

Partial atrioventricular canal

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Abstract

Partial atrioventricular canal is due to a defective fusion of the endocardial cushions with the atrial septum primum. In the normal heart this fusion constitutes the atrioventricular septum where the mitral and tricuspid annuli insert, dividing the septum into an interatrial and an atrioventricular component. It accounts for about 4% of all congenital heart defects and its incidence is estimated to be 2 per 10,000 live births. It includes a spectrum of anomalies that differ from those of the complete form because the ventricular septal defect is absent. In partial atrioventricular canal two separate atrioventricular valve annuli are usually present: forms with a common annulus are referred as intermediate. Patients with these lesions may be asymptomatic or present with a variety of symptoms depending mostly on the function of the atrioventricular valves and the associated anomalies. Treatment of partial atrioventricular canal is invariably surgical and can be done electively in the first few years of life (uncomplicated forms) or in the first few months of life when symptoms are severe. Operative risk of all atrioventricular septal defects is approximately 3%. Twenty-year survival is 96%.

Keywords

Endocardial cushion defect, atrioventricular canal, atrioventricular septal defect, congenital heart disease

Disease name and synonyms

- Partial atrioventricular (AV) canal
- Ostium primum atrial septal defect
- Partial atrioventricular septal defect
- Endocardial cushion defect

Differential diagnosis

Patients with anomalous pulmonary venous return or secundum atrial septal defect may resemble clinically those with partial AV canal. In total anomalous pulmonary venous connection, cyanosis is always present. It may be sometimes difficult to differentiate also on echocardiography between partial or intermediate AV canal and

pulmonary hypertension with a complete form where the ventricular septal defect (VSD) may be obscured by the septal insertion of anterior and posterior common leaflets. In rare instances partial AV canal with intact atrial and ventricular septal structures have been reported (Silverman). In isolated cleft of the mitral valve the cleft is oriented toward the aortic annulus because the aorta maintains its wedged position into the atrioventricular groove (di Segni).

Frequency

Among congenital heart diseases, incidence of atrioventricular canal defects is reported to be 17% in fetal life (Cook) and 4-5% in patients

born with a congenital heart defects. Occurrence is estimated to be 0.19 in 1,000 live births. Forty percent of subjects with Down's syndrome have congenital heart disease and, among these, 40% have AV canal usually in the complete form. Partial AV canal is common in non Down's patients who present with peculiar features.

Clinical description

Anatomical description

The defect is due to a deficient development of superior and inferior endocardial cushions and their fusion with the septum primum. Several consequences derive from this anomaly: the left AV valve annulus is displaced downwards and mitral and tricuspid valve annuli insert at the same level on the interventricular septum and therefore the AV portion of the septum is missing; the inlet portion of the ventricular septum is shortened and the ratio inlet/outlet septum is less than 1 (normally about 1); the aorta loses its wedged position onto the heart and the left ventricular outflow tract has the typical elongated appearance called "goose-neck deformity"; the lack of fusion of the atrial septum primum with the endocardial cushions constitutes the interatrial communication and may vary in size from very small to a single atrial cavity with small strands of remnant tissue crossing the atrium. In these instances development of the septum secundum is also abnormal; the septal leaflets of the mitral and the tricuspid valves look split into an anterior (or superior) and posterior (or inferior) component. The gap between the two left-sided components is the "cleft" of the mitral valve. The distal ends of the anterior mitral leaflet insert onto the antero-lateral and postero-medial papillary muscles, but the free ledges of the cleft are free-floating or insert with short chordae on the ventricular septum. These abnormal attachments may limit the leaflet coaptation and increase further the mitral valve insufficiency. The septal leaflet of the tricuspid valve is also abnormal with absent anterior portion of the septal leaflet and widened antero-septal commissure. Due to the split septal leaflet of the mitral valve the papillary muscles are also abnormal, usually closer to each other and sometimes fused in a single papillary muscle group. The tissue of the atrioventricular valves may be very abnormal with increased thickness, redundant tissue and myxomatous appearance. The redundant tissue may be responsible for left ventricular outflow tract obstruction. Together with the anatomy of the papillary muscles these features represent the major determinants of the clinical picture and of the surgical outcome. In the so-called intermediate form, there is a common atrioventricular valve orifice with a scooped-down interventricular septum, but the insertion of the anterior and posterior common leaflet

prevents the occurrence of an interventricular communication.

In partial atrioventricular canal we include the intermediate forms and the single atrium without anomalous pulmonary or systemic veins connections. Complete forms, AV canal type of ventricular septal defect, isolated cleft of the mitral valve are excluded. AV canal associated with heterotaxy is also excluded.

In this review paper the lesion will continue to be referred as partial AV canal and the AV valves will be called mitral and tricuspid valves.

Clinical evaluation

When prenatal diagnosis has not been made, uncomplicated partial AV canal may go unrecognized till later in childhood or young adult life. Most commonly diagnosis is made early in infancy when a complete cardiac examination is requested because of the presence of a heart murmur. Patients with Down's syndrome are more likely to be diagnosed in the prenatal period or early, *i.e.* soon after birth. The physical examination does not show anything particular except for the presence of a pansystolic murmur due to mitral valve regurgitation and/or an ejection systolic murmur along the left sternal border due to increased pulmonary blood flow. Second heart sound may be widely split and fixed with respiration.

Complicated forms of partial AV canal are those with severe mitral valve regurgitation, left ventricular outflow tract obstruction, coarctation of the aorta or interrupted aortic arch. The clinical picture of partial AV canal with interrupted aortic arch is that of a ductus-dependent systemic circulation. All other complicated forms of partial AV canal show symptoms of congestive heart failure, failure to thrive and secondary pulmonary hypertension. Symptoms may occur as early as the first few weeks in those with more severe mitral valve insufficiency or left ventricular outflow tract obstruction. The young baby looks pink or presents with mild cyanosis, tachypnea, dyspnea with hyperinflated chest: turgor of the jugular vein with prominent V wave pulsation is observed; hepatosplenomegaly and poor peripheral pulses (if coarctation is present, femoral pulses may be absent) occur, precordium is mildly hyperactive and a right ventricular impulse is palpable. A right ventricular heave reveals pulmonary hypertension. The second sound is widely split with a loud second component; there is a pansystolic murmur of different intensity with or without a thrill best heard at the apex and radiating both to the left axilla and toward the right upper sternal border. This is due to the direction of the mitral valve regurgitation through the cleft anterior leaflet and directed from left ventricle (LV) to right atrium. There may be a gallop rhythm due to both reduced ventricular

compliance and increased diastolic flow from the AV valve regurgitation and left-to-right shunt. Auscultation of lungs may reveal fine rales or bronchospasm like in bronchiolitis.

Diagnosis

Diagnosis of partial AV canal can be established in fetal life by echocardiography performed with trans-abdominal approach at 16-18 weeks gestation. Likewise, it is the major postnatal diagnostic tool. The use of all ultrasound techniques (2-D imaging, M-Mode, wave and color Doppler) allows a complete anatomical and physiological evaluation of the malformation. Surgery is only recommended on a clinical and echocardiographic basis.

Partial AV canal has to be suspected in all neonates or infants with Down's syndrome. In non syndromatic children, clinical features suggestive of partial AV canal are: pansystolic murmur of mitral regurgitation, widely-split second heart sound, failure to thrive, overt heart failure.

Several methods can be used:

Chest X-rays

Uncomplicated forms may be unnoticed or show mild enlargement of all cardiac chambers due to the dilation of the right heart because of the atrial septal defect and of the left ventricle because of mitral valve regurgitation.

Chest X-rays are performed whenever required. In complicated forms it may be needed to rule out pulmonary infection which is sometimes difficult to exclude in an infant with heart failure. In non complicated forms chest X-rays add little to the clinical and echo picture and is usually performed only preoperatively.

Echocardiography (ECG)

Constant ECG features are left axis deviation and first-degree AV block (due to an abnormal course of the conduction tissue within the ventricular septum, secondary to the absence of the AV septum) and incomplete right bundle branch block (like in secundum atrial septal defect). Complicated forms may present with biventricular hypertrophy, right ventricular hypertrophy, right or biatrial enlargement.

Echocardiography, especially in young infants or children, provides all the information needed to take the necessary measures for each case. In an asymptomatic infant clinical and echocardiographic follow-up will be established at an appropriate frequency until the elected age for the surgical correction is reached. For an infant with severe mitral valve insufficiency, hospital admission will be required to start medical treatment and take other appropriate measures to stabilize the clinical conditions before surgery is undertaken.

Echo features

Morphology

All echo views should be studied carefully.

The most important echo feature of partial AV canal is the lack of offsetting of the septal insertion of the 2 atrial valves. The interatrial communication is always present and it varies in size, from very small to an almost complete absence of interatrial septum. Few strands of tissue may cross the atrial cavity as a remnant of the atrial septum. Systemic and pulmonary venous return is normal. Atrioventricular and ventricular-arterial concordance is observed. The mitral valve always shows the typical cleft which is best seen in the left parasternal short axis view as an opening within the septal leaflet directed toward the interventricular septum. The free edges of the cleft do not attach into the papillary muscles, but may attach to the septum thanks to short chordae tendineae causing reduced valve excursion and further increasing mitral insufficiency. The tricuspid valve is also abnormal with reduced anterior component of the septal leaflet and a widened antero-septal commissure. The distance between the 2 papillary muscles in the left parasternal short axis view is reduced and the chordae may be shortened and thickened. An important feature to describe is the presence of one papillary muscle group. The tissue of the mitral leaflet may be also redundant, thickened with myxomatous edges. The short axis view shows the relative and absolute size of the 2 ventricular chambers. Volumes of the left ventricle should be measured carefully in both the short and apical 4 chamber view when true dominant right ventricular forms are suspected. The best view to analyze the AV valves and their connection within the ventricular chambers is the apical 4 chamber view. True partial (2 AV valve annuli) and the intermediate form of AV canal may also be apparent in this view, but discrimination is not always easy. Dominant left or right partial AV canal is also possible. The dominant right forms are those with small left AV valve orifice, single papillary muscle, obstruction to the left ventricular outflow tract and are more common in non-Down's patients. Left ventricular outflow tract obstruction due to redundant tissue of the abnormally displaced mitral valve or to subaortic diaphragms is best seen in the left parasternal or apical long axis views. The typical reduced ratio between the inflow and outflow septum is also apparent in these views. The aorta is usually morphologically normal but its dimensions are in the lower range. Coarctation of the aorta or interrupted aortic arch should be ruled out because they are not uncommon associated lesions.

Physiology

Physiology of partial AV canal is evaluated mainly by M-mode and Doppler echocardiography.

The M-mode confirms the right ventricular volume overload typical of other forms of left to right shunt at atrial level. Increased right ventricle (RV) dimension with paradoxical septal motion is observed. The dimensions of the LV should be in the normal range.

Wave and color Doppler are essential to evaluate the AV valve function and to semiquantitate the insufficiency. The regurgitation of the mitral valve may be mainly through the cleft or also centrally from lack of coaptation of the leaflets. Regurgitation from the cleft is often directed from left ventricle to right atrium. This feature may prevent the correct measurement of the RV pressure through the tricuspid valve regurgitation. In the apical 4-chamber views it may be possible to separate the 2 jets and make a correct prediction of the RV and pulmonary artery pressure. Peak and mean gradients across the left ventricular outflow tract or across the aortic arch may be measured with continuous wave Doppler.

Cardiac catheterization

Cardiac catheterization and angiography should never be performed in uncomplicated partial AV canal. Invasive study is required occasionally to confirm associated anomalies such as coarctation of the aorta or to measure pulmonary artery pressure and resistance when doubts exist as to the presence of pulmonary vascular disease or to rule out peripheral pulmonary artery stenosis.

Other diagnostic techniques

Magnetic resonance imaging (MRI)

New advances in cine magnetic resonance make MRI a useful technique in the diagnosis of AV canal defect and its associated anomalies. The major problems are the costs and reproducibility that make ultrasound techniques preferable. The application of MRI is useful in older subjects especially to define associated malformations such as aortic arch anomalies.

Radionuclide angiography

It has had little application and is almost completely abandoned in the diagnosis of partial AV canal.

In summary, echocardiogram is the gold standard tool for the diagnosis of partial AV canal both for the uncomplicated and complicated forms of the defect. Decision as to when to recommend surgical correction is made on clinical grounds. Antifailure measure is started in symptomatic young infants with severe mitral valve insufficiency before undergoing surgical repair. The age for the operation is determined by the clinical conditions and progress. In some instances it is necessary to intervene as early as 3-6 months of life. The determinants for surgical repair are refractory heart failure and failure to thrive. Asymptomatic children are operated upon between 2-5 years of

age depending on the surgical experience and results of the centre.

Natural history

Patients with untreated partial AV canal develop symptoms earlier than those with secundum atrial septal defect. As mentioned before symptoms such as congestive heart failure, failure to thrive and low cardiac output may develop in the first few weeks or months of life when mitral valve malfunction is relevant. In older patients, arrhythmias, right heart failure, pulmonary hypertension may develop.

Treatment

Medical treatment

Medical treatment is indicated in all infants showing symptoms of congestive heart failure early in life before they undergo surgical correction. This is a small portion of babies with partial AV canal (about 10%) in whom surgery is delayed because of the technical difficulty for the correction of the mitral valve insufficiency and the risk for mitral valve replacement. The mortality for surgical repair of partial AV canal is 3% and the determinants of hospital mortality are: heart failure, failure to thrive, age at operation and moderate to severe mitral valve insufficiency. The medical treatment consists in load reducers such as ACEIs (Angiotensin Converting Enzyme Inhibitors) and diuretics. The dose for captopril is 2-3mg/Kg/dose: 3-4 doses daily; frusemide is given at 1-2 mg/Kg/die in one or two doses; other measures include gavage feeding or continuous enteral nutrition to reduce energy consumption and high-calorie formula. Too severe fluid restriction is not recommended.

Surgical palliation

Pulmonary artery banding in partial AV canal with severe heart failure is not recommended and has been abandoned. If aortic coarctation is present, surgical repair of the coarctation is done first.

Surgical repair

The mortality for surgical repair of partial AV canal is 3% and the determinants of hospital mortality are: heart failure, failure to thrive, age at operation and moderate to severe mitral valve insufficiency.

The mortality is significant in a subgroup of partial AV canal usually not associated with Down syndrome. This group of patients develops congestive heart failure and failure to thrive early in life, requires intensive medical treatment and often undergoes surgical repair in the first year of life (Giamberti).

In uncomplicated partial AV canal, surgical correction is made in the preschool age. Because of the advances in the surgical techniques, heart preservation and postoperative intensive care, recommended age for repair of partial AV canal is between 2-4 years depending

on the experience and results of the centre. The operation consists in the patch closure of the atrial septal defect and the suture of the cleft mitral valve. This leads to a reconstitution of a bileaflet mitral valve. The belief that the cleft left unsawn is a better option has been abandoned by many surgeons, but individualized correction is needed because of the high variability in valvar morphology and function. Other manoeuvres on the mitral valve such as annular reduction, chordal shortening or elongation may be needed. Intraoperative transesophageal or epicardial echocardiography has added greatly to the immediate assessment of the AV valve function after correction. Mitral valve replacement in a young infant is always avoided even if the mitral valve repair is suboptimal. In the few instances where mitral valve replacement has been required the choice is between a mechanical valve or a biological valve. A new operation called Ross-Kabani uses the pulmonary autograft in a reverse position to replace the mitral valve and an homograft on the pulmonary valve. This operation uses the same principle as the Ross operation for aortic valve disease in pediatric age, but has been performed in a very limited number of cases and only in one case in an infant of 3 Kg of weight.

Reported hospital mortality for correction of partial AV canal is about 3%, but lower mortality is achieved in more recent series (1.6% San Donato Hospital-Personal communication). Outcome reveals a 96% survival at 20 years (King) Reoperation is required in 9% of the patients for mitral valve regurgitation or subaortic stenosis.

Etiology, genetic counselling and antenatal diagnosis

Research is being carried out to identify possible genetic causes. Studies of the offspring of affected adults have shown a 10% incidence of recurrent heart disease in offspring of adults with AV canal defects. Computer modelling suggests that partial AV canal is more likely to be due to a single gene than several genes (Burn). Genetic counselling is recommended. As mentioned in the diagnostic criteria, antenatal diagnosis is possible starting from the 16-18 weeks gestation using 2-D echocardiography by transabdominal approach. Earlier diagnosis is possible using transvaginal probes. The diagnosis of partial AV canal in a fetus requires further investigation such as chromosomal assessment because of the high incidence of trisomy 21. Genetic syndromes such as DiGeorge syndrome and Ellis-van Creveld syndrome should also be considered. A higher incidence of AV canal defect has been reported in fetal series (17%) (Cook). Of these, X% died during intrauterine death, X% underwent termination of pregnancies.

Unresolved questions

- Nomenclature: the inclusion in the AV canal defect of isolated cleft of the mitral valve and single atrium.
- Surgery: the timing for correction of severe form of AV canal defect, the treatment of parachute mitral valve; the choice of the prosthetic mitral valve when replacement is needed.

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