Plasma Lipid Profiling of Gaucher Disease: Biochemical Markers to Evaluate Therapeutic Intervention.

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Gaucher disease is a lysosomal storage disorder characterised by a deficiency of the enzyme acid -glucosidase. Clinical symptoms include hepatosplenomegaly, haematological involvement and bone lesions. Neurological impairment may also occur in certain patients. The advances in the treatment of Gaucher disease have highlighted the need to monitor the complex biochemical changes associated with the disease and the response of these changes to therapy. In this study the suitability of plasma lipids as biochemical markers of Gaucher disease was evaluated. Electrospray ionisation-tandem mass spectrometry was used to characterise and quantify plasma sphingolipids and phospholipids from Gaucher and control patients. Molecular species of glucosylceramide and G₃M3 ganglioside were elevated in Gaucher disease, whereas species of dihexosylceramide and sphingomyelin were decreased. This strategy was further employed to assess the response of patients to enzyme replacement therapy. The findings suggest that plasma lipid profiling may be a suitable strategy to monitor the efficacy of therapies for Gaucher disease.