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Minimal disease activity in Gaucher disease: criteria for definition.

Di Rocco M[1], Andria G, Bembi B, Carubbi F, Giona F, Giuffrida G, Linari S, Sibilio M, Spina V, Cappellini MD.

Author information

Abstract

Gaucher disease type I is a metabolic disorder caused by a genetic deficiency of lysosomal β-glucocerebrosidase that leads to accumulation of glucocerebroside in macrophages, thus causing damage in different organ systems. Enzyme replacement therapy with imiglucerase improves organ impairment and clinical manifestations, but patients differ in response to treatment. While clinical remission is the most desirable therapeutic outcome, a more realistic goal in patients with high disease burden is reasonably good clinical status despite persistence of residual biochemical or imaging abnormalities. Therefore, the concept of minimal disease activity--used in certain haematological or rheumatologic conditions--needs to be introduced in Gaucher disease, with a level of disease activity that patients and physicians consider a useful treatment target. In this paper, we propose specific parameters and criteria for defining minimal disease activity in Gaucher disease and its stability over time, based on three major systemic domains typically involved: haematological, visceral, and skeletal. Biomarker parameters were not included as criteria, because currently they do not adequately reflect disease evolution in individual patients. Neurological and respiratory domains were also excluded, as their involvement per se indicates severe disease unlikely to respond to enzyme replacement therapy and achieve minimal disease status. Our goal in defining minimal disease activity and stability is to identify a tool to facilitate treatment decisions in clinical practice.

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