Hyperchylomicronemia (Type I Familial Dyslipidemia)

Hyperchylomicronemia is a condition that is inherited in an autosomal recessive manner. It can be caused by lipoprotein lipase deficiency or by an altered apolipoprotein C-II. Lab values show increased chylomicrons, increased triglycerides, and increased cholesterol. Signs and symptoms include pancreatitis, hepatosplenomegaly, eruptive or pruritic xanthomas and a milky white appearance in blood samples. Unlike other dyslipidemias, patients with this disease have no increased risk for atherosclerosis.

PLAY PICMONIC

INHERITANCE

Autosomal Recessive

Recessive-chocolate

Hyperchylomicronemia demonstrates an autosomal recessive inheritance pattern. Recessive inheritance means both genes in a pair must be abnormal to cause disease. People with only one defective gene in the pair are called carriers. These people are most often not affected with the condition. However, they can pass the abnormal gene to their children.

PATHOGENESIS

Lipoprotein Lipase Deficiency (LPL)

Lip-protein-trooper with Lip-laser with Down-arrow

Lipoprotein lipase is also known as capillary lipoprotein lipase (CPL). Lipoprotein lipase breaks down fats. When it is deficient, it causes triglycerides to increase.

Altered Apolipoprotein C-II (Apo C-II)

Altered Apo-lip-protein-trooper with C-shaped-hook and 2-tutu

Apolipoprotein C-II is secreted in plasma where it is a component of very low density lipoproteins and chylomicrons. This protein activates the enzyme lipoprotein lipase in capillaries, which hydrolyzes triglycerides. When apolipoprotein C-II is altered, triglycerides accumulate.

Lab Findings

Increased Chylomicrons

Up-arrow Kylo

Chylomicrons are found in the blood and lymphatic fluid where they serve to transport fat from its port of entry in the intestine to the liver and to adipose tissue. Lipoprotein lipase and apolipoprotein C-II are involved in the break down of fatty acids, when these are deficient or altered, chylomicrons are accumulated and levels increase.

Increased Cholesterol

Up-arrow Cholesterol-burger

Lipoprotein lipase and apolipoprotein C-II are involved in the break down of fatty acids, when these are deficient or altered, cholesterol is accumulated and levels increase.

Increased Triglycerides

Up-arrow Triceratops

Lipoprotein lipase and apolipoprotein C-II are involved in the break down of fatty acids, when these are deficient or altered, triglycerides are accumulated and levels increase.

Symptoms/Findings

Pancreatitis

Pancreas-on-fire

The accumulation of chylomicrons can reduce blood flow through the pancreas, leading to acute pancreatitis.

Hepatosplenomegaly

Liver-and-spleen-balloons

Patients with hyperchylomicronemia may also have an enlarged liver and spleen (hepatosplenomegaly). The higher the levels of fat in the body, the larger the liver and spleen become. As fat levels rise, certain white blood cells called macrophages take in excess fat in an attempt to rid fat from the bloodstream. After taking in fat, the macrophages travel to the liver and spleen, where the fatty cells accumulate.

No Increased Risk for Atherosclerosis

X on top of Clogged-Artery-guy with Up-arrow-risk on Monitor

Unlike other familial dyslipidemias, patients with hyperchylomicronemia do not have an increased risk for atherosclerosis.

Eruptive/Pruritic Xanthomas

Zen-master-Jedi eating a Cholesterol-lava-burger with his Pruritic-prairie-dog

Eruptive/pruritic xanthomas are benign skin lesions that can be described as red-yellow dermal papules during examination that are caused by localized deposition of lipids in the dermis.

Milky White Appearance of Blood When Drawn

Spilled-milk and Syringe

When blood is drawn the plasma may have a milky appearance due to excessive lipids in blood.