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Late Survival in Ellis Van-Creveld Syndrome in an Indian - A Case Report

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Abstract: ***Ellis Van Creveld (EVC) syndrome also called as chondroectodermaldysplasia [1] is a rare genetic disorder [1] characterised by Chondrodystrophy, Polydactyly, Ectodermal dysplasia and cardiac anomalies. It is an important phenotype for congenital heart disease as it is present in 50% of patients with this syndrome and half of them have a common atrium [2, 5]. These patients rarely survive till adulthood. Here we report a case of EVC syndrome presenting in middle age. A 40 year old male presented with history of progressive breathlessness of five years duration and on examination he was found to be in cardiac failure. He had Short stature, Central cyanosis, Clubbing, Genu valgum, Polydactyly of both hands and feet, Clinodactyly and Common atrium as proved by echocardiogram.***

Keywords: Chondro ectodermal dysplasia, Polydactyly, EVC, Common atrium

1. Introduction

Ellis Van-Creveld (EVC) Syndrome is a very rare Autosomal Recessive Disorder caused by mutation in genes EVC and EVC 2 located on chromosome 4P16[1]. It was first described by Ellis and Simon Van- Creveld in 1940 with the frequency of 1/60000 to 1/200000 newborns [1, 4], only about 150 cases have been reported in clinical literature. It is more prevalent in Amish population of USA [1, 5, 8] where the largest pedigree has been described. Now it is known to affect all the races [8]. The exact prevalence still remains unknown [1]. There is no gender difference in incidence [6].

2. Case Report

A 40 year old male, born to second degree consanguineous parents presented with breathlessness for the past five years associated with orthopnea and PND. He was diagnosed to have a cardiac problem at the age of 15 and not under follow up and treatment. H/O Recurrent respiratory tract infections + and no H/O cyanotic spells or squatting. On examination the patient was short statured. There was B/L Polydactyly in both upper and lower limbs (6 digits), Clinodactyly and Genu valgum. There was central cyanosis, clubbing, B/L pitting pedal edema and high arched palate. Examination of CVS revealed precordial bulge ,parasternal heave, elevated JVP , apical impulse felt on left 7th intercostal space in anterior clavicular line-hyperdynamic in nature, Pulmonary area - fixed and wide split of S2 and loud P2. Tricuspid area - Grade III pan systolic murmur best heard during inspiration . RS- B/L basal crepts+,Abd- tender hepatomegaly ,CNS clinically normal.

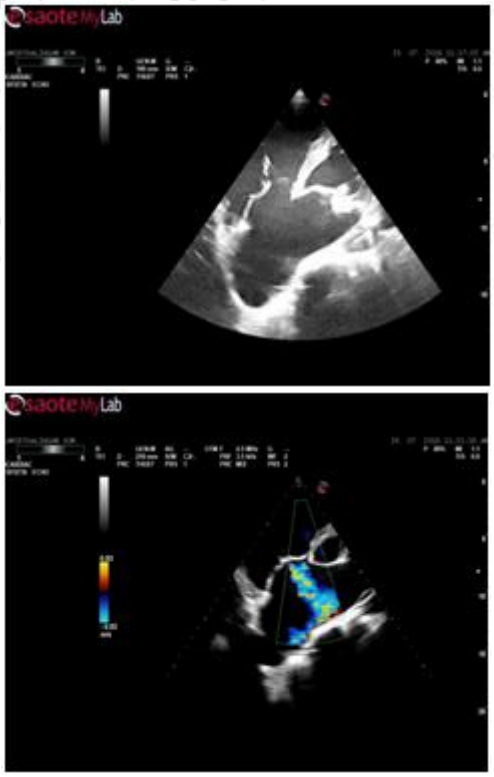
3. Investigations Done

ECG – Left axis deviation,RVH and RBBB.

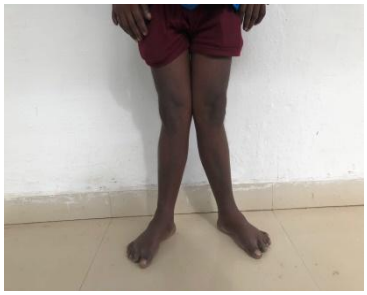
CXR PA - Cardiomegaly, Pulmonary vessel prominence,RA RV LV dilatation

X-ray Wrist - Left capitate hamate doubtful fusion+, polydactyly+, fusion of 5th and 6th metacarpalsB/L USG abdomen -congestive liver.

Echo shows Common atrium, Severe PAH, TR (moderate), MR (moderate), no pericardial effusion with EF 35%.



ECHO SHOWING COMMON ATRIUM



Genu valgum

Post Axial Polydactyly Of Both Hands

As the patient had common atrium with the classical skeletal anomalies, the diagnosis was made as EVC syndrome and the patient was treated for cardiac failure.

4. Discussion

Ellis van creveld(EVC)syndrome is a rare Autosomal Recessive Disorder. H/O parental consanguinity is common in 30% of cases [8],in our case also there was H/O second degree consanguinity.It is also called as chondroectodermal dysplasia as the condition affects the skeleton(chondro)and skin(ectoderm).The cardinal clinical features are dwarfism, polydactyly, ectodermal dysplasia and congenital heart diseases. Our patient was short statured. He had post axial polydactyly (additional finger next to little finger) in both hands which is an invariable finding(100%) in EVC [1] was present in our patient. Polydactyly of the feet only in 10%[1] which is also present in our case. Clinodactyly (bent fingers) syndactyly (interdigital webbing) said to be the common features in EVC [2]also present in our patient. Genu valgum, an inconstant finding also present in our case.Among the ectodermal dysplasia,hypoplasia of nail and high arched palate were present. Congenital heart disease occurs in about 50-60% of EVC,hence it is considered as a phenotype [2] .Among the cardiac anomalies, the most common anomaly is common atrium(50%)[1,4]which is present in our case. The other less common anomalies are VSD and hypoplasia of aorta[5,9].Cardiomegaly was reported in EVC[2,6] and in our case also there was cardiomegaly clinically and radiologically. Enlargement of left atrial portion of common atrium is seldom seen in the x-ray even in the presence of significant MR[2], the same was observed in our case .Left axis deviation of the QRS with counter clock wise depolarisation[2] and 1st degree AV block [7]are common ECG findings in common atrium and both are present in our case.

The clinical spectrum of EVC syndrome is wide[1].There are no specific constant feautures in EVC Syndome some may be absent,which doesn't exclude the diagnosis[1].like our patient where there was no teeth anomalies like missing teeth, natal teeth, peg shaped teeth and genitourinary anomalies. Mortality rate is high in infancy about 50%[5]probably due to cardiorespiratory failure and those who survive can have a normal life expectancy as our case[1].

The diagnosis of EVC can be made prenatally or immediately after birth[5]. Prenatal diagnosis is possible by USG after 18 weeks of gestation[3,5,7].Diagnosis at birth can be made by typical symptoms and by skeletal study[1].No definitive cure for EVC syndrome. Treatment is symptomatic and needs multidisciplinary approach with pulmonologist, cardiologist, maxillofacial surgeon and orthopedician as per the need of the affected individuals[1,3].

5. Conclusion

This case is reported because of the rare incidence of EVC syndrome, that too occurring in an Indian population and also to highlight the rare survival upto middle age. Antenatal screening in genetically susceptible individuals and genetic counselling can be done as a preventive measure.

References

[1] Orphanet Journal of Rare diseases,Ellis van creveld syndrome.ojrd.biomedcentral.com/articles/10.1186/1750-1172-2-27.
[2] Perloff's clinical recognition of congenital heart diseases-common atrium,6th edition,chapter 15,page 239-243.
[3] Indian association of oral and maxillofacial pathologists-EVC syndrome:a rare clinical entity,year 2013,vol:17,issue:1.
[4] Journal of cardiovascular ultrasound-well tolerated and undiscovered common atrium until lateadulthood,2016 sep,24(3):243-246
[5] Indian journal of radiology and imaging-EVC Syndrome:year 2002,vol 12,issue 4,page 549-550.
[6] Indian society of pedodontics and preventive dentistry publication – EVC Syndrome :2007,vol 25,issue :5,page 5-7.
[7] Heart India journal-Common atrium with unusual ECG in EVC syndrome,2015vol :3,issue:4.
[8] Late survival in EVC syndrome,2012 cardiology society,pubmed journal.
[9] EVC syndrome in adulthood:extending the clinical spectrum-Singapore medical journal,june 2015;56(6):e110-e111.