

# Dysbetalipoproteinemia

Template:Zkontrolováno

**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<sub>2</sub>), which binds poorly to the liver receptors. As a result, chylomicron residues accumulate as well as cholesterol-rich VLDL ( $\beta$ -VLDL)<sup>[1]</sup>.

## Clinical manifestations

- Various forms of xanthomas:
  - 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"  $\beta$ -fraction (merging pre- $\beta$  and  $\beta$  fractions).

There is an abnormal fraction between VLDL and LDL (so-called.  $\beta$ -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 – PRŮŠA, Richard. Patobiochemie metabolických drah. 2. vydání. Univerzita Karlova, 2004. 208 s.

## Reference

1.

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

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