Familial Hypertriglyceridemia

Alternative Names
FHTG

Record Category
Disease phenotype

WHO-ICD
Endocrine, nutritional and metabolic diseases > Metabolic disorders

Incidence per 100,000 Live Births
Unknown

OMIM Number
145750

Mode of Inheritance
Autosomal dominant

Gene Map Locus
15q11.2-q13.1

Description
Familial hypertriglyceridemia is an uncommon primary (genetic) dyslipidemia characterized by moderately elevated serum VLDL triglycerides, larger triglyceride-rich particles, and an increased triglyceride-to-apoB ratio usually in the absence of significant hypercholesterolemia (lipoprotein phenotype IV) and rarely manifests in childhood. VLDL-triglyceride turnover studies showed that the metabolic defect in patients with familial hypertriglyceridemia is triglyceride overproduction driving an increase in large triglyceride-enriched VLDLs.

Generally, hypertriglyceridemia is a heterogeneous anomaly, not only due to different underlying pathophysiological mechanisms, but also in terms of cardiovascular risk. However, in familial hypertriglyceridemia, cardiovascular risk is apparently only moderately affected.

Molecular Genetics
Familial hypertriglyceridemia has been suggested to be an autosomal dominant condition with low penetrance before age 30, but so far the underlying defective gene has not been elucidated. Candidate genes for FHTG have included those most directly involved in the production or catabolism of triglyceride-rich lipoproteins. Of these, LPL and apolipoprotein A-I/C-III/A-IV genes might be involved in familial clustering of hypertriglyceridemia.

Epidemiology in the Arab World
Kuwait
Tas (1989) studied the association between a single nucleotide substitution in the 3-prime untranslated region of the APOC3 gene with hypertriglyceridemia in Arabs living in Kuwait. In a three-generation family, the allele was found to be co-inherited with increased serum triglycerides and demonstrated a gene dosage effect.

References
Taş S. Strong association of a single nucleotide substitution in the 3'-untranslated region of the apolipoprotein-CIII gene with common hypertriglyceridemia in Arabs. Clin Chem. 1989; 35(2):256-9. PMID: 2914370

Related CTGA Records
Apolipoprotein C-III

External Links
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=413

Contributors
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