Geneticheck - Genetic Report

Gene

ATM

Associated Diseases

Breast Cancer
Ataxia-telangiectasia
Mantle Cell Lymphoma
Ataxia-telangiectasia

Phenotype

Non-Hodgkin lymphoma
A type of lymphoma characterized microscopically by the absence of multinucleated Reed-Sternberg cells.

Sinusitis
Inflammation of the paranasal sinuses owing to a viral, bacterial, or fungal infection, allergy, or an autoimmune reaction.

Hypoplasia of the thymus
Underdevelopment of the thymus.

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.

Recurrent respiratory infections
An increased susceptibility to respiratory infections as manifested by a history of recurrent respiratory infections.

Abnormality of the gastrointestinal tract
An abnormality of the gastrointestinal tract.

Elevated hepatic transaminase
Elevations of the levels of SGOT and SGPT in the serum. SGOT (serum glutamic oxaloacetic transaminase) and SGPT (serum glutamic pyruvic transaminase) are transaminases primarily found in the liver and heart and are released into the bloodstream as the result of liver or heart damage. SGOT and SGPT are used clinically mainly as markers of liver damage.

Recurrent bronchitis
An increased susceptibility to bronchitis as manifested by a history of recurrent bronchitis.

Defective B cell differentiation
Reduced functionality of the process in which a precursor cell type acquires the specialized features of a B cell. A B cell is a lymphocyte of B lineage with the phenotype CD19-positive and capable of B cell mediated immunity.

Gait disturbance

The term gait disturbance can refer to any disruption of the ability to walk. In general, this can refer to neurological diseases but also fractures or other sources of pain that is triggered upon walking. However, in the current context gait disturbance refers to difficulty walking on the basis of a neurological or muscular disease.

Neurological speech impairment

Cellular immunodeficiency
An immunodeficiency characterized by defective cell-mediated immunity or humoral immunity.

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).

Choreoathetosis
Involuntary movements characterized by both athetosis (inability to sustain muscles in a fixed position) and chorea (widespread jerky arrhythmic movements).

Weight loss
Reduction of total body weight.

Telangiectasia of the skin
Presence of small, permanently dilated blood vessels near the surface of the skin, visible as small focal red lesions.

Failure to thrive
Failure to thrive (FTT) refers to a child whose physical growth is substantially below the norm.

Breast carcinoma
The presence of a carcinoma of the breast.

Immunodeficiency
Failure of the immune system to protect the body adequately from infection, due to the absence or insufficiency of some component process or substance.

Abnormality of the immune system
An abnormality of the immune system.

Delayed puberty
Passing the age when puberty normally occurs with no physical or hormonal signs of the onset of puberty.

Abnormality of chromosome stability
A type of chromosomal aberration characterised by reduced resistance of chromosomes to change or deterioration.

Short stature

A height below that which is expected according to age and gender norms. Although there is no universally accepted definition of short stature, many refer to "short stature" as height more than 2 standard deviations below the mean for age and gender (or below the 3rd percentile for age and gender dependent norms).

Ataxia
Cerebellar ataxia refers to ataxia due to dysfunction of the cerebellum. This causes a variety of elementary neurological deficits including asynergy (lack of coordination between muscles, limbs and joints), dysmetria (lack of ability to judge distances that can lead to under- or overshoot in grasping movements), and dysdiadochokinesia (inability to perform rapid movements requiring antagonizing muscle groups to be switched on and off repeatedly).

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.

Nystagmus
Rhythmic, involuntary oscillations of one or both eyes related to abnormality in fixation, conjugate gaze, or vestibular mechanisms.

Splenomegaly
Abnormal increased size of the spleen.

Leukemia
A cancer of the blood and bone marrow characterized by an abnormal proliferation of leukocytes.

T lymphocytopenia
An abnormally low count of T cells.

Lymphadenopathy
Enlargment (swelling) of a lymph node.

Hypopigmentation of hair

Mucosal telangiectasiae
Telangiectasia of the mucosa, the mucous membranes which are involved in absorption and secretion that line cavities that are exposed to the external environment and internal organs.

Abnormal hair morphology
An abnormality of the hair.

Cognitive impairment
Abnormal cognition with deficits in thinking, reasoning, or remembering.

Glucose intolerance
Glucose intolerance (GI) can be defined as dysglycemia that comprises both prediabetes and diabetes. It

includes the conditions of impaired fasting glucose (IFG) and impaired glucose tolerance (IGT) and diabetes mellitus (DM).

Neoplasm
An organ or organ-system abnormality that consists of uncontrolled autonomous cell-proliferation which can occur in any part of the body as a benign or malignant neoplasm (tumour).

Decreased circulating antibody level
An abnormally decreased level of immunoglobulin in blood.

Female hypogonadism
Decreased functionality of the female gonads, i.e., of the ovary.

Polycystic ovaries

Dysarthria
Dysarthric speech is a general description referring to a neurological speech disorder characterized by poor articulation. Depending on the involved neurological structures, dysarthria may be further classified as spastic, flaccid, ataxic, hyperkinetic and hypokinetic, or mixed.

Type II diabetes mellitus
A type of diabetes mellitus initially characterized by insulin resistance and hyperinsulinemia and subsequently by glucose interolerance and hyperglycemia.

Prematurely aged appearance

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

Elevated alpha-fetoprotein
An increased concentration of alpha-fetoprotein.

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

Conjunctival telangiectasia
The presence of small (ca. 0.5-1.0 mm) dilated blood vessels near the surface of the mucous membranes of the conjunctiva.

Decreased circulating IgA level
Decreased levels of immunoglobulin A (IgA).

Reduced tendon reflexes
Diminution of tendon reflexes, which is an invariable sign of peripheral nerve disease.

Decreased proportion of CD4-positive helper T cells
A decreased proportion of circulating CD4-positive helper T cells relative to total T cell count.

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

Bronchiectasis
Persistent abnormal dilatation of the bronchi owing to localized and irreversible destruction and widening of the large airways.

Decreased circulating IgG2 level
A reduction in immunoglobulin levels of the IgG2 subclass in the blood circulation.

Tremor
An unintentional, oscillating to-and-fro muscle movement about a joint axis.

Fever
Body temperature elevated above the normal range.

Multiple cafe-au-lait spots
The presence of six or more cafe-au-lait spots.

B-cell lymphoma
A type of lymphoma that originates in B-cells.

Spasticity
A motor disorder characterized by a velocity-dependent increase in tonic stretch reflexes with increased muscle tone, exaggerated (hyperexcitable) tendon reflexes.

Premature graying of hair
Development of gray hair at a younger than normal age.

Aplasia/Hypoplasia of the skin

Myoclonus
Very brief, involuntary random muscular contractions occurring at rest, in response to sensory stimuli, or accompanying voluntary movements.

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.

Aplasia/Hypoplasia of the thymus
Absence or underdevelopment of the thymus.

Abnormality of eye movement
An abnormality in voluntary or involuntary eye movements or their control.

Lymphoma
A cancer originating in lymphocytes and presenting as a solid tumor of lymhpoid cells.

Strabismus
A misalignment of the eyes so that the visual axes deviate from bifoveal fixation. The classification of strabismus may be based on a number of features including the relative position of the eyes, whether the deviation is latent or manifest, intermittent or constant, concomitant or otherwise and according to the age of onset and the relevance of any associated refractive error.

Abnormality of movement
An abnormality of movement with a neurological basis characterized by changes in coordination and speed of voluntary movements.

Heterogeneous

Abnormal testis morphology
An anomaly of the testicle (the male gonad).

Lymphopenia
A reduced number of lymphocytes in the blood.

Hodgkin lymphoma
A type of lymphoma characterized microscopically by multinucleated Reed-Sternberg cells.

Cafe-au-lait spot
Cafe-au-lait spots are hyperpigmented lesions that can vary in color from light brown to dark brown with smooth borders and having a size of 1.5 cm or more in adults and 0.5 cm or more in children.

Abnormal spermatogenesis
Incomplete maturation or aberrant formation of the male gametes.

Diabetes mellitus
A group of abnormalities characterized by hyperglycemia and glucose intolerance.

Skeletal muscle atrophy
The presence of skeletal muscular atrophy (which is also known as amyotrophy).

Dystonia
An abnormally increased muscular tone that causes fixed abnormal postures. There is a slow, intermittent twisting motion that leads to exaggerated turning and posture of the extremities and trunk.

Decreased circulating IgG level
An abnormally decreased level of immunoglobulin G (IgG) in blood.