Supplementary Table 1a – summary of individual ADTKD studies. Patient, cohort and test characteristics per study are presented. Diagnostic yield and percentage of diagnostic yield that is explained by CNVs are summarized. In grey font additional info on phenotypes, extrapolated cohort and diagnostic yield for both of these are presented if available. A summary of noteworthy details is presented per study, including patient characteristics that are expected to have positively impacted the diagnostic yield. Abbreviations are listed in Supplementary Table 2.

ADTKD	phenotype	n of population	pediatric / adult at time of study	positive family history (n, %)	consang. (n, %)	extra-renal features (n, %)	adult-onset disease (n, %)	ESKD (n, %)	type of variants tested	n genes analyzed	diagnostic yield (n, %)	percentage of yield by CNV (%)
[Ayasreh et al., 2018]	referred with ADTKD	56 families	NR	NR	NR	NR	NR	NR	SNVs + CNVs	4	25/56 (44.6%)	0%
Noteworthy: phenotype data only reported for mutation positive families therefore not included in table												
[Gast et al., 2018]	genetic testing performed in patients with no established conflicting genetic diagnosis ADTKD n=28; unknown familial nephropathy n=44 of which n=33 consistent with ADTKD; others genotyped included GN; FSGS, IgA and reflux	113 (from CKD3-5 cohort of n=3770, n=399 MKD suspected 2027 replied to questionnaire)	adult	min. 217/399 (54.4%) 217/2027 (10.7%)	NR	NR	NR, mean 68 years for all responders at time of questionnaire	269/399 (67.4%) 72/2027 (38.1%) | 1425/3770 (37.8%)	SNVs	1 or custom gene panel in n=3	35/113 (31.0%) 35/3770 (0.9%) of CKD 3+ | 29/1425 (2.0%) of ESKD | 35/399 (8.7%) of inherited kidney disease (24% of inherited kidney disease minus ADPKD) unknown familial nephropathy 13/44 (29.5%); unknown with ADTKD 13/33 (39.4%); all ADTKD 35/61 (57.4%)	NP
Noteworthy: 252/399 (63.2%) ADPKD | also other (suspected) genetic diagnosis reported (without causative gene): most prevalent AS in 25/399 (6.3%) & FSGS/SRNS in 13/399 (3.3%) | authors reported only yield for UMOD| yield impacted by: family history; ESKD												
[Bleyer et al., 2020]	patient referred with ADTKD (either by health care professional or self-referral n=176)	665 individuals (from 828 referrals of which 77 had prior genetic diagnosis) - genetic testing performed in 275	adult for first affected contacts	min. 456/629 (72.4%)	NR	NR	NR	NR	SNVs	5	172/275 (62.5%) 172/665 (25.9%) of potential participants | 249/823 (30.3%) of referred patients	NP
Noteworthy: limited information provided for type of genetic test performed and unclear if more than 5 genes were tested | criteria for variant classification NR												
[Olinger et al., 2020]	presumptive diagnosis of ADTKD	585 families (726 individuals)	NR	NR	NR	NR	NR, median 45 years (IQR 31, 58)	216/503 (42.9%)	SNVs + CNVs	1 and/or 2	309/585 (52.8%)	1.3%
Noteworthy: UMOD testing n=562 (solely UMOD in 357); MUC1 testing n=228 (solely MUC1 in 23)												

Supplementary Table 1b – summary of individual CAKUT studies. Patient, cohort and test characteristics per study are presented. Diagnostic yield and percentage of diagnostic yield that is explained by CNVs are summarized. In grey font additional info on phenotypes, extrapolated cohort and diagnostic yield for both of these are presented if available. A summary of noteworthy details is presented per study, including patient characteristics that are expected to have positively impacted the diagnostic yield. Abbreviations are listed in Supplementary Table 2.

CAKUT	phenotype	n of population	pediatric / adult at time of study	positive family history (n, %)	consang. (n, %)	extra-renal features (n, %)	adult-onset disease (n, %)	ESKD (n, %)	type of variants tested	n genes analyzed	diagnostic yield (n, %)	percentage of yield by CNV (%)
[Sanna-Cherchi et al., 2012]	Renal agenesis or hypodysplasia	522	pediatric	96/388 (24.7%)	NR	142/522 (27.2%)	0%	NR	CNVs	genome wide	55/522 (10.5%)	100%
Noteworthy: combination of n=192 discovery cohort, n=196 and n=134 replication cohort | None of the cases was screened for mutations in known genes, such as PAX2, HNF1B, EYA1 | yield impacted by: extra-urinary tract malformations												
[Hwang et al., 2014]	isolated CAKUT VUR n=288; renal hypodysplasia n=120; unilateral renal agenesis n=90	650 families (749 individuals)	both	100/650 (15.4%)	NR	0%	0%	NR	SNVs	17	41/650 (6.3%)	NP
Noteworthy: patients with syndromic CAKUT were excluded												
[Kohl et al., 2014]	isolated CAKUT	590 families (672 individuals)	NR	NR	NR	NR	0%	NR	SNVs	12 (AR genes)	15/590 (2.5%)	NP
Noteworthy: mutations in 17 known autosomal dominant CAKUT-causing genes were excluded before this study												
[Caruana et al., 2015]	CAKUT - consecutive series MCDK n=10; PUV n=29; VUR n=29; hypo/dysplasia n=23; hydronephroureterosis n=17; PUJ obst n=26; duplex kidney n=25; agenesis n=10; VUJ obst n=9	195 unrelated families (201 individuals) - test performed in 178	pediatric	31/195 (16.4%)	NR	20.5% (n/n NR)	0%	NR	CNVs	genome wide	7/178 (3.9%) 7/195 (3.6%) in entire cohort VUR 2/29 (6.9%); PUV 2/29 (6.9%); hydronephroureterosis 2/17 (11.8%); MCDK 1/10 (10.0%)	100%
Noteworthy: patients were not screened for mutations in any known CAKUT-causing genes| yield impacted by: phenotype MCDK and PUV higher incidence of CNV												
[Nicolaou et al., 2016]	CAKUT	453	pediatric	44/453 (9.7%)	NR	64/453 (14.1%)	0%	NR	SNVs + CNVs	208	6/453 (1.3%)	0%
Noteworthy: using a burden test, no significant excess of rare variants in any of the genes in our cohort compared with controls was found | paper also describes candidate variants of pathogenicity, these are not included in the yield reported here| yield impacted by: family history												

[Xi et al., 2016]	MCDK isolated MCDK n=33; non-isolated MCDK n=4	37	pediatric	NR	NR	4/37 (10.8%)	0%	NR	CNVs	genome wide	5/37 (13.5%) isolated 4/33 (12.1%); non-isolated 1/4 (25.0%)	100%
Noteworthy: yield impacted by: extra-renal features												
[Faure et al., 2016]	PUV	45	pediatric	NR	NR	NR	0%	7/45 (15.6%)	CNVs	genome wide	2/45 (4.4%)	100%
Noteworthy: patients with known chromosomal abnormalities were excluded| yield impacted by: CKD5												
[Fu et al., 2016]	MCDK	72 (of 370 with CAKUT in consecutive cohort)	fetuses	NR	NR	10/72 (13.9%)	0%	NR	CNVs	genome wide	8/72 (11.1%) 8/370 (2.2%) in entire CAKUT cohort	100%
Noteworthy: karyotyping performed in n=72, CMA performed in n=30; yield karyotyping: 3/72 (4.2%) - CMA: 5/30 (16.7%) | yield impacted by: bilateral MCDK; extra renal abnormalities												
[Vivante et al., 2017]	CAKUT	33 families (33 individuals)	NR	NR	100%	NR	0%	NR	SNVs	exome wide	9/33 (27.1%)	NP
Noteworthy: mutations in 17 genes known to be mutated in isolated CAKUT in humans were excluded previously in this cohort | A clinical diagnosis of a known monogenic syndrome had not been previously made in any of the patients | AGXT, AQP2, CTNS, and PKHD1 cause, if mutated, a kidney disease that may represent a phenocopy of CAKUT.												
[Bekheirnia et al., 2017]	CAKUT	62 families	both	10/62 (16.1%)	NR	19/31 (30.6%)	0%	NR	SNVs + CNVs	35	7/62 (11.3%)	57.1%
*Noteworthy: exclusion criterium: individuals with syndromic forms of CAKUT in which an underlying genetic etiology was known and individuals with nonsyndromic and nonfamilial forms of VUR | *two more syndromic families were identified after WES results became available; accordingly, total syndromic = 21 (34%) | FOXP1 identified as CAKUT candidate gene (not included in yield)*												
[Heidet et al., 2017]	CAKUT (bilateral or unilateral with extra-renal defects or positive family history) + 11 patients with BOR without a renal phenotype - severe fetal cases n=93 (45%)	204 + 11 BOR without renal phenotype	both	70/204 (34.3%)	NR	79/204 (38.7%)	0%	NR	SNVs + CNVs	330	36/204 (17.6%) 3/11 (27.3%) for BOR without renal phenotype	44.4%
Noteworthy: patients with posterior urethral valves were not included in the study | 50/204 patients had been previously tested negative for mutations in HNF1B and/or PAX2 (involved in papillorenal syndrome) and/or EYA1 by Sanger sequencing; 9 fetuses had been tested for mutations in RET												
[Lei et al., 2017]	CAKUT	30	fetuses	NR	0%	8/30 (26.7%)	0%	N/A	SNVs	exome wide	4/30 (13.3%) isolated CAKUT 2/22 (9.1%); CAKUT with other abnormalities 2/8 (25%)	NP
Noteworthy: fetuses had normal findings upon karyotyping and chromosome microarray analysis| yield impacted by: extra-renal features												

[Rasmussen et al., 2018]	bilateral kidney anomalies	56 families (62 fetuses)	fetuses	11/56 (19.6%)	0%	NR	0%	NR	SNVs + CNVs	108 or exome wide	7/56 (12.5%)	0%
Noteworthy: GREB1L and ROBO1 identified as novel disease genes, adding these as solved patients increases the yield to 11/56 (19.6%)												
[Unzaki et al., 2018]	clinically diagnosed BOR syndrome	36 families (51 individuals)	both	18/36 (50.0%)	NR	100%	0%	NR	SNVs + CNVs	depending on findings: 2 to max 172 genes	26/36 (72.2%)	30.7%
Noteworthy: tiered approach												
[Boissel et al., 2018]	CAKUT (severe; from terminated pregnancies) isolated bilateral renal agenesis or dysgenesis n=11; VACTERL n=9; cerebral anomalies n=36; suspected ciliopathies n=5; miscellaneous patterns of multiple malformations n=32; fetal akinesia n=8	101	fetuses	9/101 (8.9%)	8/101 (7.9%)	90/101 (89.1%)	0%	N/A	SNVs	exome wide	19/101 (18.8%) ciliopathies (80%); cerebral anomalies (19%); multiple malformations (19%); fetal akinesia (25%); renal a/dysgenesis (0%); VACTERL association (11%)	NP
Noteworthy: neither karyotyping (n = 58 cases) nor chromosomal microarray analysis (n = 84 cases) showed any candidate or pathogenic CNVs | GREB1L identified as candidate gene (not included in yield) | yield impacted by: ciliopathies												
[Van Der Ven et al., 2018]	CAKUT	232 families (488 individuals: 319 affected; 169 unaffected)	both	40/232 (17.2%) multiplex families included	reported 50/232 (21.6%); likely consanguinity 43/232 (18.5%); combined 93/232 (40.0%)	79/319 (24.7%)	0%	NR	SNVs + CNVs	40/219/ 404 genes or exome wide	32/232 (13.8%)	3.1%
Noteworthy: genetic test depending on findings/homozygosity mapping/availability parental DNA family: tiered approach | In 61/232 (26.3%) candidate mutations | In 15/155 families with isolated CAKUT deleterious mutations detected in syndromic CAKUT genes | yield impacted by: syndromic, consanguinity												
[Li et al., 2019]	fetuses with CAKUT referred for invasive prenatal diagnosis	123	fetuses	NR	NR	87/123 (70.7%)	0%	NR	CNVs	genome wide	17/123 (13.8%)	100%
Noteworthy: incremental yield of CMA over karyotyping was 3.6% | meta-analysis indicates that the incremental yield of CMA over karyotyping was 3.8% | yield impacted by: non-isolated CAKUT												
[Verbitsky et al., 2019]	CAKUT	2824	both	413/2824 (14.6%)	4/125	570/2824 (20.2%)	0%	NR	CNVs	genome wide	159/2824 (5.6%)	100%

Noteworthy: affected individuals carried a significant burden of rare exonic CNVs and were enriched for known genomic disorders (GD). Kidney anomaly cases were most enriched for exonic CNVs, encompassing GD-CNVs and novel deletions; obstructive uropathy had a lower CNV burden and an intermediate prevalence of GD-CNVs; and vesicoureteral reflux had the fewest GD-CNVs but was enriched for novel exonic CNVs, particularly duplications. | yield impacted by: extra-renal features and multiple urinary tract infections, phenotype: kidney anomalies												
[Lin et al., 2019]	CAKUT - consecutive cohort	331	fetuses	NR	NR	123/331 (37.2%)	0%	NR	CNVs	genome wide	25/331 (7.6%) isolated CAKUT 10/208 (4.8%); non-isolated CAKUT 15/123 (12.2%)	100%
Noteworthy: fetuses with a previously known family history of autosomal recessive or dominant polycystic kidney disease were not included in the study (n not reported) | yield impacted by: extra-renal features												
[Cai et al., 2020a]	CAKUT	147	fetuses	NR	NR	22/147 (15.0%)	0%	NR	CNVs	genome wide	6/147 (4.1%) isolated CAKUT 4/125 (3.2%) non-isolated CAKUT 2/22 (9.1%)	100%
Noteworthy: pathogenic CNV inherited from unaffected mother might present incomplete penetrance | yield impacted by: extra-renal features												
[Ahn et al., 2020]	CAKUT	94	both	5/94 (5.3%)	NR	62/94 (66.0%)	0%	40/94 (42.6%)	SNVs + CNVs	60	13/94 (13.8%)	46.2%
Noteworthy: only CNVs involving captured areas of TES were searched| yield impacted by: syndromic features												
[Lei et al., 2020]	CAKUT (tertiary level referral center) isolated CAKUT n=125; multisystem anomalies n=37	163 fetus-parental trios (of 725 CAKUT cases in total cohort)	fetuses	1/163 (0.6%)	0%	37/163 (22.7%)	0%	NR	SNVs	exome wide	20/163 (12.3%) 105/725 (14.5%) in entire CAKUT cohort isolated CAKUT 10/125 (8.0%); multisystem anomalies 10/37 (27.0%)	NP
Noteworthy: familial cases were excluded, however later found out that mother of one case had polycystic kidneys | previously normal findings upon karyotyping and CMA in this cohort (from larger cohort of 725 fetuses with CAKUT, 33 had abnormal karyotype, 52 had abnormal CMA, 457 declined genetic testing/CMA/WES, 20 insufficient DNA). Yield extrapolated to cohort where genetic testing was performed (CMA/karyotype included): 105/248 (42.3%); yield in entire cohort: 105/725 (14.5%) | yield impacted by: extra-renal features												
[Zhou et al., 2020]	unexplained isolated CAKUT bilateral kidney anomalies n=19; unilateral fetal renal abnormalities n=22	41	fetuses	1/41 (2.4%)	0%	0%	0%	NR	SNVs	exome wide	3/41 (7.3%) bilateral kidney anomalies 3/19 (15.8%); unilateral fetal renal abnormalities 0/22 (0%)	NP
Noteworthy: fetuses whose tests revealed aneuploidy or CNVs which justified the fetal anomalous phenotypes, were excluded from analyses | 28 cases with parent/fetus trio; 13 cases only proband | yield impacted by: bilateral kidney anomalies												
[Cai et al., 2020b]	RHD isolated RHD n=103; non-isolated RHD n=17	120	fetuses	NR	NR	17/120 (14.2%)	0%	NR	CNVs	genome wide	11/120 (9.2%) isolated RHD 10/103 (9.7%);	100%

											non-isolated RHD 5/17 (29.4%)	
Noteworthy: yield impacted by: extra-renal features												
[Zhou et al., 2021]	fetuses with solitary functioning kidney on ultrasound - including MCDK, RHD, URA, ectopic kidney, duplex kidney	99	fetuses	NR	NR	NR	0%	NR	CNVs	genome wide	14/99 (14.1%)	100%
Noteworthy: QPRT identified as candidate gene												

Supplementary Table 1c – summary of individual ciliopathy studies. Patient, cohort and test characteristics per study are presented. Diagnostic yield and percentage of diagnostic yield that is explained by CNVs are summarized. In grey font additional info on phenotypes, extrapolated cohort and diagnostic yield for both of these are presented if available. A summary of noteworthy details is presented per study, including patient characteristics that are expected to have positively impacted the diagnostic yield. Abbreviations are listed in Supplementary Table 2.

ciliopathies	phenotype	n of population	pediatric / adult at time of study	positive family history (n, %)	consang. (n, %)	extra-renal features (n, %)	adult-onset disease (n, %)	ESKD (n, %)	type of variants tested	n genes analyzed	diagnostic yield (n, %)	percentage of yield by CNV (%)
ADPKD												
[Rossetti et al., 2012]	ADPKD	230	NR	NR	NR	NR	NR	NR	SNVs	2	115/183 (62.8%)	NP
Noteworthy: yield is based on 183/230 patients with typical ADPKD												
[Hwang et al., 2016]	ADPKD	220 families (of cohort of 288)	adult	NR	NR	NR	100%,	31.6%	SNVs + CNVs	2	186/220 (84.5%) 188/288 (65.3%) in entire at risk or affected with ADPKD cohort	1.6%
Noteworthy: exclusion criterium: serum creatinine >1.4 mg/dl at presentation; entire cohort of 288 subjects at risk or affected with ADPKD (of which 2 were found to harbor HNF1B mutations) | yield impacted by: ESKD												
[Jin et al., 2016]	ADPKD	148	both	82/148 (55.4%)	NR	NR	NR, mean 34 years (± 10.1 SD, range 12-66)	NR	SNVs	2	76/148 (51.4%)	NP
Noteworthy: yield impacted by: disease severity												
[Kinoshita et al., 2016]	ADPKD	101	adult	NR	NR	NR	NR, all >19 at time of testing	NR	SNVs + CNVs	2	94/101 (93.1%)	4.3%
[Xu et al., 2018]	ADPKD	120 families	NR	NR	NR	NR	NR	42/73 (57.5%) before age 60	SNVs + CNVs	2	98/120 (81.7%)	2.0%
Noteworthy: yield based on probably pathogenic and definite pathogenic mutations. 69.9% had a definite pathogenic variant												
[Fujimaru et al., 2018]	ADPKD without positive family history	53	adult	0%	NR	40/53 (75.5%) liver cysts	NR, mean 56 years (46-68) at time of genetic testing	10/53 (18.9%)	SNVs + CNVs	69	35/53 (66.0%)	0%
Noteworthy: those with extra-renal clinical findings related to renal cystic disease other than ADPKD (eg, hepatic fibrosis and retinitis pigmentosa) and those younger than 20 years of age were excluded | CKD5 reported separately 4/53 (7.5%) from ESKD| yield impacted by: polycystic liver disease												
[Zhang et al., 2019]	ADPKD	62	NR	NR	NR	NR	NR, mean age: PKD1 mutation: 43.5 years (± 10.6); PKD2	12/62 (19.4%)	SNVs + CNVs	3	56/62 (90.3%)	1.8%

							mutation: 44.7 years (± 6.6)					
Noteworthy: article includes VUS in detection rate of 95.2%. Mentioned diagnostic yield in this table based on LP and P mutations.												
[Mochizuki et al., 2019]	ADPKD	111	NR	NR	NR	NR	NR	NR	SNVs + CNVs	2	102/111 (91.9%)	4.9%
Noteworthy: detection rate of NGS alone: 86.5%												
[Mantovani et al., 2020]	suspected ADPKD	191 (+21 validation cohort)	adult	148/193 (76.7%)	NR	128/159 (80.5%)	NR, median 25 years	42/165 (25.5%)	SNVs + CNVs	16	119/191 (62.3%)	6.7%
Noteworthy: validation cohort n=21; confirmation cohort severe ADPKD n=36, discovery cohort n=155 (total n=212). Patient characteristics are based on entire cohort. Yield based on confirmation cohort + discovery cohort | yield impacted by: severe disease (ESKD)												
[Schönauer et al., 2020]	ADPKD	100 families (122 individuals)	NR	61/69 (88.4%)	NR	93/122 (76.2%) liver cysts	NR, mean current age of 56.7 years	68/122 (55.7%)	SNVs + CNVs	5 or exome wide in unsolved families	84/100 (84.0%)	7.1%
Noteworthy: four potential candidate genes identified (TSC2, GLI2, ALG6, LRP5)												
[Mallawaarachchi et al., 2021]	ADPKD (52% atypical) referred for diagnostic WGS	144	both	64/141 (45.4%)	NR	47/144 (32.6%) liver cysts	132/144 (91.7%) at time of referral	NR	SNVs + CNVs	13	69/144 (47.9%)	5.8%
Noteworthy: diagnostic rate of 81% in patients with typical ADPKD (98% with PKD1/PKD2 variants) and 60% in those with atypical features (56% PKD1/PKD2; 44% PKHD1/HNF1B/GANAB/ DNAJB11/PRKCSH/TSC2) | yield impacted by: typical ADPKD												
[Nielsen et al., 2021]	suspected ADPKD	118 (cohort of 147 with genetic testing for PKD1 / PKD2 / GANAB)	both	NR	NR	NR	135/147 (91.8%)	NR	SNVs + CNVs	3	103/118 (87.3%) 103/147 (70.0%) in entire cohort where genetic testing was performed	min. 6.8%
Noteworthy: paper also reports possibly pathogenic variants in diagnostic yield, which also includes VUS as classified bij ACMG | yield impacted by: patients specifically indicated to be suspected of ADPKD												
[Durkie et al., 2021]	Patients referred for ADPKD genetic testing with onset before age of 18 months	36 (from cohort of 51 with VEO-ADPKD)	perinatal / pediatric	NR	NR	NR	0%	NR	SNVs	2 or 17 (depending on time of testing); n=1 WES; n=1 WGS	21/36 (58.3%) 36/51 (70.6%) in entire cohort VEO-ADPKD	NP
Noteworthy: patients with a clinical history not suggestive of ADPKD or with an alternative genetic diagnosis (n=15: 9 biallelic PKHD1 / 6 HNF1B deletion) were not included in sequencing. Yield 21/36 based on likely biallelic PKD1/2 (suppl table). In 20/21 extended cystogenes panel / WES / WGS performed which was negative | Nine infants had a single PKD1 variant (5P; 4 likely hypomorphic), these infants were reported as genetically unresolved												
other/mixed ciliopathies												

[Bachmann-Gagescu et al., 2015]	JS	375 families (440 individuals) (from cohort of 440 families (532 individuals))	both	79/440 (17.9%)	84/440 (19.1%)	100%	NR, mean 13.1 years (SD 1.9) at time of analysis	NR	SNVs	27	232/375 (61.8%) 232/440 (52.7%) in entire cohort	NP
Noteworthy: phenotypic analysis revealed that only 34% of individuals have a 'pure JS' phenotype. Retinal disease is present in 30% of individuals, renal disease in 25%, coloboma in 17%, polydactyly in 15%, liver fibrosis in 14% and encephalocele in 8%. | Despite satisfying our criteria (MAF<0.2%, CADD>11), the variants in 12 families did not meet the ACMG variant interpretation categories 1, 2 or 3 (2007 guidelines, comparable to P, LP and VUS current guidelines) --> these are still included in reported yield, yield would be 220/375 (58.7%) if these were to be excluded | yield impacted by: positive family history and consanguinity												
[Knopp et al., 2015]	JS, JSRD, MKS, BBS MKS n=88, JS/JSRD n=61, BBS n=66	215 families - in 140 test performed	both	NR	57/215 (26.5%)	100%	NR	NR	SNVs or CNVs	4 or 5 genes for SNVs depending on genotype	73/140 (52.1%) 73/215 (22.4%) in entire cohort BBS 26/66 (39.4%), JSRD 14/31 (45.2%), MKS 33/43 (76.7%); yield in consanguineous families 16/26 (61.5%)	0%
Noteworthy: detection rate in consanguineous families 62%| yield impacted by: consanguinity, phenotype MKS												
[Braun et al., 2016]	suspected NPHP-RC based on renal ultrasound	79 families (103 individuals)	NR	19/79 (24.1%)	60/79 (75.9%)	NR	0%	NR	SNVs	exome wide - additional analysis of 90 genes	50/79 (63.3%)	NP
Noteworthy: suspicion of NPHP-RC based on renal ultrasound: chronically increased echogenicity, loss of cortico-medullary differentiation, and/or ≥2 renal cysts | in all families, a homozygous deletion in the NPHP1 gene had been excluded prior to study inclusion. Inclusion was based on suspicion of NPHP-RC, however 18/50 individuals (36%) harbored a mutation in a gene that causes a monogenic kidney disease different from NPHP-RC: 8/50 renal tubulopathies, 4/50 AS, 3/50 CAKUT, 2/50 ARPKD, 1/50 APECED | yield impacted by: consanguinity												
[Schueler et al., 2016]	NPHP-RC	384 families	NR	84/384 (21.9%)	67/384 (17.4%)	NR	NR	NR	SNVs + CNVs	34	81/384 (21.1%)	0%
Noteworthy: prior to study enrolment, individuals with homozygous deletions of NPHP1 were excluded by using a multiplex PCR-based deletion analysis.												
[Al-Hamed et al., 2016]	Cystic, enlarged or echogenic kidneys on fetal ultrasound	44 families	fetuses	26/44 (59.1%)	38/44 (86.4%)	38/44 (86.4%)	0%	NR	SNVs	90	28/44 (63.6%)	NP
Noteworthy: in group B, where DNA was available from both parents but not the affected child (10 families), homozygous mutations were inferred by finding identical heterozygous variants in both parents in seven cases, consistent with the known parental consanguinity | yield impacted by: consanguinity												
[Lindstrand et al., 2016]	BBS	92	both	NR	NR	NR	NR	NR	CNVs + SNVs in CNV positive cases	94	17/92 (18.5%)	100%*
Noteworthy: criteria for variant classification NR; 5 patients were compound heterozygotes for CNV+SNV												

[Vilboux et al., 2017]	JS and related disorders	86 families (100 individuals)	both	NR	1/100 (1%)	100/100 (100%)	0%	NR	SNVs	27 followed by WES	81/86 (94.1%)	NP
Noteworthy: in this article, for simplicity, "JS" includes Senior-Løken and COACH syndromes												
[Stokman et al., 2018]	(suspected) nephronophthisis related ciliopathy	36 families (40 individuals) - current genetic testing performed in 12 families (13 patients)	both	NR	NR	22/40 (55.0%)	NR, mean 9 years (range isolated 5-26; range syndromic 5-33)	24/40 (60.0%)	SNVs + CNVs	15 or WES	4/12 (33.3%) based on current testing 24/36 (66.7%) families || 28/40 (67.5%) individuals including predetermined genetic diagnoses	100% (in n=12) 54.2% in entire cohort
Noteworthy: diagnosis already established in n=23 individuals, of which one was comp. het. with VUS | 12 additional patients tested with ciliopathy and renal features that did not fulfill clinical cirteria of NPH (yield in overal cohort: 39/52 (75%) individuals)												
[Szabó et al., 2018]	ARPKD	36 families (37 individuals	NR	NR	0%	NR	0%	10/37 (27%)	SNVs + CNVs	4813	35/36 (97.2%)	22.9%
Noteworthy: no extra-renal and hepatic involvement suggestive of other ciliopathies was part of inclusion criteria | criteria for variant classification NR | phenocopies: PKD1, HNF1B, NPHP1, TMEM67, PKD1/TSC2												
[Al Alawi et al., 2019]	presumed inherited cystic kidney disease ADPKD n=16; ARPKD n=16; NPHP-RC n=12; ciliopathy syndromes n=5; unspecified cystic kidney disease n=4	53	both	39/53 (73.6%)	11/53 (20.8%)	NR	NR, median age at study inclusion 10 years (range 0-63)	25/53 (47.2%)	SNVs + CNVs	49	39/53 (73.6%) ADPKD 12/16 (75%); ARPKD 16/16 (100%); NPHP-RC 8/12 (66.7%); ciliopathy syndromes 1/5 (20%); unspecified cystic kidney disease 3/4 (75%)	7.5%
Noteworthy: authors reported higher yield (75%), including 1 VUS in WRD19 which segregated with phenotype | Molecular genetic testing changed the diagnosis in 6% and reveled a diagnosis in 6% with unspecified cystic kidney disease | No difference in yield in pediatric versus adult patients | yield impacted by: phenotype: ARPKD												
[Liang et al., 2020]	clinically suspected of cilia-related kidney disorders both ADPKD and syndromal ciliopathies; % NR	33 families (44 individuals)	both	20/33 (60.6%)	NR	23/33 (69.7%) | 26/44 (59.1%)	NR	6/44 (13.6%)	SNVs	88	21/33 (63.6%)	NP
[Obeidova et al., 2020]	cystic kidney diseases (ARPKD, ADPKD, NPHP,	31	pediatric	2/31 (6.5%) parents showed	NR	23/31 (74.2%)	0%, 15/31 neonatal (prenatal); 8/31	NR	SNVs + CNVs	118 or 153 (updated version)	22/31 (71.0%)	9.1%

	RCAD syndrome, BBS) ARPKD n=20; NPHP n=1; ADPKD (VEO) n=6; RCAD syndrome n=3; Bardet-Biedl syndrome n=1			ADPKD on ultrasound			infantile; 8/31 childhood					
Noteworthy: clinically based diagnosis changed in 16% of patients												
[Yue et al., 2020]	NPHP	48 families (55 individuals)	pediatric	NR	NR	23/48 (47.9%)	0%, median age 7 years (range 6 days - 17 years)	NR	SNVs + CNVs	63 + additional genes in some cases	19/48 (39.6%)	36.8%
[Al Alawi et al., 2020]	ARPKD	32 families (40 individuals)	pediatric	24/32 (75.0%)	21/32 (65.6%)	12/40 (30.0%)	0%	12/40 (30.0%)	SNVs	1 or 49	30/32 (93.8%)	NP
Noteworthy: diagnosis already established in n=18 individuals which are included in yield for newly sequenced 22 patients only PKHD1 exons 3, 6, 32 and 58 were analyzed												
[Benson et al., 2021]	PKD	148 families (169 individuals)	adult	131/169 (77.5%)	NR	85/169 (50.3%) liver cysts	NR, mean 36 years (4-79)	114/169 (67.4%)	SNVs + CNVs	227 (n=14 only 11 genes)	100/148 (67.6%)	3.0%
Noteworthy: yield using Mayo Clinic pathogenicity guidelines = 120/148 (81.1%) | Cases with previous genetic diagnosis of PKD not included | yield impacted by: age at diagnosis												

Supplementary Table 1d – summary of individual glomerulopathy studies. Patient, cohort and test characteristics per study are presented. Diagnostic yield and percentage of diagnostic yield that is explained by CNVs are summarized. In grey font additional info on phenotypes, extrapolated cohort and diagnostic yield for both of these are presented if available. A summary of noteworthy details is presented per study, including patient characteristics that are expected to have positively impacted the diagnostic yield. Abbreviations are listed in Supplementary Table 2.

glomerulo-pahies	phenotype	n of population	pediatric / adult at time of study	positive family history (n, %)	consang. (n, %)	extra-renal features (n, %)	adult-onset disease (n, %)	ESKD (n, %)	type of variants tested	n genes analyzed	diagnostic yield (n, %)	percentage of yield explained by CNV (%)
nephrotic syndrome												
[McCarthy et al., 2013]	childhood SRNS	36	pediatric	10/36 (27.8%)	NR	NR	0%	12/36 (33.3%)	SNVs	446	11/36 (30.6%)	NP
Noteworthy: reported yield based on 24/446 genes | yield impacted by: lower age of onset & familial disease												
[Al-Hamed et al., 2013]	CNS, infantile NS and childhood SRNS	49 families (62 cases)	both	12/49 (24.5%)	37/49 (75.5%)	NR	0%	20/49 (40.8%)	SNVs	9	25/49 (51.0%)	NP
Noteworthy: yield impacted by: family history, consanguinity												
[Kari et al., 2013]	children with SRNS >1 year with genetic testing performed where renal biopsy was performed primary SRNS n=36; secondary SRNS n=8	44 (from cohort of 242 children with NS of which 214 age >1 year; including 150 SSNS; 64 SRNS (incl. 11 secondary SRNS))	pediatric	NR	NR	NR	0%	NR	SNVs	3	5/44 (11.4%) 5/64 (7.8%) SRNS; 5/214 (2.3%) NS >1 year; 5/242 (2.0%) NS any age	NP
Noteworthy: children excluded with (a) an underlying, secondary cause for NS (such as lupus nephritis, infections, or neoplasm), (b) congenital and infantile NS, or (c) steroid-sensitive nephrotic syndrome (SSNS) | criteria for variant classification NR | yield impacted by: no response to immunsuppressives												
[Giglio et al., 2015]	sporadic, nonsyndromic, and non-congenital, nephrotic syndrome SRNS n=31; SSNS n=38	69	pediatric	0%	0%	0%	0%	12/69 (17.4%)	SNVs	46	10/69 (14.5%) SRNS 10/31 (32.3%); SSNS 0/38 (0%)	NP
Noteworthy: patients who exhibited extra-renal symptoms, had a familial history, or had a congenital onset were excluded | yield impacted by: lack of response to immunosuppressive agent												
[Trautmann et al., 2015]	SRNS; congenital NS; presistent subnephrotic proteinuria of likely genetic origin SRNS in first 5 years 64%; congenital NS 6%	1174 (from cohort of 1655)	both	260/1014 (25.6%)	306/1070 (28.6%)	287/1655 (17.3%)	NR, all <20 years	422/1544 (27.3%)	SNVs	NR (1-31 mentioned; 14 genes with reported mutations)	277/1174 (23.6%) 277/1655 (16.7%) in entire cohort	NR
Noteworthy: limited information provided for type of genetic test performed and number of genes tested | criteria for variant classification NR | yield impacted by: age of onset; lower post-transplant disease recurrence												

[Sadowski et al., 2015]	SRNS	1783 families (2016 individuals)	both	NR	372/1783 (20.9%)	NR	NR, mean 3.4 years (0-63)	NR	SNVs	27	526/1783 (29.5%)	NP
Noteworthy: yield impacted by: lower age of onset & consanguinity												
[Bierzynska et al., 2017]	primary SRNS, congenital and/or familial nephrotic syndrome (presumed SRNS), secondary SRNS, or FSGS on biopsy and syndromic proteinuric nephropathy	187	pediatric	22/186 (11.8%)	13/180 (7.2%)	43/187 (22.9%	0%	69/187 (36.9%)	SNVs	53	49/187 (26.2%)	NP
Noteworthy: yield impacted by: positive family history + CKD5												
[Wang et al., 2017a]	SRNS + isolated proteinuria SRNS n=110; isolated proteinuria n=10	120	pediatric	19/109 (17.4%)	NR	NR	0%	16/120 (13.3%)	SNVs + CNVs	28	34/120 (28.3%)	0%
Noteworthy: patients were excluded if their age at onset of disease was over 18 years or if they were diagnosed as having Alport syndrome | genetic testing results for pediatric SRNS patients vary with different ethnicities | yield impacted by: age of onset; family history												
[Wang et al., 2017b]	SRNS	60	pediatric	0%	0%	0%	0%	0%	SNVs	Sanger 5; NGS 17	19/60 (31.7%)	NP
Noteworthy: secondary nephrotic syndrome and those from consanguineous families were excluded | yield impacted by: lack of response to immunosuppressive agents												
[Warejko et al., 2018]	SRNS or nephrotic range proteinuria with histology of FSGS or diffuse sclerosis	300 families (335 individuals)	both	93/300 (31.0%)	146/300 (49%)	91/335 (27.2%)	8/335 (2.4%)	5/300 (1.7%)	SNVs	33 followed by WES	74/300 (24.7%)	NP
Noteworthy: in 11 families (3.7%) a mutation in a gene that causes a phenocopy of steroid-resistant nephrotic syndrome was detected | yield impacted by: consanguinity												
[Tan et al., 2018]	consecutive cohort of SRNS or nephrotic range proteinuria with histology of FSGS or diffuse sclerosis	72 families (77 individuals)	pediatric	NR	8/72 (11.1%)	NR	NR, median 3.5 years (0.1-18.8)	NR	SNVs	24	8/72 (11.1%)	NP
Noteworthy: unknown if cases were excluded with previous diagnosis, it is unlikely that all consecutive patients were unsolved | yield impacted by: age of onset, consanguinity												
[Bezdíčka et al., 2018]	SRNS congenital NS n=11; infantile n=10; child-hood onset n=52	70 families (74 individuals)	Perinatal /pediatric	NR	1/70 (1.4%)	24/74 (32.4%)	0%	28/74 (37.8%)	SNVs	tier 1: 3; tier 2: 48	25/70 (35.7%)	NP
Noteworthy: two-tiered approach (first most frequent genes (NPHS2, WT1, NPHS1), second panel of 48 genes | yield impacted by: ESKD												
[Gribouval et al., 2018]	non-syndromic, biopsy-proven FSGS or SRNS in the absence of known FH	135	Adult	0%	NR	NR	100%	67/135 (49.6%)	SNVs	35	16/135 (11.9%)	NP

Noteworthy: 14/135 (10.4%) presented with APOL1 high-risk alleles, we excluded these from the diagnostic yield and report this number here separately | paper concludes with molecular diagnosis in 30 patients (22.2%) based on pathogenic mutations in known monogenic SRNS genes and APOL1 high-risk alleles | yield impacted by: age of onset, ESRD												
[Landini et al., 2020]	NS SRNS n=64; SSNS n=47	111 (from consecutive cohort of 252 referred with nephrotic syndrome)	NR	0%	NR	0%	5/111 (4.5%)	30/111 (27.0%)	SNVs + CNVs	298	37/111 (33.3%) 37/252 (14.7%) in entire cohort (of which 141 were SSNS)	2.7%
Noteworthy: patients excluded with known family history of nephrotic syndrome OR syndromic nephrotic syndrome / extra-renal involvement. Unknown if genetic testing was performed in patients with known family history (therefore not included in extrapolated yield) | reverse phenotyping of patients and family members let to diagnosis of phenocopies in 28% of cases | VUS includes LP/P variants that don't explain patient's phenotype (CUBN/SOX17) | yield impacted by: ESRD for monogenic podocytopathies												
[Nagano et al., 2020]	congenital/infantile NS, SRNS or FSGS	230	both	30/230 (13.0%)	NR	27/230 (11.7%)	10/230 (4.3%)	NR	SNVs + CNVs	60	69/230 (30.0%)	0%
Noteworthy: remission of proteinuria in patients with unknown causative mutations was observed in 26% of patients with various immunosuppressive or renoprotective therapies, whereas only 5% of patients with monogenic disease-causing mutations exhibited complete remission | yield impacted by: age of patients												
other/mixed gomerulopathies												
[Fallerini et al., 2014]	clinical suspicion of AS	87 families (271 individuals)	both	NR	NR	49/176 (27.8%)	NR, median 36 years (range 1-82) at inclusion	42/176 (23.8%)	SNVs	3	48/87 (55.2%)	NP
Noteworthy: inheritance: XL semidomant in 65%; AR in 4% and AD in 31%												
[Morinière et al., 2014]	hematuric nephropathy AS n=90; benign familial hematuria n=10	101	both	77/96 (80.2%)	0%	min. 47/101 (46.5%)	NR, mean 11 years (range <1-54)	26/101 (25.7%)	SNVs + CNVs	3	81/101 (80.2%)	8.6%
[Nabais Sá et al., 2015a]	AS (at least 1 diagnostic criterium)	60	Adult	NR	NR	NR	NR	NR	SNVs + CNVs	1	22/60 (36.7%)	4.5%
Noteworthy: yield impacted by: number of AS criteria met												
[Nabais Sá et al., 2015b]	AS/TBMN	40	Adult	NR	2/40 (5.0%)	NR	NR	NR	SNVs	2	25/40 (62.5%)	NP
Noteworthy: probands diagnosed with AS/TBMN (at least one diagnostic criteria of AS), either with a family history suggestive of autosomal inheritance of kidney disease (n= 5) or without detectable pathogenic mutations in COL4A5 (n= 35) | yield impacted by: number of AS criteria met												
[Gast et al., 2016]	FSGS, SRNS FSGS n=80; SRNS n=1	76 (81 individuals)	Adult	24/76 (31.5%)	NR	NR	70/81 (86.4%)	NR	SNVs	39	15/76 (19.8%)	NP
Noteworthy: patients with positive family history of renal disease were prioritized in recruitment. Only 1 patient with SRNS was included. | yield impacted by: positive family history and childhood onset												
[Bu et al., 2016]	patients screened with the Genetic Complement-	193	both	NR	NR	NR	119/193 (61.7%) = current age	NR	SNVs + CNVs	11	78/193 (40.4%)	21.8%

	Mediated Renal Disease Panel TMA: aHUS n=118; TTP n=6; other TMA n=11; aHUS/TTP n=12 | C3G: C3GN n=30; DDD n=5; C3GN/DDD n=2 | other: untargeted diseases n=9, (SLE/IgAN/ARF/Acute GN)										C3GN&DDD 16/37 (43.2%); aHUS&TTP 56/136 (41.2%); other TMA 4/11 (36.4%); untargeted 2/9 (22.2%)	
Noteworthy: yield also includes VUS, data not provided for calculating yield based on LP+P | from 91 identified variants: 68 VUS; 11 P; 12 LP, yield based on LP+P is therefore expected to be much lower than the yield presented here | yield impacted by: phenotype C3G												
[Sen et al., 2017]	patients referred for gene panel testing of SRNS/collagen-related genes SRNS n=255, SSNS n=12, hematuria/AS n=35	302	both	58/183 (31.7%)	17/141 (12.1%)	NR	62/302 (20.5%)	NR	SNVs + CNVs	37	71/302 (23.5%) SRNS 54/255 (21.2%); SSNS 0.12 (0%); AS 17/35 (48.6%)	2.8%
Noteworthy: yield impacted by: age of onset, phenotype SRNS												
[Yao et al., 2019]	FSGS	179 families (193 individuals)	Adult	43/193 (22.3%)	NR	NR	135/161 (83.9%)	72/147 (49.0%)	SNVs + CNVs	109	37/179 (20.1%)	2.7%
Noteworthy: additional category of possible pathogenic added by authors, not included in diagnostic yield reported in this table, likely pathogenic is included in table | yield impacted by: positive family history												
[Schapiro et al., 2019]	onset of both proteinuria and hematuria before age 25 years	362 families (371 individuals)	NR	NR	56/362 (15.5%)	NR	NR, all before age 25	NR	SNVs	34	51/362 (14.1%) AS 17/362 (4.7%); aHUS 5/362 (1.4%); TTP 0/362 (0%); SRNS 29/362 (8.0%)	NP
Noteworthy: 11 genes screened in all families and 23 genes in families not previously screened for monogenic forms of SRNS | yield impacted by: consanguinity												
[Yamamura et al., 2019]	suspected AS referred for genetic diagnosis	441	NR	NR	NR	NR	NR	NR	SNVs + CNVs	NGS: 45; sanger: 3	397/441 (90.0%)	5.0%
Noteworthy: 4 patients had pathogenic mutations in NPHS1, EYA1, LAMB2, AND CLCN5												
[Ozdemir et al., 2020]	patients with glomerular proteinuria and/or hematuria	320	pediatric	NR	NR	NR	0%	NR	SNVs	47	87/320 (27.2%) had COL4A mutations, rest NR	NP
Noteworthy: only yield for COL4A mutations is reported | criteria for variant classification NR												

Supplementary Table 1e – summary of individual nephrolithiasis/urolithiasis studies. Patient, cohort and test characteristics per study are presented. Diagnostic yield and percentage of diagnostic yield that is explained by CNVs are summarized. In grey font additional info on phenotypes, extrapolated cohort and diagnostic yield for both of these are presented if available. A summary of noteworthy details is presented per study, including patient characteristics that are expected to have positively impacted the diagnostic yield. Abbreviations are listed in Supplementary Table 2.

nephrolithiasis / urolithiasis	phenotype	n of population	pediatric / adult at time of study	positive family history (n, %)	consang. (n, %)	extra-renal features (n, %)	adult-onset disease (n, %)	ESKD (n, %)	type of variants tested	n genes analyzed	diagnostic yield (n, %)	percentage of yield explained by CNV (%)
[Daga et al., 2018]	nephrolithiasis / nephrocalcinosis before age 25	51 families (65 individuals)	both	29/51 (56.9%)	8/51 (15.7%)	NR	NR, all before <25 years, median age of onset with monogenic cause 3 years; median age without monogenic cause 7 years	NR	SNVs	30, followed by 117 in unsolved families	16/51 (31.4%)	NP
Noteworthy: in nine of 15 families, the genetic diagnosis may have specific implications for stone management and prevention | yield impacted by: age of onset, positive family history, consanguinity												
[Amar et al., 2019]	nephrolithiasis confirmed by abdominal ultrasound (hospitalized patients)	235 families (440 individuals: 235 proband; 115 affected family members) (from cohort of 159)	both	109/229 (47.6%)	122/229 (53.3%)	NR	155/229 (67.7%) onset >20 years	NR	SNVs	30	17/235 (7.2%) 17/259 (6.6%) in entire cohort (10 no consent; 7 evident secondary cause; 7 inadequate DNA sample)	NP
Noteworthy: 49% of probands reported a history of recurrent stones| yield impacted by: family history, age of onset												
[Ziyadov et al., 2021]	patients that underwent surgery for urinary tract stone disease	48	pediatric	28/48 (58.3%)	NR	NR	0%	NR	SNVs	30	18/48 (37.5%)	NP
Noteworthy: children with known metabolic disease were excluded | Clinical significance of 6/18 detected variants were unknown, however not reported in how many patients this was present, therefore yield includes also variants of unknown significance. However, article also reports: 18 variants with known clinical significance were detected in 16 of 46 patient; in the remaining two patients, genetic changes were detected in more than one gene; therefore unclear.												
[Zhao et al., 2021]	urolithiasis referred for genetic testing	104 families (105 individuals) (from cohort of 199 that received urolithiasis treatment)	pediatric	NR	2/105 (1.9%)	NR	0%	NR	SNVs	34	38/105 (36.2%) 38/199 (19.1%) in entire cohort	NP
Noteworthy: high conformity (100%) was obtained between the molecular diagnoses and metabolic evaluation | 94 patients refused genetic testing												

Supplementary Table 1f – summary of individual tubulopathy studies. Patient, cohort and test characteristics per study are presented. Diagnostic yield and percentage of diagnostic yield that is explained by CNVs are summarized. In grey font additional info on phenotypes, extrapolated cohort and diagnostic yield for both of these are presented if available. A summary of noteworthy details is presented per study, including patient characteristics that are expected to have positively impacted the diagnostic yield. Abbreviations are listed in Supplementary Table 2.

tubulopathies	phenotype	n of population	pediatric / adult at time of study	positive family history (n, %)	consang. (n, %)	extra-renal features (n, %)	adult-onset disease (n, %)	ESKD (n, %)	type of variants tested	n genes analyzed	diagnostic yield (n, %)	percentage of yield explained by CNV (%)
[Palazzo et al., 2017]	dRTA referred for molecular diagnosis	89	both	NR	4/89 (4.5%)	69/89 (77.5%)	8/89 (9.0%)	NR	SNVs	3	64/89 (71.9%)	NP
Noteworthy: yield impacted by: age of onset, extra-renal features												
[Ashton et al., 2018]	clinical diagnosis of tubulopathy	384 families (410 individuals)	pediatric	NR	NR	NR	0%	NR	SNVs + CNVs	37	245/384 (63.8%)	NR
Noteworthy: genetic testing changed the clinical diagnosis in 16 cases and provided insights into the phenotypic spectrum of the respective disorders | %CNV determined by ExomeDepth unclear												
[Adalat et al., 2019]	patients with chronic kidney disease stage 1-3 tested for HNF1B	199	pediatric	NR	NR	NR	0%	NR	SNVs + CNVs	1	52/199 (26.1%)	63.5%
Noteworthy: limited information provided for type of genetic test performed| yield impacted by: absence of hypomagnesemia												
[Hureaux et al., 2019]	clinical diagnosis of tubular dysfunction	1033	adult	NR	NR	NR	100%	NR	SNVs + CNVs	46	269/1033 (26.0%)	0%
Noteworthy: a total of 16 patients (2.1%) had their initial clinical diagnosis revised by the panel analysis| yield impacted by: childhood onset												
[Mori et al., 2021]	clinical diagnosis of GS	70	adult	NR	NR	NR	100%	NR	SNVs + CNVs	168	30/70 (42.9%)	0%
Noteworthy: yield impacted by: age of diagnosis												

Supplementary Table 1g – summary of individual ESKD studies. Patient, cohort and test characteristics per study are presented. Diagnostic yield and percentage of diagnostic yield that is explained by CNVs are summarized. In grey font additional info on phenotypes, extrapolated cohort and diagnostic yield for both of these are presented if available. A summary of noteworthy details is presented per study, including patient characteristics that are expected to have positively impacted the diagnostic yield. Abbreviations are listed in Supplementary Table 2.

ESKD	phenotype	n of population	pediatric / adult at time of study	positive family history (n, %)	consang. (n, %)	extra-renal features (n, %)	adult-onset disease (n, %)	ESKD (n, %)	type of variants tested	n genes analyzed	diagnostic yield (n, %)	percentage of yield explained by CNV (%)
[Snoek et al., 2018]	adult-onset ESRD	5606	adult	NR	NR	NR	NR, mean 30 years for ESRD onset (18-61)	100%	CNVs	20	26/5606 (0.5%)	100%
Noteworthy: only 12% of the patients with homozygous NPHP1 full gene deletion were clinically diagnosed as having NPH | Prevalence of homozygous NPHP1 deletions was 0.9% in recipients between 18 and 50 years old at the start of first RRT (24/2794) and even higher (2.1%) in recipients ages 18–29 years old | yield impacted by: age onset ESRD												
[Mann et al., 2019]	kidney transplant recipient <25 years old urinary stone disease n=3; renal cystic ciliopathies n=9; SRNS n=21, CAKUT n=55, chronic glomerulonephritis n=7; unknown etiology ESRD n=9	104 (from cohort of 272)	both	23/104 (22.1%)	9/104 (8.7%)	55/104 (52.9%)	0%	100%	SNVs + CNVs	396	34/104 (32.7%) 34/272 (12.5%) from kidney transplant recipients <25 year urinary stone disease 3/3 (100%), renal cystic ciliopathies 7/9 (77.8%), SRNS 9/21 (42.9%), CAKUT 10/55 (18.2%), chronic glomerulonephritis 1/7 (14.3%), unknown etiology ESRD 4/9 (44.4%)	14.7%
Noteworthy: for probands in whom clinical SNP arrays revealed a pathogenic CNV and WES evaluation for SNVs and small insertions/deletions was negative, CNV analysis on WES data was performed using CoNIFER software in order to verify the clinical findings. WES was not utilized to identify novel CNVs because of the relatively low sensitivity of this technique | yield impacted by: consanguinity + extra-renal features + family history												
[Ottlewski et al., 2019]	waitlisted for KTx with undetermined ESRD	50 (from cohort of 142; of which 57 had undetermined ESRD)	adult	NR	NR	NR	NR, median age at first ESRD/RRT 43.4 (15.4-66.5)	100%	SNVs + CNVs	209	6/50 (12.0%) in undetermined ESRD 35/142 (24.6%) - in entire cohort of waitlisted patients AS n=2; FSGS n=4 (in predetermined hereditary ADPKD n=24; COL4-NP/AS n=5)	0%
Noteworthy: part of larger adult KTx waitlisted cohort with 29/142 (20.4%) patients with predetermined genetic disease												
[Schrezenmeier et al., 2021]	waitlisted for KTx with undetermined KF <40 year or biopsy suggestive for FSGS/aHUS	126 (from cohort of 635; unknown origin in 137)	adult	NR	NR	NR	100% KF onset >18 years	100%	SNVs + CNVs	600	14/126 (11.1%) 133/635 (20.9%) in entire KTx waitlisted cohort	7.1%

	ESRD<40 n=87 (11 no consent); FSGS n=29; aHUS suspicion n=21										undetermined ESRD<40 10/86 (11.6%); FSGS 3/29 (10.3%); aHUS suspicion 1/21 (4.8%)	
Noteworthy: part of larger KTx waitlisted cohort of KF >18 years with 119/635 (18.7%) patients with predetermined genetic disease. ADPKD in 104/635 (16.4%)												
[Snoek et al., 2022]	kidney transplant recipient <50 year due to ESKD of any cause (clear-cut non-genetic disease excluded) ciliopathy n=22; glomerular disease n=42; CAKUT n=13; TSC n=4; renal carcinoma n=3; vascular disease n=8; tubular disease n=2; CKD with unknown cause n=4; other n=12	110 (from cohort of 273 transplant patient)	adult	NR	NR	NR	NR, mean 23 years (range 0-47)	100% (before age 50)	SNVs + CNVs	379	56/110 (50.9%) 56/273 (20.5%) in entire transplant cohort ciliopathy 22/22 (100%); glomerular disease 19/42 (45.2%); CAKUT 1/13 (7.7%); TSC 4/4 (100%); renal carcinoma 3/3 (100%); vascular disease 0/8 (0%); tubular disease 2/2 (100%); CKD with unknown cause 1/4 (25%); other 4/12 (33.3%)	5.4% detected prior to study, ExomeDepth yield 0%
Noteworthy: reclassification original diagnosis in 6% of cohort and in 11% of cases with genetic diagnosis | Extrapolated to the 273 patient cohort who did not all fit the inclusion criteria: diagnostic yield still 21% | Excluding cases with childhood-onset with mutation identified: min. yield 33/250 (13.2%) in adult-onset cohort.												

Supplementary Table 1h – summary of individual mixed phenotype studies. Patient, cohort and test characteristics per study are presented. Diagnostic yield and percentage of diagnostic yield that is explained by CNVs are summarized. In grey font additional info on phenotypes, extrapolated cohort and diagnostic yield for both of these are presented if available. A summary of noteworthy details is presented per study, including patient characteristics that are expected to have positively impacted the diagnostic yield. Abbreviations are listed in Supplementary Table 2.

mixed kidney disease phenotypes	phenotype	n of population	pediatric / adult at time of study	positive family history (n, %)	consang. (n, %)	extra-renal features (n, %)	adult-onset disease (n, %)	ESKD (n, %)	type of variants tested	n genes analyzed	diagnostic yield (n, %)	percentage of yield explained by CNV (%)
[Alkanderi et al., 2017]	patients attending a multidisciplinary renal genetics clinic in a tertiary center familial hematuria n=30; cystic kidney disease n=25; CAKUT n=7; tubulopathy n=7; ciliopathy n=4; TSC n=3; congenital NS n=2; early-onset hypertension n=2; of which known molecular genetic diagnosis n=18	80 probands (244 individuals) of which 18 had previous genetic diagnosis	both	NR	2/80 (2.5%)	min. 25/80 (31.3%)	NR, mean 19 years at referral; 30/80 (37.5%) <18 at clinical review	NR	NR	1 to multiple ("small panels")	25/62 (40.3%) 43/80 (53.8%) - including known molecular genetic diagnosis referred for counseling familial hematuria n=15; cystic kidney disease n=4; CAKUT n=1; tubulopathy n=2; ciliopathy n=2; TSC n=0; congenital NS n=1; early-onset hypertension n=0	NR
Noteworthy: authors reported higher yield (42%), including 1 VUS in COL4A5 which segregated with phenotype | criteria for variant classification NR												
[Mallett et al., 2017]	clinical diagnoses of inherited kidney disease - referred for diagnostic genetic sequencing AS/TBMN n=27; aHUS-C3GN n=33; ADTK n=4; ARPKD n=1; BBS n=1; CAKUT n=13; cystinosis n=1; NPHP-RD n=17; NS n=28; tubular disorders n=10	135 families (140 individuals)	both	NR	NR	NR	NR, mean 19.3 years (range 0-71.3) at time of genetic testing	NR	SNVs + CNVs	207	58/135 (43.0%) ADTKD 1/4 (25.0%); aHUS 10/33 (30.3%), ARPKD 0/1 (0%); AS 22/27 (81.5%); BBS 1/1 (100%); CAKUT 1/13 (7.7%); cystinosis 1/1 (100%); NS 9/28 (32.1%); NPHP-RD 5/17 (29.4%); tubular disorders 8/10 (80.0%)	8.6%
Noteworthy: diagnostic rate same in adults and children | yield impacted by: genepanels: AS; tubular disorders												
[Lata et al., 2018]	CKD of unknown cause or familial nephropathy of unclear cause or CKD with a clinical diagnosis compatible with a Mendelian genetic disease glomerular disease n=50; tubulointerstitial disease n=10; developmental disorders n=11; hypertension n=5; undiagnosed disease/other n=16	92 (81 from 344 patient seen at outpatient nephrology clinics + 11 from other institutions)	adult	53/92 (57.6%); in out-patient cohort 106/344 (30.8%)	NR	0%	NR, mean 42 years (SD 17 years)	20/92 (21.7%)	SNVs	287 followed by WES	22/92 (23.9%) 22/344 (6.4%) for entire cohort CKD of unknown cause 9/16 (56.3%)	NP

Noteworthy: since PKD1 is not well-captured by WES, patient fulfilling clinical diagnostic criteria for ADPKD were not included

[Bullich et al., 2018]	suspected cystic/glomerular inherited kidney disease referred for genetic testing syspected cystic n=207; suspected glomerular n=98	305	both	44%	NR	NR	155/305 (50.8%)	NR	SNVs + CNVs	140	222/305 (72.8%) Cystic 161/207 (77.8%); glomerular 61/98 (62.2%)	10.4%

Noteworthy: of genetically diagnosed patients, 15% were referred with an unspecified clinical diagnosis and in 2% genetic testing changed the clinical diagnosis. Therefore, in 17% of cases genetic analysis was crucial to establish the correct diagnosis | yield impacted by: adult-onset (likely explained by high percentage of clinical diagnoses of AS)

[Groopman et al., 2019]	Two subcohorts of CKD subcohort 1: ESRD 50-80 years; subcohort 2: genetic and research biobanking study with the aim of elucidating genetic basis of CKD - all patients with clinical diagnosis of CKD eligible congenital/cystic renal disease n=531; glomerulopathy n=1411; diabetic nephropathy n=370; hypertensive nephropathy n=319; tubulointerstitial disease n=244; other n=159; nephro-pathy of unknown origin n=281	3315 subcohort 1: 1128; subcohort 2: 2187	mostly adult	subcohort 1: NR; subcohort 2: 619/2187 (28.3%)	NR	NR	3037/3315 (91.6%) >21 years at time of study entry subcohort 1: 100% >45 years at time of study entry; subcohort 2: 87.3% >21 years at time of study entry	2144/3315 (64.7%) subcohort 1: 1128/1128 (100%); subcohort 2: 1016/2187 (46.5%)	SNVs	625 kidney disease genes + other mendelian disease associated genes	307/3315 (9.3%) subcohort 1: 140/1128 (12.4%); subcohort 2: 167/2187 (7.6%) congenital or cystic renal disease 127/531 (23.9%); glomerulopathy 101/1411 (7.2%); diabetic nephropathy 6/370 (1.6%); hypertensive nephropathy 8/319 (2.5%); tubulointerstitial disease 11/244 (4.5%); other 6/159 (3.8%); nephropathy of unknown origin 48/281 (17.1%)	NP

Noteworthy: in the majority of these patients (122 of 167 [73%]), the genetic diagnosis gave new clinical insight. In 18 patients the genetic findings reclassified the disease. In 39 patients with unknown origin of disease a molecular cause was established. In 88 the genetic diagnosis could initiate referral and evaluation for previously unrecognized extra-renal features of the associated diseases. For 84 patients the genetic diagnosis could inform therapy | Difference in yield between cohorts explained by number of ADPKD cases in subcohort 1 | yield impacted by: family history, diagnosis of congenital/cystic disease, nephropathy of unknown origin

[Connaughton et al., 2019]	enrolled adult patients with CKD presenting to nephrology (mostly familial or with extra-renal features) - selection of consecutive cohort IKGP cystic kidney / renal ciliopathies n=12; CAKUT n=45; glomerular n=7; tubulointerstitial kidney disease n=7; SRNS n=7; renal tubulopathies n=2; CKD unknown etiology n=34	114 families (138 individuals) (from IKGP cohort of 1840)	adult	78/114 (68.4%)	NR	16/114 (14.0%)	85/138 (61.6%)	90/138 (65.2%)	SNVs	478	42/114 (36.8%) min. 195/629 (31.0%) in cohort with positive family history - 206/1840 (11.2%) of consecutive IKGP cohort cystic kidney / renal ciliopathies 10/12 (83.3%); CAKUT 10/45 (22.2%); glomerular 2/7 (28.6%); tubulointerstitial kidney disease 2/7 (28.6%); SRNS 0/7 (0%); renal tubulopathies 2/2 (100%); CKD of unknown etiology 16/34 (47.1%)	NP

Noteworthy: in 9 of 42 families (22%) the molecular genetic diagnosis resulted in correction of the clinical diagnosis, whereas in 16 families with CKD of unknown origin (38%), WES established a new molecular genetic diagnosis | Patients with ADPKD, confirmed AS or confirmed mutations in MUC1/UMOD were excluded | yield impacted by: positive family history and extra-renal features

[Rao et al., 2019]	clinical suspicion of genetic kidney disease glomerulopathy n=554; CAKUT n=159; renal cystic disease n=83; renal tubular disease/renal calcinosis/nephrolithiasis n=159; CKD3-5 with unknown origin n=46	1001	pediatric	NR	3/1001 (0.3%)	NR	0%	NR	SNVs + CNVs	2703 + Mendelian disease associated genes in WES	421/1001 (42.1%)* glomerulopathy 213/510 (41.8%) - from which 117/212 (55.2%) with AS; SRNS 94/281 (33.5%); CAKUT (17.0%); renal cystic disease (61.4%); renal tubular disease/renal calcinosis/nephrolithiasis (62.3%); aHUS (43.2%) CKD3-5 with unknown origin (26.1%)	2.1%

*Noteworthy: *VUS of known disease causing genes included in diagnostic yield, through discussion combined with genotype and phenotype | 106 distinct monogenetic disorders detected, 15 accounted for 60.7% of genetic diagnoses | yield impacted by: age of onset, family history (ethnic background)*

[Thomas et al., 2020]	patients referred to renal genetics clinic to establish genetic diagnosis CAKUT n=4; ciliopathy n=15; glomerular n=12; tubular transport n=8; tubulointerstitial n=2; AS n=1	43 patients underwent genetic testing (from cohort of 88 families (111 individuals))	both	31/42 (73.8%)	NR	NR	NR, mean 39.9 years at genetic evaluation	NR	SNVs	264	26/43 (60.5%) 45/88 (51.1%) of renal genetics clinic cohort including already known genetic disease (n=19) + evaluation living donor candidates AS n=9; ADPKD n=7; FSGS n=2; PAX2-mediated CAKUT n=2; ARPKD n=1; Dent n=1; Frasier n=1; Gordon n=1; Gitelman n=1; Zellweger n=1	NP

Noteworthy: 19 patients referred with known genetic disease; 10 kidney transplant recipients (yield genetic testing 3/10 = 30%), four living kidney donors referred for APOL1 screening, 2 tested positive for two high-risk alleles.

[Benson et al., 2020]	CKD patients referred for renal biopsy - (clear-cut non-genetic disease excluded) on biopsy: IgA nephropathy n=20; glomerulonephritis n=8; arteriosclerosis n=6; TMA n=8; TBMN n=5; AS n=1; mixed findings n=2	50 (from cohort of 153 native renal biopsies)	adult	NR	NR	NR	NR, median age at biopsy was 48 years	NR	SNVs	227	2/50 (4.0%) 2/153 (1.3%) in entire cohort of renal biopsies AD COL4A4 n=2	NP

Noteworthy: patients not clinically screened for suspected heritable forms of kidney disease

[Riedhammer et al., 2020]	Tentative clinical diagnosis of hereditary kidney disease - genetically unsolved cases	174	NR	69/174 (39.7%)	10/174 (5.7%)	NR	NR, median age genetic testing: 19	NR	SNVs + CNVs	exome wide	52/174 (29.9%) AS 16/34 (47.1%); ADTKD 3/6 (50.0%); CAKUT 8/30 (26.7%);	3.8%

	AS n=34; ADTKD n=6; CAKUT; n=30; ciliopathy n=19; FSGS/SRNS n=49; VACTERL n=9; other n=27						years range (IQR:7-35)				ciliopathy 9/19 (47.4%); FSGS/SRNS 8/49 (16.3%); VACTERL 0/9 (0%); Other 8/27 (29.6%)	
Noteworthy: 19% of diagnosed cases was a phenocopy | targeted mtDNA analyzed in one patient | yield impacted by: phenotype ADTKD, AS, ciliopathy												
[Murray et al., 2020]	Suspected familial kidney disease who had undergone percutaneous native renal biopsy TIKD n=18; Glomerulonephritis n=15; FSGS/AS n=11; TMA n=17; non-specific n=14	47 families (75 patients)	adult	69/75 (92.0%)	NR	NR	NR, median age at biopsy: 36 years range (7-69)	52/75 (69.3%)	SNVs	227 or WES or 1 (MUC1)	39/75 (52.0%) TIKD 13/18 (72.2%); glomerulonephritis 4/15 (26.7%); FSGS/AS 6/11 (54.5%); TMA 10/17 (58.8%); non-specific 6/14 (42.9%)	NP
Noteworthy: genetic testing resulted in changes in understanding of disease mechanism in 21 individuals (54%) in 12 families (57%). Treatment would have been altered in at least 26% of cases (10/39) | yield impacted by: phenotype TIKD												
[Jayasinghe et al., 2021]	clinical presentation consistent with likely monogenic cause referred to renal genetics clinic AS n=43; CAKUT n=14; complement abnormality n=6; cystic n=65; nephrotic n=39; tubular disease n=18; other n=14; nephropathy of unknown origin n=5	204 (from cohort of 225 referred patients)	both	117/204 (57.4%)	11/204 (5.4%)	53/204 (26.0%)	123/204 (60.3%)	52/204 (25.5%)	SNVs + CNVs	depending on phenotype, if negative: panel of 336 kidney genes; panel of ~4000 in patients with extra-renal / syndromic	80/204 (39.2%) 81/225 (36.0%) in entire cohort of referred patients AS 24/43 (55.8%); CAKUT 3/14 (21.4%); complement abnormality 0/6 (0%); cystic 31/65 (47.7%); nephrotic 7/39 (17.9%); tubular disease 11/18 (61.1%); other 4/14 (28.6%); Unknown 0/5 (0%)	NR*
*Noteworthy: *CMA routinely performed therefore unable to quantify contribution of CNVs to diagnostic yield (as reported by authors) | Phenotypes (e.g. CAKUT) with low likelihood of monogenic cause, were only included if they had extra-renal features. Patients with a pre-existing molecularly confirmed genetic diagnosis or a phenotype and family history suggestive of typical ADPKD were excluded. | 31/80 (39%) had a change in their clinical diagnosis. WES diagnosis was considered to have contributed to management in 47/80 (59%), including negating the need for diagnostic renal biopsy in 10/80 (13%), changing surveillance in 35/ 80 (44%), and changing the treatment plan in 16/80 (20%) | yield impacted by: age of onset, positive family history*												
[Mansilla et al., 2021]	consecutive patients who had samples sent in for genetic testing; various phenotypes: CAKUT, cystic diseases, tubulointerstitial disease, transport disorders and glomerular disease tubular transport n=29; CAKUT n=13; ciliopathy/tubulo-interstitial n=32; glomerulopathy n=43; unclassified n=10	127	both	25/127 (19.7%)	NR	NR	38/127 (29.9%), NR	8/127 (6.3%)	SNVs + CNVs	177	54/127 (42.5%) tubular transport 13/29 (44.8%); CAKUT 7/13 (53.8%); ciliopathy/tubulointerstitial 17/32 (53.1%); glomerulopathy 14/43 (32.6%); unclassified 3/10 (30.0%)	18.5%

Noteworthy: 10.2% of cases genetic testing changed clinical diagnosis | n=9 were X-linked, n=22 were autosomal dominant and n=22 were autosomal recessive | yield impacted by: age of testing

[Domingo-Gallego et al., 2021]	early-onset CKD <30 years with suspected monogenic cause referred for genetic testing PKD n=208, glomerulopathies n=131, CAKUT n=82, tubulopathies n=33, susp ADTKD n=6	460	both	226/460 (49.1%)	NR	165/460 (35.9%)	88/460 (19.1%)	NR	SNVs + CNVs	316	300/460 (65.2%) cystic kidney diseases 160/208 (76.9%); glomerulopathies 80/131 (61.1%); CAKUT 31/82 (37.8%); tubulopathies 25/33 (75.8%); ADTKD 4/6 (66.7%)	10.3%

Noteworthy: among the 300 genetically diagnosed patients, the clinical diagnosis was confirmed in 77%, a specific diagnosis within a clinical diagnostic group was identified in 15%, and 7% of cases were reclassified. Therefore, in 22% of cases, genetic analysis was crucial in defining the precise etiology of CKD | 55% autosomal dominant disease, 31% autosomal recessive disease and 10% X-linked disease. The remaining 4% presented a suspected complex inheritance pattern, of whom six had a dual molecular diagnosis | yield impacted by: family history, extra-renal features

[Oh et al., 2021]	non-specific nephrogenic symptoms (structural abnormalities, urinalysis abnormalities, electrolyte imbalance, renal failure) structural abnormalities n=16 (n=13 PKD); proteinuria and/or hematuria n=21; electrolyte imbalances n=12; renal failure n=2	51	both	5/51 (9.8%)	NR	NR	NR, 11.6 years age at inclusion (range 0-46 years)	NR	SNVs + CNVs	203	20/51 (39.2%) structural abnormalities 9/16 (56.3%); proteinuria and/or hematuria 4/21 (19.0%); electrolyte imbalances 7/12 (58.3%); renal failure 0/2 (0%)	20.0%

Noteworthy: initial clinical impression and molecular diagnosis were matched in 11 patients (11/20, 55%) | yield impacted by: positive family history

[Tanudisastro et al., 2021]	patients with suspected genetic kidney disease referred for renal gene panel testing requested gene panel: cystinosis n=2; AS n=86; tubulopathies n=68; aHUS/C3 GN n=152; ADTKD n=35; CAKUT n=39; BORS n=5; ARPKD n=12; NS n=106; NPHP&ciliopathies n=66; other n=12	542 families (552 individuals)	both	NR	NR	NR	NR, median 17 years old at time of test request (IQR 30) | median pediatric age 6 years | median adult age 36	NR	SNVs + CNVs	depending on requested panel (1-69)	189/542 (34.9%) cystinosis 2/2 (100%); AS 56/86 (65.1%); tubulopathies 30/68 (44.1%); aHUS/C3 GN 28/152 (18.4%); ADTKD 6/35 (17.1%); CAKUT 5/39 (12.8%); BORS 1/5 (20.0%); ARPKD 3/12 (25.0%); NS 33/106 (31.3%); NPHP&ciliopathies 22/66 (33.3%); other 4/12 (33.3%)	CNV 5% in pediatric cases; 15% in adult cases

Noteworthy: yield impacted by: pediatric cases; phenotype cystinosis & Alport

[Amlie-Wolf et al., 2021]	patients referred by nephrologists to Genetic Testing Stewardship Program requested gene panel: C3GN n=3; FSGS n=7; hematuria n=12; hyperoxaluria/oxalosis/cystinosis	76 (from cohort of 102)	NR	NR	NR	NR	NR	NR	SNVs	depending on phenotype - no extraneous genes	28/76 (36.8%) 28/102 (27.5%) in entire cohort requested gene panel: C3GN 0/3 (0%); FSGS 3/7 (42.9%); hematuria 5/12 (41.7%);	NP

	n=5; hypertension n=10; hypophosphatemia n=6; kidney stones n=9; nephrocalcinosis n=4; PKD n=10; renal tubular acidosis n=2; aHUS n=1; other n=2; cascade testing n=15										hyperoxaluria/oxalosis/ cystinosis 3/5 (60.0%); hypertension 0/10 (0%); hypophosphatemia 0/6 (0%); kidney stones 1/9 (11.1%); nephrocalcinosis 1/4 (25.0%); PKD 5/10 (50.0%); renal tubular acidosis 0/2 (0%); aHUS 1/1 (100%); other1/2 (50.0%); cascade testing 6/15 (40.0%)	
Noteworthy: in 27/28 patients the result led to changes in medical management | yield impacted by: phenotype: hematuria, polycystic kidney disease												
[Vaisitti et al., 2021]	clinical suspicion of a monogenic condition or without a well-defined diagnosis ciliopathies n=32; glomerular disease n=21; tubular disease n=11; nephrolithiasis/ nephrocalcinosis n=2; HUS n=4; unknown origin n=60; other n=5; CAKUT n=3	138 (from cohort of 160 in which genetic testing was requested; 22 excluded after a second re-evaluation	both	67/138 (48.6%)	NR	49/138 (35.5%)	NR, pediatric mean age 3 years (0-14) | adult mean age 37 (0-80)	22/138 (15.9%)	SNVs + CNVs	max 225	78/138 (56.5%) 78/160 (48.8%) in entire cohort glomerular disease 14/21 (66.7%); ciliopathies 22/32 (68.8%); tubular diseases 4/11 (36.4%); HUS ¼ (25.0%); organ-failure of unknown origin 32/60 (53.3%)	3.8%
Noteworthy: authors reported yield including VUS in 35/138 (25.4%) – VUS associated with diseases with autosomal dominant or X-linked recessive (in males) mode of inheritance, while VUS in genes associated with diseases having AR mode of inheritance were reported only if they were in line with the clinical phenotype | yield calculated including only LP+P 43/138 (31.2%) | yield impacted by: phenotype: glomerular disease, ciliopathies												