

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 49852

Title: Recognizable type of pituitary, heart, kidney and skeletal dysplasia mostly caused by SEM A mutation: A case report

Reviewer's code: 02691156

Reviewer's country: Greece

Science editor: Ying Dou

Reviewer accepted review: 2019-06-28 14:40

Reviewer performed review: 2019-07-03 07:10

Review time: 4 Days and 16 Hours

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

SPECIFIC COMMENTS TO AUTHORS

Thank you for asking me to review the manuscript entitled "A recognizable type of pituitary, heart, kidney and skeletal dysplasia mostly caused by SEMA3A mutation: A case report". The title is referring directly to the problem at hand. The abstract is

sufficient Introduction is adequate and explanatory. The SEMA3A gene is located at 7q21.11 and has a length of 496,947 bp. Semaphorin-3A which is important in the development and migration of hypothalamic neurons. Case Report: You report a patient who exhibited pituitary, heart, kidney and skeletal dysplasia caused by the new mutation of the SEMA3A gene, growth hormone deficiency, short stature, hypogonadotropic hypogonadism, heart dysplasia, kidney dysplasia and skeletal dysplasia. TREATMENT The patient was treated with recombinant human growth hormone injections. He refuses further treatment about gonad. OUTCOME AND FOLLOW-UP His height has increased by 3 cm. In addition, he has a good appetite and reduced subcutaneous fat over 3 months of recombinant human growth hormone injections therapy. They will continue to observe his height, bone density and gonads. Discussion Two similar case reports have been published before their case. All patients not only exhibited short stature, facial dysmorphism and skeletal system anomalies but also had cardiovascular, urogenital, hearing, olfactory, visual, motor development and cognitive development defects. There were some differences between their patient and the two patients presented previously. No genes related to pituitary dysplasia were discovered in whole gene exome detection. Pituitary dysplasia might occur mainly because of defective hypothalamic neurons. You Conclude that Patients who come to a hospital because of their short stature must undergo gene detection if they have other simultaneous abnormal phenotypes. References: 8 papers are included in order to support the authors' views. Illustrations: 1 figure and 2 tables are included. This is an extremely rare case of abnormalities that should be investigated in any case of short stature admitted to the hospital for further investigation. Such very rare cases should be presented in order for the medical community to enrich its knowledge for the benefit of the patients. I wonder, in order to protect the personality of the patient, if it is better for the patient's eyes to be covered.



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INITIAL REVIEW OF THE MANUSCRIPT

Google Search:

- ☐ The same title
- ☐ Duplicate publication
- ☐ Plagiarism
- ☐ No

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- ☐ The same title
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