Geneticheck - Genetic Report

Gene

LMNA

Associated Diseases

Emery-dreifuss Muscular Dystrophy 3, Autosomal Recessive
Hutchinson-gilford Progeria Syndrome
Familial Isolated Dilated Cardiomyopathy
Familial Partial Lipodystrophy, Dunnigan Type
Restrictive Dermopathy, Lethal
Hutchinson-gilford Progeria Syndrome
Atypical Werner Syndrome
Dilated Cardiomyopathy-hypergonadotropic Hypogonadism Syndrome
Lmna-related Cardiocutaneous Progeria Syndrome
Congenital Muscular Dystrophy Due To Lmna Mutation
Malouf Syndrome
Restrictive Dermopathy
Lipodystrophy, Familial Partial, Type 2
Familial Dilated Cardiomyopathy With Conduction Defect Due To Lmna Mutation
Cardiomyopathy, Dilated, 1a
Familial Partial Lipodystrophy, Köbberling Type
Mandibuloacral Dysplasia With Type A Lipodystrophy
Autosomal Recessive Emery-dreifuss Muscular Dystrophy
Mandibuloacral Dysplasia
Autosomal Semi-dominant Severe Lipodystrophic Laminopathy
Charcot-marie-tooth Disease, Axonal, Type 2b1
Muscular Dystrophy, Congenital, Lmna-related
Heart-hand Syndrome, Slovenian Type
Heart-hand Syndrome, Slovenian Type
Autosomal Dominant Emery-dreifuss Muscular Dystrophy
Emery-dreifuss Muscular Dystrophy 2, Autosomal Dominant

Phenotype

Feeding difficulties
Impaired ability to eat related to problems gathering food and getting ready to suck, chew, or swallow it.

Widely patent fontanelles and sutures
An abnormally increased width of the cranial fontanelles and sutures.

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).

Increased intraabdominal fat
An abnormal increase in the amount of intraabdominal fat tissue.

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).

Abnormality of skeletal muscle fiber size
Any abnormality of the size of the skeletal muscle cell.

Patchy alopecia
Transient, non-scarring hair loss and preservation of the hair follicle located in in well-defined patches.

Rimmed vacuoles
Presence of abnormal vacuoles (membrane-bound organelles) in the sarcolemma. On histological staining with hematoxylin and eosin, rimmed vacuoles are popcorn-like clear vacuoles with a densely blue rim. The vacuoles are often associated with cytoplasmic and occasionally intranuclear eosinophilic inclusions.

Delayed cranial suture closure
Infants normally have two fontanels at birth, the diamond-shaped anterior fontanelle at the junction of the coronal and sagittal sutures, and the posterior fontanelle at the intersection of the occipital and parietal bones. The posterior fontanelle usually closes by the 8th week of life, and the anterior fontanel closes by the 18th month of life on average. This term applies if there is delay of closure of the fontanelles beyond the normal age.

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).

Hyperkeratosis
Hyperkeratosis is thickening of the outer layer of the skin, the stratum corneum, which is composed of large, polyhedral, plate-like envelopes filled with keratin which are the dead cells that have migrated up from the stratum granulosum.

Congenital muscular dystrophy

Scleroderma
A chronic autoimmune phenomenon characterized by fibrosis (or hardening) and vascular alterations of the skin.

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

Dystrophic toenail
Toenail changes apart from changes of the color of the toenail (nail dyschromia) that involve partial or complete disruption of the various keratinous layers of the nail plate.

Ventricular hypertrophy
Enlargement of the cardiac ventricular muscle tissue with increase in the width of the wall of the ventricle and loss of elasticity. Ventricular hypertrophy is clinically differentiated into left and right ventricular hypertrophy.

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.

Second degree atrioventricular block
An intermittent atrioventricular block with failure of some atrial impulses to conduct to the ventricles, i.e., some but not all atrial impulses are conducted through the atrioventricular node and trigger ventricular contraction.

Proximal upper limb muscle hypertrophy

Short lingual frenulum
The presence of an abnormally short lingual frenulum.

Splenomegaly
Abnormal increased size of the spleen.

Bird-like facies

Abnormality of the nasal tip
An abnormality of the nasal tip.

Hypoplasia of teeth
Developmental hypoplasia of teeth.

Muscle hypertrophy of the lower extremities
Muscle hypertrophy primarily affecting the legs.

Neoplasm of the oral cavity
A tumor (abnormal growth of tissue) of the oral cavity.

Pes cavus
The presence of an unusually high plantar arch. Also called high instep, pes cavus refers to a distinctly hollow form of the sole of the foot when it is bearing weight.

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

Hypogonadism
A decreased functionality of the gonad.

Sensorineural hearing impairment
A type of hearing impairment in one or both ears related to an abnormal functionality of the cochlear nerve.

Decreased motor nerve conduction velocity
A type of decreased nerve conduction velocity that affects the motor neuron.

Neck muscle weakness
Decreased strength of the neck musculature.

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

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

Decreased calvarial ossification
Abnormal reduction in ossification of the calvaria (roof of the skull consisting of the frontal bone, parietal bones, temporal bones, and occipital bone).

Abnormal foot morphology
An abnormality of the skeleton of foot.

Skin ulcer
A discontinuity of the skin exhibiting complete loss of the epidermis and often portions of the dermis and even subcutaneous fat.

Microcolon
A colon of abnormally small caliber.

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

Restricted neck movement due to contractures

Reduced subcutaneous adipose tissue
A reduced amount of fat tissue in the lowest layer of the integument. This feature can be appreciated by a reduced skinfold thickness.

Laryngomalacia
Laryngomalacia is a congenital abnormality of the laryngeal cartilage in which the cartilage is floppy and prolapses over the larynx during inspiration.

Peripheral axonal atrophy
Atrophic changes of axons of the peripheral nervous system.

Increased facial adipose tissue
An increased amount of subcutaneous fat tissue in the face.

Hypergonadotropic hypogonadism
Reduced function of the gonads (testes in males or ovaries in females) associated with excess pituitary

gonadotropin secretion and resulting in delayed sexual development and growth delay.

Postnatal growth retardation
Slow or limited growth after birth.

Abnormality of the nail
Abnormality of the nail.

Distal lower limb amyotrophy
Muscular atrophy of distal leg muscles.

Increased intramuscular fat
An abnormal increase in the amount of intramuscular fat tissue.

Peroneal muscle weakness
Weakness of the peroneal muscles.

Generalized lipodystrophy
Generalized degenerative changes of the fat tissue.

Loss of eyelashes
This term refers to the loss of eyelashes that were previously present.

Chondrocalcinosis
Radiographic evidence of articular calcification that represent calcium pyrophosphate depositions in soft tissue surrounding joints and at the insertions of tendons near joints (Entheses/Sharpey fibers) .

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

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

Insulin resistance
Increased resistance towards insulin, that is, diminished effectiveness of insulin in reducing blood glucose levels.

Alopecia totalis
Loss of all scalp hair.

Premature arteriosclerosis
Arteriosclerosis occurring at an age that is younger than usual.

Ankylosis
A reduction of joint mobility resulting from changes involving the articular surfaces.

Pubertal developmental failure in females

Osteopenia
Osteopenia is a term to define bone density that is not normal but also not as low as osteoporosis. By definition from the World Health Organization osteopenia is defined by bone densitometry as a T score -1 to -2.5.

Glomerulopathy
Inflammatory or noninflammatory diseases affecting the glomeruli of the nephron.

Hydropic placenta
An abnormality of the placenta in which there are numerous cystic spaces within the placenta as well as placental enlargement.

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

Distal muscle weakness
Reduced strength of the musculature of the distal extremities.

Basal cell carcinoma
The presence of a basal cell carcinoma of the skin.

Ventricular arrhythmia

Waddling gait
Weakness of the hip girdle and upper thigh muscles, for instance in myopathies, leads to an instability of the pelvis on standing and walking. If the muscles extending the hip joint are affected, the posture in that joint becomes flexed and lumbar lordosis increases. The patients usually have difficulties standing up from a sitting position. Due to weakness in the gluteus medius muscle, the hip on the side of the swinging leg drops with each step (referred to as Trendelenburg sign). The gait appears waddling. The patients frequently attempt to counteract the dropping of the hip on the swinging side by bending the trunk towards the side which is in the stance phase (in the German language literature this is referred to as Duchenne sign). Similar gait patterns can be caused by orthopedic conditions when the origin and the insertion site of the gluteus medius muscle are closer to each other than normal, for instance due to a posttraumatic elevation of the trochanter or pseudarthrosis of the femoral neck.

Upper limb muscle weakness
Weakness of the muscles of the arms.

Abnormality of complement system
An abnormality of the complement system.

Limb muscle weakness
Reduced strength and weakness of the muscles of the arms and legs.

Polyhydramnios
The presence of excess amniotic fluid in the uterus during pregnancy.

Increased anterioposterior diameter of thorax

Patchy hypo- and hyperpigmentation

Accelerated atherosclerosis
Atherosclerosis which occurs in a person with certain risk factors (e.g., SLE, diabetes, smoking, hypertension, hypercholesterolaemia, family history of early heart disease) at an earlier age than would occur in another person without those risk factors.

Axonal degeneration/regeneration
A pattern of simultaneous degeneration and regeneration of axons (see comment).

Osteoarthritis
Degeneration (wear and tear) of articular cartilage, i.e., of the joint surface. Joint degeneration may be accompanied by osteophytes (bone overgrowth), narrowing of the joint space, regions of sclerosis at the joint surface, or joint deformity.

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.

Eclampsia
An acute and life-threatening complication of pregnancy, which is characterized by the appearance of tonic-clonic seizures, usually in a patient who had developed pre-eclampsia. Eclampsia includes seizures and coma that happen during pregnancy but are not due to preexisting or organic brain disorders.

Narrow nasal tip
Decrease in width of the nasal tip.

Transient ischemic attack

Hypoplastic male external genitalia
Underdevelopment of part or all of the male external reproductive organs (which include the penis, the scrotum and the urethra).

Delayed menarche
First period after the age of 15 years.

Enlarged peripheral nerve
Increase in size of a peripheral nerve. This finding can be appreciated by palpation along the axis of the nerve.

Decreased fetal movement
An abnormal reduction in quantity or strength of fetal movements.

Distal sensory impairment
An abnormal reduction in sensation in the distal portions of the extremities.

Increased LDL cholesterol concentration
An elevated concentration of low-density lipoprotein cholesterol in the blood.

Sclerosis of hand bone
Osteosclerosis affecting one or more bones of the hand.

Absent muscle fiber emerin
Immunohistochemistry shows complete lack of emerin protein in the muscle biopsy.

Short clavicles
Reduced length of the clavicles.

Adipose tissue loss
A loss of adipose tissue.

Skin erosion
A discontinuity of the skin exhibiting incomplete loss of the epidermis, a lesion that is moist, circumscribed, and usually depressed.

Proximal muscle weakness
A lack of strength of the proximal muscles.

Developmental cataract
A cataract that occurs congenitally as the result of a developmental defect, in contrast to the majority of cataracts that occur in adulthood as the result of degenerative changes of the lens.

Distal amyotrophy
Muscular atrophy affecting muscles in the distal portions of the extremities.

Decreased adiponectin level
A reduced circulating concentration of adiponectin, a 30-kDa complement C1-related protein that is the most abundant secreted protein expressed in adipose tissue.

Short nail
Decreased length of nail.

Proximal muscle weakness in lower limbs
A lack of strength of the proximal muscles of the legs.

Severe muscular hypotonia
A severe degree of muscular hypotonia characterized by markedly reduced muscle tone.

Osteoporosis
Osteoporosis is a systemic skeletal disease characterized by low bone density and microarchitectural deterioration of bone tissue with a consequent increase in bone fragility. According to the WHO criteria, osteoporosis is defined as a BMD that lies 2.5 standard deviations or more below the average value for young

healthy adults (a T-score below -2.5 SD).

Hirsutism
Abnormally increased hair growth referring to a male pattern of body hair (androgenic hair).

Dermal translucency
An abnormally increased ability of the skin to permit light to pass through (translucency) such that subcutaneous structures such as veins display an increased degree of visibility.

Hyperpigmentation of the skin
A darkening of the skin related to an increase in melanin production and deposition.

Aplasia/Hypoplasia of the skin

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

Telecanthus
Distance between the inner canthi more than two standard deviations above the mean (objective); or, apparently increased distance between the inner canthi.

Pili torti
Pili (from Latin pilus, hair) torti (from Latin tortus, twisted) refers to short and brittle hairs that appear flattened and twisted when viewed through a microscope.

Myalgia
Pain in muscle.

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

Heterogeneous

Corneal opacity
A reduction of corneal clarity.

Lipodystrophy
Degenerative changes of the fat tissue.

Increased adipose tissue around the neck
An increased amount of subcutaneous fat tissue around the neck.

Absent eyelashes
Lack of eyelashes.

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

("excavatum") appearance.

Ventricular escape rhythm
A ventircular escape rhythm occurs whenever higher-lever pacemakers in AV junction or sinus node fail to control ventricular activation. Escape rate is usually 20-40 bpm, often associated with broad QRS complexes (at least 120 ms).

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

Advanced eruption of teeth
Premature tooth eruption, which can be defined as tooth eruption more than 2 SD earlier than the mean eruption age.

Flexion contracture
A flexion contracture is a bent (flexed) joint that cannot be straightened actively or passively. It is thus a chronic loss of joint motion due to structural changes in muscle, tendons, ligaments, or skin that prevents normal movement of joints.

Scaling skin
Refers to the loss of the outer layer of the epidermis in large, scale-like flakes.

Arthralgia
Joint pain.

Sprengel anomaly
A congenital skeletal deformity characterized by the elevation of one scapula (thus, one scapula is located superior to the other).

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.

Overtubulated long bones
Overconstriction, or narrowness of the diaphysis and metaphysis of long bones.

Impaired glucose tolerance
An abnormal resistance to glucose, i.e., a reduction in the ability to maintain glucose levels in the blood stream within normal limits following oral or intravenous administration of glucose.

Precocious atherosclerosis

Transposition of the great arteries
A complex congenital heart defect in which the aorta arises from the morphologic right ventricle and the pulmonary artery arises from the morphologic left ventricle.

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

Severe failure to thrive

Abnormality of the musculature
Abnormality originating in one or more muscles, i.e., of the set of muscles of body.

Hypercholesterolemia
An increased concentration of cholesterol in the blood.

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

Left ventricular systolic dysfunction
Abnormality of left ventricular contraction, often defined operationally as an ejection fraction of less than 40 percent.

Steppage gait
An abnormal gait pattern that arises from weakness of the pretibial and peroneal muscles due to a lower motor neuron lesion. Affected patients have footdrop and are unable to dorsiflex and evert the foot. The leg is lifted high on walking so that the toes clear the ground, and there may be a slapping noise when the foot strikes the ground again.

Areflexia
Absence of neurologic reflexes such as the knee-jerk reaction.

Pancreatitis
The presence of inflammation in the pancreas.

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

Epidermal hyperkeratosis

Rocker bottom foot
The presence of both a prominent heel and a convex contour of the sole.

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.

Abnormal cellular phenotype
An anomaly of cellular morphology or physiology.

Sparse hair
Reduced density of hairs.

Decreased HDL cholesterol concentration
An decreased concentration of high-density lipoprotein cholesterol in the blood.

Limited shoulder movement
A limitation of the range of movement of the shoulder joint.

Congenital adrenal hypoplasia
A type of adrenal hypoplasia with congenital onset.

Hyperinsulinemia
An increased concentration of insulin in the blood.

Prematurely aged appearance

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

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

Short umbilical cord
Decreased length of the umbilical cord.

Breast aplasia
Failure to develop and congenital absence of the breast.

Acanthosis nigricans
A dermatosis characterized by thickened, hyperpigmented plaques, typically on the intertriginous surfaces and neck.

Decreased number of peripheral myelinated nerve fibers
A loss of myelinated nerve fibers in the peripheral nervous system (in general, this finding can be observed on nerve biopsy).

Left ventricular diastolic dysfunction
Abnormal function of the left ventricule during left ventricular relaxation and filling.

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.

Abnormality of circulating leptin level
An abnormal concentration of leptin in the blood.

Muscle weakness
Reduced strength of muscles.

Mitral stenosis
An abnormal narrowing of the orifice of the mitral valve.

Intervertebral disc degeneration
The presence of degenerative changes of intervertebral disk.

Abnormal thorax morphology
Any abnormality of the thorax (the region of the body formed by the sternum, the thoracic vertebrae and the ribs).

Retinal degeneration
A nonspecific term denoting degeneration of the retinal pigment epithelium and/or retinal photoreceptor cells.

Atrial fibrillation
An atrial arrhythmia characterized by disorganized atrial activity without discrete P waves on the surface EKG, but instead by an undulating baseline or more sharply circumscribed atrial deflections of varying amplitude an frequency ranging from 350 to 600 per minute.

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.

Generalized hyperkeratosis

Aplasia of the phalanges of the 3rd toe

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

Short distal phalanx of finger
Short distance from the end of the finger to the most distal interphalangeal crease or the distal interphalangeal joint flexion point. That is, hypoplasia of one or more of the distal phalanx of finger.

Minimal subcutaneous fat

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

Structural foot deformity
A foot deformity resulting due to an abnormality affecting the bones of the foot (as well as muscle and soft tissue). In contrast if only the muscle and soft tissue are affected the term positional foot deformity applies.

Exercise-induced myalgia
The occurrence of an unusually high amount of muscle pain following exercise.

Third degree atrioventricular block
Third-degree atrioventricular (AV) block (also referred to as complete heart block) is the complete dissociation of the atria and the ventricles. Third-degree AV block exists when more P waves than QRS complexes exist and no relationship (no conduction) exists between them.

Abnormality of the intrahepatic bile duct
An abnormality of the intrahepatic bile duct.

Premature loss of teeth
Exfoliation of a tooth more than 2 SD earlier than the normal age for the deciduous teeth and not related to traume or neglect. Exfoliation of a permanent tooth is per se abnormal.

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

Loss of subcutaneous adipose tissue in limbs
Loss (disappearance) of previously present subcutaneous fat tissue in arm or leg.

Type 1 muscle fiber atrophy
Atrophy (wasting) affecting primary type 1 muscle fibers. This feature in general can only be observed on muscle biopsy.

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.

Cyanosis
Bluish discoloration of the skin and mucosa due to poor circulation or inadequate oxygenation of arterial or capillary blood.

Ichthyosis
An abnormality of the skin characterized the presence of excessive amounts of dry surface scales on the skin resulting from an abnormality of keratinization.

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).

Thin ribs
Ribs with a reduced diameter.

Lack of skin elasticity

Shallow orbits

Reduced depth of the orbits associated with prominent-appearing ocular globes.

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.

Prominent umbilicus
Abnormally prominent umbilicus (belly button).

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

Osteolytic defects of the phalanges of the hand
Dissolution or degeneration of bone tissue of the phalanges of the hand.

Loss of truncal subcutaneous adipose tissue
Loss (reduction of previously present) of subcutaneous adipose tissue in the region of the trunk.

Abnormal eyebrow morphology
An abnormality of the eyebrow.

Conductive hearing impairment
An abnormality of vibrational conductance of sound to the inner ear leading to impairment of sensory perception of sound.

Sparse body hair
Sparseness of the body hair.

Cellulitis
A bacterial infection and inflammation of the skin und subcutaneous tissues.

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

Carotid artery occlusion
Complete obstruction of a carotid artery.

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.

Full cheeks
Increased prominence or roundness of soft tissues between zygomata and mandible.

Lipoatrophy
Localized loss of fat tissue.

Supraventricular arrhythmia
A type of arrhythmia that originates above the ventricles, whereby the electrical impulse propagates down the normal His Purkinje system similar to normal sinus rhythm.

Dental crowding
Changes in alignment of teeth in the dental arch

Absent eyebrow
Absence of the eyebrow.

White forelock
A triangular depigmented region of white hairs located in the anterior midline of the scalp.

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

Calcinosis
Formation of calcium deposits in any soft tissue.

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.

Congenital pseudoarthrosis of the clavicle
The two portions of the clavicle (corresponding to the two primary ossification centers of the clavicle) are connected by a fibrous bridge that is contiguous with the periosteum, and a synovial membrane develops, resulting in a clavicle with a bipartite appearance radiographically. Congenital pseudarthrosis of the clavicle generally presents as a painless mass or swelling over the clavicle.

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.

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.

Aplasia/Hypoplastia of the eccrine sweat glands
Absence or developmental hypoplasia of the eccrine sweat glands.

Hyperglycemia
An increased concentration of glucose in the blood.

Large fontanelles
In newborns, the two frontal bones, two parietal bones, and one occipital bone are joined by fibrous sutures, which form a small posterior fontanelle, and a larger, diamond-shaped anterior fontanelle. These regions allow for the skull to pass the birth canal and for later growth. The fontanelles gradually ossify, whereby the posterior fontanelle usually closes by eight weeks and the anterior fontanelle by the 9th to 16th month of age.

Large fontanelles are diagnosed if the fontanelles are larger than age-dependent norms.

Scapular winging
Abnormal protrusion of the scapula away from the surface of the back.

Avascular necrosis
A disease where there is cellular death (necrosis) of bone components due to interruption of the blood supply.

Secondary amenorrhea

Cardiac conduction abnormality
Any anomaly of the progression of electrical impulses through the heart.

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

Abnormal hair whorl
An abnormal hair whorl (that is, a patch of hair growing in the opposite direction of the rest of the hair).

Entropion
An abnormal inversion (turning inward) of the eyelid (usually the lower) towards the globe. Entropion is usually acquired as a result of involutional or cicatricial processes but may occasionally be congenital.

Webbed neck
Pterygium colli is a congenital skin fold that runs along the sides of the neck down to the shoulders. It involves an ectopic fibrotic facial band superficial to the trapezius muscle. Excess hair-bearing skin is also present and extends down the cervical region well beyond the normal hairline.

Subcutaneous calcification
Deposition of calcium salts in subcutaneous tissue (i.e., the the lowermost layer of the integument).

Limb-girdle muscular dystrophy
Muscular dystrophy affecting the muscles of the limb girdle (the hips and shoulders).

Hypodontia
The absence of five or less teeth from the normal series by a failure to develop.

Axial muscle weakness
Reduced strength of the axial musculature (i.e., of the muscles of the head and neck, spine, and ribs).

Prominent superficial blood vessels

Dystrophic fingernails
The presence of misshapen or partially destroyed nail plates, often with accumulation of soft, yellow keratin between the dystrophic nail plate and nail bed, resulting in elevation of the nail plate.

Temporomandibular joint ankylosis
Bony fusion of the mandibular condyle to the base of the skull, resulting in limitation of jaw opening.

Weight loss
Reduction of total body weight.

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

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.

Abnormality of the dentition
Any abnormality of the teeth.

Limb-girdle muscle weakness
Weakness of the limb-girdle muscles (also known as the pelvic and shoulder girdles), that is, lack of strength of the muscles around the shoulders and the pelvis.

Multiple joint contractures

Narrow face
Bizygomatic (upper face) and bigonial (lower face) width are both more than 2 standard deviations below the mean (objective); or, an apparent reduction in the width of the upper and lower face (subjective).

Impacted tooth
A tooth that has not erupted because of local impediments (overcrowding or fibrous gum overgrowth).

Dermal atrophy
Partial or complete wasting (atrophy) of the skin.

Abnormal mitral valve morphology
Any structural anomaly of the mitral valve.

Prominent ear helix
Abnormally prominent ear helix.

Spinal rigidity
Reduced ability to move the vertebral column with a resulting limitation of neck and trunk flexion.

Aortic atherosclerotic lesion
The presence of atheromas or atherosclerotic plaques in the aorta.

Stiff skin
An induration (hardening) of the skin

Narrow chest
Reduced width of the chest from side to side, associated with a reduced distance from the sternal notch to the tip of the shoulder.

Ureteral duplication
A developmental anomaly characterized by the presence of two, instead of one, ureter connecting a kidney to the bladder.

Abnormal hair morphology
An abnormality of the hair.

Squamous cell carcinoma of the skin
Squamous cell carcinoma of the skin is a malignant tumor of squamous epithelium.

Myotonia
An involuntary and painless delay in the relaxation of skeletal muscle following contraction or electrical stimulation.

Coxa valga
Coxa valga is a deformity of the hip in which the angle between the femoral shaft and the femoral neck is increased compared to age-adjusted values (about 150 degrees in newborns gradually reducing to 120-130 degrees in adults).

Calf muscle hypertrophy
Muscle hypertrophy affecting the calf muscles.

Heart block
Impaired conduction of cardiac impulse occurring anywhere along the conduction pathway.

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.

Proximal lower limb amyotrophy
Muscular atrophy affecting proximally located muscles of the legs, i.e., of the thigh.

Polycystic ovaries

Osteolytic defects of the distal phalanges of the hand

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.

Hypertriglyceridemia
An abnormal increase in the level of triglycerides in the blood.

Abnormal atrioventricular conduction
An impairment of the electrical continuity between the atria and ventricles.

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

Hyporeflexia
Reduction of neurologic reflexes such as the knee-jerk reaction.

Neoplasm of the small intestine
The presence of a neoplasm of the small intestine.

Sinus bradycardia
Bradycardia related to a mean resting sinus rate of less than 50 beats per minute.

Mitral valve calcification
Abnormal calcification of the mitral valve.

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

Foot dorsiflexor weakness
Weakness of the muscles responsible for dorsiflexion of the foot, that is, of the movement of the toes towards the shin. The foot dorsiflexors include the tibialis anterior, the extensor hallucis longus, the extensor digitorum longus, and the peroneus tertius muscles.

Hyperlordosis
Abnormally increased cuvature (anterior concavity) of the lumbar or cervical spine.

Wormian bones
The presence of extra bones within a cranial suture. Wormian bones are irregular isolated bones which appear in addition to the usual centers of ossification of the cranium.

Pulmonary hypoplasia

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

Peroneal muscle atrophy
Atrophy of the peroneous muscles, peroneus longus (also known as Fibularis longus), Peroneus brevis (also known as fibularis brevis, and Peroneus tertius (also known as fibularis tertius).

Prominent superficial veins
A condition in which superficial veins (i.e., veins just under the skin) are more conspicuous or noticable than normal.

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.

Sparse scalp hair
Decreased number of hairs per unit area of skin of the scalp.

Down-sloping shoulders
Low set, steeply sloping shoulders.

Osteosarcoma
A malignant bone tumor that usually develops during adolescence and usually affects the long bones including the tibia, femur, and humerus. The typical symptoms of osteosarcoma comprise bone pain, fracture, limitation of motion, and tenderness or swelling at the site of the tumor.

Decreased serum leptin
A decreased concentration of leptin in the blood.

Natal tooth
A tooth present at birth or erupting within the first month of life.

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

Micrognathia
Developmental hypoplasia of the mandible.

Pulmonary carcinoid tumor
A malignant neuroendocrine tumor of the lung. According to histopathologic criteria (WHO 2004), carcinoids are divided into four groups i.e. typical and atypical carcinoids, large cell neuroendocrine carcinoma and small cell lung carcinoma.

Autosomal recessive inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in individuals with two pathogenic alleles, either homozygotes (two copies of the same mutant allele) or compound heterozygotes (whereby each copy of a gene has a distinct mutant allele).

Acute pancreatitis
A acute form of pancreatitis.

Sinoatrial block
Disturbance in the atrial activation that is caused by transient failure of impulse conduction from the sinoatrial node to the cardiac atria.

Alopecia universalis
Loss of all hair on the entire body.

Kyphosis

Exaggerated anterior convexity of the thoracic vertebral column.

Craniofacial disproportion

Vocal cord paralysis
A loss of the ability to move the vocal folds.

Limitation of movement at ankles
An abnormal limitation of the mobility of the ankle joint.

Aortic valve calcification
Deposition of calcium salts in the aortic valve.

Mottled pigmentation
Patchy and irregular skin pigmentation.

Alopecia
A noncongenital process of hair loss, which may progress to partial or complete baldness.

Atrioventricular block
Delayed or lack of conduction of atrial depolarizations through the atrioventricular node to the ventricles.

Delayed eruption of teeth
Delayed tooth eruption, which can be defined as tooth eruption more than 2 SD beyond the mean eruption age.

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

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

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

Thoracic kyphoscoliosis

Obesity
Accumulation of substantial excess body fat.

Respiratory insufficiency due to muscle weakness

Intracranial hemorrhage
Hemorrhage occurring within the skull.

Convex nasal ridge
Nasal ridge curving anteriorly to an imaginary line that connects the nasal root and tip. The nose appears often

also prominent, and the columella low.

Distal lower limb muscle weakness
Reduced strength of the distal musculature of the legs.

Dilated cardiomyopathy
Dilated cardiomyopathy (DCM) is defined by the presence of left ventricular dilatation and left ventricular systolic dysfunction in the absence of abnormal loading conditions (hypertension, valve disease) or coronary artery disease sufficient to cause global systolic impairment. Right ventricular dilation and dysfunction may be present but are not necessary for the diagnosis.

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

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

Renal neoplasm
The presence of a neoplasm of the kidney.

Large placenta
Increased size of the placenta.

Myocardial infarction
Necrosis of the myocardium caused by an obstruction of the blood supply to the heart and often associated with chest pain, shortness of breath, palpitations, and anxiety as well as characteristic EKG findings and elevation of serum markers including creatine kinase-MB fraction and troponin.

Decreased skull ossification
A reduction in the magnitude or amount of ossification of the skull.

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

Proximal upper limb amyotrophy
Muscular atrophy affecting proximally located muscles of the arms.

Small placenta
Reduced size of the placenta.

Abnormality of retinal pigmentation

Osteolysis
Osteolysis refers to the destruction of bone through bone resorption with removal or loss of calcium.

Aplasia/Hypoplasia of the clavicles
Absence or underdevelopment of the clavicles (collar bones).

Decreased fertility

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

Fragile nails
Nails that easily break.

Labial pseudohypertrophy

Respiratory insufficiency

Acroosteolysis of distal phalanges (feet)

Elevated circulating creatine kinase concentration
An elevation of the level of the enzyme creatine kinase (also known as creatine phosphokinase, CPK; EC 2.7.3.2) in the blood. CPK levels can be elevated in a number of clinical disorders such as myocardial infarction, rhabdomyolysis, and muscular dystrophy.

Achilles tendon contracture
A contracture of the Achilles tendon.

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

Corneal ulceration
Disruption of the epithelial layer of the cornea with involvement of the underlying stroma.

Premature 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 before age of 45.

Premature ovarian insufficiency
Amenorrhea due to loss of ovarian function before the age of 40. Primary ovarian inssuficiency (POI) is a state of female hypergonadotropic hypogonadism. It can manifest as primary amenorrhea with onset before menarche or secondary amenorrhea.

Abnormality of the pinna
An abnormality of the pinna, which is also referred to as the auricle or external ear.

Toe walking

Atrial flutter

A type of atrial arrhythmia characterized by atrial rates of between 240 and 400 beats per minute and some degree of atrioventricular node conduction block. Typically, the ventricular rate is half the atrial rate. In the EKG; atrial flutter waves are observed as sawtooth-like atrial activity. Pathophysiologically, atrial flutter is a form of atrial reentry in which there is a premature electrical impulse creates a self-propagating circuit.

Abnormal aortic valve morphology
Any abnormality of the aortic valve.

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

Ascending tubular aorta aneurysm
An abnormal localized widening (dilatation) of the tubular part of the ascending aorta.

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).

First degree atrioventricular block
Delay of conduction through the atrioventricular node, which is manifested as prolongation of the PR interval in the electrocardiogram (EKG). All atrial impulses reach the ventricles.

High pitched voice
An abnormal increase in the pitch (frequency) of the voice.

Glycosuria
An increased concentration of glucose in the urine.

Onion bulb formation
Repeated episodes of segmental demyelination and remyelination lead to the accumulation of supernumerary Schwann cells around axons, which is referred to as onion bulb formation. This finding affects peripheral nerves.

Persistence of primary teeth
Persistence of the primary teeth beyond the age by which they normally are shed and replaced by the permanent teeth.

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).

Thin skin
Reduction in thickness of the skin, generally associated with a loss of suppleness and elasticity of the skin.

Fasting hyperinsulinemia
An increased concentration of insulin in the blood in the fasting state, i.e., not as the response to food intake.

Generalized amyotrophy
Generalized (diffuse, unlocalized) amyotrophy (muscle atrophy) affecting multiple muscles.

Abnormal hair quantity
An abnormal amount of hair.

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

Low-frequency sensorineural hearing impairment
A form of sensorineural hearing impairment that affects primarily the lower frequencies.

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

Hypermelanotic macule
A hyperpigmented circumscribed area of change in normal skin color without elevation or depression of any size.

Limited hip movement
A decreased ability to move the femur at the hip joint associated with a decreased range of motion of the hip.

Poor head control
Difficulty to maintain correct position of the head while standing or sitting.

Premature rupture of membranes
Premature rupture of membranes (PROM) is a condition which occurs in pregnancy when the amniotic sac ruptures more than an hour before the onset of labor.

Aplasia/Hypoplasia involving the nose
Underdevelopment or absence of the nose or parts thereof.

Syncope
Syncope refers to a generalized weakness of muscles with loss of postural tone, inability to stand upright, and loss of consciousness. Once the patient is in a horizontal position, blood flow to the brain is no longer hindered by gravitation and consciousness is regained. Unconsciousness usually lasts for seconds to minutes. Headache and drowsiness (which usually follow seizures) do not follow a syncopal attack. Syncope results from a sudden impairment of brain metabolism usually due to a reduction in cerebral blood flow.

Retrognathia
An abnormality in which the mandible is mislocalised posteriorly.

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

Abnormality of the voice

Xanthomatosis

The presence of multiple xanthomas (xanthomata) in the skin. Xanthomas are yellowish, firm, lipid-laden nodules in the skin.

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

Upper airway obstruction
Increased resistance to the passage of air in the upper airway.

Ectopic calcification
Deposition of calcium salts in a tissue or location in which calcification does not normally occur.

Mildly elevated creatine kinase

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

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 electrophysiology of sinoatrial node origin
An abnormality of the sinoatrial (SA) node in the right atrium. THe SA node acts as the pacemaker of the heart.

Progressive clavicular acroosteolysis
Progressive bone resorption in the distal part of the clavicle.

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

Increased bone mineral density
An abnormal increase of bone mineral density, that is, of the amount of matter per cubic centimeter of bones which is often referred to as osteosclerosis. Osteosclerosis can be detected on radiological examination as an increased whiteness (density) of affected bones.

Hepatomegaly
Abnormally increased size of the liver.

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

Abnormality of the pulmonary artery
An abnormality of the pulmonary artery.

Abnormal myocardium morphology
A structural anomaly of the muscle layer of the heart wall.

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

Ankyloglossia
Short or anteriorly attached lingual frenulum, associated with limited mobility of the tongue.

Insulin-resistant diabetes mellitus
A type of diabetes mellitus related not to lack of insulin but rather to lack of response to insulin on the part of the target tissues of insulin such as muscle, fat, and liver cells. This type of diabetes is typically associated with increases both in blood glucose concentrations as will as in fasting and postprandial serum insulin levels.

Cranial nerve paralysis

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

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.

Limited wrist movement
An abnormal limitation of the mobility of the wrist.

Decreased body weight
Abnormally low body weight.

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.

Neoplasm of the lung
Tumor of the lung.

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

Muscular dystrophy
The term dystrophy means abnormal growth. However, muscular dystrophy is used to describe primary myopathies with a genetic basis and a progressive course characterized by progressive skeletal muscle weakness and wasting, defects in muscle proteins, and histological features of muscle fiber degeneration (necrosis) and regeneration. If possible, it is preferred to use other HPO terms to describe the precise

phenotypic abnormalities.

Supraventricular tachycardia
Supraventricular tachycardia (SVT) is an abnormally increased heart rate (over 100 beats per minute at rest) with origin above the level of the ventricles.

Premature birth
The birth of a baby of less than 37 weeks of gestational age.

Hyperlipidemia
An elevated lipid concentration in the blood.

Sparse or absent eyelashes

Hepatic steatosis
Steatosis is a term used to denote lipid accumulation within hepatocytes.

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

Emphysema

Atherosclerosis
A condition characterized by patchy atheromas or atherosclerotic plaques which develop in the walls of medium-sized and large arteries and can lead to arterial stenosis with reduced or blocked blood flow.

Premature skin wrinkling
The presence of an increased degree of wrinkling (irregular folds and indentations) of the skin as compared with age-related norms.

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.

Arthrogryposis multiplex congenita
Multiple congenital contractures in different body areas.

Relative macrocephaly
A relatively mild degree of macrocephaly in which the head circumference is not above two standard deviations from the mean, but appears disproportionately large when other factors such as body stature are taken into account.

Adrenal hypoplasia
Developmental hypoplasia of the adrenal glands.

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

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.

Sparse eyelashes
Decreased density/number of eyelashes.

Abnormality of the Achilles tendon
An abnormality of the Achilles tendon.

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.

Hip dysplasia
The presence of developmental dysplasia of the hip.

Pericardial effusion
Accumulation of fluid within the pericardium.

Short palm
Short palm.

Proximal muscle weakness in upper limbs
A lack of strength of the proximal muscles of the arms.

Hyporeflexia of lower limbs
Reduced intensity of muscle tendon reflexes in the lower limbs. Reflexes are elicited by stretching the tendon of a muscle, e.g., by tapping.

Narrow nasal ridge
Decreased width of the nasal ridge.

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.

Hypertrophic cardiomyopathy
Hypertrophic cardiomyopathy (HCM) is defined by the presence of increased ventricular wall thickness or mass in the absence of loading conditions (hypertension, valve disease) sufficient to cause the observed

abnormality.

Abnormality of neutrophils
A neutrophil abnormality.

Shuffling gait
A type of gait (walking) characterized by by dragging one's feet along or without lifting the feet fully from the ground.

Abnormality of the vasculature
An abnormality of the vasculature.

Papillary renal cell carcinoma
The presence of renal cell carcinoma in the renal papilla.

Decreased cervical spine flexion due to contractures of posterior cervical muscles

Elbow flexion contracture
A chronic loss of elbow joint motion due to structural changes in muscle, tendons, ligaments, or skin that prevent normal movement of the joints of the elbow.

EMG: myopathic abnormalities
The presence of abnormal electromyographic patterns indicative of myopathy, such as small-short polyphasic motor unit potentials.

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

Abnormal lymphocyte physiology
Any anomaly of lymphocyte function.

Arrhythmia
Any cardiac rhythm other than the normal sinus rhythm. Such a rhythm may be either of sinus or ectopic origin and either regular or irregular. An arrhythmia may be due to a disturbance in impulse formation or conduction or both.

Thin clavicles
Abnormally reduced diameter (cross section) of the clavicles.

Wide nasal bridge
Increased breadth of the nasal bridge (and with it, the nasal root).

High-frequency sensorineural hearing impairment
A form of sensorineural hearing impairment that affects primarily the higher frequencies.

Raynaud phenomenon

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

Absence of subcutaneous fat
Lack of subcutaneous adipose tissue.

Dextrocardia
The heart is located in the right hand sided hemithorax. That is, there is a left-right reversal (or "mirror reflection") of the anatomical location of the heart in which the heart is locate on the right side instead of the left.

Dysmenorrhea
Pain during menstruation that interferes with daily activities.

Sparse eyebrow
Decreased density/number of eyebrow hairs.

Progeroid facial appearance
A degree of wrinkling of the facial skin that is more than expected for the age of the individual, leading to a prematurely aged appearance.

Premature delivery because of cervical insufficiency or membrane fragility

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

Aplasia of the middle phalanx of the hand
Absence of one or more middle phalanx of a finger.

Kyphoscoliosis
An abnormal curvature of the spine in both a coronal (lateral) and sagittal (back-to-front) plane.

Nocturnal lagophthalmos
The inability to close the eyelids during sleep.

Finger clinodactyly

Generalized osteoporosis