

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 43213

Title: Recurrent acute liver failure associated with novel SCY Y mutation: Case report

Reviewer's code: 03727922

Reviewer's country: Brazil

Science editor: Xue-Jiao Wang

Date sent for review: 2018-12-03

Date reviewed: 2018-12-03

Review time: 4 Hours

SCIENTIFIC QUALITY	LANGUAGE QUALITY	CONCLUSION	PEER-REVIEWER STATEMENTS
<input type="checkbox"/> Grade A: Excellent	<input type="checkbox"/> Grade A: Priority publishing	<input type="checkbox"/> Accept	Peer-Review:
<input checked="" type="checkbox"/> Grade B: Very good	<input checked="" type="checkbox"/> Grade B: Minor language	(High priority)	<input checked="" type="checkbox"/> Anonymous
<input type="checkbox"/> Grade C: Good	polishing	<input type="checkbox"/> Accept	<input type="checkbox"/> Onymous
<input type="checkbox"/> Grade D: Fair	<input type="checkbox"/> Grade C: A great deal of	(General priority)	Peer-reviewer's expertise on the
<input type="checkbox"/> Grade E: Do not	language polishing	<input type="checkbox"/> Minor revision	topic of the manuscript:
publish	<input type="checkbox"/> Grade D: Rejection	<input checked="" type="checkbox"/> Major revision	<input checked="" type="checkbox"/> Advanced
		<input type="checkbox"/> Rejection	<input type="checkbox"/> General
			<input type="checkbox"/> No expertise
			Conflicts-of-Interest:
			<input type="checkbox"/> Yes
			<input checked="" type="checkbox"/> No

SPECIFIC COMMENTS TO AUTHORS

The present manuscript entitled "Recurrent acute liver failure associated with novel SCYL1 mutation: Case report" is intreresting. However, I believe that needs improvement in some topics of your manuscript to a better interpretation and to emphasize its importance. Introduction - -Clarify the hypothesis stated and the



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purpose of the study. Case presentation - Information on approval of a Local Ethical Committee should also be provided in a specific section, if it possible describe the number and year. Provide the information on patients informed consent. Should describe clearly the the others ALF treatment? Has genetic evaluation influenced the treatment of the patient? What about costs? Discussion:What were the limitations of your study? What are the real benefits of your study to the clinical and scientific practice? What is the literature regarding this specific evaluation?

INITIAL REVIEW OF THE MANUSCRIPT

Google Search:

- ☐ The same title
- ☐ Duplicate publication
- ☐ Plagiarism
- ☐ No

BPG Search:

- ☐ The same title
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- ☐ No

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 43213

Title: Recurrent acute liver failure associated with novel SCY Y mutation: Case report

Reviewer's code: 03471268

Reviewer's country: Japan

Science editor: Xue-Jiao Wang

Date sent for review: 2018-12-03

Date reviewed: 2018-12-14

Review time: 11 Days

SCIENTIFIC QUALITY	LANGUAGE QUALITY	CONCLUSION	PEER-REVIEWER STATEMENTS
<input type="checkbox"/> Grade A: Excellent	<input type="checkbox"/> Grade A: Priority publishing	<input type="checkbox"/> Accept	Peer-Review:
<input type="checkbox"/> Grade B: Very good	<input type="checkbox"/> Grade B: Minor language	(High priority)	<input type="checkbox"/> Anonymous
<input type="checkbox"/> Grade C: Good	polishing	<input type="checkbox"/> Accept	<input type="checkbox"/> Onymous
<input type="checkbox"/> Grade D: Fair	<input type="checkbox"/> Grade C: A great deal of	(General priority)	Peer-reviewer's expertise on the
<input type="checkbox"/> Grade E: Do not	language polishing	<input type="checkbox"/> Minor revision	topic of the manuscript:
publish	<input type="checkbox"/> Grade D: Rejection	<input type="checkbox"/> Major revision	<input type="checkbox"/> Advanced
		<input type="checkbox"/> Rejection	<input type="checkbox"/> General
			<input type="checkbox"/> No expertise
			Conflicts-of-Interest:
			<input type="checkbox"/> Yes
			<input type="checkbox"/> No

SPECIFIC COMMENTS TO AUTHORS

This article revealed mutations of SCYL1 for Chinese patients of RALF with whole exome sequencing. Searching for mutations in minimal cases with neurologic or skeletal abnormalities is rare and may be an aid for future diagnosis. It is the first report in east Asia, and it is considered to be a useful case report.



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- ☐ No

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 43213

Title: Recurrent acute liver failure associated with novel SCY Y mutation: Case report

Reviewer's code: 03251829

Reviewer's country: Greece

Science editor: Xue-Jiao Wang

Date sent for review: 2018-12-03

Date reviewed: 2018-12-14

Review time: 11 Days

SCIENTIFIC QUALITY	LANGUAGE QUALITY	CONCLUSION	PEER-REVIEWER STATEMENTS
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<input checked="" type="checkbox"/> Grade C: Good	polishing	<input type="checkbox"/> Accept	<input type="checkbox"/> Onymous
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publish	<input type="checkbox"/> Grade D: Rejection	<input checked="" type="checkbox"/> Major revision	<input type="checkbox"/> Advanced
		<input type="checkbox"/> Rejection	<input checked="" type="checkbox"/> General
			<input type="checkbox"/> No expertise
			Conflicts-of-Interest:
			<input type="checkbox"/> Yes
			<input checked="" type="checkbox"/> No

SPECIFIC COMMENTS TO AUTHORS

This case report reports a novel homozygous insertion mutation in a patient diagnosed with pediatric recurrent acute liver failure (RALF). The authors have exploited WES to describe a 7bp insertion (GGGCCCT). The authors claim to expand the clinical mutational spectrum of SCLY1 disease. The authors report the mutation is not reported

in datasets such as 1000 genomes, the NHLBI Exome Sequencing Project (ESP) Exome Variant Server (6,500 exomes), or the Exome Aggregation Consortium Browse Comments: -To strengthen their argument they should genotype/confirm the mutation with a PCR assay with allele specific primers and conventional DNA sequencing in both the parents and child. For instance new Scyl1 mutations have been detected by WES and confirmed by PCR in the following publications: EMBO Rep. 2007 Jul;8(7):691-7. Am J Hum Genet. 2015 Dec 3;97(6):855-61. -In page 6 line 27 (Identification of a novel homozygous mutation in SCYL1) the authors should report the exact position of the mutation mentioning the exon or intron of the insertion. -In page 7 line 6 the authors write "The variant appears novel". The words "appears" denotes uncertainty, is not ideal. They should state "The variant is novel". -The authors should state if the mutation detected by exome sequencing concurs with Sanger sequencing

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