Reviewer's report

Title: Polymorphisms in the glucocerebrosidase gene and pseudogene urge caution in clinical analysis of Gaucher disease allele L444P (1448C)

Version: 2 Date: 4 May 2006

Reviewer: Bruno Bembi

Reviewer's report:

General
Authors report the usefulness of long amplicons for Gaucher allele L444P in a multiplexed suspension array assay and provide further recommendations for accurate discrimination between GBA gene and pseudogene. However there are some points that can be improved with further discussion:
1. Gaucher disease type 1 is highly prevalent in Ashkenazi Jewish population, being the N370S the most frequent mutant allele (almost 75%). Therefore, why did the authors choose the L444P for the screening of Gaucher disease?
2. Up to date, there are several screening panels for Ashkenazi Jews (Genet Med. 2005 7(3):185-90). Authors should discuss the advantages and disadvantages of the test presented here, at least regarding Gaucher disease evaluation.

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

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Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions

Level of interest: An article of limited interest

Quality of written English: Acceptable

Statistical review: No