

# Glossary of Terms

Name	Description
Carrier	Someone who has a specific gene change associated with a condition, but who does not have the condition themselves. They may pass on the gene to their children
Ceramide glucosyltransferase	An enzyme that helps to produce glucocerebroside. Glucocerebroside builds up in Gaucher cells. Some therapies for Gaucher disease reduce or limit the amount of ceramide glucosyltransferase, so that less glucocerebroside is produced
Enzyme	A substance in the body that helps to bring about a specific biochemical reaction
Enzyme replacement therapy	This type of treatment for Gaucher disease aims to replace the missing enzyme glucocerebrosidase
ERT	An abbreviation for Enzyme Replacement Therapy
Gene mutation	As genes are passed down through generations, they may change slightly. A changed gene is said to have mutated. The changed version of the gene is then passed on to children
Glucocerebrosidase	The enzyme that breaks down glucocerebroside. People with Gaucher disease don't have the glucocerebrosidase needed to break down the glucocerebroside

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Intravenous	Into the body via a drip or needle into a vein. A way of giving people fluids or drugs
Lymphatic system	A network of tissues, nodes and organs that transports lymph around the body. Lymph is a fluid that helps fight infection. The system as a whole works to process and rid the body of toxins, waste, and other unwanted materials
Lysosome	Compartment inside a cell that contains the enzyme glucocerebrosidase. Undigested glucocerebrosides builds up in the lysosomes. This is why Gaucher disease is known as a lysosomal storage disorder
Macrophage	Cells that 'eat' worn-out cells and break them down for re-use
Mutation	See Gene mutation
SRT	See substrate reduction therapy
Substrate reduction therapy	This type of treatment aims to reduce the amount of glucocerebroside that is produced by the body
Glucocerebroside	The substance that is usually broken down into glucose (sugar) and a fat called ceramide. It is broken down by glucocerebrosidase. In Gaucher disease, glucocerebrosides builds up in the cells
Haemoglobin	A protein which is found in red blood cells which transports oxygen around the body