Fabry Disease Glossary

A

Acroparesthesia: A tingling sensation in the hands and feet.

Angiokeratoma: Localized collection of thin-walled blood vessels covered by a cap of warty material. Also described as purple, non-blanching, maculopapular lesions.

Anhidrosis: Absence of sweating in the presence of an appropriate stimulus for sweating, such as heat.

Arrhythmia: Any deviation from the normal rhythm of the heart.

Arthralgia: Severe pain in a joint, without swelling or other signs of arthritis.

Atrioventricular block: Partial or complete block of electrical impulses originating in the atrium or sinus node, preventing them from reaching the AV node and ventricles.

Attenuated Fabry disease phenotype: Characterized by late onset and slow progression of disease.

B

Bioavailability: The proportion of a drug that is delivered to its site of action in the body.

Bronchitis: Inflammation of the bronchi.

C

Cardiac tamponade: A build-up of fluid around the heart within the pericardial sac.

Cardiomyocyte: Involuntary muscle comprising the myocardium and the walls of the pulmonary veins and superior vena cava.

Cardiomyopathy: Any chronic disorder affecting the muscle of the heart.

Celiac disease: A disease of the digestive system that damages the small intestine and interferes with the absorption of nutrients from food.

Congestive heart failure: Failure of the heart to maintain adequate circulation of blood in the tissues.

C

Corneal verticillata: Whorl-like opacities in the cornea.

Cryptogenic stroke: Stroke of unknown cause.

D

Dyspnea: Labored breathing or difficulty in breathing.

E

Elimination: The process of excretion of metabolic waste products from the body.

Endoplasmic reticulum: A system of membranes present in the cytoplasm of the cell. It is the site of the manufacture of proteins and lipids and is concerned with the transport of these products within the cell.

Exogenous: Originating outside the body or part of the body.

F

Fabry crises: Sharp attacks of pain which may persist for minutes to days.

Fibroblast: A widely distributed cell in connective tissue that is responsible for the production of both the ground substance and precursors of collagen, elastic fibers, and reticular fibers.

Fibromyalgia: A disorder characterized by pain in the fibrous tissue components of muscles without any inflammation.

Fibrosis: A thickening and scarring of connective tissue, most often a consequence of inflammation or injury.

G

Genotype: The genetic constitution of an individual or group as determined by the particular set of genes it possesses.

G

Glomerular filtration rate (GFR): The rate at which substances are filtered from the blood of the glomeruli into the Bowman's capsules of the nephron.

Glycosphingolipids: A heterogeneous group of membrane lipids formed through the covalent link of a glycan moiety to ceramide.

Golgi apparatus: A collection of vesicles and folded membranes in a cell that is usually connected to the endoplasmic reticulum. It stores, matures, and transports the proteins manufactured in the endoplasmic reticulum.

H

Hemizygote: Genes that are carried on an unpaired chromosome.

Hepatocytes: The principal cell type in the liver.

Heterozygote/heterozygous: An individual in whom the members of a pair of genes determining a particular characteristic are dissimilar.

Homogenate: Material that has been reduced to uniform consistency.

Hypertrophy: Increase in the size of a tissue or organ brought about by the enlargement of cells rather than cell multiplication.

Hypohidrosis: A reduction in sweating in the presence of an appropriate stimulus, such as heat.

I

Immunoglobulin (Ig): One of a group of structurally related proteins that act as antibodies. Several classes of Ig with different functions are distinguished–IgA, IgD, IgE, IgG, and IgM.

Infiltrative hypertrophic cardiomyopathy: Deposition of substances that cause the ventricular walls to become rigid.

Interstitial cells: Cells that form the part of the connective tissue (interstitium) between other tissues.

L

Left ventricular hypertrophy: The enlargement and thickening of the walls of the left ventricle.
Leukocytes (white blood cell): Any blood cell that contains a nucleus. There are three major subdivisions: granulocytes, lymphocytes, and monocytes.

Lymphedema: Accumulation of lymph in the tissues, producing swelling. The legs are most often affected.

Lyonization (X-chromosome inactivation): A process by which one of the two copies of the X-chromosome is inactivated. Which one is inactivated can vary between tissues within the same organism.

Lysosome: An organelle in the cytoplasm of the cell that contains enzymes responsible for breaking down substances in the cell and is bound by a single membrane.

M

Maculopapular: A rash that consists of both macules and papules.

Mesangial cells: Forms the central stalk of the glomerulus and interacts closely with endothelial cells and podocytes.

Microalbuminuria: The presence of albumin in the urine at levels that are higher than normal but lower than those detected by the standard protein dipstick test.

Multisystemic disease: A disease that affects many systems in the body.

Myocardial fibrosis: Fibrosis of cardiac muscle. (see **Fibrosis**)

Myocardial infarction: Death of a segment of heart muscle, which follows interruption of its blood supply.

N

Nasopharyngitis: Inflammation of the nasopharynx.

O

Obligate heterozygotes: An individual who may be clinically unaffected but who must carry a gene mutation based on analysis of the family history.

Organelle: A structure within the cell that is specialized for a specific function.

P

Phenotype: The observable characteristics of an individual, which result from interactions between genes and the environment.

Podocyte: An epithelial cell in the glomerulus of the kidneys that spreads over the capillary basement membrane.

Point mutation: A mutation that changes one nucleotide in a gene by substitution, deletion, or addition.

Proteinuria: The presence of protein in the blood.

R

Raynaud's disease: A condition of unknown cause in which the arteries of the fingers are unduly reactive and enter spasm when the hands are cold.

Recombinant DNA: DNA that contains genes from different sources that have been combined by genetic engineering rather than breeding.

Rheumatoid arthritis: The second most common form of arthritis that typically involves the joints of the fingers, wrists, feet, and ankles, with later involvement of the hips, knees, shoulders, and neck.

S

Sinusitis: Inflammation of one or more of the mucous-membrane–lined air spaces in the facial bones that communicate with the nose (the paranasal sinuses).

Stroke: Sudden attack of weakness usually affecting one side of the body as a consequence of an interruption of blood flow to the brain.

Systemic lupus erythematosus: A chronic inflammatory autoimmune disease of connective tissue, affecting the skin and various internal organs.

T

Telangiectasia: A localized collection of distended blood capillary vessels. It is recognized as a red spot, sometimes spidery in appearance that blanches on pressure.

Tinnitus: The sensation of sound in the ears, head, or around the head in the absence of an external sound source.

Transfection: The direct transfer of DNA molecules into a cell.

Transient ischemic attack: The result of temporary disruption of the circulation to part of the brain due to embolism, thrombosis to brain arteries, or spasm of the vessel walls.

V

Vertebrobasilar: The vertebrobasilar system contains three blood vessels: the two vertebral arteries and a single basilar artery that is located toward the back of the brain and provides approximately 20% of the intracranial blood supply.

Vertigo: Disabling sensation in which the individual feels that their surroundings are in a state of constant movement.

Vessel tortuosity: The bending or twisting of a blood vessel.

W

White matter: Nerve tissue of the central nervous tissue that is paler in color than the associate grey matter because it contains more nerve fibers and thus larger amounts of the insulating material, myelin.