

Dysbetalipoproteinemie

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

Related articles

- Disorders of lipid metabolism (detailed)
- Lipoproteins

Source

- MASOPUST, Jaroslav - PRUSA, Richard. Pathobiochemistry of metabolic pathways. 2nd edition. Charles University, 2004. 208 p.

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.

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