Reviewer's report

Title: 47,XXY/48,XXXY/49,XXXXY mosaic with hydrocephaly: case report and literature review.

Version: 2 Date: 1 August 2007

Reviewer: Guillermo Romero

I am familiar with the literature and believe that this case meets one of the 7 criteria for evaluation in the journal: New associations or variations in disease processes

Has the case been reported coherently?: Yes

Is the case report authentic?: Yes

Is this case worth reporting?: Yes

Is the case report persuasive?: Yes

Does the case report have explanatory value?: No

Does the case report have diagnostic value?: Yes

Will the case report make a difference to clinical practice?: Yes

Comments to authors:

General
Authors presents a variant of the Klinefelter's syndrome name by some geneticists as syndrome of Fracaro, which is a rare but interesting clinical condition. Moreover, as stated in the MS, it seems to be the fourth case reported in literature of the 47,XXY/48,XXXY/49,XXXXY mosaic.

The paper and describes the new clinical findings of hydrocephaly and cardiopathy adequately. This information could encourage geneticists to discard hydrocephaly complication in new Klinefelter patients.

Revisions necessary for publication
The MS is well written, but in the ABSTRACT, “X and Y polisomy and mosaicism” should be corrected as “X and Y polysomies and mosaicism”. In the BACKGROUND section the word “X and Y polisomy” should be change by “X and Y polysomies”.
What next?: Accept

Quality of written English: Acceptable