

عنوان مقاله:

Report of Four Children with Gaucher Disease and Review of Literature

محل انتشار:

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خلاصه مقاله:

Gaucher Disease (GD) is the most common type of Lysosomal Storage Disorder and it is divided into three distinct subtypes. The authors here report four different cases of Gaucher Disease, with varying clinical manifestations, and the diagnosis of each established by the low level of Beta-Glucosidase enzyme as well as genetic DNA testing. The study also highlights the importance of early diagnosis of the disease in order to initiate the appropriate therapeutic management to help prevent further progression of the disease.

کلمات کلیدی:

Children, case report, Gaucher disease, Lysosomal storage diseases

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