

Case Report

Ellis-van Creveld syndrome. A case report

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Abstract Ellis-van Creveld syndrome is a very rare skeletal dysplasia. It has not been previously reported in Pakistan.

Key words

Ellis-van Creveld syndrome

Introduction

Ellis-van Creveld syndrome, also called chondroectodermal dysplasia, is a disease complex consisting of bilateral ulnar polydactyly, chondrodysplasia of long bones resulting in disproportionate dwarfism, hydrotic ectodermal dysplasia affecting principally the nails, teeth and congenital heart defects.¹

Case report

A 15-year-old girl reported to Dermatology OPD with complaints of short height, abnormal teeth and nails, short fingers and toes and an extra digit of each hand (she had extra digit of right hand surgically removed 2 years back). Her parents were first cousins. There was no family history of such disorder. On examination, she was 4 feet tall with normal facial and trunk development but small distal extremities especially fingers and toes. She had bilateral ulnar polydactyly (**Figure 1**), dystrophic nails (**Figures 2 and 3**), lumbar lordosis, genu

valgum. Her teeth were small, peg shaped; the palate was high arched, the sulcus between upper lip and gum was obliterated while the short upper lip was bound down by multiple frenula (**Figure 4**). Hair and skin was normal and she had normal sweating. The auscultatory findings of heart were normal. Rest of the systemic examination was normal. A clinical diagnosis of Ellis-van Creveld syndrome was made. Her renal and liver function tests, echocardiography and abdominal ultrasound were normal. Radiologically, her tibia was short with bilateral exostosis and hypoplastic lateral tibial condyles (**Figure 5**); x-ray hands showed short middle and terminal phalanges, ulnar polydactyly and fused hamate and capitate bones (**Figure 6**). X-ray feet also showed short phalanges while X-ray spine was normal. These findings supported our initial diagnosis.

Discussion

Ellis-van Creveld syndrome is a skeletal dysplasia, first described in 1940 by Richard W. Ellis and Simon van Creveld who coined the term "chondro-ectodermal dysplasia".² It is a genetic disorder transmitted in autosomal recessive mode.² The EVC gene

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Figure 1 Ulnar polydactyly in left hand.



Figure 2 Dystrophic fingernails.



Figure 3 Dystrophic toenails.



Figure 4 Obliterated gingivolabial sulcus and peg-shaped teeth.



Figure 5 Hypoplastic lateral tibial condyles.



Figure 6 short middle and terminal phalanges, ulnar polydactyly and fused hamate and capitate bones

has been mapped to chromosome band 4p16 whose mutations were identified in these

patients. Mutations in a second gene, called EVC2 also give rise to the same phenotype of the syndrome.³ The parents are of normal stature and both are carriers of the mutation. There is a 25% chance of further

pregnancies resulting in a child with the same condition.² In the US, the frequency is 1 case per 60,000 live births and is equal in males and females.³

This syndrome is a tetrad with chondrodysplasia,² polydactyly, ectodermal involvement and congenital heart defects.⁴ Chondrodystrophy means disproportionate dwarfism i.e. normal trunk, with symmetrical shortening of distal extremities (average adult height being 109-155 cm).³ Patients also have lumbar lordosis and genu valgum.⁵ Polydactyly is a constant finding, usually bilateral and on the ulnar side.⁵ It is observed in the hands in most cases but in the feet in only 10% of cases.³ Ectodermal dysplasia (observed in up to 93% of cases) includes hypoplastic, dystrophic, friable or even absent nails. Tooth involvement may include neonatal teeth, partial anodontia, small teeth, and delayed eruption. Hair may occasionally be sparse in these patients.³ Cardiac abnormalities (in 60 %) include a common atrium (40 %), AV canal, ventricular septal defect, atrial septal defect, and patent ductus arteriosus. The cardiac anomaly is the major cause of shortened life expectancy.³ Other anomalies which may be present include musculoskeletal anomalies i.e. low-set shoulders, a narrow thorax frequently leading to respiratory difficulties, broad hands and feet, and sausage-shaped fingers.³ Teeth are small, defective, may be peg-shaped. The mucobuccal fold may be absent while the short upper lip is bound down by multiple frenula.⁵ Nails are small, thinned, short and ridged. Hair and sweating are usually normal.⁵ Occasionally, there are genitourinary anomalies i.e. hypospadias, epispadias, hypoplastic penis, cryptorchidism, vulvar atresia, focal renal

tubular dilation in medullary region, nephrocalcinosis, renal agenesis, and megaureters.³ Liver and CNS abnormalities may also be present.⁵

Our patient had all these three cardinal features of chondrodystrophy, polydactyly and ectodermal dysplasia. Cardiac anomalies were absent in our patient which is the cause of normal life expectancy in her. In the prenatal period, intrauterine growth retardation, skeletal malformations, and cardiac defects can be predicted on ultrasound images.³ 50% of patients die in infancy due to thoracic dysplasia leading to respiratory insufficiency and cardiac anomalies. Patients who survive infancy have a normal life expectancy.^{3,5}

The lack of local literature about this condition means either non occurrence or clinician's ignorance about this condition. It is necessary to identify this disease at its early stage in order to refer the case to a cardiologist, cardiac surgeon and orthopedic surgeon for corrective surgeries¹ and so render prompt treatment.

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