

# 30 YILDAN 30 MAKALE

2007

## Gaucher'de kemikte etkili bir tedavi yaşam kalitesini artırır.

Weinreb N et al. Imiglucerase (Cerezyme) improves quality of life in patients with skeletal manifestations of Gaucher disease. Clin Genet 2007;71:576-88.

Health-related quality of life (HRQOL) can be diminished in patients with type 1 Gaucher disease (GD) owing to the debilitating clinical manifestations of this chronic disease. This study investigates the impact of imiglucerase treatment on HRQOL of patients with type 1 GD and bone involvement. Thirty-two previously untreated type 1 GD patients with skeletal manifestations including bone pain, medullary infarctions, avascular necrosis, and lytic lesions received biweekly imiglucerase (at 60 U/kg). The Short Form-36 Health Survey (SF-36) was administered at regular intervals to assess HRQOL. Mean baseline SF-36 physical component summary (PCS) scores were diminished relative to US general population norms. Low PCS scores were more common in patients with medullary infarction, lytic lesions, and higher bone pain severity scores. Statistically significant improvements were observed for all eight SF-36 subscales after 2 years of treatment. Mean PCS and mental component summary (MCS) scores increased to within the normal range after 2 years of treatment and were maintained through year 4. Large HRQOL gains were observed even in patients with the most advanced disease and lowest baseline PCS scores. Imiglucerase treatment has a significant positive impact on HRQOL of type 1 GD patients with skeletal disease, including those with bone infarctions, lytic lesions, and avascular necrosis.

