Replagal Enzyme Replacement Therapy for Adults With Fabry Disease

National Institute of Neurological Disorders and Stroke (NINDS), Bethesda, Maryland, United States

This study will determine the safety and effectiveness of the drug Replagal for treating people with Fabry disease, an inherited metabolic disorder. In this disease, an enzyme called alpha-galactosidase A, which normally breaks down a lipid (fatty substance) known as ceramidetrihexoside, is missing or does not function properly. As a result, the lipid accumulates in the body, causing problems with the kidneys, heart, nerves, and blood vessels. This study will examine whether replacing the missing alpha-galactosidase A with a genetically engineered form of the enzyme called Replagal can reverse the illness.

Patients with Fabry disease who are 18 years of age or older and have completed 10 weeks of Replagal therapy as participants in protocol TKT027 may be eligible for this 6-month study extension.

Participants undergo the following tests and procedures:

- Intravenous (IV) infusions of Replagal every other week over 25 weeks for a total of 13 infusions, with close monitoring during and after the infusions.

- Brief safety evaluations at the time of each infusion, including a check of vital signs (blood pressure, pulse, breathing rate, temperature), review of any side effects, and review of medications.

- Comprehensive evaluations at baseline (before starting Replagal therapy), after 13 and 25 weeks of therapy, and 30 days after completing therapy. These include a medical history and physical examination, symptoms and pain questionnaire, blood and urine tests, check of vital signs, electrocardiogram (EKG), 2-hour Holter monitor, and sweat test (QSART).