InTheKnow Glossary

A

Achondroplasia: A type of skeletal dysplasia (a condition that affects the bones and cartilage). While the most visible effects are in the arms, legs, and face, nearly all of the bones in the body are affected.[1]

Acromelia: The shortening of the extremities affecting primarily the distal parts of the limbs (hands and feet) in relation to the other segments of the limbs.[2]

Adenoids: Small lumps of tissue at the back of the nose, above the roof of the mouth. Adenoids are part of the immune system, which helps fight infection and protects the body from bacteria and viruses.[3]

Amino acids: The building blocks of peptides and proteins.[4]

Apnoea: Temporary cessation of breathing that occurs most often during sleep.[4]

Arthritis: Inflammation of joints due to infectious, metabolic, or constitutional causes.[5]

Audiogram: A special graph that is used by audiologists to represent information collected from a pure-tone hearing test. It can be used to identify the exact type, degree, and configuration of hearing loss (or hearing ability).[6]

Autosomal: Autosomal dominance is a pattern of inheritance characteristic of some genetic diseases. “Autosomal” means that the gene in question is located on one of the numbered, or non-sex, chromosomes.[7]

B

Brachydactyly: Having abnormally short fingers and toes.[4]

C

Cartilage: Cartilage is a tough, flexible tissue found throughout the body. It covers the surface of joints, acting as a shock absorber and allowing bones to slide over one another.[8]

Cell: The basic membrane-bound unit that contains the fundamental molecules of life and of which all living things are composed.[9]

Cervicomedullary junction: The region where the skull meets the spine.[4]

Chondrocyte: A cartilage cell.[4]

Chromosome: A rod-like structure of DNA tightly coiled around histone proteins that carries genetic information in the form of genes; chromosomes are found in the nucleus of cells.[4]

Cleft palate: A congenital fissure of the roof of the mouth.[10]

Clinical geneticist: Clinical geneticists investigate, diagnose, and counsel people who may have a genetic condition, and their families. Clinical genetics encompasses a wide range of conditions, and they provide care for both adults and children.[11]

Clonus: A series of involuntary, rhythmic muscular contractions and relaxations that occur in the form of convulsive spasms in some neurological conditions.[4]

Compartment syndrome: A painful and potentially serious condition caused by bleeding or swelling within an enclosed bundle of muscles.[4]

Computed tomography: A medical imaging technique that produces high-resolution images of the body.[4]

Congenital: At or before birth. Congenital anomalies are also known as birth defects, congenital disorders, or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies that can be identified prenatally, at birth, or sometimes may only be detected later in infancy, such as hearing defects.[12]

Connective tissue: The group of tissues in the body that maintain the form of the body and its organs and provide cohesion and internal support.[13]

Consultant: An experienced doctor with a high position, who specialises in one area of medicine.[14]

Craniectomy: The surgical removal of an area of the skull.[4]

CT scanner: A large device consisting of integrated X-ray and computing equipment that produces CT scans and is used chiefly in medical diagnosis.[15]

C-type natriuretic peptide (CNP): CNP is produced by the endothelium (lining of the blood vessels) and the heart and appears to play a prominent role in vascular and cardiac function, both physiologically and pathologically.[16]

D

Detached retina: A condition of the eye in which the retina has separated from the choroid.[17] Retinal detachment describes an emergency situation in which a thin layer of tissue (the retina) at the back of the eye pulls away from its normal position.[18]

Dominant genes: A dominant gene, or a dominant version of a gene, is a particular variant which expresses itself more strongly than any other version of the gene which the person is carrying.[19]

Dwarfism: A condition (such as achondroplasia) of people marked by unusually small size or short stature.[20]

E

Ear, nose, and throat (ENT) specialist or audiologist: Otorhinolaryngologists (commonly referred to as ENT specialists) deal with the diagnosis, evaluation and management of diseases of head and neck and principally the ears, nose and throat.[21] Audiologists are experts who can help to prevent, diagnose, and treat hearing and balance disorders for people of all ages.[22]

Endochondral ossification: The process by which growing cartilage is systematically replaced by bone to form the growing skeleton.[23]

Endocrinologist: A doctor that treats diseases related to problems with hormones.[24]

Endocytosis: The process by which a cell takes in external material; the plasma membrane of the cell folds inward, fusing to form a vacuole or vesicle containing the engulfed material.[4]

Endoplasmic reticulum: A system of interconnected vesicular and lamellar cytoplasmic membranes that functions especially in the transport of materials within the cell.[4]

F

Fertility: The ability to produce offspring.[25]

FGFR3 (fibroblast growth factor receptor 3): FGFR3 has major roles in endochondral bone formation, active mutations in FGFR3 are the cause of achondroplasia, hypochondroplasia and thanatophoric dysplasia.[26]

Foramen magnum stenosis: Typically, the brain stem and spinal cord pass through the foramen magnum, an opening at the base of the skull. Foramen magnum stenosis happens when the opening narrows, putting pressure on the brain or spinal cord.[27]

G

Gait: A manner of walking or moving on foot.[28]

Genes: A specific sequence of nucleotides in DNA or RNA that is located usually on a chromosome. It is the functional unit of inheritance controlling the transmission and expression of one or more traits. This is done by specifying the structure of a particular polypeptide and especially a protein or controlling the function of other genetic material.[29]

Gene testing: Genetic testing looks for changes, sometimes called mutations or variants, in the DNA.[30]

Genetic counselling: Genetic counselling is the process through which knowledge about the genetic aspects of illnesses is shared by trained professionals with those who are at an increased risk or either having a heritable disorder or of passing it on to their unborn offspring.[31]

Genetic disorders: A disease caused in whole or in part by a change in the DNA sequence away from the normal sequence.[32]

Genome: The complete set of genetic material present in an organism.[4]

Genotype: The genetic makeup, as distinguished from the physical features (phenotype), of a cell or an organism; usually used with reference to a single trait or set of traits.[4]

Genu varum: The medical term for bowleg – a leg bowed outward at or below the knee.[33]

Glue ear: A condition affecting the middle ears of children in which the free movement of the chain of bones linking the eardrum to the inner ear is impeded by sticky mucus.[34]

G380R: The G380R mutation in the transmembrane domain of FGFR3 is a germline mutation responsible for most cases of achondroplasia.[35]

H

Hereditary: Genetically transmitted or transmittable from parent to offspring.[36]

Heterogeneity: The state of consisting of different elements or being variable.[4]

Hydrocephalus: An abnormal increase in the amount of cerebrospinal fluid within the cranial cavity that is accompanied by expansion of the cerebral ventricles and often increased intracranial pressure, skull enlargement, and cognitive decline.[37]

Hypermobile: Describing an increase in the range of movement of which a joint is usually capable.[4]

Hyperreflexia: Overactivity of reflexes.[4]

Hypertension: High blood pressure.[4]

Hypertrophy: The increase in the volume of an organ or tissue due to the enlargement of its component cells.[4]

Hypotonia: Decreased muscle tone.[4]

Hypoxia: A deficiency of oxygen reaching the tissues of the body.[4]

K

Kyphosis: Exaggerated outward curvature of the thoracic (middle) region of the spine.[4]

L

Laminectomy: A type of surgery that removes a portion of the vertebral bone called the lamina.[4]

Leg and tibial bowing: A condition in which a person's legs appear bowed (bent outward).[38]

Ligament: A tough fibrous band of connective tissue that connects bones to other bones.[4]

Lordosis: Exaggerated curvature of the cervical (upper) and lumbar (lower) regions of the spine.[4]

M

Macrocephaly: An abnormally large head with a circumference that is more than two standard deviations larger than the average for a given age and sex.[4]

Malunion: Incomplete or faulty union of fragments of fractured bone.[4]

Medical geneticist: A doctor who specialises in medical conditions that have a genetic influence. They evaluate, diagnose, research, and manage patients with hereditary conditions and/or congenital malformations.[39]

Mesomelia: A shortening of the middle (intermediate) portion of a limb.[40]

Mid-face hypoplasia: Where the upper jaw, cheekbones and eye sockets have not grown as much as the rest of the face.[41]

Mutable: Prone to change/mutation.[4]

N

Neonatal / Neonate: Relating to a new-born infant up to one-month old.[4]

Neurogenic claudication: Walking-induced leg pain associated with spinal stenosis.[4]

Neurologist: A physician skilled in the diagnosis and treatment of disease of the nervous system.[42]

Neuro paediatrician: Specialist clinicians who have expert knowledge and training in the diagnosis and management of neurological disorders affecting children and young people.[43]

Neurosurgical decompression / Microvascular Decompression: Microvascular decompression is a procedure which aims to relieve pressure from an artery on a cranial nerve — nerves which enter and leave the brain stem directly and pass through the skull base.[44]

Neurosurgeon: Neurosurgeons assess, diagnose, and undertake surgery to treat disorders of the central nervous system, including the brain and spine.[45]

NPRB receptors (natriuretic peptide receptor B): Part of a family of peptides that play an essential role on the regulation of blood pressure, the intravascular volume, and electrolyte homeostasis.[46]

O

Occupational therapist: Occupational therapists work with adults and children of all ages with a wide range of conditions to provide practical support to help people of all ages overcome the effects of disability caused by illness, ageing or accident so that they can carry out everyday tasks or occupations.[47]

Ophthalmologist: A doctor who treats eye diseases.[48]

Orthodontist: A branch of dentistry dealing with irregularities of the teeth (such as malocclusion) and their correction (as by braces).[49]

Orthopaedic surgeon: Doctors who specialise in the musculoskeletal system - the bones, joints, ligaments, tendons, and muscles that are so essential to movement and everyday life.[50]

Ossification: The process of bone formation usually beginning at particular centers in each prospective bone and involving the activities of special cells that segregate and deposit inorganic bone substance around themselves.[4]

Osteopenia: A condition caused by low bone density, although not as severe as osteoporosis.[4]

Osteotomy: A surgical operation in which a bone is divided, or a piece of bone is excised to correct a deformity.[4]

P

Paediatric endocrinologist: A doctor that treats diseases related to problems with hormones,[51] who specialises in the treatment of children and young people.[4]

Paediatrician: A doctor who has special training in medical care for children.[52]

Pathogenic variant: A genetic alteration that increases an individual's susceptibility or predisposition to a certain disease or disorder.[53]

Peroneal nerve: A nerve that branches from the sciatic nerve and provides sensation to the front and sides of the legs and to the top of the feet.[54]

Peroneal palsy: Paralysis of the peroneal nerve.[4]

Physiotherapist: Physiotherapists help people affected by injury, illness or disability through movement and exercise, manual therapy, education, and advice.[55]

Polysomnography: A technique to make a continuous record of multiple physiological variables (such as breathing, heart rate and muscle activity) during sleep.[4]

Primary care doctor: Doctors specifically trained for and skilled in comprehensive first contact and continuing care for persons with any undiagnosed sign, symptom, or health concern (the "undifferentiated" patient) not limited by problem origin (biological, behavioural, or social), organ system, or diagnosis.[56]

Psychologist: A person who specialises in the study of mind and behaviour or in the treatment of mental, emotional, and behavioural disorders.[57]

Pulmonologist: A physician who specialises in the respiratory system.[58]

R

Receptor: A chemical group or molecule (such as a protein) on the cell surface or in the cell interior that has an affinity for a specific chemical group, molecule or virus.[4]

Recessive genes: Recessive is a quality found in the relationship between two versions of a gene and refers to a type of allele which will not be manifested in an individual unless both of the individual's copies of that gene have that particular genotype.[59]

Rheumatologist: Rheumatologists are doctors who investigate, diagnose, manage, and rehabilitate patients with disorders of the musculoskeletal system, such as the locomotor apparatus, bone, and soft connective tissues.[60]

Rhizomelia: A disproportion of the length of the proximal limb usually shortening or deformity affecting the shoulder and arm or hip and thigh.[61]

S

Scoliosis: A lateral curvature of the spine.[4]

Skeletal dysplasia: The medical term for a group of about 400 conditions that affect bone development, neurological function, and cartilage growth, including its most common form, achondroplasia.[62]

Spinal fusion: Spinal fusion is surgery to permanently connect two or more vertebrae in the spine, eliminating motion between them in order to improve stability, correct a deformity or reduce pain.[63]

Spinal stenosis: A narrowing of the spaces within the spine, which can put pressure on the nerves that travel through the spine.[64]

Stenosis: A narrowing or constriction of the diameter of a bodily passage or orifice.[4]

T

Talipes: Also known as club foot, is a deformity of the foot and ankle that a baby can be born with that causes the foot to point downwards at their ankle.[65]

Tissue: An aggregate of cells usually of a particular kind together with their intercellular substance that form one of the structural materials of a plant or an animal.[66]

Trachea: The airway that leads from the larynx (voice box) to the bronchi (large airways that lead to the lungs). Also called windpipe.[67]

Tracheostomy: Surgical construction of a respiratory opening into the trachea through the neck to allow the passage of air.[4]

V

Vertebra: The 33 individual, interlocking bones that form the spinal column.[68]

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