Reviewer's report

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

Version: 4 Date: 28 November 2011

Reviewer: Karoly Szuhai

Reviewer's report:

In the revised manuscript "Mechanisms of ring chromosome formation, ring instability and clinical consequences." by Guilherme et al. the authors addressed previous comments.

Two minor remarks remained:

On page 7 of the submitted manuscript in the second paragraph of the Results section authors wrote:

"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)." As SNP does not provide information about the orientation of the alterations, as authors did show in fact in figure 3, FISH test should be performed to conclude the nature of the duplication (direct or inverted). This sentence should be corrected accordingly.

Lastly, the reference list does not have a uniform style, see the journal name types of for example ref 30 and 31, this inconsistent for many others. The names in reference 28 should be re-checked

Level of interest: An article of outstanding merit and interest in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests.