

A

Alkaline Phosphate: An enzyme made by the liver or bone indicating liver problem, Vitamin K deficiency

Allele: Any one of the alternative forms of a specified gene. Different alleles usually have different effects on the phenotype

Alopecia Areata: A condition characterized by irregular areas of baldness. Slow enlargement but eventual re-growth within one year is common, but relapse is common and occasionally total loss of hair may follow, especially in children

Amblyopia: When one eye develops proper sight, and the other eye does not develop proper sight. Amblyopia can develop into loss of vision if not treated. Also referred to as lazy eye.

Angioplasty: Surgical repair of diseased blood vessels. See also Balloon Angioplasty

Anterior Uveitis: Inflammation of the front layer of the pigmented membrane in the eye

Apnea: Temporary halting of the breathing

Astigmatism: Poorly focused vision due to a misshapen eye, the eye is oval in shape instead of round.

Atrial Septal Defect: A hole between the two upper chambers of the heart

Autism: Behavioral disorders related to Autism. Disturbed processing of information;

Disturbed social functioning; Disturbed communication and use of language;

Disturbed

Incapability for imagination and fantasy;

Abnormally limited area of interest and activities;

Disturbed emotional development;

Unbalanced development profile;

Unbalanced reactions to unexpected situations;

Unbalanced motor development;

Medical aspects: Inability to communicate physical illness

Sometimes: Epilepsy See also: PDD-NOS,

IFA

Autoimmune Thyroiditis: Autoimmune reaction attacks the thyroid gland

Autosomal Dominant: Inheritance

Inheritance whereby a gene, if present, always manifests itself phenotypically, e.g., if a gene for NS is passed on to a child by either parent, then the child will have NS. The gender of the child makes no difference, and there is no carrier state.

B

Balloon Angioplasty : A less invasive surgical repair of diseased blood vessels which opens the collapsed vessel with a balloon on the end of a catheter

Behavioral: Short attention span; Considerate. Easily confused; Imaginative. Squirmy/Fidgety; Affectionate; Overly Sensitive/Insecure; Sociable. Overactive/Underactive; Attentive. Obsessive behavior/Stubborn; Affable.

Bleeding Diathesis: A variety of defects in the coagulation and platelet systems including abnormalities in the intrinsic pathway - partial factor XI: C, XII: C and VIII:c

Bruises: Bruises easily

BT: Bleeding time, a test that determines if the blood appears to clot within a normal time.

C

CBC: Complete blood count, test that helps to confirm any abnormalities in the number of red cells, white cells and platelets.

Combined Coagulation Deficiencies: See Bleeding Diathesis

Café-au-lait Spots: Coffee colored spots on the skin

Cardio: Referring to the heart

Cervical spine fusion: Short neck, fused with spine

CFC Syndrome: Cardio-facio-cutaneous Syndrome; with characteristic symptoms in heart, facial features and skin

Cherubism: Cheekbone deformity causing lowering of the eyelid

Chromosomes: Referring to the genetic make-up. There are 23 pairs of chromosomes, which are made up of smaller cells referred to as genes.

Chyle: is the "fatty" portion of the lymph fluid which gives the fluid its milky appearance

Chylothorax: An accumulation of lymph (milky) fluid in the pleural space (between the lungs and the ribs), usually caused by a rupture or obstruction of thoracic duct with normally carries lymph through the chest area.

Clinobrachyactyly: Shortening and banding of a finger.

Collumella: The central axis of the nose.

Coarctation of the aorta: An abnormality, present from birth, in which there is a localized narrowing of the aorta (the large artery that supplies blood from the left side of the heart to the rest of the body).

Cryptorchidism: Undescended testicles

Cubitus Valgus: Elbow Deformity

Cutaneous Lymphangioma: Wart-like growth on the skin, very small, made up of abnormal lymph vessels

Cystic Hygroma: Cystic swelling of the neck

D

Delayed puberty: See also: Hypogonadism

Diastasis Recti: Wide distance between the straight belly-muscles

Distended abdomen: "Balloon Belly"

Dyspraxia: Inability to perform coordinated motor actions

E

Ears: Low set, rotated ears, incomplete folding of ears, thick helix/ outer rim

Eating: Faddy eater; Forceful vomiting; Swallowing difficulties; Frequent choking. See also: Failure to thrive.

Edema: Accumulation of fluid in the tissues

Epicanthal Fold: An extra fold of skin at the inner corner of the eye

Epistaxis: Nosebleeds

Esophagitis / Oesphagitis: Reddening and swelling of the Esophagus.

Esophagus: Gullet

F

Failure to thrive: Poor weight gain, malnutrition usually referring to infancy stage of life. Considered under the 5% percentile for weight at that given age.

Familial: A term applied to a characteristic or disorder that runs in families.

Feeding Tubes: J-Tube; G-Tube; NG-Tube

Fetal Lymphedema / Lymphoedema: Swelling due to lymph flow obstruction

Fetal Ultrasound Discoveries: Cervical hygroma, Posterior Cervical Hygroma, Retarded bone age, Webbed neck, etc.

Fundoplication: Surgery to tighten the Lower Esophageal Sphincter. The top of the stomach is wrapped around the base of the Esophagus. See also: GER, Heartburn, LES, Milk Scan, Esophagitis, Reflux, and Scintigraphy

G

Genotype: The particular alleles at specified loci present in an individual; the genetic constitution
G:ER: See: Reflux
Glue ear: Thick glue-like fluid behind eardrum can occur after recurrent ear infections
Growth Hormone: A body chemical that helps in promoting physical growth.
G-Tube: Feeding tube that is surgically inserted through the tummy into the stomach (G = Gastro, i.e. of the stomach)

H

Hair: Alopecia Areata, Curly hair; Keratosis Pilaris; Low posterior hairline, excessive hair growth on body parts.
Hands: Bluntly ended fingers; Extra padding on fingers and toes;
HCM: Hypertrophic Cardiomyopathy
Head : Cervical hygroma, Posterior Cervical
Hygroma: Large Head/ Small head; Short neck, Webbed neck, Triangular face shape
Heart Burn: When stomach acid flows into the Esophagus it causes a burning sensation
Heart Murmur: Abnormal heart sounds caused by turbulent blood flow.
Hepatomegaly: Enlarged liver
Hepatosplenomegaly: Enlarged liver and spleen
Heterogenesis: Referring to spontaneous nature of mutation of affected offspring of non-affected parents
Hirsutism: Excessive hair growth on body parts
HFA: High Functioning Autism
Hydrocephaly: Fluid on the brain
Hydrops Fetalis: Accumulation of fluid by the fetus
Hyperkeratosis: Over development of outer skin layer
Hypermasality: Nasal tone of voice referred to as farsightedness.
Hyperparathyroidism: Affects calcium balance in the body
Hypertelorism: Eyes set widely apart, syn. Telecanthus
Hypertrophic: "very thick" (Hyper=very; trophic=thick)
Hypertrophic Cardiomyopathy: Thickening of the heart muscles with possible disorganization of the muscle cells; see also **Hypertrophic, Cardilo, and Myopathy:**
Hypogonadism: Underactivity of the gonads (testes or ovaries). Abnormally decreased functioning of the testicles, and retardation of growth or sexual development.
Hypoplastic Nipples: Incomplete development of the

M

Macrocephaly/Megacephaly: A condition in which the head is abnormally large.
Malignant Hyperthermia: Abnormal response to anesthesia causing extreme high temperature
Malignant Schwannoma: Cancerous cells that are intrusive through soft tissue structure. Most often found in the trunk, and limbs.
Malocclusion: Teeth missing from set
Megakaryocytic Thrombocytopenia: A condition of abnormally large cells in the bone marrow, the mothercells of the thrombocytes; Thrombocyte=blood platelet
MCA: Multiple Congenital Abnormalities
Microcephaly: A condition in which the head is abnormally small.
Micrognathia: Small jaw
Milk Scan: See: Scintigraphy
Mitral Incompetence: The Mitral Valve is the delicate tissue that opens and closes allowing blood flow from the right ventricle to the right atrium. Incompetence refers to not proper working of the valve, which may prevent blood from entering the atrium.
MMR: Mild Mental Retardation
Motor Development: Delayed gross and/or fine motor development
Mouth: Small mouth; small lower jaw; deeply grooved philtrum or lip line.
Myopathic: Any affection of the muscles or muscular systems.
Myopia: Near sighted

N

Nasolabial Folds: Folds of skin from the nose to the upper lip
Neonatal: The period of four weeks following birth.
Neurofibromatosis: Soft celled tumors that may become cancerous and spread through the blood, lymph, and spinal fluids. There are 2 different types of NF. NF1 is characterized as classic, and accompanied by Café-au-lait spots and freckling. NF2 is usually restricted to the acoustic nerve and central nervous system.
Nevus: Birthmark in form of sharply defined red patch in skin; mole.

I

Hypotonia: Low muscle tone
Hypospadias: Urethral opening misplaced
Idiopathic: "without known cause"
IHSS: Idiopathic Hypertrophic Subaortic Stenosis
Inguinal Hernia: Protrusion of part of the intestine into the muscles in the groin.

J

Joint contractures: Tightness of joints
Joint hyperextensibility: Looseness of joints
J-Tube: Feeding tube that is surgically inserted into the Jejunum (small intestine) rather than G-tube into the stomach.

K

Karyotype: The appearance, number and arrangement of the chromosomes in the cells of an individual.
Keloids: Excessive buildup of scar tissue on the skin following a cut or surgery
Keratoconus: A conical protrusion of the cornea.
Keratosis Pilaris: Excessive keratinization of the hair follicles
King Syndrome: See: Malignant Hyperthermia
Kyphoscoliosis: A curvature of the thoracic (chest region) spine.

L

Language: Speech delayed; Sign language;
Learning disabilities: See also: PDD-NOS
Lentigines: Numerous pigmented dots, resembling freckles. (singular: lentigo)
LES: Lower Esophageal Sphincter; Ring of muscles where the stomach and Esophagus join.
Lymphedema: Build-up of fluid in the subcutaneous (under the skin) tissue caused by obstruction of lymphatic drainage; visible as swelling.
Lytes: Electrolytes, active chemicals found in body fluids. See SMAC under S.

NG-Tube: Feeding tube that is brought into the stomach through the nose via the gullet can be placed by the parents at home

Nystagmus: Involuntary jerking movement of the eyes.

O

Otitis Media: Ear Infection

Osteoporosis: Fragility of bones

P

Patent Ductus Arteriosus: A condition where a channel between the pulmonary artery fails to close after birth.

Pathogenesis: Origin of disease.

Palate: Roof of the mouth. High arched, Palate and/or False Palate: Roof of the mouth miss-formed, missing, or appearing normal but not having normal structure underneath.

Palpebral Fissures: Fissures of either eyelid: upper or lower.

PDD/PDD-NOS: Pervasive Developmental Delay (-Not Otherwise Specified)

Pectus carinatum: Prominence of breastbone

Pectus excavatum: Depression of breastbone

Phenotype: The observable characteristics of an organism as determined by the interaction of its genotype and its environment.

Philtrum: Prominent grooved upper lip

Pigmented Nevii: Birth marks

Polydactyly: Having extra fingers or toes

Polyhydramnios: Excessive amniotic fluid

Posterior Cervical Hygroma: Webbed neck, extra skin \nuchal skin on the back of the neck

Primary sex organs: Underdeveloped primary sex organs

Prothrombin: A test that may aid in determining if a blood-clotting problem exists.

PT: Prothrombin

PPT: Partial Thromboplastin Time - A test that may aid in determining if a blood-clotting problem exists

Pterygium Colli: See: Webbed neck

Ptosis: Drooping of the eyelids

Pulmonary Valve Stenosis: Narrowing and sometimes malformation of valve that takes the blood from the heart to the lungs for oxygenation

Q

Reflux: Term used to describe a condition in which contents - food and gastric acid - frequently flow back out of the stomach into the Esophagus.

Refractive Errors: Occurs when the cornea and lens of the eye fail to focus light rays onto the retina. (see separate terms: Amblyopia, Astigmatism, Hyperopia, and Myopia.

Retarded bone age: Delayed development of skeleton

Retrognathia: Back-placing of the lower jaw

S

SMAc, SMA-12 SMA-6 Lytes: Various types of blood chemistry test which look for any abnormalities in chemical make up of blood. I.e. sodium, calcium, potassium, etc.

Scapula: - Shoulder blade.

Scintigraphy: A series of scans using a mildly radioactive compound that shows how food moves out of the stomach.

Scoliosis: Sideways curvature of the spine

Shield Chest: See Pectus carinatum

Shortness of stature: Average adult height of 1.50 m, can benefit by Growth Hormone therapy if a GH deficiency exists.

Simian Creases: Deeply grooved palms and bottoms of feet

Sleep Apnea: Episodes of absence of breathing during periods of sleep

Syndrome: Complex of symptoms

Sporadic Mutation: A change in form, quality or some other characteristic without an apparent cause.

Strabismus: Greek for a "squint". An uncontrollable

relaxation of the muscles which may appear as cross-eyes.

Stenosis: "a narrowing"

Subaortic: "below the aortic valve"

Syringomyelia: A progressive disorder characterized by the development of tubular cavities in the spinal cord, which may be due to trauma, tumor, infarction, hemorrhage or without known cause

T

Telecanthus: Eyes set widely apart

Tongue: Poor tongue control

Thrombocytopenia: A condition in which there is an abnormally small number of platelets in the circulating blood.

Trimethylaminuria: A condition where the urine has a

fishy odor due to the large excretion of trimethylamine in the urine

Turriticephaly: Also known as acrocephaly, or oxycephaly- a condition in which the of the head is pointed

Turner Syndrome: Genetic Disorder similar to Noonan but is caused by a missing sex chromosome

U

Umbilical Hernia: An abnormal protrusion of internal abdominal contents through a defect in the 'belly button'.

Uveitis: Inflammation of the front part of the eyes Corneas, etc

V

Vasculitis: A clinical and pathological term for several illnesses caused by inflammation of the blood vessel walls

Ventricular Septal Defect: A hole between the two lower chambers of the heart

Vermilion Border: The red border of the lip

Vitigilo: White patches on skin, lack of pigment cells

Von Willebrand Disease: A disorder of the VIII platelet in which the 'sticky' part of the platelet is missing, and therefore other blood cells cannot stick or clot together.

W

Webbed neck: Fold of skin between the mastoid bone behind the ear and the acromion bone of the shoulder, along which the hair growth from the head is partially continued.

Willebrand: See Von Willebrand disease.

X

Y

Z