Safety, Tolerability, Ascending Dose and Dose Frequency Study of rhHNS Via an IDDD in MPS IIIA Patients

Emma Children's Hospital, Academic Medical Center, Amsterdam, , Netherlands
St. Mary's Hospital, Manchester, , United Kingdom

Sanfilippo syndrome, or Mucopolysaccharidosis (MPS) III, is a rare lysosomal storage disease (LSD) caused by loss in activity of 1 of 4 enzymes necessary for degradation of the glycosaminoglycan (GAG) heparan sulfate (HS) in lysosomes. MPS IIIA results from deficiency of the enzyme heparan N-sulfatase (sulfamidase). MPS IIIA symptoms arise on average at 7 months of age, with the average age of diagnosis at 4.5 years for the majority of patients. The central nervous system (CNS) is the most severely affected organ system in patients with MPS IIIA, evidenced by deficits in language development, motor skills, and intellectual development. In addition, there are abnormal behaviors including but not limited to aggression and excess motor activity/hyperactivity that contribute to disturbances in sleep.Overall, individuals with MPS IIIA have a marked developmental delay and significantly reduced lifespan of 15 years of age on average.

The purpose of this study is to determine the safety and tolerability of rhHNS via ascending doses administered via an a surgically implanted intrathecal drug delivery device (IDDD) intrathecal (IT) route once monthly (or every two weeks) for 6 months in patients with MPS IIIA.