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A PATIENT OF FRACTURE NECK OF FEMUR WITH PECTUS EXCAVATAM AND ACYANOTIC CONGENITAL HEART DISEASE - A CASE REPORT

Medicine

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ABSTRACT

"Pectus Excavatum" is characterised by a depression of the anterior chest wall thus producing funnel chest. Approximately 95 % of congenital wall anomalies are due to Pectus excavatum. This defect is due to involvement of third to seventh costocartilages or ribs and at the level of xiphisternum. This deformity is commonly asymmetrical but symmetrical involvement is also seen. This may be appreciated during infancy or noticed later during childhood. Various case reports are there regarding this deformity and association with variable cardiac and respiratory involvement which may or may not directly related to this .

Here a patient is presented which was basically approached in the orthopedic department for the complain of fracture neck of femur, on examination noticed to be a case of Pectus excavatum and underlying congenital acyanotic heart disease Atrial Septal Defect. Patient had minimal recognisable symptoms due to the deformity or heart disease.

KEYWORDS

Atrial septal defect, Ostium secundum defect, Pectus excavatum, echocardiography

INTRODUCTION

Pectus excavatum is a congenital deformation of the chest which presents as a funnel shaped depression in chest between the fourth and seventh rib. Exact prevalence of this deformity is unknown. Several rare syndromes has been studied found correlated with Ehlers Danlos, Marfan syndrome or Poland syndrome [23, 24, 25]. In 15% patients Mitral valve prolapse occurred.[23,28]. But prevalence of correlation with ASD is not known. There was a descriptive, prospective study done in 42 senior patients with Pectus excavatum which showed complaints of Fatigue , low exercise tolerance, shortness of breath , palpitations, inspiratory obstruction and chest discomfort or pain. A scoring was done called SPES score and Pectus Evaluation Index score(PEI score). SPES ranges from 1 to 10 .The results of the visual inspection of the funnel chest by the surgeon and the radiological analysis explained PEI score. The 0 score is flat or no pectus excavatum and 5 is deep or wedge formed excavation.The Haller Index is derived by dividing the chest diameter by the anteroposterior diameter [26,27].

SPES index>5, PEI score>3 and signs of cardiac compression were found to be eligible for surgery.

SPES SCORING

Clinical complaints	Score		
	1	2	3
Dyspnea	Minor	Moderate	Severe
Palpitations	Minor	Severe	
Low exercise intolerance	Minor/ moderate	Severe	
Chest pain	Present ±	Present	
Ventricular arrhythmia,AV block	Present	Severe	
Mitral Valve insufficiency/ Prolapse/TV insufficiency/ Enlarged RA	Minor	Present	
Treadmill ECG	Stopped in view of exhaution	Present	
Pulmonary function test / inspiratory obstruction	Moderate	Present	

Atrial sepal defect is a congenital defect where there is interatrial communication is present. Types of ASD are ostium secundum , Ostium primum and Sinus venosus . There are rare coronary sinus defects which are closely related [1]. This is one of the common congenital abnormalities seen by cardiologists.

ASD is classified as –

1. Secundum ASD (FOSSA OVALIS) [2,3]
a. The fossa ovalis may be large or the septum primum deficient.
b. 75% of ASDs and 7% of congenital heart defects rising to 40% after 40 years of age.
c. An isolated abnormality.

2. Primum ASD (endocardial cushion defect)
a. 15-20% of ASDs
b. Strong association with trisomy 21
c. A spectrum from primum ASD with cleft mitral valve to atrioventricular septal defect with common AV valve(AV canal) [4]
d. 50% mortality within 1 year if untreated.

3. Sinus venosus defects
a. Inferior defect is associated with anomalous drainage of the right inferior pulmonary vein into the IVC
b. <10%
c. Superior is much more common/ associated with anomalous drainage of the right superior pulmonary vein into the SVC

4. Unroofed coronary sinus
a. Association with persistent left SVC
b. <1%
c. Coronary sinus drains to left atrium

A patent foramen ovale (PFO) is a persistent small tunnel from the fossa ovalis to the foetal ostium secundum. These can be the source of paradoxical emboli when right atrial pressure is elevated (for example Valsalva, pulmonary hypertension).

The prevalence of diagnosed ASDs was found to be 3.89 per 1000 children while 0.88 per 1000 adults. But there may be unidentified and hidden cases due to its clinically silent behaviour. [5]

In this article , clinical manifestations , echocardiography and clinical practice guidelines has been discussed.

The American College of Cardiology and American Heart Association (ACC/AHA) has provided guidelines on the adult congenital heart disease on ASDs [1].There is variable clinical course of ASD [1,6]. Clinical complains from the patients are shortness of breath ,fatigue, exercise intolerance or palpitations.[1]. Few patients complaints of syncope or pedal edema due to right side heart failure and recurrent pulmonary infections.

Various atrial tachyarrhythmias like atrial fibrillation and flutter has been reported preoperatively in 1/5th of adults with ASDs.[7]. However postoperatively Atrial fibrillation and atrial flutter tends to occur in patients above age 40 years.[2]

Those patients with stroke, transient ischemic attack or peripheral arterial embolization has this etiology of paradoxical embolization

always in the question for pathophysiologic mechanism.[9,10]

Due to increased right sided pressure e.g. due to coughing can produce mobile venous thrombus or air embolus to enter into the arterial system if right to left shunt shunt is present across the interatrial septum. In a study of case series of 103 patiens with paradoxical embolization, 12% had ASD.[11]

CASE REPORT

A female patient aged 35 year old came to the orthopedic department for the c/o left sided fracture femur of neck (Intracapsular) . She was a thin built patient . On examination mild pallor was present . BP was 140/88 mm Hg , Pulse 102/min, Spo2 96%. On auscultation , normal vesicular breath sounds with bilateral crepitations was present. On Cardiac auscultation an audible murmur over pulmonary area was present, there was suspicion of defective mitral valve. There was occasional exercise intolerance which was not progressive. Patient was evaluated by an anesthetist for pre-anaesthetic clearance (PAC). Further reference was sent to General physician for evaluation of on and off shortness of breath and possible cardiac abnormality. Following investigation was done.

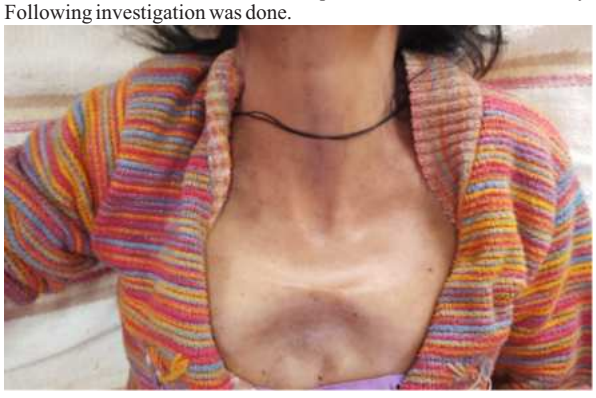

Figure 1 A 35 year old female patient with Pectus excavatum with acyanotic congenital heart disease ASD (large) Ostium Secundum type ASD

ECG showed normal sinus rhythm and Right axis deviation . There was Rsr' pattern and V1 showing excessive overload of left atrium.

HRCT Thorax findings –

Soft tissue and bony cage including sternum and vertebral column normal, no significant mediastinal or hilar lymphadenopathy, no axillary lymphadenopathy, Mediastinal contents including unopacified heart and major mediastinal vessels appear normal. No pericardial effusion/pericardial thickening. Tracheo-bronchial tree appear normal .**Fibrotic bands noted in bilateral lower lobes**. No obvious lung lesion/septal thickening or bronchiectatic changes.

Colour doppler 2D echo –

M-MODE MEASUREMENTS
Mitral study – DE- 1.4 cms
DF-0.05 mm/sec
EPSS – 0.6 mms

Ao ROOT 1.9 cm
LA- 2.0 cm
AO VALVE OPENING – 1.4 cm
LV study- IVS(d)= 0.6 cm IVS (s)=0.9 cm
LVID(d)= 3. Cm LVID (s) 1.4CM
LVPW(d) 0.6 cm LVPW(s) = 1.2 cm
LVEF= 88%
LVFS = 57%

2D OBSERVATION – Left Ventricle normal
Right Ventricle dilated
Left atrium Dilated
Atrium septum ASD +

Mitral valve e- 0.58 m/sec
a - 0.51 m/sec
Aortic valve – 0.75 m/sec
Tricuspid valve e- 0.77 m/sec
a-0.43 m/sec

Pulmonary valve- 1.53 m/sec
SD Flow 1.03/4
TR 3.03/36

Impression- Congenital Acyanotic heart disease
Atrial Septal Defect (large)
Ostium Secundum type ASD (2.8 cm)
Left to Right Shunt
Pulmonary/systemic blood flow (Qp/Qs) is 1.8:1
Pulmonary Hypertension RVSP >46 mm Hg
Ejection Fraction 88%

BLOOD REPORTS

Hb- 10.2 gm/dl , TLC 13,400 /cmm

platelet count 6.85 $\times 10^3$ µl, Serum protein 5.38gm%, corrected reticulocyte count 1.2 %

GBP- Reduced red cell density,mild anisocytosis,predominantly normocytic normochromic with microcytic hypochromiccells, neutroplhilic leucocytosis, thrombocytosis.

Serum uric acid 2.59mg/dl
Serum Albumin- 2.34 g/dl
PT/INR 12/1.42 sec
KFT normal
LFT normal
Troponin I 7.0 ng/L
HIV -ve, HBsAg -ve, HCV -ve
HsCRP 112.42 mg/l
Serum LDH 259.50 U/L
Serum calcium total 8.7 mg/dl
Serum Calcium (Ca++) 1.05 mmol/l
Serum Alkaline Phosphatase 116 IU/ML
Serum sodium 144 mmol/l
Serum potassium 3.78 mmol/l

As patient was having fracture of femur, she was undertaken for operation under spinal anesthesia which was uneventful and there was no post operative complications . The patient was further advised to visit to cardiovascular thoracic surgeon for ASD closure.

DISCUSSION

In this patient , there was on and off h/o shortness of breath which was non-progressive. On physical examination there was parasternal heave , systolic flow murmur at pulmonary area. There was fixed P2A2 i.e. reversible splitting of S2.

There was pulmonary hypertension in this patient. If fixed pulmonary hypertension develops , this marks a late disease and decreased survival. In a sample of 295 Belgian patients where 19% women with age 46±21 years with isolated secundum ASD had prevalence of pulmonary hypertension in 16% of cases.[12] Pulmonary hypertension has increased mortality, atrial arrhythmia and right heart failure. Eisenmenger syndrome, has combination of pulmonary hypertension and right to left shunting which leads to increased incidence of thrombosis, bleeding and pregnancy complications. [13]

Here in this patient, ECG was suggestive of right axis deviation, right atrial enlargement , right ventricular hypertrophy and RBBB. This was in favour of secundum ASD.[1.2] characteristics of Familial ASD.[14]

Echo Evaluation –

The echo guidelines needs demonstration of shunting across the interatrial septum, and evaluating right heart and associated abnormalities.[1].

Following are the elements of a comprehensive echo evaluation for ASD-
1. Visualisation of ASD and its size.
2. Examination of right heart
3. Direction of flow
4. Measurement of pulmonary artery pressure

5. Estimating pulmonary/systemic flow ratio
6. Associated abnormalities (mitral valve prolapse, partial anomalous right pulmonary veins- present in sinus venosus ASDs and some secundum ASDs and cleft anterior mitral valve leaflet- primum ASDs.

Though full 2D TTE with doppler does not establish the diagnosis of ASD , an agitated saline contrast study can be performed to detect an ASD[1] and 100% of ASD will be detected with a high quality agitated saline study including maneuvers(Valsalva,cough) to promote transient right to left shunting [15]. But in present study , 2D doppler echo has demonstrated ASD and other related findings.

There is potentially useful non-invasive Doppler estimate , ratio of the pulmonary/systemic blood flow (Qp/Qs): [(pulmonary artery diameter)^2× left ventricular outflow tract velocity time integral].

Guidelines recommend use of Parasternal short-Axis view at the base of the heart [16], but subcostal view may be considered also. Though Qp/Qs is equal to cardiac catheterisation . [17]

In patients of diagnostic confusion, cardiac CT and MRI can be the modality of choice.[18,19]

Exercise testing is recommended who have mild to moderate pulmonary arterial hypertension but not in severe pulmonary hypertension.[1].

Here patient was advised not to plan for pregnancy and limiting activity [13,21]

MANAGEMENT

Therapy was directed towards management of atrial fibrillation.[1]. Advising anticoagulation and restoration of sinus rhythm. Patient was advised to ASDs closure.

The primary indication for ASD closure was right heart volume overload(right atrial or ventricular enlargement) [1]. Recommendation of intervention is required when right heart is involved even in absence of symptoms. Other indications for ASD closure to be considered if left to right shunting, pulmonary artery pressure or pulmonary vascular resistance < 2/3 systemic levels.In this patient Qp/Qs was 1.8:1 . A standard approach was to repair if Qp/Qs>2 but some experts were in opinion to go for repair at lower threshold of 1.7[22] or 1.5 [8]. In such cases endothelin -1 can be the useful diagnostic test when evaluating ASD closure along with other clinical symptoms.

Surgical closure is recommended whenever tricuspid valve repair or replacement is planned. Also Ostium primum, sinus venosus ASD or coronary sinus defect indicates surgical intervention.

Post surgery, aspirin 81-325 mg daily was recommended for next 6 months with clopidogrel 75mg daily . Endocarditis prophylaxis was also indicated for 6 months post surgery.

Learning Points-

The external appearance of deformity does not always relates to the severity of disease. There was underlying congenital heart disease but patient remained asymptomatic till investigations made the diagnosis of ASD. There is study on association with Marfan syndrome but any genetic basis with Acyanotic heart disase is there , these needs further studies. Here in the CT thorax report there was no mention of Haller Index . Only fibrotic bands were reported in HRCT thorax. This could be misleading about the severity of underlying disease. There was start of respiratory symptoms as fibrosis bilaterally . Therfore any patient with Pectus Excavatum must be corrected surgically as soon as possible due to increasing complications with increasing age.

ETHICAL APPROVAL-

The ethical approval was not required as patient's identity was not disclosed. Consent was taken from the patient.

CONFLICTS OF INTEREST

The authors declare that there is no conflicts of interest.

Author's Contribution

Jyoti Verma wrote the manuscript , did the conception and design of study ,and did timed reference to CTVS department , Indreshwar verma operated for the fracture of neck of femur and took responsibility in gathering the data and wrote the critical review , Gopal Singh contributed in literature review and involved in patients care.

All authors contributed equally to the article.

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