Overview of the Most Important Heart Defects

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Ideal for studying: We have compiled an overview of the most important heart defects for you.

Patent Ductus Arteriosus

Auscultatory findings
**Small Ductus**

- Systolic murmur in the parasternal 2nd ICS

**Bigger Ductus**

- “Machinery murmur” with systolic crescendo and diastolic decrescendo

**Etiology**

- Congenital
- Up to 10% of all congenital heart defects in adult age
- In premature babies often because of immaturity of the child, in mature newborns structural anomalies more likely

**Clinic**

- Dyspnea
- Weight faltering
- Lower nourishment (it causes brain bleeding and acute renal failure in premature babies, for example)

**Particular Specialties**

It is a left-to-right-shunt, typically with a quick and high pulse. The ECG shows a left ventricular volume load. A spontaneous closure is often in premature babies, but rare in mature newborns.
Atrial Septal Defect (ASD)

Auscultatory Findings

- Systolic murmur in the left parasternal 2nd ICS
- Fixedly split 2nd heart sound in the left 2nd ICS
- Early diastolic interval noise in the left 4th ICS

Etiology

- Congenital
- 10% of the congenital heart defects in adult age (60-70% ASD II (ostium secundum defect), 30% ASD I (ostium primum defect), the rest rare defects)

Clinic

Variable picture due to severity:

- Frequent bronchopulmonary infects
- Restriction in power and rapid fatigability
- Exertional dyspnea
- Palpitations
- Chest pain
- Cerebral insults
- Right cardiac insufficiency
- No cyanosis!

Particular Specialties

It is a left-to-right-shunt. In 80% of the cases of an ASD smaller than 5 mm, a spontaneous closure happens within the first four years of life. The ECG shows a right axis deviation with right heart load. The right ventricle is enlarged in the echo. The pulmonary
trunk is prominent in radiographs and the vascular drawing of the lungs increased. Small shunts often stay without symptoms.

Ventricular Septal Defect

Auscultatory Findings

Depending on size of VSD (the smaller, the louder):

**Small defect:**
- Properly split 2nd heart sound in the left 2nd ICS
- Early diastolic rough pressed-steal sound in the left parasternal 3rd to 4th ICS

**Medium-sized defect:**
- 3rd heart sound
- Systolic flow noise in the left parasternal 3rd to 4th ICS
- Early diastolic murmur at the cardiac apex

**Eisenmenger’s VSD:**
- Trumpeting 2nd heart sound in the left 2nd ICS
- 4th heart sound
- Pulmonary ejection sound
- Mesosystolic interval murmur (left parasternal 2nd to 3rd ICS)
- Graham-Steel murmur in pulmonary valve insufficiency
Etiology

- Congenital
- Most common heart defect with about 35% of all congenital heart defects

Clinic

- Cardiac insufficiency
- Tachycardia and tachypnea
- Sweating
- Sucking weakness
- Weight faltering
- Exertional dyspnea
- Recurring bronchopulmonary infects
- Cyanosis
- Dysrhythmia
- Syncopes
- Cerebral abscesses

Particular Specialties

It is a left-to-right-shunt with recirculation of the blood through the pulmonary circulation while the volume of the shunt depends on VSD size and resistance among others. The right ventricle has to cope with higher compressive and volume loads, the left ventricle with higher volumes.

A shunt inversion can appear if the pressure in the right ventricle is bigger than in the left ventricle. A central cyanosis (Eisenmenger’s reaction) emerges that is irreversible in fixed pulmonary hypertonia. An ECG can show right heart loads, as well as left heart loads. A spontaneous closure is possible in small, perimembraneous VSDs.

Pulmonary Stenosis
Auscultatory Findings

- Pulmonary ejection click, early diastolic
- Heart sound widely split and silent participation of the pulmonary valve
- Systolic ejection murmur (p.m. left parasternal 2nd to 3rd ICS, depending on the localization of the stenosis) or systolic vessel murmur at the lungs
- Murmur referred to the back

Etiology

- Congenital
- 10% of congenital heart defects (valvular stenosis)
- 3% of all congenital heart defects (sub/supravalvular stenosis)

Clinic

Clinic due to a permanent smaller cardiac output (depending on the severity of the stenosis):

- Physical fatigability
- Exertional dyspnea
- Heart insufficiency
- Vertigo

Particular Specialties

Left parasternal systolic whirr and elevating pulsations at the inferior sternal border are typical. The ECG can be normal depending on the severity, or show a right axis deviation.
with signs of right heart load.

Tetralogy of Fallot

Auscultatory Findings

- Singular 2nd heart sound
- Rough 2/6 – 3/6 systolic murmur, on auscultation in the left parasternal 2nd to 3rd ICS
- Diastolic reflux noise at the cardiac apex, possibly audible in aortic valve insufficiency or calcified pulmonary valve

Etiology

- Most common congenital cyanotic heart defect (10%)
- Microdeletions on chromosome 22q11 in 15% of the patients

Clinic

- Dyspnea, particularly in exertion (in newborns sucking or crying, for example)
- Hypoxic attacks up to unconsciousness, restriction in power
- Slight developmental delay
- Cyanosis
- Tachypnea
- Drumstick fingers

Particular Specialties

It is a right-to-left-shunt. A right axis deviation typically shows in the ECG and a right heart hypertrophy, as well as the “coeur-en-sabot” in chest radiographs. The crouched position is typical (often in children) to increase the systemic resistance and promote the
pulmonary perfusion. Operative primary corrections are conducted in the age of 6 – 18 months.

Ebstein’s Anomaly

Image: “Internal view of the left ventricle shows an anomalous muscular band that joins the free wall with the ventricular septum (stylus) in a heart with Ebstein’s anomaly.” by Luis Muñoz-Castellanos et al. License: CC BY 2.0

Auscultatory Findings

- SI and SII are widely slit
- SIII and SIV can be heard
- Serial clicks and a mid-diastolic murmur can be auscultated

Etiology

- Congenital
- Rare heart defect (< 1% of the congenital heart defects)

Clinic

Variable picture of a cyanotic heart defect:

- Dyspnea
- Fatigue
- Restriction in power
- Heart pain
- Palpitations
- Hepatomegaly
- “Quiet thorax” (precordium often normal)
Particular Specialties

A right-to-left-shunt that goes with a relocation of the tricuspid valves into the right ventricle. A variable transversal diameter of the heart (extreme cardiomegaly, bocksbeutel-shape possible) can be measured. The anomaly is associated with cardiac dysrhythmia and accessory pathways (WPW syndrome).

Aortic Isthmus Stenosis

![Image: “Aortic Coarctation.” by BioMed Central Ltd., Ntsinjana et al. License: CC BY 2.0]

Auscultatory Findings

- Properly split 2nd heart sound
- Early systolic click
- Mesosystolic murmur (p.m. at the left parasternal 3rd to 4th ICS)
- Murmur also on auscultation between the bladebones
- Diastolic decrescendo in insufficiency of the aortic valve
- Murmurs that can continuously be heard in the area of collateral vessels

Etiology

- Congenital
- 7% of all congenital heart defects

Clinic

Symptoms due to hypertonus of the upper half and hypotonus of the lower half of the body:

- Headache
- Nosebleed
- Vertigo
- Tinnitus
- Warm hands and cold feet up to an intermittent claudicatio

Particular Specialties

Possibly associated with anomalies like a bicuspid aortic valve. An aortic valve insufficiency can emerge out of it. Epsilon sign/triple sign in kinking or double contours of
the ascendant aorta visible in radiographs. Blood pressure differences over 20 mmHg between the upper and lower or right and left half of the body are typical.

Congenital Valvular Aortic Stenosis

Auscultatory Findings

Rough systolic murmur (p.m. in the right parasternal 2nd ICS)
- Whirr in the jugulum
- Systolic murmur referred to the carotids
- Paradoxically split heart sound
- Early systolic ejection click

Etiology

- Congenital
- 6 – 10% of the congenital heart defects

Clinic

Symptoms depending on severity:
- Mild stenosis are often without symptoms
- Exertional dyspnea
- Cardiac dysrhythmia
- Anginous pain
- Vertigo and syncopes
Particular Specialties

The critical aortic stenosis of newborns expresses itself within the first days to weeks of life and leads to a cardiac insufficiency. Signs of this valvular high-grade stenosis are a greyish skin color and flat pulses.

Transposition of the Great Arteries (TGA)

Auscultatory Findings

Complete transposition:
- Quiet systolic murmur in the left 2nd ICS
- Loud or singular 2nd heart sound
- Further auscultatory findings depending on further existing anomalies

Etiology

- Congenital
- 5% of all congenital heart defects (complete transposition) and 1% of all
congenital heart defects (corrected transposition)

Clinic

Complete transposition:

- Cyanosis
- Right cardiac insufficiency within the first days of life

Corrected transposition:

- Normal during childhood
- Cardiac dysrhythmia later (AV block, WPW syndrome)

Particular Specialties

It is a right-to-left-shunt. The ductus arteriosus has postnatally to be kept open with prostaglandin E to connect both circulations until an arterial switch surgery is conducted. “Egg-shape” with bilateral extension of the heart in radiographs.

Aortic Valve Insufficiency

Auscultatory Finding

- Whispering or pouring-flowing decrescendo murmur directly after the 2nd heart sound (best to be heard in the left parasternal 3rd ICS in forwards bent patients)
- Fusiform systolic murmur, possibly audible due to increased stroke volume
- Austin-Flint murmur (rumbling late diastolic murmur) possibly audible due to obstruction of the anterior mitral valve cusp
Etiology

Acute aortic valve insufficiency:
- Aneurysm/dissection of the proximal aorta
- Thorax trauma
- Acute endocarditis

Chronic aortic valve insufficiency:
- Structural valvular changes (e.g. bicuspid aortic valve, rheumatic fever)
- Functional insufficiency due to dilatation of the aortic valve ring (e.g. arterial hypertonia, aortic aneurysm, syphilis)
- Prolapse of an aortic valve cusp/valve rupture

Clinic

Acute aortic valve insufficiency:
- Acute left cardiac insufficiency
- Lung edema
- Cardiogenic shock

Chronic aortic valve insufficiency:
- Without symptoms for a long time
- Angina pectoris
- Exertional dyspnea
- Palpitations
- Syncope (rare)
- Dysrhythmia (rare)
- Sudden cardiac death (rare)

Particular Specialties

In the acute form, the ventricle has no time to adapt to the increased volume load. Quick and high pulse, as well as a high blood pressure amplitude with isolated systolic hypertonia and visible pulsation of the carotids, are typical. The radiograph shows a cardiomegaly in “coeur-en-sabot shape”. If a higher-grade insufficiency exists for a long period of time, also a surgical valve replacement cannot stop the cardiac insufficiency because of too big myocardial damage.

We also recommend the full article about aortic valve insufficiency.

Aortic Valve Stenosis
Auscultatory Findings

- Rough and fusiform systolic murmur with p.m. in the right parasternal 2nd ICS
- Murmur referred to the carotids
- Aortic valve closing sound is weakened with eventual paradox splitting of the 2nd heart sound in higher-grade stenosis
- Heart sound is breath-variably split

Etiology

Valvular aortic valve stenosis:

- Calcifications in old age
- Early degeneration of a congenital bicuspid valve
- Postrheumatic

Subvalvular aortic valve stenosis:

- Thickened septum in HOCM
- Fibrous ring with restraint of the outflow tract of the left ventricle

Supravalvular stenosis:

- Williams-Beuren’s syndrome (calcium metabolic disorder)
- Hourglass-shapedly constrained ascending aorta

Clinic

- Low-grade stenosis, often without symptoms (symptoms in an opening area < 1.0 cm²)
- Limited resilience
- Exertional dyspnea
- Angina pectoris
- Vertigo and syncopes
- Sometimes palpitations
Particular Specialties

Slow and flat pulse and elevating apical impulse are typical. Arterial blood pressure and blood pressure amplitude decrease with falling stroke volume. The radiograph shows a sinistral extension only in decompensation. If an aortic valve stenosis develops symptoms, the prognosis is very bad (two-year survival rate lower than 50%).

We also recommend the full article about aortic valve stenosis.

Mitral Valve Stenosis

Auscultatory findings

- Trumpeting 1st heart sound
- Diastolic decrescendo murmur (p.m. at the cardiac apex)
- Afterwards presystolic crescendo murmur (only in sinus rhythm)
- Mitral opening sound
- Graham-Steel murmur in pulmonary insufficiency

Etiology

- Rarely congenital
- Rheumatic fever (may lag years)

Clinic

- Symptoms depending on the remaining opening area
- Loss of power
- Fatigue
- Dysrhythmia
- Cyanosis (peripheral) and mitral face (flush of the cheeks)
- Dyspnea
• Congested liver and kidney
• Cardiac asthma
• Jugular vein congestion
• Peripheral edemas

Particular Specialties

The left atrium can be enlarged in radiographs, as well as show double contours at the right heart border. The picture of a “standing egg-shape” results from this enlargement, as well as from hypertrophy of the right ventricle and a widened pulmonary artery.

We also recommend the full article about mitral valve stenosis.

Mitral Valve Insufficiency

Auscultatory Findings

• Quiet 1st heart sound
• Systolic murmur directly after the 1st heart sound (high frequency, ribbon-like)
• P.m. at the cardiac apex
• Murmur referred to the axilla
• Short diastolic interval murmur in higher-grade insufficiency
• 3rd heart sound can be on auscultation
Etiology

Relative mitral valve insufficiency:

- Dilated mitral valve ring, for example, in dilated cardiomyopathy or left cardiac insufficiency

Acute mitral valve insufficiency:

- Avulsion of tendinous chords or papillary muscles: e.g. in myocardial infarction, mitral valve prolapse, trauma
- Rheumatic or bacterial endocarditis

Chronic mitral valve insufficiency:

- Chronic-degenerative changes
- Calcification
- CHD
- Cardiomyopathy
- Autoimmunological or collaginous diseases

Clinic

Acute mitral valve insufficiency:

- Acute cardiac insufficiency
- Lung edema
- Cardiogenic shock

Chronic mitral valve insufficiency:

- Without symptoms for a long time
- Dyspnea
- Palpitations
- Cough attacks at night
- Fatigue

Particular Specialties

The function, as well as morphology of the left ventricle, can be evaluated with echocardiography. The additional color Doppler function gives information about systolic reflux.

We also recommend the full article about mitral valve insufficiency.

Mitral Valve Prolapse
Auscultatory Findings

- Late systolic murmur (p.m. at the cardiac apex)
- Middle to late diastolic click (time-wise shifting depending on the patient’s position or different maneuvers possible)

Etiology

Idiopathic (primary) mitral valve prolapse:

- Imbalance of the sizes of valves, valve supporting structures and ventricles

Secondary mitral valve prolapse:

- CHD
- Atrial septal defect
- Dilated or hypertrophic cardiomyopathy
- Myocarditis
- Marfan’s or Ehler-Danlos's syndrome (weakness of the connective tissue)

Clinic

- Often without symptoms
- Dysrhythmia with extra systoles and tachycardia
- Palpitations
- Syncope
- Dyspnea
- Decreased resilience
- Fatigue
- Angina pectoris

Particular Specialties

Sure diagnosis only possible with echocardiography. The “hammock phenomenon” shows here if the thickened mitral valve cusps prolapse into the left atrium during systole. Women are more often affected than men.

We also recommend the full article about mitral valve prolapse syndrome.
Pulmonary Valve Insufficiency

Auscultatory Findings

- Diastolic murmur (p.m. in the left parasternal 2nd ICS)
- Widely split 2nd heart sound

Etiology

- Changed valvular morphology (e.g. endocarditis from intravenous drug abuse)
- Dilated ventricle
- Relative pulmonary insufficiency in pulmonary hypertonia

Clinic

- Signs of right cardiac insufficiency
- Venous congestion
- Hepato- or splenomegaly
- Gastrointestinal disorders
- Fatigue
- Ascites
- Edema
Particular Specialties

The diagnosis is made with echocardiography. The pulmonary main stem is dilated.

Course tip: [Cardiology by Prof. Joseph Alpert, Lecturio USMLE](#)

References

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