

# **Revised Cover Letter**

**Manuscript No.** 49852

**Title:** A recognizable type of pituitary, heart, kidney and skeletal dysplasia mostly caused by *SEMA3A* mutation: A case report

**Author:** Fang Hu and Liao Sun

**Dear professors,**

Thank you very much for your letter and thanks a lot for the reviewers' comments and kind suggestions of our manuscript. As suggested, we have carefully revised the manuscript and uploaded our revised manuscript. We hope it is acceptable for publication in the journal.

**With best regards,**

**Sincerely yours,**

**Liao Sun**

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## **Replies to the Editorial Comments**

*1. Please provide the author contributions. See the format in the attachment file-revision policies. The format of this section should be like this: Author contributions: Wang CL and Liang L contributed equally to this work; Wang CL, Liang L, Fu JF, Zou CC, Hong F and Wu XM designed research; Wang CL, Zou CC, Hong F and Wu XM performed research; Xue Jz and Lu JR contributed new reagents/analytic tools; Wang CL, Liang L and Fu JF analyzed data; and Wang CL, Liang L and Fu JF wrote the paper.*

**Response:** Thank you for your kind reviewing. We have rewritten this part. (line 24-25 on page 1)

*2. Please write a summary of less than 100 words to outline the most innovative and important arguments and core contents in your paper to attract readers*

**Response:** Thank you for your reviewing. We have rewritten this part. (line 1-7 on page 3)

*3. 请提供每张图片的原图，并整理在 PPT 中上传。（注意遮盖图 a 患者双眼部位）*

**Response:** Thank you for your suggestion. We have uploaded the pictures and covered the patient's eyes. We will pay more attention to protecting the privacy of patients in the future.

## **Replies to the Reviewer Report**

*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.*

**Response: Thank you very much for your careful review, helpful suggestions and encouragement. Your encouragement has made me realize the value of being a doctor and inspired me to work harder in the diagnosis and treatment of short stature. The manuscript has been revised thoroughly according to your advices and we have covered the patient's eyes to protect his privacy. We will pay more attention to protecting the privacy of patients in the future.**

**Again, we are grateful to the editor and the reviewers for the professional comments and constructive suggestions to improve our paper.**

**With best regards,**

**Sincerely yours,**

**Liao Sun**

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