Reviewer’s report

Title: Novel mutations of PKD genes in Czech population with autosomal dominant polycystic kidney disease

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Reviewer: Fiona Karet

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Here a cohort of Czech patients with apparent PKD have been mutationally assessed and in ~70%, probable pathogenic PKD1 or PKD2 alterations were found. No clinical information is provided.

Major Compulsory Revisions
1. No clinical details are provided (eg if individual patients have or have not reached End-Stage), which would be useful.
2. That the rate of mutation identification of ~70% could have been due either to missed changes or the original diagnosis might have been incorrect is not discussed. This is particularly relevant to the 20 where ‘unequivocal linkage to PKD1’ had previously been found, in whom 15 mutations were identified.
3. Saying that missense alterations segregate with disease in families is only useful if we are told how many affected and no-affected family members were assessed in each case.
4. Unless the distribution of age at ESRD was normal, non-parametric statistical comparisons should be used.
5. The results and discussion are conflated (eg listing some mutations in the discussion). The paper is also very long.
6. The English needs improving (eg "Important role may play also hypomorphic variants or oligogenic inheritance [14, 16]. Arguable is the proportional representation of hypothetical PKD3 mutations in our set of patients" and numerous other grammatical errors).

Level of interest: An article of insufficient interest to warrant publication in a scientific/medical journal

Quality of written English: Not suitable for publication unless extensively edited
**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.