An Open-Label Maintenance Study of the Enzyme Replacement Therapy Replagal in Patients With Fabry Disease

National Institutes of Health Clinical Center, 9000 Rockville Pike, Bethesda, Maryland, United States

This study will continue to evaluate the safety of using intravenous doses of Replagal for two patients with Fabry disease. Fabry disease is a genetic disorder inherited as an X-linked recessive trait. It causes a deficiency in the enzyme alpha galactosidase, which normally breaks down a lipid, or fatty substance called ceramidetrihexoside, a building block in all cells of the body. The deficiency in breaking down the lipid eventually causes that lipid to accumulate and injure cells. Vascular, renal, and neurological problems are the results. It is not known exactly how lipid accumulation brings about such problems, studies of another lipid storage disorder.

Two patients 7 to 17 years of age who have Fabry disease and have been receiving intravenous infusions of Replagal at a dose of 0.2 mg/kg of body weight every 2 weeks may be eligible for this study.

Participants will undergo the following tests and procedures:

- Physical examination.
- Neurological examination.
- Medical and medication history.
- Vital signs.
- Assessment of height and weight.
- Blood tests to determine complete blood count and chemistries.
- Electrocardiogram.
- Doppler blood flow study.

Participants will go through a baseline evaluation, over a period of about 1 day. They will receive an intravenous infusion of Replagal every other week, at the dose of 0.2 mg/kg of body weight. Vital signs will be measured before the infusion and immediately and after and 1 hour afterward. There will be careful monitoring for allergic reactions and side effects. The infusion time takes approximately 40 minutes.

This study will last at least 1 year, or until the sponsor doing the investigating or the drug manufacturer decides to withdraw support of the study.