

Lysosomal Storage Disorders A Practical Guide

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Lysosomal storage diseases are a group of about 50 rare inherited metabolic disorders that result from defects in lysosomal function. Lysosomes are sacs of enzymes within cells that digest large molecules and pass the fragments on to other parts of the cell for recycling. This process requires several critical enzymes. If one of these enzymes is defective due to a mutation, the large molecules accumulate within the cell, eventually killing it. Lysosomal storage disorders are caused by lysosomal

Lysosomal storage disease - Wikipedia
In each case, lysosomal storage diseases are caused by an inborn error of metabolism that results in the absence or deficiency of an enzyme, leading to the inappropriate storage of material in various cells of the body. Most lysosomal storage disorders are inherited in an autosomal recessive manner.

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It is quite difficult to diagnose this disorder as the symptoms vary from one type to another type of Lysosomal Storage Disorders. There are a few diagnoses carried out in a specialized laboratory. Tissue biopsies are also used for diagnosis of Lysosomal Storage Disorder in the patient by analyzing the sample of tissue or cells collected from the patient's body for analysis.

Lysosomal Storage Disease -Types, Symptoms, Causes and its ...
Lysosomal storage diseases (LSDs) are due to the inherited deficiency of one of over 40 lysosomal enzymes, and lead to accumulation of undegraded substrate in a range of organs and tissues. They are multisystem and progressive disorders.

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