Reviewer's report

Title: Discontinuous microduplications at chromosome 10q24.3 identified in a Chinese family with split hand and foot malformation

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Reviewer: Giovanni Neri

Reviewer's report:

The authors report on an interesting genomic rearrangement in a family segregating SHSF malformation. They state that all family members were carefully evaluated clinically. Did that evaluation include X rays? However, molecular analysis seems to have been conducted only on three (unspecified) unaffected individuals. If that is the case, I recommend that the molecular analysis be extended to all family members, given that a defect of penetrance is well known in SHSF. In the clinical report, the authors state that there are six affected individuals in the family, but in the pedigree I see only five. On p. 7 the authors refer to the posterior part of the FBXW4 gene. What does posterior mean? The MS is generally well written, but the English can benefit of some improvement. No evaluation by a statistician is required.