Geneticheck - Genetic Report

Gene

MYBPC3

Associated Diseases

Familial Isolated Dilated Cardiomyopathy
Cardiomyopathy, Familial Hypertrophic, 4
Left Ventricular Noncompaction 10

Phenotype

Abnormality of neutrophils
A neutrophil abnormality.

Ventricular fibrillation
Uncontrolled contractions of muscles fibers in the left ventricle not producing contraction of the left ventricle. Ventricular fibrillation usually begins with a ventricular premature contraction and a short run of rapid ventricular tachycardia degenerating into uncoordinating ventricular fibrillations.

Cardiac arrest
An abrupt loss of heart function.

Lipoatrophy
Localized loss of fat tissue.

Dilated cardiomyopathy
Dilated cardiomyopathy (DCM) is defined by the presence of left ventricular dilatation and left ventricular systolic dysfunction in the absence of abnormal loading conditions (hypertension, valve disease) or coronary artery disease sufficient to cause global systolic impairment. Right ventricular dilation and dysfunction may be present but are not necessary for the diagnosis.

EMG abnormality
Abnormal results of investigations using electromyography (EMG).

Myopathy
A disorder of muscle unrelated to impairment of innervation or neuromuscular junction.

Cardiomegaly
Increased size of the heart, clinically defined as an increased transverse diameter of the cardiac silhouette that is greater than or equal to 50% of the transverse diameter of the chest (increased cardiothoracic ratio) on a posterior-anterior projection of a chest radiograph or a computed tomography.

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

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).

Pulmonary edema
Fluid accumulation in the lungs.

Pericardial effusion
Accumulation of fluid within the pericardium.

Hepatomegaly
Abnormally increased size of the liver.

Stroke
Sudden impairment of blood flow to a part of the brain due to occlusion or rupture of an artery to the brain.

Ventricular septal hypertrophy
The dividing wall between left and right sides of the heart, thickens and bulges into the left ventricle.

Chest pain
An unpleasant sensation characterized by physical discomfort (such as pricking, throbbing, or aching) localized to the chest.

Atrioventricular block
Delayed or lack of conduction of atrial depolarizations through the atrioventricular node to the ventricles.

Palmoplantar keratoderma
Abnormal thickening of the skin of the palms of the hands and the soles of the feet.

Ventricular hypertrophy
Enlargement of the cardiac ventricular muscle tissue with increase in the width of the wall of the ventricle and loss of elasticity. Ventricular hypertrophy is clinically differentiated into left and right ventricular hypertrophy.

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.

Transient ischemic attack

Elevated circulating creatine kinase concentration
An elevation of the level of the enzyme creatine kinase (also known as creatine phosphokinase, CPK; EC 2.7.3.2) in the blood. CPK levels can be elevated in a number of clinical disorders such as myocardial infarction, rhabdomyolysis, and muscular dystrophy.

Left bundle branch block

A conduction block of the left branch of the bundle of His. This manifests as a generalized disturbance of QRS morphology on EKG.

Left ventricular noncompaction
Left ventricular noncompaction (LVNC) is defined by 3 markers: prominent left ventricular (LV) trabeculae, deep intertrabecular recesses, and the thin compacted layer.

Right bundle branch block
A conduction block of the right branch of the bundle of His. This manifests as a prolongation of the QRS complex (greater than 0.12 s) with delayed activation of the right ventricle and terminal delay on the EKG.

Ascites
Accumulation of fluid in the peritoneal cavity.

Hypertrophic cardiomyopathy
Hypertrophic cardiomyopathy (HCM) is defined by the presence of increased ventricular wall thickness or mass in the absence of loading conditions (hypertension, valve disease) sufficient to cause the observed abnormality.

Sensorineural hearing impairment
A type of hearing impairment in one or both ears related to an abnormal functionality of the cochlear nerve.

Syncope
Syncope refers to a generalized weakness of muscles with loss of postural tone, inability to stand upright, and loss of consciousness. Once the patient is in a horizontal position, blood flow to the brain is no longer hindered by gravitation and consciousness is regained. Unconsciousness usually lasts for seconds to minutes. Headache and drowsiness (which usually follow seizures) do not follow a syncopal attack. Syncope results from a sudden impairment of brain metabolism usually due to a reduction in cerebral blood flow.