Congenital

Heart Diseases

Etiology

- **The causes of congenital heart disease are unknown in 90% of cases; they are very likely multifactorial with genetic and environmental inputs.**
- **5% of cases are associated with chromosomal abnormalities**

Etiology

Less than 1% of congenital defects are clearly environmental:

- maternal rubella in the first trimester
- Excessive alcohol consumption
- Excessive cigarette smoking
- Thalidomide

Etiology

- **The most critical juncture is embryologic cardiac development in gestational weeks 3- 8.**

Clinical consequences

Children with significant congenital anomalies have:

- Hemodynamic sequel
- Failure to thrive
- Retarded development
- cyanosis

Clinical consequences

- Increased risk of chronic or recurrent illness
- Infective endocarditis (due to abnormal valves or to endocardial injury from jet lesions)

The various congenital anomalies are two types: shunts

obstructions

Shunts

- **Denotes abnormal communication between heart chambers, between vessels, or between chambers and vessels. Depending on pressure relationships, blood may be shunted from left to right (more common) or right to left**

Right - to – left shunts

- **Right-to-left shunts (cyanotic congenital heart disease) cause cyanosis from the outset as poorly oxygenated blood passes into the systemic circulation. They also permit emboli from venous sources to pass directly into systemic circulation (paradoxic embolism)**

Left – to – right shunts

- **Left-to-right shunts include chronic right heart overload with secondary pulmonary hypertension and right ventricle hypertrophy, but eventually right-sided exceeds left-sided pressure and the shunt becomes right to left. Cyanosis appears late.**

Left – to right shunt

- **Once significant irreversible pulmonary hypertension develops, the structural defects of congenital heart disease are considered irreversible**

Shunts

Secondary findings in long-standing cyanotic heart disease include:

- clubbing of the fingers and toes
- hypertrophic osteoarthropathy
- polycythemia

Obstructions

Typically:

- coarctation
- Valvular stenoses
- or atresias
- **These do not cause cyanosis**

Left-to right shunts:late cyanosis

- Atrial septal defect
- Ventricular septal defect
- Patent ductus arteriosus

Atrial Septal Defect (ASD)

- **Definition :** a secundum atrial defect is a hole in the septum primum (at the site of the foramen secundum) not covered by the septum secundum
- Anatomy: the defect is high in the atrial septum, and may vary from dime size to virtual absence of the atrial septum.

Physiology

- In the vast majority instances, the shunt across to defect is from the left to right atrium in diastole. The two atria act as a single filling chamber with identical pressures if the hole is at least 1 cm in diameter; the flow in diastole is toward the ventricular chamber, which is thinner walled and more compliant,i.e., RV

Physiology ASD

- The pulmonary arterial presser is normal in spite of the huge flow, owing to the distensibility of the normal pulmonary arterioles. Increased pressure work is a late phenomenon if arteriolar obstruction appears, at which time to shunt becomes bidirectional or even net right-to-left (pulmonary hypertension)

Diagnosis

- Patients with secundum atrial defect do not become symptomatic until childhood or adolescence. This phenomenon may explain the late discovery of the malformation.
- The symptoms consist principally of failure to thrive, dyspnea, and palpitations

Physical Examination

- Patient is usually a tall, thin girl (almost 2:1)
- Cyanosis is rare and almost invariably indicates right ventricular outflow tract obstruction (pulmonary stenosis or pulmonary vascular obstructive disease)
- Jugular veins are strongly pulsatile

Physical examination

- Left chest prominence

  Auscultation: the first sound tends to be loud;
- Almost pathognomonic feature is a widely split second sound with a pulmonary closure of normal intensity, barely moving with respiration

Auscultation

- There is a soft ejection murmur at the second left interspace (louder if there is associated pulmonary stenosis)
- And a low-frequency early diastolic rumble at the lower left sternal border
- The sounds, the murmurs, and palpable impulse with left chest prominent all indicate a hyperkinetic circulation

Electrocardiography (ECG)

- Right ventricular hypertrophy
- Severe right ventricular hypertrophy indicates obstruction of the right ventricular outflow tract

Radiography

- The chest film shows mild-to-moderate right ventricular and right atrial enlargement with pulmonary vascular engorgement and a prominent main pulmonary artery segment.
- The sinus venosus defect is characterized by the absence of the right superior vena cava shadow and entrance of the horizontal pulmonary vein into the right upper cardiac shadow

Echo-Doppler Study

- The Doppler echocardiogram, particularly in color, gives a good estimate of the size and direction of the shunt
- A warning note should be sounded here lest a patient be referred for surgery on the basis of a false-positive echocardiogram without supporting data from the physical examination,chest film,and ECG

Management

- It has been to close surgically all clinically significant secundum atrial defects any time on diagnosis
- Patients operated on in childhood and early adulthood may look forward to normal lives

Ventricular Septal Defects (VSD)

- **Definition:** an opening in the ventricular septum that allows communication between the right and left ventricles
- **Anatomy:** the defect are of variable size and may be located in any part of the ventricular septum as single or multiple lesions.

Anatomy VSD

Location of VSD:

- Muscular (defect in the lower trabecular septum)
- Perimembranou
- Subpulmonary

Physiolodgy VSD

- A ventricular septal opening allows shunting of left ventricular blood into the right ventricle. The amount of shunting depends on the size of the defect and the relative pulmonary and systemic resistances. A large left-to-right shunt is associated with increased water in the lung, accounting for the symptom of tachypnea.

Diagnosis

- A ventricular septal defect is most often detected by the discovery of a murmur on routine examination. The absence of a murmur at birth, and its appearance a few days later, is characteristic of ventrilar septal defects. By contrast, the murmur of infundibular pulmonary stenosis (which is virtually identical on auscultation) is heard at birth

Physical Examination

- An infants with a small ventricular septal defects and other cardiac problems appears normal. There is loud murmur, usually loudest at the lower left sternal border.
- The infant with a large ventricular defect is often scrawny, with discordant height and weight, although both measure may be below the fifth percentile.

Physical Examination

- **Tachypnea as high as 100 breaths/min is common**
- **Peripheral pulses are small**
- **Liver is often enlarged**
- **Cardiac impulse is visibly**

Auscultation

- Pansystolic murmur
- Loudest at the lower left sternal border
- Without treatment, the heart rate is fast and a gallop rhythm may be present at the apex
- The gallop sound is diastole at a fast heart rate may become a mid-diastolic rumble as the heart slows with digoxin therapy
- Pulmonary rhonchi and rales are common

Electrocardiography (ECD)

- In the patient with a small ventricular septal defect, the ECD is normal
- With increasing larger defects, there is, first, left ventricular hypertrophy, and then, with the largest defects, both ventricular hypertrophy

Radiography

- Both the heart size and the pulmonary vasculature are normal in infants with small ventricular defects
- With larger defects, the heart is proportionally large, with increased pulmonary vascularity
- The left atrial shadow may be large.
- Pneumonitis, atelectasis, or aspiration may be evident

Echo-Doppler Study

- A Doppler search for additional associated ventricular septal defects should be made in all cases.
- Differences between right ventricular and left ventricular pressures are recorded and possible pressure gradients between the ight ventricle and pulmonary artery are estimated

Management

- Small ventricular defects. The majority of patients with ventricular septal defects are and remain asymptomatic because they defects are small. Only 15% of all patients with a VSD require surgical intervention; even among the symptomatic infants, only 30% come to surgery.

Management of the Large Ventricular Defect

- The most common problems are congestive heart failure and failure to thrive.
- Congestive heart failure is managed initially with digoxin and diuretics, but only rarely is there more than minimal relief of symptoms
- Surgical correction is desirable before right heart overload and pulmonary vascular disease develop

Patent Ductus Arteriosus (PDA)

- **Definition.** In the fetus, the ductus arteriosus permits blood flow between the aorta(distal to the left subclavian artery) and the pulmonary artery
- At term, and under the influence of relatively high oxygen tension and reduced local PG synthesis, muscular contraction closes the ductus within 1 to 2 days of life.

PDA

- Persistent patency beyond that point is generally permanent.
- About 85% to 90% of PDAs occur as isolated defects. The length and diameter (up to 1 cm) are variable.
- There is associated left ventricular hypertrophy and pulmonary artery dilatation

PDA

- Although initially asymptomatic, and notable only for a prominent heart murmur (described as “machinery-like”), long-standing PDA induces pulmonary hypertension with subsequent right ventricular hypertrophy and finally right-to-left shunting to produce late cyanosis

PDA

- Early closure of a PDA (either surgically or with prostaglandin administration in otherwise normal infants) is therefore advocated

Right-to-left Shunts: Early Cyanosis

- Tetralogy of Fallot
- Transposition of the great vessels
- Truncus arteriosus

Tetralogy of Fallot

- Ventricular septal defect (VSD)
- Dextroposed aorta overriding the VSD
- Pulmonic stenosis with right ventricular outflow obstruction
- Right ventricular hypertrophy
- Cyanosis is present from birth or soon after

Diagnosis

- Newborns (often) and children (less commonly) may be admitted with evidence of left-side failure, indistinguishable from that seen in patients with large ventricular defect. These patients have only mild pulmonary stenosis at this time

Diagnosis of tetralogy Fallot

- Older children and adults with tetralogy of Fallot who have not undergone surgical repair almost never show evidence of congestive heart failure unless there are complicating factors (bacterial endocarditis, anemia, aortic regurgitation). They have cyanosis and exercise intolerance of varying degrees; they have moderate or severe pulmonary stenosis.

Diagnosis of tetralogy Fallot

- They are cyanotic and have clubbing of the fingers and toes; after running, even walking, they may assume a squatting position

Diagnosis

- Hypercyanotic spells occur mostly in infants; these consist of uncontrollable crying with increasing cyanosis, tachycardia, tachypnea, leading occasionally to unconsciousness, and sometimes even to a cerebral vascular accident.

Diagnosis

- The frightening part of these “spells”is that they may occur in otherwise healthy looking, pink infants. The conventional explanation, based on less than unassailable evidence,is that the attacks are due to infundibular spasm. Usually, these attacks occur in the morning and last from several minutes to an hour or more

Physical Examination

- Results of the physical examination of patients having tetralogy with mild pulmonary stenosis are virtually indistinguishable from those for patients with large ventricular defects. In patients with moderate-to-severe pulmonary stenosis, cyanosis and clubbing dominate the picture.

Physical Examination

- A systolic thrill may be palpable at the left sternal border, transmitting to the suprasternal notch, but usually not to the carotids.
- On auscultation: is usually an apical click (large aorta), a single loud second sound,and grade IV-VI systolic murmur at the lower left sternal border transmitting well to the suprasternal notch

Physical Examination

- No diastolic murmur is heard

Minor laboratory tests:

- Increased hematocrit (50-75%) is characteristic of cyanotic heart disease in children and adults.

Minor laboratory test

- Severe polycythemia, unusual in infancy, may increase the viscosity of the blood to a level that would impede oxygen delivery to tissues; the usual manifestation of this are symptoms of the CNS (dizziness, headaches, blackouts).

Electrocardiography

- ECG always shows right ventricular hypertrophy often associated with peaked P waves (P pulmonale)

Radiography

- The film in a patient with a right – to- left shunt shows a normal-sized heart with right ventricular contour (“boot-shaped”, like a Dutch wooden shoe), a large aorta (right aortic arch in 20%), and normal or decreased pulmonary vasculature. The main pulmonary artery segment on the left border of the heart is diminished and may even be concave

Echo – Doppler Study

- The echo-Doppler study demonstrates the subaortic ventricular defect and the infundibular stenosis, with establish the morphologic diagnosis of tetralogy of Fallot.

Cardiac Catheterization

- Cardiac catheterization with angiography provides the morphologic and physiologic details and is usually performed prior to surgical repair.

Management

- The treatment for tetralogy of Fallot is surgery. The questions remaining are the nature and limiting of the operation. There are distinct indications even today for Blalock-Taussig shunts in the treatment of tetralogy of Fallot: a) conus coronary artery, and (b) as an emergency measure for severe hypoxic spells, not manageable by medical means.

Transposition of Great Arteries

- Origin of the aorta from the right ventricle and pulmonary artery from the left ventricle. The patient with transposition of the great arteries has two parallel circulations; blood may recirculate repeatedly through the same side of the heart before returning either to the systemic or the pulmonary capillaries.

Continuation

- The unoxygenated systemic venous return passes through the right ventricle to the aorta and back to the systemic capillaries. The oxygenated pulmonary venous return passes through the left ventricle and returns to the lungs. It is obvious that the transposed circulation is incompatible with life unless there is communication between the two circuits (VSD, ASD)

Obstructive congenital anomalies

- **Coarctation of Aorta –** is a narrowing or constriction of the aortic isthmus. It is a common, potentially fatal congenital cardiac malformation, clearly progressive through the years. 50% occur as isolated defects, the remainder with multiple other anomalies.

Coarctation of aorta

- Clinical manifestations depend on the location and severity of the constriction. Most occur lust distal to the ductus/ligamentum arteriosus (postductal)
- Preductal coarctation manifests early in life and may be rapidly fatal. Survival depends on the ability of the ductus arteriosus to sustain blood flow to the distal aorta and

Continuation

and lower body adequately. Even then, there tends to be lower body cyanosis. This form usually involves a 1-to-5 cm segment of the aortic root and is often associated with fetal RV hypertrophy and early right heart failure.

Continuation

- Postductal coarctation is generally asymptomatic unless very severe. It usually leads to upper extremity hypertension but low flow in the lover extremities, causing arterial insufficiency (claudication, cold sensitivity). Collateral flow around the coarctation generally develops, with intercostal rib notching(noted on X-ray views)

Contunuation

and internal mammary and axillary artery dilation

Management. The treatment of complex corctation is surgical:

- resected and end-to-end anastomosis
- aortoplasty, using the left subclavian artery
- balloon dilation of uncomplicated coartation