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Case Report

Ellis–Van Creveld (EVC) syndrome: A rare case report

Shagufta Rather, Kurat Sajjad, Nazim Malik

Department of Dermatology, Government Medical College, Srinagar and Jammu, Jammu and Kashmir, India

Corresponding author: Shagufta Rather, MD, E-mail: shagufta.giri@gmail.com

ABSTRACT

Ellis–Van Creveld (EVC) syndrome is a rare autosomal recessive ciliopathic disorder characterized clinically by short stature, bilateral postaxial polydactyly, bone abnormalities, retarded growth, ectodermal, and congenital heart defects. The disorder arises from the mutation of the EVC and EVC 2 genes located on the 4p16 chromosome. More than 300 cases of EVC have been recorded in the literature, with an estimated prevalence of the condition being 7 per 100,000. Herein, we present the rare case of a twelve-year-old male child diagnosed with EVC syndrome, based on oral manifestations, polydactyly, and nail dystrophy.

Key words: Ellis–Van Creveld syndrome, Polydactyly, Chondroectodermal dysplasia, Congenital heart defects

INTRODUCTION

Ellis–Van Creveld syndrome (EVCS), also known as chondroectodermal dysplasia or mesoectodermal dysplasia, was first described by Richard W.B. Ellis and Simon Van Creveld in 1940 [1]. It is a complex and rare autosomal recessive genetic disorder with less than 25 cases reported in India and 300 cases across the world [2].

Mutations in the EVC and EVC 2 genes present in a head-to-head configuration on chromosome 4p16 play an important role in the development of skeletal and endochondral abnormalities of disease [3]. The affected individuals of EVC with mutations in either the EVC or EVC 2 genes have been shown to be phenotypically indistinguishable. Besides, the same mutation is responsible for ciliary dysfunction in EVC syndrome [3]. EVCS has a multisystemic involvement ranging from heart and bone abnormalities to stomatologic ones [4]. The tissues involved in this disorder (particularly nails, long bones of the skeleton and teeth) are covered under the term *chondroectodermal dysplasia* while mesoectodermal dysplasia includes the associated cardiac disorders [4]. Herein, we report a twelve-year-old child presenting at the dermatology outpatient department with characteristic features of EVCS.

CASE REPORT

A twelve-year-old male child of Indian origin, born out of a consanguineous marriage, was brought by the parents to the outpatient department of dermatology with distinctive clinical features, the chief complaints of malformation of the teeth in the upper and lower jaws and deformities of the hands, feet, and nails. There was no history of associated fever, chest pain, or recurrent respiratory tract infections. He was second in birth order, born through normal vaginal delivery at full-term without complications and with a normal birth weight. Her psychosomatic and mental developmental were within normal limits. There was no significant family history, and the other sibling was normal. He was immunized as per the national immunization schedule.

General examination revealed the patient to be of short stature with 117 cm height, with short lower limbs, and a narrow chest (thoracic dysplasia). There was bilateral extra-axial polydactyly of the hands along with hypoplastic and dystrophic nails of both hands and feet. The hair was normal. His hands and feet were broad and square with short fingers and toes. Lower limbs were deformed with outward bending of the knees (*genu valgum*), and there was lateral deviation of the great

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toe with medial deviation of the first metatarsal bone (hallux valgus deformity) (Figs. 1a and 1b).

The examination of intraoral soft tissue revealed a short frenulum and positive labiogingival adherence. Hard tissue examination revealed hypoplasia of the enamel, conical teeth, and hypodontia. Multiple teeth were missing and various malformed teeth existed in the maxillary as well as mandibular arches. The upper lip was normally developed, and the presence of neonatal teeth or any kind of premature eruption was not seen (Figs. 1c – 1e). His neurological, cardiac, opthalmological, and auditory assessments were normal.

Laboratory investigations, including a hemogram, liver function tests, renal function tests, urine analysis, electrocardiogram, echocardiogram, and abdomen ultrasound were normal. Based on the clinical and radiological findings, we diagnosed the case as Ellis–Van Creveld syndrome (Figs. 2a – 2e). Genetic analysis was not conducted as it was unavailable in our hospital.

Parents were counseled about the genetic basis of the disease. The patient was advised to return for regular follow-up with the dentist, orthopedic surgeon, and physiotherapist for further management.

DISCUSSION

In 1933, McIntosh reported the first case of EVC syndrome, yet in 1940, Richard W.B. Ellis of Edinburgh and Simon Van Creveld of Amsterdam first described this condition as EVC syndrome [1]. It is also known as chondroectodermal dysplasia and mesoectodermal dysplasia [2]. EVC syndrome is one of the ciliopathies, and the ciliary dysfunction has been linked to a mutation in two adjacent genes, EVC and EVC2 [3].

There is no sex predilection reported and parental consanguinity has been reported in 30% of cases, which was present in our case. The tetrad of disproportionate dwarfism, bilateral postaxial polydactyly, ectodermal dysplasia, and congenital heart malformation is used to describe the characteristic signs and symptoms [4,5]. The typical syndromic facial characteristics of coarse face, conical anterior teeth, dental agenesis, multiple small extra-labial, non-midline frenula with a high arched palate and a large maxillary labial frenulum were present in our case. Other abnormalities observed comprised short limb dwarfism, a dysplastic genu valgum, an elongated trunk with severe lordosis, pectus excavatum, hypoplastic fingernails and toenails, wide, noticeably malformed hands and feet, and bimanual hexadactyly on the ulnar side of her wrists, bearing resemblance to other studies in the past [5,6]. However, anomalies of other body organs reported in these patients such as strabismus, pulmonary malformations, congenital heart defects, hematologic abnormalities, genitourinary abnormalities, and intellectual disability were absent in our patient [7,8].

EVC syndrome may be diagnosed during pregnancy, prenatally or soon after delivery [4,5]. However, the diagnosis was delayed in our case as the patient belonged to a remote hilly area with no access to

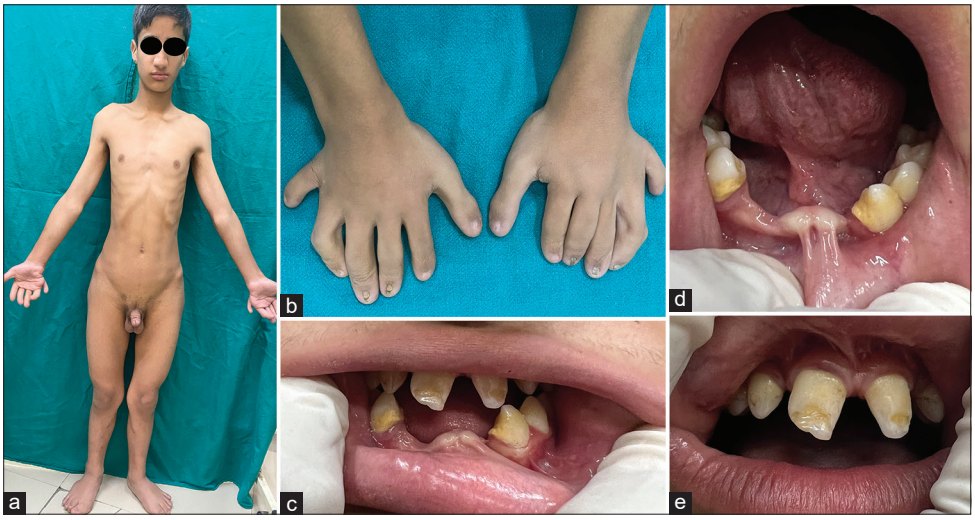

Figure 1: (a) 12-year-old boy showing chondrodysplasia, a narrow chest, deformities of the limbs, short forearms and lower limbs, bent lower limbs, and hallux valgus deformity of the great toes, with normal skin and hair. (b) Showing polydactyly of both hands and shortening of the digits in both hands and feet. Also, nail hypoplasia is seen. (c) Oral cavity showing malocclusion, hypodontia, absent maxillary lateral incisors and mandibular incisors. (d) Oral cavity showing malocclusion, hypodontia, absent maxillary lateral incisors and mandibular incisors. (e) Large maxillary labial frenum, multiple accessory labial frenula, mandibular anterior ridge defect, and crossbite.

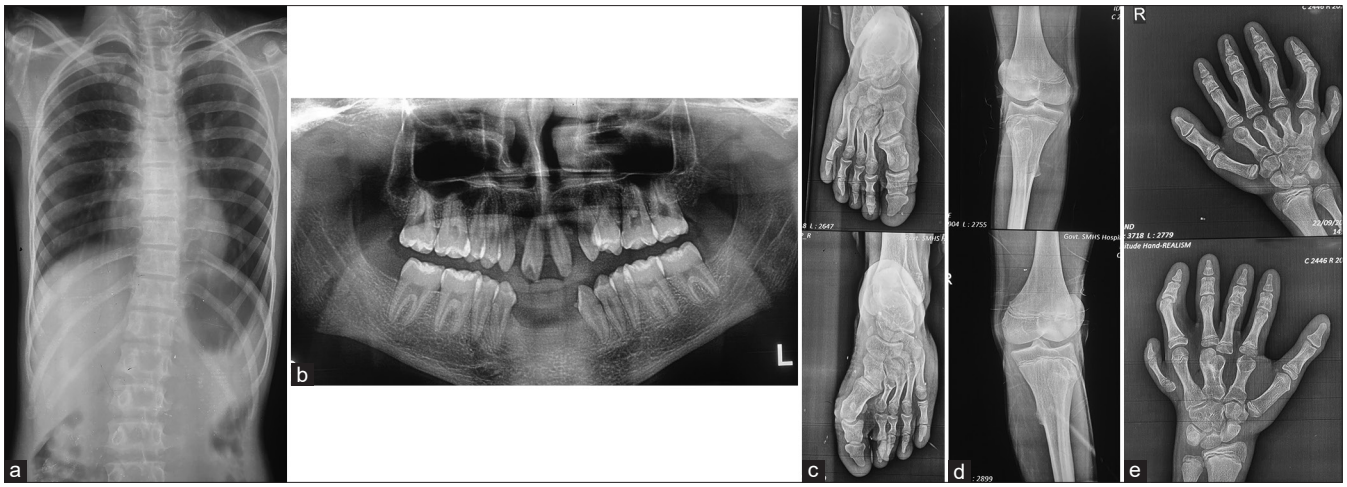

Figure 2: (a) PA view of the chest X-ray showing a narrow chest and short ribs. (b) Panoramic radiograph showing agenesis of the maxillary lateral incisors, mandibular incisors, and all four third molars. (c) X-rays of the hands showing postaxial polydactyly, short middle, and distal phalanges with cone-shaped epiphyses and carpal fusion. (d) X-rays of the feet showing bilateral hallux valgus deformity of the great toes. (e) X-rays of the legs showing lateral tibial metaphysis slanted giving rise to a genu valgum deformity.

healthcare. Ultrasonography after the 18th gestation week is used to make the prenatal diagnosis, which reveals cardiac defects, a narrow thorax, shortening of the long bones, and hexadactyly of hands and feet. At birth, clinical examination aided by an X-ray of bones may be used to diagnose the disease. The DNA mapping technique based on homozygosity for a mutation in the EVC and EVC2 genes may be used to make a reliable diagnosis [4-8]. In the present case, the diagnosis was made based on typical oral manifestations and polydactyly of the hands combined with nail dystrophy.

Since no definite cure exists for EVC syndrome, treatment is mainly symptomatic. Early dental care should be sought out by patients to restore the missing teeth, and correction of other deformities. Parental dental health education should cover oral hygiene advice, diet counseling, and physiotherapy [5-8].

CONCLUSION

EVC syndrome is a rare autosomal recessive disorder. Diagnosis is mainly clinical in cases with delayed presentations. Besides having some constant clinical features, there may be other variable components present in different patients. Awareness among clinicians about this rare entity is important, and a multidisciplinary treatment approach is recommended.

Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

REFERENCES

1. Ellis RW, van Creveld S. A syndrome characterized bvecto-dermal dysplasia. polydactyly, chondrodysplasia, and congenital morbus cordis. Arch Dis Child. 1940;15:65-84.
2. Ghosh S, Setty S, Sivakumar A, Pai KM. Report of a new syndrome: Focus on differential diagnosis and review of Ellis-van Creveld, Curry-Hall, acrofacial dysostosis, and orofacial digital syndromes. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2007;103:670-6.
3. Ibarra-Ramirez M, Campos-Acevedo LD, Lugo-Trampe J, Martínez-Garza LE, Martinez-Glez V, Valencia-Benitez M, et al. Phenotypic variation in patients with homozygous c.1678G>T mutation in EVC gene: Report of two Mexican families with Ellis–Van Creveld syndrome. Am J Case Rep. 2017;18:1325-29.
4. Shetty P, Shetty D, Priyadarshana PS, Bhat S. A rare case report of Ellis Van Creveld syndrome in an Indian patient and literature review. J Oral Biol Craniofac Res. 2015;5:98-101.
5. Ghassemi M, Goodarzi A, Seirafianpour F, Mozafarpoor S, Ziaeifar E. Rare clinical features of the Ellis van Creveld syndrome: A case report and literature review. Dermatol Ther. 2021;34:e14664.
6. Winter GB, Geddes M. Oral manifestations of chondroectodermal dysplasia (Ellis-van Creveld syndrome): Report of a case. Br Dent J. 1967;122:103-7.
7. Ghosh B, Sahai I, Agrawal G, Acharya S, Christopher J. Ellis-Van Creveld syndrome: a rare case report of an Indian child with rare cardiac anomalies and normal intelligence. Cureus. 2022;14:e29846.
8. Katsouras CS, Thomadakis C, Michalis LK. Cardiac Ellis–Van Creveld syndrome. Int J Cardiol. 2003;87:315-6.

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