

aCGH = array comparative genomic hybridization
ACGS = association for clinical genetic science
ACMG = American college of medical genetics and genomics
AD = autosomal dominant
ADPKD = autosomal dominant polycystic kidney disease
ADTKD = autosomal dominant tubulo-interstitial kidney disease
aHUS = atypical hemolytic–uremic syndrome
APECED = autoimmune polyendocrinopathy-candidiasis–ectodermal dystrophy
AR = autosomal recessive
ARPKD = autosomal recessive polycystic kidney disease
AS = Alport syndrome
BBS = Bardet-Biedl syndrome
BOR(S) = branchio-oto-renal (syndrome)
C3GN = C3 glomerulopathy
CAKUT = congenital anomalies of kidney and urinary tract
CKD = chronic kidney disease
CMA = chromosomal microarray
CNS = congenital nephrotic syndrome
CNV = copy number variations
COL4-NP = COL4-nephropathy
DDD = dense deposit disease
DP = definitely pathogenic
dRTA = distal renal tubular acidosis
ESKD = end-stage kidney disease
FH = family history
FSGS = focal segmental glomerulosclerosis
FU = follow-up
GD = genomic disorders
GN = glomerulonephritis
GS = Gitelman syndrome
GWAS = genome-wide association study

HLP = highly likely pathogenic

IgA = immunoglobulin A nephropathy

IQR = interquartile range

JS = Joubert syndrome

JSRD = Joubert syndrome related disorders

LP = likely pathogenic

LR = long-range

KF = kidney failure

KTx = kidney transplantation

MCDK = multicystic dysplastic kidney

Min. = minimum

MKS = Meckel syndrome

MLPA = multiplex ligation-dependent probe amplification

mtDNA = mitochondrial DNA

N/A = not applicable

NGS = next generation sequencing

NP = not performed

NPHP = nephronophthisis

NPHP-RC = nephronophthisis-related ciliopathies

NPHP-RD = nephronophthisis-related disorder

NR = not reported

NS = nephrotic syndrome

P = pathogenic

PCR = polymerase chain reaction

PKD = polycystic kidney disease

PUJ = pelviureteric junction

PUV = posterior urethral valves

QMPSF = quantitative multiplex polymerase chain reaction of short fluorescent

RCAD = renal cyst and diabetes syndrome

RHD = renal hypodysplasia

RRT = renal replacement therapy

SD = standard deviation

SNP-array = single nucleotide polymorphism microarray

SNV = single nucleotide variant

SRNS = steroid-resistant nephrotic syndrome

SSNS = steroid-sensitive nephrotic syndrome

TBMN = thin basement membrane nephropathy

TIKD = tubulointerstitial kidney disease

TMA = thrombotic microangiopathy

TSC = tuberous sclerosis complex

TTP = thrombotic thrombocytopenic purpura

URA = unilateral renal abnormality

VACTERL = acronym including vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities

VEO = very early onset

VNTR = variable number of tandem repeats

VUJ = vesicoureteric junction

VUR = vesicoureteral reflux

VUS = variant of unknown/uncertain significance

WES = whole exome sequencing

WGS = whole genome sequencing

XL = X-linked