Preventive and Primary Care of The Adult With Congenital Heart Disease

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Introduction

Due to remarkable advances in medical and surgical care over the past six decades, a significant and growing population of specialized patients with repaired and palliated congenital heart disease has grown into adulthood. In the United States alone, there are an estimated 800,000 adults with congenital heart disease. Currently, over 85% of the approximately 25,000 infants born each year with congenital cardiac anomalies will survive into later life, many with fairly normal life expectancies. In 1959, the first comprehensive tertiary care facility for these patients was established as the Toronto Congenital Cardiac Centre for Adults at the Toronto General Hospital (now the Toronto Hospital). Other multidisciplinary programs with ambulatory care clinics and training programs for specialists in the care of these patients were begun at the National Heart Hospital in London (now the Royal Brompton and National Heart Hospitals), UCLA, the University of Iowa, and the Mayo Clinic. Several other programs have been initiated at academic medical and regional medical centers. Ideally, all adults with congenital heart disease would receive medical care at these specialized centers. However, current estimates suggest that less than 20% of these complex patients are cared for at centers specializing in adult congenital heart disease.

Many adults with congenital heart disease receive both primary and specialty care in their communities rather than at tertiary centers for several reasons. Because adult congenital heart disease (ACHD) programs are regionally distributed, in part to control cost while optimizing care with highly trained staff, specialist care is not as accessible as might be desirable. As these patients transition into adulthood, they wish to see adult doctors in adult settings and to adopt more "normal" patterns of referral for medical care; that is, they want their primary care physician and general adult cardiologist to manage their general medical and cardiovascular problems with referral to ACHD experts reserved for specific cardiac problems. This desire has been noted as occurring as early as adolescence in one survey of preferences for primary and specialist care of parents of teenagers with congenital heart disease. Many young adults with congenital heart disease, like other young adults who may or may not have health insurance, adopt high-risk behaviors such as smoking, excessive alcohol use and poor nutrition and seek medical care only when acutely ill. Finally, health insurers of these patients may limit referrals to ACHD programs to address cardiovascular problems only.

Table 1. Health Maintenance Needs

Vaccinations
Cholesterol screening
Hypertension screening
Cancer screening
Assessment of tobacco, alcohol and drug use
Nutritional counseling

Unfortunately, incomplete knowledge of the health needs of these patients can result in clinical deterioration, unnecessary hospitalization and even premature death. In a study from the Royal Brompton and National Heart Hospital in London, 21% of the deaths of 341 adults with congenital heart disease in their database were avoidable. Many of the deaths were due to lack of knowledge by primary care physicians and general cardiologists of common illnesses, medications and arrhythmia management in these specialized patients or were from late or non-referral to ACHD specialists. Primary care physicians and general adult cardiologists must have knowledge of the preventive health needs and special issues in the general medical management of these complex patients. Importantly, physicians must recognize when to promptly refer these patients to an ACHD consultant.

Preventive Medicine and Health Maintenance

Assessment of the health maintenance status of these patients should be routine (Table 1). In addition to problems resulting from their cardiac lesion, these adults may acquire usual and unusual adult diseases, including lipid disorders, hypertension, coronary artery disease, and cancers. At appropriate ages as guided by patient risk and practice guidelines, these adults should have routine blood pressure checks, cholesterol screening, and cancer screening including mammography, pap smears, prostate specific antigen measurement and fecal occult blood tests. Cyanotic patients and other patients with complex congenital heart disease should be referred to an ACHD specialist prior to endoscopy. The patient should be queried about the use of alcohol, illegal drugs, prescription medications, over-the-counter medications, herbal or homeopathic remedies, and the use of tobacco. When appropriate, counseling for high-risk behaviors should be offered. Nutritional guidance should be available, especially for patients with special nutritional needs such as sodium-restricted or low cholesterol diets. Maintenance of ideal body weight should be encouraged.

In the pediatric population with congenital heart disease, routine vaccinations are often off-schedule or missed entirely due to illness or hospital admission. Adults are often lax in keeping vaccinations up-to-date. Physicians

1062-1458/01/$20.00
PII S1062-1458(01)00356-7

should review vaccination records with the ACHD patient with attention given to the need for boosters of the tetanus-diphtheria toxoid and measles-mumps-rubella vaccine. All adults and adolescents with congenital heart disease should receive the currently available 23-valent pneumococcal polysaccharide vaccine if they have not been previously vaccinated. Immunization with the influenza vaccine should be administered yearly.

Infective Endocarditis Prophylaxis

The need for antibiotic prophylaxis for infective endocarditis in these patients depends on the underlying cardiac anatomy and physiology and the type of procedure to be performed. At highest risk are patients with a prior history of endocarditis, prosthetic valves, systemic to pulmonary shunts, conduits, and those with complex cyanotic congenital heart disease (single ventricle-type anatomy, unrepaired Tetralogy of Fallot, etc.). Patients with an isolated secundum atrial septal defect, either unrepaired or repaired, repaired ventricular septal defect or patent ductus arteriosus (all without residua more than 6 months post-operatively) are at very low risk and do not require antibiotic prophylaxis. Most other patients with congenital heart disease are considered to be at moderate risk and endocarditis prophylaxis is recommended. In general, chemotherapeutic prophylaxis is recommended for dental cleaning and extractions, dental procedures when bleeding is anticipated, tonsillectomy and/or adenoidectomy, and certain other procedures involving respiratory mucosa, the biliary tract, intestinal mucosa, or genitourinary tract. The American Heart Association Guidelines for the Prevention of Bacterial Endocarditis should be consulted for full recommendations. Of note, endocarditis prophylaxis is not recommended for Cesarean section in any patient and is considered optional for vaginal delivery in patients at high risk.

Physicians should emphasize the need for routine dental care and good dental hygiene, with regular check-ups and cleaning every 6 months. Cyanotic patients are prone to very friable gums and periodontal disease and require special attention. Local prophylactic measures such as rinsing with antiseptic oral solutions prior to dental cleanings may decrease the risk of bacterial endocarditis in these patients.

Since the skin is a common portal of entry for bacterial endocarditis, patients require meticulous care of nails and skin and aggressive management of acne. Manipulation of pustules should be discouraged and early referral to a dermatologist considered.

Issues in Contraception

Only in very few instances should pregnancy be proscribed in women with congenital heart disease. Specifically, patients with Eisenmenger's syndrome, complicated coarctation of the aorta, and dilated aorta from Marfan's syndrome carry a maternal mortality risk of 25–50% and should avoid pregnancy. In these women, sterilization by tubal ligation or sterilization of their sexual partner is preferred over other contraceptive methods.

Several issues are important when discussing contraception with adolescents and women of childbearing age with other forms of congenital heart disease. These include failure rates of the methods and risks of the method to the health of the patient. Spermicidal topical agents used alone, the vaginal sponge, and barrier methods including the cervical cap, condoms and the diaphragm have failure rates of 10–25% per use. In women with right to left shunts and, particularly, cyanotic congenital heart disease, the risk of thromboembolism must be considered. Although the risk of thromboembolism is low with currently available low-dose estrogen-containing oral contraceptives, they are probably contraindicated in these patients. Progesterone-only contraceptive pills, intramuscular medroxyprogesterone injections, and implanted levonorgestrel do not carry the risk of thromboembolism and have a low failure rate. However, the incidence of breakthrough uterine bleeding is significant and bleeding may be severe in cyanotic patients with erythrocytosis and abnormal hemostasis. Intrauterine contraceptive devices (IUD) have only rarely been associated with infective endocarditis, but should probably be avoided in women at moderate or high risk, especially patients with systemic to pulmonary shunts, prosthetic valves, and complex congenital heart disease. Patients who choose to use the IUD for contraception should receive endocarditis prophylaxis prior to IUD placement.

Travel to High Altitude and Fitness to Fly

Patients with Eisenmenger's syndrome who travel to high-altitude locales are at high risk for acute right heart failure and potentially sudden death. Lower inspired oxygen tension with resulting increased hypoxia may precipitate pulmonary vasoconstriction with worsened right to left shunting, more severe systemic hypoxemia, and right heart strain. Cyanotic patients with Eisenmenger's may become very symptomatic at minimal levels of exertion when at high altitudes. Therefore, for those patients already acclimated to lower altitudes, travel to high altitude locations should be discouraged.

However, physicians can reassure cyanotic patients requesting advice about their fitness to fly that travel in commercial jet aircraft with pressurized cabins is generally safe. In one study of 10 cyanotic adults who had transcutaneous oxygen saturation and capillary blood gas monitored during a 2.5-hour commercial air flight to a maximum cabin altitude of 6000 feet, the mean oxygen saturation decreased from 86±5.2% at sea level to 83±5.5% at maximum cabin altitude and returned to 85±5.3% immediately after reaching the destination gate, also at sea level. Mean capillary pO_2 at maximum cabin altitude did not change from that at sea level (44±11.8 vs. 45±6.7 mm Hg). No clinical problems were noted during the flights. Some authors recommend supplemental oxygen and con-

tinuous oxygen saturation monitoring for cyanotic patients who travel on commercial pressurized aircraft.

Special Issues in the Medical Care of ACHD Patients

Evaluation and Management of the Febrile, Ill Patient

Patients with fever present a special challenge to their physicians, as do patients who complain of constitutional symptoms but are afebrile or who have only low-grade fevers. Delay in and failure of diagnosis of endocarditis is responsible for significant morbidity including stroke and the requirement for reoperation, and for high mortality in these patients. Infection of right-sided lesions (e.g., ventricular septal defects) and of shunts and conduits is particularly difficult to diagnose and should be considered in each and every adult with this type of congenital heart disease who presents with fever and/or constitutional symptoms. Endocarditis should be immediately suspected in the febrile patient with dental or periodontal disease or skin infection. Blood cultures should be obtained, especially if a source of fever is not readily apparent. Antibiotics should not be prescribed if a definite source of fever is not identified.

Fever and acute gastrointestinal illnesses can precipitate symptoms and complications of hyperviscosity in cyanotic patients with erythrocytosis because of dehydration. Volume status should be assessed and oral hydration encouraged. Patients unable to drink adequately due to vomiting or excessive diarrhea should receive intravenous hydration. They should be questioned closely and evaluated carefully for signs and symptoms of marked hyperviscosity including persistent headache, diplopia or blurred vision, lassitude, impaired alertness or mentation, tinnitus, paresthesias of the digits or lips, and myalgias or muscle weakness. Intravenous hydration should be considered for these patients to avert stroke. Of note, patients with right to left shunt who require intravenous therapy should have all fluids administered through an in-line filter to prevent air embolism.

Evaluation of Palpitations, Dizziness and Syncope

Arrhythmias are exceedingly common after intracardiac surgery in these patients and often herald progressive structural failure, such as obstruction of intraatrial baffles and conduits, hemodynamic deterioration, ventricular failure, and sudden cardiac death (Table 2). Intraatrial repairs are associated with supraventricular arrhythmias, including sinus node dysfunction, atrial fibrillation and atrial flutter, in over 50% of patients with atrial switch procedures for transposition of the great arteries, i.e., the Mustard and Senning operations. These arrhythmias occur in approximately 25% of patients after the Fontan procedure and are associated with a significant risk (2–8%) of sudden cardiac death. Operations done with ventricular incisions (e.g., repair of many patients with Tetralogy of Fallot) predispose to serious ventricular arrhythmias and complete heart block in approximately 30% of patients.

Table 2. Diagnoses Associated with Cardiac Arrhythmias

Supraventricular arrhythmias (sinus node dysfunction, atrial flutter, atrial fibrillation)
Tricuspid atresia/Fontan
Transposition of the great arteries/Mustard or Senning
Double inlet or outlet left or right ventricle
Tetralogy of Fallot
Atrial septal defect
Ventricular arrhythmias
Tetralogy of Fallot
Ventricular septal defect
Atrioventricular canal (endocardial cushion defects)
Ebstein's anomaly
Coronary artery anomalies

Therefore, new symptoms of brief palpitations or poorly characterized dizziness should be evaluated aggressively with appropriate monitoring such as a Holter monitor or continuous loop recorder and with careful evaluation of the patient's current hemodynamic status by surface or transesophageal echocardiography or catheterization when indicated. Diagnosis of the arrhythmia may require electrophysiologic testing.

Treatment of arrhythmias in these patients is complex, often differs from treatment of arrhythmias in patients with other types of cardiac disease, and should involve consultation with an expert in ACHD. Acceptable management of adults with acquired cardiac disease such as hypertension or coronary disease and atrial flutter often includes medications for rate control and antiarrhythmic agents. This practice could prove catastrophic in patients after Fontan procedure. In these patients, sinus rhythm must be restored expeditiously, the integrity of baffles and conduits assessed, and recurrence of the arrhythmia prevented with appropriate medications that do not depress ventricular function. Additionally, chronic anticoagulation with warfarin is necessary even after cardioversion due to the high incidence of Fontan thrombus, which may embolize to the pulmonary or central circulation. Electrophysiologic testing after repair of Tetralogy of Fallot may reveal ventricular tachycardia originating in the right ventricular outflow tract, which may be ablated. In patients requiring temporary or permanent pacemakers, the approach to pacemaker implantation is not straightforward due to abnormal intracardiac anatomy and may require transthoracic temporary pacing followed by operative placement of epicardial electrodes.

Use of "Routine" Medications

Medications in common use either by prescription or over-the-counter can have disastrous consequences in these patients (Table 3). Nonsteroidal anti-inflammatory agents may cause deterioration of renal function and precipitate hemorrhage in cyanotic patients with erythrocytosis and underlying renal impairment, which is common and commonly unrecognized. In patients with systemic ventricular dysfunction, these agents may trigger decompensated congestive heart failure due to fluid retention. Hormonal treatments for infertility or endometriosis such as danazol and

Table 3. Potential Complications of Common Medications

Medication	Complication	Mechanism
Aspirin	hemorrhage	platelet inhibition
	renal insufficiency	inhibition of prostacyclin-regulated renal blood flow
Nonsteroidal anti-inflammatory agents	hemorrhage	platelet inhibition
	renal insufficiency	inhibition of prostacyclin-regulated renal blood flow
	congestive heart failure	sodium and water retention
Estrogen	thromboembolic events	hypercoagulability
Progesterone	uterine hemorrhage	breakthrough bleeding
Danazol	dehydration	vomiting
	congestive heart failure	sodium and water retention
Beta-blockers	congestive heart failure	negative inotropy
Phenothiazines	ventricular arrhythmias	QT prolongation
Angiotension converting enzyme inhibitors	worsening cyanosis, dyspnea	systemic > pulmonary vasodilation increases right to left shunt
Vasodilators	worsening cyanosis, dyspnea	systemic > pulmonary vasodilation increases right to left shunt

estrogens may also cause fluid retention, bring about electrolyte imbalance and provoke vomiting and dehydration. Psychotropic medications may cause QT prolongation and aggravate arrhythmias. Vasodilator medications, often prescribed for treatment of congestive heart failure, may cause worsening cyanosis and dyspnea in cyanotic patients with single ventricle anatomy and pulmonary hypertension.

Medical Issues Specific to Cyanotic and Eisenmenger's Patients

(See Table 4.) Erythrocytosis is an adaptive response to tissue hypoxemia in these patients and is not, by its mere existence, an indication for phlebotomy, regardless of hemoglobin or hematocrit level. Patients with stable hematocrit levels, even as high as 75%, who are iron replete, and who have no or only mild symptoms of hyperviscosity are considered "compensated" and do not have an increased risk of stroke. In these patients, cerebrovascular events were associated with history of phlebotomy and microcytosis from iron deficiency, but not with the degree of erythrocytosis, ejection fraction, or use of aspirin or warfarin. Phlebotomy should not be routinely performed for an elevated hematocrit. Patients with unstable, rising hematocrit levels that are not controlled by usual feedback inhibition are considered to be "decompensated" and often have severe symptoms of hyperviscosity that interfere with most or all of their activities. Often these patients have iron deficiency and microcytosis. Hyperviscosity results from impaired deformability of red blood cells as well as other factors including plasma volume. In patients with iron deficiency and hematocrit <65%, iron replacement will alleviate symptoms of hyperviscosity and is administered in small doses (65 mg of elemental iron once daily) with frequent monitoring of the hematocrit. Iron supplementation is discontinued at the first measurable rise in the hematocrit, which often occurs in the first or second week of treatment. Phlebotomy is only indicated for patients with marked to severe symptoms of hyperviscosity and hematocrit levels over 65% who are not dehydrated. In these patients, phlebotomy is limited only to the amount of blood that provides relief of symptoms to avoid iron deficiency. Whole blood is replaced with saline or dextran.

Table 4. Medical Issues in Cyanotic Patients

Erythrocytosis
Compensated (Stable Hct, iron replete, symptoms of hyperviscosity mild or absent)
Decompensated (Rising Hct, iron deficient, symptoms of hyperviscosity severe)
Cholelithiasis
Abnormal hemostasis
Thrombocytopenia
Hemorrhage
Renal dysfunction
Hyperuricemia and gout
Hypertrophic osteoarthropathy

Hct = hematocrit.

Cholelithiasis with calcium bilirubinate stones is not uncommon and may necessitate cholecystectomy. Also, these patients have abnormal hemostasis usually with a mild bleeding diathesis resulting in easy bruising and bleeding of the gums. They usually have thrombocytopenia with platelet counts in the 60,000 to 80,000 range. Antiplatelet agents including nonsteroidal anti-inflammatory agents and aspirin may potentiate the bleeding diathesis and should be reserved for clear-cut indications. Serious hemorrhage may occur after accidental trauma and simple noncardiac surgery including oral surgery such as wisdom tooth extraction. An ACHD specialist should, therefore, be consulted prior to any noncardiac surgery, including sterilization, laparoscopy, and dental extraction in these patients.

Renal dysfunction (some with evidence of glomerulopathy) occurs in more than one third of these adults. Patients will often have proteinuria and abnormal urinalyses with only mild elevations of serum creatinine. Nonsteroidal anti-inflammatory agents, radiocontrast agents, dehydration and other medications may cause renal failure. Additionally, decreased renal clearance of uric acid along with increased production results in hyperuricemia, which may

Table 5. 10 Reasons to Refer to an Expert in ACHD

1. Complex or cyanotic congenital heart disease
2. Any change in functional status or effort tolerance
3. A new or changing murmur
4. Pregnant women
5. Before noncardiac surgery or procedure
   - Cholecystectomy
   - Sterilization
   - Wisdom tooth extraction
   - Endoscopy
6. Palpitations or arrhythmias
7. CNS symptoms
8. Decompensated erythrocytosis
9. Suspected endocarditis (after blood cultures)
10. Whenever the provider feels uncomfortable!

precipitate symptomatic gout. Acute gout is usually best managed with intravenous colchicine, though it must be used with care because of the side effects of vomiting and dehydration. Oral or intra-articular steroids may also be used. Nonsteroidal anti-inflammatory agents should be avoided. Reduced doses of allopurinol or colchicine may be given as prophylaxis of recurrent gouty arthritis. Of note however, is that many patients have chronic joint pain, not from gout, but from hypertrophic osteoarthropathy. The joint pain rarely requires specific treatment.

Indications for Prompt Referral to the ACHD Specialist

Most physicians have limited familiarity and experience with caring for this new population of patients, the adults with congenital heart disease. In addition to complex cardiac anatomy, these patients possess complex physiology and may have unusual or unexpected devastating responses to usual medications, usually "simple" surgeries and what may be benign arrhythmias in other patients. In addition, one of five deaths of ACHD patients is avoidable. Hence, cardiologists and primary care physicians must know when to refer to an ACHD specialist without delay. In each of the instances outlined below, delay in referral may result in calamity (Table 5).

All adults with complex or cyanotic congenital heart disease should be referred. Somerville states that all adults with any of these words in their diagnosis should be managed under the direct supervision of an ACHD expert: atresia, single/double inlet or outlet, transposition of the great arteries, corrected transposition, valved conduit, cyanotic and Eisenmenger—and I will add shunt, Fontan, single ventricle and Mustard. Patients with a recent change in functional status, ability to work or effort level may be exhibiting signs of ventricular failure, complications or sequelae of their surgery or life-threatening arrhythmias and require prompt consultation. Any patient with a change in or diagnosis of a new murmur should be referred. Pregnant women should be referred to an ACHD specialist and obstetrician experienced in managing high-risk pregnancies as soon as pregnancy is confirmed. This will optimize the chances of the pregnancy resulting in both a healthy mother and healthy baby and permits early genetic counseling. Likewise, all patients undergoing even minor noncardiac surgery (wisdom tooth extraction, sterilization, etc.) or endoscopic procedure should undergo preoperative evaluation by a specialist. All patients with palpitations, syncope, documented arrhythmias and stroke should be cared for by or with the ACHD consultant. Patients with subtle central nervous system complaints may be manifesting hyperviscosity or brain abscess and should be promptly evaluated. Any adult suspected to have endocarditis should be immediately cultured and referred. Finally, the physician should refer any patient with whom he or she feels uncomfortable.

Summary

As more adults who have survived early operation for congenital heart disease enter the patient panels of general cardiologists and primary care physicians, their doctors must become educated about preventive and general medical issues specific to their patients. Physicians must understand how ACHD patients differ from more routine adult patients with acquired heart disease. Most importantly, physicians must be acutely conscious of the indications for rapid consultation with an expert in the diagnosis and management of congenital heart disease in the adult, and must refer them.

Suggested Reading

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