Clinical Genetics

AB029. Rare condition of hepatic Gaucheroma in a type I Gaucher patient with enzyme replacement therapy

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Background: Gaucher disease is a lysosomal storage disorder, characterized by hepatosplenomegaly, pancytopenia, and neurological manifestation. Herein, we present a 3-year-old boy with type I Gaucher disease who had been treated with enzyme replacement therapy (ERT), and subsequently developed a focal Gaucheroma in the liver after 19 months of ERT.

Methods: Medical records and laboratory data were reviewed.

Results: The patient first presented at 15 months of age with anemia, thrombocytopenia, and hepatosplenomegaly. Gaucher disease was confirmed by leukocyte enzyme assay and *GBA* gene mutation test. ERT was administered after diagnosis. At the age of 3 years, a lobulated mass was discovered on regular MRI follow-up of abdomen. Biopsy and histological examination of the mass disclosed Gaucheroma.

Conclusions: For type I Gaucher disease, ERT should be started as soon as possible. The age at starting treatment might be the critical factor of long-term prognosis. Gaucheroma is a rare condition found in patients receiving ERT. A regular imaging study is necessary for type I Gaucher patients. A thorough survey should be done in regards to malignancy risk.

Keywords: Gaucher disease; Gaucheroma; enzyme replacement therapy (ERT)

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