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OPEN PEER REVIEW

The Concordance of Paternal and Maternal Retinoblastoma-1 Gene Mutation Pattern with Clinical Manifestation and Disease Staging in Patients Suffering from Retinoblastoma

Shadan Shahraki Moghadam, Mohammad Faranoush, Mohammad Mahdi Johari Moghadam, Ahad Sedaghat, Mohammad Jafar Ayatollahi

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1 Original submission

2 comments · 25 Jul 2021 – 26 Jul 2021

Abdolhassan Kazemi Reviewer

MINOR REVISION 25 Jul 2021

The study entitled "The concordance of paternal and maternal retinoblastoma-1 gene mutation pattern with clinical manifestation and disease staging in patients suffering retinoblastoma" is novel and interesting. However, some minor concerns should be considered to improve the originality as well as the flow of the manuscript:

Title: The current title is acceptable, but I could not find a running title.

Abstract: The current abstract is acceptable.

Keywords: The current keywords need to be (or could be) rewritten as: Mutation, retinoblastoma-1 gene, phenotype, retinoblastoma, Iran

Introduction:
The introduction section should briefly highlight the importance and necessity of this study.
M & M: The current M & M is acceptable.

Results: The current results are acceptable.

Discussion & Conclusion:
These sections could be more scientific through a broader literature review and an explanation of related studies and domestic research, which should then be cited in the Bibliography/References.
Bibliography/References: More domestic references are necessary. The authors should conduct a broader literature review to identify related domestic articles and therefore include more domestic references. (Using domestic references is part of the scientific discipline of publishing academic data.)

Final decision: The manuscript is acceptable for publication after minor correction.

I can suggest some articles from a literature review and survey, as listed below:
Ghassemi F, Bazvand F, Makateb A
Lesions Simulating Retinoblastoma at a Tertiary Care Center.
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Associate Editor

MINOR REVISION 26 Jul 2021

Reviewer Comments to Author(s):
Reviewer #1
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[Paternal GNAS mutations: Which phenotypes? What genetic counseling?].
Annales d'endocrinologie. 2015

2

Revision 1

AUTHORS' RESPONSE TO REVIEWERS

Authors' reply to reviewers

The next pages contain the authors' point-by-point reply to the comments of this revision round, as submitted with the revised manuscript. It is published as the authors wrote it.

Reply to reviewer

Reviewer 1:

#Q1: keywords

R1:The keywords of manuscript are changed as bellow:

Mutation, retinoblastoma-1 gene, phenotype, retinoblastoma, Iran

#Q2: Introduction: highlight the more importance and necessity of this study

R2:

Retinoblastoma is a most common intraocular malignancy among children , that is mainly diagnosed during routine eye examination with a white pupil reflex, known as leukocoria.

In the nonhereditary form of retinoblastoma, the risk of disease transmission is only 5%, while in the hereditary form, the risk increase to 50%.

#Q3: Discussion & Conclusion: using more and wider literature review.

R3: we use more domestic survey as bellow:

Nabie et al. showed that frequency of unilateral RB1 presentation (57.7%) was slightly higher than bilateral presentation (42.5%) in children with RB1 of north-west side of Iran, however, result of this study showed that bilateral presentation (82.60%) was significantly compared to unilateral presentation of RB1 (17.39%).

Shahraki et al. performed a comprehensive genetic screening on 106 patients whom were diagnosed with retinoblastoma in Iran. Based on their results, the mean age at the time of diagnosis was 12.7±10.5 for RB with a positive family history, they also demonstrated 38 different causative RB1 mutations among patients, with an RB1 mutation detection rate of 65.8%. In this study, we detected more than 19 different mutations among RB1 patients.

Ghassemi and colleagues showed that the most common presenting stage of RB1 patients at IAC therapy was group D reaching to up to 75%, followed by group E and group C at the first visit, Interestingly, clinical examination of left eye in our patients demonstrated that up group D is the most common diagnosed class (47.8%) that was followed by group E (21.7%).

#Q4: references: more domestic Ref. is necessary

R4: we add more domestic references which are references number 16, 17 and 18

16. Nabie R, Taheri N, Fard AM, Fouladi RF. Characteristics and clinical presentations of pediatric retinoblastoma in North-western Iran. International journal of ophthalmology. 2012;5(4):510.

17. Shahraki K, Ahani A, Sharma P, Faranoush M, Bahoush G, Torktaz I, Gahl WA, Naseripour M, Behnam B. Genetic screening in Iranian patients with retinoblastoma. Eye. 2017 Apr;31(4):620-7. 17.

18. Ghassemi F, Ghanaati H, Karkhaneh R, Boujabadi L, Tabatabaie SZ, Rajabi MT. Outcome of retinoblastoma following limited sessions of intra-arterial chemotherapy in Iran. Iranian Journal of Radiology. 2014 Aug;11(3).

2 Revision 1

1 comment · 4 Sep 2021

Referee 1 Reviewer

ACCEPT 4 Sep 2021

The revision of the manuscript in accordance with the reviewer(s)' suggestions is acceptable and addresses the necessary items; therefore, the manuscript is acceptable for publication.