

International Journal of Advanced Research in Science, Communication and Technology (IJARSCT)

International Open-Access, Double-Blind, Peer-Reviewed, Refereed, Multidisciplinary Online Journal

Volume 5, Issue 3, January 2025

A Review on Mucopolysaccharidosis

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Abstract: Mucopolysaccharidosis (MPS) refers to a group of rare genetic disorders caused by the deficiency of specific enzymes required to break down glycosaminoglycans (GAGs), which are long chains of sugar molecules. These disorders lead to the accumulation of GAGs in various tissues and organs, resulting in progressive damage and a range of symptoms.

There are several types of MPS, including MPS I, II, III, IV, VI, and VII, each associated with different enzyme deficiencies and varying severity of symptoms. Common features of MPS include skeletal abnormalities, cardiovascular issues, respiratory problems, vision and hearing loss, and developmental delays.

Diagnosis typically involves clinical evaluation, biochemical tests to measure enzyme activity, and genetic testing to identify specific mutations. Treatment options vary depending on the type of MPS and may include enzyme replacement therapy, hematopoietic stem cell transplantation, and supportive care to manage symptoms.

Early diagnosis and intervention are crucial for improving the quality of life and outcomes for individuals affected by MPS. Ongoing research aims to develop new therapies and improve understanding of these complex disorders..

DOI: 10.48175/568

Keywords: Mucopolysaccharidosis



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