

Report of Ellis–Van Creveld Syndrome: A Chondroectodermal Dysplasia

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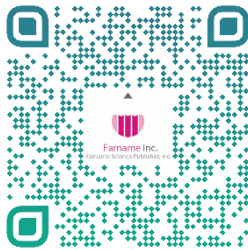
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ABSTRACT

Ellis-Van Creveld (EVC) syndrome, otherwise known as chondroectodermal dysplasia, is a genetic disorder with cardiac and skeletal manifestations. The four main characteristics of EVC syndrome include chondroectodermal dysplasia, polydactyly, congenital heart defects, and hypoplastic nails and teeth. Changes in the mechanical properties of the chest wall in infants with skeletal dysplasia reduce the functional residual capacity and cause respiratory failure. Here, we describe the case of a female patient with symptoms, including fever, poor feeding, and frequent cough, for one week before admission to a hospital in Sabzevar, Iran. The patient was a four-month-old girl with a medical history of EVC syndrome. She complained of fever, poor feeding, and repeated cough for one week. She had multiple labial frenal attachments in the upper and lower jaws, as well as long bones. The iliac bones were short and square-shaped, with the acetabulum in a transverse orientation. Slight elongation of the chest, along with short ribs, was also observed. Based on the findings, EVC syndrome should be considered as a pediatric disorder, which can damage multiple organs, including the skeletal system, and cause respiratory disorders and congenital heart disease. If this syndrome is not diagnosed in childhood, due to poor available treatments, patients will be at risk of disorders at older age.

Keywords: Ellis-Van Creveld syndrome, Mesoectodermal dysplasia, Genetic disease, Case report



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Introduction

Ellis-Van Creveld (EVC) syndrome or chondroectodermal dysplasia is a rare genetic disorder. The prevalence of this syndrome ranges from 1:1,000,000 in the general population to 7:1,500,000 in the Amish population (as the most susceptible group) (1, 2). Richard Ellis and Simon Van Creveld discovered EVC syndrome for the first time in 1940 and published a report and essay on this syndrome. The EvC1 and EvC2 genes, located on chromosome 4p16, are responsible for this disorder (3).

The EVC syndrome is characterized by a tetrad of disproportionate dwarfism, bilateral polydactyly, ectodermal dysplasia, and congenital heart disease (CHD) (3). It has a wide range of oral manifestations, the most common of which are natal teeth in the frontal area of the lower jaw and absence of mucosal folds due to the fusion of the upper lip to the margin of the upper jaw. Radiographic findings, such as X-ray, ultrasound, and echocardiography, are the main diagnostic modalities for EVC syndrome. Herein, we describe the case of a newborn with EVC syndrome.

Case Presentation

In this report, we describe the case of a four-month-old girl with symptoms, such as fever, poor nutrition, and frequent cough, for one week before admission to Heshmatie Hospital of Sabzevar (Razavi Khorasan Province, northeast of Iran) in 2020. Fever was consistent with the symptoms of coryza. She was born at 39 weeks of gestation, and her five-minute Apgar score was nine following birth to healthy parents. Her father was 30 years old when she was born, and her mother was 26 years old. Her parents were not consanguineously married, and their two previous children were healthy. Her mother had no history of radiation exposure or drug abuse during pregnancy.

The patient was born through normal vaginal delivery, and there were no complications during labor and delivery. Her weight was 2,950 g at birth, and her height was 54 cm. Her uncle was diagnosed with Down's syndrome. The symptoms of EVC syndrome were suspected in childhood when she was evaluated for

polydactyly (extra digits of the hands) (Figure 1). She had been hospitalized twice since birth due to aspiration pneumonia and atelectasis. In the oral examination, multiple labial frenal attachments were observed in the upper and lower jaws. Her mother noticed shortening of her limbs on ultrasonography in the gestational period. The radiographic findings indicated shortening of long bones. The iliac bones were short and square-shaped, with the acetabulum in a transverse orientation. She showed a slightly elongated chest with short ribs (Figure

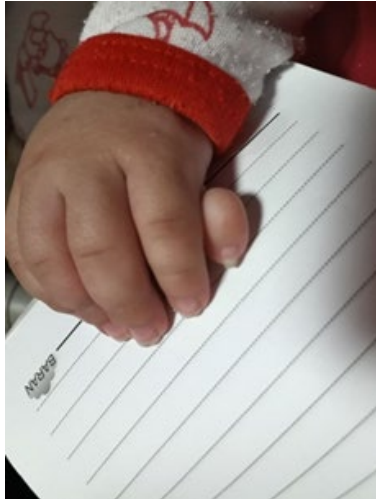


Figure 1. Extra digits in hands in a 4-month-old girl with EVC

Discussion

The EVC syndrome or chondroectodermal dysplasia is a rare disease, with an autosomal recessive inheritance pattern (4). This disorder is caused by mutations in two genes, namely, *EvC* and *EvC2*, located on chromosome 4 (3). Different organs in the body are affected by EVC syndrome with variable phenotypes (5). The prevalence of this disease at birth is estimated at seven cases per 1,000,000 population. Evidence suggests that it is more common in the older Amish population of Lancaster County (Pennsylvania, USA) (3).

In this case report, we presented the case of a four-month-old girl, whose parents were healthy and non-consanguineously married. Generally, patients with EVC syndrome may present with abnormalities of the thoracic cage, which can cause respiratory disorders (6). Findings suggest a reduction in the functional capacity of the lungs and respiratory failure in infants with skeletal dysplasia, such as EVC syndrome (7). Other unusual findings have been also reported in infants with polydactyly and shortening of the acetabulum in the pelvic cavity. Our patient had a small dorsal vertebral lumbar spine, with short and irregular metaphysics. She had acromesomelic dwarfism, with hexadactyly in the ulnar side of the left hand; there was no hexadactyly in the lower extremities. Some studies suggest that these children have disproportionate limbs, polydactyly, short long bones, and partial chest

2). Based on the cardiac auscultation, her heart sounds were normal, and there was no murmur or echo (normal). Ventricular contraction was also normal, and there were no valve abnormalities or septal defects.

The present study was approved by the Research Ethics of Committee of Sabzevar University of Medical Sciences, Sabzevar, Iran (code: IR.MEDSAB.REC.1400.074).



Figure 2. The iliac bones, acetabula, thorax and ribs in a 4-month-old girl with EVC.

elongation with short ribs (7-9). Besides, CHD is prevalent in approximately 50-60% of these cases (10). However, our patient had normal preoperative echocardiographic findings.

Some patients with EVC syndrome show hepatic and renal involvement (11); however, our patient had normal hepatic and renal functions. Previous studies used different approaches to treat this disease, and experts from different specialties, including dentists, nutritionists, radiologists, cardiologists, pulmonologists, orthopedists, urologists, pediatricians, ophthalmologists, pediatric neurologists, psychologists, and clinicians, have contributed to research on this disease. Oral problems are among important findings in the diagnosis of this syndrome.

Conclusion

The risk of mortality due to EVC syndrome is high at birth owing to cardiovascular and respiratory complications. Timely treatment protocols can improve the quality of life of these patients and their families and prevent various disorders.

Acknowledgments

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Conflict of Interest

There is no conflict of interest.

References

1. Tuna EB, Koruyucu M, Kürklü E, et al. Oral and craniofacial manifestations of Ellis-van Creveld syndrome: Case series. *J Cranio Maxillofac Surg.* 2016; 44(8):919-24. [DOI:10.1016/j.jcms.2016.04.025] [PMID]
2. Shetty P, Shetty D, Priyadarshana P, 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(2):98-101. [PMID] [DOI:10.1016/j.jobcr.2015.05.003] [PMCID]
3. Tahririan D, Eshghi A, Givehchian P, Tahririan MA. Chondroectodermal dysplasia: a rare syndrome. *J Dentist Tehran Univ Med Sci.* 2014; 2014:361-4.
4. Peña-Cardelles JF, Domínguez-Medina DA, Cano-Durán JA, Ortega-Concepción D, Cebrián JL. Oral manifestations of ellis-van creveld syndrome. A rare case report. *J Clin Experiment Dentist.* 2019;11(3):e290. [DOI:10.4317/jced.55543] [PMID] [PMCID]
5. Baujat G, Le Merrer M. Ellis-van Creveld syndrome. *Orphanet J Rare Dis.* 2007; 2(1):27. [DOI:10.1186/1750-1172-2-27] [PMID] [PMCID]
6. Naqash TA, Alshahrani I, Simasetha S. Ellis-van Creveld Syndrome: a rare clinical report of oral rehabilitation by interdisciplinary approach. *Case Rep Dentist.* 2018;2018. [DOI:10.1155/2018/8631602] [PMID] [PMCID]
7. Alapati D, Shaffer TH. Skeletal dysplasia: Respiratory management during infancy. *Respiratory Med.* 2017;131:18-26. [PMCID] [DOI:10.1016/j.rmed.2017.07.063] [PMID]
8. Valencia M, Tabet L, Yazbeck N, et al. Ellis-van Creveld syndrome: mutations uncovered in Lebanese families. *Case Rep Genet.* 2015;2015. [DOI:10.1155/2015/528481] [PMID] [PMCID]
9. Iwakura H, Fujii K, Furutani Y, et al. Ellis-van Creveld syndrome associated with chronic intestinal pseudo-obstruction. *Pediatr Int.* 2016; 58(1):64-6. [DOI:10.1111/ped.12846] [PMID]
10. Cahuana A, Palma C, Gonzáles W, Geán E. Oral manifestations in Ellis-van Creveld syndrome: report of five cases. *Pediatr Dentist.* 2004; 26(3): 277-82.
11. Katti S, Oswal P, Vas A. Ellis-van creveld syndrome: A rare case report. *J Interdisciplin Dentist.* 2019;9(1):35. [DOI:10.4103/jid.jid_45_18]

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