Complex Genetic Disorders, Genetic Susceptibility to Infections

AB120. Cholelithiasis in a Filipino child with chronic neuronopathic Gaucher disease: report of a case

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Background: Gaucher disease is the most common lysosomal storage disease. It is caused by a defect in the lysosomal enzyme β-glucocerebrosidase, resulting in multi-organ involvement. The presence of cholelithiasis has been rarely observed among patients with non-neuronopathic type of Gaucher disease and the exact pathophysiology is still unknown.

Methods: Clinical, laboratory and radiological data of a Filipino child with chronic neuronopathic Gaucher disease were reviewed.

Results: The patient was accidentally noted to have cholelithiasis on routine whole abdominal ultrasonography as part of the regular monitoring of the disease.

Conclusions: Cholelithiasis can be found in both neuronopathic and non neuronopathic Gaucher disease.

Keywords: Cholelithiasis; Gaucher disease

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