Geneticheck - Genetic Report

Gene

SMAD4

Associated Diseases

Hereditary Hemorrhagic Telangiectasia
Myhre Syndrome
Juvenile Polyposis/hereditary Hemorrhagic Telangiectasia Syndrome
Familial Pancreatic Carcinoma
Myhre Syndrome
Generalized Juvenile Polyposis/juvenile Polyposis Coli
Familial Thoracic Aortic Aneurysm And Aortic Dissection
Pancreatic Cancer
Juvenile Polyposis Syndrome

Phenotype

High hypermetropia
A severe form of hypermetropia with over +5.00 diopters.

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

Skeletal muscle hypertrophy
Hypertrophy (increase in size) of muscle cells (as opposed to hyperplasia, which refers to an increase in the number of muscle cells).

Inguinal hernia
Protrusion of the contents of the abdominal cavity through the inguinal canal.

Duodenal adenocarcinoma
A malignant epithelial tumor with a glandular organization that originates in the duodenum.

Thin vermilion border
Height of the vermilion of the medial part of the lip more than 2 SD below the mean, or apparently reduced height of the vermilion of the lip in the frontal view. The vermilion is the red part of the lips (and confusingly, the vermilion itself is also often referred to as being equivalent the lips).

Prenatal maternal abnormality

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

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.

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

Mitral valve prolapse
One or both of the leaflets (cusps) of the mitral valve bulges back into the left atrium upon contraction of the left ventricle.

Overlapping toe
Describes a foot digit resting on the dorsal surface of an adjacent digit when the foot is at rest.

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

Gastrointestinal carcinoma

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.

Severe short stature
A severe degree of short stature, more than -4 SD from the mean corrected for age and sex.

Rectal polyposis
The presence of multiple rectal hyperplastic/adenomatous polyps.

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).

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).

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.

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.

Colon cancer

Bruising susceptibility
An ecchymosis (bruise) refers to the skin discoloration caused by the escape of blood into the tissues from ruptured blood vessels. This term refers to an abnormally increased susceptibility to bruising. The corresponding phenotypic abnormality is generally elicited on medical history as a report of frequent ecchymoses or bruising without adequate trauma.

Gastrointestinal hemorrhage
Hemorrhage affecting the gastrointestinal tract.

Lymphadenopathy
Enlargment (swelling) of a lymph node.

Stiff skin
An induration (hardening) of the skin

Unilateral cleft lip
A non-midline cleft of the upper lip on one side only.

Hypogonadism
A decreased functionality of the gonad.

Hypoalbuminemia
Reduction in the concentration of albumin in the blood.

Esophageal varix
Extreme dilation of the submucusoal veins in the lower portion of the esophagus.

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.

Dilatation of the cerebral artery
The presence of a localized dilatation or ballooning of a cerebral artery.

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.

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.

Limitation of joint mobility
A reduction in the freedom of movement of one or more joints.

Descending aortic dissection
A separation of the layers within the wall of the descending aorta. Tears in the intimal layer result in the propagation of dissection (proximally or distally) secondary to blood entering the intima-media space.

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

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

Cirrhosis
A chronic disorder of the liver in which liver tissue becomes scarred and is partially replaced by regenerative nodules and fibrotic tissue resulting in loss of liver function.

Multiple gastric polyps

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.

Increased level of L-fucose in urine
An increase in the level of L-fucose in the urine.

Microcytic anemia
A kind of anemia in which the volume of the red blood cells is reduced.

Carotid artery dilatation
A dilatation (balooning or bulging out of the vessel wall) of a carotid artery.

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

Aortic root aneurysm
An abnormal localized widening (dilatation) of the aortic root.

Hepatic arteriovenous malformation

Camptodactyly
The distal interphalangeal joint and/or the proximal interphalangeal joint of the fingers or toes cannot be extended to 180 degrees by either active or passive extension.

Pulmonary arterial hypertension
Pulmonary hypertension is defined mean pulmonary artery pressure of 25mmHg or more and pulmonary capillary wedge pressure of 15mmHg or less when measured by right heart catheterisation at rest and in a supine position.

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.

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).

Cerebral arteriovenous malformation
An anomalous configuration of blood vessels that shunts arterial blood directly into veins without passing through the capillaries and that is located in the brain.

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

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.

Short long bone
One or more abnormally short long bone.

Short philtrum
Distance between nasal base and midline upper lip vermilion border more than 2 SD below the mean. Alternatively, an apparently decreased distance between nasal base and midline upper lip vermilion border.

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

Abnormality of epiphysis morphology
An anomaly of epiphysis, which is the expanded articular end of a long bone that developes from a secondary

ossification center, and which during the period of growth is either entirely cartilaginous or is separated from the shaft by a cartilaginous disk.

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.

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

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.

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

Enlarged vertebral pedicles
Increased size of the vertebral pedicle.

Transient ischemic attack

Gingival cleft
A fissure in the gingiva (gums), i.e., the mucosal tissue that lies over the mandible and maxilla.

Abnormal penis morphology
Abnormality of the male external sex organ.

Retinal telangiectasia
Dilatation of small blood vessels of the retina.

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

Generalized muscle hypertrophy
Hypertrophy (increase in size) of muscle cells in a generalized (not localized) distribution.

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.

Thin upper lip vermilion
Height of the vermilion of the upper lip in the midline more than 2 SD below the mean. Alternatively, an apparently reduced height of the vermilion of the upper lip in the frontal view (subjective).

Portal hypertension
Increased pressure in the portal vein.

Abnormality of the pubic bone
An anomaly of the the pubic bone, i.e., of the ventral and anterior of the three principal components (publis, ilium, ischium) of the hip bone.

Abnormality of the ribs
An anomaly of the rib.

Nephrolithiasis
The presence of calculi (stones) in the kidneys.

Epistaxis
Epistaxis, or nosebleed, refers to a hemorrhage localized in the nose.

Arachnodactyly
Abnormally long and slender fingers ("spider fingers").

Cerebral hemorrhage
Hemorrhage into the parenchyma of the brain.

Cardiomegaly
Increased size of the heart, clinically defined as an increased transverse diameter of the cardiac silhouette that is greater than or equal to 50% of the transverse diameter of the chest (increased cardiothoracic ratio) on a posterior-anterior projection of a chest radiograph or a computed tomography.

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

Fine hair
Hair that is fine or thin to the touch.

Prominent nasal bridge

Anterior positioning of the nasal root in comparison to the usual positioning for age.

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

Visceral angiomatosis

Short neck
Diminished length of the neck.

Ventricular septal defect
A hole between the two bottom chambers (ventricles) of the heart. The defect is centered around the most superior aspect of the ventricular septum.

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).

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

Respiratory insufficiency

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

Broad ribs
Increased width of ribs

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

Cholelithiasis
Hard, pebble-like deposits that form within the gallbladder.

Pulmonary arteriovenous malformation
Pulmonary arteriovenous malformation, a condition most commonly associated with hereditary hemorrhagic telangiectasia, is an abnormal communication between the pulmonary artery and pulmonary vein without an

intervening capillary communication. HRCT images usually show a coarse spidery appearance of the peripheral vascular markings in the lungs. More specific findings are obtained in the pulmonary angiogram where the normally invisible capillary phase is replaced by irregular vascular channels bridging the peripheral branches of pulmonary arteries and veins.

Birth length less than 3rd percentile

Abnormal cerebral vascular morphology
An anomaly of the cerebral blood vessels.

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).

Short toe
A toe that appears disproportionately short compared to the foot.

Thickened calvaria
The presence of an abnormally thick calvaria.

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.

Midface retrusion
Posterior positions and/or vertical shortening of the infraorbital and perialar regions, or increased concavity of the face and/or reduced nasolabial angle.

Anemic pallor
A type of pallor that is secondary to the presence of anemia.

Laryngotracheal stenosis

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.

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.

Edema
An abnormal accumulation of fluid beneath the skin, or in one or more cavities of the body.

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.

Microtia
Underdevelopment of the external ear.

Hepatosplenomegaly
Simultaneous enlargement of the liver and spleen.

Clubbing
Broadening of the soft tissues (non-edematous swelling of soft tissues) of the digital tips in all dimensions associated with an increased longitudinal and lateral curvature of the nails.

Abdominal aortic aneurysm
An abnormal localized widening (dilatation) of the abdominal aorta.

Hypokalemia
An abnormally decreased potassium concentration in the blood.

Aortic dissection
Aortic dissection refers to a tear in the intimal layer of the aorta causing a separation between the intima and the medial layers of the aorta.

Descending thoracic aorta aneurysm
An abnormal localized widening (dilatation) of the descending thoracic aorta.

Abnormality of the metaphysis
An abnormality of one or more metaphysis, i.e., of the somewhat wider portion of a long bone that is adjacent to the epiphyseal growth plate and grows during childhood.

Epispadias
Displacement of the urethral opening on the dorsal (superior) surface of the penis.

Coarctation of aorta
Coarctation of the aorta is a narrowing or constriction of a segment of the aorta.

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

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.

Retrognathia
An abnormality in which the mandible is mislocalised posteriorly.

Sparse hair
Reduced density of hairs.

Abnormal cardiovascular system physiology
Abnormal functionality of the cardiovascular system.

Abnormality of the voice

Duodenal polyposis
Presence of multiple polyps in the duodenum.

Large iliac wing
Increased size of the ilium ala.

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

Spontaneous hematomas
Spontaneous development of hematomas (hematoma) or bruises without significant trauma.

Clinodactyly
An angulation of a digit at an interphalangeal joint in the plane of the palm (finger) or sole (toe).

EMG abnormality
Abnormal results of investigations using electromyography (EMG).

Anemia
A reduction in erythrocytes volume or hemoglobin concentration.

Thickened skin
Laminar thickening of skin.

Mitral regurgitation
An abnormality of the mitral valve characterized by insufficiency or incompetence of the mitral valve resulting in retrograde leaking of blood through the mitral valve upon ventricular contraction.

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).

Abnormal lip morphology
An abnormality of the lip.

Deeply set eye
An eye that is more deeply recessed into the plane of the face than is typical.

Craniofacial hyperostosis
Excessive growth of the craniofacial bones.

Stroke
Sudden impairment of blood flow to a part of the brain due to occlusion or rupture of an artery to the brain.

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.

Ischemic stroke
Acute ischemic stroke (AIS) is defined by the sudden loss of blood flow to an area of the brain with the resulting loss of neurologic function. It is caused by thrombosis or embolism that occludes a cerebral vessel supplying a specific area of the brain. During a vessel occlusion, there is a core area where damage to the brain is irreversible and an area of penumbra where the brain has lost function owing to decreased blood flow but is not irreversibly injured.

Abnormality of connective tissue
Any abnormality of the soft tissues, including both connective tissue (tendons, ligaments, fascia, fibrous tissues, and fat).

Abnormal sternum morphology
An anomaly of the sternum, also known as the breastbone.

Radial deviation of finger
Bending or curvature of a finger toward the radial side (i.e., towards the thumb). The deviation is at the metacarpal-phalangeal joint, and this finding is distinct from clinodactyly.

Small for gestational age
Smaller than normal size according to sex and gestational age related norms, defined as a weight below the 10th percentile for the gestational age.

Hepatic failure

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

abnormalities.

Migraine
Migraine is a chronic neurological disorder characterized by episodic attacks of headache and associated symptoms.

Platyspondyly
A flattened vertebral body shape with reduced distance between the vertebral endplates.

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.

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.

Peripheral arterial stenosis
Narrowing of peripheral arteries with reduction of blood flow to the limbs. This feature may be quantified as an ankle-brachial index of less than 0.9, and may be manifested clinically as claudication.

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

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

Hypovolemia
An decrease in the amount of intravascular fluid, particularly in the volume of the circulating blood.

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.

Ascending aortic dissection
A separation of the layers within the wall of the ascending aorta. Tears in the intimal layer result in the propagation of dissection (proximally or distally) secondary to blood entering the intima-media space.

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.

Small intestinal polyposis
The presence of multiple polyps in the small intestine.

Paroxysmal dyspnea

A sudden attack of dyspnea that occurs while the affected person is at rest.

Subarachnoid hemorrhage
Hemorrhage occurring between the arachnoid mater and the pia mater.

Submucous cleft hard palate
Hard-palate submucous clefts are characterized by bony defects in the midline of the bony palate that are covered by the mucous membrane of the roof of the mouth. It may be possible to detect a submucous cleft hard palate upon palpation as a notch in the bony palate.

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

Blepharophimosis
A fixed reduction in the vertical distance between the upper and lower eyelids with short palpebral fissures.

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

Hypoplastic iliac wing
Underdevelopment of the ilium ala.

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.

Abnormal left ventricular function
Inability of the left ventricle to perform its normal physiologic function. Failure is either due to an inability to contract the left ventricle or the inability to relax completely and fill with blood during diastole.

Pericardial effusion
Accumulation of fluid within the pericardium.

Amblyopia
Reduced visual acuity that is uncorrectable by lenses in the absence of detectable anatomic defects in the eye or visual pathways.

Short palm
Short palm.

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).

Mucoid extracellular matrix accumulation
An increase of medial mucoid extracellular matrix creating translamellar and/or intralamellar expansions including extracellular pools as noted on an H&E stain and/or a stain to highlight extracellular matrix material (Movat's pentachrome, Alcian blue, etc.).

Vertebral fusion
A developmental defect leading to the union of two adjacent vertebrae.

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.

Aortic regurgitation
An insufficiency of the aortic valve, leading to regurgitation (backward flow) of blood from the aorta into the left ventricle.

Cleft lip
A gap in the lip or lips.

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

Intestinal polyposis
The presence of multiple polyps in the intestine.

Pancreatic adenocarcinoma
The presence of an adenocarcinoma of the pancreas.

Short finger
Abnormally short finger associated with developmental hypoplasia.

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.

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.

Bifid uvula
Uvula separated into two parts most easily seen at the tip.

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.

Narrow palpebral fissure
Reduction in the vertical distance between the upper and lower eyelids.

External genital hypoplasia
Underdevelopment of part or all of the external reproductive organs.

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

Dural ectasia
A widening or ballooning of the dural sac surrounding the spinal cord usually at the lumbosacral level.

Cholecystitis
The presence of inflammatory changes in the gallbladder.

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

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.

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.

Bicuspid aortic valve
The presence of an aortic valve with two instead of the normal three cusps (flaps). Bicuspid aortic valvue is a malformation of a commissure (small space between the attachment of each cusp to the aortic wall) and the adjacent parts of the two corresponding cusps forming a raphe (the fused area of the two underdeveloped cusps turning into a malformed commissure between both cusps; the raphe is a fibrous ridge that extends from the commissure to the free edge of the two underdeveloped, conjoint cusps).

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.

Coronary artery atherosclerosis
Reduction of the diameter of the coronary arteries as the result of an accumulation of atheromatous plaques within the walls of the coronary arteries, which increases the risk of myocardial ischemia.

Diarrhea
Abnormally increased frequency of loose or watery bowel movements.

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

Thick eyebrow
Increased density/number and/or increased diameter of eyebrow hairs.

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

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.

2-3 toe syndactyly
Syndactyly with fusion of toes two and three.

Peripheral arteriovenous fistula

Aortic valve stenosis
The presence of a stenosis (narrowing) of the aortic valve.

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.

Venous thrombosis
Formation of a blood clot (thrombus) inside a vein, causing the obstruction of blood flow.

Abnormality iris morphology
An abnormality of the iris, which is the pigmented muscular tissue between the cornea and the lens, that is perforated by an opening called the pupil.

Juvenile gastrointestinal polyposis
The presence of multiple juvenile polyps in the stomach and intestine. The term juvenile polyps refer to a special histopathology and not the age of onset as the polyp might be diagnosed at all ages. The juvenile polyp has a spherical appearance and is microscopically characterized by overgrowth of an oedematous lamina propria with inflammatory cells and cystic glands. Juvenile polyps are a specific type of hamartomatous polyps.

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

Malar flattening
Underdevelopment of the malar prominence of the jugal bone (zygomatic bone in mammals), appreciated in profile, frontal view, and/or by palpation.

Femoral hernia
A hernia which occurs just below the inguinal ligament, where abdominal contents pass through a naturally occurring weakness called the femoral canal.

Peritoneal abscess
The presence of an abscess of the peritoneum.

Cone-shaped epiphysis
Cone-shaped epiphyses (also known as coned epiphyses) are epiphyses that invaginate into cupped metaphyses. That is, the epiphysis has a cone-shaped distal extension resulting from increased growth of the central portion of the epiphysis relative to its periphery.