Acyanotic Congenital Heart Lesions

Developed by Herman Bami and Dr. Michael Grattan for PedsCases.com
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Part 1:

My name is Herman Bami and I'm a second year medical student from Schulich School of Medicine and Dentistry at the University of Western Ontario. This podcast was supervised by Dr. Michael Grattan, a pediatric cardiologist at the London Health Sciences Center. This is the first in a 2-part podcast series on acyanotic heart lesions. The objectives of the first podcast are to:

1. Define congenital heart disease and classify it as cyanotic vs. acyanotic
2. Describe the pathophysiology of left-to-right shunts
3. Review the pathophysiology, clinical presentation and management of a ventricular septal defect, atrial septal defect, patent ductus arteriosus and atrio-ventricular septal defect.

In the second podcast, we will discuss a second category of acyanotic heart lesions: outflow tract obstructions.

But before we begin, let's start with a case. Joey is a 2 year-old male who presents for assessment of a murmur. While he seems otherwise healthy, with no obvious shortness of breath or exercise intolerance, he was noted to have a hyperactive precordium with a soft systolic ejection murmur at the left sternal border. Upon further inspection and careful auscultation, a fixed and widely split second heart sound is also noted. We'll return to this case towards the end of the podcast, and further discuss the relevant diagnostic and therapeutic modalities.

Classification

Congenital heart disease, excluding bicuspid aortic valve, occurs in approximately 8 per 1000 births and has a broad range of clinical manifestations[1]. Generally, congenital heart disease is divided into cyanotic or acyanotic heart disease, a classification which is based on the level of hemoglobin saturation in the systemic circulation. Additionally, congenital heart defects can be further divided into three main pathophysiologic groups: left-to-right

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shunts, right-to-left shunts and outflow tract obstructions[2]. Acyanotic congenital heart lesions include both left-to-right shunts and outflow tract obstructions. Left-to-right shunts cause an increase in pulmonary circulation and include: ventricular septal defects (VSDs), atrial septal defects (ASDs), patent ductus arteriosus (PDAs), and atrioventricular septal defects (AVSDs). Increased pulmonary circulation can cause pulmonary edema, and increased flow through different chambers of the heart can lead to dilation and hypertrophy of the heart resulting in congestive heart failure. On the other hand, outflow tract obstructions will usually result in normal pulmonary blood flow and include: aortic stenosis (AS), pulmonary stenosis (PS), and coarctation of the aorta.

We're now going to explore both groups in further detail covering briefly their etiology, clinical manifestations, and relevant diagnostic and therapeutic modalities. Let's start by examining the specific pathologies under the branch of left-to-right shunts.

Ventricular Septal Defect

Ventricular septal defects (VSDs) are the most common congenital heart defect and comprise up to 20% of all cardiac anomalies[2-3]. While a VSD is a common isolated malformation, it can also be a component in several other cardiac defects that extend beyond the scope of this podcast. The ventricular septum is a complex structure and includes four components: the muscular, posterior or inlet, supracristal and membranous septa. VSDs occur when one of the ventricular septum's four components fail to develop normally. Although there are many different classification systems for VSDs, they can broadly be discriminated into two groups based on their location. The first type is muscular VSDs, which are found within the muscular septum. The second type is perimembranous VSDs that are found at the margins of the muscular septum, just below the aortic valve.

While the location of the VSD plays an important role in prognostication and treatment, the amount of flow through the VSD depends on the size of the defect and pulmonary vascular resistance (PVR). The size of the VSD also greatly effects the clinical presentation. For instance, small VSDs with little shunt can create very turbulent flow and present with a loud murmur but often be asymptomatic. Moderate to large VSDs can actually have a quieter murmur, as blood can more easily flow through a larger defect without as much turbulence. Larger defects can cause pulmonary hypertension and congestive heart failure, and may present with fatigue, diaphoresis and respiratory distress with feedings, and failure to thrive. The typical clinical finding with a VSD would be a pansystolic murmur, usually loudest at the lower left sternal border. This murmur is secondary to left-to-right flow from the high-pressure left ventricle into the low-pressure right ventricle. Larger VSDs are associated with higher right ventricular pressures, and do not cause pansystolic murmurs. Instead, these larger defects cause systolic ejection murmurs related to increased blood flow across the pulmonary valve. Increased flow across the mitral valve due to a large shunt may also cause a mid-diastolic rumble at the apex. Flow through the left-to-right shunt in a VSD occurs during systole when both ventricles are contracted. The contracted right ventricle acts as a conduit for blood, and an increased volume is seen in the lungs and left heart. Over time, this leads to dilation of the left heart and left ventricular hypertrophy. A long-standing untreated VSD can also result in pulmonary hypertension

leading to right ventricular hypertrophy. Significant shunts can also result in signs of congestive heart failure including a hyperdynamic precordium, hepatomegaly and increased work of breathing.

Electrocardiogram (ECG) and chest X-ray findings depend on the size of the defect. For instance, small VSDs will often present with normal studies. Larger VSDs however can cause volume overload to the left side of the heart, with ECG findings of left atrial enlargement and left ventricular hypertrophy. A chest X-ray can also be used to show cardiomegaly, left ventricular enlargement and increased pulmonary blood flow. Right ventricular enlargement can also occur due to pulmonary hypertension and increased pulmonary vascular resistance. The mainstay of diagnosis however resides in echocardiography. Echocardiography can be used to provide information such as the size and location of the defect, as well as any valvular deficiencies or associated pathologies. In terms of treatment, small VSDs usually close spontaneously and even in the absence of spontaneous closure, surgical closure of small VSDs may not be necessary. Moderate to large VSDs on the other hand may require treatment with diuretics, afterload reduction, and supplemental calories. However, the continued presence of symptoms, such as continued poor growth and pulmonary hypertension, necessitate closure. This is usually performed surgically, though some VSDs can be closed using devices placed with cardiac catheterization.

Atrial Septal Defect

Atrial septal defects (ASDs) are another type of left-to-right shunt and represent approximately 15-20% of all congenital heart defects[1-2,4]. During normal embryonic development, a septum grows towards the endocardial cushions to divide the atria; ASDs result from a failure of septal growth or excessive tissue reabsorption. The most common type of ASD is a secundum defect, which is a hole in the region of the foramen ovale. A primum ASD is located near the endocardial cushions, inferior to the true atrial septum, and is included in the spectrum of atrioventricular septal defects, which will be discussed later. The least common ASDs are sinus venosus defects and coronary sinus defects. These are not true defects in the atrial septum, but nonetheless result in shunting from the left atrium to the right atrium. The sinus venosus defect occurs due to an unroofed right pulmonary vein as it passes behind the superior vena cava. This defect always involves anomalous pulmonary venous drainage from at least one right pulmonary vein to the superior vena cava. The coronary sinus defect occurs due to unroofing of the coronary sinus as it passes posterior to the left atrium.

The pathophysiology and amount of shunting depend significantly on the size of the ASD and the compliance of the ventricles. The left-to-right shunt of an ASD shunts blood into a relaxed right heart in ventricular diastole. This leads to eventual dilatation of the right heart. Even large ASDs with significant shunts are rarely symptomatic in childhood, although significant symptoms can develop in older adults including intractable arrhythmias, heart failure and more rarely, pulmonary hypertension. Notable clinical signs include a prominent right ventricular impulse at the left lower sternal border as well as a systolic ejection murmur in the area of the right ventricular outflow tract. Another possible

manifestation includes a fixed, widely-split S_2 resulting from right ventricular overload and prolonged ejection into the pulmonary circulation.

ECG and chest X-ray findings are a result of the increased volume in the right side of the heart and pulmonary circulation. The ECG may show right axis deviation as well as right ventricular enlargement and incomplete right bundle branch block. Additionally, certain ECG changes may indicate a specific type of ASD, although this is beyond the scope of this module. A chest x-ray may illustrate cardiomegaly as well as right heart enlargement and a prominent pulmonary artery. Echocardiography can also be utilized to indicate the type and size of the ASD, as well as the level of flow across the defect. While medical management is rarely initiated, closure is recommended if a significant shunt is still present around 3-5 years of age. Secundum ASDs can often be closed with devices placed through cardiac catheterization; however, primum, sinus venosus and coronary sinus defects require surgical closure.

Patent Ductus Arteriosus

Patent ductus arteriosus (PDA) is a third type of left-to-right shunt and represents approximately 10 to 15% of congenital heart disease[1-2,5]. It has a significantly higher prevalence in premature neonates. The ductus arteriosus is a normal fetal structure that allows blood to flow from the pulmonary artery to the aorta. Normally this structure spontaneously closes within 24-74 hours, however failure of this vessel to close results in a patent ductus arteriosus. A PDA allows left-to-right shunting from the high-pressure aorta to the lower pressure pulmonary artery once the pulmonary vascular resistance decreases after birth.

The size of the PDA as well as the pulmonary vascular resistance determine the amount of shunting that occurs, which in turn is reflected in the symptom profile. Small PDAs are often asymptomatic but larger shunts can cause congestive heart failure symptoms as the pulmonary vascular resistance falls. These symptoms include failure to thrive, increased work of breathing, recurrent upper respiratory tract infections and fatigue with exertion. In premature infants, significant PDAs may lead to difficulty in weaning ventilation. Clinically, a widened pulse pressure and a continuous, machine-like murmur at the left infraclavicular area can be detected. The murmur normally radiates along the pulmonary arteries and can be heard over the left side of the back. The murmur may only be audible in systole, or may be absent altogether in very small PDAs or in large PDAs with associated pulmonary hypertension. Significant shunting can cause increased flow across the mitral valve resulting in a mid-diastolic murmur at the apex, although this may be difficult to auscultate in the presence of a continuous murmur. Significant shunts can result in signs of congestive heart failure including a hyperdynamic precordium, hepatomegaly and increased work of breathing.

While imaging findings are normal with small PDAs, larger shunts can cause a full pulmonary artery silhouette and increased pulmonary vascularity to be visible on a chest radiograph. Additionally, potential ECG findings include evidence of left ventricular hypertrophy and left atrial enlargement and, if pulmonary hypertension is present, right

ventricular hypertrophy as well. Transthoracic echocardiography is often the diagnostic method that can best determine ductal anatomy and characterize level of flow. Spontaneous closure of a PDA is uncommon after a few weeks of age. In premature neonates with moderate to large PDAs, short courses of NSAIDs or acetaminophen can augment physiologic PDA closure through the inhibition of endogenous prostaglandins. In older children, elective closure of small, hemodynamically insignificant PDAs is controversial. Moderate to large PDAs are often first managed with diuretics but eventually require closure, which can often be done in the catheterization laboratory.

Atrioventricular Septal Defect

The fourth and last type of left-to-right shunt that we'll be covering is atrioventricular septal defects, also known as endocardial cushion defects[2,6-7]. These lesions result from failure of the septum to fuse with the endocardial cushion. The failure of the septum to properly fuse also causes abnormal atrioventricular valve development. A complete atrioventricular septal defect involves: a primum ASD, inlet VSD, common atrioventricular valve and a tri-leaflet or cleft left atrioventricular valve. Thus, in addition to shunting, there may be atrioventricular valve insufficiency. There is a strong association between AVSD and trisomy 21 or Down's syndrome. AVSD is the most common congenital heart defect in infants with Down syndrome and 40-50% of infants with AVSD identified prenatally will go on to have Down syndrome.

As the pulmonary vascular resistance falls over the first few months of life, congestive heart failure symptoms such as increased work of breathing, fatigue and failure to thrive can develop. This is mainly due to the VSD, although atrioventricular valve insufficiency can exacerbate these symptoms. The presence of other signs, such as murmurs, depends on the level and extent of the shunting in both the atrial and ventricular defects.

Diagnosis of atrioventricular defects is normally made with echocardiography, which can also be used to better identify the appropriate therapeutic intervention. Furthermore, chest radiographs can be used to indicate cardiomegaly and increased vascularity. A left superior axis as well as ventricular hypertrophy may be detectable by ECG. In terms of treatment, diuretics, afterload reduction and supplemental calories are often used to reduce congestive heart failure symptoms. Ultimately though, surgical repair of the defect must be employed.

Conclusion

Now that we've gone through the different types of acyanotic congenital heart disease presenting with left-to-right shunts, let's return back to the case. Joey is a 2 year-old male presenting with a systolic ejection murmur heard best at the left sternal border. With further inspection, a hyperactive precordium as well as a fixed and widely split second heart sound were noted. While systolic ejection murmurs are quite common in healthy children, the fixed splitting of S2 offers some cause for concern, especially of a possible atrial septal defect. To further investigate the cause of the murmur, ECG, chest X-ray and echocardiography are conducted. The ECG showed some evidence of right axis deviation

consistent with right atrial enlargement. While the chest X-ray is inconclusive, diagnosis of a secundum atrial septal defect with RV dilatation is confirmed through echocardiography. Based on the symptoms and clinical findings, it is determined that elective closure will eventually be required. However, this patient will be followed until he is large enough for device closure via cardiac catheterization, likely around four to five years of age.

That's the end of our first part of the podcast series on acyanotic congenital heart lesions. The second part of this series will be focusing on outflow tract obstructions. Before we continue on though, let's summarize the key points from this podcast:

- Congenital heart disease is a term encompassing many pathologies and can be broadly classified into three main groups: left-to-right shunts, right-to-left shunts and outflow tract obstructions.
- Acyanotic congenital heart disease includes left-to-right shunts and outflow tract obstructions.
- Left-to-right shunts include: atrial septal defects, ventricular septal defects, patent ductus arteriosus and atrioventricular septal defects.
- The clinical presentation of these defects varies greatly based on the level of shunting and other factors, requiring careful consideration and follow-up to address present symptoms and prevent any future cardiovascular deterioration.

Thanks for listening to this first part of the PedsCases podcast series on acyanotic congenital heart disease.

Part 2:

Thanks for joining us for the second part of our series on acyanotic heart lesions. My name is Herman Bami and I'm a second-year medical student from the Schulich School of Medicine and Dentistry at the University of Western Ontario. This podcast was supervised by Dr. Michael Grattan, a pediatric cardiologist at the London Health Sciences Center.

Our last podcast discussed the definition of congenital heart disease, how it is classified and the first of two types of acyanotic congenital heart lesions: left-to-right shunts. If you have any questions about those, or simply want to refresh your memory, please feel free to revisit that podcast. In this podcast, we will be going through the second category of acyanotic congenital heart disease: outflow tract obstructions.

The objectives of this podcast are to:

1. Describe the pathophysiology of obstructive acyanotic heart lesions
2. Review the pathophysiology, clinical presentation and management of pulmonary stenosis, aortic stenosis and coarctation of the aorta

Before we begin though, let's go through a case presentation. Maya is a 10-year-old girl who presents to your family clinic with a history of headache associated with exercise. While she has no history of hypertension, she is noted to have a blood pressure of 130/90 mmHg in both arms. When investigating further, it is noted that she has weaker femoral pulses. After discussing the different types of obstructive lesions, we'll come back to this case to discuss next steps in the evaluation of this patient.

Pulmonary Stenosis

Let's start now with the first type of outflow tract obstructions. Pulmonary stenosis accounts for approximately 10 to 15% of all congenital heart disease and can be further subclassified based on location into: valvular, subvalvular and supravalvular stenosis[1-2]. There are multiple possible etiologies including failure of the development of valve leaflets, insufficient resorption of infundibular tissue and insufficient canalization of peripheral arteries.

Clinically, mild pulmonary stenosis is often asymptomatic but moderate to severe stenosis can manifest in exertional dyspnea as well as easy fatigability. Severe stenosis in the newborn can also cause cyanosis due to right-to-left shunting at the level of the atria. A characteristic systolic ejection murmur can be detected at the second left intercostal space, often associated with a systolic ejection click. The duration of the murmur and pitch or harshness may increase with worsening stenosis. Furthermore, with severe pulmonary stenosis, a compensatory right ventricular hypertrophy may develop resulting in an impulse or right ventricular heave at the lower left sternal border.

Mild stenosis will often be undetectable via ECG and X-ray. However, moderate to severe stenosis may cause right axis deviation and right ventricular hypertrophy. Additionally,

post-stenotic dilation of the main pulmonary artery may be detectable on chest X-ray. Echocardiography provides the most information, especially in terms of the site and severity of stenosis, valvular morphology and degree of right ventricular hypertrophy. Although valvular pulmonary stenosis often doesn't progress beyond infancy, balloon valvuloplasty can be used to reduce the pressure gradient in cases of more severe pulmonary stenosis. Surgical repair can be employed if balloon valvuloplasty is unsuccessful or if subvalvular or supravalvar stenosis is present.

Aortic Stenosis

The next type of obstructive lesion we'll be discussing is aortic stenosis, which represents approximately 5% of congenital heart disease[1-2]. It can also be divided into valvular, subvalvular or supra valvular stenosis. The type of lesion itself depends on the level of leaflet development or resorption of perivalvular tissue. One common cause of aortic stenosis in children is bicuspid aortic valve, a condition in which the aortic valve is comprised of 2 instead of 3 leaflets[8]. It is the most common congenital heart defect with an estimated prevalence between 0.5 to 2%. Although bicuspid aortic valves in most children function well, some can cause significant aortic stenosis and insufficiency requiring intervention. Adult patients with bicuspid aortic valve have a significantly increased risk for the development of degenerative aortic stenosis and aortic insufficiency. All patients with bicuspid valve are at risk of ascending aorta dilatation and associated complications.

While mild to moderate obstructions can be asymptomatic, more severe stenosis can cause exertional dyspnea, syncope, reduced exercise tolerance, and even sudden death. Neonates with severe aortic stenosis may have cardiovascular collapse when their ductus arteriosus closes. A systolic ejection murmur can be detected at the second right intercostal space, radiating into the neck. As the degree of stenosis increases, the murmur becomes higher pitched, harsher and later peaking. The murmur may remain quiet if the left ventricular function is compromised. Additionally, with valvular stenosis, a systolic ejection click can be heard and a thrill may be present at the right upper sternal border or suprasternal notch.

ECG and chest X-ray findings are often normal with mild stenosis; however, severe stenosis can cause a compensatory left ventricular hypertrophy which can be detectable via ECG and chest X-ray. Furthermore, chest radiographs can be used to detect associated dilatation of the ascending aorta or aortic knob. Echocardiography is employed to show the site of stenosis, valvular morphology, ascending aorta dilatation and presence of left ventricular hypertrophy or dysfunction. Echocardiography can also be used to estimate the pressure gradient. While the degree of stenosis progresses with age, aortic insufficiency can also manifest as a significant complication. Although balloon valvuloplasty can be the first-line intervention used, it is not as successful as pulmonary balloon valvuloplasty. Thus, surgical management is often required for cases of significant valvular insufficiency or when balloon valvuloplasty is unsuccessful.

Coarctation of the Aorta

The last obstructive lesion we'll be discussing is coarctation of the aorta, which occurs in approximately 5 to 8% of all congenital heart defects[2,9-10]. The etiology behind coarctation of the aorta is a failure of the aortic isthmus to develop properly, or the presence of ductus arteriosus tissue within this area that leads to a constriction during the closure of the PDA postnatally. This causes a constricted aortic segment comprised of localized medial thickening.

As infants presenting with coarctation of the aorta often have other associated defects - such as hypoplastic aortic arches, abnormal aortic valves and VSDs – the timing and presentation varies greatly. Certain infants with severe coarctation may be dependent on a patent ductus arteriosus for descending aortic flow, with severe symptoms occurring shortly after the ductus arteriosus closes. Such symptoms would include poor feeding, respiratory distress, and shock.

Less severe obstructions may be asymptomatic and adequate blood flow in these cases is not dependent on a patent ductus arteriosus. In this situation, children may present older and with symptoms including: headache, hypertension of the upper extremities, claudication and a murmur best heard in the left interscapular area of the back. A bicuspid aortic valve is present in approximately half of these cases resulting in a systolic ejection click and possible systolic ejection murmur of aortic stenosis. Additionally, other classical findings of coarctation of the aorta include: weaker or absent femoral pulses compared with the right radial pulses, as well as a relative hypotension in the lower extremities.

In neonates, imaging studies can be used to show evidence of right ventricular hypertrophy as well as cardiomegaly and pulmonary edema. Additionally, echocardiography can be used to locate the site and extent of the coarctation, as well as associated lesions. Findings of left ventricular hypertrophy and cardiomegaly can also be observed via ECG and chest radiographs in older children. Rib notching, a radiological finding indicating the presence of collateral blood vessels bypassing the coarctation, may also be found in older children. Initial management strategies of a coarctation depend on the patient age. In neonates with severe coarctation, strategies include intravenous infusion of prostaglandin E_1 to open the ductus arteriosus, inotropic agents, diuretics and other supportive agents. Neonates with severe coarctation usually require surgical repair. Older children may require medications like beta-blockers to control systemic hypertension. Balloon angioplasty can be performed in older infants, while stenting is the preferred management choice in older children and adolescents. Even after successful coarctation repair, patients may suffer from persistent hypertension and an increased risk of heart disease and stroke.

Conclusion

Since we've now gone through the different types of outflow tract obstructions, let's come back to the case. Maya is a 10-year-old girl that has come to your clinic with a history of headaches associated with exercise. While she had no previous history of hypertension, her blood pressure was found to be 130/90 mmHg in both arms with slightly weaker femoral pulses. On further investigation, her blood pressure is found to be 100/60 mmHg in both legs with a 30 mmHg gradient between her arm and leg systolic blood pressures. An

echocardiogram and ECG are ordered, which both show mild left ventricular hypertrophy. In addition, echocardiography reveals a bicuspid aortic valve and a juxtaductal coarctation of the aorta. The patient is prescribed a beta-blocker for antihypertensive therapy and eventually undergoes successful stenting of her coarctation. Careful clinical and imaging follow-up will be conducted to ensure proper management of ongoing systemic hypertension, to evaluate for recurrent coarctation and to follow for complications related to her bicuspid aortic valve.

We've now reached the end of our two-part series on acyanotic congenital heart lesions but before we finish up, let's summarize the main messages from this podcast:
- Outflow tract obstructions are a second type of acyanotic heart lesions and consist of: aortic and pulmonary stenosis as well as coarctation of the aorta.
- Echocardiography is a key tool in the diagnosis of outflow tract obstructions with treatment often involving either balloon valvuloplasty or surgical repair, depending on the extent of the obstruction.

Thanks for listening to this PedsCases podcast series on acyanotic congenital heart disease.

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