Author's response to reviews

Title: Analysis of cystic fibrosis gene mutations in children with Cystic Fibrosis and in 964 infertile couples, first report from the Region of Basilicata, Italy.

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Version: 6 Date: 28 July 2014

Author's response to reviews: see over
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Author's response to reviews: see over
Juli26, 2014

The Biomed Central Editorial Team

Object: MS: 5853324011258451 - Analysis of cystic fibrosis gene mutations in children with Cystic Fibrosis and in 964 infertile couples, first report from the Region of Basilicata, Italy. Dr. Domenico Dell'Edera et al.

Thank you for consideration of our manuscript for publication in your journal.

We have reviewed the above manuscript according to your reviewer’s comments.

Reviewer # 1 (sherif shehata)

**Question number 1:**
Do they think to make DNA SEQUENCING FOR ALL CASES (even the positive reverse dot blot) will be an additional proof as could differentiate the innocent polymorphism cases.

Answer number 1:
The DNA sequencing is not essential, since the detection of innocent polymorphisms is not important to control the disease.

**Question number 2**
there is many descrepancies to the former study in table 5 when compared without explanation

Answer number 2:
Castaldo’s study refers to few mutations found in the CFTR gene and in a small number of patients with cystic fibrosis. Our study was performed on patients with cystic fibrosis and patients came to our attention in order to IVF cycles.

**Question number 3**
figure 1 is very complex

**Question number 4**
despite that the aim of the study to establish priority risk of CF carriers, the results of the study is lacking that evidence.

Answer number 4:
Our pilot study has the purpose of understanding the frequency of mutations found in the population we studied, so as to bring out the most frequent ones. The knowledge of the frequency of prevalent mutations in CFTR genes in the province of Matera allows development of kits for their detection.
Answer number 5:
Grammar has been revised

Reviewer #2 (Sameh Shehata)
The reviewer number 2 considered the work interesting and did not any questions.