AIIMS, Mangalagiri successfully treats a child with rare disease (MPS- type 1) by enzyme replacement therapy for the first time in Andhra Pradesh

Department of Pediatrics, AIIMS Mangalagiri has successfully treated a 3 yr old child with a rare disease, mucopolysaccharidosis Type I with enzyme replacement therapy. This is the first time such treatment is being given to a child in the state of Andhra Pradesh. MPS-1 is a rare metabolic disease with estimated incidence of 1 case for every 100,000 births. This disease occurs due to the deficiency of L-iduronidase enzyme which is required for breaking down of an endogenous complex carbohydrate known as Glycosaminoglycans (CAG). Deficiency of this enzyme leads to accumulation of GAG fragments inside the lysosomes of cells leading on to symptoms. MPS-1 is a severe, progressive disorder with involvement of multiple organs and carries a very poor prognosis without treatment.

Bone marrow transplantation is curative in some children if done early before 2 years of age. For most others, the main treatment is lifelong 'enzyme replacement therapy'. In 'enzyme replacement therapy (ERT)' the deficient enzyme is given exogenously as injections at frequent intervals. The ERT drug for MPS-1 is known as Inj. ALDURAZYME, is marketed by Genzyme Sanofi pharmaceuticals in India. For our child, the entire cost of treatment for one year (Approx 85 lakhs) is being funded by ESI hospital Vijayawada under ESI scheme.