ACHD

29

Amanda Green and Jorge Alegria

Simple Defects

Atrial Septal Defect (ASD) A communication between the atria, allowing blood flow between the systemic and pulmonary circulations [1]. These can occur in isolation or as a part of a more complex diagnosis/constellation of defects.

Anatomy and Physiology

Defect in the atrial septum. These can be in the septum primum, secundum, or associated with anomalous pulmonary veins in the case of a sinus venosus defect (superior or inferior location).

The degree of shunting across the ASD is determined by the size of the defect as well as the degree of ventricular compliance. In older adults who have decreased ventricular compliance and a stiffer ventricle, there is a greater risk of transient heart failure after closure of the atrial defect, given that the stiff ventricle now must accept a greater volume load and no longer has the "pop off" of the atrial septum [1, 2]. The development of left ventricular diastolic dysfunction with subsequent increase in left atrial pressure may result in an increase left to right shunt in adults, especially in the presence of hypertension or coronary artery disease.

Types of ASD

Secundum ASD: Located in septum primum, in the region of the fossa ovalis. This is more common in females who make up 65–75% of the patient population with secundum ASD [3]. Depending on the defect size and pulmonary vascular resistance, it can often be closed by transcatheter techniques.

Primum ASD: Also known as endocardial cushion or AV septal defects and are associated with abnormalities of the atrioventricular valves [3]. Anatomically located in the septum secundum, these ASDs usually require surgical repair. In the electrocardiogram a first-degree atrioventricular block and left axis deviation can be found. Left ventricular outflow tract obstruction can be present. Long-term complications in adults may result in need of permanent pacemaker, left sided atrio-ventricular valve replacement.

Sinus Venosus ASD: These are located along the superior or inferior portion of the atrial septum near the junction of the SVC or IVC. They are often associated with partial

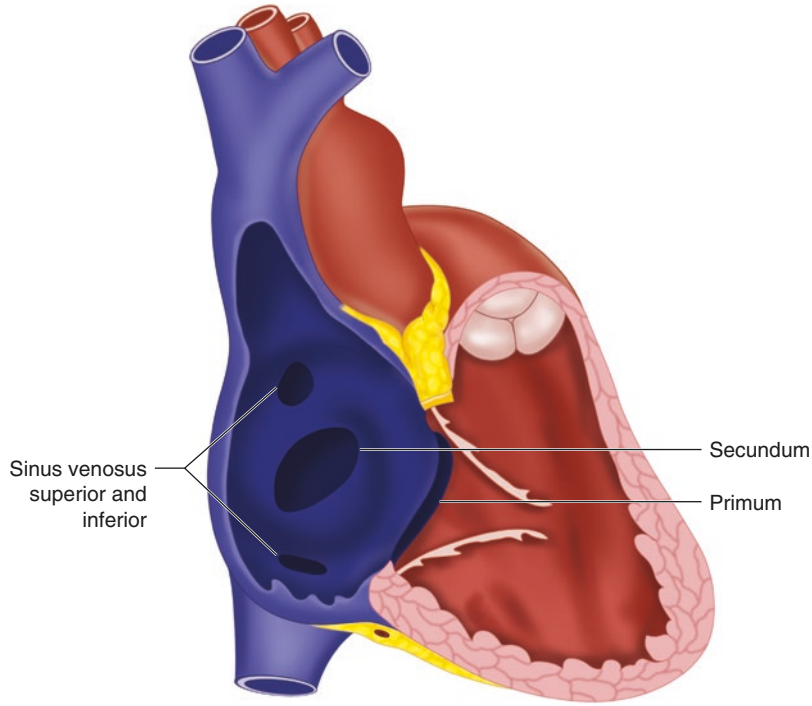
A. Green
Atrium Health/Sanger Heart and Vascular Institute, Greenville, SC, USA
e-mail: amanda.green1@atriumhealth.org

J. Alegria (✉)
Atrium Health/Sanger Heart and Vascular, Congenital Heart Center, Charlotte, NC, USA
e-mail: Jorge.Alegria@atriumhealth.org

R. Musialowski, K. Allshouse (eds.), *Cardiovascular Manual for the Advanced Practice Provider*,
https://doi.org/10.1007/978-3-031-35819-7_29

anomalous pulmonary venous return. Superior sinus venosus is the most common form of the two, and accounts for 5–10% of all ASDs [3]. These commonly requires surgical repair of defect and baffling of the pulmonary veins to the left atrium.

Coronary Sinus Septal Defect: This is a defect in the wall of the coronary sinus also known as an "unroofed coronary sinus," which can result in left to right shunting (LA CS defect RA) [3]. This is also commonly associated with a persistent left superior vena cava.



Physical Exam Correlations

On physical exam, there may or may not be a murmur if the ASD is small with a low degree of shunting. If a higher degree of left to right shunt is present, there may be a soft systolic ejection murmur given the increased blood flow across the pulmonary valve annulus. S2 may have fixed splitting but this finding is not always present [3]. In patients with pulmonary hypertension, P2 may be loud or "snappy" [3].

Adults with undiagnosed ASDs may present with a chief complaint of fatigue, exercise intolerance, and/or palpitations [1]. Paradoxical embolism may also occur.

EKG may demonstrate a right bundle branch block or rSr' pattern in secundum atrial septal defect [1, 3].

Pathology/Description

This defect may be associated with Down Syndrome, Holt Oram syndrome, DiGeorge syndrome, and Ellis Van Creveld syndrome [2]. There is an approximate 10% inheritance risk from a parent with an ASD to their child [2].

Imaging: CMR, Cardiac CT, and/or TEE are useful to evaluate ASD size, shape, rim tissue, and pulmonary venous connections in adults with

ASD. These imaging modalities are helpful in the decision-making regarding ASD closure type and timing [3].

Management

Management of the ASD depends on the type and location of the defect. Secundum ASDs can often be closed with transcatheter techniques and devices, while primum ASDs, coronary sinus septal defects, and sinus venosus ASDs require surgical repair.

The ACC/AHA guidelines recommend that adults with an unrepaired ASD or a repaired ASD with residual shunt have pulse oximetry at rest and during exercise performed. This will help assess for systemic desaturation, which would indicate the presence of right to left shunting across the defect.

Indications for Repair [4]:

- Qp:Qs (pulmonary: systemic blood flow) of >/= 1.5:1 and/or right heart enlargement.
- ASDs should not be closed in adults where the pulmonary arterial systolic pressure is >2/3 of systemic, PVR greater than 2.3 systemic, and/or there is a net right to left shunt.

Long-term surveillance for ASD depends on the type of ASD and repair needed and are specified in the AHA/ACC Guidelines for the Management of Adults with Congenital Heart Disease.

Simple (I): a small, isolated ASD. This includes a native isolated small ASD, or repaired secundum ASD or sinus venosus defect without significant residual shunt or chamber enlargement.

Moderate Complexity (II): Primum ASD, moderate to large unrepaired secundum ASD.

ACHD expertise may improve outcomes in procedures such as diagnostic and interventional cardiology procedures including EP procedures [4].

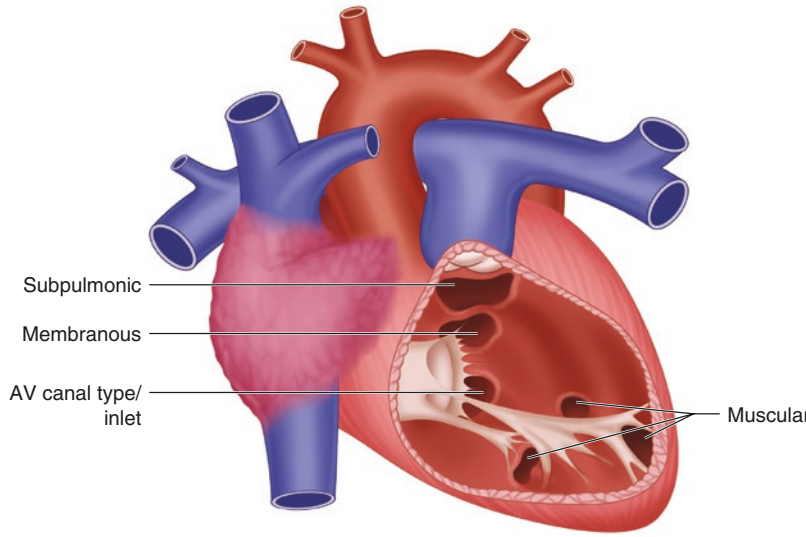
Pearls

- Approximately 34% of adults with unrepaired ASD have complaints of palpitations, which typically is sinus tachycardia [1]. In patients aged 40–60 years, 15% may have atrial flutter [1].
- ASD may have fixed splitting of S2, more common in defects with greater shunting (higher Qp:Qs) [3].
- If pulmonary hypertension is present, may have loud P2 [3].
- RV lift may be felt in the subxiphoid area on deep inspiration [3].
- EKG may show a rSr' pattern in the right precordial leads, and right bundle branch block may also be seen [1, 3].

Ventricular Septal Defect (VSD)

Anatomy and Physiology

A VSD is a hole/communication between the ventricular septum (wall that separates right and left ventricles) that allows for blood communication from the left sided systemic circulation to the right sided circulation or pulmonary circulation. Ventricular septal defects can occur in various parts of the ventricular septum and are classified based on their location [5]. In both children and adults (excluding bicuspid aortic valve in the adult population), VSDs are the most common congenital heart defect [5]. In many cases the VSD may close spontaneously from either muscle occlusion in muscular defects, or closure from aneurysmal tricuspid valve tissue in perimembranous defects. With reduction in size, the burden of shunt is also decreased [5].



Physical Exam Correlations

- Systolic Murmur typically present.
  - Very small Muscular VSD - Murmur can be a short "squeaky" type murmur as the defect may close part way through systole.
  - Muscular VSD: This will be a pansystolic murmur, usually harsh in quality, along the second to third left intercostal spaces. Patient may have an associated thrill, so the provider should always palpate precordium to determine if this is present [6].

    The louder the murmur, typically the more restrictive the defect (greater drop in pressure across the defect = louder murmur).
  - Large VSD: Systolic murmur may not be as loud if there is not a dramatic pressure gradient change across the ventricular septum. If the VSD is large, patient will have a soft murmur, may have fixed splitting of S2 or "snappy" P2 component of S2 given the elevated pulmonary arterial pressures [7].

    If there is significant volume overload from left to right shunting, patient may also have a diastolic rumble.
  - Nonrestrictive VSD: Typically, this is a large VSD with equalization of pressures in both ventricles and associated pulmonary hypertension. If this is long- standing, patients can develop Eisenmenger syndrome which is classified as irreversible pulmonary hypertension with cyanosis. These patients may have a left to right shunting at rest with reversal to right to left shunting with ambulation. Consider ambulatory saturation testing to evaluate for desaturation with activity [7].

Pathology/Description

VSD is a defect located in the ventricular septum which initially creates a volume load on the left heart. Long-standing VSDs can ultimately lead to elevation in pulmonary artery pressures and pulmonary hypertension.

Imaging

VSD can be evaluated by echocardiogram. Modalities such as Cardiac MRI may also be helpful as this will give you detailed anatomic information as well as information regarding the degree of shunting. Cardiac CT can be used to obtain information regarding anatomy (size and location) of the defect [5, 7].

If there is a concern for cyanosis, ambulatory saturation monitoring with a hall walk test is helpful.

Right heart catheterization is helpful to gather information regarding the degree of pulmonary hypertension, PH reversibility testing, as well as degree of shunting (Qp:Qs) [5, 7].

Management

- Most isolated muscular and Perimembranous VSDs close spontaneously.
- If repair is indicated:
- Membranous, Inlet, Subpulmonary, all require surgical repair if hemodynamically significant.
  - Clinically symptomatic
  - Qp:Qs ≥ 1.5:1
  - Chamber enlargement (left heart)
- Muscular VSD can be repaired in the cath lab or surgically depending on anatomy/size.
- Closure of a large VSD with associated pulmonary hypertension may not be recommended if pulmonary vascular bed not responsive or if there is a concern that the pulmonary vascular resistance is too high, although in some cases a "fenestrated" patch or device can decompress the right heart [4].

Pearls

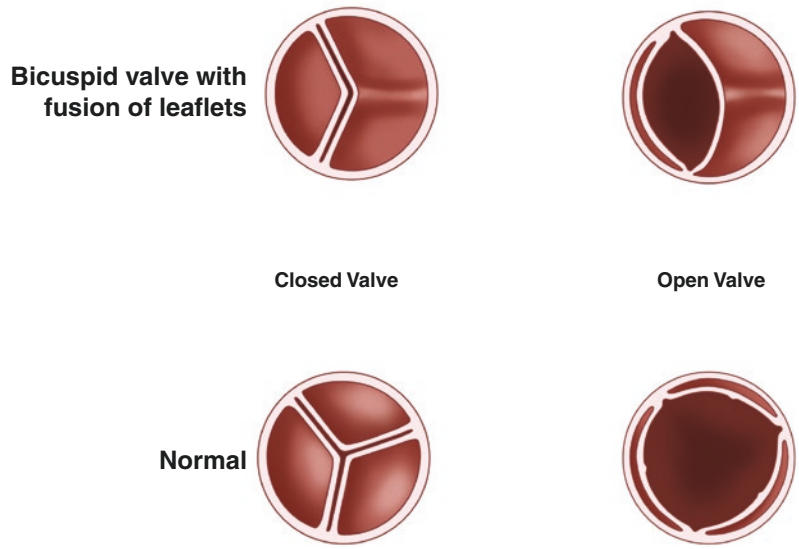
- In patients with unrepaired VSD, there is an increased risk of infective endocarditis. This will typically affect the tricuspid and pulmonary valves [4, 8].
- Survival in Unoperated VSD:
  - Small restrictive defects have a high 25-year survival of 87% [4, 8]. In patients with a small VSD, low Qp:Qs <1.5:1 and low pulmonary vascular resistance had survival rate of 96% [9].
  - Moderate and large defects have a lower 25-year survival of 86 and 61%, respectively [4].
  - Patients with large shunt and Eisenmenger syndrome had even lower 25-year survival at 42% [4].
- Survival in repaired VSD: Significantly improved but remains abnormal [4].
- The majority of small restrictive VSDs located in the membranous or muscular septum can be observed and do not need intervention [4]. For the small number of patients who develop progressive aortic insufficiency related to their perimembranous VSD, surgical repair may be indicated [4, 8].
- Patients with unrepaired VSDs, that are moderately restrictive, may have mild-moderate pulmonary hypertension. Some patients with large nonrestrictive defects may develop Eisenmenger syndrome, and have shunt reversal and systemic desaturation [4, 8].
- Patients with previous closure of their VSD may have patch leak or residual VSD [4].

Bicuspid Aortic Valve (BAV)

Bicuspid AV is the most common congenital heart defect with incidence of 4.6 per 1000 live births. It is 1.5 times more prevalent in males than females. It has a high degree of disease progression leading to aortic stenosis and/or regurgitation, as well as association with aortic root dilation, aortic dissection, and thoracic aortic aneurysm [4, 10].

Anatomy and Physiology

Bicuspid AV is caused by fusion of the aortic valve leaflet commissure creating a two-leaflet valve instead of a three-leaflet valve. This results in a "fish mouth"- appearing opening of the aortic valve rather than a round unobstructed opening. The most common anatomic forms of bicuspid aortic valve include fusion of the right and left coronary commissures or right and non-coronary commissures. Fusion of the right and non-coronary commissure is associated with a more rapid deterioration of the valve leading to stenosis or insufficiency, whereas fusion of the right and left coronary commissure has a greater incidence of association with coarctation of the aorta [10, 11].



Physical Exam Correlations

Patients may have an audible click (systolic ejection noise) [12]. Murmur may only be present if there are other associated diagnoses such as coarctation of the aorta, or valvular disease with obstruction or insufficiency [12]. Providers need to assess for coarctation of the aorta. Coarctation of the aorta and bicuspid valve are commonly associated [10, 11].

Pathology/Description

BAV has aortic valve morphology but with two functional leaflets of unequal size, instead of three, due to incomplete commissural separation during fetal development [3, 5]. There is a familial heritability in an autosomal dominant pattern. In one study, the incidence of asymptomatic bicuspid aortic valve in first-degree relatives is 9% with a 32% incidence of those first-degree relatives having an abnormal aorta [4, 12].

Given the abnormal flow pattern of blood across the aortic valve leaflets in bicuspid aortic valve, there may be calcification or obstruction of the leaflets, which can happen at a higher rate than for those with a tricuspid aortic valve [10]. There may also be incomplete coaptation of the valve leaflets resulting in aortic insufficiency.

Imaging

Evaluation of bicuspid aortic valve is with transthoracic echocardiogram. The valve can be evaluated in more detail with TEE, Cardiac MRI and Cardiac CT to look at valve morphology and evaluate degree of aortic stenosis and regurgitation.

If AS or AI is present, stress testing can also be used to evaluate and risk stratify the patient's need for intervention, either transcatheter or surgical [4, 12].

Management

Surgical and/or transcatheter management of bicuspid valves can be considered in patients with severe obstruction or regurgitation. In the younger population, balloon aortic valvuloplasty in the cath lab is more common; however, in adults, transcatheter valve therapies or surgical valve repair/replacement is more common [12, 13].

Clinical Pearls

- Bicuspid aortic valves can often be asymptomatic but can also present with aortic stenosis or insufficiency earlier in life. One study found that patients with bicuspid aortic valve

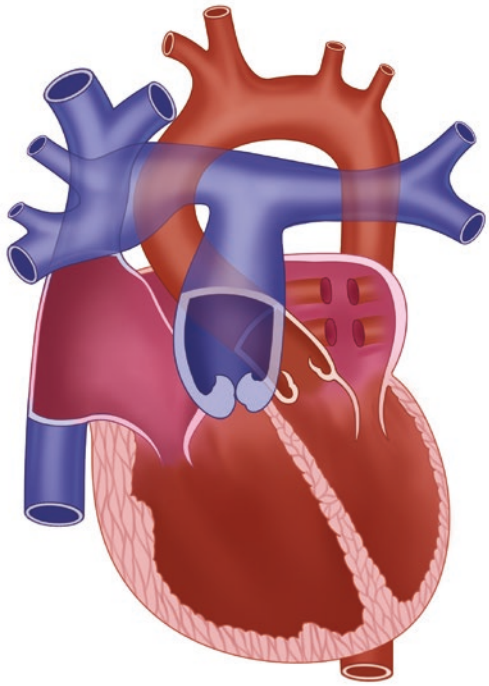
presented at age 40 ± 20 years vs. 67 ± 16 years for patients with tricuspid valve [12].
- If a Bicuspid valve found, assessment for coarctation of the aorta (supine 4 extremity BPs and imaging) and other aortopathies (ascending aorta/aortic root, thoracic aorta) should be undertaken [10–12].
- First-degree relatives of patients with bicuspid valve should have routine screening echocardiograms [4].

Valvar Pulmonary Stenosis (PS)

PS is typically associated with a conical or dome-shaped pulmonary valve or can be a result of thickening of the pulmonary valve leaflets. The narrowing of the opening of the valve between the right ventricle and pulmonary arteries with obstruction at the level of the valve, results in hypertension of the right ventricle [14].

Anatomy and Physiology

Valvar PS occurs in approximately 7% of children born with CHD. PS can be isolated or can occur in combination with other defects, such as in Tetralogy of Fallot [14]. Pulmonary stenosis is associated with Noonan syndrome, Alagille syndrome, Williams syndrome, and congenital rubella [14]. Isolated valvar PS can be associated with a dilated main PA and dysplastic valve leaflets [4]. Valvar PS results in RV hypertrophy and hypertension, which varies based on the degree of obstruction [14].



Valvar pulmonary stenosis with dysplastic /thickened leaflets of the pulmonary valve, and right ventricular hypertrophy, as well as dilation of the main and branch pulmonary arteries which can be secondary to the high velocity jet across the pulmonary valve leaflets.

Physical Exam Correlations

Physical exam findings may include a crescendo-decrescendo systolic murmur which radiates out into the axillae or to the back. The murmur varies in intensity and can have associated thrill [14]. If the patient has pulmonary insufficiency, you may also hear a diastolic murmur or a "to-fro" murmur.

Pathology/Description

Pulmonary valve stenosis is primarily a congenital diagnosis and includes pulmonary valve abnormalities such as uni-commissural, dome-shaped, dysplastic, and bicuspid [4, 14] valves. Pulmonary stenosis can be seen in Noonan, Alagille, and Williams syndromes, and with congenital rubella [4, 14].

Imaging

Echocardiograms are done for routine surveillance of the degree of obstruction and/or insufficiency. Cardiac MRI can be helpful to evaluate RV size and volumes as well as the dimensions of the pulmonary valve and main pulmonary artery if interventions are being considered [4]. EKG is also recommended [4].

Management

Intervention for valvar pulmonary stenosis can often be done by transcatheter techniques, balloon pulmonary valvuloplasty. If failure of balloon pulmonary valvuloplasty occurs or there is progressive insufficiency, surgical intervention may be considered [4].

Clinical Pearls

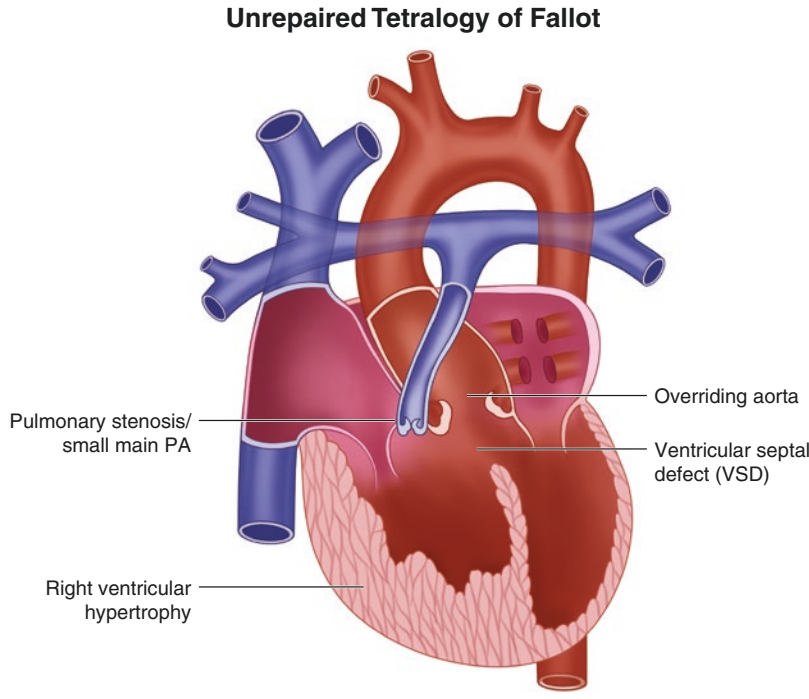
- Adults with the history of pulmonary stenosis require ongoing cardiac follow-up and monitoring for evidence of progressive valve stenosis or regurgitation, RV hypertrophy, heart failure, and arrhythmias [4, 14].
- Patients with PS (mild, moderate, and severe)—usually have a good long-term outcome. Some will require intervention in adulthood either for progressive PS or significant pulmonary insufficiency due to prior intervention (Transcatheter or surgical) [4, 14].

Tetralogy of Fallot (TOF)

The most common cyanotic congenital heart defect is Tetralogy of Fallot (TOF), and it accounts for 7% to 10% of congenital heart defects [15].

Anatomy and Physiology

Tetralogy of Fallot (TOF) is comprised of four defects including a large VSD, overriding aorta, pulmonary stenosis, and RV hypertrophy. Unrepaired TOF can either be "pink" (acyanotic) or "blue" (cyanotic) depending on the degree of pulmonary stenosis and amount of pulmonary blood flow/right to left shunting across the VSD. Repair of TOF is typically done when the patient is an infant, and consists of VSD closure, transannular patch, and resection of RV muscle bundles [4]. This repair typically leaves the patient with little to no PS and free pulmonary insufficiency. In the present day, TOF is repaired in infancy. However, there may be adult patients that have not had complete repair given when/where they were born.



Physical Exam Correlations

In repaired TOF, the physical exam may consist of a to-fro systolic murmur. This is secondary to the movement of blood back and forth across the RVOT (systolic murmur secondary to pulmonary stenosis) and diastolic murmur from the pulmonary insufficiency.

Pathology/Description

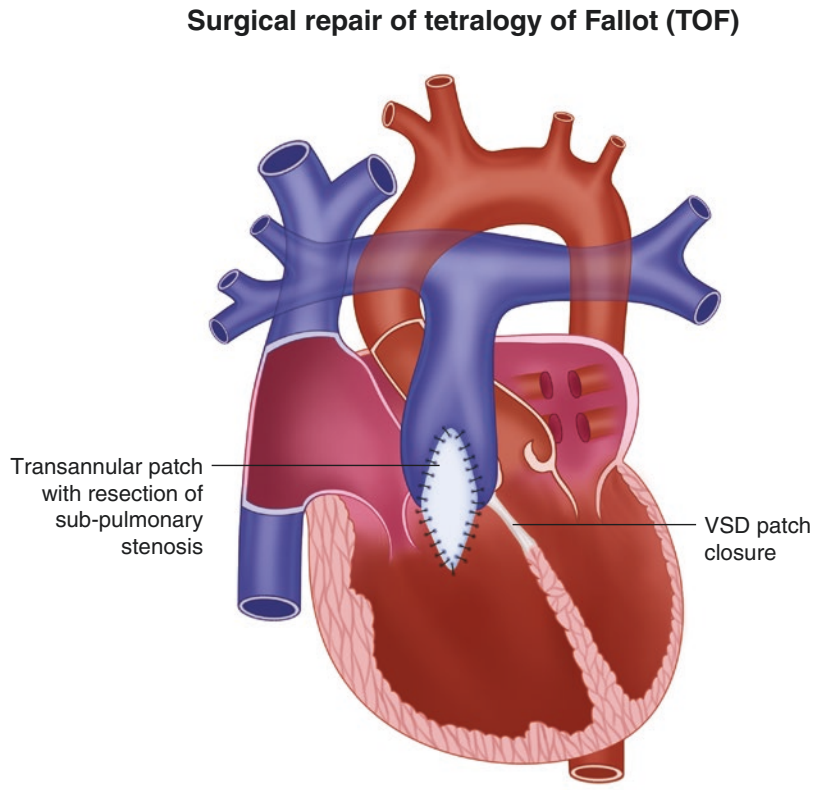
In tetralogy of Fallot, the aorta overrides the ventricular septum, which "crowds out" the pulmonary artery and subpulmonic area, resulting in a small MPA, potentially small branch PA's, valvar, and subvalvar pulmonary stenosis [15]. Adults with tetralogy of Fallot who have undergone complete repair may require pulmonary valve replacement in adulthood given ongoing PS/PI and RV dilation and dysfunction [4].

Imaging

EKG, Echo, Cardiac MRI, and stress testing are recommended at certain intervals. Cardiac MRI is helpful for quantification of RV size and function, especially when considering the timing of pulmonary valve replacement [4].

Management

Repair of tetralogy of Fallot usually consists of resection of RV muscle bundles, and use of a patch to enlarge the main pulmonary artery and branch pulmonary arteries. The VSD is closed with a patch and any residual ASD is closed [4]. Adults with repaired tetralogy of Fallot may often require pulmonary valve replacement. This can either be done via surgical techniques or in the cath lab via transcatheter techniques. There are now FDA approved transcatheter valve therapies for replacement of pulmonary valve in the native RVOT [4].



Clinical Pearls

- Cardiac MRI is the gold standard for evaluation of RV size, function, and valve regurgitation in patients with repaired TOF [4].
- Prior to any intervention, the proximal coronaries and their origins need to be outlined [4].
- Leading causes of mortality in adults with repaired TOF are arrhythmia, heart failure, and complications from reoperations [15].
- In adults with TOF, inducible VT is associated with increased risk of clinical VT or SCD. Programmed ventricular stimulation is useful in risk-stratifying patients who are at moderate risk of SCD, rather than as a routine surveillance tool in low-risk patients [4].
- Bundle Branch Block >180 ms is associated with higher risk of sudden death.
- Syncope in a repaired TOF must be considered VT until proven otherwise.
- Pulmonary hypertension can occur in repaired TOF.

Coarctation of the Aorta

Anatomy and Physiology

Coarctation is a narrowing or stricture in the aorta. This most commonly occurs near the ductal remnant and takeoff of the left subclavian artery (proximal descending aorta) [4, 16]. It is frequently associated with a bicuspid aortic valve and may be seen in Turner's Syndrome. First-degree relatives of patients diagnosed with obstructive left heart disease are at a significantly higher risk (as high as 10%) for coarctation and other left heart lesions [16].

Physical Exam Correlations

Hypertension is common in both repaired and unrepaired patients with coarctation of the aorta [4, 16]. If a patient has recurrent obstruction, four extremity blood pressures, done with the patient laying supine, may reveal decreased BP in the

lower extremities. A systolic murmur may be heard at the left mid-clavicular line and/or posteriorly along the spine. Brachial femoral pulse delay may be present if there is obstruction [4].

Pathology/Description

There are several theories of why coarctation occurs:

1. Ductal tissue extends into the aortic arch and as the ductus closes and becomes a ligament the aorta is also constricted.
2. Flow: poor flow through the aortic arch during fetal development secondary to other left heart lesions resulting in poor growth of the aorta this is often associated with hypoplasia of the aortic arch, not just coarctation.

Post-repair of coarctation, aneurysm formation in the proximal descending aorta at the site of repair can occur. Dissection can also occur, primarily in the setting of uncontrolled hypertension [4]. Ascending aortic aneurysm can occur in those patients with bicuspid aortic valve.

Imaging

Transthoracic echocardiogram, Cardiac CT and MRI can all be helpful for evaluating the structure of the aortic arch and gradient across the coarctation. Upper and lower extremity BPs are also helpful in evaluating gradient (using the right arm for upper extremity measurement). EKG and stress testing are used in evaluation with the EKG evaluating for LVH and stress testing is used to evaluate BP measurements with exercise as well as look for any evidence of ischemia [4].

Management

Surgical repair or stent angioplasty in the cath lab is recommended for adults with hypertension and significant native or recurrent coarctation of the aorta [4, 17]. Hypertension management is critical. Complications of coarctation repair include re-coarctation, pseudoaneurysm, dissection. 11% of patients may require reintervention for restenosis seen by CMR or CTA and supported by physical exam findings [4]. Patients who have undergone surgical patch repair are at a higher risk of developing aneurysms [4, 18].

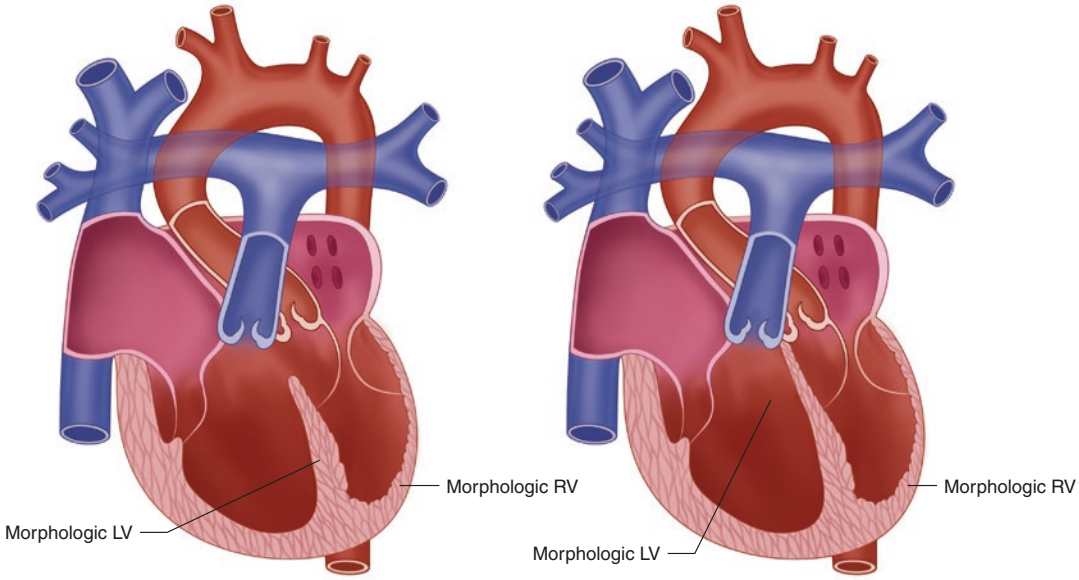
Clinical Pearls

- Recurrent obstruction is defined as an upper extremity to lower extremity resting pressure gradient of >20 mmHg (i.e., right arm Systolic BP of 150 mmHg and left leg systolic BP of 110 mmHg—in this example the peak to peak gradient is 40 mmHg), and/or mean Doppler systolic gradient >20 mmHg [4].
- Recurrent obstruction may be amenable to balloon aortoplasty.
- If LV systolic function is decreased or aortic regurgitation is present, recurrent obstruction is defined as an upper extremity to lower extremity gradient of >10 mmHg or mean Doppler gradient >10 mmHg with collateral flow. (CMR or CT is used to help assess for anatomic evidence of recurrent Coarctation of the aorta) [4].
- Cerebral aneurysm can be present in up to 10% of patients with coarctation of the aorta [4, 19].

L-TGA

Anatomy and Physiology

Congenitally corrected transposition of the great arteries (CCTGA) or L-TGA describes the condition in which the right sided (subpulmonary) ventricle has the shape of the anatomic left ventricle (morphologic LV), and the left sided (sub aortic) ventricle has the shape of the anatomic right ventricle (morphologic RV). This physiology results in the patient having a systemic right ventricle. This defect can occur in isolation or can be associated with other defects such as atrial or ventricular septal defects or pulmonary stenosis. There is also a high incidence of Ebstein-like malformation of the left sided AV-valve, which is the anatomic tricuspid valve. This incidence is felt to be as high as 90%. This malformation can lead to progressive tricuspid (systemic) valve regurgitation [4, 20]. There is a 1% per year or 10% per decade risk of complete heart block [4, 21] with this condition.



L-TGA with VSD **L-TGA with no other defects**

Physical Exam Correlations

Physical exam findings depend on other lesions present or prior surgery/palliation. If no other lesions, physical exam may be normal. Abnormalities in physical exam may show the presence of a systolic murmur in patients with PS and/or VSD.

Pathology/Description

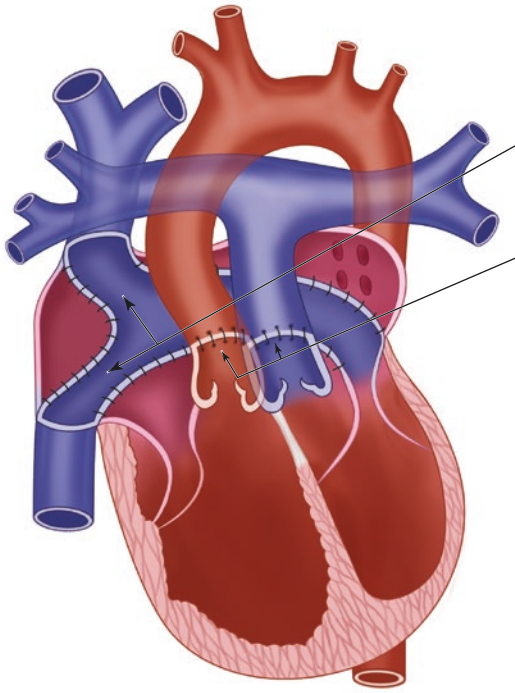
Isolated L-TGA without other defects may present in the third or fourth decade with a new diagnosis of heart failure. More than 1/3 of patients with L-TGA and no other significant defects presents by the fifth decade with significant heart failure (systemic RV) or may present in complete heart block with a 10% per decade risk (i.e., 40% risk by age 40 years) [22, 23]. Patients with L-TGA with other significant associated defects and history of prior heart surgery have a significant risk of systemic ventricular failure, with 2/3 of these patients presenting by age 45 with heart failure [23].

Imaging

Cardiac MRI or Cardiac CT, and echocardiography are all used to evaluate the ventricular function, AV-valve regurgitation, as well as any other intracardiac lesions or anatomic repairs/palliations [4]. Given the likelihood for conduction abnormalities, patients will also need routine EKG monitoring as well as Holter monitoring [4].

Management

Surgical management of patients with L-TGA varies based on the additional defects involved. One operation that can be done for patients with L-TGA with or without VSD is called a "double switch." This operation consists of an atrial switch (Senning or Mustard procedure) to re-route the venous return to the appropriate ventricle, and then arterial switch to switch the great vessels so that they are then aligned with the re-routed systemic venous return to the appropriately shaped ventricle [4, 23]. Progressive systemic atrioventricular valve regurgitation (tricuspid valve) is common and tricuspid valve replacement should be considered early when there is severe tricuspid valve regurgitation to prevent further decline in systemic ventricular dysfunction (right ventricle).



Double Switch Surgical Procedure:
Atrial switch with Mustard/Senning to re-route systemic venous return to the left sided tricuspid valve and morphologic RV; and pulmonary venous return to the mitral valve and morphologic right sided LV
Jatene/Arterial Switch with switch of the great vessels so that they are then aligned with the appropriate venous return and morphologic ventricle.
If VSD present, VSD closure can also be performed during this operation

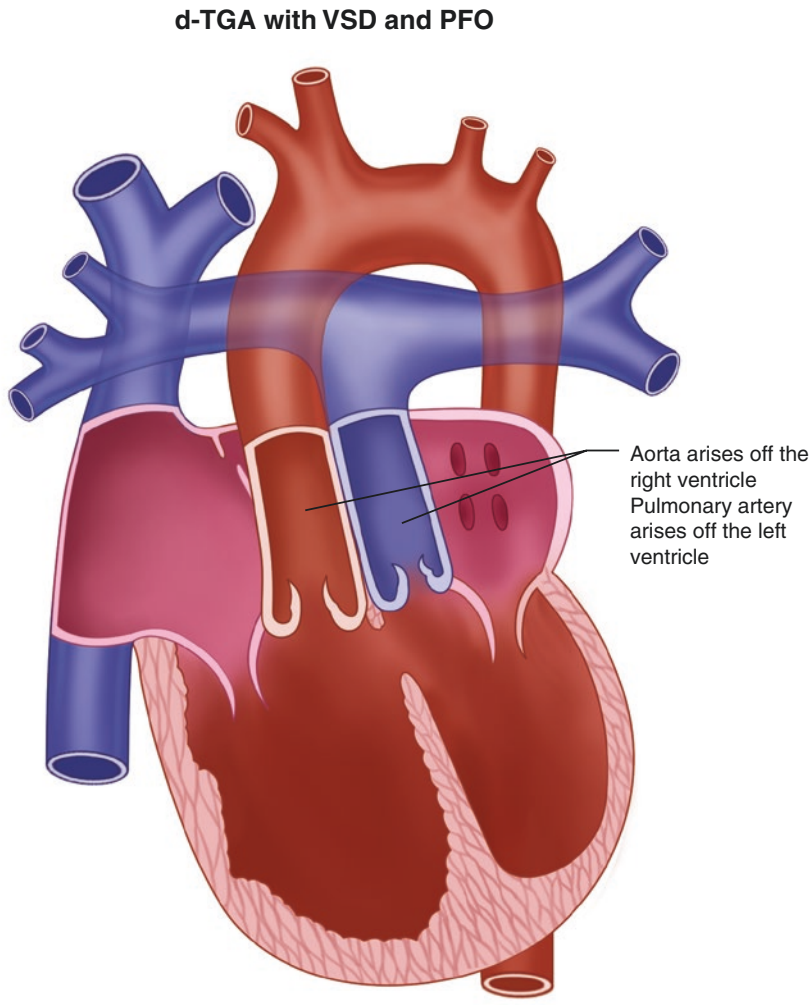
Clinical Pearls

- Disease progression and clinical course is primarily related to the presence and type of other defects and their severity. The presence of other defects also determines the type and complexity of the surgical repair/palliations needed.
- Tricuspid valve replacement in L-TGA should be considered at the earliest sign of RV dysfunction.
- These patients have a high risk of heart block as well as atrial arrhythmias.
- Failure of the systemic ventricle is higher with concomitant tricuspid regurgitation [23].
- May present later in life after asymptomatic period after birth.

D-TGA

Anatomy and Physiology

In d-TGA (dextro-transposition of the great arteries) the pulmonary artery and aorta are "switched," with the pulmonary artery arising from the left ventricle and the aorta arising from the right ventricle. This defect is often accompanied by an atrial septal defect and/or PFO. It may also have associated ventricular septal defect and coronary artery abnormalities [23, 24].



Physical Exam Correlations

A patient with repaired transposition, either with an arterial switch or an atrial switch, may have a normal physical exam. However, exam findings may include a single S2 (A2) (given anterior location of the aorta), and a systolic murmur if there is associated main or branch pulmonary artery stenosis [24]. Other physical exam findings are based on residual defects. In patients with atrial switch, assess ambulatory saturations to assess for baffle leaks and possible right to left shunting and desaturation [23].

Pathology/Description

In unrepaired d-TGA, there are two parallel circuits in which the deoxygenated systemic venous return is recirculated in the systemic circuit and oxygenated pulmonary venous return is recirculated in the pulmonary circuit. These parallel circuits are not compatible with life unless there is mixing, which can occur across an atrial defect, a VSD or PDA [23, 24]. The patent ductus arteriosus (PDA) needs to remain open in the neonatal period, which can be accomplished by an infusion of prostaglan-

din. Additionally, creation of an atrial defect with a balloon atrial septostomy procedure may need to be done if there is inadequate mixing prior to surgical repair [24].

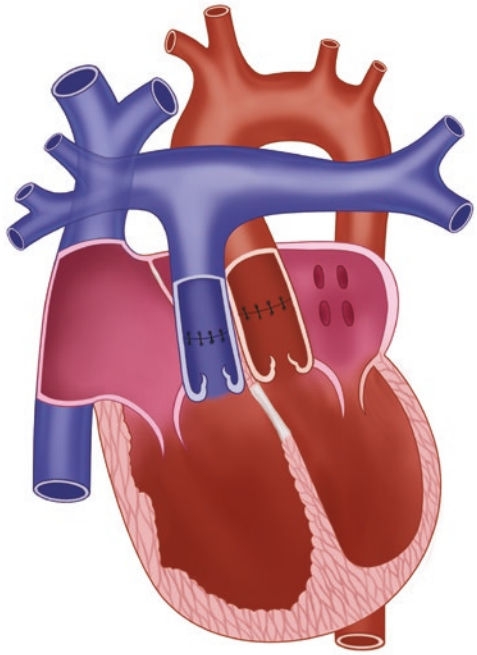
Imaging

Echo, EKG, Cardiac MRI, Cardiac CT, and exercise testing are all used at routine intervals to evaluate patients with both arterial and atrial switch for d-TGA. The timing and type of additional testing such as Holter monitors, pulse oximetry vary between the two repair types [4].

Management

There are two common surgical repairs seen in adults who underwent repair as a child—the Arterial switch (Jatene switch with LeCompte Maneuver) and Atrial switch (also known as a Mustard or Senning operation) [4, 23].

Arterial (Jatene) switch and VSD closure



Present day Surgical Repair – arterial switch (Jatene switch) done around 3-5 days of age.

Jatene switch was developed and 1st done successfully in 1975, gained popularity and widespread use for d-TGA repair in the 1980's
VSD/ASD/PFO closed (if present)
Aorta and pulmonary artery transected above the sinus.
Coronary buttons taken off and re-implanted on the neo-aorta.
Pulmonary arteries are brought anterior to the aorta, and draped over the ascending (LeCompte Maneuver)

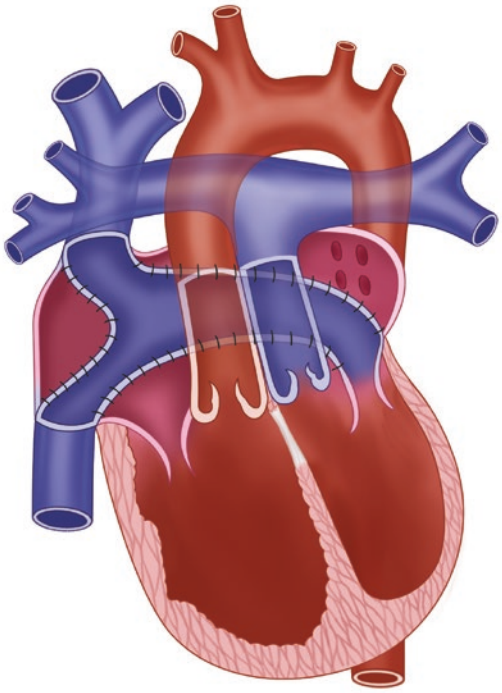
Long Term Consequences of the Arterial Switch

Stenosis at the arterial anastomotic sites, most commonly supravalvular PS
Branch pulmonary artery stenosis
Neoaortic root dilation
Neoaortic valve regurgitation (native pulmonary valve)
Coronary ostial stenosis/occlusion

Clinical Pearls

- Some patients with d-TGA s/p arterial switch who have early problems with pulmonary stenosis undergo RV to PA conduit placement (Rastelli), and can need further surgical revision of this conduit either in an open or transcatheter procedure [4, 24].
- Patients post Le-Compte maneuver as a part of the arterial switch may need branch PA plasty, which is often done in the cardiac cath lab using stent angioplasty technique [4].
- Patients post-arterial switch can develop coronary artery stenosis or occlusion given that the coronary arteries are moved as buttons in the arterial switch procedure [4].
- Patients undergoing arterial switch are at higher risk for neurodevelopmental problems and ADHD [24, 25].

Atrial Switch (Mustard/Senning)



Senning: Developed by Dr. Ake Senning in 1957. Uses a complex reconstruction utilizing flaps from the atrial septum and atrial tissue to create the baffles

Mustard: Developed in 1963 by Dr. William Mustard. Resects the atrial septum and uses pericardial patch to create the baffles

Long Term Consequences of the Atrial Switch (Mustard/Senning)

Baffle leaks
Obstruction of the venous pathways
Arrhythmias
Need for pacemakers/defibrillators
Systolic dysfunction of the systemic right ventricle.

Clinical Pearls

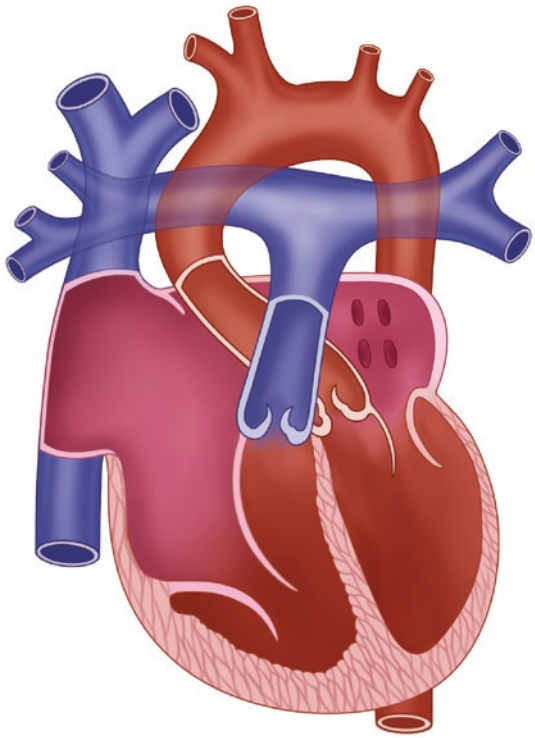
- Patients post-atrial switch can develop leak or obstruction of their systemic venous or pulmonary venous baffles. If systemic SVC venous baffle obstruction occurs, they may present with SVC type syndrome—JVD, prominent veins on upper limbs and chest. Systemic IVC baffle obstruction may present with abdominal distension/ascites, prominent veins on abdomen, lower extremity edema, and no significant upper extremity symptoms. Baffle leak may present with systemic desaturation. Assessment for baffle leak can be done with agitated saline injection but must be done from upper and lower extremities to rule out upper (SVC) and lower (IVC) baffle leaks [4, 26].
- These patients can have a significant burden of atrial arrhythmias given multiple atrial suture lines [4, 26].
- Patients with d-TGA post Atrial switch may present in heart failure given failure of the systemic RV and require transplant evaluation [4].

Ebstein's Anomaly

Anatomy and Physiology

Ebstein's anomaly of the tricuspid valve is an uncommon congenital heart defect occurring in about 0.005% of live births [4]. It is a malformation of the tricuspid valve and right ventricle with varying severity. This defect is classically described as apical displacement of the septal and posterior tricuspid valve leaflets, leading to "atrialization" of the right ventricle [27]. It is associated with an ASD in more than 80% of patients [28]. Ebstein malformation of the tricuspid valve is often found in patients who also have L-TGA (congenitally corrected TGA).

Ebstein Anomaly



Apical displacement of the tricuspid valve leaflets with atrialization of the right ventricle

Physical Exam Correlations

The exam in these patients varies based on severity of the anatomy. Exam may include systolic AV-valve murmur secondary to the tricuspid regurgitation. The patient may have cyanosis secondary to right to left shunting across the atrial septum, if ASD/PFO present.

Pathology/Description

Ebstein's anomaly has apical displacement of the septal and posterior tricuspid valve leaflets. The valve may be tethered/have restricted motion or have a sail like appearance with abnormal chordal attachments, which can contribute to inappropriate coaptation leading to significant regurgitation [27]. The functional right ventricle can be very small and consist only of the RVOT in cases of severe apical displacement [27]. Conduction system abnormalities are common, and as many as 1/3 of patients with Ebstein anomaly have more than one accessory pathway. 5–25% of patients with Ebstein anomaly have Wolff Parkinson White syndrome [28, 29].

Imaging

EKG, Holter, Cardiac MRI, 2D, and 3D echocardiogram including TEE may all be helpful.

Management

Management for Ebstein Anomaly depends on the severity of the lesion. Intervention may include ablation of accessory pathways, or surgical management of these pathways at the time of surgery. Surgical repair can include tricuspid valve repair or replacement, plication of the atrialized right ventricle (Cone procedure), reduction atrioplasty, closure of atrial defect, and arrhythmia management [4]. Poorer outcomes

are associated with delay of surgery until presentation of HF symptoms or RV systolic dysfunction [4].

Pearls

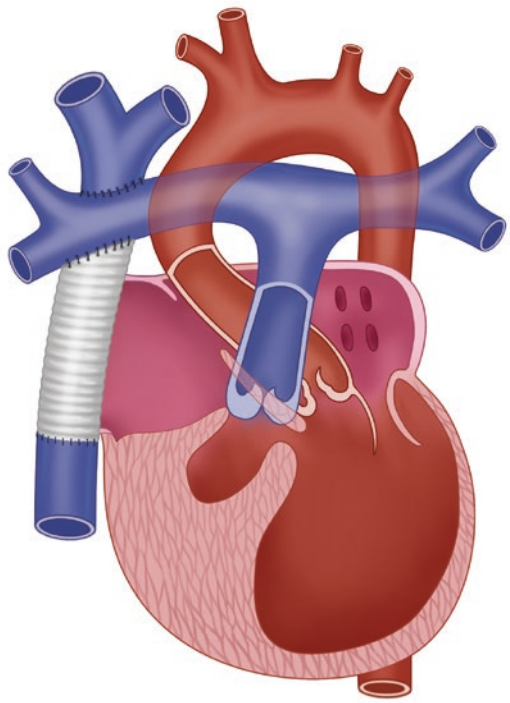
- Over half of the adult patients' initial presenting symptom is palpitations or arrhythmia [29].
- Patients can present with cyanosis, fatigue, dyspnea, arrhythmia, and/or symptoms of right heart failure [27].
- There is an association with accessory pathway tachycardia (WPW).

Single Ventricle/Fontan Physiology

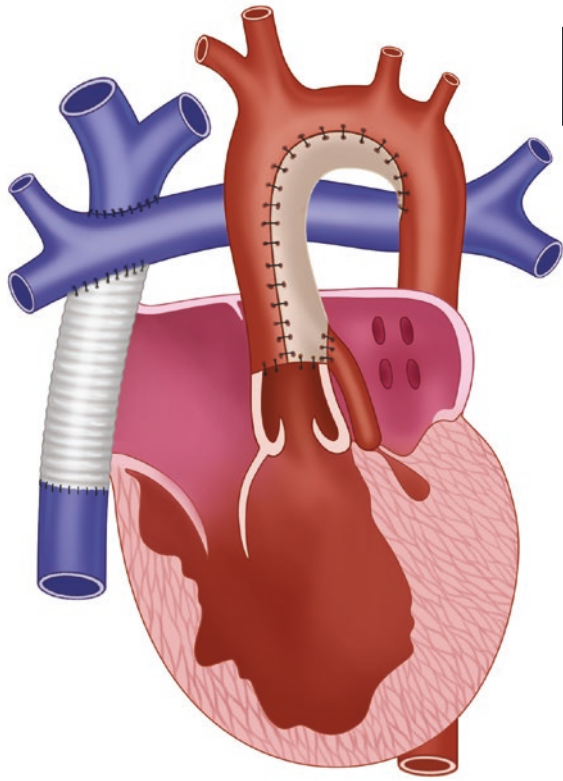
Anatomy and Physiology

The term single ventricle physiology or Fontan physiology refers to complex intracardiac anatomy that is not amenable to 2-ventricule circulation, thus leaving the patient with a "single ventricle" for systemic circulation. This may be either a systemic right ventricle or systemic left ventricle.

1. Defects include, but are not limited to:
   (a) Hypoplastic left heart syndrome (HLHS)
   (b) Double inlet left ventricle (DILV)
   (c) Tricuspid atresia
   (d) Double outlet right ventricle (DORV)
   (e) Pulmonary atresia with intact ventricular septum (PA/IVS)
   (f) Ebstein anomaly
   (g) AV canal (unbalanced)
2. Fontan palliation is typically a three-stage operation over the first few years of life.
   (a) Stage I is typically creation of a shunt—either Blalock-Taussig (BT) or Sano to the pulmonary arteries.
   (b) Stage II is a bidirectional Glenn operation in which the SVC is disarticulated from the RA and sewn directly into the pulmonary arteries. The shunt, which was placed in stage I is taken down.
   (c) Stage III—or Fontan Completion—is the baffling of the IVC return to the pulmonary arteries via a conduit. There are many variations of this with the most common including: Classic Fontan, Lateral Tunnel, and extra-cardiac.



Hypoplastic Right ventricle with Tricuspid atresia, severe pulmonary stenosis, and VSD – s/p Bidirectional Glenn and Extra-cardiac Fontan completion



Hypoplastic Left ventricle with Aortic and mitral atresia – s/p Norwood, Bidirectional Glenn and Extra-cardiac Fontan completion

Physical Exam Correlations

In a well-functioning Fontan, the patient may have normal physical exam findings with normal oxygen saturation. Alternatively, there may be a murmur if there is significant valve disease, recurrent coarctation of the aorta, or a significant burden of aorto-pulmonary collaterals. Oxygen saturation may not be normal and may range between 88% and 92%, in the absence of significant venous abnormalities, depending on the physiology. Prominent abdominal vessels, distended abdomen, or lower extremity edema may be present if the Fontan pressures are elevated or if there is obstruction in the Fontan circuit. If a fenestration is present, the oxygen saturations may drop with ambulation.

Pathology/Description

In the unoperated patient, stage I or II single ventricle physiology, there is mixed blood circulation (mixed systemic venous and systemic arterial blood) through the body. This mixing occurs depending on structural abnormality but is typically across an atrial septal defect (ASD), ventricular septal defect (VSD) or patent ductus arteriosus (PDA). This mixing of the blood is necessary to maintain cardiac output [30]. Over the three palliative surgeries, the circulation is separated such that the venous blood is routed to the lungs by passive/gravitational flow and arterial blood is pumped to the body without mixing, allowing for the patient to have a relatively normal oxygen saturation. Adults with Fontan physi-

ology have chronically low cardiac output given the passive cavo-pulmonary flow of the Fontan circuit. The Fontan circuit is not able to deliver normal amount of volume across the pulmonary vascular bed, which results in reduced ventricular filling and low stroke volume. This physiologic state is not able to augment stroke volume normally with exercise or other states of increased demand [31, 32]. Atrial tachycardias occur in 60% of adults with Fontan palliation. They are poorly tolerated and can be difficult to manage, thus they should be addressed promptly [4]. Sinus node dysfunction occurs in up to 45% of adults with Fontan palliation [4].

Imaging

EKG, transthoracic, Cardiac MRI, Cardiac CT, and stress testing are all used to evaluate the patient with a Fontan. Holter monitoring for arrhythmias is also used for evaluation.

Clinical Pearls

- Cardiac output for patients with Fontan physiology is not normal. They are limited by the passive flow to the lungs and the abnormal blood throughput in the pulmonary circuit. Over time the chronic volume depletion causes progressive decline in ventricular function, resulting in a cycle of increased end-diastolic pressure, systemic venous congestion, and low cardiac output [31, 32].
- Failing Fontan physiology:
  - Cyanosis: Patients with Fontan physiology are often mildly hypoxemic. This is caused by the presence of a surgically created fenestration or leaks in the fontan baffle itself, coronary sinus venous return to the atrium, pulmonary AV-malformations, and veno-venous collaterals which can drain into the pulmonary veins or directly into the left atrium [31].
  - Protein losing enteropathy (PLE) occurs in 5–15% of patients with Fontan physiology. This refers to the loss of serum proteins into the lumen of the gut leading to chronic diarrhea, abdominal discomfort, and peripheral edema. Lab values indicative of PLE are decreased serum albumin <3.5 g/dL and Total protein <6.0–6.3 g/dL, as well as augmented enteric protein loss with Fecal alpha-1-antitrypsin clearance >56 ml/24H (with diarrhea) and >27 ml/24 h (without diarrhea) [31].
    Management: decrease resistance in the Fontan circuit by alleviating any obstruction, reducing PVR, unfractionated heparin or budesonide to reduce enteric inflammation. Other medications that can help improve endothelial cell function and reduce inflammation such as Spironolactone and Octreotide (limited evidence) [31, 33].
  - Hepatic dysfunction: Elevated central venous pressure and systemic hypoperfusion lead to congestive hepatopathy, liver fibrosis to cirrhosis and even hepatocellular carcinoma [31].
  - Screening for hepatocellular carcinoma should be done with Alpha fetoprotein (AFP) and liver imaging (Ultrasound, liver MRI with Evoist or CT).
  - Thromboembolic complications: Risk of thromboembolism as high as 20% in patients with Fontan physiology. This is felt to be caused by lack of pulsatile flow in the pulmonary circuit and ensuing venous stasis leading to a hypercoagulable state which is made worse by deficiency of protein C, S, antithrombin III and increased platelet reactivity [31, 34].

Infective Endocarditis Prophylaxis

It should be discussed in detail with the patient the importance of infective endocarditis prevention. Infective endocarditis can be a potentially life-threatening condition with 10–20% mortality and prevention along antibiotic prophylaxis when indicated and early diagnosis is of paramount relevance. Recommendations of proper dental hygiene including teeth brushing three times a day, flossing once a day, visiting the dentist twice a year and following infective endocarditis prophylaxis guidelines with antibi-

otics when indicated. Also, for other procedures, infective endocarditis prophylaxis may need to be considered. Patients need to receive education regarding early symptoms of infective endocarditis that may include fever, myalgias, headache, arthralgias, or other symptoms. The condition could be confused with flu or COVID. Also, a low threshold for blood cultures in the absence of a clear diagnosis with fever and contacting the ACHD Care Center in collaboration with primary care providers to facilitate diagnostic and care pathways as needed. AHA infective endocarditis prophylaxis card must be given.

(Please see Chap. 19 for more details.)

References

1. Le Gloan L, Legendre A, Iserin L, Ladouceur M. Pathophysiology and natural history of atrial septal defect. J Thorac Dis. 2018;10(Suppl 24):S2854–63. https://doi.org/10.21037/jtd.2018.02.80. PMID: 30305945; PMCID: PMC6174151.
2. Redfield MM, Jacobsen SJ, Borlaug BA, et al. Age- and gender-related ventricular-vascular stiffening: a community-based study. Circulation. 2005;112:2254–62. https://doi.org/10.1161/CIRCULATIONAHA.105.541078.
3. Webb G, Gatzoulis MA. Atrial septal defects in the adult: recent progress and overview. Circulation. 2006;114(15):1645–53. https://doi.org/10.1161/CIRCULATIONAHA.105.592055. PMID: 17030704.
4. Stout KK, Daniels CJ, Aboulhosn JA, Bozkurt B, Broberg CS, Colman JM, Crumb SR, Dearani JA, Fuller S, Gurvitz M, Khairy P, Landzberg MJ, Saidi A, Valente AM, Van Hare GF. 2018 AHA/ACC guideline for the management of adults with congenital heart disease: a report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. J Am Coll Cardiol. 2019;73(12):e81–e192. https://doi.org/10.1016/j.jacc.2018.08.1029. Epub 2018 Aug 16. Erratum in: J Am Coll Cardiol. 2019 May 14;73(18):2361–2362. PMID: 30121239.
5. Minette MS, Sahn DJ. Ventricular septal defects. Circulation. 2006;114(20):2190–7. https://doi.org/10.1161/CIRCULATIONAHA.106.618124. Erratum in: Circulation 2007 Feb 20;115(7):e205. PMID: 17101870.
6. Tretter JL, Benson L, Crucean A, Spicer DE, Anderson R. Ventricular septal defect. In: Wernovsky G, Anderson RH, Kumar K, Mussatto KA, Redington AN, Tweddell JS, editors. Anderson's pediatric cardiology. 4th ed. Phillidelphia, PA: Elsivier; 2020. p. 557–84.
7. Dakkak W, Oliver TI. Ventricular septal defect. [Updated 2022 May 10]. In: StatPearls [Internet]. Treasure Island, FL: StatPearls Publishing; 2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK470330/.
8. Corone P, Doyon F, Gaudeau S, et al. Natural history of ventricular septal defect. A study involving 790 cases. Circulation. 1977;55:908–15.
9. Mattila S, Kostiainen S, Kyllönen KE, et al. Repair of ventricular septal defect in adults. Scand J Thorac Cardiovasc Surg. 1985;19:29–31.
10. Mathieu P, Bossé Y, Huggins GS, Della Corte A, Pibarot P, Michelena HI, Limongelli G, Boulanger MC, Evangelista A, Bédard E, Citro R, Body SC, Nemer M, Schoen FJ. The pathology and pathobiology of bicuspid aortic valve: state of the art and novel research perspectives. J Pathol Clin Res. 2015;1(4):195–206. https://doi.org/10.1002/cjp2.21. PMID: 27499904; PMCID: PMC4939890.
11. Fernandes SM, Sanders SP, Khairy P, Jenkins KJ, et al. Morphology of bicuspid aortic valve in children and adolescents. J Am Coll Cardiol. 2004;44(8):1648–51. https://doi.org/10.1016/j.jacc.2004.05.063.
12. Siu SC, Silversides CK. Bicuspid aortic valve disease. J Am Coll Cardiol. 2010;55(25):2789–800. https://doi.org/10.1016/j.jacc.2009.12.068. PMID: 20579534.
13. Vincent F, Ternacle J, Denimal T, Shen M, Redfors B, Delhaye C, Simonato M, Debry N, Verdier B, Shahim B, Pamart T, Spillemaeker H, Schurtz G, Pontana F, Thourani VH, Pibarot P, Van Belle E. Transcatheter aortic valve replacement in bicuspid aortic valve stenosis. Circulation. 2021;143(10):1043–61. https://doi.org/10.1161/CIRCULATIONAHA.120.048048. Epub 2021 Mar 8. PMID: 33683945.
14. Ruckdeschel E, Kim YY. Pulmonary valve stenosis in the adult patient: pathophysiology, diagnosis and management. Heart. 2019;105:414–22.
15. Diaz-Frias J, Guillaume M. Tetralogy of Fallot. [Updated 2022 Jan 18]. In: StatPearls [Internet]. Treasure Island, FL: StatPearls Publishing; 2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK513288/.
16. Law MA, Tivakaran VS. Coarctation of the aorta. [Updated 2022 May 15]. In: StatPearls [Internet]. Treasure Island, FL: StatPearls Publishing; 2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK430913/.
17. Qureshi AM, McElhinney DB, Lock JE, et al. Acute and intermediate outcomes, and evaluation of injury to the aortic wall, as based on 15 years experience of implanting stents to treat aortic coarctation. Cardiol Young. 2007;17:307–18.
18. Hager A, Kanz S, Kaemmerer H, et al. Coarctation long-term assessment (COALA): significance of arterial hypertension in a cohort of 404 patients up to 27 years after surgical repair of isolated coarctation of the aorta, even in the absence of restenosis and prosthetic material. J Thorac Cardiovasc Surg. 2007;134:738–45.

19. Wiebers DO, Whisnant JP, Huston J, et al. Unruptured intracranial aneurysms: natural history, clinical outcome, and risks of surgical and endovascular treatment. Lancet. 2003;362:103–10.
20. Prieto LR, Hordof AJ, Secic M, et al. Progressive tricuspid valve disease in patients with congenitally corrected transposition of the great arteries. Circulation. 1998;98:997–1005.
21. Huhta JC, Maloney JD, Ritter DG, et al. Complete atrioventricular block in patients with atrioventricular discordance. Circulation. 1983;67:1374–7.
22. Graham TP Jr, Bernard YD, Mellen BG, Celermajer D, Baumgartner H, Cetta F, Connolly HM, Davidson WR, Dellborg M, Foster E, Gersony WM, Gessner IH, Hurwitz RA, Kaemmerer H, Kugler JD, Murphy DJ, Noonan JA, Morris C, Perloff JK, Sanders SP, Sutherland JL. Long-term outcome in congenitally corrected transposition of the great arteries: a multi-institutional study. J Am Coll Cardiol. 2000;36:255–61.
23. Warnes CA. Transposition of the great arteries. Circulation. 2006;114(24):2699–709. https://doi.org/10.1161/CIRCULATIONAHA.105.592352. PMID: 17159076.
24. Szymanski MW, Moore SM, Kritzmire SM, et al. Transposition of the great arteries. [Updated 2022 May 11]. In: StatPearls [Internet]. Treasure Island, FL: StatPearls Publishing; 2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK538434/.
25. Marino BS, Lipkin PH, Newburger JW, Peacock G, Gerdes M, Gaynor JW, Mussatto KA, Uzark K, Goldberg CS, Johnson WH, Li J, Smith SE, Bellinger DC, Mahle WT, American Heart Association Congenital Heart Defects Committee, Council on Cardiovascular Disease in the Young, Council on Cardiovascular Nursing, and Stroke Council. Neurodevelopmental outcomes in children with congenital heart disease: evaluation and management: a scientific statement from the American Heart Association. Circulation. 2012;126(9):1143–72.
26. Broberg CS. Cardiac magnetic imaging of the patient with an atrial switch palliation for transposition of the great arteries. Prog Pediatr Cardiol. 2014;38:49–55.
27. Brown ML, Dearani JA, Danielson GK, Cetta F, Connolly HM, Warnes CA, Li Z, Hodge DO, Driscoll DJ, Mayo Clinic Congenital Heart Center. The outcomes of operations for 539 patients with Ebstein anomaly. J Thorac Cardiovasc Surg. 2008;135(5):1120–36, 1136.e1–7.
28. Khositseth A, Danielson GK, Dearani JA, Munger TM, Porter CJ. Supraventricular tachyarrhythmias in Ebstein anomaly: management and outcome. J Thorac Cardiovasc Surg. 2004;128(6):826–33.
29. Fuchs MM, Connolly HM. Ebstein anomaly in the adult patient. Cardiol Clin. 2020;38(3):353–63.
30. Heaton J, Heller D. Single ventricle. [Updated 2022 May 1]. In: StatPearls [Internet]. Treasure Island, FL: StatPearls Publishing; 2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK557789/.
31. Mazza GA, Gribaudo E, Agnoletti G. The pathophysiology and complications of Fontan circulation. Acta Biomed. 2021;92(5):e2021260. https://doi.org/10.23750/abm.v92i5.10893. PMID: 34738582; PMCID: PMC8689331.
32. Gewillig M, Brown SC, Eyskens B, Heying R, et al. The Fontan circulation: who controls cardiac output? Interact Cardiovasc Thorac Surg. 2010;10:428–33.
33. John AS, Johnson JA, Khan M, Driscoll DJ, Warnes CA, Cetta F. Clinical outcomes and improved survival in patients with protein-losing enteropathy after the Fontan operation. J Am Coll Cardiol. 2014;64:54–62.
34. Monagle P, Karl TR. Thromboembolic problems after the Fontan operation. Semin Thorac Cardiovasc Surg Pediatr Card Surg Annu. 2002;5:36–47.