CURRENT FACTS IN THE TREATMENT OF GAUCHER DISEASE

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Background. Gaucher disease (GD) is a rare metabolic disease with autosomal recessive transmission, caused by the mutation of the GBA gene, which causes a deficient synthesis of the enzyme β-glucocerebrosidase. As result, glucocerebrosides is accumulated throughout the body, especially in the bone marrow, spleen and liver. Three forms of Gaucher disease have been identified, distinguished by the absence or presence and extent of neurological complications. Aim of the study is to evaluate the current therapies for the treatment of Gaucher disease.
Material and methods. Narrative synthesis of specialized literature from scientific databases: *PubMed, Gene Cards, National library of medicine, Google Scholar* and *Hinari* of the last 10 years.
Results. Enzyme replacement therapy is an effective way to treat Type 1 Gaucher disease. Treatment is done via infusion of imiglucerase – a synthetic glucocerebrosidase, to ensure the breakdown of accumulated lipids. Substrate reduction therapy uses a small molecule drug miglustat and eliglustat tartrate that inhibits the first committed step in glycosphingolipid biosynthesis. Chaperone therapy with non-inhibitory chemical chaperones can increase glucocerebrosidase levels and activity in lysosomes. Gene therapy as a potential therapeutic approach for treatment of GD type 1. Ex vivo autologous bone-marrow-derived GD 1 hematopoietic stem cells were genetically corrected by infection with self-inactivating lentiviral vectors expressing WT GBA1 induced by different cellular promoters. Hematopoietic stem cell transplantation, involving the replacement of affected stem cells with healthy stem cells is a treatment that can provide a permanent source of enzyme to people with Gaucher disease and is a considerably less expensive procedure. People with Type 3 Gaucher disease showed no further neurological deterioration. The important limitations of HSCT are the mortality and morbidity associated with the procedure and the non-availability of HLA matched donors.
Conclusions. The treatment of Gaucher disease is a subject under constant research, and research advances offer promising improvement in the life quality of patients with GD. Treatment should be personalized according to the severity of the disease and other associated medical conditions. Enzyme replacement therapy remains the most widely used and well tolerated form of treatment, and patients should be carefully supervised to prevent any unexpected complications.
Keywords: enzyme replacement therapy, Gaucher disease, glucocerebrosidase, lysosomal disease, GBA gene.