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PEER-REVIEW REPORT

Name of journal: *World Journal of Gastroenterology*

Manuscript NO: 82137

Title: Peutz-Jeghers Syndrome without STK11 mutation may Correlate with Less Severe Clinical Manifestations in Chinese Patients

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06129219
Position: Peer Reviewer
Academic degree: MD, PhD
Professional title: Associate Professor
Reviewer's Country/Territory: Japan
Author's Country/Territory: China

Manuscript submission date: 2023-01-20

Reviewer chosen by: AI Technique

Reviewer accepted review: 2023-01-26 08:30

Reviewer performed review: 2023-02-08 10:13

Review time: 13 Days and 1 Hour

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Novelty of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No novelty
Creativity or innovation of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No creativity or innovation

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Scientific significance of the conclusion in this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No scientific significance
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
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

General comments: In this retrospective study, to investigate whether PJS patients with known STK11 mutations have a more severe spectrum of clinical phenotypes compared to those without, Genomic DNA samples were extracted from peripheral blood samples from 92 PJS patients with and without STK11/LKB1 mutations, and pathogenic germline mutations of STK11 were detected by high-throughput next-generation gene sequencing (NGS), comparing their clinical-pathologic manifestations. The manuscript is informative and well presentation. It's written well, the Introduction give a good overview about the study background and the authors raised clearly the aim of the study. The description of material studied is accurate and allows to draw the conclusions. Please add limitations of your study to the discussion. Thank you for giving opportunity to review your study.

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Reviewer's code: 06129218
Position: Peer Reviewer
Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Japan

Author's Country/Territory: China

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Reviewer accepted review: 2023-01-26 08:31

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Novelty of this manuscript	[Y] Grade A: Excellent [ ] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No novelty
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Re-review	[Y] Yes [ ] No
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

Thank you very much for asking me to review this manuscript by Lixin Jiang et al. This is a retrospective study to investigate whether PJS patients with known STK11 mutations have a more severe spectrum of clinical phenotypes compared to those without. The result of the study is of interest and may help evaluate the status and prognosis of Peutz-Jeghers Syndrome and provide an objective reference for diagnosis and treatment. Overall, this study was well conducted with good methodology and intelligible English. For rare diseases, the number of participants in the study is enough. Furthermore, minor comment that I would to proposed: 1. Title: Proper and cover all the core result from the study. 2. Abstract: Address all of the important component from the study. 3. Key words: could cover this study. 4. Introduction: Describe the overall basic knowledge for this study. Moreover, the aim of the study is clear. 5. Method: The present study is methodologically well conducted. 6. Results: The result of this study is of interest. However, in "2.2 Comparison of general information, diagnosis and treatment, pathology and examinations (Table 5)", it is recommended not to only show the results in the table, important findings also need to be described in the text. 7. Discussion: The

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manuscript clearly interprets the finding adequately and appropriately. In addition, the manuscript could highlight the key points clearly. It is recommended to add a discussion of the limitations of the study to the discussion section. 8. Tables and figures: I congratulate the authors for the captions to the tables and figures very explicative and complete.

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Reviewer's code: 06129258
Position: Peer Reviewer
Academic degree: MD, PhD
Professional title: Associate Professor, Senior Lecturer
Reviewer's Country/Territory: South Korea
Author's Country/Territory: China

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Re-review	[ ] Yes [Y] No
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

Dear authors, thank you for submitting your paper to the World Journal of Gastroenterology. In your retrospective study, a total of 92 patients with PJS from 2010 to 2022 were selected. Genomic DNA samples were extracted from peripheral blood samples, and pathogenic germline mutations of STK11 were detected. They found that 73 patients with PJS had STK11 gene mutations and 19 patients had no STK11 gene mutations, of which 6 had no other gene mutations and 13 had other gene mutations. Finally, you found compared with PJS patients with STK11 mutations, those without tended to be older at the age of initial treatment, age of first intussusception and age of initial surgery. They also had a lower number of total hospitalizations relating to intussusception or intestinal obstruction, and a lower load of small intestine polyps. Your study is a well-written, good structured recommendation for the evaluate and prognosis of PJS. Introduction give a good overview about the study background and the authors raised clearly the hypothesis of the study. The description of material studied is accurate. The aim of the study is fulfilled. The material studied is large enough and allows to draw the conclusions. The Results are presented clearly and have been

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discussed well. All recommendations are good for clinical use. Thank you for a useful and important synopsis of this important topic.