

## Response to Reviewers Comments

We thank the reviewers for their thoughtful and supportive comments. We have revised our manuscript in response to their suggestions and hope that this improved manuscript is acceptable for publication in *World Journal of Clinical Cases*

Reviewer #1:

Specific Comments to Authors:

- The sequencing and analysis were approved by Beijing Mygenostics, Beijing, China. ....please elaborate on this agency
- The second generation sequencer Illumina NextSeq 500 was used to sequence the captured region at two ends, with a reading length of 150 bp.....who and where is the manufacturer?
- One photograph or more of the most important physical findings will attract the attention of the readers

Reply: Thank you for your valuable suggestions.

For your opinion, all the authors discussed the content of the article and made the following response here:

- MyGenostics Inc was originally established in Beijing China in November 2011, Which is a high-tech biotechnology company that provides life science instruments, reagents and technical services. The products and services cover scientific research, clinical and diagnostic industries.
- The Illumina NextSeq™ 500 system, used for genetic testing, was developed by Illumina (Corporate Headquarters: 5200 Illumina Way, San Diego, CA 92122). The system enables exome, genome-wide, and transcriptome sequencing in a single run, while also adjusting the sequencing fluxes as needed.
- We contacted the patient's family, who had agreed to and authorized our public publication of the genetic testing results, but we did not obtain patient public authorization for images of facial features and other pathological

features. We will then continue the treatment and follow-up of this patient to obtain complete clinical data.

We have adjusted the content of the article, added the specific data of the case and related discussions, and standardized the structure of the article.