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ESPS PEER REVIEW REPORT

Name of journal: World Journal of Gastroenterology
ESPS manuscript NO: 12025
Title: Early hepatic complications of Lysosomal acid lipase deficiency in Mexican siblings with new mutations in LIPA gene
Reviewer code: 02860875
Science editor: Yuan Qi
Date sent for review: 2014-06-18 16:22
Date reviewed: 2014-07-08 04:38

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
[ ] Grade A: Excellent	[ ] Grade A: Priority publishing	Google Search:	[Y] Accept
[ ] Grade B: Very good	[ ] Grade B: Minor language polishing	[ ] Existing	[ ] High priority for publication
[Y] Grade C: Good	[Y] Grade C: A great deal of language polishing	[ ] No records	
[ ] Grade D: Fair	[ ] Grade D: Rejected	BPG Search:	[ ] Rejection
[ ] Grade E: Poor		[ ] Existing	[ ] Minor revision
		[ ] No records	[ ] Major revision

COMMENTS TO AUTHORS

Thank you for asking me to review this manuscript. Santillán-Hernández et al have produced a case report of two siblings who were found to have cholesterol ester storage disorder. They have described the clinical scenario and identified that the two cases have a novel compound heterozygote genotype for the LIPA gene. The cases are described adequately, diagnosed appropriately and the novel genotype adds to the previously published literature. I have the following comments: Major 1. The written English needs to be improved. There are numerous spelling and grammatical errors. 2. I am not sure that we can describe the clinical course here as unique; Bernstein et al described a spectrum of clinical courses that almost certainly reflects the functionality of the different mutations of the LIPA gene. I would tone down the references to early presentation and prolonged clinical course as somehow noteworthy. 3. Please redraft figure 3; I cannot see how a complete series of US images helps the description of the case. Similarly I am not sure that a complete description of the velocity of venous blood flow within the abdomen for both cases helps the flow of the report. 4. You should emphasise that the blood spot LAL values for the parents are sub-normal for the mother and low normal for the father. Have you performed an US on either parent? Minor 1. The OMIM number in the abstract is wrong; it should be 278000. 2. Please could we have reference values for your laboratory (Table 1) 3. Did the CT scan of sibling 1 at age 4 demonstrate adrenal calcification?

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ESPS PEER REVIEW REPORT

Name of journal: World Journal of Gastroenterology
ESPS manuscript NO: 12025
Title: Early hepatic complications of Lysosomal acid lipase deficiency in Mexican siblings with new mutations in LIPA gene
Reviewer code: 02861012
Science editor: Yuan Qi
Date sent for review: 2014-06-18 16:22
Date reviewed: 2014-07-13 22:21

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
[ ] Grade A: Excellent	[ ] Grade A: Priority publishing	Google Search:	[Y] Accept
[ ] Grade B: Very good	[Y] Grade B: Minor language polishing	[ ] Existing	[ ] High priority for publication
[Y] Grade C: Good	[ ] Grade C: A great deal of language polishing	[ ] No records	
[ ] Grade D: Fair	[ ] Grade D: Rejected	BPG Search:	[ ] Rejection
[ ] Grade E: Poor		[ ] Existing	[ ] Minor revision
		[ ] No records	[ ] Major revision

COMMENTS TO AUTHORS

This study is very clear and well written. The authors show two new mutations in exon 4 of the LIPA gene, which encodes for lysosomal acid lipase (LAL). Clinically LAL deficiency is reported as having one of the two principal phenotypic presentations: the early onset, called Wolman disease (WD) and the late onset called cholesteryl ester storage disease (CESD). The new mutations described here cause a total deficiency of LAL activity and lead to a CESD presentation with early symptomatology and complications. Comments to authors: The authors should report the frequency of these new mutations in healthy controls, and whether there is difference in the presence of these mutations in individuals of different origin.

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ESPS PEER REVIEW REPORT

Name of journal: World Journal of Gastroenterology
ESPS manuscript NO: 12025
Title: Early hepatic complications of Lysosomal acid lipase deficiency in Mexican siblings with new mutations in LIPA gene
Reviewer code: 02860966
Science editor: Yuan Qi
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CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
[ ] Grade A: Excellent	[ ] Grade A: Priority publishing	Google Search:	[ ] Accept
[Y] Grade B: Very good	[Y] Grade B: Minor language polishing	[ ] Existing	[ ] High priority for publication
[ ] Grade C: Good	[ ] Grade C: A great deal of language polishing	[ ] No records	
[ ] Grade D: Fair		BPG Search:	[ ] Rejection
[ ] Grade E: Poor	[ ] Grade D: Rejected	[ ] Existing	[Y] Minor revision
		[ ] No records	[ ] Major revision

COMMENTS TO AUTHORS

Thank you for the opportunity to review this interesting paper.