Hypertriglyceridemia (Type IV Familial Dyslipidemia)

Hypertriglyceridemia (Type IV Familial Dyslipidemia) is a disease that presents an autosomal dominant inheritance pattern. The cause of this disease is hepatic overproduction of VLDL and it is related to insulin resistance. Lab findings in patients with this disease are increased VLDL and increased triglycerides (> 1000 mg/dL). Symptoms and findings in Hypertriglyceridemia include acute pancreatitis and eruptive xanthomas. This disease can present an increased risk for coronary artery disease (CAD) and an increased risk of peripheral vascular disease.

PLAY PICMONIC

INHERITANCE

Autosomal Dominant

Domino-road

Hypertriglyceridemia presents an autosomal dominant inheritance pattern. This means that the mutated gene is a dominant gene located on one of the nonsex chromosomes (autosomes). You need only one mutated gene to be affected by this type of disorder. A person with an autosomal dominant disorder, the father has a 50 percent chance of having an affected child with one mutated gene (dominant gene) and a 50 percent chance of having an unaffected child with two normal genes (recessive genes).

pathogenesis

Hepatic Overproduction of VLDL

Liver-ship with several Veiled-Ladybug-devil guards coming out of it

The actual gene defect has not been determined in hypertriglyceridemia, but patients with this condition have an overproduction of VLDL by the liver that leads to many signs and symptoms of importance, such as pancreatitis and heart disease.

Related to Insulin Resistance

Insect-syringe swatted by the Stormtrooper's Resistance-swatter

Insulin usually suppresses hepatic VLDL production. When there is insulin resistance, like in patients with diabetes type 2, insulin is not able to do this, which leads to further overproduction of hepatic VLDL.

Lab Findings

Increased VLDL

Up-arrow Veiled-Ladybug-devil guards

Patients with hypertriglyceridemia will have increased levels of VLDL in the blood due to the unregulated hepatic overproduction.

Increased Triglycerides (> 1000 mg/dL)

Up-arrow Triceratops with >1000-glasses

Blood tests showing moderate to high increase in triglycerides (often > 1000 mg/dL) even after a fast of 12 to 14 hours, with relatively normal cholesterol, are highly suggestive of hypertriglyceridemia.

Symptoms/Findings

Acute Pancreatitis

Acute-angle Pancreas-on-fire

Increased fatty acids such as triglycerides can lead to plasma viscosity, which may induce ischemia in pancreatic tissue and trigger organ inflammation.

Eruptive Xanthomas

Zen-master Kenobi with Eruptive-lava-lamp

Eruptive xanthomas are benign skin lesions that are caused by localized deposition of lipids in the dermis.

Increased Risk for Coronary Artery Disease (CAD)

Up-arrow Risk and Crown-heart Stormtrooper

Elevated levels of fatty acids and lipoprotein remnants such as VLDL can lead to plaque formation that can lead to coronary artery disease.

Increased Risk of Peripheral Vascular Disease

Up-arrow Risk and Clogged Artery-archer in the Periphery

Elevated levels of fatty acids and lipoprotein remnants such as VLDL can lead to plaque formation that can lead to peripheral vascular disease.