Author’s response to reviews

Title: Fanconi anemia with sun-sensitivity caused by a Xeroderma pigmentosum-associated missense mutation in XPF

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Version: 1 Date: 26 Sep 2017

Author’s response to reviews:

Professor Matteo Pasini

Editor, BMC Medical Genetics

Manchester, 25th Sept 2017

Re: MGTC-D-17-00053
Fanconi anemia with sun-sensitivity caused by a Xeroderma pigmentosum-associated missense mutation in XPF

Dear Professor Pasini,

Attached please find the revised version of the above manuscript with respect to your consideration for publication in BMC Medical Genetics.

In the course of revision the first author, Isabell Rost, got married and changed her name such that our manuscript should now be held as Popp et al. Secondly, we would like to change the former title

“Fanconi anemia with sun-sensitivity caused by a Xeroderma pigmentosum-associated missense mutation in XPF – case report of a third FA-Q patient” to the shorter title above as we think it is self-evident that it is a case report, even though an extended one.

We would like to thank the Editor and the Reviewers for critical examination of our manuscript and for providing constructive criticism. As a result we are now submitting a revised and much improved manuscript. We have addressed the Reviewers’ comments and provide additional experimental data and revised figures. The text is changed accordingly at the relevant sections as indicated.

In detail, the additional work comprises the demonstration that XPF/FANCQ/ERCC4 with the Arg589Trp mutation and ERCC1 interact, that the mutations are expressed and serial dilution studies to estimate the degree of reduction of XPF/FANCQ/ERCC4 protein in 3104 cells.

We do hope that our manuscript is now suitable for publication in BMC Medical Genetics.

Sincerely,

Stefan Meyer

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