Congenital heart defects

Cardiovascular defects are by far the commonest major group of congenital anomalies. Development of the heart and blood vessels is complex and extends across several weeks of early embryonic life, providing significant opportunities for disruption to normal development. The resulting defects vary from minor abnormalities that may cause no problems, to life threatening heart malformations that may ultimately prove fatal.

DEVELOPMENT OF THE HEART

Embryology

The vascular system starts to develop around day 17 of embryonic life when clusters of angioblast cells appear. These rapidly grow and merge to develop a mesh of small blood vessels from which the heart and major vessels form. The heart initially develops as a tube that starts to pump blood through the developing vessels. The tube starts to bulge and fold to form primitive right and left atrial and ventricular sections by day 28. The following 2-3 weeks are a critical period during which the chambers of the heart and associated major vessels undergo considerable development with formation of cardiac septa and valves and definition of the major vessels.

The normal heart

The heart is divided into right and left sides, with 2 chambers on each side. Blood collects in the upper chamber, or atrium, before passing into the lower chamber, or ventricle. The ventricles are made of thick heart muscle, which contract to pump the blood out of the heart.

Blood returning from the body collects in the right atrium before passing into the right ventricle. As the heart muscle contracts the right ventricle pumps blood out through the pulmonary artery to the lungs. As it passes through the lungs the blood picks up oxygen before returning to the left side of the heart. Blood leaves the left side of the heart via the aorta and is pumped around the body where the blood delivers oxygen to the body tissues. Depleted of oxygen, the blood then returns to the right side of the heart and the process begins again. Valves are situated between the atria and ventricles and between the ventricles and arteries. These consist of a set of flaps or cusps that open and close to let the blood flow in only one direction when the heart muscle contracts. The 4 main heart valves are:

- The tricuspid valve, separating the right atrium and ventricle. This allows blood into the ventricle from the atrium but stops flow back into the atrium when the ventricle contacts, so that the only way the blood can leave the ventricle is through the pulmonary valve into the pulmonary artery.
- The pulmonary valve, separating the right ventricle from the pulmonary artery. This allows blood into the pulmonary artery when the right ventricle contracts, but stops blood flowing back from the pulmonary artery into the ventricle when it relaxes.
- The mitral valve, separating the left atrium and ventricle. This allows blood into the left ventricle from the atrium but stops flow back into the atrium when the ventricle contacts, so that the only way the blood can leave the ventricle is through the aortic valve into the aorta.
- The aortic valve, separating the left ventricle from the aorta. This allows blood into the aorta when the left ventricle contracts, but stops blood flowing back from the aorta into the ventricle when it relaxes.

Figure 17 illustrates the main structures of the normal heart.

Figure 17: The normal heart Copyright AHA

Blue - oxygen depleted blood
Red - oxygen rich blood
Fetal circulation

The growing fetus relies on the mother’s circulatory system and the placenta to provide nutrition and oxygen and to remove waste products and carbon dioxide via the umbilical vessels. Blood bypasses the fetal lungs before birth by means of the ductus arteriosus, a vessel connecting the pulmonary artery to the aorta. In addition there is an opening (the foramen ovale) in the atrial septum that allows blood to pass directly from the right to the left atrium. After birth the ductus arteriosus and foramen ovale start to close up as the baby’s lungs and cardiovascular system take over. As this happens, some heart defects (that were not problematic with the fetal circulation) become more apparent days or even weeks after birth.

THE SIZE OF THE PROBLEM

Reported rates for congenital heart malformations vary, depending on exactly which defects are considered, and whether terminations of pregnancy or spontaneous fetal losses (miscarriages) are included. CARIS has received reports of 1181 cases in Wales in the period 1998-2001 (see Table 1), with a rate of over 9 per 1000 live/stillbirths/terminations of pregnancy. This is nearly twice the corresponding rate of 5.4 per 1000 recently reported by EUROCAT.14 The Welsh rate for liveborn babies with congenital heart defects is 7.4 per 1000 livebirths, which is comparable to estimates by the American Heart Association that 8 per 1000 livebirths have a heart anomaly.15

Table 3b shows how gross rates for congenital heart defects vary across Wales with high rates in Iechyd Morgannwg. This is probably a reflection of reporting differences discussed elsewhere in this report. As more cases become available from the paediatric cardiology unit in Cardiff, this picture may change.

CAUSES OF CONGENITAL HEART DEFECTS

It has been estimated that 8% of heart malformations are due to genetic factors and 2% to environmental agents. In the majority of cases, the cause of a congenital heart defect is either not clear, or is thought to be due to complex interactions involving many different factors.

Classic examples of environmental causes for congenital heart defects include fetal exposure during pregnancy to:

- Rubella virus
- Drugs including sodium valproate, trimethadione, lithium, amphetamines and warfarin
- Vitamin A
- Alcohol
- Maternal medical conditions such as diabetes.

The CARIS database has details of at least 6 reported cases of congenital heart defect associated with drugs known to cause problems, including sodium valproate (4 cases) lithium (1 case) and warfarin (1 case). 1 case of fetal alcohol syndrome also had a heart defect, as did at least 31 babies with evidence of maternal diabetes during pregnancy.

Chromosomal anomalies represent the most significant genetic causes. 6-10% of newborns with heart defects have a chromosomal anomaly. Conversely, about one third of children born with chromosomal anomalies have a heart defect (up to 100% for cases of trisomy 18). Other significant genetic conditions associated with heart defects include DiGeorge and Goldenhar syndromes.

The CARIS database shows 218/1181 (18.5%) cases of congenital heart defects also had a chromosomal anomaly. Among liveborn babies with congenital heart malformations, 113/944 (12%) also had a chromosomal anomaly. 70% of chromosomal anomalies were Trisomy 21, 18 or 13.

Non autosomal syndromes associated with cases of heart anomalies reported to CARIS are shown in Figure 18.

Figure 18: Non-autosomal syndromes associated with cases of congenital heart defects reported to CARIS, 1998-2001

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14 Covering 6 million births 1980 - 1999, from various registries across Europe (in print)
15 American Heart Association 2002 (www.americanheart.org/presenter.jhtml?identifier=1086)
16 Sadler TW. Langman’s Medical Embryology. 7th Edn. Williams & Wilkins. Baltimore 1995
DIAGNOSIS AND GENERAL MANAGEMENT

Antenatal

Antenatal diagnosis of cardiac defects is a growing area of expertise, resting routinely on ultrasound scanning. A fetal anomaly scan at 18-20 weeks should try to include a 4 chamber view of the heart (Figure 19) and outflow vessels, although this will not exclude all cardiac anomalies. Any suspicion of a defect can be investigated further in some centres through cardiotocography and more detailed cardiac scanning. Antenatal detection allows arrangements to be made for babies with serious defects to be delivered in specialist centres. For very serious defects, the option to terminate the pregnancy may also be considered.

Figure 19: 4 chamber view of the heart on antenatal fetal ultrasound scan

Postnatal

The first suspicion after birth of a previously undiagnosed heart defect may come from observation of feeding difficulties or shortness of breath, evidence of cyanosis and/or physical signs on routine neonatal examination. Heart murmurs associated with many defects are not always present in the first few days of life when the fetal circulation may partly persist. Cyanosis arises when deoxygenated blood is present in arteries, giving the lips and skin a bluish tinge. In congenital heart disease this occurs when blood is shunted past the lungs and is prevented from absorbing more oxygen. Placing an infant or child in an oxygen rich environment has little effect on the cyanosis as this does not in itself improve pulmonary blood flow. Diagnoses are usually confirmed using echocardiography, chest X-ray and ECG. Cardiac catheterisation may be required for further study of blood flow or to undertake some operative procedures.

For some cases, defects are minor and may need no active treatment. For most defects, the risk of bacterial endocarditis is increased and antibiotic cover is required for various situations and surgical procedures. For symptomatic cases of congenital heart disease, general management may include energy rich / tube feeds to help babies in heart failure. Drugs may be required to treat heart failure until surgery is feasible. Surgical procedures are often staged to get optimal results.

The recent enquiries into paediatric cardiac services have dramatically illustrated the need for health professionals to recognise better the rights and needs of children with heart defects and their families.
SPECIFIC DEFECTS

Congenital heart anomalies can be variously classified into broad groups of defects. Some of the main examples are considered in this report.

- **Obstruction of normal blood flow** e.g. pulmonary stenosis, aortic stenosis or coarctation of the aorta.
- **Septal defects**, in which holes in the atrial or ventricular septa allow abnormal blood flow direct from one side of the heart to the other.
- **Cyanotic heart defects**, as a result of which the blood pumped to the body has less than the normal amount of oxygen, causing cyanosis. These defects include Fallot's tetralogy, transposition of the great arteries, tricuspid atresia, pulmonary atresia, total anomalous pulmonary venous connection and hypoplastic left heart syndrome.
- **Other complex defects**. These occur rarely and include single ventricle, double outlet ventricle and hypoplastic heart.

**Patent Ductus Arteriosus (PDA)**

If the ductus arteriosus fails to close in the newborn, oxygenated blood leaving the left side of the heart will shunt back into the pulmonary artery rather than continue through the aorta to the rest of the body (Figure 20). This can lead to an excess of blood passing through the lungs and ultimately to congestive cardiac failure. Mild cases may only be detected clinically by the characteristic murmur whilst more severe cases may show signs of heart failure. PDAs are particularly common in premature babies, in which they are not considered a congenital anomaly. In keeping with other registries, CARIS does not collect data on isolated PDAs in babies born at less than 37 weeks gestation (unless they are associated with other heart defects).

Treatment may be with drugs that help to close the ductus. Surgical options include blocking the vessel during cardiac catheterisation or tying the vessel off directly.

179 cases of PDA were reported to CARIS for 1998-2001, of which 43 were isolated defects in babies born at 37+ weeks gestation. The remainder were associated with other anomalies. The rate among live and stillbirths was 14/10,000 LB/SB. 150/177 liveborn cases (85%) survived to age one year. Most of the cases that did not survive infancy were associated with other major heart malformations.

**Pulmonary and Aortic Stenosis**

Stenotic heart valves do not form properly. The cusps that form the valves are often fused or are thick and stiff, so that the heart has to pump harder to get blood through them.

Many children with **congenital aortic stenosis** have few symptoms although, in severe cases, symptoms occur in infancy (chest pain, tiring easily, dizziness or fainting). Children may develop enlargement (hypertrophy) of the left side of the heart. These findings arise from the extra work required of the left ventricle to pump blood out through the stenotic valve into the aorta, together with the effect of a reduced volume of blood being pumped with each heart beat, compromising blood flow to the brain.

Treatment usually involves enlarging the valve opening to relieve symptoms, either through open surgery or the newer technique of balloon valvuloplasty during cardiac catheterisation. Stenosis often worsens over time and ultimately, replacement of the aortic valve may be required.
**Pulmonary stenosis** prevents blood flowing easily from the right ventricle into the pulmonary arteries and on through the lungs. Children often have no symptoms, but in severe cases, infants may be cyanosed as a result of reduced blood flow through the lungs and right ventricular hypertrophy may occur. In young children, the condition can be relieved by balloon valvuloplasty. Both aortic and pulmonary stenosis are reported to occur in about 3-4 per 10,000 live/stillbirths. The comparable rate in Wales (1998-2001) was 1.9 per 10,000 for aortic stenosis and 3.3 per 10,000 for pulmonary stenosis. This apparently lower than expected rate for aortic stenosis may be because of the way the data is coded or may relate to delay in diagnosis until after the first year of life. In 33/43 (79%) cases of pulmonary stenosis known to CARIS, cardiac defects were the only congenital malformations reported. 41 cases were liveborn, with 39 (95%) surviving to the end of the first year of life.

**Coarctation**

In this condition there is a severe constriction of the aorta leading to raised arterial blood pressure behind the constriction. Congestive cardiac failure may also occur. Recent developments in treatment include stretching the constricted area using a balloon during cardiac catheterisation. Traditionally however, the condition is treated surgically through replacing or patching the affected area of the aorta. After treatment, children usually do well, but need lifelong follow up as the coarctation or associated high blood pressure may recur. In 1998-2001, 46 cases of coarctation were reported to CARIS. The rate among live/stillbirths and terminations. (95% CIs 2.5 - 4.6). This is similar to the EUROCAT rate of 2.9 / 10,000. 39 of the 44 liveborn cases in Wales survived to the end of the first year of life (85%).

**Atrial septal defects**

Atrial septal defects (ASDs) are common heart defects that involve a defect in the septum between the right and left atria, most often due to failure of closure of the foramen ovale after birth. This allows blood to shunt between the atria, usually from left to right (Figure 21). The volume of blood in the right atria builds up causing increased blood flow through the right ventricle and the lungs. In most children the defect causes no symptoms although a large defect may cause congestive cardiac failure with associated shortness of breath, tiredness and poor growth. If left untreated, the defect may often cause problems in adulthood, including pulmonary hypertension, heart failure, arrhythmias and an increased risk of stroke.

Some ASDs are picked up on routine antenatal scanning or more detailed fetal echocardiography. After birth, the condition can be diagnosed from the typical heart murmur arising from increased blood flow through the pulmonary valve and a "split" second heart sound. Further echocardiography and electrocardiography (ECG) will then help determine the size of the hole together with increased blood flow through the lungs and the effect on the right side of the heart.

Approximately 40% of ASDs will close spontaneously in the first year of life. If the defect is still present at the age of 2, it will probably not close on its own. Some defects can then be "patched" during cardiac catheterisation. Others may require open-heart surgery. Heart surgery is usually successful in over 99% of cases. After closure in childhood, the heart will return to its normal size and the long-term outlook is excellent.
CARIS has received reports of 362 cases of ASD for 1998-2001. In 20% of cases, the ASD was the only defect. In a third of cases it was associated with other cardiac defects, but no other problems. A fifth of cases was associated with chromosomal defects, especially aneuploidies. ASD is more common in females. This is reflected (but not markedly) among CARIS cases where there is a ratio of 1 male to 1.2 female.

The rate for cases resulting in live/still birth or termination of pregnancy was 28/10,000 LB/SB/TOPs (95% CIs 25-31). This is higher than the reported EUROCAT rate of 10.75/10,000 LB/SB/TOPs. 338 cases were liveborn, of which 305 (90%) survived to the end of the first year of life.

Ventricular septal defects

A ventricular septal defect (VSD) constitutes a hole in the septum between the right and left ventricles, allowing blood to shunt from left to right (Figure 22). Its size and position in the septum determine the consequences of the lesion. A small hole will often close on its own, especially if it is in the muscular portion of the septum. Such holes rarely cause problems, even if they do not close spontaneously. Larger defects are more serious and may cause problems before birth or in the first few weeks of life, as large amounts of blood flow back into the right side of the heart and through the lungs, causing congestive cardiac failure. The baby may then be short of breath and fail to thrive in the first few months of life.

As with ASDs, a VSD may be detected antenatally through ultrasound scanning. After birth the defect may be characterised clinically by a typical pansystolic murmur, although, in cases of larger VSDs no murmur may be heard initially. Conversely small defects may produce very loud murmurs. Further echocardiograms, ECG and chest X-ray will help determine the size, position and consequences of the defect. Occasionally, cardiac catheterisation may be required to help assess the need for surgery in moderate defects.

In many cases, follow up in the early years may be all that is required, and the hole may close without treatment. Heart failure may be treated initially by drugs, together with high calorie feeds and/or tube feeding to reduce nutrition problems. If these measures are insufficient, surgical closure of the defect may be indicated.

The long term outlook for many cases of VSD is good, although children may require follow up as other cardiac defects may present in later life. More severe defects can lead to a number of serious problems although, with modern methods of diagnosis and treatment, these are much less common. In these more severe cases, children are prone to lung infections and failure to thrive in the first few months of life may affect brain growth. Prolonged lung exposure to increased blood flow can result in pulmonary vascular disease or pulmonary hypertension (Eisenmenger’s syndrome). In time, the pressure in the right ventricle may rise so high that blood flows through the VSD from the right side of the heart to the left, bypassing the lungs and causing cyanosis. If these pressures become irreversible, the only treatment option is heart and lung transplantation.

CARIS has received reports of 418 cases of VSD in 1998-2001 (35% of all cases of congenital heart defect). In over a third of cases, the VSD was the only malformation. In a further 28% it was associated with other cardiac defects, but no other problems. Nearly one fifth of cases were associated with chromosomal defects, especially aneuploidies.

The rate for cases of VSD resulting in live/still birth or termination of pregnancy was 32/10,000 LB/SB/TOPs (95% CIs 29-35). (The reported EUROCAT rate was 23/10,000 LB/SB/TOPs). 357 of the CARIS cases were liveborn, of which 335 (94%) survived to the end of the first year of life.
Atrio-ventricular septal defect

Occasionally a very large defect occurs, involving the atrial and ventricular septa (AVSD). This allows blood to pass freely between the atria and ventricles. The atrio-ventricular valves are deformed or absent. This condition gives rise to significant shunting of blood through the lungs and congestive cardiac failure, requiring surgical treatment in early life. The condition is relatively rare, and is associated with chromosomal defects such as Down syndrome. 76 cases were reported to CARIS for 1998-2001; 54% were associated with chromosomal anomalies. The rate for live/stillborn births and terminations for congenital anomaly was 5.8/10,000 LB/SB/TOPs (95% CIs 4.5 – 7.1), which is comparable to the EUROCAT rate of 3.3/10,000 LB/SB/TOP. 39 cases (51%) were liveborn. Of these, 27 (69%) survived to age 1 yr.

Fallot’s tetralogy

Tetralogy of Fallot is the term used to describe a combination of 4 related heart defects that commonly occur together. These are illustrated in Figure 23 and comprise:

- Pulmonary stenosis
- Ventricular septal defect
- Overriding aorta (the aortic valve is enlarged and appears to arise from both the right and left ventricles)
- Right ventricular hypertrophy

A small number of cases may have additional defects such as an ASD.

In Fallot’s tetralogy, the pulmonary stenosis limits blood flow to the lungs so that oxygen depleted blood builds up on the right side of the heart. Right ventricular pressure rises, shunting the de-oxygenated blood through the VSD and over-riding aorta and out to the body tissues. The right ventricular hypertrophy seen in Fallot’s develops as the right ventricle tries to push an increased volume of blood through the pulmonary valve.

At birth, severe cyanosis is rare as babies usually have a patent ductus arteriosus that improves pulmonary blood flow. As this closes cyanosis and a loud harsh murmur develop. Once suspected, the elements of the tetralogy can be diagnosed by echocardiography. Cardiac catheterisation may be required to study pulmonary blood flow.

Fallot’s tetralogy may sometimes be diagnosed in utero by antenatal ultrasound. Of the cases reported to CARIS19 where time of diagnosis is known, only 1/25 (4%) was diagnosed before 24 weeks gestation and 2/25 (8%) by birth. 13/25 (52%) were diagnosed in the first week of life with the remainder identified later in infancy.

Once diagnosed, initial management focuses on maintaining adequate blood oxygenation. Surgical correction is usually undertaken at about 6 months of age although earlier intervention may be required if oxygen levels are low. Survival rates for children with Fallot’s tetralogy have improved dramatically over recent decades. In the absence of additional risk factors, over 95% of infants with the condition successfully undergo surgery in the first year of life and long term cardiac function is excellent20. Later complications may include a leaking pulmonary valve or recurrence of the pulmonary stenosis, both of which may require further surgery.

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19 excluding spontaneous fetal losses or cases where pregnancy was terminated for other anomalies
20 Information from Cincinnati Children’s Hospital Medical Centre (www.cincinnatichildrens.org)
35 cases of Fallot’s tetralogy were reported to CARIS for 1998-2001; in two thirds of cases, cardiac defects were the only malformations. The rate for live/stillborn births and terminations for congenital anomaly was 2.5 / 10,000 LB/SB/TOP (95% CIs 1.6 - 3.3), which is exactly comparable to the EUROCAT rate (also 2.5/ 10,000 LB/SB/TOP). Among the CARIS cases, 32/35 (91%) were liveborn. Of these, 29 (91%) survived to age 1 yr. The percentage surviving to 1 year is slightly less than that given above for infants surviving surgery. However, numbers are small and the Welsh cases may well include babies with additional risk factors.

**Transposition of the great arteries**

In this condition, the aorta and pulmonary arteries are “transposed” from their normal position and arise from the wrong ventricle, causing two separate circulations of blood. The aorta arises from the right side of the heart and carries de-oxygenated blood back to the body, instead of the lungs, causing cyanosis. The pulmonary artery arises from the left side and carries oxygenated blood back to the lungs instead of to the rest of the body. Unless a connection is made between these 2 circulations, oxygenated blood cannot reach the body organs. In 25% of cases a connection is formed through an associated VSD. Persistence of the ductus arteriosus will also allow some initial mixing of blood but, as this closes down, cyanosis will worsen (Figure 24).

![Figure 24: transposition of the great vessels](image)

Cases of transposition can be sometimes identified during pregnancy by antenatal ultrasound. Of 34 cases reported to CARIS where time of diagnosis is known, 7 (21%) are reported as diagnosed before 24 weeks gestation and 8 (24%) by birth. Diagnosis in the newborn is confirmed by echocardiography. After birth, immediate management involves establishing safe oxygenation levels. If there is inadequate communication between the 2 circulations, a balloon atrial septostomy is performed during cardiac catheterisation to enlarge the small naturally occurring ASD and allow mixing of oxygenated and de-oxygenated blood. Surgical correction of the defect occurs using an arterial switch operation in which the two arteries are reconnected correctly to the ventricles. Without surgery, over 50% of children die before the age of 2. With surgery, outcomes have improved dramatically in recent years. In the absence of other risk factors, over 95% of infants successfully undergo surgery in the newborn period, although lifelong follow up and further treatment of additional complications is required.

51 cases of transposition of the great vessels were reported to CARIS for 1998-2001; in over two thirds of cases, the defect was not associated with other anomalies outside the cardiovascular system. The rate for live/stillborn births and terminations for congenital anomaly was 3.8 / 10,000 LB/SB/TOP (95% CIs 2.7-4.9), which is slightly higher than the EUROCAT rate of 2.5/ 10,000 LB/SB/TOP. Among the CARIS cases, 37/51 cases (73%) were liveborn. Of these, 32 (87%) survived to age 1 yr. Again the percentage surviving to 1 year is less than that given above for newborns surviving surgery. However, numbers are small and the Welsh cases may well include babies with additional risk factors or babies in whom the condition was not immediately recognised.
Babies born with this condition may only be slightly cyanosed at birth but this worsens over the next few days as the ductus arteriosus closes. The degree of cyanosis or congestive cardiac failure varies, depending on the pattern of blood flow. Management includes treatment of cardiac failure and drugs to maintain the ductus arteriosus. Surgical repair involving a number of stages is required to improve pulmonary circulation, including attachment of the superior vena cava to the pulmonary artery. By the ages of 2-5yrs children undergo the Fontan procedure in which the inferior vena cava is also connected directly to the pulmonary artery, forcing all blood returning to the heart to pass through the lungs, thus resolving the cyanosis. The response of children to treatment is usually good, with expected survival of all stages of surgical intervention of 75-95%. Long term problems often persist, including cardiac arrhythmias and long term lung dysfunction.

CARIS can identify 15 cases of Tricuspid atresia/stenosis reported for the years 1998-2001. In 2/3rds (10) cases the heart defects were not associated with anomalies in other body systems. One case was thought to be associated with lithium teratogenicity. The rate for live/stillbirths and terminations for congenital anomaly was 1.2/10,000 LB/SB/TOP (95% CIs 0.6 –1.8). The corresponding EUROCAT rate is 0.99/10,000 LB/SB/TOP.

Time of diagnosis is known for 10/15 cases. 3/10 (30%) were diagnosed before 24 weeks gestation. 5 (50%) were diagnosed in the first week of life and the remaining 2 were identified later in infancy. Four cases (including 1 detected antenatally) resulted in termination of pregnancy whilst the remaining 11 were liveborn. Of these, 9 (82%) were still alive at one year of age.

Tricuspid atresia

In tricuspid atresia, the valve between the right atrium and ventricle fails to develop, so that blood returning from the body to the right atrium cannot enter the right ventricle (which remains small). During fetal life the atretic valve can be bypassed by blood passing directly through the ostium secundum from right to left atrium. After birth, survival depends upon there being an opening through which blood can pass to allow it to flow through the lungs – usually from the left side of the heart through a VSD into the right ventricle or via a patent ductus arteriosus into the pulmonary artery (Figure 25). If the VSD is large, too much blood may flow into the lungs, causing congestive cardiac failure. If transposition of the great vessels also occurs, blood can easily get to the lungs via the left side of the heart and aorta.
Hypoplastic left heart

Hypoplastic left heart accounts for about 10% of critical congenital heart defects in infants. In its most common form the defect consists of an underdeveloped left ventricle, mitral valve and aortic root (Figure 26). Blood flow through the heart is maintained via a patent ductus arteriosus with death occurring should this close. Without treatment, survival is for no longer than the first few months of life at most. The condition usually occurs as an isolated cardiac defect without serious associated non-cardiac anomalies.

In the past, most children with hypoplastic left heart were managed with terminal supportive care. In the early 1980s Norwood described a surgical technique for repair of hypoplastic left heart which has subsequently been modified to a 3 stage surgical procedure. Pioneered in the United States, the technique is now increasingly used in the UK where up to 70% survival to 5 years of age has been reported with some patients now reaching their early teens. Surgery and the postoperative period can be difficult and places parents under considerable pressure. Concern has also been expressed about the possibility of neurodevelopmental problems as long-term adverse effects of treatment. It has been suggested that many parents continue to opt for termination of pregnancy rather than put their child through multiple operations with an uncertain long-term outlook. Cardiac transplant has been used as an alternative treatment, but the severe shortage of donor organs precludes widespread use.

International rates for hypoplastic left heart at birth are reported at around 1.5-3 / 10,000 total births / year. EUROCAT report a figure of 2.08/10,000 LB/SB/TOPs. CARIS received reports on 43 cases of hypoplastic left heart born in 1998-2001. In 26 (60%) cases the congenital defects were confined to the cardiovascular system. The CARIS rate for live/stillbirths and terminations for congenital anomaly was 3.3/10,000 LB/SB/TOP (95% CIs 2.3 - 4.3).

Time of diagnosis is known for 41/43 cases, of which 3 were diagnosed after termination of pregnancy for other anomalies. Of the remainder, 20/38 (53%) were diagnosed before 24 weeks gestation and a total of 26/38 (68%) were diagnosed before birth.

No spontaneous fetal losses are recorded but 56% (24) cases resulted in termination of pregnancy and 1 was stillborn. 7/18 (39%) liveborn cases survived to the end of the first year of life.

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O'Kelly SW, Bove EL. Hypoplastic left heart syndrome (Editorial) BMJ 1997; 314:87 (11 January)