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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 46809

Title: c.753_754delAG, a novel CFTR mutation found in a Chinese patient with cystic fibrosis: A case report and review of literature

Reviewer's code: 03647617

Reviewer's country: Slovenia

Science editor: Ying Dou

Reviewer accepted review: 2019-04-25 13:24

Reviewer performed review: 2019-05-01 21:15

Review time: 6 Days and 7 Hours

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[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish	[ ] Grade A: Priority publishing [Y] Grade B: Minor language polishing [ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection	[ ] Accept (High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

SPECIFIC COMMENTS TO AUTHORS

Dear authors, Your paper titled c.753_754delAG, a novel CFTR mutation found in a Chinese patient with cystic fibrosis: A case report and review of literature has many good points, especially the new identified mutation and the table with all published

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patients with CF in China and their mutations. On the other hand a drawback of this paper is the fact that the diagnosis CF was not made independently with another diagnostic method, e.g. pilocarpine iontophoresis, but due to the scarcity of cases in China and, therefore also inaccessibility to this or other methods to diagnose CF, this may be overlooked. You appropriately discussed this problem in China in your paper. There are some data missing and some conclusions cannot be made according to the results. I list them one by one. Please, correct them or comment on them: • p. 1: in the title, maybe, it is better to replace "review of literature" with "review of published CF patients in China", if it is not too long • p. 6, physical examination: you should add the girl's height and percentiles, and calculate also body mass index and percentiles • p. 6, laboratory examination: did you perform any test for pancreatic insufficiency e.g. stool elastase or faecal fat excretion test? • p. 7, imaging: can you explain a bit more this focal liver lesion, as it is not typical for CF. Did you perform liver biopsy? • p. 7, imaging: instead of "Echocardiography of pancreas …" you probably meant "Ultrasonography … " or "Echography of …". • p. 8, Fig. 3: You show 3 different DNA sequences and wrote that the mutation of CF patient and mother is seen. But the pattern is similar but not exactly the same between patient and father. Is the figure right? Can you explain? • p. 9, treatment: which antibiotic did you prescribe and for how long? • p. 9, nutritional support: can you describe "nutritional support" more in detail. Which kind of food and advice was given? • p. 10, follow-up: what was nutritional status (body weight, height, and BMI)? Did you manage to eradicate P. aeruginosa infection? How did you check for pulmonary disease regression/progression? Did you introduce any regular physiotherapy or some treatment for pulmonary disease? • p. 16, discussion: you have to adapt the last two sentences. As it is not clear from your case if the patient had pancreatic sufficiency or insufficiency you cannot conclude like this. Elevated serum lipase, which has not been mentioned before, is not a sign of severe mutation, more of

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possible pancreatitis which is more commonly seen in heterozygous CF carriers or in those with milder mutations and pancreatic sufficiency. Please, discuss and/or adapt. • p. 18 and p. 24; references: references no. 5 and 41 are the same. Please, correct. I hope that you will be able to adapt the paper for the publication as it carries an important message for the readers. Yours sincerely, Reviewer

INITIAL REVIEW OF THE MANUSCRIPT

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