Author's response to reviews

Title: Peters Anomaly with Post axial Polydactyly, Bilateral Camptodactyly and Club Foot in a Kenyan Neonate: A Case Report.

Authors:

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Version: 3 Date: 11 September 2011

Author's response to reviews: see over
Dear editor,

Thank you for the correspondence and for the reviews to my manuscript.

I have taken positively the critique and suggestions recommended by the reviewers and this re-submission bears corrections to the same.

Please find herewith my specific corrections, explanations and adjustments as itemized in the bullets below:

1. The section on authors’ contribution has been added at the bottom of the document. M.F and D.C are acknowledged and are not co-authors

2. The ethnicity of the patient has been included in the new version of revised manuscript

3. Appropriate wording has been used in the statements in sections on genetic linkage, inheritance and bilaterality. Appropriate citation has been included in these cases (referee 1 recommendation)

4. Review of literature and a tabular presentation of all relevant case reports for the last ten years as searched from pubmed™. These results have helped the author demonstrate easily the nature of reported extra ocular associations of Peters anomaly and bring to the fore the unique array of associations (skeletal) in this case report.

As earlier regretted, the patient presented in this case report died and therefore the priceless idea of a second level investigation cannot be materialized.

Thank you for giving me the honor of having my case peer reviewed in this highly ranked journal, I look forward to hearing from you.
Reviewer's report

Title: Peters Anomaly with Post axial Polydactyly, Bilateral Camptodactyly and Club Foot in a Kenyan Neonate: A Case Report.

Version: 1 Date: 4 August 2011

Reviewer: Sara Alquati

Which of the following following best describes what type of case report this is?: An unexpected association between diseases or symptoms

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?: Yes

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?: No

Will the case report make a difference to clinical practice?: No

Is the anonymity of the patient protected?: Yes

Comments to authors:

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In the present manuscript (MS ID:6815595935624033) Dr Aruyaru has described the interesting case of a newborn who presented at birth with Peters anomaly associated with musculoskeletal abnormalities such as polydactyly,
camptodactyly, and club foot.

The author states that this association has never been reported.

Overall, in my opinion, this case has been described clearly and coherently.

Herewith some specific observations:

1. The author states that this particular clinical presentation could represent a new syndrome. In my opinion, one case could only be fruit of a coincidence. Additional cases and investigations are needed to confirm this suggestion.

The reviewer’s concern has been noted and the author has deleted the statement that indicates this collection of symptoms could point to a new syndrome.

2. No second level clinical investigations or post-mortem examination have been carried out to describe the detail this pathological picture. This could represent a limitation of this reported case.

The author notes and similarly regrets that the patient died before further investigation and a postmortem examination was not possible due to socioethical concerns. This limits any further assessment that the reviewers would recommend at this point in time.

3. In order to highlight the uniqueness of this clinical combination of different anomalies, a careful review of the English-language literature on reported cases of Peters anomaly would significantly improve the value of this manuscript. I suggest to organise and show the result of this review in a tabular form.

This is an invaluable advice that the author has taken up and in the revised manuscript, a tabular presentation of the case reports of peters anomaly and their attendant associations (both gross and genetic) has been provided in the revised manuscript.

What comes out from this inclusion, thanks to the advice, is that the array of skeletal
associations I have reported does not appear elsewhere. As such if this case is reported it could form the first of a series of others in an attempt to determine the possibility of a new syndrome.

Quality of written English: Acceptable

Declaration of competing interests:

I declare that I have no competing interests
Reviewer's report

Title: Peters Anomaly with Post axial Polydactyly, Bilateral Camptodactyly and Club Foot in a Kenyan Neonate: A Case Report.

Version: 1 Date: 1 August 2011

Reviewer: Gerald W Zaidman

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?: Yes

Is this case worth reporting?: No

Is the case report persuasive?: Yes

Does the case report have explanatory value?: Yes

Does the case report have diagnostic value?: No

Will the case report make a difference to clinical practice?: No

Is the anonymity of the patient protected?: Yes

Comments to authors:

This case report presents the case of bilateral Peters anomaly associated with polydactyly, camptodactyly, and clubbed foot. Peters anomaly is the most common cause of congenital corneal opacities. It is usually not associated with any extraocular manifestations. However when Peters anomaly is a part of the
"Peters plus syndrome," non-ocular anomalies are seen.

The first requirement of the authors is to correctly diagnose their patient. From their description and their photographs, it is not clear what type of Peters anomaly the patient had or if the patient truly has Peters anomaly. The only way to make a diagnosis of Peters anomaly in an infant is through an exam under anesthesia. The exam under anesthesia would allow the ophthalmologist to complete the slit-lamp examination and determine the severity of the ocular abnormalities. In this way, the ophthalmologist could determine if the patient has corneal scarring alone, corneal scarring with adhesions from the iris to the cornea, involvement of the lens, etc. It is not clear from the author’s case report if a complete eye exam other than a brief penlight exam was performed.

Under the assumption that a good and comprehensive eye exam was performed, then there are several errors in the text. First, Peters anomaly is rarely inherited. It is the major cause of congenital corneal opacities and is usually sporadic

The reviewer’s observation that Peters anomaly is rarely inherited has been taken into account and the statement that had that implication has been reworded appropriately.

Type 2 Peters anomaly is not usually associated with the PAX6 or any other gene. The word ‘usually’ has been deleted and appropriate citation is provided to refer to literature that shows association between type 2 peters anomaly and PAX6 gene.

Also Type 2 Peters anomaly can occur either as a bilateral or unilateral disorder.
The same is true for Type 1. The authors make the assumption that the bilaterality of the disease helps diagnosing the type of Peters anomaly present. This is incorrect since both Type 1 and Type 2 can be unilateral or bilateral.

The reviewer’s observation has been borne into mind and the necessary change in the wording has been included in the revised manuscript.

Genetic testing is not necessary in cases of Peters anomaly since most of the children have sporadic mutations and do not have genetic syndrome. The reviewer is true and the statement to this effect (necessity of genetic testing) has been deleted in the revised manuscript.

Finally, if the authors are convinced that there is no lens involvement in their patient’s eye, then the absence of lens involvement classifies the case as Peters anomaly Type 1. This is why a thorough exam is needed.

Once the authors correct these mistakes in their manuscript and clarify what type of Peters anomaly is present, then the editors can proceed with publication if they so desire.

Quality of written English: Acceptable

Declaration of competing interests:

'I declare that I have no competing interests'