

Challenges Posed by Adults With Repaired Congenital Heart Disease

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“Pediatric Congenital Cardiac Becomes a Postoperative Adult: The Changing Population of Congenital Heart Disease,” which was written in 1973,¹ was the first publication devoted to what was destined to become a new field of special cardiovascular interest. Subsequent decades witnessed the maturity of this new subspecialty, which was formally recognized at the 22nd Bethesda Conference in 1990.² It is altogether fitting for *Circulation* to include the present article in “Clinical Cardiology: New Frontiers.” The present article can be considered a sequel to the 1973 article, which dealt with the types of surgical interventions, the effects of those interventions on survival patterns, the desirability of primary anatomic repair in infancy, and the importance of postoperative residua and sequelae.¹ The 1973 article opened by stating: “It is now possible to perform palliative or corrective surgery on almost all congenital cardiac anomalies, even the most complex . . . Survival patterns are affected, often profoundly. We are therefore confronted with a changing population of congenital cardiacs . . . However, we are obliged to look beyond the present and define our ultimate goal. What do we seek to accomplish? The answer is clear. Our efforts should focus on the quality of long-term survival.”¹

Long-term survival is chiefly concerned with the growing number of postoperative patients who require continuing medical surveillance.³ What is needed is a new generation of cardiologists with a career interest in adult congenital heart disease; these highly trained specialists function best within tertiary care facilities designed for the comprehensive care of adult patients. With few exceptions, such facilities are part of major university hospital systems. Major facilities draw patients regionally, nationally, and internationally. The geographic distribution of tertiary care centers will best be determined by the quality of the facility and the services offered rather than by external constraints or government mandate.

Historical Perspective

Ligation of patent ductus arteriosus,⁴ resection of aortic coarctation,⁵ and the legendary Blalock-Taussig anastomosis⁶ had a tremendous impact on the emerging field of cardiac surgery. In 1956, Andre F. Cournand, Dickenson W. Richards, and Werner Forssmann shared the Nobel Prize in

Medicine and Physiology “for their discoveries concerning heart catheterization and pathological changes in the circulatory system.”⁷ John H. Gibbon’s heart/lung bypass machine, which was developed in the early 1950s, set the stage for intracardiac surgery using a mechanical pump-oxygenator⁸ that permitted “accurate visualization of structures within the heart for a period sufficient to permit precise corrective measures.”⁹ Each of these innovations addressed congenital cardiac malformations and heralded one of the most successful rehabilitation programs in medical history. The last half of the 20th century witnessed remarkable diagnostic and therapeutic developments in this field, permitting accurate anatomic and physiological cardiac diagnoses and astonishing feats of reparative surgery. Because of the impact on survival, congenital heart disease should be looked on not only in terms of age of onset, but as a continuum from fetal life to advanced age.³

Congenital heart disease in adults is a recognized cardiovascular subspecialty. The stage was formally set for it to become a subspecialty by the 22nd Bethesda conference in 1990, “Congenital Heart Disease After Pediatrics: An Expanding Patient Population.”² A decade later, the 32nd Bethesda conference, “Care of the Adult with Congenital Heart Disease” followed suit. Worldwide recognition is reflected by the existence of the International Society for Adult Congenital Cardiac Disease.

Incidence

The incidence of congenital heart disease, both worldwide and regionally, is necessarily an estimate that, not surprisingly, is being reassessed even as we write. Incidence must be judged according to certain constraints, namely, live births versus stillborn fetuses, full-term versus preterm births, inclusion or exclusion of relatively high-incidence malformations (such as the bicuspid aortic valve or ventricular septal defect destined to close spontaneously within 6 months of life), whether or not congenital heart disease is confined to gross structural abnormalities (ie, congenital complete heart block), and the diagnostic technique used to identify a given malformation. Despite these qualifications, the worldwide and geographically specific incidence of congenital heart

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disease among live-born, full-term infants has averaged 4 to 9 per thousand (0.4% to 0.9%).^{10–17}

There are ≈32 000 new cases of congenital heart disease per year in the United States and ≈1.5 million new cases worldwide.^{11,12,16} Approximately 20 000 open-heart operations are performed annually in the United States on patients with these congenital defects, and >85% of infants so afflicted can now expect to reach adulthood.¹⁰ These favorable figures can be attributed chiefly to the success of cardiac surgery in neonates, infants, and children. Interestingly, the patient population reaching reproductive age is yielding a maternal recurrence rate of 2.5% to 18% and a paternal recurrence rate of 1.5% to 3%, which is higher than the prevalence in the general population. Maternal exposure to environmental factors is held responsible for no more than 2% of congenital cardiovascular malformations.¹⁵

The benefits of surgery are evident. Let us now turn to postoperative residua and sequelae and the challenges they pose.

Residua

A residuum, according to the *Oxford English Dictionary*, is “that which remains, a residue, what is left over.” Therefore, residua are extrinsic to, that is, apart from the operative design and are intentionally left behind at the time of reparative cardiac surgery. With few exceptions, these residua are obligatory and are not the result of surgery having fallen short of its objective, at least in a technical sense. Residua after reparative surgery for congenital heart disease are electrophysiological, valvular, ventricular, vascular, and noncardiovascular (Table 1).

Electrophysiological Residua

There are 2 major categories of electrophysiological residua: (1) disturbances in rhythm and conduction that are inherent components of certain unoperated malformations and that necessarily persist after surgery and (2) electrophysiological abnormalities that develop as a consequence of the hemodynamic or hypoxic stress imposed on the heart by the unoperated malformation that may or may not persist after operation.

Left axis deviation¹⁸ is an electrocardiographic feature of atrioventricular septal defects, double-outlet right ventricle, tricuspid atresia, univentricular hearts of the left ventricular type, congenitally corrected transposition of the great arteries, and Ebstein's anomaly of the tricuspid valve. When cardiac surgery leaves the basic electrophysiological mechanism unaltered, left axis deviation persists as a postoperative residuum that is usually benign. Conversely, cardiac surgery or interventional catheterization can eliminate the basic electrophysiological mechanism, as in Ebstein's anomaly in which right-sided accessory pathways that express themselves as left axis deviation are interrupted.

Preoperative abnormalities of atrioventricular conduction in the form of PR interval prolongation, second-degree atrioventricular block, and complete heart block persist after reparative cardiac surgery for congenitally corrected transposition of the great arteries.¹⁹ Less common and less important is the mild to moderate PR interval prolongation that persists after closure of an ostium secundum atrial septal defect.

TABLE 1. Residua After Reparative Surgery for Congenital Heart Disease

Electrophysiological
Valvular
Ventricular
Chamber morphology
Chamber mass
Chamber function
Vascular
Anatomic anomalies or defects
Elevated systemic/pulmonary arterial pressure/resistance
Noncardiovascular
Development abnormalities
Mental retardation
Physical retardation (dwarfism)
Somatic abnormalities (facial, musculoskeletal)
Central nervous system abnormalities
Focal neurological deficits
Seizures
Senses
Visual abnormalities
Auditory abnormalities
Dental abnormalities
Medical disorders

Potentially graver is the residual postoperative first-degree atrioventricular block that sometimes accompanies familial ostium secundum atrial septal defect.²⁰

Abnormalities of impulse formation that remain as postoperative residua often reside in the sinus node. The normal sinus node is located at the junction of superior vena cava and morphological right atrium. A sinus venosus atrial septal defect of the superior vena cava occupies the site of the normal sinus node which may be absent, resulting in an ectopic atrial focus recognized in the scalar ECG as left-axis deviation of the P wave.¹⁹ Left isomerism is characterized by bilateral morphological left atria. Accordingly, there is no junction between the superior vena cava and a morphological right atrium and no anatomic substrate for a sinus node.

Preoperative atrial tachyarrhythmias represented by supraventricular tachycardia, atrial fibrillation, and atrial flutter persist as postoperative residua in adults with ostium secundum atrial septal defect, Ebstein's anomaly of both right and left atrioventricular valves, and atrial enlargement due to large left-to-right shunts or atrioventricular valve regurgitation.

In Ebstein's anomaly of the tricuspid valve, the complex electrophysiological properties of the atrialized right ventricle remain as postoperative residua depending on the technique of repair. Foci of slow conduction reside in the atrialized right ventricle; when that substrate is triggered (excited), the result is polymorphic ventricular tachycardia/fibrillation because the micromorphology cannot anchor reentrant waves, which break up immediately. If intracardiac repair eliminates the atrialized right ventricle, no portions remain as residual electrophysiological foci of slow conduction.

Valvular Residua

Structural and functional abnormalities of cardiac valves that persist after surgery or interventional catheterization are represented by (1) unrepaired malformed valves that function normally, (2) repaired malformed cardiac valves that function variably, and (3) intrinsically normal cardiac valves rendered incompetent by the physiological stress of the congenital malformation that prompted surgical intervention. A bicuspid aortic valve that is functionally normal or rendered so by balloon dilatation or surgical repair remains bicuspid and is in itself an important postoperative residuum in addition to the accompanying medial abnormalities of the ascending aorta.²¹

Ventricular Residua

Ventricular residua can be permanent, such as inherent chamber morphology, or they can change with the passage of time, such as chamber mass and function (Table 1). Inherent ventricular morphological residua include the univentricular heart and an anatomic right ventricle in the systemic location after an atrial switch operation or after an operation for congenitally corrected transposition of the great arteries.

An increase in ventricular mass in excess of the normal growth process is prompted by myocardial immaturity at the time the inciting stimulus is imposed, the type of inciting stimulus (hemodynamic or hypoxic), the duration of the stimulus, and the myocardial cell type that is involved in the increase in mass.^{22,23} When hemodynamic overload or hypoxia is imposed on an immature heart, an increase in ventricular mass is due chiefly to myocyte hyperplasia (replication).²³ Within a few months after birth, maturing myocytes become terminally differentiated, ie, lose their capacity to replicate.^{22,23} Pressure or volume overload then results in an increase in ventricular mass due to hypertrophy (enlargement) of existing myocytes, and hypoxia then exerts a deleterious ischemic effect. An important objective of reparative cardiac surgery is removal of the stimulus responsible for the preoperative increase in ventricular mass, whether the increase was due to myocyte hyperplasia or myocyte hypertrophy.^{22–25} Regression of ventricular mass implies either a decrease in the size of the enlarged (hypertrophied) myocytes or a decrease in the size of the excessive numbers of normal-sized myocytes (hyperplasia). These distinctive patterns of regression have important functional consequences.²³

Left ventricular function in tricuspid atresia is usually better than that in a single ventricle of the left ventricular type, which in turn is better than that in a single ventricle of the right ventricular type.²⁶ In hearts equipped with 2 ventricles, the ejection fraction of a morphological right ventricle is inherently lower than the ejection fraction of a morphological left ventricle, whether the right ventricle is subpulmonary or subaortic. A case in point is the inherently low ejection fraction of a subaortic morphological right ventricle after an atrial switch operation or after reparative surgery for congenitally corrected transposition of the great arteries.

Preoperative left ventricular systolic function that exceeds normal may persist as a presumably innocent postoperative ventricular residuum.²⁷ Witness the supranormal left ventricular systolic function that results when congenital aortic valve stenosis imposes increased afterload before terminal differentiation of cardiomyocytes.²⁸ The increase in left ventricular

mass is largely due to myocyte hyperplasia, which is accompanied by proportionate replication of the microvascular bed that preserves normal capillary density.²⁸ These features set the stage for low left ventricular systolic wall stress and supranormal ejection performance. After relief of the aortic stenosis, left ventricular mass decreases, but supranormal ejection fraction usually persists as a postoperative functional residuum.²⁷

Vascular Residua

There are 2 principal categories of vascular residua: (1) abnormalities of cerebral arteries, coronary arteries, or great arterial walls and (2) elevated resistance or pressure in the systemic or pulmonary circulation (Table 1). An aneurysm of the circle of Willis is an important vascular residuum after repair of coarctation of the aorta. Predisposition to rupture persists and may announce itself in normotensive patients long after successful coarctation repair.¹⁹ Coronary arterial residua may be functionally benign as an anomalous origin of a coronary artery in Fallot's tetralogy or the dilated coronary arteries in adults with cyanotic congenital heart disease.²⁹ Not so benign are the intimal proliferation, medial thickening, and premature coronary atherosclerosis that remain as residua after the repair of aortic coarctation or supravalvular aortic stenosis.¹⁹ As the age of the patient at the time of coarctation repair increases, so does the likelihood of postoperative hypertension. This is usually represented by a disproportionate increase in systolic pressure, especially during exercise, implying a residual decrease in the compliance of major proximal systemic arterial walls.^{19,21}

Systemic hypertension occasionally persists as a residuum after the repair of supravalvular aortic stenosis in Williams syndrome. Great arterial medial abnormalities of smooth muscle, elastic fibers, collagen, and ground substance persist as postoperative residua in a variety of congenital cardiac diseases.²¹ Aortic medial abnormalities may predispose to dilatation, aneurysm, and rupture.²¹ Pulmonary trunk medial abnormalities may predispose to dilatation and aneurysm formation in mobile pulmonary valve stenosis or Fallot's tetralogy with absent pulmonary valve.²¹ Early surgical correction reduces the probability of postoperative pulmonary vascular disease, which otherwise may result in late aneurysmal dilatation and the rupture of a hypertensive pulmonary trunk.²¹

Noncardiovascular Residua

Noncardiovascular residua include developmental abnormalities; abnormalities of the central nervous system, the senses, and dentition; and medical disorders (Table 1).³⁰ Developmental abnormalities are represented by mental retardation (Down syndrome), physical retardation (dwarfism in Turner syndrome and Ellis-van Creveld syndrome), and somatic abnormalities, such as facial dysmorphism, cleft lip or palate, or skeletal abnormalities, as in the Holt-Oram syndrome.³¹ Central nervous system abnormalities that persist as postoperative residua include focal neurological deficits (paradoxical cerebral embolus), seizure disorders (brain abscess), and spinal cord injury (coarctation repair) (Table 1). Disturbances of the senses that remain as postoperative residua are repre-

TABLE 2. Sequelae After Reparative Surgery for Congenital Heart Disease

Electrophysiological
Atriotomy
Intra-atrial repair
Intraventricular repair
Ventriculotomy
Incision site
Intracardiac repair
Valvular
Left ventricular or right ventricular outflow repair
Left ventricular or right ventricular inflow repair
Prosthetic materials
Patches
Valves
Conduits
Myocardial and endocardial
Vascular
Neurological

sented by visual and auditory abnormalities, such as the cataracts and deafness of rubella syndrome. Conversely, postoperative abnormalities of the senses are sometimes salutary, such as the hyperacusis and exceptional auditory and musical skills that occur in Williams syndrome.³² Residual dental abnormalities include premature eruption of malformed maxillary incisors in Ellis-van Creveld syndrome and the malformed teeth of Williams syndrome.³²

Postoperative medical disorders may or may not be related to the congenital cardiac malformation for which surgery was performed, and they may or may not be cardiovascular. Examples include diabetes, acquired coronary artery disease, and essential hypertension (which are age-related) and autoimmune hypothyroidism in Down syndrome, which may express itself as a late postoperative residuum.³³ Cyanotic adults have an increased incidence of calcium bilirubinate gallstones that may announce themselves as acute cholecystitis years after surgery has eliminated the cyanosis.³⁴

Sequelae

A sequel (plural: sequelae) is defined as "what follows or arises out of an earlier event." Sequelae are intrinsic to the operative design and are represented by alterations or disorders that are intentionally incurred, occasionally or invariably, at the time of surgery and are looked on as necessary, if not entirely acceptable, consequences of the operation. Sequelae of reparative surgery for congenital heart disease may be electrophysiological or may involve the cardiac valves, prosthetic materials, myocardium/endocardium, the vascular bed, and the nervous system (Table 2).

Electrophysiological Sequelae

Electrophysiological sequelae after atriotomy are due less to the atrial incision per se (scarring) than to the intra-atrial or intraventricular repair for which the atriotomy provides access. The propensity to develop abnormal atrial rhythms is

also a consequence of damage to the sinus node and sinus node artery, abnormal atrial wall stress, and changes in atrial refractoriness. The incidence of atrial tachyarrhythmias after surgical closure of an ostium secundum atrial septal defect is influenced by the presence of preoperative arrhythmias, age at repair, and the duration of postoperative follow-up. Sinus bradycardia is an occasional sequel of intra-atrial repair of the ostium secundum atrial septal defect. A sinus venosus atrial septal defect of the superior vena caval type abuts or occupies the site of the normal sinoatrial node.¹⁹ If sinus rhythm is present before the operation, intraoperative injury to the node may result in postoperative ectopic atrial bradycardia. Surgical closure of an ostium primum atrial septal defect is accompanied by an increased incidence of impaired atrioventricular conduction because of the proximity of the atrioventricular node and His bundle to the site of repair. The extensive reconstruction that characterizes atrial switch operations sets the stage for postoperative sinus bradycardia, atrial flutter, atrial fibrillation, junctional tachycardia, and high-degree heart block.

The Fontan procedure has undergone many modifications and is now applied to a variety of complex cyanotic malformations. The Fontan operation typically results in a circulation in series without a functional subpulmonary ventricle, and it is the procedure of choice for single ventricle (univentricular atrioventricular connection) and tricuspid atresia. Postoperative atrial tachyarrhythmias adversely affect left ventricular function, provoking a rise in left atrial pressure that impedes the Fontan circulation and risks hemodynamic deterioration. Atrial tachyarrhythmias and sinus node dysfunction may be less frequent after total cavopulmonary connections.

In congenitally corrected transposition of the great arteries, repair of a ventricular septal defect risks complete heart block because the nonpenetrating atrioventricular conduction bundle runs along the superior margin of the septal defect. Patients who escape intraoperative heart block are apparently not at risk for late postoperative heart block.

After ventriculotomy, tachyarrhythmic sudden death remains an incompletely resolved problem, but important electrophysiological and hemodynamic risk factors have been identified.³⁵ QRS duration ≥ 180 ms and the rate of increase in QRS duration may be predictors of ventricular tachyarrhythmic sudden death.³⁵ There must, however, be a susceptible substrate characterized by slow conduction capable of supporting reentry, which is the basic electrophysiological prerequisite for monomorphic ventricular tachycardia, the usual tachyarrhythmic precursor of sudden death. The signal-averaged ECG, as currently modified, can identify postventriculotomy slow conduction that is potentially capable of sustaining reentrant monomorphic ventricular tachycardia.³⁶ If slow conduction is identified, especially in concert with a QRS duration ≥ 180 ms, an intracardiac electrophysiological study serves to determine whether monomorphic ventricular tachycardia can be induced by stimulating the slow conduction zone. The site can be mapped and, if located (usually along the ventriculotomy scar), eliminated by radiofrequency ablation.

Sequelae Involving Cardiac Valves

Surgical repair of complex obstruction to right ventricular outflow, such as Fallot's tetralogy, tends to be followed by pulmonary valve regurgitation, the importance of which depends on the degree of regurgitant flow and the functional and electrical state of the incised right ventricle. Accordingly, if an electrophysiological substrate of slow conduction and QRS prolongation coincide with the hemodynamic substrate of pulmonary regurgitation, reoperation serves to restore pulmonary valve function and eliminate the area of slow conduction by revising the ventriculotomy scar.

Sequelae of inflow valve repairs are more prevalent than are sequelae of outflow repairs because the mitral and tricuspid mechanisms are more complex than are the aortic and pulmonary valve mechanisms. Even if complete relief of regurgitation is achieved after repair of a malformed mitral valve associated with an atrioventricular septal defect, left ventricular inflow remains guarded by a morphologically abnormal mitral apparatus whose ultimate competence is influenced by changes in left ventricular geometry and function that accrue with time. The abnormal morphology of a reconstructed Ebstein tricuspid valve is an acceptable sequel if competence is established.

Prosthetic Materials

Patches, valves, and conduits represent a special category of postoperative sequelae. The devices or materials selected must achieve an immediately successful technical result and have acceptable long-term effects on morbidity and mortality.

An autograft or autologous graft refers to tissue derived from the individual receiving the graft; it is usually made up of pericardium, arteries, or valves. Endogenous material tends to have strength, compliance, and handling properties similar to those of the structure that it replaces or repairs and, in addition, is nonthrombogenic and nonantigenic because the endogenous material is derived from the host. Living endothelial cells and fibroblasts permit endogenous materials to retain their substance and configuration, to resist infection, and to grow. Important sequelae of using endogenous materials include limited availability and the effects on the host of materials removed from their normal location. The amount of remaining pericardium limits availability if reoperation is required. An endogenous valve removed from its normal position, as in the Ross procedure, requires replacement with an exogenous biological or synthetic prosthesis, which adds to the complexity of the operation and exerts an impact on long-term outcome.

Homografts or allografts are exogenous bioprosthetic materials derived from an individual of the same species but of disparate phenotype. Xenografts or heterografts refer to tissue derived from an individual of a different species. Exogenous bioprosthetic materials are secured from human cadavers (homografts) or animal sources (xenografts). Xenograft valve durability is significantly coupled with patient age at insertion. In addition, fixation alters the natural characteristics of the tissue, rendering it prone to fibrocalcific degeneration, fusion, and disruption. Dacron fabric and polytetrafluoroethylene (Gore-Tex fabric) are synthetic materials available as a

TABLE 3. Additional Relevant Topics

Infective endocarditis
Comorbidities and coexisting cardiac and noncardiac diseases
Cardiac catheterization as a therapeutic intervention
Transplantation
Diagnostic techniques
Noncardiac surgery
Reproduction
Research

flat sheet or tube graft. Long-term results with Gore-Tex seem to be better than those with Dacron.

Mechanical prosthetic valves include caged-ball and tilting discs; the latter are either monoleaflet or bileaflet. Despite excellent engineering modifications, the thromboembolic complications of mechanical valves have not been resolved; thus, they require the use of anticoagulants.

Neurological Sequelae

The incidence of neurological sequelae has declined substantially as surgical techniques have improved. Only a small percentage of patients sustain permanent neurological sequelae (seizures, motor disorders) or disorders of higher cortical function (mental retardation, learning disabilities).^{37,38} However, total circulatory arrest in infancy may be followed by impaired motor coordination,³⁹ and the impact of cardiopulmonary bypass on the developmental outcome of children who undergo open heart surgery for closure of secundum atrial septal defect compares unfavorably with the developmental outcome after device closure.⁴⁰ Cognitive ability after a Fontan operation is lower than that of the general population.⁴¹

Additional Relevant Topics

Infective Endocarditis

The clinical and epidemiological manifestations of infective endocarditis (Table 3) have evolved considerably, but what has not changed are the 2 major predisposing factors: (1) a susceptible cardiac or vascular substrate and (2) a source of bacteremia.⁴² Surgical interventions have had a significant impact on the risk of endocarditis. Operations using prosthetic valves or conduits substantially increase the risk, whereas other operations (ie, division of a patent ductus arteriosus) eliminate the risk. Advances in imaging techniques, especially transesophageal echocardiography, have provided rapid and accurate diagnosis, and advances in bacteriological assessment and antibiotic availability have been major therapeutic steps forward.

Comorbidities and Coexisting Cardiac and Noncardiac Diseases

The postoperative adult not only confronts the surgically modified congenital malformation, but is susceptible decade by decade to acquired cardiovascular disease, such as ischemic heart disease and systemic hypertension, and to noncardiac diseases such as diabetes mellitus. The interplay between

the postoperative congenital malformation, its residua and sequelae, and acquired cardiac and noncardiac diseases increases the complexity of the challenges posed by adults with repaired congenital heart disease.

Cardiac Catheterization as a Therapeutic Intervention

Cardiac catheterization techniques can be corrective, reparative, or palliative; can serve as alternatives to high-risk surgery; or can be used to compliment and enhance surgical results. Balloon valvuloplasty for typical, mobile pulmonary valve stenosis has replaced surgical repair and is now the procedure of choice for all ages. It usually leaves behind virtually no residua or sequelae. However, balloon valvuloplasty for typical, mobile, bicuspid aortic stenosis can, at best, achieve mechanical results analogous to those of a functionally normal bicuspid aortic valve, including the risk of infective endocarditis and the risk inherent in the accompanying medial abnormality of the ascending aorta.²¹ Balloon dilatation of unoperated aortic coarctation damages the inherently abnormal paracoarctation aorta that contains medial abnormalities of smooth muscle, collagen, and elastin.²¹ An ideal repair eliminates both the obstruction and the abnormal paracoarctation aortic media. Surgical division of a patent ductus arteriosus relieves left ventricular volume overload and eliminates the ductus as a substrate for infective endocarditis; transcatheter occlusion competes with these results on both counts. Device closure of an ostium secundum atrial septal defect is beginning to compete with surgery, eliminating both the shunt and the substrate for paradoxical embolization.

Transplantation

Lung and heart/lung transplantation tend to be the major transplantation options for congenital heart disease. In patients with Eisenmenger syndrome, the least complex option is single lung transplantation with intracardiac repair of an isolated interatrial or interventricular septal defect. The operative risk of transplantation is greater than in acyanotic primary pulmonary hypertension because of the incidence of major thromboses of dilated hypertensive proximal pulmonary arteries in Eisenmenger syndrome.²² Heart/lung transplantation poses the dual problems of donor organ availability and more complex immunological management after transplantation.

Diagnostic Techniques

Cardiac catheterization and angiocardiology were the first major diagnostic steps forward, followed by transthoracic and transesophageal echocardiography. By providing exquisite anatomic detail and hemodynamic information, echocardiography often obviates the need for cardiac catheterization. Cine MRI has added considerably to the diagnostic armamentarium and, as a complementary imaging modality, it may largely supplant echocardiography.

Noncardiac Surgery

When adults with congenital heart disease undergo noncardiac surgery, perioperative safety can be appreciably increased if the risks inherent in that patient population are anticipated.³⁴ A cardiac anesthesiologist with experience in congenital heart disease is pivotal. It is the anesthesiologist

who is largely responsible for the physiological integrity of the patient during noncardiac surgery and who plays a major postoperative role, especially in pain management. Central to risk stratification and perioperative planning are the type of congenital heart disease, coexisting acquired cardiovascular or medical disorders intrinsic to or apart from the congenital cardiac malformation, and whether the noncardiac operation is elective or urgent, major or minor. High risk patients are best managed in a tertiary care facility. The cardiac anesthesiologist and the attending cardiologist are more important than the noncardiac surgeon.

Reproduction

The postoperative woman with congenital heart disease now constitutes a major category of those who are pregnant and have heart disease.⁴² Successful cardiac surgery improves fertility in women whose heart disease had reduced sexual and ovarian function. Women who were previously ill-equipped to bear children or who may not have reached reproductive age are now presenting for obstetric and cardiological care after reparative surgery. Central to this topic is the intricate interplay between maternal circulatory and respiratory physiology and maternal congenital heart disease and the effects of this interplay on the fetus, which is exposed to risks that threaten its intrauterine viability and risks that are subsequently expressed as developmental defects or genetically transmitted anomalies of the heart and circulation.

Gravidas with functionally important unoperated or postoperative congenital cardiac disease should be managed in a tertiary care facility by a high-risk pregnancy obstetrician in collaboration with an experienced cardiologist.⁴³ Risks are appreciably reduced by meticulous attention to gestation, labor, delivery, and the puerperium. Determination of fetal lung maturity with amniocentesis, controlled induced vaginal delivery, lumbar epidural anesthesia, and meticulous postpartum care have substantially lowered maternal risk.⁴³ Management of the fetus includes intrauterine echocardiography and selective amniocentesis. Fetal viability is threatened by the functional class of the mother, maternal cyanosis, and oral anticoagulants. Remote risks take the form of genetic parental transmission, teratogenic effects of certain cardiac drugs, and the harmful effects of certain environmental toxins and exposures.

Potential parents should be provided with genetic counseling regarding recurrence risk that varies according to the relative (parent or sibling) and according to the type of congenital heart lesion in the relative. A common concern voiced by parents of a child with congenital heart disease is the probability of recurrence during subsequent pregnancies. Recurrence with one previously affected sibling is 2.3% and with two previously affected siblings, it is 7.3%.⁴³ If the mother has congenital heart disease, the recurrence risk in her offspring is 6.7%, and if the father is affected, the risk is 2.1%. An important aspect of recurrence is concordance, ie, the tendency for repetition to be in the same category of congenital malformation.⁴³ However, the concordant malformation may differ appreciably in severity and complexity, as in conotruncal malformations. Fetal echocardiography has been a major step forward in providing potential parents with a basis for judgment.

Research

Research is an obligatory commitment prompted by a desire to resolve questions posed by an adult congenital heart disease population, and it is a necessary experience for fellows with a career interest in adult congenital heart disease. Investigations generally require collaboration with colleagues in a number of other disciplines, thus stimulating valuable interdisciplinary interchange.

It is not idle to consider certain key issues that are likely to assume importance during the coming decade. Genetics and molecular biology loom large. Examples include (1) identification of genes that dictate left-right/anterior-posterior patterning in the heart, (2) identification of genes that make the left and right ventricles morphologically distinct, (3) identification of modifier genes and environmental cues that determine the variation in disease expression despite a similar basic genetic abnormality, (4) determination of subtle variations in gene mutations that lead to wide variations in phenotype, and (5) determination of genetic abnormalities that may in part be responsible for postoperative predisposition to arrhythmias. Advances in cardiothoracic surgery include (1) transplantation biology, especially lung; (2) development of more durable valve and vascular replacements by bioengineering and tissue engineering; and (3) robotic and nanotechnology designed to minimize, if not preclude, conventional surgical intervention.

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