

## PEER-REVIEW REPORT

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

**Manuscript NO:** 66540

**Title:** Congenital disorder of glycosylation caused by mutation of ATP6AP1 gene (c.1036G>A) in a Chinese infant: A case report

**Reviewer's code:** 01518946

**Position:** Editorial Board

**Academic degree:** MD, PhD

**Professional title:** Professor, Research Fellow, Senior Consultant Dermatologist

**Reviewer's Country/Territory:** Japan

**Author's Country/Territory:** China

**Manuscript submission date:** 2021-04-12

**Reviewer chosen by:** AI Technique

**Reviewer accepted review:** 2021-04-28 08:43

**Reviewer performed review:** 2021-04-29 02:34

**Review time:** 17 Hours

<b>Scientific quality</b>	<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 Grade E: Do not publish
<b>Language quality</b>	<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
<b>Conclusion</b>	<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
<b>Re-review</b>	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No
<b>Peer-reviewer statements</b>	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**

This manuscript describes a case report of congenital disorder of glycosylation (CDG) caused by mutation of ATP6AP1 gene (c.1036G>A) in a Chinese infant. CDG patients have recently been increased with analysis of next-generation DNA sequences. The cause of CDG is various, including glyco-genes and their chaperon. ATP6AP1 gene is one of the causative genes for CDG. There are some reports on ATP6AP1 gene mutation on CDG patients. While c.1036G>A mutation in ATP6AP1 gene was reported in CDG patients previously, this report is the first case in Asian people. The authors described a history and clinical examination very clearly and discussed well according to many reports on CDG patients with ATP6AP1 gene mutation. Therefore, this manuscript is acceptable for publication as a case report in the world journal of gastroenterology.

## PEER-REVIEW REPORT

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

**Manuscript NO:** 66540

**Title:** Congenital disorder of glycosylation caused by mutation of ATP6AP1 gene (c.1036G>A) in a Chinese infant: A case report

**Reviewer's code:** 02942437

**Position:** Peer Reviewer

**Academic degree:** MD

**Professional title:** Doctor

**Reviewer's Country/Territory:** Japan

**Author's Country/Territory:** China

**Manuscript submission date:** 2021-04-12

**Reviewer chosen by:** AI Technique

**Reviewer accepted review:** 2021-04-30 12:47

**Reviewer performed review:** 2021-05-06 17:10

**Review time:** 6 Days and 4 Hours

<b>Scientific quality</b>	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input checked="" type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
<b>Language quality</b>	<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
<b>Conclusion</b>	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input type="checkbox"/> Minor revision <input checked="" type="checkbox"/> Major revision <input type="checkbox"/> Rejection
<b>Re-review</b>	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No
<b>Peer-reviewer statements</b>	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**

Yang X et al. presented a very rare case of ATP6AP1-CDG and review of related-literatures. Patients with ATP6AP1-CDG have been previously reported, therefore I'm afraid that there is a lack of novelty in this manuscript. Major points 1. ATP6AP1-CDG is very rare and the authors demonstrate that the present case is the first report in East Asia; however, 18 cases with ATP6APA-CDG have and 6 cases with c.1036G>A mutation have been reported in the world wide. Thus, I'm afraid that there is a lack of novelty in this manuscript. The authors should present novelty other than that this case is the first report in East Asia countries. 2. The authors should describe the clinical information in more detail. How did transaminase and ammonia levels decrease after treatment? The authors should indicate blood examinations in details. Platelet counts, fibrosis marker (type IV collagen 7S and M2BPGi), LDH, PT%, albumin, ChE, T-Cho, ALP, and GGT... 3. The author should present the images of hepatosplenomegaly (US or CT) which is an important manifestation in patients with c.1036G>A mutation type. Minor point 1. In Table 2, the abbreviation is incorrect regarding "CL".

## RE-REVIEW REPORT OF REVISED MANUSCRIPT

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

**Manuscript NO:** 66540

**Title:** Congenital disorder of glycosylation caused by mutation of ATP6AP1 gene (c.1036G>A) in a Chinese infant: A case report

**Reviewer's code:** 02942437

**Position:** Peer Reviewer

**Academic degree:** MD

**Professional title:** Doctor

**Reviewer's Country/Territory:** Japan

**Author's Country/Territory:** China

**Manuscript submission date:** 2021-04-12

**Reviewer chosen by:** Chen-Chen Gao

**Reviewer accepted review:** 2021-07-05 09:46

**Reviewer performed review:** 2021-07-05 11:51

**Review time:** 2 Hours

<b>Scientific quality</b>	<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
<b>Language quality</b>	<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
<b>Conclusion</b>	<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
<b>Peer-reviewer statements</b>	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

The authors well responded to reviewer's comments; however, I request again as follows. Although some blood examinations were added in Table 1, the authors did not indicate the transaminase levels and ammonia levels after treatment. I request detailed values again.