Geneticheck - Genetic Report

Gene

PTEN

Associated Diseases

Meningioma, Familial, Susceptibility To
Prostate Cancer
Cowden Syndrome
Proteus Syndrome
Proteus-like Syndrome
Macrocephaly/autism Syndrome
Bannayan-riley-ruvalcaba Syndrome
Juvenile Polyposis Of Infancy
Bilateral Frontoparietal Polymicrogyria
Lhermitte-duclos Disease
Cowden Syndrome 1
Hereditary Breast And Ovarian Cancer Syndrome
Segmental Outgrowth-lipomatosis-arteriovenous Malformation-epidermal Nevus Syndrome

Phenotype

Irregular hyperpigmentation

Long face
Facial height (length) is more than 2 standard deviations above the mean (objective); or, an apparent increase in the height (length) of the face (subjective).

Narrow internal auditory canal
Reduction in diameter of the internal auditory canal.

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

Retinal detachment
Separation of the inner layers of the retina (neural retina) from the pigment epithelium.

Broad forehead
Width of the forehead or distance between the frontotemporales is more than two standard deviations above the mean (objective); or apparently increased distance between the two sides of the forehead.

Abnormality of the uterus
An abnormality of the uterus.

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

Abnormality of the parathyroid gland

An abnormality of the parathyroid gland.

Calvarial hyperostosis
Excessive growth of the calvaria.

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

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.

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).

Thick nasal alae
Increase in bulk of the ala nasi.

Abnormal large intestine morphology
Any abnormality of the large intestine.

Myofibrillar myopathy
Myofibrillar structural changes characterized by abnormal intracellular accumulation of the intermediate filament desmin and other proteins.

Downslanted palpebral fissures
The palpebral fissure inclination is more than two standard deviations below the mean.

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).

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.

Subcutaneous lipoma
The presence of subcutaneous lipoma.

Splenomegaly
Abnormal increased size of the spleen.

Acrokeratosis
Overgrowth of the stratum corneum characterized by flesh-coloured or slightly pigmented smooth or warty papules on the upper surface of hands and feet.

Hypoplasia of the maxilla
Abnormally small dimension of the Maxilla. Usually creating a malocclusion or malalignment between the upper and lower teeth or resulting in a deficient amount of projection of the base of the nose and lower midface region.

Abnormality of cardiovascular system morphology
Any structural anomaly of the heart and great vessels.

Limbal dermoid
A benign tumor typically found at the junction of the cornea and sclera (limbal epibullar dermoid).

Gastrointestinal hemorrhage
Hemorrhage affecting the gastrointestinal tract.

Thyroid carcinoma
The presence of a carcinoma of the thyroid gland.

Myopia
An abnormality of refraction characterized by the ability to see objects nearby clearly, while objects in the distance appear blurry.

Skeletal dysplasia
A general term describing features characterized by abnormal development of bones and connective tissues.

Goiter
An enlargement of the thyroid gland.

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

Hypopigmented skin patches

Hypoalbuminemia
Reduction in the concentration of albumin in the blood.

Skin tags
Cutaneous skin tags also known as acrochorda or fibroepithelial polyps are small benign tumours that may either form secondarily over time primarily in areas where the skin forms creases, such as the neck, armpit or groin or may also be present at birth, in which case they usually occur in the periauricular region.

Motor delay
A type of Developmental delay characterized by a delay in acquiring motor skills.

Low-set ears
Upper insertion of the ear to the scalp below an imaginary horizontal line drawn between the inner canthi of the eye and extending posteriorly to the ear.

Polycystic ovaries

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).

Abnormal dental enamel morphology
An abnormality of the dental enamel.

Delayed skeletal maturation
A decreased rate of skeletal maturation. Delayed skeletal maturation can be diagnosed on the basis of an estimation of the bone age from radiographs of specific bones in the human body.

High, narrow palate
The presence of a high and narrow palate.

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.

Upper limb asymmetry
Difference in length or size between the right and left arm.

Mandibular prognathia
Abnormal prominence of the chin related to increased length of the mandible.

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.

Angioid streaks of the fundus
Irregular lines in the deep retina that are typically configured in a radiating fashion and emanate from the optic disc. Angioid streaks are crack-like dehiscences in abnormally thickened and calcified Bruch's membrane, resulting in atrophy of the overlying retinal pigment epithelium. They may be associated with a number of

endocrine, metabolic, and connective tissue abnormalities but are frequently idiopathic.

Thrombophlebitis
Inflammation of a vein associated with venous thrombosis (blood clot formation within the vein).

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".

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

Excessive wrinkled skin

Broad phalanx of the toes
Increased width of phalanx of one or more toes.

Abnormality of the nail
Abnormality of the nail.

Palmoplantar keratoderma
Abnormal thickening of the skin of the palms of the hands and the soles of the feet.

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.

Cerebellar vermis hypoplasia
Underdevelopment of the vermis of cerebellum.

Hypoplasia of the brainstem
Underdevelopment of the brainstem.

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

Generalized hirsutism
Abnormally increased hair growth over much of the entire body.

Papilloma
A tumor of the skin or mucous membrane with finger-like projections.

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

Exostoses
An exostosis is a benign growth the projects outward from the bone surface. It is capped by cartilage, and

arises from a bone that develops from cartilage.

Postnatal macrocephaly
The postnatal development of an abnormally large skull (macrocephaly).

Epidermal nevus
Epidermal naevi are due to an overgrowth of the epidermis and may be present at birth (50%) or develop during childhood.

Colorectal polyposis
Multiple abnormal growths that arise from the lining of the large intestine (colon or rectum) and protrude into the intestinal lumen.

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.

Fibroma
Benign tumors that are composed of fibrous or connective tissue. They can grow in all organs, arising from mesenchyme tissue. The term "fibroblastic" or "fibromatous" is used to describe tumors of the fibrous connective tissue. When the term fibroma is used without modifier, it is usually considered benign, with the term fibrosarcoma reserved for malignant tumors.

Transitional cell carcinoma of the bladder
The presence of a carcinoma of the urinary bladder with origin in a transitional epithelial cell.

Myopathy
A disorder of muscle unrelated to impairment of innervation or neuromuscular junction.

Micrognathia
Developmental hypoplasia of the mandible.

Increased head circumference
An abnormally increased head circumference in a growing child. Head circumference is measured with a nonelastic tape and comprises the distance from above the eyebrows and ears and around the back of the head. The measured HC is then plotted on an appropriate growth chart.

Conjunctival hamartoma
A hamartoma (disordered proliferation of mature tissues) of the conjunctiva.

Furrowed tongue
Accentuation of the grooves on the dorsal surface of the tongue.

Abdominal wall muscle weakness
Decreased strength of the abdominal musculature.

Colonic diverticula
The presence of multiple diverticula of the colon.

Kyphosis
Exaggerated anterior convexity of the thoracic vertebral column.

Carious teeth
Caries is a multifactorial bacterial infection affecting the structure of the tooth. This term has been used to describe the presence of more than expected dental caries.

Intestinal bleeding
Bleeding from the intestines.

Short nose
Distance from nasion to subnasale more than two standard deviations below the mean, or alternatively, an apparently decreased length from the nasal root to the nasal tip.

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).

Intussusception
An abnormality of the intestine in which part of the intestine invaginates (telescopes) into another part of the intestine.

Nevus
A nevus is a type of hamartoma that is a circumscribed stable malformation of the skin.

Capillary hemangioma
The presence of a capillary hemangioma, which are hemangiomas with small endothelial spaces.

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

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.

Joint stiffness
Joint stiffness is a perceived sensation of tightness in a joint or joints when attempting to move them after a

period of inactivity. Joint stiffness typically subsides over time.

Melena
The passage of blackish, tarry feces associated with gastrointestinal hemorrhage. Melena occurs if the blood remains in the colon long enough for it to be broken down by colonic bacteria. One degradation product, hematin, imbues the stool with a blackish color. Thus, melena generally occurs with bleeding from the upper gastrointestinal tract (e.g., stomach ulcers or duodenal ulcers), since the blood usually remains in the gut for a longer period of time than with lower gastrointestinal bleeding.

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

Open bite
Visible space between the dental arches in occlusion.

Angina pectoris
Paroxysmal chest pain that occurs with exertion or stress and is related to myocardial ischemia.

Hypoplasia of the pons
Underdevelopment of the pons.

Abnormal penis morphology
Abnormality of the male external sex organ.

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.

Polymicrogyria
Polymicrogyria is a congenital malformation of the cerebral cortex characterized by abnormal cortical layering (lamination) and an excessive number of small gyri (folds).

Abnormality of the metacarpal bones
An abnormality of the metacarpal bones.

Macrodactyly
Significant increase in the length and girth of most or all of a digit compared to its contralateral digit (if unaffected) or compared to what would be expected for age/body build. The increased girth is accompanied by an increase in the dorso-ventral dimension AND the lateral dimension of the digit.

Rectal prolapse
Protrusion of the rectal mucous membrane through the anus.

Obesity
Accumulation of substantial excess body fat.

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.

Gait imbalance

Bilateral tonic-clonic seizure with generalized onset
A bilateral tonic-clonic seizure with generalized onset is a type of bilateral tonic-clonic seizure characterised by generalized onset; these seizures rapidly engage networks in both hemispheres at the start of the seizure.

Hyperthyroidism
An abnormality of thyroid physiology characterized by excessive secretion of the thyroid hormones thyroxine (i.e., T4) and/or 3,3',5-triiodo-L-thyronine zwitterion (i.e., triiodothyronine or T3).

Intracranial hemorrhage
Hemorrhage occurring within the skull.

Abnormality of the thyroid gland
An abnormality of the thyroid gland.

Varicocele
A varicocele is a widening of the veins along the spermatic cord, leading to enlarged, twisted veins in the scrotum, and manifested clinically by a painless testicle lump, scrotal swelling, or bulge in the scrotum.

Thyroiditis
Inflammation of the thyroid gland.

Lipoma
Benign neoplasia derived from lipoblasts or lipocytes of white or brown fat. May be angiomatous or hibernomatous.

Renal cyst
A fluid filled sac in the kidney.

Biparietal narrowing
A narrowing of the biparietal diameter (i.e., of the transverse distance between the protuberances of the two parietal bones of the skull).

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

Protein-losing enteropathy
Abnormal loss of protein from the digestive tract related to excessive leakage of plasma proteins into the lumen of the gastrointestinal tract.

Hamartoma
A disordered proliferation of mature tissues that is native to the site of origin, e.g., exostoses, nevi and soft tissue hamartomas. Although most hamartomas are benign, some histologic subtypes, e.g., neuromuscular hamartoma, may proliferate aggressively such as mesenchymal cystic hamartoma, Sclerosing epithelial hamartoma, Sclerosing metanephric hamartoma.

Retinal nonattachment
Failure of attachment of the retina during development.

Hashimoto thyroiditis
A chronic, autoimmune type of thyroiditis associated with hypothyroidism.

Visceral angiomatosis

Trichilemmoma
A benign tumour originating from the outer root sheath of the hair follicle.

Abnormality of retinal pigmentation

Asymmetry of the thorax
Lack of symmetry between the left and right halves of the thorax.

Meningioma
The presence of a meningioma, i.e., a benign tumor originating from the dura mater or arachnoid mater.

Tall stature
A height above that which is expected according to age and gender norms.

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

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.

Esotropia
A form of strabismus with one or both eyes turned inward ('crossed') to a relatively severe degree, usually defined as 10 diopters or more.

Hypothyroidism
Deficiency of thyroid hormone.

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

Cerebral dysmyelination
Defective structure and function of myelin sheaths of the white matter of the brain.

Long philtrum
Distance between nasal base and midline upper lip vermilion border more than 2 SD above the mean. Alternatively, an apparently increased distance between nasal base and midline upper lip vermilion border.

Bone cyst
A fluid filled cavity that develops with a bone.

Intellectual disability, mild
Mild intellectual disability is defined as an intelligence quotient (IQ) in the range of 50-69.

Macroorchidism
The presence of abnormally large testes.

Sirenomelia
A developmental defect in which the legs are fused together.

Abnormality of the wrist
Abnormality of the wrist, the structure connecting the hand and the forearm.

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

Pectus excavatum
A defect of the chest wall characterized by a depression of the sternum, giving the chest ("pectus") a caved-in ("excavatum") appearance.

Refractory anemia

Uterine neoplasm
A tumor (abnormal growth of tissue) of the uterus.

Palmoplantar hyperkeratosis
Abnormal thickening of the skin localized to the palm of the hand and the sole of the foot.

Hypoglycemia
A decreased concentration of glucose in the blood.

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

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.

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).

Communicating hydrocephalus
A form of hydrocephalus in which there is no visible obstruction to the flow of the cerebrospinal fluid between the ventricles and subarachnoid space.

Endometrial carcinoma
A carcinoma of the endometrium, the mucous lining of the uterus.

Hemimegalencephaly
Enlargement of all or parts of one cerebral hemisphere.

Hand polydactyly
A kind of polydactyly characterized by the presence of a supernumerary finger or fingers.

Progressive macrocephaly
The progressive development of an abnormally large skull.

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

Sudden cardiac death
The heart suddenly and unexpectedly stops beating resulting in death within a short time period (generally within 1 h of symptom onset).

Atonic seizure
Atonic seizure is a type of motor seizure characterized by a sudden loss or diminution of muscle tone without apparent preceding myoclonic or tonic event lasting about 1 to 2 seconds, involving head, trunk, jaw, or limb musculature.

Macrocephaly
Occipitofrontal (head) circumference greater than 97th centile compared to appropriate, age matched, sex-matched normal standards. Alternatively, a apparently increased size of the cranium.

Hematochezia
The passage of fresh (red) blood per anus, usually in or with stools. Most rectal bleeding comes from the colon, rectum, or anus.

Genu recurvatum
An abnormally increased extension of the knee joint, so that the knee can bend backwards.

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.

Neurological speech impairment

Cerebellar dysplasia
Cerebellar dysplasia (abnormal growth or development) is defined by abnormal cerebellar foliation, white matter arborization, and gray-white matter junction. Cerebellar dysplasia is a neuroimaging finding that describes abnormalities of both the cerebellar cortex and white matter and is associated with variable neurodevelopmental outcome. Dysplasia may globally involve the cerebellum or affect only one cerebellar hemisphere. In addition, cerebellar dysplasia may be associated with cortical/subcortical cysts.

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

Delayed gross motor development
A type of motor delay characterized by a delay in acquiring the ability to control the large muscles of the body for walking, running, sitting, and crawling.

Breast carcinoma
The presence of a carcinoma of the breast.

Narrow palate
Width of the palate more than 2 SD below the mean (objective) or apparently decreased palatal width (subjective).

Chorioretinal coloboma
Absence of a region of the retina, retinal pigment epithelium, and choroid.

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).

Ovarian cyst
The presence of one or more cysts of the ovary.

Abnormal cerebellum morphology
Any structural abnormality of the cerebellum.

Hemangioblastoma
A hemangioblastoma is a benign vascular neoplasm that arises almost exclusively in the central nervous system. Hemangioblastomas consist of a tightly packed cluster of small blood vessels forming a mass of up to 1 or 2 cm in diameter.

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

Abnormal lung lobation

A developmental defect in the formation of pulmonary lobes.

Central heterochromia
The presence of distinct colors in the central (pupillary) zone of the iris than in the mid-peripheral (ciliary) zone.

Neoplasm of the thyroid gland
A tumor (abnormal growth of tissue) of the thyroid gland.

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

Round face
The facial appearance is more circular than usual as viewed from the front.

Midclavicular hypoplasia
Underdevelopment of the middle portion of the clavicle.

Asymmetric growth
A growth pattern that displays an abnormal difference between the left and the right side.

Bronchogenic cyst
A rare congenital cystic lesion of the lungs in the mediastinum.

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

Intellectual disability, severe
Severe mental retardation is defined as an intelligence quotient (IQ) in the range of 20-34.

Abnormality of finger
An anomaly of a finger.

Metatarsus valgus
A condition in which the anterior part of the foot rotates outward away from the midline of the body and the heel remains straight.

Abnormality of the neck
An abnormality of the neck.

Hallux valgus
Lateral deviation of the great toe (i.e., in the direction of the little toe).

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

Anemia
A reduction in erythrocytes volume or hemoglobin concentration.

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

Hydrocele testis
Accumulation of clear fluid in the between the layers of membrane (tunica vaginalis) surrounding the testis.

Thickened skin
Laminar thickening of skin.

Venous insufficiency

Abnormal subcutaneous fat tissue distribution

Neoplasm of the adrenal cortex
The presence of a neoplasm of the adrenal cortex.

Narrow mouth
Distance between the commissures of the mouth more than 2 SD below the mean. Alternatively, an apparently decreased width of the oral aperture (subjective).

Short chin
Decreased vertical distance from the vermilion border of the lower lip to the inferior-most point of the chin.

Depressed nasal bridge
Posterior positioning of the nasal root in relation to the overall facial profile for age.

Hepatomegaly
Abnormally increased size of the liver.

Lower limb asymmetry
A difference in length or diameter between the left and right leg.

Melanocytic nevus
A oval and round, colored (usually medium-to dark brown, reddish brown, or flesh colored) lesion. Typically, a melanocytic nevus is less than 6 mm in diameter, but may be much smaller or larger.

Congestive heart failure
The presence of an abnormality of cardiac function that is responsible for the failure of the heart to pump blood at a rate that is commensurate with the needs of the tissues or a state in which abnormally elevated filling pressures are required for the heart to do so. Heart failure is frequently related to a defect in myocardial contraction.

Intention tremor
A type of kinetic tremor that occurs during target directed movement is called intention tremor. That is, an oscillatory cerebellar ataxia that tends to be absent when the limbs are inactive and during the first part of voluntary movement but worsening as the movement continues and greater precision is required (e.g., in touching a target such as the patient's nose or a physician's finger).

Muscle weakness
Reduced strength of muscles.

Joint hyperflexibility
Increased mobility and flexibility in the joint due to the tension in tissues such as ligaments and muscles.

Generalized myoclonic seizure
A generalized myoclonic seizure is a type of generalized motor seizure characterised by bilateral, sudden, brief (<100 ms) involuntary single or multiple contraction of muscles or muscle groups of variable topography (axial, proximal limb, distal). Myoclonus is less regularly repetitive and less sustained than is clonus.

Abnormal facial shape
An abnormal morphology (form) of the face or its components.

Cranial nerve paralysis

Abnormal form of the vertebral bodies
Abnormal morphology of vertebral body.

Papule
A circumscribed, solid elevation of skin with no visible fluid, varying in size from a pinhead to less than 10mm in diameter at the widest point.

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

Abnormal heart morphology
Any structural anomaly of the heart.

Recurrent infections
Increased susceptibility to infections.

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

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

Neoplasm of the central nervous system
A neoplasm of the central nervous system.

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.

Generalized hyperkeratosis

Large forehead

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.

Glaucoma
Glaucoma refers loss of retinal ganglion cells in a characteristic pattern of optic neuropathy usually associated with increased intraocular pressure.

Buphthalmos
Diffusely large eye (with megalocornea) associated with glaucoma.

Abnormal pupil morphology
An abnormality of the pupil.

Recurrent fractures
The repeated occurrence of bone fractures (implying an abnormally increased tendency for fracture).

Cutis marmorata
A reticular discoloration of the skin with cyanotic (reddish-blue appearing) areas surrounding pale central areas due to dilation of capillary blood vessels and stagnation of blood within the vessels. Cutis marmorata generally occurs on the legs, arms and trunk and is often more severe in cold weather.

Hemiplegia/hemiparesis
Loss of strength in the arm, leg, and sometimes face on one side of the body. Hemiplegia refers to a severe or complete loss of strength, whereas hemiparesis refers to a relatively mild loss of strength.

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

Talipes
A deformity of foot and ankle that has different subtypes that are talipes equinovarus, talipes equinovalgus, talipes calcaneovarus and talipes calcaneovalgus.

Abnormality of the optic nerve
Abnormality of the optic nerve.

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

Enlarged polycystic ovaries

Brachydactyly
Digits that appear disproportionately short compared to the hand/foot. The word brachydactyly is used here to describe a series distinct patterns of shortened digits (brachydactyly types A-E). This is the sense used here.

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.

Thyroid adenoma
The presence of a adenoma of the thyroid gland.

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).

Tooth agenesis
The absence of one or more teeth from the normal series by a failure
to develop

Anteverted nares
Anteriorly-facing nostrils viewed with the head in the Frankfurt horizontal and the eyes of the observer level with the eyes of the subject. This gives the appearance of an upturned nose (upturned nasal tip).

Hyperostosis
Excessive growth or abnormal thickening of bone tissue.

Enlarged cerebellum
An abnormally increased size of the cerebellum compared to other brain structures.

Follicular thyroid carcinoma
The presence of an follicular adenocarcinoma of the thyroid gland.

Macroglossia
Increased length and width of the tongue.

Language impairment
Language impairment is a deficit in comprehension or production of language that includes reduced vocabulary, limited sentence structure or impairments in written or spoken communication. Language abilities are substantially and quantifiably below age expectations.

Intestinal polyposis
The presence of multiple polyps in the intestine.

Hypotonia
Hypotonia is an abnormally low muscle tone (the amount of tension or resistance to movement in a muscle). Even when relaxed, muscles have a continuous and passive partial contraction which provides some resistance to passive stretching. Hypotonia thus manifests as diminished resistance to passive stretching. Hypotonia is not the same as muscle weakness, although the two conditions can co-exist.

Vascular skin abnormality

Increased intracranial pressure
An increase of the pressure inside the cranium (skull) and thereby in the brain tissue and cerebrospinal fluid.

Hamartomatous polyposis
Polyp-like protrusions which are histologically hamartomas. These can occur throughout the gastrointestinal tract. Hamartomatous polyps are composed of the normal cellular elements of the gastrointestinal tract, but have a markedly distorted architecture.

Generalized hyperpigmentation

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).

Generalized hypotonia
Generalized muscular hypotonia (abnormally low muscle tone).

Decreased muscle mass

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

Diabetes insipidus
A state of excessive water intake and hypotonic (dilute) polyuria. Diabetes insipidus may be due to failure of vasopressin (AVP) release (central or neurogenic diabetes insipidus) or to a failure of the kidney to respond to AVP (nephrogenic diabetes insipidus).

Abnormality of the vasculature
An abnormality of the vasculature.

Adenomatous colonic polyposis
Presence of multiple adenomatous polyps in the colon.

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

Abnormal pyramidal sign
Functional neurological abnormalities related to dysfunction of the pyramidal tract.

Telangiectasia
Telangiectasias refer to small dilated blood vessels located near the surface of the skin or mucous membranes, measuring between 0.5 and 1 millimeter in diameter. Telangiectasia are located especially on the tongue, lips, palate, fingers, face, conjunctiva, trunk, nail beds, and fingertips.

Freckled genitalia
One or more brown punctate macules on the skin of the genitalia.

Lymphangioma
Lymphangiomas are rare congenital malformations consisting of focal proliferations of well-differentiated lymphatic tissue in multi cystic or sponge like structures. Lymphangioma is usually asymptomatic due to its soft consistency but compression of adjacent structures can be seen due to the mass effect of a large tumor.

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

Abnormality of the kidney
An abnormality of the kidney.

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.

Heterochromia iridis
Heterochromia iridis is a difference in the color of the iris in the two eyes.

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.

Thymus hyperplasia
Enlargement of the thymus.

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

Long penis
Penile length more than 2 SD above the mean for age.

Broad thumb
Increased thumb width without increased dorso-ventral dimension.

Diarrhea
Abnormally increased frequency of loose or watery bowel movements.

Gynecomastia
Abnormal development of large mammary glands in males resulting in breast enlargement.

Gray matter heterotopia
Heterotopia or neuronal heterotopia are macroscopic clusters of misplaced neurons (gray matter), most often situated along the ventricular walls or within the subcortical white matter.

Cavernous hemangioma
The presence of a cavernous hemangioma. A hemangioma characterized by large endothelial spaces (caverns) is called a cavernous hemangioma.

Testicular neoplasm
The presence of a neoplasm of the testis.

Pulmonary embolism
An embolus (that is, an abnormal particle circulating in the blood) located in the pulmonary artery and thereby blocking blood circulation to the lung. Usually the embolus is a blood clot that has developed in an extremity (for instance, a deep venous thrombosis), detached, and traveled through the circulation before becoming trapped in the pulmonary artery.

Typical absence seizure
A typical absence seizure is a type of generalised non-motor (absence) seizure characterised by its sudden onset, interruption of ongoing activities, a blank stare, possibly a brief upward deviation of the eyes. Usually the patient will be unresponsive when spoken to. Duration is a few seconds to half a minute with very rapid recovery. Although not always available, an EEG would usually show 3 Hz generalized epileptiform discharges during the event.

Dysplastic gangliocytoma of the cerebellum
It is a rare, slowly growing tumor of the cerebellum, a gangliocytoma sometimes considered to be a hamartoma, characterized by diffuse hypertrophy of the granular layer of the cerebellum.

Lymphopenia
A reduced number of lymphocytes in the blood.

Disproportionate tall stature
A tall and slim body build with increased arm span to height ratio (>1.05) and a reduced upper-to-lower segment ratio (<0.85), i.e., unusually long arms and legs. The extremities as well as the hands and feet are unusually slim.

Abnormal fallopian tube morphology
An abnormality of the fallopian tube.

Macrotia
Median longitudinal ear length greater than two standard deviations above the mean and median ear width greater than two standard deviations above the mean (objective); or, apparent increase in length and width of the pinna (subjective).

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

Craniosynostosis
Craniosynostosis refers to the premature closure of the cranial sutures. Primary craniosynostosis refers to the

closure of one or more sutures due to abnormalities in skull development, and secondary craniosynostosis results from failure of brain growth.

Wide nose
Interalar distance more than two standard deviations above the mean for age, i.e., an apparently increased width of the nasal base and alae.

Carcinoma
A malignant tumor arising from epithelial cells. Carcinomas that arise from glandular epithelium are called adenocarcinomas, those that arise from squamous epithelium are called squamous cell carcinomas, and those that arise from transitional epithelium are called transitional cell carcinomas (NCI Thesaurus).

Fibroadenoma of the breast
A benign biphasic tumor of the breast with epithelial and stromal components.

Subcutaneous hemorrhage
This term refers to an abnormally increased susceptibility to bruising (purpura, petechiae, or ecchymoses).