

Holes in the heart

= a basic guide

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Who can forget their undergraduate experience of congenital heart disease; all too often on bland descriptions in pathology books with confusing Latin terminology. The truth however is that congenital heart disease is logical and relatively straightforward and clinically exciting. Despite the bewildering array of technological advances of the past few decades, the diagnosis and management of congenital heart patients is based primarily on the clinician's eyes, ears, hands and stethoscope and probably in that order. The chest x-ray and electrocardiogram are helpful and the echocardiogram is a wonderful tool, but these are merely adjuncts in the overall management.

This short account is intended as a brief and practical review of congenital cardiac septal defects. I will deal with ventricular septal defects (VSDs), atrial septal defects (ASDs) and atrioventricular septal defects (AVSDs).

Physiology

At the risk of losing your interest already, please allow me a short paragraph on some basic physiology. The body is divided, from the circulatory point of view, into two circuits. Firstly, the low resistance pulmonary circuit which allows deoxygenated blood to be pumped at low pressure from the right atrium to the right ventricle and then to the lungs via the pulmonary arteries.

Secondly, the high resistance systemic circuit, which requires oxygenated blood to be pumped from the left atrium to the left ventricle at much higher pressure to the aorta, supplying the various organs and tissues of the body. These separate circuits are normally separated by an intact atrial septum and ventricular septum and no mixing or shunting occurs. If a hole exists in the atrial or ventricular septum, blood will naturally flow from the higher pressure systemic (left-sided) circuit to the lower pressure pulmonary (right-sided) circuit. A septal defect will therefore produce a left to right shunt and will result in an abnormally high pulmonary blood flow. This will cause the lungs to be stiffer and wetter than usual, the result being tachypnoea, dyspnoea and a tendency to respiratory tract infections. It may also expose the lungs to higher perfusion pressures than nature intended (pulmonary hypertension) and active changes, eventually irreversible, may occur.

So, how do the various organs of the body cope with some of their blood supply being 'stolen' as a result of the left to right shunt? Remembering that the cardiac output is the product of stroke volume and heart rate, the body compensates by increasing the heart rate and therefore the systemic cardiac output (4 litres/m²/minute) is met by an increase in the heart rate. This results in the clinical finding of tachycardia, sweating and a limitation in the ability to exercise. In babies and

infants, feeding is a major source of exercise and the baby will feed poorly and fail to thrive.

Ventricular septal defect

This is simply a hole in the wall between the right and left ventricles. It may be small, medium or large. It is usually single but multiple holes may be present. VSDs are the commonest congenital heart defect seen in our clinics. The ventricular septum is divided into an inlet (muscular) and outlet (membranous) portion, and holes may occur in any of these locations. The commonest are membranous and muscular VSDs. As many as 50% of all VSDs will undergo spontaneous closure and still more may become smaller and less significant with time.

Even relatively large defects in the newborn period may become trivial or close spontaneously with time. The challenge is to identify those who will require cardiac surgery before damage is done to the pulmonary vascular bed. VSDs are rarely identified in the early newborn period. This is because the pulmonary resistance is generally high for the first few days of life and the pressure in the right ventricle is therefore equal to left ventricular pressure and little or no flow across the VSD occurs. A murmur is generally not heard until after three or four days of life and symptoms rarely occur before a week or two of life, even with large VSDs.

As the pulmonary resistance falls, an increasing volume of blood flows from the left ventricle to the right ventricle and the symptoms and signs described above begin to appear. The baby is noted to be tachypnoeic and increasingly unable to finish a bottle and may fall asleep during feeds. They may become anxious when awake, increasingly lethargic and sweating, weight gain is poor. A finger on the pulse reveals a modest tachycardia with a normal pulse volume. The precordium is hyperdynamic and the liver is enlarged. There is a pan systolic murmur heard throughout the precordium, particularly at the lower left sternal edge. When the left to right shunt is large, the excessive pulmonary blood flow also causes a mid diastolic murmur at the apex caused by the excessive pulmonary venous return to the left atrium rushing across the mitral valve (similar to the murmur heard with mitral stenosis).

With large VSDs the high pressure in the left ventricle is transmitted to the right ventricle and from there to the pulmonary arteries, causing the pulmonary valve to close against abnormally high pressure and therefore the P2 is loud. A small VSD, on the other hand, will produce little by way of symptoms, as there is relatively little extra blood flow to the lungs. There will still be a murmur heard as there is considerable tur-

bulence produced by blood flowing from a high pressure left ventricle to a low pressure right ventricle. Because the hole is small, the murmur will be higher in pitch than its larger counterpart and will not be associated with the hyperdynamic precordium, loud P2 or diastolic murmur mentioned above.

Practice points

A keen pair of eyes and a hand on the precordium will therefore usually distinguish between the child with a large and a small VSD without resorting to the stethoscope. An important practice point is that the loudness of the murmur alone bears little correlation with the size of the VSD. Water flowing out of a large tap will produce little noise. However, if you partly occlude the tap with your thumb, the water will flow with a louder pitched hissing sound. Infants with a moderate to large sized VSD will be at their most symptomatic at three months of age, when the pulmonary resistance is at its lowest and compensatory mechanisms have not yet occurred.

Thereafter the lungs begin to develop compensatory changes, eventually leading to irreversible arterial scarring in later childhood. As this process occurs, the child becomes less symptomatic for a while until eventually the pulmonary arterials are so scarred that the right ventricular pressure is now higher than the left ventricular pressure and the direction of shunting is reversed, producing central cyanosis (Eisenmenger syndrome). This is a very rare occurrence these days.

Treatment

Treatment involves strategies to improve feeding in infancy (high calorie feeds and occasionally nasogastric tube feeding), anti-failure medication (frusemide, spironolactone, captopril and digoxin) and early intervention for respiratory infections. If by six months to a year of age, the VSD has shown no signs of closing and signs of pulmonary hypertension or failure to thrive are present, surgical closure by way of an open heart procedure is required.

Although open heart surgery (particularly on an infant) is never undertaken lightly, the results have improved dramatically in the past two decades. A large team of cardiac surgeons, nurses, anaesthetists, intensivists, cardiologists, liaison nurses, cardiac technicians, pump technicians and administrative staff are required to ensure that all goes well and standards are maintained.

Reoperation either early or late is rarely required these days, although the patient requires regular supervision throughout childhood. The surgery usually involves sewing a patch of synthetic material over the hole. Although this rapidly becomes covered and hidden by the patient's own endothelium, it is still a wise precaution in most children to recommend endocarditis prophylaxis prior to dental procedures, general anaesthesia and other potentially septic procedures. The long-term prognosis is usually excellent with little impact on sports activities, career choices or life insurance later in life.

Finally, it is worth repeating that 50% or more of VSDs

(particularly those in the muscular septum) are small and will eventually become tiny or undergo spontaneous closure. While still patent, their importance lies in the small but definite risk of endocarditis. Endocarditis prophylaxis is universally recommended for as long as the VSD is present.

Atrial septal defect

We are all born with a communication (foramen ovale) between the right and left atrium. This allows blood to flow from the right to the left atrium in foetal life and usually closes spontaneously in the first few months of life. An atrial septal defect is a hole in the wall between the right and left atrium. It usually occurs at the site of the foramen ovale (secundum ASD), but may also occur high in the atrial septum (sinus venosus ASD) or in the lower atrial septal septum (primum ASD or partial atrioventricular septal defect).

Whatever the site of the defect, the clinical signs are usually similar. ASDs are slightly less troublesome than VSDs but they are often difficult to detect clinically. The pressure in the

left atrium is higher than in the right atrium, and a hole in the atrial septum will cause a left to right shunt as with VSDs. However, the atria operate at much lower pressures than the ventricles and the pressure difference between the left atrium and right atrium is much smaller. The flow of blood across an atrial septal defect will therefore be much less turbulent.

The murmur of an ASD is caused not by the blood flow across the ASD, but as a result of extra blood being pumped from the right ventricle across the pulmonary valve. This causes a soft to moderate ejection systolic murmur in the pulmonary area (upper left sternal edge)

with wide fixed splitting of the second heart sound. If the ASD is large, the pulmonary blood flow (and the resultant pulmonary venous return to the left atrium) will be excessive and will pass again across the ASD from left to right atrium and across the tricuspid valve. This causes a diastolic murmur, this time heard best at the lower left sternal edge. The extra volume load on the right ventricle will be felt as a right ventricular heave.

The signs of excessive pulmonary blood flow (tachypnoea, sweating, poor feeding etc.) will also be evident, but usually less dramatic than with VSDs. These signs are often subtle and are easily overlooked. A history of frequent respiratory infections with mild shortness of breath on exertion in early school-going years is typical. Often the murmur is first picked up at a school medical examination and the history is evident only with hindsight.

Small ASDs may produce no clinical signs of note and may be evident only on echocardiography. They rarely warrant intervention in the asymptomatic patient. It is rare for an uncomplicated ASD to cause problems in infancy. A secundum ASD may undergo spontaneous closure in the first few years of life. If by three or four years of age, spontaneous closure is not evident, surgical or, more recently, catheter closure of the ASD is indicated. An ASD in a location other than the

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usual secundum ASD will never undergo spontaneous closure and requires surgical closure in early childhood.

In contrast to VSDs, ASDs (even large ones) will produce only mild or moderate pulmonary hypertension. The changes of pulmonary vascular disease, as described with VSDs, will occur at a much slower pace and rarely before the third decade of life. Surgery in early childhood is advocated not only to prevent the onset of pulmonary vascular disease but also to prevent later occurrence of atrial arrhythmias. Surgery, performed usually between two and four years of age, involves an open heart procedure under cardiopulmonary bypass. It is usually possible to close the ASD with direct suturing. Sometimes, if the defect is large or in an unusual position, a patch may be required.

The surgical results are excellent and long-term sequelae are relatively few. Because surgery on the atrium may cause scarring around the sinus node, a small percentage of patients may require pacemaker insertion for sinus node disease in late teenage or early adult life. Long-term cardiac follow-up is therefore recommended for all such patients. Endocarditis prophylaxis however is usually not required, except where the surgical repair is more complicated than usual.

Atrioventricular septal defect

This type of septal defect is variously termed 'AV canal', 'endocardial cushion defect' and, more recently, 'atrioventricular septal defect (AVSD)'. It is the mother of all septal defects. Although less common than either ASDs or VSDs, it still accounts for significant morbidity and mortality and alone accounts for a large proportion of the workload of most paediatric cardiac centres. It is particularly associated with patients with Down syndrome, although as many as 50% of all AVSDs occur in non-Down syndrome patients.

The embryologists have shown us that the central part of the heart (i.e. the mitral and tricuspid valves, the lower part of the atrial septum and the posterior part of the ventricular septum) is formed from the endocardial cushion; an entity distinct from the rest of the heart. A defect in the formation of the endocardial cushion, as occurs so often in Down syndrome patients, produces therefore a hole in the lower part of the atrial septum and posterior part of the ventricular septum (the inlet septum) and defective mitral and tricuspid valves.

The result of this unfortunate sequence is a large left to right shunt at atrial and ventricular levels, as well as incompetence of the atrial and tricuspid valves. In fact the mitral and tricuspid valves, which normally form two distinct rings, usually consist of just one common valve ring with leaflets shared by both the tricuspid and mitral valve. These patients present with signs of congestive heart failure (as described in the VSD section) in early infancy and develop pulmonary vascular changes before the second year of life.

In addition, the atrioventricular valve tissue is often deficient and a variable degree of mitral and tricuspid valve regurgitation is often present. This contributes further to the signs of congestive heart failure. It is extremely rare for these defects

to become smaller with time and surgery is usually necessary in the first 12 to 18 months of life. With improvements in diagnosis, surgical techniques and intensive care, it is now possible to repair these defects in infancy. Due to the relatively large number of Down syndrome babies in Ireland, our experience in the management of this condition has grown.

The surgical mortality is less than 10% and the long-term prognosis, if operated early in infancy, is excellent.

Controversy still exists on the ethics of major surgical intervention in patients with Down syndrome where the natural life expectancy, even with a normal heart, is shortened. However, our experience and that of most other centres, suggest that the quality of life and life expectancy is enhanced significantly by successful surgical intervention. Reoperation may be required for progressive mitral or tricuspid regurgitation and the need for endocarditis prophylaxis for dental and other potentially septic procedures is life long.

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First non-surgical closure of ASDs in Ireland

Until very recently, the repair of an ASD has required open heart surgery under cardiopulmonary bypass. Despite many advances in this technique, it remains a major surgical procedure involving a long stenotomy scar, the potential complications of open heart surgery, not to mention the pain, discomfort and cost of intensive care and up to 10 to 14 days in hospital. A remarkable advance in modern biotechnology may provide a simpler solution to this problem.

Over the past number of years, several groups throughout the world have been working on a variety of implantable devices designed to seal the ASD in the cardiac catheterisation laboratory without the need for surgical incision or indeed cardiopulmonary bypass. One of these devices has recently passed clinical trials and appears set to revolutionise the management of patients with ASDs. Called an Amplatzer device, named after its inventor Dr Kurt Amplatz, it consists of a double disk button-shaped device made of nitinol.

Recently, paediatric cardiologists at Our Lady's Hospital for Sick Children, Crumlin, with the help of Dr Kevin Walsh of Alder Hey Hospital in Liverpool, one of the principal investigators in the clinical trials, inserted the Amplatzer device into seven children with ASDs. The procedures were carried out in the cardiac catheterisation laboratory under general anaesthesia and took one to two hours each. In each case, the ASD was successfully closed and there were no complications. The patients were back on their feet in a few hours and able to go home the following day. Although the early results are promising, the cardiac team at Our Lady's Hospital are anxious to stress that experience with the new device is limited and careful follow-up and audit of results will be required.

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