Reviewer’s report

Title: Breakpoint Associated with a novel 2.3 Mb deletion in the VCFS region of 22q11 and the role of Alu (SINE) in recurring microdeletions.

Version: 1 Date: 15 October 2005

Reviewer: Tadao Arinami

Reviewer’s report:

General
The authors screened 22q11 deletion in 379 patients from Southwestern Ontario with suspected schizophrenia by FISH and found 11% of them with microdeletion in 22q11. The authors determined the deletion break points in one patient and found a novel 2.3 Mb deletion. Molecular characterization of the sequences at the breakpoints revealed that the Alu like shared sequence of the breakpoint regions may be a preferred sequence in the breakpoint regions. This paper contains two major findings of population and mechanism of the deletion. In addition, they presented the screening methods of hemizygous regions.

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)
1. The purpose of this study is unclear.
2. The characterization of the patients screened need to be described in more details.
3. Page 2, line 3, what is unusual?
4. Were deleted regions were determined in 41 patients with 22q11 deletion?
5. Figure legends for supplementary figure 1 and figure 3 are insufficient.

Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No

Declaration of competing interests:
I declare that I have no competing interests.