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Flat C, 23/F., Lucky Plaza,
315-321 Lockhart Road,
Wan Chai, Hong Kong, China

ESPS Peer-review Report

Name of Journal: World Journal of Gastroenterology

Ms: 2597

Title: Different regional distribution of SLC25A13 mutations in Chinese patients with neonatal intrahepatic cholestasis

Reviewer code: 02444760

Science editor: x.z.huang@wjgnet.com

Date sent for review: 2013-03-03 18:35

Date reviewed: 2013-03-17 18:01

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A (Excellent)	<input type="checkbox"/> Grade A: Priority Publishing	Google Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B (Very good)	<input checked="" type="checkbox"/> Grade B: minor language polishing	<input type="checkbox"/> Existed	<input type="checkbox"/> High priority for publication
<input checked="" type="checkbox"/> Grade C (Good)	<input type="checkbox"/> Grade C: a great deal of language polishing	<input type="checkbox"/> No records	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade D (Fair)		BPG Search:	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade E (Poor)	<input type="checkbox"/> Grade D: rejected	<input type="checkbox"/> Existed	<input checked="" type="checkbox"/> Minor revision
		<input type="checkbox"/> No records	<input type="checkbox"/> Major revision

COMMENTS



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COMMENTS TO AUTHORS:

The manuscript of ‘Different regional distribution of SLC25A13 mutations in Chinese patients with neonatal intrahepatic cholestasis’ focuses on the mutation spectra of SLC25A13 gene in different parts of China. As defined by the mutation point, c.851_854del, c.1638_1660dup23, c.615+5G>A and c.1750+72_1751-4dup17insNM_138459.3:2667 are the hotspots. Thirteen other mutation types have also been recorded. As compared to that of border and northern regions, mutant alleles seem to be more common in the southern region. These findings may shed light on the genetic characteristics of neonatal intrahepatic cholestasis in Chinese patients and, as a result, serve as potential tool for the clinical diagnosis. Major comments 1. This research investigates the frequency of SLC25A13 mutations in 3 regions of China. In result, the mutation spectrum of SLC25A13 gene varies considerably among different regions of China. Four common mutations display higher frequency in the southern region than in border and northern regions. However, only 61 patients enrolled in the study are from northern China, which is much lower than that of border and southern regions. Subsequently, there are merely 4 mutant alleles found in this population. Then whether the bias of sampling should be responsible for the genetic difference between 3 regions of China? Additional data and, at least, detailed discussion are suggested. 2. Different frequency of common mutations has been reported in various areas of China. Is there any inconsistency among previous reports and the present study? Thorough comparison and rational explanation would be appreciated. Minor comments 1. Some expression, such as ‘In north the mutation c.851_854del accounted for 50% (2/4) of the mutant alleles.’ (P. 11), ‘with different mutations having a higher proportion in the border and north regions than in the south regions’ (P. 12), seem to be unsuitable and even mistake. 2. The manuscript illustrates the distribution of mutant alleles that have been enrolled in the study. Unfortunately, the reviewer can only find 2 parts, not 3 parts of China, as separated by the Yangtze River. The southern, border and northern areas would be better clearly distinguished, such as by different colors. What is worse, both Liaoning and Henan have been labeled in the wrong place.



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Name of Journal: World Journal of Gastroenterology

Ms: 2597

Title: Different regional distribution of SLC25A13 mutations in Chinese patients with neonatal intrahepatic cholestasis

Reviewer code: 00053433

Science editor: x.z.huang@wjgnet.com

Date sent for review: 2013-03-03 18:35

Date reviewed: 2013-03-18 20:33

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A (Excellent)	<input type="checkbox"/> Grade A: Priority Publishing	Google Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B (Very good)	<input type="checkbox"/> Grade B: minor language polishing	<input type="checkbox"/> Existed	<input type="checkbox"/> High priority for publication
<input type="checkbox"/> Grade C (Good)	<input type="checkbox"/> Grade C: a great deal of language polishing	<input type="checkbox"/> No records	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade D (Fair)	<input type="checkbox"/> Grade D: rejected	BPG Search:	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade E (Poor)		<input type="checkbox"/> Existed	<input type="checkbox"/> Minor revision
		<input type="checkbox"/> No records	<input type="checkbox"/> Major revision

COMMENTS

COMMENTS TO AUTHORS:

This is a retrospective study aimed at investigating the regional distribution of SLC25A13 mutations in Chinese patients with neonatal intrahepatic cholestasis. The topic is relevant, since biochemical diagnosis of citrin deficiency is not widely available and mutation analysis of the SLC25A13 gene is crucial to diagnosis. The study was well-conducted and the manuscript is reasonably well written (although English review is obviously needed), with good scientific value. However, some issues should be addressed by the authors. 1) Authors are strongly suggested to ask a native English speaker to check the manuscript for spelling and grammar. Text comprehension is compromised in many parts of the manuscript. 2) For the sake of clarity, the results of the inclusion process and mutation identification should be presented in a flow-chart. 3) Authors should clarify if a WRITTEN informed consent was obtained from all participants or their guardians. 4) Table 1. Authors should indicate the reference number for each study.