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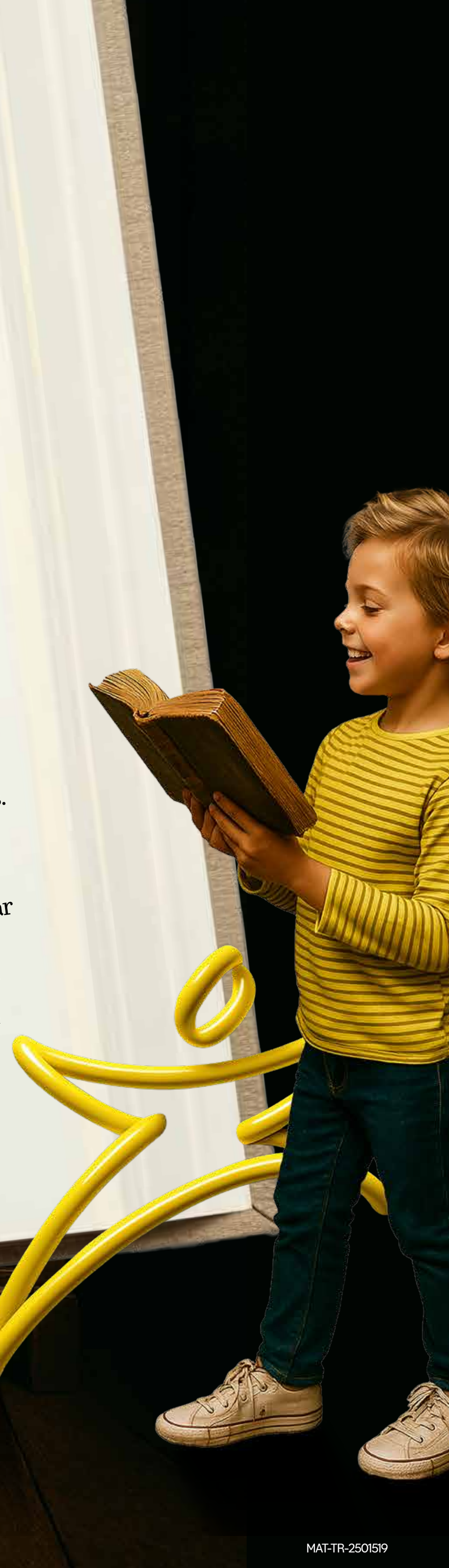
Gaucher'de vestibüler tutulum görülebilir.

Onan E et al. Audiovestibular findings in Gaucher disease Types I and III: Evidence of vestibular involvement in GD1. J Inherit Metab Dis 2025;48:e70046.

Gaucher disease (GD), the most prevalent lysosomal storage disorder, is characterized by varying levels of systemic and neurological involvement. This study aims to investigate audiovestibular system involvement in patients with Gaucher disease type I (GD1) and type III (GD3) using audiometric and vestibular evaluations. We conducted a retrospective analysis of 42 patients diagnosed with GD who presented to the Department of Otorhinolaryngology at Çukurova University Faculty of Medicine between January 2001 and September 2023. The evaluations included pure tone audiometry (PTA), speech discrimination scores (SDS), acoustic impedance tests, and Video Head Impulse Test (vHIT) assessments. Of the 42 patients, 18 were diagnosed with GD1, and 24 with GD3. Audiovestibular anomalies were identified in 11 patients (26.2%). Sensorineural hearing loss (SNHL) was detected in 9 patients, including 4 GD1 and 5 GD3 patients, with bilateral involvement in 5 cases. The severity of hearing loss ranged from mild to moderately severe. Vestibular impairment, demonstrated by reduced vestibulo-ocular reflex (VOR) gain and catch-up saccades (CUS), was observed in 5 patients, predominantly among GD3 cases. Notably, concurrent audiovestibular dysfunction was observed in three patients, one with GD1 and two with GD3. This study is the first to describe the vestibular involvement in GD1. Audiovestibular abnormalities can manifest in both GD1 and GD3 patients, with distinct patterns of involvement. Regular auditory and vestibular assessments are essential to identify sensory deficits early, guide rehabilitation strategies, and enhance the quality of life for GD patients.



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