Geneticheck - Genetic Report

Gene

LDLR

Associated Diseases

Homozygous Familial Hypercholesterolemia
Hypercholesterolemia, Familial, 1

Phenotype

Abnormal nervous system physiology
A functional anomaly of the nervous system.

Renal steatosis
Abnormal fat accumulation in the kidneys.

Xanthomatosis
The presence of multiple xanthomas (xanthomata) in the skin. Xanthomas are yellowish, firm, lipid-laden nodules in the skin.

Tendon xanthomatosis
The presence of xanthomas (intra-and extra-cellular accumulations of cholesterol) extensor tendons (typically over knuckles, Achilles tendon, knee, and elbows).

Hyperlipidemia
An elevated lipid concentration in the blood.

Arthralgia
Joint pain.

Heart murmur
An extra or unusual sound heard during a heartbeat caused vibrations resulting from the flow of blood through the heart.

Hepatic steatosis
Steatosis is a term used to denote lipid accumulation within hepatocytes.

Sudden cardiac death
The heart suddenly and unexpectedly stops beating resulting in death within a short time period (generally within 1 h of symptom onset).

Precocious atherosclerosis

Cerebral artery atherosclerosis
Atherosclerosis (HP:0002621) of a cerebral artery.

Dyspnea

Difficult or labored breathing. Dyspnea is a subjective feeling only the patient can rate, e.g., on a Borg scale.

Mitral regurgitation
An abnormality of the mitral valve characterized by insufficiency or incompetence of the mitral valve resulting in retrograde leaking of blood through the mitral valve upon ventricular contraction.

Myocardial infarction
Necrosis of the myocardium caused by an obstruction of the blood supply to the heart and often associated with chest pain, shortness of breath, palpitations, and anxiety as well as characteristic EKG findings and elevation of serum markers including creatine kinase-MB fraction and troponin.

Abnormal tendon morphology
An abnormality of the structure or form of the tendons, also often called sinews.

Autosomal recessive inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in individuals with two pathogenic alleles, either homozygotes (two copies of the same mutant allele) or compound heterozygotes (whereby each copy of a gene has a distinct mutant allele).

Calcification of the aorta
Calcification, that is, pathological deposition of calcium salts in the aorta.

Corneal arcus
A hazy, grayish-white ring about 2 mm in width located close to but separated from the limbus (the corneoscleral junction). Corneal arcus generally occurs bilaterally, and is related to lipid deposition in the cornea. Corneal arcus can occur in elderly persons as a part of the aging process but may be associated with hypercholesterolemia in people under the age of 50 years.

Coronary artery atherosclerosis
Reduction of the diameter of the coronary arteries as the result of an accumulation of atheromatous plaques within the walls of the coronary arteries, which increases the risk of myocardial ischemia.

Abnormal internal carotid artery morphology
An abnormality of an internal carotid artery.

Abnormal left ventricular function
Inability of the left ventricle to perform its normal physiologic function. Failure is either due to an inability to contract the left ventricle or the inability to relax completely and fill with blood during diastole.

Xanthelasma
The presence of xanthomata in the skin of the eyelid.

Hypercholesterolemia
An increased concentration of cholesterol in the blood.

Supravalvular aortic stenosis
A pathological narrowing in the region above the aortic valve associated with restricted left ventricular outflow.

Autosomal dominant inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in heterozygotes. In the context of medical genetics, an autosomal dominant disorder is caused when a single copy of the mutant allele is present. Males and females are affected equally, and can both transmit the disorder with a risk of 50% for each child of inheriting the mutant allele.

Abnormal eye physiology
A functional anomaly of the eye.

Optic neuropathy

Angina pectoris
Paroxysmal chest pain that occurs with exertion or stress and is related to myocardial ischemia.

Myocardial steatosis
Steatosis in the myocardium.

Coronary artery aneurysm
Enlargement of the diameter (cross-section) of a coronary artery as defined by a focal dilation of a segment at least 1.5 times larger than the reference vessel.

Renal artery stenosis
The presence of stenosis of the renal artery.

Increased LDL cholesterol concentration
An elevated concentration of low-density lipoprotein cholesterol in the blood.

Aortic atherosclerotic lesion
The presence of atheromas or atherosclerotic plaques in the aorta.

Hypertension
The presence of chronic increased pressure in the systemic arterial system.

Premature coronary artery atherosclerosis
Reduction of the diameter of the coronary arteries as the result of an accumulation of atheromatous plaques within the walls of the coronary arteries before age of 45.

Peripheral arterial stenosis
Narrowing of peripheral arteries with reduction of blood flow to the limbs. This feature may be quantified as an ankle-brachial index of less than 0.9, and may be manifested clinically as claudication.

Premature arteriosclerosis
Arteriosclerosis occurring at an age that is younger than usual.