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.
- 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.

Development of the Cardiovascular System

Development of the heart
- Parallel tubes convolute to form chambers
- Septa and valves form from endocardial cushion

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
Fetal circulation- anatomy

- foramen ovale
- ductus venosus
- ductus arteriosus
- pre-ductal flow
- placenta

Click to see a diagram of fetal circulation

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

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

Congenital Heart Disease

Etiologic Factors

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

Medical history

- failure to thrive
- retarded growth, development
- decreased exercise tolerance
- squatting
- fainting
**Medical history**
- chronic pulmonary infections
- chronic cough
- feeding difficulties
- headaches
- epistaxis (nosebleeds)
- 'noisy breathing'

**Physical examination**
- small stature, underdeveloped
- color- may be cyanotic
- clubbing

**Physical examination**
- color- may be cyanotic
- clubbing
- heart murmurs- abnormal
  ◆ blood flow
  ◆ valve activity

Click for information on the physiology of heart murmurs
http://www.wilkes.med.ucla.edu/Physiology.htm

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

**Physical examination**
- wheezing- CHD often mistaken for asthma
- tachypnea
- tachycardia

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

- Radiography
  - chest radiograph
  - angiography
- Echocardiography - replaced catheterization for many defects

Click for information on echocardiography and CHD
http://www.echoincontext.com/advanced/chd_01.asp

Diagnosis

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

FYI - Click for article on diagnostic cardiac catheterization and CHD
http://www.ncbi.nlm.nih.gov/pmc/articles/PMC479386/

Categories

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

Acyanotic Cardiac Anomalies

Acyanotic Anomaly Types

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

Acyanotic Anomaly Types

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

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

Persistent Fetal Structures

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

Click to see persistent fetal circulation
http://www.kumc.edu/Institution/medicine/pedcard/cardiology/pedcardio/pfcdiagram.gif

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

Patent Ductus Arteriosus

**Complications**
- excessive workload on left ventricle
- pulmonary artery disease (Eisenmenger's)
- chronic pulmonary infections

Patent Ductus Arteriosus

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

Click for more information and pictures of PDA
http://www.pted.org/?id=patentductus1
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

FYI - Click for article on PDA closure
http://pediatriccct.surgery.ucsf.edu/conditions-procedures/patent-ductus-arteriosus.aspx

Septal Defects

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

Click to see evolution of Eisenmenger's complex

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)

Click to see evolution of Eisenmenger’s complex

Septal Defects

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

Small VSD

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

Click to hear VSD murmur
http://www.wilkes.med.ucla.edu/Systolic.htm

Small VSD

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

Click to hear VSD murmur
http://www.wilkes.med.ucla.edu/Systolic.htm
Small VSD - Left-to-Right Shunt

VSD- Right-to-Left Shunt

Large VSD

VSD diameter > aortic valve
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

Manifestations
Murmur
CHF
Cyanosis with pulmonary hypertension
LV hypertrophy

Diagnosis
Echocardiography
Heart catheterization
Angiography

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

Click for more information and pictures of VSD
http://www.pted.org/?id=ventricularseptal1

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Pulmonary Artery Banding

- Palliative procedure to reduce pulmonary blood flow

Click for article about Flowatch PA band with illustrations
http://circ.ahajournals.org/content/110/11_suppl_1/II-158.full

Atrial Septal Defect

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

Atrial Septal Defect

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

Click to hear murmur with ASD
http://www.wilkes.med.ucla.edu/Systolic.htm

Atrial Septal Defect

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

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

Atrial Septal Defect

- Manifestations
  - murmur
  - 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

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
Atrial Septal Defect
- left-to-right shunt

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

Endocardial Cushion Defect
- Hemodynamics-- depend on specific defects
  - 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

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

Endocardial Cushion Defect
- Complete AV canal

Endocardial Cushion Defect
- Management
  - palliative PA banding
  - heart failure management
    - diuretics
    - digitalis
  - surgical correction
    - septal defect closure- Dacron patch
    - valvuloplasty- technically difficult
Obstructive Anomalies

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

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

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

Coarctation of the Aorta
- Manifestations
  - Reduced pulses, blood pressure in lower extremities
  - Headaches
  - Epistaxis
  - Leg cramps

Coarctation of the Aorta
- Manifestations
  - Left ventricular hypertrophy
  - CHF, pulmonary edema
  - Neonates - lower body cyanosis
    - Pre-ductal coarctation
    - In presence of PDA

Click for more information and pictures of aortic stenosis: http://www.pted.org/?id=aorticstenosis1

Click for more information and pictures of coarctation: http://www.pted.org/?id=coarctation1
Coarctation of the Aorta

- Pre-ductal coarctation proximal to ductus arteriosus

![Coarctation Diagram]

Management
- Avoid heavy exercise
- Balloon dilatation with stent
- Resection - may require graft

Coarctation of the Aorta

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

Cyanotic Anomalies

Categories:
- Increased pulmonary blood flow
- Decreased pulmonary flow

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

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

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

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

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

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

Click for more information and pictures of TOF
http://www.pted.org/?id=tetralogyfallot1

Manifestations
- Cyanosis- "tet spells" with exertion
- Squatting to relieve exertional spells
- Clubbing
- Growth retardation
- Systolic ejection murmur

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

Click to see 'boot-shaped' heart on x-ray
http://www.bcm.edu/radiology/cases/pediatric/text/3a-desc.htm

Management of tet spells
- Fetal positioning
- Morphine
- Oxygen- an exception for supplemental O2
- Bicarbonate
- Propranolol
- Vasoconstrictors

Palliation- arterial to pulmonary artery shunts
- Bypass stenotic pulmonary valve
- Increase pulmonary blood flow
- Total correction
  - Excision of PV obstruction
  - Patch closure of VSD
Tricuspid atresia

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

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

Click for more information and pictures of tricuspid atresia
http://www.pted.org/?id=tricuspidatresia1

Signs
- Early cyanosis (from birth)
- Worsening, death on closure of ductus arteriosus
- Growth retardation
- Squatting
- Clubbing

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

Tricuspid atresia

- Management
  - Corrective - Fontan
    - High risk, high failure rate
    - Bypass RV by directing blood from RA to PA
    - Pulmonary blood flow becomes dependent on passive venous return.

Click to see pictures of the Fontan procedure
http://www.pted.org/?id=fontan1

Bicuspid atresia - hypoplastic LV

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

Hypoplastic LV

- Signs
  - Early cyanosis
  - Shock
  - Worsening, death with DA closure

Click to see hypoplastic LV
http://www.pted.org/?id=hypoplasticleft1

Hypoplastic LV

- Echocardiogram
  - Diminutive left ventricle
  - Absent bicuspid echo

Hypoplastic LV

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

- **Defects**
  - Single artery for LV & RV
  - VSD

**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

**Decreased PVR ==> excessive pulmonary blood flow ==>**
- High output LV failure (CHF)
- Pulmonary vascular dx
- Increased PVR ==> reduced in pulmonary blood flow ==> hypoxemia

**Diagnosis**
- CXR - cardiomegaly
- ECG - combined hypertrophy
- Echocardiogram
  - Visualize vessel origins
  - One semilunar valve
- Catheterization - equal LV & RV pressures

**Manangement**
- Heart failure
  - Digoxin
  - Diuretics
- Palliative - reduce PA flow
  - PA banding
  - Subambient FIO2

Click to see chest xray of patient with truncus arteriosus
http://cardiopedia.wdfiles.com/local--files/truncus-arteriosus/truncus%20arteriosus%20type%201.jpg
**Persistent truncus arteriosus**
- Corrective surgery
- Main trunk moved to left
- Creation of outflow tube from RV to PAs
- Closure of VSD

**Transposition of great arteries (TGA)**

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

- **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

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

- **Management**
  - Palliative
    - Maintain PDA
    - Balloon septostomy

**TGA**

- **Signs**
  - Diabetic mother -- high risk
  - Early cyanosis
  - CHF

Click for more information and pictures of TGA
http://www.pted.org/?id=transpositiond1
**TGA**

▲ Management

◆ Corrective

➢ Mustard -- baffle in atria

➢ Jatene (switch) -- vessels switched to correct ventricles

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**Therapeutics**

---

**Maintaining a PDA**

▲ Indication -- ductal dependent cardiac anomaly; e.g.:

◆ transposition of great arteries

◆ tricuspid atresia

◆ mitral atresia

▲ Methods

◆ stent

◆ alprostadil (Prostin)

◆ subambient O2

---

**Subambient O2 Therapy**

▲ goals

◆ increase pulmonary vascular resistance to reduce pulmonary blood flow

➢ large VSD

➢ endocardial cushion defect

➢ persistent truncus arteriosus

◆ prevent closure of ductus arteriosus

➢ transposition of great arteries

➢ tricuspid 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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**Post-surgical Considerations**

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Response</th>
<th>Action</th>
</tr>
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<tbody>
<tr>
<td>DA closure PIP</td>
<td>Increase CL</td>
<td>Decrease</td>
</tr>
<tr>
<td>PA band</td>
<td></td>
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<tr>
<td>A-PA shunts PIP</td>
<td>Decrease CL</td>
<td>Increase</td>
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<tr>
<td>DA patency</td>
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</table>
Special issues
- Transplant organs difficult to obtain.
- Patients' hearts can outgrow synthetic structures, like valves.
- Oxygen therapy can kill patients with ductal dependent anomalies.

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

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

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

Summary & Review
- Acyanotic defects
  - management
    - palliation with PA bands
    - total correction

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

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

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

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

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

References

END