ACYANOTIC HEART DEFECTS

Acyanotic congenital heart defects may be due to obstructive lesions (stenosis) or left-to-right shunts. Lesions with left-to-right shunts include atrial septal defect, ventricular septal defect, and patent ductus arteriosus. Obstructive lesions include pulmonary stenosis, aortic stenosis, and coarctation of the aorta. These babies are “pink” and present with symptoms of congestive heart failure, which may include poor weight gain, feeding or exercise intolerance (baby sweats when he eats), or prolonged recovery from simple respiratory infections. These defect should be dealt with prior to the development of Eisenmenger syndrome, irreversible pulmonary vascular damage (elevated PVR and pulmonary pressures), which causes reversal of the shunt (right to left becomes left to right).

Atrial Septal Defect (ASD)

ASDs constitute 8-13% of all congenital heart defects. There are three main types of ASDs. The most common is the ostium secundum, which arises within the boundaries of the fossa ovalis. A primum ASD results from persistence of the ostium primum and is associated with AV canal defects. A sinus venosus defect usually occurs high on the septum close to the orifice of the SVC and is associated with PAPVR. A rare type of ASD is the coronary sinus defect, which results from unroofing of the coronary sinus as it courses along the left atrial floor.

Patients with isolated ASD are usually asymptomatic. On exam there is a systolic ejection murmur due to flow across the pulmonary valve, and a widely split and fixed S2. CXR shows cardiomegaly with prominent pulmonary vasculature and a prominent PA bulb. EKG may reveal RVH, and incomplete RBBB. Echocardiogram is used to define the defect and can be enhanced with bubble contrast. If performed, cardiac catheterization would show a “step-up” in oxygen saturation between the SVC and RA due to mixing.

In 1952, Drs Lillihei and Lewis performed the first ASD closure, by using cooling blankets to lower the child’s core body temperature to temporarily stop the heart. The
surgery had to be completed in under ten minutes. Today we have the luxury of the cardiopulmonary bypass machine, and can attempt more complex repairs. Closure is indicated for all significant ASDs ($Qp:Qs \geq 1.5$) and should be performed by 4-5 years of age to prevent pulmonary vascular disease. Contraindications to closure include small, hemodynamically insignificant ASDs without cardiomegaly, severe LV failure, and irreversible pulmonary hypertension (PVR of 8-12 Woods units, despite aggressive vasodilator challenge). Device closure at catheterization has become increasingly popular while surgical closure primarily or with a patch (‘tanned’ pericardium) can be accomplished with low morbidity.

**Ventricular Septal Defect (VSD)**

VSD is the most common congenital heart defect, comprising 20-25% of all lesions and occurring in association with other defects about 50% of the time. Classification is based upon the location of the defect in the interventricular septum. The most common type is the **membranous defect**, located in the membranous septum directly under the aortic valve, near the bundle of His. **Muscular defects** are located in the muscular and apical portion of the ventricle and are the most common type of uncomplicated VSD (isolated VSD without associated lesions). Multiple muscular defects may be present, constituting a “Swiss-cheese” type of VSD. **Infundibular defects** (also referred to as conal, subpulmonary, subarterial, and supracristal) are located just below the pulmonic valve. This type may cause aortic insufficiency.

![Diagram of Normal Heart and Hearts with Ventricular Septal Defects](image)

Patients with small defects may be asymptomatic and diagnosed only secondary to a loud holosystolic murmur at the left lower sternal border (smaller VSD = usually louder murmur). The murmur is usually not audible just after birth as the PVR is too high to allow for a significant left-to-right shunt. Patients with larger defects may present with heart failure. The EKG may be normal or show LVH or RVH. CXR will show increased pulmonary
vascularity and possible cardiomegaly. Echocardiography is used to define the defect(s) and cardiac catheterization is not routinely performed but can be used for device closure or to investigate pulmonary vascular resistance or response to vasodilators.

The majority of small membranous and muscular VSDs will close spontaneously, so asymptomatic patients may just be followed. However, infants with heart failure symptoms despite medical therapy require VSD closure. Children older than one year of age with PA pressure greater than one half that of systemic pressure require repair. All VSDs associated with aortic regurgitation should be repaired. In adults, VSDs should be repaired when symptoms are present or the Qp:Qs ratio is $> 1.5:1$. Fixed pulmonary vascular hypertension precludes operative repair. Similar to ASDs, device closure is possible for an increasing proportion of VSDs.

**Patent Ductus Arteriosus (PDA)**

PDA results from persistent patency of the fetal ductus arteriosus, which is necessary during fetal circulation to divert deoxygenated blood from the PA to the descending aorta, away from the unventilated pulmonary vasculature. After birth, the shunt becomes left-to-right and under normal conditions is completely closed in the first 2-3 weeks of life. PDA is common in premature infants with low birth weight. Dr Robert Gross first performed surgical closure of PDA in the 1938 and is regarded as a pioneer in the field, being one of the first to attempt surgery on the major vessels in a neonate.

Symptoms vary from asymptomatic to findings of heart failure and increased incidence of respiratory infections, depending on the amount of shunting. A continuous “machinery” murmur is present at the LUSB and the CXR may show cardiomegaly and increased pulmonary vascular markings. The EKG may show LA/LV enlargement or biventricular hypertrophy. Diagnosis is confirmed by echocardiography, which can also be used to determine the size of the duct and estimate the degree of shunting.

Closure is felt to be indicated for all isolated PDAs to eliminate heart failure, prevent pulmonary hypertension, and avoid endocarditis. Medical closure with indomethacin or
ibuprofen is usually effective in neonates. Surgical ligation or division of the PDA is indicated in neonates for failure of medical treatment, contraindications to indomethacin treatment, and for infants with necrotizing enterocolitis. The approach is through left thoracotomy incision. Treatment via VATS has been used. Complications of surgery include recurrent laryngeal nerve injury, phrenic nerve injury, and chylothorax. Transcatheter closure has gained acceptance and many devices are available.

**Pulmonary Stenosis (PS)**

Pulmonary stenosis at the valvar level accounts for 8-10% of all congenital heart defects. Presentation depends on the severity of the stenosis and the presence of a PFO or ASD.

Most patients present after the neonatal period with a harsh systolic ejection murmur and a thrill. EKG reveals right-axis deviation, prominent P waves, and RVH. CXR may show a prominent pulmonary artery shadow secondary to poststenotic dilation. Echocardiography is used to establish the diagnosis and severity, and associated abnormalities. Cardiac catheterization may be performed for balloon valvotomy.

Treatment is indicated if symptoms are present or the transvalvar pressure gradient is significant. Catheterization with balloon valvotomy is the preferred initial treatment, and may be attempted in cases of recurrent stenosis. Most patients will have pulmonary insufficiency after valvotomy, but it is usually well tolerated. Open pulmonary valvotomy is indicated in cases of failed balloon valvotomy. A transanular patch can be placed to enlarge a small anulus. Long-term outcomes are excellent.

**Coarctation of the Aorta**

Coarctation of the aorta is a narrowing that usually occurs near the site of insertion of the ductus arteriosus. It frequently occurs in association with other heart defects, including bicuspid aortic valve in 50% of patients and VSD in 30-60%.

Nearly half of patients develop symptoms within the first month of life, often in conjunction with closure of the PDA. They present with systemic hypoperfusion, metabolic...
acidosis, and congestive heart failure. They have prominent upper extremity pulses and weak lower extremity pulses. Patients with less severe coarctation or sufficient collateral development (internal mammary, posterior intercostal, anterior spinal) may present as late as adolescence, usually with a murmur or hypertension, exercise intolerance, headaches, or angina. The murmur is systolic ejection and best heard over the left interscapular region. The EKG may show LVH and CXR may show rib notching from hypertophic intercostal arteries. Diagnosis is confirmed by echocardiography, which will also show associated defects. A peak gradient of > 20 mmHg by echo suggests significant coarctation.

Neonates presenting with profound acidosis should be treated similarly to interrupted aortic arch (see below section on IAA) to be stabilized medically prior to operative intervention. Surgical repair is via left thoracotomy (right thoracotomy for a rightward aortic arch). For neonates, primary treatment is extended end-to-end anastomosis, whereas adolescents and adults usually require patch angioplasty or interposition graft due to limited aortic mobility. Another technique is subclavian artery flap angioplasty, where the left subclavian artery is divided and the proximal portion is turned down onto the aorta for reconstruction. This, however, affects limb development. Recoarctation occurs in about 10-30% of patients following repair as an infant. Balloon angioplasty has a 90% success rate when employed for recoarctation, with a 50% reintervention rate at 20 years.

Atrioventricular Septal Defect (AVSD)

Atrioventricular septal defects are also referred to as AV canal defects and endocardial cushion defects. They represent a spectrum of anomalies characterized by varying degrees of incomplete development of the septal tissue surrounding the AV valves in addition to abnormalities of the valves themselves. Partial or incomplete AVSDs have a crescent-shaped primum ASD, positioned just above the AV valve. Complete AVSDs have both defects in the atrial septum just above the AV valves and defects in the ventricular septum just below the AV valves. There is a single AV valve bridging the left and right sides of the heart. Intermediate or transitional AVSDs have both an ASD just above and a VSD just below the AV valves but have two distinct left and right AV valve orifices. As many as 50-75% of patients with complete AVSD have Down syndrome, and AVSD is the most common
congenital heart defect in Down syndrome. They are also present in almost all patients with asplenia.

Presentation depends on the amount of left-to-right shunting and therefore on the pulmonary vascular resistance. Patients with partial AVSDs, and no AV valve regurgitation, may be asymptomatic for years. Patients with complete AVSDs have more left-to-right shunting because of the large VSD. As PVR decreases in the first few weeks of life, patients present with worsening symptoms, especially if there is AV valve regurgitation. There may be a systolic ejection murmur due to increased flow across the pulmonary valve, a holosystolic apical murmur from left AV valve regurgitation, and murmurs from the ASD and VSD. CXR reveals an enlarged PA and RVH. EKG shows RVH and sometimes LVH. Echocardiography is the diagnostic test of choice. Cardiac cath may be used to measure PVR and determine operability (see below).

Similar to secundum ASD, patients with asymptomatic partial AVSD should be repaired electively by about 5 years of age, and sooner if significant left AV valve regurgitation is present. Repair is typically by closure of the left AV valve cleft and patch closure of the primum ASD. Infants with complete AVSD should undergo repair at 2-4 months of age, and sooner if symptomatic. Contraindications include PVR > 10 units/m² or PVR:SVR ratio > 0.7. Repair of the complete AVSD must be taken with care to avoid damaging the AV node.

**Aortic Stenosis (AS)**

Congenital stenosis of the aortic valve is the most common form of LVOT obstruction in the neonate or infant. They will often have a “unicuspid” valve orifice with no appreciable leaflets. The valve itself may be normal, as in supravalvar aortic stenosis, which is associated with Williams syndrome (Chromosome 7). Diagnosis is by echocardiography. They present with profound congestive heart failure and cyanosis. Balloon dilation and operative valvotomy or transventricular dilation have similar risks and their use varies by institution.
Older children with valvar AS usually have a bileaflet valve with fused commissures. They may have exercise intolerance, angina, and syncope or near-syncope. Operation is indicated for a symptomatic child with echocardiography showing a transvalvar gradient > 40 mmHg, LVH, or ischemia, or an asymptomatic patient with gradient > 50-75 mmHg. Open surgical valvotomy is effective for most children with congenital valvar AS, although at least half of patients will require subsequent reoperation. Valve replacement (the Ross procedure) is seldom necessary except in patients with severe aortic insufficiency or for rheumatic heart disease.

Cyanotic congenital heart defects cause right-to-left shunting and include tetralogy of Fallot, transposition of the great arteries, tricuspid atresia, total anomalous pulmonary venous connection, truncus arteriosus, hypoplastic left heart syndrome, double-outlet right ventricle, interrupted aortic arch, pulmonary atresia with or without intact ventricular septum, double-inlet left ventricle, and absent pulmonary valve syndrome. (Note that most of the cyanotic heart defects start with a "T"). These patients present with varying degrees of cyanosis. Until the late 1940s these children were all destined for early death. Drs Alfred Blalock and Helen Taussig and lab assistant Vivian Thomas performed the first surgery for “blue babies” in 1946 at Johns Hopkins (see below, Modified Blalock-Taussig Shunt).

Tetralogy of Fallot (TOF)

Tetralogy of Fallot refers to the constellation of VSD, RVOT obstruction (ie: pulmonary stenosis), RV hypertrophy, and overriding aorta. The VSD is usually large and unrestrictive, and the direction and severity of shunting depend upon the degree of RVOT obstruction. The higher the obstruction to blood flow exiting the RV to the PA, the more blood which will be forced across the VSD. This in turn determines the degree of cyanosis and the age of presentation. Patients with mild RVOT obstruction may have normal O_2 saturations and predominantly left-to-right shunting through the VSD (acyanotic or “pink” tetralogy), while those with severe RVOT obstruction will have significant right-to-left shunting and present early in infancy with cyanosis. Cyanosis is usually mild at birth and gradually progresses with age as RVH and RVOT obstruction worsen. Most patients present by infancy, so the classic description of the older child with TOF squatting to increase SVR, and thereby increase pulmonary blood flow, is seldom seen. Cyanotic “tet spells” are
periods of profound hypoxemia that may be induced by dehydration, respiratory infection, or peripheral vasodilation from medications, resulting in a marked decrease in pulmonary blood flow and increase in the right-to-left shunt across the VSD.

Patients usually have a midsystolic ejection murmur best heard at the left 3rd intercostal space, and is related to the VSD. CXR may show a “boot-shaped” heart, and a right-sided aortic arch is present 25% of the time. The EKG will show RVH and right axis deviation. Echocardiogram will show the typical malalignment VSD with aortic override and RVOT obstruction. Coronary abnormalities may be present and can often be seen on echocardiogram. Cardiac cath is usually not performed prior to surgery unless specific questions arise. Lab studies to detect a deletion on chromosome 22 are also performed, as conotruncal anomalies may arise as part of a syndrome, the most severe form being DiGeorge syndrome.

Most children have adequate O₂ saturations at birth and can be allowed to grow and await elective repair. Prior to repair, children should be kept well hydrated and free from viral infections to avoid spells. If saturations fall below 75-80%, or if patients are experiencing cyanotic spells, surgical repair should be performed. Otherwise, elective repair is recommended by 6 months of age.

Palliative surgery may be performed by using a number of different shunts, all of which have the common goal of establishing blood flow to the pulmonary arteries (see most common, the modified Blalock Taussig shunt, below).

**Modified Blalock Taussig Shunt used for palliation:**
The subclavian artery gives blood to pulmonary artery via Gor-Tex graft.
Complete repair addresses the features of TOF and includes patch repair of VSD, ligation of any existing systemic to pulmonary shunts (congenitally and/or surgically created), and alleviation of the RVOT obstruction.

**Transposition of the Great Arteries (TGA)**

This is the most common of the cyanotic heart diseases. In transposition of the great arteries, the aorta arises from the RV and the PA arises from the LV. Survival is dependent upon mixing, usually by an atrial level connection, most often a PFO. Up to 50% of patients will also have a VSD. The aorta is usually anterior and to the right of the PA. There are a number of configurations of the coronary arteries which must be considered carefully when planning repair.

Presentation depends on the presence of associated cardiac anomalies. If there is no VSD, cyanosis will be more pronounced and may be present at birth. On the other hand, if there is a significant VSD, cyanosis will develop later and heart failure may be the predominant clinical finding. Most patients with TGA and intact ventricular septum have a soft systolic murmur. The EKG will show increasing RVH or biventricular hypertrophy over time. CXR shows an egg-shaped heart, narrow superior mediastinum, and increased pulmonary vascular markings with cardiomegaly. Echocardiography confirms the diagnosis and defines associated atrial and ventricular defects, valve function, pulmonary stenosis, and coronary configuration. Cath is usually reserved for those patients requiring balloon septostomy (patients born without mixing defects like an ASD or VSD- see below).

TGA is the most common congenital heart defect requiring early intervention. Early repairs (Mustard, Senning) re-routed venous return into the atria (atrial switch procedures) but those operations had a high incidence of atrial arrhythmias and late RV failure. Currently, anatomic correction is performed early in life using the arterial switch operation. Since the LV is ejecting against the lower resistance pulmonary circulation, repair must be carried out early or the LV will fail to thicken and be unable to support the systemic
circulation following the switch. If LV pressure has already fallen to less than 60% of systemic, a two-staged approach involving initial PA banding may be used. The PA band restricts pulmonary blood flow and causes the LV to work harder, allowing for growth and thickening. For infants with hemodynamic instability or persistent hypoxemia or acidemia shortly after birth, balloon atrial septostomy may be employed to improve mixing. The arterial switch operation involves division of the PDA, transection of the great vessels, transfer of the coronary buttons to the new aorta, and closure of any defects.

**Tricuspid Atresia**

Tricuspid atresia is the most common type of single ventricle defect. It is characterized by absence of the tricuspid valve and hypoplasia of the RV. *Type I* tricuspid atresia refers to those with normal relation of the great arteries (80%), *type II* those with transposition, and *type III* those with other complex anomalies. There is almost always a VSD, which is frequently restrictive. Blood passes from the RA through a PFO or ASD to the LA, and from there to the LV. If the great arteries are normally related, pulmonary blood flow occurs through a VSD to the hypoplastic RV. The amount of pulmonary blood flow is determined by the VSD, RVOT obstruction, and the pulmonary vascular resistance. The VSD generally gets progressively smaller and the patients become increasingly cyanotic. If the great arteries are transposed, blood flow becomes even more complex. Aortic coarctation is also more common in this group, and leads to CHF quickly. The degree of illness and of symptoms (cyanosis versus congestive heart failure) depend on the amount of pulmonary blood flow.

Most patients present in early infancy with cyanosis or a moderate systolic murmur, while few present with excessive pulmonary blood flow and symptoms of congestive failure. EKG may show right atrial enlargement and left superior axis deviation. CXR will show a small or minimally enlarged heart, except in those patients with increased pulmonary blood flow. Echocardiogram readily demonstrates the anatomic features of tricuspid atresia and associated defects. Cardiac cath is not routinely performed.

The goal of early management is to allow these patients to be satisfactory candidates for the “Single Ventricle Pathway” with the final step being the Fontan procedure. The ultimate goal of this pathway is to re-route all systemic venous blood (from
the cavae) to the pulmonary arteries, bypassing the right side of the heart. A typical pathway would include a modified Blalock-Taussig shunt (see TOF, above for explanation) as a neonate, followed by a bidirectional Glenn shunt (see below) around 6 months, and the completion Fontan (see below). Selection criteria required for a successful Fontan procedure include pulmonary arteriolar resistance < 4 units/m², mean PA pressure < 15 mmHg, normal ventricular function, competent left AV valve, and no PA distortion from previous operations.

Total Anomalous Pulmonary Venous Return (TAPVR)

In TAPVR the entire pulmonary venous circulation drains to the RA, either directly or by systemic vein or sinus connecting with the RA via the SVC, IVC, or coronary sinus. A PFO or ASD is necessary to deliver oxygenated blood to the left heart. TAPVR is classified as supracardiac, cardiac, or infracardiac. In the most common variant of supracardiac TAPVR, all four pulmonary veins drain into a common, left vertical vein which then drains into the innominate vein, azygous vein, SVC, or RA. The classic CXR finding in this type is that of a “snowman” or “figure-of-eight” due to the large vertical vein on the left and SVC on the right. In the cardiac subtype, pulmonary venous drainage is into the coronary sinus. In the infracardiac variant, a vertical vein descends below the diaphragm, where it usually connects with the portal vein and then into the IVC.

In cases without obstruction there is a large left-to-right shunt and patients present with heart failure months after birth up to early childhood. There may be a gallop or faint systolic murmur secondary to increased flow across the tricuspid valve, and a prominent fixed S2. Patients with nonobstructed TAPVR are repaired electively when diagnosed, usually when symptoms develop around 6 months of age.

When there is pulmonary venous obstruction, infants present in extremis within hours to days of birth. They are profoundly cyanotic and in severe congestive heart failure. There is marked pulmonary vasculature and edema on CXR. Diagnosis and mapping of the anomalous connections is possible by echocardiography. Cardiac catheterization is not
commonly performed but it may show equal or similar oxygen saturations in all four cardiac chambers. Obstructed TAPVR is a surgical emergency. Patients require intubation with 100% oxygen, hyperventilation, and correction of acidosis. ECMO has been used as a temporizing measure. The operation depends on the anatomic variant of TAPVR but usually requires deep hypothermic circulatory arrest in neonates. Approximately 5-15% of patients develop recurrent pulmonary venous obstruction within the first year post-op.

**Truncus Arteriosus**

In truncus arteriosus a single vascular trunk arises from the heart and gives rise to the aorta, brachiocephalic, pulmonary, and coronary arteries. There is almost always a VSD, usually a PFO, and an ASD in 10-20% of cases. There is an association with interrupted aortic arch 10-20% of the time, rightward arch 20-35% of the time, and aberrant right subclavian artery about 5-10% of the time. The truncus may equally override both ventricles (~2/3) or be deviated over the RV (~1/3) or LV (5%). Coronary abnormalities are relatively common.
The amount of pulmonary blood flow is mainly affected by PVR. Most patients show only mild cyanosis at birth. As PVR decreases in the first few days of life, pulmonary overcirculation ensues and neonates present with congestive heart failure and cyanosis decreases. In cases with truncal valve regurgitation, heart failure is worsened as coronary blood flow is compromised. Physical exam will reveal a single loud S2 and possibly an S3. There is a pansystolic murmur at the left sternal border and a low-pitched diastolic murmur at the apex from increased flow across the mitral valve. Truncal valve regurgitation will produce a high-pitched diastolic murmur along the left sternal border. The peripheral pulse pressure is widened due to diastolic runoff into the pulmonary bed. The CXR shows moderate cardiomegaly, prominent pulmonary vasculature, and possibly a rightward aortic arch. Echocardiogram is the diagnostic study of choice. Cardiac cath is not routinely performed.

The natural history if untreated is 90% mortality at one year. Neonatal repair is the treatment of choice. After arrest the pulmonary arteries are removed and the defect repaired. In cases of moderate to severe truncal valve regurgitation, repair is performed. An RV infundibulotomy is then made with care to avoid aberrant coronary arteries and the VSD is closed. An RV-to-PA conduit is performed (ie: Sano Shunt, see below).

Sano Shunt: PTFE graft from RV to PA

Hypoplastic Left Heart Syndrome (HLHS)

HLHS includes a wide spectrum of diseases characterized by a varying degree of underdevelopment of the left-sided heart structures, including the LV, mitral valve, aortic valve, and aorta. The RV therefore supports both the pulmonary and systemic circulations, the latter being entirely ductal dependent. Pulmonary venous return mixes in the right atrium via an ASD, PFO or anomalous venous connection. Associated noncardiac abnormalities are common (25%), and 5% will have a trisomy syndrome.

Neonates may appear normal at birth, but become tachypneic and pale within hours to days, and may progress rapidly to acidosis, cyanosis, and hemodynamic collapse. Symptoms are dependent upon the size of the interatrial communication. Patients with severe restriction at the atrial septum will present with respiratory distress, cyanosis, and acidosis immediately after birth (functionally equivalent to obstructed TAPVR) and require immediate balloon atrial septostomy. On exam there is a dominant RV impulse, single S2, and nonspecific soft systolic murmur. EKG reveals RA enlargement and RVH. CXR shows mild cardiomegaly and increased pulmonary vasculature. Echocardiography is the diagnostic study of choice and provides anatomic details about the aortic valve, aorta,
coronary anatomy, atrial septum, and other valvular function. Cardiac cath is not routinely performed.

Patients should be treated with PGE₁ to maintain ductal patency and supplemental oxygen should be avoided since it causes pulmonary vasodilation and decreases systemic perfusion in the single ventricle child. Surgical treatment is either by staged palliation ending with the Fontan procedure or by transplantation (see above, Tricuspid Atresia and the "Single Ventricle Pathway"). The first stage in palliation is the Norwood procedure.

The procedure takes place under deep hypothermic arrest. The main PA is transected and the diminutive aorta is incised longitudinally. The proximal aorta is anastomosed to the proximal main PA and the aortic arch is augmented with a homograft patch. PA blood flow is supplied by either a right modified BT shunt (innominate artery to right PA) or a Sano shunt (RV to main PA- see above). Second and third stage palliation with bidirectional Glenn/hemi-Fontan and completion Fontan are completed as described for tricuspid atresia above.
Double Outlet Right Ventricle (DORV)

In DORV, the PA and at least 50% of the aorta arise from the RV. There is almost always a VSD. Four groups exist, based on the relationship of the VSD to the great arteries. The VSD can be subaortic, subpulmonary, doubly committed, or noncommitted. Therefore, DORV encompasses a spectrum of disease that ranges anatomically and physiologically from TOF to TGA.

Interrupted Aortic Arch (IAA)

In interrupted aortic arch there is an absence of luminal continuity between the ascending aorta and the descending aorta. There are three types of IAA, and each describes where the interruption takes place. Most common type is B, where the defect lies between the left carotid and left subclavian artery. There is almost always a large VSD. An aberrant right subclavian artery arising from the descending thoracic aorta and passing posterior to the esophagus is common, especially in type B. Blood reaches the descending aorta via the ductus.

Most patients present within the first 2 weeks of life. As PVR decreases and pulmonary blood flow increases, patients develop signs of congestive heart failure, and these worsen as the ductus begins to close. The lower extremities may also become mottled or gray. There is eventually circulatory collapse and profound shock. PGE$_1$ should be infused to maintain ductal patency. Hyperventilation should be avoided so as to not increase pulmonary blood flow and further worsen systemic perfusion. Echocardiography
is used for diagnosis and can identify the site of the interruption as well as an anomalous right subclavian artery and any associated cardiac defects, including the VSD. DiGeorge syndrome is present about 27% of the time and may be indicated by hypocalcemia.

Repair is accomplished through a median sternotomy, and the arch is reconstructed under deep hypothermic circulatory arrest, using a piece of homograft for augmentation if necessary. The PDA and the VSD are both closed in the same procedure.