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Commentary

Enzyme Deficiencies and its Causes

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DESCRIPTION

Enzymes are proteins that help the body split down food components into energy during metabolism, which is how it acquires energy for regular growth and development. Enzyme deficiencies, or a lack of certain enzymes, are genetic abnormalities that can lead to a variety of life-altering or life-threatening illnesses, including: Lysosomal storage disorders (LSDs) are metabolic inborn abnormalities characterised by the excessive accumulation of substrates in the cells of diverse organs due to lysosome dysfunction. They cause organ dysfunction and contribute to high morbidity and mortality in the organs where they accumulate. LSDs (lysosomal storage disorders) are diseases caused by single-gene abnormalities. Nearly seventy percent of LSDs are caused by enzyme deficiencies, with the rest being caused by abnormalities in enzyme activator or related proteins.

Incorrect enzyme-coding leads in inactive enzymes; a gene on a certain chromosome locus transcribes a specific enzyme. Defective activators are also caused by mutations in activator genes. So far, seventy LSDs have been identified, with many more anticipated to be discovered in the future. One out of every 5,000 newborns born in the United States is thought to have some kind of LSD. Intellectual and developmental problems, clouded corneas, small stature, stiff joints, incontinence, speech and hearing impairment, chronic runny nose, hernia, heart disease, hyperactivity, depression, pain, and a severely decreased life span are all common in those who are affected.

The mucopolysaccharidoses (MPS) are a class of lysosomal storage disorders that are hereditary. Within cells, lysosomes serve as the principal digesting units. Lysosomes contain enzymes which break down or digest specific elements, such as carbs and lipids. Individuals with MPS disorders have an abnormal deposition of some complex carbohydrates (mucopolysaccharides or glycosaminoglycans) in their arteries, skeleton, eyes, joints, ears, skin, and/or teeth due to a deficit or dysfunction of particular lysosomal enzymes. The respiratory system, liver, spleen, central nervous system, blood, and bone marrow are all places where these levels can be detected. Cells, tissues, and organ systems in the body suffer progressive damage as a result of this accumulation. Mucopolysaccharidosis is divided into numerous kinds and subtypes.

NPC (Niemann-Pick disease type C) is a slowly progressing lysosomal condition with age-dependent symptoms. Hepatosplenomegaly, jaundice, and (in certain cases) pulmonary infiltrates are the most common visceral symptoms in the perinatal period and infancy. The presentation is dominated by neurologic symptoms from late infancy onwards. Hypotonia and developmental delay are common in early children, with ataxia, dysarthria, dysphagia, and, in certain cases, epileptic seizures, dystonia, and gelastic cataplexy developing later. Mutations in the SMPD1 gene cause Niemann-Pick disease types A and B. This gene directs the production of an enzyme known as acid sphingomyelinase. This enzyme can be found in lysosomes, which are cellular compartments that break down and recycle various chemicals.

Acid sphingomyelinase is responsible for converting sphingomyelin, a lipid, into ceramide, a different type of lipid. Mutations in SMPD1 cause a deficit of acid sphingomyelinase, resulting in diminished sphingomyelin breakdown and fat accumulation in cells. Cells malfunction and eventually die as a result of the fat buildup. Cell loss affects the function of tissues and organs in people, including the brain, lungs, spleen, and liver, over time.

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