A Multicenter, Multinational, Cross-sectional Clinical Assessment Study of Subjects with Mucopolysaccharidosis IVA (Morquio Syndrome)

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Synopsis:
MOR-001 is a multicenter, multinational, cross-sectional clinical assessment study of subjects diagnosed with Mucopolysaccharidosis IVA (Morquio Syndrome).

Mucopolysaccharidosis IV type A (MPS IVA, also known as Morquio syndrome) is one of the rarest lysosomal storage disorders in the United States. Reliable incidence figures are not available, but estimates have varied between 1 in 200,000 live births to 1 in 300,000 live births. MPS IVA is an inherited, autosomal recessive disease belonging to the group of lysosomal storage diseases and is characterized by deficient activity of lysosomal enzymes that are required for the degradation of the glycosaminoglycan (GAG) keratan sulfate (KS). As a result, KS accumulates in many tissues and organs and is excreted in the urine. There are 2 different enzyme activity deficiencies that define the 2 types of Morquio syndrome: in type A, N-acetylgalactosamine-6-sulfatase (galactose-6-sulfatase; GALNS) activity is deficient and in type B, beta-galactosidase activity is deficient. This study is examining MPS IV, type A, only. The excessive lysosomal storage of keratan sulfate (KS) causes systemic skeletal dysplasia, short stature, and joint abnormalities, all of which limit mobility and endurance.

This multicenter, multinational, cross-sectional study in subjects diagnosed with MPS IVA will be the first clinical assessment study conducted in the world based on direct observation and testing of a substantial number of subjects affected with MPS IVA. The study will enroll MPS IVA subjects without limitations on age or symptom severity. The sample size of up to 300 subjects will represent 5-10% of the estimated total disease population in the world. The objective of the study is to quantify endurance and respiratory function in subjects with MPS IVA and to better characterize the spectrum of symptoms and biochemical abnormalities in MPS IVA disease.