COMMON ATRIUM ASSOCIATED WITH POLYDACTYLY AND SHORT STATURE IN A MIDDLE AGE MALE PATIENT - A CASE REPORT

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Abstract:
Polydactyly associated with short stature may serve as a hint for the presence of additional cardiac abnormalities, thus rousing the demand for a detailed cardiac and genetic investigation. In our case diagnosis of most likely Ellis-van Creveld syndrome. We may conclude that prenatal diagnosis of the syndrome can be readily achieved by fetoscopy, fetal echocardiography, and molecular genetic testing by amniocentesis or DNA extracted from chorionic villus samples. Prenatal diagnosis can also be established using mutation analysis of EVC gene from fetal DNA. These cases emphasize the importance of fetal examination for accurate diagnosis of rare syndromes. Education of the general public, especially parents, on congenital anomalies as well as improvement of medical and diagnostic facilities is therefore suggested even if not demanded.

Keyword: Ellis-van Creveld syndrome, polydactyly, common atrium.

INTRODUCTION
Common atrium is a rare variety of interatrial communication characterized by absence or virtual absence of the atrial septum, vestigial remnants of which occasionally remain as diaphanous strands of tissue. The right-sided portion of the common chamber has features of a morphologic right atrium (crista terminalis, pectinate muscles, right atrial appendage) and receives the superior and inferior vena cavae and the coronary sinus. The left-sided portion of the common chamber has features of a morphologic left atrium (smooth nontrabeculated walls, left atrial appendage) and receives the pulmonary veins. Common atrium therefore differs from atrial isomerism with a single atrial chamber that is either a bilateral morphologic right atrium or a bilateral morphologic left atrium. Absence of the atrial septum includes the ostium primum (atrioventricular septum) location, so there is a common atrioventricular valve or a cleft anterior mitral leaflet. Physiologic consequences of common atrium resemble non-restrictive atrial septal defect except...
for obligatory venoarterial mixing. Common atrium therefore a cyanotic malformation with increased pulmonary arterial blood flow. Majority of patients are symptomatic during first year of life with dyspnoea on effort, fatigue, respiratory tract infection, mild cyanosis, physical underdevelopment. Symptoms of common atrium resembles that of nonrestrictive atrial septal defect but are earlier in onset and more pronounced although occasional patients are relatively well into late childhood or early adolescence. Cyanosis sometimes absent or insignificant when the patient is at rest but almost always present after excersise. We describe here such a case of common atrium who presents his symptomatology at adult age.

**CASE REPORT**

A 45 year old male patient presented with insidious onset of easy fatigability, exertional dyspnoea (NYHA Class IV) for past 2 months, associated with swelling of legs for 2 weeks and abdominal distension for 1 week. Patient also gives H/o orthopnea is present. There was no H/o PND, hemoptysis, fever, no past H/o CVA or seizures. There is no clinically relevant family history. On general examination patient was conscious, oriented, afebrile, no pallor, not icteric, ocular fundus normal, short statured, had polydactyly all four limbs, and left lower limb shortening was present. Patient was dyspnoeic, jugular venous pressure elevated, peripheral cyanosis, pan digital clubbing grade - 3, bilateral pitting pedal edema were present. Oral cavity and dentition were normal. Patient’s vital signs normal except respiratory rate is 26 per minute. Patients height 145cm, upper span 71cm, lower span 155cm, arm span 155cm, weight 45kg. Cardiovascular examination on inspection apical impulse apical impulse confirmed in the above position, hyperdynamic in character, palpable P2, parasternal heave present, no thrill felt. On auscultation mitral area S1, S2 heard soft systolic murmur of grade - 3 heard with radiation to axilla. Aortic area S1, S2 heard systolic murmur with no radiation to carotids. Pulmonary area S1, S2 heard with fixed split and loud P2, and an Ejection systolic murmur of grade-3 was present. Left parasternal area Ejection systolic murmur heard. Other system examination Respiratory system on examination showed decreased air entry on the right mammary, infra axillary, infra scapular area. Gastrointestinal system on examination showed distended abdomen, free fluid, and tender hepatomegaly. Central nervous system was clinically normal. This patient remained asymptomatic until adulthood. Routine blood examination was within normal limit. The chest radiograph revealed gross cardio-megaly with bilateral plethoric lung fields and right sided pleural effusion. The ECG revealed left axis deviation, right ventricular hypertrophy and right atrial enlargement. Echocardiogram revealed situs solitus, levocardia, single atrium, inlet VSD closed by septal leaflet of tricuspid valve producing intact ventricular aneurysm, Mitral regurgitation grade - II, Tricuspid regurgitation trivial, cleft anterior mitral leaflet, TRPG 48mm Hg, moderate PHT, coronary sinus grossly dilated probably due to left SVC opening into coronary sinus. Ultra sonogram revealed hepatomegaly, ascites, and right pleural effusion present. Final diagnosis was made as a case of congenital heart disease, atrio-ventricular canal deformity, common atrium, and inlet closed VSD, probable left SVC opening into coronary sinus with polydactyly and short stature. Patient was treated with antifailure
measures and improved symptomatically.

Fig-3, showing patient with common atrium

Fig-1, Picture showing six digits in both hands
Fig-2, Picture showing foot with six toes and left leg shortening

Fig-4, showing no evidence of hypertelorism
Fig-5, showing no evidence low set ears

Fig-6, showing no evidence of dental anomalies

DISCUSSION
Complete absence of atrial septum is rare and least common variety of ASD. Common atrium denotes complete absence of the atrial septum, malformation of the atrioventricular valves, with or without interventricular communication.
Symptoms of fatigue, dyspnea and peripheral cyanosis are the usual manifestations. Physical findings include fixed split, soft systolic murmur over pulmonary area, systolic murmur over apex, radiating towards axilla. ECG shows prolonged PR interval, LAD, RVH. Pulmonary hypertension occurs earlier. Mixing between systemic venous and oxygenated pulmonary venous blood at the atrial level is common due to associated AV regurgitation if any.

Single atrium denotes complete absence of atrial septum without endocardial cushion defect. It is rare variety usually asymptomatic physical findings same as common atrium except for the absence of apical systolic murmur. That is similar to large ASD at the level of fossa ovalis. In these patients pulmonary hypertension had develops at late stage. Complete mixing between systemic venous and oxygenated pulmonary venous blood at the atrial level is rare.

**Fig-7, ECHO showing common atrium**

**Fig-8, ECHO showing gross dilatation of coronary sinus**

An atrioventricular canal defect (Endocardial cushion defects) constitutes a spectrum of lesions including partial AV canal (separate AV orifices), complete AV canal (common AV orifice) and isolated inlet VSD. A persistent left SVC is the most common congenital anomaly involving the systemic veins. It constitutes 3-10% of patients with CHD. In most cases left SVC drains into right atrium by way of coronary sinus. Coronary sinus dilatation (CSD) usually results from anomalous drainage to the sinus from a persistent left SVC or an anomalous pulmonary vein. CSD also occurs due to elevated right atrial pressure contrast enhanced echocardiography (Agitated Saline contrast Echocardiography) is of great value in the differential diagnosis of CSD.

In our case, echocardiography findings led to the diagnosis of most likely Ellis-van Creveld syndrome or chondroectodermal dysplasia which is an important phenotype because 50% of patients with the syndrome have congenital heart disease and half of those have common atrium. It is a tetrad of chondrodysplasia, ectodermal dysplasia, polydactyly and congenital heart disease. The exact prevalence is unknown, but the syndrome seems more common among the Amish community. Many Indian cases have also been reported.

It is an autosomal recessive disorder characterized by short stature with polydactyly of the hands that is invariable and polydactyly of the feet in 10% of cases. Premature eruption of malformed maxillary incisors, gingival...
hypertrophy, and multiple frenula are distinctive features. The syndrome can be caused by mutations in the EVG gene or the EVC2\textsuperscript{10,11} gene. The genes are located close to each other in a head-to-head configuration on chromosome 4p16.\textsuperscript{12} Clinical diagnosis can be confirmed by DNA analysis.

Our patient had all features of Ellis-van Creveld syndrome in the form of polydactyly in all four limbs, shortening of left lower limb, common atrium, atrioventricular canal defect, cleft anterior mitral leaflet except hypoplastic nails dental anomalies with an additional feature of inlet closed VSD.

So our interest of presenting this case is: 1. It is a rare variety of congenital anomaly. 2. Late presentation of clinical manifestation beyond its natural course. Patient had features of nonrestrictive ASD but became symptomatic in fourth decade of life which is beyond the natural history found in literature.

**ABBREVIATION**


**REFERENCES**


