**Supplementary table 1.** Summary of the measurement characteristics when determining TKV using MRI

1A 3T Siemens verio magnet (Munich, Germany) was used in all patients using a cardio specific array with 32 channels covering from liver to iliac crests. Patients were all introduced head first in a supine position. T1 in/out phase axial sequences, T2 in axial, coronal and sagittal planes, and axial T1 and T2 fat sat sequences were performed in all cases.

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
|  | **Sequence** | **Slices** | **Thickness** | **TR** | **TE** | **Bandwidth** |
| **T1 in/out** | 3D Vibe caypidixon | 144 | 2mm | 4.21 | 1.35/2.58 | 210 |
| **T2 axial** | HASTE | 65 | 1.1mm | 1240 | 88 | 1184 |
| **T2 coronal** | HASTE | 25 | 4mm | 1240 | 88 | 1184 |
| **T2 sagittal** | HASTE | 25 | 4mm | 1240 | 88 | 1184 |
| **T1 fat sat** | 3D vibe caypidixon | 144 | 2mm | 4.21 | 1.35/2.58 | 210 |
| **T2 fat sat** |  | 35 | 4mm | 2500 | 80 | 254 |

**TE, echo time; TR, repetition time**

**Supplementary information on genetic testing**

Genetic testing was performed by targeted next-generation sequencing of *PKD1* and *PKD2*, as previously described (24). Briefly, capture enrichment of *PKD1* and *PKD2* genomic regions was performed using custom NimbleGen SeqCap EZ Developer Libray (Roche NimbleGen; Madison, WI, USA) and sequenced on a HiSeq instrument (Illumina). Data analysis was performed using open source in-house pipeline. Analysis of copy number variants (CNVs) was performed using the CONTRA tool (28). Single nucleotide pathogenic variants were validated by Sanger sequencing and CNVs by multiplex ligation-dependent probe amplification (MLPA) analysis. Segregation analysis was assessed for all available family members. *PKD1* mutations were divided into two categories: truncating mutations (including nonsense, frameshift, splicing mutations and large rearrangements) and nontruncating mutations (including missense variants and in-frame short deletions and insertions).