

ATRIOVENTRICULAR SEPTAL DEFECTS

DEFINITION AND CLASSIFICATION

The primordial single atrium divides into right and left sides by formation and fusion of the septum primum and septum secundum. The septum primum grows from the primordial atrial roof toward the endocardial cushions, and the septum secundum grows from the ventrocranial atrial wall on the right side of the septum primum. The true septum between the two cavities, however, is not nearly as extensive as generally believed.¹ Only the floor of the oval fossa, derived from the *primary atrial septum*, or "*septum primum*", and its *antero-inferior buttress*, formed by muscularisation of the vestibular spine and mesenchymal cap, which then represents the second atrial septum, are made up of walls that directly separate the atrial cavities.² The superior margin of the oval fossa, often described as the "septum secundum", is no more than a deep interatrial fold.

Atrioventricular septal defects (AV canal or endocardial cushion defects) are **complete** (large VSD, common AV junction, and common AV valve with five leaflets), or **partial** (ostium primum ASD with a common AV junction but two separate AV valves) (Figures 1 and 2).^{3-5 6}

Ostium primum defect at the lower part of the atrial septum is also, rather incorrectly, called partial atrioventricular septal defect, and may or may not have a VSD component (15% of ASDs) (Figure 2).

Ostium secundum defect involves the region of the fossa ovalis (80%). Of note, the interatrial channels described as being "secundum" defects represent the secondary interatrial foramen as formed during cardiac development.¹ They are the consequence, nonetheless, of deficiency of the primary atrial septum, which forms the floor of the oval fossa. They are not defects of the true second atrial septum.

Vestibular defects exist in the true second atrial septum, which is the antero-inferior buttress of the fossa. Such defects are more intimately positioned near the atrioventricular node, potentially increasing the risks in disturbing the conduction system.²

Sinus venosus defect at the junction of the right atrium and superior vena cava (5%).

Coronary sinus defect ("unroofed" coronary sinus) are very rare (<1%). Sinus venosus and coronary sinus defects are actually interatrial channels rather than septal defects.

Patent foramen ovale is the incomplete septal partition (usually an oval-shaped window) at the point where the septum secundum overlaps perforations of the septum primum (ie the foramen secundum).

Complete AV canals rarely reach adulthood without Eisenmerger syndrome. Partial AV canal defects (including ostium primum ASD) are not uncommon in adults.

OSTIUM PRIMUM ASD

Epidemiology

All kinds of AVSDs represent one third of the cases of congenital heart disease detected in adults, with an estimated incidence of 5.6 per 10000 live births.^{4,7}

Aetiology

Approximately 40% of patients with Down syndrome have an AV septal defect, usually complete.³ Primum ASD may also be associated with DiGeorge, Ellis-Van Creveld, and Noonan syndromes. Gender distribution is equal for ostium primum ASD. Adults with AV septal defects have an approximate 3-10% risk of recurrence in their offspring (excluding familial ASD and heart-hand syndromes with autosomal dominant inheritance).⁸

Abnormalities in genes essential to cardiac septation have been associated with atrial septal defects, including mutations in the cardiac transcription factor gene *NKX2.5* (associated with conduction abnormalities), *GATA(4-6)*, *TBX5*, *MYH6*, and other genes.^{3,9}

Pathophysiology

In ostium primum ASD there is a cleft (trileaflet) mitral valve and results in variable degrees of regurgitation.^{3,8} The shorter distance from the left AV valve annulus to the left ventricular apex compared to that from the apex to the aortic annulus combined with the cleft mitral valve creates the characteristic “gooseneck” deformity that used to be a major diagnostic feature on left ventriculography. The elongation of the left ventricular outflow tract as well as the chordal attachments of the left AV valve to the ventricular septum is the reason for the development of LV outflow tract obstruction which may occur even late after successful repair of the defect and require reoperation. The abnormal AV junction affects the AV conduction tissue, which in turn produces the characteristic left-axis deviation and predisposes these patients to heart block. Most primum ASDs are relatively large and lead to right heart dilation. Right atrial dilation and stretching predispose to atrial flutter and fibrillation. The pathophysiology of isolated primum ASD is similar to this of a large secundum ASD.

Presentation

Depending on the severity of dysfunction of the left AV valve, adult patients with ostium primum ASD may become symptomatic at a much younger age than patients with other types of ASD.

Physical Examination

Physical signs as in secundum ASD (see below) but there is usually an additional **pansystolic murmur** due to MR (or TR). If there is also a ventricular defect, signs resemble those found in large VSD with MR or TR. **Cyanosis** suggests pulmonary hypertension or pulmonary stenosis.

Investigations

Chest radiography in ostium primum ASD may be normal; otherwise resembles that of secundum ASD. Coexistent VSD usually is associated by cardiomegaly and pulmonary plethora.

ECG Long PR (unless if AF or flutter), left axis deviation, RBBB with RV hypertrophy. Development of right axis in primum ASD suggests pulmonary hypertension. Patients should be monitored with annual ECG and periodic monitoring for dysrhythmias (AHA 2015 scientific statement, on ACHD, I-C).¹⁰

Echocardiography and **cardiac magnetic resonance** have replaced angiography as the main diagnostic tool for documentation of the type and size of the ASD, direction of the shunt and pulmonary venous return.

Maximal **exercise testing** can be useful to document exercise capacity in patients with symptoms that are discrepant with clinical findings or to document changes in oxygen saturation in patients with pulmonary hypertension.

Cardiac catheterization is used only to assess pulmonary hypertension and test vasoreactivity in patients with repaired or unrepaired ASD (Table 1).

Therapy

Primary surgical repair is recommended for partial AV canals provided there is no irreversible pulmonary hypertension (Tables 2 and 3). Peri-operative mortality is 3.2 (0-6.5)%.¹¹ In cases of residual interatrial or interventricular communications, endocardial pacing causes an elevated risk of paradoxical emboli, and epicardial pacing may be required.

Endocarditis prophylaxis

It is recommended only in high-risk patients with repaired ASDs. Revised indications are discussed in Chapters 1 and 82 on Endocarditis.

Pregnancy

After ASD repair, pregnancy is usually well tolerated (WHO risk class II–III), although arrhythmias and worsening atrioventricular valve regurgitation have been described (Table 4).¹² All women with a history of AVSD should be evaluated before conception to ensure that

there are no significant residual hemodynamic lesions that might complicate the management of pregnancy. The issue of pregnancy risk and preventive measures should be discussed with women with Down syndrome and their caregivers. The recurrence risk of congenital defects is up to 14%,¹³ and genetic counseling is indicated.

References

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