Author's response to reviews

Title: Mechanisms of ring chromosome formation, ring instability and clinical consequences.

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Version: 5 Date: 13 December 2011

Author's response to reviews: see over
Subject: Revised Manuscript: Mechanisms of ring chromosome formation, ring instability and clinical consequences

December 12, 2011

Dear Sirs

We were glad to know our MS “Mechanisms of ring chromosome formation, ring instability and clinical consequences” has been peer reviewed and considered for publication.

Enclosed please find the revised version of the manuscript taking into consideration the comments received.

Based on the reviewer’s comments, we changed the following points:

- We included “and FISH” in the sentence "The SNP array results obtained for patient VI who has an r(13) revealed a 43.5 Mb inverted duplication at 13q21.33q34 followed by a 364 kb terminal deletion at 13q34 (Figure 3)” changing it to “The SNP array and FISH results obtained for patient VI who has an r(13) revealed a 43.5 Mb inverted duplication at 13q21.33q34 followed by a 364 kb terminal deletion at 13q34 (Figure 3)”, considering that FISH was performed to conclude the nature of the duplication, information that was missing as noticed by the referee 1.

- The reference was redone in a uniform style, because the previously one was done using an End Note without style edition.

- Regarding cases VII and XII who do not present any deletion, we have discussed that the clinical consequences may derived from configuration of the ring chromosome, thus affecting gene expression. Considering that this is one among other possibilities, as mentioned by reviewer 2, the sentence “Another factor that can influence the phenotype is the configuration of the ring chromosome that can change the gene expression and cause clinical abnormalities…” was changed to “Another factor that could influence the phenotype is the configuration of the ring chromosome that could change the gene expression and cause clinical abnormalities …” to become clearer.

We hope that our paper now meets all the requirements for publication.

Thank you for all your attention.

Yours sincerely,

Roberta S. Guilherme