Geneticheck - Genetic Report

Gene

ACTC

Associated Diseases

Atrial Septal Defect, Ostium Secundum Type
Cardiomyopathy, Dilated, 1r
Atrial Septal Defect 5
Familial Isolated Dilated Cardiomyopathy
Cardiomyopathy, Familial Hypertrophic, 11

Phenotype

Pulmonary arterial hypertension
Pulmonary hypertension is defined mean pulmonary artery pressure of 25mmHg or more and pulmonary capillary wedge pressure of 15mmHg or less when measured by right heart catheterisation at rest and in a supine position.

Exertional dyspnea
Perceived difficulty to breathe that occurs with exercise or exertion and improves with rest.

Atrial flutter
A type of atrial arrhythmia characterized by atrial rates of between 240 and 400 beats per minute and some degree of atrioventricular node conduction block. Typically, the ventricular rate is half the atrial rate. In the EKG; atrial flutter waves are observed as sawtooth-like atrial activity. Pathophysiologically, atrial flutter is a form of atrial reentry in which there is a premature electrical impulse creates a self-propagating circuit.

Supraventricular tachycardia
Supraventricular tachycardia (SVT) is an abnormally increased heart rate (over 100 beats per minute at rest) with origin above the level of the ventricles.

Restrictive cardiomyopathy
Restrictive left ventricular physiology is characterized by a pattern of ventricular filling in which increased stiffness of the myocardium causes ventricular pressure to rise precipitously with only small increases in volume, defined as restrictive ventricular physiology in the presence of normal or reduced diastolic volumes (of one or both ventricles), normal or reduced systolic volumes, and normal ventricular wall thickness.

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

First degree atrioventricular block
Delay of conduction through the atrioventricular node, which is manifested as prolongation of the PR interval in the electrocardiogram (EKG). All atrial impulses reach the ventricles.

Pedal edema
An abnormal accumulation of excess fluid in the lower extremity resulting in swelling of the feet and extending upward to the lower leg.

Bundle branch block
Block of conduction of electrical impulses along the Bundle of His or along one of its bundle branches.

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

Ventricular arrhythmia

Recurrent bacterial infections
Increased susceptibility to bacterial infections, as manifested by recurrent episodes of bacterial infection.

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.

Cyanosis
Bluish discoloration of the skin and mucosa due to poor circulation or inadequate oxygenation of arterial or capillary blood.

Right ventricular failure
Reduced ability of the right ventricle to perform its function (to receive blood from the right atrium and to eject blood into the pulmonary artery), often leading to pitting peripheral edema, ascites, and hepatomegaly.

Right ventricular dilatation
Enlargement of the chamber of the right ventricle.

Left-to-right shunt
Pattern of blood flow in the heart that deviates from the normal circuit of the circulatory system from the left side of the heart to the right.

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 mitral valve morphology
Any structural anomaly of the mitral valve.

Transient ischemic attack

Breathing dysregulation

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 atrial enlargement
Increase in size of the right atrium.

Left ventricular hypertrophy
Enlargement or increased size of the heart left ventricle.

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.

Tricuspid regurgitation
Failure of the tricuspid valve to close sufficiently upon contraction of the right ventricle, causing blood to regurgitate (flow backward) into the right atrium.

Abnormality of neutrophils
A neutrophil abnormality.

ST segment depression
An electrocardiographic anomaly in which the ST segment is observed to be located inferior to the isoelectric line.

Exercise intolerance
A functional motor deficit where individuals whose responses to the challenges of exercise fail to achieve levels considered normal for their age and gender.

Palpitations
A sensation that the heart is pounding or racing, which is a non-specific sign but may be a manifestation of arrhythmia.

Pneumonia
Inflammation of any part of the lung parenchyma.

Lipoatrophy
Localized loss of fat tissue.

Supraventricular arrhythmia

A type of arrhythmia that originates above the ventricles, whereby the electrical impulse propagates down the normal His Purkinje system similar to normal sinus rhythm.

Fatigue
A subjective feeling of tiredness characterized by a lack of energy and motivation.

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.

Increased pulmonary vascular resistance
Pulmonary vascular resistance (PVR) more than 3 wood units, as defined by the current definition of pulmonary hypertension. 95% of individuals have a PVR of less than 2.4 wood units.

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

Arrhythmia
Any cardiac rhythm other than the normal sinus rhythm. Such a rhythm may be either of sinus or ectopic origin and either regular or irregular. An arrhythmia may be due to a disturbance in impulse formation or conduction or both.

Atrial septal defect
Atrial septal defect (ASD) is a congenital abnormality of the interatrial septum that enables blood flow between the left and right atria via the interatrial septum.

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.

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

Congestive heart failure
The presence of an abnormality of cardiac function that is responsible for the failure of the heart to pump blood at a rate that is commensurate with the needs of the tissues or a state in which abnormally elevated filling pressures are required for the heart to do so. Heart failure is frequently related to a defect in myocardial contraction.

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

Airway obstruction
Obstruction of conducting airways of the lung.

Systolic heart murmur
A heart murmur limited to systole, i.e., between the first and second heart sounds S1 and S2.

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.

Atrial fibrillation
An atrial arrhythmia characterized by disorganized atrial activity without discrete P waves on the surface EKG, but instead by an undulating baseline or more sharply circumscribed atrial deflections of varying amplitude an frequency ranging from 350 to 600 per minute.

Orthopnea
A sensation of breathlessness in the recumbent position, relieved by sitting or standing.