Author’s response to reviews

Title: Partial trisomy 16q21–qter due to an unbalanced segregation of a maternally inherited balanced translocation 46,XX,t(15;16)(p13;q21): A case report and review of literature

Authors:

Rupesh Mishra (rupeshmm015@gmail.com)
Chamara Paththinige (paththinige@yahoo.com)
Nirmala Sirisena (nirmalasirisena@yahoo.com)
Shakila Nanayakkara (nanayakkara.shakila@gmail.com)
Imalki Kariyawasam (imalki.kariyawasam@gmail.com)
Vajira Dissanayake (vajirahwd@hotmail.com)

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Author’s response to reviews:

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Editor BMC-Pediatrics

Re: BPED-D-16-00591R1 : Responses to reviewers’ comments

Thank you very much for reviewing our manuscript titled, “Partial trisomy 16q21–qter due to an unbalanced segregation of a maternally inherited balanced translocation 46,XX,t(15;16)(p13;q21): A case report and review of literature” by Rupesh et al., submitted for publication in the BMC Pediatrics journal.

We hereby submit our responses to the editorial and reviewers’ comments provided.

Response to reviewers’ comments

Editor Comments: None

Reviewer 1:

1. Line 60 there is a space between partial trisomy and 16
Space added

2. Line 123 conclusion should be replaced by Discussion
Revised; deleted “Conclusions”, added “Discussion”

3. Line 127 references should be added
References added

4. Line 130 reference should be added
References added (line 130 and 131)

5. Line 136 reference should be added
References added (line 137)

6. Line 162 reference should be added
References added (line 163)

- Reference list (Page 9 and page 10, line 208-266) was revised according the changes done in 3-6
- Reference numbers in the Table 1 were changed according to the above changes

7. Fig 2 is hazy and not clear
The figure and the legends were modified according to the suggestions made by Reviewer 2

Reviewer 2:

1. Line 67: small and downslanting palpebral fissures?
Revised as “small and downslanting palpebral fissures”

2. Line 76: replace Hypertelorism by hypertelorism.
Corrected as “hypertelorism”

3. Line 77: delete pathognomonic
   Deleted

4. Line 89: Put the phrase "The proband was referred for genetic evaluation due to multiple congenital abnormalities at the age of 24 days." at the beginning of the session "Case presentation"
   Revised as suggested

5. Line 90: delete "she was the product of the third pregnancy." This is already described in the opening paragraph.
   Deleted

6. Line 94: Modify to "small and downslanting palpebral fissures"
   Revised as suggested

7. Line 101: Delete "by peripheral venepuncture"
   Deleted

8. Line 128/129 "small and downslanting palpebral fissures"
   Revised as suggested

9. Line 175: replace advanced by additional
   Revised as suggested

Suggestion: Create a figure with the normal chromosome and ideogram to the side, demonstrating the hybridization points of the FISH probes, and do the same with the chromosome altered.

Figure 2 was modified according to these suggestions. Figure legend (page 12, line 281-287) was revised accordingly.
Thank you.

Yours Sincerely,

Dr. Rupesh Mishra

Human Genetics Unit
Faculty of Medicine
University of Colombo
Sri Lanka