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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 74260

Title: MutL homolog 1 germline mutation c.(453+1_454-1)_(545+1_546-1)del identified in

lynch syndrome: A case report and review of literature

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06083984 Position: Peer Reviewer

Academic degree: BSc, MSc, PhD

Professional title: Research Scientist

Reviewer's Country/Territory: Belarus

Author's Country/Territory: China

Manuscript submission date: 2021-12-18

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-12-18 20:01

Reviewer performed review: 2021-12-26 15:30

Review time: 7 Days and 19 Hours

Scientific quality	[] Grade A: Excellent [Y] Grade B: Very good [] Grade C: Good [] Grade D: Fair [] Grade E: Do not publish
Language quality	[] Grade A: Priority publishing [Y] Grade B: Minor language polishing [] Grade C: A great deal of language polishing [] Grade D: Rejection
Conclusion	[] Accept (High priority) [] Accept (General priority) [Y] Minor revision [] Major revision [] Rejection
Re-review	[Y]Yes []No



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Peer-reviewer

Peer-Review: [Y] Anonymous [] Onymous

statements Conflicts-of-Interest: [] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The the "MLH1 germline work presented in mutation c.(453+1_454-1)_(545+1_546-1)del-induced lynch syndrome: A case report" is new and interesting, described data could be further used in the medical area and clinical practice. I have some minor comments, please, find them below. 1. Please, de-crypt all used abbreviations upon the first use (including genes) and add the abbreviation list. 2. Please, explain briefly the function of the MLH1 gene. Where described mutation is located? How does it affect proteins' function? 3. Please, consider the title correction. The mutation induces or causes LS? 4. Please, provide a more detailed explanation of used gene detection steps (methods, bioinformatic tools, equipment).



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lynch syndrome: A case report and review of literature

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03735621 Position: Peer Reviewer

Academic degree: MD, PhD

Professional title: Staff Physician

Reviewer's Country/Territory: Japan

Author's Country/Territory: China

Manuscript submission date: 2021-12-18

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-01-10 03:05

Reviewer performed review: 2022-01-12 07:54

Review time: 2 Days and 4 Hours

Scientific quality	[] Grade A: Excellent [] Grade B: Very good [] Grade C: Good [Y] Grade D: Fair [] Grade E: Do not publish
Language quality	[] Grade A: Priority publishing [] Grade B: Minor language polishing [Y] Grade C: A great deal of language polishing [] Grade D: Rejection
Conclusion	[] Accept (High priority) [] Accept (General priority) [] Minor revision [Y] Major revision [] Rejection
Re-review	[]Yes [Y]No



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Peer-reviewer

Peer-Review: [Y] Anonymous [] Onymous

statements Conflicts-of-Interest: [] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The authors provided detailed information on a Chinese woman with MLH1 germline mutation c.(453+1 454-1) (545+1 546-1)del-induced LS. This reviewer wishes to express some reservations about this manuscript prior to publication. Comments #1. According to the authors, deletion of MLH1 exon 6 (c.(453+1 454-1) (545+1 546-1)del) infrequently caused LS in Chinese. However, the Discussion made no mention of previously reported cases with the same MLH1 genetic variant. The authors should compare the clinicopathological characteristics of this case to those of previously reported ones. #2. The final pathological diagnosis should be made using the most recent WHO classification. Due to the fact that there are several histological subtypes of endometrial cancer, the term "endometrial cancer" alone is insufficient. #3. The results of the IHC staining appear to be somewhat peculiar. "Immunohistochemical (IHC) staining results of pathological findings showed MLH1 (-), PMS2 (-), MSH6 (partial lesions +), and MSH2 (partial lesions weak +)," the authors wrote. If the tumor had MLH1 gene mutations, both MLH1 and PMS2 should be lost in the majority of such cases. However, both MSH6 and MSH2 expression appeared to be abnormal. The authors should provide a rationale for this occurrence. Additionally, readers should be able to recognize intact MMR protein expression in internal control cells via microphotographs of IHC. The present form of microphotographs seems to be inappropriate. #4. Some typographical errors are present.