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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Pediatrics*

Manuscript NO: 100493

Title: Molecular profiles and long-term outcomes of Thai children with hepatic glycogen storage disease in Thailand

Provenance and peer review: Invited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 08246378
Position: Editorial Board
Academic degree: MD, PhD
Professional title: Associate Professor
Reviewer's Country/Territory: China
Author's Country/Territory: Thailand

Manuscript submission date: 2024-08-18

Reviewer chosen by: Meng-Hua Liu (Quit 2024)

Reviewer accepted review: 2024-09-05 14:48

Reviewer performed review: 2024-09-06 14:02

Review time: 23 Hours

Scientific quality	[Y] Grade A: Excellent [ ] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish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

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Scientific significance of the conclusion in this manuscript	[Y] Grade A: Excellent [ ] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No scientific significance
Language quality	[Y] Grade A: Priority publishing [ ] 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

This study provides valuable insights into the clinical, biochemical, and molecular features of glycogen storage diseases (GSD) in Thai children, an understudied population. The authors' efforts in conducting whole-exome sequencing (WES) to identify causative genetic variants and assessing long-term outcomes are commendable. However, a few aspects of the study design, data interpretation, and presentation could be strengthened to enhance the rigor and impact of the findings. 1. The study includes only 8 patients, which limits the statistical power and generalizability of the findings. Consider discussing the limitations associated with the small sample size and strategies for future studies to increase the cohort size. 2. Provide more detailed clinical descriptions of the patients, including age at onset, family history, and any comorbidities. 3. Clarify the criteria used for assessing developmental delay and the specific biochemical markers of hypoglycemia, transaminase, and lipid abnormalities. 4. Describe the bioinformatic pipeline used for WES data analysis, including variant filtering criteria and validation methods. 5. For the two novel variants identified in AGL, provide additional information such as allele frequency in control databases, potential

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functional impacts, and whether they have been reported in other GSD cases. 6. Provide more details on the specific dietary interventions (e.g., dosage, frequency, and compliance) and their impact on biochemical markers and clinical symptoms. Discuss potential confounding factors that may have influenced the observed improvements in height, liver enzymes, blood sugar, and lipid profiles. 7. Compare the clinical and molecular findings with those reported in other populations to highlight any unique features of Thai GSD patients. Discuss how the long-term outcomes compare to similar studies with larger sample sizes and different treatment protocols. 8. Acknowledge the limitations of the study, including sample size, potential biases, and lack of genetic counseling information. 9. Suggest future research directions, such as expanding the cohort size, investigating genotype-phenotype correlations, and exploring novel therapeutic approaches. 10. In summary, this study represents an important step forward in understanding GSD in Thai children. With some modifications to address the above recommendations, the authors have the potential to produce a more impactful manuscript that will contribute significantly to the field of pediatric metabolic disorders. I recommend this study for publication pending the authors' incorporation of the suggested revisions.

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Peer-review model: Single blind

Reviewer's code: 07961025
Position: Peer Reviewer
Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: China

Author's Country/Territory: Thailand

Manuscript submission date: 2024-08-18

Reviewer chosen by: Meng-Hua Liu (Quit 2024)

Reviewer accepted review: 2024-09-04 23:39

Reviewer performed review: 2024-09-13 01:51

Review time: 8 Days and 2 Hours

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

SPECIFIC COMMENTS TO AUTHORS

The current study entitled "Molecular profiles and long-term outcomes of Thai children with hepatic glycogen storage disease in Thailand " is meaningful. To further improve the quality of the manuscript, only small modifications are suggested: Pathological section images need to add a scale bar to increase readability.