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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 72923

Title: CY Y7 mutation in a case of cerebrotendinous xanthomatosis: a case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03789236
Position: Peer Reviewer
Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Indonesia

Author's Country/Territory: China

Manuscript submission date: 2021-11-04

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-12-04 15:57

Reviewer performed review: 2021-12-05 00:04

Review time: 8 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [ ] Grade C: Good [Y] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[Y] Grade A: Priority publishing [ ] Grade B: Minor language polishing [ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [Y] Major revision [ ] Rejection
Re-review	[Y] Yes [ ] No
Peer-reviewer	Peer-Review: [ ] Anonymous [Y] Onymous

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statements	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

This article is interesting and well written, but it has some limitations: 1. Introduction: Please cite the related studies (these should be discussed at the discussion section). 2. Case: Please clearly elaborate why do a patient with such an almost-lifelong illness finally underwent genetic testing, even 2 years after a major surgery was done. A strong rationale must evidently be elaborated on each additional examination/investigation, not just random employment of new technology. 3. Discussion: Apart from CDCA, is there no other possible treatment for such a rare, devastating, and progressive disease? If none, please clearly state so. 4. Conclusion: The conclusion of "Prompt diagnosis and treatment of CTX improve patient outcomes" does NOT reflect the rather-gloomy outcome in this patient (After 1 year of treatment, the patient felt that the symptoms of weakness in both lower limbs had improved slightly); nor the 33-year duration from onset to diagnosis. 5. Illustrations: More comprehensive clinical pictures are absolutely needed to give the readers more insight about this rare, devastating disase. Especially if the disease is severely pronounced, such as in this case. Considering these changes be incorporated in the revised manuscript version, the article can be accepted for publication in this reputed journal - for now some "major revisions" are needed.

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Reviewer's code: 05314933
Position: Peer Reviewer
Academic degree: MD

Professional title: Associate Professor, Doctor

Reviewer's Country/Territory: India

Author's Country/Territory: China

Manuscript submission date: 2021-11-04

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-12-02 13:58

Reviewer performed review: 2021-12-20 01:44

Review time: 17 Days and 11 Hours

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[Y] Grade A: Priority publishing [ ] Grade B: Minor language polishing [ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [Y] Minor revision [ ] Major revision [ ] Rejection
Re-review	[Y] Yes [ ] No
Peer-reviewer	Peer-Review: [Y] Anonymous [ ] Onymous

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statements	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The manuscript is well written. I have few minor comments which will enhance the quality of manuscript and give a semblance of completeness to it. 1. Kindly discuss the neurological and MSK features of CTX on MRI briefly in discussion. 2. It would be worthwhile to discuss the biochemical tests like lipid profile. 3. The resolution of MRI images is not up to the mark. If available kindly add images of higher resolution. 4. If known kindly add what is the normal function of CYP27A1 gene in human body.