Author’s response to reviews

Title: Molecular Analysis Using DHPLC of Cystic Fibrosis: Increase of the Mutation Detection Rate Among the affected Population in Central Italy.

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Dear Prof. Matthew Hodgkinson,

Thank you very much for your letter of January 23, 2004 concerning our paper “Molecular Analysis Using DHPLC of Cystic Fibrosis: Increase of the Mutation Detection Rate Among the affected Population in Central Italy”.

We appreciated the Reviewer’s comments. The manuscript has been amended accordingly. In particular, we performed the following changes:

Reviewer: Paolo Arosio

- We changed the title as suggested;
- We described briefly the class of CFTR mutations in the Background Section;
- The sentence regarding the CF frequency has been modified as suggested;
- Nucleotide substitutions as well as amino acid mutations were included to describe the variant detected;
- The exact figure of samples ran on DHPLC was indicated;
- The sentence concerning previous Reports using this DHPLC technique has been modified as suggested;
- Table 2 has been in part modified as suggested. Now, it include the nucleotide substitution, and the detection system used. We think that the inclusion in this Table of polymorphisms, adds further difficulties in interpretation, therefore this suggestion was not accepted;
- The discussion has been improved and new sentences were added.
Reviewer: Claude Ferec

- The incidence of the disease was modified;
- The size of the gene has been corrected in the text;
- The nomenclature of the major CF mutation has been changed;
- The introduction has been modified as suggested;
- Previous reports are now quoted appropriately;

I hope that this revised version meets your approval for its publication in BMC Medical Genetics as Technical Advance article

Yours sincerely

Maria Rosaria D’Apice