Author’s response to reviews

Title: Intergenerational and Intrafamilial Phenotypic Variability in 22q11.2 Deletion syndrome Subjects

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Version: 6 Date: 22 December 2013

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Dear Editor,

enclosed please find the manuscript entitled “Intergenerational and intrafamilial phenotypic variability in 22q11.2 subjects”, revised as suggested, that we hope is now suitable for publication in BMC Medical Genetics.

All the authors have read and approved the submission to BMC Medical Genetics.

Looking forward to hearing from you,

Sincerely yours,
Claudio Pignata, M.D.

Point by point reply

1. In the abstract, p. 2, Conclusions, the sentence "In addition, the identification of adults with a milder phenotype deserves careful attention because the early recognition of these features could benefit from an early treatment and genetic counseling." has been deleted.

2. In the abstract, the sentence, "Both ascertainment bias related to patients selection or to the low rate of reproductive fitness of adults with a more severe phenotype, and/or several not well defined molecular mechanism, could explain intergenerational and intrafamilial phenotypic variability in this syndrome" has been amended into "Both ascertainment bias related to patient selection or to the low rate of reproductive fitness of adults with a more severe phenotype, and several not well defined molecular mechanism, could explain intergenerational and intrafamilial phenotypic variability in this syndrome.”