COURSE OF A PATIENT DURING IMIGLUCERASE SHORTAGE.

APROPOS OF A CASE

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BACKGROUND

Gaucher disease is a hereditary metabolic disorder characterized by deficiency of the lysosomal enzyme beta-glucocerebrosidase which catalyzes the hydrolysis of glucosylceramide to glucose and ceramide. This causes accumulation of glucosylceramide within the lysosomes with systemic manifestations consisting of bone and hematological abnormalities and visceromegaly.

PURPOSE

To describe the course of a patient diagnosed with Gaucher disease in whom the treatment she was receiving with imiglucerase was replaced by miglustat, an oral drug also indicated for Gaucher disease.

MATERIALS AND METHODS

Review of patient history and recording of biomarkers of the disease: Chitotriosidase activity (nM/mL.h) and CCL-18 PARC concentration (ng/mL). Blood count data were collected: platelets, hemoglobin, and white blood cells

RESULTS

Miglustat treatment was between November 2009 and March 2010, reintroducing imiglucerase in April 2010. Before starting miglustat the patient had the following values: chitotriosidase 1432 nM/mL.h and CCL-18 PARC of 367ng/mL, 127000 platelets/µL, hemoglobin 13.4 g/dL and 4200 WBC/µL with good general condition. In March 2010, laboratory tests were: chitotriosidase 2546 nM/mL.h and CCL-18 PARC of 561 ng/mL, 113000 platelets/µL, hemoglobin 12.4 g/dL and 4400 WBCs/µL.

During this period, the patient did not worsen clinically but showed tremor, flatulence, and mild diarrhea during treatment with miglustat.

CONCLUSIONS

The increase in markers after switching shows disease worsening. Switching to the oral route may seem an improvement in quality of life.

*Conflict of interest: nothing to disclose