Reviewer's report

Title: Beta-defensin genomic copy number is not a modifier locus for cystic fibrosis.

Version: 1 Date: 1 June 2005

Reviewer number: 1

Reviewer’s report:

General
This paper is clear and well written. The authors address the question as to whether copy number of defensin as determined in a large cohort of CF patients relates to severity of lung disease. No significant association is found. The results seem very clear but initial date of colonisation was not recorded and this could be a significant parameter.

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)
This paper would be greatly improved by determining expresssion levels of the gene product. This could be done from a few individuals taken from each cohort to see whether copy number related to expression levels. The conclusion currently could be that increased expression of DEFB4 is not a major modifier for lung disease in CF, or it may be that increased copy number does not predict disease status. Until expression levels are examined the two cannot be separated. Commercial antibodies are available for DEFB4/2 so it wouldn't be too hard to do.

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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)
This paper would be greatly improved by determining expression levels of the gene product. This could be done from a few individuals taken from each cohort to see whether copy number related to expression levels.

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Discretionary Revisions (which the author can choose to ignore)
This paper would be greatly improved by determining expression levels of the gene product. This could be done from a few individuals taken from each cohort to see whether copy number related to expression levels.

What next?: Unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: Yes, but I do not feel adequately qualified to assess the statistics.