Burger-Grutz Syndrome

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History:
Hyperlipidemia, hyperlipoproteinemia, or hyperlipidaemia (British English) is the condition of abnormally elevated levels of any or all lipids and/or lipoproteins in the blood. It is the most common form of dyslipidemia (which also includes any decreased lipid levels). Lipids (fat-soluble molecules) are transported in a protein capsule, and the size of that capsule, or lipoprotein, determines its density. The lipoprotein density and type of apolipoproteins it contains determines the fate of the particle and its influence on metabolism. Lipid and lipoprotein abnormalities are common in the general population, and are regarded as a modifiable risk factor for cardiovascular disease due to their influence on atherosclerosis. In addition, some forms may predispose to acute pancreatitis. Classification Hyperlipidemias may basically be classified as either familial (also called primary) caused by specific genetic abnormalities, or acquired (also called secondary) when resulting from another underlying disorder that leads to alterations in plasma lipid and lipoprotein metabolism. Also, hyperlipidemia may be idiopathic, that is, without known cause. Hyperlipidemias may also be classified directly into which ...

Symptoms:
Abdominal pain (from pancreatitis), lipemia retinalis, eruptive skin xanthomas, hepatosplenomegaly

Diagnosis:
The diagnosis of familial LPL deficiency is based on the assay of LPL enzyme activity in plasma following intravenous administration of heparin. Detection of very low or absent LPL enzyme activity in an assay system that contains either normal plasma or apoprotein C-II and excludes hepatic lipase is diagnostic of familial LPL deficiency. Molecular genetic testing of LPL is available on a clinical basis.

Interesting Facts!
Mutations in the LPL gene cause familial lipoprotein lipase deficiency. The LPL gene provides instructions for producing an enzyme called lipoprotein lipase. This enzyme helps break down fat-carrying molecules (lipoproteins). Lipoproteins normally carry fat molecules from the intestine into the bloodstream. As lipoproteins are broken down, they release fats that the body uses for energy or puts into storage. Mutations in the LPL gene prevent the enzyme from breaking down lipoproteins effectively. As a result, fatty substances build up in the bloodstream, leading to the signs and symptoms of familial lipoprotein lipase deficiency. This condition affects about 1 per million people worldwide. It is much more common in some areas of the province of Quebec, Canada.

Resources:
- en.wikipedia.org/wiki/Burger-Grutz_syndrome
- www.wrongdiagnosis.com/medical/Burger_grutz_syn drome.htm
- www.steadyhealth.com/encyclopedia/Burger_Grutz_syn drome
- www.steadyhealth.com/whatis_burger_grutz_syn drome_1199014.html