Geneticheck - Genetic Report

Gene

BRCA2

Associated Diseases

Familial Pancreatic Carcinoma
Breast Cancer
Breast-ovarian Cancer, Familial, Susceptibility To, 2
Hereditary Breast And Ovarian Cancer Syndrome
Prostate Cancer
Fanconi Anemia, Complementation Group D1
Glioma Susceptibility 3
Nephroblastoma
Fanconi Anemia
Medulloblastoma
Pancreatic Cancer, Susceptibility To, 2
Wilms Tumor 1

Phenotype

Irregular hyperpigmentation

Jaundice
Yellow pigmentation of the skin due to bilirubin, which in turn is the result of increased bilirubin concentration in the bloodstream.

Abnormality of the urinary system
An abnormality of the urinary system.

Dolichocephaly
An abnormality of skull shape characterized by a increased anterior-posterior diameter, i.e., an increased antero-posterior dimension of the skull. Cephalic index less than 76%. Alternatively, an apparently increased antero-posterior length of the head compared to width. Often due to premature closure of the sagittal suture.

Renal insufficiency
A reduction in the level of performance of the kidneys in areas of function comprising the concentration of urine, removal of wastes, the maintenance of electrolyte balance, homeostasis of blood pressure, and calcium metabolism.

Abnormality of the eye
Any abnormality of the eye, including location, spacing, and intraocular abnormalities.

Abnormality of vision
Abnormality of eyesight (visual perception).

Poor appetite
A reduced desire to eat.

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.

Abnormal aortic valve morphology
Any abnormality of the aortic valve.

Astigmatism
A type of astigmatism associated with abnormal curvatures on the anterior and/or posterior surface of the cornea.

Aplasia/Hypoplasia of the radius
A small/hypoplastic or absent/aplastic radius.

Facial asymmetry
An abnormal difference between the left and right sides of the face.

Abnormality of the uterus
An abnormality of the uterus.

Acute myeloid leukemia
A form of leukemia characterized by overproduction of an early myeloid cell.

Neoplasm of the pancreas
A tumor (abnormal growth of tissue) of the pancreas.

Ptosis
The upper eyelid margin is positioned 3 mm or more lower than usual and covers the superior portion of the iris (objective); or, the upper lid margin obscures at least part of the pupil (subjective).

Microcephaly
Head circumference below 2 standard deviations below the mean for age and gender.

Weight loss
Reduction of total body weight.

Nausea and vomiting
Nausea is a commonly encountered symptom that has been defined as an unpleasant painless subjective feeling that one will imminently vomit. Vomiting has been defined as the forceful expulsion of the contents of the stomach, duodenum, or jejunum through the oral cavity. While nausea and vomiting are often thought to exist on a temporal continuum, this is not always the case. There are situations when severe nausea may be present without emesis and less frequently, when emesis may be present without preceding nausea.

Epicanthus
A fold of skin starting above the medial aspect of the upper eyelid and arching downward to cover, pass in front of and lateral to the medial canthus.

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

High palate
Height of the palate more than 2 SD above the mean (objective) or palatal height at the level of the first permanent molar more than twice the height of the teeth (subjective).

Breast carcinoma
The presence of a carcinoma of the breast.

Sloping forehead
Inclination of the anterior surface of the forehead from the vertical more than two standard deviations above the mean (objective); or apparently excessive posterior sloping of the forehead in a lateral view.

Intermittent diarrhea
Repeated episodes of diarrhea separated by periods without diarrhea.

Aplasia/Hypoplasia of fingers
Small/hypoplastic or absent/aplastic fingers.

Aniridia
Abnormality of the iris characterized by, typically bilateral, complete or partial iris hypoplasia. The phenotype ranges from mild defects of anterior iris stroma only to almost complete absence of the iris.

Hepatosplenomegaly
Simultaneous enlargement of the liver and spleen.

Hearing abnormality
An abnormality of the sensory perception of sound.

Hematuria
The presence of blood in the urine. Hematuria may be gross hematuria (visible to the naked eye) or microscopic hematuria (detected by dipstick or microscopic examination of the urine).

Aganglionic megacolon
An abnormality resulting from a lack of intestinal ganglion cells (i.e., an aganglionic section of bowel) that results in bowel obstruction with enlargement of the colon.

Melanoma
The presence of a melanoma, a malignant cancer originating from pigment producing melanocytes. Melanoma can originate from the skin or the pigmented layers of the eye (the uvea).

Abnormality of the upper limb
An abnormality of the arm.

Bicornuate uterus
The presence of a bicornuate uterus.

Abnormality of skin pigmentation
An abnormality of the pigmentation of the skin.

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.

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

Clubbing of toes
Terminal broadening of the toes (distal phalanges of the toes).

Abnormality of the ulna
An abnormality of the ulna bone of the forearm.

Colon cancer

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

Lymphadenopathy
Enlargment (swelling) of a lymph node.

Umbilical hernia
Protrusion of abdominal contents through a defect in the abdominal wall musculature around the umbilicus. Skin and subcutaneous tissue overlie the defect.

Toe syndactyly
Webbing or fusion of the toes, involving soft parts only or including bone structure. Bony fusions are referred to as "bony" Syndactyly if the fusion occurs in a radio-ulnar axis. Fusions of bones of the toes in a proximo-distal axis are referred to as "Symphalangism".

Hypopigmented skin patches

Hypogonadism
A decreased functionality of the gonad.

Abnormal eyelid morphology
An abnormality of the eyelids.

Short thumb
Hypoplasia (congenital reduction in size) of the thumb.

Pes planus
A foot where the longitudinal arch of the foot is in contact with the ground or floor when the individual is standing; or, in a patient lying supine, a foot where the arch is in contact with the surface of a flat board

pressed against the sole of the foot by the examiner with a pressure similar to that expected from weight bearing; or, the height of the arch is reduced.

Renal hypoplasia/aplasia
Absence or underdevelopment of the kidney.

Clinodactyly of the 5th finger
Clinodactyly refers to a bending or curvature of the fifth finger in the radial direction (i.e., towards the 4th finger).

Abnormality of femur morphology
Any anomaly of the structure of the femur.

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

Oligohydramnios
Diminished amniotic fluid volume in pregnancy.

Abnormal foot morphology
An abnormality of the skeleton of foot.

Reduced bone mineral density
A reduction of bone mineral density, that is, of the amount of matter per cubic centimeter of bones.

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.

Anemia
A reduction in erythrocytes volume or hemoglobin concentration.

Finger syndactyly
Webbing or fusion of the fingers, involving soft parts only or including bone structure. Bony fusions are referred to as "bony" Syndactyly if the fusion occurs in a radio-ulnar axis. Fusions of bones of the fingers in a proximo-distal axis are referred to as "Symphalangism".

Intrauterine growth retardation
An abnormal restriction of fetal growth with fetal weight below the tenth percentile for gestational age.

Fever
Body temperature elevated above the normal range.

Abnormal renal morphology
Any structural anomaly of the kidney.

Abnormal aortic morphology
An abnormality of the aorta.

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.

External ear malformation
A malformation of the auricle of the ear.

Abnormal carotid artery morphology
Any structural abnormality of the carotid arteries, including the common carotid artery and its' arterial branches.

Abnormal preputium morphology
An abnormality of the retractable fold of skin that covers the tip of the penis.

Upslanted palpebral fissure
The palpebral fissure inclination is more than two standard deviations above the mean for age (objective); or, the inclination of the palpebral fissure is greater than typical for age.

Cranial nerve paralysis

Neoplasm of the liver
A tumor (abnormal growth of tissue) of the liver.

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

Frontal bossing
Bilateral bulging of the lateral frontal bone prominences with relative sparing of the midline.

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

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

Prostate cancer
A cancer of the prostate.

Intellectual disability
Subnormal intellectual functioning which originates during the developmental period. Intellectual disability, previously referred to as mental retardation, has been defined as an IQ score below 70.

Neoplasm of the lung
Tumor of the lung.

Duodenal stenosis
The narrowing or partial blockage of a portion of the duodenum.

Choanal atresia
Absence or abnormal closure of the choana (the posterior nasal aperture).

Anal atresia
Congenital absence of the anus, i.e., the opening at the bottom end of the intestinal tract.

Cleft palate
Cleft palate is a developmental defect of the palate resulting from a failure of fusion of the palatine processes and manifesting as a separation of the roof of the mouth (soft and hard palate).

Chronic fatigue
Subjective feeling of tiredness characterized by a lack of energy and motivation that persists for six months or longer.

Absent testis
Testis not palpable in the scrotum or inguinal canal.

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.

Medulloblastoma
A rapidly growing embryonic tumor arising in the posterior part of the cerebellar vermis and neuroepithelial roof of the fourth ventricle in children. More rarely, medulloblastoma arises in the cerebellum in adults.

Ventriculomegaly
An increase in size of the ventricular system of the brain.

Micrognathia
Developmental hypoplasia of the mandible.

Hydroureter
The distention of the ureter with urine.

Hearing impairment
A decreased magnitude of the sensory perception of sound.

Intestinal pseudo-obstruction
A functional rather than mechanical obstruction of the intestines, associated with manifestations that resemble those caused by an intestinal obstruction, including distension, abdominal pain, nausea, vomiting, constipation or diarrhea, in an individual in whom a mechanical blockage has been excluded.

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).

Exocrine pancreatic insufficiency
Impaired function of the exocrine pancreas associated with a reduced ability to digest foods because of lack of digestive enzymes.

Patent ductus arteriosus
In utero, the ductus arteriosus (DA) serves to divert ventricular output away from the lungs and toward the placenta by connecting the main pulmonary artery to the descending aorta. A patent ductus arteriosus (PDA) in the first 3 days of life is a physiologic shunt in healthy term and preterm newborn infants, and normally is substantially closed within about 24 hours after bith and completely closed after about three weeks. Failure of physiologcal closure is referred to a persistent or patent ductus arteriosus (PDA). Depending on the degree of left-to-right shunting, PDA can have clinical consequences.

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

Recurrent urinary tract infections
Repeated infections of the urinary tract.

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).

Azoospermia
Absence of any measurable level of sperm in his semen.

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

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

Hip dislocation
Displacement of the femur from its normal location in the hip joint.

Abnormal cardiac septum morphology
An anomaly of the intra-atrial or intraventricular septum.

Tracheoesophageal fistula
An abnormal connection (fistula) between the esophagus and the trachea.

Arteriovenous malformation
An anomalous configuration of blood vessels that shunts arterial blood directly into veins without passing

through the capillaries.

Cataract
A cataract is an opacity or clouding that develops in the crystalline lens of the eye or in its capsule.

Scoliosis
The presence of an abnormal lateral curvature of the spine.

Extrahepatic cholestasis
Impairment of bile flow due to obstruction in large bile ducts outside the liver.

Triphalangeal thumb
A thumb with three phalanges in a single, proximo-distal axis. Thus, this term applies if the thumb has an accessory phalanx, leading to a digit like appearance of the thumb.

Leukopenia
An abnormal decreased number of leukocytes in the blood.

Aplasia/Hypoplasia of the uvula
Underdevelopment or absence of the uvula.

Hydrocephalus
Hydrocephalus is an active distension of the ventricular system of the brain resulting from inadequate passage of CSF from its point of production within the cerebral ventricles to its point of absorption into the systemic circulation.

Pancreatic adenocarcinoma
The presence of an adenocarcinoma of the pancreas.

Short palpebral fissure
Distance between the medial and lateral canthi is more than 2 SD below the mean for age (objective); or, apparently reduced length of the palpebral fissures.

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.

Decreased fertility in males

Abnormality of the liver
An abnormality of the liver.

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).

Abnormal thumb morphology

An abnormal structure of the first digit of the hand.

Chromosomal breakage induced by crosslinking agents
Increased amount of chromosomal breaks in cultured blood lymphocytes or other cells induced by treatment with DNA cross-linking agents such as diepoxybutane and mitomycin C.

Meckel diverticulum
Meckel's diverticulum is a congenital diverticulum located in the distal ileum.

Almond-shaped palpebral fissure
A shape created by an acute downward arching of the upper eyelid and upward arching of the lower eyelid, toward the medial canthus, which gives the outline of the palpebral fissures the configuration of an almond. Thus, the maximum distance between the fissures is offset from, and medial to, the center point.

Glioblastoma multiforme
A tumor arising from glia in the central nervous system with macroscopic regions of necrosis and hemorrhage. Microscopically, glioblastoma multiforme is characterized by regions of pseudopalisading necrosis, pleomorphic nuclei and cells, and microvascular proliferation.

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

Abnormal nervous system morphology
A structural anomaly of the nervous system.

Primary peritoneal carcinoma
A type of cancer that originates in the peritoneum. It is to be distinguished from metastatic cancer of the peritoneum. Peritoneal cancer can occur anywhere in the abdominal space, and affects the surface of organs contained inside the peritoneum.

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

Global developmental delay
A delay in the achievement of motor or mental milestones in the domains of development of a child, including motor skills, speech and language, cognitive skills, and social and emotional skills. This term should only be used to describe children younger than five years of age.

Thrombocytopenia
A reduction in the number of circulating thrombocytes.

Pyridoxine-responsive sideroblastic anemia
A type of sideroblastic anemia that is alleviated by pyridoxine (vitamin B-6) treatment.

Growth delay
A deficiency or slowing down of growth pre- and postnatally.

Multiple cafe-au-lait spots

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

Astrocytoma
Astrocytoma is a neoplasm of the central nervous system derived from astrocytes. Astrocytes are a type of glial cell, and thus astrocytoma is a subtype of glioma.

T-cell acute lymphoblastic leukemias
Acute lymphoblastic leukemia of T-cell origin. It comprises about 15% of childhood cases and 25% of adult cases. It is more common in males than females.

Hypospadias
Abnormal position of urethral meatus on the ventral penile shaft (underside) characterized by displacement of the urethral meatus from the tip of the glans penis to the ventral surface of the penis, scrotum, or perineum.

Visual impairment
Visual impairment (or vision impairment) is vision loss (of a person) to such a degree as to qualify as an additional support need through a significant limitation of visual capability resulting from either disease, trauma, or congenital or degenerative conditions that cannot be corrected by conventional means, such as refractive correction, medication, or surgery.

Hypertelorism
Interpupillary distance more than 2 SD above the mean (alternatively, the appearance of an increased interpupillary distance or widely spaced eyes).

Spina bifida
Incomplete closure of the embryonic neural tube, whereby some vertebral arches remain unfused and open. The mildest form is spina bifida occulta, followed by meningocele and meningomyelocele.

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.

Functional intestinal obstruction

Ovarian neoplasm
A tumor (abnormal growth of tissue) of the ovary.

Tetralogy of Fallot
A congenital cardiac malformation comprising pulmonary stenosis, overriding aorta, ventricular septum defect, and right ventricular hypertrophy. The diagnosis of TOF is made if at least three of the four above mentioned features are present.

Bone marrow hypocellularity
A reduced number of hematopoietic cells present in the bone marrow relative to marrow fat.

Heterogeneous

Microphthalmia
A developmental anomaly characterized by abnormal smallness of one or both eyes.

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 fallopian tube morphology
An abnormality of the fallopian tube.

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

Multifactorial inheritance
A mode of inheritance that depends on a mixture of major and minor genetic determinants possibly together with environmental factors. Diseases inherited in this manner are termed complex diseases.

Ovarian carcinoma
A malignant neoplasm originating from the surface ovarian epithelium.

Hypoplasia of the ulna
Underdevelopment of the ulna.

Peritoneal abscess
The presence of an abscess of the peritoneum.

Abnormality of the hypothalamus-pituitary axis
Abnormality of the pituitary gland (also known as hypophysis), which is an endocrine gland that protrudes from the bottom of the hypothalamus at the base of the brain. The pituitary gland secretes the hormones ACTH, TSH, PRL, GH, endorphins, FSH, LH, oxytocin, and antidiuretic hormone. The secretion of hormones from the anterior pituitary is under the strict control of hypothalamic hormones, and the posterior pituitary is essentially an extension of the hypothalamus, so that hypothalamus and pituitary gland may be regarded as a functional unit.

Hyperreflexia
Hyperreflexia is the presence of hyperactive stretch reflexes of the muscles.

Abnormal localization of kidney
An abnormal site of the kidney.