Plasma lipids and lipoproteins

By Dr. Yasmine Sami

Medical biochemistry 2022-2023

Intended learning outcomes

- Describe disorders of lipid metabolism
- Illustration of case study

Lipid profile

- Lipids are essential for health, but raised plasma **cholesterol** and **triglyceride** concentrations are associated with an increased incidence of **cardiovascular disease**.
- Conversely, plasma **HDL cholesterol** is cardioprotective, partly because of its central role in **reverse cholesterol** transport returning cholesterol from the tissues to the liver.

Disorders of lipid metabolism

- There are many cases of **secondary hyperlipidaemias**, including obesity, alcohol excess, diabetes mellitus, hypothyroidism, chronic kidney disease and cholestasis.
- The genetic hyperlipidaemias include:
  - Familial hypercholesterolaemia (FH)
  - Familial combined hyperlipidaemia (FCH)
  - Type III hyperlipoproteinaemia.
- Familial hypercholesterolaemia usually results from a defect of the LDL receptor.

Disorders of lipid metabolism

Some causes of raised plasma high-density lipoprotein (HDL) cholesterol

Primary

- Hyperalphalipoproteinaemia
- Cholesterol ester transfer protein deficiency

Secondary

- High ethanol intake
- Exercise
- Certain drugs, e.g. estrogens, fibrates, nicotinic acid, statins

Disorders of lipid metabolism

Some important causes of secondary hyperlipidaemia

Predominant hypercholesterolaemia

- Hypothyroidism
- Nephrotic syndrome
- Cholestasis, e.g. primary biliary cirrhosis
- Acute intermittent porphyria
- Anorexia nervosa/bulimia
- Certain drugs or toxins, e.g. ciclosporin and chlorinated hydrocarbons

Disorders of lipid metabolism

Some important causes of secondary hyperlipidaemia

Predominant hypertriglyceridaemia

- Alcohol excess and Obesity
- Diabetes mellitus and metabolic syndrome
- Certain drugs, e.g. estrogens, protease inhibitors and glucocorticoids
- Chronic kidney disease
- Some glycogen storage diseases, e.g. von Gierke’s type I
- Systemic lupus erythematosus
- Paraproteinaemia

Case 1: Description

A 23-year-old woman had her plasma lipids checked by her general practitioner because:

- **Her father had died of a myocardial infarction aged 44 years.**
- **Her 24-year-old brother had hyperlipidaemia.**
- **Her renal, liver and thyroid function tests were normal, as was her blood glucose.**

Results of laboratory tests

Plasma (fasting)

Cholesterol 11.4 mmol/L (3.5–5.0)

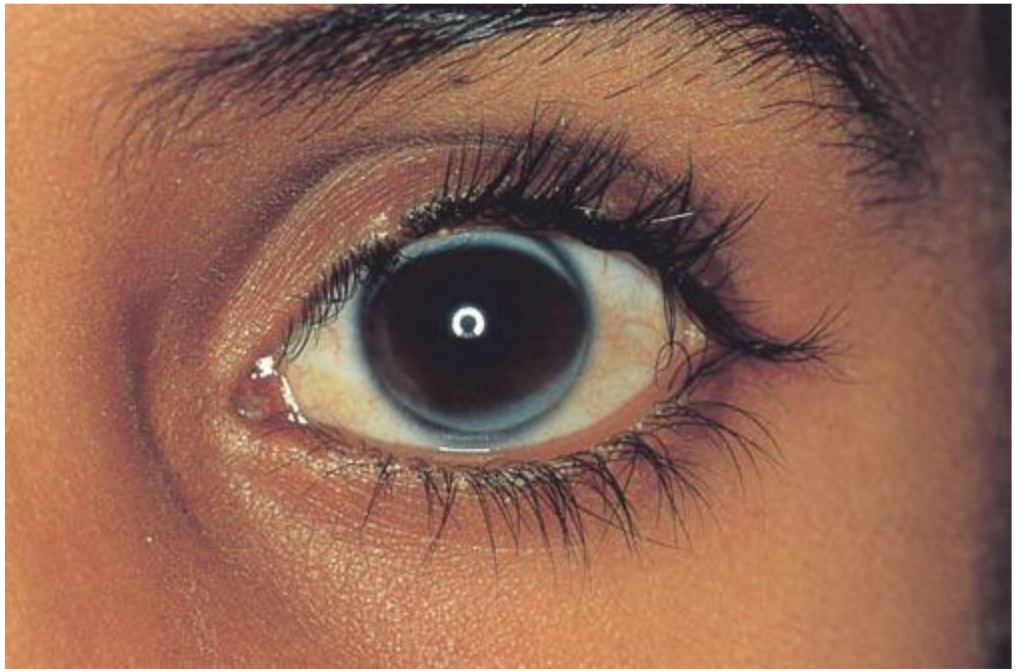
Triglyceride 1.1 mmol/L (0.3–1.5)

HDL cholesterol 1.2 mmol/L (1.0–1.8)

On examination, she had tendon xanthomata on her Achilles tendons and bilateral corneal arci.

Examination

Tendinous xanthomas in familial hypercholesterolaemia



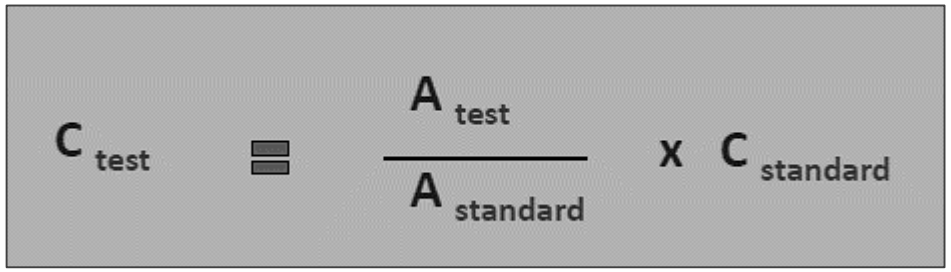
Corneal arcus in familial hypercholesterolaemia

Discussion of the case

- Note the considerably **raised plasma cholesterol** concentration. The absence of an obvious secondary hyperlipidaemia, in conjunction with the **family history** of a first-degree relative with premature cardiovascular disease and hyperlipidaemia, suggests a genetic hyperlipidaemia.
- The presence of **tendon xanthomata** and **premature corneal arci** supports the diagnosis of familial hypercholesterolaemia.
- This is usually an autosomal dominant disorder and usually a defect of the low-density lipoprotein (LDL) receptor

Important definitions

Calculation of cholesterol, triglyceride and HDL:



- VLDL = TG/5

- LDL = TG – (HDL + TG/5)

THANK YOU !

ANY QUESTIONS ??

PLEASE ASK