Reviewer's report

Title: Array based characterization of the terminal deletion of chromosome 15q26.2: An emerging syndrome associated with growth retardation, cardiac defects and developmental delay

Version: 2 Date: 14 October 2007

Reviewer: Jacqueline Schoumans

Reviewer's report:

General

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

In this case report by Davidson et al a patient with cardiac defect, growth and developmental delay is presented due to a rare unbalanced rearrangement consisting of a dicentric chromosome 15, resulting in a 6.48 Mb terminal deletion. This case is interesting because very few such cases have been described so far nor have the 15q deletions been characterized in details. Only one case with a dicentric 15 has previous been reported which arose from a maternal paracentric inversion. Subtelomeric deletions of chromosome 15q are also rare as only one single case was found among 11 688 patients that were screened for subtelomeric aberrations by Ravnan et al.

Minor essential revisions:

According to the authors subtelomeric aberrations are found in approximately 5% of patient with mental retardation. This high amount is based on early studies and is dependent on patient selection. Subtelomeric screening has been used in diagnostic laboratories for almost a decade and the extensive experience as well as the large study reported by Ravnan et al showed that the yield is approximately 2.5%. The authors should update this information in the abstract and background and add the above reference.

The authors describe the abnormal chromosome as a “marker” chromosome throughout the text. This should be changed in derivative chromosome according to the ISCN nomenclature.

Parental samples displayed normal karyotypes and microsatellite analysis was not performed to determine the parental origin of the dicentric chromosome. It would be interesting to add whether the authors investigated the parental chromosomes 15 for inversions and explain why they did not investigate whether the maternal or the paternal chromosome 15 was abnormal in the child.
What next?: Accept after minor essential revisions

Level of interest: An article whose findings are important to those with closely related research interests

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