CARDIAC STRUCTURAL DEFECTS

(CHAPTER 25)

QUESTION 1

List some genetic factors leading to congenital structural disorders.

CHD can be caused by any type of **genetic mutation**:

deletion – GA**T**GCA → GAGCA

insertion – GATGCA → GAT**C**GCA

substitution – GA**T**GCA → GA**C**GCA

duplication – GA**TGC**A → GA**TGCTGC**A

etc...

QUESTION 2

List some environmental factors that can cause structural disorders.

CHD can also be caused by **environmental factors** that the fetus is exposed to during development.

The heart is one of the **first organs** to develop, roughly during weeks **3-7** of gestation.

Maternal use of **drugs** and **alcohol** can contribute, including **tobacco** (even second-hand.)

QUESTION 3

List some maternal factors that can cause structural disorders.

Viral infection during early pregnancy, particularly with rubella/CMV, is a known risk factor.

Obesity, **diabetes mellitus**, and other metabolic disorders can contribute to structural disorders.

Advanced maternal age (above 35) also confers a greater risk of birth defects.

QUESTION 4

Describe the cause of a heart murmur.

Heart murmurs are caused by **turbulent blood flow** through one or more heart valves, which can be caused by a heart defect.

The **pattern** and **amplitude** of the murmur are determined by the nature of the abnormality.

Functional (benign) **murmurs** can occur in infants in the absence of any defect, are mild, and usually resolve over time.

QUESTION 5

What is the most reliable diagnostic tool for congenital heart disease (CHD?)

Echocardiography (ultrasound) is the preferred technique for diagnosing heart defects.

Transthoracic echocardiography (TTE) is non-invasive, but transesophageal echocardiography (TEE) often produces better images and may be more useful diagnostically.

QUESTION 6

What is the most common type of critical congenitive heart disease (CCHD,) and which defects cause it?

The most common forms of CCHD are **increased pulmonary blood flow defects**, meaning that they cause **more** blood than normal to be forced into the lungs.

These all involve some form of **left-to-right shunting** in which blood is able to move from the **left** side of the heart to the **right** side.

Three of these defects are described in the next question.

QUESTION 7

Describe atrial septal defect (ASD,) ventricular septal defect (VSD,) and patent ductus arteriosus (PDA.) Why are these defects so problematic?

Septal defects refer to disorders where the **septum** (wall) separating the left and right sides of the heart is incomplete.

An **atrial** septal defect (ASD) allows communication between the left and right **atria**.

A **ventricular** septal defect (VSD) allows communication between the left and right **ventricles**.

The left side of the heart is **stronger** than the right side, so when the heart contracts, the left side will overpower the right and some of the blood will be forced **through** the defect and into the right atrium/ventricle.

Before we talk about the third defect (PDA,) let's talk about what the **ductus arteriosus** actually is.

When a fetus is in the womb, it (obviously) **doesn't breathe**, because it is submerged in amnotic fluid.

All of its oxygen comes from the **mother's lungs** and passes through the placenta and the umbilical vein.

Because all its O_2 comes from the mother, the fetus's lungs remain **non-functional** until it is born.

The blood leaving the right ventricle doesn't need to go to the lungs because it is **already oxygenated**.

A small passage called the **ductus arteriosus** connects the **pulmonary artery** to the **aorta**, bypassing the lungs and allowing both ventricles to pump blood into the systemic circulation **together**.

The ductus arteriosus normally **closes** shortly after birth, when the baby's lungs start working.

In some cases, it can remain **patent** (open,) allowing **deoxygenated** blood to circumvent the lungs.

All three of these (ASD, VSD, and PDA) are an issue because they break down the separation between **oxygenated** and **deoxygenated** blood, allowing them to mix with each other.

QUESTION 8

Describe defects that lead to decreased pulmonary blood flow. Why are these defects so problematic?

A few heart defects have the opposite effect of septal defects: **decreasing** the amount of blood flowing into the lungs from the heart.

These include two forms of **atresia**, in which a particular heart valve is **missing**.

Pulmonary atresia allows the force of the left atrium to push blood in the pulmonary artery back into the right ventricle.

Tricuspid atresia allows the force of the right ventricle to push blood back into the right atrium.

In **pulmonary stenosis**, the pulmonary artery is **narrowed**, decreasing the amount of blood that can be pushed through into the lungs.

This is one of the four defects seen in the **tetralogy of Fallot**, which we'll cover in the next question.

All of these defects **reduce** pulmonary blood flow, making it more difficult to oxygenate blood.

QUESTION 9

Describe the tetralogy of Fallot.

The **tetralogy of Fallot** ("tetra-" as in **four**) is a set of four closely-linked heart defects which often occur together, especially in cases of Down syndrome and DiGeorge syndrome.

The four defects are:

- **VSD** – "hole" between the left and right ventricles
- **pulmonary stenosis** – narrowed pulmonary artery
- **right ventricular hypertrophy** (RVH) – thickened wall of RV (more muscle, smaller chamber)
- **overriding aorta** – aorta connects to **both** ventricles

QUESTION 10

Describe hypoplastic left heart syndrome (HLHS.)

Hypoplastic left heart syndrome is another complex set of heart deformities, which includes:

- **ASD** – "hole" between the left and right atria
- **PDA** – "hole" between pulmonary artery and aorta
- **hypoplastic** (small and underdeveloped) left ventricle

QUESTION 11

What does "mixed" refer to in mixed blood flow defects?

Mixed blood flow refers to defect such as PDA, septal defects, and overriding aorta in which deoxygenated and oxygenated blood **mix together**, leading to inefficient oxygenation of tissues.

QUESTION 12

What does total anomalous pulmonary venous return (TAPVR) refer to? Which type of hemoglobin would you expect to see in the pulmonary veins?

TAPVR is a **very rare** heart defect in which all four **pulmonary** veins (carrying oxygenated blood) communicate with the **systemic** veins (carrying deoxygenated blood.)

This results in oxygenated blood leaking into the systemic venous circulation, meaning **less** oxygenated hemoglobin and **more** deoxygenated hemoglobin entering the left atrium.

QUESTION 13

List the signs and symptoms of critical congenital heart disease (CCHD.)

- **Decreased oxygen saturation** and associated **dyspnea**
- **Tachpynea** + **dyspnea** → **impaired feeding**
- **Cyanosis** from poor blood perfusion

(Occasional fatigue, dypsnea, or feeding problems are not uncommon in infants, but these will be **persistent**)

QUESTION 14

How does rheumatic fever affect the heart with repeated infection?

Rhuematic heart disease occurs when an **inflammatory response** causes permanent damage (**scarring**) to the heart valves.

It is a sequela of **rheumatic fever**, a disease which can develop in the weeks following an untreated **streptococcal URI** (strep throat.)

QUESTION 15

Describe endocarditis.

Endocarditis in an inflammation of the **inner layer** of tissue lining the chambers of the heart, often also involving the **heart valves**.

It can either be caused by **infection** (typically bacterial) or non-infective causes (such as hypercoagulation or lupus.)

QUESTION 16

Why would aortic valve calcification be a problem?

Calcification of the aortic valve can lead to **aortic stenosis**, which diminishes the bloodflow out of the left ventricle and into the systemic circulation.

Aortic stenosis also significantly increases the risk of other cardiovascular problems, such as **myocardial infarction** and **cardiac-related mortality**.