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Drug improves measures of genetic disease that affects liver, spleen

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Among previously untreated adults with Gaucher disease type 1, a genetic disease in which there is improper metabolism due to a defect in an enzyme, treatment with the drug eliglustat resulted in significant improvements in liver and spleen size hemoglobin level, and platelet count, according to a study in the February 17 issue of *JAMA*.

Gaucher disease type 1 is characterized by enlargement of the spleen and liver, anemia, low blood platelets, chronic bone pain, and the failure to grow properly. Untreated Gaucher disease type 1 is a chronic and progressive disorder associated with disability, reduced life expectancy, and, in some patients, life-threatening complications. The current standard of care is enzyme replacement therapy, which requires lifelong intravenous infusions every other week. A safe, effective oral therapy is needed, according to background information in the article.

Pramod K. Mistry, M.D., Ph.D., F.R.C.P., of the Yale University School of Medicine, New Haven, Conn., and colleagues randomly assigned 40 untreated adults with Gaucher disease type 1 to receive eliglustat (twice daily; n = 20) or placebo (n = 20) for 9 months. Eliglustat is a novel oral medication, which showed favorable results for patients with this disease in a phase 2 trial. This phase 3 trial was conducted at 18 sites in 12 countries.

The researchers found that administration of eliglustat resulted in a reduction in spleen volume of approximately 30 percent compared with placebo, as well as improvements in hemoglobin level, decreased liver

volume (-6.6 percent), and increased platelet count (41 percent). No serious adverse events occurred. No patient discontinued treatment over the course of the 9-month study because of a treatment-emergent adverse event.

The authors add that more definitive conclusions about clinical efficacy and utility will require comparison with the standard treatment of enzyme replacement therapy as well as longer-term follow-up.

More information: *JAMA*, DOI: 10.1001/jama.2015.459