ATRIOVENTRICULAR SEPTAL DEFECT

Synonyms:
- Endocardial cushion defect
- AV canal

Definition:
Atrioventricular septal defect is a type of heart malformation affecting the development of the lower part of the interatrial septum, the posterior and superior area of the interventricular septum and the atrioventricular valves. This group of defects can be further divided in partial, transitional and complete.

In the complete type of defect, the key element is the presence of a unique atrioventricular valve separating both atria from the ventricles, instead of 2 valves (mitral and tricuspid). At the same time, there is an ostium primum atrial septal defect and an inlet ventricular septal defect located perpendicularly to the valve, one above and one below.

Atrioventricular septal defect. Anatomy.

In the partial type, there is no ventricular septal defect, and instead of a single valve, there are two, but the mitral valve has an indent dividing its anterior leaflet in two. This is called mitral cleft. This type, which is the mildest form of the disease, is then an ostium primum atrial septal defect and a mitral cleft.

The transitional atrioventricular septal defect is a rare variety found between the partial and complete forms. It has 2 openings separated at the level of the atrioventricular valves such as the partial form, but it has an atrial and a ventricular septal defect (generally small) such as the complete form.
Clinical Picture:

Clinical presentation of the atrioventricular septal defect shall depend mainly on the type of defect it is (See Clinical Picture in Diagnosis and Treatment).

Many children with partial atrioventricular septal defect do not present symptoms and their disease is diagnosed in a routine preschool health check. Some patients show mild symptoms of heart failure (i.e. agitation and fatigue) and frequent respiratory infections. This is due to the blood passage from the left to the right atrium, which draws flow to the left ventricle deviating it towards the right ventricle and the pulmonary circuit. However, the mitral cleft usually gives place to mitral regurgitation (blood back flow from the left ventricle to the left atrium) of variable magnitude. When it is severe, symptoms appear early, and heart failure signs show during the first months of life.

In the complete atrioventricular septal defect, heart failure appears during the first weeks or months of life, sometimes with severe decompensation. Blood flow from the left to the right side and atrioventricular valve regurgitation has 2 consequences. On the one hand, part of the blood that should be directed to the aorta and the organs "escapes" towards the right side, determining heart failure by inability to deliver enough oxygen and nutrients to the body. On the other hand, blood excess reloading the lungs little by little makes them ill definitively. This pathology is known as pulmonary hypertension, and it is initially due to the increased blood volume in the lungs. When this condition continues, the muscle of the arterial walls in the lungs enlarges and becomes too resistant, which gives place to fixed pulmonary hypertension with poor prognosis.

Diagnosis:

Diagnosis is suspected by clinical evaluation (See Diagnosis in Diagnosis and Treatment). A significant murmur is auscultated in the chest. The x-ray shows the dilated heart and the electrocardiogram can show the axis deviated to the left with a first degree atrioventricular block.

Echocardiography is the best diagnostic method. It helps to show the location and size of the atrial and ventricular septal defects, to calculate the amount of the pulmonary blow flow and to define the anatomy of the common atrioventricular valve or the mitral and tricuspid valves in the case of the partial defect. It also evaluates the presence of valve regurgitation, pulmonary hypertension and related lesions.

Cardiac catheterization may be necessary in borderline cases to measure intracardiac pressures, especially in the pulmonary artery, to define if surgery is suitable or not (See Haemodinamycys in Diagnosis and Treatment).

This entity is frequently associated with Down syndrome, and as much as one third of these children can suffer from it. This is why it is recommended to complete a comprehensive cardiologic evaluation in these patients in search of congenital heart defects. At the same time, more than a half of children with complete atrioventricular septal defect suffer from Down syndrome. Patients without Down syndrome may have more deteriorated valves which are more difficult to repair than those of patients without the syndrome.

Treatment:

While the only definitive treatment for this disease is corrective surgery, selecting the most adequate time for the operation shall depend mostly on the type of defect the patient has.

For the complete form of the atrioventricular septal defect it is important not to delay the surgery and schedule it during the first few months of life, since the excessive pulmonary blood flow these children suffer may cause pulmonary hypertension, which decreases significantly the opportunity of long term survival. This correction is carried out
with cardiopulmonary bypass (See Surgery in Diagnosis and Treatment). Access to the common atrioventricular valve is through an incision in the right atrium. There are two different surgical techniques, with one or two patches. In the two patches technique, the ventricular septal defect is closed with a patch whose upper limit is stitched to the valve, dividing it in a right and a left component. Once the valve is divided, the repair of the left component is completed in order to prevent mitral regurgitation. Later, the function of the mitral valve is tested to verify it is competent (that is to say, closes well). Lastly, the atrial septal defect is closed with an autologous pericardial patch (a small piece of the own pericardium).

![Complete AV canal repair](image)

For the partial atrioventricular septal defect, since there is no ventricular septal defect, the pulmonary blood flow and the risk of pulmonary hypertension are smaller than for the complete form. Corrective surgery is carried out during the first few years of life, in general before the school period. After accessing the heart in the same way than the complete defect, the mitral valve is repaired closing the cleft with separate stitches. After
testing the valve to verify it closes properly, the ostium primum atrial septal defect is closed with a patch taken from the own pericardium.

Prognosis:
In partial atrioventricular septal defect, the prognosis is excellent once the surgery is overcome, which is usually a low risk procedure (See Postoperative in Diagnosis and Treatment). The main determining factor of evolution in the long term is the degree of mitral regurgitation, which in some cases progresses to important degrees and may require repair or replacement.

In the case of complete atrioventricular septal defect, surgical risk is higher, although mortality is below 5% in most of the centers. The risk can increase significantly in patients with severe impairment of the clinical condition or with increased pulmonary vascular resistances. This explains the importance of early surgical intervention for these
children. In the same way as in the partial atrioventricular septal defect, prognosis and life quality shall depend mostly on mitral valve function. During evolution, many patients may require one or more surgeries to repair or even replace the mitral valve. On the other hand, the development of subaortic stenosis (obstruction of the left ventricular outflow tract) during the post-operative period may influence the general condition and determine the need to perform another surgery (See Follow-up and Control in Diagnosis and Treatment).

However, despite these complications, long term survival and quality of life are excellent in most of the cases.