Addendum: Sanofi announces additional data on avalglucosidase alfa in Pompe disease

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Pompe Disease

Pompe disease is a progressive, debilitating and often fatal neuromuscular disease caused by a genetic deficiency or dysfunction of the lysosomal enzyme acid alpha-glucosidase (GAA) that results in the buildup of glycogen in the body's cells. The disease affects an estimated 50,000 people worldwide and can occur at any age from infancy to late adulthood.

Pompe disease is often classified as late-onset (LOPD) or infantile-onset Pompe disease (IOPD). Patients with LOPD typically present any time after the first year of life to late adulthood The hallmark symptom of LOPD is skeletal muscle weakness, which often leads to walking disability and reduced respiratory function. Patients often require wheelchairs to assist with mobility and may require mechanical ventilation to help with breathing. Pompe disease is classified as IOPD when symptoms begin within a few months of birth and there is impact to the heart in addition to skeletal muscle weakness.

Avalglucosidase Alfa Clinical Development Program

Avalglucosidase alfa is an investigational enzyme replacement therapy that has been designed for selective receptor targeting and enzyme uptake, with the aim of enhancing glycogen clearance.

The original NEO1 study included a total of 24 LOPD patients, including 10 with no prior treatment experience and 14 who had been in treatment with alglucosidase alfa for at least nine months. Following completion of treatment in NEO1, 19 of 24 patients were enrolled in the NEO-EXT study, of which 17 are still enrolled. Patients in NEO-EXT are treated with avalglucosidase alfa at 20 mg/kg dose every other week for a planned six-year follow-up.

Mini-COMET is a Phase 2, open-label, ascending-dose, 3-cohort study, evaluating safety, pharmacokinetics, and preliminary efficacy of avalglucosidase alfa in patients aged <18 years with IOPD previously treated with alglucosidase alfa demonstrating clinical decline (Cohorts1&2) or sub-optimal response (Cohort3).

In addition to the NEO-EXT trial and mini-COMET, avalglucosidase alfa is being evaluated in the COMET trial, a currently recruiting Phase 3 study to compare the efficacy and safety of bi-weekly infusions of avalglucosidase alfa and alglucosidase alfa in patients with LOPD who have not previously been on treatment. For more information on these trials, please visit https://www.clinicaltrials.gov/.