Canadian Cardiovascular Society 2009 Consensus Conference on the management of adults with congenital heart disease: Outflow tract obstruction, coarctation of the aorta, tetralogy of Fallot, Ebstein anomaly and Marfan's syndrome

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With advances in pediatric cardiology and cardiac surgery, the population of adults with congenital heart disease (CHD) has increased. In the current era, there are more adults with CHD than children. This population has many unique issues and needs. Since the 2001 Canadian Cardiovascular Society Consensus Conference report on the management of adults with CHD, there have been significant advances in the field of adult CHD. Therefore, new clinical guidelines have been written by Canadian adult CHD physicians in collaboration with an international panel of experts in the field. Part II of the guidelines includes recommendations for the care of patients with left ventricular outflow tract obstruction and bicuspid aortic valve disease, coarctation of the aorta, right ventricular outflow tract obstruction, tetralogy of Fallot, Ebstein anomaly and Marfan's syndrome. Topics addressed include genetics, clinical outcomes, recommended diagnostic workup, surgical and interventional options, treatment of arrhythmias, assessment of pregnancy risk and follow-up requirements. The complete document consists of four manuscripts that are published online in the present issue of *The Canadian Journal of Cardiology*. The complete document and references can also be found at www.ccs.ca or www.cachnet.org.

Key Words: *Adult congenital heart disease; Bicuspid aortic valve; Coarctation of the aorta; Congenital heart disease; Ebstein anomaly; Marfan's syndrome; Outflow tract obstruction; Tetralogy of Fallot*

La conférence consensuelle 2009 de la Société canadienne de cardiologie sur la prise en charge des adultes ayant une cardiopathie congénitale : L'obstruction de la chambre de chasse, la coarctation de l'aorte, la tétralogie de Fallot, la maladie d'Ebstein et le syndrome de Marfan

Étant donné les progrès de la cardiologie pédiatrique et de la chirurgie cardiaque, la population d'adultes ayant une cardiopathie congénitale (CPC) a augmenté. Il y a maintenant plus d'adultes que d'enfants ayant une CPC. Cette population a de nombreux problèmes et besoins uniques. Depuis le rapport de la conférence consensuelle 2001 de la Société canadienne de cardiologie sur la prise en charge des adultes ayant une CPC, on constate d'importantes avancées dans le domaine des CPC chez les adultes. Par conséquent, de nouvelles lignes directrices cliniques ont été rédigées par des médecins canadiens s'occupant des CPC chez les adultes, en collaboration avec un groupe d'experts internationaux dans le domaine. La partie II des lignes directrices contient des recommandations sur les soins aux patients ayant une obstruction de la chambre de chasse du ventricule gauche et une bicuspidie valvulaire aortique, une coarctation de l'aorte, une obstruction de la chambre de chasse du ventricule droit, une tétralogie de Fallot, une maladie d'Ebstein et un syndrome de Marfan. Les sujets abordés incluent la génétique, les issues cliniques, les bilans diagnostiques recommandés, les possibilités chirurgicales et d'intervention, le traitement des arythmies, l'évaluation des risques de la grossesse et de la contraception et les recommandations de suivi. Le document complet se compose de quatre manuscrits publiés par voie électronique dans le présent numéro du *Journal canadien de cardiologie*. Le document complet et les références figurent également aux adresses www.ccs.ca et www.cachnet.org.

LEFT VENTRICULAR OUTFLOW TRACT OBSTRUCTION

Part 1. Background information

The present section concerns left ventricular outflow tract obstruction (LVOTO) in the setting of concordant atrioventricular and ventriculoarterial connections. (Neither hypertrophic cardiomyopathy nor interrupted aortic arch will be considered here.)

LVOTO can occur at several levels:

- Supravalvar LVOTO may occur rarely in isolation as an hourglass deformity. However, it is more often diffuse, involving the major arteries to varying degrees, and begins at the superior margin of the sinuses of Valsalva. The origin of the coronary arteries is usually proximal to the obstruction.
- Valvar LVOTO in the adult patient with congenital heart disease

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(CHD) is usually due to bicuspid aortic valve (BAV) (rheumatic and trileaflet calcific aortic stenosis are not discussed here). It usually occurs in isolation but is associated with other abnormalities, the most common being coarctation of the aorta, patent ductus arteriosis (PDA) or ascending aortopathy.

- Subvalvar LVOTO is usually either a discrete fibromuscular ridge, which partially or completely encircles the left ventricular (LV) outflow tract, or is a long fibromuscular narrowing beneath the base of the aortic valve. Occasionally, there is a tunnel-like narrowing of the whole LV outflow tract, with a small aortic root. Rarely, abnormal insertion of the mitral valve or accessory mitral leaflet may cause significant obstruction.

The concurrence of both LV inflow tract obstruction (including supravalvar mitral ring or parachute mitral valve) and LVOTO (including subvalvar LVOTO, BAV and aortic coarctation) is known as Shone's syndrome.

Part II. Prevalence and genetics

Supravalvar LVOTO is usually part of Williams syndrome, which is an autosomal dominant contiguous gene deletion syndrome. It is associated with neurodevelopmental and multisystem manifestations, and is caused by a microdeletion on 7q11.23, which encompasses the elastin gene (1,2). The incidence of Williams syndrome is estimated to be one in 10,000 births. It is characterized by cardiac defects, infantile hypercalcemia, skeletal and renal anomalies, cognitive deficits, 'social' personality and elfin facies. It is readily detectable by fluorescence in situ hybridization (FISH) in 99% of cases. Familial supravalvar LVOTO may be associated with point mutations in the elastin gene in the absence of other features of Williams syndrome, or with the fetal rubella syndrome.

BAV is the most common congenital cardiac anomaly, occurring in 1% to 2% of the population, with a male predominance (4:1 ratio). BAV is sometimes inherited as an autosomal dominant trait with variable penetrance. BAV may be associated with chromosome abnormalities, Noonan syndrome, Turner syndrome and Williams syndrome. Aortic atresia can be associated with a deletion of 11q (Jacobsen syndrome), Turner syndrome, trisomy 13, trisomy 18, or a deletion of 4p (Wolf-Hirschhorn syndrome).

Subvalvar LVOTO also has a male predominance (2:1 ratio). In some instances, particularly in Shone's syndrome, the condition may be familial (3).

Part III. History and management of unoperated patients

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

Valvar LVOTO commonly progresses as the patient ages, but the rate is variable. Some patients with BAV will not experience any related problem, although there is a lifelong risk of endocarditis. Others will develop aortic stenosis (especially after calcification of the valve in the sixth decade) (4,5), aortic regurgitation, aortic dissection or aneurysmal aortic root dilation (irrespective of altered hemodynamics or age) due to aortic medial abnormalities (6-9).

Subvalvar LVOTO often progresses, but the rate is variable; low gradients may remain for many years (10,11), seldom becoming more than moderate. It is often associated with aortic regurgitation (up to 60% of cases) through an otherwise normal valve, which has been damaged by the subvalvar jet of blood (12). There may be associated small ventricular septal defects (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 including VSDs, atrioventricular septal defects (AVSDs) or conotruncal abnormalities, and may develop after repair of a VSD or AVSD (13).

Part IV. Diagnostic workup

An adequate initial workup should include the following:

- Documentation of the level(s) of obstruction.
- Quantification of the severity and anatomy of the obstruction(s).
- Identification of associated abnormalities including aortic regurgitation, proximal aortic root dilation, aortic coarctation and the associated anomalies of Williams and Shone's syndromes.

The diagnostic workup should include the following (at a minimum):

- A thorough clinical assessment.
- Electrocardiogram (ECG).
- Chest x-ray.
- Transthoracic echocardiogram (TTE)-Doppler examination by an appropriately trained individual to determine the level(s) of obstruction, septal thickness, the size of aortic root, the ascending aorta and associated abnormalities.

The diagnostic workup may require the following:

- Transesophageal echocardiogram (TEE) to define the anatomy precisely if unclear from the TTE.
- Exercise testing to determine whether there is coronary artery ischemia. Myocardial perfusion imaging with exercise/pharmacological stress or stress echocardiography (echo) may be helpful because these patients frequently have abnormal resting ECGs.
- A heart catheterization with or without provocative testing to assess the hemodynamics and severity of obstruction if echo is not definitive.
- Coronary angiography and aortography if intervention is being planned.
- Magnetic resonance imaging (MRI) and/or computed tomography (CT) to assess associated lesions (such as pulmonary artery stenoses or coarctation) and aortic dilation, to measure LV mass and function, and to identify significant renal or other arterial stenoses.
- FISH testing for microdeletion at 7q11.23 should be considered in all patients with supravalvar aortic stenosis if not tested previously, and especially if associated with additional features characteristic of Williams syndrome.

Part V. Indications for intervention/reintervention/medical therapy

1. Supravalvar LVOTO

Operative intervention is recommended for patients with supravalvar LVOTO with symptoms and/or a mean echo or a mean catheter gradient of greater than 50 mmHg or a peak instantaneous echo gradient of greater than 70 mmHg if the obstruction is discrete.
Class I, level C (14)

Criteria for intervention for diffuse obstruction are not well defined but are probably similar because the end effect on the coronary arteries and the myocardium is the same.

2. Valvar LVOTO

Valvar LVOTO requires intervention for symptoms (dyspnea, angina, presyncope or syncope) and significant left-sided outflow obstruction (mean echo gradient of greater than 40 mmHg or aortic valve area of less than 1.0 cm^2 or less than 0.6 cm^2/m^2). Gradients may be lower if there is significant LV systolic dysfunction. **(Level C)**
Patients with BAVs may also require intervention for symptoms and severe regurgitation, severe aortic regurgitation with LV end-systolic dimensions of greater than 55 mm, end-diastolic diameter of greater than 75 mm or LV ejection fraction of less than 50%. **(Level B)**

Aortic root replacement is required for ascending aortic dissection and should be considered prophylactically for proximal aortic dilation (greater than 50 mm) or progressive dilation of greater than 5 mm/year. **(Level B)**
Class I, level B or C as indicated (15-21)

Intervention may be considered for asymptomatic patients with 'critical' aortic stenosis (valve area of less than 0.6 cm^2) and/or mean Doppler gradient of greater than 60 mmHg. Intervention may be indicated occasionally for other reasons (eg, person with a lesser degree of obstruction who wishes to play vigorous sports or to become pregnant).
Class IIb, level C (20-22)

3. Subvalvar LVOTO

Intervention is indicated for patients with subvalvar LVOTO with symptoms and a peak instantaneous echo gradient of greater than 50 mmHg or a mean echo gradient of greater than 30 mmHg, or if combined with progressive aortic regurgitation. If there is an associated VSD, the gradient may be underestimated and important subvalvar LVOTO may become manifest only after VSD closure.
Class I, level C (23-30)

In adults, intervention for LVOTO is not indicated solely for the prevention of aortic regurgitation.

Reinterventions

Reoperation is indicated after valvotomy or after surgery for:
- Recurrent LVOTO (same criteria as above).
- Severe aortic regurgitation.
- Combined restenosis with moderate or greater regurgitation, especially if symptoms or progressive LV dilation are present.

Class I, level C (16-19,31)

Part VI. Interventional options

Patients who require operation for supravalvar or subvalvar LVOTO should be operated on by congenital heart surgeons with experience with the technique.
Class I, level C (32,33)

Supravalvar LVOTO requires patch aortoplasty or, rarely, replacement of the proximal ascending aorta. Reconstruction of the coronary ostia may be necessary (14). Concomitant surgery or stent placement for branch pulmonary artery stenosis may be performed. Catheter intervention for supravalvar LVOTO is not appropriate.

Valvar LVOTO may be treated with balloon valvuloplasty (if the valve is noncalcified) – especially in young adults (4) – open aortic valvotomy, or valve replacement using a mechanical valve, biological valve or pulmonary autograft (Ross procedure consists of replacing the aortic valve with the patient's pulmonary valve and implanting a homograft in the pulmonary position). The choice depends on the availability and skills of the team, and the preference of the patient.

Pulmonary autograft (Ross procedure) and balloon valvuloplasty for valvar LVOTO should be performed in centres and by physicians with substantial experience in these procedures.
Class I, level C

Discrete subvalvar LVOTO requires surgical resection almost invariably associated with myomectomy or myotomy. 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 (aortoventriculoplasty with aortic valve replacement) or other modifications for enlargement of the outflow tract. In the past, an LV 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 is unacceptable and the procedure has been abandoned. Some of these patients are still alive.

Part VII. Interventional outcomes

Supravalvar LVOTO should have a low operative mortality. Recurrence of obstruction is uncommon. 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 may be associated with progressive recurrent stenosis and calcification, and/or progressive regurgitation, and may eventually require valve replacement. Surveillance should include assessment for aortic dilation/aneurysm and endocarditis.

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

Patients with pulmonary autografts in the aortic position have excellent hemodynamic characteristics, require no anticoagulation, and have much reduced risk of thromboembolism. However, the autograft may deteriorate with time, as well as the pulmonary homograft, leading to stenosis and/or regurgitation. Progressive neoaortic root dilation and regurgitation may develop (6,34-36).

Patients with pulmonary autografts in the aortic position need careful long-term follow-up.
Class IIa, level C (6,34-36)

Recurrence of fibromuscular subvalvar LVOTO is not uncommon (up to 20% over a decade), particularly if the aortic root is small or the preoperative peak gradient is greater than 60 mmHg (30).

Tunnel-like subvalvar LVOTO with extensive repair, with or without aortic valve replacement, has a high risk of recurrence and progressive aortic regurgitation (37).

After the Konno or Ross-Konno procedure, excellent early outcomes have been reported but with recurrent subaortic stenosis as high as 86% at five years (13).

Clinically important aortic regurgitation following subvalvar LVOTO repair is not uncommon (up to 25% of patients).

Part VIII. Arrhythmias

In adults with unoperated LVOTO, high-grade ventricular ectopy is common. Repolarization anomalies and risk for sudden cardiac death may persist after surgery. Patients should be carefully monitored for the early detection of arrhythmia, particularly when symptoms arise, with resting and/or stress ECGs and periodic ambulatory rhythm recordings.

Part IX. Pregnancy and contraception

Most LVOTO lesions in women of childbearing age are due to BAV disease. Patients with mild to moderate LVOTO and normal LV function can usually be managed conservatively through the entire pregnancy. In patients with more significant obstruction or symptoms, maternal cardiac complications can occur (38-41). Global (ventricular systolic dysfunction and history of cardiac complications) and lesion-specific risks need to be taken into account when determining the risk of pregnancy (42).

Women with symptomatic LVOTO should be advised to delay conception until relief of LVOTO is accomplished. In women with suitable valves, balloon dilation of a severely stenotic bicuspid valve can be performed during pregnancy, but due to the associated risk, should only be performed when necessary (43,44). Similarly, valve surgery during pregnancy should be contemplated only for the control of refractory functional class III or IV symptoms because fetal mortality during cardiac surgery is high.

The presence of a BAV and ascending thoracic aorta dilation may predispose to further dilation or spontaneous aortic dissections. Some experts recommend beta-blocker therapy during pregnancy in this setting.

Part X. Follow-up

All patients should have regular cardiology follow-up. Patients with Williams or Shone's syndrome, and those with complex LVOTO, with or without repair, should be followed by an adult CHD (ACHD) cardiologist. Particular attention should be paid to the following:

- Progressive/recurrent stenosis at any level.
- Aortic regurgitation.
- Ventricular function and/or dilation.
- Aortic root and ascending aortic dilation.
- Specific complications after the Ross procedure include right ventricle-to-pulmonary artery conduit and pulmonary aortograft degeneration, neoaortic dilation, neoaortic valve regurgitation and coronary abnormalities.
- Risk for heart block, ventricular arrhythmias and sudden death.

Class IIa, level C

Endocarditis prophylaxis is not recommended in patients with LVOTO unless a prosthetic valve has been inserted.
Class III, level B (45)

Early diagnosis and identification of complications associated with Williams syndrome is important. This includes treatment of hypercalcemia and hypercalciuria to prevent nephrocalcinosis, screening for thyroid and renal anomalies, close blood pressure monitoring for early-onset systemic hypertension, as well as appropriate testing of other family members, and genetic and reproductive counselling.

COARCTATION OF THE AORTA

Part I. Background information

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

In the absence of an extensive collateral circulation, significant aortic coarctation can be defined as the presence of upper limb hypertension and an associated significant gradient (simultaneous pressure gradient at catheterization or upper-lower extremity limb gradient of at least 20 mmHg). However, if there is extensive collateral circulation, a significant aortic coarctation may have minimal or no pressure gradient, and the diagnosis of a 'significant coarctation' is based on evidence of significant coarctation and collateral flow by radiographic imaging (cardiac MRI or CT).

Associated cardiovascular abnormalities include the following:

- BAV.
- Intracranial aneurysms (3% to 10%) (46).
- Anomalies of the brachiocephalic circulation, such as anomalous origin of the right subclavian artery distal to the coarctation segment and involvement of the left subclavian artery in the coarctation or the arch origin of the left vertebral artery may influence stent choice for intervention.
- Collateral circulation that is both anterior (involving the internal mammary arteries) and posterior (involving the intercostal arteries).
- Aortic medial disease in the paracoarctation aorta (8), and in the ascending aorta.
- Aortic arch hypoplasia.
- VSD.
- PDA.
- Subaortic stenosis.
- Mitral valve abnormalities.

Part II. Prevalence and genetics

Coarctation of the aorta is more common in males, with a male to female ratio of 1.5:1. (47,48). It is usually sporadic, but genetic influences can play a role (10% to 15% of Turner syndrome [45,X] females have aortic coarctation) (49). It is also seen in maternal phenylketonuria syndrome and in Kabuki syndrome.

Part III. History and management of unoperated patients

There is a spectrum of disease with, at one end, severe coarctation characterized by marked anatomical narrowing, presence of collaterals, elevated gradient and significant hypertension; and at the other end, mild coarctation characterized by mild hypoplasia, absence of collaterals, little or no gradient and no hypertension.

In a patient with hemodynamically significant coarctation, presentation in adolescence or adulthood is usually with upper limb hypertension, differential arm-leg pulses, exertional leg fatigue, or an incidental murmur. Symptoms are often absent. Rarely, presentation may be with an intracerebral hemorrhage. An occasional patient may be diagnosed from the typical x-ray appearance.

The mean survival of patients with untreated aortic coarctation before widespread surgical repair and modern diagnostic method was 35 years, with 75% mortality by 46 years of age (50). Most developed systemic hypertension, typically during childhood, and ultimately, by the fifth decade of life, they had LV failure.

Death in untreated aortic coarctation is usually due to the following:

- Heart failure.
- Aortic rupture/dissection.
- Infective endarteritis/endocarditis.
- Cerebral hemorrhage.
- Premature coronary artery disease.
- Concomitant aortic valve disease (usually involving a BAV).
- Sudden cardiac death of presumed arrhythmic etiology.

Part IV. Diagnostic workup

An initial diagnostic workup should document the following:

- The location and type of aortic coarctation together with its severity.
- The presence (or absence) and severity of other intracardiac lesions (BAV, mitral valve abnormalities, subaortic stenosis, VSD).
- LV function and the presence (or absence) of LV hypertrophy.
- The presence (or absence) of other extracardiac cardiovascular anomalies such as collateral circulation, involvement of other vessels (subclavian/carotid stenoses) and associated aneurysms.
- Presence and severity of hypertension including 24 h ambulatory blood pressure monitoring when necessary.

The diagnostic workup should include the following (at a minimum):

- A thorough clinical assessment, including upper and lower limb blood pressure measurement, determination of radiofemoral pulse delay, palpation of femoral and distal pulses, and auscultation for collaterals around the scapula. The clinical assessment of the arm-leg gradient is nonstandardized and nonsimultaneous – when coarctation is suspected, gradients should be clarified invasively.
- ECG, which may show signs of LV hypertrophy with or without 'strain'.
- Chest x-ray, which may show the '3 sign' (caused by indentation of the aorta at the site of the aortic coarctation, combined with dilation before and after the coarctation) or 'rib notching' (caused by erosion of the inferior border of the posterior ribs by enlarged intercostal arteries).
- Echo Doppler evaluation by an appropriately trained individual. Some echo windows – in particular, the suprasternal arch view – may be difficult in older subjects.
- MRI to delineate the coarctation anatomy, presence of collaterals, associated vascular anomalies and flow abnormality. If MRI is not possible, CT angiography is an alternate approach.

The diagnostic workup may require the following:

- Invasive angiography with hemodynamic measurements to assess the aortic coarctation gradient, nature of the obstruction and the presence/absence of collaterals or aneurysm formation if appropriate information cannot be obtained by MRI, if MRI is

not available, or if percutaneous intervention is not planned. If percutaneous intervention is planned, angiography can be performed at the time of the procedure.
- Coronary angiography because of the increased risk of premature coronary artery disease in these patients, if a clinical indication exists, if the patient is older than 40 years of age (or younger if major coronary risk factors) or if there is any evidence of LV dysfunction.
- Cerebral MRI or CT angiography to rule out associated intracranial aneurysms.
- Chromosome analysis if Turner syndrome is suspected.

Part V. Indications for intervention/reintervention/medical therapy

All patients with significant coarctation (native or recoarctation postrepair) should be considered as candidates for treatment.
Class I, level C (50)

There is consensus that intervention is indicated for young adults with significant coarctation in the setting of hypertension. Intervention is associated, in the majority (65%) of cases, with significant improvement in blood pressure and is believed to modify the poor natural history of this lesion. However, in less straightforward cases, such as the elderly, absence of hypertension, mild coarctation and presence of significant comorbidity, decision making needs to be individualized because risks of intervention may outweigh benefits.

Part VI. Surgical/interventional options

For significant native aortic coarctation, a surgical or percutaneous approach (if the anatomy is suitable) is reasonable. The preferred approach should reflect centre expertise and patient preference.
Class I, level B (51-55)

For significant recoarctation postrepair, a percutaneous approach (if the anatomy is suitable) is the preferred initial intervention.
Class I, level B (56,57)

Surgical repair of aortic coarctation or recoarctation in adults should be performed by congenital heart surgeons with expertise in the procedure.
Class I, level B (58)

Percutaneous interventions should be performed in centres and by individuals with expertise in the procedure.
Class I, level C

Surgical repairs that have been used for coarctation of the aorta in children include the following:
- Interposition graft.
- Resection with end-to-end anastomosis (usually the preferred method for initial repair).
- Patch aortoplasty.
- Arch augmentation.
- Jump graft bypassing the aortic coarctation segment.
- Subclavian flap aortoplasty.

In the adult with native coarctation, the interposition graft, the jump graft and end-to-end anastomosis tend to be the techniques of choice.

Part VII. Surgical/interventional outcomes

Following surgical repair of simple aortic coarctation, the obstruction is usually relieved with minimal mortality (less than 1%). Mortality is higher for reoperation (59) and in adults. Recurrent coarctation is more common when initially repaired in infancy.

Complications of surgical repair include the following:
- Paraplegia due to spinal cord ischemia, which is uncommon, but recognized, particularly in patients who do not have well-developed collateral circulation.
- Rebound paradoxical hypertension in the early postoperative phase. This may be due to rebound sympathetic activation and activation of the renin-angiotensin system. It usually responds to beta blockade.
- Recurrent laryngeal nerve palsy.
- Phrenic nerve injury with diaphragmatic paralysis.
- Aneurysm and pseudoaneurysm formation (60,61).
- Arm claudication (rare) if subclavian flap aortoplasty has been used.

Stenting in many centres has replaced balloon dilation as the percutaneous intervention of choice in the adult with coarctation of the aorta. Suboptimal anatomy for percutaneous interventions includes long segments, vessel tortuosity and transverse arch hypoplasia (62). These appear to be less of an issue with covered stents. Although only early to midterm data are available for stenting in adults, in well-selected patients, procedural success at relieving obstruction is very good, with very low mortality and morbidity (53-55,62). Percutaneous intervention is the treatment of choice in recoarctation postrepair in which the presence of scar tissue may protect from aneurysm formation (56,57). In patients with native coarctation, there is more concern regarding aneurysmal formation following the procedure. Predilation and balloon oversizing contribute to aneurysm formation, and should be avoided.

Complications of stenting include the following:
- Recoarctation.
- Pseudoaneurysm formation.
- Femoral artery injury/thrombosis.
- Stroke (rare).
- Aortic rupture (rare).

Hemoptysis from a leaking/ruptured aneurysm into a bronchus is a life-threatening complication, and requires immediate investigation and treatment.

Long-term follow-up after surgical repair has shown an increased incidence of premature cardiovascular disease and death (63).

In many patients, hypertension resolves in the early postrepair period. However, with long-term follow-up, patients are at an increased risk of hypertension (64). Recoarctation must be ruled out first. However, in the majority of cases, there is no residual gradient and the cause of hypertension is believed to be multifactorial, including arch hypoplasia, abnormal aortic compliance, vascular stiffness and abnormal baroreceptor function (65).

Late development of LV dysfunction may occur. Multiple mechanisms are possible, including residual increased afterload, coronary artery disease and hemodynamic effects of chronic valvular dysfunction. The abnormal ventricular substrate increases the risk of arrhythmia and sudden death.

A diffuse arteriopathy in the upper part of the body seems to be present in a majority of patients, despite successful surgical repair (66).

Late strokes may occur, notably in those who underwent surgical repair as adults and in those with residual hypertension. Cerebral hemorrhage due to a ruptured berry aneurysm can occur late after repair, even in the absence of systemic hypertension. Screening for berry aneurysms is controversial.

Endocarditis/endarteritis can occur at the aortic coarctation site or involving associated lesions. If at the coarctation site, embolic manifestations are usually restricted to the abdominal viscera and legs.

Part VIII. Pregnancy and contraception

Most women of childbearing age with coartation of the aorta have undergone repair. The majority of women with coarctation of the aorta do well during pregnancy (67,68,41). Hypertension worsens in some patients and the spontaneous abortion rate is increased. The most feared complication is acute aortopathy (dissection or rupture), which, although rare, is catastrophic. Patients with significant aortic dilation should be counselled against pregnancy until repair has been performed.

Women with significant aortic coarctation contemplating pregnancy should undergo repair before pregnancy.
Class I, level C (67,68)

Part IX. Arrhythmias
Although sudden death from presumed arrhythmias is uncommon during the first 20 years after repair, the incidence increases thereafter, particularly in those who develop ventricular dysfunction.

Part X. Follow-up

All patients require periodic follow-up by an ACHD cardiologist. All patients should have a periodic MRI following repair of aortic coarctation to document the postrepair anatomy and mechanical complications (restenosis or aneurysm/pseudoaneurysm formation). CT is an alternative if MRI cannot be used (claustrophobic patient or implanted devices, etc). In patients with stents, a focused CT examination is required to interrogate the stented region to rule out aneurysm formation.
Particular attention should be directed toward the following:
- Residual hypertension, heart failure, coronary artery disease or other cardiac disease.
- Associated BAV, which may develop stenosis or regurgitation later in life.
- Recurrent aortic coarctation or significant arm-leg blood pressure gradient at rest.
- Ascending aortopathy, especially in the presence of bicuspid aortic valve.
- New or unusual headaches because of the possibility of intracerebral aneurysms.
- Late dissection proximal or distal to the repair site.
- Aneurysm formation at the site of aortic coarctation repair.

Class I, level C (63,69)

Endocarditis prophylaxis is not recommended in patients with coarctation of the aorta, except within the first six months after interventions (interpositional graft surgery or stent placement).
Class III, level B (45)

RIGHT VENTRICULAR OUTFLOW TRACT OBSTRUCTION

Part I. Background information
Supravalvar right ventricular outflow tract obstruction (RVOTO) seldom occurs in isolation. It may occur in tetralogy of Fallot, Williams syndrome, Noonan syndrome, VSD, arteriohepatic dysplasia or congenital rubella syndrome.

Valvar RVOTO, the most common form of RVOTO, is almost always congenital in origin. Typically, the stenotic pulmonic valve is a thin, pliable, dome-shaped structure, with a narrow opening at its apex. In 10% to 15% of cases, the valve is dysplastic with thickened and immobile cusps (this is frequent in association with Noonan syndrome). In adults, the valve may calcify late in life. A sometimes markedly dilated main pulmonary artery is a common association. The dilated pulmonary artery typically does not rupture.

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

A separate but somewhat similar entity is 'double-chambered right ventricle' with midcavity obstruction, often from a prominent moderator band. This may be associated with a VSD.

Branch pulmonary artery stenosis is not considered here.

Hemodynamic severity grading: The following grading of severity of RVOTO is based on the echo peak instantaneous Doppler gradient (70):
- Mild: Lower than 36 mmHg (peak velocity of less than 3 m/s)
- Moderate: 36 mmHg to 64 mmHg (peak velocity 3 m/s to 4 m/s)
- Severe: Greater than 64 mmHg (peak velocity greater than 4 m/s)

In some patients – for instance, those with long-segment supravalvar stenosis or patients with stenotic conduits – Doppler-derived gradients may not accurately estimate the severity of RVOTO and the estimated right ventricular (RV) systolic pressures derived from assessment of tricuspid regurgitation jet velocity may be a helpful marker of disease severity.

Part II. Prevalence and genetics
Valvar pulmonary stenosis is usually an isolated lesion and occurs in approximately 7% to 12% of all CHD. Patients with Noonan syndrome (autosomal dominant inheritance) may have pulmonary stenosis, atrial septal defect (ASD) and hypertrophic cardiomyopathy. Developmental delay, learning disabilities, facial dysmorphisms, short stature, thoracic/penile and/or testicular abnormalities as well as congenital lymphedema may also be present. Noonan syndrome can be caused by mutations in *PTPN11*, *KRAS*, *SOS1* or *RAF1* genes for which clinical genetic testing is available. Mutations in these genes are identified in 68% to 88% of patients with Noonan syndrome, and are more readily found in familial cases (71,72). Mutations in the *RAF1* gene are found in patients with Noonan syndrome (73).

Williams syndrome is a contiguous gene deletion syndrome caused by a deletion at chromosome 7q11.23, which is associated with cardiac (pulmonary stenosis, pulmonary artery stenosis and supravalvar aortic stenosis), neurodevelopmental (mental retardation and 'cocktail personality') and multisystem manifestations (abnormal facies, short stature and hypercalcemia). Patients with Alagille syndrome (autosomal dominant inheritance – also called arteriohepatic dysplasia) may have pulmonary stenosis, pulmonary arterial stenosis and abnormal facies (triangular facies and deep-set eyes). Mutations in the *JAG1* gene at 20p12 are identified in 90% of patients who meet clinical diagnostic criteria for Alagille syndrome.

Part III. History and management of unoperated patients
Supravalvar RVOTO may progress in severity and should be monitored. Supravalvar obstruction can occur after interventions such as pulmonary artery banding. This lesion usually does not progress.

Patients with trivial valvar RVOTO who are asymptomatic do not become worse with time as adults and will not require treatment, unless endocarditis occurs. Some female patients may present to physicians during pregnancy because of an increase in the loudness of their murmur. Others may present because of enlarged pulmonary arteries detected on chest x-ray.

Mild valvar RVOTO may progress in 20% of unoperated patients. Moderate obstruction may progress in up to 70% of unoperated patients. Some of these patients will also become symptomatic later in life because of atrial arrhythmias.

RVOTO secondary to a double-chambered right ventricle usually progresses in severity and often leads to the development of worsening RV hypertrophy, symptoms and significant gradients requiring surgical repair.

Part IV. Diagnostic workup
An adequate diagnostic workup includes the following:
- Documentation of the level(s) of obstruction.
- Quantification of the severity of the obstruction(s).
- Identification of associated abnormalities such as ASD, PDA, VSD and tetralogy of Fallot.

The diagnostic workup should include the following (at a minimum):
- A thorough clinical assessment, paying particular attention to the 'a' wave on the venous pulse, the length of the murmur, the pulmonary component of the second sound and RV hypertrophy.
- ECG.
- Chest x-ray, paying particular attention to valvar calcification on the lateral film.
- Echo Doppler examination by an appropriately trained individual, paying particular attention to the supravalvar pulmonary artery, the pulmonary valve, the RV outflow tract in multiple projections and the presence or absence of pulmonary regurgitation.

The diagnostic workup may require the following:

- Oximetry (rest and exercise) to determine whether there is cyanosis because of associated abnormalities (ASD or VSD).
- Coronary angiography in patients at risk for coronary artery disease, or in patients older than 40 years of age in whom intervention is being planned.
- MRI to assess associated lesions such as pulmonary artery stenoses, coexisting pulmonary regurgitation and RV function if unable to properly assess these by echo or angiography.
- CT angiography if unable to assess the pulmonary arteries.
- Radionuclide multigated imaging to assess RV function if MRI is not possible (intracardiac devices or claustrophobic patient).
- Heart catheterization (including angiocardiography) to assess hemodynamics and severity of the obstruction and pulmonary artery abnormalities is not indicated unless percutaneous catheter intervention is being performed.
- Patients suspected of having Alagille syndrome should undergo a karyotype and FISH analysis to detect a 20p12 rearrangement or deletion. For those with a normal karyotype and FISH, *JAG1* mutation analysis is now clinically available. All patients with documented *JAG1* mutations or suspected Alagille syndrome should have evaluations for cardiac, hepatic, pigmentary retinal, orthopedic, hematological and renal anomalies (74).

Part V. Indications for intervention/reintervention/medical therapy

In symptomatic patients with valvar RVOTO, a domed pulmonary valve and peak instantaneous Doppler gradients of greater than 50 mmHg or mean echo gradients of greater than 30 mmHg, balloon valvotomy is recommended.
In asymptomatic patients with valvar RVOTO, a domed pulmonary valve and peak instantaneous Doppler gradients of greater than 60 mmHg or mean gradients of greater than 40 mmHg, balloon valvotomy should be considered.
The surgical approach is recommended for patients with significant RVOTO and dysplastic pulmonary valves, subvalvar or supravalvar pulmonary stenosis, associated pulmonary hypoplasia or severe pulmonary regurgitation.
Class I, level C (21,75-77)

In patients with valvar RVOTO, intervention is also probably indicated if there are the following:

- Important arrhythmias (usually sustained atrial flutter).
- An associated ASD or VSD, especially if there is right-to-left shunting.
- Recurrent endocarditis.

Class IIa, level C

In patients with a double-chambered right ventricle with significant midcavity obstruction (pullback gradient at catheter of greater than 50 mmHg), surgery should be considered
Class IIa, level C (78)

Reintervention is indicated for the following:

- Recurrent RVOTO after previous surgery or balloon valvotomy (same criteria as above).
- Severe pulmonic regurgitation associated with reduced exercise capacity of cardiovascular cause or deteriorating RV function or substantial tricuspid regurgitation or sustained atrial flutter/fibrillation or sustained ventricular tachycardia.

Class I, level C (79)

Part VI. Surgical/interventional options

Balloon valvuloplasty is the treatment of choice for valvar RVOTO. Occasionally, valve replacement may be necessary. **(Level B)**

Balloon valvuloplasty for valvar RVOTO should still be performed only in centres and by teams with experience in this technique.
(Level C)
Class I, level B (33,32,77,80-82)

Relief of obstruction in a double-chambered right ventricle is accomplished by surgical resection of RV muscle bands.

Patients who require operation for supravalvar or subvalvar RVOTO should be operated on by congenital heart surgeons.
Class I, level C (32,33)

Part VII. Surgical/interventional outcomes

The long-term results of surgical pulmonary valvotomy are well known. Surgical outcomes are good, but patients require continued surveillance for progressive pulmonary regurgitation, RV outflow tract stenosis or associated lesions. Atrial arrhythmias are common. Long-term survival in surgical patients when valvar RVOTO occurs as an isolated lesion is close to normal. Long-term mortality may be increased, however, with greater age (older than 21 years) at the time of surgery (80).

Patients treated with balloon valvuloplasty, in the absence of a dysplastic valve, have the same prognosis as those who have had surgical valvotomy, at least in the medium term (77,83,84).

Dynamic subvalvar RVOTO often resolves when coexistent valvar stenosis is treated with balloon valvuloplasty.

Subvalvar and supravalvar RVOTO seldom recur after adequate repair.

Part VIII. Pregnancy and contraception

The increased hemodynamic load of pregnancy may precipitate right heart failure, atrial arrhythmias or tricuspid regurgitation in patients with significant RVOTO, irrespective of the presence or absence of symptoms before pregnancy. Patients with moderate-to-severe RVOTO should, therefore, be considered for RVOTO relief before conception.

Balloon valvuloplasty for valvar pulmonary stenosis may be used during pregnancy if the stenosis is severe and symptoms due to pulmonary stenosis develop. When possible, intervention should be delayed until after the first trimester.

Pregnancy in patients with mild RVOTO or RVOTO that has been alleviated by valvuloplasty or surgery, with or without pulmonary regurgitation, is generally well tolerated. Global (ventricular systolic dysfunction and history of cardiac complications) and lesion-specific risk factors for adverse events during pregnancy should be taken into account when determining the risk of pregnancy (42,85).

Part IX. Arrhythmias

Atrial flutter is the most common arrhythmia. Following repair, the onset of new atrial arrhythmias should prompt a search for residual RVOTO and/or pulmonary regurgitation.

Part X. Follow-up

Patients with trivial RVOTO (peak-to-peak catheter gradient of less than 25 mmHg) do not require ACHD cardiology follow-up.
Patients with mild or greater RVOTO, or moderate to severe pulmonary regurgitation require monitoring by an ACHD cardiologist because intervention may be required.
Particular attention should be paid to the following:

- Progressive/recurrent stenosis, especially at the subvalvar level.
- RV size and function in the context of significant pulmonary/subpulmonary stenosis and/or regurgitation.
- Tricuspid regurgitation (often reflecting RV dysfunction).
- Atrial and occasionally ventricular (usually postoperative) arrhythmias (sustained).

- Evidence of intracardiac shunting, especially right to left.

Class I, level C

Endocarditis prophylaxis is not recommended in patients with RVOTO, unless a prosthetic valve has been inserted.
Class III, level B (54)

TETRALOGY OF FALLOT

Part I. Background information

The defect is due to anterocephalad deviation of the outlet septum, resulting in an unrestricted large anterior malalignment subaortic VSD; RVOTO, which may be infundibular, valvar, supravalvar or a combination of all; consequent RV hypertrophy; and an overriding aorta (less than 50%). Accompanying features can include additional muscular VSDs, AVSDs most commonly in patients with Down's syndrome, anomalous coronary arteries, a right-sided aortic arch, PDA, main and branch pulmonary artery anomalies including hypoplasia and stenosis, and aortopulmonary collaterals (mainly seen in patients with pulmonary atresia/VSD, which is not discussed here) and a dilated thoracic aorta.

The so-called pentalogy of Fallot also has an ASD or patent foramen ovale (PFO).

Part II. Prevalence and genetics

Approximately 15% of patients with tetralogy of Fallot have the 22q11 deletion syndrome (86). The clinical spectrum is highly variable, ranging from normal individuals to severely affected newborns with CHD, clefting defects, thymic hypoplasia, dysmorphic facial features and hypocalcemia (Cardiac defect, Abnormal facies, Thymic hypoplasia, Cleft palate, Hypocalcemia and 22q11 deletion). These patients have an elevated risk of late psychiatric disorders (87-89). The 22q11 deletion syndrome occurs de novo in 96% of cases, with the offspring of affected individuals having a 50% risk of inheriting the transmission. In the remaining 6% of cases, the condition is inherited from a parent as an autosomal dominant trait. FISH testing for 22q11 deletion is clinically available and is increasingly offered in patients with conotruncal defects including tetralogy of Fallot. Other syndromes associated with tetralogy of Fallot include fetal alcohol syndrome, maternal phenylketonuria syndrome, Alagille syndrome and cat-eye syndrome.

Part III. History and management of unoperated patients

The pathophysiology varies depending on the degree of RVOTO.

With relatively mild obstruction, the presentation is of increased pulmonary blood flow and minimal or no cyanosis, so-called 'pink tetralogy' or 'acyanotic Fallot'. This occasionally presents in adulthood.

Most children will have had reparative surgery.

Rarely, adults present who are unoperated. For them, surgical repair is still recommended because the results are gratifying and the operative risk is comparable with pediatric series, provided there is good LV function and no serious coexisting morbidity (90,91).

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

- Blalock-Taussig shunt or modification (subclavian artery to pulmonary artery).
- 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.
- Right ventricle to pulmonary artery conduit without VSD closure or with fenestrated closure.

Reparative surgery involves closing the VSD and relieving the RVOTO. The latter may involve the following:

- Resection of infundibular muscle.
- RV subannular outflow tract patch.
- Transannular patch (a patch across the pulmonary valve annulus that disrupts the integrity of the pulmonary valve and causes substantial pulmonary regurgitation). This technique has now been largely abandoned for a less aggressive surgical approach. Avoidance of free pulmonary regurgitation and subsequent RV dilation at the expense of some, albeit not severe, residual pulmonary stenosis is now a key therapeutic goal of reparative surgery.
- An extracardiac conduit placed between the right ventricle and pulmonary artery (in cases of anomalous coronary artery crossing the RVOT).
- Replacement of the pulmonary valve.
- Pulmonary valvotomy.
- Pulmonary arterioplasty.
- Repair of PFO, secundum ASD or AVSD, if present.
- Additional lesions, such as aortic regurgitation or muscular VSD, which may also need to be addressed.

Part IV. Investigational workup in operated patients

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

All patients should have the following (at a minimum):

- A thorough clinical assessment.
- ECG.
- Chest x-ray.
- Echo-Doppler examination by an appropriately trained individual to detect and quantify residual pulmonary stenosis and regurgitation, residual VSD, RV and LV size and function, aortic regurgitation and aortic root size.

All patients may require the following:

- MRI for the assessment of pulmonary artery or aortic anomalies, pulmonary regurgitant fraction as well as RV size and function. CT angiography is an alternative if MRI is unable to assess the pulmonary arteries and/or if radionuclide multigated imaging/MRI are unable to assess RV function.
- Exercise testing (or cardiopulmonary exercise testing) to assess functional capacity and to detect possible exertional arrhythmias.
- Holter monitoring.
- Quantitative lung perfusion scans in patients with suspected pulmonary artery branch stenosis (this information can be obtained through MRI).
- Heart catheterization if adequate assessment of the hemodynamics is not obtainable by noninvasive means, including pulmonary angiography in patients with suspected pulmonary artery branch stenosis and coronary angiography if surgical reintervention is planned.
- Electrophysiological study for those being evaluated because of suspected or sustained atrial or ventricular arrhythmias, or for risk stratification for sudden cardiac death.

FISH testing for 22q11 deletion is recommended for any adult with tetralogy of Fallot, truncus arteriosus, interrupted aortic arch, VSD or aortic arch anomaly not previously tested, especially if they have at least one other feature of the 22q11 deletion syndrome (92-94).

For patients who have had previous palliation, assessment of pulmonary artery pressure and anatomy is mandatory at some point because these shunts have inherent complications (distortion of the pulmonary arteries, stenosis or aneurysm in the shunt or at the site of anastomosis, development of pulmonary hypertension and volume overloading of the left heart).

The following issues may need to be addressed following a palliative shunt:

- Determine whether complete repair is possible.
- Explain increasing cyanosis with erythrocytosis.
- Determine whether pulmonary hypertension is present (unilateral or bilateral).

- Explain the reduction or absence of the continuous shunt murmur (suspected shunt stenosis or occlusion).
- Determine whether there is aneurysm formation in the shunt.
- Determine whether there is an anomalous coronary artery crossing the RV outflow tract.

Patients with repaired or palliated tetralogy of Fallot should be followed up regularly by a cardiologist with expertise in ACHD. Echo, advanced imaging (MRI, CT and nuclear), heart catheterization and arrhythmia interventions should be performed by staff with expertise in ACHD.
Class I, level C (32,33)

Part V. Indications for intervention/reintervention/medical therapy

Following palliative surgery, complete intracardiac repair should be considered in all patients, in the absence of severe irreversible pulmonary hypertension or unfavourable anatomy (inadequate pulmonary arteries). The following situations particularly warrant complete repair:
- Worsening symptoms.
- Cyanosis with erythrocytosis.
- Reduction or absence of the continuous shunt murmur (suspected shunt stenosis or occlusion).
- Aneurysm formation in the shunt.
- LV dilation due to aortic regurgitation or a residual shunt.

Class IIa, level C (91)

Reoperation is only necessary in approximately 10% to 15% of patients following reparative surgery over a 20-year follow-up period. This frequency may increase with time, but longer-term follow-up data are currently not available.

The following situations may warrant intervention following repair:
- Free pulmonary regurgitation associated with progressive or moderate to severe RV enlargement (RV end-diastolic volume of greater than 170 mL/m^2), moderate to severe RV dysfunction, important tricuspid regurgitation, atrial or ventricular arrhythmias, or symptoms such as deteriorating exercise performance.
- Residual VSD with a shunt of greater than 1.5:1.
- Residual pulmonary stenosis with an RV pressure of at least two-thirds the systemic pressure (either the native RV outflow or valved conduit if one is present).
- Significant aortic regurgitation associated with symptoms and/or progressive LV systolic dysfunction.
- Aortic root enlargement of at least 55 mm in diameter.
- A large RV outflow tract aneurysm or evidence of infection or false aneurysm.
- Sustained clinical arrhythmias, most commonly either atrial flutter or fibrillation, or sustained monomorphic ventricular tachycardia (VT). When any of these arrhythmias occur, the patient should also be evaluated for a treatable hemodynamic cause of the arrhythmia.
- The combination of residual VSD and/or residual pulmonary stenosis and regurgitation – all mild-moderate but leading to substantial RV enlargement, reduced RV function or symptoms.

Class IIa, level C (79,95-100)

Part VI. Surgical/interventional options

Patients with tetralogy of Fallot who require intervention should be operated on by congenital heart surgeons.
Class I, level C (32,33)

The following are possible intervention strategies:
- Reoperation to insert a new pulmonary valve (either homograft or porcine) may be necessary for severe pulmonary regurgitation leading to RV dilation, sustained arrhythmias and/or symptoms. Tricuspid valve (TV) annuloplasty may also be necessary when at least moderate tricuspid regurgitation is present.
- Percutaneous pulmonary valve insertion, as an alternative to surgery, may be used for residual pulmonary stenosis or pulmonary regurgitation but can only be performed in the setting of a conduit of suitable size (currently 22 mm or smaller).
- Surgery for residual significant RVOTO involving resection of residual infundibular stenosis or placement of an RV outflow or transannular patch.
- Aortic valve and/or root replacement may be necessary for those with aortic valve regurgitation and/or root dilation.
- Suture or patch closure of a residual VSD if the shunt is 1.5:1 or greater, or if the patient is undergoing cardiac reoperation for other reasons.
- Branch pulmonary artery stenosis may be managed with balloon dilation, stent insertion or surgery.
- Radiofrequency or surgical cryoablation for atrial flutter and sustained VT; Maze procedure including pulmonary vein isolation for atrial fibrillation.
- Automatic implantable cardioverter defibrillator (AICD) for primary and secondary prevention of sudden cardiac death in high-risk patients.
- Closure of ASD or PFO, especially if there is persistent cyanosis or paradoxical embolism.

Part VII. Surgical/interventional outcomes

The overall survival of patients who have had operative repair is excellent, provided the VSD has been closed and the RVOTO has been relieved. A 36-year survival of 85% has been reported (95). Death may occur from reoperation, endocarditis or congestive heart failure. Sudden cardiac death is the leading cause of mortality following tetralogy of Fallot repair, although the overall incidence is low (approximately 0.15% per year) (101).

Surgical pulmonary valve replacement for chronic significant pulmonary regurgitation can be performed with a low mortality, and may lead to improvement in RV dimension and performance if performed before marked RV dilation or dysfunction supervenes (102-104).

Percutaneous pulmonary valve replacement can be performed with similar mortality and favourable hemodynamic short- and intermediate-term results with less morbidity than surgical pulmonary valve replacement (105), but should be done only in ACHD centres with expertise in the procedure (106). At present, these therapies are reserved primarily for patients with circumferential right ventricle to pulmonary artery conduits (ie, homografts and valved conduits) measuring less than 22 mm.

Part VIII. Arrhythmias

Frequent ventricular ectopy and nonsustained ventricular arrhythmias on Holter monitoring are common. Sustained VT has been reported in 12% of patients 35 years after repair (107). Ventricular tachyarrhythmias relate to surgical sequela (eg, ventriculotomy incisions) and abnormal hemodynamics, usually from RV dilation secondary to pulmonary regurgitation and/or tricuspid regurgitation. QRS duration on the surface ECG correlates with RV size and, when prolonged (QRS of 180 ms or greater), is an independent predictor of sustained VT and sudden death (108).

VT: *Primary prevention:* Inducible sustained VT on electrophysiological study identifies patients at higher risk for clinical VT and sudden cardiac death (109). Although the selection of appropriate candidates for risk stratification with electrophysiological study remains controversial, it is not recommended as a routine screening test in asymptomatic patients. It may be most beneficial in 'moderate-risk' patients (history of palpitation or syncope, older age at repair, transannular patch, QRS of 180 ms or greater, nonsustained VT, etc). AICDs appear to be effective in recognizing and terminating ventricular arrhythmias in patients with tetralogy of Fallot. The selection of appropriate candidates for primary prevention AICDs remains to be refined. AICDs are probably most beneficial in 'high-risk patients'

(eg, previous palliative shunt, QRS of greater than 180 ms, nonsustained VT, inducible VT and LV dysfunction) and are probably best reserved for those with a high annual risk (3.5% or greater per year) of sudden cardiac death (110). The benefits of AICDs in this patient population must be weighed against the high complication rate, including inappropriate shocks (111).

Restoration of hemodynamics through pulmonary valve implantation, TV repair or RV outflow tract aneurysm resection, with concomitant intraoperative ablation, leads to a recurrence rate of VT between 0% and 10%. AICD implantation following corrective surgery may still have a role, particularly in patients with inducible VT postoperatively.

In patients with sustained ventricular tachyarrhythmia and/or resuscitated from sudden cardiac death with no clear identified reversible cause, implantable cardioverter defibrillators are indicated for secondary prevention.
Class I, level B (111)

Patients deemed to be at particularly high risk for sudden cardiac death may benefit from implantable cardioverter defibrillators for primary prevention.
Class IIa, level B (111)

Atrial flutter and fibrillation: The most common atrial arrhythmias are typical atrial flutter and incision-related macro-reentrant circuits. Atrial fibrillation occurs less frequently. Development of atrial tachyarrhythmias may herald worsening ventricular function and tricuspid regurgitation. Hemodynamic compromise may be precipitated in some and acute management is dictated by the level of clinical stability. Reassessment of the repair, and consideration for electrophysiological testing and ablation, may be indicated. In patients undergoing cardiac surgery for another indication, concomitant surgical ablation (eg, right atrial maze for macro-reentrant tachyarrhythmias and biatrial maze for atrial fibrillation) may be considered.

Atrioventricular block: With improved surgical techniques, the incidence of atrioventricular block postrepair in the current era is low, but postoperative and late heart block can occur; these have been implicated as potential causes of sudden death.

Part IX. Pregnancy and contraception

The risk of pregnancy in surgically repaired patients depends on the hemodynamic status. Pregnancy in unoperated patients constitutes a considerable risk of maternal and fetal complications and death. This risk is greater when resting oxygen saturations are lower than 85%.

The risk is low in women with good underlying hemodynamics. In women with significant residual RVOTO, severe pulmonary regurgitation with or without tricuspid regurgitation and RV dysfunction, the increased volume load of pregnancy may lead to right heart failure and arrhythmias (41,112). Global risk factors for adverse events during pregnancy (ventricular dysfunction, degree of cyanosis and history of cardiac complications) need to be incorporated into decision making (42).

All patients with tetralogy should have cardiology counselling preconception and be followed up by an ACHD cardiologist during pregnancy. Preconception screening for features of 22q11 deletion syndrome is recommended.

Part X. Follow-up

All patients with tetralogy of Fallot should have regular cardiology follow-up by an ACHD cardiologist.
Endocarditis is recommended in patients with unrepaired tetralogy of Fallot, for six months following surgical repair, if there is a prosthetic valve or if there is a VSD patch leak.
Class I, level C (45)

Endocarditis prophylaxis is not recommended in patients with repaired tetralogy of Fallot unless a prosthetic valve has been inserted.
Class III, level B (45)

EBSTEIN ANOMALY

Part I. Background information

Ebstein anomaly is rare, occurring in one to five per 200,000 births (accounting for less than 1% of all CHD). The cardinal feature is incomplete delamination of the septal and posterolateral leaflets of the TV; the leaflets arise from the ventricular wall below the atrioventricular junction.

The consequences of this may include the following:

- Atrialization of the inflow of the right ventricle to varying degrees with a resultant smaller 'functional' right ventricle.
- Varying degrees of tricuspid regurgitation (exceptionally, the TV is stenotic).
- Enlargement of the right atrium.
- Varying degrees of anatomical and physiological RV inflow or outflow tract obstruction.
- Varying impairment of LV function due to abnormal septal motion and abnormal RV function (113-115).

Common associations include the following:

- A shunt at the atrial level, either PFO or secundum ASD, in approximately 50% with varying degrees of cyanosis.
- One or more accessory conduction pathways in 25% of patients, which increases the risk of atrial tachyarrhythmias and sudden death.

Part II. Genetics

Most cases are sporadic, although rare familial occurrences have been reported. Chromosome abnormalities are rare. Animal studies implicate several possible candidate genes on chromosome 17q. There is a small risk of Ebstein anomaly in fetuses exposed to lithium during the pregnancy.

Part III. History and management of unoperated patients

Patients with mild Ebstein anomaly may be asymptomatic with no functional limitation. Survival to the ninth decade has been reported. Patients with severe Ebstein anomaly usually present at birth or even in utero. Patients with moderate Ebstein anomaly may become symptomatic during late adolescence or young adult life. The most common symptoms in adults are exercise intolerance (dyspnea and fatigue) and symptomatic supraventricular arrhythmias. Heart block occasionally occurs. When an atrial defect is present, patients may be cyanotic to a varying degree – particularly during exercise – and are at risk of paradoxical embolus resulting in transient ischemic attack (TIA) and/or stroke. Alternatively, they may have a left-to-right shunt at rest, which can reverse on effort. End-stage disease with severe tricuspid regurgitation and RV dysfunction may manifest as right-sided cardiac failure, which may be precipitated by an arrhythmia such as atrial flutter or fibrillation. Sudden death (presumed arrhythmic in nature) may occur at any age and is more likely if accessory pathway(s) is/are present (116-123).

Part IV. Diagnostic workup

An adequate diagnostic workup includes the following:

- Documentation of the anatomical severity of the TV abnormality (degree of apical displacement of the valve leaflets), together with the resultant degree of right-sided chamber abnormality (right atrial enlargement and RV diminution) and the physiological consequences in terms of RV dysfunction, tricuspid regurgitation/stenosis and RVOTO.
- Determination of whether the TV has the potential for surgical repair. This depends on the anterior leaflet size and degree of tethering, as well as the relative size of the 'functional' right ventricle.
- Documentation of the presence or absence of an atrial communication, and whether there is right-to-left shunting.
- Assessment of LV function and identification of any mitral valve abnormalities.

- Definition, if possible, of the presence or absence of accessory pathways.
- Determination of the presence or absence of associated lesions.
- Determination of the amount of functional limitation, if any.

The initial workup should include the following (at a minimum):
- A thorough clinical assessment.
- ECG.
- Chest x-ray.
- Transthoracic echo-Doppler evaluation by an appropriately trained individual.
- Oximetry with exercise.

The diagnostic workup may require the following:
- Exercise test/cardiopulmonary assessment to clarify the severity of functional limitation.
- TEE Doppler examination if the anatomical information is not provided by TTE.
- Holter monitor.
- An electrophysiology study if there is a history of syncope or palpitations, or ECG evidence of arrhythmias or accessory pathway(s).
- Coronary angiography in patients at risk of coronary artery disease or in patients older than 40 years of age if surgical repair is planned.
- MRI may be helpful to quantify the degree of left-to-right shunting, or the severity of mitral and tricuspid regurgitation.

Part V. Indications for intervention/reintervention/medical therapy

The following situations warrant intervention:
- Limited exercise capacity (New York Heart Association class greater than II).
- Increasing heart size (cardiothoracic ratio greater than 65%).
- Important cyanosis (resting oxygen saturations of less than 90%).
- Severe tricuspid regurgitation with symptoms.
- TIA or stroke.

Class I, level B (124,125)

Part VI. Interventional options

Ebstein anomaly should only be repaired by congenital heart surgeons who have substantial specific experience and success with this operation. Every effort should be made to preserve the native TV.
Class I, level C (126-130)

When the anterior TV leaflet is mobile and can serve as a monocusp valve, and the functional right ventricle is of adequate size (greater than one-third of the total right ventricle), valve repair may be possible and may be preferable to valve replacement (131). If the TV is not repairable, valve replacement is necessary. Atrial communication, if present, should be closed. Improvement in symptoms and exercise capacity can occur from closure of the interatrial shunt; this percutaneous option may be offered to selected patients. A test occlusion is mandatory to assure that there is no drop in cardiac output or increase in right atrial pressure.

Given normal pulmonary artery pressures, in patients with an inadequate right ventricle (because of size or function), severe tricuspid regurgitation and/or chronic supraventricular arrhythmias, a bidirectional cavopulmonary connection may be used to supplement the intracardiac repair.

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

It is controversial whether the atrialized portion of the right ventricle should be plicated to improve hemodynamics and reduce the risk of atrial arrhythmias.

Atrial arrhythmias are often dealt with at the time of surgery. Radiofrequency or operative cryoablation has been successful in preventing atrial flutter. A biatrial maze procedure, including pulmonary vein encirclement, may be helpful to prevent and treat atrial fibrillation.

Nonsurgical interventions may include the following:
- Percutaneous closure of an interatrial communication in the setting of a TIA/stroke or exercise-induced cyanosis in an individual who is otherwise well without significant hemodynamic compromise or symptoms.
- Transvenous radiofrequency or cryomapping and ablation of atrial arrhythmias when there is no other indication for surgery or prior to surgery if surgery is indicated.

Part VII. Interventional outcomes

With satisfactory valve repair, with or without bidirectional cavopulmonary connection, medium-term prognosis is excellent. Late arrhythmias, most commonly atrial tachyarrhythmias and, seldomly, complete atrioventricular block, may occur.

Repeat surgery may be necessary because of a failing bioprosthesis or thrombosed mechanical valve. There is a high incidence of complete heart block with TV re-replacement (132-136).

Part VIII. Arrhythmias

First-degree atrioventricular block often results from intra-atrial conduction delay. Accessory atrioventricular or atriofascicular pathways are found in 25%, and are frequently right sided and multiple. Supraventricular tachyarrhythmias, including accessory pathway-mediated tachycardia, focal atrial tachycardia and atrial fibrillation or flutter, occur in 30% to 40% of cases, constituting the most common presentation in adolescents and adults (117). Sudden death is relatively uncommon, and may be preceded by cyanosis or rapid conduction of atrial fibrillation or flutter to the ventricles via high-risk or multiple pathways. In general, the complexity of catheter mapping and ablation procedures is increased by associated malformations, multiple pathways and signals that may be difficult to interpret within the 'atrialized' ventricle.

Part IX. Pregnancy and contraception

In the absence of maternal cyanosis, right-sided heart failure or arrhythmias, pregnancy is usually well tolerated (41,137,138). Global risk factors (ventricular function, degree of cyanosis and history of cardiac complications) for adverse events during pregnancy should be incorporated into the overall risk assessment (42,85).

Part X. Follow-up

All patients with Ebstein anomaly should receive regular cardiology follow-up by an ACHD cardiologist. Particular attention should be paid to the following:
- Cyanotic patients.
- Substantial cardiomegaly (cardiothoracic ratio greater than 64%).
- RV function, which may worsen and cause congestion.
- Tricuspid regurgitation or tricuspid stenosis in the previously operated patient.
- Degeneration and/or infection of a bioprosthetic valve, or thrombosis and/or infection of a mechanical valve.
- Recurrent atrial arrhythmias.
- Ventricular arrhythmias.
- Complete heart block.

Class I, level C

Endocarditis prophylaxis is not recommended in patients with Ebstein anomaly, unless the patient is cyanotic, within the first six months after a repair or if a prosthetic valve has been inserted.
Class III, level B (45)

MARFAN'S SYNDROME

Part I. Background information

Marfan's syndrome is an autosomal, dominant, inherited connective tissue disorder. The cardinal features involve the skeletal, ocular and cardiovascular systems. The pulmonary and integumentary systems and dura may also be involved.

TABLE 1
Ghent diagnostic criteria for Marfan's syndrome

System	Major criteria	Minor criteria
Skeletal		
Major criterion: ≥4 major	Pectus carinatum	Pectus excavatum (moderate)
Involvement: ≥2 major or 1 major + 2 minor	Pectus excavatum (severe)	Joint hypermobility
	Long limbs (dolichostenomelia)*	High arched palate and dental crowding
	Long fingers (arachnodactyly)†	Typical facial features:
	Scoliosis >20° or spondylolisthesis	Long face (doliocephaly)
	Reduced elbow extension <170°	Flat cheek bones (malar hypoplasia)
	Pes planus	Deep-set eyes (enophthalmos)
	Protrusio acetabulae (on hip x-ray)	Undershot lower jaw (retrognathia)
		Down-slanting palpebral fissures
Ocular		
Major criterion: 1 major	Ectopia lentis	Flat cornea
Involvement: ≥2 minor		Elongated globe
		Myopia
Cardiovascular		
Major criterion: ≥1 major	Aortic root dilation	Mitral valve prolapse
Involvement: ≥1 minor	Ascending aortic dissection	Main pulmonary artery dilation <40 years
		Mitral annulus calcification <40 years
		Descending and/or abdominal aorta
		Dilation/dissection <50 years
Pulmonary		
Major criterion: None	None	Spontaneous pneumothorax
Involvement: 1 minor		Apical bulla on chest x-ray or computed tomography
Skin/integument		
Major criterion: None	None	Unexplained stretch marks (striae atrophicae)
Involvement: ≥1 minor		Recurrent or incisional hernias
Dura		
Major criterion: 1 major	Lumbosacral dural ectasia by computed tomography or magnetic resonance imaging	None
Family/genetic history		
To be contributory = 1 major	Parent, child or sibling who meets diagnostic criteria independently	None
	Presence of *FBN1* mutation known to cause Marfan's syndrome	
	Presence of haplotype around *FBN1* inherited by descent, known to be associated with unequivocally diagnosed Marfan's syndrome in the family (linkage)	

**Reduced upper to lower segment ratio for age or arm span to height ratio greater than 1.05; †Wrist and thumb sign positive. Data from reference 149*

Part II. Prevalence and genetics

Prevalence has been estimated to be one in 3000 to one in 5000, with no sex or ethnic predispositions. It is caused by mutations in the *FBN1* gene located on chromosome 15, which encodes the connective tissue protein fibrillin-1, a structural and regulatory glycoprotein component of the extracellular microfibrils. New mutations account for 25% to 30% of cases. No robust genotype-phenotype correlations have emerged, despite more than 1000 disease-causing *FBN1* mutations having been reported. Age at onset, tissue distribution and severity of clinical manifestations are highly variable both among and within affected families (139-142).

Part III. History and management of unoperated patients

The prognosis of patients with Marfan's syndrome is mainly determined by aortic root abnormalities, which predispose to progressive dilation and dissection, and lead to aortic regurgitation. The mean survival time of untreated patients is 40 years, but the variance is large. Patients with a dilated aorta are usually asymptomatic. Symptoms of a type A dissection with the initial tear in the ascending aorta can be variable; however, classically, it presents with acute onset of severe chest pain going through to the back. Type B dissection, typically with the initial tear in the proximal descending thoracic aorta, accounts for approximately 10% of acute dissection in Marfan's syndrome. The presence of aortic regurgitation or mitral valve prolapse with regurgitation may lead to signs or symptoms of LV volume overload. The risk of type A dissection clearly increases with increasing aortic root diameter in Marfan's syndrome, as in other dilated aortopathies (143-146).

Between 1972 and 1995, the life expectancy for patients with Marfan's syndrome improved substantially. This was likely, to a large degree, due to earlier clinical recognition and benefits arising from cardiovascular surgery (147,148).

Part IV. Diagnostic workup

An adequate diagnostic workup includes the following:

- Documentation of the basis for the diagnosis of Marfan's syndrome, using the Ghent criteria (Table 1) (149).
- Determination of the diameter, and search for the dissection of the aortic root and all other parts of the aorta.
- Determination of the presence of mitral valve prolapse, mitral regurgitation, calcification of the mitral annulus, presence of TV prolapse and tricuspid regurgitation.
- Determination of the presence and degree of aortic regurgitation and its mechanism, in particular, considering the possibility of dissection.

To meet the Ghent criteria for the clinical diagnosis of Marfan's syndrome requires either of the following:

- Major criterion in the first system, involvement in the second system and a family/genetic history; or

- Major criterion in the first two systems and involvement in the third system.

Class I, level C (149)

Clinical screening of patients with suspected Marfan's syndrome and their relatives should be through a multidisciplinary clinic involving professionals with specific training and experience in Marfan's syndrome. Accuracy of diagnosis is critical, and requires a rigorous approach. Genetic screening with *FBN1* mutational analysis should yield a result in approximately 95% of patients with classic Marfan's syndrome according to the Ghent criteria (150). Genetic testing in a family with a known disease-causing *FBN1* mutation is of most benefit because it allows for presymptomatic and prenatal screening. Genetic testing for an individual patient with suspected Marfan's syndrome, but not enough features for a clinical diagnosis and no (or an uncertain) family history may be of minimal benefit (139). Exceptions include atypical presentations in an individual patient with a personal or family history of early aortic or arterial dissection; in these cases, screening and genetic testing for the following conditions should now be considered: Loeys-Dietz syndrome (151,152), Ehlers-Danlos syndrome type IV or vascular type (153) and familial thoracic aortic aneurysm (154-156).

When the diagnosis of Marfan's syndrome has been established, the diagnostic workup should include the following (at a minimum):

- A thorough clinical assessment.
- ECG.
- Chest x-ray.
- Echo-Doppler evaluation, especially for measurements of the aortic root and ascending thoracic aorta, assessment for aortic regurgitation, mitral valve prolapse and degree of mitral regurgitation.
- MRI scanning for measurements of the entire aorta and its branches, and assessment for lumbosacral dural ectasia. If MRI is not possible, CT is an alternate approach. Abdominal ultrasound remains an alternative imaging modality for the abdominal aorta, especially in claustrophobic patients, but cannot assess the descending thoracic aorta well.

A diagnostic workup may require the following:

- Coronary arteriography in patients older than 50 years of age (or younger if there are risk factors for coronary artery disease) in whom surgery is being planned. Because of the specific risks of catheter manipulation in patients with fragile aortas, thresholds for angiography in this population differ from other lesions.
- CT scanning or TEE if aortic dissection is suspected.

Part V. Indications for intervention/reintervention/medical therapy

Beta-adrenergic blockade currently remains the 'standard of care' for patients with Marfan's syndrome, and may work by reducing heart rate and the rate of increase in aortic pressure over time (139,157,158). Rigorous blood pressure control is important, especially in patients with dilated aortic roots. Recent experimental evidence and data from a small pediatric series have led to clinical trials with the hypothesis that losartan, an angiotensin II type 1 receptor blocker, may play a role in slowing the rate of aortic root dilation in Marfan's syndrome (159,160).

All patients with Marfan's syndrome should be advised to take beta-blockers and to remain on them unless side effects preclude their use. This is especially true, usually in association with other blood pressure-lowering agents, if dissection has occurred.

Class IIa, level B (139,157,158)

Exercise and activity recommendations in patients with Marfan's syndrome should be individualized according to the underlying severity of aortic root dilation and/or a malignant family history of aortic dissection as indicated below (139,161,162).

Patients should avoid the following:

- Heavy isometric exercise, such as weight lifting, which can markedly increase peripheral vascular resistance and proximal aortic wall stress.
- Competitive aerobic sports, which can markedly increase cardiac output and aortic rate of increase in aortic pressure over prolonged periods of time.
- Activities that involve risk of bodily collisions.
- Marked changes in ambient air pressure (as in scuba diving or sudden changes in altitude in a nonpressurized aircraft) because they can predispose to pneumothorax (163).

Patients with Marfan's syndrome, especially those with a significantly dilated aortic root and/or a malignant family history of aortic dissection, should be advised to avoid heavy isometric exercise, competitive sports, contact sports and other activities that involve risk of sudden impact.

Class I, level C (139,161,162)

The following situations warrant surgical intervention:

- A maximal aortic root/ascending aorta diameter of greater than 50 mm.
- A maximal aortic root/ascending aorta diameter of greater than 45 mm to 50 mm with the following:
  1. Rapid aortic root growth of more than 5 mm per year.
  2. Progressive aortic regurgitation, especially if the surgeon believes the aortic valve can be spared and an aortic valve-sparing procedure is planned.
  3. Family history of premature aortic dissection of less than 50 mm.
  4. Severe mitral valve regurgitation that requires surgery.
- A maximal aortic root/ascending aorta diameter of greater than 44 mm if pregnancy is desired.
- A maximal dimension of other parts of the aorta of 50 mm to 60 mm or progressive dilation.
- Severe mitral regurgitation with symptoms or progressive LV dilation/dysfunction as per the current guidelines on valvular heart disease.

Class I, level B (20,139-146,164-169)

Women have aortas that are, on average, 5 mm smaller than those of men (170). Some experts believe that in small individuals, the use of an indexed diameter adjusted for body surface area may be useful in operative decision making (146).

Part VI. Surgical options

For aortic root replacement, the surgical options include a composite aortic valve and root replacement (modified Bentall procedure – using a mechanical, bioprosthetic or homograft aortic valve prosthesis) (171), and an aortic valve-sparing procedure (172). In patients with primary normal valves, in whom aortic insufficiency is due to the dilated annulus or dissection, valve-sparing operations with root replacement by a Dacron prosthesis and with reimplantation of the coronary arteries into the prosthesis (David operation) or remodelling of the aortic root (Yacoub operation) have now become the preferred choice of surgery. If necessary, all other parts of the aorta can be replaced. Surgery should be performed in a centre and by surgeons with substantial experience performing these types of surgeries.

Although there are few reports of short-term success after endovascular stent grafting of the descending thoracic aorta, stent grafting in patients with Marfan's syndrome is not recommended unless the risk of conventional surgical repair is deemed prohibitive (173).

Part VII. Surgical outcomes

The Bentall procedure has well-established, long-term durability in Marfan's syndrome (174), but the newer aortic valve-sparing procedure has now evolved to a point that similar short- and long-term

outcomes have emerged, without the additional need for long-term anticoagulation (175-178). Bentall patients, as expected due to the need for anticoagulation, had higher rates of thromboembolic complications. Valve-sparing patients had an increased need for reoperation on the aortic root, which was associated with the remodelling rather than the reimplantation approach, as in previous series (178). Progressive aortic insufficiency has been reported.

Following aortic root replacement, new dilation or dissection in more distal segments of the aorta may develop, necessitating long-term surveillance of the entire aorta.
Class I, level C (139,179)

Part VIII. Arrhythmias

Arrhythmias are not a feature of Marfan's syndrome itself. They may occur as a consequence of mitral valve prolapse and/or mitral regurgitation, myocardial ischemia/infarction due to dissection, or ventricular dysfunction. Ventricular arrhythmias have been demonstrated to be particularly associated with LV dilation and abnormalities of repolarization; therefore, Marfan's syndrome patients with these features warrant increased surveillance.

Part IX. Pregnancy and contraception

For women with Marfan's syndrome, pregnancy presents a twofold problem: transmission to offspring, with a 50% chance that the child will be affected; and an increased (but unquantified) risk of aortic dissection during pregnancy and for up to six months postpartum.

Women with an aortic diameter above 44 mm should be strongly discouraged from becoming pregnant without repair.
An aortic diameter below 40 mm rarely presents a problem, although a completely safe diameter does not exist.
Surgical replacement of the aortic root does not completely normalize the risk because dissection can occur elsewhere in the aorta.
Class I, level B (166-169)

Part X. Follow-up

Whenever possible, Marfan's patients should be under the care of professionals with specific training/experience in Marfan's syndrome. Ideally, this should be through a multidisciplinary clinic.
During follow-up, the aortic root and the entire aorta should be regularly evaluated with echo (usually annually), MRI/CT of the entire aorta and/or abdominal ultrasound examinations every three years. Annual MRI/CT of the entire aorta may be indicated if any aortic segment is dilated, approaching surgical indication, within approximately one year following aortic surgery and for two to three years postaortic dissection to monitor stability, especially of a residual dissected aorta.
Patients with mitral valve prolapse, and moderate or more severe mitral regurgitation, should also be followed with serial echo.
Class I, level C

Endocarditis prophylaxis is not recommended in patients with Marfan's syndrome, except for six months following surgery or if a prosthetic valve has been inserted.
Class III, level B (45)

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