GENETICS

MANAGEMENT OF GENETIC DISORDERS

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Abstract: *The treatment of inherited metabolic diseases and other genetic disorders have been limited primarily to symptomatic and supportive care. In the last two decades, advances in understanding the pathogenesis of the diseases and biotechnology has helped to develop novel therapies for genetic disorders like enzyme replacement therapy. Hemetopoietic stem cell transplantation is the state of the art treatment for hemoglobinopathies and some metabolic disorders. Enzyme replacement therapy is a reality for Gaucher disease, Fabry disease, mucopolysaccharidosis I and VI. Cost is prohibitive for clinical use especially in developing countries and enough facilities are not available. Bisphosphonates in osteogenesis imperfecta is the standard of care to prevent recurrent fractures. Gene therapy is envisioned as a potentially definitive treatment for a variety of diseases that have a genetic etiology. However, additional clinical and basic research is needed to determine the future role of gene therapy. This review discusses the various modalities of treatment of genetic disorders like metabolic correction, hematopoietic stem cell transplantation, enzyme replacement therapy, pharmacological therapy and gene therapy.*

Keywords: *Hematopoietic stem cell transplantation, Enzyme replacement therapy, Gene therapy, Metabolic correction.*

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Points to Remember

- ***Management of genetic disorders can be considered as a biological model viewed from clinical phenotype and working back to the molecular level.***
- ***Metabolic disorders can be treated with substrate limited therapy (dietary restriction) and pharmacological therapy to reduce the toxicity of the accumulated metabolite.***
- ***Enzyme replacement therapy and enzyme enhancement therapy is showing promising results in the management of lysosomal storage disorder.***
- ***Hematopoietic stem cell transplantation (HSCT) is an effective treatment for various genetic disorders like thalassemia, osteopetrosis and primary immunodeficiency disorders.***
- ***Gene therapy is a promising technology for the definitive treatment of genetic disorders.***

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