Medical Terminology Guide

This glossary explains key terms related to GM1 Gangliosidosis and other lysosomal storage diseases to help you communicate with healthcare providers and explain the condition to others.

Lysosome	Small sac-like structures within cells that contain enzymes that break down various substances and recycle cellular materials. Often called the cell's "recycling center."
Enzyme	A protein that speeds up chemical reactions in the body. In lysosomal storage diseases, specific enzymes are missing or don't work properly.
Beta-galactosidase (β-galactosidase)	The specific enzyme that is deficient in GM1 Gangliosidosis. It normally breaks down certain lipids and sugars in the cell.
Ganglioside	A type of complex fat (lipid) found in nerve tissue, especially abundant in the brain.
Lysosomal Storage Disease (LSD)	A group of about 70 rare inherited metabolic disorders caused by defects in lysosomal function, resulting in abnormal buildup of toxic materials in various organs.
Autosomal recessive	The pattern of inheritance for GM1 Gangliosidosis. A child must inherit two copies of the mutated gene (one from each parent) to develop the condition. Parents who carry one copy of the mutation typically do not show symptoms.
GLB1 gene	The gene that provides instructions for making the beta-galactosidase enzyme. Mutations in this gene cause GM1 Gangliosidosis
Carrier	A person who has one copy of a mutated gene for an autosomal recessive condition but doesn't show symptoms of the disorder. When two carriers have a child, there's a 25% chance the child will have the disorder.

Type I (Infantile form)	The most severe form of GM1 Gangliosidosis, with symptoms appearing by age 6 months, rapid progression, and limited life expectancy.
Type II (Juvenile form)	Symptoms typically appear between ages 1-5 years, with a more gradual progression than Type I. Life expectancy ranges from childhood to early adulthood.
Neurodegeneration	Progressive damage and death of nerve cells in the brain and spinal cord, leading to loss of function.
Developmental regression	Loss of previously acquired developmental skills and milestones.
Hepatosplenomegaly	Enlargement of both the liver and spleen.
Hypotonia	Decreased muscle tone, often presenting as "floppiness.
Dystonia	Involuntary muscle contractions causing repetitive or twisting movements.
Seizure	Sudden, uncontrolled electrical disturbance in the brain that can cause changes in behavior, movements, feelings, and levels of consciousness.
Cherry-red spot	A characteristic eye finding in some forms of lysosomal storage diseases, appearing as a red spot surrounded by whitish tissue when examining the retina.
Enzyme assay	A test that measures the activity of a specific enzyme. For GM1, it measures beta-galactosidase activity.
Genetic testing	Analysis of DNA to identify mutations in the GLB1 gene.
Biopsy	Removal of a small sample of tissue for examination under a microscope
Biomarker	A measurable substance that indicates the presence of a disease state.
Enzyme Replacement Therapy (ERT)	Treatment that provides a functional version of the missing or deficient enzyme, typically administered intravenously.
	Uses medications to reduce the production of substances that build up

Substrate Reduction	in cells.
Gene Therapy	Experimental approach that aims to treat the disease by replacing, inactivating, or introducing genes into cells.
Hematopoietic Stem Cell Transplant (HSCT)	Transplantation of blood-forming stem cells to potentially provide cells that can make the missing enzyme.
Pharmacological Chaperone Therapy	Uses small molecules to stabilize and improve the function of the affected enzyme.
Palliative care	Medical care focused on providing relief from symptoms and improving quality of life for people with serious illnesses.
Clinical trial	Research study performed with human participants to evaluate medical, surgical, or behavioral interventions.
Natural history study	Research that follows a group of people over time to understand how a disease progresses without treatment
Biorepository	A facility that collects, processes, stores, and distributes biological samples for research purposes.

This glossary provides basic definitions of common terms related to GM1 gangliosidosis and lysosomal storage diseases. For more detailed information, please consult with healthcare providers or visit curegm1.org.