Author's response to reviews

Title: X linked congenital adrenal hypoplasia associated with hypospadias in an Egyptian infant : a case report

Authors:

Kotb A Metwalley (kotb72@yahoo.com)
Hekma S Farghaly (hekma73@hotmail.com)

Version: 2 Date: 5 October 2012

Author's response to reviews: see over
Dear sir

Greetings

Sorry for late replay

We tried our best to improve the language of the manuscript via professional copyediting office

Regards

Kotb

In addition to the reviewers' and the Associate Editor's comments, please also address the following formatting changes required for your paper:

1. Please format your title to follow the structure: A presenting with B in C: a case report
   
   Title was changed to: X linked congenital adrenal hypoplasia associated with hypospadias in an Egyptian infant: a case report

2. Please include the ethnicity and sex of the patient in the abstract
   
   Ethnicity and sex of the patient were added in the abstract (Egyptian boy)

3. Please include the sex of the patient in the case presentation section of the manuscript.
   
   Sex of the patient in the case presentation section of the manuscript was added (boy).

++++++++++++++++++
Editor's comment
Title: Hypospadias in an Egyptian infant with X linked congenital adrenal hypoplasia: a possible new syndrome.
Authors:
Kotb A Metwalley (kotb72@yahoo.com)
Hekma S Farghaly (hekma73@hotmail.com)
Version: 1 Date: 18 September 2012

Dear sir

Thank you for your comments

Editor's comment:
Your manuscript is interesting, thank you, however you have to respond to remarks of both Reviewers
Done (please see the revised manuscript and my relay to both reviewers)

in all points they have raised both in core of the manuscript as well as revising language status.

Language were revised as the editor indicate
Dear sir

Thank you for your comments

The authors should state that presented case may be also co-existing clinical situation. Because there is not enough evidence to show the genetical relationship between two clinical disturbances.

The following paragraph was added to the manuscript as the reviewer indicate:

We reported an Egyptian infant with an association of congenital adrenal hypoplasia due to DAX1 mutation and hypospadias due to mutation of MAMLD1 gene. Early diagnosis of this association and determining its optimal treatment are very important in helping to avoid its fatal course.

There are many syntax errors in the text and English must be improved.

Syntax errors in the text and English were improved as the reviewer indicate.

The pictures of the patient are irrelevant and reductant.

The pictures were reduced to one picture only.
Reviewer's report
Title: Hypospadias in an Egyptian infant with X linked congenital adrenal hypoplasia: a possible new syndrome.
Version: 1 Date: 31 August 2012
Reviewer: Tsutomu Ogata

Dear sir
Thank you for your comments

Comments to authors:
This manuscript describes an interesting association of DAX1 mutation and CXorf6 mutation.

I have several comments that hopefully help to improve this manuscript.

1. The word [syndrome] must be deleted throughout the manuscript. This boy has an association of DAX1 mutation and CXorf6 mutation, rather than a syndrome.

   Word :syndrome was deleted throughout the manuscript as the reviewer indicate

2. The word [CXorf6] must be replaced with [MAMLD1]. MAMLD1 is the formal name for this gene.
   The word [CXorf6] was replaced with[MAMLD1]. As the reviewer indicate

3. ARG327PRO should be rewritten as [p.Arg327Pro], and 325delG should be rewritten as c.325delG.

   ARG327PRO was changed to [p.Arg327Pro], and 325delG was changed to  c.325delG. as the reviewer indicate

4. Do you have some data showing that p.Arg327Pro is a pathologic mutation rather than a polymorphism.

   Sorry I do not have these data

5. Figure should be reduced in number. Fig. 3 only would be enough.
   All figures were deleted except figure 3 which changed to figure 1 as the the reviewer indicate

6. English and typing are both poor.
English and typing were improved as the reviewer indicate

7. Please describe about ethical approval.

_The following paragraph was added as the reviewer indicate:_

**Ethical approval**  
This report was approved by the Hospital Institutional Review Board and Ethical Commettee

**Quality of written English:** Needs some language corrections before being published  
**Declaration of competing interests:**  
I have nothing to declare.