

Ellis-van Creveld syndrome (chondro-ectodermal dysplasia) in two siblings

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Abstract

Two male siblings aged 12 and 15 years (Fig 1) presented with growth retardation, limb abnormalities and, defective teeth and nail since childhood. There was no history of similar defects in other family members. On examination their height was 114cm and 130 cm (expected height; 169 cm and 150 cm) respectively. There were hypoplastic and dystrophic nails, and microdontia and hypodontia on dental examination. There were post-axial polydactyly in both the hands and left foot. Skeletal survey revealed short forearms, legs & distal phalanges (Acromesomelia). Cardiac or other organ system defects were not noticed on clinical examination or echocardiogram. Clinical picture was suggestive of a diagnosis of Chondroectodermal dysplasia (Ellis van Creveld syndrome). These cases have been reported for their rarity and for the presence of some rare features of this syndrome.

Ectodermal dysplasias are a group of hereditary diseases which involve one or more structures originating from the ectoderm such as hair, teeth, nail and sweat glands. They are rare and many of them may also involve other structures like bones. Chondroectodermal dysplasia (Ellis van Creveld syndrome) is a unique among them that may involve skeletal system, heart and other organs along with structures like nail and teeth. It is a very rare syndrome with frequency of 1 per 60,000 live births in the general population and among persons from the Old Order Amish, 5 per 1000 live births¹. Apart from some of the usual features, we also found some rarer manifestations of this syndrome in our cases.

Case report

Two male siblings aged 12 and 15 years (Fig 1) were presented with growth retardation, limb abnormalities and, defective teeth and nail since childhood. Third sibling (male) aged 8 years was normal. There was no history of similar defects in other family members.

On examination their height was 114cm and 130 cm (expected height; 169 cm and 150 cm) respectively. Both

were having hypoplastic and dystrophic nails involving all finger nails and toe nails (Fig 2, 3). There were post-axial (ulnar) polydactyly in both the hands and left foot (Fig 3). Skeletal survey revealed short forearms, legs & distal phalanges (Acromesomelia). Bilateral genu valgum were also present. There was microdontia and hypodontia on dental examination (Fig 4). Cardiac or other organ system defects were not noticed on clinical examination or echocardiogram.

Presence of ectodermal dysplasia (nail and dental defects), chondrodystrophy and polydactyly are characteristic features of Chondroectodermal dysplasia (Ellis van Creveld syndrome). A cardiac defect, the fourth feature of this syndrome was not seen in these patients. This can occur in up to half of all patients¹. Other condition considered as a differential diagnosis was Patau's syndrome which manifests with Polydactyly and congenital heart defects². Presence of severe mental deficiency, Holoprosencephaly, rocker-bottom feet, facial clefting and other features distinguishes it from Ellis van Creveld syndrome.

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Fig 1: Case 1 and 2 showing polydactyly, short stature and genu valgum



Fig 2: Hypoplastic and dystrophic finger nails

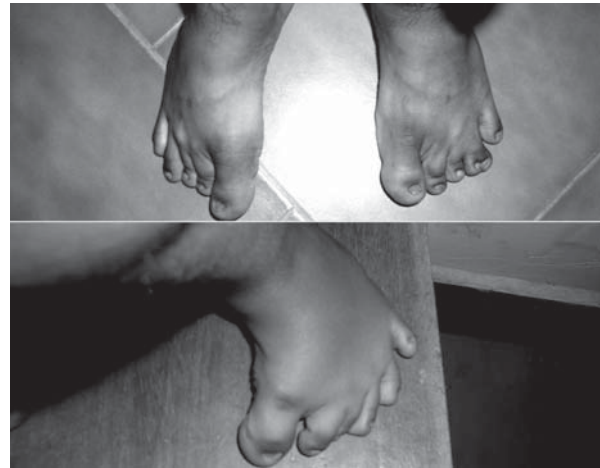


Fig 3: Left foot with Polydactyly

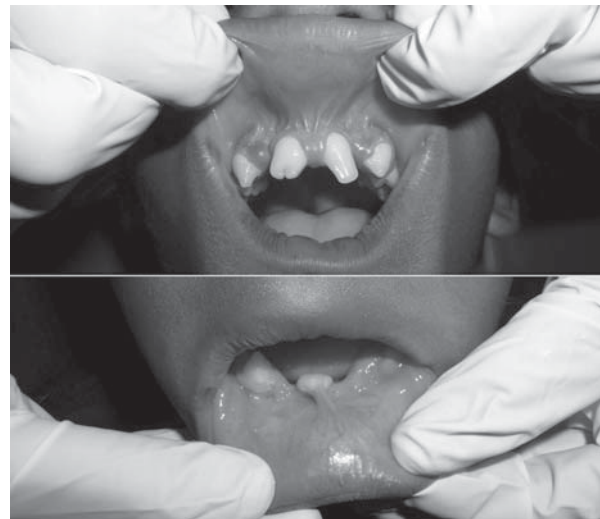


Fig 4: Hypodontia and Microdontia

Discussion

Richard W.B. Ellis of Edinburgh and Simon van Creveld of Amsterdam met in a train compartment in the late 1930s. They discovered that both had a patient with disproportionate dwarfism, polydactyly, ectodermal dysplasia, small chest, and a high frequency of congenital heart defects. In 1940, they termed this syndrome as Chondro-ectodermal dysplasia¹.

Autosomal recessive inheritance has been noticed and the EVC gene is located on the chromosome 4p16¹. Ectodermal dysplasias are rare heterogeneous group of disorders in which there are defects in the hair, tooth, nail and sweat gland. Classification (Freire - Maia and Pinheiro) is based on the type of ectodermal structures affected². Accordingly '1' indicates hair defect, '2' indicates tooth defect, '3' indicates nail defect and '4' indicates sweating defect². Ellis van Creveld syndrome

(Chondro-ectodermal dysplasia) can be categorized in to the sub group 1 - 2- 3.

Ellis van Creveld syndrome is characterized by a tetrad of ectodermal dysplasia, chondrodysplasia, polydactyly and cardiac defects. Ectodermal dysplasia in EVC has been noticed in up to 93% of cases. It is characterized by nail, dental and in some patients by the hair defects. Nails are hypoplastic and dystrophic but also there may be anonychia. Dental defects include neonatal teeth, anodontia, microdontia and enamel hypoplasia. Hairs may occasionally be sparse. Disproportionate dwarfism and progressive distal limb shortening are the results of Chondrodysplasia. Polydactyly, which is the most constant finding, is bilateral and postaxial. Hands are affected in most but feet in only 10%¹. Our both cases had polydactyly of their left foot. Cardiac anomalies such

as common atrium and ASD are seen in only about 60% of cases³. Other anomalies involving Musculoskeletal, Urogenital and central nervous system may also be seen. Management primarily includes dental care, nail care, and surgical correction of orthopedic, cardiac and other defects. Partial dentures, Implants and prosthetic rehabilitation are done to correct dental defects. Nail care to prevent nail and paronychia infections, and acrylic nail prosthesis to improve cosmetic appearance are advised. Genetic counseling is important since the recurrence risk in patient's siblings is 25%.

Both cases reported here were essentially similar in their clinical presentation. Both did not have any cardiac anomalies. They had a rare polydactyly of foot. It's a rare syndrome and apart from these cases there are a few other case reports in Indian literature too^{3,4}. Multidisciplinary approaches including orthopedic surgeon, dental surgeon, dermatologist, cardio-thoracic surgeon, pediatrician and others to manage various defects and improve appearance of these patients are the services that health care workers should provide.

References

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