Congenital Heart Diseases

By
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Congenital Heart Disease

• Most common type of heart disease among children
  a) ~ 1% of live births
  b) Causes in most cases are unknown; but
     - ~ 10% genetic: trisomy 21 (Down syndrome)
     - congenital defect in parent or sibling is greatest risk factor
- Types:
  
a) Left to Right shunt

b) Right to Left shunt

c) Obstructions
1- Left to Right shunts

a) ASD, VSD, PDA
   i) ↑ pulmonary blood flow (ASD): no cyanosis

b) ↑ RV pressures and Vol. (VSD,PDA)
   i) hypertrophy
   ii) ↑ pulmonary vascular resistance (PVR)
Left to Right Shunts

- Most common: ventricular septum defect (VSD),
- Atrial septum defect (ASD), Patent ductus arteriosus (PDA)
- and atrio-ventricular defect (AVSD)
i) VSD most common
   - close spontaneously (50%)

ii) ASD usually not symptomatic before 30 yrs
iii) **PDA:** DA remains open after birth

- ~ 90% occur as isolated anomaly

- reversal of flow with $\uparrow$ PVR causes cyanosis

iv) **Complete atrioventricular canal defect**

- all 4 chambers freely communicate (Down syndrome).
2- Right to Left shunt

a) ↓ pulmonary blood flow → Cyanosis (blueness of skin).

b) examples:

i) tetralogy of Fallot
ii) great vessel transposition
iii) truncus arteriosus
iv) tricuspid atresia
v) anomalous pulmonary venous connection
long standing cyanosis is associated with “clubbing” of the tips of the fingers and toes
Tetralogy of Fallot

Most common form of cyanotic congenital heart disease

- **Defects:**
  - a) VSD
  - b) Pulmonary artery stenosis
  - c) aorta that overrides VSD
  - d) RV hypertrophy
Tetralogy of Fallot

Normal Heart
- Right atrium
- Pulmonary artery
- Right ventricle
- Left atrium
- Aorta
- Left ventricle

Heart with Defects Found with Tetralogy of Fallot
- Narrowing of pulmonary passageway
- Misaligned aorta
- Ventricular septal defect
- Enlarged right ventricle
Transposition of the Great Arteries (TGA)

a) Aorta arises from right ventricle

b) Pulmonary artery arises from left ventricle

c) total separation of pulmonary and systemic circulations

   i) Need a shunt to survive following birth

      - VSD → stable shunt
Transposition of the Great Vessels

Normal

Transposition of the Great Vessels
Truncus Arteriosus

• Failure of separation into aorta and pulmonary artery
  
a) results in single great artery
    
i) receives blood from both ventricles
      - early systemic cyanosis
  
b) accompanying VSD
  
c) ↑ pulmonary blood flow
    
i) danger of irreversible pulmonary hypertension
Tricuspid Atresia

• Clinical Presentation:
  a) Cyanosis is almost always present at birth and is progressive.
  b) Etiology/Pathophysiology:
    i) Due to absence of the tricuspid valve.
    This leads to an inter-atrial right to left shunt, usually through a patent foramen ovale.
ii) It is classified by the absence of tricuspid valve, pulmonary stenosis, and VSD.

iii) The most common form is also associated with a hypoplastic right ventricle.
TRICUSPID ATRESIA

1 - Atrial Septal Defect
2 - Absent Tricuspid Valve
3 - Ventricular Septal Defect

Blood is shunted through an atrial septal defect to the left atrium and through the ventricular septal defect to the pulmonary artery. The shaded arrows indicate mixing of the blood.
3- Obstructions (of flow)

a) Coarctation of the aorta

b) Valvular stenosis
   i) aortic
   ii) pulmonary

c) Complete obstruction is called “Atresia”
Coarctation of the Aorta

• Narrowing

• Males : Females  2:1
  females with Turners frequently have coarctatio

• LV hypertrophy

• 2 types:
  a) infantile (with PDA; poor outcome)
  b) adult (without PDA)
Coarctation of Aorta
Pulmonary Stenosis and Atresia

• Obstruction at pulmonary valve

• May occur as isolated defect or with:
  - Tetralogy of Fallot or b) TGA

• Right Ventricular hypertrophy
  - usually there is post-stenotic dilatation

• Relatively frequent defect
Valvular (severe) aortic stenosis or atresia

a) outflow obstruction
   i) hypoplastic LV and ascending aorta

b) PDA must be present to allow blood flow to:
   i) aorta
   ii) coronary arteries
   iii) always fatal in first week of life when ductus closes!!
Aortic stenosis (valvular)

1 - narrowed aortic valve
Flow patterns are normal but blood flow to the aorta is reduced as indicated by the broken white arrows
Congestive Heart failure
Congestive Heart Failure

- Cardiac output insufficient for metabolic requirements of the body
- Systolic dysfunction – myocardial contractility
- Diastolic dysfunction – insufficient expansion for ventricular volume
- Problems are exaggerated by increased demand – high output heart failure
CHF

• Signs and symptoms:
  1) Tachycardia
  2) Dyspnea
  3) Edema

• Hypertrophy and dilatation
CHF — Body’s Compensation

- Tachycardia
- Increased End Diastolic Volume
- Myocardial hypertrophy
- Increase oxygen extraction from hemoglobin
Hypertrophy

• Pressure Overload (Concentric)
• Volume Overload (CHF)
• LVH, RVH, atrial, etc.
CHF: Autopsy Findings

- Cardiomegaly
- Chamber Dilatation
- Hypertrophy of myocardial fibers.
(Hypertrophy of cardiac muscle fibers); Note the thick fibers and thick nuclei.
Brain in CHF – cerebral hypoxia

- Irritability
- Loss of attention span
- Restlessness
- Stupor
- Coma
CHF – final pathway to death

• Ischemic heart disease
• Hypertensive heart disease
• Valvular heart disease
• Cardiomyopathy
• Myocarditis
• Specific heart muscle diseases
Left-sided Heart Failure

Causes:

• Ischemic heart disease

• Hypertension

• Aortic and mitral valve disease

• Myocardial disease
Left Sided Failure

• Low output

• Lungs
  – pulmonary congestion and edema
  – heart failure cells

• Kidneys
  – salt and fluid retention

• Brain:
  – Irritability, decreased attention, stupor/coma
HYPERTENSION

Pressure overload

⇒ Cardiac work

⇒ Wall stress

⇒ Cell stretch

Hypertrophy and/or dilation

Characterized by
- ↑ heart size and mass
- ↓ protein synthesis
- induction of immediate-early genes
- induction of fetal gene program
- abnormal proteins
- fibrosis
- inadequate vasculature

MYOCARDIAL INFARCTION

Pressure and/or volume overload

Regional dysfunction with volume overload

CARDIAC DYSFUNCTION

Characterized by
- heart failure (systolic/diastolic)
- arrhythmias
- neurohumoral stimulation
Symptoms

- Dyspnea: on exertion at rest
- Orthopnea: dyspnea lying down
- Paroxysmal Nocturnal Dyspnea
- Cough with Blood tinged sputum
- Cyanosis
- Elevated pulmonary pressure
Lungs – Pulmonary edema

- Dyspnea – breathlessness
- Orthopnea – dyspnea lying down
- Paroxysmal nocturnal dyspnea – extreme dyspnea
Lung – Pulmonary Edema – pale pink edema fluid filling alveoli
Lung – alveolar hemorrhage, heme-filled macrophages "heart failure cells", with iron stain to right
Right-sided heart failure

Causes:

– Pure cor pulmonale

– Consequence of left-sided failure

– Myocardial: myocarditis, cardiomyopathy, constrictive pericarditis
Symptoms and signs

- Liver and spleen
  - passive congestion of the liver
  - congestive spleenomegaly
  - ascites

- Pleura/Pericardium
  - pleural and pericardial effusions
  - transudates

- Peripheral tissues: edema
Symptoms and signs

- Fatigue
- Dependent edema
- Jugular Vein Distension
- Hepatomegaly (congestion)
- Ascites, Pleural Effusion
- GI disorders
- Cyanosis
- Increased peripheral venous pressure
Right failure - systemic effects

- Liver – chronic passive congestion
- Spleen – congestive splenomegaly
- Kidneys – congestion and hypoxia
- Peripheral edema and anasarca
- Pleural space – effusions
- Brain – venous congestion and hypoxia
- Ascites
Liver – chronic passive congestion – blood pools near the central veins
Liver – chronic passive congestion – blood pools near the central veins
Liver – chronic passive congestion – red cell pooling near central veins and pericentral necrosis of the hepatocytes.
Thank you