Fig. S1 Distribution of kidney volumes within the different patient groups. Filled circles give values of right kidney (A) and total kidney (both organs) (B) as body surface-area related kidney volumes (RV/BSA) [21]. Median values of cohorts and normal range of body-surface-area related renal volume for one organ (grey bar) are indicated. Statistics for (A) p<0.05, Kruskal-Wallis, Dunn’s and (B) p<0.05, ANOVA, Tukey’s confirm increased kidney size for both organs in ARPKD as compared to NPH and BBS cohorts. Distribution of values corresponds to expected range for respective patient cohorts.
### Distribution of gene variants

| Patient   | Phenotype   | Gene variant(s)                                                                 | eGFR [mL/min/1.73m²] |
|-----------|-------------|--------------------------------------------------------------------------------|-----------------------|
| ARPKD 1   | ARPKD       | compound heterozygote PKHD1; missense (p), splice defect (m)                     | 35                    |
| ARPKD 2   | ARPKD       | compound heterozygote PKHD1; missense (p, m)                                    | 45                    |
| ARPKD 3   | ARPKD       | compound heterozygote PKHD1; missense (p, m)                                    | 40                    |
| ARPKD 4   | ARPKD       | compound heterozygote PKHD1; nonsense (p), in-frame deletion, (m)               | 32                    |
| ARPKD 5   | ARPKD       | compound heterozygote PKHD1; missense (p, m)                                    | 30                    |
| ARPKD 6   | ARPKD       | compound heterozygote PKHD1; missense (p, m)                                    | 137                   |
| ARPKD 7   | ARPKD       | compound heterozygote PKHD1; missense (p), nonsense (m)                         | 119                   |
| ARPKD 8   | ARPKD       | not available                                                                    | 45                    |
| ARPKD 9   | ARPKD       | compound heterozygote PKHD1; nonsense, missense (no segregation)                | 40                    |
| ARPKD 10  | ARPKD       | not available                                                                    | 35                    |
| NPH 1     | NPH1        | homozygote NPHP1, deletion exons 1-20                                            | 16                    |
| NPH 2     | NPH1        | homozygote NPHP1, deletion                                                       | 13                    |
| NPH 3     | NPH1        | homozygote NPHP1, deletion                                                       | 21                    |
| NPH 4     | NPH1        | compound heterozygote NPHP3, missense (p), nonsense (m)                          | 5                     |

Table S1-1
### Distribution of gene variants (continued)

| patient | phenotype            | gene variant(s)                          | eGFR [mL/min/1.73m²] |
|---------|----------------------|------------------------------------------|---------------------|
| BBS 1   | Bardet Biedl Syndrome| homozygote BBS7, missense                | 46                  |
| BBS 2   | Bardet Biedl Syndrome| homozygote BBS7, missense                | 69                  |
| BBS 3   | Bardet Biedl Syndrome| homozygote BBS10, out-of-frame deletion, STOP | 64                  |
| BBS 4   | Bardet Biedl Syndrome| homozygote BBS1, missense                | 103                 |
| BBS 5   | Bardet Biedl Syndrome| homozygote BBS4, deletion exons 7+8     | 86                  |

Table S1-2