Geneticheck - Genetic Report

Gene

KCNQ1

Associated Diseases

Atrial Fibrillation, Familial, 3
Long Qt Syndrome 1
Jervell And Lange-nielsen Syndrome
Romano-ward Syndrome
Familial Short Qt Syndrome
Beckwith-wiedemann Syndrome
Jervell And Lange-nielsen Syndrome 1
Short Qt Syndrome 2

Phenotype

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.

Torsade de pointes
A type of ventricular tachycardia characterized by polymorphioc QRS complexes that change in amplitue and cycle length, and thus have the appearance of oscillating around the baseline in the EKG.

Seizure
A seizure is an intermittent abnormality of nervous system physiology characterised by a transient occurrence of signs and/or symptoms due to abnormal excessive or synchronous neuronal activity in the brain.

Omphalocele
A midline anterior incomplete closure of the abdominal wall in which there is herniation of the abdominal viscera into the base of the abdominal cord.

Hepatoblastoma
A kind of neoplasm of the liver that originates from immature liver precursor cells and macroscopically is composed of tissue resembling fetal or mature liver cells or bile ducts.

Prominent metopic ridge
Vertical bony ridge positioned in the midline of the forehead.

Cryptorchidism
Testis in inguinal canal. That is, absence of one or both testes from the scrotum owing to failure of the testis or testes to descend through the inguinal canal to the scrotum.

Midface retrusion
Posterior positions and/or vertical shortening of the infraorbital and perialar regions, or increased concavity of the face and/or reduced nasolabial angle.

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).

Shortened QT interval
Decreased time between the start of the Q wave and the end of the T wave as measured by the electrocardiogram (EKG).

Overgrowth
Excessive postnatal growth which may comprise increased weight, increased length, and/or increased head circumference.

Vesicoureteral reflux
Abnormal (retrograde) movement of urine from the bladder into ureters or kidneys related to inadequacy of the valvular mechanism at the ureterovesicular junction or other causes.

Adrenocortical cytomegaly
The presence of large polyhedral cells with eosinophilic granular cytoplasm and enlarged nuclei in the adrenal cortex.

Ventricular arrhythmia

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).

Adrenocortical carcinoma
A malignant neoplasm of the adrenal cortex that may produce hormones such as cortisol, aldosterone, estrogen, or testosterone.

Bilateral sensorineural hearing impairment
A bilateral form of sensorineural hearing impairment.

Overgrowth of external genitalia

Tachycardia
A rapid heartrate that exceeds the range of the normal resting heartrate for age.

Thromboembolic stroke
A cerebrovascular accident (stroke) that occurs because of thromboembolism.

Nevus flammeus
A congenital vascular malformation consisting of superficial and deep dilated capillaries in the skin which produce a reddish to purplish discolouration of the skin.

Posterior helix pit
Permanent indentation on the posteromedial aspect of the helix that may be sharply or indistinctly delineated.

Nephroblastoma
The presence of a nephroblastoma, which is a neoplasm of the kidney that primarily affects children.

Dandy-Walker malformation
A congenital brain malformation typically characterized by incomplete formation of the cerebellar vermis, dilation of the fourth ventricle, and enlargement of the posterior fossa. In layman's terms, Dandy Walker malformation is a cyst in the cerebellum (typically symmetrical) that is involved with the fourth ventricle. This may interfere with the ability to drain cerebrospinal fluid from the brain, resulting in hydrocephalus. Dandy Walker cysts are formed during early embryonic development, while the brain forms. The cyst in the cerebellum typically has several blood vessels running through it connecting to the brain, thereby prohibiting surgical removal.

Abnormal autonomic nervous system physiology
A functional abnormality of the autonomic nervous system.

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

Hypokalemia
An abnormally decreased potassium concentration in the blood.

Loss of consciousness

Diastasis recti
A separation of the rectus abdominis muscle into right and left halves (which are normally joined at the midline at the linea alba).

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.

Macroglossia
Increased length and width of the tongue.

Abnormal cardiac exercise stress test
Abnormal results of exercise on heart function.

Gonadoblastoma
The presence of a gonadoblastoma, a neoplasm of a gonad that consists of aggregates of germ cells and sex cord elements.

Abnormal T-wave

An abnormality of the T wave on the electrocardiogram, which mainly represents the repolarization of the ventricles.

Iron deficiency anemia

Congenital sensorineural hearing impairment
A type of hearing impairment caused by an abnormal functionality of the cochlear nerve with congenital onset.

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.

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

Nephrolithiasis
The presence of calculi (stones) in the kidneys.

Accelerated skeletal maturation
An abnormally increased rate of skeletal maturation. Accelerated skeletal maturation can be diagnosed on the basis of an estimation of the bone age from radiographs of specific bones in the human body.

Prominent occiput
Increased convexity of the occiput (posterior part of the skull).

Prolonged QTc interval
A longer than normal interval (corrected for heart rate) between the Q and T waves in the heart's cycle. Prolonged QTc can cause premature action potentials during late phase depolarizations thereby leading to ventricular arrhythmias and ventricular fibrillations.

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.

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.

Bradycardia
A slower than normal heart rate (in adults, slower than 60 beats per minute).

Nephrocalcinosis

Nephrocalcinosis is the deposition of calcium salts in renal parenchyma.

Pancreatic hyperplasia
Hyperplasia of the pancreas.

Hepatomegaly
Abnormally increased size of the liver.

Profound sensorineural hearing impairment
Complete loss of hearing related to a sensorineural defect.

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

Coarse facial features
Absence of fine and sharp appearance of brows, nose, lips, mouth, and chin, usually because of rounded and heavy features or thickened skin with or without thickening of subcutaneous and bony tissues.

Cardiomyopathy
A myocardial disorder in which the heart muscle is structurally and functionally abnormal, in the absence of coronary artery disease, hypertension, valvular disease and congenital heart disease sufficient to cause the observed myocardial abnormality.

Sinus bradycardia
Bradycardia related to a mean resting sinus rate of less than 50 beats per minute.

Neonatal hypoglycemia

Large fontanelles
In newborns, the two frontal bones, two parietal bones, and one occipital bone are joined by fibrous sutures, which form a small posterior fontanelle, and a larger, diamond-shaped anterior fontanelle. These regions allow for the skull to pass the birth canal and for later growth. The fontanelles gradually ossify, whereby the posterior fontanelle usually closes by eight weeks and the anterior fontanelle by the 9th to 16th month of age. Large fontanelles are diagnosed if the fontanelles are larger than age-dependent norms.

Postexertional malaise
A subjective feeling of tiredness characterized by a lack of energy and motivation and that is induced by exertion or exercise.

Heterogeneous

Renal cortical cysts
Cysts of the cortex of the kidney.

Abnormality of prenatal development or birth
An abnormality of the fetus or the birth of the fetus, excluding structural abnormalities.

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.

Enlarged kidney
An abnormal increase in the size of the kidney.

Proptosis
An eye that is protruding anterior to the plane of the face to a greater extent than is typical.

Hemihypertrophy
Overgrowth of only one side of the body.

Prolonged QT interval
Increased time between the start of the Q wave and the end of the T wave as measured by the electrocardiogram (EKG).