

CHALLENGES IN THE CARE OF ADULT PATIENTS WITH CONGENITAL HEART DEFECTS

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This article will endeavour to accomplish four things: to point out some of the more common challenges in the assessment and management of adult patients with congenital heart defects (CHD); to review in general which adult patients with CHD do not need to be seen in specialised adult CHD centres; to review which patients should be seen in such specialised centres; and to review the resources which should thereby be available to them in the expert centres.

TIPS IN THE MANAGEMENT OF ADULT PATIENTS WITH CHD

Atrial septal defects

Even the humble atrial septal defect (ASD) has its complexities. First, not all ASDs need to be closed. As a rule, an ASD worth closing should be at least 10 mm in diameter and be associated with clear right ventricular dilation.

There is a consensus view that all ASDs meeting these criteria should be closed unless there are reasons not to do so.¹ Moreover, the ASD should be closed when diagnosed, and preferably before the age of 25 years if the goal of treatment is to avoid premature mortality and morbidity.²

The dilemma as to whether to recommend ASD closure in the asymptomatic patient has eased greatly now that device closure of ASDs can be done so successfully and easily. That said, one needs to have access to a skilled interventionalist and modern devices to make this promise relevant.

Estimation of the size, the number, and type(s) of atrial defects can be difficult at times. While transthoracic echocardiography (TTE) will usually confirm the diagnosis of an ASD, it will seldom identify whether multiple ASDs are present, an issue that must be evaluated by transoesophageal echocardiography (TOE) before considering device closure of the defect. Anomalous pulmonary venous drainage (APVD) must also be excluded before proceeding to device closure.

While most isolated ASDs are of the secundum type, it is important to recognise if a primum (~7%) or sinus venosus defect (~8%) is present, since device closure is inappropriate, and since referral to a congenital heart surgeon should be considered.

The presence of associated anomalous pulmonary venous drainage should be looked for in all ASDs, usually with TOE or magnetic resonance imaging (MRI) to ensure that all four pulmonary veins drain into the left atrium. The surgeon or the interventional cardiologist should have this information before the procedure. If anomalous pulmonary venous drainage is present, device closure is not appropriate, and referral to a congenital heart surgeon should be considered.

All patient candidates for transvenous pacing should be evaluated for an ASD or possibly even a patent foramen ovale, since they are at increased risk of strokes and systemic embolic events through the embolisation of pacing lead thrombi.

Ventricular septal defects

While small ventricular septal defects (VSDs) may be seen in practice, it is important to look for associations that may require repair. First, check that there is no more than mild associated aortic regurgitation. In some patients outlet VSDs³ occur which can lead to aortic cusp prolapse and clinically important aortic regurgitation. Such patients should be evaluated as possible surgical candidates. Other patients with what is believed to be small VSDs may also have important right ventricular outflow tract obstruction causing right ventricular hypertrophy, symptoms, and atrial flutter. They too are surgical candidates.

Other patients believed to have small VSDs⁴ may dilate the left heart chambers in follow up, which in turn may lead to atrial fibrillation and congestive heart failure. A periodic TTE and awareness of this issue should avoid this happening.

Patent ductus arteriosus

Isolated patent ductus arteriosus (PDA) occurs in four forms:

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- ▶ silent PDAs (discovered on echocardiography usually, with no chamber dilation and no murmur, which probably should not be followed)
- ▶ small PDAs exposing the patient to the risk of endarteritis (no or mild chamber enlargement but with a (preferably continuous) murmur—traditionally considered for closure to reduce a very low endarteritis risk)
- ▶ moderate PDAs with the typical murmur plus dilation of the left heart chambers (requiring consideration of closure for haemodynamic reasons)
- ▶ large PDAs which have caused severe pulmonary hypertension and usually Eisenmenger syndrome (who are too late for closure).

Physicians should note that left-to-right shunt calculations based on oxygen saturations at heart catheterisation are not valid in PDA patients because of streaming, and should not be used in decision making. Regardless of left-to-right shunt quantification, PDAs associated with evidence of significant shunting are candidates for closure.

Modern treatment in many countries is with device closure because of its ease and high success rate in expert hands. Surgery in the older adult is more difficult because of ductal calcification and friability.

Aortic coarctation

Making the diagnosis of aortic coarctation can be an uplifting experience. This can be achieved by finding some combination of the following: right arm hypertension; radiofemoral delay; weak leg and foot pulses; low popliteal or pedal pressures; and murmurs or bruits over the thorax. As we all were taught, we will find what we look for.

Most patients have a bicuspid aortic valve. Even after repair of a coarctation, the physician should know that recurrent or persistent coarctation⁵ can occur, and may need special management. Patients who have had coarctation repairs need the same physical exam manoeuvres conducted, and imaging via MRI or computed tomographic (CT) scanning or angiography looking for coarctation, hypoplastic aortic arch (both of which can cause hypertension), and aneurysm formation.⁶

ISSUES TO BE AWARE OF FOR PATIENTS ALSO UNDER EXPERT CARE

Repaired tetralogy of Fallot

The most common issue in these patients is residual pulmonary regurgitation. Echo labs without specific expertise in CHD should not evaluate patients who may have complex CHD. Competent echocardiographic evaluation can quite accurately determine the presence of severe pulmonary regurgitation by finding a laminar flow pattern, and can also evaluate lesser degrees of pulmonary regurgitation. In the opinion of several experts, echocardiography is not very good at assessing right ventricle size, or systolic function. Fortunately, MRIs and even radionuclide angiography (RNA) in good facilities with well trained staff are better at assessing these issues.⁷ This same principle applies to all right ventricles, regardless of whether they are subpulmonary or subaortic (for example, Mustard or Senning repair, or congenitally corrected transposition).

Sustained atrial or ventricular arrhythmias should be seen to reflect haemodynamic problems in these patients.⁸ The arrhythmia needs to be managed, but a complete and expert review of haemodynamics needs to be undertaken as well.

The availability of radiofrequency ablation techniques in atrial flutter, of surgical cryoablation for atrial flutter, and of a surgical maze procedure for atrial flutter (right atrial) and

fibrillation (both atria) may need consideration.^{9,10} Such therapy should be performed by individuals with special training and experience in these techniques.

Clinicians should be “on the look-out” for tetralogy patients with a CATCH-22 deletion. Pediatricians are usually most attuned to this possibility. Knowing that this is present should translate into counselling that the patient has a 50% chance of passing on the 22q deletion to any child he/she may have.

Mustard/Senning repair of transposition of the great arteries

Systemic right ventricular function is best evaluated in expert cardiac MRI and RNA centres with specific expertise in adult CHD. Systemic tricuspid regurgitation may impose an additional load on the ventricle. There is no consensus as to whether patients with a systemic right ventricle should be treated with angiotensin converting enzyme inhibitors or other agents.

Mustard and Senning patients typically have a slow heart rate. Asymptomatic bradycardia and “pauses” in the asymptomatic patient may lead to unnecessary pacemaker insertion. If pacing is needed, implantation should be performed by skilled personnel who have had experience navigating the baffles to implant and secure the ventricular lead in the morphologic left ventricle.

Most Mustard and Senning patients are able to live fully or almost fully active lives, including sporting activities and the ability to carry a pregnancy (although we recommend such women be evaluated by an expert before conception).

Congenitally corrected transposition of the great arteries

The diagnosis of congenitally corrected transposition of the great arteries (ccTGA) may not be made until adult life. If there are no associated lesions, such patients may present with “mitral regurgitation” or atrioventricular block (complete or only first degree) or with a pacemaker without the physician being aware that the fundamental problem is ccTGA.

Fibromuscular subaortic stenosis

In the unoperated patient, aortic regurgitation, as well as the degree of obstruction, will determine whether surgical evaluation by an expert may be required. After resection, obstruction will recur in about 30%,¹¹ and should be picked up by periodic evaluation.

Fontan procedure

These patients have truly unique physiology (imagine living with only one ventricle) and have one of several types of connections to create their Fontan circulation. They are potentially very fragile patients, and the physician should instinctively seek quick expert attention if the patient develops a sustained atrial arrhythmia, develops oedema or ascites, becomes pregnant, is in need of surgery, or experiences a failing physical capacity.¹²

Physicians should note that the right atrial and jugular venous pressure should be elevated in Fontan patients, typically to 10–15 cm above the sternal angle. Diuretics should be avoided.

Ebstein anomaly

These patients are rare. They may present with a wide range of severity—being pink or blue, having normal heart size or a huge heart, with normal or abnormal physical abilities, and with the presence or absence of arrhythmias. They need expert assessment unless the condition is clearly mild.

Cyanotic patients with congenital heart disease

All cyanotic CHD patients should be seen as fragile and high risk, and should be seen regularly in expert centres. A few points of general interest can be made. Therapeutic phlebotomy or venesection should only be offered to cyanotic patients with symptoms of hyperviscosity. These symptoms should disappear after adequate phlebotomy, and the same symptoms anticipated if further phlebotomy were to be necessary. The haemoglobin concentration should not be < 170 g/l (10.6 mmol/l) in most patients with saturations $< 90\%$.

Cyanotic patients should avoid iron deficiency, a state associated with: an increased risk of stroke; the development of hyperviscosity symptoms at a lower haemoglobin concentration; and with lessened functional capacity. While iron replacement will usually lead to a higher haemoglobin, hyperviscosity symptoms will often not reappear or may only after a higher haemoglobin concentration is reached.

Oxygen saturations should not be measured until the cyanotic patient has been resting for at least five minutes. Cyanotic patients should have bubble traps or air filters in any intravenous lines to prevent paradoxical emboli. The basis for cyanosis in each patient should be understood, with a particular emphasis on finding treatable causes. Cyanotic patients can be destabilised very easily by respiratory infections or arrhythmias, and should immediately receive appropriate treatment.

The cyanotic woman should know that the chances of her producing a healthy living baby are directly related to her saturation levels, and that success—if her saturation is less than 85%—is likely to be very low.¹³ If she conceives, expert care is recommended in a high risk pregnancy unit.

Eisenmenger syndrome

This is a special group of cyanotic patients. In addition to the considerations just mentioned, the following points are important:

- ▶ all patients with this diagnosis may die at any time
- ▶ any surgical intervention or anaesthetic must be planned carefully
- ▶ haemoptysis may be life threatening
- ▶ seemingly pedestrian respiratory infections may cause death
- ▶ pregnancy must be avoided.

All such patients should have been evaluated by an adult CHD expert, and should ideally be monitored by them to anticipate and prevent health threats.¹⁴

Pregnant patients

The physician should ensure that women with CHD receive appropriate counselling regarding pregnancy once they are, or expect to become, sexually active. The physician should consider which conditions may be genetically transmitted, and counsel or arrange appropriate counsel re the recurrence risk of CHD and other events for the aspiring couple/person. Inherent in such counselling must be an understanding of the likely life and health expectations of the affected parent, so that this can be included in the discussion.¹⁵

There is growing literature on the safety of pregnancy in various conditions. Validated risk measures have been published¹⁶ that can be applied fairly easily. Maternal risk factors include:

- ▶ prior cardiac event (heart failure, transient ischaemic attack, or stroke before pregnancy) or arrhythmia
- ▶ baseline New York Heart Association functional class $> II$ or cyanosis

Table 1 Adult congenital heart patients not needing care in an expert centre

"Simple" valves	<ul style="list-style-type: none"> ▶ Isolated aortic valve disease ▶ Isolated mitral valve disease (except parachute or cleft mitral valves) ▶ Isolated tricuspid valve disease (except Ebstein anomaly) ▶ Mild pulmonary valve stenosis
"Simple" shunts	<ul style="list-style-type: none"> ▶ ASDs—small, or repaired without residual problems (e.g. pulmonary hypertension and arrhythmias) ▶ VSDs—small and isolated, or repaired without residual problems ▶ PDAs—small and closure not planned, or closed without residual problems

ASD, atrial septal defect; PDA, patent ductus arteriosus; VSD, ventricular septal defect.

- ▶ left heart obstruction (mitral valve area < 2 cm², aortic valve area < 1.5 cm², or peak left ventricular outflow tract gradient > 30 mm Hg by echocardiography)
- ▶ reduced systemic ventricular systolic function (ejection fraction $< 40\%$).

In this trial, four indicators to assess risk were used. Predictors of primary cardiac events were incorporated into a revised risk index in which each pregnancy was assigned 1 point for each of the four predictors present. The estimated risk of a cardiac event in pregnancies with 0, 1, and > 1 points was 5%, 27%, and 75%, respectively. Referral of women with risk factors to an expert in high risk pregnancy should be considered.

GENERAL CHALLENGES

Diagnostic facilities for adult patients with complex CHD must have contemporary equipment and expert staff. This is true for echocardiography, MRI, and heart catheterisation in particular. As a rule, these facilities will probably be found at an expert centre for adult CHD or at a paediatric cardiology facility.

Patients and family members should also educate themselves and take an active role in their own management as best they can. They can access guidelines for the assessment and management of patients such as themselves. A big step forward would be taken if more complex patients with CHD carried a "health care passport"¹⁷ or copies of their own medical reports.

Which adult CHD patients do not need expert care?

Many patients having what I call "simple valves" and "simple shunts" do not need follow up in expert adult CHD centres (table 1).

Which adult CHD patients do need expert care?

All other patients with CHD (table 2) should be assessed periodically in an expert centre, with the visit frequency determined on the basis of the patient's estimated stability, his or her likelihood of developing new problems, and the severity of the consequences if the condition destabilises.

Looked at another way, the patients with CHD who should be seen in the expert centres are those who face a significant prospect of premature death, reoperation/reintervention, or complications of their condition(s), and for whose care the community cardiologist has not been well trained (table 2).

As Professor Jane Somerville has said, most cardiologists should shy away from caring for congenital cardiac conditions

Table 2 Types of patients who should be seen at expert centres

- ▶ Congenital cardiovascular conditions not previously seen in one's practice
- ▶ Conditions sufficiently unfamiliar that the practitioner should consult a textbook or colleague
- ▶ Adult congenital cardiovascular patients:
 - with pulmonary hypertension
 - with poor functional class
 - who are pregnant or considering becoming so
 - who are cyanotic
 - who may have inherited conditions (e.g. CATCH-22)
 - with transposition, coarctation, moderate or severe pulmonary valve disorders, and conduits
- ▶ Adult congenital cardiovascular patients with named conditions:
 - Mustard
 - Senning
 - Rastelli
 - Jatene (arterial switch)
 - Fontan
 - Fallot
 - Ebstein
 - Eisenmenger
 - Marfan
 - Holt-Oram

bearing men's names. These conditions are generally complex and prone to serious complications (table 2).

Practical clues to patient management

Physicians who would like to access expert recommendations as the need arises can do so at www.achd-library.com (a site for professionals) or at www.cachnet.org (a site for patients and family). In these consensus conference recommendations regarding the management of adult CHD, one can find quick summaries of the issues regarding the major conditions, recommendations for care, and references supporting the care plans.

Cardiologists and sometimes primary care physicians should establish a referral relationship with an expert adult CHD centre.¹⁸ This relationship should permit access to timely telephone advice and informal consultations, as well as referral for consultation, assumption of care, and plans for joint follow up, when appropriate.

The Grown-Up Congenital Heart (GUCH) Working Group of the European Society of Cardiology will soon publish a supplementary set of European recommendations for the care of adult CHD or GUCH patients. An organisational and practical set of recommendations to advance the care of these patients in the USA were recently published,^{14 17 18} and a report of the British Cardiac Society working group on GUCH has also been published recently.¹⁹

The basic problem: more complex patients, few expert cardiologists

Thanks to the treatment successes of the past 40 years resulting in the saving of many lives of children with CHD, the number of adult patients needing surveillance continues to grow. The health care systems in most developed countries have not yet properly addressed the needs of these adult patients. There is a serious shortage of professionals, including paediatric cardiologists and adult cardiologists, trained to take responsibility for these adult patients. There are too few centres of excellence to act as the anchors for this care.

Instead, there are too many groups and units caring for these patients in most developed countries. Consolidation to help develop a regional adult CHD centre of excellence should

Table 3 Expert skills and services required in an adult congenital cardiac expert centre*

- ▶ Cardiology (preferably both paediatric and adult trained)
- ▶ Cardiac surgery (congenital heart surgery is usually based at a children's hospital)
- ▶ Diagnostic and interventional catheterisation
- ▶ Echocardiography
- ▶ Electrophysiology
- ▶ Special imaging (especially expert magnetic resonance imaging)
- ▶ Nursing
- ▶ High risk pregnancy and reproductive counselling
- ▶ Special intensive care and anaesthesiology

*Any team member may fill one or more functions if qualified to do so.

be a priority. Professional and institutional interests often seem to trump patients' interests. Medical leaders, insurers, and governments have seldom shown a commitment to high quality care for these patients, and need to be educated on these matters to plan the needed services.

The solution in brief

The care of rare and dangerous conditions should be regionalised. The principle that regionalisation of care is needed for adult patients with complex CHD has been widely accepted. The only way to offer high quality care for the complex patients at a national or regional level is as follows. For any given country, train and retain enough experts of various types (table 3) to meet the anticipated needs of these patients. Decide which centres should be the regional anchors or expert centres for the country. See that patients and their physicians can access these needed skills. Ensure that these regional centres establish strong relationships with physicians and hospital units in their area, so that cooperation in patient care is encouraged.

The expert multidisciplinary adult CHD teams in strategically located units should each be associated with a strong paediatric cardiac group. The expert adult CHD centre should serve a population of 5–10 million people so the team becomes well honed and best able to meet the needs of the patients referred to them.

The ultimate test of the appropriateness of the care provider is the question "What's in the best interests of this patient?"

CONCLUSION

Physicians of adult patients with CHD can have an important role in their management. They may care for the patient if the condition is simple, or see that the patient gets into the right expert hands. They should know enough to help protect the patient from unwise initiatives. They can ensure the patients present for regular visits, have appropriate pre-conception counselling, and are seen in a high risk pregnancy unit if this seems appropriate.

The care of complex adult patients with congenital cardiovascular disease should be regionalised in all developed countries. Champions must emerge to make this happen. They and their colleagues can make major contributions to ACHD care in their country and beyond.

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