

Supplementary Table S1. Three heterozygous missense variants of *NR1P1* (NM_003489.3) in three individuals with CAKUT

| FAMILY | Genomic position | Transcript position | Amino acid change (Het/hom) | Segregation | gnomAD (hom/het / WT allele count) | PPH2 | SIFT | Mutation Taster | <i>M.m.</i> | <i>G.g.</i> | <i>X.t.</i> | <i>D.r.</i> | ACMG classification | CAKUT Phenotype | Gender | Ethnicity |
|--------------|------------------|---------------------|-----------------------------|-------------|------------------------------------|------|------|-----------------|-------------|-------------|-------------|-------------|---------------------|------------------------------------|--------|------------|
| B633 | chr21:16,340,58 | c.456A>C | p.Gln152His (Het) | Unknown | 0/6/282,350 | 0.98 | D | DC | Q | Q | Q | Q | VUS | Left multicystic dysplastic kidney | F | Caucasian |
| A3460 | chr21:16,339,544 | c.970C>T | p.His324Tyr (Het) | Unknown | 0/0/Never reported | 0.74 | T | DC | H | H | P | Q | VUS | Left renal agenesis | M | Macedonian |
| A782 | chr21:16,339,171 | c.1343G>A | p.Arg448Gln (Het) | Unknown | 0/2/251,250 | 0.92 | D | DC | R | R | R | R | VUS | Right renal agenesis | M | Macedonian |

ACMG, American College of Medical Genetics and Genomics; CAKUT, congenital anomalies of kidneys and urinary tract; D, deleterious; DC, disease causing; Dr, *Danio rerio*; F, female; Gg, *Gallus gallus*; gnomAD, genome aggregation database (<https://gnomad.broadinstitute.org/>); Het, heterozygous; Hom, homozygous; M, male; Mm, *Mus musculus*; PPH2 score, PolyPhen-2 prediction score (0.0–1.0; i.e., tolerated to deleterious; variants from 0.85 to 1 are more confidently predicted to be damaging) (<http://genetics.bwh.harvard.edu/pph2/>); SIFT, sorting intolerant from tolerant (<https://sift.bii.a-star.edu.sg/>); T, tolerated; VUS, variants with unknown significance; Xt, *Xenopus tropicalis*; WT, wild type.

Red background represents deleterious prediction by the in silico algorithm. Blue background represents tolerated prediction by the in silico algorithm. The genomic coordinates are based on genome build GRCh37 (hg19), and transcript used is NM_003489.3.