

The National MPS Society Glossary of Terms

Revised 04.04



Acetyl CoA: α -glucosaminide acetyltransferase: Lysosomal enzyme deficient in MPS III-C.

Adenoids: The collection of lymphatic tissue at the rear of the nose. Enlargement of the adenoids may cause obstruction of breathing through the nose.

Adenoidectomy: a surgical procedure to remove adenoid growth.

α -L-Iduronidase: Lysosomal enzyme deficient in MPS I.

α -N-Acetylglucosaminidase: Lysosomal enzyme deficient in MPS III-B.

Amino Acid: A class of chemical compounds that can be built up to form larger polymers called proteins. In most biological systems there are 20 common amino acids that can be linked in various combinations to generate larger molecules containing 100-10,000 amino acids. These larger molecules, or proteins, carry out most of the active functions within a cell or an organism.

Amniocentesis: Procedure involving withdrawal of amniotic fluid, the fluid that surrounds the growing fetus in the uterus, generally performed between the 15th-20th week of pregnancy by inserting a needle through the abdominal wall into the uterus. Cells that are contained in the fluid can be isolated and used for prenatal diagnosis of gender and for particular genetic conditions (including MPS/ML).

Anterior: Front

Arylsulfatase B: Lysosomal enzyme deficient in MPS VI.

Atrophy: A wasting of tissues, organs, or the entire body, as from death and reabsorption of cells, diminished cellular proliferation, decreased cellular volume, pressure, ischemia, malnutrition, lessened function, or hormonal changes. It is often associated with brain disease in MPS/ML children.

Autosomal Recessive Inheritance: A pattern of inheritance in which a nondominant (recessive) gene on a non-sex determining chromosome (autosome) results in a person either being a carrier of a trait or being affected. Males and females are affected with equal frequency. There is usually no family history of the trait. Instead, it is revealed when two unaffected parents who are both carriers of a particular recessive gene have a child who receives two copies of the recessive gene.

β -Galactosidase: Lysosomal enzyme deficient in MPS IV-B.

β -Glucuronidase: Lysosomal enzyme deficient in MPS VII.

Blood-Brain Barrier: The blood vessels of the brain (and the retina) are much more impermeable to large molecules than blood vessels elsewhere in the body. This has important implications for the ability of the organism to mount an immune response and to provide protection to these tissues, although the basis for the difference in blood vessel permeability is

not well understood. The implications for human genetic disease are that it is far more difficult to provide therapeutic treatment to neural (brain) tissues than in other tissues in the body. Since many lysosomal storage diseases have a specific involvement in the neural tissues it is critical to provide access to these tissues during treatment.

Bone Marrow Transplant: See **Stem Cell Transplant**.

Bone Marrow: Tissue found in the center of most bones. It is the site in which most blood cells are made, including red blood cells involved in transport of oxygen in the blood and white blood cells.

BPAP: Bilevel positive airway pressure, often used for people with sleep apnea to open the airway at nighttime using two pressure settings.

Carpal Tunnel: The space between the carpal bones of the wrist and the connective tissue over the flexor tendons. The wrist or carpus consists of 8 small bones known as carpals, which are joined by a band of fibrous proteins called ligament. Nerves have to pass through the wrists in the space between the carpal bones and the ligaments.

Carpal Tunnel Syndrome: Thickening of the ligaments in the carpal tunnel that causes pressure on the nerves. This can cause irreversible nerve damage if not surgically corrected. In children with MPS/ML carpal tunnel syndrome occurs because of accumulation of GAG deposits.

Carrier: An individual who has a recessive, disease-causing version of a gene at a particular site on one chromosome of a pair and a normal version of a gene at that locus on the other chromosome. By definition, carriers of a recessive condition do not have clinical signs and symptoms of the condition.

Cerebrospinal Fluid (CSF): The fluid that surrounds the brain and spinal cord and that is produced in the ventricles of the brain.

Chorionic Villus Sampling (CVS): Prenatal diagnostic procedure involving sampling the chorionic villi, generally performed between the tenth and twelfth weeks of pregnancy. The test can reveal many, but not all, genetic abnormalities. The decision to have prenatal testing, and the appropriate method of prenatal diagnosis should be discussed with your health care provider. Currently amniocentesis is more widely available than CVS for prenatal testing for MPS conditions.

Chromosome: The linear, double stranded structural unit of genetic material consisting of DNA and supporting proteins called chromatin. Human cells are expected to contain 46 chromosomes identified as 23 pairs; 22 pairs are autosomes and one pair are the sex chromosomes.

Contracture: Muscle shortening resulting in loss of motion of the joint.

Cord Blood Transplant: See **Stem Cell Transplant**

Cornea: The transparent circular part of the front of the eye.

Corneal Clouding: Disruption of the clear layers of the cornea in individuals with MPS/ML due to storage of GAG, causing a milky appearance of the eye, decreased vision, and sensitivity to light. Cloudy corneas can be replaced via a corneal transplant.

Corneal Transplant: Surgical procedure to remove cloudy cornea and replace with a healthy, donated cornea.

CPAP: Continuous positive airway pressure, often used for people with sleep apnea to open the airway at nighttime using a constant pressure setting.

Cranium: The part of the skeleton that encloses the brain.

Deposits: See **Glycosaminoglycans (GAGs)**

DNA: The molecule which encodes the genes responsible for the structure and function of an organism and allows for transmission of genetic information to the next generation.

Dysostosis: The abnormal formation of bone caused by the lack of proper ossification.

Echocardiogram: Ultrasound of the heart to evaluate for heart valve and heart muscle function.

Electroencephalogram (EEG): A record of the electric potentials in the brain recorded by attaching electrodes on the scalp. Often this procedure is used to look for seizure activity.

Electrocardiogram (EKG or ECG): A study of the currents in the heart that control its contraction.

Electromyography (EMG): Continuous recording of the electrical activity of a muscle by means of electrodes inserted into the muscle fibers. Used, although not required, to diagnose carpal tunnel syndrome (can be diagnosed by nerve conduction studies).

Enamel: The hard outer covering of the crown of a tooth.

Enzyme Replacement Therapy: A therapeutic approach for a genetic disorder whereby the missing protein is manufactured separately and given intravenously to the patient on a regular basis.

Enzyme: A protein that facilitates a biological reaction without itself being used up in the reaction (i.e. it acts as a catalyst). An enzyme acts by binding with the substance involved in the reaction (the substrate) and converting it into another substance (the product of the reaction).

Fontanelle: The soft spot on a baby's head.

Galactose 6-sulfatase: Lysosomal enzyme deficient in MPS IV-A.

Gastrostomy (G-Tube): A surgical procedure in which an opening is made into the stomach from the outside. It is usually performed to allow nutrition and/or medications to be administered directly into the stomach when swallowing is difficult because of disease or obstruction of the esophagus.

Gene: Basic unit of heredity that codes for a specific protein leading to a particular characteristic or function .

Gene Therapy: A therapeutic approach to a genetic disorder whereby treatment is achieved by inserting a corrected copy of the gene or a new gene to replace the incorrect version.

Genetic Code: Information carried by the DNA molecules that decides the physical traits of an offspring. The code fixes the pattern of amino acids that build body tissue proteins within a cell.

Genu Valgum: knock-knee (knees curving inward in relation to the thigh).

Gibbus: kyphosis.

Glaucoma: A condition in which loss of vision occurs because of an abnormally high pressure in the eye.

Glycosaminoglycans (GAGs): Long repeating chains of complex sugar molecules. See **Mucopolysaccharide**.

Heparan N-sulfatase: Lysosomal enzyme deficient in MPS III-A.

Hepatomegaly: enlargement of the liver.

Hepatosplenomegaly: Enlargement of the liver and the spleen. (Hepatomegaly: enlargement of the liver; Splenomegaly: enlargement of the spleen).

Hernia: Protrusion of a part or structure through the tissues normally containing it.

Heterozygote: An individual possessing a variant gene and a normal gene at identical sites of homologous chromosomes. (adjective: heterozygous).

Heterogeneity: Variations in clinical features(characteristics) within a specific disease.

Homologous Chromosomes: A pair of chromosomes, one from each parent, having the same gene loci in the same order.

Homozygote: An individual possessing a pair of identical pair of genes, either both normal or both variant, at identical sites on homologous chromosomes.

Hurler Syndrome: Severe end of a clinical spectrum of MPS I. See **MPS I**

Hurler-Scheie Syndrome: Clinical spectrum that is intermediate between Hurler and Scheie syndromes. See **MPS I**

Hunter Syndrome: See **MPS II**

Hyaluronidase: Lysosomal enzyme deficient in MPS IX.

Hydrocephalus: An abnormal increase in the amount of cerebrospinal fluid within the ventricles of the brain. Communicating hydrocephalus or increased pressure may be caused by obstruction to the outflow of cerebrospinal fluid from the ventricles or a failure of its reabsorption into the cerebral sinuses. It can be treated using a ventriculoperitoneal shunt.

Hypoxia: A deficiency of oxygen in the tissue or blood.

Inguinal Hernia: Hernia occurring in the lower abdomen and groin.

I-Cell Disease: See **ML II**

Iduronate sulfatase: Lysosomal enzyme deficient in MPS II.

Individualized Education Program (IEP): An IEP is a program designed for each child within the public school system that receives special educational services with goals to improve teaching, learning and appropriate goal setting for each individual. Often, a team including members from the school system and the family are involved in designing the IEP, and federal legislation is in place to guide the development of appropriate IEPs.

Intubation: The placement of a breathing tube during anesthesia.

Joint Contracture: Fibrosis of a muscle tissue producing shrinkage and shortening of the muscle without generating any strength. It is usually a consequence of pain in or disuse of a muscle or limb.

Kyphosis: Abnormal angular curve of the vertebrae of the spine (syn: gibbus).

Lumber Puncture: A procedure in which cerebrospinal fluid is withdrawn by means of a needle inserted into the membrane space in the region of the lower back. This procedure may be performed to measure intracranial pressure to aid in diagnosing hydrocephalus.

Lysosomal Enzyme: A protein found within the cytoplasm of most cells, especially leukocytes, kidney and liver cells, which are key components in the function of digestive processes within the cell.

Lysosomal Storage Disorder (LSD): An inborn error of metabolism resulting in a particular lysosomal enzyme deficiency. At this time there are more than 40 identifiable lysosomal storage disorders.

Lysosome: A specialized compartment (organelle) in the cytoplasm of cells that contains enzymes responsible for breaking down substances in the cell.

Melatonin: A compound involved in circadian rhythms (biological variations during a 24 hour period) sometimes used as a sleep aid for those with MPS/ML

Maroteaux-Lamy Syndrome: See **MPS VI**

Mitral Valve Prolapse: Flaps between 2 parts of the heart, the left atrium and the left ventricle, don't close evenly allowing a small amount of blood to leak back into the left atrium.

ML II: Also called **I-Cell Disease**, caused by a deficiency of the lysosomal enzyme N-acetylglucosaminyl-1-phosphotransferase. Autosomal recessive disease characterized by severe psychomotor retardation and by many of the clinical features seen in severe MPS I.

ML III: Also called **Pseudo-Hurler Polydystrophy**, caused by a deficiency of the lysosomal enzyme N-acetylglucosaminyl-1-phosphotransferase. Autosomal recessive disease with less severe disease course than ML II, presenting later in life with survival into adulthood.

Morquio Syndrome: See **MPS IV**

MPS I: Also called **Hurler, Hurler-Scheie and Scheie**, caused by a deficiency of the lysosomal enzyme α -L-Iduronidase. Autosomal recessive, heterogeneous disease characterized by a wide range of clinical involvement, including corneal clouding, bone changes, stiff joints, large liver and spleen, and heart disease.

MPS II: Also called **Hunter syndrome**, caused by a deficiency of the lysosomal enzyme Iduronate sulfatase. X-linked recessive, heterogeneous disease characterized by a wide range of clinical involvement, including large liver and spleen, stiff joints, bone changes, and heart disease.

MPS III: Also called **Sanfilippo syndrome**, an autosomal recessive disease classified into 4 types based on the enzyme deficiency. The features in each type are similar and characterized by severe central nervous system degeneration but only mild somatic (relating to the body) problems.

MPS III-A: Caused by a deficiency of the lysosomal enzyme heparan N-sulfatase.

MPS III-B: Caused by a deficiency of the lysosomal enzyme α -N-acetylglucosaminidase.

MPS III-C: Caused by a deficiency of the lysosomal enzyme acetyl CoA: α -glucosaminide acetyltransferase.

MPS III-D: Caused by a deficiency of the lysosomal enzyme N-acetyl glucosamine 6-sulfatase.

MPS IV: Also called **Morquio syndrome**, autosomal recessive disease classified into 2 types based on the enzyme deficiency, each with a wide range of clinical manifestations. Both types are characterized by short trunk dwarfism, fine corneal deposits and preservation of intelligence.

MPS IV-A: Caused by a deficiency of the lysosomal enzyme galactose 6-sulfatase.

MPS IV-B: Caused by a deficiency of the lysosomal enzyme β -galactosidase.

MPS VI: Also called **Maroteaux-Lamy syndrome**, caused by a deficiency of the lysosomal enzyme arylsulfatase B. Autosomal recessive disease with bone abnormalities, corneal clouding and normal intelligence.

MPS VII: Also called **Sly syndrome**, caused by a deficiency of the lysosomal enzyme β -Glucuronidase. This is an autosomal recessive disease characterized by large liver and spleen, bone abnormalities and a wide spectrum of severity.

MPS IX: An autosomal recessive disease, caused by a deficiency of the lysosomal enzyme Hyaluronidase, characterized by short stature, soft-tissue masses and normal joint movement and intelligence.

Mucopolipidosis: Term coined to denote diseases that combined clinical features common to both the mucopolysaccharidoses and the sphingolipidoses (diseases characterized by abnormal lipid or fat metabolism, affecting nerve tissue). See **ML II and ML III**

Mucopolysaccharide: A complex carbohydrate molecule that is a common constituent of secretions and the connective tissue between cells. Although the molecules were originally called "mucopolysaccharides" because of their ability to form viscous, mucin-like solutions, the terminology was revised to "proteoglycans" and subsequently to "glycosaminoglycans" in the last decades.

Mutation: A change in the genetic material (DNA) of a cell that alters expected genetic processes.

N-acetylglucosamine 6-sulfatase: Lysosomal enzyme deficient in MPS III-D.

N-acetylglucosaminyl-1-phosphotransferase: Lysosomal enzyme deficient in **ML II/III**.

Odontoid Dysplasia: Malformation in the bones that stabilize the connection between head and neck.

Otitis Media: inflammation of the middle ear occurring commonly in children as a result of an infection and often causing pain and temporary hearing loss.

Papilledema: Swelling around the optic disc.

Port-a-Cath: Brand name for a long-term indwelling catheter into a central vein with access through the skin.

Posterior: Back

Precocious Puberty: The early onset of sexual maturation.

Preimplantation Genetic Diagnosis (PGD): Also known as preimplantation testing. A procedure used to decrease the chance of a particular genetic condition for which a fetus is specifically at risk by testing one cell from embryos from in vitro fertilization for the DNA mutation known in the family. Only embryos found not to carry the DNA mutation are transferred to the mother's uterus.

Pseudo-Hurler Polydystrophy: See **ML III**

Recessive Disease:

Autosomal recessive: A pattern of inheritance requiring the presence of two copies of a particular gene mutation in order to have express clinical signs and symptoms of a condition. Pattern of inheritance seen in all MPS/ML disorders with the exception of MPS II.

X-linked recessive: A mode of inheritance in which a mutation in a gene on the X chromosome causes males to have clinical features of a particular condition as they only have one X chromosome. Pattern of inheritance seen in MPS II.

Recombinant DNA: DNA that contains genes from different sources that have been combined by the techniques of genetic engineering.

Rhinorrhea: Thick, chronic discharge of mucous from the nose.

Scaphocephalic: Having a long, narrow head shape.

Sanfilippo Syndrome: See **MPS III**

Scheie Syndrome: Mild end of a clinical spectrum of MPS I. See **MPS I**

Scoliosis: Lateral (sideways) deviation of the spine.

Seizures: Disruption of electrical signals in the brain. Seizures may cause brief changes in a person's body movements, awareness, emotions, or senses such as taste, smell, vision, or hearing.

Splenomegaly: enlargement of the spleen.

Trigger Finger: Caused by a thickening of the tendon that bends the fingers, often experienced as swelling in the palm of the hand as the finger is moved.

Ventriculoperitoneal shunt: A thin tube that drains fluid from the brain into the abdominal cavity used in the treatment and management of hydrocephalus.

Sleep apnea: A temporary cessation of breathing during sleep generally caused by obstruction of the airway.

Sly Syndrome: See **MPS VII**

Spinal Fusion: Surgery to connect the spinal bones to each other to prevent slippage.

Spleen: A large organ situated on the left side of the body below and behind the stomach.

Stem Cell Transplant: A therapeutic treatment for patients where stem cells from bone marrow, peripheral blood, or from umbilical cord blood are infused into the bloodstream after the original bone marrow cells have been ablated (destroyed) by either chemotherapy and/or radiation therapy. The cells migrate to the interior of certain bones and begin producing immature cells called “committed progenitors”. These committed progenitors produce colonies of cells that eventually mature into red blood cells, white blood cells, or platelets. The purpose is to allow the donor cells to re-populate the bone marrow and various other tissues of the recipient. If the cells can also provide the missing gene and function to the recipient then it can sometimes improve clinical symptoms. It is important to note that the process of destroying the recipient's bone marrow cells is extremely invasive and leaves the individual immunocompromised and susceptible to life threatening infections. Also it is critical to have donor cells come from an individual with compatible tissue types in order to avoid rejection of the donor cells after the transplant bloodstream and the immune cells that provide protection from infection.

Stem Cells: A cell whose daughter cells have the potential to develop into a variety of specialized cell types.

Sternum: A long flat bone, articulating with the cartilages of the first seven ribs and with the clavicle, forming the middle part of the anterior wall of the thorax.

Swallowing study (modified barium swallow study): Videotaped X ray of a individual's oral (mouth) and pharyngeal (throat) mechanism during eating or drinking. This procedure is often ordered to evaluate for obstruction or aspiration. The results from this procedure may allow for a therapist to better identify ways to safely feed the individual and ways to help the family make appropriate modifications.

Trachea: The air tube extending from the larynx to the thorax (level of the fifth or sixth thoracic vertebra) where it divides into the right and left main bronchi.

Tracheostomy: A surgical procedure in which a hole is made into the trachea through the neck to relieve obstruction to breathing. A curved metal, plastic, or rubber tube is usually inserted through the hole.

Umbilical Hernia: A hernia in which bowel or omentum protrudes through the abdominal wall under the skin at the umbilicus (navel).

Revised 4-04