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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 62413

Title: Idiopathic basal ganglia calcification associated with new MYORG mutation site: A case report

Reviewer's code: 04409361
Position: Peer Reviewer
Academic degree: MD
Professional title: Consultant Physician-Scientist
Reviewer's Country/Territory: Japan
Author's Country/Territory: China

Manuscript submission date: 2021-01-11
Reviewer chosen by: AI Technique
Reviewer accepted review: 2021-04-09 09:14
Reviewer performed review: 2021-04-09 10:28
Review time: 1 Hour

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[ ] Grade A: Priority publishing [Y] 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 statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

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SPECIFIC COMMENTS TO AUTHORS

Thank you for the opportunity to review this manuscript. The authors present an impressive case report with pedigree and I am very interested in the patients with IBGC associated with novel mutations in MYORG. This case report is worth publishing worldwide, but the discussions mentioned below can be improved. Major comments #1 I wonder if the mutations c.1438T>G and c.1271_1272 TGGTGCGC are in one allele or two alleles in Patient II-7. Given the patients III-3 and III-4 has only c.1271_1272 TGGTGCGC mutation and the patient III-5 has only c.1438T>G mutation, I suspect that the two mutations exist in separate alleles in II-7. III-3, III-4, III-5 have no clinical symptoms or calcifications on CT assumingly because they are still 30es. On the other hand, the patient II-5 (66-year-old) has only c.1271_1272 TGGTGCGC mutation and has SLIGHT calcifications on CT WITHOUT clinical symptoms. Moreover, the patient II-7 has PROMINENT calcifications on CT and PROMINENT clinical symptoms. I suspect that MYORG may be autosomal dominant gene, but II-7 accidentally affected by mutations on both alleles. So, could the authors elaborate the discussion about Mendelian inheritance and the corresponding severity of this family? #2 Total Calcification Score (Nicolas et al., Brain 2013, doi:10.1093/brain/awt255) can be assessed and incorporated into the Table1 (all family members whose cranial CT examined). This may increase the readability and may help interpret the discussion regarding #1. #3 Is there a previous evidence of relationship between mutation sites and phenotypes in MYORG, like SLC20A2 (Nishii et al., Sci Rep 2019, doi: 10.1038/s41598-019-53401-0)? If the novel mutations in the present reports are presented with known mutation sites and discussed, the impact of the present report probably increases. #4 Could the author add the discussion of the needs of the genetic counseling and the further survey of the family member without prominent clinical symptoms? I believe that there are human

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rights not to know their genetic sequences ethically. (But this is, without fear of misunderstanding, the opposite attitude from a scientific viewpoint.) Minor comments

#5 Is the patient 65-year-old (summary), 61-year-old (Chief complaints), 63-year-old (Age at evaluation in Table 1)? If the first visit and the evaluation time are different, could the authors revise the case presentation for the readers to understand easily? #6 Gene names should be written in italics.

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RE-REVIEW REPORT OF REVISED MANUSCRIPT

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Professional title: Consultant Physician-Scientist
Reviewer's Country/Territory: Japan
Author's Country/Territory: China

Manuscript submission date: 2021-01-11
Reviewer chosen by: Jia-Ru Fan
Reviewer accepted review: 2021-05-08 03:50
Reviewer performed review: 2021-05-08 04:14
Review time: 1 Hour

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I read through your polite letter and revision. Only one thing I am afraid is that Total calcification scores of 54 and 10 in Table 1 may be misplaced. After the author addressing the point, I endorse for publication.