42 - 280 Congenital Heart Disease in the Adult

280 Congenital Heart Disease in the Adult

There is a paucity of evidence to inform practice guidelines for surgical and/or transcatheter valve intervention in patients with multiple or mixed valve disease. When there is a clear, dominant lesion, as for example in a patient with severe AS and mild AR, indications for intervention are straightforward and follow those recommended for patients with AS (Chap. 272). In other patients, however, there is less clarity, and decisions regarding intervention should be based on several considerations, including those related to lesion severity, ventricular remodeling, functional capacity, and PA pressures. In this regard, it is important to realize that patients with multiple and/or mixed valve disease may develop limiting symptoms or signs of physiologic impairment even with moderate valve lesions. Concomitant aortic and mitral valve replacement surgery is associated with a significantly higher perioperative mortality risk than replacement of either valve alone; therefore, operation should be carefully considered. Double valve replacement surgery is usu ally performed for treatment of severe (unrepairable) valve disease at both locations and for the combination of severe disease at one location with moderate disease at the other to avoid the hazards of reoperation in the intermediate to late term for progressive disease of the unoperated valve. In addition, the presence of a prosthesis in the aortic position significantly restricts surgical exposure of the native mitral valve. The need for double valve replacement may also impact the decision regarding the type of prosthesis (i.e., mechani cal vs tissue). In selected patients, TAVI for severe AS followed by edge-to-edge clip repair for severe MR can be accomplished during the same procedure. Tricuspid valve repair for moderate or severe secondary (func tional) TR at the time of left-sided valve surgery is now common place, particularly if there is dilation of the tricuspid annulus (>40 mm). The addition of tricuspid valve repair, consisting usu ally of insertion of an annuloplasty ring, adds little time or com plexity to the procedure and is well tolerated, but may be associated with an excess risk of heart block and the need for a permanent pacemaker. Reoperation for repair (or replacement) of progres sive TR years after initial surgery for left-sided valve disease, on the other hand, is associated with a relatively high perioperative mortality risk. Mitral valve repair or replacement for moderate or severe secondary MR at time of AVR for AS can usually be undertaken with acceptable risk for perioperative death or major complication. The presence of moderate or severe MR in patients with rheu matic MS is a contraindication to percutaneous mitral balloon commissurotomy (PMBC). TAVI can be performed for mixed AS and AR when the anatomic findings related to annulus size, coro nary height, and the distribution of calcium are favorable. Trans catheter management of both severe AS and severe primary or secondary MR (with deployment of an edge-to-edge clip) has been undertaken with increasing

frequency in appropriately selected patients with prohibitive or high surgical risk. Further advances in transcatheter treatments for multiple and mixed valve disease are anticipated. ■ ■FURTHER READING Alaour B et al: Combined significant aortic stenosis and mitral regur gitation: Challenges in timing and type of intervention. Can J Cardiol 40:235, 2024. Egbe AC et al: Outcomes in moderate mixed aortic valve disease: Is it time for a paradigm shift? J Am Coll Cardiol 67:2321, 2016. Gammie JS et al: Concomitant tricuspid repair in patients with degenerative mitral regurgitation. N Engl J Med 286:327,

)22.
Otto CM et al: 2020 AHA/ACC guidelines for management of patients with valvular heart disease. A report of the American Heart Association Joint Commission on Clinical Practice Guidelines. Circulation 143:e72, 2021.

Anne Marie Valente, Michael J. Landzberg

Congenital Heart Disease

in the Adult CHAPTER 280 ■ ■PREVALENCE The number of adults with congenital heart disease (CHD) living in the United States is estimated to be at least 1.4 million. It is now projected that >10% of adults living with CHD in Europe will be over 60 years old by 2030. The majority of adults with CHD were diagnosed in childhood, although a substantial percentage may have CHD first recognized as adults. Lifelong follow-up in coordination with, or directly by, clinicians with expertise in adult congenital heart disease (ACHD) is recommended. In this chapter, we will review the cur rent field of ACHD, with an introduction to CHD nomenclature and cardiac development. This is followed by a summary of the more com mon CHD lesions that may be diagnosed in adulthood. Lastly, some of the common repaired CHD lesions that are encountered in adults are discussed. Throughout the chapter, to aid in the understanding of con genital cardiac anatomy and physiology, we include figures displaying the passage of blood flow between blood vessels and cardiac chambers in various disorders (Fig. 280-1). Congenital Heart Disease in the Adult ■ ■THE CHANGING LANDSCAPE OF ADULT CHD A Relatively New Subspecialty in Cardiovascular Disease

Over the past two decades, the field of caring for adults with CHD has blossomed, and several nationwide initiatives have been initiated to standardize care. The American College of Cardiology and American Heart Association developed guidelines for the care of adults with CHD, first published in 2008 and revised in 2018, which emphasize the need for collaboration among primary care practitioners, cardiologists, and ACHD subspecialty cardiologists. The body of medical knowledge and competencies attendant with ACHD combined with skill acquisition in coordination of complex care over a patient’s medical lifetime led in 2015 to ACHD board certification examinations by the American Board of Medical Subspecialties, as well as the establishment of requirements for advanced fellowship training in ACHD care by the Accreditation Coun cil for Graduate Medical Education. In temporal association, the Adult Congenital Heart Association (ACHA) developed a process for ACHD care program accreditation based on standardization of infrastructural components felt requisite to achieve quality outcomes for ACHD. ■ ■SPECIAL CONSIDERATIONS FOR THE ACHD PATIENT Due to the need for lifelong care, it is essential that pediatric cardiology programs partner with ACHD programs to provide successful transi tion of patients. However, gaps in care are common during transition, much of which may be due to

disparities in social determinants of health, such as race, ethnicity, socioeconomic status, access to insur ance, and residence in geographically remote locations. Additionally, adults with CHD may not recognize subtle changes in their exercise capacity, some of which are associated with worse survival; by the time symptoms are recognized, irreversible physiologic changes may have occurred. ACHD patients are, therefore, advised to undergo regular evaluations for surveillance of anatomic, hemodynamic, and electro physiologic sequelae that may be present. In addition, specific situa tions may arise in which it is prudent to review care in consultation with an ACHD specialist, several of which are outlined below. Noncardiac Surgery Nearly all adults with CHD can be classi fied with stage A (harboring risk) or greater degrees of heart failure. As such, adults with CHD may demonstrate limited hemodynamic reserve to altered myocardial perfusion or loading conditions and may have subclinical organ dysfunction that is not recognized by standard laboratory assessment. Comprehensive, multispecialty assessment and care strategy review are recommended in advance of invasive or

Normal Heart Pulmonary artery PART 6 Disorders of the Cardiovascular System Aorta Right pulmonary veins Left pulmonary veins Left atrium Superior vena cava Right atrium Mitral valve Pulmonary valve Left ventricle Right ventricle Inferior vena cava Aortic valve Tricuspid valve FIGURE 280-1 Normal heart. Understanding of congenital cardiac anatomy and physiology is facilitated by use of box diagrams, displaying passage of blood flow between blood vessels and cardiac chambers. Labeling (e.g., structure names, arrows to denote direction of flow, coloring to represent oxygen saturation, connections or obstructions, chamber or vascular pressures, oxygen saturations) can aid in representation. Ao, aorta; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary artery; PV, pulmonary veins; RA, right atrium; RV, right ventricle; SVC, superior vena cava. operative procedures for adults with CHD. Figure 280-2 illustrates the multiorgan considerations that should be considered in adults with CHD during perioperative resuscitation and convalescence. Anesthetic management requires knowledge of anatomy, physiologic consequence of underlying defects, myocardial and vascular performance, presence and nature of previous palliative procedures and residual shunts, altera tion of venous or arterial pathways within the circulation, and status of noncardiovascular organ physiology. Pregnancy Women with CHD should receive counseling regard ing both maternal and fetal risks prior to conceiving a pregnancy and should be cared for in institutions with experience in treating CHD during pregnancy. Preconception evaluation includes detailed medical history, with particular attention to the women’s functional capacity, which is closely linked to maternal and fetal outcomes. Table 280-1 lists the World Health Organization classification of risk during preg nancy in women with heart disease; women at risk should be strongly counseled about the significant risks of morbidity and mortality during pregnancy and the postpartum period. Normal physiologic hemody namic changes of pregnancy are significant, occur over a relatively Hepatic Neurologic Congestive hepatopathy Neurocognitive dysfunction Hepatic fibrosis Stroke Renal Mood disorders (anxiety, depression) Chronic kidney disease Malignancy Endocrine Long-term exposure to ionizing radiation Hepatocellular carcinoma Diabetes Hypercholesterolaemia Obesity Immune/Infection Calcium metabolism and bone density SBE Abnormal immune function Thyroid disorders FIGURE 280-2 Noncardiac considerations in adults with congenital heart disease (CHD). SBE, subacute bacterial endocarditis. (Modified from J Buber et al: Common congenital heart problems in acute and intensive care. Eur Heart J Acute Cardiovasc Care 12:267, 2023.)

condensed period of time, and may be compounded in adults with CHD. Silversides and colleagues have devel oped a weighted-risk score for pregnant women with heart disease, based on a large registry known as CARPREG 2. The highest-weighted risk factors (weight of 3 points) include a prior his tory of cardiac events or arrhythmias, decreased functional status (New York Heart Association class ≥III), and pres ence of a mechanical heart valve. Risk factors that account for 2 points include ventricular dysfunction, high-risk leftsided valve disease/left ventricular outflow tract obstruction, pulmonary hypertension, coronary artery disease, and high-risk aortopathy. One point is assigned for late pregnancy assessment or no prior cardiac intervention. In this cohort, 16% of women experienced an adverse cardiac outcome, primarily heart failure and arrhythmia related. The predicted risks for cardiac events stratified according to point score were as follows: ≤1 point, 5%; 2 points, 10%; 3 points, 15%; 4 points, 22%; and >4 points, 41%. PV SVC IVC RA LA Mitral valve Tricuspid valve RV LV Pulmonary valve Aortic valve PA Ao Prepregnancy medications should be reviewed to ensure their safety in pregnancy. Alternatives to angiotensinconverting enzyme (ACE) inhibitors, angiotensin receptor blockers, direct oral anticoagulants, and endothelin receptor blockers should be considered, as these agents are teratogenic and contraindicated during pregnancy and should be discontinued. Women requiring anticoagula tion must be advised of the challenges of managing anticoagulation during pregnancy, and individualized strategies should be developed. A fetal echocardiogram between 18 and 22 weeks of gestation is advised for patients with CHD. Additionally, both men and women with CHD should be counseled regarding the risk of CHD in their offspring. ■ ■CONGENITAL TERMINOLOGY, DEVELOPMENT, AND GENETICS Congenital Nomenclature One of the challenges in caring for adults with CHD is the inconsistent terminology used to describe the congenital heart lesions. Several classification systems have been pro posed, from the initial descriptions by Maude Abbott, Maurice Lev, and Jesse Edwards, to the extensive characterizations by Stella and Richard Van Praagh and Robert Anderson. In this chapter, we follow a seg mental approach. The heart is composed of several segments that are Genetic Gastrointestinal Malnutrition Specific genetic syndromes associated with CHD Malabsorption Haematologic Airway Anaemia Thrombosis Congenital anomalies Small airways disease Coagulopathy Bleeding diathesis Pulmonary Frailty Restrictive lung disease Sarcopenia Obstructive sleep apnoea Deconditioning Pulmonary hypertension

TABLE 280-1 Modified World Health Organization (mWHO) Classification of Heart Disease in Pregnancy mWHO I mWHO II mWHO II–III mWHO III mWHO IV Small or mild Pulmonary stenosis Patent ductus arteriosus Mitral valve prolapse Successfully repaired simple lesions (atrial or ventricular septal defect, patent ductus arteriosus, anomalous pulmonary venous drainage) Atrial or ventricular ectopic beats, isolated Unoperated atrial or ventricular septal defect Repaired tetralogy of Fallot Most arrythmias (supraventricular arrhythmias) Turner syndrome without aortic dilatation Diagnosis (if otherwise well and uncomplicated) Risk No detectable increased risk of maternal mortality and no/mild increased risk in morbidity Small increased risk of maternal mortality or moderate increase in morbidity Abbreviations: ASI, aortic size index; EF, ejection fraction; HTAD, heritable thoracic aortic disease. analyzed separately before formulating a comprehensive diagnosis. The principal segments are the atria, the ventricles, and the great arteries, which are joined together by the atrioventricular canal and the conus (infundibulum). In the normal heart, the right ventricle (RV) is rightsided and organized inflow-to-outflow from right to left, while the left ventricle (LV) is left-sided and organized inflow-to-outflow from left to right. It is important to determine the segmental alignments, that is, what drains into what. For example, in the normal heart, the right atrium (RA) is aligned with the RV and the LV with the aorta. Finally, the

segmental connections, the way in which adjacent segments are physically linked to each other, are described. For example, in the normal heart, the pulmonary artery (PA) is connected to the RV by a complete muscular conus (infundibulum), while the aorta is connected to the LV by aortic-mitral fibrous continuity (without a complete conus). Alignment and connection are different concepts, and both are important, especially in complex defects. Cardiac Development The heart starts to form in the third week of gestation and is nearly fully formed by 8 weeks' gestation. Mesoder mal precardiac cells migrate to form the cardiac crescents (primary heart fields) in anterior lateral plate mesoderm, which are then brought together to form a primary linear heart tube by ventral closure of the embryo. Cells of the second heart field continue to proliferate outside the heart and are added to the heart tube over the course of embryo genesis, contributing to the atria, the RV, and outflow tract. Addition ally, cardiac neural crest cells migrate into the developing heart in the 5th–6th weeks and are essential for septation of the outflow, formation of the semilunar valves, and patterning of the aortic arches. Once formed, the heart tube grows and elongates by addition of cells from the second heart field. The ends of the heart tube are relatively fixed by the pericardial sac so that as it elongates it must loop (bend), and in the vast majority of hearts, the loop falls to the right (D-loop). Fur ther elongation pushes the mid-portion of the tube (future ventricles)

Mild left ventricular impairment (EF >45%) Hypertrophic cardiomyopathy Native or tissue valve disease not considered WHO I or IV (mild mitral stenosis, moderate aortic stenosis) Marfan or other HTAD syndrome without aortic dilatation Aorta <45 mm in bicuspid aortic valve pathology Repaired coarctation Atrioventricular septal defect Moderate left ventricular impairment (EF 30–45%) Previous peripartum cardiomyopathy without any residual left ventricular impairment Mechanical valve Systemic right ventricle with good or mildly decreased ventricular function Fontan circulation Fontan circulation with good clinical course and without associated comorbidities Unrepaired cyanotic heart disease Other complex heart disease Moderate mitral stenosis Severe asymptomatic aortic stenosis Moderate aortic dilatation

(40–45 mm in Marfan syndrome or other HTAD; 45–50 mm in bicuspid aortic valve, Turner syndrome ASI 20–25 mm/m2, tetralogy of Fallot <50 mm) Ventricular tachycardia Pulmonary arterial hypertension Severe systemic ventricular dysfunction (EF <30% or NYHA class III–IV) Previous peripartum cardiomyopathy with any residual left ventricular impairment Severe mitral stenosis Severe symptomatic aortic stenosis Systemic right ventricle with moderate or severely decreased ventricular function Severe aortic dilatation (>45 mm in Marfan syndrome or other HTAD,

> 50 mm in bicuspid aortic valve, Turner syndrome ASI >25 mm/m2, tetralogy of Fallot >50 mm) Vascular Ehlers-Danlos Severe (re)coarctation Fontan with any complication CHAPTER 280 Congenital Heart Disease in the Adult Intermediate increased risk of maternal mortality or moderate to severe increase in morbidity Significantly increased risk of maternal mortality or severe morbidity Extremely high risk of maternal mortality or severe morbidity inferior or caudal to the inflow, resulting in the normal relationship between the atria and ventricles. Further growth pushes the outflow medially and is associated with outflow

rotation, both processes essen tial for normal alignment of the outflow. Finally, the proximal part of the outflow is incorporated in the RV, shortening the outflow in asso ciation with further rotation. While this remodeling is occurring, the outflow is undergoing septation under the influence of cardiac neural crest cells. Septation proceeds from distal to proximal, culminating in formation and muscularization of the infundibular, or muscular, outflow septum, which inserts onto the superior endocardial cushion at the rightward rim of the outflow foramen, walling the aorta into the LV via the outflow foramen and the PA directly into the RV. Genetic Considerations CHD is the most commonly occur ring birth defect; etiologic contributors are increasingly recognized, although often speculated to be multifactorial. Children born with trisomy 21 have a 50% chance of having CHD, most commonly defects in the atrioventricular canal. Conotruncal defects are associated with several chromosomal abnormalities, most notably a deletion at chro mosome 22q11 (DiGeorge syndrome). Echocardiographic clues to this association in patients with a conotruncal defect include an associated right aortic arch or aberrant subclavian artery. Many adults currently living with conotruncal defects may not have undergone testing for DiGeorge syndrome. This condition is important to recognize because a variety of psychiatric disorders and disabilities in cognitive function may be present and go untreated. Patients with Noonan syndrome commonly have a dysplastic pulmonary valve and have facial and lymphatic abnormalities. Several defects in specific genes have been associated with Noonan syndrome, most notably PTPN11. Adults with Williams syndrome (7q11.23 deletion) commonly have supravalvar aortic stenosis and diffuse arteriopathy, with a “cocktail-like” personal ity and hypercalcemia. There is a growing importance of genome-wide analyses in subjects with CHD.

TABLE 280-2 Congenital Etiologies of Right Heart Dilation Congenital tricuspid valve disease Tricuspid valve dysplasia with regurgitation Ebstein anomaly Congenital pulmonary valve regurgitation Pulmonary arterial hypertension Myocardial abnormalities Arrhythmogenic RV cardiomyopathy Uhl’s anomaly Shunt lesions Partial anomalous pulmonary venous return Primum ASD Secundum ASD Sinus venosus defect Coronary sinus septal defect Gerbode defect (LV-RA shunt) Coronary artery fistula to the RA, CS Postoperative residual shunts PART 6 Disorders of the Cardiovascular System Abbreviations: ASD, atrial septal defect; CS, coronary sinum; LV, left ventricle; RA, right atrium; RV, right ventricle. ■ ■SPECIFIC CHD LESIONS Dilated Right Heart There are many congenital etiologies for right heart dilation (Table 280-2). These include congenital valvular anomalies (such as Ebstein anomaly or pulmonary regurgitation), intrinsic RV myocardial anomalies (arrhythmogenic RV dysplasia, Uhl’s anomaly), or shunt lesions occurring proximal to the tricuspid valve (atrial septal defects or partial anomalous pulmonary veins). Cardiac imaging is critical in determining the etiology of right heart dilation, and knowledge of the anatomy and physiology of various shunt lesions is essential. Atrial Septal Defect One of the most common etiologies of right heart dilation is presence of an atrial septal defect (ASD; Fig. 280-3A). Intracardiac communications allow blood transmission between chambers or spaces based on relative resistance, propulsion, and flow patterns. Patients with large ASDs often present in child

hood; however, many ASDs are not discovered until adult life. The physiology of an ASD is predominantly that of a “left-to-right” shunt Atrial Septal Defect SVC IVC x x+y PA Ao Right PVs SVC x Left PVs x x+y ASD y LA x RA x+y x LV x+y RV IVC A FIGURE 280-3 A. Atrial septal defect. In the presence of an atrial septal defect, the difference in compliance between the (RA + RV) as compared to the (LA + LV), combined with the size of the defect itself, allows for a “shunt” of flow (“y”) of “red” (oxygenated) blood from the left side of the heart to the right side (deoxygenated). Systemic venous return of pure deoxygenated blood (“x”) is increased by the oxygenated shunted blood (“y”) to increase volume of blood (“x + y”) in the RA, RV, and total blood flow to the lungs. If the volume or the sequelae of the shunted blood are sufficient, RA and RV can dilate (hashed lines), and arrhythmias or shortness of breath (and occasionally pulmonary hypertension) can ensue. Ao, aorta; ASD, atrial septal defect; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary artery; PV, pulmonary veins; RA, right atrium; RV, right ventricle; SVC, superior vena cava. B. Diagrammatic representation of the location of various atrial septal defects. ASD 1, primum atrial septal defect; ASD 2, secundum atrial septal defect. (Part B used with permission from Emily Flynn McIntosh, illustrator.)

(flow of pulmonary venous, or oxygenated, blood toward systemic venous, or deoxygenated, chambers or vessels). The degree of left-toright shunting determines the amount of right heart volume loading and is dictated by the size of the defect as well as the diastolic prop erties of the heart. As patients age, several factors, such as diabetes mellitus, systemic hypertension, and atherosclerosis, may contribute to decreased compliance of the left-sided cardiac chambers and contribute to increased left-to-right shunting and symptomatology. The classic physical examination finding is a wide, fixed splitting of the second heart sound, which is due to prolonged RV ejection and increased PA capacitance, which, in turn, delay pulmonary valve closure. The surface electrocardiogram (ECG) commonly displays an incomplete right bundle branch block. Symptoms, when they occur, most commonly include exercise intolerance, arrhythmia, and dyspnea with exertion. It is not uncommon for adults to have incidentally noted asymptomatic ASD during evaluation of other comorbid issues. Right heart dilation, without additional etiology for such, in the setting of unrepaired ASD is considered a risk for pro gression toward symptomatic right heart failure, atrial arrhythmias, and potential development of pulmonary arterial hypertension (if such is not already present). Therefore, a patient with an ASD and right heart dilation, particularly with symptoms attributable to such, should be offered ASD closure. Pulmonary vascular disease leading to pulmonary hypertension develops in up to 10% of patients with unrepaired ASD, and Eisenmenger syndrome (ES) is a rare complica tion (see below). Management of patients with concomitant ASD and pulmonary hypertension should be coordinated with both ACHD and pulmonary hypertension experts. Figure 280-3B illustrates the locations of various ASDs. The most common type of an ASD is a secundum ASD, which is a defect, or true deficiency in the atrial septum, in the region of the fossa ovalis. This should be differentiated from a patent foramen ovale (PFO), which is persistence of patency of the flap valve of the fossa ovalis (not associ ated with right-sided cardiac dilation) and persists in up to 25% of adults. Secundum ASDs can often be closed with occluder devices placed percutaneously. However, certain anatomic determinants make percutaneous closure less favorable, including large defects, inade quate tissue rims surrounding the defect, and concomitance of anom alous draining pulmonary veins. A primum ASD is a deficiency of the atrioventricular (AV) canal portion of the atrial septum; primum ASD is always associated with abnormal development of the AV valves, PV ASD y RA LA x+y x Sinus venosus defect ASD 1° RV LV x+y x x+y x PA Ao ASD 2° B

most commonly resulting in a cleft in the mitral valve. A coronary sinus defect is rare and involves an opening between the coronary sinus and the left atrium. A sinus venosus defect is not a defect in the atrial septum but, rather, a defect between either the right superior vena caval–atrial junc tion and the right upper pulmonary vein(s) or, less commonly, the inferior vena caval–atrial junction and the right lower pulmonary veins. Surgical closure is required for primum ASDs, sinus venosus defects, and coronary sinus septal defects. APV APV Right PVs Partial Anomalous Pulmonary Venous Return Partial anomalous pulmonary venous return (PAPVR) is occasionally discovered in adults with right heart dilation or incidentally on cross-sectional imaging (Fig. 280-4). There are several possible anomalous connections, with the most common being a left upper pulmonary vein to an ascending vertical vein into the innominate vein or the right upper pulmonary vein draining to the superior vena cava. In the latter case, care ful attention should be paid to ensure that there is not an associated sinus venosus defect. Con comitant pulmonary hypertension can occur but is uncommon. Symptomatology may be absent, and a decision to repair isolated PAPVR should include variance in anatomy, lung ventilation and perfusion, hemodynamic response to shunt, symptoms, and surgical experience. FIGURE 280-4 Partial anomalous pulmonary venous return. In the presence of an anomalously draining pulmonary vein (typically to a systemic vein such as the left innominate vein, SVC, or rarely IVC), an obligate “shunt” of flow (“y”) of “red” (oxygenated) blood from the affected pulmonary vein to the right heart (deoxygenated) ensues. Systemic venous return of pure deoxygenated blood (“x”) is increased by the oxygenated shunted blood (“y”) to increase volume of blood (“x + y”) in the SVC, RA, RV, and total blood flow to the lungs. If the volume or the sequelae of the shunted blood are sufficient, RA and RV can dilate (hashed lines), or shortness of breath can ensue. Ao, aorta; APV, anomalous pulmonary vein; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary arteries; PV, pulmonary veins; RA, right atrium; RV, right ventricle; SVC, superior vena cava. Ebstein Anomaly Ebstein anomaly (Fig. 280-5) is the result of embryologic failure of delamination, or “peeling away,” of the tricuspid valve leaflets from the ventricular myocardium, resulting in adher ence of the valve leaflets to the underlying myocardium. This results in a wide variety of abnormalities, including apical and posterior Ebstein Malformation SVC IVC PA Ao Right PVs x+y x+z x+z x SVC Left PVs x LA PFO x+z z RA x+y x+z y LV RV x+y IVC FIGURE 280-5 Ebstein malformation. In the presence of Ebstein anomaly, the tricuspid valve leaflets can be redundant, fenestrated, and sail-like (typically seen in the anterior leaflet*) or adherent to the underlying myocardium with apical displacement of the nonadherent components (typically the septal and posterior leaflets). Location and degree of leaflet coaptation are variable and account for varying degrees of tricuspid regurgitation, shift of the functional tricuspid valve anterior from the anatomic annulus into the right ventricle, “atrialization” of the right ventricle, and most commonly angulation of the tricuspid valve into the RV outflow tract. RA and RV dilation (hashed lines) can occur due to the effects of combined volume from systemic venous return (“x”) and tricuspid regurgitant flow (“y”). PFO is frequent; worsening compliance and elevation of pressure in the RA as compared to the LA can lead to increasing “right-to-left” (deoxygenated to oxygenated) shunt and cyanosis. RV myocardial function may be abnormal. Ao, aorta; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary arteries; PFO, patent foramen ovale; PV, pulmonary veins; RA, right atrium; RV, right ventricle; SVC, superior vena cava; *, anterior tricuspid valve leaflet.

Partial Anomalous Pulmonary Venous Return PV SVC CHAPTER 280 y x IVC x x+y y RA LA PA Ao x+y x x x+y Left PVs SVC Congenital Heart Disease in the Adult LA RV LV x x+y x RA x+y x x+y x x+y LV PA Ao RV IVC displacement of the dilated tricuspid valve annulus, dilation of the “atrialized”

portion of the RV, and fenestrations, redundancy, and tethering typically of the anterior leaflet of the tricuspid valve. The malformed tricuspid valve is usually regurgitant but may occasionally be stenotic. The clinical presentation of Ebstein anomaly in the adult depends on several factors, including the extent of tricuspid valve leaflet distortion, degree of tricuspid regurgitation (TR), right atrial pressure, and presence of an atrial level shunt. The physical examination of a patient with Ebstein anomaly may vary depending on the severity of disease. In more severe cases, the first heart sound may be split and the second component of the first heart sound may have a distinctive snapping quality (known as the sail sign, due to the redundancy of the anterior tricuspid valve leaflet). Patients with significant TR may have prominent “v” waves of the jugular venous pulsations; however, this find ing is often absent due to abnormal right atrial compliance. The ECG is often abnormal, with right atrial and ventricular enlargement. Up to 20% of patients have evidence of ventricular pre excitation (Wolff-Parkinson-White pattern). Sur gical treatment includes a tricuspid valve repair or replacement, closure of any atrial level defects, and arrhythmia ablative procedures. PV PFO x x z RA LA y * RV LV x+z x+y x+z x PA Ao Shunt Lesions Causing Left Heart Dilation Intracardiac shunts or intravascu lar passages that occur below the level of the tricuspid valve result in left heart dilation. The two major types of congenital shunts that result in left heart dilation are a ventricular septal defect (VSD; Fig. 280-6A) and patent ductus arteriosus (PDA; Fig. 280-7). Ventricular Septal Defects VSDs are the most common congenital anomaly recognized at

Ventricular Septal Defect SVC IVC x x+y PART 6 Disorders of the Cardiovascular System PA Ao Right PVs SVC x x x+y Left PVs LA x+y RA x y x+y LV RV x IVC A FIGURE 280-6 A. Ventricular septal defect. In the presence of a ventricular septal defect, the difference in pressure and outflow resistance in systole (and the difference in compliance in diastole) between the RV and LV, combined with the size of the defect itself, allow for a “shunt” of flow (“y”) of “red” (oxygenated) blood from the left side of the heart to the right side (deoxygenated). Systemic venous return of pure deoxygenated blood (“x”) is increased by the oxygenated shunted blood (“y”) to increase volume of blood (“x + y”) through the outflow of the RV into the lungs, and in the left atrium and left ventricle. If the volume or the sequelae of the shunted blood are sufficient, LA and LV can dilate (hashed lines), and arrhythmias or shortness of breath (and occasionally pulmonary hypertension) can ensue. Ao, aorta; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary arteries; PV, pulmonary veins; RA, right atrium; RV, right ventricle; SVC, superior vena cava; VSD, ventricular septal defect. B. Diagrammatic representation of the location of various ventricular septal defects. AV, atrioventricular. (Part B used with permission from Emily Flynn McIntosh, illustrator.) birth; however, they account for only ~10% of CHD in the adult, due to the high rate of spontaneous closure of small VSDs during the early years of life. Large VSDs usually cause symptoms of heart failure and poor somatic growth and are most often surgically closed before adult hood. Several classification systems for VSDs exist. Figure 280-6B illustrates various locations of VSDs; the most common location is in the membranous septum (also referred to as perimembranous or outlet defects). Muscular defects that persist into adult life are often pressure and flow restricted, resulting in no significant hemodynamic conse quence. AV canal defects, also referred to as inlet defects, are located in the crux of the heart and are associated with abnormalities of the AV valve leaflets. Subpulmonary defects, also known as conal septal defects, are commonly associated with prolapse of the right coronary Patent Ductus Arteriosus y Ao PA Right PVs x SVC x LA x+y RA x x+y x LV RV IVC FIGURE 280-7 Patent ductus arteriosus. In the presence of a patent ductus arteriosus, the difference in pressure and resistance in both systole and diastole between the pulmonary arteries and the aorta, combined with the size of the ductus

itself, allow for a “shunt” of flow (“y”) of “red” (oxygenated) blood from the aorta to the pulmonary arteries (deoxygenated). Systemic venous return of pure deoxygenated blood (“x”) is increased by the oxygenated shunted blood (“y”) to increase volume of blood (“x + y”) in the lungs, the left atrium, the left ventricle, and out the aortic valve. If the volume or the sequelae of the shunted blood are sufficient, LA and LV can dilate (hashed lines), and arrhythmias or shortness of breath (and occasionally pulmonary hypertension) can ensue. Ao, aorta; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary arteries; PDA, patent ductus arteriosus; PV, pulmonary veins; RA, right atrium; RV, right ventricle; SVC, superior vena cava.

PV RA LA x x+y Subpulmonary RV LV Membranous x+y y x VSD x x+y AV canal type PA Ao Muscular B cusp and aortic insufficiency. The outcome for adults with small VSDs without evidence of ventricular dilation or pulmonary hypertension is generally excellent. Patent Ductus Arteriosus A PDA courses between the aortic isthmus and the origin of one of the branch PAs. Small PDAs are often silent to auscultation and do not cause hemodynamic changes. The classic murmur is heard best just below the left clavicle and typically extends from systole past the second heart sound into diastole, reflect ing flow turbulence and gradient between the aorta and the PAs (result ing in left-to-right shunting). Large PDAs will lead to left heart dilation and may lead to chronically elevated pulmonary vascular resistance, including the potential for ES. PV SVC PDA IVC x x+y RA LA x x+y Left PVs RV LV x x+y x x+y y PDA x x+y PA Ao

■ ■MODERATE AND COMPLEX CHD Tetralogy of Fallot Tetralogy of Fallot (TOF) is the most common form of cyanotic CHD, occurring in 0.5 per 1000 live births. It involves anterior deviation of the conal septum, resulting in RV outflow tract (RVOT) obstruction, a VSD, RV hypertrophy, and an overriding aorta (Fig. 280-8A, B). There is a large spectrum of severity of disease in TOF, from patients who have only mild pulmonary stenosis to those with complete pulmonary atresia (TOF/PA). Current surgical strategies involve primary repair in infancy (Fig. 280-8C); however, many adults Tetralogy of Fallot (unrepaired) PV SVC IVC x x-y RA LA x x-y Right PVs

SVC RV LV x x-y x y RVH

-

VSD RA x x-y x x PA Ao IVC A Tetralogy of Fallot (repaired) PA Ao Right PVs x SVC x x x x+y RA y x x+y LV RV IVC C FIGURE 280-8 A. Tetralogy of Fallot involves anterior and superior malalignment of a bar of tissue (conal septum) (see *in part B, which presents a cut-away view through the anterior surface of the RV, into the RV outflow), partially obstructing the right ventricular outflow (under the pulmonary valve, i.e., “subpulmonary stenosis”; labeled as 1), and leaving a gap in the interventricular septum (VSD). The pulmonary valve annulus is typically hypoplastic. Outflow obstruction prevents regression of right ventricular hypertrophy (#), which was present in utero. The difference in pressure and outflow resistance in systole (and the difference in compliance in diastole) between the obstructed RV and the LV allows for a “shunt” of flow (“y”) of “blue” (deoxygenated) blood from the right side of the heart to the left side (oxygenated). Systemic venous return of pure deoxygenated blood (“x”) is decreased by the shunted blood (“y”), leading to a total decrease in the volume of blood (“x – y”) passing beyond into the lungs. The deoxygenated shunted blood (“y”) mixes with fully oxygenated blood in the LV, contributing to systemic arterial cyanosis. C. Tetralogy of Fallot—repaired. After modern repair of tetralogy of

Fallot, VSD has been patched closed, and outflow tract obstruction has been surgically removed, frequently at the expense of a patch enlarging the pulmonary valve annulus at the expense of sacrificing the integrity of the pulmonary valve (causing pulmonary regurgitation). The pulmonary regurgitant volume (“y”) is added to systemic venous return (“x”), contributing to RV chamber enlargement (hashed lines) and may be associated with tricuspid annular dilation and valve regurgitation, resulting in RA enlargement. Ao, aorta; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary arteries; PV, pulmonary veins; RA, right atrium; RV, right ventricle; RVH, right ventricular hypertrophy; SVC, superior vena cava; VSD, ventricular septal defect.

may have first undergone palliative procedures (Blalock-Taussig, Potts, Waterston shunts) prior to a complete repair. The goal of surgical repair is to alleviate the pulmonary stenosis and close the VSD. Up to 10% of patients with TOF have an anomalous coronary artery, most com monly, an anomalous left anterior descending coronary artery from the right coronary cusp. Patients with an anomalous coronary as well as those with TOF/PA may require an RV-to-PA conduit.

CHAPTER 280 Adults with repaired TOF often have hemodynamic sequelae that may require reintervention in adulthood (Table 280-3). Pulmonary Congenital Heart Disease in the Adult Conal Anatomy PA Ao x-y Left PVs LA * x-y VSD *

x-y

LV y x-y RV x

B PV SVC IVC x x RA LA x+y x Left PVs RV LV x+y x LA x VSD patch VSD patch x+y x x PA Ao

TABLE 280-3 Potential Sequelae of Repaired Tetralogy of Fallot Right atrial dilation Right ventricular dilation Right ventricular dysfunction Right ventricular outflow tract obstruction Pulmonary regurgitation Branch pulmonary artery stenosis Tricuspid regurgitation Residual ventricular septal defect Left ventricular dysfunction Aortic root dilation Atrial arrhythmias Ventricular arrhythmias Sudden cardiac death PART 6 Disorders of the Cardiovascular System regurgitation is common following TOF repair and is usually associ ated with RV dilation. Accurate quantification of RV size, function, and mass is particularly important in adults after repair of TOF, as RV dilation, dysfunction, and hypertrophy are associated with adverse out comes in these patients. Patients may also have residual RVOT obstruc tion, which may occur beneath the pulmonary valve, at the valve level, above the valve, or in the branch PAs. Cardiac magnetic resonance imaging is routinely used in the surveillance of these patients. Left ven tricular dysfunction is present in at least 20% of adults with repaired TOF, particularly those who were repaired later in life, had prior pal liative shunts, or have concomitant RV dysfunction. As patients age with repaired TOF, both atrial and ventricular arrhythmias occur with increasing frequency. A QRS duration on a D-loop Transposition PA Ao Right PVs SVC LA RA RV IVC A FIGURE 280-9 A. Transposition of the great arteries. When the great arteries are transposed, the aorta arises from the RV, and the pulmonary artery arises from the LV, leaving deoxygenated blood circulating from systemic veins to systemic arteries in separated fashion from oxygenated blood, which circulates from pulmonary veins to pulmonary arteries. Without interchamber or intravascular communications, this circulation is incompatible with life. Presence of an atrial septal defect (ASD), depicted here, ventricular septal defect (VSD),

or patent ductus arteriosus (PDA) allows for some interchamber or intravascular mixing and, at best, partial relief of cyanosis and sustenance of life, at the expense of increased pulmonary blood flow. B. Atrial switch. Atrial level switch procedures (Mustard and Senning) were the first standardized surgeries to alter the natural course of complex congenital heart disease, utilizing intracardiac rerouting via a “baffle” to redirect blood flow. The atrial switch simulates inverted trousers, with each “pants leg” (*) attaching to either the SVC or the IVC, transporting deoxygenated blood through the interior of the trousers to the “waist of the trousers” and directing blood through the mitral valve to the LV and out the PA. Surgical removal of the atrial septum allows pulmonary venous return to traverse from posterior left atrium through the space between the pants legs of the baffle, through the tricuspid valve to the RV (serving as the “systemic ventricle,” i.e., that pumps to the systemic arterial circulation), through the aorta. Non-infrequent sequelae include sinus node dysfunction, atrial arrhythmias, systolic dysfunction of the RV, tricuspid regurgitation (from RV to LA), leaks in the baffle material allowing shunting of blood, and obstruction of the systemic or pulmonary venous baffles. C. Arterial switch. The arterial switch operation allowed both anatomic and physiologic correction for D-loop transposition of the great arteries. Successful surgical switching of the PA and the Ao above the level of the native roots (hashed lines) necessitated ability to transfer coronary artery origins contained within a button of tissue (*) back to the neo-aorta (now supported by the LV). Deoxygenated blood flow from SVC and IVC passes from RA to RV to PA, and oxygenated blood passes from PV to LA to LV to Ao. Uncommon sequelae include obstruction at any of the surgical sites (supravalvar PA or Ao stenosis, coronary orifice obstruction) or more distal obstructions due to tension placed on the PA, Ao, or coronary arteries. Ao, aorta; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary arteries; PV, pulmonary veins; RA, right atrium; RV, right ventricle; SVC, superior vena cava.

resting ECG of 180 ms or more has been associated with increased risk of ventricular tachycardia and sudden death in this patient popula tion. In one prospective follow-up study of 144 adults with repaired TOF, there was a 72% survival at 40 years, but only a 25% cumula tive event-free survival. These events include need for reintervention (most commonly pulmonary valve replacement [PVR]), symptomatic arrhythmias, and heart failure. The most common reintervention in a repaired TOF patient is a PVR. However, optimal timing of PVR in asymptomatic patients with repaired TOF remains unclear. Traditionally, PVR has been accomplished with a surgical procedure; however, percutaneous implantation of pulmonary valves is becoming increasingly utilized in clinical practice. Patients with repaired TOF may also undergo interventions includ ing closure of residual VSDs, dilation and/or stenting of the RVOT or branch PAs, and tricuspid valve repair. Patients with clinically signifi cant arrhythmias may benefit from catheter ablation. Transposition of the Great Arteries Transposition of the great arteries (TGA) is defined by the great arteries arising from the opposite side of the ventricular septum than normal; as such, the aorta arises from the RV and the PA from the LV. The more common form of TGA, known as D-loop TGA, involves AV concordance and ventriculararterial discordance, resulting in a physiology that allows two circuits to be in parallel rather than in series (Fig. 280-9A) and intense cyano sis shortly after birth. This physiology is not compatible with long-term survival without surgical intervention. Patients with TGA may be born with additional congenital defects (most commonly a VSD). The surgical repairs for D-loop TGA have evolved over time. In the late 1950s through the 1970s, the atrial switch procedure (Mustard, Senning procedures) was performed (Fig. 280-9B). These atrial switch PV SVC ASD IVC RA LA Left PVs RV LV Ao PA LV

Atrial Switch PA Ao Right PVs SVC RA RV IVC B Arterial Switch neo Ao Right PVs neo PA LA SVC oo oo * * RA RV IVC C FIGURE 280-9 (Continued) procedures relieved the cyanosis but left the patient with a systemic RV. Despite moderate-term survival over decades, there are multiple longterm sequelae that may present following the atrial switch procedure. The most worrisome complication is that of systemic RV dysfunc tion. The prevalence of RV dysfunction in this population is not well defined. Limited study has failed to reveal medical therapies effective for systemic RV dysfunction. A subset of patients with D-loop TGA, VSD, and PS may have undergone a Rastelli procedure. This intervention involves placing an RV-to-PA conduit and routing the LV to the aorta through the VSD, which results in relief of cyanosis and the benefit of a systemic LV. In the 1980s, the arterial switch operation (ASO; Fig. 280-9C) became the surgical procedure of choice for D-loop TGA. This pro cedure involves transecting the great arteries above the sinuses and placing the PAs anteriorly to come into alignment with the RV, result ing in draping of the branch PAs over the ascending aorta. A coronary artery translocation is performed. The ASO has resulted in substantial long-term survival. The potential long-term sequelae of the various surgical procedures for D-loop TGA are listed in Table 280-4.

PV LA CHAPTER 280 SVC IVC Left PVs RV LV LA Congenital Heart Disease in the Adult Ao PA LV PV SVC IVC RA LA Left PVs RV LV neo PA neo Ao LV The less common form of TGA, known as L-loop TGA (physiologi cally corrected or “congenitally corrected” TGA; Fig. 280-10), may not require surgical intervention but is presented here in relation to other forms of TGA. L-loop TGA involves both AV discordance (RA allow ing passage of deoxygenated systemic venous return to the LV, and TABLE 280-4 Long-Term Sequelae of D-Loop TGA Surgery ATRIAL SWITCH ARTERIAL SWITCH RASTELLI PROCEDURE Systemic venous baffle Arterial anastomosis stenosis Subaortic stenosis Pulmonary venous baffle Branch PA stenosis RV-PA conduit obstruction RV (systemic) dysfunction Neo-aortic root dilation Pulmonary regurgitation Tricuspid regurgitation Neo-aortic regurgitation Ventricular dysfunction Baffle leaks Coronary artery stenosis LVOT obstruction (PS) LV dysfunction Abbreviations: LV, left ventricle; LVOT, left ventricular outflow tract; PA, pulmonary artery; PS, pulmonary stenosis; RV, right ventricle; TGA, transposition of the great arteries.

Congenitally (L-loop transposition) Corrected TGA PART 6 Disorders of the Cardiovascular System PA Ao Right PVs SVC LA RA LV IVC FIGURE 280-10 Congenitally corrected transposition of the great arteries. Physiologically corrected transposition of the great arteries (also known as congenitally corrected transposition of the great arteries) is characterized by atrioventricular discordance and ventriculoarterial discordance. Systemic venous blood passes from the right atrium (RA) through the mitral valve into the morphologic left ventricle (LV) to the pulmonary artery (PA). Oxygenated blood then returns to the lungs to the left atrium (LA) through the tricuspid valve into the morphologic right ventricle (RV) and then out the aorta (Ao). IVC, inferior vena cava; PV, pulmonary veins; SVC, superior vena cava. conversely, the left atrium conducting oxygenated pulmonary venous blood to the RV) as well as ventriculoarterial discordance (connections of LV to PA, RV to aorta). This results in normal arterial oxygen satu ration, yet an RV associated with the aorta. Patients with L-loop TGA commonly have associated congenital anomalies, including dextrocar dia, ASDs, a dysplastic tricuspid valve, and pulmonary stenosis. Con duction disturbances are common, and complete heart block occurs in up to 30% of patients. Those patients without associated defects may not present until later in life, most commonly with heart failure, TR, or newly recognized conduction disease. Coarctation of the Aorta Adults with coarctation of the aorta (Fig. 280-11) typically have a shelf-like obstruction at the level of the descending aorta that passes just

posterior to the junction of the main and left PA; obstruction less commonly involves the transverse aortic arch. On physical examination, the lower extremity blood pres sure and pulses are lower than (and delayed in timing, in contrast to) the upper extremity values, unless significant aortic collaterals have developed. A continuous murmur over the scapula may be present due to the collateral blood flow. Significant coarctation increases afterload to all proximal structures in the path of oxygenated blood, from LV and coronary arteries to ascending and transverse aorta, to cerebral and arm vessels and proximal descending aorta. Bicuspid aortic valve (typically with right-left commissural fusion) is a common association. In women with short stature, webbed neck, lymphedema, and primary amenorrhea, a concomitant diagnosis of Turner syndrome should be considered, the presence of which indicates greater degree of, and risks from, sequelae from seemingly similar anatomy and physiology. Patients who have undergone surgical repair in general have a good prognosis; however, they remain at risk for systemic hypertension, premature atherosclerosis, LV failure, and aortic aneurysm, dissection, and recurrent coarctation. Single Ventricle The term single ventricle heart disease is impre cise but useful in some settings, as it refers to congenital heart condi tions in which one ventricle or its valves preclude surgical creation of a biventricular circulation. Common congenital diagnoses in this category include tricuspid atresia, double inlet LV, and hypoplastic left heart syndrome. Most patients with single ventricle physiology undergo a series of surgeries culminating in a Fontan procedure (Fig. 280-12A, B). Since its initial use for tricuspid valve atresia in

PV SVC IVC RA LA Left PVs LV RV PA Ao RV 1971, multiple modifications of this procedure have occurred, with common features of near complete separation of the pulmonary and systemic circulations. The Fontan procedure utilizes the single ventri cle to pump pulmonary venous (oxygenated) blood through the aorta to the body and allows for “passive” flow of systemic venous return of deoxygenated blood through surgically created connections to the lungs. Patients who have undergone a Fontan procedure are at risk for multiple comorbidities in adulthood, including atrial arrhythmias, heart failure, renal and hepatic dysfunction, and both venous and arte rial thrombosis and embolism. Coarctation of the Aorta: Sequelae/Associations

PA *

LA Ao

RA

LV

RV FIGURE 280-11 Aortic coarctation (*). Bicuspid aortic valve (1) is most common concomitant lesion. Sequelae from aortic coarctation (unrepaired or repaired) include systemic arterial hypertension, ascending (2) or descending (3) aortic enlargement or aneurysm formation, left ventricular (LV) hypertrophy (4), LV diastolic and systolic heart failure, accelerated coronary (5) or cerebral (6) atherosclerosis, cerebral aneurysm formation, and recurrence of coarctation after repair. Ao, aorta; PA, pulmonary arteries.

Fontan PA Ao RA LA SVC RA * LV RV IVC A Atriopulmonary Fontan Classic Fontan Extracardiac Fontan Lateral tunnel Fontan B FIGURE 280-12 A. Fontan surgery creates a unique circulation in

which deoxygenated blood is directed to the PAs from the SVC and IVC in a fashion that bypasses any pumping chamber. The SVC and IVC are connected (*) via either an internal “tunnel” or an extracardiac conduit that guides flow to the PA. Pulmonary venous (oxygenated) return courses from PV to LA to LV to aorta. In contrast to physiology in normal adults (where pressure is generated by an RV to propel blood flow from a lower pressure RA to a higher pressure LA), in Fontan circulation, by definition, due to the absence of a pumping chamber to the PA, RA pressure is greater than LA pressure, permitting flow through the lungs. Ao, aorta; IVC, inferior vena cava; LA, left atrium; LV, left ventricle; PA, pulmonary arteries; PV, pulmonary veins; SVC, superior vena cava; *, Fontan baffle. B. Diagrammatic representation of the location of various types of Fontan operations. (Part B used with permission from Emily Flynn McIntosh, illustrator.)

PV SVC Extracardiac conduit CHAPTER 280 IVC LA * * LV Congenital Heart Disease in the Adult PA Ao

---

Revision #1
Created 2026-01-06 16:33:55 UTC by Omar Ayman
Updated 2026-01-06 16:33:55 UTC by Omar Ayman