Dysbetalipoproteinemia (Type III Familial Dyslipidemia)

Dysbetalipoproteinemia is a condition that is inherited in an autosomal recessive manner. It caused by a defective apolipoprotein E. Lab values show increased chylomicrons and increased VLDL. Signs and symptoms include premature atherosclerosis, tuberoeruptive xanthomas and palmar xanthomas.

PLAY PICMONIC

INHERITANCE

Autosomal Recessive

Chewie eating Recessive-chocolate

Dysbetalipoproteinemia 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

Defective Apolipoprotein E (Apo E)

Defective Apo-lip-protein-trooper due to E-electric-shot

The apolipoprotein E gene provides instructions for making a protein called apolipoprotein E (apoE). This protein combines with fats (lipids) in the body to form molecules called lipoproteins. Lipoproteins are responsible for packaging cholesterol and other fats and carrying them through the bloodstream. Chylomicrons and VLDL utilize apoE as their main ligand for clearance from the circulation. In dysbetalipoproteinemia, there is a defective apolipoprotein E, which causes chylomicrons and VLDL to accumulate.

Lab Findings

Increased Chylomicrons

Up-arrow Kylo-microphone-lightsaber

Chylomicrons are lipoprotein remnants 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. Chylomicrons utilize apoE as their main ligand for clearance from the circulation. In dysbetalipoproteinemia, there is a defective apolipoprotein E, which impairs the clearance of chylomicrons from the circulation, causing their levels to increase.

Increased VLDL

Up-arrow Veiled-Ladybug-devil guard

Like chylomicrons, another lipoprotein remnant that is accumulated in dysbetalipoproteinemia is VLDL (very low density lipoproteins). VLDL utilizes apoE as their main ligand for clearance from the circulation. In dysbetalipoproteinemia, there is a defective apolipoprotein E, which impairs VLDL clearance from the circulation, causing their levels to increase.

Symptoms/Findings

Premature atherosclerosis

Clogged-artery-guy with diapers

The remnant particles accumulated in dysbetalipoproteinemia are susceptible to uptake by macrophages in peripheral tissues, which may become cholesterol-laden foam cells and lead to the development of atherosclerosis earlier in life than usual. Premature atherosclerosis can lead to complications such as myocardial infarction, claudication, and stroke.

Tuberoeruptive xanthomas

Zen-master Yoda on an Inflatable-tube on top of an Eruption

Tuberoeruptive xanthomas are dermal papules with an inflammatory component due to fatty accumulation under the skin. In dysbetalipoproteinemia tuberoeruptive xanthomas usually develop on the elbows.

Palmar xanthomas

Palm-tree and Zen-Master-Snoke

Yellowish lipid deposits in the palmar crease due to fatty accumulation under the skin are characteristic of dysbetalipoproteinemia patients.