

**ADULT CONGENITAL HEART DISEASE  
IN THE NETHERLANDS  
GUIDELINES 2000**

**Derived from the 1996 Consensus Conference  
on Adult Congenital Heart Disease  
Commissioned by the Canadian Cardiovascular Society**

**Updated and adapted by :  
workinggroup "Congenitale Cardiologie bij Volwassenen" of the Netherlands  
Society of Cardiology**



## Leesinstructies bij de behandelingsrichtlijnen voor volwassenen met aangeboren hartafwijkingen.

1. De richtlijnen geven een beknopte beschrijving van de achtergronden, hemodynamiek, diagnostische en therapeutische mogelijkheden van iedere afwijking, zonder de pretentie te hebben een tekstboek te kunnen vervangen.
2. Behandelingsadviezen zijn gedrukt in *grijze kaders*. De mate waarin het advies wetenschappelijk kan worden onderbouwd en daarmee samenhangend het gewicht dat aan het advies moet worden toegekend, zijn weergegeven onderin het grijze kader (“grade” en “level”). De betekenis van deze *grades en levels* wordt beschreven in appendix II.
3. Omwille van de gebruiksvriendelijkheid zijn adviezen die voor meerdere aandoeningen van belang zijn, bij elk van deze aandoeningen opnieuw afgedrukt. Dit leidt tot herhalingen die storend kunnen zijn voor de lezer die de guidelines van de eerste tot de laatste bladzijde doorleest. Om de leesbaarheid te bevorderen zijn adviezen die meer dan eenmaal zijn opgenomen *cursief* gedrukt. Cursivering duidt dus niet op een extra groot belang van de betreffende tekst.
4. Kleine aanpassingen voor de Nederlandse situatie door de bewerkers aangebracht zijn aangegeven door middel van een ander `lettertype(Gothic)`. Uitgebreidere aanpassingen of veranderingen zijn aangegeven met ♥ (Marfan syndroom, Aritmieën, General Recommendations en Endocarditis profylaxe).

## **TABLE OF CONTENTS**

| <b>Section</b> | <b>Title</b>   | <b>Page</b> |
|----------------|--|-------------|
|                | Foreword   |             |
|                | Introduction   |             |
| I              | General Recommendations  |             |
|                | Part I – Organisation  |             |
|                | Part II – Specific Issues in the Care of patients with ACHD        |             |
|                | Part III – Care of the Cyanotic Patient                            |             |
|                | Part IV – Arrhythmias in Adult Congenital Heart Disease            |             |
| II             | Atrial Septal Defect   |             |
| III            | Ventricular Septal Defect  |             |
| IV             | Atrioventricular Septal Defect                                     |             |
| V              | Patent Ductus Arteriosus   |             |
| VI             | Left Ventricular Outflow Tract Obstruction & Bicuspid Aortic Valve |             |
| VII            | Right Ventricular Outflow Tract Obstruction                        |             |
| VIII           | Tetralogy of Fallot  |             |
| IX             | Coarctation of the Aorta   |             |
| X              | Ebstein Anomaly  |             |
| XI             | Complete Transposition of the Great Arteries                       |             |
| XII            | Congenitally Corrected Transposition of the Great Arteries         |             |
| XIII           | Eisenmenger Syndrome & Pulmonary Vascular Obstructive Disease      |             |
| XIV            | Fontan Operation   |             |
| XV             | Marfan syndrome  |             |
| XVI            | Coronary Anomalies   |             |
| XVII           | Transplantation Issues   |             |
| XVIII          | Reproductive Issues  |             |
| XIX            | Infective Endocarditis   |             |
| XX             | Psychosocial Issues  |             |
|                | Appendix I – Endocarditis Prophylaxis Recommendations              |             |
|                | Appendix II – Levels of Evidence                                   |             |
|                | Appendix III – Glossary of Terms                                   |             |
|                | Appendix IV – Panel members and workinggroup members               |             |
|                | Bibliography   |             |

## **FOREWORD**



In the past decades prognosis and life expectancy of patients with congenital heart disease have dramatically improved, as a result of improved surgical techniques. Increasing numbers of these patients are now reaching adulthood, which has led to a new population of patients consisting of adults with a surgically corrected heart defect. In the Netherlands the number of adults with congenital heart disease is estimated to be around 20.000 to 25.000, increasing at a rate of 5% per year. 75% of these patients have undergone a palliative or corrective heart operation in the past.

However, postoperative morbidity is substantial and sufficient knowledge and experience in the care of these patients is still lacking.

Several years ago the Canadian Cardiovascular Society invited a panel to make recommendations for diagnosis and treatment of this group of patients. In 1996, the Consensus Conference Report on Adult Congenital Heart Disease has been published. The panellists are mostly international experts in the field of congenital heart disease and therefore their recommendations are the best available given present knowledge. The Consensus is written for members of the Canadian Cardiovascular Society, but with some adaptations especially concerning the organisation, the Consensus is very suitable for members of the Dutch Society of Cardiology. Updating and adapting have been performed by the members of the workinggroup “Congenitale Cardiologie bij Volwassenen” of the Dutch Society of Cardiology (NVVC).

To optimise knowledge and experience with this new group of patients, concentration of these patients in specialised centers is of great importance. This has also been stated in a recently published report of the “Commissie Congenitale Cardiologie” (Committee for Congenital Cardiology) consisting of members of the Dutch Society of Cardiology (NVVC), the Dutch Society of Paediatrics (NVK), the Dutch Society of Thoracic Surgery (NVT) and the Dutch Heart Foundation (NHS). This committee has made a survey of the actual state of organisation and care for adults with congenital heart disease in the Netherlands. Concentration of care for this group of patients in specialised centers is necessary to reach optimal knowledge and experience and so optimal care for these patients. A uniform database should be developed for all patients in the Netherlands and research and education should be coordinated by the centers. Collaboration with other hospitals remains essential because many patients would benefit from periodic contact with a cardiologist in their community along with their specialist at the referral center.

These recommendations are of temporary nature and it is clear that high standards of evidence can not yet be reached. The recommendations need to and will be refined and strengthened substantially in the next few years. This population of patients is young and new and persuasive clinical research is greatly needed.

Workinggroup “Congenitale Cardiologie bij Volwassenen” of the Dutch Society of Cardiology

## **INTRODUCTION**

We, the authors of this Consensus Conference Report on Adult Congenital Heart Disease (ACHD), are grateful to the Canadian Cardiovascular Society (CCS) and its Council for the opportunity to assemble the document which follows.

To our knowledge, such recommendations have never before been compiled. Consequently, we expect that this document will attract international interest. Because of this, we have assembled some highly respected international members to join in the process. While most of the contributing panelists are CCS members serving the purposes of our society, we are especially grateful to Drs. Richard Liberthson, Joseph Perloff, Francisco Puga, Jane Somerville, and Carole Warnes for contributing their time, talents, and wisdom to this project.

The recommendations which follow are the best available given present knowledge. We believe these will be refined and strengthened substantially in the next few years. With the support of the Canadian Cardiovascular Society, we as a group will periodically update this information, so that this document will live and evolve over time.

These recommendations have been written for “members of the Canadian Cardiovascular Society”. This is important to state, since an audience more or less knowledgeable about the subject would require a different amount of background information and depth of treatment of the material.

The recommendations are timely, since ever-increasing numbers of people with congenital heart defects are reaching adulthood. Canada as a society will decide how this challenge will be met. We physicians, surgeons, and other caregivers will decide how best to do our part.

We panelists are convinced that the interests of any but the most simple patients are best served by involving what we call “regional or supraregional referral centres”. The knowledge and experience in the care of these patients should be focused, so that competence and skill become available as quickly as possible. This recommendation is not intended to stand in the way of involving local physicians in the care of these patients as collaborating members of a team with the best interests of the patients at heart. Almost all these patients require primary care. Many would benefit from periodic contact with a cardiologist in their community, along with their specialist at the regional referral centre.

As time passes, there will be a liberalisation of the care of less complex patients. Each community will find its own way to deal with these patients. We hope to be able to address the changing situation in later reports.

One of the problems and challenges of ACHD is the large number of different lesions and situations one may encounter. Those of us working hard in the area have fought for our competence and confidence, and regularly continue to come up short in managing individual patients. We have great respect for the seemingly endless scenarios we encounter.

Patients with congenital heart disease (CHD) are interesting to have in one's practice. This should be done either collaboratively with a regional referral centre, or after one has concluded that the patient does not need such a referral. The natural interest in “collecting a few congenitals” should be resisted until this test has been run. This principle applies as much in a surgical as a medical practice.

Canada is fortunate to have a national group of regional referral centres called the CACH

Network. We encourage Canadian readers to make use of these facilities and the skills and experience they represent. More information can be obtained on the Internet at

<http://www.cachnet.org>

Another unique aspect of this consensus conference is that it is available on the Internet. We have thus been able to receive input from a remarkable number of people, with different backgrounds and interests.

The Grown Up Congenital Heart Working Group of the European Society of Cardiology has endorsed the document.

Equally unique is the fact that we did not meet as a group. We did this for the first time in Montreal when we presented this report to a plenary session of the CCS. The wonders of e-mail and faxes, supplemented by conference calls, and energised by individual contributions made a personal meeting unnecessary.

We decided to write this material in as user-friendly a fashion as possible. We envisaged a clinician looking up a lesion, and wishing to see the recommendations “at a glance”, rather than having to refer to other sections of the report. This has led to some repetition for the reader who begins at the beginning, and ends at the end. *The repetitive portions are printed in italics to reduce frustrations resulting from this style.*

We also committed to NOT writing a textbook, even though a good and current one is needed for an audience such as ours. We have focused on the principles of management of these patients, leaving latitude where possible for the clinician to exercise judgement. We wish to guide, but not to constrain unduly.

We have given weight to our management recommendations. The scales which we used are shown in Appendix II. We accepted Dr. David Naylor’s advice in this regard, and use standards similar to those used in past CCS Consensus Conferences. Indeed, we wish we could meet the high standards of evidence met by these earlier reports. We cannot. Our field and our patients are young. Persuasive clinical research is greatly needed. We will mature. Our evidence will become stronger in the next decade.

We hope these recommendations will be found helpful to the patients in whose interests we have written them, and to those who care for them. Canadians have made important contributions to the management of patients with CHD. We hope this report will follow in this tradition.

### Choice of Panelists

The CCS invited Dr. Gary Webb (president of the Canadian Adult Congenital Heart Network) to lead the process. Drs. Gilles Dagenais (representing the CCS), William G. Williams (representing surgery), and Webb selected the primary and secondary panels. The panels were selected so as to receive input from various interested groups (adult/medical cardiology, pediatric cardiology, cardiac surgery, patients), the various regions of Canada, and both English and French-speaking Canadians.

The international panelists are world renowned experts in the field of ACHD, both medically (Drs. Richard Liberthson, Joseph Perloff, Jane Somerville, Carole Warnes) and surgically

(Dr. Francisco Puga).

Dr. Michael Connelly was the 'lead writer' of the document. Drs. Webb and Connelly worked closely in producing it. Most of the panellists worked very hard reviewing many drafts, and offering suggestions for improvement. The panel had almost no difficulty in reaching agreement on the statements made. Debate occurred only where there was insufficient data to decide a point.

The French version of the document was proofed by Drs. Lise-Andrée Mercier and Georges Delisle.

## **SECTION I - GENERAL RECOMMENDATIONS**



### **Part I - organisation**

The recommendations in the Consensus Conference Report are focused on the Canadian situation. Therefore some adaptations concerning the organisation in The Netherlands are necessary.

In 1998 a report of the “Commissie Congenitale Cardiologie” (Committee for Congenital Cardiology) has been published, called “cardiologische zorg voor patiënten met aangeboren hartafwijkingen: van kind naar volwassenheid”. In the Committee members of the Dutch Society of Cardiology (NVVC), the Dutch Society of Paediatrics (NVK), the Dutch Society of Thoracic Surgery (NVT) and the Dutch Heart Foundation (NHS) were assembled. In the published report recommendations are given for the organisation of the care for adults with congenital heart defects in the Netherlands. A summary of these recommendations is presented.

### **Proposal for the organisation of congenital cardiology**

The Committee is convinced that the care for adolescents and adults with congenital heart defects has to be concentrated in specialised centers or clusters (co-operations between academic hospitals). Congenital cardiologists working in these centers should have special facilities and they are primarily responsible for the care of patients with a congenital heart defect. Diagnosis and therapy for any but the most simple patients should take place in these centers.

Collaboration with other hospitals remains essential because many patients would benefit from periodic contact with a cardiologist in their community along with their specialist at the referral center. Not all hospitals, however, will be able to deal with the specific problems of these patients. Therefore the Committee made a list of types of care which are to be expected in these patients:

1. Care for acute complications, such as rhythm disturbances or acute heart failure.
2. Care for additional cardiac disorders, for example treatment for angina pectoris or chronic rhythm disturbances.
3. General specialist care, such as regulation of diabetes mellitus or peri-operative advice for non-cardiac surgery.
4. Regular follow up in the out-patient clinic.
5. Care in the terminal phase.

For acute complications (1) patients should be treated in their own neighborhood. Therefore, in each hospital primary care for acute complications should be available and after treatment, contact should be made with the specialist in the referral center for further treatment and follow up.

For the other types of care (2-5) special affiliated hospitals should be created, where a cardiologist with special interest and some basic training and facilities collaborates with a center or cluster.

In this model of shared care the amount and intensity of participation of the interested cardiologist will be individually determined, dependent on the special interest and expertise of each cardiologist. Therefore, an exact list of types of patients who may be cared for exclusively in the affiliated hospitals, or who should be seen at referral centers can not be given. Each center should collaborate with different affiliated hospitals in a different way according to clear, individually made agreements. The specialist in the referral center will advise individual patients how and where follow up will take place. A general design of conditions and facilities necessary for the care of patients with congenital heart disease in different types of hospitals is derived from the report.

### General design for the care of adults with congenital heart disease

|  | Center/Cluster | Affiliated | Local |
|--|----------------|------------|-------|
| Cardiac surgery                                | X              |            |       |
| Congenital heart surgery                       | X              |            |       |
| Intervention cardiology                        | X              |            |       |
| Congenital cardiologist                        | X              |            |       |
| Interested cardiologist                        |                | X          |       |
| Cardiologist                                   |                |            | X     |
| Out-patients care $\geq$ 20 min/patient        | X              | X          |       |
| Structured meetings<br>(surgeon, cardiologist) | X              |            |       |
| Psychosocial team                              | X              | X          |       |
| Electrophysiology                              | X              |            |       |
| Interested gynaecologist                       | X              | X          |       |
| Interested pathologist                         | X              |            |       |
| Genetic counseling                             | X              |            |       |
| Congenital database                            | X              | X          |       |
| Regular visitation                             | X              | X          |       |

## **Part II - Specific Issues in the Care of Patients with ACHD**

### **Non-cardiac Surgery**

Performance of any surgical procedures in most adult patients with CHD carries a greater risk than in the normal population. Evaluation in a center for congenital cardiology prior to surgery is recommended, and in the case of unoperated or complex ACHD, it is recommended (where feasible) that the surgery be carried out in the center for congenital cardiology, utilizing experienced cardiac anesthetists. This is strongly recommended for cyanotic patients, patients with pulmonary hypertension, or with some rhythm abnormalities. Pregnant women with CHD should be managed by the patient's obstetrician and ACHD cardiologist together with a cardiac anesthetist if necessary. In complex cases, an obstetrician knowledgeable in the management of ACHD is optimal. Post-operatively, the patients with CHD *may* need ICU/monitoring facilities even for relatively minor procedures.

### **Dental Care**

Regular dental care, often in a hospital setting, is mandatory for adult patients with CHD to decrease the likelihood of caries, abscesses or periodontal disease, all of which contribute to increased incidence of infective endocarditis. There is justification for government subsidization of dental care in those patients unable to afford it. Endocarditis prophylaxis is recommended except for 'fillings above the gum line'.

### **Informed Consent**

Despite its lifelong presence, most adolescents and young adults with CHD have inadequate knowledge about their cardiac conditions. Health care providers must assess each patient's knowledge of his or her condition and give appropriate information to enable independent decision-making about choices in care. Adults with CHD should be encouraged to understand not only their disease, but the medications they use. They should be involved in major management decisions or decisions involving invasive procedures. Further involvement of patients in evaluation of processes, programs, and in the planning of research trials within the constraints of their motivation and capacity to understand them is ideal. Patients should be encouraged to inform their specialists themselves of any new events which may occur.

### **Advance Directives and Palliative Care**

Patients should be made aware of the utility of advance directives which are legally binding. Their use may reduce uncertainty when caring for critically ill individuals. Likewise the role of non-intervention, or of palliative care, as a treatment modality should be presented in a realistic, unbiased, and acceptable manner as one of the options to patients making decisions about interventions or procedures. The probable result of this clinical pathway should be objectively explained with comparison of outcomes with other interventions when this information is known.

### **Part III - Care of the Cyanotic Patient**

Erythrocytosis is expected in cyanotic patients. Phlebotomy should be considered in any patient with erythrocytosis and hyperviscosity symptoms in the absence of dehydration. A phlebotomy of no more than 500 c.c. is usual in adults, combined with volume replacement - especially in pulmonary hypertensive patients. It should be performed in hospital. The purpose of the phlebotomy is to relieve hyperviscosity symptoms.

“Desired hematocrit” levels are rarely less than 65%, unless the patient has hyperviscosity symptoms at this level, in which case, a level of 60% is chosen. In the rare situation of hematocrits at or above 75%, serial phlebotomy to 65% over several days may be necessary. In general serial phlebotomies should be avoided because of the iron deficiency anemia which may result .

Iron deficiency anemia, even in the face of erythrocytosis, is undesirable because of the reduced oxygen carrying capacity and deformability of the red cells and an increased risk of stroke. Both the peripheral film and serum ferritin will confirm this diagnosis. Patients should be treated with the careful administration of oral iron supplements for one to four weeks with weekly monitoring of hemoglobin, hematocrit and reticulocyte count. There may be a rapid increase in hematocrit levels, which may require discontinuation of iron therapy.

Avoidance of iron deficiency is important. If iron deficiency is anticipated (e.g. periodic phlebotomies, GI or GU blood loss), iron supplements should be prescribed (under close supervision).

#### **Non-Cardiac Sequelae of Erythrocytosis**

Abnormal hemostasis, including reduced platelet count, abnormal platelet function, decreased von Willebrand factor (multimeric forms), and other factors in the blood, is frequent in cyanotic patients. It may be manifest through easy bruising, frequent and recurrent epistaxis, gingival bleeding, or traumatic bleeding (including peri-operative bleeding). Spontaneous bleeding is rare but spontaneous intravascular thrombosis may occur. Desmopressin is usually effective for epistaxis, hemoptysis, and peri-operative bleeding (including post-cardiac bypass) but may have undesirable renal side effects.

Hyperuricemia is common in cyanotic patients. Rarely does it result in uric acid kidney stones. Hyperuricemia with recurrent gouty arthritis is best managed with allopurinol or combined therapy.

Glomerular sclerosis, secondary to cyanosis, is usually manifest initially as proteinuria. This may cause problems with radiopaque contrast media and dehydration leading to uremia, oliguria and even anuria. Thus patients should be hydrated prior to procedures involving contrast media.

Hypertrophic osteoarthropathy accompanied by arthralgia may require analgesia.

The frequency of calcium bilirubinate gallstones and consequent cholecystitis is increased in patients with erythrocytosis, even in children and adolescents.



## Part IV - Arrhythmias in adult congenital heart disease

Rhythm disturbances and conduction defects are important problems in adults with congenital heart disease.

Causes for arrhythmias are:

- abnormal anatomy
- disturbed hemodynamics
- intracardiac surgery

A subdivision can be made in:

- supraventricular rhythm disturbances
- ventricular rhythm disturbances
- conduction defects

The aim should be to keep these patients in sinus rhythm as long as possible.

### Diagnostic recommendations

An adequate diagnostic work up for patients with arrhythmias contains:

- a thorough anamnesis and physical examination
- accurate and complete file of the anatomic defect, including surgical reports
- electrocardiogram, on indication 24-hour holter monitoring
- echocardiogram, and/or MRI for functional assessment
- exercise test
- laboratory test (electrolytes)

Invasive tests like electrophysiological studies and angiography are only advised on indication.

### Treatment

Before medical treatment of a patient is started, the underlying hemodynamic situation must be optimised with medication, or if necessary surgery.

Electrolyte disturbances should be corrected.

Furthermore, the following items should be checked before starting any medication:

1. ventricular function (anti-arrhythmic agents are often negative inotropes)
2. the risk of sinus node dysfunction and conduction defects which can be aggravated by the medication
3. the risk of pro-arrhythmic effects

### Supraventricular arrhythmias

Supraventricular arrhythmias (atrial fibrillation, atrial flutter and atrial tachycardia) are often seen in patients with congenital heart disease especially in patients with enlargement of the atria or scars in the atria. Examples are patients with an atrial septal defect, Morbus Ebstein or with Fontan or Mustard operation. Atrial arrhythmias contribute substantially to the morbidity but also to the mortality of these patients. Active treatment of the supraventricular arrhythmias is warranted beginning with the correction of underlying abnormalities (hemodynamics, electrolytes etc.).

If chronic atrial fibrillation develops anticoagulation therapy is indicated.

After electrical cardioversion sometimes long sinus arrests are seen and temporary pacing

may be necessary. Be sure an open route to the right ventricle is available for eventual pacing.

Patients who have evidence of mild sinus node dysfunction or atrioventricular conduction impairment may require antiarrhythmic therapy to prevent tachyarrhythmias as after a Senning or Mustard repair. In these patients the antiarrhythmic therapy should be started in a monitored setting where prompt withdrawal of the drug and institution of supportive care can be provided if bradyarrhythmias emerge. Implantation of a permanent pacemaker may be required to continue the antiarrhythmic therapy. In patients with the so-called "brady-tachy syndrome", pacing of the atrium can lower the incidence of tachyarrhythmias.

### **Ventricular arrhythmias**

Ventricular arrhythmias may be present without symptoms but have a negative influence on the prognosis. A high risk group for sudden death can be identified in patients after surgery of tetralogy of Fallot who have a widened QRS complex on the ECG, abnormal hemodynamics (high right ventricular systolic pressure and right ventricular dilatation), or ventricular arrhythmias during exercise test. In this high risk subgroup prophylactic treatment might be justified, although in general, treatment of non-sustained ventricular arrhythmias does not improve survival. Ref . 106

Patients who present with sustained ventricular tachycardia or syncope or who are resuscitated from ventricular fibrillation require extensive evaluation of their hemodynamic status. The possibilities of either medical anti-arrhythmic therapy, catheter ablation, implantation of an internal cardiac defibrillator (ICD) or surgical revision with potential excision of the ventricular tachycardia focus should be evaluated.

Anti-arrhythmic drug therapy in these patients should be initiated in the hospital.

### **Third degree AV block**

#### Congenital

The natural history of congenital complete heart block shows that long existing bradycardia can induce progressive left ventricular dilatation.

Most patients require permanent implantable pacemakers before age 50.

Generally accepted indications for pacemaker implantation in these patients are:

1. symptomatic AV block, including symptoms caused by left ventricular dysfunction
2. escape rhythm with widened QRS complex or QTc segment of more than 450 msec.
3. when bradycardia-induced ventricular arrhythmias occur.

In all other adult patients with congenital complete heart block there may be an indication for pacemaker implantation, especially when the exercise test shows a reduced exercise capacity. Ref . 107

#### Post surgery

Permanent pacemaker implantation is required in 2 to 4 percent of patients who undergo intracardiac repair of congenital cardiac malformations. In the early postoperative period, the most common indication for pacemaker implantation is surgically induced heart block that persists longer than 2 weeks after surgery. If a third degree AV block persists a pacemaker should be implanted, even if an adequate escape rhythm is present. If postoperative AV block is temporary present a third degree AV block may develop in the future.

If symptomatic bradyarrhythmias exist prior to surgical repair and an endocardial pacing approach is difficult or impossible, epicardial leads should be implanted at the time of surgical repair. Atrioventricular sequential pacing may improve cardiac output and sustain ventricular performance better than ventricular pacing alone.

## **SECTION II - ATRIAL SEPTAL DEFECT**

## Part I - Background Information

Atrial septal defect (ASD) includes the following types: ostium secundum, sinus venosus and coronary sinus. Ostium primum [partial atrioventricular septal defect (AVSD)] is discussed in Section IV.

A “significant” ASD:

- Causes volume and sometimes pressure overload.
- May cause exercise limitation.
- May be associated with atrial arrhythmias.
- May cause late right heart failure.
- May permit paradoxical embolism resulting in TIA/CVA.

## Part II - Unoperated History and Management

Most patients with “significant” ASDs (see above) will eventually develop symptoms, although the timing of symptom development is unpredictable and may be after the 5<sup>th</sup> decade.

The most common symptoms are exercise intolerance (dyspnea and fatigue) and symptomatic supraventricular arrhythmias (usually atrial fibrillation, less often atrial flutter or sick sinus syndrome).

If atrial fibrillation occurs, both anticoagulants and antiarrhythmic therapy are usually indicated.

**Grade: A**

**Level: I**

**Refs: 1**

Preoperative arrhythmias often persist after surgery, but are associated with fewer symptoms and less disability after the ASD has been closed.

*Transvenous pacing should be avoided where possible in patients with ASDs, since paradoxical emboli may occur. For the same reason venous thromboemboli from any source are a potential hazard. If found, anticoagulation is recommended and ASD closure should be considered.*

**Grade: C**

**Level: V**

**Refs: 2,3**

Any condition causing reduced left ventricular compliance (e.g. left ventricular hypertrophy due to hypertension, cardiomyopathy or myocardial infarction) will tend to increase the left-to-right shunt through an ASD. Their prevention and/or early treatment should be attempted.

Similarly, mitral stenosis and/or regurgitation will increase the left-to-right shunt through an ASD. The combination is usually poorly tolerated clinically.

ASD with Eisenmenger syndrome may have a better prognosis than VSD or PDA with Eisenmenger syndrome. *Poor prognostic features are felt to be syncope, heart failure and hemoptysis.*

*Phlebotomy with volume replacement is indicated in Eisenmenger syndrome if the patient is **symptomatic** from erythrocytosis. See general recommendations part III*

**Grade: C**

**Level: V**

**Refs: 4**

*Pregnancy is contraindicated in Eisenmenger syndrome because of the high maternal (up to 50%) and fetal (up to 60%) mortality.*

**Grade: C**

**Level: V**

**Refs: 5**

### **Part III - Diagnostic Recommendations**

*An adequate diagnostic workup:*

- Documents the presence and type of ASD.
- Determines the size (defect diameter) of the defect.
- Determines the functional importance of the defect by:
  - left-to-right shunt estimate (if no evidence of volume overload).
  - right ventricular size, function and volume overload.
  - pulmonary artery pressures and resistance.
- Identifies other associated conditions that may influence management (anomalous pulmonary venous return, common in the presence of a sinus venosus defect; significant valve disease; or coronary artery disease).

*The diagnostic workup should include at a minimum:*

- *A thorough clinical evaluation.*
- *ECG.*
- *Chest X-ray.*
- *Echo-Doppler evaluation by a skilled individual.*
- *Resting oxygen saturation.*

*The diagnostic workup may require:*

- *Transesophageal (TEE) echo/Doppler examination to prove the existence of an ASD or better define its/their location(s) and size(s); to assess pulmonary venous return; to evaluate the cardiac valves, if this information is not provided by transthoracic echocardiography (TTE).*
- *A complete heart catheterization (for the reasons listed for TEE above plus determination of pulmonary artery pressures and resistances if this is of concern; to assess right-to-left shunting).*
- *Coronary angiography if there is suspicion of coronary artery disease or in patients over the age of 40 years and surgical repair is planned.*
- *Magnetic resonance imaging (MRI) occasionally to prove the existence of an ASD or to assess pulmonary venous return if doubts remain after other imaging modalities.*
- *An oxygen saturation with exercise if there is any suggestion of pulmonary hypertension. If there is severe pulmonary hypertension or resting desaturation of <85%, the patient should not be exercised.*
- *Open lung biopsy should only be considered when the reversibility of the pulmonary hypertension is uncertain from the hemodynamic data. It is potentially hazardous and should be done only at centres with experience in CHD.*

### **Part IV - Indications for Intervention**

Indications for closure are still being debated in the literature.

The following situations warrant intervention (usually operative closure):

- The mere presence of a “significant” ASD. [In older patients (>60 years of age), absence of symptoms does not preclude closure if there is a significant shunt (>2:1)].
- *If pulmonary hypertension is present [pulmonary artery pressure (PAP)>2/3 systemic arterial blood pressure (SABP), or pulmonary arteriolar resistance > 2/3 systemic arteriolar resistance], there must be a net left-to-right shunt of at least 1.5:1; or evidence of pulmonary artery reactivity when challenged with a pulmonary vasodilator (e.g. oxygen or nitric oxide); or lung biopsy evidence that pulmonary arterial changes are potentially reversible (Heath Edwards grade II or less).*
- *Possibly:* a cryptogenic cerebrovascular event in the presence of a small ASD or PFO and right-to-left shunting demonstrated on contrast echo.

**Grade: C**

**Level: III**

**Refs: 6-10**

## Part V - Surgical/Interventional Technical Options

For secundum ASD without pulmonary hypertension, surgical closure should result in little or no operative mortality.

Patients with a sinus venosus ASD (with or without partial anomalous pulmonary venous connection) or ostium primum ASD should be repaired by surgeons with an interest in and experience with CHD.

**Grade: C**

**Level: V**

**Refs: 11,12**

The availability of an inframammary or thoracotomy approach to a typical secundum ASD should be made known to potentially interested patients (especially young women) although the morbidity may be higher.

The use of devices to close ASDs percutaneously is experimental at present, and available only to patients with secundum ASDs. There are limits to the size of the defect which may be closed by device (currently 18-20 mm) and a rim must be present around the defect. The relative risks and long-term results cannot yet be compared to surgical results.

**Grade: C**

**Level: V**

**Refs: 13,14,108**

Notwithstanding, device closure may be attractive to a patient wishing to avoid the consequences of surgery (general anesthesia, pain, disfigurement), or to a patient felt not to be a good surgical candidate. Its ultimate role is still evolving.

Device closure of ASDs should only be performed in centers and by individuals with a commitment to the technique and to its clinical evaluation.

**Grade: Consensus**

## Part VI - Surgical/Interventional Outcomes

Following surgical repair, pre-operative symptoms, if any, decrease or abate. Pre-existing atrial fibrillation often persists but is better tolerated and is managed with standard medical therapy. Late atrial fibrillation may occur in up to 1/3 of patients.

Patients with persistent atrial fibrillation should be anticoagulated post-operatively.

**Grade: A**

**Level: I**

**Refs: 1**

Patients with pulmonary vascular disease should be anticoagulated post-operatively.

**Grade: Consensus**

Post-operative pericardial tamponade should be sought by clinical examination and confirmed by echocardiogram if suspected. It may occur up to several weeks after surgery.

**Grade: Consensus**

Left ventricular failure may occur in patients with associated cardiovascular disease (e.g. coronary artery disease, hypertension)

## **Part VII - Follow Up**

The following patients require periodic follow up by a cardiologist:

- Those repaired as adults.
- Elevated pulmonary artery pressures at the time of surgery.
- Atrial arrhythmias pre- or post-operatively.
- Ventricular dysfunction pre-operatively.
- Co-existing heart disease (e.g. coronary artery disease, valvular heart disease, hypertension).

**Grade: Consensus**

## **SECTION III - VENTRICULAR SEPTAL DEFECT**

### **Part I - Background Information**

#### **Hemodynamic Severity Grading of Isolated Ventricular Septal Defects (VSDs) in Adults**

|              |   |
|--------------|---|
| Small:       | Pulmonary/aortic systolic pressure ratio < 0.3, and pulmonary/systemic flow ratio (Qp/Qs) < 1.4 |
| Moderate:    | Systolic pressure ratio > 0.3, and Qp/Qs 1.4 to 2.2   |
| Large:       | Systolic pressure ratio > 0.3, and Qp/Qs > 2.2  |
| Eisenmenger: | Systolic pressure ratio > 0.9, and Qp/Qs < 1.5  |

#### **Surgical Classification of Isolated VSD in Adults**

|                  |   |
|------------------|---|
| Restrictive:     | RV pressure < LV pressure in the absence of right ventricular outflow tract obstruction.                  |
| Non-restrictive: | Equal right and left ventricular pressures in the absence of right ventricular outflow tract obstruction. |

#### **Clinical Severity Grading of Isolated VSDs in Adults**

|           |  |
|-----------|--|
| Small:    | Causes negligible hemodynamic changes. LV size is usually normal without any pulmonary hypertension.   |
| Moderate: | Causes enlargement of left ventricle and usually some pulmonary hypertension (reversible).   |
| Large:    | Results in pulmonary vascular obstructive disease and Eisenmenger physiology unless there is coexistent right ventricular outflow tract obstruction. |

#### **Pathologic Classification**

|             |                                     |
|-------------|-------------------------------------|
| Inlet:      | Perimembranous or muscular.         |
| Trabecular: | Muscular - anterior, mid or apical. |
| Outlet:     | Doubly-committed sub-arterial.      |
| Multiple:   | combination of the above.           |

Only isolated VSDs will be considered.

VSDs may co-exist with other cardiac lesions (especially valvar or subvalvar pulmonary stenosis) or result in secondary infundibular hypertrophy and right ventricular outflow obstruction.

### **Part II - Unoperated History and Management**

Small VSDs are associated with a relatively high risk of endocarditis and otherwise normal life expectancy. Spontaneous closure can still occur occasionally in adult life. Moderate VSDs are unusual in the adult but may occur when a prolapsing aortic valve cusp partially obstructs the defect. They are associated with the development of left heart dilation and shunt-related pulmonary hypertension (which often reverses with correction of the defect), and resultant congestive heart failure and atrial fibrillation, as well as the risk of endocarditis.

Large VSDs without pulmonary hypertension exist in adults only when associated with obstruction to right ventricular outflow and are rare. All such patients are at risk for endocarditis. Some are cyanotic because of more severe pulmonary stenosis.

VSD with Eisenmenger syndrome may have a worse prognosis than ASD with Eisenmenger syndrome although survival to old age is possible. *Poor prognostic features are felt to be syncope, heart failure and hemoptysis.*

*Phlebotomy with volume replacement is recommended in Eisenmenger syndrome if the patient is **symptomatic** from erythrocytosis. See general recommendations part III*

**Grade: C**

**Level: V**

**Refs: 4**

*Pregnancy is contraindicated in Eisenmenger syndrome because of high maternal (up to 50%) and fetal (up to 60%) mortality.*

**Grade: C**

**Level: V**

**Refs: 5**

5% of VSDs develop aortic valve regurgitation. Patients with doubly-committed sub-arterial VSDs are more likely to develop aortic regurgitation from progressive prolapse of the aortic valve cusps compared to those with a perimembranous VSD.

*Transvenous pacing should be avoided where possible in all patients with VSDs since paradoxical emboli may occur. For the same reason, venous thromboemboli from any source are a potential hazard. If found, anticoagulation is recommended.*

**Grade: C**

**Level: V**

**Refs: 2**

### **Part III - Diagnostic Recommendations**

An adequate diagnostic workup:

- Documents the number and type of VSD.
- Determines the size (restrictive/non-restrictive and/or pulmonary-to-systemic flow ratio) and functional importance (left-to-right shunt estimate; assesses left and right ventricular size/function and volume and pressure overload; pulmonary artery pressure and resistance) of the defect.
- Identifies other associated conditions that may influence management (aortic regurgitation; sub-aortic stenosis; right ventricular outflow obstruction; significant valve disease; or coronary artery disease).

*The diagnostic workup should include at a minimum:*

- *A thorough clinical evaluation.*
- *ECG.*
- *Chest X-ray.*
- *Echo-Doppler evaluation by a skilled individual.*

*The diagnostic workup may require:*

- Oximetry
- A complete heart catheterization (to determine pulmonary artery pressures and resistances [ $\pm$  reversibility using oxygen or nitric oxide]; to assess intracardiac shunting; to evaluate associated lesions, particularly if aortic regurgitation is present; to exclude multiple VSDs).
- *Coronary angiography if there is suspicion of coronary artery disease or in patients over the age of 40 years if a surgical repair is planned.*
- *Open lung biopsy should only be considered when the reversibility of the pulmonary hypertension is uncertain from the hemodynamic data. It is potentially hazardous and should be done only at centres with experience in CHD.*

#### Part IV - Indications for Intervention

The following situations warrant operative closure:

- The presence of a “significant” VSD [symptomatic; Qp/Qs of 2/1; elevated pulmonary artery pressures (pulmonary artery systolic pressure >50 mm Hg)]; deteriorating ventricular function due to volume (LV) or pressure (RV) overload; a cardiothoracic (CT) ratio greater than 50%.
- *In the presence of severe pulmonary hypertension (PAP>2/3 SABP or pulmonary arteriolar resistance greater than 2/3 systemic arteriolar resistance), there must be a net left-to-right shunt of at least 1.5:1; or evidence of pulmonary artery reactivity when challenged with a pulmonary vasodilator (e.g. oxygen, nitric oxide); or lung biopsy evidence that pulmonary arterial changes are potentially reversible (Heath Edwards grade II or less).*
- Significant right ventricular outflow tract obstruction.
- An inlet or outlet VSD with more than mild aortic incompetence.
- An outlet VSD together with aortic valve prolapse. (An inlet VSD with aortic valve prolapse can be observed unless there is deterioration.)

|                 |                  |                    |
|-----------------|------------------|--------------------|
| <b>Grade: C</b> | <b>Level: IV</b> | <b>Refs: 15-20</b> |
|-----------------|------------------|--------------------|

Endocarditis (especially recurrent) **may** be an indication for operative closure.

#### Part V - Surgical/Interventional Technical Options

Device closure of VSDs is an experimental procedure.

Patients with an isolated VSD with or without associated lesions (right ventricular outflow tract obstruction, aortic valve prolapse, sub-aortic stenosis or infective endocarditis) should be repaired by surgeons with an interest in and experience with CHD.

|                 |                 |                    |
|-----------------|-----------------|--------------------|
| <b>Grade: C</b> | <b>Level: V</b> | <b>Refs: 11,12</b> |
|-----------------|-----------------|--------------------|

#### Part VI - Surgical/Interventional Outcomes

Successful closure is associated with excellent survival if ventricular function is normal, although late ventricular arrhythmias are a potential risk especially in patients repaired late in life.

Elevated pulmonary artery pressures preoperatively may progress, regress or remain unchanged post-operatively.

#### Part VII - Follow Up

Patients with the following problems require periodic cardiac evaluation:

- Patch leaks or residual VSDs (which seldom require reoperation).
- Elevated PA pressures at the time of surgery.
- Aortic valve surgery.
- Late repair of moderate or large defects.
- Significant atrial or ventricular arrhythmias.
- Associated cardiac lesions (e.g. right ventricular outflow tract obstruction or aortic regurgitation).

**Grade: Consensus**

## **SECTION IV - ATRIOVENTRICULAR SEPTAL DEFECT**

### **Part I - Background Information**

#### **Definition**

This term covers a spectrum caused by maldevelopment of the atrioventricular junction. There may be atrial septal defect (ostium primum ASD), ventricular septal defect, both atrial and ventricular septal defect or neither. The atrioventricular valves are fundamentally abnormal being derived from five leaflets (a right antero-superior leaflet, a right inferior leaflet, a superior bridging leaflet, an inferior bridging leaflet and a left mural leaflet). This may result in separate right and left AV valves (which are abnormal) or a common valve. The left AV valve is invariably abnormal, having a “cleft” at the conjunction of the superior and inferior bridging leaflets. (N.B. This is different from an isolated cleft mitral valve which is unrelated to AVSD.)

#### **Classification:**

**Complete AVSD:** There is a non-restrictive VSD. There is usually a primum ASD but rarely the atrial septum may be intact. There is a common AV orifice.

**Partial AVSD:** The ventricular septum is intact. There is almost always a primum ASD.  
Very rarely, there is no ASD and the only abnormality is the ‘cleft’ in the left AV (mitral) valve.

**Intermediate AVSD:** The spectrum between complete and partial AVSD. Often the VSD is restrictive. There is usually a primum ASD and the left AV (mitral) valve has a ‘cleft’.

Most complete forms will have been repaired in childhood and if presenting de novo as an adult will be associated with pulmonary vascular disease. Presentation of partial AVSD (ostium primum ASD) as an adult is not uncommon.

#### **Clinical Issues**

This depends on whether there is an ASD, VSD, or both, and upon the competence of the left AV (mitral) valve.

Clinical presentation may take several forms:

- The shunt, ranging from heart failure to pulmonary vascular disease.
- AV valve insufficiency leading to pulmonary congestion, hepatic congestion and peripheral edema, or both.
- Arrhythmias.
- Sub-aortic stenosis (which may develop or progress in up to 5% of patients after repair, particularly in patients with primum ASD and some complete defects, especially if the left AV (mitral) valve has been replaced).

AVSD may coexist with other lesions, both non-cardiac and cardiac. Down syndrome occurs in 35% of patients with AVSD. Most complete AVSDs occur in Down patients (>75%). Most partial AVSDs occur in non-Down patients (>90%). AVSD may occur in association with tetralogy of Fallot and other forms of complex CHD including univentricular hearts.

## Part II - Unoperated History and Management

Most patients with complete defects will have been repaired in infancy although some may have been palliated in the past with pulmonary artery bands and have variable degrees of pulmonary vascular obstructive disease.

Where surgical repair has not been possible because of unfavourable anatomy such as straddling AV valve, marked disproportion of ventricular size (so called “unbalanced AVSD”) or established pulmonary hypertension, care should be provided in a center for congenital cardiology.

**Grade: Consensus**

Regurgitation through the left AV (mitral) valve is common because of its abnormal morphology (the so-called ‘cleft’). It may worsen with time necessitating valve repair or replacement. Sub-aortic stenosis may or may not be present initially but may develop or progress.

Patients with established pulmonary vascular disease are considered in section XIII. Most patients will have left axis deviation on their ECG. First degree AV block is common and complete AV block occurs spontaneously or after repair (up to 3%). Sick sinus syndrome may also occur after repair and may lead to supraventricular tachycardia (SVT). Arrhythmias in the adult are not uncommon.

AVSD with Eisenmenger syndrome has a worse prognosis than ASD, VSD or PDA with Eisenmenger syndrome. *Poor prognostic features are felt to be syncope, heart failure and hemoptysis.*

*Phlebotomy with volume replacement is indicated in Eisenmenger syndrome if the patient is **symptomatic** from erythrocytosis. See general recommendations part III.*

**Grade: C**

**Level: V**

**Refs: 4**

*Pregnancy is contraindicated in Eisenmenger syndrome because of the high maternal (up to 50%) and fetal (up to 60%) mortality.*

**Grade: C**

**Level: V**

**Refs: 5**

*Transvenous pacing should be avoided if there are residual intra-atrial or intra-ventricular communications since paradoxical emboli may occur. For the same reason, venous thromboemboli from any source are a potential hazard. If found, anticoagulation and closure of the defects are recommended.*

**Grade: C**

**Level: V**

**Refs: 2**

## Part III - Diagnostic Recommendations

*An adequate diagnostic workup:*

- Documents the presence of each component of the AVSD and whether or not the ventricular chamber sizes are “balanced” (although this is usually a pediatric issue).
- Assesses the magnitude and direction of intracardiac shunting.
- Documents the pulmonary artery pressure.
  
- Documents abnormalities of the atrio-ventricular valves and their connections (straddling/over-riding) and assesses the severity of AV valve regurgitation, if any.

- Documents the presence/absence of sub-aortic stenosis. This may occasionally require provocative testing with isoproterenol although it may be impossible to document a gradient in the presence of a VSD.
- Identifies the presence of associated abnormalities (cardiac and non-cardiac) which may impact upon management (e.g. pulmonary hypertension, tetralogy of Fallot, PDA, other muscular VSDs, coarctation or Down syndrome).

*The diagnostic workup should include at minimum:*

- *A thorough clinical examination paying particular attention to AV valve regurgitation.*
- *ECG.*
- *Chest X-ray.*
- *Echo-Doppler evaluation by a skilled individual.*

*The diagnostic work-up may require:*

- TEE to determine the exact anatomy (if unclear after TTE); the presence of intracardiac shunts; chordal attachments; the presence and severity of left AV (mitral) valve regurgitation (or stenosis if previous valve repair has been undertaken); the presence and severity of right AV valve regurgitation and sub-aortic stenosis.
- A complete heart catheterization to determine: the presence and magnitude of intracardiac shunts; pulmonary artery pressures and resistances; the severity of pulmonary vascular disease; the presence and severity of left AV (mitral) valve regurgitation (or stenosis, if previous valve repair has been undertaken); the presence and severity of sub-aortic stenosis (provocative testing may be necessary).
- *Coronary angiography if there is suspicion of coronary artery disease or in patients over the age of 40 years if a surgical repair is planned.*
- *Open lung biopsy should only be considered when the reversibility of the pulmonary hypertension is uncertain from the hemodynamic data. It is potentially hazardous and should be done only at centres with experience in CHD.*
- Holter monitoring to assess AV block.

## **Part IV - Indications for Intervention**

The following situations warrant intervention:

- The unoperated or newly diagnosed AVSD with significant hemodynamic defect (manifest by atrial arrhythmias, impaired ventricular function, right ventricular volume overload in primum ASD [i.e. the same criteria as for secundum ASD]), attributable symptoms or reversible pulmonary hypertension.
- Persisting or new hemodynamically significant defects arising after the original repair.
- Left AV (mitral) valve regurgitation (or stenosis from previous repair) causing symptoms, atrial arrhythmia or deterioration in ventricular function.
- Significant sub-aortic obstruction (gradient of 50mm Hg at rest or on provocative testing with isoproterenol).

**Grade: C**

**Level: V**

**Refs: 21-23**

## **Part V - Surgical Technical Options**

AVSD, including ostium primum ASD, left AV (mitral) valve repair, sub-aortic stenosis or residual defects should be operated on by surgical teams with a commitment to and experience with CHD most commonly in a center for congenital cardiology.

**Grade: C**

**Level: V**

**Refs: 11,12,109**

When mitral valve repair is not possible, mitral valve replacement may be necessary. It should have a similar operative risk as routine mitral valve replacement although the risk of complete AV block may be higher. It may cause left ventricular outflow tract obstruction, especially if a bioprosthetic valve is used.

### **Part VI - Surgical Outcomes**

In the short term, the results of repair of partial AVSD are similar to those following closure of secundum ASD, but sequelae of left AV (mitral) valve regurgitation, sub-aortic stenosis and AV block may develop or progress.

Repair of the abnormal left AV (mitral) valve may result in a stenotic valve.

The long term results of repair of complete AVSD are not well known but similar problems as with partial AVSD are likely.

### **Part VII - Follow Up**

All patients require periodic follow up by a cardiologist because of the possibility of progressive AV valve regurgitation (or stenosis); the development of sub-aortic stenosis; the development of significant atrial arrhythmias, or progression of the commonly present 1<sup>st</sup> degree AV block.

Particular attention should be paid to those with pulmonary hypertension pre-operatively.

**Grade: Consensus**

**Refs: 110**

## SECTION V - PATENT DUCTUS ARTERIOSUS

### Part I - Background Information

#### Clinical Severity Grading of Patent Ductus Arteriosus (PDA) in adults

|              |   |
|--------------|---|
| Silent:      | Detected only by non-clinical means (usually echo).   |
| Small:       | Audible continuous murmur. Causes negligible hemodynamic change. Normal LV size without any pulmonary hypertension.   |
| Moderate:    | Audible continuous murmur. Wide pulse pressure (as in aortic regurgitation). Causes enlargement of the left ventricle and some pulmonary hypertension (usually reversible). |
| Large:       | Usually does not exist without Eisenmenger physiology. Diastolic murmur may be absent. Causes enlargement of the left ventricle and left atrium and pulmonary hypertension. |
| Eisenmenger: | Continuous murmur is absent. Causes substantial pulmonary hypertension and differential hypoxemia and often differential cyanosis.  |

PDA is usually an isolated lesion.

### Part II - Unoperated History and Management

The risk of endarteritis with silent PDA is unknown but is likely very low (only sporadic case reports exist). No intervention is indicated if a silent PDA is detected.

All other PDAs are associated with a risk of endarteritis (which may increase with increasing age).

Small PDAs have a normal life expectancy.

Moderate PDA is unusual in the adult. It is associated with the development of left heart dilation and shunt-related pulmonary hypertension (which often reverses with correction of the defect). The majority of patients are symptomatic from dyspnea or palpitations (atrial arrhythmias) although frank heart failure is unusual.

Large PDA is rare in the adult, most having been corrected in infancy and childhood.

Pulmonary hypertension is usual and may not reverse entirely with closure of the defect.

Most patients are symptomatic from dyspnea or palpitations. Aneurysm formation of the duct is an uncommon but important complication.

Eisenmenger PDA has a similar prognosis to Eisenmenger VSD although symptoms may be less marked and exercise tolerance better. *Poor prognostic features are felt to be syncope, heart failure and hemoptysis.*

*Phlebotomy with volume replacement is recommended in Eisenmenger syndrome if the patient is **symptomatic** from erythrocytosis. See general recommendations part III*

**Grade: C**

**Level: V**

**Refs: 4**

*Pregnancy is contraindicated in Eisenmenger syndrome because of the high maternal (up to 50%) and fetal (up to 60%) mortality.*

**Grade: C**

**Level: V**

**Refs: 5**

### Part III - Diagnostic Recommendations

*An adequate diagnostic workup:*

- Documents the presence of PDA.
- Determines the size (systemic-to-pulmonary shunt estimate) and functional importance (pulmonary artery pressures) of the defect. Shunt estimates may be inaccurate because of the difficulty in obtaining a representative pulmonary blood sample for saturation assessment.
- Identifies whether a ductal aneurysm is present.
- Identifies whether the duct is calcified if surgical repair is planned.

*The diagnostic work up should include at a minimum:*

- *A thorough clinical examination.*
- *ECG.*
- *Chest X-ray.*
- *Echo-Doppler evaluation by a skilled individual.*
- *Oximetry.*

The diagnostic workup may require:

- A complete heart catheterization (to determine pulmonary artery pressures and resistances [ $\pm$  reversibility using oxygen or nitric oxide]).
- *Coronary angiography if there is suspicion of coronary artery disease or in patients over 40 years if a surgical repair is planned.*
- *Open lung biopsy should only be considered when the reversibility of the pulmonary hypertension is uncertain from the hemodynamic data. It is potentially hazardous and should be done only at centres with experience in CHD.*
- MRI or CT scan to define the anatomy.

#### **Part IV - Indications for Intervention**

The following situations warrant intervention:

- The presence of a PDA (except the silent duct at one extreme and the presence of severe, irreversible pulmonary vascular disease at the other extreme).
- The occurrence of an episode of endarteritis on a clinically silent PDA.
- *If pulmonary hypertension is present ( $PAP > 2/3$  SABP or pulmonary arteriolar resistance exceeds  $2/3$  systemic arteriolar resistance), there must be a net left-to-right shunt of at least 1.5:1, or evidence of pulmonary artery reactivity when challenged with a pulmonary vasodilator (e.g. oxygen, nitric oxide) or lung biopsy evidence that pulmonary arterial changes are potentially reversible (Heath Edwards grade II or less).*

**Grade: C**

**Level: V**

**Refs: 16,17,24,25**

#### **Part V - Surgical/Interventional Technical Options**

Device closure is the preferred method in centres with demonstrated experience (< 1% mortality) and when possible should be planned at the same time as the diagnostic catheterization. Antibiotic prophylaxis is continued for 6 months after a successful procedure.

**Grade: Consensus**

**Refs: 26,27**

Surgical closure should be reserved for those in whom the PDA is too large for device closure or if facilities for device closure are not available. Cardiopulmonary bypass may be necessary in the presence of aneurysm, prior endocarditis, elevated pulmonary artery pressure or calcification. Due to the increased risk of operative difficulties if the duct is calcified, operation should be undertaken by a surgeon with an interest in and training in ACHD.

**Grade: Consensus**

## **Part VI - Surgical/Interventional Outcomes**

More than 85% of ducts are closed by one year following device placement. In a small proportion of patients, a second or even a third device may need to be placed. This is usually deferred for at least 6 months. Recanalization is unusual but recognized.

More than 95% of ducts are closed by surgery. Recanalization is unusual but recognized. In patients with a residual shunt (detected clinically or by echocardiography), antibiotic prophylaxis is continued until 6 months after the shunt is eliminated.

## **Part VII - Follow Up**

Patients should have periodic cardiac examination because recanalization can occur or residual defects may be present. Patients with devices in situ should be followed because the natural history of these devices is unknown.

Patients with a silent PDA do not require follow up.

**Grade: Consensus**

**Refs: 111**

## **SECTION VI - LEFT VENTRICULAR OUTFLOW TRACT OBSTRUCTION & BICUSPID AORTIC VALVE**

### **Part I - Background Information**

#### **Definition**

The information refers to concordant atrio-ventricular and ventriculo-arterial connections. *Note that neither hypertrophic cardiomyopathy nor interrupted aortic arch are considered here.*

Left ventricular outflow tract obstruction (LVOTO) can occur at several levels:

- Supravalvar LVOTO seldom occurs in isolation: it is usually part of Williams syndrome, but may be familial with normal facies, or associated with rubella syndrome. It is rarely localised. It is usually diffuse, involving the major arteries to varying degrees and begins at the superior margin of the sinuses of Valsalva.
- Valvar LVOTO in the adult patient with CHD is usually due to bicuspid aortic valve (rheumatic and senile calcific aortic stenosis are excluded here). Bicuspid aortic valve is the most common congenital cardiac anomaly occurring in 1-2% of the population with a male preponderance (4:1). It usually occurs in isolation but is associated with other abnormalities in 20%, the most common being coarctation of the aorta (which should be sought) and PDA.
- Subvalvar LVOTO is usually a discrete fibromuscular ridge partially or completely encircling the left ventricular outflow tract or a long fibromuscular narrowing beneath the base of the aortic valve. Occasionally, there is a tunnel-like narrowing of the whole left ventricular outflow tract with a small aortic root. There is a male predominance (2:1). Family members may be affected. Rarely, abnormal insertion of the mitral valve or accessory mitral leaflet may cause significant obstruction.

The concurrence of subvalvar LVOTO, coarctation and mitral stenosis (parachute mitral valve and supramitral ring) is known as Shone syndrome.

### **Part II - Unoperated History and Management**

Supravalvar LVOTO is usually progressive and aortic regurgitation is common. With Williams syndrome, there are often associated peripheral pulmonary artery or systemic arterial (including coronary ostial) stenoses, which may worsen, resolve or remain unchanged. There is often systemic hypertension.

Valvar LVOTO commonly progresses as the patient grows but the rate is variable. Most patients with bicuspid aortic valve will not experience any problems, although there is the lifelong risk of endocarditis. Some however, may develop aortic stenosis (especially after calcification of the valve in the sixth decade), aortic regurgitation, aortic dissection or aneurysmal aortic root dilation. If there is associated coarctation, this should usually be dealt with first (unless there is critical LVOTO: if both are severe, they may be dealt with at one operation).

Subvalvar LVOTO usually progresses. It is often associated with aortic regurgitation through an otherwise normal valve which has been damaged by the subvalvar jet of blood. It may also progress but is seldom more than moderate. There may be associated small VSDs. Tunnel-like subvalvar LVOTO is progressive and requires surgery for relief of obstruction although this may be technically difficult because the aortic root is small. Subvalvar LVOTO may occur with a variety of associated lesions.

### Part III - Diagnostic Recommendations

*An adequate diagnostic workup:*

- Documents the level(s) of obstruction.
- Quantitates the severity and anatomy of the obstruction(s).
- Identifies associated abnormalities including aortic regurgitation, proximal aortic dilation, coarctation and the associated anomalies of Williams and Shone syndromes.

*The diagnostic workup should include at a minimum:*

- A thorough clinical examination.
- ECG.
- Chest X-ray.
- Echo-Doppler examination by a skilled individual to determine the level of obstruction, septal thickness, size of the aortic root.

*The diagnostic workup may require:*

- TEE to define precisely the anatomy if unclear from TTE.
- Exercise testing.
- A complete heart catheterization ± provocative testing to assess the hemodynamics and severity of obstruction.
- Coronary angiography and aortography if surgery is being planned.
- MRI to assess associated lesions such as pulmonary artery stenoses or coarctation.
- Abdominal aortography to identify significant renal or other arterial stenoses.

### Part IV - Indications for Intervention/Re-intervention

Supravalvar LVOTO requires intervention for a gradient of >50 mm Hg if the obstruction is discrete. Criteria for intervention for diffuse obstruction are not well defined but are probably similar since the end effect on the coronary arteries and the myocardium are the same.

**Grade: C**

**Level: V**

**Refs: 28**

Valvar LVOTO requires intervention for symptoms (dyspnea, angina, arrhythmia, presyncope or syncope) or "critical" aortic stenosis (valve area <0.6cm<sup>2</sup>). Intervention may be indicated occasionally for other reasons (e.g. a person with a lesser degree of obstruction who wishes to play vigorous sports or wishes to become pregnant).

**Grade: C**

**Level: IV**

**Refs: 29-33,112**

Subvalvar LVOTO requires intervention for a resting gradient >50 mm Hg, symptoms or if aortic regurgitation is moderate-severe or progressive. If there is an associated VSD, the gradient may be underestimated and important subvalvar LVOTO may become manifest after VSD closure.

**Grade: C**

**Level: IV**

**Refs: 34-36**

Bicuspid aortic valve requires intervention for stenosis (as above), moderate or severe regurgitation associated with deteriorating ventricular function or a dilating ventricle; proximal aortic dilation (>55 mm); or aortic dissection.

**Grade: C**

**Level: IV**

**Refs: 29,37,38**

Re-intervention is indicated after valvotomy or after surgery for:

- Recurrent LVOTO (same criteria as above).
- Severe aortic regurgitation.
- Combined restenosis with moderate or greater regurgitation.

**Grade: C**

**Level: IV**

**Refs: 30-33,37**

## Part V - Surgical/Interventional Options

Patients who require operation for supra- or subvalvar LVOTO should be operated on by surgical teams with a commitment to and experience with CHD.

**Grade: C**

**Level: V**

**Refs: 11,12**

Valvar LVOTO may be treated with balloon valvuloplasty (if the valve is non-calcified); open aortic valvotomy; or valve replacement using a mechanical valve, biological valve or pulmonary autograft. The choice depends on the availability and skills of the team involved and preference of the patient.

Balloon valvuloplasty for valvar LVOTO is an experimental technique and should be performed in centres and by people with experience in and a commitment to evaluation of this method. There may be a role for compassionate or emergency use (e.g. a pregnant female with severe aortic stenosis).

**Grade: Consensus**

Supra- or subvalvar LVOTO requires patch aortoplasty or, rarely, replacement of the proximal ascending aorta.

Discrete subvalvar LVOTO requires surgical resection almost invariably associated with myomectomy. In older patients, the aortic valve may also need to be replaced or repaired because of significant aortic regurgitation.

Tunnel-like subvalvar LVOTO may require augmentation of the LVOTO using the Konno procedure or other modifications for enlargement of the outflow tract. Rarely a left ventricular apex-to-aorta valved conduit was implanted if it was impossible to relieve the LVOTO adequately by any other means but the long term durability was unacceptable and the procedure has been abandoned.

Subvalvar LVOTO associated with repair of AVSD often recurs if the fibromuscular tissue alone is excised. Patch enlargement of the infundibular septum and patch enlargement of the superior bridging leaflet of the left AV (mitral) valve or left AV (mitral) valve replacement may be required.

## Part VI - Surgical/Interventional Outcomes

Supravalvar LVOTO should have an operative mortality of <2%. Recurrence of obstruction should be very low. The long term durability of the patches or conduits used to relieve the obstruction may be a problem and surveillance should include assessment for aneurysm and endocarditis.

Valvar LVOTO treated by valvotomy or valvuloplasty is associated with progressive recurrent stenosis and calcification and may require eventual valve replacement.

Patients who require valve replacement will have a similar course to those who have valve replacement for acquired valvar LVOTO.

Recurrence of fibromuscular subvalvar LVOTO is not uncommon (up to 20% over a decade, particularly if the aortic root is small).

Tunnel-like subvalvar LVOTO with extensive repair with or without aortic valve replacement has a high recurrence risk although newer techniques may improve this.

## Part VII - Follow up

All patients should have regular cardiology follow up. Some require follow up in a center for congenital cardiology e.g. patients with Williams or Shone syndrome, those with complex LVOTO with or without repair and those with pulmonary autografts.

Particular attention should be paid to:

- progressive/recurrent stenosis at any level.
- aortic regurgitation.
- ventricular function and/or dilation.
- aortic root dilation.
- right ventricle-pulmonary artery conduit stenosis (and coronary abnormalities) following a pulmonary autograft operation.

**Grade: Consensus**

## **SECTION VII - RIGHT VENTRICULAR OUTFLOW TRACT OBSTRUCTION**

### **Part I - Background Information**

Right ventricular outflow tract obstruction (RVOTO) can occur at any level.

Supravalvar RVOTO seldom occurs in isolation. It may occur in tetralogy of Fallot, Williams syndrome, Noonan syndrome, VSD or arteriohepatic dysplasia.

Valvar RVOTO, the most common form of RVOTO, is almost always congenital in origin. In 15% of cases, the valve is dysplastic as well as stenotic. In adults, the valve may calcify from the fourth decade onwards.

Subvalvar (infundibular) RVOTO usually occurs in combination with other lesions, particularly ventricular septal defect, and as part of tetralogy of Fallot.

RVOTO (either valvar or subvalvar) may occur in association with sub-aortic stenosis.

A separate but somewhat similar entity is “double-chambered right ventricle” with mid-cavity obstruction. This may be associated with a small VSD (see indications for intervention).

Branch pulmonary artery stenosis is not considered here.

### **Hemodynamic Severity Grading**

(Based on peak systolic pressure gradients at heart catheterization or echocardiography)

|                     |             |
|---------------------|-------------|
| Trivial:            | <25 mm Hg   |
| Mild:               | 25-49 mm Hg |
| Moderate:           | 50-79 mm Hg |
| Severe or Critical: | >80 mm Hg   |

### **Part II - Unoperated History and Management**

Patients with trivial and mild valvar RVOTO are asymptomatic, do not become worse with time as adults and do not require active treatment, apart from possible endocarditis prophylaxis.

**Grade: C**

**Level: IV**

**Refs: 39**

Female patients often present to physicians during pregnancy because of an increase in the loudness of the murmur. Others may present because of enlarged pulmonary arteries detected on chest X-ray.

Moderate valvar RVOTO may progress in 20% of unoperated patients especially in adults because of calcification of the valve. Some of these patients will also become symptomatic particularly in later life because of atrial arrhythmias and consequent tricuspid regurgitation. When the gradient is severe or the patient is symptomatic, valvotomy is recommended.

**Grade: C**

**Level: IV**

**Refs: 39**

Patients with severe valvar RVOTO should be referred for valvotomy (balloon or rarely surgical).

**Grade: C**

**Level: IV**

**Refs: 39**

Supravalvar RVOTO may progress in severity and should be monitored.

Subvalvar RVOTO progresses in severity and often leads to the development of worsening right ventricular hypertrophy, symptoms and critical gradients requiring surgical repair.

### Part III - Diagnostic Recommendations

*An adequate diagnostic workup:*

- Documents the level(s) of obstruction.
- Quantitates the severity of the obstruction(s).
- Identifies associated abnormalities such as ASD, PDA, VSD and tetralogy of Fallot.

*The diagnostic workup should include at a minimum:*

- *a thorough clinical examination*, paying particular attention to the 'a' wave on the venous pulse, the length of the murmur, the pulmonary component of the second sound and right ventricular hypertrophy.
- *ECG.*
- *Chest X-ray*, paying particular attention to valvar calcification on the lateral.
- *Echo Doppler examination by a skilled individual.*

*The diagnostic workup may require:*

- oximetry (rest and exercise) to determine if there is cyanosis because of associated abnormalities (ASD or VSD).
- a complete heart catheterization ( $\pm$  provocative testing with Isoproterenol) to assess the hemodynamics and severity of obstruction including assessment of possible sub-aortic stenosis or tricuspid stenosis.
- *coronary angiography if there is suspicion of coronary artery disease, or in patients over the age of 40 years in whom surgery is being planned.*
- MRI (very rarely) to assess associated lesions such as pulmonary artery stenoses and aortic coarctation.

### Part IV - Indications for Intervention/Re-intervention

Intervention is indicated if:

- The combined gradient across the RVOT is  $>50$  mm Hg at rest.
- Symptoms are present (exertional dyspnea, angina, presyncope or syncope).

**Grade: C**

**Level: IV**

**Refs: 39,40**

- There are important arrhythmias.
- There is associated ASD, especially if there is right-to-left shunting.
- There is a double chambered right ventricle with significant mid-cavity obstruction.

**Grade: Consensus**

Intervention may be indicated occasionally for other reasons (e.g. a person with a lesser degree of obstruction who wishes to play vigorous sports, scuba dive or become pregnant).

Intervention may be indicated following an episode of endocarditis.

Re-intervention is indicated for:

- Recurrent RVOTO after prior surgery or balloon valvotomy (same criteria as above).
- Severe pulmonic regurgitation associated with reduced exercise capacity of cardiovascular cause or evidence of deteriorating right ventricular function especially with the onset of tricuspid regurgitation.

**Grade: C**

**Level: V**

**Refs: 40,41,113**

## Part V - Surgical/Interventional Options

Patients who require operation for supra- or subvalvar RVOTO should be operated on by surgical teams with a commitment to and experience with CHD.

**Grade: C**

**Level:**

**Refs: 11,12**

Balloon valvuloplasty is the treatment of choice for valvar RVOTO but may be unsuccessful if the valve is calcified or dysplastic in which case surgical valvotomy, valvectomy or occasionally valve replacement may be necessary.

**Grade: C**

**Level: IV**

**Refs: 42,43**

Surgery is necessary if there is double chambered right ventricle with significant obstruction.

Balloon valvuloplasty for valvar RVOTO is an established technique but should still be performed generally in centres and by teams with experience in this technique.

**Grade: Consensus**

**Refs: 42,43**

## Part VI - Surgical/Interventional Outcomes

The long term results of surgical pulmonary valvotomy are established. Clinical outcomes are excellent. Relief of valvar RVOTO is usually maintained, but residual obstruction may progress. Occasionally pulmonary regurgitation may progress and become severe enough to warrant re-intervention. Long term survival in surgical patients when RVOTO occurs as an isolated lesion is close to normal. Long-term mortality is increased however with greater age (>21 years) at time of surgery.

Patients treated with balloon valvuloplasty, in the absence of a dysplastic valve, probably have the same prognosis as those who have had surgical valvotomy at least in the medium term.

Subvalvar and supra- or subvalvar RVOTO seldom recur after adequate repair.

## Part VII - Follow Up

Patients with trivial or mild RVOTO do not require specialist follow-up by a congenital cardiologist. Follow-up by a general physician and/or internist/cardiologist is sufficient.

Patients with moderate or greater RVOTO or moderate or severe pulmonary regurgitation require careful monitoring as intervention may be required. In most instances, this should be at a center for congenital cardiology.

Particular attention should be paid to:

- Progressive/recurrent stenosis, especially at the subvalve level.
- Right ventricular size and function in the context of significant pulmonary/subpulmonary stenosis and/or regurgitation.
- Tricuspid regurgitation (often reflecting right ventricular dysfunction).
- Atrial and occasionally ventricular (usually post-operative) arrhythmias.
- Evidence of intracardiac shunting, especially right-to-left.

**Grade: Consensus**

## **SECTION VIII - TETRALOGY OF FALLOT**

### **Part I - Background Information**

#### **Definition**

The defect is due to antero-cephalad deviation of the outlet septum resulting in: (1) an unrestricted sub-aortic VSD, (2) right ventricular outflow tract obstruction which may be infundibular, valvar or a combination of both, (3) an overriding aorta that has deviated to the right, and (4) right ventricular hypertrophy. Some patients also have supravalve or branch pulmonary artery stenosis.

The so-called pentalogy of Fallot has in addition an ASD or PFO.

### **Part II - Unoperated History and Management**

The pathophysiology varies depending on the degree of right ventricular outflow obstruction.

With mild obstruction, the presentation is of increased pulmonary blood flow and minimal cyanosis, so-called “pink tetralogy” or “acyanotic Fallot”. This occasionally presents in adulthood.

Most young patients, however, have significant RVOT obstruction with consequent right-to-left shunt. Most adults will have had surgery, either palliative or, more commonly, reparative.

Rarely, adults present who are unoperated. For them, surgical repair is still recommended since the results are gratifying and the operative risk is comparable to pediatric series (provided there is no serious co-existing morbidity).

Some patients reach adulthood with previous palliation only. The types of palliative procedures include:

- Blalock-Taussig shunt or modification (subclavian artery-to-pulmonary artery shunt).
- Waterston shunt (ascending aorta-to-right pulmonary artery).
- Potts shunt (descending aorta-to-left pulmonary artery).
- Central interposition tube graft.
- Infundibular resection (Brock procedure) or pulmonary valvotomy.
- RV-to-PA conduit without VSD closure or with fenestrated closure.

Reparative surgery involves closing the ventricular septal defect and relieving the RVOT obstruction. The latter may involve:

- Resection of infundibular muscle.
- Right ventricular outflow tract patch.
- Transannular patch (a patch across the pulmonary valve annulus which disrupts the integrity of the pulmonary valve and causes pulmonary regurgitation).
- An extracardiac conduit placed between the right ventricle and pulmonary artery.
- Replacement of the pulmonary valve.
- Excision of the pulmonary valve, especially if calcified.

A patent foramen ovale or secundum ASD needs closure if present.

Additional treatable lesions such as aortic regurgitation or muscular VSDs may also need to

be addressed.

### **Part III - Investigational Recommendations in Operated Patients**

Investigations are directed toward the postoperative sequelae and will vary according to the type of operation performed.

*All patients should have, at a minimum:*

- *A thorough clinical examination.*
- *ECG.*
- *Chest X-ray.*
- *Echo Doppler examination by a skilled individual to detect and quantify residual pulmonary stenosis and regurgitation, residual VSD, right and left ventricular function, aortic regurgitation, and aortic root size. The RVOT may be difficult to visualize.*
- *Exercise testing to assess functional capacity and to evaluate possible exertional arrhythmias.*
- *Holter monitoring.*

*and may require:*

- *Quantitative lung perfusion scan in patients with suspected pulmonary artery branch stenosis.*
- *Complete heart catheterization including coronary angiography if surgical re-intervention is planned or if adequate assessment of the hemodynamics is not obtainable by non-invasive means.*
- *EPS for those being evaluated because of major cardiac arrhythmias.*
- *MRI occasionally for some pulmonary artery or aortic anomalies.*

For those patients who have had previous palliation, assessment of pulmonary artery pressure and anatomy is mandatory at some point, since these shunts have inherent complications (distortion of the pulmonary arteries, development of pulmonary hypertension, and volume overloading of the left heart).

Patients presenting as adults who have not been repaired may have elevated pulmonary artery pressures despite severe RVOT obstruction.

### **Part IV - Indications for Re-intervention**

The following situations may warrant investigation following a palliative shunt:

- To determine whether complete repair is possible.
- Increasing cyanosis with erythrocytosis.
- Suspicion of pulmonary hypertension.
- Reduction or absence of the continuous shunt murmur (suspected shunt stenosis or occlusion).
- Aneurysm formation in the shunt.

Reoperation is only necessary in approximately 10% of patients following reparative surgery over a 20 year follow up.

The following situations may warrant intervention following repair:

- Residual VSD with a shunt >1.5:1.
- Residual pulmonary stenosis with RV pressure  $\geq$  2/3 of systemic pressure (either the native RV outflow or valved conduit if one is present).
- Free pulmonary regurgitation associated with progressive RV enlargement, progressive tricuspid regurgitation, arrhythmias, or symptoms such as deteriorating exercise performance.
- Significant aortic regurgitation associated with symptoms and/or progressive left ventricular dilation or deterioration of left ventricular function.
- Aortic root enlargement  $\geq$  55 mm in diameter.
- Rapid enlargement of an RV outflow tract aneurysm or evidence of infection or false aneurysm.
- The development of major cardiac arrhythmias, most commonly atrial flutter or fibrillation, or ventricular tachycardia which may reflect hemodynamic deterioration.
- The combination of residual VSD, residual pulmonary stenosis and regurgitation, all mild-moderate but leading to progressive RV enlargement, reduced RV function or symptoms.

**Grade: C**

**Level: V**

**Refs: 41,44-49,114-116**

## Part V - Surgical/Interventional Options

Patients who require intervention should be treated at a center for congenital cardiology by cardiologists and cardiac surgeons with appropriate expertise.

**Grade: C**

**Level: V**

**Refs: 11,12**

The following are possible intervention strategies:

- Surgery may be necessary for residual pulmonary stenosis; this may involve resection of residual infundibular stenosis or placement of an RV outflow or transannular patch. Occasionally a valved extracardiac conduit may be necessary.
- Aortic valve and/or root replacement may be necessary for those with aortic valve regurgitation and/or root dilation.
- Reoperation to insert a new pulmonary valve may be necessary for severe pulmonary regurgitation or a grossly calcified pulmonary valve. Tricuspid valve annuloplasty may also be necessary when at least moderate tricuspid regurgitation is present.
- Suture or patch closure of a residual VSD if the shunt is  $\geq$  1.5:1 or if the patient is undergoing reoperation for other reasons.
- Branch pulmonary artery stenosis may be managed with balloon dilation  $\pm$  stent insertion or surgery.
- Ablative therapy of arrhythmias, either atrial or ventricular.
- Closure of ASD or PFO if there is persistent cyanosis.

## Part VI - Surgical/Interventional Outcomes

The overall survival of patients who have had operative repair is excellent, provided the VSD has been closed and the RVOT obstruction has been relieved. A 25 year survival of >94% has been reported. Death may occur from reoperation or congestive heart failure. The reported incidence of sudden, presumed arrhythmic, death is approximately 3% which, although low, accounts for approximately one third of late deaths.

## Part VII - Follow Up

All patients should have regular cardiology follow-up by a cardiologist with an interest in and a dedication to the management of adult patients with CHD, preferably at a center for congenital cardiology.

**Grade: Consensus**

## **SECTION IX - COARCTATION OF THE AORTA**

### **Part I - Background Information**

Coarctation of the aorta is a stenosis usually but not always in the region of the ligamentum arteriosum. It may be discrete or associated with diffuse hypoplasia of the aortic arch and isthmus. The specific anatomy, severity and degree of hypoplasia proximal to the coarctation is highly variable and may involve the origin of the left subclavian artery.

'Simple' coarctation refers to coarctation in the absence of other intracardiac lesions. It is the most common form detected de novo in adults. "Complex' coarctation is used to describe coarctation in the presence of other important intracardiac anomalies (e.g. VSD, LVOTO, mitral stenosis) and is usually detected in infancy.

A significant coarctation is one with a gradient of more than 20 mm Hg across the coarctation site (at heart catheterization) in the absence of a well developed collateral circulation around the coarctation site. If there is extensive collateral circulation, there may be minimal or no pressure gradient and acquired aortic atresia.

A significant coarctation causes pressure elevation proximally with left ventricular hypertrophy and ultimately heart failure.

Premature coronary artery disease is common if the coarctation was not adequately repaired in childhood.

Associated abnormalities include:

- Bicuspid aortic valve (up to 85%).
- Intracranial aneurysms (most commonly of the circle of Willis).
- VSD.
- Acquired intercostal artery aneurysms.

Coarctation is common in XO Turner syndrome (35%).

### **Part II - Unoperated History and Management**

The mean survival of patients with untreated coarctation is 35 years with 75% mortality by 50 years of age. Most develop systemic hypertension, typically during childhood, and ultimately, by the 5th decade, left ventricular failure.

Death is usually due to:

- Aortic rupture/dissection.
- Cerebral hemorrhage.
- Infective endarteritis/endocarditis.
- Heart failure (usually beyond 30 years of age).
- Premature coronary artery disease.
- Concomitant aortic valve disease.

Symptoms may be absent and diagnosis should be suspected in any patient with upper limb hypertension, differential pulses in the upper and lower limbs or unexplained intracerebral hemorrhage. An occasional patient will be diagnosed from the typical X-ray appearances.

### **Part III - Diagnostic Recommendations**

*An adequate diagnostic workup documents:*

- The location and type of coarctation together with its severity.
- The presence or absence of significant collateral vasculature.
- Associated stenoses in other great vessels (carotids and subclavian arteries).
- Associated aneurysms, especially of intercostal arteries.
- Other cardiac lesions - notably aortic, mitral or sub-aortic abnormality and the status of left ventricular function.

*The diagnostic workup should include at a minimum:*

- a *thorough clinical examination* including upper and lower limb blood pressure measurement, palpation of femoral pulses and auscultation for collaterals around the scapula.
- *ECG.*
- *Chest X-ray.*
- *Echo Doppler evaluation by a skilled individual*, as the echo window for adults with coarctation may be elusive.
- Either MRI or angiography to delineate the coarctation (see below).

The diagnostic workup may require:

- Exercise test to detect exercise-induced hypertension and/or arm-leg pressure gradients.
- Invasive angiography with hemodynamic measurements to assess the gradient and nature of the obstruction and to determine the presence/absence of collaterals or aneurysm formation.
- MRI which provides good anatomical detail and may obviate the need for angiography.
- Digital subtraction angiography (DSA) which provides good anatomical detail and may obviate the need for invasive arteriography.
- Complete heart catheterization with aortography if associated cardiac disease (most commonly aortic valve disease) is present.
- Coronary angiography because of the risk of premature coronary artery disease if surgery is planned, if the patient is over 40 years, or if there is any evidence of left ventricular failure.

#### **Part IV - Indications for Intervention**

All patients with significant coarctation or re-coarctation, including those with long standing hypertension (regardless of age), whether symptomatic or asymptomatic, warrant intervention.

**Grade: C**

**Level: V**

**Refs: 50**

Patients with significant aortic valve stenosis may also require valve surgery which may or may not be done at the same time as coarctation repair. If operated upon separately, the sequence depends upon the severity of each of the lesions, the more severe being dealt with first.

Females contemplating pregnancy should have repair prior to pregnancy. The risk of complications (dissection/aneurysm rupture) during pregnancy is low. Management of hypertension in the unoperated pregnant patient may be problematic because too low a pressure below the coarctation site may result in abortion or death of the fetus.

**Grade: C**

**Level: V**

**Refs: 51-54**

## Part V - Surgical/Interventional Options

Surgical repair of coarctation in adults is more hazardous than in children. It should be performed by surgical teams with a commitment to and experience with CHD, preferably in a center for congenital cardiology. It may involve cardiopulmonary bypass.

**Grade: C**

**Level: V**

**Refs: 11,12**

For native or re-coarctation, surgical repair may involve:

- End-to-end anastomosis.
- Interposition graft.
- Arch augmentation.
- Jump graft by-passing the stenosis if it not surgically correctable.
- (Subclavian flap aortoplasty may be used in children but is not recommended in adults because of concern about the arterial supply to the arm).

Balloon dilation with or without stent insertion is an accepted procedure in re-coarctation, but is an experimental procedure in native coarctation and should only be performed in centers and by individuals with a commitment to the technique and to its clinical evaluation. There may be a role for compassionate use in patients thought to have an unacceptably high operative risk or who have serious co-morbid conditions.

**Grade: Consensus**

**Refs: 55,56,117**

If there is aortic stenosis and the coarctation is relieved first, re-evaluation of the aortic gradient is needed; this may usually be accomplished with echo Doppler.

## Part VI - Surgical/Interventional Outcomes

Following surgical repair of simple coarctation, the obstruction is usually relieved with minimal mortality (<1%). Paraplegia due to spinal cord ischemia is uncommon but recognized, particularly in patients who do not have well developed collateral circulation.

Late dissection at the repair site is a rare complication but false aneurysms may occur.

Aneurysm formation is well recognized following prosthetic patch aortoplasty (particularly Dacron) and occurs opposite the patch.

Following balloon dilation, aneurysm formation at the site of coarctation is a recognized complication, but its significance is unknown. The incidence is low in patients who have had prior coarctation surgery. This complication appears to be reduced if stents are used.

Hemoptysis from a leaking/ruptured aneurysm is a serious complication and requires urgent investigation. MRI or DSA is optimal because of the risk of rupture with aortography and failure to visualize the aneurysm.

Prior hypertension resolves in up to 50% of patients but this may depend on the length of follow up and age at repair. If it fails to resolve, it is generally responsive to standard therapy. Systolic hypertension is common with exercise. It may be related to residual arch hypoplasia.

Heart failure usually resolves.

Late cerebrovascular events occur, notably in those repaired as adults and in those with residual hypertension.

Endocarditis/endarteritis can occur at the coarctation site or on intracardiac lesions. If at the coarctation site, embolic manifestations are restricted to the legs.

## Part VII - Follow Up

All patients require periodic follow up by a cardiologist, preferably at a center for congenital cardiology.

All patients should have at least one MRI or angiogram following repair of the coarctation.

Particular attention should be directed towards:

- Residual hypertension, heart failure, or intracardiac disease.
- Other correctable risk factors for coronary artery disease, (hypercholesterolaemia, smoking).
- Associated bicuspid aortic valve which may develop stenosis later in life.
- Recurrent coarctation or significant arm-leg blood pressure gradient at rest or with exercise.
- Ascending aortopathy especially in the presence of bicuspid aortic valve.
- New or unusual headaches because of the possibility of berry aneurysms.
- Late dissection proximal or distal to the repair site.
- Aneurysm formation at the site of coarctation repair, especially if a Dacron patch has been used.

**Grade: Consensus**

Refs: 118-119

## **SECTION X - EBSTEIN ANOMALY**

### **Part I - Background Information**

Ebstein anomaly is rare. It encompasses a wide spectrum of anatomic and functional abnormalities of the morphological tricuspid valve (TV) which have certain features in common:

- Apical displacement of the septal and postero-lateral leaflets of the TV below the atrioventricular junction into the right ventricle.
- Resultant 'atrialization' of the inflow of the right ventricle to varying degrees.
- Varying degrees of tricuspid regurgitation (exceptionally the tricuspid valve is stenotic).
- Enlargement of the right atrium.
- A shunt at atrial level, either PFO or secundum ASD, in approximately 50%.
- One or more accessory conduction pathways, increasing the risk of atrial tachycardias, in 25% of cases.
- Varying degrees of anatomical and physiological right ventricular inflow or outflow tract obstruction.
- Varying impairment of left ventricular function.
- Varying degrees of cyanosis.

Associated lesions include:

- VSD.
- PS.
- Occasionally others e.g. coarctation or mitral valve prolapse.

### **Part II - Unoperated History and Management**

Patients with mild Ebstein anomaly may be asymptomatic with no functional limitation. They may be cyanosed or acyanotic. Survival to the ninth decade has been reported. However, they may die or become symptomatic at any age.

The most common symptoms in adults are exercise intolerance (dyspnea and fatigue) and symptomatic supraventricular arrhythmias (usually atrial fibrillation). Heart block occasionally occurs.

When an atrial defect is present, patients may be cyanotic to a varying degree (particularly on exercise), and are at risk of a paradoxical embolus resulting in TIA/CVA. Alternatively, they may have a significant left-to-right shunt which can reverse on effort.

End-stage disease with severe tricuspid regurgitation and right ventricular dysfunction may manifest as right-sided cardiac failure. It is usually precipitated by an arrhythmia such as atrial fibrillation. Sudden death may occur at any time.

### **Part III - Diagnostic Recommendations**

*An adequate diagnostic workup:*

- Documents the anatomic severity including the degree of right-sided enlargement, RV dysfunction and degree of TR.
- Determines whether the TV has the potential for surgical repair.
- Documents the presence or absence of an atrial communication and whether there is right-to-left shunting.
- Determines the presence or absence of associated lesions.

- Measures left ventricular function and mitral valve abnormalities.
- Defines, if possible, the presence or absence of an accessory pathway.
- Determines the amount of functional limitation, if any.

*The diagnostic workup should include at a minimum:*

- *A thorough clinical evaluation.*
- *ECG.*
- *Chest X-ray.*
- *Echo Doppler evaluation by a skilled individual.*
- *Oximetry.*
- *Exercise test.*

*The diagnostic workup may require:*

- TEE Doppler examination if the anatomic information is not provided by TTE.
- Holter monitor.
- An electrophysiological study if there is a history or ECG evidence suggestive of arrhythmias.
- *Coronary angiography if there is a suspicion of coronary artery disease or in patients over the age of 40 years if surgical repair is planned.*

#### **Part IV - Indications for Intervention**

The following situations warrant intervention:

- Deteriorating exercise capacity.
- Increasing heart size.
- Progressive cyanosis.
- Severe tricuspid regurgitation with increasing symptoms.
- A TIA or CVA.
- Atrial arrhythmias secondary to an accessory pathway which are not well controlled by medical therapy or which have the potential for sudden death.

**Grade: C**

**Level: V**

**Refs: 57-60**

#### **Part V - Surgical/Interventional Options**

Patients with Ebstein anomaly should only be repaired by surgeons with an interest in and experience with CHD, ideally with specific experience in this operation. Every effort should be made to preserve the native tricuspid valve.

**Grade: C**

**Level: V**

**Refs: 11,12**

When the anterior TV leaflet is mobile and not inserted into the ventricular outlet, valve repair may be possible and is preferable to valve replacement.

If the TV is not repairable, valve replacement will be necessary.

An atrial communication, if present, should be closed.

If an accessory pathway is present, this should be mapped and obliterated usually at the time of surgical repair but occasionally pre-operatively in the catheter laboratory. In patients with an inadequate RV (because of size or function) a bi-directional cavo-

pulmonary connection may supplement the intracardiac repair.

Occasionally a Fontan operation may be the best option in patients with tricuspid stenosis and/or hypoplastic RV.

It is controversial whether the atrialized portion of the right ventricle should be plicated.

## **Part VI - Surgical/Interventional Outcomes**

With satisfactory valve repair, with or without bi-directional cavo-pulmonary connection, medium term prognosis is excellent. Late arrhythmias, including atrio-ventricular block may occur.

With valve replacement, results are less satisfactory. Valve re-replacement may be necessary because of a failing bioprosthesis or thrombosed mechanical valve. Long term anti-coagulation with mechanical valves is mandatory. There is a high incidence of heart block.

## **Part VII - Follow Up**

All patients should have regular follow-up in a center for congenital cardiology.

Particular attention should be paid to:

- Recurrent tricuspid regurgitation in the previously operated patient.
- Degeneration of a bioprosthetic valve.
- Right-sided ventricular function which may worsen and cause congestion.
- Recurrent atrial arrhythmias.
- Ventricular arrhythmias.
- Complete heart block.

**Grade: Consensus**

# SECTION XI - COMPLETE TRANSPOSITION OF THE GREAT ARTERIES

## Part I - Background Information

### Definition

There is atrio-ventricular concordance and ventriculo-arterial discordance i.e. the right atrium connects to the right ventricle which gives rise to the aorta and the left atrium connects to the left ventricle which gives rise to the pulmonary artery.

Approximately 2/3 of patients have no major associated abnormalities ("simple" transposition). Approximately 1/3 have associated abnormalities ("complex" transposition). The most common associated abnormalities are VSD and pulmonary/sub-pulmonary stenosis.

## Part II - Unoperated/Operated History and Management

Unoperated (simple) transposition is a lethal condition with 90% mortality by one year. Thus nearly all patients seen as adults will have had intervention.

The most common surgical procedure in patients who are currently adults is the **atrial** switch operation. Most will have had a Mustard operation, although a few may have had a Senning operation. Blood is redirected at the **atrial** level using a baffle (Mustard operation) or atrial flaps (Senning operation), achieving physiological correction, but the right ventricle continues to support the systemic circulation.

Now, the atrial switch operation has been supplanted by the **arterial** switch operation (Jatene) but few of these patients have yet become adults. Blood is redirected at the **great artery** level by switching the aorta and pulmonary arteries such that the left ventricle supports the systemic circulation. The coronary arteries are translocated to the neo-aorta (formerly the pulmonary artery). The tissue loss in the sinuses of the neo-pulmonary artery is made good with a pericardial patch.

In a small proportion of patients (<10%) who have VSD and pulmonary/sub-pulmonary stenosis, a **Rastelli** operation will have been done. Blood is redirected at the **ventricular** level (with the left ventricle tunnelled to the aorta) and a valved conduit is placed from the right ventricle to the **pulmonary artery**. The left ventricle supports the systemic circulation.

Rarely, in patients with a large VSD and established pulmonary vascular disease, a **palliative atrial** switch operation will have been done to improve oxygenation. This is an **atrial** switch operation but the VSD is left open (or enlarged). These patients resemble Eisenmenger VSDs and should be managed as such.

*Transvenous pacing should be avoided in all patients with residual intracardiac communications since paradoxical emboli may occur. For the same reason, venous thromboemboli from any source are a potential hazard in these patients. Anticoagulants are recommended if a potential source of venous thromboembolism is found.*

**Grade: C**

**Level: V**

**Refs: 2**

In patients with an **atrial** switch operation, transvenous pacing leads must traverse the upper limb of the atrial switch to enter the morphological left ventricle. Active fixation is required.

## Part III - Investigational Recommendations in Operated Patients

Since all patients will have had some operation, investigations are directed towards post-operative sequelae and will vary according to the type of operation performed.

*All patients should have at a minimum:*

- A thorough clinical examination.
- ECG.
- Chest X-ray.
- Oximetry.

Patients who have had an **atrial** switch operation also require:

- *Echo-Doppler examination by a skilled individual* to detect baffle obstruction or baffle leak, AV valve regurgitation, and to assess systemic ventricular function and sub-pulmonary obstruction.
- A Holter monitor because of the high prevalence of sick sinus syndrome and atrial arrhythmias and possible ventricular arrhythmias in older patients.

*and may require:*

- TEE if there is inadequate visualisation of the intra-atrial baffle on the TTE.
- Nuclear cardiology assessment of ventricular function, if inadequate assessment is obtained from TTE or TEE.
- Complete heart catheterization including coronary angiography if there are doubts about additional lesions and surgical re-intervention is planned; or if adequate assessment of the hemodynamics is not obtained by non-invasive means.
- Exercise testing to evaluate heart rate, blood pressure response, oximetry, and to assess whether arrhythmias may be provoked.
- MRI to evaluate ventricular function and baffle function (obstruction or leakage) if not well visualized by other means.

Patients who have had an **arterial** switch operation also require:

- *Echo-Doppler examination by a skilled individual* to assess right ventricular outflow tract obstruction (the most common problem), ventricular function, possible neo-aortic valve regurgitation, and coronary ostial status (although the last may be difficult to see in adults).
- Exercise ECG at least once post-operatively because coronary ischemia is possible due to reimplantation or redirection of the coronary arteries.

*and may require:*

- Holter monitoring if arrhythmia is suspected. (The natural history following the arterial switch is unknown, but arrhythmias appear to be less common than after the atrial switch operation).
- Nuclear cardiology assessment of myocardial perfusion (if ischemia is suspected).
- Coronary arteriography if ischemia is documented on non-invasive testing.
- Complete heart catheterization if adequate assessment of the hemodynamics are not obtained by non-invasive means or additional lesions are suspected.

Patients who have had a **Rastelli** operation also require:

- *Echo-Doppler examination by a skilled individual* to assess right ventricle-to-pulmonary artery conduit stenosis/regurgitation, sub-aortic stenosis, aortic regurgitation, ventricular function and AV valve regurgitation.
- Assessment of the conduit gradient may be difficult but it is usually possible to measure the right ventricular systolic pressure from the tricuspid regurgitation jet and this may be a

useful surrogate in the absence of pulmonary hypertension.

and may require:

- MRI if inadequate information is obtained from TTE.
- A complete heart catheterization to determine the severity of conduit stenosis or regurgitation and the status of the distal pulmonary arteries if inadequate information is obtained from non-invasive testing and surgery is contemplated.

Patients who have had a **palliative atrial** switch operation also require:

- *Echo-Doppler examination by a skilled individual* to detect baffle obstruction or baffle leak, AV valve regurgitation and to assess systemic ventricular function.
- A Holter monitor because of the high prevalence of sick sinus syndrome and atrial arrhythmias.
- Oximetry at rest and possibly on exercise.
- Hemoglobin and assessment of iron stores. See general recommendations part III

#### Part IV - Indications for Re-intervention

The following situations may warrant re-intervention following the **atrial** switch procedure:

- Significant systemic (tricuspid) AV valve regurgitation.
- Severe right or left ventricular dysfunction.
- Symptomatic bradycardia, tachyarrhythmias or sick sinus syndrome.
- Baffle leak resulting in a significant left-to-right shunt (>1.5:1), any right-to-left shunt, symptoms or ventricular dysfunction.
- SVC or IVC pathway obstruction.
- Pulmonary venous obstruction (although this is usually seen early and will have been re-operated upon in childhood).

**Grade: C**

**Level: V**

**Refs: 61-66,120,121**

The following situations may warrant intervention following an **arterial** switch procedure:

- Significant right ventricular outflow tract obstruction at any level (gradient >60 mm Hg or RV:LV pressure ratio >0.6).
- Myocardial ischemia from coronary artery obstruction.
- Neo-aortic valve regurgitation.

**Grade: C**

**Level: V**

**Refs: 67-69**

The following situations may warrant re-intervention following the **Rastelli** procedure:

- Significant right ventricle-to-pulmonary artery conduit stenosis (>60 mm Hg) or significant regurgitation.
- Sub-aortic obstruction across the left ventricle-to-aorta tunnel.
- Residual VSD.
- Branch pulmonary artery stenosis.
- Aortic regurgitation.

**Grade: C**

**Level: V**

**Refs: 70,71**

## Part V - Surgical/Interventional Options

Patients who require re-intervention should only be treated at a center for congenital cardiology by cardiologists and cardiac surgeons with appropriate experience.

**Grade: C**

**Level:**

**Refs: 11,12**

The following are possible intervention strategies:

- Surgery may be necessary for baffle stenosis or leakage in patients with an **atrial** switch procedure. Balloon dilation of SVC or IVC stenosis is an option but success is limited in adults. Pathway obstruction is less common after the Senning operation than after the Mustard operation and is usually amenable to balloon dilation. SVC stenosis is usually benign, unlike IVC stenosis which may be life threatening. Stent insertion may be considered for SVC or IVC stenosis but is experimental.
- Patients with an **atrial** switch procedure and severe systemic (tricuspid) AV valve regurgitation may need valve replacement if systemic ventricular function is adequate.
- Patients with severe systemic (right) ventricular dysfunction and/or severe systemic (tricuspid) AV valve regurgitation following an **atrial** switch procedure may require consideration of heart transplantation. A conversion procedure to an **arterial** switch may be considered but this is experimental with little data available in adults.
- Patients who have had an **arterial** switch operation may require coronary artery bypass grafting (preferably with arterial conduits) for myocardial ischemia.
- Patients who have had an **arterial** switch operation may require augmentation of the right ventricular outflow tract for outflow tract obstruction.
- Patients who have had a **Rastelli** operation will need conduit replacement at some time.
- Patients who have had a **Rastelli** operation may need left ventricle-to-aorta baffle revision because of obstruction.
- Patients who have had a **palliative atrial** switch operation may require consideration of lung or heart-lung transplantation.
- Transvenous pacemaker insertion for symptomatic bradycardia or anti-tachycardia pacing for some atrial arrhythmias.
- Trans-catheter ablation procedures for intractable rhythm problems is experimental.

The role of afterload reduction with ACE inhibitors to preserve systemic right ventricular function is as yet unknown in clinical trials but the effects on dysfunctional systemic left ventricles are favourable and anecdotally it appears to have similar beneficial effects on systemic right ventricles.

## Part VI - Surgical/Interventional Outcomes

The overall survival of patients who have had an **atrial** switch procedure is approximately 65% at 25 years, with increased likelihood of survival with later year of operation. Patients who have “simple” transposition have a better survival (80% at 25 years) than those with “complex” transposition (45% at 25 years). Causes of death include sudden unexpected (presumed arrhythmic) death, ventricular failure and baffle obstruction.

The long term survival following the **arterial** switch is as yet unknown. Neo-aortic valve regurgitation, right ventricular outflow tract obstruction and coronary artery stenosis/occlusion are recognized complications.

Following the **Rastelli** operation repeated conduit changes will be necessary and there is a risk of deteriorating ventricular function. Ventricular and supraventricular tachycardia may be a problem.

Patients who have had a **palliative atrial** switch probably have a prognosis similar to Eisenmenger VSD but specific information is lacking. Quality of life is generally improved, however.

## **Part VII - Follow Up**

All patients should have regular cardiology follow up by a cardiologist with an interest in and dedication to the management of adult patients with CHD preferably at centers for congenital cardiology.

**Grade: Consensus**

## **SECTION XII - CONGENITALLY CORRECTED TRANSPOSITION OF THE GREAT ARTERIES**

### **Part I - Background Information**

#### **Definition**

There is atrio-ventricular discordance and ventriculo-arterial discordance. Systemic venous return to the right atrium enters the left ventricle which ejects blood into the pulmonary artery. Pulmonary venous return is to the left atrium and then via the right ventricle to the aorta. The circulation is physiologically corrected but the systemic circulation is supported by the morphologic right ventricle.

Congenitally corrected transposition may exist in the setting of univentricular heart, but this is not considered further here.

It is rare (<1% of CHD) but accounts for a high percentage of cyanotic patients undergoing surgery as adults. Associated anomalies occur in up to 98% in some series and consist of VSD (3/4 of cases), pulmonary or subpulmonary stenosis (3/4 of cases) and systemic (tricuspid) valve anomalies (1/3 of cases). Congenital complete heart block occurs in 5%.

### **Part II - Unoperated/Operated History and Management**

Patients with no associated abnormalities may survive until the 6<sup>th</sup> or 7<sup>th</sup> decade and may go unrecognized until problems arise. In addition to congenital complete atrio-ventricular block, acquired complete atrio-ventricular block continues to develop at 2% per year. Progressive systemic (tricuspid) AV valve regurgitation and systemic (right) ventricular dysfunction which may present as acute pulmonary edema precipitated by atrial arrhythmias tend to occur from the 4<sup>th</sup> decade onwards. Atrial arrhythmias are common from the 5<sup>th</sup> decade onwards.

Patients with VSD and pulmonary/subpulmonary stenosis are frequently cyanotic and may have been **palliated** with systemic-to-pulmonary artery shunts in childhood. **Repair** may involve implantation of a valved conduit from the pulmonary (left) ventricle to the pulmonary artery and repair of the VSD(s). Alternatively, the Ilbawi approach involves tunnelling the left ventricle to the aorta, a right ventricle-to-pulmonary artery conduit and an atrial switch (Mustard) with/without a bi-directional cavo-pulmonary anastomosis. Data in adults is lacking.

Complete atrio-ventricular block requires the insertion of a permanent pacemaker. The optimal modality is DDD but is not always possible. Active fixation electrodes are required.

*Transvenous pacing should be avoided if there are intracardiac shunts since paradoxical emboli may occur. Epicardial leads are preferred under these circumstances. For the same reason, venous thromboemboli from any source are a potential hazard. Anticoagulants should be used if a source of venous thromboembolism is found.*

**Grade: C**

**Level: V**

**Refs: 2**

If moderate or severe systemic (tricuspid) AV valve regurgitation develops, valve replacement is usually required. Valve repair is usually unsuccessful because of the abnormal often “Ebstein-like” anatomy of the valve.

**Grade: C**

**Level: V**

**Refs: 72**

Data in the adult using the Ilbawi approach (in the setting of VSD and pulmonary stenosis) or a double switch (i.e. Mustard operation and an arterial switch if there is no pulmonary stenosis) is lacking.

Patients with severe systemic (right) ventricular dysfunction (which may appear after surgical repair) may require consideration for transplantation.

If atrial arrhythmias occur, both anticoagulants and anti-arrhythmic therapy are usually required. Back-up pacing may also be necessary.

Pregnancy may be associated with a marked deterioration in ventricular function and/or development or worsening of systemic (tricuspid) AV valve regurgitation.

Pulmonary (mitral) AV valve regurgitation may occasionally occur and sub-pulmonary (morphological left) ventricular dysfunction or outflow tract obstruction may also develop and progress.

### **Part III - Diagnostic Recommendations**

*An adequate diagnostic workup:*

- Documents the anatomy described above.
- Identifies and quantitates associated abnormalities which may influence management (VSD, pulmonary/subpulmonary stenosis, systemic (tricuspid) AV valve regurgitation, ventricular function and AV block).

*The diagnostic workup should include at a minimum:*

- *A thorough clinical evaluation.*
- *ECG.*
- *Chest X-ray.*
- *Echo-Doppler examination by a skilled individual.*
- *Holter monitor.*
- *Exercise test with oximetry.*

*The diagnostic workup may require:*

- TEE examination to assess ventricular function, AV valve regurgitation and pulmonary outflow tract if this information is not provided by a TTE study, particularly in the operated patient. These patients are often very difficult to image on TTE because of a poor echo window.
- A complete heart catheterization to assess the hemodynamics, especially in the operated patient who has a conduit between the left ventricle and pulmonary artery or the unoperated patient who is being considered for surgery.
- *Coronary angiography if there is suspicion of coronary artery disease or if the patient is over the age of 40 years and surgery is planned.*
- Nuclear cardiology to assess ventricular function if inadequate information is available

from echocardiography.

- Rarely MRI to confirm/assess anatomy.
- Hemoglobin and iron studies if the patient is cyanosed.

#### Part IV - Indications for Intervention/Re-intervention

The outcome of patients with pulmonary stenosis/VSD who have a balanced pulmonary circulation (without excessive pulmonary blood flow on the one hand or excessive pulmonary stenosis on the other hand) is similar with or without operation.

The following situations may warrant intervention:

- The presence of VSD.
- Pulmonary or subpulmonary stenosis (gradient >60 mm Hg).
- The presence of moderate or greater systemic (tricuspid) AV valve regurgitation.
- Complete AV block which requires pacemaker implantation for symptoms, progressive or profound bradycardia, poor exercise heart rate response or cardiac enlargement.

**Grade: C**

**Level: V**

**Refs: 72-77, 122**

The following situations may warrant re-intervention:

- Stenosis across a prior left ventricle-to-pulmonary artery conduit (gradient >60mm Hg).
- Moderate or worse systemic (tricuspid) AV valve regurgitation following prior surgical repair.
- Deteriorating systemic (right) ventricular function.
- Residual VSD.
- Progressive pulmonary/subpulmonary stenosis not previously dealt with.
- Failing pacemaker.

**Grade: C**

**Level: V**

**Refs: 70,78,79**

The role of afterload reduction with ACE inhibitors to preserve systemic right ventricular function is as yet unknown in clinical trials but the effects on dysfunctional systemic left ventricles are favourable and anecdotally it appears to have similar beneficial effects on systemic right ventricles.

#### Part V - Surgical/Interventional Options

Patients who require intervention or re-intervention should be treated at a center for congenital cardiology by cardiologists and cardiac surgeons with appropriate experience.

**Grade: C**

**Level: V**

**Refs: 11,12**

VSD closure alone is almost always done in childhood with the VSD patch placed to avoid atrioventricular block.

Balloon dilation of pulmonary stenosis may lead to complete AV block and is not recommended.

VSD and pulmonary/subpulmonary stenosis are not infrequently repaired in adulthood. The VSD is patched and a conduit (usually valved) is placed from the pulmonary (left) ventricle to the pulmonary artery.

The native pulmonary trunk may or may not be left open.

Now, the Ilbawi approach may be used for VSD and pulmonary stenosis: the morphological

left ventricle is tunnelled through the VSD to the aorta and a valved conduit is placed between the morphological right ventricle and pulmonary artery. An **atrial** switch operation is also necessary. Thus the morphological left ventricle and mitral valve support the systemic circulation. These patients have not yet become adults and so long term outcome data is not available.

Patients with significant systemic (tricuspid) regurgitation will usually require valve replacement. Repair is usually impractical because the valve is often morphologically abnormal. Surgery should be performed before systemic ventricular function deteriorates. The Ilbawi approach, leaving the regurgitant tricuspid valve on the pulmonary side may be an option.

**Grade: C**

**Level: V**

**Refs: 72,76**

In isolated pulmonary/subpulmonary stenosis, direct enlargement of the outflow tract and valve is seldom possible because of the wedging of the outflow tract and the close relation to the conducting system and left coronary artery, and a pulmonary (left) ventricle-to-pulmonary artery conduit is required.

Complete AV block is not uncommon after surgery and necessitates pacing. However, it may occur at any time.

Occasional patients may be unsuitable for repair because of small pulmonary arteries, small systemic (right) ventricles or straddling AV valves and may require palliative shunt procedures.

Patients may require reoperation for conduit stenosis (gradient >60 mm Hg) or progressive systemic (tricuspid) AV valve regurgitation, either of a native valve or following prior valve repair/replacement.

Patients with deteriorating systemic (right) ventricular function should be treated aggressively with medical therapy but may need to be considered for transplantation. Deterioration may be rapid.

## **Part VI - Surgical/Interventional Outcome**

Of patients with congenitally corrected transposition operated or unoperated, who reach adulthood, median survival is 40 years. Survival is better if there are no associated anomalies. Usual causes of death are sudden (presumed arrhythmic) and progressive ventricular dysfunction with systemic (tricuspid) AV valve regurgitation.

Following surgical repair of VSD and/or subpulmonary stenosis, rapidly progressive systemic (tricuspid) AV valve regurgitation is well recognized. Medical therapy is often tried, but valve replacement is usually required.

Atrial arrhythmias are common in operated patients and may be related to systemic (tricuspid) AV valve regurgitation.

## **Part VII - Follow Up**

All patients should have regular cardiology follow up.

Follow up should be by a cardiologist with an interest in and dedication to the management of adult patients with CHD preferably at a center for congenital cardiology.

Particular attention should be paid to:

- Ventricular function. (Deteriorating systemic ventricular function may require consideration of transplantation).
- Systemic (tricuspid) AV valve regurgitation.
- Pulmonary (mitral) AV valve regurgitation.
- Progressive AV block.
- Atrial arrhythmias.

**Grade: Consensus**

## **SECTION XIII - EISENMENGER SYNDROME & PULMONARY VASCULAR OBSTRUCTIVE DISEASE**

### **Part I - Background Information**

#### **Definition**

Eisenmenger Syndrome [an extreme form of pulmonary vascular obstructive disease (PVOD)] is irreversible pulmonary hypertension at or near systemic levels due to high pulmonary vascular resistance with reversed or bi-directional shunting at great vessel, ventricular, and/or atrial levels.

The high pulmonary vascular resistance is usually established early in infancy (except in ASDs) and may sometimes be present from birth.

### **Part II - Unoperated History and Management**

Patients with large defects which allow free communication between the pulmonary and systemic circuits at the aortic or ventricular levels usually have a fairly healthy childhood, and gradually become progressively cyanotic during their second or third decade. Exercise intolerance (dyspnea and fatigue) is proportional to the degree of hypoxemia or cyanosis. In the absence of complications, these patients generally experience a slowly progressive decline in their physical capacity over the years.

Patients with an Eisenmenger duct may present later, having had a normal childhood, and not having had problems until either pregnancy or some other cardiovascular stress occurs.

In patients with medium or large ASDs, Eisenmenger physiology is usually acquired later, often after pregnancies or recurrent thromboembolism. Such additional factors may be required in ASD patients to develop this physiology. Early PVOD may occur with ASD, possibly in association with living at high altitude; congenital lung disease; associated abnormalities such as VSD or PDA; or obstructive left sided lesions.

Eisenmenger patients are prone to the following complications:

- Erythrocytosis.
- Hemoptysis.
- Bleeding disorders.
- Hyperuricemia/gout.
- Arrhythmias.
- Heart failure.
- Syncope.
- Sudden death.
- Stroke.
- Paradoxical emboli.
- Infective endocarditis.
- Angina pectoris.
- Brain abscess.

Heart failure seldom occurs before age 40 except in Eisenmenger AVSD.



*An adequate diagnostic workup:*

- Documents the presence of one or more communications between the systemic and pulmonary circuits at the great vessel, ventricular or atrial level.
- Documents the existence of severe pulmonary hypertension with significant right-to-left shunting (saturation <90%).
- Identifies other factors affecting the clinical condition of the patient (see complications and clinical sequelae).

*The diagnostic workup should include at a minimum:*

- *A thorough clinical examination* including examination of the toes.
- *ECG.*
- *Chest X-ray.*
- *Echo Doppler evaluation by a skilled individual.*
- Oximetry at rest, and occasionally with exertion (if the saturation at rest is more than 90%).
- Blood work (CBC, clotting profile, ferritin, uric acid).

*The diagnostic workup may occasionally require:*

- MRI to visualize the defect(s) between the pulmonary and systemic circuits or to define better its/their location(s) and size(s).
- TEE (rarely) to visualize defects between the pulmonary and systemic circuits or to define better its/their location(s) and size(s). Caution should be exercised with sedation.
- a complete heart catheterization with pulmonary vasodilators primarily to determine pulmonary artery pressures and resistances if these have not been adequately defined by other investigations and to rule out potentially reversible pulmonary vascular disease.
- *Open lung biopsy should only be considered when the reversibility of the pulmonary hypertension is uncertain from the hemodynamic data. It is potentially hazardous and should be done only at centres with experience in CHD.*

## **Part IV - Indications for Intervention**

The underlying principle of clinical management is to avoid any factors which may destabilize the delicately balanced physiology. In general an approach of non-intervention is recommended.

The main interventions, therefore, are directed to prevent complications (e.g. flu shots to reduce the morbidity of respiratory infections) or to restore the physiologic balance (e.g. iron replacement for iron deficiency; antiarrhythmic management of atrial arrhythmias).

Beyond this, the main intervention available is lung or heart-lung transplantation. This is generally reserved for those individuals without contra-indications who are felt to have a probability of one year survival of less than 50%. However, such assessment is fraught with difficulty because of the unpredictability of the time course of the disease and the risk of sudden death.

## **Part V - Surgical/Interventional Options**

Phlebotomy with fluid replacement  $\pm$  iron supplementation may be necessary in patients who are symptomatic from erythrocytosis. Prevention of iron deficiency is important.

**Grade: C**

**Level: V**

**Refs: 4,84,85**

Lung transplant may be undertaken in association with repair of existing cardiovascular defect(s).

Heart-lung transplantation may be required if the intracardiac anatomy is not correctable.

Contraception is extremely important in female patients. Sterilization is generally preferred (but is not without risks), and should be conducted with skilled anesthetic and intensive care support after full consultation with the patient. Intra-uterine contraceptive devices and the combined oral contraceptive pill are best avoided although progesterone-only pills and depot injection may be acceptable after the adolescent period.

**Grade: Consensus**

Supplemental oxygen, even at home, may reduce episodes of dyspnea although its routine use is not recommended. Prolonged supplementation especially at night can perhaps help raise oxygen saturation and reduce symptomatology, although its impact on survival is unclear.

**Grade: Consensus**

**Refs: 87**

The use of oral anticoagulants or aspirin is controversial.

As a general rule, the first episode of hemoptysis should be considered an indication for hospital admission and investigation. While often self-limiting, each such episode should be regarded as potentially important and a treatable cause sought. Some patients have recurrent small hemoptyses which seems to have a better prognosis. These may be due to pulmonary artery thromboses and consequent pulmonary infarcts.

Patients with PVOD and Eisenmenger syndrome should generally be given the following advice:

- Take medication only after consultation with your physician.
- Avoid pregnancy.
- Avoid dehydration.
- Avoid smoking.
- Tell your consultant cardiologist if you need non-cardiac surgery or are involved in an accident.
- Avoid excessive physical activity.
- Ask to be referred to a personal physician who understands and has experience in management of the Eisenmenger Syndrome.
- Avoid high altitude exposure.
- Consider supplemental oxygen on commercial airline travel and have assistance at airports.

**Grade: Consensus**

**Refs: 5,81,88**

## Part VI - Surgical/Interventional Outcomes

The outcome of transplantation in these patients is generally less satisfactory than for patients without Eisenmenger syndrome. Results of lung transplantation with primary intracardiac repair are worse than for lung transplantation in patients with primary pulmonary

hypertension or other primary lung disease. The outcome after heart-lung transplantation is even worse than lung transplantation alone. These options may, however, be relatively attractive to individuals confronting mortality. This poor outlook underscores the importance of preventing such pathological physiology through early diagnosis and treatment.

## **Part VII - Follow Up**

All such patients should be cared for by a cardiologist working in a, preferably center for congenital cardiology. They require expert supervision because of the precarious hemodynamics.

They may also benefit from the involvement of other specialists within such a center for congenital cardiology (respirology, psychology/psychiatry, hematology, gynecology, anesthesia, social worker).

**Grade: Consensus**

Refs:124

## SECTION XIV - FONTAN OPERATION

### Part I - Background Information

#### Definition

The Fontan operation is a palliative procedure for patients with a univentricular circulation. There is diversion of all of the systemic venous return to the pulmonary arteries, usually without employing a subpulmonary ventricle.

Originally described for patients with tricuspid atresia, it has now been extended to most forms of single ventricle circulation.

There are numerous variations in the surgical approach which may be a single or staged procedure.

### Part II - Operated History and Management

Patients who have had a Fontan operation are at risk from the following:

#### Arrhythmias

- Atrial flutter/fibrillation is common and increases with increasing duration of follow-up. This can be associated with profound hemodynamic deterioration. Attempts should be made to restore sinus rhythm.
- Heart block may also occur late and is often associated with hemodynamic deterioration.

When arrhythmias are present, an underlying hemodynamic cause should always be sought, and in particular, obstruction of the Fontan circuit needs to be excluded.

**Grade: C**

**Level: V**

**Refs: 89**

#### Thromboembolism (both systemic and pulmonary):

- May be associated with atrial fibrillation.
- May be related to sluggish circulation, especially in the systemic veins and right atrium.
- May be related to clotting abnormalities.

#### Protein-losing enteropathy (PLE):

- Occurs in up to 10% of postoperative Fontan patients.
- Is associated with ascites, peripheral edema, pleural and pericardial effusions.

#### Progressive deterioration of ventricular function:

- This may be part of the natural history of a patient with a single ventricle.

#### Hepatic dysfunction:

- Usually due to hepatic congestion.

#### Right pulmonary vein compression/obstruction:

- Due to compression from the enlarged right atrium or atrial baffle bulging into the left atrium.

#### Re-intervention:

- Pacemaker insertion or implantation.
- Revision of RV-PA, RA-PA or RA-RV connection especially if a conduit was used.
- Systemic AV valve repair or replacement.
- Repair of residual shunts.
- Intervention with balloons, stents or closure devices.

Pregnancy may carry additional risks to the mother because of the increased hemodynamic burden on the single ventricle and atrium. There is an increased risk of:

- Deterioration in ventricular function.
- Systemic AV valve regurgitation.
- Atrial arrhythmias.
- Thrombosis.

### Part III - Investigational Recommendations

All patients who have had a Fontan operation should be followed at a center for congenital cardiology.

**Grade: Consensus**

Particular attention should be paid to:

- Ventricular function, both systolic and diastolic.
- Obstruction at the Fontan anastomosis.
- Residual shunts.
- Systemic AV valve regurgitation.
- The detection of thrombus within the right atrium.
- Increasing cyanosis.
- The development of arrhythmias.
- The detection of pulmonary arterio-venous malformations resulting in increased cyanosis.
- Serum protein and albumin levels.
- Hepatic function.

Investigations are directed towards postoperative sequelae and will vary according to the type of operation performed.

*All patients should have, at a minimum:*

- *A thorough clinical examination.*
- *Electrocardiogram.*
- *Chest X-ray.*
- *Echo Doppler examination by a skilled individual to assess systemic ventricular function, AV valve regurgitation, the presence or absence of residual shunts, the presence or absence of obstruction in the Fontan circuit, and of spontaneous contrast ('smoke') in the atrium.*
- Serum protein and albumin measurement. If low, increased  $\alpha_1$  anti-trypsin clearance in the stool documents the presence of PLE.

*The diagnostic workup may require:*

- Echocardiography with a bubble study to rule out pulmonary arterio-venous malformations or venous collateral channels.
- TEE if there is inadequate visualization of the Fontan anastomosis or to exclude thrombus in the atrium.
- MRI if the Fontan anastomosis cannot be assessed reliably by TEE
- Complete heart catheterization if surgical re-intervention is planned or if adequate assessment of the hemodynamics is not obtained by non-invasive means. Even small gradients between the atrium and pulmonary artery (or outflow chamber) may suggest important obstruction across the Fontan anastomosis.

### Part IV - Indications for Re-intervention

The following situations may warrant re-intervention:

- Residual atrial septal defect resulting in a significant right-to-left shunt, symptoms or cyanosis.
- Residual shunt secondary to a previous palliative surgical shunt or residual ventricle-to-pulmonary artery connection.
- Significant systemic AV valve regurgitation.
- Obstruction in the Fontan circuit.
- Development of venous collateral channels or pulmonary arterio-venous malformations.
- Development of serious atrial arrhythmias.
- Development of PLE.
- Pulmonary venous obstruction.
- Pacemaker insertion or replacement.
- Planned closure of a fenestrated Fontan (trans-catheter).

**Grade: C**

**Level: V**

**Refs: 90-93,125**

## Part V - Surgical/Interventional Options

Patients who require re-intervention should be treated at a center for congenital cardiology by cardiologists and cardiac surgeons with appropriate experience.

**Grade: C**

**Level:**

**Refs: 11,12**

The following are possible intervention strategies:

- If permanent pacing is required, epicardial A-V sequential pacing should be employed whenever possible.
- Patients with systemic AV valve regurgitation may require AV valve repair or replacement:(caution: AV valve regurgitation may be the result of systemic ventricular failure).
- Patients with residual shunts of significance may require closure of the residual shunt.
- Patients with significant obstruction at the Fontan anastomosis may be candidates for balloon angioplasty, stenting or surgical intervention.
- Patients whose anastomosis is a valved conduit may need conduit replacement.
- Patients with venous collateral channels or arterio-venous malformation may need trans-catheter occlusion or possibly creation of an axillary arterio-venous fistula.
- Conversion of a classical Fontan to a lateral tunnel or external conduit may be considered for serious atrial arrhythmias or for very poor, deteriorating hemodynamics with a giant right atrium in a classical Fontan.
- Patients with PLE may be candidates for creation of a fenestration in the atrial septum.
- Transplantation may be necessary for systemic ventricular failure or intractable PLE.
- Patients with poorly controlled arrhythmias may be candidates for catheter ablation.

## Part VI - Surgical/Interventional Outcomes

The Fontan operation is palliative.

The reported average 10 year survival following Fontan operation is approximately 60% rising to 80% under ideal circumstances.

If PLE develops, the 5 year survival is approximately 50%. Reoperation following the Fontan procedure carries a high mortality, and with PLE the mortality may be as high as 75%. If obstruction in the Fontan circuit is the cause of the PLE, however, successful revision of the Fontan anastomosis may cure the PLE.

Usual causes of death are those related to ventricular failure, atrial arrhythmias, reoperation and PLE.

A beneficial role for ACE inhibitors is as yet unproven.

The role of long term anticoagulation is contentious. It is recommended that patients with a history of documented arrhythmias, fenestration in the Fontan connection, or spontaneous contrast ('smoke') in the right atrium on echocardiography be anticoagulated.

**Grade: Consensus**

## Part VII - Follow Up

All such patients should be cared for at a center for congenital cardiology.

**Grade: Consensus**

## SECTION XV MARFAN SYNDROME

### ♥ Part I – Background Information

#### Definition

The Marfan Syndrome is an autosomal dominantly inherited disorder of connective tissue in which cardiovascular, skeletal and ocular abnormalities may be present to a highly variable degree.

Prevalence has been estimated to be 1 in 3-5.000, 25-30% of which represent new mutations. The clinical features are the result of a weakening of the supportive tissues, related to a deficiency of fibrillin, a glycoprotein in the extracellular matrix. The underlying genetic defect is located on chromosome 15, where the mutations in the fibrillin gene may vary.

#### Part II - Unoperated History and Management

Prognosis of patients with Marfan syndrome is mainly determined by progressive aortic root dilation, potentially leading to aortic type A dissection and aortic rupture, which are the major causes of death. The mean survival of untreated patients is 40 years, but the variance is large. Not only the aortic root, but also other parts of the aorta may be dilated. Patients with a dilated aorta are usually asymptomatic. The presence of aortic valve insufficiency or mitral valve prolapse with insufficiency may lead to signs or symptoms of left ventricular volume overload.

Both medical and surgical therapies have improved life expectancy substantially, from a mean survival of 40 years in 1972 approximately to 60 years in 1993. The risk of type A dissection clearly increases with increasing aortic root diameter. Nonetheless, patients with no or only mild aortic dilatation occasionally dissect. A beneficial effect of  $\beta$ -adrenergic blockade has been shown in slowing the rate of aortic dilatation and reducing the risk of dissection.

In addition to reducing aortic stress by pharmacological means, avoiding both physical and emotional situations that increase blood pressure and heart rate is advisable. Furthermore, patients have to be strongly recommended to avoid exertion at maximal capacity or to perform contact sports and isometric sports.

**Grade : C**

**Level : V**

Refs:126-129

For women with Marfan syndrome, pregnancy presents a two-fold problem: first the genetic problem (a 50% chance that the child will be affected) and secondly, a high risk (up to 50%) of aortic dissection during or (especially) shortly after pregnancy.

Women with an aortic diameter above 50 mm are strongly discouraged regarding pregnancy. An aortic diameter below 40 mm rarely presents a problem, although a completely safe diameter does not exist.

**Grade : C**

**Level : V**

Refs:130

### Part III – Diagnostic Recommendations

## An adequate diagnostic workup

- Documents the presence of Marfan syndrome;  
Elucidation of the molecular mechanisms behind Marfan syndrome will allow improvement in diagnostic testing, but so far the diagnosis Marfan syndrome has to be made on clinical manifestations. Because of the variability in clinical expression, a multidisciplinary approach in a center for Marfan screening is required for complete evaluation of a patient and screening of the patients' relatives for Marfan syndrome. A definite diagnosis requires occurrence of a major manifestation in two different organ systems and involvement of a third organ system (table 1, ref 131)
- Determines the diameter of aortic root and all other parts of the aorta;
- Determines the presence of mitral valve prolapse and/or insufficiency, calcification of the mitral annulus and the diameter of the pulmonary artery;

*When the diagnosis Marfan syndrome has been established the diagnostic workup should include at a minimum:*

- *A thorough clinical evaluation,*
- *Electrocardiogram (ECG),*
- *Chest x-ray,*
- *Echo-Doppler evaluation especially for measurements of all parts of the aorta.*

*A diagnostic workup may require:*

- MRI to assess all aortic diameters,
- Coronary arteriography in patients over the age of 50 years (or younger if there are severe risk factors for coronary artery disease) in whom surgery is being planned.

Table 1

### Diagnostic Criteria for Marfan Syndrome

---

| Criteria                      | Major   | Minor   |
|-------------------------------|---|---|
| - family history/<br>genetics | parents, child, sibling<br>mutation FBN1  | -   |
| - cardiovascular              | aortic root dilatation<br>dissection ascending aorta  | aortic regurgitation<br>mitral valve prolapse<br>calcification of the mitral<br>valve (< 40 yrs)<br>dilatation pulmonary<br>artery<br>dilatation/dissection<br>descending aorta |
| - ocular                      | ectopia lentis  | ( $\geq 2$ ) : <sup>2</sup><br>flat cornea<br>myopia<br>elongated globe   |
| - skeletal                    | ( $\geq 4$ ): <sup>1</sup><br><br>pectus carinatum<br>severe pectus excavatum<br>wrist and thumb sign<br>scoliosis > 20 °<br>armspan-length ratio > 1,05<br>protrusio acetabulae<br>diminished extension<br>elbows (<170 °) | ( $\geq 2$ major, or 1 major<br>and 2 minor signs) : <sup>3</sup><br>pectus excavatum<br>hypermobility<br>high narrowly arched<br>palate<br>typical facies                      |
| - pulmonary                   | -   | spontaneous<br>pneumothorax   |
| - skin                        | -   | striae<br>recurrent herniae   |
| - central nervous system      | dural ectasia<br>on CT or MRI   | -   |

1. At least four manifestations should be present for a major criterium.
2. At least two manifestations should be present for a minor criterium.
3. The presence of at least two manifestations of the “major criteria” list forms one minor criterium. The presence of at least one manifestation of the “major criteria” list and two manifestations of the “minor criteria” list may also form one minor criterium.

#### Part IV – Indications for Intervention

The following situations warrant surgical intervention:



Three conditions are included:

**Ectopic origin of either the right or left coronary artery from the opposite sinus of Valsalva.**

- The ectopic artery passes between the aorta and right ventricular outflow tract (RVOT) and may be hypoplastic and/or kinked. It may be mechanically compressed by the great vessels particularly during exercise or with hypertension.

**Anomalous origin of the left coronary artery (LCA) from the pulmonary artery.**

- This occurs in 1/300,000 births.
- The anomalous LCA has a variable number of intercoronary collaterals which connect to the RCA.
- There may be segmental left ventricular dysfunction and papillary muscle dysfunction with associated mitral regurgitation.
- Pulmonary hypertension with an intracardiac shunt may mask the presence of the anomalous LCA which then presents as mitral regurgitation.
- May present as a left ventricular aneurysm.

**Congenital coronary arteriovenous fistula (CCAVF).**

- These are usually isolated anomalies involving communications of various size and number between a coronary artery and cardiac chamber, great artery or vena cava.
- Both coronary arteries arise normally.
- The coronary segment proximal to the fistula is variably dilated and tortuous and may become aneurysmal.
- The coronary artery distal to the origin of the fistula may be normal or small.
- The magnitude of flow through the fistula is determined by its size and by the presence or absence of restriction to flow at its drainage site.
- Two thirds of fistulae originate from the RCA, one quarter from the LCA, and the remainder from both arteries. Ninety percent of fistulae drain into the right atrium, coronary sinus or right ventricle.
- Multiple fistulae may originate from a single coronary artery and are typically restrictive.
- In older adults, fistulae may cause heart failure secondary to chronic volume overload and myocardial ischemia due to excessive diastolic run-off. Those entering the atrium or coronary sinus may also predispose to atrial fibrillation.

**Part II - Unoperated History and Management**

**Ectopic origin of coronary artery from the aorta:**

- May present in children and young adults (<40 years) with exertional chest pain, ventricular arrhythmias or sudden death (up to 20% of sudden deaths in young athletes in some series). In these individuals, the ectopic artery is typically dominant with a hypoplastic proximal portion and proximal compression.
- In older individuals (> 40 years) it may be discovered incidentally at the time of coronary angiography. Here there is typically no proximal obstruction and no related distal myocardial compromise. Hence there is no clinical significance.

**Anomalous origin of the LCA from the pulmonary artery:**

- Patients are generally identified as infants. They present with myocardial infarction, heart failure or mitral regurgitation. Intervention may allow partial or complete recovery of myocardial function and survival to adult age.

- 5-10% survive by natural mechanisms (optimal collateral/myocardial perfusion), and are identified during adulthood by the presence of mitral insufficiency, a remote Q wave on the ECG, an LV aneurysm, angina pectoris, ventricular arrhythmias or sudden death. There may be a continuous murmur from collaterals masquerading as a patent ductus arteriosus.

#### **CCAVF:**

- Rarely presents early in life with heart failure unless very large.
- Is usually diagnosed after detection of a continuous murmur in children or adults and less often during evaluation of atrial fibrillation or heart failure.
- May be discovered incidentally by coronary angiography (usually small, insignificant fistulae) especially in patients with cyanotic heart disease.
- May be a site for endocarditis.

### **Part III - Diagnostic Recommendations**

*An adequate diagnostic workup includes:*

#### **For ectopic origin of a coronary artery from the aorta:**

- Delineation of the origin and proximal course of the ectopic coronary artery, its caliber, relationship to the aorta and RVOT, its distal anatomy and size, and the status of the myocardium it perfuses notably in response to exercise. Exercise-induced ventricular tachycardia must also be sought.

#### **For anomalous origin of the LCA from the pulmonary artery:**

- Delineation of the origin, size and distribution of the anomalous coronary artery and the RCA and the intercoronary collateral vessels, the status of the left ventricle and mitral valve, the shunt size (typically small), the pulmonary artery pressure (typically normal), and the status of previously performed surgical interventions.

#### **For CCAVF:**

- Delineation of the specific anatomy [site of origin and exit(s), size and number of fistulae], the status of the coronary artery distal to fistula takeoff, the status of the receiving chambers, shunt size (typically small) and pulmonary artery pressure (usually normal), and the status of the remaining coronary arteries. An associated PDA should be excluded.

*The diagnostic workup should include at a minimum:*

- *A thorough clinical evaluation.*
- *ECG.*
- *Chest X-ray.*
- *Echo Doppler evaluation by a skilled individual* although fistulae may not be seen (Stress ultrasound for wall motion abnormality may be helpful).
- *Holter monitoring for ventricular arrhythmias.*
- *Thallium stress testing and sometimes ECG-gated exercise scan for regional wall motion assessment.*
- *Heart catheterization and selective coronary angiography in nearly all patients.*
- *MR angiography can be very helpful to delineate the course of ectopic originating coronary arteries.*

### **Part IV - Indications for Intervention**

#### **For ectopic origin of coronary artery from the aorta:**

Patients who have demonstrable myocardial ischemia in the distribution of the ectopic artery, who were resuscitated from cardiac arrest, or who have had serious ventricular arrhythmias or angina.

**Grade: C**

**Level: V**

**Refs: 94, 138**

**For anomalous origin of the LCA from the pulmonary artery:**

All patients with anomalous origin of LCA from the pulmonary artery.

**Grade: C**

**Level: V**

**Refs: 95**

**For CCAVF:**

Fistulae causing symptoms, hemodynamic compromise or arrhythmia.

Fistulae detected in young patients should be closed prophylactically because they enlarge with time.

**Grade: C**

**Level: V**

**Refs: 96**

## **Part V - Surgical/Interventional Options**

**For ectopic origin of coronary artery from the aorta:**

- Coronary artery bypass grafting using an arterial conduit whenever possible. (There has been a small experience with reimplanting or unroofing the ectopic artery internally for selected individuals. It is not yet clear whether ligation of the proximal ectopic artery should be performed at the time of bypass to avoid competitive flow.)
- If the decision to revascularize is unclear, at the least beta blockers and/or calcium channel blockers with frequent re-evaluation are appropriate.

**For anomalous origin of the LCA from the pulmonary artery:**

Establishment of an antegrade two coronary system is ideal. This may be achieved by:

- Re-implantation of the LCA into the aorta.
- Intra-pulmonary artery tunnel from the aorta to the origin of the LCA,
- Anastomosis of the left subclavian artery to the origin of the LCA.
- Coronary artery bypass grafting if re-implantation is not possible.

Ligation of the ectopic origin has largely been abandoned as a technique but this will nevertheless stop the coronary steal into the pulmonary artery.

Surgical attention to the mitral valve or a large ventricular aneurysm may also be needed.

**For CCAVF:**

- Transcatheter occlusion in experienced centres is increasingly an option if the anatomy is favourable and shunt size is significant.
- Surgical closure may be necessary if device occlusion is not possible.

## **Part VI - Surgical/Interventional Outcomes**

The risk of sudden death is probably reduced in patients with ectopic coronary artery origin after successful revascularization. Late bypass patency varies with surgical approach and the distal coronary artery status. Data are still not available as to late patency in these

generally young individuals. Ischemia, ventricular tachyarrhythmia and angina are usually relieved.

Angina, ventricular arrhythmias and the risk of sudden death are all usually improved in patients with anomalous origin of the LCA from the pulmonary artery following successful surgical repair. Mitral valve intervention, if needed, or aneurysmectomy carry routine surgical risks as does bypass grafting. Left ventricular function may return to normal but may remain abnormal and present as late cardiomyopathy.

Patients with CCAVF improve markedly following either transcatheter or surgical occlusion. Heart size, symptoms of preceding heart failure or, if present, myocardial ischemia (which is rare) abate. Atrial tachyarrhythmia which is peculiar to the adult group may persist and requires coumadin anticoagulation. Regardless of transcatheter or surgical closure, residual or recurrent fistulae may be seen.

## **Part VII - Follow Up**

All patients, except those with incidental small fistulae, warrant continued cardiac follow up, preferably in a center for congenital cardiology with particular attention being paid to:

- Recurrent myocardial ischemia.
- Deterioration of LV function or mitral regurgitation.
- Atrial (and possibly ventricular) arrhythmias.
- Recurrence of CCAVF after initial therapy.

**Grade: Consensus**

## **SECTION XVII - TRANSPLANTATION ISSUES**

### **Part I - Background Information**

Cardiac transplantation for patients with non-CHD may achieve excellent results with very low (<1%) operative mortality and 10 year adult survival rates of up to 50-60%.

This may also be true for patients suffering from CHD not amenable to surgical repair or palliation. For those patients with failure of repaired CHD the operative risk is higher (10-20%).

Lung transplantation (single or double) has been performed for patients with advanced (plexiform) PVOD secondary to CHD. While the procedure is effective in reducing pulmonary arterial pressure and relieving right ventricular hypertension, long-term results of pulmonary transplantation continue to be disappointing because of complications of obliterative bronchiolitis. Indications for pulmonary transplantation for PVOD are not fully established.

En bloc cardio-pulmonary transplantation can be performed on patients with PVOD (Eisenmenger syndrome), but given the scarcity of organs for transplantation, single or double lung transplant together with intracardiac repair may be a more reasonable option.

Cardiopulmonary transplantation would appear particularly suitable for those patients with congenital malformations affecting the central and peripheral pulmonary arteries. Patients with pulmonary atresia/VSD, absent or hypoplastic central pulmonary arteries and major systemic-to-pulmonary arterial collaterals would appear as the best candidates for this procedure if unoperated but multiple previous palliative procedures increases the mortality because of bleeding from bronchial/pleural collaterals. Long-term results are worse than those of isolated lung transplantation because of a higher incidence of obliterative bronchiolitis.

### **Part II - Population**

AChD patients represent a significant pool of potential transplant patients. Patient groups can be separated according to the following categories:

- Those with untreated CHD. There continues to be a small group of patients who are diagnosed late in life or for whom treatment became available when the patient was no longer operable. Transplantation may be indicated in those symptomatic patients who are not candidates for palliative or reconstructive surgery.
- Those with palliated CHD. There is a significant population of patients who had palliative surgery in infancy, childhood or adolescence who are not candidates for further reconstructive procedures.
- Those with “repaired” CHD. Most patients with “repaired” CHD require continued vigilance by medical teams experienced with this pathology. Failure of so called “curative” procedures is relatively common as the years of follow-up accumulate for those patients despite a technically adequate procedure.
- The patient with CHD transplanted in the pre-adult age group. A significant number of

infants, children and adolescents are undergoing transplantation for otherwise untreatable CHD. Some will reach adulthood and will continue to experience the problems inherent to patients receiving long-term immunosuppressive agents: rejection, infections, coronary atherosclerosis, malignancies, etc. Re-transplantation may become an important issue in these patients.

### **Part III - Indications for Transplantation**

Cardiac and/or pulmonary transplantation can be considered for patients with CHD when their estimated survival is less than that expected for transplanted patients.

Transplantation is a reasonable option when the patient's symptoms and quality of life have deteriorated to an intolerable degree.

Cardio-pulmonary testing may be useful in assessing patients for transplantation, but many patients who are symptomatically well have maximal oxygen consumption/exercise capacity within the same range as patients awaiting transplantation for other reasons (i.e. ischemic/dilated cardiomyopathy).

Adults with CHD undergoing cardiac transplantation have usually had previous palliative and/or reconstructive surgery. The most common indications include: dysfunction of the systemic ventricle resulting in congestive heart failure, and inability to palliate further, either medically or surgically, patients with anomalies not suitable for definitive correction.

A common reason for cardiac transplantation is in patients with failure of the systemic right ventricle i.e. complete transposition post Mustard or Senning procedures; congenitally corrected transposition of the great arteries. Reconstructive procedures designed to retrain the morphologic left ventricle and use it as the systemic ventricle have been tried "but have not been successful in adults so far". Their role in the management of these patients remains unclear and currently transplantation may be a better option.

An increasingly important indication for cardiac transplantation is patients with failing Fontan procedures. Indications include systemic ventricular failure and protein losing enteropathy. The latter may be reversed with cardiac transplantation although mortality may be high.

Patients with advanced (plexiform) PVOD resulting from congenital heart lesions have been treated successfully by transplantation, either using heart-lung transplantation or single/double lung transplantation with intracardiac repair (if the cardiac anatomy is suitable).

Bilateral lung transplantation appears preferable to single lung transplantation for patients with PVOD. Single lung transplantation may result in overperfusion of the transplanted lung with resulting ventilation-perfusion mismatch.

Patients with pulmonary atresia, VSD and major aorto-pulmonary arterial collaterals present a significant problem for transplantation, particularly when they have undergone multiple palliative and/or reconstructive procedures. The profuse mediastinal and pleural arterial collaterals in these patients are responsible for severe hemorrhage during attempts at pulmonary or cardiopulmonary transplantation. This problem is exacerbated by the presence of vascular adhesions resulting from previous surgical procedures. Selection of these patients for transplantation remains questionable.

### **Part IV - Evaluation of Patients for Transplantation**

The decision to pursue transplantation of heart and/or lungs in adult patients with CHD is complex. It is best done in collaboration with centres with experience in the management of

these patients. It requires contributions from cardiologists with special interest in CHD, a congenital heart surgeon, and a transplant surgeon. In addition to medical issues, socioeconomic issues play an important role in this decision.

Evaluation of patients for cardiac and/or pulmonary transplantation requires:

- A thorough medical evaluation.
- Multi-organ physiologic assessment.
- Cardiac catheterization for morphologic and hemodynamic assessment.
- Infectious disease evaluation.
- Psychosocial evaluation.

Neurological, hepatic and renal function must be thoroughly evaluated. Impairment in multi-organ function must be reversible after transplantation.

Hemodynamic assessment in preparation for cardiac transplantation should include the estimation of pulmonary artery resistance and its response to pharmacologic agents; nitroprusside, prostaglandin, nitric oxide, etc.

Assessment of cardiac and pulmonary arterial morphology is indispensable for the proper planning of the transplantation procedure. Of particular interest is the spatial arrangement of those cardiac structures that will remain after excision of the diseased heart. Thus, the transplant surgeon will be interested in the presence or absence of anomalies of cardiac position, anomalies to the systemic and/or pulmonary venous drainage, transposition of the great vessels, aortic arch anomalies, central pulmonary artery anomalies, etc. Imaging techniques such as TTE and TEE, CT and MRI must be available for this purpose and will be used in selected circumstances.

## **Part V - Technical Aspects of Cardiopulmonary Transplantation**

A normal donor heart can be adapted to most congenital heart recipients. Harvesting the donor heart with long segments of attached aorta and pulmonary arteries allows the surgeon to transplant patients with transposition anomalies and those with abnormalities of the central pulmonary arteries. Similarly, patients with anomalous systemic and pulmonary venous connections can be transplanted using special reconstructive techniques. Nevertheless, a few patients with major, complex spatial abnormalities (cardiac malformations associated with asplenia and polysplenia syndromes) may not be amenable to cardiac transplantation.

Pulmonary transplantation for patients with PVOD can be performed with concomitant repair of cardiac lesions (atrial or ventricular septal defects, PDA or aortopulmonary window) in those patient with well preserved cardiac function.

In general terms, orthotopic cardiac transplantation is preferred over heterotopic transplantation for patient with CHD.

Transplantation should be considered (and performed) before renal and/or hepatic dysfunction becomes established.

Multiple previous cardiac surgical procedures have not represented a significant obstacle to cardiac transplantation or to patients undergoing isolated lung transplants. Previous lateral thoracotomy has increased significantly the risk of en bloc cardiopulmonary transplant due to uncontrollable hemorrhage. Thus the likelihood of possible eventual transplantation should be considered before performing a palliative procedure using a lateral thoracotomy.

## **Part VI - Post-transplantation Course**

Postoperative management of the transplant patient is best done by a multidisciplinary group including the cardiac transplant surgeon, cardiac anesthetist, transplant cardiologist and infectious disease specialist. For those patients undergoing pulmonary transplantation, the team must include a pulmonary specialist. Surveillance requires regular biopsies and the contribution of a pathologist well versed in problems of cardiac and/or pulmonary rejection.

Common complications that affect the course of transplanted patients include acute rejection, sepsis, and manifestations of chronic rejection, such as coronary atherosclerosis and pulmonary fibrosis. Late malignancies have been reported in these patients. Surveillance of these patients must continue in the hands of the transplant team.

## **SECTION XVIII - REPRODUCTIVE ISSUES**

### **Part I - Preconception Counseling - Maternal and Fetal Risks**

Maternal risk for pregnancy - morbidity and mortality - varies with functional class, but there is no consensus on which functional classification to use.

The following adaptation of the New York Heart Association Functional Classification is used in this document about adults with CHD and refers to those who are cyanotic or acyanotic as well as those who are unoperated or who have undergone intervention:

- Class 1 - Asymptomatic at all levels of activity.
- Class 2 - Symptoms are present but do not curtail average, everyday activity.
- Class 3 - Symptoms significantly curtail most but not all average, everyday activity.
- Class 4 - Symptoms significantly curtail virtually all average, everyday activity and may be present at rest.

Maternal risk is low if not absent for women in Class 1, and high for women in Class 3 or 4.

Irrespective of functional disability, the presence of significant pulmonary vascular disease (PAP>2/3 SABP ) carries high maternal risk for death (up to 50% in Eisenmenger syndrome).

Cyanosis with pulmonary hypertension constitutes a grave risk to both the mother (pulmonary vascular disease) and fetus (hypoxemia). Cyanosis itself threatens the growth, development, and viability of the fetus, and increases fetal loss, dysmaturity, and prematurity.

Intrauterine viability is threatened by the functional class of the mother, by maternal cyanosis, and by maternal oral anticoagulants.

Remote risks to the fetus include genetic parental transmission (excepting autosomal conditions), and teratogenic effects of certain cardiac drugs.

Fetal mortality ranges from virtually zero in Class 1 mothers to up to 50% in Class 4 mothers.

Anticoagulants should be avoided whenever possible in women of childbearing age. This includes both warfarin (fetal skeletal abnormalities in the first trimester) and long term heparin (maternal osteoporosis).

**Grade: C**

**Level: III**

**Refs: 97,98**

This principle emphasizes the importance, when possible, of valve reconstruction or consideration of a bioprosthesis rather than replacement with a mechanical prosthesis in women having preconception cardiac surgery. However, there may be rapid deterioration of bioprostheses in young people.

If anticoagulants are required during pregnancy, four options are available:

Preferably:

- Continuing warfarin during the pre-conception period with frequent pregnancy tests,

switching to *fraxiparin* as soon as pregnancy is confirmed and continuing through the first trimester, then returning to warfarin through the 32<sup>nd</sup> week when *fraxiparin* is re-employed in anticipation of labour.

Other options are:

- If conception is unplanned, replacing warfarin with *fraxiparin* as soon as pregnancy is established, continuing *fraxiparin* throughout pregnancy or using warfarin from the second trimester through the 32<sup>nd</sup> week before re-employing *fraxiparin*.
- Replacing warfarin with subcutaneous *fraxiparin* *prior* to conception, continuing *fraxiparin* during the first trimester, then returning to warfarin through the 32<sup>nd</sup> week when *fraxiparin* is re-employed in anticipation of labour.
- Replacing warfarin with *fraxiparin* *before* conception and continuing *fraxiparin* throughout pregnancy.

## Part II - Preconception Counseling - Genetic Counseling

Genetic counseling focuses upon fetal risks of recurrence of CHD, which are best related to a risk of 0.4 to 0.8 % in the general population.

- Risk with one offspring previously affected averages 2.3% (1.5 to 5.0%) and depends on the lesion.
- Risk with two affected siblings averages 7.3% (5 to 10%).
- Risk if the mother is affected averages 6.7 % (2.5 to 18%).
- Risk if the father is affected averages 2.1% (1.5 to 3%).

The defect in a sibling or offspring is not necessarily the same type of cardiac malformation as in a previously affected individual.

There is a higher recurrence risk for left heart obstructive lesions.

Some conditions show an autosomal dominant pattern of inheritance (e.g. velo-cardio-facial syndrome, Marfan syndrome) with a risk of 50% in a second offspring.

## Part III - Post Conception Counseling

In families with CHD, either in the parents or offspring, fetal echocardiography should be performed (at 20-22 weeks) to exclude whether or not the fetus is affected with CHD. If the fetus is affected, advice should be offered as to the outlook for the affected fetus and, if appropriate, discussion should involve the possibility of termination of the pregnancy.

## Part IV - Management of Gestation, Labour, and Delivery

In most Class 1 women, management of labour and delivery is the same as normals, except for the risk of infective endocarditis. Antibiotic prophylaxis should be considered under most circumstances.

In many Class II-III women, no additional measures for the management of labour and delivery are necessary.

**Grade: Consensus**

Irrespective of functional class, cesarean section should be advised for major **obstetrical** indications and should possibly be used for patients with Marfan syndrome with a dilated aortic root (because of the risk of dissection/rupture).

**Grade: Consensus**

**Refs: 51, 99**

In high risk pregnancies, labour should be induced, with delivery carefully planned after amniocentesis has determined fetal lung maturity. An obstetrician skilled in high risk pregnancies, neonatologist, and cardiologist should be accessible. An anesthetist should be consulted prior to the onset of labour. Cervical ripening can be performed using either intracervical or intravaginal prostaglandins. Induction or stimulation of labour can be achieved with oxytocin infusion.

**Grade: Consensus**

In high risk pregnancies, women should labour in a lateral decubitus position to attenuate hemodynamic fluctuations associated with major contractions in the supine position. Care must be exercised with regard to volume status. The application of lumbar epidural anesthesia is often advisable to reduce tachycardia and hypertension. The fetus should pass through the pelvis in response to the force of uterine contractions, unaugmented as much as possible with straining. Delivery is assisted by vacuum extraction and/or low forceps. Following delivery, careful monitoring needs to be maintained for up to 72 hours, occasionally in an intensive care unit environment. Routine pediatric cardiac assessment of the newborn infant is advised.

**Grade: Consensus**

**Refs: 100**

## Part V - Contraception

Apart from barrier methods and tubal ligation, levonorgestrel (Norplant) may be a safe and efficacious contraceptive and can possibly be used for women with cyanotic CHD and pulmonary vascular disease.

**Grade: Consensus**

Medroxyprogesterone (Depoprovera) and progesterone-only pills are inappropriate for patients with heart failure because of the tendency for fluid retention. In adolescents, progesterone-only pills may cause depression. Progesterone-only pills have a higher failure rate than combined oral contraceptive pills but can be considered.

**Grade: Consensus**

Combined oral contraceptive pills with low estrogen components (including triphasic pills) may be safe and less thrombogenic than those with high estrogen components, with high efficacy provided there is strict patient compliance, but data are lacking.

**Grade: Consensus**

Intra uterine devices may increase the risk of endocarditis and, particularly in cyanotic women, cause increased bleeding.

**Grade: Consensus**

**Refs: 101,102,139**

## **Part VI - Sterilization and Termination of Pregnancy**

Pregnancy is contraindicated and sterilization should be offered:

- As an option to prevent conception in Class 3 or 4 women.
- Irrespective of class, in women with pulmonary vascular disease (pulmonary vascular resistance  $>2/3$  of systemic).
- In cyanotic women with pulmonary hypertension.
- When systemic ventricular dysfunction is likely to be exacerbated by pregnancy.
- As an option to prevent recurrence of autosomal dominant conditions.

**Grade: Consensus**

**Refs: 5,103**

Tubal ligation is probably the safest and most appropriate form of sterilization.

**Grade: Consensus**

Under exceptional circumstances, vasectomy may be considered for the spouses of females with CHD.

**Grade: Consensus**

## **Part VII - Specific High Risk Settings that Contraindicate Pregnancy and in which Termination should be offered**

Pregnancy is contraindicated in the following settings, and termination should be offered:

- Pulmonary vascular disease, especially with cyanotic heart lesion, particularly Eisenmenger syndrome.
- Grade 4 systemic ventricular function.
- Marfan syndrome with aortic aneurysm.

In patients with severe left ventricular outflow obstruction, surgery or balloon dilatation should be applied prior to conception.

**Grade: C**

**Level: V**

**Refs: 5,104,105**

Termination may also be considered if fetal echocardiography demonstrates severe CHD.

**Grade: Consensus**

## **SECTION XIX - INFECTIVE ENDOCARDITIS**

### **Part I - General Settings that Incur Risk**

There are two major predisposing components of infective endocarditis: a susceptible cardiac or vascular lesion (substrate), and a source of bacteremia.

Lesions that impart risk are characterized by high velocity flow, jet impact, turbulence and focal increases in the shear rate.

Sources of bacteremia may include the oral cavity (teeth, gums), skin, fingernails/periungual area, genitourinary tract, labour and delivery, indwelling vascular catheters, nasotracheal intubation or suctioning, and certain types of non-cardiac surgery (e.g. bowel surgery).

The rate of infection incurred by intrauterine devices is only 1.4 times normal, provided the sexual relationships are monogamous. Antibiotic prophylaxis should be given at the time of insertion of the intrauterine device. Right-sided endocarditis may occur secondary to pelvic inflammatory disease, especially in sexually promiscuous patients.

Fiberoptic or flexible endoscopy does not incur risk of bacteremia unless traumatic (except genito-urinary endoscopy in the presence of infection). Cardiac catheterization and transesophageal echocardiography incur only trivial risk and prophylaxis is not recommended.

Cardiac surgery predisposes to bacteremia because of the invasive peri-operative supportive measures in the immediate postoperative period (urethral catheterization in the male, indwelling flotation catheters, traumatic nasotracheal intubation and suction) and the open wound. Prolonged intensive care unit stay may be associated with increased risk of endocarditis on native or newly implanted valves.

### **Part II - Prophylaxis**

Both non-chemotherapeutic and chemotherapeutic prophylaxis are important.

Non-chemotherapeutic prophylaxis includes proper day-to-day oral hygiene, skin and nail care.

Chemotherapeutic prophylaxis depends upon the risk category and the potential sources of bacteremia, and also upon potential antibiotic sensitivity.

Refs: 140, 141

Patients should be educated about the importance of avoiding antibiotics until blood cultures have been taken.

Some conditions do not need antibiotic prophylaxis. (e.g. ASD, mild pulmonary stenosis)

### **Part III - Criteria for the Diagnosis of Infective Endocarditis**

There is legitimate suspicion in a patient with a susceptible substrate, a source of bacteremia, and symptoms and signs; or echo or surgical findings suggestive of endocarditis. Confirmation ideally rests upon positive blood cultures but culture negative endocarditis is recognized. This is usually the result of indiscriminate use of antibiotics prior to appropriate investigation i.e. blood cultures.

### **Part IV - Management**

Management should be in collaboration with an infectious disease specialist. Unusual organisms may be involved. The crucial element in treatment is identification of the organism and selection of the appropriate antibiotics. To reduce cost without risking efficacy, home administration of antibiotics should be considered for completion of the course after an initial in-patient stay.

Management decisions about infected prosthetic valves or infected conduits in which the duration of preoperative antibiotic therapy must be balanced against the anticipated risk incurred by deferring surgical replacement of the infected prosthetic material should be made in collaboration with a cardiac surgeon and should be considered after 24-48 hours of appropriate intravenous antibiotics.

The indications for prophylaxis of endocarditis as well as the specific antibiotic regimes are advised by the Dutch Heart Foundation(Ref: 142).See appendix I

## **SECTION XX - PSYCHOSOCIAL ISSUES**

### **Part I - Background**

CHD has implications for mental health, neuropsychological functioning and quality of life. Many adults present with psychologic, psychiatric and neuropsychological morbidities which can impact upon medical management.

Adults with CHD face the challenge of maintaining employment, inter-personal relationships and assuming responsibility for their own health care. Long-term disturbances in psychological adjustment range from frank psychiatric disorders to subclinical distress (secondary to the stress of chronic illness & disability, parental overprotection). Disturbances in neurocognitive functioning (secondary to ischemia/hypoxia and/or cardiopulmonary bypass in childhood) also occur.

Acute exacerbations of illnesses and hospitalisations in individuals without frank psychiatric illness can precipitate psychological difficulties that require intervention in their own right. They can exacerbate cardiac symptomatology and can interfere with the individual's adjustment to their illness/hospitalisation and acceptance of care. Additionally, individuals with heart defects associated with heart failure and/or hypoxemia can present with new abnormalities in neuropsychological functioning (sometimes superimposed on long-term neurocognitive sequelae of early hypoxia/ischemia) due to cardiac dysfunction in the absence of frank CVA or brain abscess.

An awareness and understanding of co-morbidities related to mental status is necessary to deliver appropriate care. Expert input should be sought when needed.

### **Part II - Psychosocial Outcomes: Impact of Congenital Heart Disease on Achievement of Developmental Milestones of Adulthood**

Psychosocial outcomes amongst adults with CHD are highly variable. They:

- Are less likely to earn a university degree.
- Are 3 to 4 times as likely to face unemployment than their peers. (This may partly be due to discrimination.)
- May underachieve because of inaccurate beliefs about their own health.
- May have different perceptions about their well-being than their cardiologist.

It is thus important to discuss functional capacity with a cardiologist, and if necessary with a psychologist, so as to facilitate optimal functioning, including employability.

**Grade: Consensus**

Young adults with CHD describe more concerns about sexuality than other groups of young adults with chronic illness: young men report relatively greater concerns related to issues of sexual arousal and performance and issues related to pregnancy are of concern to a significant proportion of affected young women.

Genetic transmission of CHD to offspring is of concern to both males and females with CHD. Many may not have had the opportunity to discuss this because in childhood such concerns were discussed with the affected individual's parents.

### Part III - Psychiatric Morbidity

Specific prevalence of frank psychiatric disorders in adults with congenital cardiac disorders is unknown but clinical impression and preliminary data suggest that anxiety disorders and personality disorders may be more prevalent than in the general population.

Individuals who present with depression and/or anxiety require diagnostic evaluation as well as psychotherapeutic and possibly pharmacological intervention. Consultation about diagnoses of psychiatric disorders, as well as therapeutic options, should be with a psychologist or psychiatrist with a special interest and expertise in CHD patients when possible.

**Grade: Consensus**

Pharmacological interventions require psychiatric consultation and monitoring. Collaboration between cardiologist and psychologist/psychiatrist may clarify diagnostic uncertainties generated by an overlap between cardiac and psychological/psychiatric symptoms.

In addition to frank psychiatric disorders, individuals with CHD appear to be at risk for subclinical depression and anxiety which may impact on their ability to work and maintain interpersonal relationships. It appears that otherwise unanticipated limited exercise tolerance may be related to self-reported anxiety/depression.

Onychophagia (persistent nail biting) which often serves to reduce anxiety appears to be more prevalent in adults with CHD. It risks infection and staphylococcal bacteremia.

Poor psychological adjustment in children and adolescents with CHD has been found to be associated with maternal pampering and overprotection.

Young adults with CHD perceive themselves to be different from their peers: to evaluate themselves more negatively, to be more dependent and overprotected, to be less well informed and to be less ambitious. Increased feelings of vulnerability, poor body concept, poor self-image and emotional immaturity have also been reported in adults with CHD. They have been found to be more likely to lead a dependent lifestyle, living at home with their parents without a marital/romantic relationship, and be more likely to show evidence of social immaturity. Clinical concerns with respect to psychological functioning are sometimes intertwined with existential issues related to the challenge of chronic illness at a young age.

There may be a higher prevalence of body image issues in adults with CHD. Body image issues may both fuel anxiety with respect to intimate relationships and also lead to excessive concern about accepting surgical interventions that leave scars.

### Part IV - Psychological Adjustment To Acute Exacerbations In Illness &/Or Hospitalization

Individuals with long-standing psychiatric difficulties (including personality disorders) may experience exacerbations during acute cardiac decompensation, increased cardiac symptomatology, or when diagnostic/therapeutic interventions or hospitalization are required.

Individuals without long-standing frank psychiatric difficulties may also experience intense anxiety and sometimes panic attacks under similar circumstances. They may become hypervigilant to their experience of pain and cardiac symptomatology. They are often very

fearful of the hospital environment, diagnostic tests & interventions. Cardiac catheterization is particularly anxiety/panic-provoking in some adults with CHD.

Anxiety and fear can exacerbate the aversiveness of cardiac symptomatology and/or impede the likelihood that interventions will be tolerated or accepted. These may be intensified by the limited experience of some individuals with congenital cardiac disorders to assume responsibility for their own health care, since parents have often assumed responsibility for these issues. Extreme dependency may ultimately hamper a sense of self-sufficiency and ability to cope with their illness.

Consultation with the psychologist and/or psychiatrist is indicated. In the case of severe agitation, psychiatric consultation regarding pharmacotherapeutic strategies is appropriate. In cases of less severe anxiety or if psychotropic medication has proved to be ineffective or is contraindicated for cardiac reasons, psychologists can provide specific and brief cognitive-behavioural strategies that will allow the individual to manage their own anxiety, panic and fear more effectively.

**Grade: Consensus**

## **Part V - Acute & Chronic Neuropsychological Abnormalities**

A significant proportion of children with untreated or palliated CHD present with neurological abnormalities on physical examination, on EEG or on neurological imaging. 1.5 to 10.9% of children born with cyanotic CHD will have cerebrovascular infarction over the course of their illness. Most occur in the first two years of life, either spontaneously or after surgery. Cyanotic cardiac anomalies with right-to-left shunts are particularly susceptible to cerebrovascular complications and brain abscesses. Under anoxic conditions, the younger child is particularly susceptible to periventricular hemorrhage with an increased likelihood of infarction of subcortical grey matter. The older the child with hypoxia/ischemia, the more likely are white matter lesions and watershed lesions.

Brain areas found to be particularly vulnerable to injury in the older child with CHD include the hippocampus, the Purkinje cells of the cerebellum and the pontine nuclei in the brainstem. Neuropathological findings have generally been attributed to the effects of prolonged hypoxia, acidosis, increased hematocrit and hypotension. Different mechanisms may underlie the increased risk of cerebrovascular infarction as the child ages.

CNS comorbidities have implications for mental status over the course of development. Structural neuroimaging may not be the most sensitive index of brain dysfunction in systemic disorders prior to the development of frank cerebral lesions. Neuropsychological examinations are an important component of the assessment for the detection of CNS comorbidity.

There is limited data in adults with CHD. Measures of intellectual functioning may be normal overall but the distribution may show a wider range. Abstract reasoning and spatial memory may be impaired. There is a high self-assessed level of depression/anxiety and cognitive difficulties.

It has been suggested that impairment in intellectual development observed in children with cyanotic CHD lesions may be secondary to the long-term effects of chronic cerebral hypoxia in infancy. Generally, children at highest risk for neurologic complications, poorer intellectual and academic outcomes were those who underwent unsuccessful palliative procedures, who were surgically repaired after the age of 2 years and who exhibited episodes of heart

failure in infancy. While each of these factors, individually, was only modestly correlated to later cognitive functioning, combinations of factors were more strongly related to childhood neurologic and psychologic outcomes.

While there is evidence that early surgical correction of cyanotic heart lesions is associated with a reduced risk of cerebrovascular infarction, several studies suggest that pediatric cardiac surgery with circulatory arrest and rapid cooling may be associated with neurocognitive loss. This may have consequences later in adult life.

## **Part VI - Neuropsychiatric Problems in the Mentally Retarded**

Adults with CHD and mental retardation associated with neurodevelopmental disorders such as Down Syndrome can present with further mental decline and increased risk for agitation as they become older. Adults with Down syndrome are at increased risk for neuro-pathological changes associated with dementia of the Alzheimer type as they become older. Agitation is a common concomitant of brain injury and can be of sufficient persistence and intensity that it requires intervention. Both pharmacological therapies (if not contraindicated from a cardiac standpoint) and behaviour modification techniques have a place in treatment. Recent onset/new mental decline in individuals with Down or other neuropsychological impairments (CNS comorbidity related to stroke or cardiac disease) may also be related to exacerbations in cardiac disease or other metabolic factors and these should be investigated.

## APPENDIX I



### ENDOCARDITIS PROPHYLAXIS RECOMMENDATIONS (1997 Dutch Heart Foundation)

#### Prophylaxis recommended in patients with

- previous endocarditis
- valve abnormalities, bicuspid aortic valve included
- mitral valve prolapse with regurgitation
- congenital heart disease (not ASD)
- coarctatio aortae
- persistent ductus arteriosus
- hypertrophic obstructive cardiomyopathy (HOCM)
- prosthetic valve
- VSD patch/outflowpatch within 6 months after implantation
- suspected residual defects after: VSD closure, valvulotomy or Waterston- and Blalock-Taussig shunts

#### Prophylaxis **not** recommended

- dental procedures not likely to cause bleeding, such as adjustment of orthodontic appliances and simple fillings above the gum line
- intra-oral injection of local anaesthetic
- functional murmur
- ASD
- mitral valve prolapse without regurgitation
- mitral annulus calcification
- ischemic heart disease
- previous coronary bypass operation
- peripheral vascular disease
- pacemaker or implantable defibrillator (AICD)
- uncomplicated delivery
- heartcatheterisation more than 6 months after valve replacement

In patients at higher-than-normal risk, such as those with previous endocarditis or prosthetic valves, prophylaxis may be chosen even for these procedures, according to clinical judgement.

#### Procedures for which prophylaxis is recommended

1. dental/oral/upper respiratory procedures
- 2a. surgical genitourinary/gastrointestinal procedures
  - b. diagnostic and instrumental procedures in infected genitourinary tract
  - c. diagnostic procedures and biopsy in gastrointestinal tract in patients with a prosthetic valve
3. procedures in infected tissue

### **1.Dental/Oral/Upper respiratory procedures**

AMOXICILLIN 3.0 g orally 1 hour before procedure  
or bicillin 1,2 million U i.m.

***Amoxicillin/Penicillin allergy or within 7 days after treatment with penicillin***

CLINDAMYCIN 600 mg orally 1 hour before procedure

### **2.Genitourinary/Gastrointestinal procedures**

AMOXICILLIN 1g i.v. 30 – 60 min before procedure

***Plus***

GENTAMICIN 3 mg/kg im\* or i.v. 30 - 60 min before procedure

followed by Amoxicillin 1g i.v. 6 hours after procedure

***Penicilin/Ampicillin allergy***

VANCOMYCIN 1g i.v. infused SLOWLY over 1 hour beginning 1 hour before  
procedure.

***Plus***

GENTAMICIN 3 mg/kg im\* or i.v. 30 – 60 min before procedure

### **3.Procedures in infected tissues**

FLUCLOXACILLIN 2 g orally 30 – 60 min before procedure.  
followed by flucloxacillin 1 g orally 6 hours after procedure

***Penicillin allergy***

CLINDAMYCIN 600 mg orally 1 hour before procedure

\*im doses should not be given if on anticoagulants

## ***APPENDIX II***

| <b>Level of Evidence</b>   | <b>Grade of Recommendation</b> |
|--|--------------------------------|
| Level I: Large randomized trials with clear-cut results, and low risk of error           | A                              |
| Level II: Randomized trials with uncertain results and/or moderate to high risk of error | B                              |
| Level III: Nonrandomized studies with contemporaneous controls                           | C                              |
| Level IV: Nonrandomized studies with historical controls                                 | C                              |
| Level V: Case series without controls  | C                              |

## ***APPENDIX III***

### **GLOSSARY OF TERMS**

#### **Abbreviations used in the text**

|          |   |
|----------|---|
| ACHD     | Adult congenital heart disease              |
| ASD      | Atrial septal defect                        |
| AV       | Atrio-ventricular                           |
| AVSD     | Atrio-ventricular septal defect             |
| CACH     | Canadian Adult Congenital Heart [Network]   |
| CBC      | Complete blood count                        |
| CCA VF   | Congenital coronary arteriovenous fistula   |
| CCS      | Canadian Cardiovascular Society             |
| CHD      | Congenital heart disease                    |
| CT       | Computed tomography                         |
| CT ratio | Cardio-thoracic ratio                       |
| CVA      | Cerebrovascular accident                    |
| DORV     | Double outlet right ventricle               |
| ECG      | Electrocardiogram                           |
| EPS      | Electrophysiologic study                    |
| GI       | Gastrointestinal                            |
| GU       | Genitourinary                               |
| ICU      | Intensive care unit                         |
| IVC      | Inferior vena cava                          |
| LCA      | left coronary artery                        |
| LV       | left ventricle                              |
| LVH      | left ventricular hypertrophy                |
| LVOTO    | left ventricular outflow tract obstruction  |
| MCHC     | Mean corpuscular hemoglobin concentration   |
| MCV      | Mean corpuscular volume                     |
| MRI      | Magnetic resonance imaging                  |
| NYHA     | New York Heart Association                  |
| PA       | Pulmonary artery                            |
| PAP      | Pulmonary artery pressure                   |
| PDA      | Patent ductus arteriosus                    |
| PFO      | Patent foramen ovale                        |
| PLE      | Protein-losing enteropathy                  |
| PS       | Pulmonary stenosis                          |
| PVOD     | Pulmonary vascular obstructive disease      |
| Qp       | Pulmonary blood flow                        |
| Qs       | Systemic blood flow                         |
| RCA      | Right coronary artery                       |
| RV       | Right ventricle                             |
| RVOT     | Right ventricular outflow tract             |
| RVOTO    | Right ventricular outflow tract obstruction |
| SABP     | Systemic arterial blood pressure            |
| SVC      | Superior vena cava                          |
| SVT      | Supraventricular tachycardia                |
| TEE      | Transesophageal echocardiogram              |

|     |                              |
|-----|------------------------------|
| TIA | Transient ischemic attack    |
| TR  | Tricuspid regurgitation      |
| TTE | Transthoracic echocardiogram |
| TV  | Tricuspid valve              |
| VSD | Ventricular septal defect    |

## Definitions of terms used in the text

|                            |   |
|----------------------------|---|
| AV septal defect           | See atrio-ventricular septal defect   |
| AV valve                   | See atrio-ventricular valve   |
| Arteriohepatic dysplasia   | Syn.: Alagille syndrome. An hereditary syndrome consisting of intrahepatic cholestasis, characteristic facies, butterfly-like vertebral anomalies and varying degrees of peripheral pulmonary artery stenoses or diffuse hypoplasia of the pulmonary artery and its branches. Associated with deletion of chromosome 20p.   |
| Atrio-ventricular valve    | The valve guarding the inlet to the ventricle. AV valves correspond with their respective ventricles; the tricuspid valve with the right ventricle, the mitral valve with the left ventricle. However, in the setting of an AVSD, there is neither a true mitral nor a true tricuspid valve. Rather, there is a single atrio-ventricular orifice, guarded by a 5-leaflet AV valve. The “left AV valve” comprises the left lateral leaflet and the left portions of the superior (anterior) and inferior (posterior) bridging leaflets, while the “right AV valve” comprises the right inferior leaflet, the right antero-superior leaflet, and the right portions of the superior and inferior bridging leaflets. |
| - common                   | Describes a 5-leaflet AV valve in complete AVSD which is related to both ventricles.  |
| - straddling               | Describes an AV valve with anomalous insertion of tendinous chords or papillary muscles into the contralateral ventricle (VSD required).  |
| - cleft                    | A defect often involving the left AV valve in AVSD formed by the conjunction of the superior and inferior bridging leaflets. May also be seen in the septal tricuspid leaflet. A similar but separate entity may involve the anterior or rarely posterior leaflet of the mitral valve in otherwise normal hearts.   |
| Atrial septal defect (ASD) | An interatrial communication, classified according to its location relative to the oval fossa:  |
| - ostium primum ASD        | Part of the spectrum of AVSD. Located anterior to the oval fossa; often in association with a “cleft” in the “anterior” mitral leaflet.   |
| - ostium secundum ASD      | Located at the level of the oval fossa.   |
| - sinus venosus ASD        | Postero-superior (or rarely postero-inferior) to the oval fossa, often associated with anomalous pulmonary venous drainage (commonly right pulmonary veins, especially the right upper pulmonary vein with the superior defect).  |
| - coronary sinus ASD:      | Inferior and slightly anterior to the oval fossa located at the anticipated site of the orifice of the coronary sinus. May be part of a complex anomaly including absence of the coronary sinus and a persistent left superior vena cava.   |
| Atrio-ventricular septum   | The atrio-ventricular septum separates the <u>left</u> ventricular inlet  |

from the right atrium. It has two parts: a muscular portion which exists because the attachment of the septal leaflet of the tricuspid valve is more towards the apex of the ventricle than the corresponding attachment of the mitral valve; and a fibrous portion superior to the attachment of the septal leaflet of the tricuspid valve. This latter part separates the right atrium from the sub-aortic left ventricular outflow tract.

Atrio-ventricular septal defect (AVSD)

A group of conditions resulting from a deficiency of the atrio-ventricular septum. They have in common: 1) a common atrio-ventricular junction with a common fibrous ring and a unique, 5-leaflet AV valve; 2) unwedging of the aorta from its usual position deeply wedged between the mitral and tricuspid valves; 3) a narrowed subaortic outflow tract; 4) disproportion between the inlet and outlet portions of the ventricular septum. A spectrum of anomalies is found, including those previously known as (and often still described as) ostium primum ASD (partial AVSD), “cleft” anterior mitral and/or septal tricuspid valve leaflet, inlet VSD, and complete AVSD (“complete AV canal defect”). Another older term describing these defects is “endocardial cushion defects”.

Baffle

A structure surgically created to divert blood flow. In the case of the Mustard operation for complete transposition of the great vessels, an intra-atrial baffle of pericardium is constructed to divert systemic venous return across the mitral valve thence to LV and pulmonary artery, and pulmonary venous return across the tricuspid valve thence to RV and aorta.

Balanced

As in “balanced circulation”, e.g. in the setting of VSD and pulmonary stenosis. The stenosis is such that there is neither excessive pulmonary blood flow (leading to pulmonary hypertension) nor inadequate pulmonary blood flow (leading to excessive cyanosis)

Blalock-Taussig shunt

A palliative operation for the purpose of increasing pulmonary blood flow, hence systemic oxygen saturation. It involves creating an anastomosis between a subclavian artery and the ipsilateral pulmonary artery either directly with an end-to-side anastomosis (classical) or using an interposition tube graft (modified).

Bridging leaflets

The superior and the inferior bridging leaflets of the AV valve are two leaflets uniquely found in association with AVSD. They “bridge”, or pass across, the interventricular septum. When the central part of the bridging leaflet tissue runs *within* the interventricular septum, the AV valve is functionally separated into left and right components; when the bridging leaflets do not run within the interventricular septum, but *pass over its crest*, a common AV valve guarding a common AV orifice (with an obligatory VSD) is the result.

Brock procedure

A palliative operation to increase pulmonary blood flow and reduce right-to-left shunting in tetralogy of Fallot. It involves resection of part of the RV infundibulum so as to reduce RVOTO, without VSD closure.

|  |   |
|--|---|
| CACH (Canadian Adult Congenital Heart) Network             | A co-operative nation-wide association of Canadian cardiologists, many of whom are situated in regional referral centres for adult congenital heart disease, dedicated to improving the care of ACHD patients.  |
| Cleft AV valve   | See atrio-ventricular valve.  |
| Common (as in: AV valve, atrium, ventricle, etc.)          | Implies bilateral structures with absent septation. Contrasts with “single”, which implies absence of corresponding contralateral structure.  |
| Complete transposition of the great arteries               | (syn.: d-transposition; d-TGA; atrio-ventricular concordance with ventriculo-arterial discordance). An anomaly wherein the aorta arises from the right ventricle and the pulmonary artery from the left ventricle.  |
| Concordant atrio-ventricular connection                    | Appropriate connection of morphologic right atrium to morphologic right ventricle and of morphologic left atrium to morphologic left ventricle.   |
| Concordant ventriculo-arterial connection                  | Appropriate origin of pulmonary trunk from morphologic right ventricle and of aorta from morphologic left ventricle.  |
| Connection   | Anatomic link between two structures (e.g.: veno-atrial, atrio-ventricular, ventriculo-arterial).   |
| Conduit  | A structure which connects parts of the cardiovascular system, allowing blood to flow between them. Often fashioned from prosthetic material. May or may not be valved.   |
| Congenital coronary arteriovenous fistula (CCAVF)          | A communication between a coronary artery and cardiac chamber, great artery or vena cava. May vary in size and number.  |
| Congenital heart disease                                   | Anomalies of the heart originating in fetal life. Their expression may, however, be delayed beyond the neonatal period, and may change with time as further post-natal physiologic and anatomic changes occur.  |
| Congenitally corrected transposition of the great arteries | (syn.: l-transposition; l-TGA; atrio-ventricular discordance with ventriculo-arterial discordance). An anomaly wherein the aorta arises from the right ventricle and the pulmonary artery from the left ventricle and, in addition, the atrio-ventricular connection is discordant such that the right atrium connects to the left ventricle and the left atrium connects to the right ventricle. There are usually associated anomalies, the most common being VSD, pulmonic stenosis, hypoplastic ventricle. The right ventricle supports the systemic circulation. |
| Conus  | see infundibulum  |
| Cyanosis   | a bluish discolouration due to the presence of an increased quantity of desaturated hemoglobin in tissues. In congenital heart  |

|  |  |
|--|--|
|  | disease generally due to right-to-left shunting through congenital cardiac defects, bypassing the pulmonary alveoli; or due to acquired intrapulmonary shunts (central cyanosis). It can also occur due to increased peripheral extraction due, for instance, to reduced cardiac output (peripheral cyanosis).   |
| Dacron   | a synthetic material often used to fashion conduits and other prosthetic devices for the surgical palliation or repair of congenital heart disease.  |
| Differential hypoxemia;<br>differential cyanosis | a difference in the degree of hypoxemia/cyanosis in different extremities due to the site of a right-to-left shunt. The most common occurrence is of greater hypoxemia/cyanosis in feet and sometimes left hand, as compared to right hand and head, in a patient with an Eisenmenger PDA.   |
| Discordant atrio-ventricular connection          | Inappropriate connection of atria and ventricles such that the morphologic right atrium connects via a mitral valve to a morphologic left ventricle, and the morphologic left atrium connects via a tricuspid valve to a morphologic right ventricle.  |
| Discordant ventriculo-arterial connection        | Inappropriate connection of the great arteries and ventricles such that the pulmonary trunk arises from the left ventricle and the aorta arises from the right ventricle.  |
| Double-chambered RV                              | Separation of the right ventricle into a higher pressure inflow chamber, and a lower pressure infundibular chamber; the separation usually being produced by hypertrophy of the “septomarginal band”. When a VSD is present, it usually communicates with the high pressure RV inflow chamber.   |
| Doubly-committed VSD                             | see ventricular septal defect.   |
| Ebstein anomaly                                  | an anomaly of the tricuspid valve in which the basal attachments of the septal (and sometimes the postero-lateral) leaflet(s) are displaced apically within the RV. Abnormal structure of all three leaflets is seen, with the anterior leaflet typically large, with abnormal attachments to the ventricular wall. Tricuspid regurgitation is common, tricuspid stenosis occurs occasionally, and right-to-left shunting through a patent foramen ovale or atrial septal defect is a regular but not invariable concomitant. Other congenital lesions are often associated, such as VSD, PS, accessory conduction pathways. |
| Eisenmenger syndrome                             | an extreme form of pulmonary vascular obstructive disease arising as a consequence of pre-existing systemic-to-pulmonary shunt, wherein pulmonary vascular resistance rises such that pulmonary pressures are at or near systemic levels and there is reversed (right-to-left) or bi-directional shunting at great vessel, ventricular, and/or atrial levels.  |

|                              |  |
|------------------------------|--|
| Erythrocytosis               | Increase in red blood cell concentration secondary to chronic Hypoxemia.   |
| Fontan procedure (operation) | a palliative operation for patients with a univentricular circulation, involving diversion of the systemic venous return to the pulmonary artery usually without the interposition of a sub-pulmonary ventricle. There are many variations, all leading to improvement or normalization of systemic oxygen saturation and elimination of volume overload of the functioning ventricle. |
| Glenn shunt (operation)      | a palliative operation for the purpose of increasing pulmonary blood flow, hence systemic oxygen saturation, in which an anastomosis is created between the superior vena cava and a pulmonary artery (usually right pulmonary artery). Acquired pulmonary arterio-venous malformations, with systemic arterial desaturation, are a long term complication.                            |
| - classic Glenn              | Anastomosis of the SVC to the distal end of the <u>divided</u> pulmonary artery with division/ligation of the SVC below the anastomosis.   |
| - bi-directional Glenn       | end-to-side anastomosis of the divided SVC to the <u>undivided</u> pulmonary artery  |
| Goretex                      | a synthetic material often used to fashion conduits and other prosthetic devices for the surgical palliation or repair of congenital heart disease.  |
| Heath-Edwards classification | a histopathologic classification, graded from I (hypertrophy of media of muscular arterioles) to VI (necrotizing arteritis), useful in assessing potential for reversibility of pulmonary vascular disease.  |
| Heterotopic                  | in transplantation refers to placement of an organ/tissue in an anatomically abnormal recipient site.  |
| Hyperviscosity               | an excessive increase in viscosity of blood, as may occur with erythrocytosis in cyanotic congenital heart disease, leading to symptoms such as headache, dizziness, fatigue, tinnitus, bleeding, and thrombosis.  |
| Ilbawi approach              | an operation for congenitally corrected transposition of the great arteries with VSD and PS, wherein a communication is established between the LV and the aorta via the VSD using a baffle within the RV. The RV is connected to the pulmonary artery using a valved conduit. An atrial switch procedure is done. The left ventricle then supports the systemic circulation.          |
| Infundibulum, infundibular   | (pertaining to) a ventricular-great arterial connecting segment. Normally sub-pulmonary, but can be sub-aortic, and may be absent. Also called the conus.  |
| Inlet VSD                    | see ventricular septal defect  |

|                                  |   |
|----------------------------------|---|
| Jatene procedure (operation)     | (syn.: arterial switch procedure). An operation used in complete transposition of the great arteries, involving removal of the aorta from its attachment to the RV, and of the pulmonary artery from the LV, and the reattachment of the great arteries to the contralateral ventricles, with reimplantation of the coronary arteries into the neo-aorta. The left ventricle supports the systemic circulation. |
| Konno procedure (operation)      | repair of tunnel-like subvalvar LVOTO by aortoventriculoplasty. The operation involves enlargement of the LV outflow tract by inserting a patch in the ventricular septum, as well as aortic valve replacement and enlargement of the ascending aorta.  |
| Ligamentum arteriosum            | a fibrous structure representing the ductus arteriosus after its spontaneous closure.   |
| Muscular VSD                     | see ventricular septal defect.  |
| Mustard procedure (operation)    | an operation for complete transposition of the great arteries, in which venous return is directed to the contralateral ventricle by means of an atrial baffle made from autologous pericardial tissue or (rarely) synthetic material, after resection of most of the atrial septum. The right ventricle supports the systemic circulation.  |
| Non-restrictive VSD              | see ventricular septal defect.  |
| Noonan syndrome                  | an autosomal dominant syndrome phenotypically somewhat similar to Turner syndrome, but with normal chromosomal complement, associated with congenital cardiac anomalies, especially dysplastic pulmonic stenosis, hypertrophic cardiomyopathy, ASD.   |
| Orthotopic                       | in transplantation refers to placement of an organ/tissue in an anatomically normal recipient site.   |
| Ostium primum ASD                | see atrial septal defect.   |
| Outlet VSD                       | see ventricular septal defect.  |
| Overriding valve                 | an AV valve which empties into both ventricles or a semilunar valve which originates from both ventricles.  |
| Palliation, palliative Operation | a procedure carried out for the purpose of relieving symptoms or ameliorating some of the adverse effects of an anomaly, which does not address the fundamental anatomic/physiologic disturbance. Contrasts with “repair” or a “reparative operation”.  |
| Partial AV septal defect         | see atrio-ventricular septal defect.  |
| Patent ductus arteriosus (PDA)   | (syn.: persistently patent ductus arteriosus, persistent arterial duct) A ductus which fails to undergo normal closure in the early   |

|                                  |   |
|----------------------------------|---|
|                                  | post-natal period.  |
| Perimembranous VSD               | see ventricular septal defect.  |
| Pentalogy of Fallot              | see tetralogy of Fallot.  |
| Pink tetralogy of Fallot         | see tetralogy of Fallot.  |
| Potts shunt                      | a palliative operation for the purpose of increasing pulmonary blood flow, hence systemic oxygen saturation. Involves creating a small communication between a pulmonary artery and the ipsilateral descending thoracic aorta. Often complicated by the development of pulmonary vascular obstructive disease if too large, or loss of the pulmonary artery if too small and distortion occurs.                 |
| Protein-losing enteropathy (PLE) | a complication seen following the Fontan operation in which protein is lost via the gut, manifesting ascites, peripheral edema, pleural and pericardial effusions. It is of unknown cause, though exacerbated by high systemic venous pressure.   |
| Pulmonary hypertension           | Elevated pulmonary arterial pressure, sometimes defined for clinical purposes as pulmonary artery pressure > 2/3 systemic artery blood pressure or pulmonary arteriolar resistance > 2/3 systemic arteriolar resistance.  |
| Rastelli procedure (operation)   | an operation for repair of complete transposition of the great arteries in association with a large VSD and pulmonic stenosis, wherein a communication is established between the LV and the aorta via the VSD using a baffle within the RV. The RV is connected to the pulmonary artery using a valved conduit, and the LV-PA connection is obliterated. The left ventricle supports the systemic circulation. |
| Restrictive VSD                  | see ventricular septal defect   |
| RV infundibulum                  | (syn.: RV conus) a connecting segment between the body of the RV and the pulmonary artery.  |
| Senning procedure (operation)    | an operation for complete transposition of the great arteries, in which venous return is directed to the contralateral ventricle by means of an atrial baffle fashioned in situ by using right atrial wall and interatrial septum. The right ventricle supports the systemic circulation.   |
| Shone complex (syndrome)         | an association of multiple levels of left ventricular inflow and outflow obstruction [subvalvar LVOTO, coarctation of the aorta and mitral stenosis (parachute mitral valve and supramitral ring)].   |
| Shunt                            | Congenitally abnormal or surgically created connection and communication between two circuits (at the level of the atria, ventricles, or great vessels).  |

|                             |   |
|-----------------------------|---|
| Sinus venosus ASD           | see ASD.  |
| Straddling AV valve         | see atrio-ventricular valve.  |
| Subpulmonary ventricle      | the ventricle which relates most directly to the pulmonary artery.  |
| Systemic AV valve           | the atrio-ventricular valve guarding the inlet to the systemic ventricle.   |
| Tetralogy of Fallot         | a congenital anomaly, the primary pathophysiologic components of which are obstruction to right ventricular outflow at the infundibular level and a large non-restrictive VSD. The other two components of the “tetralogy” are an over-riding aorta and concentric right ventricular hypertrophy. Valvar RVOTO (pulmonic stenosis) may also be present.) The essential morphogenetic anomaly is malalignment of the infundibular (outlet) septum such that it fails to unite with the trabecular septum (hence the VSD) due to anterior deviation (hence the RV outflow tract obstruction). |
| - pentalogy of Fallot       | tetralogy of Fallot with an associated ASD or PFO   |
| - pink tetralogy of Fallot  | (syn.: acyanotic Fallot.) Tetralogy of Fallot presenting with increased pulmonary blood flow and minimal or no cyanosis because of a lesser degree of RVOT obstruction  |
| Trabecular VSD              | see ventricular septal defect.  |
| Transannular                | crossing the annulus. In connection with the RV outflow tract in tetralogy of Fallot, the term refers to the pulmonary valve annulus, which often must be enlarged by a transannular patch, with consequent obligatory pulmonary insufficiency.   |
| Tricuspid atresia           | a congenital anomaly in which there is no physiologic or gross morphologic connection between the right atrium and right ventricle and there is an interatrial connection allowing mixing of systemic and pulmonary venous return at the atrial level. There is varying hypoplasia of the RV. The LV and mitral valve are normal.   |
| Turner syndrome             | 45 X0 karyotype with characteristic but variable phenotype, and association with congenital cardiac anomalies, especially post-ductal coarctation of the aorta and other left-sided obstructive lesions, as well as partial anomalous pulmonary venous drainage without ASD.  |
| Univentricular connection   | both atria are connected to only one ventricle. Note: the connection is univentricular, not the heart.  |
| Velo-cardio-facial syndrome | (syn.: Shprintzen syndrome.) Syndrome of cleft palate, abnormal facies (square nasal root, long nose with narrow alar base, long face with malar hypoplasia, long philtrum, thickened helix or low-set ears), velopharyngeal incompetence and congenital cardiac  |

|                                 |  |
|---------------------------------|--|
|                                 | defects (conotruncal anomalies, isolated VSD, tetralogy of Fallot). Associated with chromosome 22q11 deletion.   |
| Venous (or pulmonary) AV valve  | the AV valve guarding the inlet to the venous, or pulmonary, ventricle.  |
| Ventricular septal defect (VSD) | a defect in the ventricular septum, such that there is direct communication between the two ventricles.  |
| - doubly-committed VSD          | a defect in the outlet septum such that there is fibrous continuity between the aortic and pulmonary valves, with the VSD situated directly beneath both semi-lunar valves.  |
| - inlet VSD                     | a defect in the lightly trabeculated inlet portion of the muscular interventricular septum.  |
| - muscular VSD                  | a defect entirely enclosed within the muscular septum.   |
| - non-restrictive VSD           | a ventricular septal defect of such a size that there is no significant pressure gradient between the ventricles. Hence the pulmonary ventricle is exposed to systemic pressure.   |
| - outlet VSD                    | a defect in the non-trabeculated outlet portion of the muscular interventricular septum, hence above the crista ventricularis. Syn.: supracristal VSD. Sometimes also described as subpulmonary, subarterial, or doubly committed subarterial VSD.   |
| - perimembranous VSD            | (syn.: membranous VSD; infracristal VSD) a VSD located in the membranous portion of the interventricular septum with variable extension into the contiguous portions of the inlet, trabecular, or outlet portions of the muscular septum, but not involving the atrio-ventricular septum.  |
| - restrictive VSD               | a ventricular septal defect of small enough size that there is a pressure gradient between the ventricles, such that the pulmonary ventricle is protected completely or partially from the systemic pressure of the contralateral ventricle.   |
| - trabecular VSD                | a defect in the heavily trabeculated central or trabecular portion of the muscular interventricular septum. May be multiple.   |
| Waterston shunt                 | A palliative operation for the purpose of increasing pulmonary blood flow, hence systemic oxygen saturation. Involves creating a small communication between the main pulmonary artery and the ascending aorta. Often complicated by the development of pulmonary vascular obstructive disease if too large. Not uncommonly caused distortion of the pulmonary artery. |
| Williams syndrome               | A congenital syndrome of heterogeneous cause, often sporadic, occasionally autosomal dominant, associated with infantile hypercalcemia, characteristic phenotype, and congenital heart disease, especially supravalvar aortic stenosis and multiple peripheral pulmonary stenoses.   |

## ***APPENDIX IV***

### **Membership list of the 1996 Consensus Conference on Adult Congenital Heart Disease**

#### **primary panel**

Dr. Ruth Collins-Nakai  
Dr. Michael Connely  
Dr. John Finley

#### **secondary panel**

Dr. Nanette Alvarez  
Mr. Tim Caley  
Dr. Georges Delisle

Dr. Victor Huckell  
Dr. Richard Liberthson  
Dr. Lise-Andree Mercier  
Dr. Joseph Perloff  
Dr. F.J. Puga  
Dr. Jane Somerville  
Dr. Carole Warnes  
Dr. Gary Webb  
Dr. W.G. Williams

Dr. John Fulop  
Dr. Peter Liu  
Dr. Francois Marcotte  
Dr. Roxane McKay  
Dr. Brian Morton  
Dr. Michael Patterson  
Dr. David Ross  
Dr. Andrew Weeks  
Dr. Anne Williams

### **Membership list of the Dutch workinggroup “Congenitale Cardiologie bij Volwassenen”**

Dr. Ad Backx  
Dr. Ger Benninck  
Dr. Ad Bogers  
Dr. Eli Broekhuis  
Dr. Johan Brügemann  
Dr. Werner Budts  
Dr. Pieter van der Burgh  
Dr. Tjark Ebels  
Dr. Nienke Elzenga  
Dr. Henry de Haan  
Dr. Hans Hamer  
Dr. Mark Hazekamp

Dr. Jan Hoorntje  
Dr. Theo Hoorntje  
Dr. Lilian Jeekel  
Dr. Paul de Jong  
Dr. Folkert Meijboom  
Dr. Huub Meijburg  
Dr. Gert Jan van Mill  
Dr. Peter Molhoek  
Dr. Barbara Mulder  
Dr. Aagje Nijveld  
Dr. Kathinka Peels  
Dr. Els Pieper

Mrs. Ivonne Plekkenpol  
Dr. Thijs Plokker  
Dr. Hans Romkes  
Dr. Jolien Roos-Hesselink  
Dr. Paul Schoof  
Dr. Silja Spitaels  
Dr. Jan Stappers  
Dr. Gerrit Veen  
Dr. Jaap Visser  
Dr. Hubert Vliegen  
Dr. Tjalling Waterbolk  
Dr. Jan van Wijngaarden

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