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Validity of β -d-glucosidase activity measured in dried blood samples for detection of potential Gaucher disease patients.

[Stroppiano M](#)¹, [Calevo MG](#)², [Corsolini F](#)¹, [Cassanello M](#)³, [Cassinero E](#)⁴, [Lanza F](#)¹, [Stroppiana G](#)¹, [Cappellini MD](#)⁵, [Filocamo M](#)⁶.

Author information

Abstract

OBJECTIVES: Gaucher disease (GD) diagnosis relies on the demonstration of deficient β -d-glucosidase (GBA) activity in cellular homogenates. Diagnosis process, however, can be delayed as (i) some GD symptoms are non-specific; and (ii) diagnostic tests are performed in specialized laboratories. These difficulties negatively impact on timely access of patients to therapy. GBA assay in dried blood spots (DBS) represents a method facilitating early identification of patients who will be finally diagnosed with gold standard assay of nucleated cells. Aim of this study is to investigate the DBS analytical performance compared with gold standard method.

DESIGN & METHODS: A cross-sectional study started by comparing data of 50 DBS and 50 homogenate samples from the same subjects (25 known-GD and 25 controls). The subsequent phase examined 443 DBS samples. Along with these, 73 blood samples were sent for leukocyte separation and/or EBV-lymphoblast cell lines, and 1 skin biopsy for fibroblast cell lines. Overall the study included a total of 493 subjects.

RESULTS: While the results from this first validation group did not yield false positive/negative values, when the analysis was extended to 443 DBS, 14.4% (64 samples) of positive results was yielded. Among these, only 15 were confirmed as GD values with gold standard test. In addition, a thorough examination of some clinical data also revealed 2 false negative results which were confirmed by both enzymatic and molecular analyses.

CONCLUSIONS: DBS test could be useful as screening method although with cautions, whereas the standardized GBA assay should remain the gold standard for laboratory diagnosis of Gaucher disease.

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KEYWORDS: Dried blood spot; False negative; False positive; Gaucher; Screening; β -d-glucosidase assay

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