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# GAUCHER DISEASE TYPE 1 AND GASTRIC CANCER: A CASE REPORT

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Gaucher disease (GD is a liposomal storage disease that is inherited in an autosomal recessive manner. The basis of the disease is a mutation of the gene that codes for the enzyme glucocerebrosidase. The clinical division of GB into type 1, 2 and 3 is based on the absence (type 1) or presence (type 2 and 3) of manifestations by the central nervous system. In order to establish a definitive diagnosis, the level of β-glucose cerebrosidase in leukocytes and the value of chitotriosidase in the serum are determined. Genotype analysis is helpful in assessing the type and severity of the disease. Since 1991, Gaucher disease has been treated with enzyme replacement therapy (EST). We present the clinical characteristics of a patient with type 1 Gaucher disease diagnosed in November 2004 in the Hematology Clinic, UKC of Serbia. The patient was a heterozygous carrier of the N307S mutation. In February 2006, treatment was started with imiglucerase (Cerezyme$^{R}$) IV at a dose of 30 U/kg body weight every two weeks. After 24 months of imiglucerase therapy, a significant improvement in the patient's condition was registered, but she complained of nausea, an urge to vomit and pain in the epigastrium. MSCT of the upper abdomen was performed, and esophagogastroduodenoscopy with a biopsy of changes in the stomach. Pathohistological findings of biopsied changes in the stomach indicated the existence of gastric adenocarcinoma. A total gastrectomy with splenectomy and cholecystectomy was performed. PH finding was adenocarcinoma ventriculi intramucosum (early cancer). After the surgical intervention, the patient continued enzyme replacement therapy with imiglucerase. Patients with GD have an increased risk of developing malignant diseases, most often lymphoproliferative, although solid tumors (hepatocellular carcinoma) have also been described. In our case, to the best of our knowledge, the association of Gaucher disease with gastric cancer has been rarely reported in the literature.

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***Key words:*** *Gaucher disease, enzyme replacement therapy, imiglucerase, gastric cancer*