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Congenital Heart Defects



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Section 1: Introduction

References: 1, 2

Congenital heart defects are abnormalities in the heart present at birth.

Congenital heart defects (CHDs) are the most common type of birth defect for newborns and are the leading cause of birth defect-associated infant illness and death. CHDs affect the structure and blood flow of the heart and can range from mild to severe. Mild CHDs often have little to no symptoms and can resolve on their own. Severe CHDs can cause serious chronic health issues and are often fatal without surgical interventions in the first few years of life.

In a healthy heart, the right side pumps oxygen-poor blood from the heart to the lungs. After the lungs oxygenate the blood, the left side of the heart pumps oxygen-rich blood to the rest of the body. The heart is divided into four chambers: two atria and two ventricles. The upper chambers of the heart are the left and right atria. The lower chambers of the heart are the left and right ventricles. CHDs commonly affect the walls of the heart between the chambers, heart valves, or the large blood vessels that carry blood to and from the heart. Early diagnosis is crucial to treating CHDs as some defects can cause serious adverse effects later in life and be fatal without early intervention. This course will review the prevalence and risk factors of CHDs, the different types of CHDs, common symptoms, diagnostic criteria, treatment options, and the impact of CHDs across the lifespan.

Section 2: Risk Factors and Prevalence

References: 1, 2, 3

According to the Centers for Disease Control and Prevention (CDC) 1% of infants born in the United States are diagnosed with a congenital heart defect and research shows that the prevalence of CHDs continues to increase. In the United

States, newborn screening for CHDs is routine shortly after birth. Routine screenings can help diagnose defects early and promote early intervention. Research has also helped develop effective treatment options for critical types of CHD. Survival rates for CHDs are improving as more long-term treatment options are becoming available. 81% of patients with CHDs are expected to survive to at least 35 years of age. Survival rates of CHD depend on the type and complexity of the defect and other comorbidities. CHDs can be categorized as cyanotic (critical) and non-cyanotic (non-critical). 97% of infants born with non-cyanotic CHDs are expected to survive to at least one year old compared to 75% of infants born with cyanotic CHDs. 4% of all infant deaths are due to CHDs and usually occur within the first 28 days of life.

Most causes of CHDs are unknown. It is common for CHD to occur due to genetic mutations not inherited from the parents. Research shows that the cause of CHDs can sometimes be a combination of genetic and other external factors. Certain risk factors have been linked to CHDs including:

- Smoking during pregnancy
- Certain medications during pregnancy
- Chronic conditions including pre-existing diabetes, obesity, and phenylketonuria
- Family history of CHD
- Preterm birth

Certain medications can put infants at higher risk for congenital heart defects. Medications that are contraindicated during pregnancy are called teratogens. Thalidomide is a teratogen that is used to treat multiple myeloma and certain skin conditions. Research shows that thalidomide is associated with severe birth defects including congenital heart defects. Isotretinoin is another medication

contraindicated in pregnancy that is used to treat severe cystic acne. Other medications that are considered teratogens include certain anticonvulsants, antiarrhythmics, and antidepressants. It is important for patients to be aware of which medications should be stopped during pregnancy to prevent CHDs and other birth defects.

Education should be provided during pregnancy about the importance of controlling existing medication conditions, such as diabetes and phenylketonuria, and avoiding environmental factors such as smoke exposure to decrease the risk of congenital heart defects. Phenylketonuria is a disorder that causes an excess amount of an amino acid called phenylalanine and has been shown to increase the risk of certain birth defects. Infants born preterm are also at greater risk of CHDs due to the underdevelopment of the fetus. While CHDs cannot always be prevented, patients should be educated on the importance of maintaining a healthy pregnancy and decreasing any existing risk factors.

Section 2 Personal Reflection

What are some things you could do in your community to spread awareness for risk factors of congenital heart defects?

Section 2 Key Words

Congenital heart defect - An abnormality of the structure of the heart that is present at birth.

Atria - The two upper chambers of the heart that receive blood from the body or the lungs.

Ventricles - The lower chambers of the heart that pump blood to the lungs or the body.

Teratogen - An agent that causes the malformation of an embryo and is therefore contraindicated during pregnancy.

Section 3: Non-cyanotic Congenital Heart Defects

References: 1, 2, 3, 4, 6, 7

Non-cyanotic or non-critical congenital heart defects affect the blood flow of the heart, are often asymptomatic, and may resolve on their own without medical interventions. Non-cyanotic CHDs do not affect the amount of oxygen in the blood. About 75% of all congenital heart defects are non-cyanotic. Types of non-cyanotic CHDs include:

- Atrial septal defect
- Ventricular septal defect
- Atrioventricular septal defect
- Coarctation of the aorta
- Aortic valve stenosis
- Pulmonary valve stenosis
- Patent ductus arteriosus

Non-cyanotic CHDs are often asymptomatic or have minimal symptoms that do not impact the infant's daily life. Diagnosis of non-cyanotic CHDs can be delayed due to the lack of symptoms. Common symptoms of non-cyanotic CHDs include:

- Heart murmur
- Hypertension

- Pulmonary hypertension
- Heart failure
- Dyspnea
- Dizziness and syncope
- Fatigue

Non-cyanotic CHDs can be diagnosed during pregnancy or after birth. Sometimes the heart defect is not diagnosed until adulthood due to the lack of symptoms.

Diagnostic criteria for non-cyanotic CHDs includes:

- Ultrasound
- Echocardiogram
- Electrocardiogram
- Chest x-ray
- Cardiac catheterization

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A more prominent non-cyanotic CHD can be detected on an ultrasound during pregnancy. An ultrasound is a procedure that uses sound waves to look at tissues and organs inside the body and is the most effective way to monitor fetal growth during pregnancy. Not all CHDs are detected during pregnancy especially if they are small. If an ultrasound shows a suspected defect, the patient will be referred to a pediatric cardiologist for further testing to determine the type and severity of the CHD and if any interventions are needed.

An echocardiogram (ECHO) uses ultrasound to create images of the heart valves and structure. An ECHO gives a detailed image and can detect abnormalities in the structure and blood flow of the heart. An ECHO can be performed during

pregnancy or after birth. Other testing such as an electrocardiogram (EKG or ECG) and cardiac catheterization are typically performed after birth for further diagnosis. An EKG measures the electrical activity of the heart and can determine if the heart is in an abnormal rhythm. A cardiac catheterization is a procedure where a catheter is peripherally inserted and advanced to the heart to help determine the adequacy of blood flow in the heart. Cardiac catheterization can measure the oxygenation of the blood and detect any blockages or abnormalities in the heart structure and valves.

Atrial Septal Defect

An atrial septal defect (ASD) is a hole in the wall of the heart that divides the atria that remains open after birth. During fetal development, the heart has several openings in the walls dividing the atria, but these holes usually close before or soon after birth. The cause is often unknown, but research shows that certain cases can be connected to genetics. 13 in every 10,000 infants are born with ASDs each year.

ASDs can be diagnosed either during pregnancy or soon after birth. In mild cases, an ASD may not be diagnosed until adulthood due to the lack of symptoms. Heart murmurs are a common sign of ASDs and are often found during routine newborn exams. An ECHO is typically ordered after the heart murmur is detected and can confirm the ASD diagnosis.

ASDs can vary in size and can sometimes resolve on their own. A smaller hole typically presents with mild or no symptoms while a bigger hole can cause significant symptoms. Common symptoms of ASDs include:

- Heart murmur
- Frequent respiratory infections

- Dyspnea
- Fatigue when feeding in infants
- Arrhythmias
- Palpitations
- Lower extremity edema
- Stroke

Treatment for ASDs depends on the defect's size, the age at diagnosis, and other health conditions. In infants, healthcare providers often monitor the ASD to see if the hole closes on its own. Medications may be prescribed to treat any present symptoms, but medications will not repair the defect. The only way to repair the defect is through surgical intervention. Surgery may be recommended within the first year of life if the hole is large or if serious symptoms are present. Surgery may also be recommended to prevent any problems later in life.

Ventricular Septal Defect

The most common type of congenital heart defect is a ventricular septal defect (VSD). VSDs occur in 37% of children with a congenital heart defect. A VSD is a hole in the wall of the heart that divides the ventricles that remains open after birth. During fetal development, the heart has several openings in the walls dividing the ventricles, but these holes usually close before or soon after birth. A VSD can cause oxygen-rich and oxygen-poor blood to mix which can cause an increased workload on the heart and the lungs. The cause of VSDs is often unknown, but research shows that certain cases can be connected to genetics.

VSDs can be diagnosed either during pregnancy or soon after birth. 90% of VSDs eventually resolve on their own. A small VSD may have mild or no symptoms, but

a large VSD can cause severe symptoms and may require surgical intervention. Heart murmurs are a common sign of VSDs and are often found during routine newborn exams. An ECHO is typically ordered after the heart murmur is detected and can confirm the VSD diagnosis. Common symptoms of VSDs include:

- Heart murmur
- Tachypnea
- Dyspnea
- Fatigue
- Poor feeding in infants
- Poor weight gain
- Pulmonary hypertension

Treatment for VSDs depends on the defect's size, location, the age at diagnosis, and other health conditions. In infants, healthcare providers often monitor the VSD to see if the hole closes on its own. Medications may be prescribed to treat any present symptoms, but medications will not repair the defect. The only way to repair the defect is through surgical intervention. Surgery may be recommended within the first year of life if the hole is large or if serious symptoms are present. A VSD can cause a leak in the aorta which can have serious consequences. Pediatric cardiologists will monitor patients with VSDs for symptoms of a leaky aorta or other serious symptoms. Patients may need surgery to repair VSDs and prevent further complications. After the initial surgery or if the VSD closes on its own, patients have a good long-term prognosis and rarely need additional medical interventions.

Atrioventricular Septal Defect

An atrioventricular septal defect (AVSD) is holes between the chambers of the right and left sides of the heart that can cause the valves between these chambers to not form correctly. AVSDs are also known as atrioventricular (AV) canal defects or endocardial cushion defects. AVSDs cause abnormal blood flow in the heart and can cause blood to have a lower amount of oxygen or a larger than normal amount of blood in the lungs. The cause of AVSDs is often unknown, but research shows that some cases can be attributed to genetics. AVSDs can be commonly found in infants diagnosed with Down syndrome.

There are two types of AVSDs: complete and partial. A complete AVSD is a large hole in the center of the heart which allows blood to flow between all four chambers. All the valves that normally control the blood flow through the heart do not form correctly causing the mixing of oxygen-rich and oxygen-poor blood. A partial AVSD occurs when only some of the valves have defects, so the abnormal blood flow is not as significant.

AVSDs can be diagnosed during pregnancy or after birth. In mild cases, AVSDs may not be diagnosed until adulthood due to the lack of symptoms. During pregnancy, routine birth defect screenings can detect AVSDs on an ultrasound. After birth, infants are often diagnosed by detecting a heart murmur during routine exams. Patients are referred to a pediatric cardiologist and an ECHO is typically ordered to confirm the AVSD diagnosis.

Complete AVSDs usually show symptoms a few weeks after birth while partial AVSDs may not show symptoms for years. Symptoms of an AVSD include:

- Heart murmur
- Dyspnea
- Weak pulses

- Ashen or bluish skin color
- Poor feeding in infants
- Slow weight gain in infants
- Lower extremity edema
- Arrhythmias
- Congestive heart failure
- Pulmonary hypertension

Patients often need surgical intervention to treat AVSDs and any correlating symptoms. Medications may be prescribed to treat symptoms and correlating congestive heart failure, but these are often only a short-term solution. Surgery can close the holes and separate the valves to allow for correct blood flow through the heart. Patients can have lifelong complications even after a successful surgery. A common complication of AVSDs is a leaky mitral valve. This occurs when the mitral valve does not close completely, allowing blood to back up in the mitral valve.

Coarctation of the Aorta

Coarctation of the aorta is the narrowing of the aorta causing blockage to normal blood flow. This can cause blood to back up in the left ventricle which can increase the workload on the heart. The aorta is the main artery of the heart that carries oxygen-rich blood to the rest of the body. If the narrowing is severe, serious problems can occur including heart failure.

Coarctation of the aorta is usually diagnosed after birth depending on the severity of symptoms. Low levels of oxygen indicated by pulse oximetry monitoring in

newborns is typically the first sign of coarctation of the aorta. Other symptoms include:

- Heart murmur
- Irritability
- Diaphoresis
- Dyspnea
- Pale skin
- Weak pulses in lower extremities

Patients with these symptoms will be referred to a pediatric cardiologist and an ECHO can confirm the diagnosis. The ECHO can also show the location and severity of the coarctation. Cardiologists may also order other diagnostic tests such as an EKG, cardiac MRI, or cardiac catheterization to measure the heart function.

Surgery is often needed soon after birth to fix the coarctation. Balloon angioplasty is often performed to correct the narrowing. Balloon angioplasty is a cardiac catheterization procedure that uses a peripherally inserted catheter to inflate a balloon at the narrow part of the aorta. The balloon expands the aorta and allows blood to flow through. If balloon angioplasty is not successful, patients may need open-heart surgery to remove the narrow portion and reconstruct the aorta. This is a more invasive procedure and is usually recommended only when balloon angioplasty is not successful or if the patient is experiencing severe symptoms. Patients with coarctation of the aorta may continue to experience high blood pressure even after surgical interventions. High blood pressure is a lifelong condition but can be controlled with medications.

Aortic and Pulmonary Valve Stenosis

Aortic valve stenosis is the narrowing of the aortic valve opening and restricts blood flow from the left ventricle into the aorta. Pulmonary valve stenosis is the narrowing of the pulmonary valve opening and restricts blood flow from the right ventricle to the pulmonary arteries. Aortic valve stenosis can be congenital or caused by damage to the valve due to aging. Pulmonary valve stenosis is often congenital but can rarely be caused by a severe infection in adults.

Patients with aortic or pulmonary valve stenosis can be asymptomatic until the restricted blood flow becomes severe. Symptoms of aortic and pulmonary valve stenosis include:

- Heart murmur
- Palpitations
- Chest pain
- Dyspnea
- Dizziness or syncope
- Lower extremity edema
- Fatigue
- Poor weight gain
- Poor feeding in infants

Patients with these symptoms will be referred to a pediatric cardiologist and an ECHO can confirm the diagnosis. Cardiologists may also order other diagnostic tests such as an EKG, cardiac MRI, or cardiac catheterization to measure the heart function.

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If patients are asymptomatic, consistent monitoring is often the only intervention needed. Cardiologists will monitor the valve stenosis to determine if the narrowing is getting worse. If symptoms worsen, further interventions may be required. Medications or surgery may be recommended to repair or replace the valves. Balloon valvuloplasty is often performed to correct stenosis. Balloon valvuloplasty is a cardiac catheterization procedure that uses a peripherally inserted catheter to inflate a balloon at the narrow part of the valves. Even if the balloon valvuloplasty is successful, there is a chance that stenosis can occur again, and further interventions may be needed. Patients may need open-heart surgery to repair or replace the valves with a mechanical or donor valve. Patients with a replacement valve need to take medications to prevent lifelong complications such as blood clots, endocarditis, right ventricular hypertrophy, and heart failure. Patients will need to have regular follow-up with their cardiologist throughout their lives.

Patent Ductus Arteriosus

Patent ductus arteriosus (PDA) occurs when the opening between the aorta and the pulmonary arteries does not close properly after birth causing excessive blood flow to the lungs. The ductus arteriosus is a normal opening between the aorta and pulmonary arteries that occurs during fetal development. The ductus arteriosus allows blood to bypass the fetus's lungs due to the placenta providing oxygen-rich blood. Normally the ductus arteriosus closes a few days after birth. A PDA is a common congenital heart defect and often occurs in premature infants. Prematurity is defined as birth more than three weeks before the due date.

A PDA is usually diagnosed soon after birth. A heart murmur and congestion in the lungs is typically heard during routine newborn exams. Healthcare providers will refer the patient to a pediatric cardiologist and an ECHO can confirm the

diagnosis.

Small openings can eventually close on their own and may not require any treatment. Patients with larger openings may exhibit more severe symptoms and need surgical interventions. Cardiac catheterization or open-heart surgery can treat PDAs. Cardiac catheterization is the most common treatment due to the decreased risks compared to open-heart surgery. A coil is threaded through a peripherally inserted catheter to close the opening. Open-heart surgery is usually only recommended in infants with very large PDAs. If a PDA is left untreated, it can cause lifelong complications including heart failure, endocarditis, pulmonary edema, and pulmonary hypertension.

Section 3 Personal Reflection

What education should be provided to parents whose child has a non-cyanotic congenital heart defect regarding different treatment options?

Section 3 Key Words

Non-cyanotic heart defect - A type of congenital heart defect that causes blood to flow through the heart abnormally but does not affect the amount of oxygen in the blood.

Heart murmur - A whooshing sound heard during a heartbeat caused by excessive blood flow through the heart valves and can indicate certain congenital heart defects.

Ultrasound - A diagnostic procedure that uses sound waves to look at tissues and organs inside the body and is the most effective way to monitor fetal growth during pregnancy.

Echocardiogram (ECHO) - A diagnostic procedure that uses ultrasound to create images of the heart valves and structure and can detect abnormalities in the structure and blood flow of the heart.

Electrocardiogram (EKG or ECG) - A diagnostic procedure that measures the electrical activity of the heart and can determine if the heart is in an abnormal rhythm.

Cardiac catheterization - A diagnostic procedure where a catheter is peripherally inserted and advanced to the heart to measure the adequacy of blood flow in the heart, the oxygenation of the blood, and detect any blockages or abnormalities in the heart structure and valves.

Atrial septal defect (ASD) - A congenital heart defect that occurs when there is a hole in the wall of the heart that separates the atria that remains open after birth.

Ventricular septal defect (VSD) - A congenital heart defect that occurs when there is a hole in the wall of the heart that separates the ventricles that remains open after birth.

Atrioventricular septal defect (AVSD) - A congenital heart defect that occurs when there is a hole in the walls of the heart that separates the atria and the ventricles causing mixing of oxygen-rich and oxygen-poor blood and abnormal blood flow through the heart.

Coarctation of the aorta - A congenital heart defect that occurs when the aorta is narrowed causing a backup of blood flow and an increased workload on the heart.

Aorta - The main artery of the heart that carries oxygen-rich blood away from the heart and to the rest of the body.

Balloon angioplasty - A cardiac catheterization procedure that uses a peripherally

inserted catheter to inflate a balloon at the narrow part of the aorta to allow blood to flow through.

Open-heart surgery - An invasive surgical procedure where the chest is opened to allow visualization of the heart and the ability to repair and reconstruct the heart structure and vessels.

Aortic valve stenosis - A congenital heart defect that occurs when the aortic valve becomes narrowed and restricts blood flow from the left ventricle into the aorta.

Pulmonary valve stenosis - A congenital heart defect that occurs when the pulmonary artery valves become narrowed and restricts blood flow from the right ventricle to the pulmonary arteries.

Balloon valvuloplasty - A cardiac catheterization procedure that uses a peripherally inserted catheter to inflate a balloon at the narrow part of the valves to promote blood flow.

Patent ductus arteriosus (PDA) - A congenital heart defect that occurs when the ductus arteriosus does not close when it is supposed to and causes excess blood to flow to the lungs.

Section 4: Cyanotic Congenital Heart Defects

References: 1, 2, 3, 5, 9, 10, 12, 13, 14, 15, 16, 17

Cyanotic or critical congenital heart defects affect the amount of oxygen-rich blood in the heart and can cause life-threatening symptoms that require immediate medical interventions. Patients with cyanotic CHDs commonly have low levels of oxygen after birth and often require surgical interventions within the first year of life. About 25% of all congenital heart defects are cyanotic. Some non-cyanotic CHDs can be deemed critical if the defect is severe such as coarctation of

the aorta and aortic or pulmonary stenosis.

There are three different types of cyanotic CHDs: left heart obstructive defects, right heart obstructive defects, and mixing defects. Left heart obstructive defects reduce blood flow between the heart and the rest of the body, right heart obstructive defects reduce blood flow between the heart and the lungs, and mixing defects cause oxygen-rich and oxygen-poor blood to blend.

Types of left heart obstructive defects include:

- Hypoplastic left heart syndrome
- Interrupted aortic arch
- Severe coarctation of the aorta
- Severe aortic valve stenosis

Types of right heart obstructive defects include:

- Pulmonary atresia
- Tricuspid atresia
- Tetralogy of Fallot
- Severe pulmonary valve stenosis

Types of mixing defects include:

- Transposition of the great arteries
- Total anomalous pulmonary venous return
- Truncus arteriosus

Cyanotic CHDs often have severe symptoms soon after birth that can be fatal without prompt interventions. Common symptoms of cyanotic CHDs include:

- Diaphoresis and irritability with feeding
- Poor weight gain
- Tachypnea
- Fatigue while feeding
- Cyanosis
- Dyspnea
- Tachycardia
- Pulmonary edema
- Heart murmur
- Hepatomegaly
- Weak femoral pulses
- Signs of poor perfusion
- Lethargy
- Exercise intolerance and palpitations in older children

Cyanotic CHDs can be diagnosed during pregnancy or soon after birth. Diagnostic criteria for cyanotic CHDs is similar to non-cyanotic CHDs and include:

- Fetal ultrasound
- ECHO
- EKG
- Pulse oximetry screening

Pulse oximetry screening on newborns can often detect the first signs of a cyanotic CHD. Pulse oximetry screening was first recommended by the American Academy of Pediatrics in 2012 and is standard for all newborns in the United States once the infant is at least 24 hours old. Pulse oximetry screening can only identify CHDs that cause cyanosis so it is not a reliable diagnostic test for all congenital heart defects. Positive pulse oximetry screening results includes at least one of the following criteria:

- Oxygen saturation less than 90%
- Greater than a 3% difference in oxygen saturation levels between the right upper and lower extremities. This must be measured at least 3 different times and at least one hour apart.
- Oxygen saturation less than 95% in both extremities. This must be measured at least 3 different times and at least one hour apart.

Even with effective initial treatment, patients with cyanotic CHDs often have lifelong complications. Multiple surgeries are often required and some patients may eventually need a heart transplant. Complications such as kidney and liver disease, chronic fatigue, arrhythmias, and heart failure are common in patients with cyanotic congenital heart defects.

Left Heart Obstructive Defects

Severe coarctation of the aorta, severe aortic valve stenosis, hypoplastic left heart syndrome (HLHS), and interrupted aortic arch (IAA) are all types of left heart obstructive defects. The left side of the heart is responsible for supplying oxygen-rich blood to the body, so left heart obstructive defects can cause significant complications. Common symptoms of left heart obstructive defects include:

- Heart murmur

- Dyspnea
- Cyanosis
- Low oxygen saturation levels
- Weak pulses in lower extremities
- Fatigue
- Poor feeding in infants

Left heart obstructive defects can be diagnosed during pregnancy or soon after birth. If a defect is suspected on a routine fetal ultrasound, an ECHO can confirm the diagnosis. Newborn pulse oximetry screening can assist in early diagnosis soon after birth.

Severe coarctation of the aorta and severe aortic valve stenosis are two CHDs that can be considered critical if the defect is serious enough to cause cyanosis. Extensive narrowing of the aorta can cause a significant increase of the workload on the heart which can ultimately lead to heart failure. More mild cases of coarctation of the aorta or aortic valve stenosis may be asymptomatic and require less invasive interventions. When these CHDs cause cyanosis, early surgical interventions are required to prevent serious complications.

Hypoplastic left heart syndrome (HLHS) is a critical congenital heart defect that occurs when the left side of the heart does not form correctly during pregnancy. This causes the inability of the left side of the heart to pump oxygen-rich blood to the rest of the body and affects a number of structures on the left side of the heart. The left ventricle, mitral valve, aortic valve, and part of the aorta are all underdeveloped and too small with HLHS.

HLHS is fatal without early surgical interventions. Medications may be prescribed to treat symptoms until the infant is strong enough for surgery, but multiple

surgeries throughout the first few years of life are necessary to restore adequate heart function. The Norwood procedure is usually performed during the first 2 weeks of life. This surgery focuses on repairing the aorta and connecting it to the right ventricle to allow blood flow to the rest of the body. The Glenn shunt procedure is usually performed during 4 to 6 months of age. This surgery focuses on reducing the workload of the right ventricle by allowing oxygen-poor blood to flow directly to the lungs. The Fontan procedure is usually performed 18 months to 3 years of age and focuses on repairing the structure of the heart to prevent oxygen-rich and oxygen-poor blood from mixing.

Interrupted aortic arch (IAA) is a rare cyanotic CHD that causes a structural defect along the aortic arch which causes an obstruction in blood flow to the rest of the body. Similar to HLHS, medications may be prescribed to treat symptoms until surgical interventions can be performed. Surgery can repair the obstruction in the aorta to allow for adequate blood flow from the heart to the rest of the body. Many times, multiple surgeries are needed if the obstruction is severe.

Right Heart Obstructive Defects

Severe pulmonary valve stenosis, pulmonary and tricuspid atresia, and Tetralogy of Fallot are all types of right heart obstructive defects. The right side of the heart is responsible for receiving oxygen-poor blood from the body and delivering it to the lungs, so right heart obstructive defects can cause significant complications. Symptoms of right heart obstructive defects are similar to symptoms of left heart obstructive defects and include:

- Heart murmur
- Dyspnea
- Cyanosis

- Low oxygen saturation levels
- Fatigue
- Poor feeding in infants

Similar to left heart obstructive defects, right heart obstructive defects can also be diagnosed during pregnancy or soon after birth. If a defect is suspected on a routine fetal ultrasound, an ECHO can confirm the diagnosis. Newborn pulse oximetry screening can assist in early diagnosis of some right heart obstructive defects soon after birth.

Severe pulmonary valve stenosis is a CHD that can be considered critical if the defect is serious enough to cause cyanosis. Extensive narrowing of the pulmonary valve can cause a restriction in blood being able to flow to the lungs to receive oxygen. More mild cases may be asymptomatic and require less interventions, but severe pulmonary stenosis requires early surgical interventions to prevent serious complications.

Pulmonary atresia causes a blockage of the pulmonary valve which blocks blood flow from entering the lungs. Tricuspid atresia causes a blockage of the tricuspid valve which blocks blood flow on the right side of the heart. Tricuspid atresia often causes the underdevelopment of the right ventricle. Both pulmonary and tricuspid atresia require early surgical intervention. Multiple surgeries are often needed to repair the valves and restore adequate blood flow.

Tetralogy of Fallot is one of the most common critical CHDs and involves four different defects including pulmonary stenosis, a ventricular septal defect (VSD), an overriding aorta, and right ventricular hypertrophy. An overriding aorta is when the aorta is positioned between the two ventricles causing oxygen-poor blood to flow directly into the aorta. Right ventricular hypertrophy is when there is an increased workload on the heart causing the right ventricular wall to thicken.

“Tet spells” are a common symptom of Tetralogy of Fallot. These instances involve a rapid drop in oxygen saturation levels causing cyanosis and usually occur when infants are crying or eating. Tet spells commonly occur during infancy and usually begin to decrease after 4 to 5 years of age. Multiple surgeries are needed to repair the multiple defects involved in Tetralogy of Fallot. A complete repair of the heart is completed in several steps which helps patch the VSD and repair or replace the pulmonary valve. A complete repair will usually help resolve the right ventricular hypertrophy as the heart no longer has to work hard to pump blood through the right ventricle.

Mixing Defects

Transposition of the great arteries, total anomalous pulmonary venous return (TAPVR), and truncus arteriosus are all types of mixing defects. Mixing defects involve structural abnormalities in the heart that cause oxygen-rich and oxygen-poor blood to mix. Symptoms of mixing defects include:

- Tachycardia
- Dyspnea
- Cyanosis
- Low oxygen saturation levels
- Fatigue
- Poor feeding in infants

Similar to other cyanotic congenital heart defects, mixing defects can be diagnosed during pregnancy or soon after birth. If a defect is suspected on a fetal ultrasound, an ECHO can confirm the diagnosis. Newborn pulse oximetry screening can detect some mixing defects early on.

Transposition of the great arteries occurs when the aorta and the pulmonary artery are in abnormal positions. This causes the aorta and the pulmonary artery to be connected to the wrong ventricles and oxygen-poor blood to flow to the rest of the body. Infants need surgical interventions in the first few weeks of life. Transposition of the great arteries is often fatal within the first year of life without surgery.

Total anomalous pulmonary venous return (TAPVR) occurs when oxygen-rich blood flows to the right side of the heart instead of the left. This causes mixing of oxygen-rich and oxygen-poor blood and not enough oxygen to the rest of the body. Surgery is needed to fix the TAPVR defect, but the age at which the surgery is recommended can vary. The severity of the defect and the specific abnormal structures of the defect will determine how soon after birth surgery needs to occur. Similar to other cyanotic CHDs, TAPVR can cause lifelong complications even after successful surgical interventions.

Truncus arteriosus occurs when the aorta and pulmonary artery fail to separate and instead create one blood vessel in the heart. Because the blood vessels fail to separate, oxygen-rich and oxygen-poor blood mix as it flows to the lungs and the rest of the body. Medications can be given to help with symptoms until the infant is strong enough for surgery, but early surgical intervention is needed within the first few months of life to create an artificial vessel. Multiple surgeries are needed to continue to replace the artificial vessels as the child grows.

Section 4 Personal Reflection

What support and resources could be provided to parents whose child needs to have surgery for a critical congenital heart defect?

Section 4 Key Words

Cyanotic congenital heart defect - A type of congenital heart defect that causes blood to flow through the heart abnormally and affects the amount of oxygen-rich blood in the heart.

Left heart obstructive defect - A type of cyanotic congenital heart defect that reduces the blood flow between the heart and the rest of the body.

Right heart obstructive defect - A type of cyanotic congenital heart defect that reduces the blood flow between the heart and the lungs.

Mixing defect - A type of cyanotic congenital heart defect that causes oxygen-rich and oxygen-poor blood to mix.

Pulse oximetry screening - A standard screening for all newborns in the United States to help identify congenital heart defects that cause cyanosis by measuring the oxygen saturation levels in the upper and lower extremities.

Coarctation of the aorta - A congenital heart defect that occurs when the aorta is narrowed causing a backup of blood flow and an increased workload on the heart.

Aortic valve stenosis - A congenital heart defect that occurs when the aortic valve becomes narrowed and restricts blood flow from the left ventricle into the aorta.

Hypoplastic left heart syndrome (HLHS) - A cyanotic congenital heart defect that occurs when the left side of the heart does not form correctly causing the inability of the heart to pump oxygen-rich blood to the body, and is fatal without surgical interventions.

Interrupted aortic arch (IAA) - A cyanotic congenital heart defect that causes a structural defect along the aortic arch and an obstruction in blood flow to the rest of the body.

Pulmonary valve stenosis - A congenital heart defect that occurs when the pulmonary artery valves become narrowed and restricts blood flow from the right ventricle to the pulmonary arteries.

Pulmonary atresia - A congenital heart defect that occurs when the pulmonary valve is blocked preventing blood flow into the lungs.

Tricuspid atresia - A congenital heart defect that occurs when the tricuspid valve is blocked preventing blood flow on the right side of the heart.

Tetralogy of Fallot - A cyanotic congenital heart defect that involves pulmonary stenosis, a ventricular septal defect, an overriding aorta, and right ventricular hypertrophy causing restricted blood flow on the right side of the heart.

Overriding aorta - A condition resulting in the aorta being positioned between the two ventricles causing oxygen-poor blood to flow directly into the aorta.

Right ventricular hypertrophy - A condition resulting in an increased workload on the heart causing the right ventricular wall to thicken.

Tet spells - A common symptom of Tetralogy of Fallot where there is a rapid drop in oxygen saturation levels causing cyanosis and usually occurs when infants are crying or eating.

Transposition of the great arteries - A cyanotic congenital heart defect that occurs when the aorta and the pulmonary artery are in abnormal positions causing oxygen-poor blood to flow to the rest of the body.

Total anomalous pulmonary venous return (TAPVR) - A cyanotic congenital heart defect that occurs when oxygen-rich blood flows to the right side of the heart instead of the left causing mixing of oxygen-rich and oxygen-poor blood and not enough oxygen to the rest of the body.

Truncus arteriosus - A cyanotic congenital heart defect that occurs when the aorta

and pulmonary artery fail to separate causing oxygen-rich and oxygen-poor blood mix as it flows to the lungs and the rest of the body.

Section 5: Impact Across the Lifespan

References: 1, 2, 4, 18, 19

Congenital heart defects can have lifelong implications. Advancements in healthcare have allowed children with CHDs to live longer and healthier lives, but serious complications can still occur. Children and adults with CHDs need lifelong medical care and often develop chronic health issues related to the complexity and severity of the CHD. Potential health issues related to CHDs include:

- Arrhythmias
- Pulmonary hypertension
- Heart failure
- Infective endocarditis
- Increased risk of blood clots and stroke
- Kidney and liver disease
- Endocrine disorders including diabetes
- Obesity
- Complications during pregnancy
- Fatigue during physical activity
- Cognitive disabilities

Arrhythmias can cause an increased risk of blood clots due to the heart not being

able to pump enough blood to the rest of the body. Infective endocarditis is an infection in the heart tissue that can become fatal if left untreated. Due to the heart already being damaged from the CHD, infective endocarditis can cause serious complications. All of these complications can ultimately lead to heart failure in patients with CHDs. According to research, heart failure is the leading cause of death for adults who have a CHD.

A CHD is one of the most common complications for someone who is pregnant. Pregnancy can cause stress on the heart which can be exacerbated by a CHD. Many times the defect does not cause any complications during pregnancy, but some patients may need to take medications to decrease the workload on the heart and be monitored closely during pregnancy.

20 to 30% of adults with CHDs have physical or cognitive disabilities. The likelihood of a disability is directly related to the complexity and severity of the CHD. Over 50% of children with a critical CHD have some form of disability while only 20% of children with a non-critical CHD have some form of disability. Children with a CHD are more likely to have developmental delays, behavioral problems, and speech disorders.

The Impact on Caregivers

Children with CHDs require frequent hospitalizations, therapy appointments, medications, and other medical care. Children often miss school and cannot participate in as many extracurricular activities due to medical restrictions. Caring for a child with a CHD can cause stress and financial strain for caregivers. Research shows that parents of children with a CHD report higher levels of stress, anxiety, and depression.

It is important to help families develop effective coping strategies when caring for a child with a CHD. Parents often grieve the lifestyle they thought their child

would have and can have a difficult time accepting their child's limitations. The financial and emotional stress of ongoing medical needs can also contribute to daily stress. It is important for caregivers to be aware of coping strategies including:

- Spirituality
- Support system
- Physical activity
- Cognitive therapy
- Self care activities

Effective coping strategies can help caregivers cope with the daily needs of a child with a CHD and help the family adjust to the needs of the child. It is important for healthcare providers to care for the patient and the family. Healthcare providers can be an important resource for the family when navigating big life changes such as caring for a child with a CHD.

Lifestyle Changes

Patients need to make many lifestyle changes to cope with potential health issues related to CHDs. Patients need to follow up with their cardiologist regularly. Many adult patients with CHDs continue to follow up with their pediatric healthcare providers throughout their lives to provide continuity of care. Even with successful interventions during infancy, additional surgeries or other procedures may be needed throughout the lifespan.

Other lifestyle changes that can help prevent potential health issues include:

- Heart healthy diet

- Regular physical activity
- No smoking
- Maintaining a healthy weight
- Managing stress

A heart healthy diet can help lower blood pressure and prevent heart failure. A heart healthy diet includes a lot of fruits, vegetables, whole grains, and lean protein. Patients on a heart healthy diet should avoid processed foods and alcohol, and limit sugar and salt intake. Patients can work with a nutritionist and a cardiologist to develop an individualized diet plan to maintain a healthy heart. Research shows that regular physical activity can help maintain a healthy weight and help manage stress. Patients with CHDs may have some physical restrictions depending on the severity of the CHD, but some type of regular physical activity is recommended to maintain a healthy heart. Patients can discuss with their cardiologist what type of physical activity is right for them.

Managing mental health is also an important part of a healthy lifestyle for patients with CHDs. Children and adults may experience anxiety, depression, and stress when dealing with lifelong complications from CHDs. Developing healthy coping skills is essential to maintaining a healthy lifestyle. Healthcare providers can help patients find coping strategies such as cognitive therapy, self care activities, and a support system. Not every coping skill will be effective for each patient, so it is important for healthcare providers to develop an individualized plan for each patient.

Congenital heart defects are lifelong health conditions that affect patients and families throughout the lifespan. Healthcare providers must be aware of the impact of CHDs and the needs of the patient and family throughout the lifespan to cope with this chronic condition.

Section 5 Personal Reflection

What does “taking care of the patient and family” mean to you as a nurse? How can you support the patient and the family when caring for a patient with a CHD?

Section 5 Key Words

Arrhythmia - An irregular heart rhythm that can be caused by many different things.

Infective endocarditis - Inflammation of the heart tissue that is caused by an infection and can cause serious complications.

Heart healthy diet - A diet focusing on fruits, vegetables, whole grains, and lean protein to help improve heart function.

Section 6: Case Study #1

The nurse is caring for a pregnant patient who has arrived at the clinic for their initial exam with the OBGYN. This is the patient’s first pregnancy. During the assessment, the patient states that they are currently on several medications. The patient states “I cannot remember the name of the medications, but one is for my seasonal allergies and the other is for my cystic acne.” The patient expresses fear that they are “worried about doing something to hurt the baby”. The patient states “my sister had a baby with a heart defect and the baby did not survive. I am so scared that I will do something during pregnancy to cause a heart defect.”

1. What are some information and statistics that could be provided to the patient to ease their concerns about their baby having a CHD?
2. What are some preventative measures the nurse could provide to the patient to help during pregnancy to decrease the risk of a CHD?

3. What education would the nurse provide to the patient about medications to avoid during pregnancy that could increase the risk of a CHD?

Section 7: Case Study #1 Review

This section will review the case studies that were previously presented in each section. Responses will guide the clinician through a discussion of potential answers as well as encourage reflection.

1. What are some information and statistics that could be provided to the patient to ease their concerns about their baby having a CHD?

In the United States, newborn screening for CHDs is routine shortly after birth. Routine screenings can help diagnose defects early and promote early intervention. Research has also helped develop effective treatment options for critical types of CHD. Survival rates for CHDs are improving as more long-term treatment options are becoming available. There are some preventative measures that can be taken during pregnancy to reduce the risk of CHDs, but many times the cause is unknown. It is important to educate parents of a child with a CHD that because the cause is often unknown, it is likely not their fault that caused the heart defect.

2. What are some preventative measures the nurse could provide to the patient to help during pregnancy to decrease the risk of a CHD?

Most causes of CHDs are unknown. It is common for CHD to occur due to genetic mutations not inherited from the parents. Research shows that the cause of CHDs can sometimes be a combination of genetic and other external factors. Certain risk factors have been linked to CHDs including smoking during pregnancy, certain medications during pregnancy, chronic conditions including pre-existing diabetes, obesity, and phenylketonuria,

family history, and preterm birth. Education should be provided during pregnancy about the importance of controlling existing medication conditions and avoiding environmental factors such as smoke exposure to decrease the risk of CHDs.

3. What education would the nurse provide to the patient about medications to avoid during pregnancy that could increase the risk of a CHD?

Certain medications can put infants at higher risk for CHDs. Medications that are contraindicated during pregnancy are called teratogens.

Thalidomide is a teratogen that is used to treat multiple myeloma and certain skin conditions. Research shows that thalidomide is associated with severe birth defects including CHDs. Isotretinoin is another medication contraindicated in pregnancy that is used to treat severe cystic acne. Other medications that are considered teratogens include certain anticonvulsants, antiarrhythmics, and antidepressants.

Section 8: Case Study #2

A mother brings her 3-month-old infant into the emergency department stating “something is wrong with my baby”. The triage nurse performs an initial assessment and notices that the patient is tachypneic and hears a heart murmur. The mother states that the infant seems to get tired while breastfeeding and always seems to be hungry even right after feeding. The consulting cardiologist tells the mother that a ventricular septal defect (VSD) is initially suspected. The cardiologist orders an ECHO to confirm the diagnosis and the patient is admitted for further evaluation and treatment.

1. Based on the available information, why would a non-cyanotic CHD be most likely?

2. What education should be provided to the mother about possible interventions for a VSD?
3. What education should be provided to the mother about lifelong effects of her child's diagnosis?

Section 9: Case Study #2 Review

This section will review the case studies that were previously presented in each section. Responses will guide the clinician through a discussion of potential answers as well as encourage reflection.

1. Based on the available information, why would a non-cyanotic CHD be most likely?

Non-cyanotic CHDs often have minimal symptoms and diagnosis can be delayed due to the lack of symptoms. Common symptoms of non-cyanotic CHDs include a heart murmur, tachypnea and dyspnea, fatigue when feeding, and poor weight gain. A VSD is the most common type of CHD and is commonly found during routine newborn exams by detecting a heart murmur.

2. What education should be provided to the mother about possible interventions for a VSD?

Treatment for VSDs depends on the defect's size, location, the age at diagnosis, and other health conditions. In infants, healthcare providers often monitor the VSD to see if the hole closes on its own. Medications may be prescribed to treat any present symptoms, but medications will not repair the defect. The only way to repair the defect is through surgical intervention. Surgery may be recommended if the hole is large or if serious

symptoms are present.

3. What education should be provided to the mother about lifelong effects of her child's diagnosis?

After the initial surgery or if the VSD closes on its own, patients have a good long-term prognosis and rarely need additional medical interventions. A VSD can cause a leak in the aorta which can have serious consequences. Pediatric cardiologists will monitor patients with VSDs for symptoms of a leaky aorta or other serious symptoms. Patients may need surgery to repair VSDs and prevent further complications.

Section 10: Case Study #3

The nurse is caring for a 10-day old infant who has been diagnosed with Tetralogy of Fallot. The patient has a heart murmur and fatigue while feeding. The nurse informs the mother that the plan of care is to keep the patient in the hospital until they are strong enough for surgery. The mother states "I just cannot imagine my baby having surgery. They are so tiny and I am terrified that something will go wrong." The mother also states that she is worried about the baby when they are crying. She states "The alarms in the room always go off when my baby starts crying. They start to look blue, and I don't know what to do."

1. What education would the nurse provide to the mother about the surgical interventions for Tetralogy of Fallot?
2. What symptom of Tetralogy of Fallot would the nurse suspect when the mother states "The alarms in the room always go off when my baby starts crying. They start to look blue, and I don't know what to do"?
3. What other symptoms of Tetralogy of Fallot would the nurse want to be

sure the mother is aware of?

Section 11: Case Study #3 Review

This section will review the case studies that were previously presented in each section. Responses will guide the clinician through a discussion of potential answers as well as encourage reflection.

1. What education would the nurse provide to the mother about the surgical interventions for Tetralogy of Fallot?

Multiple surgeries are needed to repair the multiple defects involved in Tetralogy of Fallot. A complete repair of the heart is completed in several steps which helps patch the VSD and repair or replace the pulmonary valve. A complete repair will usually help resolve the right ventricular hypertrophy as the heart no longer has to work hard to pump blood through the right ventricle. These surgeries will need to be spaced out to ensure the patient is strong enough for each surgery.

2. What symptom of Tetralogy of Fallot would the nurse suspect when the mother states “The alarms in the room always go off when my baby starts crying. They start to look blue, and I don’t know what to do”?

“Tet spells” are a common symptom of Tetralogy of Fallot. These instances involve a rapid drop in oxygen saturation levels causing cyanosis and usually occur when infants are crying or eating. Cyanosis is the lack of oxygen circulating in the body which causes the bluish color of the skin. Tet spells commonly occur during infancy and usually begin to decrease after 4 to 5 years of age.

3. What other symptoms of Tetralogy of Fallot would the nurse want to be

sure the mother is aware of?

In addition to “tet spells”, symptoms of right heart obstructive defects are similar to symptoms of left heart obstructive defects. These include heart murmur, dyspnea, cyanosis, low oxygen saturation levels, fatigue, and poor feeding. Some patients may not have all of these symptoms, but it is important to be aware of all of the symptoms in order to prevent further complications.

Section 12: Case Study #4

The nurse is caring for a 5-year-old patient diagnosed with a congenital heart defect (CHD). The patient has had multiple surgeries and is coming to see the cardiologist for a follow up appointment. The nurse notices the parents constantly checking the time and looking anxious. The nurse asks the parents if there is anything they can do and the mother bursts into tears. The parents state “We just cannot keep up with all the appointments. We are already late for physical therapy and have several other appointments this week.” The parents state that they were recently told that the patient may have a speech delay and may not be able to start school next month.

1. What may be some reasons the parents are struggling with their child’s diagnosis?
2. What education can the nurse provide to the parents about coping strategies for caregivers of children with CHDs?
3. What information can the nurse provide to the parents about coping strategies for the patient as they grow and continue to cope with their chronic health conditions related to the CHD?

Section 13: Case Study #4 Review

This section will review the case studies that were previously presented in each section. Responses will guide the clinician through a discussion of potential answers as well as encourage reflection.

1. What may be some reasons the parents are struggling with their child's diagnosis?

Children with CHDs require frequent hospitalizations, therapy appointments, medications, and other medical care. Children often miss school and cannot participate in as many extracurricular activities due to medical restrictions. The patient has already had multiple surgeries and may need more in the future. Caring for a child with a CHD can cause stress and financial strain for caregivers. Research shows that parents of children with a CHD report higher levels of stress, anxiety, and depression. The parents are struggling to manage the patient's frequent appointments and outpatient care. The parents are also dealing with the patient's potential speech delay and uncertainty of attending school. Parents often grieve the lifestyle they thought their child would have and can have a difficult time accepting their child's limitations.

2. What education can the nurse provide to the parents about coping strategies for caregivers of children with CHDs?

It is important to help families develop effective coping strategies when caring for a child with a CHD. Parents may be struggling to accept their child's limitations and the financial and emotional stress of ongoing medical needs can contribute to daily stress. It is important for caregivers to be aware of coping strategies including spirituality, a good support system, physical activity, cognitive therapy, and self care activities. Effective coping

strategies can help caregivers cope with the daily needs of a child with a CHD and help the family adjust to the needs of the child. It is important for healthcare providers to care for the patient and the family. Healthcare providers can be an important resource for the family when navigating big life changes such as caring for a child with a CHD.

3. What information can the nurse provide to the parents about coping strategies for the patient as they grow and continue to cope with their chronic health conditions related to the CHD?

Managing mental health is an important part of a healthy lifestyle for patients with CHDs. Children and adults may experience anxiety, depression, and stress when dealing with lifelong complications from CHDs. Developing healthy coping skills is essential to maintaining a healthy lifestyle. Healthcare providers can help patients find coping strategies such as cognitive therapy, self care activities, and a support system. Not every coping skill will be effective for each patient, so it is important for healthcare providers to develop an individualized plan for each patient.

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