Reviewer's report

Title: Seizures as the first manifestation of chromosome 22q11.2 deletion syndrome in a 40-year-old man.

Version: 2 Date: 7 May 2007

Reviewer: syed M jalal

I am familiar with the literature and believe that this case meets one of the 7 criteria for evaluation in the journal: An unexpected association between diseases or symptoms

Has the case been reported coherently?: Yes
Is the case report authentic?: Yes
Is this case worth reporting?: Yes
Is the case report persuasive?: No
Does the case report have explanatory value?: Yes
Does the case report have diagnostic value?: No
Will the case report make a difference to clinical practice?: No

Comments to authors:

Revisions necessary for publication

Major concern:
1. To prove the point that seizures are the first manifestations of the syndrome, especially in adults, (1) It should be clarified when seizures were first observed in this patient. In the case presentation the point is unclear. (2) A careful literature search needs to be done to clarify if seizures are indeed one of the first indications of the disease. There are some 10 publications on the topic that are not cited in this paper. A few are:
   Would it not be more appropriate to suggest both seizures and hypocalcemia as the major features in adults?
2. Appropriate changes in the abstract and discussion should be made in response to item 1.

Minor concerns:
1. Change 22q11.2 to 22q11.2 in the title
2. In figure 1, normal and abnormal brain tomography should be so labeled.
3. Del22q11.2 is not an acceptable cytogenetic expression. Change it to del(22)(q11.2) or microdeletion of 22q11.2. Technically, the deletion is del(22)(q11.2q11.2).

What next?: Accept after minor revisions

Quality of written English: Acceptable