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Abstract Topic:- Rare disease therapeutics

Abstract Title:- Our experience of enzyme replacement therapy (ERT) in Gaucher disease and Hunter disease (MPS-II) patients

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Aims:-Gaucher and Hunter disease are the common Lysosomal storage disorders (LSDs) seen in India. Current approaches for treatment of LSDs includes- enzyme replacement therapy (ERT), Bone marrow transplantation (BMT), Pharmacological Chaperon therapy (PCT), Substrate reduction therapy (SRT) and Gene therapy. Significant improvement in clinical outcome of the LSD's patients have been observed after emergence of ERT. This is considered the standard of care for many LSDs and is presently approved for the treatment of Fabry disease, Gaucher disease, Pompe disease, Mucopolysaccharidosis (MPS) type I, II, IVA, VI and VII, acid lipase deficiency, and late infantile neuronal ceroid lipofuscinosis type 2 (CLN2). In this study, we elaborate our experience of the outcomes of ERT in 3 patients each with Infantile Gaucher disease, Adult onset Gaucher disease and Hunter disease respectively.

Methods:- We have carried out a long-term follow up study in three of our patients who received ERT approval from Takeda. 1st patient (P-I) affected with Infantile Gaucher disease was 3 years old male child at the starting of treatment. 2nd patient(P-II), a case of adult onset Gaucher disease was 22 years old female at the starting. Both the cases were confirmed by haematological, biochemical and molecular parameters. Intravenous infusion of Velaglucerase Alfa with dose of 60 units/kg body weight of every 15 days has been given and assessed for all the haemotogical and biochemical parameters at the interval of 12 months for 6 years. 3rd patient (P-III) is a severe case of Hunter disease (MPS-II) diagnosed biochemically and by molecular study at the age of 3 years. He has been given intravenous infusion of Elaprase 0.5 mg/kg body weight every week for past 10 years and measured all the physical and biochemical parameters every year.

Results:- P-I is currently 9 years old male child affected with Gaucher disease now going to school and performs all his work without any complaint. P-II was an adult case of Gaucher disease, is now mother of two healthy female children with no complications (prenatal and post-natal). In both the cases, spleen size and plasma chitotriosidase levels has been significantly decreased in size and his haematological factors like- Haemoglobin and Platelets count have been markedly increased. P-III was a 9-year-old male child with severe form of Hunter disease. He has shown significant impact on visceral organs and also improvement in behavioural issues. Though his cognition is not much improved. His total Glycosaminoglycans (GAG) has been significantly reduced with notably increased height.

Conclusions:- Our study demonstrates the benefits of early initiative of ERT in patients with infantile Gaucher disease, adult onset Gaucher disease and Hunter disease. None of the patients has developed any infusion related complication till date.

Keywords:- Enzyme replacement therapy, Gaucher disease, MPS-II, LSD