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Matamoros, Tamps, México, October 06, 2022.

Distinguished Dr. Jin-Lei Wang, Company Editor-in-Chief
World Journal of Clinical Cases

Thank you for your soon reply about the manuscript No. 79958, entitled “**Novel *TINF2* gene mutation in dyskeratosis congenita with extremely short telomeres: A case report**”.

Here, I am returning to you the amended version of our MS, modified according with the comments and suggestions of the reviewers. Principal changes are highlighted in bold type in the MS.

Next, I attach the reviewers' comments and our response to every suggestion.

Thanks for your attention.

Sincerely,

Juan Pablo Meza E.

Dr. Juan Pablo Meza-Espinoza
Corresponding author



COMMENTS

Reviewer #1:

Scientific Quality: Grade B (Very good)

Language Quality: Grade B (Minor language polishing)

Conclusion: Minor revision

Specific Comments to Authors:

Acknowledgments. Thanks for the review.

Comment 1. This is a good paper, on a very interesting topic. Since the condition is rare and genetic, a table with review of most important literature sources would be good for readers. Also, there is no language editing certificate for non-native English authors included in the system. Can you provide one?

Reply 1: The most important details of dyskeratosis congenita are included in the text. However, two tables were included: a about the genetic spectrum of the disease and the other with the frameshift mutations of the *TINF2* gene.

Comment 2. Also, there is no language editing certificate for non-native English authors included in the system. Can you provide one?

Reply 2: An editing English certificate was provided.

Reviewer #2:

Scientific Quality: Grade C (Good)

Language Quality: Grade B (Minor language polishing)

Conclusion: Major revision

Specific Comments to Authors: This is an excellent case report of dyskeratosis congenita with a novel genetic variant. The case presentation and discussion are generally well described. I would like to make a few recommendations to improve this case report.

Acknowledgments. Thanks for the comments.

Comment 1 (C1). The authors describe dyskeratosis congenita as a rare genetic disease. For readers who do not specialise in this field, the frequency of this disease should also be explained.

Reply 1 (R1). The prevalence of dyskeratosis congenita was added.

Comment 2. Collagen diseases such as dermatomyositis and systemic lupus erythematosus also come up in the differential as diseases that generally present with haematopoietic and skin disorders in young females. Have screening tests such as antinuclear antibodies been performed for these diseases?

Reply 2. Many studies of laboratory, including tests for antinuclear antibody were performed. The test for antinuclear antibodies was negative, therefore systemic



lupus erythematosus and mixed connective tissue disease were discarded. This information was already included in the manuscript.

Comment 3. The patient was treated with high-dose prednisolone. Prednisolone can cause a high risk of infection. Was fungal infection considered as a differential for the white lesions on the skin and tongue in that patient?

Reply 3. Yes, fungal infections, such as *Candida albicans*, were discarded.

Comment 4. In Figure 1C, light reflections and leukokeratosis plaques on the surface of the tongue are difficult to distinguish. Also, the oral cavity is dark and difficult to see. Therefore, a photograph in which the palate or posterior wall of the pharynx is illuminated rather than the surface of the tongue is more desirable.

Reply 4. Another photograph was included. However, it may not meet the required quality.

Comment 5. It is excellent that an accurate diagnosis was made promptly in this case. As the authors point out, there is no fundamental treatment for this disease. Has appropriate mental care and support been provided for these patients? For example, were regular genetic counselling or consultation to a psychiatrist considered?

Reply 5. Genetic counselling was provided to the family. Moreover, the girl has been in psychotherapy since the onset of puberty, because she does not accept the disease.