Primary mixed hyperlipidemia

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It is the most common genetic disorder of lipoprotein metabolism . The frequency is estimated at 1: 100 to 1:50. Heredity is usually marked as autosomal recessive.

Clinical manifestations

It often occurs in obese and diabetics . There are no xanthomas or arcus corneae; pathological manifestations of atherosklerosis (coronary heart disease, lower limb ischemia) do not begin until adulthood.

Biochemical findings

An abnormal lipid finding is usually not detected until adulthood. The serum is clear or opalescent. VLDL (pre-β-lipoproteins), also LDL (β-lipoproteins) and apoprotein B are elevated, [cholesterol]] is between 10-15 mmol / l, triacylglycerols are between 2.26-5.65 mmol / l. HDL-cholesterol and apoprotein C-II and C-III are usually reduced. Lipoprotein electrophoresis shows familial combined hyperlipoproteinemia such as type IIb, IV or even IIa or V. Sometimes another fraction of pre-α (pre-α1 and pre-α2) is evident, caused by an increase in lipoprotein (a) [Lp (a)]. Chylomicrons are not detected on an empty stomach.

Pathobiochemistry

The cause is thought to be an abnormally high synthesis of Apo B in the liver, accompanied by increased VLDL production.

Prognosis

A common complication is myocardial infarction before the age of 60; an association with diabetes and obesity is common.

Healing

- Above all, lifestyle modification: weight reduction, diet with lower fat content (preference for fat with unsaturated fatty acids instead of saturated ones) - reduction of cholesterol intake.
- Drug therapy only in patients for whom lifestyle modification has not been shown to be helpful; fibrates, are most often used , ev.resins, (e.g. Lipanthyl® or Gevilon® in combination with Colestide®); sometimes nicotic acid helps.

Familial dysbetalipoproteinemia (ie type III hyperlipoproteinemia, increase in β-VLDL)

Dysbetalipoproteinemia (type III hyperlipidemia) is a rare inherited disorder characterized by a defect in the removal of chylomicron and VLDL residues. The underlying disorder is homozygosity for the mutant form of apo E (apo E 2), which binds poorly to liver receptors. As a result, chylomicron residues accumulate as well as cholesterol-rich VLDL (β-VLDL)[1].

Clinical manifestations

- Various forms of xanthomas dominate :
  - tuberous xanthomas (in 80%),
  - palmar xanthomas (70%) - are characteristic,
  - tendon xanthomas (30%),
  - eruptive xanthomas (rare).
- Hyperuricaemia and diabetes are observed in about half of patients.
- Early atherosclerotic changes first affect the lower limbs and coronary arteries (in men before the age of 40, in women before the age of 50).

Biochemical findings

Opalescent serum; increased both cholesterol and triacylglycerols: S-cholesterol usually above 7.5 mmol / l, sometimes up to 25 mmol / l, S-triacylglycerols 2-10 mmol / l, rarely 20 mmol / l.

Characteristic appearance of ELFO-lipoproteins: "broad" β-fraction (merging pre-β and β fractions). There is an abnormal fraction between VLDL and LDL (so-called β-VLDL) on the polyacrylamide gel. An increase in the cholesterol / triacylglycerol ratio to> 0.30, a decrease in HDL and LDL cholesterol and, conversely, an increase in VLDL, IDL and chylomicron residues are characteristic.

Links

Source

- MASOPUST, Jaroslav and Richard PRŮŠA. Pathobiochemistry of metabolic pathways. 1st edition. Prague:

Charles University, 1999. 182 pp. ISBN 80-238-4589-6 .

Reference

1. BURTIS, Carl A, Edward R ASHWOOD a David E BRUNS. *Tietz textbook of clinical chemistry and molecular diagnostics.* 4. vydání. St. Louis, Mo : Elsevier Saunders, 2006. 2412 s. s. 930. ISBN 978-0-7216-0189-2.