Reviewer's report

Title: Unilateral hypoplastic kidney - a novel highly penetrant feature of familial juvenile hyperuricaemic nephropathy

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Reviewer: Luca Rampoldi

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The authors have properly addressed most of the points that were raised. They added significant work, as exome sequencing data, that exclude the potential involvement of known FJHN genes / loci in the reported pedigree.

There is one major remaining concern. The key message of the manuscript is that unilateral renal hypoplasia could be a new feature associated with FJHN. Although this is of potential great interest, the authors do not seem to provide sufficient data to support their conclusions.

Unfortunately, the authors could not provide uric acid values for the other affected members of the family.

We are given uric acid serum level for the index case only, and we know that indiv. 5 was reported to suffer from gout, and indiv. 7,8 have raised uric acid levels (on which bases?). Despite this limited information, Table 1 reports that all affected individuals (# 5,6,7,8,11) have “raised uric acid level”. This seems rather misleading and should be corrected.

Overall, one wonders if the available data are enough to support the diagnosis of FJHN. The authors should better explain the criteria that in their opinion support the FJHN diagnosis.

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

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.