

OSTIUM PRIMUM TYPE OF ATRIAL SEPTAL DEFECT WITH CLEFT ANTERIOR MITRAL LEAFLET – A RARE CASE REPORT

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ABSTRACT

The atrial septal defect of ostium primum type with cleft anterior mitral leaflet is a rare congenital cardiac defect. It contributes for about 4% of congenital heart diseases and occurs in 2 in 10000 live births. A 12 year old girl's echocardiography showed this anomaly when the patient presented with breathlessness to the Department of Radiology, Chettinad Medical College and Research Institute, Chennai. This study focuses on the anatomical knowledge along with embryologic development of inter atrial septum and endocardial cushions which provide the basis for better understanding of pathophysiology and clinical manifestations of this rare anomaly.

Key words : *Atrial septal defect (ASD), ostium primum, cleft mitral leaflet, endocardial cushion, atrioventricular (AV)*

INTRODUCTION

Atrial septal defect is a hole in the inter-atrial septum. When ASD is present, the blood flows through the defect primarily from the left to right atrium. This shunting increases the blood volume in right atrium which means more blood flows through the lungs than would normally. The mitral leaflets are originally derived from the endocardial cushions¹. The congenital cleft formation in otherwise normal mitral valve usually presents with concomitant cardiac anomaly, mainly atrial septal defect. The congenital cleft of mitral leaflet is a rare cause of mitral insufficiency resulting from various

degrees of failure of fusion of endocardial cushions². Hence the present study provides a detailed study of the anatomical and embryological aspects of ASD of ostium primum type with associated cleft anterior mitral leaflet.

MATERIAL AND METHOD

A 12 year old girl's echocardiography in Chettinad Hospital and Research Institute, Chennai was used for the present study.

RESULTS

2D Echocardiography showed ostium primum type of atrial septal defect of 1.8mm, severe tricuspid regurgitation and mild mitral regurgitation with cleft anterior mitral leaflet (Figure 1). In the short axis, the left atrioventricular valve assumes a triangular rather than a 'fish mouth' appearance in diastole.

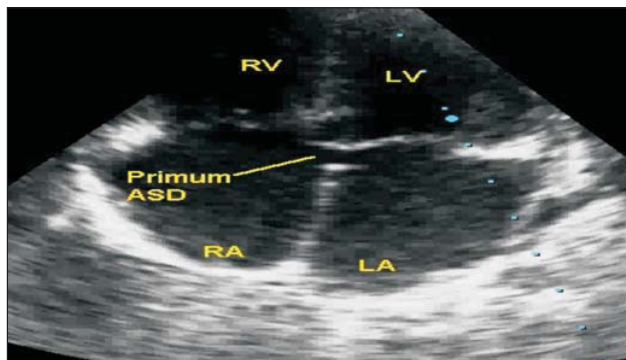


Figure 1: Echo cardiography showing ASD.
RA – Right atrium; RV- Right Ventricle; LA – Left atrium; LV – Left ventricle;
Primum ASD – ostium primum type of atrial septal defect

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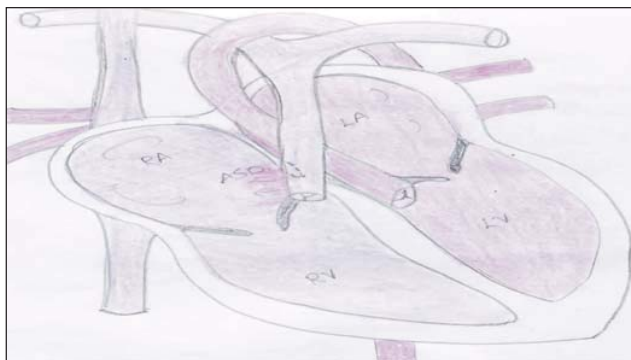


Figure 2: Schematic diagram showing ASD.
 RA – Right atrium; RV- Right Ventricle; LA – Left atrium; LV
 – Left ventricle;
 Primum ASD – ostium primum type of atrial septal defect

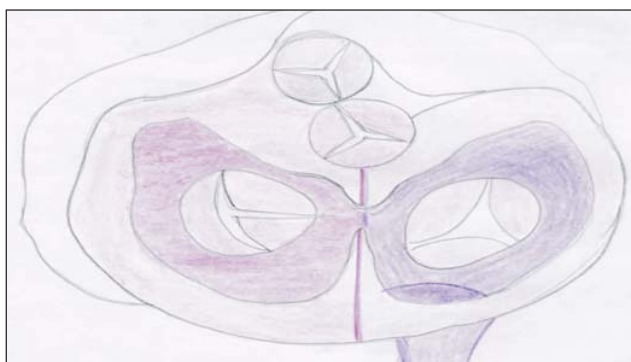


Figure 3: Schematic diagram showing
 PV – Pulmonary valve; AV – Aortic valve; TV – Tricuspid
 valve; CMV- Cleft mitral
 Valve; ASD – Atrial septal defect; LA- Left atrium; RA- Right
 atrium

DISCUSSION

This rare anomaly results due to the defective fusion of endocardial cushions with the septum primum. It occurs in 4% of all congenital heart defects, the incidence being 2 in 10000 live births. Patients with this anomaly may be asymptomatic or present with symptoms of complications. Down's syndrome is the common non-cardiac anomaly which is usually associated with ostium primum type of ASD and cleft anterior mitral leaflet³. Genetic syndromes such as DiGeorge syndrome, Ellis-van Creveld syndrome should also be considered. A higher

incidence of this anomaly has been reported in foetal series (17%)⁴.

Anatomical Description:

In this anomaly, the defect is due to deficiency in the development of anterior and posterior endocardial cushions and their fusion with septum primum. The left AV valve annulus is displaced downwards. So when there are two separate atrioventricular valves, absence of the atrioventricular septum is identified by origin of the leaflets of atrioventricular valves at the same level from the crest of ventricular septum. When the bridging leaflets are attached to the crest of ventricular septum either directly or through fused chords, an isolated atrial shunt exists. But there can be no interventricular shunt. This variety of atrioventricular septal defect has been called as 'ostium primum' (figure2) and typically co-exists with a morphologically abnormal and usually functionally incompetent left atrioventricular valve. The septal leaflet of mitral valve appears to be split into anterior and posterior components. The gap between the two left sided components is the "cleft" of mitral valve (figure 3). The cleft in the anterior mitral leaflet is oriented towards the ventricular septum because the 'cleft' is a functional commissure between the anterior and posterior bridging leaflets. The free edges of the cleft of mitral valve are either floating free or are attached with the small chordae on the crest of ventricular septum. These abnormal attachments limit the coaptation of mitral valve leaflet and further increase the mitral insufficiency. As the septal leaflet of mitral valve is split, the papillary muscles are also abnormal, mostly lying closer and sometimes fused in a single group. The thickness of tissues of mitral valve is increased. These factors may determine the clinical picture and also the surgical outcome.

Embryological description:

On the 28th day of gestation, the common atrial cavity is divided into two parts by the formation of atrial septum. The subendocardial tissue proliferates all around the opening of the AV canal, so that it becomes surrounded by a ridge of tissue which pushes up the endocardium to form the atrioventricular cushions. A septum from the roof of the common atrial chamber called septum primum grows downwards on Day 35 towards the AV cushions and a foramen is formed between its lower free margin and AV cushions called "ostium primum". Before it meets AV cushions, there is a local failure of the septum primum in the upper part forming a foramen called "ostium secundum". The septum primum fuses with the part of the AV cushions in the middle of the common AV canal. At the same time, the septum secundum is formed from the roof of the common atrial chamber on the right side of septum primum and this result in the formation of "foramen ovale" by means of which the right and left atria are kept in communication throughout the whole of foetal life. When there is deficiency in the development of anterior and posterior endocardial cushions and their fusion with septum primum, atrial septal defect (ostium primum type) with associated cleft anterior mitral valve exists.

CONCLUSION:

In this type of anomaly, the clinical presentation is governed by the magnitude of left to right shunt and the degree of mitral regurgitation. The surgical repair of this malformation is well described. The success of the surgery depends on the repair of mitral regurgitation since it may remain even after surgery. Computer modeling suggests that this anomaly is more likely to be due to single gene

rather than several genes⁵. Antenatal diagnosis is possible during 16-18 weeks of gestation using 2-D echocardiography by transabdominal approach. Finally genetic counseling is recommended.

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