Familial Hypercholesterolemia (Type IIa Familial Dyslipidemia)

Familial hypercholesterolemia is a common life-threatening genetic condition that causes high cholesterol. Untreated, it leads to early heart attacks and heart disease. Patients with with familial hypercholesterolemia have a high amount of low density lipoprotein (LDL) or “bad cholesterol” due to a mutation in one of the genes that controls the way cholesterol is cleared by the body. As a result, cholesterol accumulates in the bloodstream and can ultimately build up in the walls of the arteries. Cholesterol build up in the artery wall can lead to problems such as heart attacks and strokes in young adults and even children. Familial hypercholesterolemia is inherited and passed down through families. When one individual with this disease is diagnosed, it is important that all family members are screened for familial hypercholesterolemia.



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INHERITANCE

Autosomal Dominant
Domino
Familial hypercholesterolemia demonstrates an autosomal dominant inheritance pattern, meaning that on average 50% of children of an affected parent will have the disease passed on to them.

PATHOGENESIS

Defective or Absent LDL Receptors
Ladybug-devil guards with Broken and Absent Receptors
Familial hypercholesterolemia patients have defective or absent LDL receptors, which means that they cannot clear LDL from the plasma, causing the LDL levels to rise.

Defective Apolipoprotein B-100 (ApoB-100)
Dizzy Apo-lip-protein trooper with B-bees circling around him while holding 100-dollar-bill
Defects in apolipoprotein B cause defective binding of ApoB-100 with LDL receptors, leading to impairments in LDL catabolism and decreased clearance of circulating LDL.

Lab Findings

Increased LDL
Up-arrow Ladybug-devil guard
Gene mutations make it impossible for patients with Familial Hypercholesterolemia to remove excess low-density lipoproteins (LDL), from the blood stream. LDL delivers hepatic cholesterol to peripheral tissues. LDL is considered as "bad cholesterol" because, over time, the elevated blood cholesterol will lead to blockages in the arteries of the heart and/or brain. The longer a person experiences high LDL, the higher the risk for heart attacks or stroke related to these blockages.

Decreased HDL
Down-arrow Halo-dog-angel
High-density lipoprotein (HDL) transports cholesterol from peripheral tissues back to the liver. It is known as “good cholesterol” because it removes excess cholesterol from the blood. In this disease, HDL levels are decreased.

Type IIb also has Increased VLDL
Luke-hamburger with two-tutu and B-bee on shoulder and Up-arrow Veiled-Ladybug-devil guard
The main difference between types IIa and IIb of Familial Hypercholesterolemia is that in type IIb there can also be an increase in VLDL.

Symptoms/Findings

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Accelerated Atherosclerosis

Clogged-artery-guy running fast

Accelerated atheroscleros is caused by very high levels of LDL. Patients with familial hypercholesterolemia may have a myocardial infarction (MI) before age 20.

Achilles Tendon Xanthomas

Achilles-trooper-tendon and Zen-master Jedi

Achilles tendon xanthomas are painless soft tissue masses occurring most commonly at the distal one-third of the tendon and are usually bilateral and symmetrical. It is characterized by localized accumulation of lipid-laden macrophages, inflammatory cells and giant cells secondary to cholesterol deposition in tissue.

Xanthelasma

Zen-master Jedi with llama

Xanthelasma (also known as xanthelasma palpebra) is a well-circumscribed flat or slightly elevated yellowish growth that typically occurs on or around the eyelids. It is made up of cholesterol deposits that accumulate underneath the skin.

Corneal Arcus

Corn-arch around eye window

Arcus is a deposition of lipid in the peripheral corneal stroma. It starts at the inferior and superior poles of the cornea and in the late stages encircles the entire circumference. Corneal arcus is sometimes indicative of a high LDL with elevated serum cholesterol, especially in patients under 40 years of age. It can be a prognostic factor for coronary artery disease in this age group.

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