

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 53340

Title: A novel frameshift mutation in the SACS gene causing Spastic Ataxia of Charlevoix-Saguenay in a consanguineous family from the Arabian Peninsula

Reviewer's code: 00646291

Position: Editorial Board

Academic degree: PhD

Professional title: Senior Lecturer

Reviewer's Country/Territory: United Kingdom

Author's Country/Territory: Kuwait

Manuscript submission date: 2019-12-23

Reviewer chosen by: Ruo-Yu Ma

Reviewer accepted review: 2020-02-07 11:55

Reviewer performed review: 2020-02-13 12:58

Review time: 6 Days and 1 Hour

SCIENTIFIC QUALITY	LANGUAGE QUALITY	CONCLUSION	PEER-REVIEWER STATEMENTS
<input type="checkbox"/> Grade A: Excellent	<input checked="" type="checkbox"/> Grade A: Priority publishing	<input type="checkbox"/> Accept	Peer-Review:
<input checked="" type="checkbox"/> Grade B: Very good	<input type="checkbox"/> Grade B: Minor language	(High priority)	<input checked="" type="checkbox"/> Anonymous
<input type="checkbox"/> Grade C: Good	polishing	<input checked="" 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 checked="" type="checkbox"/> General
			<input type="checkbox"/> No expertise
			Conflicts-of-Interest:
			<input type="checkbox"/> Yes
			<input checked="" type="checkbox"/> No



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SPECIFIC COMMENTS TO AUTHORS

The case study is informative, clearly presented and provides novel findings. Minor spelling errors need to be corrected (page 8: CASE PRESENTAION and page 24: blew).

INITIAL REVIEW OF THE MANUSCRIPT

Google Search:

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

BPG Search:

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

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 53340

Title: A novel frameshift mutation in the SACS gene causing Spastic Ataxia of Charlevoix-Saguenay in a consanguineous family from the Arabian Peninsula

Reviewer's code: 00722239

Position: Editorial Board

Academic degree: MD, PhD

Professional title: Associate Professor

Reviewer's Country/Territory: Japan

Author's Country/Territory: Kuwait

Manuscript submission date: 2019-12-23

Reviewer chosen by: Le Zhang

Reviewer accepted review: 2020-03-12 03:49

Reviewer performed review: 2020-03-15 12:26

Review time: 3 Days and 8 Hours

SCIENTIFIC QUALITY	LANGUAGE QUALITY	CONCLUSION	PEER-REVIEWER STATEMENTS
<input checked="" 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 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 checked="" 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 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

The authors have reported a case of autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). ARSACS is very rare disease and this is first case in Arabian Peninsula. In addition, they identified a novel pathogenic frameshift mutation in the SACS gene by whole exome sequencing. The value and clinical implications of this case report is very high. I recommend publication of this report after minor revision. Although the authors referred to “Table 2 and 3” in Page 10, I could not find Table 2 nor Table 3. Please explain it. Figure 1a and Figure 1b appeared in wrong order in text. As Figure 1a (imaging findings) and Figure 1b (family pedigree) is different sort of Figures, they should be separated as independent figures. Please describe clinical course and treatment of the patient before discussion section.

INITIAL REVIEW OF THE MANUSCRIPT

Google Search:

- ☐ The same title
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- ☐ Plagiarism
- ☒ No

BPG Search:

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- ☐ Duplicate publication
- ☐ Plagiarism
- ☒ No