Author’s response to reviews

Title: Phenotype variability in a large Spanish Familiy with Alport syndrome associated with novel mutations in COL4A3 gene

Authors:

Cristina Cervera (ccervera@riojasalud.es)
Ana Coloma (acoloma@riojasalud.es)
Emma Huarte (mehuarte@riojasalud.es)
Milagros Sierra (mmsierrac@riojasalud.es)
Elena Domínguez (edominguez@riojasalud.es)

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Jens Michael Hertz (Reviewer 2):

No information regarding hearing loss and renal symptoms are provided about the family to the mothers of V.1 and V.2. Even though the two mothers (IV.2 and IV.4) are without hematuria, this might not be the case for their relatives? Have they been analyzed for hematuria only once?:

Both mothers were donors, they had a complete screening before transplantation

No mutations were detected in COL4A4, and the pedigree isn't in accordance with X-linked inheritance, but has COL4A5 been sequenced in any of the family members? yes, sister of V.1

Line 275: A paper is referred (ref. 29) that suggest MYO1E or other non-COL4 podocyte genes should be screened if the clinical phenotype is more severe than expected. Have MYO1E or other relevant genes been screened? Yes, no mutation detected

The rest comments are corrected in the review paper