Reviewer’s report

Title: BAC array CGH in patients with VCFS-like features reveals genomic aberrations on chromosome region 1q21.1

Version: 8 Date: 11 August 2009

Reviewer: Stefania Gimelli

Reviewer’s report:

Concerning article 1544534040248275:
BAC array CGH in patients with VCFS-like features reveals genomic aberrations on chromosome region 1q21.1

Comments from Dr. Stefania Gimelli (stefania.gimelli@gmail.com):

The manuscript reports on a CNV analysis of 18 VCFS-like patients, not presenting the typical 22q11 microdeletion, using a whole genome and a chromosome 22-specific BAC array. The author discuss on the possible association of 1q21.1 duplication with heart defects which has been never described before and, in addition, refers on the potential role of some CNVs in the VCFS-like phenotype. The author has done some important modifications giving a new statement to the paper that appears now better organized and discussed with a less confusing message. Moreover the new discussion, more focalised on the possible correlation between the the 1q21.1 duplication and CHD, makes this study worthy of publication.

1) Major Compulsory Revisions:
Considering the high phenotypic variability of patients with VCFS and the apparent rarity of 1q21.1 duplications, I don’t agree with the author’s conclusion suggesting to consider this region in the MLPA screening of patients showing a VCFS-like phenotype as well as the ones with psychiatric diseases. I would better suggest a whole genome array-CGH should be proposed to such type of patients at least until more cases will be described, giving a better delineation on a possible correlation between the the 1q21.1 duplication and the VCFS phenotype.

2) Minor Essential Revisions
I would shorten the FISH materials and methods part, writing something like: “Fluorescent in situ hybridization (FISH) analysis was performed on metaphase chromosome spreads following standard procedures”

Page 10 last line: I suggest to write the name of the patient (V5) instead of “this patient”
Page 11 line 19 “screened”
Page 12 line 10 loci
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' below. If your reply is yes to any, please give details below.