Reviewer's report

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

Version: Date: 20 June 2006

Reviewer: Renzo Guerrini

Reviewer's report:

General

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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)

The authors evaluated a multiplexed suspension array assay to characterise alleles covering 8 diseases highly prevalent in the Ashkenazi population. In particular, the study is focused on wild-type and mutant genetic determinations for Gaucher disease allele L444P. This assay reduces the false positive frequency of the L444P allele in the Gaucher disease.

The paper is well written and the molecular experiments are fully described and supported by tables and literature references. However, the article is too long; the initial parts of the introduction and discussion are redundant and should be considerably reduced.

Minor comments:
- In the abstract background the authors should specify what are the 8 prevalent diseases in the Ashkenazi population.
- A standard nomenclature for mutations should be used (i.e. 1448T®C should be c.1448T>C)

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Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions