Developmental anatomy of the heart and the embryological basis for cardiac defects

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http://www.childrenheartinstitute.org/educate/defects/tetra1.htm

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Formation of Endocardial Tube

- Separate endocardial tubes that fuse
- Single endocardial tube elongates, forms dilations & constrictions
- Sinus venosus, atrium, ventricle, bulbus cordis, truncus arteriosus

From Moore & Persaud 1998
General Organization

From Moore & Persaud 1998
Blood Flow and Embryological Fates

- Solid arrows: circulation
- Dotted arrows: embryological derivatives
Endocardial cushions

- dorsal & ventral swellings
- fuse, dividing the single AV canal into paired canals
- involved in formation of interatrial & interventricular septa
- derived from neural crest
- involved in many CHDs

From Moore & Persaud 1998
Atrial Partitioning I

- Septum primum grows from atrial roof toward endocardial cushions
- Foramen primum: shunt that closes
- Foramen secundum: perforates septum primum, allowing shunt
- Septum secundum grows down, overlapping foramen secundum

From Moore & Persaud 1998
Atrial Partitioning II

- Septum secundum grows down, overlapping foramen secundum
- Foramen ovale: between septum primum & septum secundum
- Remaining portion of septum primum forms valve of foramen ovale

From Moore & Persaud 1998
Atrial Partitioning III

- **Fetus**
  - right side high pressure (high pulmonary resistance, etc.)
  - well oxygenated blood streams through foramen ovale
  - valve of foramen ovale closes with left atrial contraction

- **After birth**
  - right side low pressure (low pulmonary resistance)
  - valve remains closed (physiological closure)
  - valve eventually fuses (anatomical closure): fossa ovalis

From Moore & Persaud 1998
Atrial Partitioning IV

postnatal  
probe patent foramen ovale (not an ASD)

From Moore & Persaud 1998
Ventricular Partitioning

• Closes in week 7: not part of fetal circulation
• Muscular IV septum grows from floor
• Membranous IV septum forms from endocardial cushions and bulbar ridges
• Closure of membranous IV is associated with partitioning of truncus arteriosus

From Moore & Persaud 1998
Partitioning of Truncus Arteriosus

- continuous set of ridges in bulbus cordis (bulbar ridges) and truncus arteriosus (truncal ridges)
- grow toward each other, spiraling 180°
- fuse to form spiraling aorticopulmonary septum, dividing aorta & pulmonary trunk
- bulbar ridges involved in formation of IV septum
- bulbar & truncal ridges derived from neural crest cells—clinical implications

From Moore & Persaud 1998
Aortic arches: Ductus arteriosus

- postnatal vessels cobbled together from aortic arches, aortic sac, TA, & dorsal aortae
- Ductus arteriosus: persistent distal portion of left 6th arch
- DA connects pulmonary trunk to aorta
- DA closes postnatally

From Moore & Persaud 1998
Congenital Heart Defects

**Acyanotic**
- Volume load
  - Left-to-right shunts
    - Atrial septal defect
    - Ventricular septal defect
    - AV canal
    - Patent ductus arteriosus
- Pressure load
  - Obstructed ventricular outflow
    - Pulmonary valve stenosis
    - Aortic valve stenosis
    - Coarctation of aorta

**Cyanotic**
- Increased pulmonary flow
  - Tetralogy of Fallot
  - Pulmonary atresia
  - Tricuspid atresia
  - Total anomalous pulmonary return with obstruction
- Decreased pulmonary flow
  - Transposition of great vessels
  - Single ventricle
  - truncus arteriosus
  - Total anomalous pulmonary return without obstruction

Modified from Bernstein (1996) and other sources
**Ventricular Septal Defects (VSD)**

- Membranous (= perimembranous, conoventricular) VSD
  - Most common CHD (males>females)
  - Endocardial cushions & bulbar ridges fail to fuse with musc. septum

- Muscular VSD
  - In muscular IV septum
  - “Swiss cheese” VSD

- Supracristal VSD
  - Least common

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Acyanotic

volume load

left-to-right shunts

- atrial septal defect
- ventricular septal defect
- AV canal
- patent ductus arteriosus

http://www.med.yale.edu/intmed/cardio/chd
Atrial Septal Defects (ASD)

Secundum ASDs
- (Ostium) Secundum ASDs
  - Most common ASD (females>males)
  - Usually due to problems with septum primum (perforated or too short), but sometimes septum secundum or both septa
- AV septal defect (AV canal)
  - Endocardial cushion problems so that septum primum never fuses with cushion tissue
  - Patent foramen (ostium) primum
  - Valve defects
  - Sometimes no fusion of endocardial cushions: AV septal defect
  - 20% of Downs patients
- Sinus venosus ASDs: very rare

Primum ASDs & AV canal
- Sinus venosus ASD

From Moore & Persaud 1998
Increased pressure load defects: Valve stenosis

- Pulmonary or aortic stenosis
- Unequal partitioning of the truncus arteriosus
- Deviation of the aorticopulmonary septum
- One side expanded, other side stenosed

Acyanotic

Pressure load

Obstr. ventric. outflow

- Pulmonary valve stenosis
- Aortic valve stenosis
- Coarctation of aorta

From Moore & Persaud 1998
Increased pressure load defects: Aortic coarctation

- Constriction of the aorta distal to the left subclavian artery
- Typically near ductus arteriosus (lig. arteriosum)
  - Preductal (= infantile)
  - Postductal (= “adult”)
  - Juxtaductal

Acyanotic

pressure load

obstr. ventric. outflow

- pulmonary valve stenosis
- aortic valve stenosis
- coarctation of aorta

From Moore & Persaud 1998

From Cahill, 1997
Increased pressure load defects: Aortic coarctation

Collateral Circulation
- Subclavian → IMA → intercostals → aorta
- Subclavian → IMA → sup. epigastr. → inf. epigastr. → iliac → aorta
- Subclavian → cervical & scap. branches → intercostals → aorta
- Subclavian → vertebral → ant. spinal → intercostals & lumbar → aorta

From Cahill, 1997
Congenital Heart Defects

Acyanotic
- volume load
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    - ventricular septal defect
    - AV canal
    - patent ductus arteriosus
- pressure load
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    - pulmonary valve stenosis
    - aortic valve stenosis
    - coarctation of aorta

Cyanotic
- ↑ pulmonary flow
  - transpos. of gr. vessels
  - single ventricle
  - truncus arteriosus
  - total anomalous pulm. return w/o obstruction
- ↓ pulmonary flow
  - tetralogy of Fallot
  - pulmonary atresia
  - tricuspid atresia
  - total anomalous pulm. return w/ obstruction

modified from Bernstein (1996) and other sources
Transposition of the Great Arteries (d-TGA)

- Most common cyanotic neonatal heart defect
- Failure of aorticopulmonary septum to take a spiraling course
- Fatal without PDA, ASD, & VSD

Increased pulmonary load defects: TGA

Cyanotic

↑ pulmonary flow

- transpos. of gr. vessels
- single ventricle
- truncus arteriosus
- total anomalous pulm. return w/o obstruction

From Moore & Persaud 1998
Increased pulmonary load defects: Truncus arteriosus

Cyanotic

- Single outflow tract from the heart
- Improper formation of truncal ridges & aorticopulmonary septum such that aorta & pulmonary trunk are not fully divided
- 1-2% of all CHDs

↑ pulmonary flow

- transpos. of gr. vessels
- single ventricle
- truncus arteriosus
- total anomalous pulm. return w/o obstruction

From Moore & Persaud 1998
Decreased pulmonary load defects: Tetralogy of Fallot

- 5-7% of all CHDs
- Four co-occurring heart defects
  - Pulmonary stenosis
  - Ventricular septal defect
  - Overriding aorta (dextroposition)
  - Right ventricular hypertrophy
- Asymmetrical fusion of bulbar & truncal ridges

Cyanotic

↓ pulmonary flow

- tetralogy of Fallot
- pulmonary atresia
- tricuspid atresia
- total anomalous pulm. return w/ obstruction

From Moore & Persaud 1998
References: print sources


References: internet

• http://www.med.yale.edu/intmed/cardio/chd
• http://www.pediheart.org/parents/defects/index.html
• http://www.childrenheartinstitute.org/educate/eduhome.htm
• http://www.tmc.edu/thi/congenit.html
• http://www.kumc.edu/kumcpeds/cardiology/cardiology.html
• http://www.congenitalheartdefects.com/typesofCHD.html