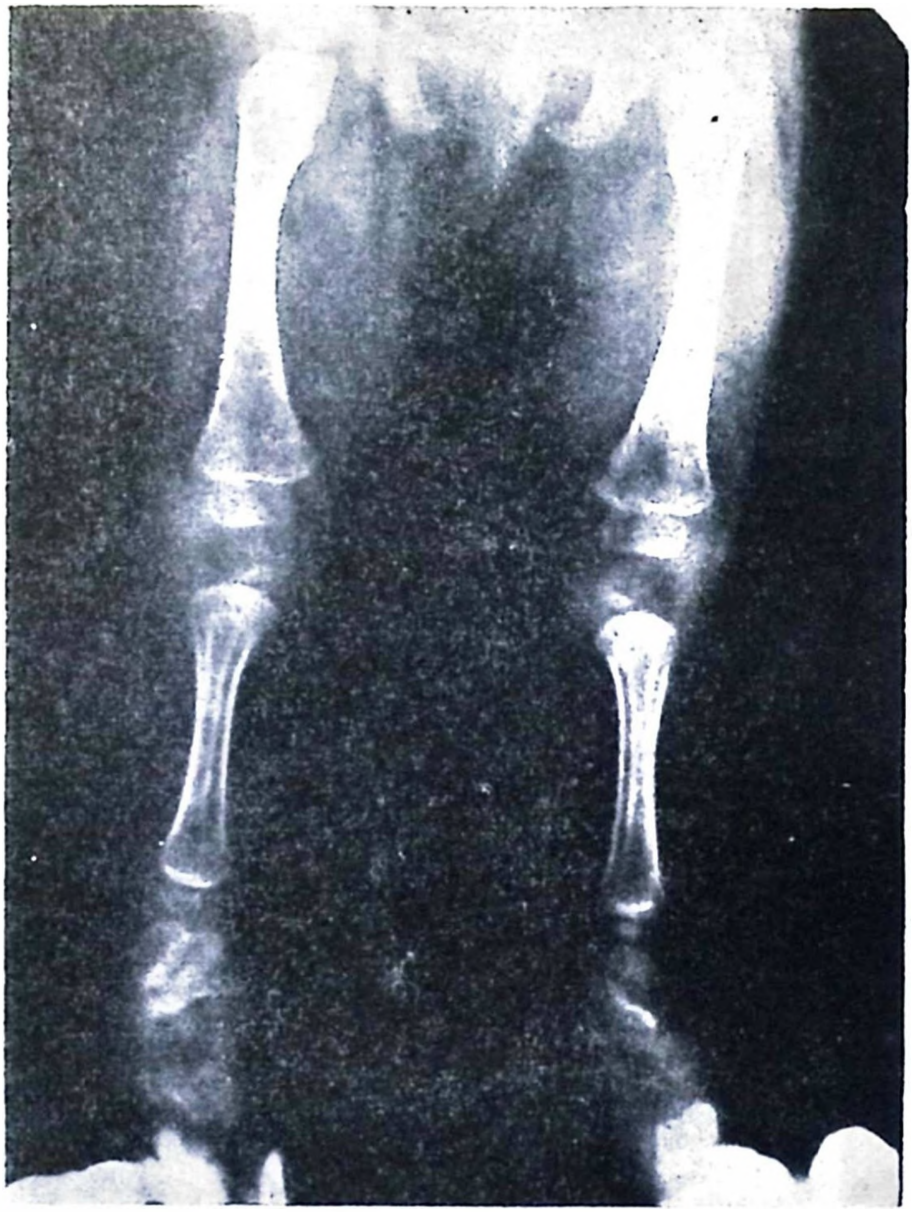


Figure 4. Shortening of distal bones.

Discussion

In our case, the constant symptoms of the syndrome were chondrodysplasia, polydactyly and ectodermal dysplasia.

Chondrodysplasia : The extremities were abnormally shortened at the distal end, which is the opposite of Morquio's disease and classical achondroplasia. The hands showed fusions between the third and fourth metacarpal bones. The humeri and femurs were bowed and both the head of the ulna and the distal end of the radius were enlarged. Caffey [8] regarded the appearance of the tibia as diagnostic : the proximal end of the tibial shafts was widened and pointed; the epiphyseal ossification center was hypoplastic and

medially displaced; the fibula showed disproportionate shortening. The spine was not affected.

Polydactyly : Both hands had six dixits on the ulnar side. The right foot showed a pes equinovarus deformity and the left foot a syndactyly between the third and fourth toes.

Ectodermal dysplasia : The nails, teeth and hair were affected, but skin and sweating were normal.

The fourth symptom, congenital heart disease, was not present in our patient. According to reports, approximately 60 per cent of E-vC cases have a congenital heart lesion;[1, 9] however, we feel this estimate to be higher than it should be, because in all reported instances the patient initially goes to the doctor with heart complications, not because of the other symptoms.

In reported cases, urogenital abnormalities (epispadias, hypospadias, hydrocolpos) and other anomalies such as coloboma of the iris, strabismus, cleft lip and palate, hepatomegaly and mental retardation are infrequent.[1, 4] Nabrady [10] indicated neuroectodermal disorders in this syndrome. Patients have been reported to have bronchial cartilage defects [11] and a defect in the respiratory system, such as malformation of the thoracic cage, lungs or bronchial tree, could be the cause of neonatal death.

Our patient showed none of the above mentioned abnormalities, but we did find megacolon due to partial anal stenosis, although we had not met this type of anomaly in the literature. Our patient came to us because of constipation, not because of any other symptom. A week after the anal ring incision, the patient's bowel movement was normal and the abdominal circumference was 45 cm.

The disorder has an autosomal and recessive character.[6, 12] Consanguinity was reported in many cases,[2, 4, 6, 12] especially by McKusick, who found that intermarriage was common in the families of the majority of his 52 patients, 25 of whom were male with nine over 19 years of age. The oldest, aged 58, was a carpenter, married, with seven children, two of whom showed the clinical features of E-vC syndrome, but were able to live independently. McKusick and his associates found the frequency of this syndrome to be two per 1000 living persons in the Amish community of Lancaster County, Pennsylvania.

The etiology is unknown, but the inheritable disorders of connective tissue and enzymatic defects have been suggested.[4, 12, 13]

Maroteaux and Lamy [13] marked an increase in the normal fraction of chondroitin sulfate A. A possible connection between this syndrome and mucoplysoccharidoses has been cited.[13] Chromosomal studies have showed no abnormalities.[1, 3]

The prognosis depends on cardiac and respiratory malformations, but patients who escape severe ones will not die in the newborn period or in early infancy and will reach middle age. The oldest reported patient is a 58-year-old male.

Summary

This further case of Ellis-van Creveld syndrome, with relative megacolon due to partial anal stenosis, is reported here to draw greater attention to this type of disorder.

REFERENCES

1. Ellis, R.W.B. and van Creveld, S : A syndrome characterized by ectodermal dysplasia and congenital morbus cordis : a report of 3 cases, Arch. Dis. Child. 15 : 65, 1940.
2. Dayer, L, : Le syndrome D'Ellis-van Creveld. Une forme de dysplasie chondroectodermique. Discussion des limites de l'affection, Bibloth. Paediatr. Karger, Bâle, New York, 1960, (Cited by ref. 4).
3. Ellis, R. W. B. and Andrew, J. D. : Chondroectodermal dysplasia, J. Bone and Joint Surg. 44 : 626, 1962.
4. McKusick, V. A., Egeland, J. A., Eldridge, R. and Krusen, D. E. : Dwarfism in the Amish I. the Ellis-van Creveld syndrome, Bull. J. Hopkins Hospital 115 : 306, 1964.
5. Reddy, D. V. and Madenlioğlu, M. : Chondroectodermal dysplasia (Ellis-van Creveld Syndrome), Clinical Pediatrics 6 : 51, 1967.
6. Weller, S. D. V. : Chondro-ectodermal dysplasia (Ellis-van Creveld syndrome), Proc. Roy. Soc. Med. 44 : 731, 1951.
7. Nelson, W.E. : Textbook of Pediatrics, Philadelphia, ed. 8, W. B. Saunders Co., 1964, p. 48.
8. Caffey, J. : Chondroectodermal dysplasia (Ellis-van Creveld disease), Amer. J. Roentgen. 68 : 875, 1952.
9. Smith, H. L. and Hand, A. M. : Chondroectodermal dysplasia (Ellis-van Creveld syndrome), Pediatrics 21 : 298, 1958.
10. Nabrady, J. : Ellis-van Creveld Syndrome and nervoectodermal injury, Ann. Paediat. 196: 18, 1961.
11. Moore, T. C. : Chondroectodermal dysplasia (Ellis-van Creveld syndrome) with bronchial malformation and neonatal tension lobar emphysema, J. Thorac. Cardiov. Surg. 46 : 1, 1963.
12. McKusick, V. A. : Genetic factors in cardiovascular disease. The four major types of cardiovascular disease, Med. Concepts Cardiovas. Dis. 28 : 535, 1959.
13. Maroteaux, P. and Lamy, M. : Hurle's disease, Morquio's disease, and related mucopolysaccharidoses, J. Pediat. 67 : 312, 1965.