Case Report

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Paediatrics Section

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

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ABSTRACT

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

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

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 IIII/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 × 10^5

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_aCO_2): 42.7 mm Hg, HCO_3:20.5mmol/l, C-reactive protein was 2+, and erythrocyte sedimentation rate was 13.

Radiography of her upper limbs with several abnormalities is shown in [Table/Fig-2,3].

[Table/Fig-4] also shows abnormal bones' deviations in lower limbs.

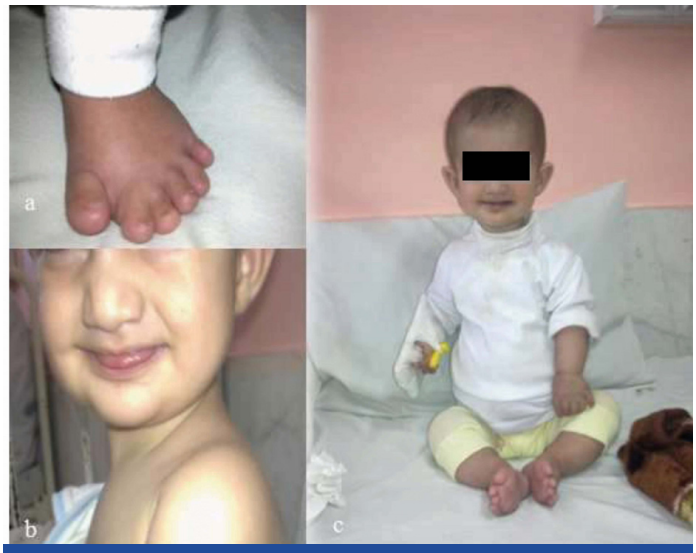
Electrocardiography showed features of left axis deviation, bi-lateral enlargement and right ventricular hypertrophy. Chest radiograph showed increased cardiothoracic ratio along with reticulonodular shadowing, lobar collapse in a lower zones of lungs that confirmed a diagnosis of pneumonia.

Echocardiography identified Cor-atrium, along with left-to-right shunt across the septal defect, mild mitral regurgitation and left-ventricular hyperthrophia. The mean pulmonary artery pressure (MPA) using Doppler Radiography, Genu valgum, shortness of metacarples and phalanges. Cranial sonography revealed mild hydrocephalus without intracranial and cerebral ventricular hemorrhage.

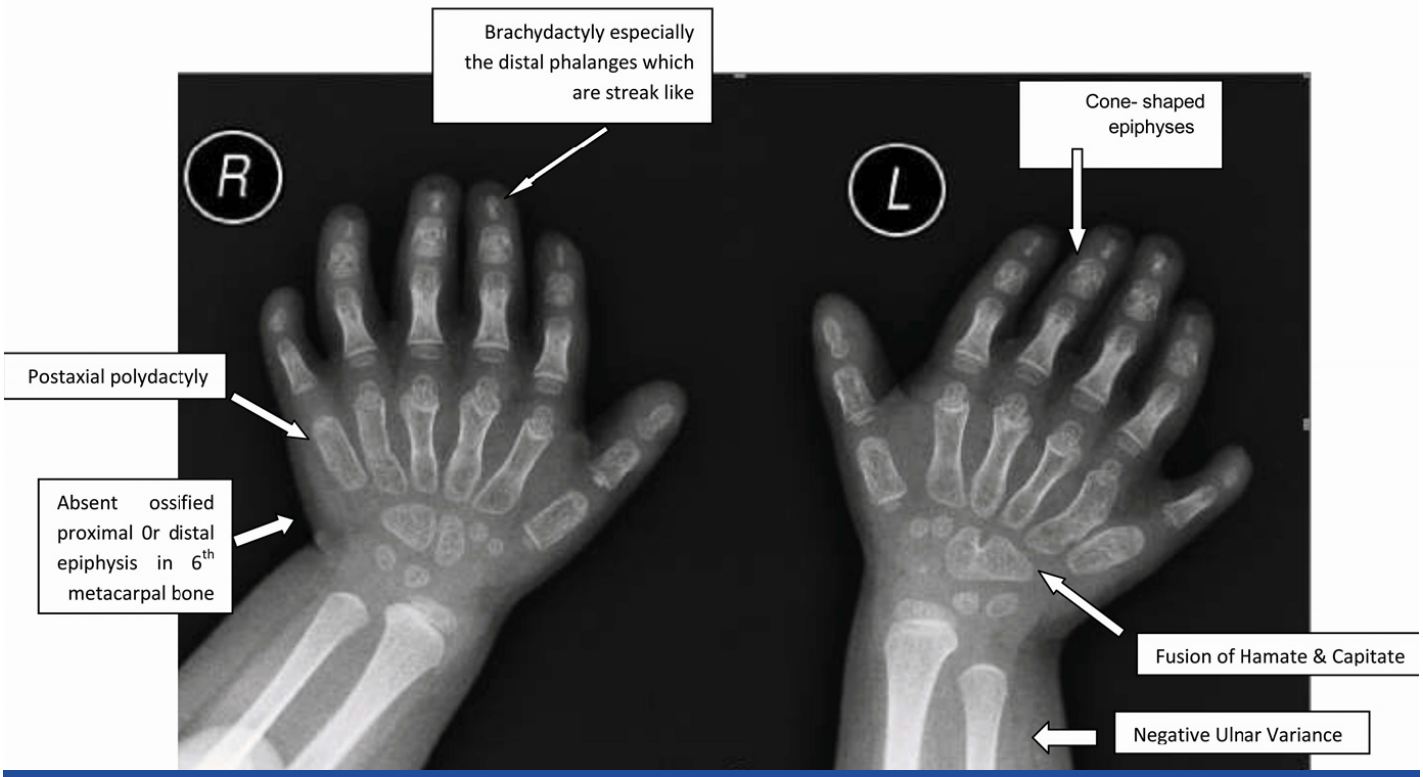
From then on the patient was under continuous observations, and now she is 3 year-old.

DISCUSSION

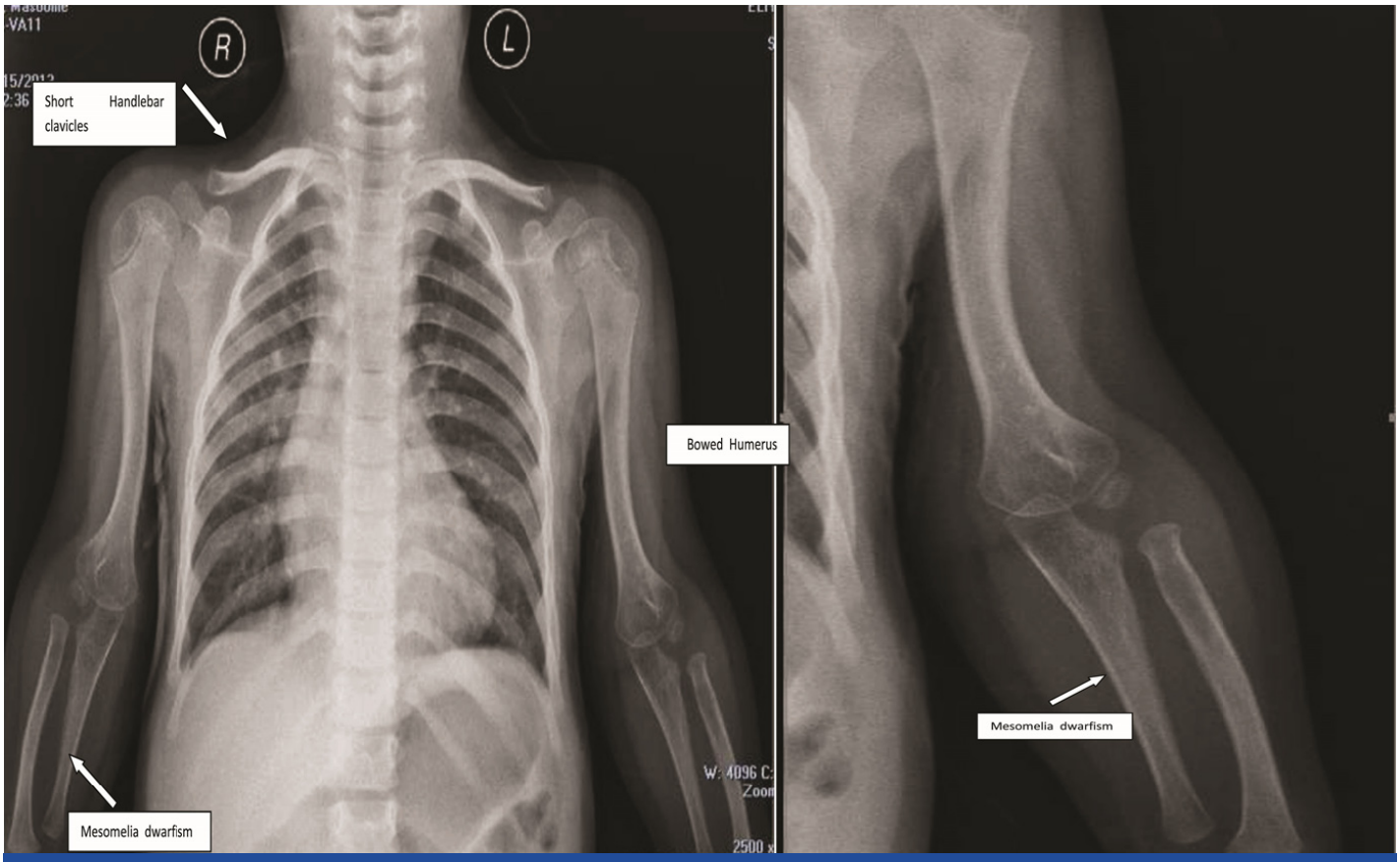
Ellis-van Creveld syndrome (EVCS) is a chondro-ectodermal dysplasia described by Richard Ellis and Simon van Creveld for the



[Table/Fig-1]: Phocomelia in all 4 limbs: (a) dysplastic nails (b) andontia and (c) hexadactyly on both hands



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

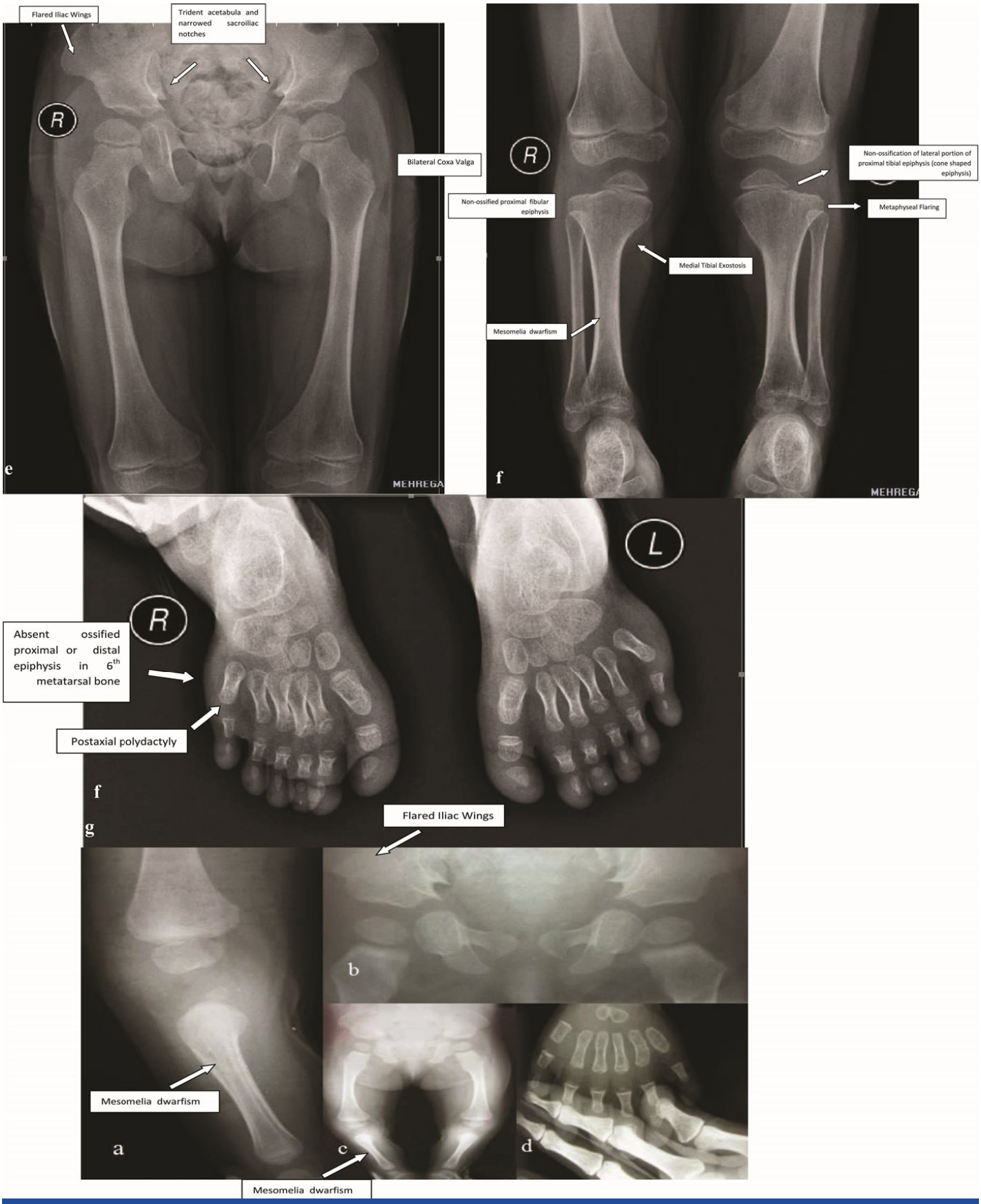


[Table/Fig-3]: Radiographic abnormalities in upper limbs

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-primun atrial septal defect



[Table/Fig-4]: Abnormalities in lower limbs: (a to d) deviation in iliac bones

as an unusual variant of EVC syndrome [5], which was seen in the present case.

DA SILVA et al., reported 15 cases of Elis-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.

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