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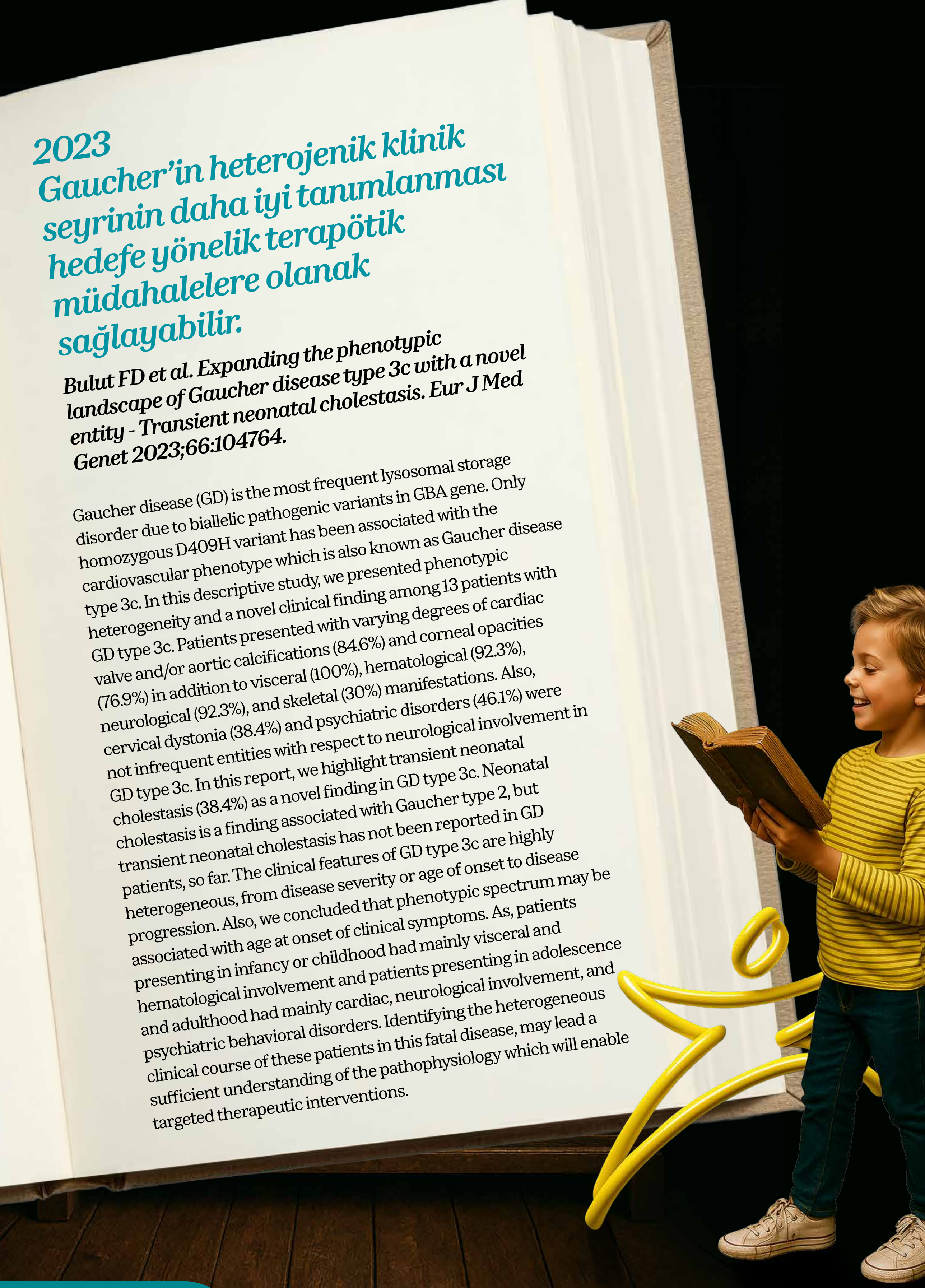
Gaucher'in heterojenik klinik seyrinin daha iyi tanımlanması hedefe yönelik terapötik müdahalelere olanak sağlayabilir.

Bulut FD et al. Expanding the phenotypic landscape of Gaucher disease type 3c with a novel entity - Transient neonatal cholestasis. Eur J Med Genet 2023;66:104764.

Gaucher disease (GD) is the most frequent lysosomal storage disorder due to biallelic pathogenic variants in GBA gene. Only homozygous D409H variant has been associated with the cardiovascular phenotype which is also known as Gaucher disease type 3c. In this descriptive study, we presented phenotypic heterogeneity and a novel clinical finding among 13 patients with GD type 3c. Patients presented with varying degrees of cardiac valve and/or aortic calcifications (84.6%) and corneal opacities (76.9%) in addition to visceral (100%), hematological (92.3%), neurological (92.3%), and skeletal (30%) manifestations. Also, cervical dystonia (38.4%) and psychiatric disorders (46.1%) were not infrequent entities with respect to neurological involvement in GD type 3c. In this report, we highlight transient neonatal cholestasis (38.4%) as a novel finding in GD type 3c. Neonatal cholestasis is a finding associated with Gaucher type 2, but transient neonatal cholestasis has not been reported in GD patients, so far. The clinical features of GD type 3c are highly heterogeneous, from disease severity or age of onset to disease progression. Also, we concluded that phenotypic spectrum may be associated with age at onset of clinical symptoms. As, patients presenting in infancy or childhood had mainly visceral and hematological involvement and patients presenting in adolescence and adulthood had mainly cardiac, neurological involvement, and psychiatric behavioral disorders. Identifying the heterogeneous clinical course of these patients in this fatal disease, may lead a sufficient understanding of the pathophysiology which will enable targeted therapeutic interventions.



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