Lysosomal Storage Disease

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Lysosomes are cytoplasmic organelles containing hydrolytic enzymes that degrade the macromolecules proteins of cellular catabolism. The causes of lysosomal enzyme disorder stem from

1. Impaired enzyme synthesis
2. Abnormal enzyme targeting
3. Defect of structure of Accessory factor which is needed for enzymes function. Clinical manifestations depend on the organ(s) involved.

Seizure disorder, mental handicap and regression are the characteristics of many lysosomal enzyme storage diseases. The lysosomal storage disease, as first described by Hers are characterized by an accumulation of undergraded macromolecules within lysosomes. These disorders are clinically heterogenous groups of inborn errors by about 50 different clinical entities. Nature of the incompletely degraded macromolecules that accumulate in various tissues include:

Tthe glycogen storage diseases

The mucopolysuccharidosis

The mucolipidosis

The glycoproteinosis

The sphingolipidosis

The acid lipase deficiency diseases

It is said the combined prevalence of all lysosomal storage disease is 1 in 6600 to 1 in 7700 live birth (1).

The disease entities result from various single gene mutation with each of the enzyme defect is induced by one of the several different abnormalities. Mutation or deletions that produce immunologically responsive or unresponsive enzyme protein. The defect can impair glycosylation of the enzyme protein or cause a failure to generate recognition markers that permits the enzyme to attach itself to the lysosomal membrane. The majority of genes that encode lysosomal various lysosomal storage disease subtypes is manifested by multi systemic involvement. Disease manifestation may be evident prenatally or at any time from birth to adulthood on the therapeutic front several modalities of treatment have been suggested Enzyme replacement therapy, enzyme enhancement therapy, substrate reduction therapy and gene therapy are the

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different approaches which at present, are being tried.

Treatment of the secondary disabilities like seizure disorder, sensory deficit, behavioral problem, and sleep-wake cycle disorder has positive impact on the patients quality of life and help address some of parental anxieties. Genetic counseling of affected individuals and their relatives is also important component of care of the patient and family.

Keywords: Lysosome; Enzyme; Gene therapy; Child

References:

1. Meikle PJ. Hopwood JJ, Clange AE, et al. prevalence of lysosomal storage disorders. Am Med Assoc 1999; 281: 249-254.
2. Hers Gh. Inborn lysosomal diseases. Gastroenterology 1963; 58:605-633.