Mucopolysaccharidosis (MPS VI), or Maroteaux-Lamy syndrome (MIM#253200), is an autosomal recessive lysosomal storage disease resulting from the deficiency of the enzyme N-acetylgalactosamine 4-sulfatase or Arylsulfatase B (ASB). This enzyme is responsible for the hydrolysis of the sulfate group of dermatan sulfate and chondroitin sulfate. Deficiency or lack of this enzyme activity leads to a progressive intracellular accumulation of the glycosaminoglycans resulting in an irreversible multisystem dysfunction and premature mortality.

MPS VI is characterized by a wide spectrum of clinical manifestations as well as all MPSs, that includes skeletal deformities (dysostosis multiplex) with stunted growth, coarse facial features, corneal opacity, hepatosplenomegaly, cardiac abnormalities, and usually a normal cognitive function (Valayannopoulos, et al., 2010). The severe phenotype may have onset at birth with progressive severe skeletal deformities and mortality within the second decade of life. A slowly progressive phenotype has a later onset in life with mild skeletal deformities and a survival up to the fifth decade of life. Since identification of the gene (ARSB), more than 150 mutations have been described. Most of these mutations are either novel, private, or compound heterozygous making phenotype-genotype correlation difficult as well as population screening.

Until recently, supportive care and bone marrow transplantation were the only treatments available for MPS VI patients. Supportive care has been focused to optimize the general well being, family support and counseling. Bone marrow or hematopoietic stem transplantation has been done for a limited number of patients worldwide. A long-term follow up study documented the restoration of normal enzyme activity with good cardiovascular function and stabilization of visual acuity. However, skeletal abnormalities remained a major problem for these patients (Herskhovitz, et al., 1999).

Naglazyme (Galafreze) was approved as an Enzyme Replacement Therapy (ERT) for MPSVI in May 2005 and was considered a safe and effective treatment secondary to complications of their underlying disease. Three siblings were started on the treatment at the age of 12 and 13 years respectively. Both died after 5 months of commencing the treatment (Harmatz P., et al., 2006). Case series show a significant improvement of cardiovascular function and stabilization of visual acuity. However, treatment before development of the anticipated skeletal deformities is mandatory in order to prevent this complication. Therefore early detection and diagnosis with immediate intervention are crucial for a better outcome.

This study reports on 18 Saudi Arab patients from six unrelated, consanguineous families seen and diagnosed at Johns Hopkins Aramco Healthcare from January 1, 1983 to December 31, 2013. Five of these families are inhabitants of the Al-Hasa area, the largest oasis in Saudi Arabia located in the Eastern Province. One family originated from Abha (in the south of Saudi Arabia). Five patients received ERT, and one received a bone marrow transplantation from a full matched heterozygous sibling. The clinical course and outcomes for this group of patients are described.