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Case Report

Ellis-Van Creveld syndrome: a case report

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ABSTRACT

Ellis-Van Creveled (EVC) syndrome, also called chondroectodermal dysplasia, is a rare genetic disorder with autosomal recessive type of inheritance with 25% risk of transmission to subsequent pregnancies. The syndrome is due to mutation of EVC1 and EVC2 genes on chromosome 4p16, characterized by acromesomalic dwarfism, bilateral postaxial polydactyly, ectodermal dysplasia affecting nails, teeth and congenital heart malformation. One third of the patients die at an early age or during infancy from cardio respiratory problems. The birth prevalence of EVC syndrome is 1/5000 live births in Amish population and 7/1,000,000 live birth in non-Amish population. There are a very few case reports in the literature. We reported a case of 14-year male presenting with typical features of this syndrome.

Keywords: Ellis Van Creveled syndrome, Ectodermal dysplasia, Postaxial polydactyly, Acromesomalic dwarfism

INTRODUCTION

EVC syndrome, also known as chondroectodermal and mesoectodermal dysplasia, is a rare congenital disorder. It follows autosomal recessive type of inheritance. Richard WB Ellis and Simon Van Creveled described EVC in the year 1940. EVC syndrome results from the genetic mutation of two genes, EVC1 and EVC2. Both of the genes are located on locus 16 of the short arm of chromosome 4 (4p16) in a head-to-head configuration.[1-3] The birth prevalence of EVC syndrome is 1/5000 live births in Amish population and 7/1,000,000 live birth in non-Amish population.[4-6] Around 300 cases of EVC are reported throughout the world, around 25 cases have been reported from India.

This syndrome is characterised by tetrad. It manifests with the following spectrum of clinical features: acromesomalic dysplasia, post axial polydactyly, ectodermal dysplasia affecting nails, teeth and congenital heart malformations. Disproportionate dwarfism occurs as a result of acromesomalic chondrodysplasia of long tubular bones, where the severity of short limbs increases from proximal to distal portions. Bilateral post axial polydactyly of hands and feet is observed, where the supernumary finger is usually situated on the ulnar side. Hidrotic ectodermal dysplasia along with dysplastic small nails, sparse thin hair and other oral manifestations are characteristic. Congenital heart malformations manifest in about 50-60% of the cases. EVC belongs to short rib polydactyly group (SRP) of syndromes. It is characterized by short ribs, associated with cardio respiratory problems which is primary cause for reduced life expectancy in these patients.[1]

CASE REPORT

A 14-year-old male presented to orthopaedic outpatient department (OPD) with a presenting complaint of progressive deformity of bilateral lower limbs which was present since birth, leading to difficulty in walking and running. Thereafter the patient was referred to the

department of paediatrics for thorough medical examination in view of corrective osteotomy surgery. A detailed history was elicited. Birth history revealed that, the patient was delivered at term through LSCS. He is the third child born out of a third-degree consanguineous marriage to clinically normal parents, with birth weight of 2.7 kg. His mother gives history of NICU admission during first week of birth; however, does not remember the reason for admission in NICU.

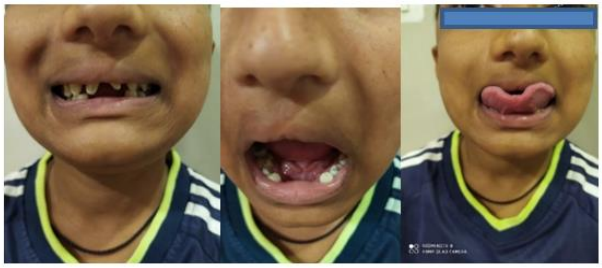

Figure 1: Oral view showing dysmorphic teeth and ankyloglossia.

On general physical examination the patient had a short stature of 125 cm (<3rd percentile) and weighed 25.7 kg (<3rd percentile) with a head circumference of 47 cm (normocephalic). The patient had thin hair which was normally distributed over scalp and sparsely distributed over the eyebrows. Hypertelorism was present with low set ears. Oro maxillary examination revealed deficient mucobuccal fold, shallow labial sulci, ankyloglossia, dysmorphic conical teeth with irregular spacing as shown in (Figure 1). The patient had bilateral post axial polydactyly of hands and could not make a complete tight fist. The finger and toe nails were hypoplastic as shown in Figure 2.

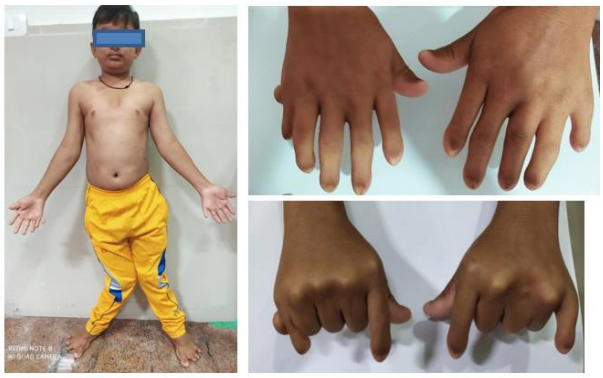

Figure 2: Frontal view of the patient with disproportionate short stature, wide gap between first and second toes, polydactyly of hands and foot with dystrophic nails.

A wide gap was present between both left and right greater toe and second toe. Family history revealed that one of his elder sisters also had polydactyly and similar oral and dental findings. Bilateral cubitus valgus and genu valgus deformities were present along with middle and distal segment shortening, leading to acromesomelic type of short limb skeletal dysplasia. Skeletal radiograph showing genu valgum is depicted in (Figure 3).

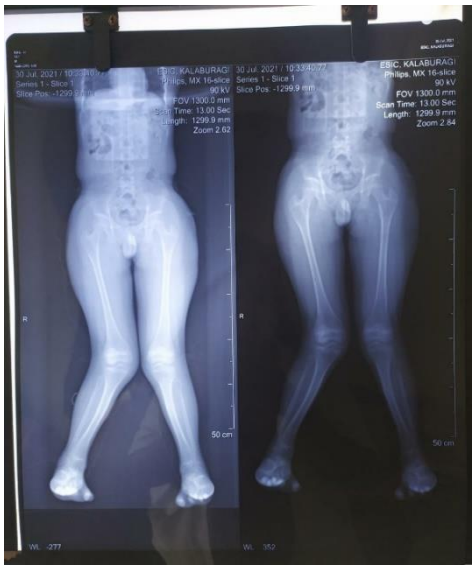

Figure 3: Skeletal radiograph showing genu valgum.

Spine and genital examination were normal. There was no cryptorchidism, epispadiasis or hypospadiasis. Other system examinations were normal with a normal cognitive and psychomotor development. Intellectual abilities were within normal limits. Abdominal and renal ultrasound, ECG and echocardiogram reported normal study. Biochemical, hormonal, haematological and routine urine tests were within normal range.

DISCUSSION

EVC is a syndrome with variable phenotype affecting multiple organs.[1,7] Hence diagnosis can be made as early as 18th week of gestation by ultrasonography when increased translucency is evident.[4,5,7,8] The association of several structural fetal defects in the late 1st trimester, including narrow thorax, short and bowed long bones, rounded metaphysis, post axial polydactyly and cardiac defects may suggest diagnosis of EVC.[9] However the diagnosis can be still made later after birth by clinical examination. The definitive diagnosis of EVC syndrome is made through direct gene sequencing which is based on the mutation of EVC1 and/or EVC2 genes. However the genetic mutations are seldom required for the clinical diagnosis as gene mutations is positive in only 2/3rd of patients.[7] Due to the lack of availability of genetic studies, the diagnosis was made clinically, based on observation of symptoms which were supported by radiological evidences and laboratory tests. Differential diagnosis includes other short rib polydactyly syndromes like Weyers acrodental dysostosis (Curry-Hall syndrome), asphyxiating thoracic dystrophy (Jeune syndrome), achondroplasia, chondroplasia punctata, orofaciodigital syndromes and Morquio's syndrome.[5,7-9] The clinical spectrum of EVC syndrome is not precisely demarcated. There are no specific or constant features to

define the syndrome. Some of them are usually present but their absence does not exclude the diagnosis. Careful elicitation of case history, examination of phenotype and correlation with the genotype of the patient with further follow-up will give supportive information and helps to distinguish EVC syndrome from the other short rib-polydactyly syndromes.

CONCLUSION

EVC is an autosomal recessive disorder with a 25% risk of transmission to subsequent pregnancies. Hence a detailed case history, family history and genetic counselling is of utmost importance. One third of the patients die at an early age or during infancy from cardio respiratory problems. To those who survive, the treatment has to be a multidisciplinary approach which includes a team of paediatrician, cardiologist, pulmonologist, clinical geneticist, pedodontist, oral and maxillofacial surgeon, prosthodontist, orthopaedic surgeon, urologist and psychiatrist. The treatment may include orthopaedic correction of deformities, surgical repair of cardiac malformations, dental procedures. Rehabilitation along with strong mental support from family and peers is equally important as it plays a significant role in their all-round development and helps them to do better.

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REFERENCES

1. Talib AN, Alshahrani I, Simasetha S. Ellis-van Creveld syndrome: a rare clinical report of oral rehabilitation by interdisciplinary approach, Case Rep Dent. 2018;23:1-5.
2. Polymeropoulos MH, Ide SE, Wright M. The gene for Ellis van Creveld syndrome is located on chromosome 4p16. Genomics. 1996;35(1):1-5.
3. Galdzicka N, Patnala S, Hirshmanetal MG. A new gene, EVC2, is mutated in Ellis van Creveld syndrome. Molec Genet Metab. 2002;77(4):291-5.
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. Hegde K, Puthran RM, Nair G, Nair PP. Ellis-Van Creveld syndromeea report of two siblings. Br Med J Case Reports. 2011:1-8.
6. Cahuana A, Palma C, Gonzales W, Gean E. Oral manifestations in Ellis-van creveld syndrome. Rep Five Cases Pediatr Dent. 2004;26:277-82.
7. Baujat G, Merrer ML. Ellis-van Creveld syndrome. Orphanet J Rare Dis. 2007;2(1):27.
8. Gopal G, Belavadi GB. Case report of a child with Ellis-Van creveld syndrome. Int J Pharm Biomed Res. 2014;5:14-s7.
9. Alves-Pereira D, Berini-Aytes L, Gay-Escoda C. Ellis-Van Creveld syndrome. Med Oral Patol Oral Cir Bucal. 2009;14:E340-3.

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