Congenital heart disease in adults

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Abstract

Major advances in paediatric cardiac surgery over recent years have resulted in an increased number of children with congenital heart disease (CHD) surviving into adulthood. Currently in Europe, the adult CHD population is greater than that of the paediatric CHD population. This changing prevalence and ageing congenital population brings with it multiple co-morbidities and new challenges in all areas of their medical, surgical and psychological management. This article gives an introduction to the common conditions presenting acutely to general physicians, long-term complications, treatment options and the long-term considerations for end-of-life care. Adult congenital heart disease patients are safest treated in a network with a multidisciplinary team approach.

Keywords Adult congenital heart disease; aortic coarctation; congenitally corrected transposition of the great arteries; Eisenmenger syndrome; Fontan circulation; septal defects; tetralogy of Fallot

Changing prevalence of adult congenital heart disease

Major advances in paediatric cardiac surgery over recent years have resulted in an increased number of children with congenital heart disease (CHD) surviving into adulthood.[1,2] Approximately 90% of those born in the 1950s with complex CHD died during infancy, whereas the majority of those born in the 1980s survived beyond the age of 18 years.[3,4] Furthermore, increased awareness of CHD in the adult and improved imaging have led to more patients being diagnosed later in life, so that currently in Europe the adult CHD population is estimated at 2.3 million, compared with a paediatric CHD population of 1.9 million patients.[5]

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What's new?

- There are more adults than children with congenital heart disease
- Increasing use of percutaneous interventions in congenital heart disease
- Cardiac transplantation available to those with congenital heart disease
- Palliative care involvement recommended early

Common conditions presenting in adulthood

Examples of mode of presentation of CHD in the adult include unexplained dyspnoea, palpitations and dyspnoea during pregnancy, drug-resistant hypertension and intermittent claudication.

The common conditions presenting for the first time in adulthood include the following.

Atrial septal defects

Secundum ASD is the most common defect seen in adults (80%).[1] Other defects are discussed in more detail in the article on CHD in childhood. Patients often present with reducing functional capacity, exertional dyspnoea, and palpitations (commonly atrial tachyarrhythmias). Left untreated, late complications include right-sided heart failure with right heart dilatation and dysfunction, severe pulmonary vascular disease (<5%),[1] increasing tachyarrhythmias and associated thrombo-embolic phenomena.[1] Percutaneous device closure is the treatment of choice in the majority of symptomatic patients with secundum defects, with serious complications observed in ≤1% of patients.[6] Surgery is appropriate if device closure is not feasible and has a low mortality (approximately 1% in patients without significant co-morbidity).[7] Indications for ASD closure include right heart dilatation in the absence of irreversible pulmonary hypertension and paradoxical emboli.[1] Surgical closure remains the treatment option for primum and sinus venosus defects.

Ventricular septal defects

Ventricular septal defects (VSD) are the most common congenital heart malformation at birth (30–40%)[1] but can also present in adulthood. VSDs are classified by location in the ventricular septum (Figure 1).

Clinical presentations include:

- restrictive VSD with insignificant left-to-right shunt and no evidence of left ventricular volume overload but a holosystolic murmur on auscultation at routine medical examination
- unrestrictive VSD with significant left-to-right shunt causing left ventricular volume overload and symptoms of left ventricular failure
- Eisenmenger syndrome from a large unrestrictive VSD with the development of severe pulmonary vascular disease resulting in shunt reversal (right-to-left shunt), which will ultimately present with severe dyspnoea and cyanosis.

Treatment options include surgical closure, which has a low operative mortality (1–2%) and good long-term results.[8] Transcatheter closure is considered in selected cases with increased

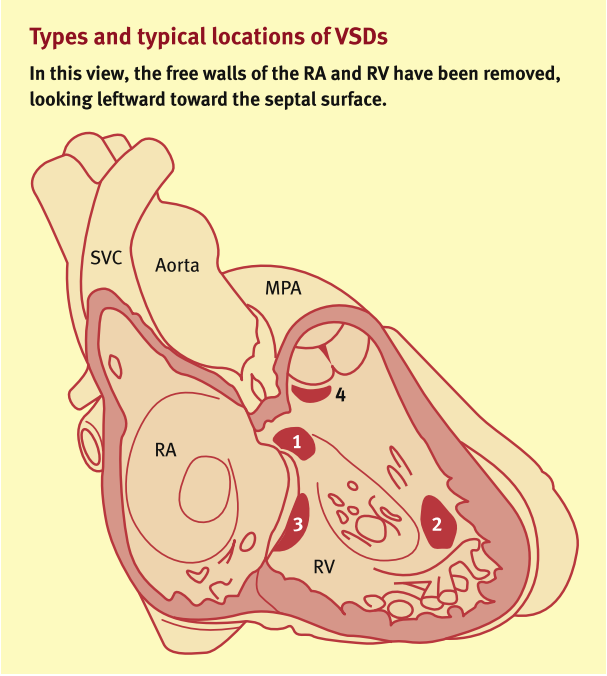

Figure 1 SVC indicates superior vena cava; 1, perimembranous VSD; 2, muscular VSD; 3, inlet/AV canal type VSD; and 4, subpulmonary VSD. AV, arteriovenous; MPA, main pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava; VSD, ventricular septal defect. Reproduced from Sommer RJ, Hijazi ZM, Rhodes JF Jr. Pathophysiology of Congenital Heart Disease in the Adult Part I: Shunt Lesions. *Circulation*. 2008; **117**(8): 1090–1099 with permission from Wolters Kluwer Health.

risk factors for surgery, including disadvantageous VSD position and accessibility.

Aortic coarctation

Aortic coarctation is a discrete or long-segment narrowing of the aorta,[1] typically located where the ductus arteriosus inserts just distal to the left subclavian artery. Aortic coarctation has a number of associated lesions, the most common being a bicuspid aortic valve (up to 85%).[1] The most common presentation is drug-resistant systemic hypertension but other symptoms may include headache, nosebleeds, dyspnoea secondary to hypertensive heart disease, abdominal angina and lower limb claudication. Left untreated, coarctation may result in left heart failure or aortic rupture/dissection. Uncontrolled hypertension may result in accelerated coronary and cerebrovascular disease.[9] Berry aneurysms are found in up to 10% of patients with coarctation.[10] Percutaneous stenting of native coarctation is the preferred treatment in adults with anatomically suitable stenosis. Angioplasty, with or without stent implantation, is effective for adults with recurrent or persistent coarctation.[11] Surgical techniques include resection with end-to-end anastomosis, prosthetic patch aortoplasty, interposition tube graft, and bypass jump grafts from ascending to descending aorta. After repair, patients require long-term follow-up for potential complications, including persistent arterial hypertension, recurrent stenosis, aneurysm formation and aortic valve disease, and, in highly selected cases, review of berry aneurysms (although there is no indication for routine screening in asymptomatic patients).

Congenitally corrected transposition of the great arteries (Figure 2)

Congenitally corrected transposition of the great arteries (ccTGA) has a prevalence of 1% of all congenital heart disease.[1] Isolated ccTGA rarely presents in adulthood but systemic ventricular dysfunction (morphological right ventricle) with dyspnoea and reduced exercise capacity is a common manifestation of this condition. Palpitations secondary to supraventricular and ventricular arrhythmias can be associated with ventricular dysfunction. Atrioventricular block may be present and transvenous permanent pacing and resynchronization pacing may be necessary. Cardiac transplantation may be indicated if severe systemic ventricular dysfunction ensues. Currently, physicians have varying opinions on the use of conventional heart failure medication in patients with systemic right ventricles.[12]

Acute presentation of previously diagnosed patients with congenital heart disease

With the increasing population of adult congenital heart disease, more and more adults with this diagnosis are presenting as general medical and surgical emergencies. Those conditions accounting for multiple hospital admissions due to late complications include:

Repaired tetralogy of Fallot (ToF)

ToF is the most common form of cyanotic CHD presenting after infancy, with an incidence approaching 10% of all forms of CHD.[1] Over 85% survive into adulthood after surgical repair.[13] Common complications of ToF in adulthood include:

- right heart failure secondary to chronic severe pulmonary regurgitation with right ventricular dilation and dysfunction
- atrial and ventricular arrhythmias related to residual haemodynamic lesions and surgical scarring
- left ventricular dysfunction secondary to longstanding preoperative cyanosis and multiple palliative procedures before late repair or inadequate myocardial protection during cardiopulmonary bypass.

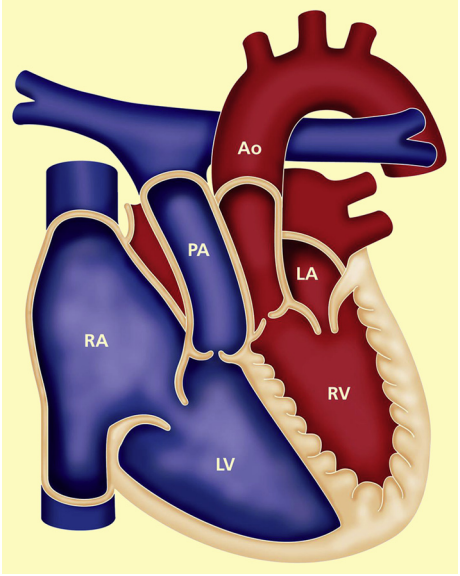

Figure 2 Congenitally corrected transposition of the great arteries (copyright of University Hospital Southampton).