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## CHAPTER 20

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# HEART DISEASE IN THE YOUNG

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## INTRODUCTION

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Congenital heart defects are relatively rare; seriously debilitating heart abnormalities rarer still. Approximately 8 babies in 1,000—representing somewhat less than 1 percent of live births—have some form of cardiac malformation at birth. In only half of these babies is the abnormality severe enough to cause symptoms that could require medical or surgical treatment. It is believed that genetic factors may play a role in the cause of these defects, but the pattern of inheritance is generally unclear. In fact, in all but about 3 percent of cases the underlying cause of the abnormality cannot be identified.

New surgical procedures have been developed in the past few years that can treat defects in children who could once be offered only palliative therapy. Now all but the most severe anomalies can be successfully treated with either medication or surgery. In many cases, open-heart surgery is being replaced by less invasive techniques involving catheterization and balloon angioplasty.

Surgical procedures are being performed on progressively younger children, which has many long-term advantages for these patients. In the 1960s the average child undergoing surgery was of school age; today, about a third of all congenital heart disease cases are being corrected within the first week of life.

The trend reflects increased awareness of the signs and symptoms of congenital heart disease among general pediatricians and family practitioners, as well as the increased number of infants who are diagnosed prior to birth and to improved surgical and postoperative care.

Although congenital heart disease is the most common type of cardiac disease affecting infants and children, it is not the only type. Acquired heart diseases may also affect children, although fortunately the two most common forms of acquired heart disease in children, rheumatic fever and Kawasaki disease, are relatively rare. High blood pressure and high blood cholesterol (hyperlipidemia) are also found in children. Children whose families have either of these conditions are at higher than normal risk of developing them.

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## THE DEVELOPING HEART

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The human embryonic heart begins to form from a single tubular structure in about the fourth week of pregnancy. Within the next four weeks, this tube gradually increases in length, and eventually a loop forms as the tube twists toward the right side. Next a wall, or septum, divides the heart into left and right

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chambers; soon there will be upper and lower chambers as well and four valves to keep blood flowing forward through these four chambers and out to all parts of the body.

It appears that multiple factors, including genetic and environmental ones, may interact to alter the formation of the heart during the first eight to ten weeks of development. A few specific environmental exposures during this critical period can cause structural abnormalities. These include exposure to certain medications (for example, anticonvulsant medications such as phenytoin, the dermatologic medication Accutane, and lithium salts), viruses (rubella, cytomegalovirus), parasites (toxoplasmosis), metabolic disorders (such as uncontrolled diabetes), and excessive alcohol consumption. Fortunately, exposure to these environmental factors does not always result in cardiac abnormalities.

To understand the nature of some congenital anomalies, it is helpful to understand a little about the fetal heart and how it works. Because the fetus uses the placenta, rather than the lungs, to obtain oxygen and to rid itself of carbon dioxide, the path of the blood flow before birth is different from what it will become afterward. To accommodate this, the fetal heart and circulation are somewhat different in structure from the mature beam. Two special blood vessels, the ductus venosus and the ductus arteriosus, and the foramen ovale, an oval-shaped hole in the atrial septum (the wall separating the upper chambers), allow this special circulatory pathway to operate. Normally, all three of these close spontaneously within hours to days after birth. (The normal path of circulating blood in the adult is described in Chapter 1.)

Because of the unique communications that exist within the fetal heart and the lack of dependence upon the lungs for respiration, it is possible for fetal hearts to develop with remarkable degrees of malformation without this causing difficulties for the fetus. Such abnormalities may become important only after the fetal circulation begins its transition to the newborn state, when the two sides of the circulation become separated from each other and the lungs and circulatory system attempt to function on their own.

Certain conditions or syndromes characteristically cause a constellation of related abnormalities in newborns; in these cases genetic abnormalities may cause specific malformations or other problems of more than one organ. For example, about half of babies with Down syndrome have cardiac malformations, and approximately 25 percent of them have an opening in the atrioventricular septum, the wall separating

the right and left atria and the ventricles. More than 90 percent of newborns with other genetic defects (trisomy 13 or 18) have ventricular septal defects. Newborns with other types of genetic abnormalities may have bicuspid aortic valves, aortic coarctation, atrial septal defects, or pulmonic stenosis. These abnormalities of the heart are frequent enough in such syndromes that when a newborn is suspected of having one of these genetic abnormalities, cardiac malformations should also be suspected, even if the baby is not yet showing signs or symptoms of heart disease.

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## THE FETUS WITH CONGENITAL HEART DISEASE

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Clinical research over the past 15 years has made considerable progress in applying ultrasound imaging techniques to perform fetal echocardiography. This diagnostic technique makes it possible to view tiny fetal hearts in action and to diagnose some complex forms of cardiac structural malformation as early as the 16th to 20th week of pregnancy.

The use of fetal echocardiography has reassured many parents that their baby's heart is normal; for others, it has afforded an opportunity to know long before birth that there is a malformation. This, in turn, allows more time for counseling about possible care and future treatment for the baby. A small number of parents have elected to terminate pregnancies when extreme forms of heart disease have been diagnosed in the fetus, especially when these are associated with abnormalities of other organ systems or of the fetus's chromosomes.

Fetal echocardiography is a specialized technique that is not part of the standard ultrasound examination many women undergo during the first or second trimester of pregnancy. Perinatal cardiologists at Yale have advocated that a screening view of the four chambers of the fetal heart be included when any fetal ultrasound study is performed after the 16th week of pregnancy. Almost 90 percent of the severest cases of structural heart disease are suspected on the basis of such studies. Once a suspicion is raised, a more complete and specialized fetal cardiac examination should be performed, ideally involving a joint evaluation by a perinatal obstetrician and a pediatric cardiologist with experience in fetal cardiac imaging. This specialized exam, available at a number of major university medical centers throughout the country, is

## Indications for Specialized Fetal Echocardiography

Specialized fetal echocardiographic studies maybe recommended if previous test have shown any of the following conditions in the fetus, or if the mother has any of the following risk factors.

### Fetal Conditions and Pregnancy Factors

Growth retardation  
Hydrops (swelling of the fetus)  
Heart rhythm disturbances  
Structural abnormalities of other fetal organs  
Abnormal appearance of heart on four-chamber screening exam  
Abnormal location of heart in the fetal chest  
Excessive amniotic fluid (polyhydramnios)

### Maternal and Family Risk Factors

Mother has congenital heart disease.  
Mother has taken any of these medications or substances:  
Anticonvulsants such as phenytoin (Dilantin)  
Isotretinoin (Accutane)  
Lithium  
Cocaine  
Excessive alcohol  
Mother has any of these infections during pregnancy:  
Rubella  
Cytomegalovirus (CMV)  
Toxoplasmosis  
HIV  
Mother has a metabolic disorder such as:  
Diabetes  
Phenylketonuria (PKU)  
Congenital heart disease is present in a previous child, the father, or other relatives.  
Genetic syndromes known to be associated with cardiac disease (e. g., tuberous sclerosis, Noonan syndrome, or Marfan syndrome) are present in the family.

appropriate when specific risk factors are present or suspected, which is the case in about 10 percent of pregnancies. (See box, "Indications for Specialized Fetal Echocardiography.")

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## DIAGNOSING HEART DISEASE IN INFANTS AND CHILDREN

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When heart disease is suspected in a child, whether newborn or older, the pediatric cardiologist generally

employs relatively standard techniques to establish a diagnosis. The evaluation begins with a history of the pregnancy and the child since birth, as well as detailed information about the family to determine if there is any family history of congenital heart disease or syndromes that may be associated with specific cardiac malformations. The rest of the evaluation will entail a physical examination and perhaps other diagnostic procedures.

### PHYSICAL EXAMINATION

Regardless of how sophisticated diagnostic tests have become, a detailed history and a thorough physical examination remain key elements of a cardiac evaluation. Often the diagnosis can be suspected on the basis of these alone, although it may need to be confirmed, or the severity of the problem more precisely delineated, by further tests.

The pediatric cardiologist will listen carefully to the child's heart and lung sounds with a stethoscope, take the pulse in various locations, and check the blood pressure, using a special child-sized cuff. He or she will note the child's skin color and palpate (feel with the hands) the child's chest and abdomen as well as the internal organs. The doctor will also look for any evidence of swelling (edema), typically in the abdomen, eyelids, liver, and, in the older child, ankles. Also in older children, the doctor may check to see if the fingers and toes are bulbous at the ends and the nails curved. This condition, known as "clubbing," is an indication of insufficient oxygen reaching the extremities.

The physician will be able to detect a wide range of conditions by observing certain signs and symptoms. For example, in cases where a cardiac abnormality causes heart failure, an accumulation of fluid in the lungs often causes rapid and labored breathing. This congestion makes it more difficult to expand and deflate the lungs, and the infant may appear to be working harder than is normal to breathe. Such babies frequently feed poorly and therefore may be underweight. These infants have more severe types of congenital heart lesions, but fortunately, these are also less common.

In severe cases with serious congenital abnormalities, where there is congestion of the circulatory system, it is common for children to develop fluid accumulation in the liver. Parents sometimes become alarmed when it is noted that the liver is enlarged, fearing liver disease in addition to a circulatory problem. Actually, the problem is parallel to that often seen in elderly people with circulatory congestion

who develop fluid accumulation in the feet, ankles, and legs at the end of the day. In young children, and especially in infants, who rarely stand, gravity does not make fluid accumulate in these areas. On the other hand, the liver of the newborn is covered with a capsule that stretches more easily than the capsule of an adult's liver. The liver, being a spongy organ with a generous blood supply, serves as a reservoir for the accumulated fluid.

In other instances, a child may have an anomaly that obstructs blood flow from the right ventricle to the pulmonary artery. If the abnormality is associated with a hole or shunt within the heart connecting the right and left sides (either ventricles or atria), oxygen-poor blood may find it easier to cross to the left side of the circulation than to pass through the obstruction to the lungs. This means that not enough blood will be sent to the lungs to pick up oxygen before being circulated to the body. Children with this condition often have a blue (cyanotic) cast to the skin and lips and tend to breathe deeply, in response to the central nervous system's detection of inadequate oxygen within the bloodstream.

The simple act of taking the blood pressure in the legs as well as the arms, in addition to taking the pulse at several points on the body, may reveal the possibility of another congenital anomaly, coarctation of the aorta. In children with this condition, the aorta is narrowed, usually between the arterial branches to the head and arms and the lower body. The pulses in the arms and neck may be easily detected, while pulses to the legs are decreased. For the same reason, the blood pressure in the arms maybe high and the pressure in the legs lower.

The stethoscope is used to listen to the heart and to breath sounds over the lungs. The physician will listen to determine whether the child is able to move air in and out of both lungs, and may detect a bubbling sound (rales) that signifies severe congestion in the lungs. Fluid leakage into the lungs causes bubbles to form when the child's breathing causes air to pass into the fluid-filled areas.

When the cardiologist listens to the heart, he or she is listening to the sounds that correspond to the closure of the valves that separate the atria and ventricles (tricuspid valve on the right and mitral valve on the left). These valves close at the onset of the contraction of the ventricles (systole), during the period when blood is being ejected from the ventricles into the pulmonary artery and aorta. This is the first heart sound. When systole ends and the ventricles relax in order to refill with blood for the next contraction, the valves between the right ventricle and

pulmonary artery (pulmonic valve) and the left ventricle and aorta (aortic valve) close and produce in the second heart sound. Under normal circumstances the cardiologist can discern a splitting of this sound, because the pulmonic valve usually closes a fraction of a second after the aortic valve. If the movement of one of these valves is restricted, or in the extreme case that one valve has not formed, the cardiologist will detect only a single second heart sound.

A murmur or a swooshing sound heard through the stethoscope may be a clue that there is a cardiac lesion. Many families become quite concerned about the presence of a heart murmur, but more often than not it turns out to be inconsequential.

It is important to recognize that a murmur is not a specific disorder, but merely a sign; it is an extra sound that the physician hears in addition to the first and second heart sounds. The stethoscope detects the presence of vibrations in the structures beneath it. These vibrations are transmitted to the listener's ear and detected as noise. Keeping in mind at the heart functions as an elaborate pump that propels blood to various parts of the body, it is not surprising that vibrations may be created, sometimes even in the perfectly normally functioning heart and circulatory system. Many vibratory murmurs that are heard during childhood are "innocent," or "functional," murmurs that do not denote any structural abnormality. Many, if not most, children maybe found to have such murmurs at sometime or other during their development, and they require no specialized tests, treatment, or follow-up.

By determining the timing of the murmur and its quality, location, and distribution, the cardiologist should be able to determine whether a given murmur is innocent or whether it is being created by a valve leak, a hole between areas of the circulation, or an obstruction in a valve or artery.

While many patients may be evaluated using these classic diagnostic techniques and may not require further testing, in many cases some laboratory evaluations are advisable. In such cases the cardiologist usually follows a logical sequence, choosing the test that is most likely to supply the needed diagnostic information for a given suspected diagnosis. In most situations the cardiologist prefers to use the least invasive and least costly tests first.

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#### ELECTROCARDIOGRAM (ECG)

This procedure involves the use of electrodes, placed in contact with the child's limbs and chest wall, which detect the electrical activity of the heart. The electro-

cardiogram is very useful for evaluating disorders of the heart rhythm as well as possible deficiencies in the delivery of blood and oxygen to the heart itself. The information may also suggest that the walls of the heart or its specific chambers are enlarged or thickened, although the echocardiogram (see below) can determine this more specifically. The test detects the heart's own electrical current and does not expose the patient to any form of energy, so there is neither risk nor pain involved.

### **CHEST X-RAY**

The chest X-ray is useful in determining the exact size and location of the child's heart. In some complex forms of heart disease the heart may be displaced from its usual position in the left and center of the chest. The chest X-ray also provides important information concerning the amount of blood flowing to the patient's lungs and may detect the presence of fluid accumulation, pneumonia, or "air trapping" in the child's lungs. This procedure is painless, but does require that the child hold still (in some circumstances a special restraint may be needed). There is no pain, and the radiation exposure is minimal.

### **BLOOD TESTS**

Blood tests may help assess the severity of certain types of congenital heart disease. In cyanotic heart disease, for example, where there is an inadequate blood supply to the lungs, the body's response is to increase the number of oxygen-carrying red blood cells, so that the same volume of blood will be able to deliver more oxygen to the tissues.

### **ECHOCARDIOGRAPHY (CARDIAC ULTRASOUND)**

This painless test uses high-frequency sound waves to provide "sonar" pictures of the heart and great arteries. These cross-sectional views of the heart and its various internal structures give a complete picture of relative sizes of the structures and wall thicknesses of the heart and its attached blood vessels. Further, echocardiography can be used to assess the functioning of the ventricles and, with the addition of Doppler techniques, to measure blood flow patterns and velocities within the heart and blood vessels. This is the most valuable noninvasive test for obtaining anatomic detail in cases of congenital heart disease. (See Chapter 10 for more details.)

Although there is no risk from echocardiography itself, it does require that the child be still for long periods. This may require sedation or, in the case of transesophageal echocardiography (see below), even anesthesia, which carries a small risk of its own.

More recently, echocardiographic equipment has been modified to allow the transducer (the hand-held microphones that both transmit and receive sound waves) to be mounted on an endoscope (a tube-shaped apparatus) that can be passed into the patient's esophagus. This is useful for visualizing hearts in patients with large chests or chest deformities that make standard echocardiograms from the chest surface difficult. When necessary, transesophageal echocardiograms can be done in children if they are large enough to have an esophagus that can accommodate the endoscope. Transducers small enough to be used safely in infants are now being tested. At this point the most compelling indication for transesophageal echocardiography is in the operating room at the time of open-heart surgery. There it is used to evaluate cardiac anatomy just prior to heart surgery and to evaluate the anatomic and functional results of the surgery immediately afterward, before the patient is sent to recovery.

### **CARDIAC CATHETERIZATION**

This procedure involves inserting a small-diameter cardiac catheter—a hollow, flexible tube—into a vein or artery and then advancing it under fluoroscopic control into various cardiac chambers and blood vessels. The physician carefully manipulates the catheter into the desired location and can then measure the blood pressure and flow in the various heart chambers and blood vessels. If a radiopaque dye is injected through the catheter, X-ray motion pictures called angiograms can be recorded. These movies demonstrate the blood flow through the normal and abnormal structures of the heart and are often ordered if cardiac surgery is being considered,

Cardiac catheterization (which is described in greater detail in Chapter 10) is a common diagnostic technique for coronary artery disease and other cardiac disorders in adults, and cardiac catheterization laboratories are available in many hospitals. It is essential, however, that pediatric cardiac catheterizations be performed in a dedicated pediatric cardiac catheterization laboratory by a team that includes physicians, nurses, and technicians specifically trained in pediatric techniques. The staff should also be able to provide extensive pre-catheterization counseling for both parents and patients, ideally aug-

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mented by written information geared to each level. If such counseling is unavailable or inadequate, parents should seriously question whether the particular facility being considered is a truly experienced pediatric cardiac center. Only a relatively small number of children with congenital heart abnormalities actually have to undergo cardiac catheterization.

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## SPECIFIC CONGENITAL MALFORMATIONS

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The prognosis for children with most of the following congenital defects is good, and for some conditions it is excellent. Most will lead full, normal lives, although some will require surgical therapy and long-term monitoring, and in a few cases, activity (specifically, competitive sports) may have to be curtailed. Regardless of the extent of recovery, however, some of these children will have a lifelong increased risk of developing bacterial endocarditis. Fortunately, the risk can be significantly decreased by taking prophylactic measures. (See box, "Preventing Bacterial Endocarditis.")

### PATENT DUCTUS ARTERIOSUS (PDA)

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The ductus arteriosus is a shunt pathway in the fetal blood circulation that connects the pulmonary artery to the descending aorta. It provides a detour for fetal blood to bypass the lungs and travel instead to the placenta, where it receives oxygen. At birth, the umbilical cord is clamped, the infant begins to breathe through the lungs, and the duct is no longer needed. As the amount of oxygen in the blood increases, the body's production of prostaglandin E<sub>1</sub>, a chemical substance that is thought to keep the ductus open, decreases. The duct closes functionally within hours after birth and permanently within the first few days of life.

In some newborns, however, this shunt does not completely close, a condition known as patent ductus arteriosus. Now, because of differences in pressure in the blood vessels, too much blood, rather than hardly any, travels to the lungs. Infants who are born extremely prematurely or those whose mothers had rubella (German measles) during pregnancy appear to be at higher risk for this condition.

In cases of patent ductus, the symptoms will depend upon the size of the duct opening as well as the age and degree of prematurity of the infant. If the

## Preventing Bacterial Endocarditis

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Children and adults with valve damage or congenital heart disease are particularly at risk of developing bacterial endocarditis, an infection of the endocardial tissue that lines the heart walls, valves, and blood vessels. Sometimes called infective endocarditis, this is a serious illness whose treatment requires intravenous antibiotics administered during a lengthy hospital stay.

Any situation—from simple teeth cleaning to complex surgery—that could result in bacteria entering the bloodstream potentially exposes the patient to this infection. Fortunately, the risk can be substantially reduced by consistent use of prophylactic antibiotics. The American Heart Association and American Dental Association recommend that antibiotics be administered an hour before and six hours following any procedure that is likely to result in exposure. This includes surgical procedures performed on any unsterile body parts—the mouth and upper respiratory, gastrointestinal, and genitourinary tracts.

Prophylaxis is recommended for all congenital cardiac malformations, with the exception of isolated *ostium secundum* atrial septal defect. Parents should discuss these preventive measures with their pediatrician, pediatric cardiologist, and dentist. Local chapters of the American Heart Association can supply free wallet cards detailing the recommended antibiotics and appropriate doses. (See Chapter 13.)

ductus is only slightly open, the amount of excess blood circulation to the lungs will be very small, and the only finding may be a characteristic continuous heart murmur, with no symptoms.

At the other end of the spectrum is the child with a completely open ductus arteriosus, which may literally flood the lungs and pulmonary blood vessels in early life. Such children usually become short of breath and show other evidence of pulmonary congestion, such as easy fatigue. They often have difficulty with feeding and may have delayed development. If the problem is not recognized promptly there may be chronic pulmonary infections as well.

Patent ductus may not be recognized initially in extremely premature infants, because many of them suffer hyaline membrane disease (the pulmonary disease of prematurity), which keeps them on a ventilator while their lungs mature. As the lung disease improves, however, the expected ability to be weaned

from the ventilator does not materialize, because the lungs rapidly become compromised by increased blood flow and the accumulation of excessive fluid (pulmonary edema).

Surgery has been used for many years successfully to ligate, or tie off, a patent ductus. The surgery (described in Chapter 25) carries minimal risk and is recommended for all children who have this condition, even if the opening is small and the child has no symptoms. This is because patent ductus, however minor, carries a risk of developing endocarditis, a bacterial infection of the heart or blood vessels that bears a much greater risk for the child than does the surgery.

For premature infants, surgery can be avoided in many cases by administering medications such as indomethacin, which interfere with the body's production of prostaglandins and cause the duct to close spontaneously. Indomethacin is unlikely to be successful in full-term infants or older children, however, because the patency is usually due to an abnormal ductus arteriosus rather than delayed closure. In these cases, surgery is required.

In recent years a special technique has been developed for closing a patent ductus arteriosus under local anesthesia in the catheterization laboratory. This procedure uses a detachable, miniature umbrella-like device made of stainless steel covered with a Dacron mesh, the same material used for cardiac surgical patches. Under fluoroscopic guidance, it is advanced inside a catheter into the open ductus and released. It remains in place permanently and is quickly covered with the patient's own tissue. Although there is no risk of rejection and little risk to the procedure at all, it is still considered investigational by the Food and Drug Administration. Many experts believe it will soon become the treatment of choice for patent ductus in all but premature infants, replacing surgery.

### SEPTAL DEFECTS

The cardiac septa are the walls within the heart that separate the right and left atria from one another, as well as the right and left ventricles from each other. Sometimes an infant is born with a hole in the septum, called either an atrial septal defect or a ventricular septal defect, depending on its location. Since intravascular pressures in the left heart chambers generally exceed those in the right, in cases of a septal defect without other complications, the blood will leak from the left into the right chamber; this is known as a left-to-right shunt.

### VENTRICULAR SEPTAL DEFECTS

The ventricular septum consists of three distinct areas that fuse together to form a single, solid muscle wall. When there is a ventricular septal defect, some of the oxygen-rich blood within the left ventricle that is supposed to be pumped out to the body through the aorta is ejected through the defect directly into the right ventricle and then to the pulmonary artery, through which it goes to the lungs rather than into the aorta. If the defect is small, there may be no symptoms and the only sign may be a loud (but harmless) murmur. The defect may ultimately close by itself or remain open with no real damage to the heart or pulmonary circulation.

If the defect is large, on the other hand, the amount of blood that recirculates through the pulmonary circulation may be great. The lungs may become congested, leading to shortness of breath and, in many cases, failure to gain weight adequately.

In the case of a large defect, the increased amount of blood returning to the heart from the lungs means that the left side of the heart also becomes overburdened. In order to provide adequate blood flow to the body, the heart pumps harder than normal and the heart rate and the force of contractions are increased. This may lead to an enlargement of the heart. The increased flow and pressure in the lungs may eventually lead to damage to the pulmonary blood vessels. If the heart is not able to sustain this extra work, the child may go into heart failure, which can lead to a backup of blood in the veins and accumulation of fluid in the lungs and other body tissues.

A newborn with a ventricular septal defect often will not develop a full set of symptoms for several days or weeks, because the left-to-right shunt is dependent upon the drop in pulmonary resistance that does not occur until some days after birth. The most compelling problems involve respiratory distress and growth failure. Medications may be used to help the heart and lungs to compensate for the extra burden that the ventricular defect and shunt place upon them. Digoxin and diuretics are most commonly prescribed, as well as vasodilators. If medical therapy is unsuccessful, or if the amount of blood flow into the lung or the blood pressure within the pulmonary vessels remains elevated for several months, surgery is recommended to close the defect with a patch. (See Chapter 25 for description.) In situations where there is only one hole, surgery is almost always successful.

Many ventricular septal defects, including some that are large enough to cause symptoms in the newborn period, tend to narrow spontaneously or even

close completely. If the defect has not completely closed by the time the child is 5 to 7 years old, it is not likely that it will. On the other hand, in the absence of a significant overcirculation to the lungs, enlargement of the heart, or increase in pulmonary artery pressure, the mere presence of a ventricular septal defect does not warrant surgical treatment.

### ATRIAL SEPTAL DEFECTS

Like the ventricular septum, the atrial septum forms during the embryonic development of the heart. Defects may develop at a number of locations in the wall (see the later discussion of atrioventricular canal defect). Most commonly, the defect involves the foramen ovale, an oval-shaped hole in the wall that is present in all children during the fetal period but closes spontaneously soon after birth. If the foramen ovale is larger than it should be or if the flap of tissue that usually closes it is displaced or deficient, the hole may remain after birth. Such a problem is referred to as an *ostium secundum* defect.

In most patients with atrial septal defects, symptoms are rare during childhood. While children with large atrial left-to-right shunts are often thin, neither respiratory symptoms nor severe growth failure is common. If anything, there may be a heart murmur, but it may not be evident until the child's second year. In itself, the murmur is not a problem, but it should be monitored periodically.

With the passage of years, however, these low-pressure shunts at the atrial level result in gradual enlargement of the right atrium and ventricle. It is not unusual for adults with this defect to develop symptoms related to cardiac rhythm disturbances such as atrial fibrillation or, later on, evidence of congestive heart failure. (See Chapters 14 and 16.)

Perhaps 25 percent or more of atrial septal defects diagnosed incidentally because a murmur is heard or through echocardiography done during infancy will close spontaneously. When the child reaches 2 or 3 years of age, most atrial defects that are producing clinical signs (enlarged right heart, substantially increased pulmonary blood flow), although not necessarily symptoms, should be repaired. This is because spontaneous closure after this age is highly unlikely and there is a risk of symptoms in later life.

Traditional surgical repair for an atrial septal defect, involving a Dacron patch, is both safe and effective. Even more promising is a new clamshell-like double-umbrella device that can be implanted under

local anesthesia via a catheter procedure (similar to the one described in the earlier discussion of patent ductus arteriosus). Although the device is still considered investigational, it is expected to be approved by the Food and Drug Administration and ultimately to replace open-heart surgery in more than half of atrial septal defect repairs. The treatment of the other forms of atrial septal defect requires surgery in all cases. (See Chapter 25.)

### ATRIOVENTRICULAR SEPTAL DEFECT (CANAL DEFECT)

This defect, also called an atrioventricular septal defect, is usually quite complex, and can be partial or complete. The complete defect, which is more common, involves the portion of the heart where the atrial septum (the wall vertically dividing the heart's upper chambers) meets the ventricular septum (separating the heart's lower chambers), as well as the valves—mitral and tricuspid—that divide these chambers horizontally. Partial defects may involve only the lower portion of the atrial septum (called *ostium primum* atrial septal defect and usually associated with a mitral valve defect) or, rarely, may involve only the ventricular septum, with or without a mitral valve abnormality.

The effect of the complete canal defect is a large hole spanning both the upper and lower parts of the septum and the presence, in place of two discrete mitral and tricuspid valves, of one large valve that spans both sides of the defect. The defect is compounded by the fact that this rudimentary valve does not always close properly, so that some of the blood flows back, or regurgitates, into one of the upper chambers. These defects usually result in an excessive amount of blood flowing to the lungs (a large left-to-right shunt) early in life, which produces severe symptoms of congestion and pulmonary hypertension. Infants with this congenital defect are often emaciated because of the hard work required to breathe and the consequent inability to take adequate nourishment. Their lips and fingernails may appear blue (cyanotic) if they have severe pulmonary edema.

Atrioventricular septal defect is often associated with Down's syndrome: Approximately 25 percent of children born with this syndrome have the defect, while approximately 50 percent of children with the defect have Down's syndrome.

Surgical intervention for atrioventricular septal defect is usually required within the first few months of life, regardless of the presence of Down's syndrome, because medical management is rarely able

to prevent severe congestive heart failure. Without surgery, there is usually irreversible damage to the walls of the pulmonary blood vessels. (See Chapter 25.)

#### SEMILUNAR VALVE STENOSIS (AORTIC STENOSIS, PULMONARY STENOSIS)

The semilunar valves are the aortic, which lies between the left ventricle and the aorta, and the pulmonary, separating the right ventricle from the pulmonary artery. They are so named because their leaflets (flaps of tissue at their openings) are shaped like crescent moons. Stenosis, or a narrowing of the opening, may occur in either of these valves because they are thicker than normal or because they have deformed or fused valve leaflets.

In a child with pulmonary stenosis, the right ventricle must pump more vigorously to send sufficient blood to the lungs to be oxygenated. As a result, its muscular walls may become enlarged, thickened, and less efficient. If the stenosis is severe, the child may tire rapidly from any sort of exertion. An infant with severe stenosis may appear quite cyanotic and need immediate treatment—the hormonelike substance prostaglandin E<sub>1</sub>, followed by balloon valvuloplasty (see below). If the impairment is only slight, treatment may not be necessary, but the child should be monitored for any indication that the condition is worsening.

In cases of aortic stenosis, the narrowed entrance to the aorta makes it more difficult for oxygenated blood to reach the body, and it may result in severe changes in the muscle of the left ventricle, gradually resulting in congestive heart failure. These babies are often found to have scarred and dilated left ventricles, in which case the prognosis is quite poor. At the other end of the spectrum are children with mild stenosis who may have no symptoms at all (although a heart murmur may be present), nor require treatment.

Although surgery has been the traditional treatment for children with either of these conditions, most newborns who have a severe obstruction in either valve now receive balloon dilation valvuloplasty. In this procedure, a balloon-tipped catheter is threaded through an artery or vein into the heart to the center of the valve. The balloon is then inflated, stretching the valve's opening.

Balloon valvuloplasty for the pulmonary valve has been very successful, and at Yale and other institutions it has completely replaced surgery for both newborns and older children. In contrast to the con-

dition in adults, aortic balloon valvuloplasty has a promising outlook, although it may carry a risk of the development of valve insufficiency (also a risk with traditional surgery), in which the valve does not close properly and blood “leaks” backward.

Children who undergo surgical or balloon valvuloplasty of the aortic valve need to be followed throughout life, because there is a tendency for the valve to calcify and degenerate, along with recurrent stenosis or progressive insufficiency or both. A complete surgical replacement of the valve may ultimately be required.

#### COARCTATION OF THE AORTA

This is a constriction, or narrowing, of a section of the aorta, the main artery carrying blood from the heart to the body. It increases pressure in the arteries closest to the heart, those serving the head and arms, while circulation to the legs remains poor.

In the newborn, coarctation may not be evident for as long as a week—until the ductus arteriosus closes. In some cases this is accompanied by a sudden obstruction to blood flow from the left ventricle, and may lead to acute heart failure and shock. Such infants may need respiratory support and will require medical treatment with prostaglandin E<sub>1</sub>, which may help reopen the ductus. Other drugs may be given to improve cardiac contraction, but ultimately, surgery will be needed.

Coarctation may also be associated with other cardiac malformations, including ventricular septal defects and aortic or mitral valve abnormalities. Infants with these multiple anomalies are often quite ill, and require aggressive medical and surgical treatment.

Surgery involves either removing the constricted section of the aorta completely, patching it with a synthetic material, or creating a patch from a section of artery from an arm. In approximately a third of surgical repairs in newborns the coarctation recurs, regardless of the type of surgery. Recurrent lesions may be manageable using balloon angioplasty.

Occasionally coarctation may not be diagnosed until later in childhood. This almost invariably is associated with hypertension in the upper body that had not been detected earlier. If the condition is severe, the child may tire quickly, have headaches, leg cramps, and a pale appearance, and develop slowly.

For children, surgery (described above) is still routinely performed, but balloon angioplasty techniques are being refined and may ultimately replace surgery. In this case, a balloon-tipped catheter is advanced

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into the aorta and the balloon inflated until it stretches the areas of constriction.

The prognosis for infants and children who have surgery is quite good. Without repair, hypertension can become severe. It also appears that the older the patient at the time of repair, the higher the likelihood that he or she will have persistent hypertension.

### CYANOTIC CONDITIONS

Children with certain types of congenital anomalies have a cyanotic appearance a blue tinge to the mucous membranes, particularly evident in the lips and the finger- and toenail beds. This is a result of lower than normal amounts of oxygen in their systemic blood (the blood that circulates from the heart to all parts of the body except the lungs). In these children, defects that result in right-to-left shunting allow blood from the veins (after it has delivered its oxygen to the cells) to mix with oxygenated blood in the arteries. Such children may have obstructions to blood flow to the lungs combined with communications (holes) within the heart that allow the right-to-left shunt. A third type of defect, transposition of the great arteries, results in systemic venous blood returning directly to the aorta, because the aorta is abnormally connected to the right ventricle, while the pulmonary artery arises from the left ventricle. (See discussions of specific conditions below.)

Children who have deep cyanosis tend to be “hyperpneic”—that is, their central nervous system response to low levels of oxygen results in deep breathing, but it is generally unlabored. This is in contrast to children with left-to-right shunts, who, because of excess fluid in the lungs, have very labored (dyspneic) breathing.

### TETRALOGY OF FALLOT

The most common cause of cyanotic congenital heart disease, tetralogy of Fallot is classically described as having four basic components: a ventricular septal defect, pulmonic stenosis (narrowing), an aorta that is unusually positioned above the ventricular septal defect, and thickening of the right ventricular muscle. In actuality, there is one primary problem with cardiac malformation: the upper portion of the ventricular septum aligns incorrectly, resulting in the presence of a large ventricular septal defect under the aorta. This is positioned in such a way that the aorta appears to arise from both the left and right ventricles. The misaligned septum narrows the outlet

from the right ventricle to the pulmonary artery, resulting in pulmonary stenosis. Because the blood flow into the pulmonary artery is obstructed, the oxygen-poor blood entering the right ventricle finds it easier to enter the aorta and body than to enter the pulmonary artery.

Infants with this defect usually develop symptoms at an early age; cyanosis may be apparent shortly after birth. Toddlers may tire easily or even faint as a result of normal exertion.

Tetralogy of Fallot is very amenable to complete surgical repair (described in Chapter 25), and children who have this operation before school age can lead relatively normal lives. Recent evidence, however, indicates that those who undergo surgery very early seem to have better heart function than children who undergo surgery at a later age. Further, palliative shunts (temporary holding measures) have been shown often to result in inadvertent scarring and deformation of the pulmonary arteries, making subsequent surgery more difficult.

### TRANSPOSITION OF THE GREAT ARTERIES

In transposition of the great arteries the aorta arises from the right ventricle, while the pulmonary artery arises from the left ventricle, instead of the reverse. The result is that blood that has already been oxygenated returns to the lungs for more oxygen, while blood lacking oxygen is circulated to the body. Without some immediate way for these two parallel circulations to mix, the infant will not survive. Ironically, what allows survival is another defect, either patent ductus arteriosus or an atrial or ventricular defect, that provides a path for some oxygenated blood to reach the body's tissues.

Until fairly recently, only infants with a second defect were able to survive. With the advent of an interventional catheterization procedure called balloon septostomy, this pathway can be created on an emergency basis. A balloon-tipped catheter is threaded through a vein into the right atrium, across the foramen ovale, into the left atrium, and then forcefully withdrawn until the inflated balloon tears a small flap of tissue, resulting in the production of an atrial septal defect.

The septostomy is only a temporary measure to provide oxygen to the body. It must be followed by surgery to correct the physiology permanently. The most recent procedure used to repair the defect is an arterial switch, in which the aorta and the pulmonary artery are retransposed with the coronary arteries reimplanted to restore normal blood flow. Although

patients who undergo this procedure seem to have a much better prognosis than those who receive the atrial switch developed some 30 years ago, the long-term results of the procedure remain to be seen.

### PERSISTENT TRUNCUS ARTERIOSUS

This extremely rare condition is the result of a misalignment of the ventricular septum. The pulmonary arteries appear to arise as branches from the ascending aorta ("truncus"). Although this condition is considered a form of cyanotic heart disease because the circulating blood has less oxygen than normal, infants with this condition seldom appear cyanotic. They do develop an extremely heavy overcirculation of blood to the lungs and are usually quite ill with congestive cardiac failure. The extra work these infants must do just to breathe makes it difficult for them to feed, and many fail to develop properly. Some of these infants develop severe insufficiency (leakage) of the valve that controls the origin of the truncus, a rare condition that can be fatal.

The prognosis for children with this defect has improved considerably in recent years with the advent of surgery for complete repair in early infancy, rather than early palliative procedures with later repair. Early surgery helps prevent severe malnutrition and pulmonary vascular damage. The condition usually requires another operation prior to starting school, and perhaps one or two further open-heart procedures in later childhood or early adulthood.

Some infants with persistent truncus arteriosus are found to have deficiencies in calcium metabolism (known as hypoparathyroidism) and cellular immunity (thymic aplasia), and they may be at risk for unusual infections.

### TRICUSPID ATRESIA

Atresia refers to the complete absence of an opening in a body organ. In tricuspid atresia the valve that allows blood to flow directly from the right atrium to the right ventricle is absent. This anomaly is usually found in conjunction with an atrial septal defect, which does allow the blood to leave the right atrium. In infants with tricuspid atresia, the systemic venous blood (returning to the heart after delivering oxygen to the body) crosses the atrial septum through the defect into the left atrium, where it mixes with oxygenated blood. This oxygen-deficient mixture then flows through the mitral valve and enters the left ventricle to be pumped out to the body, resulting in cyanosis.

In some cases, the great arteries arise normally—the aorta from the left ventricle and the pulmonary artery from the right ventricle, which is usually hypoplastic (underdeveloped). In such cases there is a ventricular septal defect as well, and the pulmonary valve, which allows blood to flow from the right ventricle to the pulmonary artery, is usually stenosed (narrowed). If the ventricular septal defect is large, the child may have excessive blood flow to the lungs, leading to congestive heart failure. If the pulmonary valve is narrowed or if the ventricular septal defect spontaneously closes, the child will develop cyanosis.

In other cases, the great arteries are transposed (see earlier discussion), producing increased pulmonary blood flow and leading to congestive heart failure, or there may be pulmonic stenosis, resulting in substantial cyanosis.

Early surgery is necessary to ensure that blood flow is as normal as possible, even though the procedure at this point is only palliative. To maintain or increase blood flow to the lungs and relieve the cyanosis, septostomy with a balloon (see the earlier discussion of transposition of the great arteries) or blade is used to enlarge or keep open the atrial septal defect. Alternatively, surgery may be performed to create a palliative shunt between pulmonary and systemic circulations to increase blood flow to the lungs until a type of surgery called a Fontan procedure can be done. Although the Fontan procedure is not a true repair, it is the best long-term solution available at this time. It has been in use for many years and allows the child to grow up with a reasonably normal, although somewhat restricted, range of activities. Long-term monitoring by a pediatric cardiologist will be required.

On the other hand, if there is too much blood flow to the lungs, resulting in severe congestive cardiac failure, the condition may be treated medically with digoxin and diuretics. If this proves unsuccessful, surgery may be used to place a constricting band around the pulmonary artery until more permanent surgery can be attempted.

### PULMONARY ATRESIA

In this congenital anomaly there is no pulmonary valve to allow blood flow from the right ventricle into the pulmonary artery. Newborns with this condition are usually quite cyanotic (blue in appearance), because not enough blood reaches the lungs to become oxygenated. Some of the blood that should flow from the right atrium into the right ventricle instead is shunted across an opening in the septum into the left

## SPECIAL SITUATIONS

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atrium. The blood that reaches the left atrium mixes with oxygenated blood from the lungs, and this oxygen-deficient mixture is circulated throughout the body, producing the symptoms of cyanosis.

Some blood does manage to reach the lungs because it flows through the ductus arteriosus, a passageway that is present during the fetal period that connects the pulmonary artery to the descending aorta. This duct usually begins to close within hours after birth, worsening the cyanosis, unless prostaglandin  $E_1$  is administered to keep it open until surgery can be performed.

The prognosis for pulmonary atresia represents a broad spectrum, depending upon the size of the right ventricle and the pulmonary arteries. If the right ventricle is very small and thick-walled, the long-term effectiveness of surgical repair is questionable. If it is slightly larger but unable to act as a pump, a Fontan procedure (described in Chapter 25) maybe used to circumvent it by creating a connection directly from the right atrium to the pulmonary arteries. If the ventricle is of adequate size, it may ultimately be included in the circulation after surgery. To increase the size of the pulmonary arteries, balloon angioplasty may be required.

Children with pulmonary atresia will require long-term follow-up by a pediatric cardiologist, even when surgery is completely successful.

### TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION

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This rare cyanotic condition involves an anomalous (out-of-place) return of oxygenated blood via the pulmonary veins from the lungs. Oxygenated blood from the lungs, rather than returning to the left atrium, flows instead through the anomalous vein directly to the right atrium or to veins returning to the right atrium. There it mixes with unoxygenated blood, and some of the mixture flows through a defect in the atrial septum and out through the aorta to the body. If the connections are narrow, this may obstruct vein return from the lungs and lead to severe pulmonary edema and cyanosis. The rest of the mixture flows into the right ventricle and travels to the lungs through the pulmonary artery. Surgery to patch the atrial septal defect and connect the pulmonary vein to the left atrium is generally successful if performed in early infancy.

### HYPOPLASTIC LEFT HEART SYNDROME

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This is potentially the most serious of congenital malformations, with the poorest prognosis. Fortunately,

it is relatively rare. In its mildest form, it includes a moderately small left ventricle with a mild degree of obstruction; at its most severe, the left ventricle is very tiny, underdeveloped, and completely isolated from the rest of the heart and circulation because both the mitral and the aortic valves are missing (a condition called atresia). Such infants may seem normal for the first several days, because the ductus arteriosus has not yet completely closed and it provides a route for systemic circulation. Blood flows from the pulmonary artery, through the ductus arteriosus, into the aorta.

Once the ductus arteriosus closes, the newborn develops shock and ultimately fatal multiorgan failure. Survival depends on maintaining the patency of the ductus arteriosus with the hormonelike substance prostaglandin  $E_1$  until definitive surgery can be performed. The most common surgery for this disorder (discussed in Chapter 25) must be considered, at best, innovative therapy that still borders on clinical investigation. Called the Norwood procedure after the physician who developed it, it is a staged operation that results initially in the infant living with a single (right) ventricle. A rudimentary aorta is constructed at the expense of the pulmonary artery, and a shunt is established to connect the systemic circulation to the pulmonary artery. This procedure is followed some months to years later by a Fontan operation, which is essentially only palliative. Current overall survival rates are in the range of 40-50 percent.

The alternative is neonatal cardiac transplantation. (See Chapter 25.) This is quite innovative, bordering on clinical experimentation, and experience is limited, but initial survival statistics are encouraging, as are midterm results. Whether, in the long term, transplant complications will require retransplantation is not yet known.

Both approaches have promise and should be offered to the families of all infants born with the severe form of hypoplastic left heart syndrome. At present, however, many families who are faced with this tragedy in the absence of encouraging survival rates appear to choose an expectant course of management, providing compassionate pain relief while allowing nature to run its course.

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## HIGH BLOOD PRESSURE

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In adults, the cause of high blood pressure is discovered in less than 10 percent of cases. In these in-

stances (known as secondary hypertension), the cause may be an abnormality of another organ, such as the kidney, or it may even be the result of a congenital heart condition, such as coarctation of the aorta. Secondary hypertension is significantly more common in children than in adults. Sometimes the underlying cause can be remedied and the blood pressure will drop to normal. The earlier high blood pressure is noted, the more likely it is that a specific cause will be found. Primary, or essential, high blood pressure accounts for the other 90 percent of cases.

For all people over age 18, high blood pressure is defined as a reading of 140/90 or more when taken several times over a period of weeks. (See Chapter 12 for information on measuring blood pressure.) For children, these numbers are lower. For example, a pressure of 130/85 would be considered high for a 12-year-old boy or girl. (See Table 20.1.)

Although somewhat under 3 percent of all children are found to have high blood pressure, the condition may be more serious if it starts in childhood and goes undetected for many years. Blood pressure should be measured as part of each child's annual physical. If it is elevated, it should be checked again within several weeks or months, preferably in a setting in which the child is relaxed and comfortable. This is especially important if either of the child's parents or another close relative has high blood pressure. At least once, the child's blood pressure should be measured in the legs as well as the arms.

If a child is found to have high blood pressure, it can sometimes be lowered with simple measures. If the child is overweight, losing the excess weight should be the first step. Ideally, this should be accomplished through exercise as well as calorie reduction. Exercise is not only an important habit to establish for life-long health, but it can have an effect on blood pressure, in addition to helping to control weight.

Reducing the amount of salt in the diet may be helpful. Salty snacks such as potato and other chips,

pretzels, and salted popcorn are some of the foods that boost sodium consumption. Unfortunately, these may not be easy for some children to give up. Parents need to be sure that there are plenty of substitutes—fresh fruit, vegetables, and juice—readily available. Cereal (without the sugar frosting) is a good food for nibbling that need not be limited to breakfast. (See Chapters 5 and 12 for additional information.)

If the blood pressure is very high or resistant to these changes, medication, such as diuretics or angiotensin-converting enzyme (ACE) inhibitors, may have to be considered.

In all cases of elevated blood pressure in children under the age of 8 to 10, some additional studies (such as X-rays of the blood vessels to the kidneys) should be done. In children often a specific cause will be found, and it can usually be treated.

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## HIGH BLOOD CHOLESTEROL (HYPERLIPIDEMIA)

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High blood cholesterol levels, like high blood pressure, can be found in children as well as in adults. In a small number of children, perhaps 1 to 2 percent, this is a result of a genetic abnormality called familial hypercholesterolemia, and the levels can be very high. Another 13 to 14 percent have elevated cholesterol readings unrelated to genetics. The typical high-fat American diet may be one of the underlying causes.

The long-term danger of high levels of cholesterol in the blood is the development of atherosclerosis, a condition in which the inner walls of the arteries thicken and become narrowed and irregular. The process starts when excess cholesterol, a fatlike substance, hardens into plaque and is deposited on the artery walls. Atherosclerosis is a major cause of coronary heart disease.

Some pediatricians and pediatric cardiologists recommend that all children have a blood cholesterol test by the time they enter school, while others feel this is only necessary for children at special risk. At the very least, children with a family history of elevated cholesterol or early heart disease (under age 65) should have an initial test by age 5, with follow-up tests every few years. A ten-year follow-up is probably sufficient for children with normal levels and no family history.

Blood (serum) cholesterol is measured in milliliters

Table 20.1  
Upper Limits of Normal Blood Pressure in Children  
(These numbers are approximate.)

Age	Pressure
Up to 6	110/75
6 to 10	120/80
11 to 14	125/85
15 to 18	135/85-90

per deciliter of blood, ml/all. For children, an ideal level is 150 ml/all or less (compared to 200 ml/all in adults). A child found to have elevated cholesterol should be treated by diet first. A heart-healthy diet recommended by Yale, the American Heart Association, and many other organizations calls for no more than 30 percent of calories from fat and no more than 300 milligrams of cholesterol. (See Chapter 5.) This is a general recommendation for healthy adults and children age 2 and over (under age 2, more fat is needed for development).

If the child's cholesterol is greatly elevated, it may be necessary to restrict fat and cholesterol further. This decision is best made in consultation with a pediatrician or pediatric cardiologist. A registered dietitian may be a useful resource for helping the entire family find practical ways to have a healthy diet.

Exercise can also be helpful in controlling cholesterol by raising the level of high-density lipoprotein (HDL), considered the beneficial form of cholesterol. If diet and exercise are unsuccessful in lowering a very high level (for a child, this is above 250 ml/all), drugs may have to be used.

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## RHEUMATIC HEART DISEASE

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Rheumatic fever has been almost eradicated in the United States in the past three decades, but recently it seems to have reappeared in several communities. Although outbreaks have traditionally been higher among urban children of lower socioeconomic status, the most recent cases have been seen primarily in suburban and rural middle-class children.

Rheumatic heart disease is the end result of rheumatic fever, which follows about 1 percent of cases of acute streptococcal (strep) infection of the throat. Rheumatic fever may be an exaggerated response of the immune system in which the body produces substances to fight off the strep infection that are toxic to its own cells instead.

The symptoms of rheumatic fever, which usually develop within two weeks but may not appear for several months, include fever, skin rash, swelling of the joints, and involuntary twitching of muscles (chorea). In about a third of cases there is inflammation of the heart tissue and valves. In other cases, symptoms may be vague, such as generalized fatigue, and go unrecognized. Most cases occur in children aged 5 to 15, although adults are occasionally affected.

The fever that gives this disease its name may last

up to two weeks and be accompanied by arthritis-like pains in the joints. The pain may migrate from one joint to another and be accompanied by redness and inflammation. A lacy rash called erythema marginatum may also be present, as well as rapid, involuntary twitching and jerking of the muscles. If there is heart involvement, the child may be short of breath, tire easily, and have a poor appetite. If the physician finds heart enlargement and a murmur, chest X-rays, an electrocardiogram, and possibly an echocardiogram will be ordered to confirm rheumatic heart disease.

Rheumatic fever is treated primarily with bed rest and a number of medications, depending on the symptoms. Aspirin or steroids such as prednisone or cortisone may be prescribed for joint pain and possible inflammation of the heart. Diuretics or a sodium-restricted diet may be necessary to reduce excess work by the heart caused by fluid retention. Sedatives or tranquilizers may be used to relieve muscle twitching.

Children who have had rheumatic fever are at risk of recurrence and are usually given prophylactic antibiotics to prevent this. Prophylactic antibiotics are also recommended before any surgery or other procedure that is likely to involve blood, especially for those who have rheumatic heart disease. This therapy helps reduce the risk of developing endocarditis, a bacterial infection of the heart valves or heart lining.

The dramatic declines in rheumatic fever have been due to better detection and to treatment with antibiotics. Prevention depends upon prompt treatment. A culture should be taken of any sore throat that comes on suddenly and is accompanied by a fever, which may be as high as 104°F. Other symptoms may include headache, nausea, vomiting, and abdominal pain. The culture will determine whether the sore throat is caused by a virus or a bacterium and, if so, which type. (Rheumatic fever can only be caused by group A hemolytic streptococcus.) Strep throat, regardless of which type of bacterium caused it, can be treated effectively with penicillin or other antibiotics; there is no effective treatment for viral sore throat.

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## KAWASAKI DISEASE (MUCOCUTANEOUS LYMPH NODE SYNDROME)

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Kawasaki disease is a rare acute inflammatory disease of children that is thought to be the result of

infection, possibly viral, although a responsible agent has not been identified. Each year in the United States approximately 1,500 children develop this inflammation of the mucous membranes and lymph glands, and 98 percent of these children recover. In about one out of five cases there is cardiac involvement, primarily the formation of aneurysms (outpouchings in the vessel walls) in the coronary arteries. Some patients may have myocarditis, pericarditis, and hepatitis. The 2 percent mortality is related to coronary arterial aneurysms and thrombosis.

The incidence of Kawasaki disease appears to be rising, but this may only be because of better recognition and reporting. The disease itself has only been identified in the past 20 years or so. Incidence is highest in Japan and among Asian children in other countries, including those in Hawaii. It is not limited to Asian children, but incidence appears to be lower among Caucasians. Boys, for unknown reasons, seem to develop Kawasaki disease twice as often as girls. The infection primarily affects children under 10,80 percent of whom are under 5.

The symptoms are varied and uncomfortable, including a sudden high fever (102 °F., spiking to 104 or 105 °F.); a deep red rash that may mimic hives, scarlet fever, or measles; swollen hands and feet that sometimes have dark red or purple blotches on the palms and soles; bulbar conjunctivitis (red eyes); swollen lymph nodes, particularly on one side of the neck; and inflammation of the mouth, with a strawberry appearance to the tongue and swelling and cracking of the lips. There may also be a stiff neck, diarrhea, or abdominal pain. About 40 percent of the children will develop painful but temporary arthritis, or swelling of joints, especially in the legs.

The acute phase lasts several days to a week, with symptoms subsiding as soon as the fever disappears. In two to three weeks, the skin on the hands and feet may peel. Treatment is aimed at ameliorating the symptoms and may require hospitalization. Aspirin may be given to reduce fever and inflammation, but a large single intravenous dose of gamma globulin seems to shorten the acute inflammatory phase of the disease and appears to cut the risk of aneurysm by more than half.

Long-term cardiac complications, if there are any, may be seen as early as two weeks after the onset of the disease. Aneurysm formation and abnormalities of heart function are usually best diagnosed by echocardiography. In approximately one-half of patients whose aneurysms remain after the acute phase of the disease has resolved, these aneurysms will disappear within two years. It is unclear, however, how many

of these children will ultimately develop coronary occlusions and have cardiac symptoms.

In some cases coronary angiography, for diagnosis and prognosis, may be warranted. Long-term antithrombotic aspirin therapy is recommended for coronary artery aneurysm follow-up care, or Coumadin if the aneurysm is uncommonly large. In rare cases, patients ultimately require coronary artery bypass surgery, and some go on to develop myocardial infarctions.

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## SUMMARY

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Most children with cardiac problems who are encountered in 1991 have some form of congenital heart disease. Only about half of the 8 cases of congenital heart disease in 1,000 liveborn children require any form of treatment.

The 1980s witnessed remarkable improvements in surgical management techniques, including the widespread use of cardiac transplants that accompanied the introduction of cyclosporin for immunosuppression. Transplants are now used in the management of complex structural disease in children and newborns. In addition, the 1980s witnessed the development of a technique for treatment of the hypoplastic left heart syndrome.

The 1990s offer the promise of more successful interventional catheterization techniques to ameliorate obstructed blood vessels, and to provide definitive procedures for the closure of intracardiac shunts, without need for open-heart surgery.

The development of new high-resolution and high-speed imaging systems shows promise that may allow MRI and CAT scanners, in tandem with ultrasound scanners, to make cardiac imaging more precise and less invasive than in the past.

The advances that have been made in fetal cardiac imaging have given birth to subspecialty care of the fetus with congenital heart disease. It is likely that future developments will include the introduction of prenatal catheterization and surgical techniques aimed at repair of the malformed fetal heart.

When parents are confronted with the fact that their newborn child has congenital heart disease, there is often a period of disbelief and denial, which is a normal response to learning bad news. Unfortunately, in some of the most severe cases of congenital heart disease, parents may be called upon to make important management decisions, the conse-

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quences of which they and other members of the family may live with for many years to come. These decisions are often made in an extremely emotionally charged atmosphere at a time when reasoned decisions may be difficult. The newborn “management

team” must try its utmost to provide accurate information to the parents regarding diagnosis, management options, and prognosis (short- and long-term) to allow the parents the best opportunity to make informed decisions.