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

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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).

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

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Introduction

Gaucher disease—Morbus Gaucher (GD) is a liposomal storage disorder inherited in an autosomal-recessive pattern, with the underlying cause being mutations within the gene which encodes enzyme glucocerebrosidase (1–5).

The accumulation of glucocerebrosidase in macrophages occurs in the liver, spleen, bone marrow, and rarely in the lungs and other organs. It results from a deficiency in enzyme synthesis, a disorder or a lack of enzyme function, or a deficiency of saposin C, which acts as an enzyme activator. Accumulation of glucocerebrosidase causes numerous multi-organ complications such as anemia, thrombocytopenia, hepatomegaly, splenomegaly, and skeletal and neurological changes (1, 6–10).

Clinical classification of GD is based on the absence or presence of central nervous system manifestations, and it is differentiated into Type 1 (characterized by the absence of central nervous system manifestations) and Types 2 and 3 (characterized by the presence of central nervous system manifestations) (1, 10).

Type 1 GD is the most common, and clinical manifestations may vary from asymptomatic forms to forms with severe complications in childhood or adulthood. It is manifested with hepato-splenomegaly, leukopenia, thrombocytopenia, skeletal changes and pulmonary disease (1).

Type 2 is characterized as an acute and lethal neuronopathic form involving the nervous system, while Type 3 GD is chronic neuronopathic form involving visceral organs, bones and the heart.

Levels of beta-glucocerebrosidase in leukocytes are measured in order to make the definitive diagnosis of GD. However, a broad spectrum of pathological features observed in GD

is not merely a consequence of accumulation and mechanical activity of glucocerebrosidase, but a consequence of macrophage activation and cytokine secretion as well.

In the serum of patients with GD, the levels of interleukin-1 beta, interleukin-6, THF-alfa, soluble interleukin-2 receptor, and CD14 are elevated. What is specific for patients with GD is the elevation of chitotriosidase activity in serum originating from the Gaucher cells, and it is considered to be a marker of macrophage activation and immune response induction. Chitotriosidase levels serve as a surrogate marker for the total amount of accumulated glucocerbrosidase and for assessing Enzyme replacement therapy (ERT) intervention as well (1, 11–18).

The presence of the Gaucher cells in the bone marrow and other tissues is not pathognomonic of GD, but it can be seen in a number of other diseases (thalassemia, acute and chronic lymphoproliferative disease, granulocytic leukemia).

Since 1991, ERT has been used for treating Gaucher disease by substituting beta-glucocerebrosidase with the enzyme produced by recombinant technology (alglucerase, imiglucerase, velaglucerase, taliglucerase alfa). The therapy has been shown to achieve effective results, corrects the anaemia, thrombocytopenia, organomegaly, improves bone status, and adverse effects are scarce (10, 19–24).

Aim

The paper aimed to 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.

Materials and Methods

Between January 2016 and January 2025, at the Clinic of Hematology, Allergology and Clinical Immunology, taliglucerase alfa was used in treating 5 patients with Type 1 Gaucher disease who did not respond to previous treatment or because the drug donation was discontinued. One patient underwent splenectomy. Since 2016, the patients were treated at the expense of the National Health Insurance Fund.

ERT taliglucerase alfa was administered at a dose of 30 U/kg body weight every other week as a 60–120 minutes intravenous infusion. The dosage of taliglucerase alfa was adjusted on an individual basis to achieve adequate therapeutic response (absence of thrombocytopenia and anemia syndrome, reduced spleen and liver volume, as well as reduction in chitotriosidase levels and improvements in skeletal system) (20–23, 25, 26).

The treatment started after the definitive diagnosis of Type 1 Gaucher disease was made; diagnostic procedure included bone marrow biopsy, confirmation of the presence of Gaucher cells, assessment of chitotriosidase levels, measurement of spleen and liver volume and identification of genotypes based on PCR and direct gene sequencing, while the gold standard for making the diagnosis of Gaucher disease is to determine the beta-glucocerebrosidase levels in leukocytes (1, 11–20).

Results

ERT Taliglucerase alfa was used to treat 5 patients with type 1 Gaucher disease, 3 male patients (60%) and 2 female patients (40%) (Figure 1).

The mean age of all the patients at the time of initiating taliglucerase alfa therapy was 39.2 years. The youngest patient was 17, and the oldest one was 63 years old. Two patients previously received 1 therapeutic line (imiglucerase within the donation programme), one patient received 3 therapeutic lines (recombinant glucocerebrosidase within the clinical study, Protalix from humanitarian aid, imiglucerase), and two patients were treated with taliglucerase alfa in the first therapeutic line.

The time period between establishing GD diagnosis and initiation of ERT with taliglucerase alfa was 44.4 months (range 1–132) (Figure 2).

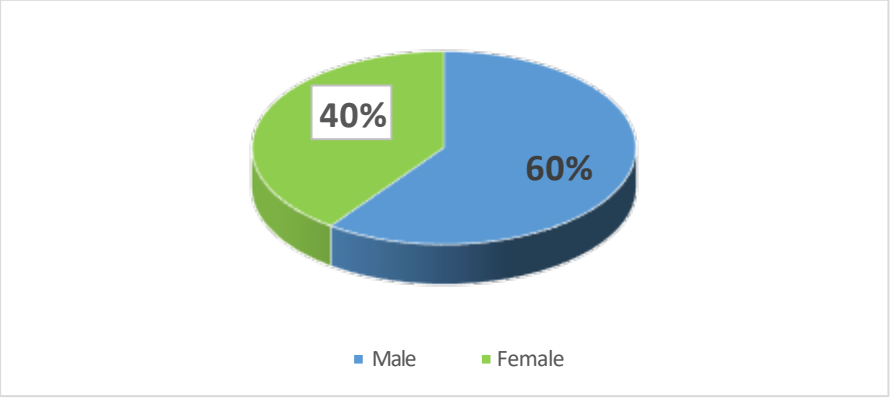

Figure 1. Patients with Gaucher disease by gender

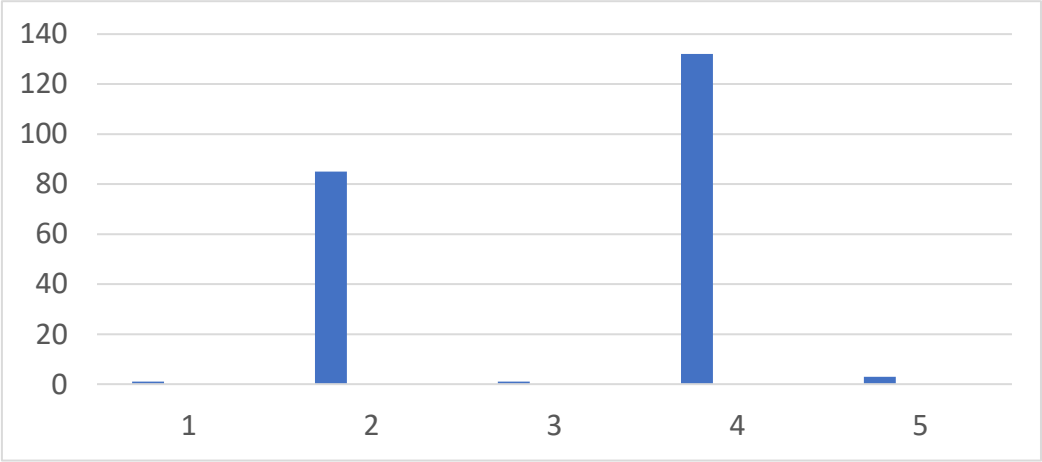

Figure 2. Time in months from diagnosis of GD to initiation of ERT

The time needed for treatment response after initiation of ERT taligluceraze alfa was 6–12 months.

The average duration of treatment with taliglucerase alfa was 61.6 months (range 13–83 months), and the average chitotriosidase level for that period was 3937.5 nmol/h/ml (range 481.4–8299.5 nmol/h/ml).

Chitotriosidase is an enzyme belonging to the family of chitinases that plays a role in innate immunity, and it is a marker of macrophage activation. Activated macrophages secrete chitotriosidase. Chitotriosidase activity level drops 3–6 months after initiation of ERT, and increases 20 weeks after ERT cessation.

Extremely elevated chitotriosidase activity level originating from the Gaucher cells is registered in the plasma of patients with GD. A direct correlation between chitotriosidase concentration and the total amount of non-degraded glucocerebroside substrate was established. That is why one of the parameters of treatment success is still serum chitotriosidase level.

The amount of glucocerebrosidase in patients with GD is increased 10–1.000 times and can make up to 2% body weight of the patient.

The analysis of chitotriosidase average value upon introducing ERT taliglucerase alfa showed the following trend: for the first patient, average chitotriosidase value was 9007.2 nmol/ml/h (range 4202–21226.3); for the second patient, the average chitotriosidase value was 4709.3 nmol/ml/h (range 2992.5–8299.5); for the third patient, the average chitotriosidase value was 683.3 nmol/ml/h (range 534–1137); for the fourth patient, the average chitotriosidase value was 970.7 nmol/ml/h (range 657.4–1443.6); and for the fifth patient, the average chitotriosidase value was 3402.8 nmol/ml/h (range 749.7–6056). Patients responded well to the therapy from its initiation throughout its course, except for two patients who responded at the level of stable disease, meaning that chitotriosidase level was maintained, as well as the volume of the liver and spleen in the previous two years. There were no adverse effects during the administration of the drug (Table 1).

Table 1. Effect of ERT taliglucerase alfa in patients with Gaucher disease

Patients with GD	Chitotriosidase value before ERT administration	Chitotriosidase value after EST application	Side effects
1.	21226.3 nmol/ml/h	4202 nmol/ml/h	No
2.	8229.5 nmol/ml/h	2992.5 nmol/ml/h	No
3.	1137 nmol/ml/h	534 nmol/ml/h	No
4.	1443.6 nmol/ml/h	657.4 nmol/ml/h	No
5.	6056 nmol/ml/h	749.7 nmol/ml/h	No

Apart from the assessment of chitotriosidase levels, our small group of patients was regularly radiologically monitored by performing whole-body NMR imaging once a year (measurements of spleen and liver volume, as well as regular monitoring of skeletal system status).

The most common clinical manifestation of Type 1 GD is painless splenomegaly, mostly detected accidentally. The spleen is sometimes palpated a few centimeters below the left rib arch, or it can be enlarged throughout the abdomen. The liver is slightly enlarged, and portal hypertension is rare.

Magnetic Resonance Imaging of the abdomen performed a year after the initiation of ERT taliglucerase alfa revealed a reduction in the spleen size in two patients: a reduction of 32%, from 201 mm to 136 mm, was registered in the first patient after a year of ERT taliglucerase alfa administration; in the second patient, the spleen volume was reduced from 134 mm to 130 mm. In one patient, the spleen was of regular size both at the time of diagnosis and at the check-up by abdominal NMR imaging. In another patient, the liver and spleen size were maintained despite the administered therapy. Additionally, one patient underwent splenectomy before establishing the diagnosis. The control magnetic resonance imaging was performed every year, aiming at detecting any changes in the liver and spleen volume, and the skeletal system as well.

The presence of anemia and thrombocytopenia is one of the criteria for establishing the diagnosis of GD. Our group of patients had regular laboratory analyses and follow-ups to assess the efficacy of ERT. At the time of diagnosis, two patients had severe anemia syndrome, two patients had moderate thrombo-cytopenia, and one patient had both anemia and thrombocytopenia.

The analysis of average number of platelets upon the introduction of ERT taliglucerase alfa showed the following: for the first patient, the average number of platelets was 122.8 x 10^9/L (range 84–149); for the second patient, the average number of platelets was 233.2 x 10^9/L (range 220–244); for the third patient, the average number of platelets was 149 x 10^9/L (range 125–180); for the fourth patient, the average number of platelets was 271.6x 10^9/L (range 210–330); and for the fifth patient, the average number of platelets was 115 x 10^9/L (range 62–209).

The values of Hgb levels monitored in patients upon ERT taliglucerase alfa introduction were as follows: for the first patient the average HGB value was 137.8 g/l (range 128–147), for the second patient, the average HGB value was 152.4 g/l (range 149–157), for the third patient, the average HGB value was 95.2 g/l (range 81–106), for the fourth patient, the average HGB value was 131.6 g/l (range 109–145), and for the fifth patient, the average HGB value was 140 g/l (range 135–148).

For the assessment of bone involvement, pelvic MRI and long bones MRI were performed. One of the important features of type 1 Gaucher disease is skeletal system involvement, as registered in 60 % of our small group of patients. Skeletal changes are not always in direct correlation with other symptoms; bone involvement is not rare, and it is often the first presenting sign of the disease. Bone involvement most commonly includes the thighbone, vertebrae, pelvis, the upper arm and forearm long bones with the development of osteoporosis, osteolysis, avascular necrosis, and pathological fractures. Episodes of bone crises are manifested as intense pain and are caused by bone microinfarction. At the time of establishing the diagnosis, in 3 out of 5 monitored patients, signal intensity changes in the bones were registered, and the regression of manifestations was observed after the administration ERT Taliglucerase alfa.

Discussion

Gaucher disease is an autosomal recessive disease, more common in females and more prevalent in Ashkenazi Jews than in the general population. At the time of diagnosis, patients' age ranges from birth to 81 years, mean age of 17.4 years, with almost half of the patients diagnosed before 10 years of age. In the current study small group, the mean age was 39.2 years (27–29).

The purpose of treating Gaucher disease is correction of anemia syndrome, thrombocytopenia, and reduction of liver and spleen volume, along with the improvement of bone status. The effects of ERT can be seen 3 to 6 months after the initiation of the treatment, while in this study group of patients, the effects were seen after 6 to 12 months of treatment, which is in accordance with literature data (18, 19, 20–23, 25, 26).

Available literature does not favour any type of ERT, but according to current experience, taliglucerase alfa exhibits lower risk of immunogenicity in comparison to imiglucerase. ERT taliglucerase alfa is well tolerated, and adverse effects are mild to moderate in intensity and transient. The most common adverse effects include headache, nasopharyngitis, hypertension, chest discomfort, nausea, vomiting, itching, and pain in extremities, although no adverse effects were registered in our small group (24, 29, 30).

Getting IV ERT treatment every other week in a hospital environment harms patients' quality of life, because it is a lifelong treatment (31).

All patients included in the present study had a good therapeutic response, except for one patient in whom there was no reduction in the liver and spleen volume over a period of over a year, and treatment modality change has been considered. Taliglucerase alfa dosage was increased in two patients in proportion to their body size. According to literature data, termination of ERT results in disease progression in most patients, as well as in chitotriosidase level increase 20 weeks after treatment discontinuation (22, 23).

Conclusion

Until the 1990s, the only treatment available for Gaucher disease was a symptomatic one, such as erythrocyte transfusion, transfusion of platelets, multivitamin infusions, NSAIDs, androgen and bisphosphonate treatment, and orthopaedic procedures. Splenectomy was accompanied by transient improvement only, and the disease was still progressing, especially in accelerating skeletal structures damage.

A significant advancement in GD treatment was the introduction of replacement therapy with alglucerase (the placenta-derived modified form of beta-glucocerebrosidase) in the 1990s. A recombinant form of alglucerase—imiglucerase, Cerezyme—was introduced as a treatment option in the late 1990s.

Based on previous experience, administration of taliglucerase alfa results in a significant reduction of anemia, thrombocytopenia and organomegaly, as well as in bone status improvement.

Gaucher disease is an example of how modern medicine may alter the course of human life.

References

1. Suvajdžić-Vuković N. Gošeova bolest tipa 1- Klinička slika, hematološki aspekti i supstituciona terapija. Bil Hematol 2004; 32:165-8.
2. Zhao H, Grabowski GA. Gaucher disease: Perspectives on a prototype lyposomal disease. Cell Mol Life Sci 2002; 59(4):694-707. [CrossRef] [PubMed]
3. Beutler E. Lysosomal storage diseases:natural history and ethical and economic aspects. Mol Genet Metab 2006;88(3): 208-15. [CrossRef] [PubMed]
4. Redzić A, Begić F. [Type I Gaucher's disease- a rare genetic metabolic disease]. Med Arh 2003; 57(3):173-6.
5. Beutler E, Gelbart T, Scott CR. Hematologically important mutations:Gaucher disease. Blood Cells Mol Dis 2005;35(3):355-64. [CrossRef] [PubMed]
6. Dokić M. Morbus Gaucher- a report of two cases. Vojnosanit Pregl 2006 ;63(12):1039-44. [CrossRef] [PubMed]
7. Wenstrup RJ, Roca-Espiau M, Weinreb NJ, Bembi B. Skeletal aspects of Gaucher disease: a review. Br J Radiol 2002;75 Suppl 1: A2-12. [CrossRef] [PubMed]
8. Stowens DW, Teitelbaum SI, Kahn AJ, Barranger JA. Skeletal complications of Gaucher disease. Medicine (Baltimore) 1985; 64:310-22. [CrossRef] [PubMed]
9. Maas M, Poll LW, Terk MR. Imaging and quantifying skeletal involvement in Gaucher disease. Br J Radiol 2002; 75(suppl 1): A13-A24. [CrossRef] [PubMed]
10. Mrsić M. [Diagnosis and treatment of Gaucher disease in Croatia]. Lijec Vjesn. 2007 May;129 Suppl 3:38-42.
11. Pandey MK, Grabowski GA. Immunological cells and functions in Gaucher disease. Crit Rev Oncog 2013;18(3):197-220. [CrossRef] [PubMed]
12. Deegan PB, Moran MT, McFarlane I, Schofield JP, Boot RG, Aerts JM, et al. Clinical evaluation of chemokine and enzymatic biomarkers of Gaucher disease. Blood Cells Mol Dis 2005; 35:259-67. [CrossRef] [PubMed]
13. Manolagas SC. The role of IL-6 type cytokines and their receptors in bone. Ann NY Acad Sci 1998;840:194-204. [CrossRef] [PubMed]
14. Barak V, Acker M, Nisman B, Kalickman I, Abrahamov A, Zimran A, et al. Cytokines in Gaucher's disease. Eur Cytokine Netw 1999;10(2):205-10.
15. Cox TM. Biomarkers in lysosomal storage diseases: a review. Acta Paediatr Suppl 2005;94(447):39-42. [CrossRef] [PubMed]
16. Aerts JM, Hollak CE, van Breemen M, Maas M, Groener JE, Boot RG. Identification and use of biomarkers in Gaucher disease and other lysosomal storage diseases. Acta Paediatr Suppl 2005;94(447):43-6. [CrossRef] [PubMed]
17. Korolenko TA, Zhanaeva SY, Falameeva OV, Kaledin VI, Filyushina EE, Buzueva II, et al. Chitotriosidase as a marker of macrophage stimulation. Bull Exp Biol Med 2000;130: 948-50. [CrossRef] [PubMed]
18. Poll IW, Maas M, Terk MR, Roca-Espiau M, Bembi B, Ciana G, et al. Response of Gaucher bone disease to enzyme replacement therapy. Br J Radiol 2002;75 Suppl 1:A25-36. [CrossRef] [PubMed]
19. Grabowski GA, Hopkin RJ. Enzyme therapy for lysosomal storage disease: principles, practice and

prospects. Annu Rev Genomics Hum Genet 2003; 4:403-36. [CrossRef] [PubMed]
20. Brady RO. Enzyme replacement for lysosomal diseases. Annu Rev Med 2006; 57:283-96. [CrossRef] [PubMed]
21. Zimran A. How I treat Gaucher disease. Blood 2011; 118(6):1463-71. [CrossRef] [PubMed]
22. Zimran A, Wajnrajch M, Hernandez B, Pastores GM. Taliglucerase alfa: safety and efficacy across 6 clinical studies in adults and children with Gaucher disease. Orphanet J Rare Dis 2018;13(1):36. [CrossRef] [PubMed]
23. Pastores GM, Shankar SP, Petakov M, Giraldo P, Rosenbaum H, Amato DJ, et al. Enzyme replacement therapy with taliglucerase alfa: 36-month safety and efficacy results in adult patients with Gaucher disease previously treated with imiglucerase. Am J Hematol 2016 Jul;91(7):661-5. [CrossRef] [PubMed]
24. Hollak CE. An evidence-based review of the potential benefits of taliglucerase alfa in the treatment of patients with Gaucher disease. Core Evid 2012;7:15-20. [CrossRef] [PubMed]
25. Charrow J, Andersson HC, Kaplan P, et al. The Gaucher Registry: Demographics and Disease Characteristics od 1698 Patients With Gaucher Disease. Arch Intern Med 2000; 160(18):2835-2843. [CrossRef] [PubMed]
26. Zimran A, Brill-Almon E, Chertkoff R, Petakov M, Blanco-Favela F, Muñoz ET, et al. Pivotal trial with plant cell-expressed recombinant glucocerebrosidase, taliglucerase alfa, a novel enzyme replacement therapy for Gaucher disease. Blood 2011;118(22):5767-73. [CrossRef] [PubMed]
27. Shemesh E, Deroma L, Bembi B, Deegan P, Hollak C, Weinreb NJ, et al. Enzyme replacement and substrate reduction therapy for Gaucher disease. Cochrane Database Syst Rev 2015 Mar 27;2015(3):CD010324. [CrossRef] [PubMed]
28. Nalysnyk L, Rotella P, Simeone JC, et al. Gaucher disease epidemiology and natural history: a comprehensive review of the literature. Hematology 2017;22(2):755–73. [CrossRef] [PubMed]
29. Zimran A, Durán G, Mehta A, Giraldo P, Rosenbaum H, Giona F, et al. Long-term efficacy and safety results of taliglucerase alfa up to 36 months in adult treatment-naïve patients with Gaucher disease. Am J Hematol 2016;91(7):656-60. [CrossRef] [PubMed]
30. Van Rossum A, Holsopple M. Enzyme Replacement or Substrate Reduction? A Review of Gaucher Disease Treatment Options. Hosp Pharm 2016;51(7):553-63. [CrossRef] [PubMed]
31. Revel-Vilk S, Mansfield R, Feder-Krengel N, Machtiger-Azoulay N, Kuter D, Szer J, et al. Real-World Experiences with Taliglucerase Alfa Home Infusions for Patients with Gaucher Disease: A Global Cohort Study. J Clin Med 2023;12(18):5913. [CrossRef] [PubMed]

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PRIMENA ENZIMSKE SUPSTITUCIONE TERAPIJE KOD PACIJENATA SA GOŠEOVOM BOLESTI TIPA 1: ISKUSTVA JEDNOG CENTRA

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Gošeova (*Gaucher*) bolest (GB) jeste lipozomalna bolest nakupljanja koja se nasleđuje autozomno recesivno, a u njenoj osnovi je mutacija gena koji kodira enzim glukocerebrozidazu. Do nagomilavanja glukocerebrozida u makrofazima jetre, slezine, koštane srži, a ređe u plućima i u drugim organima, dolazi usled smanjene sinteze enzima, nedostatka ili poremećaja funkcije enzima ili deficita sapozina C (aktivatora enzima). Klinička podela GB-a zasniva se na odsustvu (tip 1) ili prisustvu (tip 2 i tip 3) manifestacija u centralnom nervnom sistemu. Nivo β-glukozocerebrozidaze u leukocitima i vrednost hitotriozidaze u serumu određuju se radi postavljanja definitivne dijagnoze Gošeove bolesti. Nagomilavanje glukocerebrozida izazviva komplikacije u većem broju organa (anemiju, trombocitopeniju, hepatomegaliju, splenomegaliju, skeletne i neurološke promene). Za lečenje Gošeove bolesti od 1991. godine koristi se enzimska supstituciona terapija. Cilj ovog rada bio je da predstavi rezultate lečenja pacijenata sa Gošeovom bolesti tipa 1 na Klinici za hematologiju, alergologiju i kliničku imunologiju Univerzitetskog kliničkog centra Niš primenom taligluceraze alfa kao enzimske supstitucione terapije. Od januara 2016. godine do januara 2025. godine primenom taligluceraze alfa lečeno je pet pacijenata sa Gošeovom bolešću tipa 1. Kod ovih pacijenata nije bilo odgovora na prethodnu terapiju ili je pak donacija leka bila obustavljena. Svi ovi pacijenti dobro su reagovali na terapiju i nije bilo neželjenih efekata. Zapaženo je da primena taligluceraze alfa dovodi do značajnog smanjenja anemije, trombocitopenije i organomegalije, kao i do poboljšanja statusa kostiju.

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***Ključne reči**: Gošeova bolest, enzimska supstituciona terapija, taligluceraza alfa*