Geneticheck - Genetic Report

Gene

CALR

Associated Diseases

Essential Thrombocythemia
Thrombocythemia 1
Primary Myelofibrosis
Myelofibrosismyelofibrosis With Myeloid Metaplasia, Included

Phenotype

Abnormal cerebral vascular morphology
An anomaly of the cerebral blood vessels.

Impaired epinephrine-induced platelet aggregation
Abnormal response to epinephrine as manifested by reduced or lacking aggregation of platelets upon addition of epinephrine.

Abnormal platelet morphology
An anomaly in platelet form, ultrastructure, or intracellular organelles.

Flank pain
An unpleasant sensation characterized by physical discomfort (such as pricking, throbbing, or aching) and perceived to originate in the flank.

Amaurosis fugax
A transient visual disturbance that is typically caused by a circulatory, ocular or neurological underlying condition.

Easy fatigability
Increased susceptibility to fatigue.

Prolonged bleeding time
Prolongation of the time taken for a standardized skin cut of fixed depth and length to stop bleeding.

Constitutional symptom
A symptom or manifestation indicating a systemic or general effect of a disease and that may affect the general well-being or status of an individual.

Increased circulating lactate dehydrogenase concentration
An elevated level of the enzyme lactate dehydrogenase in the blood circulation.

Ecchymosis
A purpuric lesion that is larger than 1 cm in diameter.

Myelodysplasia
Clonal hematopoietic stem cell disorders characterized by dysplasia (ineffective production) in one or more

hematopoietic cell lineages, leading to anemia and cytopenia.

Hepatosplenomegaly
Simultaneous enlargement of the liver and spleen.

Poikilocytosis
The presence of abnormally shaped erythrocytes.

Hemangioma
A hemangioma is a benign tumor characterized by blood-filled spaces lined by benign endothelial cells. A hemangioma characterized by large endothelial spaces (caverns) is called a cavernous hemangioma (in contrast to a hemangioma with small endothelial spaces, which is called capillary hemangioma).

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

Arterial thrombosis
The formation of a blood clot inside an artery.

Bone marrow hypercellularity
A larger than normal amount or percentage of hematopoietic cells relative to marrow fat.

Acute leukemia
A clonal (malignant) hematopoietic disorder with an acute onset, affecting the bone marrow and the peripheral blood. The malignant cells show minimal differentiation and are called blasts, either myeloid blasts (myeloblasts) or lymphoid blasts (lymphoblasts).

Leukocytosis
An abnormal increase in the number of leukocytes in the blood.

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.

Splenomegaly
Abnormal increased size of the spleen.

Extramedullary hematopoiesis
The process of hematopoiesis occurring outside of the bone marrow (in the liver, thymus, and spleen) in the postnatal organisms.

Transient ischemic attack

Pancytopenia

An abnormal reduction in numbers of all blood cell types (red blood cells, white blood cells, and platelets).

Lymphadenopathy
Enlargment (swelling) of a lymph node.

Petechiae
Petechiae are pinpoint-sized reddish/purple spots, resembling a rash, that appear just under the skin or a mucous membrane when capillaries have ruptured and some superficial bleeding into the skin has happened. This term refers to an abnormally increased susceptibility to developing petechiae.

Low-grade fever
Mild fever that does not exceed 38.5 degrees centigrade.

Portal hypertension
Increased pressure in the portal vein.

Impaired collagen-induced platelet aggregation
Abnormal response to collagen or collagen-mimetics as manifested by reduced or lacking aggregation of platelets upon addition collagen or collagen-mimetics.

Pallor
Abnormally pale skin.

Anorexia
A lack or loss of appetite for food (as a medical condition).

Abnormal thrombocyte morphology
An abnormality of platelets.

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

Increased megakaryocyte count
Increased megakaryocyte number, i.e., of platelet precursor cells, present in the bone marrow.

Abnormal thrombosis
Venous or arterial thrombosis (formation of blood clots) of spontaneous nature and which cannot be fully explained by acquired risk (e.g. atherosclerosis).

Abnormal megakaryocyte morphology
Any structural anomaly of megakaryocytes. Mature blood platelets are released from the cytoplasm of megakaryocytes, which are bone-marrow resident cells.

Myelofibrosis
Replacement of bone marrow by fibrous tissue.

Anemia
A reduction in erythrocytes volume or hemoglobin concentration.

Paresthesia
Abnormal sensations such as tingling, pricking, or numbness of the skin with no apparent physical cause.

Abnormality of bone marrow cell morphology
An anomaly of the form or number of cells in the bone marrow.

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.

Thrombocytopenia
A reduction in the number of circulating thrombocytes.

Myeloproliferative disorder
Proliferation (excess production) of hemopoietically active tissue or of tissue which has embryonic hemopoietic potential.

Fever
Body temperature elevated above the normal range.

Hepatomegaly
Abnormally increased size of the liver.

Somatic mutation
A mode of inheritance in which a trait or disorder results from a de novo mutation occurring after conception, rather than being inherited from a preceding generation.

Hematological neoplasm
Neoplasms located in the blood and blood-forming tissue (the bone marrow and lymphatic tissue).

Impaired ADP-induced platelet aggregation
Abnormal platelet response to ADP as manifested by reduced or lacking aggregation of platelets upon addition of ADP.

Venous thrombosis
Formation of a blood clot (thrombus) inside a vein, causing the obstruction of blood flow.

Thrombocytosis
Increased numbers of platelets in the peripheral blood.

Abnormality of blood and blood-forming tissues
An abnormality of the hematopoietic system.

Purpura
Purpura (from Latin: purpura, meaning "purple") is the appearance of red or purple discolorations on the skin that do not blanch on applying pressure. They are caused by bleeding underneath the skin. This term refers to an abnormally increased susceptibility to developing purpura. Purpura are larger than petechiae.

Cachexia
Severe weight loss, wasting of muscle, loss of appetite, and general debility related to a chronic disease.

Abnormal bleeding
An abnormal susceptibility to bleeding, often referred to as a bleeding diathesis. A bleeding diathesis may be related to vascular, platelet and coagulation defects.