Geneticheck - Genetic Report

Gene

SH2B3

Associated Diseases

Essential Thrombocythemia
Thrombocythemia 1
Myelofibrosismyelofibrosis With Myeloid Metaplasia, Included
Erythrocytosis, Familial, 1

Phenotype

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

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.

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.

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

Abnormal thrombocyte morphology
An abnormality of platelets.

Vertigo
An abnormal sensation of spinning while the body is actually stationary.

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.

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

Cerebral hemorrhage
Hemorrhage into the parenchyma of the brain.

Myelofibrosis
Replacement of bone marrow by fibrous tissue.

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

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.

Increased hematocrit
An elevation above the normal ratio of the volume of red blood cells to the total volume of blood.

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.

Myelodysplasia
Clonal hematopoietic stem cell disorders characterized by dysplasia (ineffective production) in one or more hematopoietic cell lineages, leading to anemia and cytopenia.

Increased hemoglobin

Increased red blood cell mass
The presence of an increased mass of red blood cells in the circulation.

Peripheral thrombosis

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.

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).

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.

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.

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.

Headache
Cephalgia, or pain sensed in various parts of the head, not confined to the area of distribution of any nerve.

Splenomegaly
Abnormal increased size of the spleen.

Plethora

Transient ischemic attack

Thrombocytosis
Increased numbers of platelets in the peripheral blood.

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.

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