# Congenital Heart Disease

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## Learning Objectives:

- Identify common etiologies and risk factors for congenital heart defects.
- Describe clinical manifestations and diagnostic methods for congenital heart defects.
- Explain the pathophysiology, manifestations, diagnosis and management of acyanotic congenital cardiac anomalies.

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## Learning Objectives:

- Explain the pathophysiology, manifestations, diagnosis and management of obstructive congenital anomalies.
- Explain the pathophysiology, manifestations, diagnosis and management of cyanotic congenital anomalies.
- Explain the implications of cardiac anomalies for respiratory care.

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## Development of the Cardiovascular System

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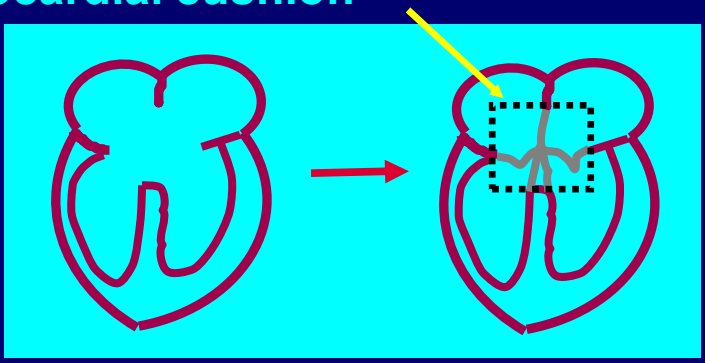
## Development of the heart

- Parallel tubes convolute to form chambers
- Septa and valves form from endocardial cushion



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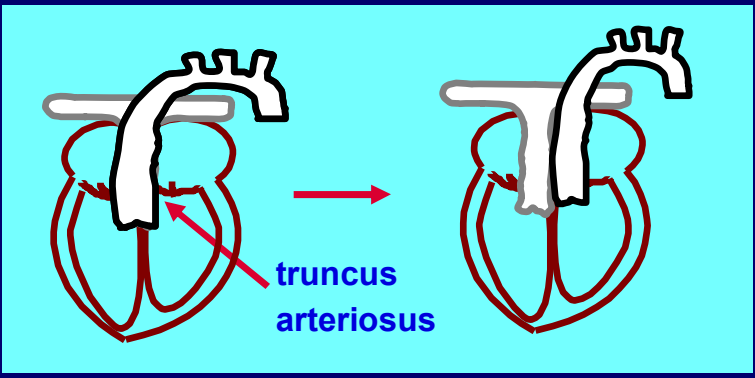
## Development of the heart

- Single artery (truncus arteriosus) divided by aorticopulmonary septum
- At eight weeks, fetal circulation is complete

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## Fetal circulation- anatomy

- Includes placental circulation- low resistance circuit
- Foramen ovale-- window between atria
- Ductus arteriosus-- vessel connecting aorta to pulmonary artery
- Ductus venosus- bypasses liver

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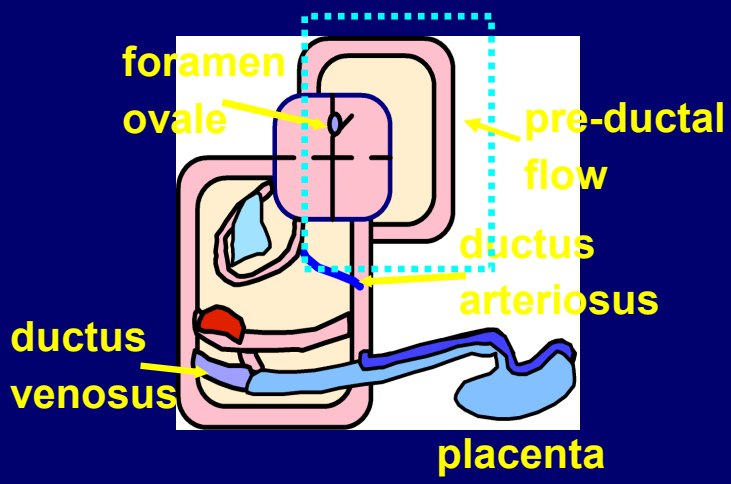
## Fetal circulation- anatomy

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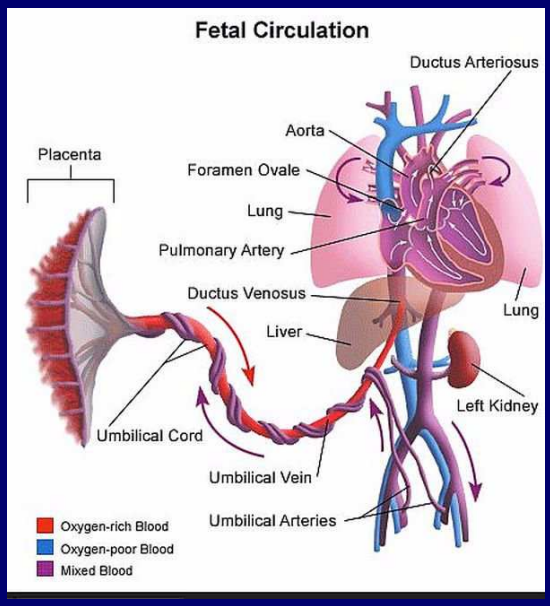


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## Fetal circulation- physiology

- High pulmonary vascular resistance
- Left side includes low resistance placental circuit
- Venous admixture at all shunts
- Pre-ductal blood with highest PaO2 to upper body

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## Changes at Birth

- Removal of placental circuit increases left-sided resistance
- Increased PaO2 lowers pulmonary vascular resistance
- Foramen ovale functionally closed-resistance on left > right
- Ductus closes due to increased PaO2, etc., about 15 hours postpartum

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## Congenital Heart Disease

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## Etiologic Factors

- maternal infections- rubella, syphilis
- maternal metabolic dx- diabetes
- maternal drug ingestion
  - phenytoin (Dilantin)
  - thalidomide
  - sex hormones

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## Medical history

- failure to thrive
- retarded growth, development
- decreased exercise tolerance
- squatting
- fainting

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## Medical history

- chronic pulmonary infections
- chronic cough
- feeding difficulties
- headaches
- epistaxis (nosebleeds)
- 'noisy breathing'

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## Physical examination

- small stature, underdeveloped
- color- may be cyanotic
- clubbing

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## Physical examination

- color- may be cyanotic
- clubbing
- heart murmurs- abnormal
  - blood flow
  - valve activity

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## Physical examination

- cyanosis
- clubbing
- heart murmurs
- displaced point of maximal impulse (PMI)
- precordial bulge

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## Physical examination

- wheezing- CHD often mistaken for asthma
- tachypnea
- tachycardia

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## Physical examination

- wheezing- CHD often mistaken for asthma
- tachypnea
- tachycardia
- blood pressure greater in arms
- weak femoral pulses
- epistaxis

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## Diagnosis

- Radiography
  - chest radiograph
  - angiography
- Echocardiography-
  - replaced catheterization for many defects
  - detects congenital defects in utero

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## Diagnosis

- Electrocardiography
- Blood gases and/or oximetry
  - pre, post-ductal SO2
  - SO2 in various compartments
- Cardiac catheterization
  - diagnostic
  - therapeutic

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## Categories

- Acyanotic CHD
- Obstructive defects
- Conduction defects
- Cyanotic CHD
- Miscellaneous
  - Dextrocardia
  - Vascular rings

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## Acyanotic Cardiac Anomalies

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## Acyanotic Anomaly Types

- Persistent fetal structures
  - patent ductus arteriosus
  - patent foramen ovale
- Septal defects
  - ventricular septal defects
  - atrial septal defects
  - endocardial cushion defects

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## Acyanotic Anomaly Types

- Obstructive defects
  - coarctation of aorta
  - aortic stenosis
- Conduction defects

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## Persistent Fetal Structures

- Types
  - patent ductus arteriosus
  - patent foramen ovale
- May persist asymptomatically, through adulthood.
  - exacerbated by pulmonary hypertension (hypoxemia)
  - shunt may change to right-to-left with PEEP, worsening hypoxemia

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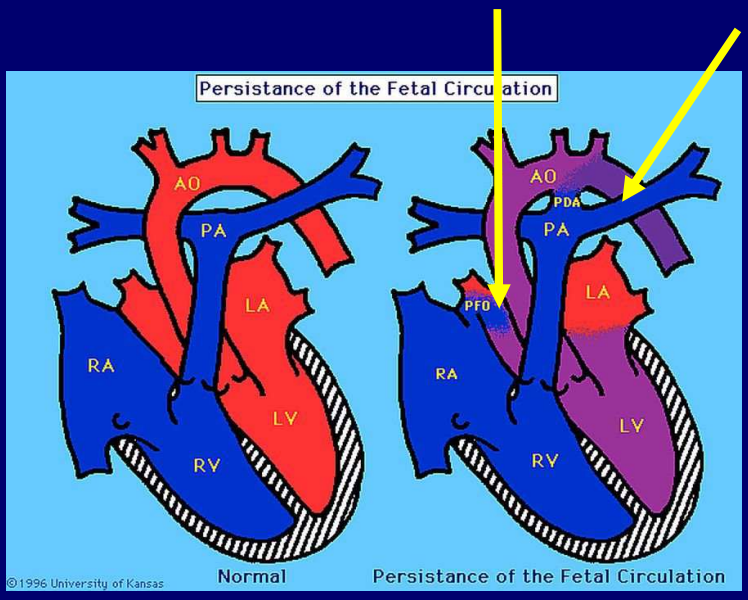


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## Persistent Fetal Structures

- Normal pulmonary vascular resistance
  - left-to-right shunt
    - no effect on arterial blood gases
    - elevated mixed venous PO2
  - increased LV work
    - LV failure
    - CHF

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## Persistent Fetal Structures

- Increased pulmonary vascular resistance
  - right-to-left shunt
  - hypoxemia, refractory to supplemental O2

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## Patent Ductus Arteriosus

- Second most common anomaly in term infants
- Etiologic factors
  - neonatal asphyxia, hypoxemia
  - maternal viral infections, e.g., rubella
  - low socioeconomic status- nutrition
- Note- patent ductus is necessary for survival in patients with ductal-dependent anomalies

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## Patent Ductus Arteriosus

- Complications
  - excessive workload on left ventricle
  - pulmonary artery disease (Eisenmenger's complex) due to excessive pulmonary blood flow
  - chronic pulmonary infections

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## Patent Ductus Arteriosus

- Manifestations
  - persistent murmur
  - decreased lung compliance ==> increased work of breathing
  - cardiomegaly
  - diagnosed by echocardiogram

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## Patent Ductus Arteriosus

- Management
  - Medical
    - ibuprofen (Advil) to close ductus
    - indomethacin to close ductus
    - intubate and ventilate with PEEP to improve oxygenation
  - Surgical
    - ligation (sometimes done in NICU)
    - division- requires thoracotomy

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## Septal Defects

- Normal pulmonary vascular resistance (PVR)
  - left-to-right shunt
    - no effect on arterial blood gases
    - elevated mixed venous PO2
  - increased LV work due to need for increased cardiac output required by loss of peripheral blood flow
    - LV failure
    - CHF

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## Septal Defects

- Normal PVR
  - left-to-right shunt
  - increased LV work
  - excessive pulmonary blood flow
    - causes chronic pulmonary infections
    - causes remodeling of pulmonary vasculature (Eisenmenger's complex)

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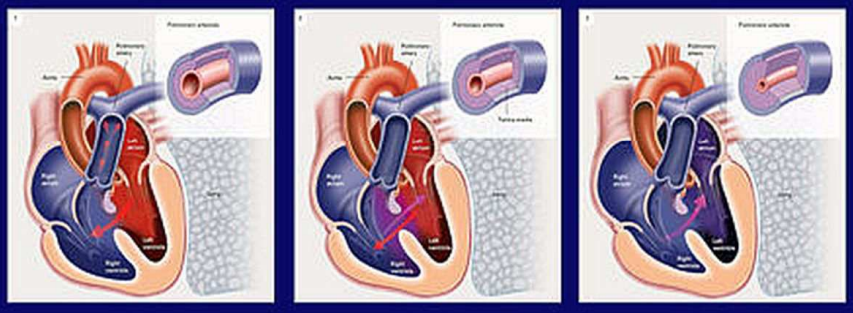
## Evolution of Eisenmenger Syndrome



ASD, VSD, or complex defect ↑ Qp and/or PAp, with L-to-R shunting

Over time, PVR ↑ resulting in bi-directional flow

PVR ↑'s: shunt reverses: R-to-L → Eisenmenger syndrome: ↑ cyanotic

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## Septal Defects

- Increased pulmonary vascular resistance
  - right-to-left shunt
  - hypoxemia, refractory to supplemental O2

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## Septal Defects

- Small VSD (less than diameter of aortic valve)
  - left-to-right shunt if VSD < 50% aortic diameter
  - RV & LV pressures normal
  - May close spontaneously

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## Small VSD

- Manifestations
  - may be asymptomatic
  - only clinical sign may be murmur
  - other data normal

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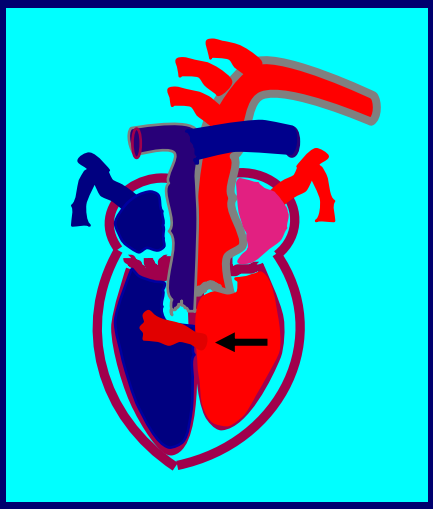
## Small VSD- Left-to-Right Shunt



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## Large VSD

- Defined as VSD diameter > aortic valve diameter
- Hemodynamics
  excessive PA flow ==> vascular remodeling ==> increased PVR ==> right-to-left shunt (hypoxemia)
- Prolonged left-to-right shunt that causes remodeling of pulmonary vessels necessitates a heart & lung transplant

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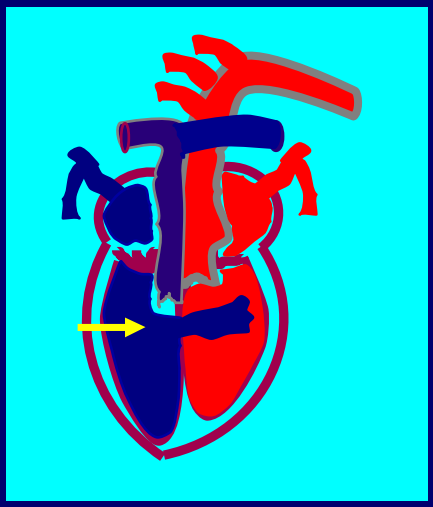
## VSD- Right-to-Left Shunt

- venous admixture to left ventricle



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## Large VSD

- Manifestations
  - Murmur
  - CHF
  - Cyanosis with pulmonary hypertension
- LV hypertrophy

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## Large VSD

- Diagnosis
  - Echocardiography
  - Heart catheterization
  - Angiography

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## Large VSD

- Management
  - Palliation, to reduce pulmonary blood flow
    - Pulmonary artery banding
    - subambient FIO2- causes pulmonary vasoconstriction
  - Correction- Gortex patch closure

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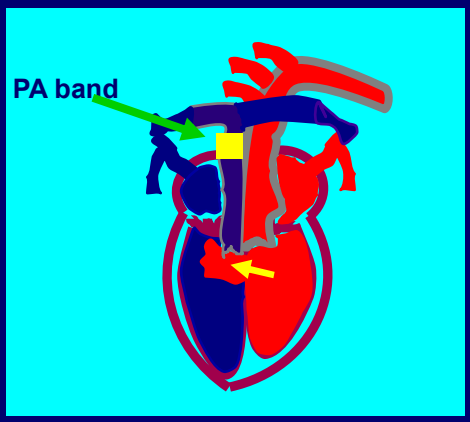
## Pulmonary Artery Banding

- Palliative procedure to reduce pulmonary blood flow

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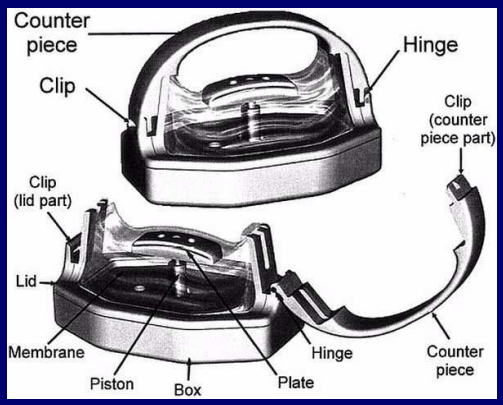
## Pulmonary Artery Banding

- Flowatch(TM) - telemetrically adjustable pulmonary artery band

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## Atrial Septal Defect

- Categories- based on position of the defect on atrial wall
  - ostium primum
  - ostium secundum
  - sinus venosus

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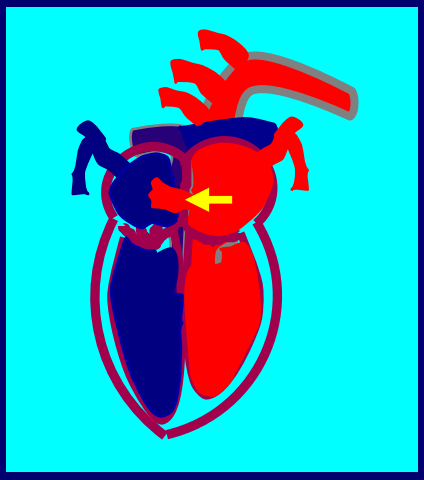
## Atrial Septal Defect

- left-to-right shunt



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## Atrial Septal Defect

- Manifestations
  - murmur
  - may be congenital PFO
  - may be otherwise asymptomatic for 20-30 years
  - normal PVR ==> left-to-right shunt ==> elevated RA and RV PO2

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## Atrial Septal Defect

- Manifestations
  - murmur
  - may be congenital PFO
  - may be otherwise asymptomatic for 20-30 years
  - normal PVR ==> left-to-right shunt ==> elevated RA and RV PO2
  - first sign may be right ventricular failure
  - may follow pathophysiology of VSD

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## Atrial Septal Defect

- Diagnosis
  - ECG - Right axis deviation
  - Echocardiography- detected with bubble test
  - Heart catheter- elevated RA, RV SO2

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## Atrial Septal Defect

- Diagnosis
  - ECG - Right axis deviation
  - Echocardiography- detected with bubble test
  - Heart catheter- high RA, RV SO2
- Treatment- closure in catheterization lab.

Click to see video on ASD repair (4 min)
http://www.youtube.com/watch?v=PzKJ_chafEU

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## Endocardial Cushion Defect

- Pathogenesis- incomplete development of ECD
- Associated with trisomy 21 (Down's syndrome)
- Defects- permutations of:
  - ASD
  - VSD
  - Cleft mitral, tricuspid valve leaflets

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## Endocardial Cushion Defect

- Hemodynamics-- depend on specific defects and their extent
  - ASD- L to R shunt
  - VSD- L to R shunt ==> left ventricular hypertrophy
  - Mitral regurgitation ==> left atrial hypertrophy
  - Increased PA flow ==> vascular remodeling & increased PVR

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## Endocardial Cushion Defect

- Manifestations
  - May be asymptomatic
  - May develop severe CHF & pulmonary edema
- Diagnosis
  - ECG- left axis deviation
  - Heart catheter- increased SaO2 in RA & RV
  - Echocardiography

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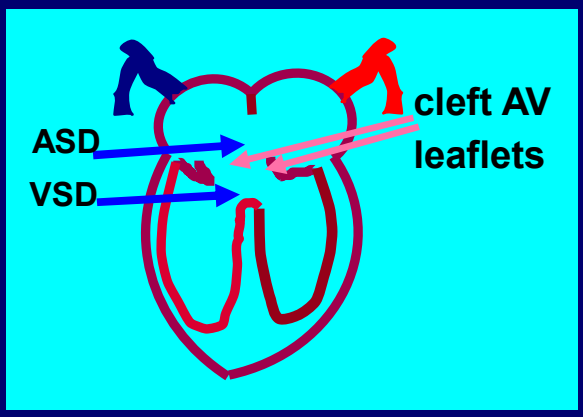
## Endocardial Cushion Defect

- Complete AV canal




Click for video of AV canal (2.3 min.)
https://www.youtube.com/watch?v=C0Gb-yScjSk

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## Endocardial Cushion Defect

- Management
  - palliative PA banding
  - heart failure managment
    - diuretics
    - digitalis
  - surgical correction
    - septal defect closure- Dacron patch
    - valvuloplasty- technically difficult

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## Obstructive Anomalies

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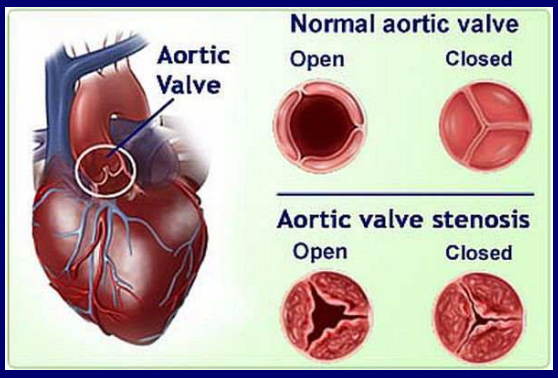
## Aortic stenosis

- Narrowed aortic outflow tract
- Hemodynamics- increased resistance to LV outflow ==> increased LV work ==> hypertrophy ==> LV failure

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## Aortic stenosis

- Manifestations
  - Ejection systolic murmur
  - Left ventricular hypertrophy
  - CHF, sudden death (severe)
- Management
  - Valvotomy, balloon valvuloplasty
  - Valve replacement

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## Coarctation of the Aorta

- Narrowing of portion of aorta
- Hemodynamics
  - aortic obstruction
  - severity dependent on degree of narrowing
- Associated with chromosomal abnormality- Turner's syndrome

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## Coarctation of the Aorta

- Manifestations
  - reduced pulses, blood pressure in lower extremities
  - headaches
  - epistaxis
  - leg cramps

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## Coarctation of the Aorta

- Manifestations
  - left ventricular hypertrophy
  - CHF, pulmonary edema
  - neonates- lower body cyanosis
    - pre-ductal coarctation
    - in presence of PDA

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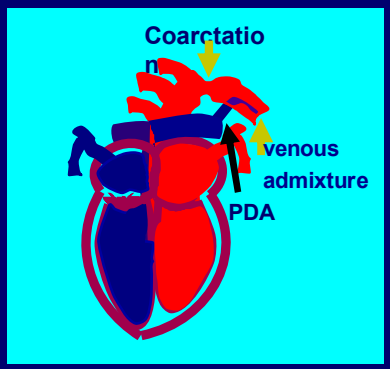
## Coarctation of the Aorta

- Pre-ductal- coarctation proximal to ductus arteriosus




Click to see video on coarctation (2.4 min)
https://www.youtube.com/watch?v=HbUrfLgE2FE

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## Coarctation of the Aorta

- Management
  - avoid heavy exercise
  - balloon dilatation with stent
  - resection- may require graft

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## Conduction defect

- Wolff-Parkinson-White syndrome
- Impulse aberrantly conducted through bundle of Kent
- Manifestations
  - PR interval < 0.12s
  - paroxysmal atrial tachycardia (PAT)
- Treatment
  - Medical- antidysrhythmics
  - Electrophysiology- ablation

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# Cyanotic Anomalies

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## Cyanotic Anomalies

- Categories:
  - increased pulmonary blood flow
  - decreased pulmonary flow

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## Cyanotic Anomalies

- Requirements for arterial desaturation
  - Communication between systemic & pulmonary circulation
    - abnormal vessels
    - septal defects
  - PVR > SVR
- Desaturation due to intracardiac shunt is unresponsive to increased FiO2

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## Cyanotic Anomalies

- Conditions with low pulmonary flow
  - Tetralogy of Fallot
  - Pulmonary atresia
  - Tricuspid atresia
- Bicuspid atresia, AKA hypoplastic left ventricle

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## Cyanotic Anomalies

- Conditions with high pulmonary flow
  - Transposition of great vessels
  - Persistent truncus arteriosus
  - Total anomalous pulmonary venous return

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## Tetralogy of Fallot

- Defects (tetra = four)
  - Pulmonary stenosis
  - Ventricular septal defect (VSD)
  - Overriding aorta-- aorta straddles both ventricles
  - Right ventricular hypertrophy

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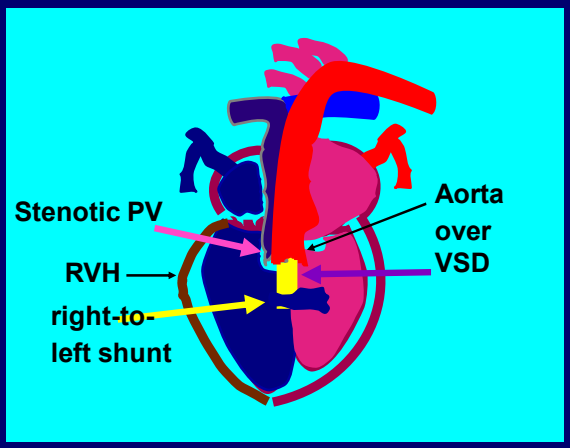
# Tetralogy of Fallot

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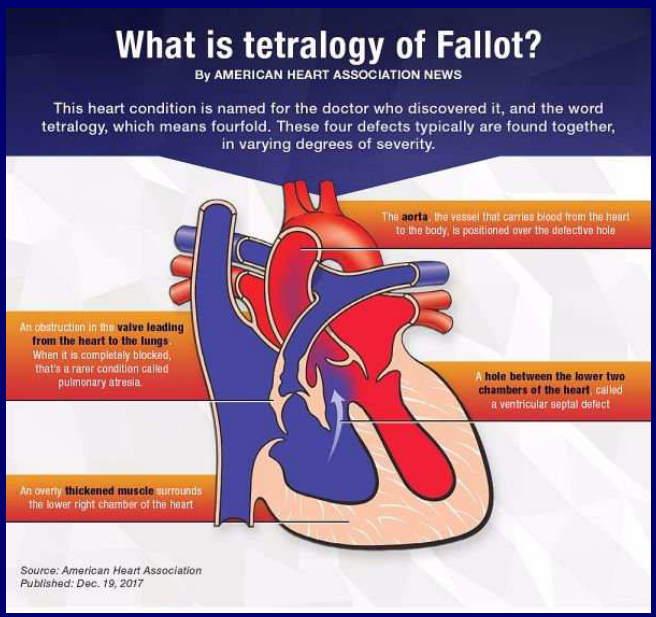


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## Tetralogy of Fallot

- Hemodynamics
  - Pulmonary stenosis
    - Determines PA resistance to flow
    - Regulates resistance to right ventricular flow
    - Determines right to left shunt
    - Leads to RV hypertrophy
    - Degree of stenosis determines urgency of surgical intervention

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## Tetralogy of Fallot

- Hemodynamics
  - VSD- channel for shunt
    - Will be left-to-right with low pulmonary resistance
    - Usually large

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## Tetralogy of Fallot

- Hemodynamics
  - Overriding aorta
    - Carries outflow from both ventricles
    - Contributes to severity of shunt
  - RV hypertrophy
    - Chronic elevated flow resistance
    - Very large VSD- equalizes pressures in LV and RV

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## Tetralogy of Fallot

- Spectrum from "pink tets" to emergent cases in neonatal stage
- May not appear until closure of PDA, then pulmonary blood flow declines

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## Tetralogy of Fallot

- Manifestations
  - cyanosis- "tet spells" with exertion
  - squatting to relieve exertional spells- increases systemic resistance to blood flow
  - clubbing
  - growth retardation
  - systolic ejection murmur

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## Tetralogy of Fallot

- Chest xray- 'boot-shaped' heart
- ECG-- right axis deviation
- Echocardiography- usually definitive
- Catheterization

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## Tetralogy of Fallot

- Management of tet spells
  - fetal positioning
  - morphine
  - low flow oxygen- an exception for supplemental O2
  - bicarbonate
  - propanolol
  - vasoconstrictors

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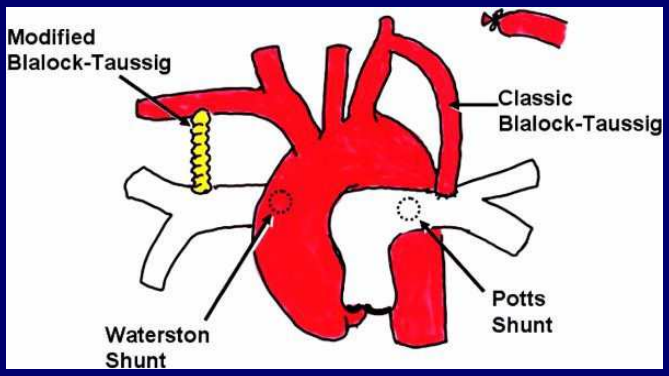
## Tetralogy of Fallot

- Palliation- arterial to pulmonary artery shunts
  - bypass stenotic pulmonary valve
  - increase pulmonary blood flow

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## Tetralogy of Fallot

- Palliation- arterial to pulmonary artery shunts
  - bypass stenotic pulmonary valve
  - increase pulmonary blood flow
- Total correction
  - Excision of PV obstruction
  - Patch closure of VSD

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## Tricuspid atresia

- Defects
  - Atretic tricuspid valve- does not open, so blocks blood flow from atrium to ventricle
  - Diminutive (small) RV
  - VSD & ASD

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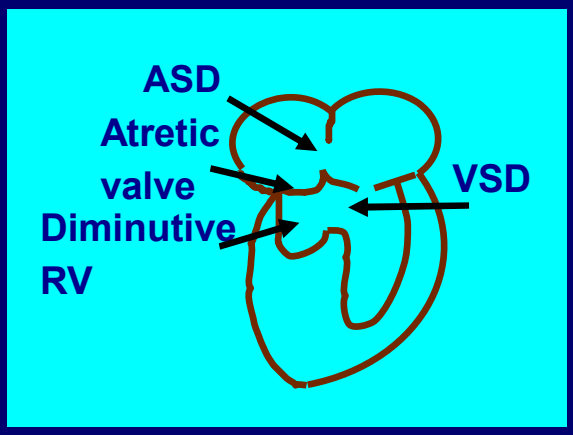
## Tricuspid atresia

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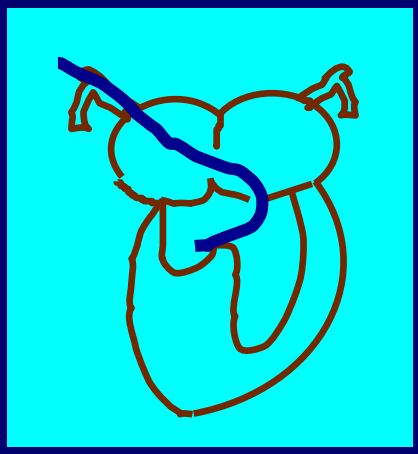
## Tricuspid atresia

- Blood flow
  - Vena cava to RA to ASD to LA to LV to RV (via VSD)



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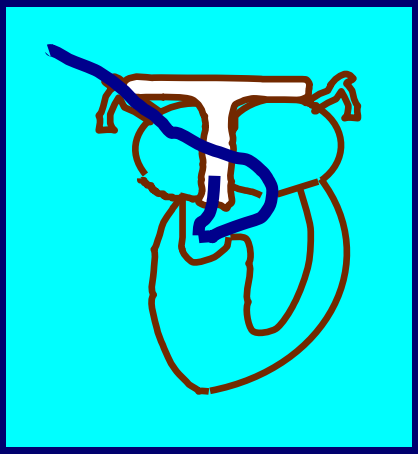
## Tricuspid atresia

- Blood flow

  Vena cava to RA to ASD to LA to LV to RV (via VSD) to PA



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## Tricuspid atresia

- Signs
  - early cyanosis (from birth)
  - worsening, death on closure of ductus arteriosus
  - growth retardation
  - squatting
  - clubbing

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## Tricuspid atresia

- Diagnosis
  - ECG- left axis deviation
  - Echocardiography
    - diminutive right ventricle
    - absent tricuspid echoes
  - Catheterization-- catheter will not enter RV

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## Tricuspid atresia

- Palliative procedures- to increase pulmonary blood flow
  - Maintain PDA
    - subambient FIO2
    - alprostadil
    - stent placement
  - Waterston shunt-- aorta to RPA
  - Blalock-Taussig (BT) shunt- from subclavian artery to PA

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## Tricuspid atresia

- Management
  - Corrective- Fontan
    - bypass RV by directing blood from RA to PA
    - pulmonary blood flow becomes dependent on passive venous return.

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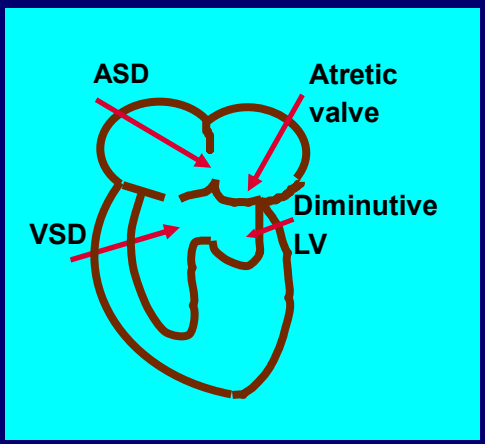
## Bicuspid atresia- hypoplastic LV

- Defects
  - Atretic bicuspid valve
  - Diminutive LV
  - VSD & ASD

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## Hypoplastic LV

- Signs
  - early cyanosis
  - shock
  - worsening, death with DA closure

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## Hypoplastic LV

- Echocardiogram
  - diminutive left ventricle
  - absent bicuspid echo

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## Hypoplastic LV

- Maintain PDA
- Surgical management
  - Norwood- multiple stage procedure
  - Fontan
  - Blalock-Taussig (BT) shunt

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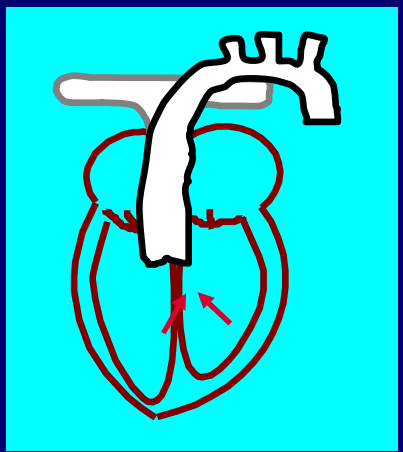
## Persistent truncus arteriosus

- Defects
  - Single artery for LV & RV
  - VSD



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## Persistent truncus arteriosus

- Hemodynamics
  - truncus carries blood to PA & aorta
  - flow is dependent upon resistance to flow at each side
    - increased SVR ==> increased pulmonary flow
    - increased PVR ==> increased systemic flow

Click for video of truncus arteriosus (2 min)
https://www.youtube.com/watch?v=193JQ6LQRwI

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## Persistent truncus arteriosus

- In presence of decreased PVR ==> excessive pulmonary blood flow ==>
  - high output LV failure (CHF)
  - pulmonary vascular dx
- In presence of increased PVR ==>
  - reduced in pulmonary blood flow==> hypoxemia

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## Persistent truncus arteriosus

- Manifestations
  - Cyanosis
  - CHF

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## Persistent truncus arteriosus

- Diagnosis
  - CXR- cardiomegaly
  - ECG- combined hypertrophy
  - Echocardiogram
    - visualize vessel origins
    - one semilunar valve
  - Catheterization- equal LV & RV pressures

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## Persistent truncus arteriosus

- Management
  - Heart failure
    - digoxin
    - diuretics
  - Palliative- reduce PA flow
    - PA banding
    - subambient FIO2

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## Persistent truncus arteriosus

- Corrective surgry
  - main trunk moved to left
  - creation of outflow tube from RV to PAs
  - closure of VSD

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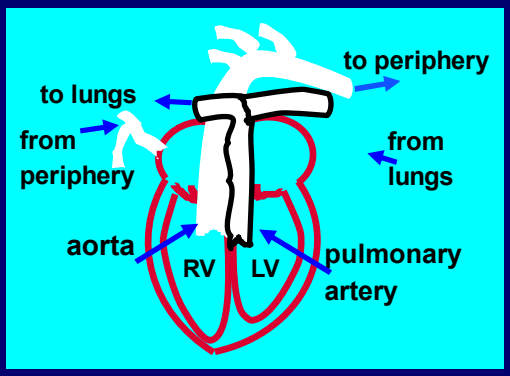
# Transposition of great arteries (TGA)

- Defects
  - Aorta arises from RV
  - Pulmonary artery arises from LV
  - ASD and/or VSD, PDA (increase chance for survival)

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# TGA

- Hemodynamics
  - Separate circulations
  - Pulmonary venous blood to LA to LV through PA to lung
  - Systemic venous return to RA to RV to aorta to system
  - Without septal defect, life impossible
  - With VSD, there is mixing

Click for video of TGA (3 min)
https://www.youtube.com/watch?v=lBGUtiBp8k8

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## TGA

- Signs
  - diabetic mother- high risk
  - early cyanosis
  - CHF

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## TGA

- Diagnosis
  - CXR-- cardiomegaly
  - Echocardiogram- visualize vessels
  - Catheterization- catheter enters aorta from RV

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## TGA

- Management
  - Palliative
    - maintain PDA
    - balloon septostomy

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## TGA

- Management
  - Corrective
    - Mustard-- baffle in atria
    - Jatene (switch)- vessels switched to correct ventricles

Click for video of Jatene switch procedure (2 min)
https://www.youtube.com/watch?v=QNUnZqoeBTo

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# Therapeutics

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## Maintaining a PDA

- Indication- ductal dependent cardiac anomaly; e.g.:
  - transposition of great arteries
  - tricuspid atresia
  - mitral atresia
- Methods
  - stent
  - alprostadil (Prostin)
  - subambient O2

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## Subambient O2 Therapy

- goals
  - increase pulmonary vascular resistance to reduce pulmonary blood flow in presence of
    - large VSD
    - endocardial cushion defect
    - persistent truncus arteriosus

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## Subambient O2 Therapy

- goals
  - increase pulmonary vascular resistance to reduce pulmonary blood flow in presence of
    - large VSD
    - endocardial cushion defect
    - persistent truncus arteriosus
  - prevent closure of ductus arteriosus in presence of:
    - transposition of great arteries
    - tricuspid atresia
    - mitral atresia

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## Subambient O2 Therapy

- methods
  - bleed-in nitrogen to ventilator circuit
  - obtain premixed subambient mixture in cylinder
  - titrate FIO2 to SaO2 80-85%
- problem- some O2 analyzers may not measure subambient FO2

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## Special issues

- Frequently, congenital defects are undetected until later childhood or adulthood
- Transplant organs difficult to obtain, especially those for infants and children.
- Patients' hearts can outgrow. synthetic structures, like valves.
- Oxygen therapy can kill patients with ductal dependent anomalies.
- Post-surgical cardiac patients are very prone to dysrhythias.

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## Summary & Review

- Development of the cardiovascular system
  - endocardial cushion
  - truncus arteriosus
  - fetal circulation with shunts
  - changes at birth

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## Summary & Review

- Congenital heart disease
  - etiologic factors
  - historical manifestations
  - physical manifestations
  - diagnostic procedures
  - categories
    - acyanotic
    - cyanotic
    - obstructive
    - conduction defects

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## Summary & Review

- Acyanotic defects
  - types
    - persistent fetal structures
    - ventricular septal defects
    - atrial septal defects
    - endocardial cushion defects
  - complications
    - remodeling of pulmonary vessels
    - left ventricular failure

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## Summary & Review

- Acyanotic defects
  - management
    - palliation with PA bands
    - total correction

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## Summary & Review

- Obstructive defects
  - types
    - aortic stenosis
    - coarctation of the aorta
  - manifestations
  - management
    - limit exercise
    - surgical repair

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## Summary & Review

- Conduction defect- WPW syndrome
  - abnormal conduction pathway
  - ECG- decreased P-R interval
  - management
    - medications for PAT
    - ablation of bundle of Kent

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## Summary & Review

- Cyanotic defects
  - types- high, vs. low pulmonary blood flow
  - ductal dependence
  - manifestations
    - cyanosis
    - tetralogy spells
    - ventricular failure

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## Summary & Review

- Cyanotic defects
  - tetralogy of Fallot
  - tricuspid atresia
  - hypoplastic left ventricle (mitral atresia)
  - Persistent truncus arteriosus
  - Transposition of great arteries

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## Summary & Review

- Subambient oxygen therapy
- Postoperative expectations
- Issues in congenital heart disease

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# END

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