

PEER-REVIEW REPORT

Name of journal: World Journal of Gastroenterology

Manuscript NO: 66814

Title: Detection and analysis of common pathogenic germline mutations of Peutz-Jeghers syndrome

Reviewer's code: 06074688

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Australia

Author's Country/Territory: China

Manuscript submission date: 2021-04-15

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-04-19 00:37

Reviewer performed review: 2021-04-19 10:29

Review time: 9 Hours

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input checked="" type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input checked="" type="checkbox"/> Grade A: Priority publishing <input type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input checked="" type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No
Peer-reviewer statements	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous Conflicts-of-Interest: <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No

SPECIFIC COMMENTS TO AUTHORS

In this study, the authors collected 24 cases from the treated PJS cases as the research objects, collected peripheral venous blood or normal tissues adjacent to polyps for high-throughput sequencing study (NGS) of 139 hereditary colorectal tumor-related genes including STK11/LKB1, in order to study the correlation between genotype and clinical phenotype of PJS, and explore the internal molecular mechanism of different clinical phenotypes. As is known, PJS is a rare disorder in which growths called polyps form in the intestines. The authors present an interesting study that is well motivated and designed. Their data is well presented and their conclusions largely follow from this data. They also do a nice job as to hypothesizing the mechanisms that may be driving their results without overstating their significance. They discovered a series of new gene mutation sites, analyzed their pathogenicity, and enriched the mutation spectrum of PJS pathogenic genes. And through the summary of the clinical phenotypes with different STK11 genotypes, to explore whether they are related, and get some tendentious research results. The detection of slx4 gene mutations in patients with PJS was reported for the first time. Although the relationship between several mutations of this gene and the occurrence of pjs is still unclear, this is a good start for explaining the genetic heterogeneity of PJS.

PEER-REVIEW REPORT

Name of journal: World Journal of Gastroenterology

Manuscript NO: 66814

Title: Detection and analysis of common pathogenic germline mutations of Peutz-Jeghers syndrome

Reviewer's code: 06074681

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Finland

Author's Country/Territory: China

Manuscript submission date: 2021-04-15

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-04-19 00:37

Reviewer performed review: 2021-04-21 11:47

Review time: 2 Days and 11 Hours

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input checked="" type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No
Peer-reviewer statements	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous Conflicts-of-Interest: <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No



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SPECIFIC COMMENTS TO AUTHORS

The manuscript "Detection and analysis of common pathogenic germline mutations of Peutz-Jeghers syndrome" by Guoli Gu investigate the correlation between the genotype and clinical phenotype of Peutz-Jeghers syndrome (PJS). It is well written and highly interesting. The study is well designed and presented with optimal analysis, discussion, tabulation and graphic display of data. Thank you for giving opportunity to review this study. However, the following points must be considered before publication. In my opinion, note that the additional expanded discussions are mandatory. In addition, I recommended to add main inclusion and exclusion criteria for the subjects included in the study.

PEER-REVIEW REPORT

Name of journal: World Journal of Gastroenterology

Manuscript NO: 66814

Title: Detection and analysis of common pathogenic germline mutations of Peutz-Jeghers syndrome

Reviewer's code: 06074684

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Denmark

Author's Country/Territory: China

Manuscript submission date: 2021-04-15

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Review time: 2 Days and 11 Hours

Scientific quality	<input checked="" type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No
Peer-reviewer statements	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous Conflicts-of-Interest: <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No

SPECIFIC COMMENTS TO AUTHORS

Peutz-Jeghers syndrome (PJS) is a rare disorder in which growths called polyps form in the intestines. A person with PJS has a high risk of developing certain cancers. In the manuscript, authors collected 24 patients with PJS. 139 common hereditary tumor-related genes including STK11/LKB1 were screened and analyzed for pathogenic germline mutations by high-throughput sequencing (NGS). And the mutation status of those common genetic genes in PJS and its relationship with clinical phenotypes were explored. They found that PJS has a relatively complicated genetic background. Changes in the sites responsible for coding functional proteins in exon 1 and exon 4 of STK11/LKB1 may be one of the main causes of PJS; the mutation of SLX4 gene may be the one reason of genetic heterogeneity in PJS. Overall, this study was well conducted with good methodology and intelligible English. The reviewer has minor comments. Comments: 1-Abstract: The Abstract is not good enough and needs to be revised. Need to add some simple background and be more organized. 2-Introduction: The background part of the text is well written. However, I noticed that the sample size of this study was only 24 cases. Because PJS is an extremely rare disease, and its prevalence is 1/200,000 ~ 1/8000. I suggest that more basic knowledge of this disease, such as epidemiology and prevalence, should be introduced in the background introduction. 3-Experimental objects and methods: the paragraphs are generally well structured and explained. 4-Result: well and clearly presented with pertinent statistics. 5-Discussion: The manuscript clearly interprets the finding adequately and appropriately. In addition, the manuscript could highlight the key points clearly.