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2025 Gaucher'de ERT kemik mineral yoğunluğunu artırır.

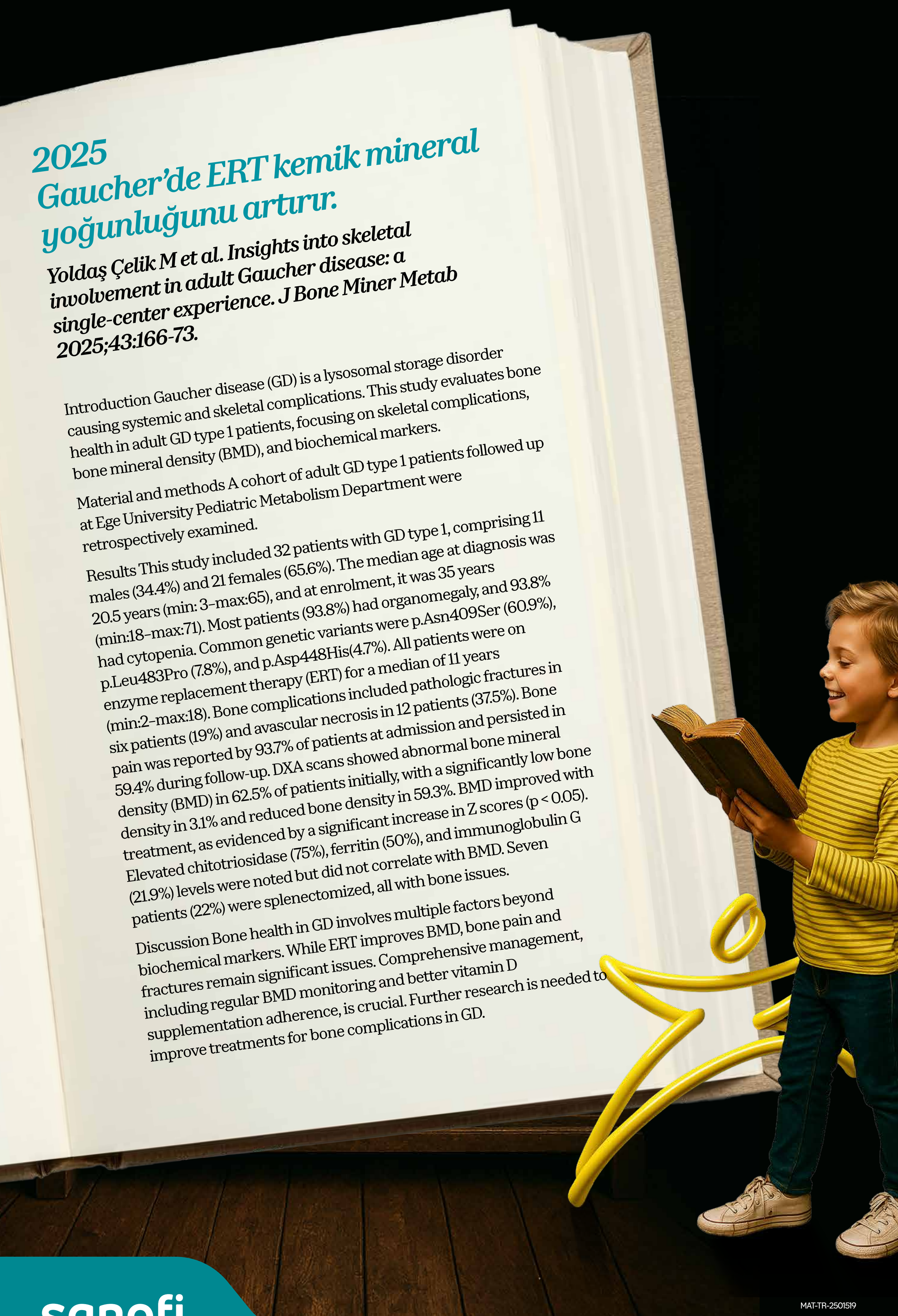
Yoldaş Çelik M et al. Insights into skeletal involvement in adult Gaucher disease: a single-center experience. J Bone Miner Metab 2025;43:166-73.

Introduction Gaucher disease (GD) is a lysosomal storage disorder causing systemic and skeletal complications. This study evaluates bone health in adult GD type 1 patients, focusing on skeletal complications, bone mineral density (BMD), and biochemical markers.

Material and methods A cohort of adult GD type 1 patients followed up at Ege University Pediatric Metabolism Department were retrospectively examined.

Results This study included 32 patients with GD type 1, comprising 11 males (34.4%) and 21 females (65.6%). The median age at diagnosis was 20.5 years (min: 3–max:65), and at enrolment, it was 35 years (min:18–max:71). Most patients (93.8%) had organomegaly, and 93.8% had cytopenia. Common genetic variants were p.Asn409Ser (60.9%), p.Leu483Pro (7.8%), and p.Asp448His(4.7%). All patients were on enzyme replacement therapy (ERT) for a median of 11 years (min:2–max:18). Bone complications included pathologic fractures in six patients (19%) and avascular necrosis in 12 patients (37.5%). Bone pain was reported by 93.7% of patients at admission and persisted in 59.4% during follow-up. DXA scans showed abnormal bone mineral density (BMD) in 62.5% of patients initially, with a significantly low bone density in 3.1% and reduced bone density in 59.3%. BMD improved with treatment, as evidenced by a significant increase in Z scores ($p < 0.05$). Elevated chitotriosidase (75%), ferritin (50%), and immunoglobulin G (21.9%) levels were noted but did not correlate with BMD. Seven patients (22%) were splenectomized, all with bone issues.

Discussion Bone health in GD involves multiple factors beyond biochemical markers. While ERT improves BMD, bone pain and fractures remain significant issues. Comprehensive management, including regular BMD monitoring and better vitamin D supplementation adherence, is crucial. Further research is needed to improve treatments for bone complications in GD.



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