Enzyme Errors: Little People and Lysosomal Storage Disorders

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Abstract

Lysosomal storage disorders (LSD) are a group of inherited metabolic disorders causing enzyme malfunction or dysfunction in key pathways, causing the the accumulation of excess substrate in lysosomes, which are the cell's "recycling center". The abnormal buildup of substances in the lysosomes can cause progressive, irreversible damage to the affected organ systems. While enzyme function assays and genetic testing are definitive, we discuss the role of electron microscopy (EM) in recognizing LSD's and illustrative cases where EM findings eventually led to a diagnosis.