

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 54228

Title: Cholesteryl ester storage disease of clinical and genetic characterisation: A case report and review of literature

Reviewer's code: 00035033

Position: Editorial Board

Academic degree: MD

Professional title: Associate Professor

Reviewer's Country/Territory: Italy

Author's Country/Territory: Denmark

Manuscript submission date: 2020-01-15

Reviewer chosen by: AI Technique

Reviewer accepted review: 2020-01-15 18:13

Reviewer performed review: 2020-01-15 20:57

Review time: 2 Hours

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 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 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

This is a very accurate and well-presented description of the clinical and genetic characterisation of two adult siblings with cholesterol ester storage disease (CESD). Both subjects presented progressive hepatic failure leading to hepatic transplantation and advanced multiorgan atherosclerosis. Both patients show signs of recurrence of CESD in the liver after transplantation. Moreover, three family members who were LIPA heterozygous had a lysosomal acid lipase (LAL) activity below the reference value. Since analyses of SNPs showed variants with an increased risk of non-alcoholic fatty liver disease and fibrosis for both patients, I suggest that the Authors discuss the clinical case also taking into consideration the following recent publications: Baratta F et al. Reduced lysosomal acid lipase activity: A new marker of liver disease severity across the clinical continuum of non-alcoholic fatty liver disease? *World J Gastroenterol.* 2019 Aug 14;25(30):4172-4180 Baratta F et al. Lysosomal acid lipase activity and liver fibrosis in the clinical continuum of non-alcoholic fatty liver disease. *Liver Int.* 2019 Dec;39(12):2301-2308 Angelico F et al. Severe reduction of blood lysosomal acid lipase activity in cryptogenic cirrhosis: A nationwide multicentre cohort study. *Atherosclerosis.* 2017; 262:179-184

INITIAL REVIEW OF THE MANUSCRIPT

Google Search:

- ☐ The same title
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- ☐ No



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- ☐ The same title
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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 54228

Title: Cholesteryl ester storage disease of clinical and genetic characterisation: A case report and review of literature

Reviewer's code: 02444978

Position: Editorial Board

Academic degree: MD

Professional title: Professor

Reviewer's Country/Territory: Italy

Author's Country/Territory: Denmark

Manuscript submission date: 2020-01-15

Reviewer chosen by: AI Technique

Reviewer accepted review: 2020-01-16 12:01

Reviewer performed review: 2020-01-22 16:41

Review time: 6 Days and 4 Hours

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 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 type="checkbox"/> Accept	<input type="checkbox"/> Onymous
<input checked="" 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

This paper describes the case of two siblings affected by CESD who underwent liver transplantation. The siblings were clinically and genetically evaluated, recognizing they were compound heterozygous for the missense variant in LIPA exon 8. Their first-degree family members were also examined. Although this paper does not contains relevant novelty about CESD, the disease is presented in a clear and complete way in the background, with a correct diagnostic and therapeutic conduct. Consequently, the paper offers an example of how to diagnose and manage this rare disease, proposing CESD as a multi-organ disease, the progression of which may occur post-liver transplantation. I have some revisions concerning the exposition of the data. In general, the description of the subjects involved in the study is confused. This part needs to be more rigorously and clearly reviewed. There is a discrepancy between table 1 and the other two tables: the number of subjects examined is different. Moreover, it is not easy to identify which subjects the genetic variations correspond to, and what are their clinical characteristics. These aspects need to be better specified. Figure 2 is missing. In table 2 and 3 does not seem to be reported the data of siblings, as stated in the text. In the Family members chapter it is stated that “None of the family members showed evidence of LAL-D in blood tests (Table 1)”, but no data on LAL activity is reported in the table. Moreover, two adult family members and one child are mentioned, but in the table appear three adults and three children. Table 3 reports the “Analysis of SNPs in family members of patients with CESD”, and not LAL activity. In the discussion, page 11, line 20, it is stated that a “significant correlation between heterozygosity and LAL activity” was found, but no data are reported on this statistical analysis.

INITIAL REVIEW OF THE MANUSCRIPT



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