Geneticheck - Genetic Report

Gene

TSC1

Associated Diseases

Focal Cortical Dysplasia Of Taylor
Tuberous Sclerosis Complex
Tuberous Sclerosis-1
Lymphangioleiomyomatosis
Lymphangioleiomyomatosis

Phenotype

Renal angiomyolipoma
A benign renal neoplasm composed of fat, vascular, and smooth muscle elements.

Infantile spasms
Infantile spasms represent a subset of "epileptic spasms". Infantile Spasms are epileptic spasms starting in the first year of life (infancy).

Adenoma sebaceum
The presence of a sebaceous adenoma with origin in the sebum secreting cells of the skin.

Autism
Autism is a neurodevelopmental disorder characterized by impaired social interaction and communication, and by restricted and repetitive behavior. Autism begins in childhood. It is marked by the presence of markedly abnormal or impaired development in social interaction and communication and a markedly restricted repertoire of activity and interest. Manifestations of the disorder vary greatly depending on the developmental level and chronological age of the individual (DSM-IV).

Subcutaneous nodule
Slightly elevated lesions on or in the skin with a diameter of over 5 mm.

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

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.

Impulsivity
Acting on the spur of the moment in response to immediate stimuli; acting on a momentary basis without a plan or consideration of outcomes; difficulty establishing or following plans; a sense of urgency and self-harming behavior under emotional distress.

Hemimegalencephaly

Enlargement of all or parts of one cerebral hemisphere.

Restrictive ventilatory defect
A functional defect characterized by reduced total lung capacity (TLC) not associated with abnormalities of expiratory airflow or airway resistance. Spirometrically, a restrictive defect is defined as FEV1 (forced expiratory volume in 1 second) and FVC (forced vital capacity) less than 80 per cent. Restrictive lung disease may be caused by alterations in lung parenchyma or because of a disease of the pleura, chest wall, or neuromuscular apparatus.

Aggressive behavior
Aggressive behavior can denote verbal aggression, physical aggression against objects, physical aggression against people, and may also include aggression towards oneself.

Retinal astrocytic hamartoma
A glial tumor of the retinal nerve fiber layer arising from a retinal astrocyte.

Dental enamel pits
The presence of small depressions in the dental enamel.

Angiofibromas
Angiofibroma consist of many often dilated vessels.

Ependymoma
The presence of an ependymoma of the central nervous system.

Respiratory failure
A severe form of respiratory insufficiency characterized by inadequate gas exchange such that the levels of oxygen or carbon dioxide cannot be maintained within normal limits.

Status epilepticus
Status epilepticus is a type of prolonged seizure resulting either from the failure of the mechanisms responsible for seizure termination or from the initiation of mechanisms which lead to abnormally prolonged seizures (after time point t1). It is a condition that can have long-term consequences (after time point t2), including neuronal death, neuronal injury, and alteration of neuronal networks, depending on the type and duration of seizures.

Abnormal urinary color
An abnormal color of the urine, that is, the color of the urine appears different from the usual straw-yellow color.

Preauricular hair displacement
An tongue-like extension of hair towards the cheeks, in which hair growth extends in front of the ear to the lateral cheekbones.

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).

Poor speech

Macule
A flat, distinct, discolored area of skin less than 1 cm wide that does not involve any change in the thickness or texture of the skin.

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.

Gingival fibromatosis
The presence of fibrosis of the gingiva.

Pulmonary infiltrates

Pituitary adenoma
A benign epithelial tumor derived from intrinsic cells of the adenohypophysis.

Atelectasis
Collapse of part of a lung associated with absence of inflation (air) of that part.

Hypomelanotic macule
Hypomelanotic macules ("ash leaf spots") are white or lighter patches of skin that may appear anywhere on the body and are caused by a lack of melanin. White ash leaf-shaped macules are considered to be characteristic of tuberous sclerosis.

Gastrointestinal hemorrhage
Hemorrhage affecting the gastrointestinal tract.

Abnormality of the lymphatic system
An anomaly of the lymphatic system, a network of lymphatic vessels that carry a clear fluid called lymph unidirectionally towards either the right lymphatic duct or the thoracic duct, which in turn drain into the right and left subclavian veins respectively.

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

Lymphadenopathy

Enlargment (swelling) of a lymph node.

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

Cough
A sudden, audible expulsion of air from the lungs through a partially closed glottis, preceded by inhalation.

Optic nerve glioma
A glioma originating in the optic nerve or optic chiasm.

Aortic aneurysm
Aortic dilatation refers to a dimension that is greater than the 95th percentile for the normal person age, sex and body size. In contrast, an aneurysm is defined as a localized dilation of the aorta that is more than 150 percent of predicted (ratio of observed to expected diameter 1.5 or more). Aneurysm should be distinguished from ectasia, which represents a diffuse dilation of the aorta less than 50 percent of normal aorta diameter.

Cardiac rhabdomyoma
A benign tumor of cardiac striated muscle.

Skin plaque
A plaque is a solid, raised, plateau-like (flat-topped) lesion greater than 1 cm in diameter.

Focal impaired awareness seizure
Focal impaired awareness seizure (or focal seizure with impaired or lost awareness) is a type of focal-onset seizure characterized by some degree (which may be partial) of impairment of the person's awareness of themselves or their surroundings at any point during the seizure.

Astrocytosis
Proliferation of astrocytes in the area of a lesion of the central nervous system.

Fever
Body temperature elevated above the normal range.

Optic atrophy
Atrophy of the optic nerve. Optic atrophy results from the death of the retinal ganglion cell axons that comprise the optic nerve and manifesting as a pale optic nerve on fundoscopy.

Attention deficit hyperactivity disorder
Attention deficit hyperactivity disorder (ADHD) manifests at age 2-3 years or by first grade at the latest. The main symptoms are distractibility, impulsivity, hyperactivity, and often trouble organizing tasks and projects, difficulty going to sleep, and social problems from being aggressive, loud, or impatient.

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.

Behavioral abnormality
An abnormality of mental functioning including various affective, behavioural, cognitive and perceptual

abnormalities.

Multiple renal cysts
The presence of many cysts in the kidney.

Repetitive compulsive behavior

Focal cortical dysplasia type II
A type of focal cortical dysplasia that is characterized by disrupted cortical lamination and specific cytological abnormalities.

Renal cell carcinoma
A type of carcinoma of the kidney with origin in the epithelium of the proximal convoluted renal tubule.

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.

Ascites
Accumulation of fluid in the peritoneal cavity.

Shagreen patch
A plaque representing a connective-tissue nevus. Connective tissue naevi are uncommon skin lesions that occur when the deeper layers of the skin do not develop correctly or the components of these layers occur in the wrong proportion. Shagreen patches are oval-shaped and nevoid, skin-colored or occasionally pigmented, smooth or crinkled, The word shagreen refers to a type of roughened untanned leather.

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.

Pneumothorax
Accumulation of air in the pleural cavity leading to a partially or completely collapsed lung.

Carcinoid tumor
A tumor formed from the endocrine (argentaffin) cells of the mucosal lining of a variety of organs including the stomach and intestine. These cells are from neuroectodermal origin.

Noncommunicating hydrocephalus
A form of hydrocephalus in which the flow of cerebrospinal fluid (CSF) within the cerebral ventricular system or in the outlets of the CSF to the arachnoid space is obstructed.

Achromatic retinal patches
Areas of the retina lacking pigmentation. Punched out areas of chorioretinal hypopigmentation less than 1 disc diameter in size and tending to be located in the midperiphery of the retina.

Cerebral calcification
The presence of calcium deposition within brain structures.

Parathyroid adenoma
A benign tumor of the parathyroid gland that can cause hyperparathyroidism.

Respiratory tract infection
An infection of the upper or lower respiratory tract.

Emphysema

Autistic behavior
Persistent deficits in social interaction and communication and interaction as well as a markedly restricted repertoire of activity and interest as well as repetitive patterns of behavior.

Dyspnea
Difficult or labored breathing. Dyspnea is a subjective feeling only the patient can rate, e.g., on a Borg scale.

Anxiety
Intense feelings of nervousness, tenseness, or panic, often in reaction to interpersonal stresses; worry about the negative effects of past unpleasant experiences and future negative possibilities; feeling fearful, apprehensive, or threatened by uncertainty; fears of falling apart or losing control.

Subependymal giant-cell astrocytoma
A demarcated, largely intraventricular tumor in the region of the foramen of Monro composed of spindle to large plump or ganglion-like cells with eosinophilic to amphophilic cytoplasm and somewhat pleomorphic nuclei with occasional prominent nucleoli. These tumors are almost always associated with tuberous sclerosis.

Chorioretinal hypopigmentation

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

Confetti-like hypopigmented macules

Pulmonary lymphangiomyomatosis
Infiltration of smooth muscle-like cells in lymph vessels as well as the lung (pleura, alveolar septa, bronchi, pulmonary vessels and lymphatics as well as lymph nodes, especially in posterior mediastinum and retroperitoneum). Focal emphysema can develop because of airway narrowing, and the thoracic duct may be obliterated. Pulmonary lymphangiomyomatosis may lead to multiple small cysts with a hamartomatous proliferation of smooth muscle in their walls.

Precocious puberty
The onset of secondary sexual characteristics before a normal age. Although it is difficult to define normal age ranges because of the marked variation with which puberty begins in normal children, precocious puberty can be defined as the onset of puberty before the age of 8 years in girls or 9 years in boys.

Chylopericardium

Accumulation of chyle (the whitish fluid taken up by the lacteals in the intestine, consisting of an emulsion of lymph and triglyceride fat thatpasses into the veins by the thoracic duct) in the pericardium. Chylopericardium is generally caused by obstruction of or trauma to the thoracic duct.

Pancreatic endocrine tumor
A neuroendocrine tumor originating in a hormone-producing cell (islet cell) of the pancreas.

Sleep disturbance
An abnormality of sleep including such phenomena as 1) insomnia/hypersomnia, 2) non-restorative sleep, 3) sleep schedule disorder, 4) excessive daytime somnolence, 5) sleep apnea, and 6) restlessness.

Wolff-Parkinson-White syndrome
A disorder of the cardiac conduction system of the heart characterized by ventricular preexcitation due to the presence of an abnormal accessory atrioventricular electrical conduction pathway.

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.

Subependymal nodules
Small nodular masses which originate in the subependymal region of the lateral ventricles and protrude into the ventricular cavity. They may represent subependymal hamartomas of tuberous sclerosis.

Internal hemorrhage
The presence of hemorrhage within the body.

Chordoma
A chordoma is a tumors that arises from embryonic remnants of the notochord along the length of the neuraxis. Chordomas generally occur in the sacrum, intracranially at the clivus, or along the spinal axis.

Hyperactivity
Hyperactivity is a state of constantly being unusually or abnormally active, including in situations in which it is not appropriate.

Specific learning disability
Impairment of certain skills such as reading or writing, coordination, self-control, or attention that interfere with the ability to learn. The impairment is not related to a global deficiency of intelligence.

Focal white matter lesions

Neurodevelopmental delay

Chylothorax
Accumulation of excessive amounts of lymphatic fluid (chyle) in the pleural cavity.

Ungual fibroma

Flesh-colored papule in or around the nail bed. Ungual fibromas may be periungual (arising under the proximal nail fold) or subungual (originating under the nail plate).

Focal-onset seizure
A focal-onset seizure is a type of seizure originating within networks limited to one hemisphere. They may be discretely localized or more widely distributed, and may originate in subcortical structures.

Self-injurious behavior
Aggression towards oneself.

Cortical dysplasia
The presence of developmental dysplasia of the cerebral cortex.

Epileptic spasm
A sudden flexion, extension, or mixed extension-flexion of predominantly proximal and truncal muscles that is usually more sustained than a myoclonic movement but not as sustained as a tonic seizure. Limited forms may occur: Grimacing, head nodding, or subtle eye movements. Epileptic spasms frequently occur in clusters. Infantile spasms are the best known form, but spasms can occur at all ages

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

Depression
Frequent feelings of being down, miserable, and/or hopeless; difficulty recovering from such moods; pessimism about the future; pervasive shame; feeling of inferior self-worth; thoughts of suicide and suicidal behavior.

Generalized abnormality of skin
An abnormality of the skin that is not localized to any one particular region.

Stage 5 chronic kidney disease
A degree of kidney failure severe enough to require dialysis or kidney transplantation for survival characterized by a severe reduction in glomerular filtration rate (less than 15 ml/min/1.73 m2) and other manifestations including increased serum creatinine.

Abnormality of the kidney
An abnormality of the kidney.

Renal cyst
A fluid filled sac in the kidney.

Renal neoplasm
The presence of a neoplasm of the kidney.

Hepatic cysts

Lymphedema
Localized fluid retention and tissue swelling caused by a compromised lymphatic system.

Cortical tubers
Cortical tubers in the brain are hamartomatous lesions typically located at the gray-white matter interface, commonly in the frontal and parietal lobes. Cortical tubers are composed of abnormal glial and neural cells, and the size, number, and location vary among patients.

Respiratory distress
Respiratory distress is objectively observable as the physical or emotional consequences from the experience of dyspnea. The physical presentation of respiratory distress is generally referred to as labored breathing, while the sensation of respiratory distress is called shortness of breath or dyspnea.

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.

Hemiparesis
Loss of strength in the arm, leg, and sometimes face on one side of the body. Hemiplegia refers to a complete loss of strength, whereas hemiparesis refers to an incomplete loss of strength.

Hemoptysis
Coughing up (expectoration) of blood or blood-streaked sputum from the larynx, trachea, bronchi, or lungs.

Pheochromocytoma
Pheochromocytomas (also known as chromaffin tumors) produce, store, and secrete catecholamines. Pheochromocytomas usually originate from the adrenal medulla but may also develop from chromaffin cells in or about sympathetic ganglia. A common symptom of pheochromocytoma is hypertension owing to release of catecholamines.

Premature chromatid separation
The presence of premature sister chromatid segregation.

Chronic kidney disease
Functional anomaly of the kidney persisting for at least three months.

Polycystic kidney dysplasia
The presence of multiple cysts in both kidneys.

Subungual fibromas
The presence of fibromata beneath finger or toenails.

Abnormal morphology of female internal genitalia
An abnormality of the female internal genitalia.

Hypothyroidism
Deficiency of thyroid hormone.

Sporadic
Cases of the disease in question occur without a previous family history, i.e., as isolated cases without being transmitted from a parent and without other siblings being affected.

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.

Parathyroid hyperplasia
Hyperplasia of the parathyroid gland.

Retinal hamartoma
A hamartoma (a benign, focal malformation consisting of a disorganized mixture of cells and tissues) of the retina.

Abnormal social behavior
An abnormality of actions or reactions of a person taking place during interactions with others.

Epidermoid cyst
Nontender, round and firm, but slightly compressible, intradermal or subcutaneous cyst measuring 0.5-5 cm in diameter. Epidermal cysts are intradermal or subcutaneous tumors, grow slowly and occur on the face, neck, back and scrotum. They usually appear at or around puberty, and as a rule an affected individual has one solitary or a few cysts.