Author's response to reviews

Title: Patient with ring chromosome15 20-years cytogenetic and molecular follow-up: a case report.

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

Thank you for considering our MS “Patient with r(15) 20-years cytogenetic and molecular follow-up: a case report” for publication:

Enclosed please find the Revised Version of the manuscript taking into consideration the comments and suggestions received.

We changed the following points:

- We removed the sub-sections/sub-headings from the case presentation section.
- We removed written consent uploaded as supplementary file.
- We revised the consent statement to confirm that informed written consent was received for publication of the manuscript and figures.
- We clarified that the deletion on 15q was the only clinically relevant genetic imbalance since the breakpoint in 15p was distal to the nucleolus organizer region. Furthermore, deletion of acrocentric short arms would not result in phenotypic effect.
- We changed the last sentence ending in the conclusion “and silencing of genes present in the ring chromosome” for “and an altered expression of genes present in the ring chromosome”, as suggested.
- In Abstract, we substituted “Karyotyping revealed one normal and one monocentric ring chromosome 15” for “Karyotyping revealed that the majority of the cells harbored one normal chromosome and one ring chromosome”, as suggested.
- We clarified the methods used at the two different patient ages.
- We clarified that the patient’s parents were cytogenetically studied and presented normal karyotypes.
- We discussed about the influence of the ring instability in the phenotype.
- The patient’s data were rewritten to make it shorter and more concise.
- The language was checked and some errors were corrected.

Hoping that now our paper meets all the requirements for publication and thanking you for all your attention.

Sincerely yours,

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