Reviewer’s report

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

Version: 0 Date: 08 Jul 2017

Reviewer: Kohichiro Yoshiura

Reviewer’s report:

Re: MGTC-D-00053
Title: Fanconi anemia with sun-sensitivity caused by a Xeroderma pigmentosum associated missense mutation in XPF - case report of the FA-Q patient.

Authors described patient's phenotype, genomic analyses and results of protein functional assay. Nonetheless, some points yet to be corrected or cleared.

1. In figure 1E, quantify the transcripts with exon5 and without exon5. Digital PCR, quantification PCR or personal type next generation sequencer can be applied for quantification. It seems exon5-skipped transcript looks like less than 1/4 compare to exon5-retaining transcript.

2. Related to 1, transcript containing missense mutation in exon8 should be quantified. Authors' discussion about the quantity of XPF/ERCC4/FANCQ is not quantitative, so please discuss quantitatively based on the mRNA and protein quantity. It looks like the quantity of FANCQ protein in cell decreased in Figure 1H. This reduction suggests that XPF/ERCC4/FANCQ with p.R589W is unstable, because less than expected quantity based on mRNA quantity. All of this kind of discussion should be based on two kinds of transcript and western blot analyses.

3. In figure 1G, this reviewer think horizontal scale should be log scale like figure 2E. Is MMC concentration correct? The unit ng/microL may be ng/mL, please make sure. In addition, concentration is shown nM in figure 2E. It may be better to consolidate the same unit.

4. In figure 2A and 2C, XP23OS is used representative to XP-F in figure 2A and XP42RO is used representative to XP-F in figure 2D. Did you use various cell lines? If you did experiment using same cell line, the data using same cell line is ideal, or show the results from both of them.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes
Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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