Reviewer's report

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

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Reviewer: Luca Rampoldi

Reviewer's report:

This is an interesting report where Plumb and colleagues describe a FJHN family with a novel phenotype, showing association of renal failure and hyperuricemia with unilateral renal hypoplasia (in 3/5 affected individuals).

Major revisions:

1. In order to consider this pedigree as potentially expanding current knowledge of the genetic heterogeneity of FJHN, all known FJHN genes/loci should be considered and excluded.

Mutations in the renin gene have also been implicated in AD pedigrees with FJHN phenotype (Zivna et al, Am J Hum Genet. 2009). Have the authors excluded this possibility?

Also, although the genetic analysis might not be informative, the authors should acknowledge the possibility that the described family is linked to the locus FJHN 3 (Piret et al, Hum Genet. 2011).

Minor revisions:

2. The conclusions in the abstract session could be misleading. It is not clear on which bases the "reported pedigree reveals the possibility" of a link between renal hypoplasia and uromodulin when UMOD mutations have in fact been excluded in the pedigree.

3. The description of clinical features in Table 1 needs improvement. For instance, what are the uric acid values in the tested individuals? What are the creatinine values that are reported in the table?

4. In the Discussion section, TCF2 and UMOD have been linked to FJHN through mutation screening, not just familial linkage analysis.

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.