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Opinion Article

Different Types of Lysosomal Storage Disorders and Treatment Methods

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DESCRIPTION

Lysosomal storage disorders are a group of more than 50 rare diseases. They affect the lysosome a structure in animal cells that breaks down substances such as proteins, carbohydrates, and old cell parts so the body can recycle them. People with these disorders are missing important enzymes (proteins that speed up reactions in the body). Without those enzymes, the lysosome isn't able to break down these substances. When that happens, they build up in cells and become toxic. They can damage cells and organs in the body. Lysosomal Storage Diseases (LSDs) are inborn errors of metabolism characterized by the accumulation of substrates in excess in various organs' cells due to the defective functioning of lysosomes. They cause dysfunction of those organs where they accumulate and contribute to great morbidity and mortality. Although rare individually, their prevalence is significant when viewed collectively.

Lysosomal storage diseases are caused by defects in individual genes. Approximately 70% of Lysosomal storage disorders are due to defects in enzymes, this occurs due to defects in enzyme activators or related proteins. Genes at specific chromosomal locations transcribe specific enzymes. Incorrect enzyme coding leads to inactive enzymes. Similarly, mutations in activator genes result in defective activators. 70 Lysosomal storage diseases have been described, and many more may be discovered in the future. Individually rare but common, they justify efforts to study these disorders as a group. These conditions cause disease in the organs where they accumulate and determine clinical signs and symptoms. Babies and children suffer more than adults. Clinical features are unique among many children and adults with the same disease. For example, children's developing brain is prone to symptoms and signs of dysfunction, whereas these are mild or absent in adults.

Some of lysosomal disorders are explained below

Fabry disease: A lysosomal disorder that affects a person's ability to make alpha-galactosidase-A enzyme. This enzyme breaks down a fatty substance called globotriaosylceramide, without this enzyme, fat accumulates within cells and damages them.

Gaucher disease: Lack of glucocerebrosidase causes this condition. This enzyme breaks down a fat called glucocerebroside. Without glucocerebroside, fat accumulates in the spleen, liver and bone marrow.

Krabbe disease: This affects the nervous system. A deficiency in the enzyme galactosylceramidase causes this condition. This enzyme helps to maintain myelin, the protective layer around nerve cells, and aids in nerve communication.

Metachromatic leukodystrophy: If a person doesn't have the enzyme arylsulfatase A, it causes Metachromatic Leukodystrophy. It usually breaks down a group of fats called sulfatides. Without enzymes, these fats accumulate in the white matter, the part of the brain that contains nerve fibers. This destroys the myelin coating that surrounds and protects nerve cells.

Treatment methods

Pharmacological chaperone therapy: The final form of the enzyme is its tertiary structure. This involves proper folding of protein molecules and resistance to degradation. Certain mutations in lysosomal-associated proteins involved in the folding process render the enzyme fragile. In such cases, the catalytic site is intact. Small molecules that penetrate the membrane of cellular organelles, increase resistance to enzymatic degradation are called chaperones. When used, they are invaluable in some LSDs.

Proteostatic regulators: Unlike Pharmacological Chaperone Therapy, proteostatic regulators are not ligands (they are not bound to enzymes), but rather shift the balance away from proteolytic (enzymatic) degradation by regulating multiple steps in the metabolic pathway.

Small molecule assisted substrate transportation: Small Molecule Assisted Substrate Transportation is a transporter-less modality in which the accumulated substrate is converted into another compound that is readily transported out of the lysosome.

Anti-inflammatory: Inflammation is involved in the pathogenesis of Lysosomal disorders. Anti-inflammatory and immunosuppressive agents reduce inflammatory cytokines such as IL-1 and TNF-α, reducing organ damage.

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