Ellis Van Creveld Syndrome: A Rare Case Report

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ABSTRACT

Ellis Van Creveld Syndrome is a rare genetic disorder having autosomal recessive inheritance, mostly affecting the Amish Population of Lancaster County in Pennsylvania in the US, with prevalence rate of 1/5000 at live birth. In non-Amish population, the birth prevalence is 7/1,000,000. Classical manifestations of the disease includes small stature, short limbs, hypoplastic finger nails, bilateral post axial polydactyly, hypoplasia of the enamel, hypodontia and malocclusion. Heart defects, especially abnormalities of atrial septation has been reported in 60 % of the cases. Very few cases of Ellis Van Creveld Syndrome have been reported in Pakistan. We hereby presents a case of 13 year old male child presenting with classical oral, chondroectodermal and cardiac manifestations of this syndrome.

INTRODUCTION

Ellis Van Cleveld Syndrome or Chondro-ectodermal dysplasia, is a rare disease complex where all three germ layers appears to be involved(1). Also known as Mesvectodermal Dysplasia, it was first described in 1940 by Richard W.B Ellis and Simon Van Creveld hence the name Ellis Van Creveld(2). A rare Autosomal Recessive disorder, it is most prevalent in the Amish population of Lancaster County, Pennsylvania, US, occurring in 1/5,000 live births, and birth in non-Amish

population is expected to be 7/1000000(2)(3). Of 300 reported cases, ectodermal dysplasia was found in up to 93% of cases(4).

EVC is cause by mutation in EVC gene located on chromosome 4p16, as well as mutation in a non-homologous gene EV2, located near EVC gene in head to head configuration, with parenteral consanguinity in about 30% of cases(1)(5)(6). This syndrome is characterized by variable spectrum of clinical findings:

a) Skeletal dysplasia is present at birth, being characterized by short limbs, especially inthe middle and distal segments, resulting in disproportionate dwarfism, and an exceptionally long trunk.(1)(7)(11).

b) Polydactyly is a constant finding, on the hands, usually bilateral on the ulnar side. Polydactyly of feet is present in only 10% of patients.(2).

c) Hypodontia involving maxillary and mandibular incisors is a consistent finding, though supernumerary teeth have been reported in some cases(2)(8). Another constant finding is fusion of anterior portion of upper lip to maxillary gingival margin as a result of which no mucobuccal fold exists, causing the upper lip to present a slight V-shaped notch in the middle.(2)(9)(10)

d)Nearly, all patients show severe dystrophy of the fingernails, which are markedly hypoplastic, thin, and often spoon-shaped.(1)

e) Approximately half of affected individuals have congenital heart disease, most commonly atrial septal defect, often with single atrium.(11)

Although most patients have normal intelligence, occasional central nervous system anomalies or mental retardation have been reported

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We hereby represents a case report that presents a typical case of Ellis Van Creveld Syndrome in a 13 year old male child from Karachi, Pakistan.

History

We have a 13 years old male child presented with complains of shortness of breath (FC II-III), decreased exercise tolerance and palpitations. According to patient's father he had these symptoms since childhood and his symptoms worsened over a period 2-3 years. There was no history of cyanosis, syncope or seizures. Upon inquiring regarding his past medical history, he had been diagnosed with congenital heart disease in early childhood but lost to follow up. There is no history of consanguineous marriages between his parents and grandparents. No significant family history of congenital heart diseases in his family.

On general physical examination, patient had a short stature with long narrow chest and bilateral post-axial polydactyly. Further examination of hands and feet revealed malformed and dysplastic nails. He had an elongated face with low set ears and oral examination showed absent upper incisor teeth bilaterally.
Chest X-ray was done and showed enlarged cardiac silhouette and dilated main pulmonary artery with peripheral pruning. Echocardiography was done and showed hugely dilated common atrium, severe pulmonary hypertension, interrupted IVC? and L-TGA?. To rule out uncertainty in echocardiographic assessment CT Angiography was done and revealed single atrium, small VSD and bilateral SVC.
Hand and wrist radiograph was done which showed complete post-axial polydactyly with no other bony deformity.



Skeletal Dysplasia(Disproportionate Dwarfism)



Polydactyly in the Hands



Dystrophy of Fingernails. Hypoplastic, thin, spoon-shaped



Polydactyly of the Feet



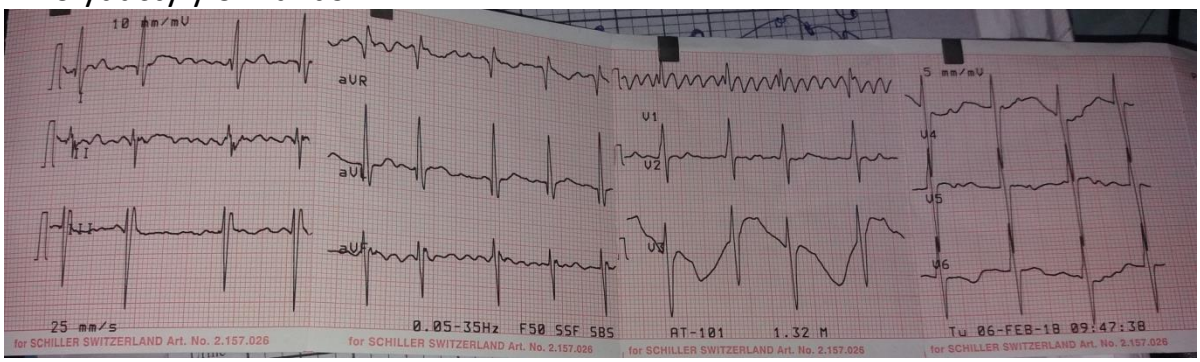
Hypodontia of teeth, Absent Incisors



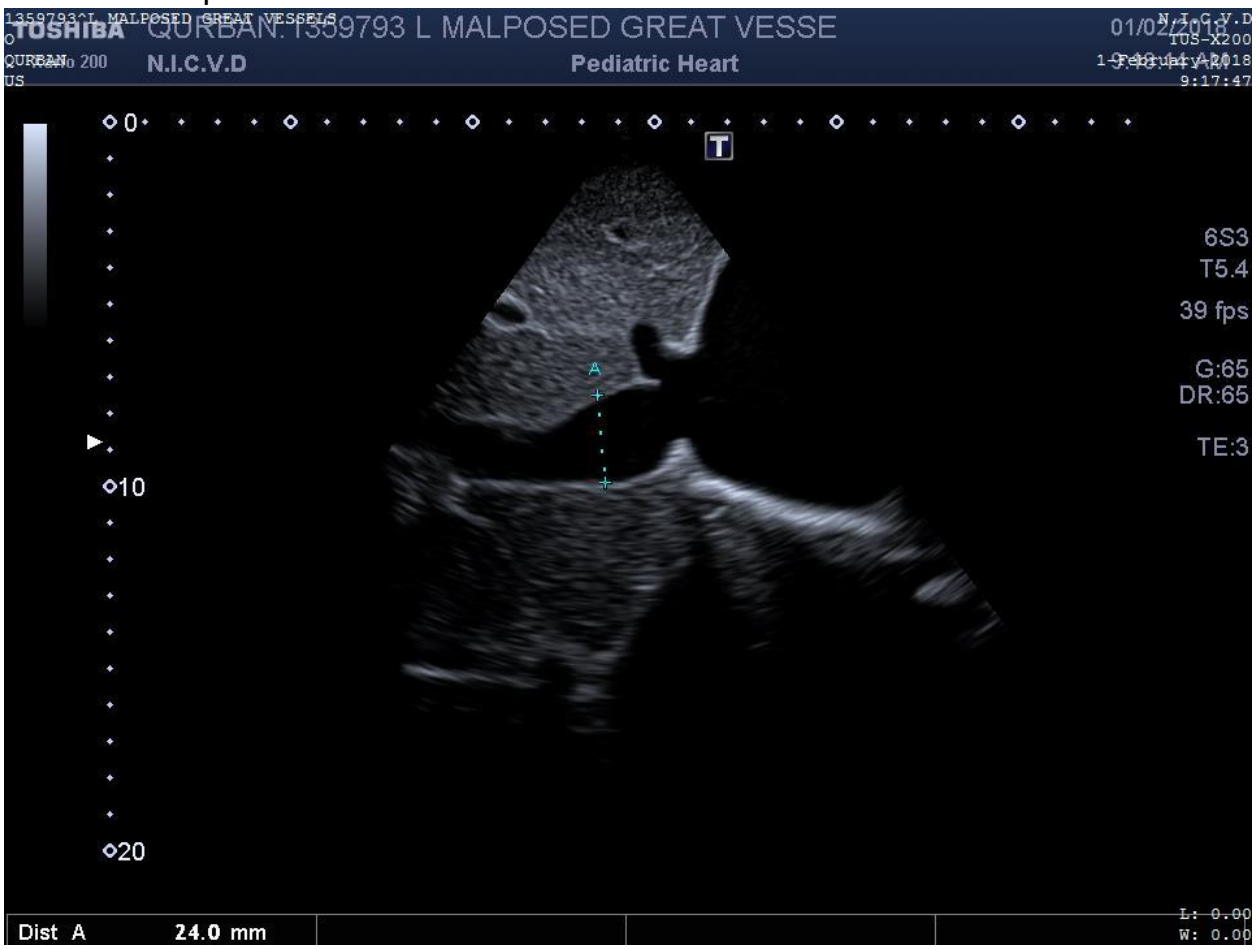
Hypoplastic, Spoon-Shaped nails


ARDIOVASCULAR
DISEASES

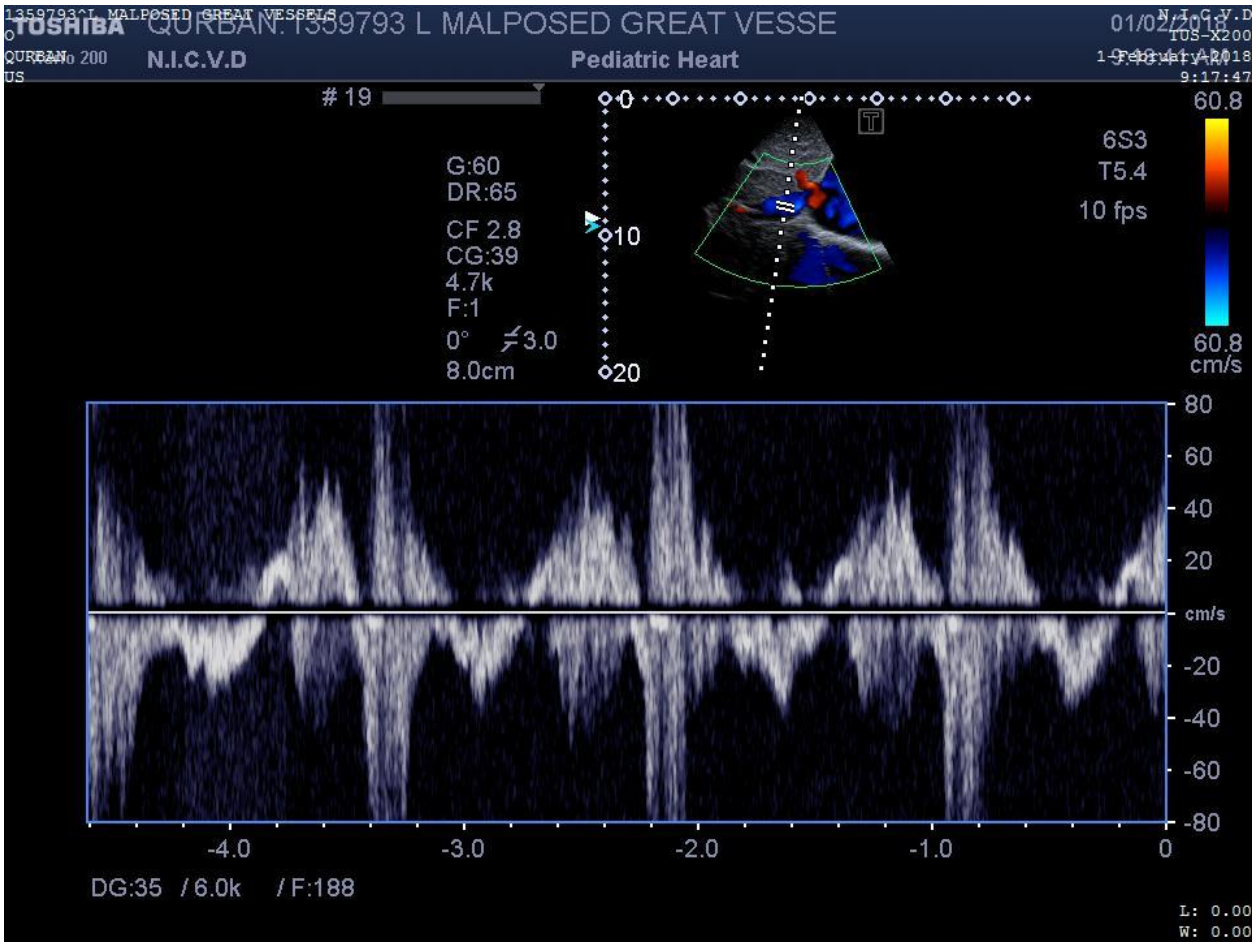
Polydactyly of hands



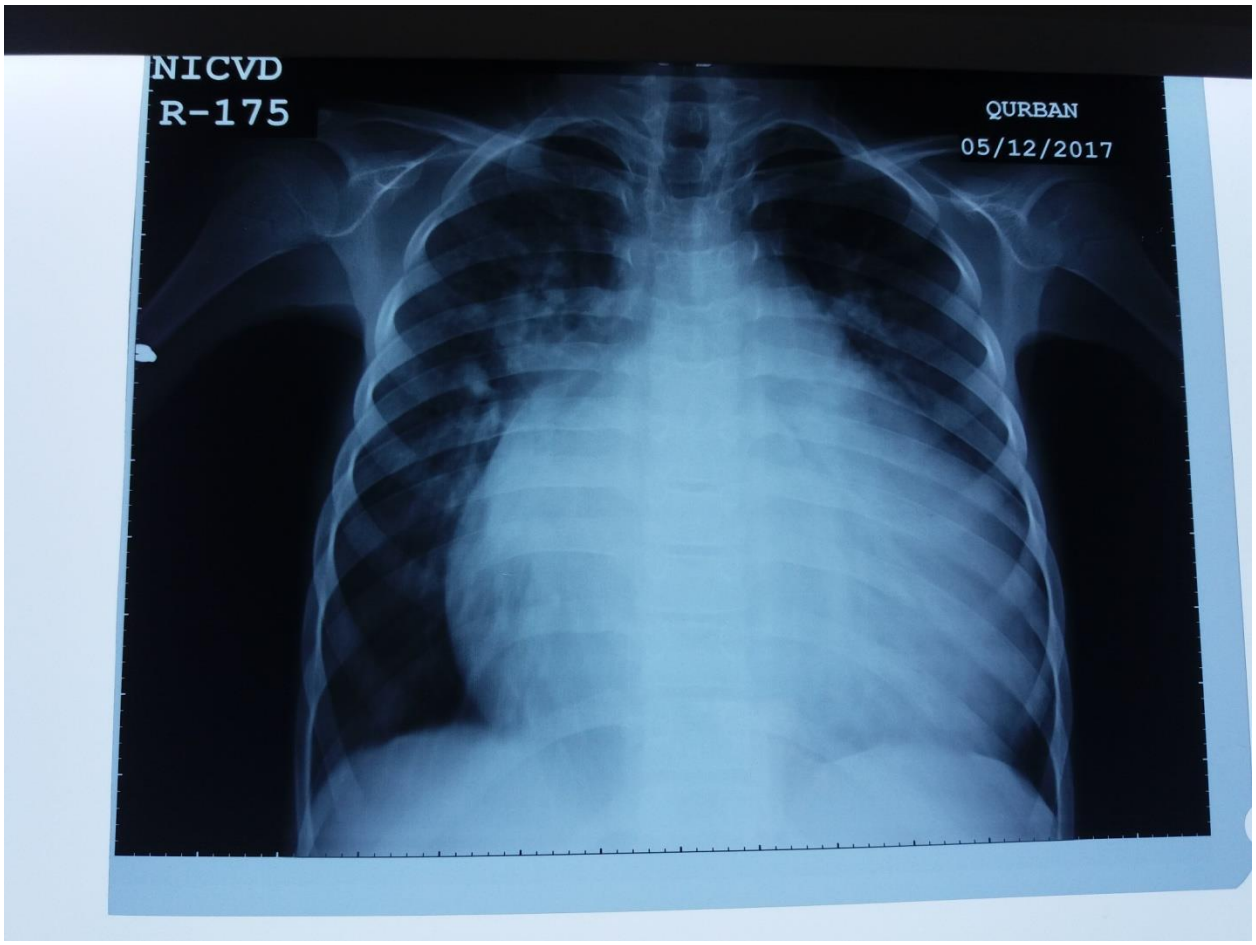
ECG of the patient



Echo of the patient



Echo of the patient



Chest X-ray of the patient



X-ray of Hand, AP and Lateral

DISCUSSION

Ellis-van Creveld syndrome (EVC) is a rare autosomal recessive genetic disorder with characteristic clinical manifestations, resulting from a genetic mutation in two genes, EVC1 *and* EVC2(5)(7).

A chondral and ectodermal dysplasia, EVC is characterized by short stature, short limbs,polydactyly, growth retardation, and ectodermal and heart defects. It is a rare disease with approximately 300 cases reported worldwide(12)

Chondral dysplasia in the form of acromesomelia, long narrow chest, with progressive shortening of distal extremities, along with post axial polydactyly of the hands, which is usually bilateral was present in this patient. Other skeletal defects found in EVC such as cubitusvalgus, genuvalgus, fused carpals, polydactyly in feet were not seen.(13)

Ectodermal Dysplasia presenting as tooth, hair and nail abnormalities were reported in 93% of published cases. Common nail abnormalities include hypoplastic/dysplastic/friable/absent nails(14) Our case presented with malformed and dysplastic nails.

Oral manifestations of EVC are diverse and includes hyperplasticfrenula, absence of mucobuccal fold, serrations of the alveolar ridge, multiple small alveolar notches, partial cleft lip, neonatal teeth, peg-shaped laterals, partial anodontia, congenitally missing incisors, conical and microdontic teeth, enamel hypoplasia and delayed eruption of teeth(15)(16). Among these, congenitally missing incisors was present in our patient.

Congenital heart malformations occurring in about 50–60% of cases and comprising of single atrium, defects of the mitral and tricuspid valves, patent ductus, ventricular septal defect, atrialseptal defect and hypoplastic left heart syndrome(13)(17)(18). Single atrium, small VSD and bilateral SVC was reported in this patient.

Conclusion :

EVC is a rare genetic disorder with no definitive cure.(19).Treatment is usually symptomatic, which can be accomplished with multidisciplinary approach. Patients generally need the consultation of Pulmonologist, Cardiologist, Orthopedist, Physiotherapist, Plastic surgeon, Dentist, Psychologist, etc.(1)(2)(7)(19).

Keywords: EVC , autosomal recessive, single atrium , short stature , chondroectodermal dysplasia.

References:

1. Ellis Van Creveld Syndrome with unusual oral and dental findings; A rare clinical entity
SameeullaShaik, Jayam Raviraj, Suresh Dirasantchu, Suman S Venkata

2. Kurian K, Shanmugam S, Harshvardhan T, Gupta S. Chondroectodermal Dysplasia(Ellis Van Creveld Syndrome): A report of three cases with review of Literature. Indian J Dent Res 2007;18;31-4

3. Stoll C, Dott B, Roth MP, Alembik Y. Birth prevalence rates of skeletal dysplasias. Clin Genet 1989;35:88-92.

4 Dandy-Walker malformation in Ellis-van Creveld syndrome.
Zangwill KM, Boal DK, Ladda RL.

5. Ellis-van Creveld syndrome in an Indian child: a case report.
K. M. Veena, H. Jagadishchandra, Prasanna Kumar Rao, Laxmikanthchatra

6. A rare case report of Ellis Van Creveld syndrome in an Indian patient and literature review.
PrashanthShetty, DeepthiShetty, P.S. Priyadarshana, SmithaBhat.

7. Ellis-van Creveld Syndrome: A Rare Clinical Report of Oral Rehabilitation by Interdisciplinary Approach.
TalimAminNaqash, Ibrahim Alshahrani, SiripanSimasetha

8. Chondroectodermal dysplasia (Ellis-van Creveld syndrome): report of two cases.
Prabhu SR, Daftary DK, Dholakia HM.

9. Babaji P. Oral abnormalities in Ellis Van Creveld Syndrome. Indian J Dent Res 2010;21 143-5

10. Chondroectodermal Dysplasia: A rare Syndrome
Dana Tahririan, Alireza Eshghi, Pirooz Givehchian, and Muhammad Ali Tahririan

11. First Trimester Prenatal Diagnosis Of Chondroectodermal Dysplasia(Ellis Van Creveld Syndrome) with Ultrasound.
L DUGOFF, G THIEME, J C HOBBINS

12 . Ellis-van Creveld Syndrome: Mutations Uncovered in Lebanese Families
Maria Valencia, Lara Tabet, Nadine Yazbeck, Alia Araj, Victor L. Ruiz-Perez, KhalilCharaffedine, Farah Fares, Rebecca Badra, and Chantal Farra.

13. Adult Survival in Ellis-van Creveld Syndrome with Common Atrium – A Rare Case Report
MugulaSudhakarRao, Devavrata Sahu, Hashir Kareem, Tom Devasia, and Kishor Kumar Shetty

14. Ellis-van Creveld syndrome presenting in the second decade
Uday Yanamandra, Prafull Sharma, AnanthaKrishnan Ramamoorthy and Vasu Vardhan

15. Oral manifestations of Ellis-van Creveld syndrome: report of two siblings with unusual dental anomalies.
Hattab FN. Yassin OM, Sasa IS

16. Oral manifestations of Ellis-van Creveld syndrome.
RiteshKalaskar, and Ashita R. Kalaskar

17. Hills, C.B. , Kochilas, L. ,Schimmenti, L.A. et al . PediatrCardiol (2011) 32: 977.
http://doi.org/10.1007/s00246-011-0006-9

18. Ellis-Van Creveld syndrome.
Genevieve Baujat and Martine Le Merrer

19. Ellis-van Creveld syndrome: A rare clinical entity
ReetKamal, Praveen Dahiya, Simerpreet Kaur, Rohit Bhardwaj, and Karun Chaudhry