Reviewer's report

Title: Analysis of patients with developmental disorder and normal array CGH results - the added value of high-resolution SNP array

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Reviewer: Nicola Brunetti-Pierri

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Linda et al. report the results of SNP array 6.0 in cases previously analyzed by array CGH and they conclude that ultra high resolution array does not improve the diagnostic yield over CGH array.

The authors do not provide any information on the ‘several new CNVs’ that they conclude were ‘insignificant in correlation to the patient’s phenotype’. The data on the CNV as well as the patients’ phenotypes should be provided to allow the reader to agree on their conclusions.

The title is a bit confusing as it seems to imply that the study by Linda et al. provide evidence that the SNP array does indeed increase the diagnostic yield.

The manuscript conclusion is that SNP array 6.0 does not improve the diagnostic yield over CGH array. This conclusion is in contrast with two previous studies both reporting the findings in larger numbers of patients. The sample size in the studies of Bernardini and Mannik is about twice the size of the study of Linda et al. and this may be another reason for explaining difference in diagnostic yields. Therefore, it is uncertain whether the findings of Linda et al. can be generalized in other contexts.

Minor issues

1. In the abstract the background section describes the study and should therefore be moved into the methods and results section.
2. In table 1, it should be clarified what the numbers in the columns named ‘patient’, ‘mother’ and ‘father’ correspond to.
3. In table 2, please change the commas for the frequency number with dots.
4. Bernardini et al. performed their first line evaluation with a 44K array and not 35K as stated in the manuscript.

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