

ENZYME REPLACEMENT THERAPY IN PATIENTS WITH TYPE 1 GAUCHER DISEASE: A SINGLE-CENTER EXPERIENCE

Ivana Golubović^{1,2}, Miodrag Vučić^{1,3}, Irena Čojbašić^{1,3}, Milan Lazarević^{2,4}, Vesna Nikolić^{1,2}, Andrijana Pejčić^{1,2}

Gaucher disease (GD) is a liposomal storage disorder inherited in an autosomal recessive pattern. The underlying cause of the disease is a mutation within the gene that encodes the enzyme glucocerebrosidase. Accumulation of glucocerebrosidase in macrophages in the liver, spleen, bone marrow, rarely in the lungs and other organs, occurs due to a deficiency of enzyme synthesis, disorder or lack of enzyme function, or a deficiency of saposin C (enzyme activator). Clinical classification of GD is based on the absence (Type 1) or presence (Types 2 and 3) of central nervous system manifestations. Levels of beta-glucocerebrosidase in leukocytes, as well as the levels of serum chitotriosidase, are measured to make the definitive diagnosis of Gaucher disease. Accumulation of beta-glucocerebrosidase causes numerous multi-organ complications (anemia, thrombocytopenia, hepatomegaly, splenomegaly, skeletal and neurological changes). Since 1991, enzyme replacement therapy (ERT) has been used for treating Gaucher disease. Show the treatment results in patients with Type 1 Gaucher disease by administering ERT taliglucerase alfa at the Clinic of Hematology, Allergology and Clinical Immunology, University Clinical Center Niš. Between January 2016 and January 2025, taliglucerase alfa was used to treat 5 patients with Type 1 Gaucher disease who did not respond to previous treatment or because the drug donation was discontinued. All our patients responded well to treatment, and there were no adverse effects (administration of taliglucerase alfa results in significant regression of anemia, thrombocytopenia and organomegaly, along with bone status improvement).

Acta Medica Medianae 2025;64(4):80–86.

Key words: *Gaucher disease, enzyme replacement therapy, taliglucerase alfa*

¹University Clinical Center Niš, Clinic of Hematology, Allergology and Clinical Immunology, Niš, Serbia

²University of Niš, Faculty of Medicine, Doctoral Studies, Niš, Serbia

³University of Niš, Faculty of Medicine, Niš, Serbia

⁴University Clinical Center Niš, Clinic of Cardiovascular Surgery, Niš, Serbia