Author's response to reviews

Title: Common sequence variants in the LOXL1 gene in pigment dispersion syndrome and pigmentary glaucoma

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Author's response to reviews:

Dear, Naushin Waseem,

thank you for your message. here we provide a revised version of our manuscript, significantly improved by the reviewers’ comments.

In particular we modified our manuscript as requested.

Below the list of the corrections suggested by reviewer 2.

1. Eighty four Italian patients with PDS/PG were recruited for this study. All samples originating from Continental Italy. We modified the section “Methods” in the abstract: “A cohort of Caucasian origin of 84 unrelated and clinically well-characterised patients with PDS/PG and 200 control subjects were included in the study.”

point 2. How many controls were included in this study? In the abstract 200 controls are mentioned. However in Methods/Genotyping sections only 100 controls were extracted. Further 100 control samples were only analysed for 3 SNPs:

rs1048661, rs3835942 and rs2304722

R. The sequencing of LOXL1 gene was performed on 100 samples. Subsequently, the genotyping phase has been extended to 100 new samples through the use of real time PCR. Therefore, 200 samples were genotyped.

3. In Table 2 and fig1 how many controls were used? Are the results for the 3 SNPs mentioned above included in this? No justification is given for screening
additional 100 controls for the three SNPs. If the authors have included these 3 SNPs in their analysis that would have created bias in the analysis.

R. In Table 2 and fig.1, 200 controls were used and all 3 SNPs were included into it.

4. How many times were the experiments performed for the real time PCR?

R. The real-time PCR experiments were performed in triplicate.

5. I understand it is difficult to obtain tissues from eye tissues but the expression of the LOXL1 in lymphocytes does not necessary mimic that of the eye. This fact should be mentioned in the discussion.

R. We have modified in the Discussion: It is to notice that the expression levels of LOXL1 were determined on c-DNA from peripheral blood lymphocytes by quantitative real-time RT-PCR. Thus it is even possible that a correlation between expression levels and genotype at LOXL1 could be restricted at eye tissues. Further experiments on eye tissues should be performed to definitely exclude a functional effect of genetic variants on local expression of LOXL1.

6. Table 2 footnote- significative should be changed to significant. In the result section, second paragraph, regulative should be changed to regulatory.

R: We have modified “significative in the significant “ (in table 2) and “regolative in the regulatory” in the results as requested.

I hope that these changes make our manuscript suitable for publication in BMC ophthalmology.

Best regards

Rome, 07th February 2014 Emiliano Giardina