Dear Editor,

We'd like to thank the reviewers for their careful readings and valuable comments concerning our manuscript entitled "**Compound heterozygous mutations in the NEU1 gene in type 1 sialidosis: Case report and literature review**" (Manuscript NO: 57892). We believe the constructive feedback will improve the manuscript and increase its potential impact to the community. We read the comments carefully and respond to the reviewer's comments as followed:

Responds to the reviewer's comments:

Reviewer #1:

Response to comment: "It is mentioned that the patient' s parents did not exhibit the same marked symptoms than the patient which is affected by 2 mutations in the gene. it is not mentioned if they present with other mild symptoms. When compared with the other patients (Table 2), most of them with a S182G mutation, it would be expected to see some symptoms in the mother. The authors might want to explain this discrepancy."

Response: Sialidosis type 1 (ST-1) is a relatively mild, late-onset, and progressive lysosomal storage disease inherited with autosomal recessive inheritance. As the Table 2 demonstrated, homozygous mutations of 544A>G (Ser182Gly) and compound heterozygous mutations (Ser182Gly and an additional pathogenic variant) were the genetic cause of ST-1 in reported Chinese cases.

We feel obliged to clarify that the patient's parents did not complain any

symptoms like visual defects, myoclonus, ataxia, seizures and sensory loss. There was only one ST-1 patient (II:3) in this family with compound heterozygous mutations (c.239C>T (p.P80L) and c.544A>G (p.S182G)) in the *NEU1* gene. And the proband's mother harboring heterozygous mutation (wide type, p.S182G) was not expected to show related symptoms.

Special thanks to you for your good comments.

Dr. Guohua Zhao

Department of Neurology

The Fourth Affiliated Hospital, Zhejiang University School of Medicine

Yiwu, Zhejiang 322000

China