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USE OF SUBSTITUTE ENZYMATIC TREATMENT AND SUBSTRATE REDUCTION IN GAUCHER DISEASE

González Rosa V, Sierra Torres MI, Fernández Espínola S, Zaragoza Rascón M, Pajares Alonso M
Servicio de Farmacia, Hospital Serranía de Ronda (Málaga)

Background

Gaucher disease is included within lipidosis that occur due to mutations in the gene encoding the enzyme ß-glucosidase. As a result of this accumulates a fatty substance, the glucocerebroside that is the cause of disease manifestations such as anemia, thrombocytopenia, hepatosplenomegaly and bone injuries. Available therapeutic options include enzyme replacement (ERT) or substrate reduction therapy (SRT) to prevent glucocerebroside accumulation.

Purpose

To describe the use of ERT and SRT in patients with Gaucher disease

Material and methods

Retrospective observational study of all patients diagnosed with Gaucher disease in our area, followed up in our hospital and in treatment with ERT or SRT. Data from the clinical history in DIRAYA and the corresponding analytics in the laboratory application were reviewed. The analyzed variables were sex, age, value of chitotriosidase, symptoms of Gaucher disease and adverse reactions to treatment.

Results

A total of 4 patients (2 men and 2 women) with an average age of 50 years were included. All patients had type 1 Gaucher disease (not neuropathic). The initial treatment was miglustat (SRT) in 3 patients and velaglucerase (ERT) in one of them.The value of chitotriosidase before the start of treatment had a mean value of 11034 nmol/h/mL (7184-12777) and after treatment it was 1536 nmol/h/mL (239-3973) (Table 1) All the patients presented at the beginning typical manifestations of type 1 Gaucher disease (Table 2)

	Value chitotriosidase (nmol/h/mL)	
Patient	Before treatment	After treatment
1	7184	1172
2	12777	750
3	12090	4089
4	12084	3973

Table 1

Adverse Reaction	Number of patients
Bone affectation	3
Hepatosplenomegaly	3
Anemia	2
Thrombopenia	2

Table 2

Regarding the safety of the SRT, treatment with miglustat was started in 3 patients. It was finished in two cases due to bone progression and in one case due to poor tolerance (paresthesia, diarrhea, tremor and weight loss) and the TES was switched to imiglucerase or velaglucerase, which were well tolerated in all patients. All the patients presented improvement in the symptoms of Gaucher disease when starting ERT.

Conclusions

TRS with miglustat is a convenient option due to its oral administration, although in 3 patients who were initially administered it had to be suspended due to poor tolerance or progression. ERT has been shown to be effective and safe, and, despite not being curative, an improvement and even remission of certain symptoms of the disease has been proven.

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