We do not know how many adolescents and adults suffer from congenital heart disease but there is good reason to believe that the numbers are rising steadily year by year. The medical and surgical care of such young people is complex, expensive and needs particular expertise and training. There is no nationwide framework to provide care for these patients despite a great deal of prompting of the Department of Health by the profession.

A changing population

Surgery for congenital heart disease has seen more dramatic advances than most disciplines of modern medicine over the last 30 years. According to the Society of Cardiac Surgeons of Great Britain and Ireland, 3,000–4,000 operations are performed on children with congenital heart disease each year with a surgical mortality now in single figures. More than 85% of children born with congenital heart disease will survive, cured or palliated into adult life. There are probably more than 60,000 patients aged 16 years or over in the UK who require medical or surgical management of their congenital heart disease. Between 5,000 and 6,000 new patients each year now survive past their 16th birthday.

A wide spectrum of anomalies

The clinician treating adult congenital heart disease (ACHD) patients has to cope with a wide variety of anomalies and pathophysiological states. There are some patients who are ‘medical survivors’, whose lesion was diagnosed in childhood and who have survived without surgery into adult life.

Ventricular septal defect

Small ventricular septal defects (VSD) are usually haemodynamically insignificant, and therefore not considered for surgery. In the adolescent population, these defects are more frequently perimembranous than muscular because muscular defects have a greater tendency to spontaneous closure in the first few years of life. Although VSD is common, we are as yet unable to predict which lesion will persist into adult life. There are, however, risks even when the defect is haemodynamically so insignificant. Patients with a VSD have a lifetime risk of bacterial endocarditis, perhaps as high as 10%. Perimembranous VSDs are often associated with the development of aortic regurgitation in childhood or adult life even after they have closed spontaneously. Therefore, even very small VSDs merit regular, if not necessarily frequent, cardiological follow-up.

Oval fossa or central atrial septal defect

Despite the frequency with which the general population undergoes clinical examination, patients still present with congenital heart disease for the first time in adult life. The commonest ‘sleeper’ of this sort is the oval fossa or central atrial septal defect which is usually asymptomatic in childhood. First presentation may follow a secondary problem occurring in adult life: either the onset of atrial fibrillation or heart failure, particularly in middle-aged women. The management of such late presenting, middle-aged patients remains a matter of controversy. Closure of atrial septal defects in children and young adults is associated with an extremely small risk (<1%), so surgical closure can be safely recommended with confidence at virtually all ages. Small central atrial septal defects with small shunt (pulmonary flow to systemic flow ratio <1.5:1) have not in the past been put up for closure in childhood or adolescence. This may well change with the advent of highly sophisticated and successful closure devices which can be introduced at the time of cardiac catheterisation. Anecdotal evidence suggests that, left unclosed, these patients may develop clinically significant left to right shunts in middle life, particularly if the compliance of the left ventricle changes with acquired heart disease. In general terms, there is little data-based evidence to guide the clinician in the decision whether or not to close the defect in adult life.

Bicuspid aortic valve

The most common congenital cardiac anomaly is the bicuspid aortic valve. It may be totally unrecognised until the onset of calcification in middle age produces a murmur or symptoms. It is, however, important to recognise even the asymptomatic bicuspid valve as it places the individual at risk of bacterial endocarditis and necessitates lifelong antibiotic prophylaxis for clinical procedures and dental extractions. Most patients with bicuspid aortic valve have mildly dilated aortic roots. In most cases this is of no importance, but there is evidence to suggest that aortic dissection is more common in patients with bicuspid aortic valve than in those with the three cusp valve. For these two good clinical reasons regular, though not necessarily frequent, follow-up with assessment of valvular competence and aortic root size is important in the presence of congenital aortic anomalies.

Surgical survivors

Cyanotic congenital heart disease

In the 1950s and 1960s, when surgery first became possible for congenital cardiac malformations, complete repair
was unusual but there was widespread palliation using aorto-pulmonary shunts in cyanosed patients. Some of these palliated patients survived against all the odds and turn up from time to time in general medical clinic outpatients. They are frequently disabled and cyanosed, the underlying pathology is always complex, and they are often suffering from pulmonary hypertension and pulmonary vascular disease. While transplantation may be their only option, they should be referred for assessment at regional centres for ACHD.

If they had been born today, many of these complex patients would have undergone total correction with surprisingly good results. For instance, the commonest form of cyanotic congenital heart disease, transposition of the great arteries, was treated by re-routing the venous and atrial blood flow but leaving untouched the underlying abnormality of the great arteries. Although this works well in the short term, we now know that it is a poor long-term option. Total correction of transposition now takes place within the first month of life, with single figure mortality and probably a long-term life expectancy. The ACHD doctors will know in a generation whether this and other new surgical advances are as good as we think they are.

Tetralogy of Fallot

Some cardiac anomalies are considered to be correctable despite the fact that they continue to pose clinical problems because of disordered function. Tetralogy of Fallot has been repaired for several decades by patching the VSD and relieving the right ventricular outflow tract obstruction with transannular patches, pulmonary valvotomy and resection of myocardium. While the cyanosis and dyspnoea are immediately relieved and the patients grow and develop, many are left with a seriously dysfunctional right ventricle. It may be fibrotic, dilated and volume loaded, restrictive or have a degree of outflow tract obstruction. Potentially lethal ventricular arrhythmias are often associated and the patients are restricted in their physical capabilities. The arrhythmia management continues to pose a problem which may not always be amenable to medication. Pulmonary valve replacement is now accepted when pulmonary regurgitation is considered to be of clinical significance. More recent correction of tetralogy in infancy has been modified to reduce the amount of damage done to the right ventricular myocardium. By closing the VSD through the tricuspid valve and by restricting the amount of ventriculotomy, it is possible that the tetralogy patients of tomorrow will have fewer problems in later life.

Coarctation of the aorta

Another lesion which merits long-term follow-up after surgical repair is coarctation of the aorta. One-third of these patients have an associated bicuspid aortic valve, leading to the possibilities of calcification, obstruction and bacterial endocarditis already mentioned. The time at which the coarctectomy is carried out has a significant bearing on mortality and morbidity. Repair of coarctation in infancy by end-to-end anastomosis or subclavian flap procedure greatly reduces the incidence of systemic hypertension in the long term. If the correction is performed later in childhood or young adult life, the incidence of persisting systemic hypertension can be very high. Long-term follow-ups have in fact shown a less than satisfactory outcome for many apparently successfully corrected coarctation patients. In addition to systemic hypertension, they have accelerated atherosclerosis and cerebrovascular accidents.

Non-surgical problems in adult patients with congenital heart disease

Even after successful palliation and repair, ACHD patients require a great deal of support when seeking employment or life insurance.

Genetic implications

The ACHD patient needs to be aware of the genetic implications of reproduction. There is now good evidence that the recurrence risks for complex congenital heart disease are elevated. A father with congenital heart disease confers a recurrence risk of about 2% to his offspring – similar to that which pertains if a previous sibling was affected. However, the mother with congenital heart disease must expect a risk as high as 5% of recurrence in her offspring, and there are individual families where the risk is even higher.

Pregnancy

Pregnancy may or may not pose significant problems for the ACHD patient. Quite severe left ventricular outflow tract obstructions such as aortic
stenosis or coarctation are compatible with a satisfactory outcome in most pregnancies. On the other hand, young women who are severely cyanosed are less likely than normal women to carry a pregnancy successfully to term. The lower the maternal arterial oxygen saturation, the higher the risk of miscarriage. Following correction of cyanotic congenital heart disease such as tetralogy of Fallot, both symptomatology in pregnancy and the outcome return towards normal.

Although most congenital heart disease therefore has little effect on the ability of women to conceive and carry to term, the major exceptions are pulmonary vascular disease and pulmonary hypertension. Here, the risk of cardiac decompensation and death during pregnancy, particularly at the time of delivery, is high. Vascular resistances in pregnancy are generally reduced and variable. If the pulmonary vascular resistance is, however, high and fixed, right to left shunting may be increased with low cardiac output and hypoxia. There is a particular hazard to life during the rapid volume changes around delivery and labour. Most cardiologists would agree that established pulmonary vascular disease should be a contraindication to pregnancy. If, however, a young woman with pulmonary vascular disease does reach the stage of delivery, the involvement of a cardiologist or an intensivist used to handling this situation is obligatory during labour.

Non-cardiac surgical risks

Surprisingly, young adults with congenital heart disease may be most at risk when undergoing non-cardiac surgery. Inappropriate anaesthesia or endocarditis may cause problems, and cyanosed patients may frequently be at risk of uncontrolled haemorrhage. It is becoming increasingly likely that in the future this sort of surgery in high risk individuals will be performed in regional centres used to treating ACHD. Most ACHD centres in this country already have their own dental clinic and perform dental extractions in their own centres.

Summary

Adolescents and young adults with congenital heart disease are increasingly commonly seen in cardiological practice. Their problems are particular and often unique. Their management requires training and experience in the management of congenital heart disease by the attending cardiologist. In these days of clinical governance, management of ACHD patients must be supervised by clinicians specifically trained in handling congenital cardiac malformation.

References


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