Ellis van Creveld Syndrome (EVCS) in an Iranian, Three-Year-old Girl: A Case Report

ABSTRACT
Ellis van Creveld syndrome (EVCS) is mainly characterized by dysplastic changes in skeletal structures along with variable inter- and intra-familial patterns. Shortening of bony structures such as rib and limbs, postaxial polydactyly, and dysplastic teeth and nails are among the clinical presentation of this autosomal recessive disease.

CASE PRESENTATION
A 3-year-old girl who was a known case of cardiac disease, admitted to our pediatric hospital for the first time with complaints of cough, wheezing and manifestations of cardiac problems when she was one year-old. Four days prior to the admission, she developed productive cough, fever, rhinorrhea and vomiting. She was the second child of consanguineous marriage. There was no history of congenital defects among close relatives. The first child of the family died at the age of nine months due to encephalopathy. Congenital cardiac defect was detected by ultrasonography at 7th month of the pregnancy, but due to the legal issues, termination of pregnancy was not possible. She was delivered through a normal term vaginal delivery. Birth weight was 2800g, height was 48cm and occipitofrontal circumference was 32cm. At the time of examination, weight was 7kg, head circumference was 44cm and height was 70cm. There was no history of paroxysmal nocturnal dyspnea, swelling of legs, syncope and hemoptysis. She has been taking Lanoxin and Captopril, and in the last three months, she was under treatment for recurrent respiratory infection with Acetaminophen, Diphenhydramin and Amoxicillin. Physical examination revealed phocomelia in all four limbs, dysplastic nails, hexadactyly in both hands and andontia. [Table/Fig-1a-c] shows her appearance at one-year-old age.

At the time of admission, her vital signs were normal with blood pressure of 110/70 mm Hg, pulse rate of 116 bpm and respiratory rate of 19/min (her vital signs such as blood pressure, pulse and respiratory rate were normal and they were respectively 110/70 mm Hg, 116 bpm and 19/min) . She was alert, seemed malnourished and was 5% dehydrated. Cardiac auscultation revealed a prominent S1 with a low-pitched grade II-III/VI mid-diastolic rumbling murmur (MDM) over the apex and a grade III/VI holosystolic murmur in apex. There was a fixed S1 and S2 splitting.

Laboratory values
White blood cells: 17.5 × 10^3/µl, Neutrophils: 36%, Lymphocytes: 64%, Platelet count: 4.27 × 10^9

Random urine analysis, blood and urine culture test, random blood glucose, serum sodium and calcium levels, liver and kidney function tests, thyroid function test all were normal.

Arterial blood gases test showed pH of 7.4, a partial pressure of carbon dioxide (P CO2): 42.7 mm Hg, HCO3:20.5mmol/L, C-reactive protein was 2+, and erythrocyte sedimentation rate was 13.

DISCUSSION
Ellis-van Creveld syndrome (EVCS) is a chondro-ectodermal dysplasia described by Richard Ellis and Simon van Creveld for the
Alaee Ehsan et al., Ellis van Creveld Syndrome (EVCS) in an Iranian, Three-year-old girl: A Case Report

**Keywords:** Epidermoid cyst, Enucleation, Impacted wisdom teeth, Mandible

---

**Table/Fig-2:** Abnormal features of hand bones in plain x-ray

**Table/Fig-3:** Radiographic abnormalities in upper limbs

---

First time [1]. EVCS is a rare syndrome and its exact prevalence is unknown, but it has been estimated to be 7/1,000,000 of live birth [2,3]. This syndrome has a variable phenotype and affects multiple organs such as heart. Congenital heart defects have been reported in about 50% of the cases [2,4]. Some cardiac manifestations of Ellis-van Creveld syndrome are as following: defects of the mitral and tricuspid valves, patent ductus arteriosus, ventricular septal defects and atrial septal defects [4].

Bassam et al., analysed molecular and clinical aspects of six children with EVC syndrome. They concluded that consanguineous marriages, which are very common in Arab and Middle East, lead to genetic disorders like EVCS and it is heterogeneous at the molecular level in the UAE population [1]. As we described before, our case was also a child of consanguineous marriage.

Chakraborty et al., reported a rare association of carpal tunnel syndrome (CTS), polydactyly and Ostium-primum atrial septal defect.
as an unusual variant of EVC syndrome [5], which was seen in the present case.

DA SILVA et al., reported 15 cases of Ellis-van Creveld syndrome. In this series, all patients had polydactyly of hands and feet. The frequency of cardiac malformation was 50%. The most frequent oral manifestation in this series was hypodontia and only one patient had all his teeth [6]. These manifestations were all seen in our case.

**REFERENCES**


**PARTICULARS OF CONTRIBUTORS:**
1. Assistant Professor, Department of Neonatology, Neonatal and Children’s Health Research Center, Golestan University of Medical Sciences, Gorgan, Iran.
2. Professor, Department of Cardiology, 5 Azar Hospital, Golestan University of Medical Sciences, Gorgan, Iran.
3. Assistant Professor, Department of Pediatric Gastroenterology, Neonatal and Children’s Health Research Center, Golestan University of Medical Sciences, Gorgan, Iran.
4. Assistant Professor, Department of Radiology, Golestan University of Medical Sciences, Gorgan, Iran.
5. Radiologist, Mehregan Medical Imaging Center, Gorgan, Iran.

**NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:**
Dr. Alaee Ehsan, Neonatologist, Assistant Professor, Neonatal and Children's Health Research Center, Taleghani Hospital, Jambazan St, Gorgan City, Iran.
Phone: +98911-171-6302, E-mail: ealaee@yahoo.com

**FINANCIAL OR OTHER COMPETING INTERESTS:** None.

---

