

Supplementary Information

Supplementary Table 1. Gene lists by panel. Genes listed in bold were not included in the TruSight One panel testing prior to April 2018. Abbreviations are as follows: ADTKD, autosomal dominant tubulointerstitial kidney disease; aHUS/C3 GN, atypical hemolytic uremic syndrome-C3 glomerulonephritis; ARPKD, autosomal recessive polycystic kidney disease; BORS, branchio-oto renal syndrome; CAKUT, congenital anomalies of the kidney and urinary tract.

Panel	Gene list
ADTKD	<i>HNF1B, REN, SEC61A1, UMOD</i>
aHUS/C3 GN	<i>ADAMTS13, C3, C5_ex21, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, DGKE, MMACHC, THBD (PLG removed)</i>
Alport syndrome	<i>COL4A3, COL4A4, COL4A5</i>
ARPKD	<i>DZIP1L, PKHD1</i>
BORS	<i>EYA1, SIX1, SIX5</i>
CAKUT	<i>ACE, AGT, AGTR1, BICC1, BMP4, BMP7, CDC5L, CHD1L, CHD7, DHCR7, DSTYK, EYA1, EZH2, FAM58A, FGF10, FGF20, FGF8, FGFR1, FGFR2, FOXC1, FOXC2, FRAS1, FREM1, FREM2, GATA3, GLI3, GPC3, GRIP1, HNF1B, HOXA13, HOXA4, HOXB6, HPSE2, ITGA8, JAG1, KAL1, KDM6A, KMT2D, LRIG2, LRP4, NIPBL, NOTCH2, NPHP3, PAX2, PBX1, REN, RET, ROBO2, ROR2, SALL1, SALL4, SEMA3A, SIX1, SIX2, SIX5, SLIT2, SOX17 SRGAP1, TBC1D1, TBX18, TFAP2A, TRAP1, UMOD, UPK3A, WNT4, WT1</i>
Cystinosis	<i>CTNS</i>
Nephronophthisis & related ciliopathies	<i>AH11, ALMS1, ARL13B, ARL6, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CCDC28B, CEP164, CEP290, CEP41, CPLANE1, DCDC2, DYNC2H1, GLIS2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, INVS, IQCB1, KIF7, LZTFL1,</i>

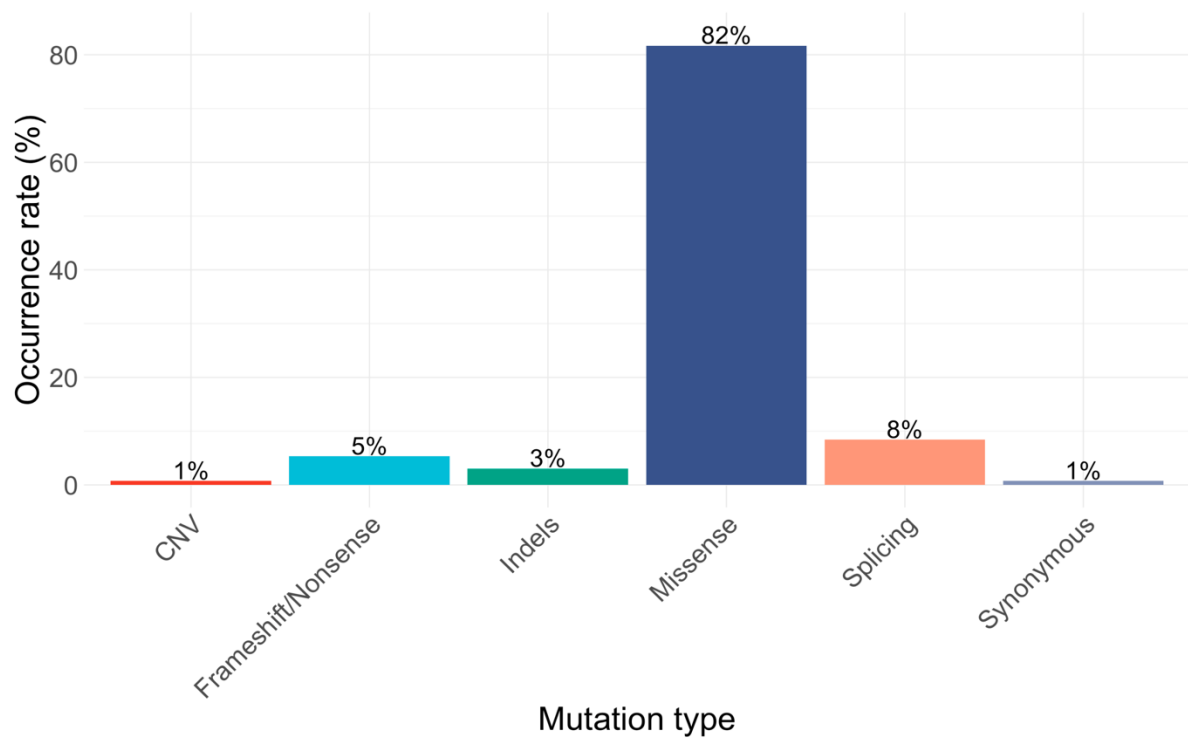
MKKS, MKSI, NEK1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TCTN1, TCTN2, TMEM138, TMEM216, TMEM237, TMEM67, TTC21B, TTC8, WDPCP, WDR19, WDR35, XPNPEP3 (EVC removed)

Nephrotic syndrome

*ACTN4, ALG1, ALMS1, ANLN, APOL1, ARHGAP24, **ARHGDI**A, CD2AP, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, **COQ7**, **COQ8B**, **CRB2**, CUBN, **DGKE**, **EMP2**, **FAT1**, INF2, ITGB4, KANK1, **KANK2**, **KANK4**, LAMB2, LMNA, LMX1B, MAGI2, MYH9, MYO1E, NPHS1, NPHS2, **NUP107**, **NUP205**, **NUP93**, OCRL, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, TTC21B, WT1, **XPO5***

Tubulopathies

*ADCY10, AGXT, ALPL, AP2S1, AQP2, APRT, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CA2, CASR, CDC73, CLCN5, CLCNKA, CLCNKB, **CLDN10**, CLDN16, CLDN19, CNNM2, CUL3, CYP24A1, CYP27B1, DMP1, **EHHADH**, ENPP1, FAH, FAM111A, FAM20A, FGF23, FXYD2, **GNA11**, GRHPR, HNF1B, HNF4A, HOGA1, HPRT1, KCNA1, KCNJ1, KCNJ10, KL, KLHL3, **MAGED2**, MAGT1, NR3C2, OCRL, PCBD1, PHEX, PTH1R, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC7A9, SLC9A3R1, TRPM6, VDR, WNK1, WNK4, XDH*



Supplementary Figure 1. Distribution of mutation types of variants of uncertain significance (VOUS) (n = 552 probands). Variant classification was based on 2015 ACMG guidelines²⁰. Abbreviated mutation types are as follows: CNV, copy number variation; indels, insertions or deletions.