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Lysosomal storage disorders (LSDs) are monogenic inborn errors of metabolism. Various groups have been delineated according to the affected pathway and the accumulated substrate, and new entities are still being identified. They are severe disorders with a heterogeneous clinical spectrum encompassin ...

[Lysosomes and lysosomal storage diseases]

FIGURE 2. Timeline for discoveries on lysosomal storage disorders (LSD) and their impact on cell biology Although some of the lipidoses and mucopolysaccharidoses (MPS) were the first of the lysosomal storage disorders to be described, much of the initial concept for the lysosome and its dysfunction came from the studies of Pompe disease.

Lysosomal Storage Disease: Revealing Lysosomal Function ...

Measuring relative lysosomal volume for monitoring lysosomal storage diseases. te Vrugte D(1), Wallom KL(1), Platt FM(1). Author information: (1)Department of Pharmacology, University of Oxford, Oxford, UK. Biomarkers are important tools in medicine, which can be used for monitoring disease progression and response to therapy.

Measuring relative lysosomal volume for monitoring ...

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Lysosomes And Lysosomal Diseases Volume 126 Methods In ...

Lysosomal storage disease. Lysosomal storage diseases comprise a group of over 70 inherited metabolic disorders caused by deficiency of certain enzymes in certain compartments of the cells that generally involve progressive neurological manifestations and that primarily affect children 1). Lysosomal storage diseases are individually rare but collectively affect 1 in 5,000 live births.

Lysosomal storage disease types, list, causes, symptoms ...

Some of the most common lysosomal storage disorders include: Gaucher disease: Gaucher disease often causes spleen and liver enlargement, blood problems and bone issues. Learn more about Gaucher disease. Fabry disease: This disorder often causes severe burning pains in hands and feet and, in some cases, a distinctive skin rash on the legs ...

Lysosomal Storage Disease & Disorder | National Gaucher ...

Lysosomes were originally described in the early 1950s by de Duve who was also the first to recognize the importance of these organelles in human disease. We know now that lysosomes are involved in numerous biological processes, and abnormalities in lysosomal function may result in a broad range of diseases.

Lysosomes, Lysosomal Storage Diseases, and Inflammation ...

Second, we quantified the endo-lysosomal volume in LPS-treated cells fixed with 4% PFA. Herein, we took advantage of that this treatment collapsed tubular endo-lysosomes into spheroid objects. As with other previous measurements, we observed higher endo-lysosome volume in fixed LPS-treated relative to resting macrophages . Collectively, these data demonstrate that the endo-lysosome volume expands in response to macrophage and dendritic cell stimulation, concurrent with tubulation.

Enhanced translation expands the endo-lysosome size and ...

In nervous system disease: Disorders of fat and fatty acid metabolism. Lysosomal diseases are caused by the accumulation of substances that are normally metabolized in the cellular structures called lysosomes. These disorders often produce symptoms of neurological involvement at birth or in the early years of childhood. Read More

Lysosomal disorder | pathology | Britannica

Lysosomes are near-ubiquitous organelles and lysosomal diseases can affect any tissue or organ system. Imbued with a wide complement of acid hydrolases, each with specificity towards a particular chemical linkage, lysosomal enzymes act in concert to degrade a diverse constellation of macromolecular complexes, which may be predominantly lipid, polysaccharide, protein or nuclear acid in nature.

The cellular pathology of lysosomal diseases - Cox - 2012 ...

Lysosomal storage diseases are a group of rare, inborn, metabolic errors characterized by deficiencies in normal lysosomal function and by intralysosomal accumulation of undegraded substrates. The past 25 years have been characterized by remarkable progress in the treatment of these diseases and by the development of multiple therapeutic approaches. These approaches include strategies aimed at ...

Lysosomal Storage Diseases: From Pathophysiology to ...

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Recent Developments in Therapeutic Approaches for ...

The problem with lysosomes is that if they are ruptured, their contents can leak out and cause serious damage to the cell. Calcium oxalate crystal-induced kidney injury, which is linked to the...