Author's response to reviews

Title: SNPs and real-time quantitative PCR method for constitutional allelic copy number determination, the VPREB1 marker case

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Author's response to reviews: see over
To Editor-in-Chief
BMC Medical Genetics

San Donato Milanese, January 17, 2011

We are submitting the revised version R1 of the manuscript entitled “SNPs and real-time quantitative PCR method for constitutional allelic copy number determination, the VPREB1 qPCR marker case” by Frigerio M et al, to BMC Medical Genetics as Original Article.

We thank the Editor and Reviewers to give us the opportunity to revise our manuscript. It has been copyedited for English grammar and style and it has been revised and changed according to the Reviewers suggestions. Here the details of the answers to Reviewers Comments follow:

To Reviewer 1, Ester Ramos:
1. We agree with Ester Ramos that data presented in the present manuscript are not consistent with the aim of the study we declared. Indeed, though our investigation began with the purpose to validate qPCR methods for 22q11.2 microdeletion detection, in the present paper submitted to BMC Medical Genetics we would like to discuss the importance of primer design for Real-Time PCR presenting our experience with Weksberg PCR method for the screening of 22q11.2 microdeletion. The text has been consequently revised and in particular the aim of the study was adequately clarified. Evaluation of the qPCR technique in healthy controls and patients with velocardiofacial/DiGeorge syndrome is programmed in next future as soon as suitable samples will be available.
2. The number of control del22q11.2 negative samples have been extended to 10.
3. The maximun Ct standard deviation has been specified at page 12.
4. As suggested, numbers have been spelled out.

To Reviewer 2, Marsha Speevak:
We thank Marsha Speevak for the revision and the manuscript has been revised according to the suggestions. The first paragraph of the Results section has been moved to the Methods section as suggested by the Reviewer.
All Authors have directly participated in the planning, execution, or analysis of this study. All Authors have read and approved the final revised version submitted. The contents of this manuscript have not been copyrighted or published previously. The contents of this manuscript are not now under consideration for publication elsewhere. The contents of this manuscript will not be copyrighted, submitted, or published elsewhere, while acceptance by BMC Medical Genetics is under consideration. There are no directly related manuscripts or abstracts, published or unpublished, by any authors of this paper. Ethical or financial conflicts of interest were disclosed by all Authors.

Looking forward to hearing from you soon

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