

Ellis Van Creveld Syndrome

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Abstract

Ellis-van Creveld syndrome (EVC) or chondroectodermal dysplasia is a rare autosomal recessive disorder. It is a tetrad of chondrodysplasia, ectodermal dysplasia, polydactyly, and congenital heart disease. In several case reports, dysplasia involving other organs has also been identified. The exact prevalence is unknown, but the syndrome seems more common among the Amish community. Many Indian cases have also been reported.

This report describes a classical case of EVC syndrome in a 22 year old woman of Indian origin born of a consanguinous marriage. The patient had chondrodysplasia of tubular bones resulting in disproportionate dwarfism, postaxial polydactyly, severely dystrophic nails, partially absent teeth, pectus excavatum with narrow chest, knock knees and AV canal defect.

Introduction

Chondroectodermal dysplasia is a rare mesenchymal - ectodermal dysplasia first described in 1940 by Richard W.B. Ellis and Simon van Creveld now known as Ellis van Creveld syndrome. It is also known as mesectodermal dysplasia. Majority of cases were characteristically seen in one particular inbred population from the Amish community of Lancaster County, Pennsylvania, U.S.A.

Ellis-van Creveld (EVC) syndrome is thought to be due to mutation in EVC and EVC-2 genes located in chromosome 4p16, characterized by short limbed dwarfism, post axial polydactyly, dysplastic teeth and nails, and cardiac defects.¹⁻³ Other features include partial harelip and multiple frenulae in lips; short ribs and narrow chest; genital abnormalities like epispadias, hypospadias and cryptorchidism; low iliac wings with spur-like projections at acetabula and genu valgum. Although most patients have normal intelligence, occasional central nervous system anomalies or mental retardation have been reported.

Case History

A 22 years old young female presented to us with chest pain, progressive exertional dyspnea and easy fatigability of two months duration with no h/o orthopnea or PND. She had no significant illness in the past except that she was born with short limbs and six fingers in each hand and had frequent respiratory tract infections. She had short stature which was apparent from one year of age. Her school performance was average. She attained menarche at the age of seventeen years and having normal menstrual cycles once in 2 months since then with no h/o any other bleeding tendencies, no passage of worms in stools. She was a vegetarian by diet. Her younger brother also, was having short stature. Her four other siblings and parents had normal height.

On examination, she was found to have a short stature with a height of 126 cm (Fig. 1). Examination of hands revealed six digits in each (Figs. 2, 3). The finger and toe nails were small and brittle (Fig. 2). Oral cavity examination revealed absent incisors

and the rest were natal teeth (Fig. 4). The patient had knock knees with pectus excavatum. The patient had severe pallor.

Examination of cardio-respiratory system revealed narrow chest and had a prominent diastolic thrill at the apex, a loud S1



Fig. 1 : Short stature & genu valgum

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Fig. 2 : Hands showing six digits and small and brittle nails



Fig. 3 : Post axial Polydactyly



Fig. 4 : Prenatal teeth & absent incisors

with a low-pitched grade 4/6 mid-diastolic rumbling murmur and a pansystolic murmur in the mitral area. The S2 was widely split, fixed with a loud P2. There was a grade 3/6 ejection systolic murmur over the left second intercostal space. Other systems were normal.

On investigation, the hematological parameters were as follows: hemoglobin 5.3 gm/dl, TLC 5700/cmm, platelet count 4.4 lacs/cmm, with peripheral smear having dimorphic blood picture and 4% band forms. Bone marrow showed predominant megaloblastic picture with hypersegmented neutrophils. The cause for anemia was probably nutritional. Serum vit B12 assay was not done because of financial constraints. Liver and kidney function tests were within normal limits. Thyroid function tests were normal.

Skeletal radiology of upper limb revealed short metacarpals, ulnar thick 5th metacarpal, ulnar polydactyly and subluxation of right inferior radioulnar joint (Fig. 3). X-ray skull showed craniofacial disproportion (Fig. 5). Radiological examination of the lower limb revealed depressed and flattened lateral



Fig. 5 : Cranio-facial disproportion



Fig. 6 : Cardiomegaly

compartment of upper end of tibia on both sides giving rise to genu valgum. Chest X-ray showed cardiomegaly (Fig. 6). Echocardiography showed cardiomegaly and AV canal defect (Figs. 7, 8). USG abdomen was normal with incidental finding of a single, small right ovarian cyst. Chromosomal analysis done was normal.

She was transfused with two pints of whole blood and was started on haematinics which was continued after discharge and she was being followed up on OPD basis. Recent follow up showed that the patient's symptoms have reduced with increased tolerance for work. Her recent hemoglobin level was 10.8 gm%. She was continued on haematinics and multivitamins.

Discussion

This syndrome was first described by Richard W. B. Ellis of Edinburgh and Simon Van Creveld of Amsterdam in 1940. In general population the incidence is reported as one per 60,000 live-births with an increased incidence of five per 1000 live-births in old Amish population of Pennsylvania. The frequency of carriers in this population may be as high as 13 percent. The sex predilection of this syndrome is the same in males and females.

