Reviewer’s report

Title: The eleventh reported case of Mulvihill-Smith syndrome in the literature

Version: 2 Date: 14 September 2013

Reviewer: Maria Cristina Digilio

Reviewer’s report:

The paper by Breinis et al. is reporting on a new patient with clinical features of Mulvihill-Smith syndrome. The case report is interesting considering the rarity of the syndrome. Nevertheless, it is not adding new information about this condition.

Major compulsory revisions
1) The paper is long, particularly the “case presentation” section. This section should be synthesized and schematised, repetitions should be avoided.
2) Last paragraph of case presentation: the number and location of dental agenesis should be specified.
3) The degree of mental retardation should be added.
4) The molecular basis of Mulvihill syndrome is unknown, but autosomal recessive mechanism of inheritance is suspected. A sentence should be added to note that the fact that the proband is the only affect child among 8 is not typical for autosomal recessive conditions.
5) Which kind of metabolic tests were performed?
6) No mention in the text about genetic examinations. Chromosome analysis and CGHarray should be included.

Minor essential revisions
7) The Discussion section should be titled.

Level of interest: An article of limited interest

Quality of written English: Needs some language corrections before being published

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests