Reviewer's report

Title: Chromosome 3q29 deletion, with GI malformation; a case report

Version: 2 Date: 7 July 2010

Reviewer: Annelies de Klein

Which of the following best describes what type of case report this is?: Unexpected or unusual presentations of a disease

Has the case been reported coherently?: No

Is the case report authentic?: No

Is the case report ethical?: Yes

Is there any missing information that you think must be added before publication?: Yes

Is this case worth reporting?: No

Is the case report persuasive?: No

Does the case report have explanatory value?: No

Does the case report have diagnostic value?: No

Will the case report make a difference to clinical practice?: No

Is the anonymity of the patient protected?: Yes

Comments to authors:

The finding of GI malformations in a 3q29 deletion case is indeed special and needs further investigation. However as presented, here it is not suitable for publication.

- Quality of provided karyogram is to low to allow a proper analysis.
- Additional investigations using high resolution techniques as SNP or oligo arrays should be performed to confirm pure deletion of 3q29 and to exclude for example a chromosome translocation, and thus a duplication of an other chromosome adding to the phenotype.
Quality of written English: Acceptable

Declaration of competing interests:
I declare that I have no competing interests