Reviewer's report

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

Version: 8 Date: 28 July 2009

Reviewer: Cédric Le Caignec

Reviewer's report:

Dear Editor,

Overall, Brunet et al. answered most of my criticisms. However, in general, I share the reservations with the two other reviewers concerning the deleterious effect of the 212 kb 1q21.1 microduplication. Patient V-5, in addition to the de novo 212 kb 1q21.1 microduplication presented with a de novo 1.15 Mb 1q21.1 microdeletion. Such microdeletion is known to be responsible for congenital heart defects (CHD). Therefore, it’s rather speculative to write that this is the 212 kb 1q21.1 microduplication that likely contribute to the CHD. The second patient (case 112) carries a 212 kb 1q21.1 microduplication inherited from his phenotypically normal father. Incomplete penetrance is possible but, once again, the role of the duplication in CHD is unproven. Nevertheless, this 212 kb 1q21.1 microduplication is extremely rare in the general population, so, his presence in these two patients is surprising. Additional studies will contribute to delineate more precisely the role of this microduplication in CHD.

Discretionary revisions

In the Title: VCFS-like may be written as velocardiofacial syndrome-like

In the Abstract: Results, “… the same 212 kb microduplication”. “reciprocal to the TAR microdeletion syndrome” may be added.

There are a number of typing errors, such as “sumbicroscopic” in the Results section, “for these reason”, “a rare the novo 1q21.1 microduplication”…

Level of interest: An article of limited interest

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.