Reviewer’s report

Title: Familial inheritance of the 3q29 microdeletion syndrome: case report and review

Version: 0 Date: 24 Oct 2018

Reviewer: David A. Koolen

Reviewer’s report:

Dear editor,

Khan et al., describe a case with the 3q29 microdeletion syndrome and familial inheritance.

The manuscript is concise and well written.

The message of the manuscript is not novel, although it might be informative to stress the importance of a molecular diagnosis for families that harbor copy number aberrations with variable expressivity. Reports of the 3q29 deletions inherited from an unaffected parent have been published before.

In the Introduction, the authors mention the "recent discovery of several ... syndromes". The references, though, are not recent.

"The majority of 3q29 microdeletion syndrome cases occur de novo [8-12]; however, inheritance from a mildly affected parent has also been reported in some families (Table 1) [13]". How many cases? Please also refer to the other papers describing familial cases.

Apparently, the 3q29 deletion is an important risk factor for schizophrenia (~40 fold). Do these schizophrenia patients have (other) features of the 3q29 microdeletion syndrome? This stresses that the penetrance is not 100%.

The authors mention that the mother carrying the deletion is healthy, but she does have a history of learning disabilities. What kind of disabilities. Does she still have a subtle cognitive impairment or signs of neuropsychiatric disorders?


The authors conclude that anemia is an uncommon feature of the syndrome. It might just be a coincidence. It seems too early to tell whether it is a rare feature or not.
Also the statement that "the familial inheritance of this pathogenic deletion is unique" is striking, because in the same sentence other examples are mentioned.

The resolution of the Figures is too low.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

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Please indicate the quality of language in the manuscript:

Acceptable

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