Reviewer's report

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

Version: 2 Date: 6 November 2013

Reviewer: Litsa Karageorgos

Which of the following best describes what type of case report this is?: Unexpected or unusual presentations of a disease

Has the case been reported coherently?: Yes

Is the case report authentic?: Yes

Is the case report ethical?: Yes

Is there any missing information that you think must be added before publication?: No

Is this case worth reporting?: Yes

Is the case report persuasive?: Yes

Does the case report have explanatory value?: Yes

Does the case report have diagnostic value?: Yes

Will the case report make a difference to clinical practice?: Yes

Is the anonymity of the patient protected?: Yes

Comments to authors:

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

The paper by Sharkia et al., describes the diagnosis of two cases of Sanfilippo syndrome type A in Israel, believed to be the first report of its kind. This report describes the difficulties that doctors experience when diagnosing patients with lysosomal storage disorders.

Comments to the Author
I have no major criticism on this case report.

Only a few descriptive changes need to be made.
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.

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.

**Quality of written English:** Acceptable

**Declaration of competing interests:**

I declare that I have no competing interests