Cardiac Disorders in People with Down’s Syndrome

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The normal heart

People with Down’s syndrome have an increased risk of cardiac disorders. These are largely congenital (present from birth) but there is also an increased risk of acquired cardiovascular disease, partly due to other conditions associated with Down’s syndrome. Congenital cardiac disorders are present in approximately 40-60 percent of babies born with Down’s syndrome. We now know a great deal about these disorders and the Down Syndrome Medical Interest Group, UK and Ireland (DSMIG), has produced guidelines in order that they can be diagnosed and treated promptly. Timely diagnosis and treatment has led to a significant improvement in the health and life expectancy of those affected.

In this article the common congenital heart disorders will be explained in addition to the screening for cardiac conditions as set out by the current DSMIG guidelines. We will then touch on the presentation and treatment of these conditions.
Which cardiac disorders are most common in people with Down’s syndrome?

An atrioventricular septal defect (AVSD)

An atrioventricular septal defect (AVSD) is the most common congenital cardiac defect in people with Down’s syndrome. An atrioventricular septal defect is when the septum or partition between the two ventricles (lower chambers) of the heart and between the two atria (upper chambers) of the heart does not develop properly and a hole is present. This allows mixing of blood from the left to the right side of the heart causing increased blood flow to the lungs and complications discussed later.

A ventricular septal defect (VSD)

A ventricular septal defect (VSD) is a hole through the part of the septum that separates the two ventricles in the heart. As with AVSD, blood from the left ventricle flows through the hole into the right ventricle but the atria are completely separated.
Secundum atrial septal defects (ASD)
In secundum atrial septal defects (ASD) there is a hole through the septum that separates the two atria, but the ventricles are separate.

Persistent ductus arteriosus (PDA)
Persistent ductus arteriosus (PDA) is persistence of a blood vessel which is a normal part of the heart structure whilst the baby is still in the womb.

It allows blood to bypass the lungs before the baby needs to breathe, by taking a short cut between the blood vessels that will later take blood to the lungs and the aorta which takes blood to the body. If it does not close, once the baby starts to breathe, the pressure in the aorta makes the blood flow back through this duct causing congestion in the lungs.
Tetralogy of Fallot

Tetralogy of Fallot is another cardiac defect commonly seen in babies with Down’s syndrome. The term “tetralogy” comes from the four malformations that occur together: a VSD, an overriding aorta, sub-pulmonary stenosis and right ventricular hypertrophy. The overriding aorta is when the aorta has connections with both the right and left ventricle, (in typical development it only connects with the left). Blood coming from the lungs carrying high levels of oxygen mixes with blood that has just come from the body (deoxygenated), so the blood going around the body carries less oxygen than it should do. There is narrowing of the outflow below the pulmonary valve that separates the right ventricle from the pulmonary artery so the right ventricle has to work harder to overcome this and becomes more muscular and thickened.

Babies with Down’s syndrome can also have less common congenital heart defects, or may have complex congenital heart disease with more than one of these problems.

Cardiac problems in older children or adults

Cardiac conditions can also occur later in life. This may include congenital heart defects that were not picked up in infancy (the person may have been born at a time, or in a country where early screening was not available), further problems in those who had defects corrected in infancy, or new problems that may arise.

Investigation of those who have never been screened, or have new symptoms should be by a cardiac specialist.

Symptoms suggestive of a heart problem may include breathing difficulties, fatigue, dizziness or faints, or chest pain. There are of course many other causes of all of these.

Children who had early correction may develop leaky valves or arrhythmias (irregular heart beat) as a result of scarring. Some may have developed pulmonary hypertension (see below), which will require specialised long-term management including medications.

New problems that may arise include mitral valve prolapse and aortic regurgitation. Mitral valve prolapse is when the mitral valve connecting the left atrium to the left ventricle does not close properly. This valve should close to stop backwards flow of blood as the heart pumps. If it does not close properly blood can leak back into the left atrium causing back pressure on the lungs.

Aortic regurgitation is commonly thought of as a ‘leaky’ aortic valve. This valve sits where the left ventricle connects to the aorta and usually comprises of three separate leaflets that open to allow blood to flow out of the left ventricle into the aorta to go to the body, and closes to stop backflow of blood from the aorta into the left ventricle. In aortic regurgitation incomplete closure allows blood to flow back into the left ventricle causing volume.

In the general population, as people age the risk of other cardiac problems such as hypertension, and coronary heart disease increases. The same applies for those with Down’s syndrome, but rates of both these problems seem to be lower. However people with the syndrome are more likely to be overweight, or have diabetes or thyroid disease, all of which can contribute to cardiac problems. Advice on healthy lifestyles, including good diet, taking exercise, and the avoidance of smoking is as important as for the general population.

Pulmonary arterial hypertension: a complication of congenital heart disorders

The congenital heart disorders seen in children with Down’s syndrome can lead to pulmonary arterial hypertension (PAH). This is when the pressure in the pulmonary artery which delivers blood to the lungs is too high.

When there is a hole between the right and left sides of the heart (as in AVSD and VSD) blood can freely move from the left side (which is at higher pressure) to the right side, increasing the blood
in the right ventricle and the blood flow through the pulmonary artery to the lungs. This makes muscle around the pulmonary arteries thicken, and some of the smaller blood vessels in the lung close, so the pressure in the blood vessels in the lungs increases, and blood flow reduces. Increased blood pressure in the lungs causes increased pressure in right side of heart.

Eventually the pressure on the right side equals or exceeds that on the left and blood flows from the right side of the heart to the left through the hole. This means deoxygenated blood (from the right side) mixes with oxygenated blood (from the left-side), and lowers the levels of oxygen in the blood circulating around the body, causing the older child to look blue or cyanotic. This situation is known as Eisenmenger syndrome, and once established will cause increasing health problems, as the child progresses into adult life, and reduced life expectancy.

This situation can be prevented by closure of the cardiac defects at the right time (depending on the type of defect), but not once Eisenmenger syndrome is established.

People with Down's syndrome are more likely to develop PAH than the general population and therefore prompt diagnosis and treatment of these heart conditions is essential.

Sometimes PAH can develop without a major congenital heart defect. The most common cause of this is upper airway obstruction, and this could develop at any stage of life.

When and how are the congenital heart disorders diagnosed?

Currently in the UK pregnant women are offered a detailed ultrasound scan at around 20 weeks’ gestation. This may highlight heart defects in the foetus which can then be investigated with more detailed scans (foetal echocardiogram) and may be what leads to the antenatal diagnosis of Down’s syndrome. When a diagnosis is made before birth, parents can be offered information about the problem with a plan for care at the time of birth, and subsequent treatment required.

What are the signs and symptoms of heart disease?

It is not always obvious that a baby with Down’s syndrome has congenital heart disease as there may be no signs or symptoms. This is why it is so important that they have proper assessment to check if the heart is healthy or there is a problem that needs treatment.

In some there will be immediate signs such as cyanosis, (blueness of the skin) or they may develop signs of heart failure, with breathing difficulties, or puffiness.

A heart murmur may be detected either during the newborn examination on the first day of life or in later health check-ups. This is an added sound when listening to the heart that we would not normally expect to be there. However some soft murmurs are “innocent” and due to the sound of blood flowing through a healthy young heart.

After the newborn period, signs suggesting heart disease can include faltering growth or signs of heart failure that include poor feeding, and breathing difficulties.

Due to the high prevalence of cardiac disorders in children with Down’s syndrome and the potentially difficult diagnosis solely on these clinical signs the current DSMIG guidelines make recommendations that include cardiac assessment and an echocardiogram (heart scan) for all babies born with Down’s syndrome within the first few weeks of life.

What is the treatment for these heart conditions?

Treatment will depend on the type of cardiac defect. Some require early surgery, some require treatment with medicines, and may need surgery later, and some may not need any treatment, but require close monitoring in case things change.

A heart specialist will decide on which management is appropriate for each individual child. They will assess the type of defect and the risk of developing PAH and irreversible pulmonary artery disease, and for AVSD early surgery will usually be within the first 3-6 months. If, in other conditions, the pressure in the pulmonary artery is not high, an operation may be carried out at a few years of age with close monitoring until then. Sometimes medical treatment will be required prior to an operation. Such treatment includes nutritional supplementation with high calorie feeds (often via naso-gastric tube) and medication to encourage fluid loss so the lungs are not as congested, breathing is easier and the baby is more comfortable.

Children with congenital heart disorders can be at increased risk of getting chest infections and so it is important to offer all the usual childhood immunizations, as well as influenza vaccine in the winter. They are also more likely to get bronchiolitis caused by the respiratory syncytial virus (RSV) and prophylaxis may be offered to some depending on the heart. Parents should also be offered advice on hygiene and ensuring the child is not around others with infections.

Infective endocarditis

Infective endocarditis is an inflammation of the endocardium (inner lining) of the heart and heart valves. It is caused by bacteria infecting the heart via the blood stream and leads to deposits of clot-like material settling on the endocardium. This can damage the heart valves or the conduction pathways of the heart.

The risk of this infection is higher in people with a known heart problem and is increased by any procedure which allows bacteria to enter the blood stream e.g. surgical or dental procedures.

Up until March 2008 most people with congenital heart defects were given treatment with antibiotics to cover some surgical and dental procedures and prevent infection. However the National Institute of Clinical Effectiveness (NICE) have since changed their guidelines so that only patients with a high risk of getting the infection or where an operation is being carried out where there is a suspected infection are given antibiotic prophylaxis. For people with Down’s syndrome who have had isolated ASD, fully repaired VSD or fully repaired PDA, antibiotic prophylaxis is no longer recommended but could still be considered for those with a valve disorder or replacement or who have had infective endocarditis in the past.

Conclusions

People with Down’s syndrome are at increased risk of congenital and acquired heart disorders. It is essential that all babies with Down’s syndrome are screened for heart defects at a very young age. The possibility of previously undiagnosed congenital disorders or new heart problems having developed, should be considered at regular medical review throughout life. Such vigilance will allow prompt diagnosis and management and lead to a decrease in the burden of cardiac disease in people with Down’s syndrome.