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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 51342

Title: Cutaneous nodules and a novel GNAS mutation in a Chinese boy with pseudohypoparathyroidism type Ⅰa: A case report and review of literature

Reviewer's code: 03883464
Position: Peer Reviewer
Academic degree: MD
Professional title: Doctor, Research Fellow
Reviewer's country: Portugal
Author's country: China

Reviewer chosen by: Artificial Intelligence Technique

Reviewer accepted review: 2019-12-01 22:05

Reviewer performed review: 2019-12-02 00:25

Review time: 2 Hours

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[ ] Grade E: Do not publish		[ ] Rejection	[ ] Advanced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

SPECIFIC COMMENTS TO AUTHORS

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The authors present a case of a 5-year-old proband and his mother, with hereditary pseudohypoparathyroidism type 1a, that presented a new GNAS mutation. The case is very well- written. I would suggest: 1. Please better describe the mechanism leading to PTH resistance in the kidney in PHP1a: As the maternal allele is the only source of Gs-alpha in the kidney (the paternal allele is normally silenced in this tissue), this maternal silencing results in marked reduction of Gs-alpha levels, leading to PTH resistance. 2. The authors present a case of hereditary PHP1a and describe a new mutation. Please present a supplementary table with other GNAS mutations previously described. What is the percentage of cases of sporadic PTH1a? 3. Please better explain heterotopic ossification in PHP1a, concerning its location and mechanism (hypercalcemia versus GNAS mutation induced osteogenic differentiation of stromal cells derived from adipose tissue while inhibiting their adipogenic differentiation).

INITIAL REVIEW OF THE MANUSCRIPT

Google Search:

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