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ELLIS VAN CREVELD SYNDROME WITH COMPLETE AV CANAL DEFECT – LATE RECOGNITION STILL REPAIRABLE

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ABSTRACT

Ellis Van Creveld syndrome (EVC) or chondroectodermal dysplasia is a rare congenital disorder of autosomal inheritance. It is commonly seen in Amish population of Pennslyvania in USA with the prevalence of 1/5,000 live births. In non- amish population the prevalence is 7/1,000,000 live births. It is caused by mutation in the EVC gene located on chromosome 4p16 and also mutation of EVC2 gene located close to EVC gene in a head to head configuration. It is characterized by bilateral postaxial polydactyly of hands, dysplasia of teeth and nails, short limbed dwarfism and congenital heart disease.
This case report describes a 19 year old year Indian female born to a consanguineous marriage with the classical features of Ellis Van Creveld Syndrome with complete AV canal defect with relatively asymptomatic childhood with classical goose neck deformity in left ventricular angiogram

Keywords: Ellis van creveld syndrome, polydactyly, chondroectodermal dysplasia, complete AV canal defect, goose neck deformity

INTRODUCTION

Robert Ellis and Simon Van Creveld in 1940 defined a syndrome termed chondro- ectodermal dysplasia comprising of certain type of chondrodysplasia, bilateral polydactyly of hands, small and dysplastic teeth and nails, congenital heart defects.(1) It is now referred to as Ellis Van creveld syndrome. It is most prevalent in the Amish population of Pennsylvania in USA occurring in 1 / 5,000 live births. In non- amish population birth prevalence is 7 / 1,000,000 live births, making it quite a rare occurrence in Indian subjects.

It is an autosomal recessive disorder caused by mutations in the EVC gene located on chromosome 4p16 and also mutation in EVC2 gene located close to EVC gene in a head to head configuration. Of the 300 cases reported ectodermal dysplasia is present in upto 93%. Five different mutations have been associated with this syndrome.(2) There is no male or female predeliction.

It usually presents as a characteristic tetrad (3)

1. Bilateral postaxial polydactyly of hands. Polydactyly of the feet is present in only 10% of the patients. Frequently the patient cannot make a tight fist.
2. Disproportionate dwarfism due to chondrodysplasia of the long bones and an exceptionally long trunk.
3. Presence of multiple frenulae tethering the upper and lower lip to the gingiva. Ectodermal dysplasia with dystrophic, small nails, thin sparse hair and hypodontic and abnormally formed teeth.
4. Congenital heart defects in 50- 60% of cases, mostly a single atrium and a Ventricular septal defect.

Patient's intelligence is usually normal. Liver and central nervous system abnormalities are rare. It is impossible to radiographically differentiate Ellis Van Creveld syndrome from similar chondrodystrophies.[2] Other skeletal anomalies such as genu valgum (knock knees) also can be seen.

CASE REPORT

A 19 year old young female presented to the outpatient department with complaints of exertional dyspnea of 1 year duration which has progressive from NYHA class 2 to NYHA class 3 within the past 2 months. There was no history of orthopnea or paroxysmal nocturnal dyspnea. She also had complaints of easy fatigability and occasional chest pain. She was told that she has some cardiac problem at birth but never had follow up due to long asymptomatic period. She had frequent upper respiratory tract infection in the past and was relatively asymptomatic throughout her childhood and early adolescence. She was born to a consanguineous marriage, attained menarche at the age of 13 years and has normal menstrual cycles. Her school performance was average. She had short stature and was born with six fingers in each hand. Her siblings and parents were normal and had no significant illness.

On examination she was short statured with a height of 135cm and well nourished. She had six fingers in both the hands (Figure 2) (extra finger in each hand on the ulnar side), small and brittle nails in the hands and toes (dysplastic nails).

Oral cavity examination revealed peg teeth with multiple frenulae in the lower lip and high arched palate. Cardiovascular examination: S1 heard normally, S2 widely split, pansystolic murmur in mitral area, no added sounds. Other systems were normal. Investigations revealed haemoglobin of 9 g/dl otherwise normal.

Chest xray showed cardiomegaly. Ultrasound abdomen was normal. Electrocardiogram showed normal sinus rhythm, first degree heart block, biatrial enlargement, right ventricular hypertrophy (Figure 5).

Echocardiography showed COMPLETE ATRIOVENTRICULAR CANAL DEFECT- large ostium primum Atrial septal defect amounting to single atrium, two separate atrioventricular valves with concominant regurgitation, small ventricular septal defect closed by septal leaflet of tricuspid valve, right atrium and right ventricle dilated, mild pulmonary artery hypertension. (Figure 6).

Catheterization study- classical Goose Neck deformity (Figure 7) due to elongated left ventricular outflow tract in left ventricular angiogram. The saturation run showed significant step up in right atrium with QP/QS- 5.05. Mean pulmonary artery pressure was recorded as 26mm Hg.

DISCUSSION

EVC syndrome belongs to a the short rib polydactyly group (SRP's) which includes Verma – Naumoff syndrome, Jeune dystrophy, Mekusick-Kaufman syndrome and Weyers syndrome, Beemer- Langer syndrome, Saldino- Noonan syndrome and Ellis Van Creveld syndrome. These short rib polydactyly group are usually among the differential diagnosis in the pernatal and postnatal period.[5]

The parents are the carriers of the mutation and there is 25% chance of further pregnancies resulting in a child with a similar problem.[5] Our patient was born to a consanguineous marriage with no similar illness among the siblings or her parents.

Chondrodysplasia means disproportionate dwarfism i.e., normal trunk with symmetrical shortening of distal extremities. Thoracic dysplasia leads to frequent respiratory infections.[3] Our patient had frequent chest infections primarily due to acyanotic heart disease with left to right shunt and was a dwarf (Figure 1) with a height of 135cms. There was no lumbar lardosis or genu valgum or pectus carinatum in the patient which could also be seen as a result of chondrodysplasia.

She had classical polydactyly (Figure 2) of Ellis Van Creveld syndrome involving both the hands on the postaxial side. There was no polydactyly of the foot which is also present in 10% of the patients. There was no syncarpalism or synmetacarpalism[3] in her which could also be possible. She could not make tight fist[3] like other patients of Ellis Van Creveld syndrome.

Ectodermal dysplasia in this patient lead to multiple frenulae connecting the lower lip to the gingiva(Figure 3), peg teeth(Figure 4) and high arched palate.[3] There was no natal or congenitally missing teeth but they were microdontic in nature.[4] She had dysplatic nails in both hands and feets (Figure 2) as seen in classical patient.

Cardiac manifestations are seen in 50-60% of cases.[5] Patient had a complete atrioventricular canal defect which is the most common congenital abnormality seen in 40% of cases followed by a ventricular septal defect or a patent ductus arteriosus.[3]

Liver and Central nervous system abnormalities could also occur rarely in Ellis Van Creveld syndrome. Other genitourinary anomalies like renal agenesis, renal tubular dilatation, nephrocalcinosis, megaureters, vulvar atresia.

50 % patients with Ellis Van Creveld syndrome usually die at infancy due to recurrent respiratory infections.[3] Those without any cardiac abnormality may have a normal life expectancy. This patient who has a complete atrioventricular canal defect has survived till 19 years without any significant pulmonary artery hypertension is quiet uncommon.

CONCLUSION

Ellis van Creveld syndrome is a rare disease in Indian population. This case report describes a 19 year old young female with Ellis van Creveld syndrome with complete atrioventricular canal defect who had a relatively asymptomatic childhood who presented in her late adolescence without any severe symptoms whose defect is still repairable.

CONFLICT OF INTEREST

None.

REFERENCES

1. Ellis RW, van Creveld S. A syndrome characterized by ectodermal dysplasia, polydactyly, chondro-dysplasia and congenital morbus cordis. Arch Dis Childhood 1940;15:65- 79.
2. K Kurian , Shanmugam S , Harsh Vardha 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. Ikramullah K, Syed A, Kiren M. Ellis van Creveld syndrome: A case report. Journal of Pakistan Association of Dermatologists 2006;16:239-242.
4. Cahuana et al. Oral manifestations in Ellis van Creveld syndrome: report of five cases. Pediatric Dentistry 2004;26:3:277-279.
5. Baujat G, Le Merrer M. Ellis van Creveld syndrome: Review. Orphanet Journal of Rare Dis 2007;2:27 1-5.

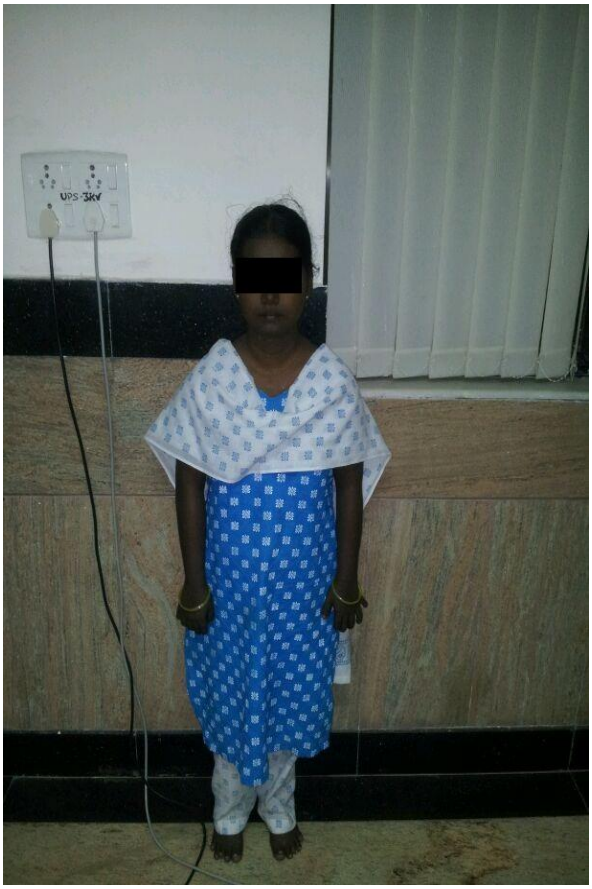

Figure 1: dwarfism

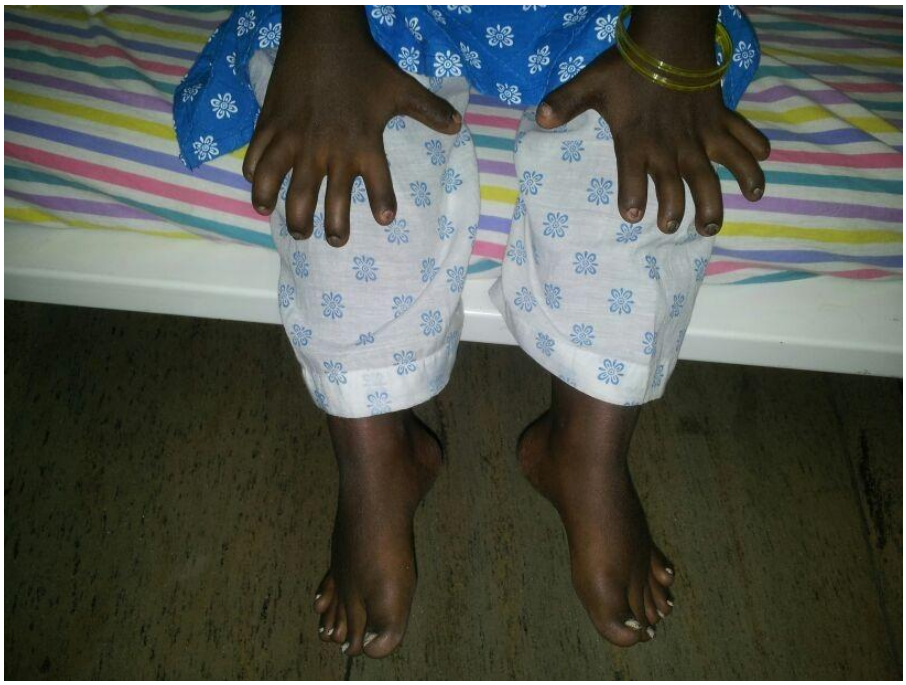

Figure 2: Bilateral postaxial polydactyly

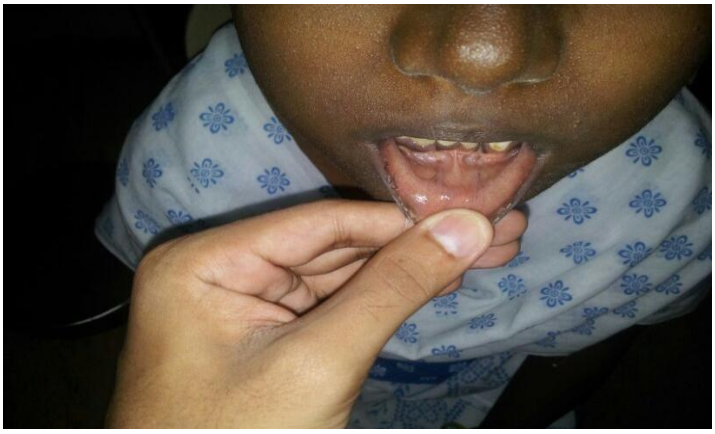

Figure 3: Multiple frenulae connecting the lower lip to gingival

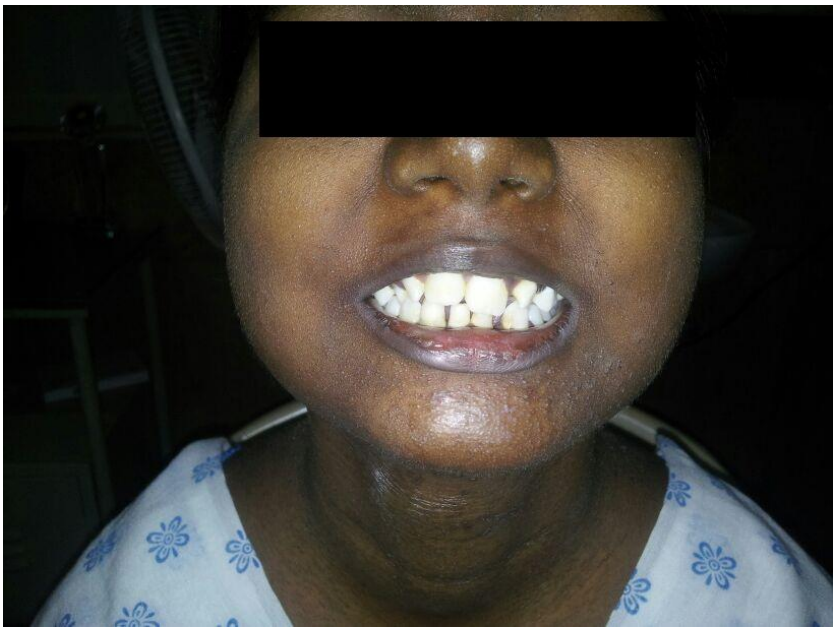

Figure 4: Peg teeth

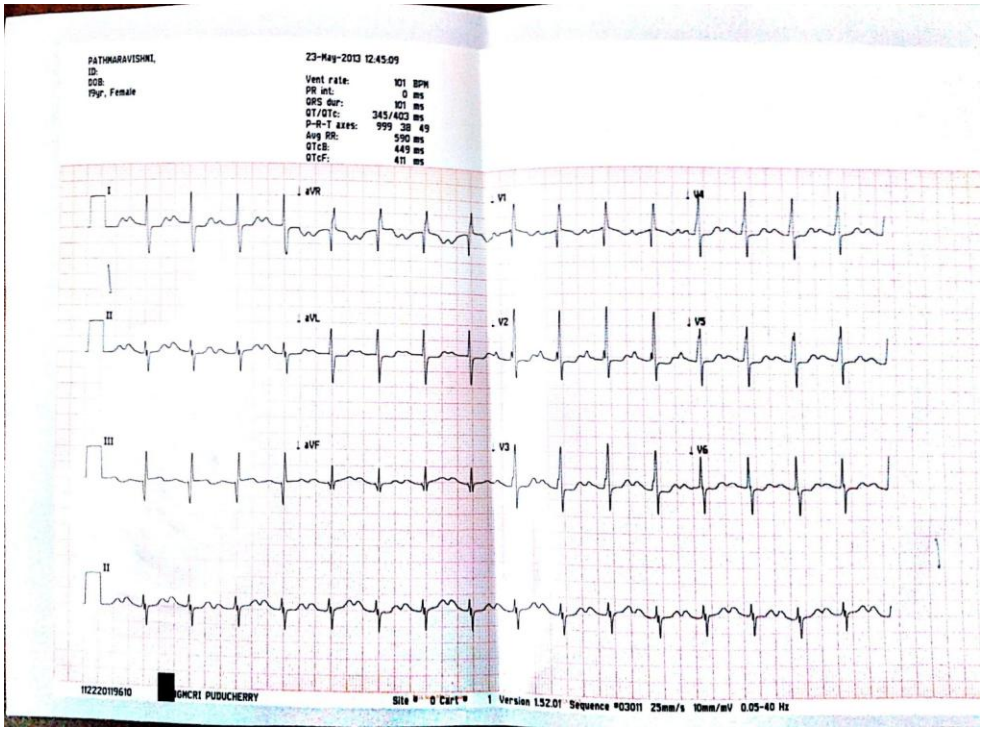

Figure 5: normal sinus rhythm, first degree heart block, biatrial enlargement, right ventricular hypertrophy

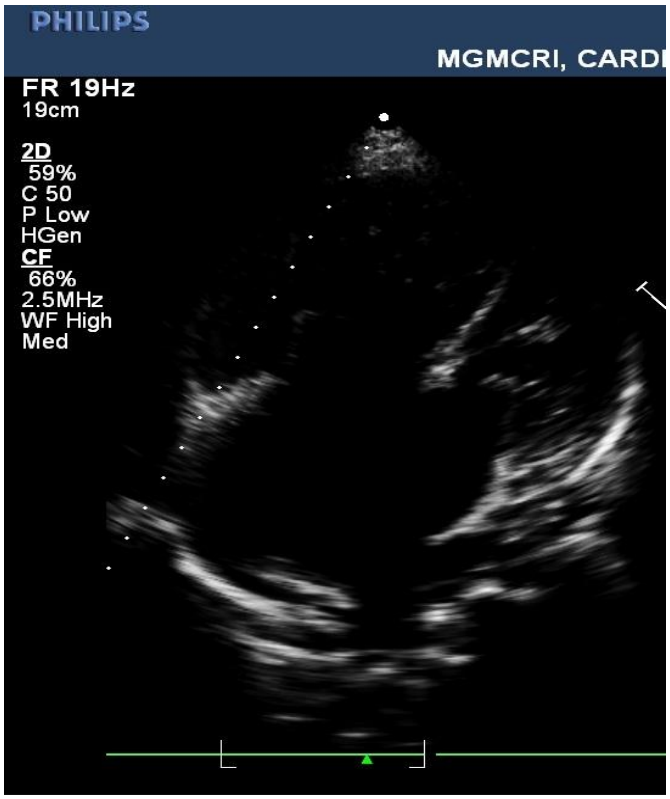

Figure 6: complete Atrioventricular canal defect

Figure 7: Goose Neck deformity due to elongated Left ventricular outflow tract in Left ventricular angiogram