S21-3 Enzyme Replacement Therapy for Mucopolysaccharidoses

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Mucopolyasaccharidoses (MPS) are genetic disorders due to the congenital deficiency of lysosomal enzymes necessary for degradation of mucopolysaccharide in lysosome. Seven different types of MPS are reported. All types are inherited by autosomal recessive trait except MPSII, inherited by X-linked recessive trait. MPS is a systemic disease and multiple organs are affected. Major clinical symptoms are mental retardation, regression, corneal clouding, otitis media, hearing loss, obstructive lung disease, valvular heart diseases, hepatosplenomegaly, dysostosis multiplex, and joint stiffness. Enzyme replacement therapy has been developed in three types of MPS (MPSI, MPSII, and MPSVII). Normalization of liver and spleen volume, improvement of respiratory function are observed in almost all cases treated by enzyme administration. However, there is no obvious therapeutic efficacy for CNS lesions. Development of therapy for CNS lesions is expected.