Cerezyme® 30 YIL

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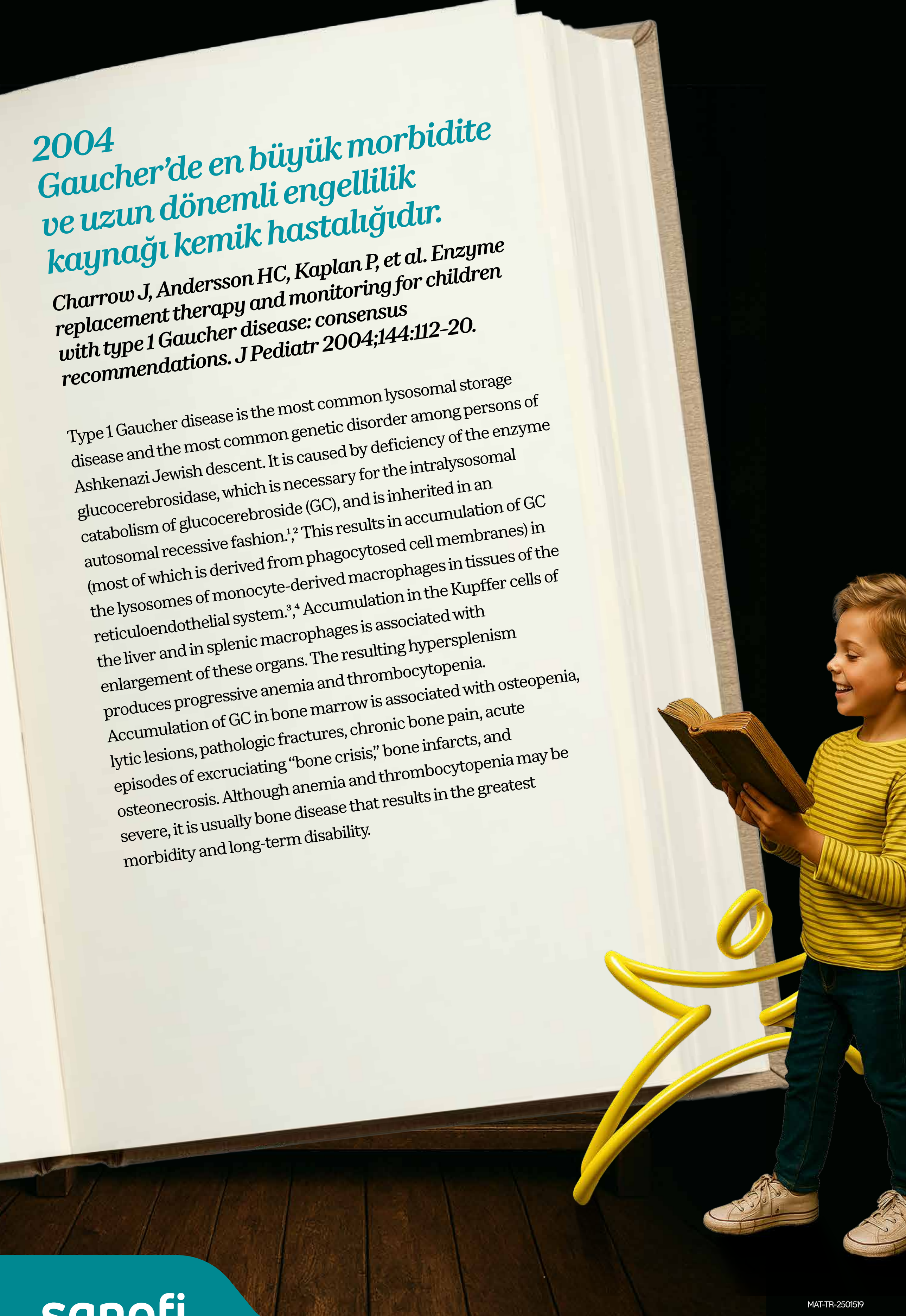
Gaucher'de en büyük morbidite ve uzun dönemli engellilik kaynağı kemik hastalığıdır.

Charrow J, Andersson HC, Kaplan P, et al. Enzyme replacement therapy and monitoring for children with type 1 Gaucher disease: consensus recommendations. J Pediatr 2004;144:112–20.

Type 1 Gaucher disease is the most common lysosomal storage disease and the most common genetic disorder among persons of Ashkenazi Jewish descent. It is caused by deficiency of the enzyme glucocerebrosidase, which is necessary for the intralysosomal catabolism of glucocerebroside (GC), and is inherited in an autosomal recessive fashion.[1,2] This results in accumulation of GC (most of which is derived from phagocytosed cell membranes) in the lysosomes of monocyte-derived macrophages in tissues of the reticuloendothelial system.[3,4] Accumulation in the Kupffer cells of the liver and in splenic macrophages is associated with enlargement of these organs. The resulting hypersplenism produces progressive anemia and thrombocytopenia. Accumulation of GC in bone marrow is associated with osteopenia, lytic lesions, pathologic fractures, chronic bone pain, acute episodes of excruciating "bone crisis," bone infarcts, and osteonecrosis. Although anemia and thrombocytopenia may be severe, it is usually bone disease that results in the greatest morbidity and long-term disability.



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