Author’s response to reviews

Title: A Novel Frame-shift deletion in FANCF Gene Causing Autosomal Recessive Fanconi Anemia: a case report

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Author’s response to reviews:

Dear Editor-in-chief:
Ref: MGTC-D-18-00466
Title: A Novel Frame-shift deletion in FANCF Gene Causing Autosomal Recessive Fanconi Anemia: a case report
Journal: BMC Medical Genetics

Thank you for excellent reviewing of this manuscript “A Novel Frame-shift deletion in FANCF Gene Causing Autosomal Recessive Fanconi Anemia: a case report “in BMC Medical Genetics Journal with Manuscript Number: MGTC-D-18-00466 and valuable views of the reviewers. The manuscript was revised and corrected according to reviewers' suggestions, which performed on particular draft. I have corrected the manuscript according to reviewer comments.

The following review was carried out.
1. A highlighted/track changes version of the latest revision of the submission
2. A clean copy of the latest version of the submission.
3. A complete point-by-point response to each reviewers’ comment.
4. All of the figures.
Reviewer reports:
Babu Rao Vundinti (Reviewer 1):

1. The manuscript entitled "A Novel Frame-shift deletion in 1 FANCF Gene Causing Autosomal Recessive Fanconi Anemia: a case report submitted by Zareifar and colleagues. The manuscript is poorly written.
Response 1:
Extensive English editing was done by Clinical Research Development Center of Shohadaye-Khalij-e-Fars Hospital and Miss Fatemeh Gholizadeh that acknowledge them.

2. It is a single case report with novel frameshift mutation. The case report is lacking genotype - phenotype correlation.
Response 2:
This patient is single case report; therefore, we cannot definitely determine the genotype - phenotype correlation. Additionally some changes were done and highlighted in yellow.

3. The chromosomal breakage frequency need be given properly.
Response 3:
It was explained in detail in the text and highlighted in yellow.

4. Discussion is lacking the mutations reported in FANCF gene and its clinical correlation.
Response 4:
One paragraph in discussion part was added and highlighted in yellow

6. Authors are not discussed about the clinical severity of the novel mutations compared to literature reports.
Response 5:
One paragraph was added and highlighted in yellow

7. The management importance this mutations need to be clearly written.
Response 7:
Because the patient is a case of Fanconi anemia, the management is the same as other case of Fanconi. Additionally, the patient is a single case, need long follow up in order to determine the prognosis, and retrospectively managed him/her.

Paula Rio (Reviewer 2): The paper by Zareifar et al describes a new frame-shift mutation in FANCF that causes Fanconi anemia. This is a case report of a patient with typical phenotype of Fanconi anemia and in which the group describes the presence of a new frame-shift mutation in homozygosis.

The paper is interesting and the description of a new mutation could be relevant for the disease and, as the authors suggest, it could be relevant to study the phenotype-genotype correlations in the disease.
Major comments:
Background: a brief description of FA patients studied by the group or if there is any register/data with Iranian FA patients.
Response:
There is no data with Iranian Fanconi anemia
It would be interested to confirm the presence of the mutation in the parents.
Response:
Due to unavailability, we cannot do the genetic mutation of parents

Figure 4: Describe in detail the number and type of the different aberrations detected.
Response Figure 4:
The details were explained in text and highlighted in yellow
The paper should be reviewed by an English speaker.
Response:
Extensive English editing was done by Clinical Research Development Center of Shohadaye-Khalij-e-Fars Hospital and Miss Fatemeh Gholizadeh that acknowledge them.

The quality of the figures should be improved.
Response:
It was improved