Phase II study of an oral therapy for Gaucher disease yields positive results

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Gaucher disease, a rare enzyme deficiency disorder, is one of many conditions with few approved treatment options for patients. In a study published online today in *Blood*, the journal of the American Society of Hematology, researchers present positive results of a Phase II clinical trial of eliglustat tartrate, an oral therapy in development to treat Gaucher disease.

Gaucher disease is a genetic disorder affecting an estimated 1 in 50,000 to 1 in 100,000 people in the U.S. population. Gaucher disease occurs when a mutation of the glucocerebrosidase gene causes low activity of that enzyme in the body. As a result, harmful fatty substances accumulate in the liver, spleen, bones, and bone marrow, preventing cells and organs from working properly. The primary treatment option is enzyme replacement therapy, which is given intravenously, to break down the accumulated fatty substances.

Eliglustat tartrate is an oral drug currently in development for Gaucher disease type 1 (GD1). As a substrate reduction therapy, the drug decreases the body's production of the fatty substances so they do not accumulate in cells. Based on Phase I trials in healthy volunteers that demonstrated positive initial safety results, the research team initiated a multinational, open-label, single-arm Phase II study of 26 GD1 patients to evaluate the efficacy, safety, and pharmacokinetics (how the body absorbs and processes the drug) of eliglustat tartrate administered orally at 50mg or 100mg doses twice daily. Eligible patients had confirmed Gaucher disease, characterized as enzyme deficiency and a spleen

volume at least 10 times greater than normal, plus abnormal values of either platelet count or hemoglobin levels.

"As with many other rare conditions, we strive to develop new options with improved efficacy and safety for our patients, but we also look for options that may be easier on the provider and the patient.In this case, we were hoping an oral alternative would be viable," said lead study author Elena Lukina, MD, of the Hematology Research Center at the Russian Academy of Medical Sciences in Moscow, Russia.

The study endpoint (improvement in at least two of the three main efficacy parameters: spleen volume, hemoglobin level, and platelet count) was met by three-fourths (77%) of all patients and nearly all (91%) of the 22 patients who completed the full 52 weeks, with the greatest overall improvements seen in hemoglobin level and spleen volume. The research team found statistically significant improvements across many disease symptoms, including mean hemoglobin level, platelet count, spleen volume, liver volume, and lumbar spine bone mineral density. Furthermore, the patients' glucosylceramide plasma levels normalized. Disease symptoms seemed to respond rapidly, and improvement was seen especially in bone mineral density. This may have resulted from the drug's small molecular size, which allows it to diffuse quickly within affected cells.

"Eliglustat tartrate provides promise for a safe, effective, and convenient oral therapy for patients with Gaucher disease type 1," said Dr. Lukina.

Further clinical development of eliglustat tartrate is already proceeding with larger, controlled Phase III studies in untreated patients and in patients previously stabilized with imiglucerase - an intravenously administered medication currently used to treat Gaucher disease.

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