Reviewer's report

Title: SRY mutation analysis by next generation (deep) sequencing in a cohort of chromosomal Disorders of Sex Development (DSD) patients with a mosaic karyotype.

Version: 2 Date: 9 July 2012

Reviewer: Ken McElreavey

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This is a straightforward study of 14 patients presenting with sex chromosome rearrangements for mutations in the SRY gene using a deep sequencing approach.

The authors did not find SRY mutations in any of the 14 cases and the authors conclude that mutations involving SRY in these cases of gonaosome mosaicism.

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