

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 57897

Title: Chinese patient with cerebrotendinous xanthomatosis confirmed by genetic testing:
A case report and review of literature

Reviewer's code: 03549400

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: China

Author's Country/Territory: China

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Reviewer chosen by: AI Technique

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Scientific quality	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input checked="" type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No
Peer-reviewer statements	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous Conflicts-of-Interest: <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No

SPECIFIC COMMENTS TO AUTHORS

Cao et al. reported a Chinese family with CTX characterized by psychiatric signs, enlarged Achilles tendons and nodules on the bilateral tibial tubercles. Besides, they summarize the clinical and genetic characteristics of all the reported CTX patients worldwide during the past 30 years. The results of this study may benefit researchers in this field. Minor points: 1. "The patient is a 47-year-old male mainly manifesting psychiatric signs but none of the cardinal features of CTX such as cataracts, cerebellar ataxia, pyramidal signs and chronic diarrhea...", this sentence is not accurate as the patient has enlarged Achilles tendons and nodules on the bilateral tibial tubercles. 2. The patient had a poor mental state, did he evaluated by any neuropsychiatric scale? 3. "Cerebrospinal fluid (CSF) was mildly elevated: 680.4 mg/L (normal range: 150-400 mg/L)." What does this sentence mean? 4. Specific abnormalities in figures should be marked with arrow(s) to more clearly point out abnormalities for non-CTX experts. Figure 2H, white matter defects 5. First-line treatment of CTX is based on chenodeoxycholic acid (CDCA) replacement therapy. Authors need to explain why this patient did not receive CDCA treatment. 6. As a Chinese CTX case report, I suggest the authors to cite some of the Chinese CTX reports, such as: Tao et al. Clinical and genetic characteristics of Chinese patients with cerebrotendinous xanthomatosis. Orphanet J Rare Dis. 2019 Dec 3;14(1):282. doi: 10.1186/s13023-019-1252-9. Chen et al. Clinical and molecular genetic features of cerebrotendinous xanthomatosis patients in Chinese families. Metab Brain Dis. 2017 Oct;32(5):1609-1618.