Response to Reviewers Comments

Reviewer #1:

Specific Comments to Authors: Dear Editor, Thanks to the authors for this case report. The abstract summarize and reflect the work described in the manuscript. The manuscript adequately describes the background, present status and significance of the study. It interprets the findings adequately and appropriately. I think that it may contribute to the literature.

Reply: Thank you for your valuable suggestions.

Reviewer #2:

Specific Comments to Authors: AUTHORS Title: A novel frameshift mutation in the AHDC1 gene in a Chinese global developmental delay patient: A case report Authors report a novel frameshift mutation of c.1155dupG (p.Arg386Alafs*3) in the AHDC1 gene, found by high-throughput whole-exosome sequencing in an 11-month-old patient with global developmental delay, hypotonia, micrognathia, and other mild dysmorphic feature. This is a very short but interesting manuscript, being also well-written using robust scientific language. I do however have one concern as I cannot see the ethics permission on the manuscript (besides permission from tutors) and sensible data has been retrieved. I advise publication after minor modifications are made and the ethics issue is surpassed. Is the case 6 or 11 months old? At the end of introduction (11 mo), on case description is 6 mo. I believe the acronym "mo" stands for months-old. This hould be explained when first mentioned in the text. "The patient had been receiving rehabilitation treatment for nearly 6 mo", should be "The patient had been receiving rehabilitation treatment for nearly 6 months"? When authors say "The patient's liver function, kidney function, electrolytes, blood glucose, and organic acids in blood and urine showed no obvious abnormalities.". Could the word "obvious" be removed? Later, authors no longer use the mo acronym: "equivalent to that of a 4-month-old child," When stated "To improve the

quality of the patient," should it be "To improve the quality of the patients life"? When defining the Xia-Gibbs syndrome you state "(XGS, OMIM: 615829) is an autosomal dominant genetic disease caused by mutation of the AHDC1 gene. Typical features include global developmental delay, intellectual disability, structural abnormalities of the brain, global hypotonia, feeding problems, sleep difficulties, apnea[6], and short stature." Why was reference 6 placed not at the end of the sentence? Short stature is not referenced by 6? If do, please place alternative reference here.

Reply: Thank you for your valuable suggestions.

We have obtained informed consent from the families of the proband and attached the corresponding documents.

We have observed and treated this patient for nearly half a year, and some statements in the article may be misleading, and we have corrected the corresponding narrative.

We have modified the words and the references.