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## Gerçek yaşam etkililik çalışmasına göre ERT Gaucher'in progresif bulgularını önlemektedir.

Weinreb NJ et al. Effectiveness of enzyme replacement therapy in 1028 patients with type 1 Gaucher disease after 2 to 5 years of treatment: a report from the Gaucher Registry. Am J Med 2002;113:112-9.

**PURPOSE:** Gaucher disease is the first lysosomal storage disorder to be treated with macrophage-targeted enzyme replacement therapy. Previous studies in relatively small numbers of patients demonstrated short-term efficacy of this treatment. This study describes the effects of 2 to 5 years of treatment on specific manifestations of type 1 Gaucher disease.

**SUBJECTS AND METHODS:** Physicians reported data from 1028 patients to the Gaucher Registry. Assessment of response included serial measurements of hemoglobin concentration, platelet count, liver and spleen volumes, and the occurrence of bone pain and bone crises.

**RESULTS:** Among anemic patients, hemoglobin concentration increased to normal or near normal within 6 to 12 months, with a sustained response through 5 years. In thrombocytopenic patients with intact spleens, the most rapid response occurred during the first 2 years, with slower improvement thereafter. The likelihood of achieving a normal platelet count decreased with increasing severity of baseline thrombocytopenia. In patients who had undergone splenectomy, platelet counts returned to normal within 6 to 12 months. Hepatomegaly decreased by 30% to 40% during follow-up; splenomegaly decreased 50% to 60%, but rarely to volumes below five times normal size. In patients with pretreatment bone pain or bone crises, 52% (67/128) were pain free after 2 years and 94% (48/51) reported no additional crises.

**CONCLUSION:** Enzyme replacement therapy prevents progressive manifestations of Gaucher disease, and ameliorates Gaucher disease-associated anemia, thrombocytopenia, organomegaly, bone pain, and bone crises.

