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A Review on Lysosomal Storage Disease

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Abstract: *Lysosomal Storage Disorders (LSDs) comprise a group of at least 50 distinct genetic diseases, each one resulting from a deficiency of a particular lysosomal protein activity, or in a few cases from nonlysosomal activities that are involved in lysosomal biogenesis or protein maturation. All share a common biochemical characteristic in that they result in accumulation of normally degraded substrates within lysosomes. That is eventually leads to an irreversible cell damage, an ultimately multi-organ dysfunction. Th e substrates stored and site of storages vary, leading to a wide spectrum of clinical manifestations. Lysosomal storage disorders are a family of over seventy rare monogenic diseases that typically present in infancy or childhood and collectively affect 1:5000 live births Most of the causative genes encode lysosomal enzymes or proteins involved in lysosomal enzyme modification or transport, but they can also encode lysosomal membrane proteins.*

Keywords: Lysosomal Storage Disorders