Reviewer's report

Title: A 120 kb microdeletion of 16p13.3 in a young girl with Rubinstein-Taybi syndrome detected by oligonucleotide-based array CGH.: a case report

Version: 1 Date: 18 September 2011

Reviewer: Beata Nowakowska

Which of the following best describes what type of case report this is?: Findings that shed new light on the possible pathogenesis of a disease or an adverse effect

Has the case been reported coherently?: Yes

Is the case report authentic?: Yes

Is the case report ethical?: Yes

Is there any missing information that you think must be added before publication?: 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?: No

Is the anonymity of the patient protected?: Yes

Comments to authors:

The manuscript by Mohd Fadly Md Ahid et al. describes 3-year-old patient with de novo 120 kb deletion in 16p13.3, identified by oligo arrayCGH. Detailed clinical description of patient likely has a considerable impact in further elucidation of variability in Rubinstein-Taybi syndrome. However, the following points should be addressed by the Authors before publication.

Major comments:

1. Authors did not mention or tried to explain inconsistence between two diagnostics methods they used. ArrayCGH revealed only partial deletion of
CREBBP gene, whereas MLPA showed complex aberration with hemizygous deletion of exons 1 - 2 and exons 6 - 31 in this gene. Especially, that Authors concluded the article with: “we also demonstrate the reliability of aCGH technique as a powerful tool for screening and detecting clinically significant genomic imbalances in the human genome.”

2. In discussion Authors described only function of the second deleted gene: TRAP1. Despite the fact that this gene has not been associated with any human disease yet, is it possible, that this gene is responsible for any features observed in the described patient?

3. The discussion lacks also novel observations.

Minor comment:

1. Since even smaller aberrations in this region were described previously, Authors should refer their work to the article published in EJHG (2010), “Exon deletions of the EP300 and CREBBP genes in two children with Rubinstein-Taybi syndrome detected by aCGH.”

Quality of written English: Acceptable

Declaration of competing interests:

I declare that I have no competing interests