Reviewer's report

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

Version: 1 Date: 25 July 2013

Reviewer: Karl Lhotta

Reviewer's report:

The authors describe a family with they label FJHN, in which 3 out of 5 affected members have a hypoplastic kidney. I agree that this is an interesting family and that identification of the causative gene defect would be an advance in understanding these diseases.

Major revisions

The authors may point out that FJHN is only a description of a clinical presentation and as the field advances these diseases are classified according to the underlying gene defect. A short description of the FJHN classification as provided in OMIM in Type 1, 2 and 3 is warranted. In addition it should be pointed out that FJHN 1 is now included in uromodulin-associated kidney disease and cases caused by HNF1 beta mutations as renal cysts and diabetes syndrome.

A low FEUA is a classical feature of these diseases. If possible provide the results of FEUA in the index case and other family members. In the table please provide eGFR in addition to serum creatinine (units for creatinine are missing) and serum uric acid levels.

Minor revision

Did the aunt also have a right small kidney? This would be another notable finding that the right kidney was affected in all 3 cases.

As mutations in UMOD and TCF2 could be excluded, the discussion on HNF1 beta and uromodulin can be shortened.

Discretionary revisions

FJHN type 3 is linked to a locus on chromosome 2p.22.1. Was a linkage analysis done? If not, please mention that in the text.

Ultrasound images of the index case's kidneys would be informative

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 interest