**Author's response to reviews**

**Title:** Sanfilippo type A: New Clinical Manifestations and Neuro-imaging findings in patients from the same family in Israel

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**Author's response to reviews:** see over
To,

The Editor, JMCR. Date: 13/11/2013

We are thankful to you and to the highly-experienced reviewers for their objective and constructive review of our manuscript, as well as for their valuable comments, which we are honored to respond to them and to incorporate the changes required into the manuscript. We are pleased to embody these changes (highlighted with green color) into our revised manuscript.

With our kind regards,

Dr. R. Sharkia and the Research Team.

Responses to Reviewers' comments:

Reviewer #1:

Comment-1: The authors may consider to include the reference of Verhoeven et al. (Acta Psychiatr Scand 122:162-165, 2009) in the section Introduction.
Response-1: The required reference (# 2) has been included in the Introduction section (page # 3, line # 4), as well as in the References section (reference # 2: page # 11, line # 6)

Reviewer #2

Comment-1: In the Abstract: Genetic examination through whole exome sequencing revealed a homozygous T139M mutation in the SGSH gene. Comment: T139M describes a mutation in a protein sequence NOT a gene sequence.
# change to; Genetic examination through whole exome sequencing revealed a homozygous mutation c.416C>T (p.T139M) mutation in the SGSH gene.
Response-1: The required change has been done in the Abstract section (page #2, line # 11).

Comment-2: In the Abstract: And in Conclusion: if similar clinical features are present during childhood, it is preferred to go directly for genetic diagnosis of Sanfilippo syndrome. Comment: Other lysosomal storage disorders (MPS I; Metachromatic leukodystrophy; Gaucher) may also present in a similar fashion in the early childhood years, hence screening for all lysosomal storage disorders may be more advantageous.
Response-2: The required change has been done in the Abstract section (page # 2, line # 18), as well as in the Conclusion section (page # 9, lines # 10 and 11).