Reviewer’s report

Title: The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome

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Reviewer: Wasim Ahmad

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Subject: The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome

The subject manuscript describes the use of WES to search for mutations in the human genes for the diagnosis purpose. Further, the authors have identified a novel frameshift mutation the WDR62 gene causing primary microcephaly.

The paper is well-written. Clinical features of the four patients have been described in detail. WES led to the identification of a disease causing mutation. This certainly endorsed the objectives presented in the manuscript that by using this method mutation can be identified in a shortest possible time.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

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