Reviewer's report

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

Version: 1 Date: 5 October 2011

Reviewer: Carmen Ramos

Reviewer's report:

To the author

The series of cases presented in your article is of great interest since it is a nice series of 14 ring chromosomes.

However there are major revisions that should be done in order to reconsider the article for publication.

The information presented in the article do not fulfils the expectancies you have when reading the title.

The mechanisms of formation are insufficiently explained and besides parental bloods in those complex rearrangements should have been studied for a proper understanding of the mechanisms by which they arose.

The cytogenetic study of the instability is not in the table and the possible correlation between the instability and phenotypic manifestation is not very much considered. Besides there is no clear explanation about what the new technologies may offer in the understanding of the phenotypic manifestations in the patients.

A deep revision for a proper phenotype-genotype correlation for the different cases, together with the the discussion of what new technologies offer for the understanding of such cases is needed.

Level of interest: An article of limited interest

Quality of written English: Needs some language corrections before being published

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'