Congenital Heart defects

Learning objectives:

1. **To know the etiology and classification of CHD**

2. **To understand the clinical features and diagnosis of CHD**

3. **How to treat patients with CHD**

Etiology: 1. Multifactorial inheritance

2-4%.

2. **Medications or alcohol or drug abuse during pregnancy (anticonvulsant, warfarin, cocaine,).**
3. **Maternal viral infection, such as rubella (German measles) in the first trimester of pregnancy.**
4. **Maternal diseases (diabetes mellitus, SLE)**
5. **Genetic or chromosomal abnormalities such as Down syndrome, Turner syndrome, Noonan, William, and Marfan syndrome.**

Non-cyanotic:

- **Ventricular septal defect (VSD), Atrial septal defect (ASD), Patent ductus arteriosus (PDA), Aortic stenosis, Pulmonic stenosis, Coarctation of the aorta, Atrioventricular canal (endocardial cushion defect)**

Cyanotic:

- **Tetralogy of Fallot , Transposition of the great vessels , Tricuspid atresia ,Total anomalous pulmonary venous return ,Truncus arteriosus , Hypoplastic left heart , Hypoplastic right heart , Ebstein's anomaly.**

Ventricular septal defect

Definition

VSD is a developmental defect of the interventricular septum whereby a communication exists between the cavities of the two ventricles.

Classifications of subtypes

1. **Perimembranous (infracristal) VSDs: They are the most common types of VSDs and account for 80% of defects**

2. **Supracristal defects comprise 5-8% of isolated VSDs**

3. **Muscular VSDs (trabecular)**

Frequency:

VSD is the most common congenital cardiac defect and account for 30 % of all lesions.

Sex: VSDs are slightly more common in females; 44% occur in males, and 56% occur in females.

Clinical presentations:
Symptoms and physical findings relate to the size of the defect and the magnitude of the left-to-right shunt.

- **Infants with small defects <5 mm ○ Mild or no symptoms**

- **Infants with moderate defects 5-10 mm**

    - **Tachypnea with increased respiratory effort, excessive sweating, and fatigue when feeding, a detailed feeding history is important.**

    - **Respiratory infections may precede these symptoms. Infants often have failure to thrive.**

- **Infants with large VSDs :> 10 mm Symptoms and signs are similar to but are more severe than those observed with moderate defects. ○**

- **Eisenmenger syndrome (VSD with severe pulmonary vascular obstruction): Symptoms include exertional dyspnea, chest pain, syncope, and hemoptysis. Cyanosis due to right to left shunting.**

Physical findings are :

- **Precordial activity is accentuated**
- **Holosystolic harsh murmur is most prominent over the lower left sternal border (LSB).**

- **A prominent S_3 with a short early middiastolic rumble is audible at the apex when pulmonary blood flow is twice systemic blood flow or greater.**

- **Infants with Eisenmenger physiology:**

  - **Cyanosis, Clubbing, Erythrocytosis, Prominent RV lift, maximal in xiphoid region revealed on palpation, Very short or no systolic murmur, Possible short pulmonary ejection murmur along upper left parasternal region Loud harsh holosystolic murmur in a patient in this physiological group Indicates tricuspid insufficiency**

  - **S_2 - Loud, single, or closely split, S_3 of right ventricular origin - May be present, Squatting in 15% of patients**

. **Hemoptysis: Hemoptysis occurs in 33% of patients.**

Diagnosis:

Imaging Studies:

- **Cardiac enlargement with increased pulmonary vascular markings**
- **The main pulmonary artery segment is prominent.**
- **Enlarged LA)**

- **Echocardiography**

- **Electrocardiography LVH, left axis deviation**

- **Catheterization:**

Treatment:

- **Children with small VSDs are asymptomatic and have excellent long-term prognoses. Neither medical therapy nor surgical therapy is indicated. Antibiotic prophylaxis against endocarditis should be provided at the time of dental or surgical procedures likely to produce bacteremia.**

- **In children with moderate or large VSDs:**

  Furosemide, captopril, and digoxin, is indicated for symptomatic congestive heart failure.

- **Furosemide in a dosage of 1-3 mg/kg/d in 2 or 3 divided doses is used.**
- **Captopril in a dosage of 0.1-0.3 mg/kg 3 times daily can be useful to reduce systemic afterload.**
- **Digoxin in a dosage of 0.005-0.01 mg/kg/day may be indicated if diuresis and afterload reduction do not relieve symptoms adequately.**

- **Indications for surgical repair**
  - **Uncontrolled congestive heart failure.**
  - **Surgical repair is indicated in older asymptomatic children with normal pulmonary pressure if pulmonary to systemic flow is greater than 2:1.**
  - **Supracristal VSD: Early repair may prevent progression of aortic insufficiency.**

Complications:

- **Eisenmenger complex.**
- **Secondary aortic insufficiency**
- **Right ventricular outflow tract obstruction:**
- **Subaortic obstruction.**
- **Infective endocarditis**

Prognosis:

Children with small VSDs are asymptomatic and have excellent long term prognoses.

Closure is earlier and most frequently observed in muscular defects (80%) followed by perimembranous defects (35-40%) in the first few years of life.

ATRIAL SEPTAL DEFECT

Types of atrial septal defects:

1. Ostium secundum defect, the most common type.

2. Ostium Primum defect.. ostium primum defects are virtually always associated with a cleft in the anterior mitral valve leaflet

3. Sinus venosus

4. Coronary sinus

ASD occur in approximately 7% of these children with CHD , The female to-male ratio is approximately 2:1.

History

- **Infants and young children with ASDs typically are asymptomatic.**
- **Most ASDs are diagnosed after a suspicious murmur is detected during a routine health-maintenance examination.**

Physical

- **A midsystolic pulmonary ejection murmur.**
- **Fixed splitting of S2.**
- **A large shunt increases flow across the tricuspid valve, and the patient with ASD is likely to have a middiastolic rumble at the left sternal border.**

Diagnosis

- **Plain radiographic findings in ASD are nonspecific but include right atrial and right ventricular dilatation, pulmonary artery dilatation, and increased pulmonary vascular markings.**
- **Echocardiography**

- **ECG: right-axis deviation, right ventricular hypertrophy, Left-axis deviation suggests an ostium primum-type ASD**
- **All types of ASD can result in prolonged PR intervals. This prolongation of internodal conduction may be related to the increased size of the atrium and a long internodal distance.**

- **Cardiac catheterization is rarely necessary in the preoperative evaluation of a child with ASD.**

TREATMENT

Medical therapy is of no benefit in children with asymptomatic ASDs.

1. **Surgery is ideally performed in children aged 2-4 years, even in asymptomatic patients to prevent adult hood complications like atrial arrhythmias.**

2. **Closure through cardiac catheterizations.**

Prognosis

- **The prognosis for a child with ASD is good; the rate of surgical mortality is less than 1%.**
- **Approximately 15% of ostium secundum defects close spontaneously. Spontaneous closure does not occur with the other types of ASDs.**

A few patients with an ostium primum ASD and an abnormal mitral valve may require a second operation for mitral valve dysfunction later in their lives.

Patent ductus arteriosus (PDA)

The main function of the ductus arteriosus is that during fetal life it shift the blood from the pulmonary artery to the descending aorta, the patency is maintained by the local secretion of PGE2.

Normally, functional closure of the ductus arteriosus occurs by about 15 hours of life in healthy infants born at term.

True anatomic closure, where the ductus loses the ability to reopen, may take several weeks.

The female-to-male ratio is 2:1, it is more common in premature neonates.

CLINICAL PRESENTATIONS:

1. SMALL PDA: The infant is asymptomatic, and on examination there is machinery continuous murmur at the left second intercostal space.

2. medium size : the symptoms appear at 2-5 month of life slow or difficult feeding , repeated chest infections, and failure to thrive

Exam. Shows collapsing pulses, wide pulse pressure (because of decrease diastolic pressure), left ventricular enlargement, systolic thrill and machinery murmur at the 2nd left intercostal space radiating to the back.

3. large size: the symptoms appears since birth which are similar to the moderate type but are more severe, the murmur may be only systolic type , with signs of sever heart failure..

Investigations:

- **Chest radiography**
- **Doppler echocardiography**

- **Electrocardiogram**

Treatment:

The premature neonate with a significant PDA usually is treated with intravenous indomethacin. This has been quite successful in most patients. Recently, intravenous ibuprofen.

Many dosage regimens exist, and dose of indomethacin is dependent on postnatal age (PNA) at time of first dose; one example is as follows:
PNA <48 hours: 0.1 mg/kg IV q12h for 3 doses

PNA 2-7 days: 0.2 mg/kg IV q12h for 3 doses
PNA > 7 days: 0.25 mg/kg IV q12h for 3 doses

- **In the symptomatic neonate, diuretics and cautious fluid restriction may be sufficient for initial therapy if symptoms are mild and the baby is not extremely premature. Spontaneous closure is common. If significant respiratory distress or impaired systemic oxygen delivery is present, Intravenous indomethacin (or the new preparation of intravenous ibuprofen) is frequently effective in closing a PDA if it is administered in the first 10-14 days of life.**
- **After the first birthday, the most common treatment for a PDA is occlusion at cardiac catheterization.**

Surgical Care

Surgical ligation or surgical ligation and division remain the standard treatment of large PDAs that require treatment in infancy.

Complications

- **Endocarditis**
- **Congestive heart failure**
- **Pulmonary vascular obstructive disease**
- **Aortic rupture**

Prognosis

- **Typically, following PDA closure, patients experience no further symptoms and have no further cardiac sequelae.**
- **Premature infants who had a significant PDA are more likely to develop bronchopulmonary dysplasia.**

Tetralogy of Fallot

Tetralogy of Fallot may account for 10-15% of all congenital (newborn) heart defects.

- **Children with TOF typically can be born with 4 abnormalities of the heart:**
    - **Obstruction of the right ventricular outflow tract.**
    - **Ventricular septal defect-VSD: It's usually large and located below the aortic valve.**
    - **Enlargement of the right ventricle (hypertrophy):**
    - **Over-riding of the ventricular septum by the aorta.**

Symptoms:

. Cyanosis

- **Failure to thrive.**
- **Episodes of extreme blue coloring (called Tet spells) usually occur in the first 2-3 years of life. The child suddenly becomes blue, has difficulty breathing, and may show such symptoms as irritability or even fainting ,syncopal attack,convulsions,or even loss of conciosness,coma and death in severe cases,. The spells occur in 20-70% of cases. They often happen during feeding, crying, straining, or on awakening in the morning. Spells can last from a few minutes to a few hours.**

If your child starts to turn blue, place the child on his or her back in the knee to-chest position (squatting position.).

- **Physical examination**
    - **Children with Tetralogy of Fallot usually will be short in stature and thin. The child's fingers may be stubby (clubbed).**

    . A second heart sound usually will be single and will represent closure of the aortic valve.

    Ejection systolic murmur at the pulmonary area. .

- **Diagnosis:**
    - **Polycythemia (high PCV, and Hb.).**
    - **ECG: This heart imaging test may show enlargement of the right ventricle.**
    - **Chest x-ray: This image may demonstrate the classic "boot shaped heart.**
    - ○ **Echocardiography:**
    - **Cardiac catheterization**

Complications of untreated TOF:

1. **CVA USUALLY <2 YEARS OF LIFE.**
2. **BRAIN ABSCESS >2 YEARS OF LIFE.**
3. **GOAT**
4. **ARRITHROPATHY AND CLUBBING**
5. **PSYCHOLOGICAL SEQUELI**
6. **LOW GRADE DIC**
7. **GUM DISEASE**
8. **HEMOPTYSIS**
9. **PREGNANCY COMPLICATIONS.**

Treatment:

Venesection, Inderal orally, iron supplement,

Hypercyanotic (Tet) spells

Squatting position

Oxygen, morphine, propranolol (or metoprolol), or in extreme cases, phenylephrine). These medications decrease the frequency and severity of the Tet spells.

Surgery is the primary way to correct the heart problem.

- **The Blalock-Taussig operation (palliative surgery).**
- **Total correction years of life.**
- **The operation has a surgical death rate of about 1-5%.**

Transposition of the great arteries

Transposition of the great arteries (TGA) is the most common cyanotic congenital heart lesion presenting in the neonate. The hallmark of TGA is ventriculoarterial discordance, in which the aorta arises from the morphologic right ventricle and the pulmonary artery arises from the morphologic left ventricle.

The following are 3 common anatomic sites for mixing of oxygenated and deoxygenated blood in TGA:

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

One or all of these lesions can be present in d-TGA, and the degree of arterial hypoxemia depends on the degree of anatomic mixing.

Frequency:

- **This lesion presents in 5-7% of all patients with congenital heart disease.**

Sex: TGA has a 60-70% male predominance.

Clinical:

History:

- **Infants with TGA usually are born at term, with cyanosis apparent within hours of birth.**

- **The clinical course and manifestations depend on the extent of intercirculatory mixing and the presence of associated anatomic lesions.**

    - **TGA with intact ventricular septum: Prominent and progressive cyanosis within the first 24 hours of life is the usual finding in infants.**

    - **TGA with large ventricular septal defect**
        - **Infants may not manifest symptoms of heart disease initially, although mild cyanosis (particularly when crying) is often noted.**
        - **Signs of congestive heart failure (tachypnea, tachycardia, diaphoresis, and failure to gain weight) may become evident over the first 3-6 weeks as pulmonary blood flow increases..**

Physical: TGA with intact ventricular septum

- **Infants typically present with progressive central cyanosis. Other than cyanosis, the physical examination is often unremarkable.**

- **TGA with large ventricular septal defect**

    - **Cyanosis may be mild initially, although it is usually more apparent with stress or crying. At presentation, infants often have an increased right ventricular impulse, a prominent grade 3-4/6 holosystolic murmur, third heart sound, mid-diastolic rumble, and a gallop rhythm.**

    - **Hepatomegaly may be present.**

Imaging Studies:

- **Chest radiography**

    - **The chest radiograph may appear normal in newborns with TGA and intact ventricular septum but may demonstrate the classic "egg on a string" appearance in approximately one third of patients. With a ventricular septal defect, cardiomegaly usually occurs with increased pulmonary arterial vascular markings.**

- **Echocardiography**

- **Cardiac catheterization**

.Medical Care:

- **Initial treatment consists of maintaining ductal patency with continuous IV prostaglandin E_1 infusion to promote pulmonary blood flow,**
- **Cardiac catheterization, The balloon atrial septostomy is used to increase the atrial level shunt and to improve mixing in patients with intact ventricular, And atrial septum.**

- **For the ill neonate, metabolic acidosis should be corrected with fluid replacement and bicarbonate administration.**
- **Treatment of congestive heart failure.**

- **Mechanical ventilation may be necessary if pulmonary edema develops in concert with severe hypoxemia.**

- **Ultimately, the patient requires surgical repair or palliation early in life.**

Surgical Care:

- **An arterial switch procedure.**

Reference:

- **Nelson Textbook of Pediatrics 20th 2015 .**