

## PEER-REVIEW REPORT

**Name of journal:** *World Journal of Clinical Pediatrics* 

Manuscript NO: 77097

Title: Three Novel Homozygous ITGB2 Mutations among Two Patients with Leukocyte

Adhesion Defect Type-1: the First Report in Thailand

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03207387

Position: Peer Reviewer

Academic degree: MD

Professional title: Dean

Reviewer's Country/Territory: China

Author's Country/Territory: Thailand

Manuscript submission date: 2022-04-22

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-04-22 05:45

Reviewer performed review: 2022-04-29 03:36

Review time: 6 Days and 21 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish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



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

## SPECIFIC COMMENTS TO AUTHORS

The authors reported two cases with leukocyte adhesion defect type 1 (LAD-1) and found they were caused by ITGB2 mutations. The two cases presented recurrent omphalitis, soft tissue 36 infection, marked leukocytosis and neutrophilia. Mutation analysis was performed using direct DNA sequencing of the ITGB2 gene. The results revealed two novel homozygous missense mutations; c.920C>T (p.Leu307Pro) in exon 8 and c.758G>A (p.Arg253His) in exon7, and one novel homozygous nonsense mutation; c.262C>T (p.Gln88Ter) in exon 4, in the genomic DNA of the first and second patients, respectively. Heterozygous of the same mutations were identified in the parents of both patients suggesting carrier status. The authors did not describe the detailed information of the two cases since the ages of onset were quite early and there were much present history were ignored. It is quite uncommon that there were two homozygous missense mutations identified in one case. At least 200-500 normal controls need to be tested.



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Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

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Reviewer's code: 04152279

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: China

Author's Country/Territory: Thailand

Manuscript submission date: 2022-04-22

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-05-17 00:47

Reviewer performed review: 2022-05-19 14:50

Review time: 2 Days and 14 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Language quality	<ul> <li>[ ] Grade A: Priority publishing [Y] Grade B: Minor language polishing</li> <li>[ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection</li> </ul>
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## SPECIFIC COMMENTS TO AUTHORS

1.If the keywords can be added "bacterial soft tissue infection" and "molecular investigation", it can make the article easier to be searched; 2.Please refine the format of the article so that it will look more formal.