Reviewer’s report

Title: Copy Number Variants (CNVs) Analysis in a Deeply Phenotyped Cohort of Individuals with Intellectual Disability (ID)

Version: 2 Date: 6 May 2014

Reviewer: Michael Gill

Reviewer’s report:

This is a detailed descriptive study of a small sample of ID individuals. Given the number of statistical tests applied it was unlikely in the first place to have produced significant findings. It will be important to be able to combine this data with similar data from other cohorts and the availability of the data to other researchers should be made explicit in the manuscript. For the purposes of collaborations or meta-analyses, the data without the modifications (High and low frequency phenotypes and the addition of additional fine phenotypes) should be available.

The CNVs identified are defined as de-novo. How is this determination made unless parent DNA was also available and studied. There is no mention of this in the methods.

The paper also makes reference to unique events and mentions controls - these are also missing from the methodology.

The Silfhout paper analyses specific phenotypic features - could these features be analysed in the present cohort, reducing the number of tests and providing comparison/replication?

The conclusions should address the lack of any findings by CNV grouping

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: No competing interests