Author’s response to reviews

Title: Divergent phenotypes in siblings with identical novel mutations in the HNF-1α gene leading to maturity onset diabetes of the young type 3

Authors:

Birgit Knebel (bknebel@ddz.uni-duesseldorf.de)

Susanne Mack (s.mack@ascklepios.com)

Jutta Haas (info@haas-metall.com)

Mona-Kathrin Herman-Friede (lab@ddz.uni-duesseldorf.de)

Simone Lange (si.lange@asklepios.com)

Oliver Schubert (oschubert@docschubert.de)

Jorg Kotzka (jktzka@ddz.uni-duesseldorf.de)

Dirk Müller-Wieland (d.mueller-wieland@asklepios.com)

Version: 1 Date: 13 Apr 2016

Author’s response to reviews:

BMC Medical Genetics MGTC-D-15-00061

Dear Dr Sands,

We would like to submit the revised manuscript.

Divergent phenotypes in siblings with identical novel mutations in the HNF-1α gene leading to maturity onset diabetes of the young type 3
We took the suggestions of the reviewer as indicated below, but according to the marginal changes to the manuscript we directly provide the plain tex.

Sincerely,

Prof Dr. D. Mueller-Wieland

Corresponding author

Reviewer reports:

Reviewer #1:

I enjoyed reading this revision, as it was easy to follow.

I have a few discretionary comments.

1. Whilst patient therapy (and response to it) is not the focus of the article, it is nevertheless of interest to clinical readers and helps place this Case Report within a clinical context. I felt this information became 'buried' by including it only in Figure 1. It should have been included in the main text.

--We added the information of medication to the footnotes of Table 2 and 3

2. Background. Regarding the statement 'MODY ...(has a)..clinical appearance similar to type 2 diabetes', maybe a better statement would be that there is overlap in the clinical characteristics of both conditions.
3. Determination of diabetes related antibodies. The Table indicates that GAD, ICA and IA2 were measured but the text (page 4 first paragraph) also mentions tyrosine phosphatase and insulin antibodies. Consistency of terminology and reporting would reduce the potential for confusion.

--We clarified this in the text “To exclude the presence of type 1 diabetes or latent autoimmune diabetes of adults, measurement of antibodies against insulin, GAD, tyrosine phosphatase (IA2), and islet cells (ICA) were determined.”

4. Results section - first line on Page 5. There is discussion about diabetic complications during pregnancy. Am assuming these are obstetric diabetic complications rather than traditional non pregnancy related complications? This sentence was confusing and could be reworded.