

# Malformations of the heart

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# Malformations of the heart

= *congenital heart defects (CHDs)*

= **most common malformations**

40% all malformations

incidence approximately 2,5% all live born children

most of them diagnosed by prenatal USG

# Malformations of the heart

## Symptoms

A. Critical congenital heart defects

B. Manifestation in childhood

C. Manifestation in adulthood

# Malformations of the heart

## Symptoms

### A. Critical congenital heart defects

= most serious malformations of the heart  
with **severe hypoxemia** and **heart failure**  
→ need urgent surgery  
(otherwise lead to death)

Transposition of the great arteries

Tetralogy of the Fallot

“Functionally single ventricle”  
(hypoplastic left heart syndrome etc.)

Severe coarctation of aorta

Pulmonary or tricuspid atresia

Total anomalous pulmonary venous return

# Malformations of the heart

## Symptoms

### **B. Manifestation in childhood**

= less severe CHDs

- murmur, growth retardation, dyspnea during feeding
- dyspnea during exercise, recurrent lung infections

# Malformations of the heart

## Symptoms

### C. Manifestation in adulthood

= usually long asymptomatic CHDs with only nonspecific symptoms like **infective endocarditis** and **arrhythmias** or **Eisenmenger syndrome**

Atrial septal defect

Patent ductus arteriosus

# Classification

## Morphologic

- Abnormal position of the heart
- **Septal defects**
- **Atrioventricular defects**
- Malformations of heart valves
- **Malformation of the great arteries**
- **Patent ductus arteriosus**
- Anomalous pulmonary venous return
- Abnormalities of coronary arteries

## Functional

- CHDs with shut
  - Right-to left shunts
  - Left-to-right shunts
- Obstructive CHDs

Isolated vs. combined CHDs vs. complex malformations of multiple organ systems !!!

# CHDs with shunt

## Right-to left shunt

- CHDs with early cyanosis (“**cyanotic** malformations”)
- Some portion of deoxygenated blood bypass lung circulation and enters systemic circulation
  - in systemic circulation flows mixed blood
    - **cyanosis**
      - growth retardation, club fingers...
  - risk of paradox embolism

**Tetralogy of Fallot**

**Transposition of the great arteries**

**Total anomalous  
pulmonary venous return**

**Truncus arteriosus**



# CHDs with shunt

## Left-to-right shunt

- CHDs with late cyanosis (initially non-cyanotic malformations)
- increased volume in right heart
  - increased volume in lung circulation
  - pulmonary hypertension
  - increased pressure in right heart
    - right ventricle hypertrophy
    - change of pressure gradient
      - change to right-to-left shunt (cyanosis)

= “Eisenmerger syndrome”

Atrial septal defect

Ventricular septal defect

Atrioventricular septal defects

Patent ductus arteriosus

# Obstructive CHDs

- Non-cyanotic
- Increased pressure in left or right ventricle
  - concentric hypertrophy
- + low perfusion behind the obstruction

Coarctation of aorta

# **Most common CHDs**

# Atrial septal defect (ASD)

= second most common CHD

- Types:
  - Ostium primum defect
  - Ostium secundum defect
  - Sinus venosus defect
  - Coronary sinus defect
  - *Foramen ovale patens vs. pervium*

*left-to-right shunt*

# Ventricular septal defect (VSD)

= most common CHD

- Types:
  - **perimembranous**
  - intramuscular

*left-to-right shunt*

# Atrioventricular septal defects

- Association with Down syndrome
- Types:
  - Incomplete
    - Partial
    - Transitional
  - Complete

*left-to-right shunt*

# Tetralogy of Fallot

= most common cyanotic CHD

- Combined CHD with 4 typical features:
  - Right ventricle hypertrophy
  - Subpulmonary stenosis
  - Ventricular septal defect
  - Dextroposition of aorta  
(overrides the VSD)
- + Atrial septal defect (= pentalogy)

*right-to-left shunt*

# Transposition of the great arteries

- Types:
  - **noncorrected**
  - corrected

*right-to-left shunt*



# Truncus arteriosus

- Failure of separation of truncus arteriosus
  - mixing of deoxygenated and oxygenated blood
  - cyanosis
- VSD
- Often insufficient or stenotic common valve

*right-to-left shunt*

# Coarctation of aorta

- association with bicuspid aortic valve and Turner syndrome
- Types:
  - **preductal (infant)**
    - right-to-left shunt + obstruction
  - **juxtaductal (adult)**
    - obstruction
    - ↑ BP in upper part vs. ↓BP in lower part of body
  - *Postductal*
    - *left-to-right shunt + obstruction*

# Patent ductus arteriosus

= persistence of ductus arteriosus after 14. day of life

- More common in preterm infants
- “Locomotive” (machinery) murmur
- Typically asymptomatic for a long time, but can later lead to heart failure

*left-to-right shunt*

# **Ebstein anomaly of tricuspid valve**

- Association with ASD
- Symptoms:
  - tricuspidal insufficiency
  - arrhythmia

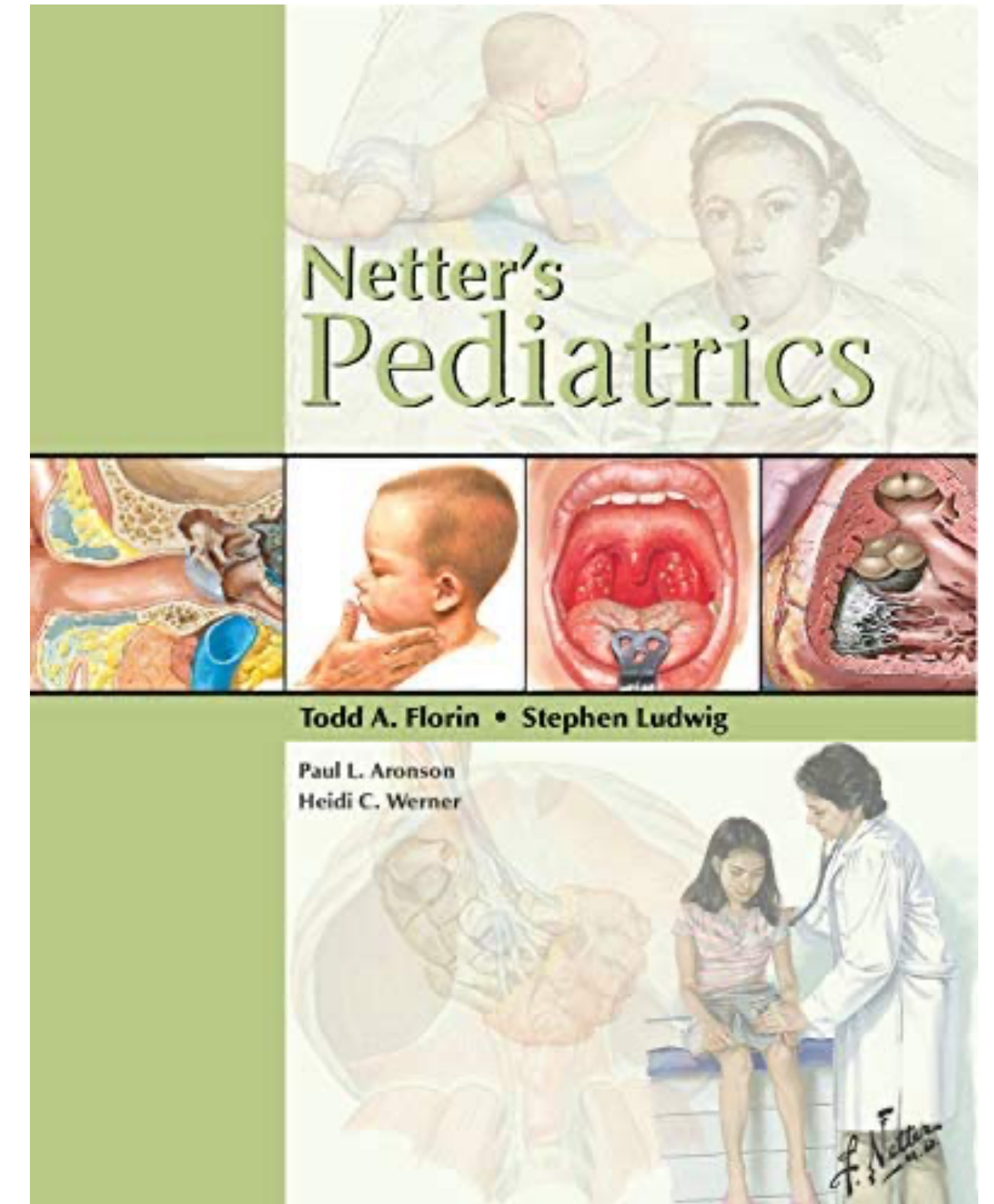
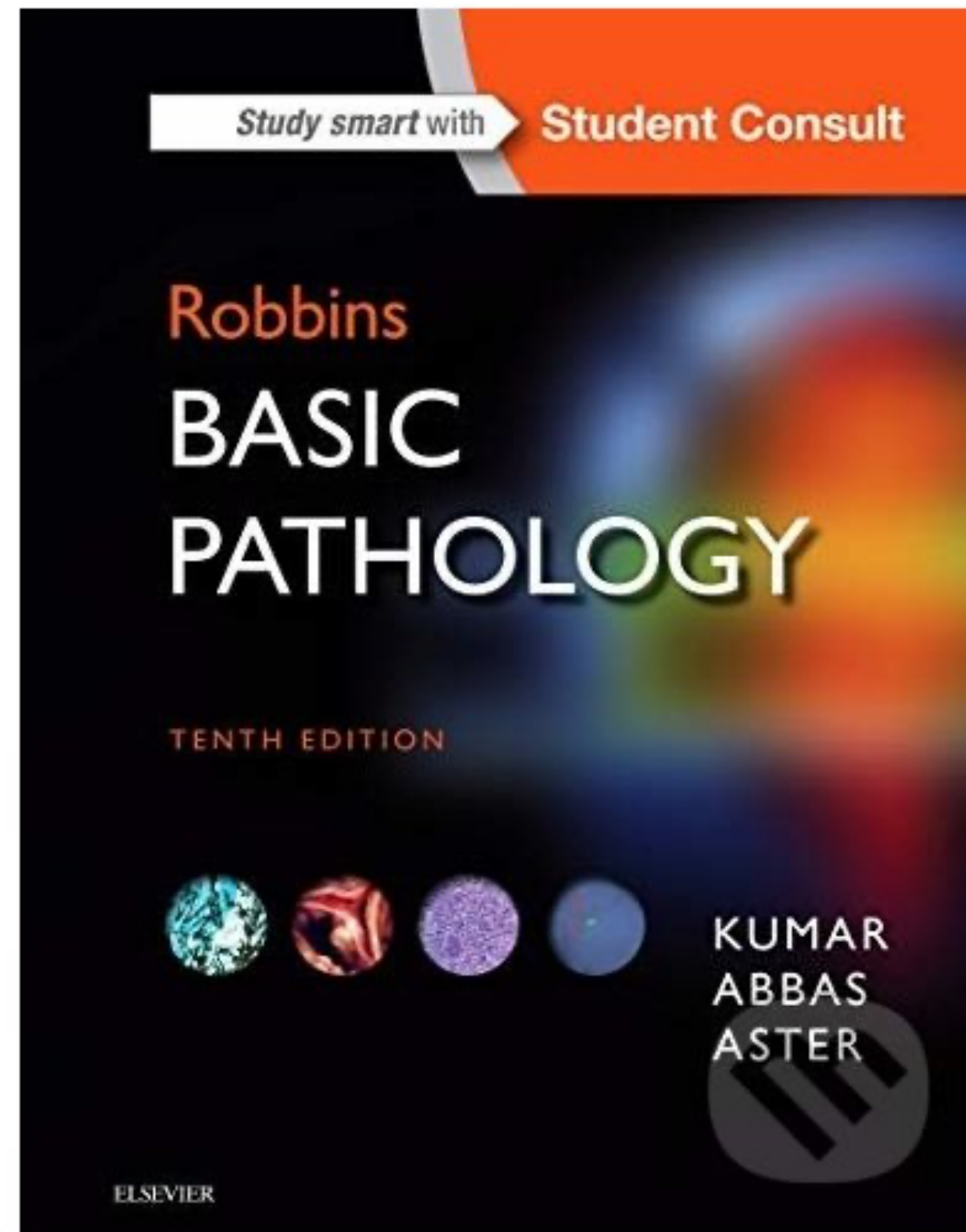
# **Anomalous pulmonary vein return**

- association with ASD

# “Functionally single ventricle”

- Group of CHDs:
  - Double inlet ventricle (DILV)
  - **Hypoplastic left heart syndrome (HLHS)**
  - Hypoplastic right heart syndrome (HRHS)

...more in...



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