Lysosomal Storage Disease (LSDs)

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A Lysosome (Derived from the Greek words Lysis, meaning to “loosen” and Soma, “body”) is a structure, covered by a membrane which exists in all animal cells except in red blood cell. This Lysosome is one of the minute bodies seen with the electronic microscope, containing various hydrolytic enzymes. Injury to a lysosome is followed by release into the cell the enzymes, which can injure the cell. The lysosomal storage diseases are a clinically different group of inborn errors of metabolism that are classified based upon the biochemical nature of incompletely degraded substances which accumulate in different tissue. The lysosome maintains an acidified environment, containing catabolic enzymes which ease degradation of various byproducts of cellular turnover delivered to the lysosome. Phagocytosis and autophagy are other avenue of delivery of these substances into the lysosome. Absence or deficiency in the hydrolytic activity within the lysosome which can be a consequence of mutation within encoding gene is the most common cause of the disease. For instance deficient glucocerebrosidase activity in Gusher’s disease results in the storage of its substrate, glygosphingolipids, glucosylceramide.

There is wide different clinical manifestation of each entity among individually affected patients. Presentation of disease manifestation may appear at any time from birth till adulthood. The variable disease severity between the individual patients seems stem from different mutations. The majority of lysosomal storage diseases are autosomal recessive disorder. Heterozygotes or carrier individuals with a single defective gene often have adequate enzyme activity generated by their other alleles. It is said lysosomal storage disease have a combined prevalence of about 1/5000-8000 live births and so far 70 of lysosomal storage diseases have been described.

As far as treatment is concerned, recombinant human enzymes succeeded introduction of enzyme replacement therapy for several disorders, like: Gaucher’s, Fabry’s, Pompe’s (Glycogene storage disease II) diseases and Haurler-Scheie (MPSI H/S), Hunter’s (MPS II) and Maroteaux-Lamy (MPS VI) syndrome.

Also, enzyme enhancement therapy and gene therapy are considered as other ways of lysosomal storage disease therapies.

Keyword: Lysosome; Metabolic disease; Storage disease