Reviewer's report

Title: Autosomal dominant polycystic kidney disease in a family with mosaicism and hypomorphic allele

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Reviewer: stephane burtey

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The manuscript entitled “Autosomal dominant polycystic kidney disease in a family with mosaicism and hypomorphic allele” described an interesting case report illustrating the usefulness of molecular diagnosis in autosomal dominant polycystic kidney disease.

This article is well written, the case description is clear, and the choice of the molecular biology tools is wise. It described two main causes of variability in the phenotype expression of Autosomal dominant polycystic kidney disease, mosaicism and mutation in the other allele of PKD1. The association is certainly infrequent but this case is interesting for the clinician in charge of ADPKD patients.

Major Compulsory Revisions
There is no major revision.

Minor Essential Revisions
Two minor modifications in the text must be done:

In the background the authors write: “Mutation either in the PKD1 (approximately 85% of patients) or PKD2 gene (approximately 15%) cause ADPKD, with an average age of 54.3 and 74 years, respectively, at the onset of ESRD (end stage renal disease) of 54.3 and 74 years, respectively [1]” there is one extra “with an average age of 54.3 and 74 years, respectively”.

In the case presentation, the authors write “and AT1 receptor blocker because of mild hypertension”. AT1 is for angiotensin II type1 not angiotensin 1 like it is stated in the abbreviation summary.

Level of interest: An article of outstanding merit and interest 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