Clinical case study
Lipid disorder

By

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CASE 1

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.

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.

DISCUSSION

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.

CASE 2

A 43-year-old man attended the vascular surgery outpatient clinic for peripheral vascular disease. He was a non-smoker but had undergone a coronary artery bypass graft the year before. Some of his laboratory results were as follows:

Plasma (fasting)

1. Cholesterol 8.7 mmol/L (3.5–5.0)
2. Triglyceride 9.1 mmol/L (0.3–1.5)
3. HDL cholesterol 0.86 mmol/L (1.0–1.8)
4. On examination, he had tuberous xanthomata and palmar striae.

DISCUSSION

The diagnosis was type III hyperlipoproteinaemia (familial dysbetalipoproteinaemia or broad beta-hyperlipidaemia). Note the mixed hyperlipidaemia (both cholesterol and triglyceride concentrations raised) in an approximately 1:1 molar ratio. The type III hyperlipoproteinaemia is associated with raised concentration of remnant lipoprotein particles, which are particularly atherogenic. Note also the characteristic lipid stigmata and premature peripheral vascular and coronary heart disease.

CASE 3

A 15-year-old woman presented to the surgical unit with acute pancreatitis. Some of her laboratory results were as follows:

Plasma (fasting)

1. Cholesterol 33.4 mmol/L (3.5–5.0)
2. Triglyceride 69.1 mmol/L (0.3–1.5)
3. HDL cholesterol 0.9 mmol/L (1.0–1.8)
4. Amylase < 20 U/L (<200)
5. On examination, she had eruptive xanthomata on her arms and thighs and fundoscopy revealed lipaemia retinalis.

DISCUSSION

This patient has grossly elevated lipid concentrations with severe hypertriglyceridaemia. The blood sample would be lipaemic and some plasma sodium assays (indirect ion electrodes) may show pseudohyponatraemia. She was found to have lipoprotein lipase deficiency when this enzyme was measured before and after heparin administration, which releases the enzyme from capillaries into the circulation. Lipoprotein lipase deficiency can result in the chylomicron syndrome and eruptive xanthomata may be present. Plasma amylase concentration is normally elevated in acute pancreatitis but, due to the gross lipaemia, the assay was unsatisfactory, giving a spuriously low result. The latter is an important practical point and a spot urinary amylase may be preferable, or assay of plasma amylase after separation from the lipid fraction, under such circumstances.

Thank you for your attention