Reviewer's report

Title: Identification of a rare 17p13.3 duplication including the BHLHA9, YWHAE genes in a family with developmental delay and behavioural problems

Version: 3 Date: 22 August 2012

Reviewer: Damien L Bruno

Reviewer's report:

The authors have addressed many of the earlier comments, but the manuscript still contains a number of grammatical errors some of which are highlighted below.

Minor Essential Revisions

Abstract

1. Replace ‘We have studied a family with a…’ with ‘We have studied a family segregating a…’
2. Replace ‘…was transmitted by…’ with ‘…was inherited from…’
3. The final sentence of the abstract should be rewritten as the assertion that a new microduplication syndrome will be delineated through characterisation of further microduplication cases is highly questionable. I suggest rewording along the lines of ‘...this and other cases will help characterise better the clinical spectrum of phenotypes associated with 17p13.3 microduplications.'

Background

1. Replace ‘...genes are the pathogenic causes of...’ with ‘...genes cause two...’
2. Replace ‘...of 17p13.3 region, overlapping the MDS critical region...’ with ‘...overlapping the MDS critical region...’
3. The last two sentences are poorly written. Suggest you replace with ‘...may also include the genomic region encompassing the CRK and YWHAE genes, and are associated with developmental delay, psychomotor delay and associated hypotonia. Here, we report a 7-year old boy and his mother with a novel class I 17p13.3 microduplication measuring about 329.5 Kb in length and involving only seven genes, including YWHAE, CRK and BHLHA9.'

Case Presentation
1. ‘The patient and his mother had a normal karyotype.’ Insert ‘G-band’ after 'normal'.

Discussion
1. The first sentence is poorly written and should be rewritten.
2. The authors refer to several of the 17p13.3 genomic disorders as syndromes. The evidence to date would appear to conflict with this interpretation.

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

**Quality of written English:** Needs some language corrections before being published

**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