This manuscript reports linkage and subsequent mutation analysis of the WDR62 gene in four Pakistani families with primary microcephaly. One novel and three previously identified mutations were detected. The authors conclude that WDR62 are a frequent cause of microcephaly in Pakistan. This is an interesting paper, since WDR62 has been only recently associated to microcephaly, and limited numbers of mutated cases are published to date. Therefore, the report of novel cases can greatly contribute to define the phenotypic spectrum and genotype-phenotype correlates in patients harboring WDR62 mutations.

Minor essential revisions:
1) The main limitation of the paper is the scarceness of neuroimaging documentation regarding the patients. From the pedigrees depicted in figure 1, it appears that there are ten living affected subjects, but only the CT scan from one affected subject is presented. Neuroimaging was not available for patients of family MCP26, but what about the others? The authors state that “the affected individuals of families MCP3 and MCP35, carrying known missense mutations, showed milder symptoms and simplified gyral patterns (data not shown)”, while it would be very useful to show these additional data and give them emphasis.
2) The authors conclude that WDR62 gene is a major contributor for autosomal recessive primary microcephaly in Parkistani population, but do not specify whether additional microcephaly families have been excluded for linkage to the WDR62 region, and therefore not considered for this paper. Also, it would be important to know the ascertainment criteria (how were the families referred to the authors?). These data would give us a more accurate estimate of the prevalence of WDR62 mutations, at least according to the recruitment method adopted.

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