ultragenyx pharmaceutical

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FOR IMMEDIATE RELEASE:

Ultragenyx Initiates Phase 2 Study of UX001 in Hereditary Inclusion Body Myopathy, a Rare Neuromuscular Disease

NOVATO, CA – July 5, 2012 - Ultragenyx Pharmaceutical Inc., a biotechnology company focused on developing treatments for rare and ultra-rare genetic disorders, today announced the dosing of the first two patients in a Phase 2 study of UX001 for hereditary inclusion body myopathy (HIBM). HIBM is a rare, severe, neuromuscular disease caused by sialic acid deficiency. UX001 is an extended-release oral tablet formulation of sialic acid (SA-ER) intended as a substrate replacement therapy for HIBM.

The Phase 2 clinical trial is an international, multi-center, randomized, double-blind, placebo-controlled, parallel group study of UX001 in HIBM patients. The study plans to enroll up to 45 patients between 18 and 65 years of age with a previously demonstrated mutation in the GNE gene causing HIBM. The subjects will receive either of two dose levels of SA-ER or placebo over 24 weeks, with all patients continuing on active treatment after 24 weeks. The study's primary objectives are evaluating safety, and improvements in sialylation biochemistry of muscle (pharmacodynamic endpoint). Clinical and patient-reported outcomes will also be evaluated though the study is not powered for these endpoints. Study sites are located in the US and Israel. The total duration of the Phase 2 study is up to 48 weeks, with data expected in 2013.

Emil D. Kakkis, MD, PhD, Chief Executive Officer of Ultragenyx commented: "The initiation of this Phase 2 study is a critical milestone for our team in developing a therapeutic for HIBM patients who currently lack treatment options for this devastating disease. It follows quickly upon the positive results from our Phase 1 trial. This Phase 2 study should help us determine if UX001 is improving the biochemistry of the muscle in these patients and help us learn more about the disease. We look forward to seeing top line results next year."

About HIBM

HIBM is also known as GNE myopathy, Quadriceps Sparing Myopathy (QSM), Inclusion Body Myopathy type 2, Distal Myopathy with Rimmed Vacuoles (DMRV) and Nonaka myopathy. HIBM is a severe, adult-onset, progressive, genetic neuromuscular disease caused by a deficiency of an enzyme in the first step of sialic acid biosynthesis needed for the modification of proteins and fats. Patients with HIBM typically begin to have weakness and abnormal walking at 18 to 30 years of age. Over the ensuing 10 to 20 years, many patients progressively lose significant functional ability and become wheelchair-bound. There are no current treatments for this disease.

About Ultragenyx

Ultragenyx is a privately held, developmental stage biotechnology company committed to bringing life-enhancing therapeutics for patients with rare and ultra-rare genetic diseases, also known as orphan and ultra-orphan diseases, to market. The company focuses on rare metabolic diseases that affect small numbers of patients, but for which the unmet medical need is high and there are no effective treatments. Ultragenyx intends to build a sustainable pipeline of safe and effective therapies to address these underserved diseases. Ultragenyx' lead program, UX001, is being evaluated as a potential treatment for hereditary inclusion body myopathy (HIBM), also known as GNE myopathy. The UX001 program has been granted orphan drug designation in the US and the EU.

The company is led by an experienced management team in rare disease therapeutics. Ultragenyx is striving toward an improved model for successful rare disease drug development, which has the potential to increase efficiency while maintaining appropriate safety and efficacy standards. The company believes that it can deliver significant value to patients by building a high-quality pipeline of rare disease therapeutics and efficiently transforming good science into great medicine.

For more information on Ultragenyx, please visit the company's website at www.ultragenyx.com.