Journal of Clinical and Translational Research
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Molecular insight of dyskeratosis congenita: Defects in telomere length homeostasis

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Handling editor:
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Review timeline:

Received: 28 February, 2021
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1st Editorial decision
19-Jun-2021

Ref.: Ms. No. JCTRes-D-21-00029
Molecular Insight of Dyskeratosis Congenita: Defects in Telomere Length Homeostasis
Journal of Clinical and Translational Research

Dear Dr oladnabi,

Reviewers have now commented on your paper. You will see that they are advising that you considerably revise your manuscript. If you are prepared to undertake the work required, I would be pleased to reconsider my decision.

For your guidance, reviewers' comments are appended below.
Please understand that the manuscript misses its aim and does not present the complete scope of the molecular background of dyskeratosis congenita and related telomere biology disorders. This fact, in addition to a significant upgrade to academic level English as stipulated in the journal's author guidelines, will require a complete overhaul of the manuscript. The editorial board encourages the authors to not take this task lightly. We will not burden our reviewers if we deem the depth of corrections insufficient and will consequently reject the manuscript.
Naturally, if the authors have any questions related to moving forward they should not hesitate to contact the editor (m.heger@jctres.com).

If you decide to revise the work, please submit a list of changes or a rebuttal against each point which is being raised when you submit the revised manuscript. Also, please ensure that the track changes function is switched on when implementing the revisions. This enables the reviewers to rapidly verify all changes made.

Your revision is due by Jul 19, 2021.

To submit a revision, go to https://www.editorialmanager.com/jctres/ and log in as an Author. You will see a menu item call Submission Needing Revision. You will find your submission record there.

Yours sincerely

Michal Heger
Editor-in-Chief
Journal of Clinical and Translational Research

Reviewers' comments:

Reviewer #1: Description
In the presented manuscript, the authors aim to review the current literature and knowledge on the molecular background of Dyskeratosis congenita.

General comments
I would generally suggest having the manuscript reviewed by a native English speaker. It could also be more concise in its structure and be shortened.
When discussing the molecular background of Dyskeratosis congenita and related Telomere Biology Disorders, the authors only mention 10 genes as disease causing in Dyskeratosis congenita. However, to date there are 15 genes described. I would suggest reviewing for example the following papers: A. Bertuch, RNA Biol. 2016 Aug 2;13(8):696-706. Gable et al. Genes Dev. 2019 Oct 1;33(19-20):1381-1396.
There are several terms used in the literature to describe the spectrum of telomere associated diseases. The term "telomere biology disorders" (TBD) is favored since it probably describes the underlying pathophysiology best.

Specific comments
1. Gene names should be written in italic. This was not consistently done throughout the manuscript.
2. In 1.3. its mentioned that 400 DC families have been reported in the literature and that Dyskeratosis congenita accounts for 1% of telomere syndromes. It would be helpful to cite the references these statements are based on.
3. The description of DC (1.3.) it remains unclear what the classic presentation of DC is. Helpful literature includes: Dokal et al. Eur J Hum Genet. 2015 Apr;23(4).
4. Section 3: The only clinically validated test to diagnose Dyskeratosis congenita is Flow-FISH (see Alter et al, Haematologica 2012;97(3):353-9).

Authors’ response

Responses to Reviewer 1:

General comments:

I would generally suggest having the manuscript reviewed by a native English speaker. It could also be more concise in its structure and be shortened.

When discussing the molecular background of Dyskeratosis congenita and related Telomere Biology Disorders, the authors only mention 10 genes as disease causing in Dyskeratosis congenita. However, to date there are 15 genes described. I would suggest reviewing for example the following papers: A. Bertuch, RNA Biol. 2016 Aug 2;13(8):696-706. Gable et al. Genes Dev. 2019 Oct 1;33(19-20):1381-1396.

There are several terms used in the literature to describe the spectrum of telomere associated diseases. The term "telomere biology disorders" (TBD) is favored since it probably describes the underlying pathophysiology best.

Answer: According to the helpful comment of the reviewer, the text was edited by an English speaker and structurally reviewed.

Based on a review of articles (A. Bertuch, RNA Biol. 2016 Aug 2; 13 (8): 696-706. Gable et al. Genes Dev. 2019 Oct 1; 33 (19-20): 1381-1396.), From 15 Genes Discussed In these articles, we have identified 13 genes that cause Dyskeratosis Congenita. so three more genes have been added to our gene list.

Comment 1: Gene names should be written in italic. This was not consistently done throughout the manuscript.

Answer: All the names of the genes mentioned in the text are italicized.

Comment 2: In 1.3. its mentioned that 400 DC families have been reported in the literature and that Dyskeratosis congenita accounts for 1% of telomere syndromes. It would be helpful to cite the references these statements are based on.

Answer: This sentence was referenced from the https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=1775

Comment 3: The description of DC (1.3.) it remains unclear what the classic presentation of DC is. Helpful literature includes: Dokal et al. Eur J Hum Genet. 2015 Apr;23(4).

Answer: Due to the authors' opinion and the uncommonness of the term "classic presentation of DC", it was removed from the text.

Comment 4: Section 3: The only clinically validated test to diagnose Dyskeratosis congenita is Flow-FISH (see Alter et al, Haematologica 2012;97(3):353-9).

Answer: Thank you for your accurate comment, the Flow-FISH technique has been clinically approved, but in this section, in addition to the approved clinical technique, we have mentioned other techniques that have been used in various non-clinical research to give a complete overview of telomere length measurement techniques.

2nd Editorial decision
29-Oct-2021

Ref.: Ms. No. JCTRes-D-21-00029R1
Molecular Insight of Dyskeratosis Congenita: Defects in Telomere Length Homeostasis
Journal of Clinical and Translational Research

Dear author(s),

Reviewers have submitted their critical appraisal of your paper. The reviewers' comments are appended below. Based on their comments and evaluation by the editorial board, your work was FOUND SUITABLE FOR PUBLICATION AFTER MINOR REVISION.

If you decide to revise the work, please itemize the reviewers' comments and provide a point-by-point response to every comment. An exemplary rebuttal letter can be found on at http://www.jctres.com/en/author-guidelines/ under "Manuscript preparation." Also, please use the track changes function in the original document so that the reviewers can easily verify your responses.

Your revision is due by Nov 28, 2021.

To submit a revision, go to https://www.editorialmanager.com/jctres/ and log in as an Author. You will see a menu item call Submission Needing Revision. You will find your submission record there.

Yours sincerely,

Michal Heger
Editor-in-Chief
Journal of Clinical and Translational Research

Reviewers' comments:

Dear authors,

Thank you for submitting a revised version of your manuscript.

The paper has now passed peer review because you have sufficiently addressed the reviewer's comments. However, before we can proceed to publication, the manuscript must be improved in terms of linguistics in order to become compliant with our author guidelines.

There are several ways that you could do this. The first and easiest way is to engage a native speaker to go through the text on your behalf. The second way is to contract a proofreading service to eliminate linguistic errors and inconsistencies. Finally, our journal can employ an editor to help you with the language polishing for a fee. Our contractors not only improve the language but also perform a deep dive on the content and text structure. If you choose the last option, please contact me (m.heger@jctres.com).

Thank you and kindest regards,

Michal Heger
Editor

Authors’response

Dear editor in chief of JCTR
The manuscript has been edited by an English-speaking native, so we hope it now matches the journal standard.

Manuscript ID: JCTRes-D-21-00029R2

Sincerely,
Morteza Oladnabi (Ph.D)
Associated professor
Department of Medical Genetics

3rd Editoral decision
03-Dec-2021

Ref.: Ms. No. JCTRes-D-21-00029R2
Molecular Insight of Dyskeratosis Congenita: Defects in Telomere Length Homeostasis
Journal of Clinical and Translational Research

Dear authors,

I am pleased to inform you that your manuscript has been accepted for publication in the Journal of Clinical and Translational Research.

You will receive the proofs of your article shortly, which we kindly ask you to thoroughly review for any errors.

Thank you for submitting your work to JCTR.

Kindest regards,

Michal Heger
Editor-in-Chief
Journal of Clinical and Translational Research

Comments from the editors and reviewers: