**SUPPLEMENTARY MATERIAL**

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**Supplementary** **Tables:**

Supplementary Table 1. Kidney-disease gene panel and mean depth of coverage for targeted genes across individuals……………………..……………………………………………………………………………………3

Supplementary Table 2. Clinical and genetic data of patients with disease-causing variants identified……………………………………………………………………………..…………………………………………………11

Supplementary Table 3. Clinical and genetic data of patients in whom VUS were identified………..……………………………………………………………………………………….……….…………….………77

**Supplementary Material and Methods:**

Bioinformatics Analysis……………………………………………………………………………………….………………….81

**Supplementary Data**

Logistic regression model of predictive clinical features ……………………………………………………..…81

**Supplementary References** ..……………………………………………….………………………………………………..82

**Supplementary Table 1.** Kidney disease gene panel and mean depth of coverage for targeted genes across individuals

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Gene** | **OMIM ID** | **Description** | **Mean depth of coverage** | | |
| *ACE* | 106180 | Angiotensin i-converting enzyme | 594x | | |
| *ACTN4* | 604638 | Actinin alpha 4 | 442x | | |
| *ADCK4* | 615567 | AarF domain containing kinase 4 | 546x | | |
| *ADCY10* | 605205 | Adenylate cyclase 10 | 668x | | |
| *AGT* | 106150 | Angiotensinogen | 597x | | |
| *AGTR1* | 106165 | Angiotensin ii receptor, vascular type 1 | 539x | | |
| *AGXT* | 604285 | Alanine-glyoxylate aminotransferase | 434x | | |
| *AHI1* | 608894 | Abelson helper integration site 1 | 704x | | |
| *ALG1* | 605907 | ALG1, yeast, homolog of | 446x | | |
| *ALG8* | 608103 | ALG8, S. cerevisiae, homolog of | 628x | | |
| *ALMS1* | 606844 | ALMS1, centrosome and basal body associated protein | 639x | | |
| *ALPL* | 171760 | Alkaline phosphatase, liver | 528x | | |
| *ANKFY1* | 607927 | Ankyrin repeats- and fyve domain-containing protein 1 | 593x | | |
| *ANKS6* | 615370 | Ankyrin repeat and sterile alpha motif domain containing 6 | 652x | | |
| *ANLN* | 616027 | Anillin actin binding protein | 770x | | |
| *AP2S1* | 602242 | Adaptor-related protein complex 2, sigma-1 subunit | 595x | | |
| *APOA1* | 107680 | Apolipoprotein A-I | 457x | | |
| *APOL1* | 603743 | Apolipoprotein L1 | 608x | | |
| *APRT* | 102600 | Adenine phosphoribosyltransferase | 415x | | |
| *AQP2* | 107777 | Aquaporin 2 | 519x | | |
| *ARHGAP24* | 610586 | Rho GTPase activating protein 24 | 794x | | |
| *ARHGDIA* | 601925 | Rho GDP dissociation inhibitor alpha | 600x | | |
| *ARL6* | 608845 | ADP ribosylation factor like GTPase 6 | 740x | | |
| *ATP6V0A4* | 605239 | ATPase, H+ transporting, lysosomal, V0 subunit A, isoform 4 | 610x | | |
| *ATP6V1B1* | 192132 | ATPase, H+ transporting, lysosomal, 56/58-KD, V1 subunit B, isoform 1 | 488x | | |
| *ATXN10* | 611150 | Ataxin 10 | 721x | | |
| *AVIL* | 613397 | Advillin | 652x | | |
| *AVPR2* | 300538 | Arginine vasopressin receptor 2 | 393x | | |
| *B2M* | 109700 | Beta-2-microglobulin | 637x | | |
| *B9D1* | 614144 | B9 domain containing 1 | 465x | | |
| *B9D2* | 611951 | B9 protein domain 2 | 566x | | |
| *BBIP1* | 613605 | BBS protein complex-interacting protein 1 | 596x | | |
| *BBS1* | 209901 | Bardet-Biedl syndrome 1 | 665x | | |
| *BBS10* | 610148 | Bardet-Biedl syndrome 10 | 846x | | |
| *BBS12* | 610683 | Bardet-Biedl syndrome 12 | 901x | | |
| *BBS2* | 606151 | Bardet-Biedl syndrome 2 | 773x | | |
| *BBS4* | 600374 | Bardet-Biedl syndrome 4 | 720x | | |
| *BBS5* | 603650 | Bardet-Biedl syndrome 5 | 713x | | |
| *BBS7* | 607590 | Bardet-Biedl syndrome 7 | 809x | | |
| *BBS9* | 600374 | Bardet-Biedl syndrome 9 | 787x | | |
| *BICC1* | 614295 | BicC family RNA binding protein 1 | 652x | | |
| *BMP4* | 112262 | Bone morphogenetic protein 4 | 753x | | |
| *BMP7* | 112267 | Bone morphogenetic protein 7 | 660x | | |
| *BSND* | 606412 | BSND Gene | 619x | | |
| *C21ORF58* |  | Chromosome 21 open reading frame 58 | 374x | | |
| *CA2* | 611492 | Carbonic anhydrase ii | 735x | | |
| *CASR* | 601199 | Calcium-sensing receptor | 720x | | |
| *CC2D2A* | 612013 | Coiled-coil and C2 domain containing 2A | 780x | | |
| *CD2AP* | 604241 | CD2-associated protein | 757x | | |
| *CDC5L* | 602868 | Cell division cycle 5-like | 749x | | |
| *CDK20* | 610076 | Cyclin-dependent kinase 20 | 554x | | |
| *CEP104* | 616690 | Centrosomal protein, 104-KD | 629x | | |
| *CEP120* | 613446 | Centrosomal protein, 120-KD | 740x | | |
| *CEP164* | 614848 | Centrosomal protein 164 | 634x | | |
| *CEP290* | 610142 | Centrosomal protein 290 | 751x | | |
| *CEP41* | 610523 | Centrosomal protein 41 | 626x | | |
| *CEP83* | 615847 | Centrosomal protein 83 | 782x | | |
| *CFH* | 134370 | Complement factor H | 809x | | |
| *CFHR1* | 134371 | Complement factor h-related 1 | 457x | | |
| *CFHR5* | 608593 | Complement factor H related 5 | 728x | | |
| *CFTR* | 602421 | Cystic fibrosis transmembrane conductance regulator | 760x | | |
| *CHD1L* | 613039 | Chromodomain helicase DNA-binding protein 1-like | 739x | | |
| *CHD7* | 608892 | Chromodomain helicase DNA-binding protein 7 | 726x | | |
| *CHRM3* | 118494 | Cholinergic receptor, muscarinic, 3 | 849x | | |
| *CLCN5* | 300008 | Chloride channel 5 | 590x | | |
| *CLCNKA* | 602024 | Chloride channel, kidney, A | 463x | | |
| *CLCNKB* | 602023 | Chloride channel, kidney, B | 449x | | |
| *CLDN14* | 605608 | Claudin 14 | 683x | | |
| *CLDN16* | 603959 | Claudin 16 | 790x | | |
| *CLDN19* | 610036 | Claudin 19 | 370x | | |
| *COL4A1* | 120130 | Collagen type IV alpha 1 chain | 759x | | |
| *COL4A3* | 120070 | Collagen type IV alpha 3 chain | 752x | | |
| *COL4A4* | 120131 | Collagen type IV alpha 4 chain | 728x | | |
| *COL4A5* | 303630 | Collagen type IV alpha 5 chain | 605x | | |
| *COL4A6* | 303631 | Collagen type IV alpha 6 chain | 545x | | |
| *COQ2* | 609825 | Coenzyme Q2, polyprenyltransferase | 766x | | |
| *COQ6* | 614647 | Coenzyme Q6, monooxygenase | 772x | | |
| *CPLANE1* | 614571 | Ciliogenesis and planar polarity effector 1 | 723x | | |
| *CPT2* | 600650 | Carnitine palmitoyltransferase II | 620x | | |
| *CRB2* | 609720 | Crumbs 2, cell polarity complex component | 490x | | |
| *CSPP1* | 611654 | Centrosome and spindle pole associated protein 1 | 622x | | |
| *CTNS* | 606272 | Cystinosin | 541x | | |
| *CUBN* | 602997 | Cubilin | 734x | | |
| *CUL3* | 603136 | Cullin 3 | 767x | | |
| *CYP24A1* | 126065 | Cytochrome P450, family 24, subfamily A, polypeptide 1 | 675x | | |
| *DAAM1* | 606626 | Dishevelled-associated activator of morphogenesis 1 | 741x | | |
| *DACH1* | 603803 | Dachshund family transcription factor 1 | 606x | | |
| *DCDC2* | 605755 | Doublecortin domain-containing protein 2 | 763x | | |
| *DCHS2* | 612486 | Dachsous cadherin-related 2 | 761x | | |
| *DDX59* | 615464 | Dead box polypeptide 59 | 708x | | |
| *DGKE* | 601440 | Diacylglycerol kinase epsilon | 787x | | |
| *DLC1* | 604258 | Deleted in liver cancer 1 | 749x | | |
| *DNAJB11* | 611341 | DNAJ/HSP40 homolog, subfamily B, member 11 | 730x | | |
| *DSTYK* | 612666 | Dual serine/threonine and tyrosine protein kinase | 704x | | |
| *DYNC2H1* | 603297 | Dynein cytoplasmic 2 heavy chain 1 | 711x | | |
| *EHHADH* | 607037 | Enoyl-CoA hydratase/3-hydroxyacyl CoA dehydrogenase | 736x | | |
| *EMP2* | 602334 | Epithelial membrane protein 2 | 605x | | |
| *ETV4* | 600711 | Ets variant gene 4 | 378x | | |
| *EYA1* | 601653 | EYA transcriptional coactivator and phosphatase 1 | 838x | | |
| *FAM20A* | 611062 | Family with sequence similarity 20, member A | 578x | | |
| *FAM58A* | 300708 | Family with sequence similarity 58, member A | 373x | | |
| *FAN1* | 613534 | FANCD2- and FANCI-associated nuclease 1 | 692x | | |
| *FANCA* | 607139 | FANCA gene | 521x | | |
| *FAT1* | 600976 | Fat atypical cadherin 1 | 849x | | |
| *FAT3* | 612483 | Fat atypical cadherin 3 | 791x | | |
| *FAT4* | 612411 | Fat atypical cadherin 4 | 839x | | |
| *FGA* | 134820 | Fibrinogen, A alpha polypeptide | 720x | | |
| *FGF20* | 605558 | Fibroblast growth factor 20 | 653x | | |
| *FGF23* | 605380 | Fibroblast growth factor 23 | 590x | | |
| *FGF8* | 600483 | Fibroblast growth factor 8 | 383x | | |
| *FGFR1* | 136350 | Fibroblast growth factor receptor 1 | 620x | | |
| *FLCN* | 607273 | Folliculin | 679x | | |
| *FMN1* | 136535 | Formin 1 | 724x | | |
| *FOXP1* | 605515 | Forkhead box P1 | 690x | | |
| *FRAS1* | 607830 | FRAS1 gene | 762x | | |
| *FREM1* | 608944 | FRAS1-related extracellular matrix protein 1 | 722x | | |
| *FREM2* | 608945 | FRAS1-related extracellular matrix protein 2 | 840x | | |
| *FXYD2* | 601814 | FXYD domain-containing ion transport regulator 2 | 365x | | |
| *GANAB* | 104160 | Glucosidase, alpha, neutral AB | 585x | | |
| *GATA3* | 131320 | Gata-binding protein 3 | 660x | | |
| *GCM2* | 603716 | Glial cells missing transcription factor 2 | 651x | | |
| *GDF11* | 603936 | Growth/differentiation factor 11 | 536x | | |
| *GDNF* | 600837 | Glial cell line-derived neurotrophic factor | 608x | | |
| *GLA* | 300644 | Galactosidase alpha | 620x | | |
| *GLI3* | 165240 | Gli-kruppel family member 3 | 700x | | |
| *GLIS2* | 608539 | GLIS family zinc finger 2 | 540x | | |
| *GNA11* | 139313 | Guanine nucleotide-binding protein, alpha-11 | 354x | | |
| *GON7* | 617436 | GON7, S. cerevisiae, homolog of | 815x | | |
| *GREB1L* | 617782 | GREB1-like protein | 706x | | |
| *GRHPR* | 604296 | Glyoxylate reductase/hydroxypyruvate reductase | 549x | | |
| *GRIP1* | 604597 | Glutamate receptor-interacting protein 1 | 798x | | |
| *HGF* | 142409 | Hepatocyte growth factor | 781x | | |
| *HNF1B* | 189907 | HNF1 homeobox B | 666x | | |
| *HNF4A* | 600281 | Hepatocyte nuclear factor 4-alpha | 468x | | |
| *HOGA1* | 613597 | 4-Hydroxy-2-oxoglutarate aldolase 1 | 465x | | |
| *HOXA13* | 142959 | Homeobox A13 | 616x | | |
| *HPRT1* | 308000 | Hypoxanthine guanine phosphoribosyltransferase 1 | 432x | | |
| *HPSE2* | 613469 | Heparanase 2 | 662x | | |
| *HSD11B2* | 614232 | 11-Beta-hydroxysteroid dehydrogenase, type II | 387x | | |
| *IFT122* | 606045 | Intraflagellar transport 122 | 600x | | |
| *IFT140* | 614620 | Intraflagellar transport 140 | 592x | | |
| *IFT172* | 607386 | Intraflagellar transport 172 | 736x | | |
| *IFT27* | 615870 | Intraflagellar transport 27 | 608x | | |
| *IFT43* | 614068 | Intraflagellar transport 43 | 531x | | |
| *IFT81* | 605489 | Intraflagellar transport 81 | 581x | | |
| *INF2* | 610982 | Inverted formin, FH2 and WH2 domain containing | 470x | | |
| *INPP5E* | 613037 | Inositol polyphosphate-5-phosphatase, 72-KD | 478x | | |
| *INVS* | 243305 | Inversin | 761x | | |
| *IQCB1* | 609237 | IQ motif containing B1 | 536x | | |
| *ITGA3* | 605025 | Integrin subunit alpha 3 | 577x | | |
| *ITGA8* | 604063 | Integrin, alpha-8 | 736x | | |
| *ITGB4* | 147557 | Integrin subunit beta 4 | 567x | | |
| *ITSN1* | 602442 | Intersectin 1 | 652x | | |
| *JAG1* | 601920 | Jagged 1 | 567x | | |
| *KAL1* | 300836 | KAL1 gene | 488x | | |
| *KANK1* | 607704 | KN motif and ankyrin repeat domains 1 | 592x | | |
| *KANK2* | 614610 | KN motif and ankyrin repeat domains 2 | 512x | | |
| *KANK4* | 614612 | KN motif and ankyrin repeat domains 4 | 615x | | |
| *KCNJ1* | 600359 | Potassium channel, inwardly rectifying, subfamily J, member 1 | 862x | | |
| *KCNJ10* | 602208 | Potassium channel, inwardly rectifying, subfamily J, member 10 | 873x | | |
| *KIF14* | 611279 | Kinesin family member 14 | 669x | | |
| *KIRREL2* | 607762 | Kin of irre-like 2 | 488x | | |
| *KLHL3* | 605775 | KELCH-Like 3 | 604x | | |
| *LAGE3* | 300060 | L antigen family, member 3 | 239x | | |
| *LAMA5* | 601033 | Laminin alpha 5 | 457x | | |
| *LAMB2* | 150325 | Laminin subunit beta 2 | 642x | | |
| *LGR4* |  | G protein-coupled receptor 48; | 664x | | |
| *LIFR* | 151443 | Leukemia inhibitory factor receptor | 664x | | |
| *LMNA* | 150330 | Lamin A/C | 450x | | |
| *LMX1B* | 602575 | LIM homeobox transcription factor 1 beta | 462x | | |
| *LRIG2* | 608869 | Leucine-rich repeats- and immunoglobulin-like domains-containing protein 2 | 710x | | |
| *LRP4* | 604270 | Low density lipoprotein receptor-related protein 4 | 581x | | |
| *LRP5* | 603506 | Low density lipoprotein receptor-related protein 5 | 496x | | |
| *LYZ* | 153450 | Lysozyme | 750x | | |
| *LZTFL1* | 606568 | Leucine zipper transcription factor like 1 | 662x | | |
| *MAGI2* | 606382 | Membrane-associated guanylate kinase, ww and pdz domains-containing, 2 | 655x | | |
| *MAPKBP1* | 616786 | Mitogen-activated protein kinase-binding protein 1 | 539x | | |
| *MEFV* | 608107 | Familial mediterranean fever gene | 603x | | |
| *MEN1* | 613733 | MEN1 gene | 481x | | |
| *MET* | 164860 | MET proto-oncogene, receptor tyrosine kinase | 867x | | |
| *MKKS* | 604896 | McKusick-Kaufman syndrome | 836x | | |
| *MKS1* | 609883 | Meckel syndrome, type 1 | 743x | | |
| *MUC1* | 158340 | Mucin 1, cell surface associated | 535x | | |
| *MYH9* | 160775 | Myosin heavy chain 9 | 586x | | |
| *MYO1E* | 601479 | Myosin IE | 755x | | |
| *NEIL1* | 608844 | Nei like DNA glycosylase 1 | 591x | | |
| *NEK1* | 604588 | NIMA-related kinase 1 | 666x | | |
| *NEK8* | 609799 | NIMA-related kinase 8 | 634x | | |
| *NFIA* | 600727 | Nuclear factor I/A | 586x | | |
| *NOS1AP* | 605551 | Nitric oxide synthase 1 (neuronal) adaptor protein | 507x | | |
| *NOTCH2* | 600275 | Notch 2 | 712x | |
| *NPHP1* | 607100 | Nephrocystin 1 | 739x | | |
| *NPHP3* | 608002 | Nephrocystin 3 | 756x | | |
| *NPHP4* | 607215 | Nephrocystin 4 | 610x | | |
| *NPHS1* | 602716 | Nephrin | 579x | | |
| *NPHS2* | 604766 | Podocin | 672x | | |
| *NR3C2* | 600983 | Nuclear receptor subfamily 3, group C, member 2 | 674x | | |
| *NRIP1* | 602490 | Nuclear receptor-interacting protein 1 | 926x | | |
| *NUP107* | 607617 | Nucleoporin 107 | 741x | | |
| *NUP205* | 614352 | Nucleoporin 205 | 760x | | |
| *NUP93* | 614351 | Nucleoporin 93 | 673x | | |
| *NXF5* | 300319 | Nuclear RNA export factor 5 | 482x | | |
| *OCRL* | 300535 | OCRL gene | 504x | | |
| *OFD1* | 300170 | OFD1, centriole and centriolar satellite protein | 412x | | |
| *OSGEP* | 610107 | O-Sialoglycoprotein endopeptidase | 655x | | |
| *OXGR1* | 606922 | G protein-coupled receptor 80 | 920x | | |
| *PAX2* | 167409 | Paired box 2 | 547x | | |
| *PAX8* | 167415 | Paired box gene 8 | 497x | | |
| *PBX1* | 176310 | Pre-B-cell leukemia transcription factor 1 | 588x | | |
| *PDE6D* | 602676 | Phosphodiesterase 6D | 611x | | |
| *PDSS2* | 610564 | Decaprenyl diphosphate synthase subunit 2 | 783x | | |
| *PHEX* | 300550 | Phosphate-regulating endopeptidase homolog, x-linked | 530x | | |
| *PKD1* | 601313 | Polycystin 1 | 823x | | |
| *PKD2* | 173910 | Polycystin 2 | 759x | | |
| *PKHD1* | 606702 | Fibrocystin/polyductin | 780x | | |
| *PLCE1* | 608414 | Phospholipase C epsilon 1 | 802x | | |
| *PMM2* | 601785 | Phosphomannomutase 2 | 586x | | |
| *POC1A* | 614783 | POC1 centriolar protein A | 468x | | |
| *PODXL* | 602632 | Podocalyxin like | 620x | | |
| *PRDM15* | 617692 | PR domain-containing protein 15 | 572x | | |
| *PRKCSH* | 177060 | Protein kinase C substrate 80K-H | 535x | | |
| *PROK2* | 607002 | Prokineticin 2 | 530x | | |
| *PROKR2* | 607123 | Prokineticin receptor 2 | 722x | | |
| *PTH* | 168450 | Parathyroid hormone | 668x | | |
| *PTPRO* | 600579 | Protein tyrosine phosphatase, receptor type O | 772x | | |
| *REN* | 179820 | Renin | 645x | | |
| *RET* | 164761 | Rearranged during transfection protooncogene | 605x | | |
| *ROBO1* | 602430 | Roundabout guidance receptor 1 | 834x | | |
| *ROBO2* | 602431 | Roundabout guidance receptor 2 | 745x | | |
| *ROR2* | 602337 | Receptor tyrosine kinase-like orphan receptor 2 | 661x | | |
| *RPGRIP1L* | 610937 | RPGRIP1 like | 808x | | |
| *SALL1* | 602218 | Sal-like 1 | 547x | | |
| *SCARB2* | 602257 | Scavenger receptor class B member 2 | 817x | | |
| *SCLT1* | 611399 | Sodium channel and clathrin linker 1 | 639x | | |
| *SCNN1A* | 600228 | Sodium channel, nonvoltage-gated 1, alpha subunit | 554x | | |
| *SCNN1B* | 600760 | Sodium channel, nonvoltage-gated 1, beta subunit | 550x | | |
| *SCNN1G* | 600761 | Sodium channel, nonvoltage-gated 1, gamma subunit | 615x | | |
| *SDCCAG8* | 613524 | Serologically defined colon cancer antigen 8 | 722x | | |
| *SDHB* | 185470 | Succinate dehydrogenase complex iron sulfur subunit B | 637x | | |
| *SDHD* | 602690 | Succinate dehydrogenase complex subunit D | 537x | | |
| *SEC61A1* | 609213 | SEC61 translocon, alpha-1 subunit | 655x | | |
| *SEC63* | 608648 | SEC63 homolog, protein translocation regulator | 562x | | |
| *SEMA3A* | 603961 | Semaphorin 3A | 834x | | |
| *SETBP1* | 611060 | Set-binding protein 1 | 595x | | |
| *SGPL1* | 603729 | Sphingosine-1-phosphate lyase 1 | 735x | | |
| *SIX1* | 601205 | SIX homeobox 1 | 648x | | |
| *SIX2* | 604994 | SIX homeobox 2 | 595,x | | |
| *SIX5* | 600963 | SIX homeobox 5 | 374x | | |
| *SLC12A1* | 600839 | Solute carrier family 12 (sodium/potassium/chloride transporter), member 1 | 660x | | |
| *SLC12A3* | 600968 | Solute carrier family 12 (sodium/chloride transporter), member 3 | 500x | | |
| *SLC22A12* | 607096 | Solute carrier family 22 (urate transporter), member 12 | 503x | | |
| *SLC2A9* | 606142 | Solute carrier family 2 (facilitated glucose transporter), member 9 | 599x | | |
| *SLC34A1* | 182309 | Solute carrier family 34 (type ii sodium/phosphate cotransporter), member 1 | 517x | | |
| *SLC34A3* | 609826 | Solute carrier family 34 (sodium/phosphate cotransporter), member 3 | 400x | | |
| *SLC3A1* | 104614 | Solute carrier family 3 (cystine, dibasic, and neutral amino acid transporter), member 1 | 728x | | |
| *SLC41A1* | 610801 | Solute carrier family 41 member 1 | 601x | | |
| *SLC4A1* | 109270 | Solute carrier family 4 (anion exchanger), member 1 | 543x | | |
| *SLC4A4* | 603345 | Solute carrier family 4 (sodium bicarbonate cotransporter), member 4 | 743x | | |
| *SLC5A2* | 182381 | Solute carrier family 5 (sodium/glucose cotransporter), member 2 | 472x | | |
| *SLC7A13* | 617256 | Solute carrier family 7, member 13 | 804x | | |
| *SLC7A9* | 604144 | Solute carrier family 7 (cationic amino acid transporter, y+ system), member 9 | 556x | | |
| *SLC9A3R1* | 604990 | Solute carrier family 9, member 3, regulator 1 | 455x | | |
| *SLIT2* | 603746 | SLIT guidance ligand 2 | 684x | | |
| *SLIT3* | 603745 | SLIT guidance ligand 3 | 535x | | |
| *SMARCAL1* | 606622 | SWI/SNF-related, matrix-associated, actin-dependent regulator of chromatin, subfamily a like 1 | 696x | | |
| *SOX17* | 610928 | SRY-Box 17 | 437x | | |
| *SOX9* | 608160 | SRY-Box 9 | 363x | | |
| *SRGAP1* | 606523 | SLIT-ROBO RHO GTPase-activating protein 1 | 673x | | |
| *TBC1D1* | 609850 | TBC1 domain family, member 1 | 686x | | |
| *TBC1D32* | 615867 | TBC1 domain family member 32 | 618x | | |
| *TBX1* | 602054 | T-Box 1 | 438x | | |
| *TBX18* | 604613 | T-Box 18 | 545x | | |
| *TBX6* | 602427 | T-Box 6 | 508x | | |
| *TCTN2* | 613846 | Tectonic family member 2 | 760x | | |
| *TCTN3* | 613847 | Tectonic family, member 3 | 657x | | |
| *TFAP2A* | 107580 | Transcription factor AP-2 alpha | 680x | | |
| *TMEM107* | 616183 | Transmembrane protein 107 | 553x | | |
| *TMEM138* | 614459 | Transmembrane protein 138 | 755x | | |
| *TMEM216* | 613277 | Transmembrane protein 216 | 681x | | |
| *TMEM231* | 614949 | Transmembrane protein 231 | 642x | | |
| *TMEM237* | 614423 | Transmembrane protein 237 | 608x | | |
| *TMEM67* | 609884 | Transmembrane protein 67 | 709x | | |
| *TNS2* | 607717 | Tensin 2 | 511x | | |
| *TNXB* | 600985 | Tenascin XB | 118x | | |
| *TP53RK* | 608679 | TP53-regulating kinase | 533x | | |
| *TPRKB* | 608680 | TP53RK-binding protein | 539x | | |
| *TRAF3IP1* | 607380 | TRAF3 interacting protein 1 | 576x | | |
| *TRAP1* | 606219 | Tumor necrosis factor receptor-associated protein 1 | 611x | | |
| *TRIM32* | 602290 | Tripartite motif containing 32 | 821x | | |
| *TRPC6* | 603652 | Transient receptor potential cation channel subfamily C member 6 | 662x | | |
| *TRPS1* | 604386 | Zinc finger transcription factor TRPS1 | 842x | | |
| *TSC1* | 605284 | TSC complex subunit 1 | 773x | | |
| *TSC2* | 191092 | TSC complex subunit 2 | 622x | | |
| *TTC21B* | 612014 | Tetratricopeptide repeat domain 21B | 756x | | |
| *TTC8* | 608132 | Tetratricopeptide repeat domain 8 | 734x | | |
| *UMOD* | 191845 | Uromodulin | 677x | | |
| *UPK3A* | 611559 | Uroplakin 3A | 608x | | |
| *VDR* | 601769 | Vitamin D receptor | 607x | | |
| *VHL* | 608537 | Von Hippel-Lindau tumor suppressor | 687x | | |
| *VWA2* | 618281 | Von willebrand factor A domain-containing protein 2 | 585x | | |
| *WDPCP* | 613580 | WD repeat containing planar cell polarity effector | 704x | | |
| *WDR19* | 608151 | WD repeat domain 19 | 771x | | |
| *WDR34* | 613363 | WD repeat domain 34 | 403x | | |
| *WDR35* | 613602 | WD repeat domain 35 | 681x | | |
| *WDR60* | 615462 | WD repeat-containing protein 60 | 623x | | |
| *WDR73* | 616144 | WD repeat domain 73 | 590x | | |
| *WFS1* | 606201 | Wolframin er transmembrane glycoprotein | 577x | | |
| *WNK1* | 605232 | Protein kinase, lysine-deficient 1 | 710x | | |
| *WNK4* | 601844 | Protein kinase, lysine-deficient 4 | 551x | | |
| *WNT4* | 603490 | Wingless-type MMTV integration site family, member 4 | 507x | | |
| *WT1* | 607102 | Wilms tumor 1 | 623x | | |
| *XDH* | 607633 | Xanthine dehydrogenase | 607x | | |
| *XPNPEP3* | 613553 | X-prolyl aminopeptidase 3 | 748x | | |
| *XPO5* | 607845 | Exportin 5 | 746x | | |
| *ZMPSTE24* | 606480 | Zinc metalloproteinase STE24 | 734x | | |
| *ZNF423* | 604557 | Zinc finger protein 423 | 669x | | |
|  |  |  | |  | |

**Supplementary Table 2.** Clinical and genetic data of patients with disease-causing variants identified.

| **Patient** | **Sex** | **fam/**  **sp** | **Clinical diagnosis** | **Age at CKD onset (death)** | **Renal presentation (onset, y)** | **Age at KFRT**  **onset (y)** | **Extrarenal alterations** | **Genetic diagnosis #OMIM** | **Zygosity (Inheritance)** | **Gene** | **Disease-causing variant/s (origin)** | **Ref** | **ACMG (HGMD)** | **Utility of genetic diagnosis** |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| P1 | U | sp | UPKD | Prenatal (TOP) | RHD, echogenic kidneys | TOP | Hepatic-pancreatic dysplasia | Renal-hepatic-pancreatic dysplasia 2; #609799 | Homo (AR) | *NEK8* | c.325\_327del p.(Phe109del)(p), c.325\_327del p.(Phe109del)(m) | [1] | LP (PM2, PM3, PM4, PP5, BP4) | Identified a specific diagnosis |
| P2 | M | fam | CAKUT | U | Right RA, intermittent Mh and prot. Normal Cr (39 y) | No at 39 y | Right testicular agenesis | Multiple OMIM classifications, \*164761 | Het (AD) | *RET* | c.2656C>T p.(Arg886Trp) | [2] | LP (PM1,PM2, PP2, PP3) | Confirmed the clinical diagnosis |
| P3 | M | sp | CAKUT | 2 mo | Bilateral RC. Left kidney hydronephrosis | U at 2 y |  | Polycystic kidney disease 1; #173900 + Alport syndrome 3; #104200 | CIP | *PKD1* | c.5080del p.(His1694Metfs\*28) | Novel | P (PVS1, PM2, PP3) | Reclassified the diagnosis |
| *COL4A3* | c.345del p.(Pro116Leufs\*37) | Novel | P (PVS1, PM1, PM2) |
| P4 | M | fam | UPKD | Birth | Bilateral renal pelvic dilatation and right RC (birth), bilateral RC (10 y). CKD G2 at 10 y | No at 10 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.826C>T (p.Arg276\*)(p) | [3] | P (PVS1, PS3, PM1, PM2, PP3, PP5) | Identified a specific diagnosis |
| P5 | F | fam | CAKUT | Prenatal (TOP) | Renal tubular dysgenesis, AH | TOP | Intestinal malrotation, pulmonary hypoplasia | Renal tubular dysgenesis; #267430 | Comp het (AR) | *ACE* | c.899del p.(Phe300Serfs\*156)(m) | Novel | P (PVS1, PM2, PM3, PP4) | Confirmed the clinical diagnosis |
| c.1361del p.(Leu454Cysfs\*2)(p) | Novel | P (PVS1, PM2, PM3, PP4) |
| P6 | M | sp | NPHP-RC | 13 y | Polyuria, polydipsia (birth), RH, loss of corticomedullary differentation, RC (13 y). CKD G4 at 18 y | No at 18 y |  | Papillorenal syndrome; #120330 | Het (AD) | *PAX2* | c.76dup p.(Val26Glyfs\*28)(de novo) | [4] | P (PVS1, PM1, PM2, PP3, PP5) | Reclassified the diagnosis |
| P7 | U | fam cons | CAKUT | Prenatal (TOP) | Renal tubular dysgenesis. Potter facies | TOP |  | Renal tubular dysgenesis; #267430 | Homo (AR) | *ACE* | c.1384dup p.(Ile462Asnfs\*19)(p), c.1384dup p.(Ile462Asnfs\*19)(m) | Novel | P (PVS1, PM2, PM3, PP4) | Confirmed the clinical diagnosis |
| P8 | M | sp | NPHP-RC | U | Biopsy: Chronic tubulointerstitial nephritis. KFRT at 1 y | 1 y |  | Renal tubular dysgenesis; #267430 | Homo (AR) | *ACE* | c.2049C>A p.(Ser683Arg)(p), c.2049C>A p.(Ser683Arg)(m) | Novel | LP (PM2, PM3, PP1, PP4, BP4) | Reclassified the diagnosis |
| P9 | M | U | CAKUT | 5 y | Bilateral RD, RC in left kidney | U at 5 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) | [5] | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P10 | M | sp | UPKD | U | Bilateral RC | No at 21 y | Seminal vesicle dysplasia, absence of ejaculation | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) | [5] | P (PVS1, PM2, PP3, PP5) | Identified a specific diagnosis |
| P11 | F | fam | CAKUT | 28 y | Bilateral RC, mild renal pelvic dilatation | No at 28 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) | [5]. | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P12 | M | sp | UPKD | Birth | Polyhydramnios (prenatal). Bilateral RC, loss of corticomedullary differentiation | U at 18 mo |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) | [5] | P (PVS1, PM2, PP3, PP5) | Identified a specific diagnosis |
| P13 | M | sp | CAKUT | Prenatal | Bilateral RD, two RC | U at 1 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) (de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P14 | M | fam | CAKUT | Childhood | Left RH, NC | U | Cryptorchidism, hypogonadotropic hypogonadism | Hypogonadotropic hypogonadism 2 with or without anosmia, #147950 | Het (AD) | *FGFR1* | c.1820dup p.(Ala608Glyfs\*48) | Novel | P (PVS1, PM1, PM2) | Confirmed the clinical diagnosis |
| P15 | U | fam | CAKUT | Prenatal (TOP) | Bilateral RA | TOP |  | Renal hypodysplasia/aplasia 3, #617805 | Het (AD) | *GREB1L* | c.5074G>T p.(Asp1692Tyr) (de novo) | Novel | LP (PS2, PM2, PP3) | Confirmed the clinical diagnosis |
| P16 | M | sp | UPKD | U | Bilateral corticomedullary RC, duplex right renal system | U at 17 y | – | Alagille syndrome 1; #118450 | Het (AD) | *JAG1* | c.2914C>G p.(Pro972Ala) (de novo) | Novel | LP (PS2, PM2, PP2, BP4) | Identified a specific diagnosis |
| P17 | F | fam | CAKUT | 3 mo | Echogenic kidneys, loss of corticomedullary differentiation. Bilateral RD. Right VUR stage I. NR prot, microh. CKD G3 at 6 y | No at 6 y | Vesicovaginal reflux | Branchiootorenal syndrome 1, with or without cataracts; #113650 | Het (AD) | *EYA1* | c.929del p.(Asp310Thrfs\*56)(p) | Novel | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P18 | M | fam | CAKUT | Prenatal | Asymmetric kidneys (prenatal), solitary functioning kidney | U at 7 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(544+1\_545-1)del p.(?)(m) | Novel | P (PVS1, PM2, PP1, PP3) | Confirmed the clinical diagnosis |
| P19 | M | sp | CAKUT | < 15 y | Oligomeganephronia, polyuria, polidipsia, hyperuricemia, gout, RC (15 y). KFRT at U y | < 43 y | Diabetes mellitus (post-transplant) | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.544C>T p.(Gln182\*) | [6] | P (PVS1, PM1, PM2,PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P20 | M | sp | CAKUT | Prenatal | RD (prenatal). Enlarged, echogenic kidneys, loss of corticomedullary differentation, RC (postnatal). Normal Cr at 5 mo | No at 5 mo | Bile duct cyst | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?)(p) | [5] | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P21 | F | sp | CAKUT | Prenatal  (TOP) | Echogenic kidneys, bilateral RC. Necropsy: MCDK | TOP |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.443C>T p.(Ser148Leu) (de novo) | [7] | LP (PM1, PM2, PM5, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P22 | M | sp | CAKUT | Prenatal | Bilateral MCKD, right kidney nephrectomy (1 y) | U at 3 y | Hypospadias | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) (de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P23 | M | fam | CAKUT | Prenatal | Right renal pelvic dilatation (prenatal), enlarged, echogenic kidneys, MCDK (birth) | U at 7 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.1340-3C>G p.(?)(m) | [1] | LP (PM2, PP1, PP3, PP5) | Confirmed the clinical diagnosis |
| P24 | M | sp | UPKD | Prenatal | Bilateral RC (prenatal), enlarged, echogenic kidneys, loss of corticomedullary differentiation (birth) | No at 7 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) (de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Identified a specific diagnosis |
| P25 | F | sp | CAKUT | Prenatal | Normal-sized, echogenic kidneys, bilateral MCDK. Normal Cr at 6 y | No at 6 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.505T>C p.(Tyr169His) (de novo) | [1] | P (PS2, PM1, PM2, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P26 | M | sp | CAKUT | U | Bilateral MCDK | U at 17 y | Hyperglycemia | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) (de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P27 | F | sp | CAKUT | Childhood | Right RA, left RC. Normal Cr at 60 y | No at 60 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) (de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P28 | M | sp | CAKUT | Prenatal | Ureterohydronephrosis and duplicated right renal collecting system (prenatal), left kidney atrophy due to VUR | U at 3 y | Neuroblastoma (3 y) | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?)(de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P29 | M | fam | CAKUT | Prenatal | MCDK (left kidney) and RHD (right kidney), bilateral VUR. CKD G2 at 4 y | No at 4 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.544C>T (p.Gln182\*)(m) | [6] | P (PVS1, PM1, PM2, PP1, PP3, PP5) | Confirmed the clinical diagnosis |
| P30 | M | sp | CAKUT | U | Bilateral RH and RC. CKD G2 at 2 y | No at 2 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?)(de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P31 | F | sp | CAKUT | U | Bilateral MCDK. CKD at 5 y | No at 5 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.910A>G p.(Arg304Gly)(p) | Novel | LP (PM1, PM2, PP2, PP3) | Confirmed the clinical diagnosis |
| P32 | M | fam | CAKUT | U | Bilateral MCKD and right urethral stricture (underwent surgery). CKD G2 at 18 mo | No at 18 mo |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.495\_496del p.(Ala166Argfs\*55)(p) | Novel | P (PVS1, PM1, PM2, PP1) | Confirmed the clinical diagnosis |
| P33 | M | sp | CAKUT | Prenatal | Hydronephrosis, RD, hypomagnesemia | U at 3 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?)(de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P34 | M | fam | CAKUT | Prenatal | Left RA (prenatal). CKD G3 at 11 y | No at 11 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.494G>A (p.Arg165His)(m) | [6] | LP (PM1, PM2, PM5, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P35 | F | sp | NPHP-RC | 2y | Echogenic kidneys, loss of corticomedullary differentiation, renal microcysts | U at 7 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) (m) | [5] | P (PVS1, PM2, PP3, PP5) | Reclassified the diagnosis |
| P36 | M | fam | CAKUT | Prenatal | Echogenic kidneys and severe right renal hydronephrosis (prenatal), bilateral RD, RH and RC (left kidney) and hydronephrosis due to urethral stricture (right kidney). Severe CKD at 11 y | No at 11 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) | [5] | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P37 | M | sp | NPHP-RC | Prenatal | Echogenic kidneys, RH (prenatal). Polyuria, polydipsia, Mh, one cyst (2 y). Mild CKD at 2 y | No at 2 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.884G>C p.(Arg295Pro) | [7] | LP (PM1, PM2, PPM5, PM6, PP2, PP3, PP5) | Reclassified the diagnosis |
| P38 | F | sp | CAKUT | Prenatal | Normal-sized, echogenic kidneys, bilateral RC. Normal Cr at 1 y | No at 1 y |  | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?)(de novo) | [5] | P (PVS1, PS2, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P39 | F | sp | CAKUT | U | Bilateral RD | U at 15 y | Coloboma | Papillorenal syndrome; #120330 | Het (AD) | *PAX2* | c.76dup p.(Val26Glyfs\*28)(de novo) | Novel | P (PVS1, PS2, PM1, PM2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| P40 | F | fam | CAKUT | U | Bilateral RH. CKD at 15 y | No at 15 y |  | Papillorenal syndrome; #120330 | Het (AD) | *PAX2* | c.1242T>A p.(Tyr414\*)(m) | Novel | P (PVS1, PM2, PP1, PP3) | Confirmed the clinical diagnosis |
| P41 | M | fam | CAKUT | U | Bilateral RH, HBP (childhood). CKD G4 at 29 y | No at 29 y | Bilateral coloboma | Papillorenal syndrome; #120330 | Het (AD) | *PAX2* | c.497-1G>A p.(?) (m) | [8] | P (PVS1, PM2, PP1, PP3, PP4) | Confirmed the clinical diagnosis |
| P42 | F | sp | ADPKD | 17 y | UTI (13 y). Normal-sized kidneys, bilateral RC, RL. Normal Cr at 19 y | No at 19 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.8897\_8898del p.(Glu2966Valfs\*6) | [1] | P (PVS1, PM2, PM6, PP5) | Confirmed the clinical diagnosis |
| P43 | M | fam | ADPKD | 27 y | Renal ultrasound compatible with ADPKD, mild prot. Normal Cr at 28 y | No at 28 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.3607C>T p.(Gln1203\*)(p) | [1] | P (PVS1, PM2, PP1, PP3) | Confirmed the clinical diagnosis |
| P44 | F | fam | ADPKD | 6 y | Enlarged kidneys, HBP, bilateral RC | No at 21 y | T1DM, hypothyroidism | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2113C>T p.(Gln705\*)(p) | [9] | P (PVS1, PM2, PP1, PP3) | Confirmed the clinical diagnosis |
| P45 | F | sp | ADPKD | 12 y | Normal-sized kidneys, bilateral RC. Normal Cr at 16 y | No at 16 y | – | Polycystic kidney disease 1; #173900 | MOSAIC 35% reads | *PKD1* | c.8945=/del p.(Pro2982=/Pro2982Argfs\*12)(de novo) | [1] | P (PVS1, PS2, PM2, PP3) | Confirmed the clinical diagnosis |
| P46 | M | fam | ADPKD | U | Enlarged kidneys, bilateral RC. Normal Cr at 28 y | No at 28 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2078\_2089dup p.(Ala696\_Gln697insArgProProAla)(m) | Novel | LP (PM2, PM4, PP1, PP4) | Confirmed the clinical diagnosis |
| P47 | M | fam | ADPKD | 30 y | Enlarged kidneys, bilateral RC. CKD G3 at 32 y | No at 32 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11628\_11629insA p.(Gly3877Argfs\*84)(p) | [1] | P (PVS1, PM2,PP1, PP5) | Confirmed the clinical diagnosis |
| P48 | F | sp | ADPKD | 21 y | Enlarged kidneys, bilateral RC. Normal Cr at 21 y | No at 21 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.380T>G p.(Phe127Cys) | [1] | LP (PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P49 | F | sp | ADPKD | 22 y | Slightly enlarged kidneys, bilateral RC (22 y). Normal Cr at 33 y | No at 33 y | HC | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.1249C>T p.(Arg417\*) | [10] | P (PVS1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P50 | F | fam | ADPKD | 25 y | Bilateral RC (25 y). Normal Cr at 27 y | No at 27 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.566C>G p.(Ser189\*)(p) | [1] | P (PVS1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P51 | F | fam | ADPKD | 23 y | Bilateral RC (23 y). Normal Cr at 26 y | No at 26 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.6493C>T p.(Gln2165\*) (m) | [1] | P (PVS1, PM1, PM2, PP1, PP3) | Confirmed the clinical diagnosis |
| P52 | F | U | ADPKD | 21 y | Normal-sized kidneys, bilateral RC (21 y), HBP. Normal Cr at 33 y | No at 33 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.1202-2A>T p.(?) | [1] | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P53 | M | fam | ADPKD | 25 y | Bilateral RC. Normal Cr at 26 y | No at 26 y | – | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.2533C>T p.(Arg845\*)(p) | [11] | P (PVS1, PM2, PP1, PP3, PP5) | Confirmed the clinical diagnosis |
| P54 | M | fam | ADPKD | 9 y | Bilateral RC (9 y). Normal Cr at 14 y | No at 14 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.7204C>T p.(Arg2402\*) | [12] | P (PVS1, PM1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P55 | M | sp | ARPKD | 13 y | Slightly enlarged kidneys, bilateral RC (13 y), UTI. CKD G3 at 36 y | No at 36 y | Infertility | Polycystic kidney disease 1; #173900 | CIP | *PKD1* | c.7921C>T p.(Gln2641\*)(de novo) | [1] | P (PVS1, PS2, PM1, PM2, PP3) | Reclassified the diagnosis |
| *PKD1* | c.5132C>T p.(Thr1711Ile)(p) | [1] | VUS (PP3, PP5, BS2) |
| P56 | F | sp | ADPKD | 12 y | Normal-sized kidneys, bilateral RC, MAU. Normal Cr at 13 y | No at 13 y | – | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.717C>A p.(Tyr239\*)(de novo) | [1] | P (PVS1, PS2, PM, PP5) | Confirmed the clinical diagnosis |
| P57 | F | fam | ADPKD | 10 y | Bilateral RC, MAU (10 y). Normal Cr at 29 y | No at 29 y | – | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.2419C>T p.(Arg807\*)(p) | [13] | P (PVS1, PM2, PP1, PP3, PP5) | Confirmed the clinical diagnosis |
| P58 | F | sp | ADPKD | 21 y | Bilateral RC (21 y), HBP (35 y). CKD G5 at 47 y | No at 47 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.5014\_5015del p.(Arg1672Glyfs\*98) | [14] | P (PVS1, PM2, PM6, PP5) | Confirmed the clinical diagnosis |
| P59 | F | fam | ADPKD | Childhood | Enlarged kidneys, bilateral RC. Normal Cr at 34 y | No at 34 y | HC, infertility | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.1669dup p.(Leu557Profs\*30)(m) | [1] | P (PVS1, PM2, PP1) | Confirmed the clinical diagnosis |
| P60 | M | fam | ADPKD | 15 y | Enlarged kidneys, bilateral RC (15 y), RL, macroh (24 y) HBP (28 y), hyperuricemia. CKD G3 at 38 y | No at 38 y | Hearing loss | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11725\_11740del p.(Leu3909Serfs\*31)(m) | [1] | P (PVS1, PM2, PP1) | Confirmed the clinical diagnosis |
| P61 | F | fam | ADPKD | U | Normal-sized kidneys, bilateral RC. CKD G3 at 33 y | No at 33 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.1543G>T p.(Gly515Trp) | [13] | LP (PM1, PM2, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P62 | M | fam | ADPKD | < 30 y | Normal-sized kidneys, bilateral RC, HBP. Normal Cr at 35 y | No at 35 y | HC | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.1445del p.(Phe482Serfs\*32)(m) | [15] | P (PVS1, PM2, PP1, PP5) | Confirmed the clinical diagnosis |
| P63 | F | sp | ADPKD | 30 y | Enlarged kidneys, bilateral RC, UTI. CKD G2 at 33 y | No at 35 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.1810C>T p.(Gln604\*) | [1] | P (PVS1, PM2, PM6, PP3) | Confirmed the clinical diagnosis |
| P64 | M | sp | ADPKD | 12 y | Bilateral RC, VUR, UTI (12 y). Normal Cr at 32 y | No at 32 y | Infertility (32 y) | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2932C>T p.(Gln978\*) | [1] | P (PVS1, PM1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P65 | M | fam | ADPKD | 29 y | Enlarged kidneys, bilateral RC, HBP. CKD G4 at 36 y | No at 36 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.8311G>A p.(Glu2771Lys) (m) | [16] | LP (PM1, PM2, PP1, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P66 | F | sp | ADPKD | 10 y | Bilateral RC and VUR (10 y), enlarged kidneys (27 y). Normal Cr at 27 y | No at 27 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.12061C>T p.(Arg4021\*) | [16] | P (PVS1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P67 | F | fam | ADPKD | 20 y | Bilateral RC, UTI (20 y). Normal Cr at 27 y | No at 27 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.6124G>C p.(Ala2042Pro)(p) | [1] | LP (PM2, PP1, PP3, PP4, PP5, BP1) | Confirmed the clinical diagnosis |
| P68 | F | sp | ADPKD | 21 y | Enlarged kidneys, bilateral RC. Normal Cr at 35 y | No at 35 y | HC (34 y) | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.380T>G p.(Phe127Cys) | [1] | LP (PM1, PM2, PM6, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P69 | F | sp | ADPKD | 12 y | Normal-sized kidneys, bilateral RC, prot. Normal Cr at 12 y | No at 12 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.3022\_3039del p.(Asn1008\_Val1013del) | [9] | LP (PM1, PM2, PM4, PM6, PP5, BP4) | Confirmed the clinical diagnosis |
| P70 | M | fam | ADPKD | 20 y | Bilateral RC (20 y), HBP (27 y), slightly enlarged kidneys (32 y). Normal Cr at 35 y | No at 35 y | HC (27 y) | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.2533C>T p.(Arg845\*)(m) | [11] | P (PVS1, PM2, PP1, PP3, PP5) | Confirmed the clinical diagnosis |
| P71 | F | sp | ADPKD | Birth | RC (birth), enlarged kidneys, echogenic kidneys, loss of corticomedullary differentiation, bilateral RC (31 y). Normal Cr at 31 y | No at 31 y | – | Polycystic kidney disease 1; #173900 | CIP | *PKD1* | c.11416T>G p.(Trp3806Gly) | [1] | LP (PM2, PM6, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| *PKD1* | c.8998C>T p.(Arg3000Cys) | [1] | VUS (PM1, PM2, PM6, BP1, BP4) |
| P72 | F | fam | ADPKD | 19 y | Bilateral RC (19 y), HBP. Normal Cr at 24 y | No at 24 y |  | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.916C>T p.(Arg306\*)(p) | [15] | P (PVS1, PM2, PP1, PP3, PP5) | Confirmed the clinical diagnosis |
| P73 | F | sp | ADPKD | 25 y | Bilateral RC (25 y). Normal Cr at 27 y | No at 27 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11341\_11343del p.(Tyr3781del)  (de novo) | Novel | LP (PS2, PM2, PM4, BP4) | Confirmed the clinical diagnosis |
| P74 | F | sp | ADPKD | < 30 y | Bilateral RC, HBP. Normal Cr at 32 y | No at 32 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.10694T>G p.(Leu3565Arg) | Novel | LP (PM2, PM6, PP3, PP4) | Confirmed the clinical diagnosis |
| P75 | F | fam | ADPKD | 24 y | Bilateral RC (24 y). HBP (29 y), left renal colics with hematuria (35 y). CKD G3 at 35 y | No at 35 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11863C>T p.(Gln3955\*) | [16] | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P76 | M | fam | ADPKD | 23 y | Bilateral RC (23 y). Normal Cr at 25 y | No at 25 y |  | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.1445del p.(Phe482Serfs\*32) | Novel | P (PVS1, PM2, PP5) | Confirmed the clinical diagnosis |
| P77 | M | fam | ADPKD | 19 y | Bilateral RC (19 y). Normal Cr at 31 | No at 31 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11249\_11263del p.(Arg3750\_Leu3754del) | [17] | LP (PM1, PM2, PM4, PP5, BP4) | Confirmed the clinical diagnosis |
| P78 | F | sp | ADPKD | 6 y | Normal-sized kidneys, bilateral RC. Normal Cr at 16 y | No at 16 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.7816C>T p.(Gln2606\*) | Mayo db | P (PVS1, PM1, PM2, PM6, PP3) | Confirmed the clinical diagnosis |
| P79 | F | sp | ADPKD | 18 y | Enlarged kidneys, bilateral RC, Pyelonephritis (18 y). CKD G4 at 38 y | No at 38 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.5014\_5015del p.(Arg1672Glyfs\*98) | [14] | P (PVS1, PM2, PM6, PP5) | Confirmed the clinical diagnosis |
| P80 | F | fam | ADPKD | Prenatal | Early-onset ADPKD. HBP | U at 8 y |  | Polycystic kidney disease 1; #173900 | CIP | *PKD1* | c.1136dup p.(Asn379Lysfs\*140)(m) | Novel | P (PVS1, PM2, PP1) | Confirmed the clinical diagnosis |
| *PKD1* | c.4826T>C p.(Ile1609Thr)(p) | Novel | VUS (PM2, PP3, BP1) |
| P81 | F | U | ADPKD | Prenatal | Early-onset ADPKD, bilateral RC (prenatal) | U at 2 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.4550dup p.(Tyr1517\*) | Novel | P (PVS1, PM1, PM2, PP3) | Confirmed the clinical diagnosis |
| P82 | M | sp | ADPKD | 21 y | Bilateral RC, RL (21 y). Pyelonephritis (18 y), HBP, CKD G2 at 41 y | No at 41 y | HC, aneurysmal dilatation of the anterior choroidal artery | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.12232del p.(Glu4078Serfs\*120) | Novel | P (PVS1, PM2, PM6, PP3) | Confirmed the clinical diagnosis |
| P83 | F | fam | ADPKD | 19 y | Bilateral RC (19 y, UTI). Normal Cr at 20 y | No at 20 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.7301\_7312dup p.(Arg2434\_Val2437dup)(m) | Novel | LP (PM1, PM2, PM4, PP1) | Confirmed the clinical diagnosis |
| P84 | F | fam | ADPKD | < 30 y | Renal ultrasound compatible with ADPKD. Normal Cr at U y | No at U y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.9486dup p.(Asn3163Glnfs\*16) | Novel | P (PVS1, PM1, PM2) | Confirmed the clinical diagnosis |
| P85 | F | fam | ADPKD | 29 y | Enlarged kidneys, bilateral RC. Normal Cr at 29 y | No at 29 y | Hepatomegaly, HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11258G>A p.(Arg3753Gln) | [16] | LP (PM1, PM2, PM5, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P86 | F | fam | ADPKD | 2 y | Renal ultrasound compatible with ADPKD, prot | No at 2 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.9202-16G>A p.(?)(p) | [9] | LP (PS3, PM2, PP1, PP5, BP4) | Confirmed the clinical diagnosis |
| P87 | F | sp | ADPKD | 15 y | Enlarged kidneys, bilateral RC. Normal Cr at 28 y | No at 29 y | Papillary thyroid carcinoma | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.896\_897del p.(Pro299Argfs\*71) | Novel | P (PVS1, PM1, PM2, PM6) | Confirmed the clinical diagnosis |
| P88 | M | fam | ADPKD | 14 y | Renal ultrasound compatible with ADPKD, HBP. Normal Cr at 27 y | No at 27 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2494dup p.(Arg832Profs\*40) (p) | [18] | P (PVS1, PM2, PP1, PP5) | Confirmed the clinical diagnosis |
| P89 | F | sp | ADPKD | 15 y | Bilateral RC. Normal Cr at 20 y | No at 20 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2542C>T p.(Gln848\*) | [19] | P (PVS1, PM2, PM6, PP6, PP5) | Confirmed the clinical diagnosis |
| P90 | F | fam | ADPKD | 29 y | Renal ultrasound compatible with ADPKD | No at 38 y | HC, iron-deficiency anemia | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.2508C>A p.(Tyr836\*) | [20] | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P91 | F | fam | ADPKD | 18 y | Normal-sized kidneys, bilateral RC (18 y), HBP (23 y). Normal Cr at 38 y | No at 38 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.4485dup p.(Ala1496Argfs\*27) | Novel | P (PVS1, PM1, PM2) | Confirmed the clinical diagnosis |
| P92 | M | fam | ADPKD | 23 y | Renal ultrasound compatible with ADPKD, HBP (24 y). CKD G2 at 37 y | No at 37 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.1831C>T p.(Arg611Trp) | [21] | LP (PM2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| P93 | F | fam | ADPKD | 3 y | Renal ultrasound compatible with ADPKD. Normal at 23 y | No at 23 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.12682C>T p.(Arg4228\*) | [17] | P (PVS1, PM1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P94 | M | U | ADPKD | 29 y | Renal ultrasound compatible with ADPKD, HBP. CKD G3 at 35 y | No at 35 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.9568+1G>C p(?) | Novel | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P95 | M | fam | UPKD | 6 y | Echogenic kidneys, right RD, left RC | No at 6 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.12373C>T p.(Gln4125\*)(p) | Mayo db | P (PVS1, PM2, PP1, PP3, PP5) | Identified a specific diagnosis |
| P96 | M | fam | ADPKD | 29 y | Enlarged kidneys, bilateral RC. Normal Cr at 30 y | No at 30 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.(287+1\_288-1)\_(12003+1\_12004-1)del p.(?) | Novel | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P97 | F | fam | UPKD | 2 y | Right renal cyst (2 y), oliguria | No at 2 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.10745dup p.(Val3584Argfs\*43) | [20] | P (PVS1, PM2, PP5) | Identified a specific diagnosis |
| P98 | M | fam | ADPKD | 13 y | Bilateral RC (13 y). CKD G4at 33 y | No at 33 y | Infertility, fatty liver disease | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.3295+1G>T p.(?) | Novel | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P99 | M | sp | ADPKD | 29 y | Bilateral RC, HBP | No at 29 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD2* | c.860T>C p.(Leu287Ser) (de novo) | Novel | LP (PS2, PM2, PP3) | Confirmed the clinical diagnosis |
| P100 | F | fam | ADPKD | 25 y | Renal ultrasound compatible with ADPKD | U at 25 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11343C>G p.(Tyr3781\*) | [22] | P (PVS1, PM2, PP5, BP4) | Confirmed the clinical diagnosis |
| P101 | F | fam | ADPKD | < 21 y | Enlarged kidneys, bilateral RC, RL. CKD G3 at 23 y | No at 23 y |  | Polycystic kidney disease 1; #173900 + Cystinuria; #220100 | CIP | *PKD1* | c.11426dup p.(Ser3810Leufs\*6)(m) | Novel | P (PVS1, PM2, PP1) | Reclassified the diagnosis |
| *PKD1* | c.127C>G p.(Pro43Ala)(p) | Novel | LB (BS2, BP1, BP4) |
| *SLC3A1* | c.647C>T p.(Thr216Met) (p), c.647C>T p.(Thr216Met)  (m) | [23] | LP (PM2, PM3, PP2, PP3, PP5) |
| P102 | F | fam | ADPKD | 14 y | Renal ultrasound compatible with ADPKD | No at 14 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.6701T>A p.(Val2234Glu) | Novel | LP (PM1, PM2, PP3, PP4) | Confirmed the clinical diagnosis |
| P103 | M | fam | ADPKD | 2 y | Enlarged kidneys, right RC. Normal Cr at 2 y | No at 2 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.9841del p.(Ala3281Profs\*35)(p) | Novel | P (PVS1, PM2, PP1) | Confirmed the clinical diagnosis |
| P104 | F | fam | ADPKD | 26 y | Enlarged kidneys, bilateral RC, HBP (26 y). CKD G1 at 43 y | No at 43 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.7984C>T p.(Gln2662\*) | Mayo db | P (PVS1, PM1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P105 | F | fam | ADPKD | 26 y | Slightly enlarged kidney, bilateral RC, HBP (26 y). Normal Cr at 46 y | No at 46 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2180T>C p.(Leu727Pro) | [16] | LP (PM2, PM5, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| P106 | M | fam | ADPKD | Prenatal | Bilateral RC. CKD G2 at 2 y | No at 2 y |  | Renal cysts and diabetes syndrome; #137920 + Polycystic kidney disease 1; #173900 | CIP | *PKD1* | c.1265T>C p.(Leu422Pro) | Novel | LP (PM1, PM2, PP3, PP4, BP1) | Reclassified the diagnosis |
| *HNF1B* | c.703C>T p.(Arg234Trp) | Novel | LP (PM1, PM2, PM5, PP2, PP3, PP5) |
| P107 | F | fam | ADPKD | 12 y | Renal ultrasound compatible with ADPKD (12 y). Normal Cr 30 y | No at 30 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.8311G>A p.(Glu2771Lys)  (m) | [9] | LP (PM1, PM2, PP1, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P108 | F | fam | ADPKD | 15 y | Bilateral RC (15 y), HBP (26 y). Normal Cr at 37 y | No at 37 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.1154T>C p.(Leu385Pro)(p) | Novel | LP (PM2, PP1, PP3, PP4) | Confirmed the clinical diagnosis |
| P109 | M | sp | ADPKD | 6 y | Normal-sized kidneys, bilateral RC, hematuria, obstruction of the proximal ureter by kidney stone. Normal Cr at 6 y | No at 6 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.12682C>T p.(Arg4228\*)(de novo) | [17] | P (PVS1, PS2, PM1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P110 | F | fam | ADPKD | 16 y | Enlarged kidneys, bilateral RC, left pyelonephritis (16 y). CKD G1 at 29 y | No at 29 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2659del p.(Trp887Glyfs\*11) | Novel | P (PVS1, PM2, PP5) | Confirmed the clinical diagnosis |
| P111 | F | sp | ADPKD | 29 y | Enlarged kidneys, bilateral RC, UTI. Normal Cr at 33 y | No at 33 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.10946C>G p.(Pro3649Arg) | Novel | LP (PM2, PM5, PM6, PP3, BP1) | Confirmed the clinical diagnosis |
| P112 | F | fam | ADPKD | 21 y | Enlarged kidneys, bilateral RC, HBP (21 y). Normal Cr at 32 y | No at 32 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2534T>C p.(Leu845Ser) | [24] | LP (PS1, PM2, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P113 | F | fam | ADPKD | 24 y | Renal ultrasound compatible with ADPKD. Normal Cr at 34 y | No at 34 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.856\_862del p.(Gly287\*) | [20] | P (PVS1, PM1, PM2, PP5) | Confirmed the clinical diagnosis |
| P114 | M | fam | UPKD | Prenatal | Renal ultrasound compatible with PKD (prenatal), increased diuresis (12 days). Normal Cr at 8 mo | No at 8 mo |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.7249dup p.(Leu2417Profs\*2) | Novel | P (PVS1, PM1, PM2) | Identified a specific diagnosis |
| P115 | F | fam cons | ADPKD | U | Normal-sized kidneys, bilateral RC. Normal Cr at 17 y | No at 17 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11426dup p.(Ser3810Leufs\*6) | Novel | LP (PVS1, PM2) | Confirmed the clinical diagnosis |
| P116 | M | fam | ADPKD | Childhood | Enlarged kidneys, bilateral RC. Normal Cr at 30 y | No at 30 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.112del p.(Pro41Glnfs\*32) | Novel | LP (PVS1, PM2) | Confirmed the clinical diagnosis |
| P117 | M | fam | ADPKD | 12 y | Bilateral RC (12 y), HBP (25 y). Normal Cr at 34 y | No at 34 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.3067C>T p.(Gln1023\*) | Novel | P (PVS1, PM1, PM2) | Confirmed the clinical diagnosis |
| P118 | F | fam | ADPKD | Childhood | Normal-sized kidneys, bilateral RC. Normal Cr at 17 y | No at 17 y | Splenomegaly | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.1831C>T p.(Arg611Trp) | [21] | LP (PM2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| P119 | M | fam | ADPKD | 21 y | Enlarged left kidney, bilateral RC, NC. Normal Cr at 23 y | No at 23 y | Slightly echogenic liver | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.10822-1G>C p.(?) | Novel | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P120 | M | fam | ADPKD | 25 y | Renal ultrasound compatible with ADPKD, HBP. | No at 31 y | HC, GGT | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.2119-2A>G p.(?) | Novel | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P121 | F | fam | ADPKD | 15 y | Renal ultrasound compatible with ADPKD. Normal Cr at 36 y | No at 36 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.12691C>T p.(Glu4231\*) | [20] | P (PVS1, PM1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P122 | F | fam | ADPKD | 12 y | Bilateral RC. Normal Cr at 18 y | No at 18 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.1700C>G p.(Ser567\*) | Novel | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P123 | M | sp | ADPKD | 14 y | Bilateral RC | No at 15 y | HC | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.11457C>G p.(Tyr3819\*)(de novo) | Novel | P (PVS1, PS2, PM2, PP3) | Confirmed the clinical diagnosis |
| P124 | M | fam | ARPKD | Prenatal (TOP) | Enlarged kidneys, microcysts in collecting ducts, Potter facies, OH. | TOP | DPM, mild bilateral ventriculomegaly, ventricular septum defect | CPT II deficiency; #608836 | Comp het (AR) | *CPT2* | c.251G>A p.(Cys84Tyr)(p) | Novel | LP (PM2, PM3, PP3, PP4) | Reclassified the diagnosis |
| c.1459G>T p.(Glu487\*)(m) | Novel | P (PVS1, PM2, PP3, PP4) |
| P125 | U | sp | UPKD | Prenatal (TOP) | Enlarged, echogenic kidneys, OH. | TOP |  | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.1305del p.(Thr436Profs\*26)(p) | [1] | P (PVS1, PM1, PM2, PM3, PP5) | Identified a specific diagnosis |
| c.1529del p.(Gly510Alafs\*25)(m) | [25] | P (PVS1, PM2, PM3, PP3, PP5) |
| P126 | U | sp | UPKD | Prenatal (TOP) | Enlarged, echogenic kidneys, OH. | TOP |  | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.5903T>G p.(Ile1968Ser)(p) | [1] | LP (PM1, PM2, PM3, PP2, PP3) | Identified a specific diagnosis |
| c.9689del p.(Asp3230Valfs\*34)(m) | [25] | P (PVS1, PM2, PP3, PP5) |
| P127 | F | fam cons | ARPKD | Birth | Renal ultrasound compatible with ARPKD. Normal Cr at 2 y | No at 2 y | Hepatic alterations compatible with ARPKD | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Homo (AR) | *PKHD1* | c.8893T>C p.(Cys2965Arg) (p), c.8893T>C p.(Cys2965Arg) (m) | [26] | LP (PM2, PM3, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P128 | F | sp | UPKD | Prenatal (TOP) | Enlarged kidneys, microcysts, OH. | TOP | CHF | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.1529del p.(Gly510Alafs\*25)(p) | ARPKD db/ | P (PVS1, PM2, PM3, PP3, PP5) | Identified a specific diagnosis |
| c.842G>A p.(Gly281Glu)(m) | [25] | LP (PM1, PM2, PM3, PP2, PP3, PP5) |
| P129 | F | sp | ARPKD | Birth | AH, enlarged, echogenic kidneys, loss of corticomedullary differentiation, RC. Normal Cr at 10 mo | No at 10 mo | Echogenic liver, HC | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.353del p.(Ser118Ilefs\*5) (p) | [27] | P (PVS1, PM2, PM3, PP5) | Confirmed the clinical diagnosis |
| c.8012G>C p.(Arg2671Pro)(m) | [1] | LP (PM2, PM3, PP2, PP5, BP4) |
| P130 | F | sp | ARPKD | Birth | Enlarged, echogenic kidneys, RC. Normal Cr at 5 mo | No at 5 mo |  | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.664A>G p.(Ile222Val) | [28] | LP (PM1, PM5, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| c.8642+1G>A p.(?) | [29] | P (PVS1, PM2, PP4, PP5) |
| P131 | M | fam cons | ARPKD | Prenatal | Enlarged, echogenic kidneys, RC, HBP. CKD G3 at 2 y | No at 5 y | Hepatomegaly, hepatic fibrosis, CD (3 y) | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Homo (AR) | *PKHD1* | c.8312 T>C p.(Val2771Ala) (p), c.8312 T>C p.(Val2771Ala) (m) | [30] | LP (PM1, PM2, PM3, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P132 | U | sp | UPKD | Prenatal (TOP) | RC, tubular epithelium detachment, OH. | TOP | Portal tract fibrosis, CD | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.5334dup p.(Asn1779\*)(p) | [1] | P (PVS1, PM2, PM3, PP5) | Identified a specific diagnosis |
| c.9689del p.(Asp3230Valfs\*34)(m) | [25] | P (PVS1, PM2, PM3, PP3, PP5) |
| P133 | M | sp | ARPKD | 2 y | Enlarged, echogenic kidneys. Normal Cr at 5 y | No at 5 y |  | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.107C>T p.(Thr36Met)(p) | [28] | LP (PM3, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| c.7592T>C p.(Leu2531Pro) (m) | Novel | LP (PM2, PM3, PP2, PP3) |
| P134 | M | sp | ARPKD | Birth | Anuria, enlarged kidneys, microcysts, loss of corticomedullary differentiation | U at birth | Echogenic liver | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.370C>T p.(Arg124\*)(m) | [31] | P (PVS1, PM1, PM2, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| c.6383del p.(Leu2128\*)(p) | [25] | P (PVS1, PM2, PM3, PP5) |
| P135 | M | fam | ARPKD | 23 y | Bilateral RC and microcysts, loss of corticomedullary differentiation. CKD G2 at 23 y | No at 23 y | Echogenic liver due to fatty liver disease | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.842G>A p.(Gly281Glu)(p) | [25] | LP (PM1, PM2, PM3, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| c.6992T>A, p.(Ile2331Lys)(m) | [28] | LP (PM3, PP2, PP3, PP5) |
| P136 | F | sp | ARPKD | 7 mo | Enlarged kidneys, hyperuricemia, HBP. Normal Cr at 2 y. | No at 2 y | CFH | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.2216C>T p.(Pro739Leu)(p) | [25] | LP (PM2, PM3, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| c.9689del p.(Asp3230Valfs\*34)(m) | [25] | P (PVS1, PM2, PM3, PP3, PP5) |
| P137 | F | sp | ARPKD | Prenatal (birth) | Enlarged kidneys, microcysts, Potter faces, AH. | Exitus at birth | CHF | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.2279G>A p.(Arg760His)(m) | [32] | LP (PM1, PM2, PM3, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| c.9689del p.(Asp3230Valfs\*34)(p) | [25] | P (PVS1, PM2, PM3, PP3, PP5) |
| P138 | M | sp | ARPKD | Prenatal | Enlarged kidneys. Bilateral pyelectasis, HBP, NC. Normal Cr at 6 y | No at 6 y | Hepatomegaly, splenomegaly | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.2980C>T p.(Arg994Trp)(m) | Novel | LP (PM1, PM2, PM3, PP2, PP3) | Confirmed the clinical diagnosis |
| c.9689del, p.(Asp3230Valfs\*34)(p) | [25] | P (PVS1, PM2, PM3, PP3, PP5) |
| P139 | M | sp | UPKD | Prenatal (TOP) | Enlarged kidneys, RD, defects compatible with renal tubular dysgenesis, Potter facies, OH | TOP |  | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Homo (AR) | *PKHD1* | c.5895dup p.(Leu1966Thrfs\*4)(p), c.5895dup p.(Leu1966Thrfs\*4)(m) | [28] | P (PVS1, PM1, PM3, PP5) | Identified a specific diagnosis |
| P140 | F | fam | ARPKD | Prenatal | Enlarged kidneys, microcysts | U | Cardiomegaly | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.790G>A p.(Val264Met)(de novo) | Novel | LP (PM1, PM2, PM3, PP2, PP3) | Confirmed the clinical diagnosis |
| c.9386del p.(Gly3129Alafs\*33)(m) | Novel | P (PVS1, PM2, PM3, PP3) |
| P141 | M | sp | ARPKD | 11 y | Renal ultrasound compatible with ARPKD. Normal Cr at 12 y | No at 12 y | Thrombocytopenia, splenomegaly, small echogenic pancreas | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.9107T>G p.(Val3036Gly) | [33] | LP (PM2, PP2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| c.9348del p.(Leu3117Phefs\*45) | Novel | P (PVS1, PM2, PP4) |
| P142 | U | sp | ARPKD | Prenatal (TOP) | Prenatal renal ultrasound and autopsy compatible with ARPKD | TOP |  | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.9689del p.(Asp3230Valfs\*34) | [25] | P (PVS1, PM2, PP3,PP4, PP5) | Confirmed the clinical diagnosis |
| c.10036T>C p.(Cys3346Arg) | [25] | LP (PS1, PP2, PP3, PP4, PP5) |
| P143 | F | sp | ARPKD | Prenatal (TOP) | Enlarged kidneys, probable bilateral RD, OH (prenatal). Autopsy: Enlarged kidneys, microcysts in collecting ducts | TOP | Bile duct malformation, PH | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Comp het (AR) | *PKHD1* | c.8407T>C p.(Cys2803Arg) (p) | [29] | P (PS1, PM1, PM2, PM3, PM5, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| c.9689del p.(Asp3230fs)(m) | [25] | P (PVS1, PM2, PM3, PP3, PP5) |
| P144 | M | sp | NPHP-RC | Prenatal (TOP) | PH, enlarged kidneys, RC | TOP | Polydactyly | Bardet-Biedl syndrome 8; #615985 | Comp het (AR) | *TTC8* | c.69del p.(Cys23Trpfs\*31)(m) | Novel | LP (PM2, PM3, PP3, PP4) | Confirmed the clinical diagnosis |
| c.(768+1\_769-1)\_(879+1\_880-1)del p.(?) (p) | Novel | P (PVS1, PM3) |
| P145 | M | sp | NPHP-RC | Childhood | Clinical features compatible with Bardet-Biedl syndrome. Enlarged kidneys. Normal Cr at 47 y | No at 47 y | Retinitis pigmentosa, low vision, hypogonadism, polydactyly, mental retardation borderline, obesity, fatty liver disease, cardiac hypertrophy | Bardet-Biedl syndrome 1; #209900 | Homo (AR) | *BBS1* | c.1169T>G p.(Met390Arg) (p), c.1169T>G p.(Met390Arg)  (m) | [34] | LP (PM3, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| P146 | U | sp | NPHP-RC | Prenatal | Enlarged, echogenic kidneys with normal amniotic fluid volume | No prenatally | – | Bardet-Biedl syndrome 12; #615989 | Comp het (AR) | *BBS12* | c.1616G>A p.(Gly539Asp)(m) | [35] | LP (PM2, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| c.1890\_1891del p.(Pro632Phefs\*7)(p) | Novel | P (PVS1, PM2, PM3) |
| P147 | M | fam | NPHP-RC | <30 y | Echogenic kidneys, renal cortical cysts. CKD G3 at 33 y | No at 33 y | – | Nephronophthisis 3; 3604387 | Homo (AR) | *NPHP3* | c.329\_331del p.(Leu110del)(p), c.329\_331del p.(Leu110del)(m) | Novel | LP (PM2, PM3, PM4, BP4) | Confirmed the clinical diagnosis |
| P148 | M | sp | NPHP-RC | U | Slightly diminished kidneys, echogenic kidneys, CKD, nocturnal enuresis, polyuria and polydipsia. CKD G2 at 16 y | No at 16 y | – | Nephronophthisis 4; #606966 | Comp het (AR) | *NPHP4* | c.517+1G>A p.(?) | [1] | P (PVS1, PM2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| c.4115T>C p.(Leu1372Pro) | [1] | LP (PM1, PM2, PP3, PP4, PP5) |
| P149 | M | sp | NPHP-RC | 19 y | HBP. CKD G3 at 19 y | No at 19 y | CHF, retinitis pigmentosa, esophageal varices | Nephronophthisis 13; #614377 Senior-Loken syndrome 8; #616307 | Comp het (AR) | *WDR19* | c.1477G>C p.(Asp493His) | [36] | LP (PM2, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| c.3112C>T p.(Arg1038\*) | [1] | P (PVS1, PM2, PM3, PP3, PP5) |
| P150 | F | sp | NPHP-RC | 16 y | Nocturnal enuresis (birth), polyuria and polydipsia, hyperuricemia (infancy), RC (16 y). CKD G4 at 16 y | No at 16 y |  | Nephronophthisis 4; #606966 | Comp het (AR) | *NPHP4* | c.518-1G>C p.(?)(m) | [1] | P (PVS1, PM2, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| c.1956-2A>C p.(?)(p) | [37] | P (PVS1, PM2, PM3, PP3, PP5) |
| P151 | M | sp | NPHP-RC | U | Normal-sized, echogenic kidneys, loss of corticomedullary differentiation, RC. CKD | No at 15 y | Growth retardation (1 y), myopia (6 y), asthenia, genu valgum, dorsal scoliosis, adynamia, renal osteodystrophy, cone-shaped epiphyses, retinitis pigmentosa | Short-rib thoracic dysplasia 9 with or without polydactyly; #266920 | Comp het (AR) | *IFT140* | c.975G>T p.(Glu325Asp) | [1] | P (PM2, PM3, PP4, PP5) | Confirmed the clinical diagnosis |
| c.2656C>T p.(Gln886\*) | [1] | P (PVS1, PM2, PM3, PP3, PP4, PP5) |
| P152 | M | sp | UPKD | Prenatal (2 y) | Enlarged, echogenic kidneys, RC, OH | Exitus at 2 y | Blindness, psychomotor delay, anemia,  failure to thrive | Joubert syndrome 5; #610188 | Comp het (AR) | *CEP290* | c.4045\_4046del p.(Met1349Glufs\*11)(m) | [1] | P (PVS1, PM1, PM2, PM3) | Identified a specific diagnosis |
| c.6869del p.(Asn2990Ilefs\*11)(p) | [1] | P (PVS1, PM1, PM2, PM3, PP5) |
| P153 | F | fam cons | NPHP-RC | 12 y | RH, HBP, loss of corticomedullary differentiation, cortical RC | 15 y | Growth retardation, psychomotor delay, hypertensive retinopathy | Nephronophthisis 12; #613820 | Homo (AR) | *TTC21B* | c.626C>T p.(Pro209Leu)(p), c.626C>T p.(Pro209Leu)(m) | [38] | LP (PM3, PP3, PP4, PP5, BP1) | Confirmed the clinical diagnosis |
| P154 | M | fam cons | NPHP-RC | U | NR prot, HBP | 20 mo | Growth retardation, anemia, metabolic acidosis, cone-shaped epiphyses, left ventricular hypertrophy | Short-rib thoracic dysplasia 9 with or without polydactyly; #266920 | Homo (AR) | *IFT140* | c.(?\_-1)\_(\*1\_?)del p.(?)(p), c.(?\_-1)\_(\*1\_?)del p.(?)(m) | Novel | P (PVS1, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| P155 | M | fam cons | NPHP-RC | 12 y | U | 15 y |  | Nephronophthisis 1, juvenile; #256100 | Homo (AR) | *NPHP1* | c.(?\_-1)\_(\*1\_?)del p.(?)(p), c.(?\_-1)\_(\*1\_?)del p.(?)(m) | [39] | P (PVS1, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| P156 | M | sp | ARPKD | 12 y | Echogenic kidneys, loss of corticomedullary differentiation, HBP, Mh, 1 cyst (13 y). CKD G3 at 17 y | No at 16 y | CHF, craniofacial dysmorphism, high-arched palate, anodontia, clinodactyly, syndactyly, splenomegaly (13 y) | Cranioectodermal dysplasia 1; #218330 | Comp het (AR) | *IFT122* | c.1049G>A p.(Gly350Asp)(p) | Novel | LP (PM2, PM3, PP3, PP4) | Reclassified the diagnosis |
| c. 1685T>C p.(Leu562Pro)(m) | Novel | LP (PM2, PM3, PP3, PP4) |
| P157 | M | sp | NPHP-RC | 1 y | Echogenic kidneys, loss of corticomedullary differentiation, 1 cyst (right kidney), HBP. CKD G5 at 1 y | No at 1 y | CHF | Nephronophthisis 3; 3604387 | Comp het (AR) | *NPHP3* | c.1871C>T p.(Ser624Phe)(m) | Novel | LP (PM2, PM3, PP3, PP4) | Confirmed the clinical diagnosis |
| c.1943dup p.(Ile649Asnfs\*22)(p) | Novel | P (PVS1, PM2, PM3, PP4) |
| P158 | M | sp | NPHP-RC | 2 mo | Cortical echogenic kidneys, peripheral microcysts, HBP | 2 y | Anemia, metabolic acidosis | Nephronophthisis 3; 3604387 | Comp het (AR) | *NPHP3* | c.372\_376del p.(Lys124Asnfs\*37)(m) | Novel | P (PVS1, PM2, PM3) | Confirmed the clinical diagnosis |
| c.3837dup p.(Ala1280Serfs\*3)(p) | Novel | P (PVS1, PM2, PM3, PP3) |
| P159 | M | sp | NPHP-RC | Childhood | Skin biopsy compatible with NPHP-RC | <30 y |  | Nephronophthisis 1, juvenile; #256100 | Homo (AR) | *NPHP1* | c.(?\_-1)\_(\*1\_?)del p.(?)(p), c.(?\_-1)\_(\*1\_?)del p.(?)(m) | [39] | P (PVS1, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| P160 | M | sp | NPHP-RC | 1 y | Renal biopsy: tubulointerstitial fibrosis | 2 y |  | Nephronophthisis 2, infantile; #602088 | Comp het (AR) | *INVS* | c.1484G>A p.(Trp495\*)(p) | Novel | P (PVS1, PM2, PM3, PP3) | Confirmed the clinical diagnosis |
| c.2695C>T p.(Arg899\*)(m) | [40] | P (PVS1, PM3, PP3, PP5) |
| P161 | M | fam cons | UPKD | Prenatal (TOP) | Enlarged kidneys, RC | TOP | DPM, facial dysmorphism | NPHP-RC; \*609884 | Homo (AR) | *TMEM67* | c.517T>C p.(Cys173Arg)(m), c.517T>C p.(Cys173Arg)(p) | [1] | LP (PM2, PM3, PP3, PP5) | Identified a specific diagnosis |
| P162 | F | sp | UPKD | Prenatal (TOP) | Enlarged kidneys, RC, AH | TOP | DPM, facial dysmorphism, PH, ventriculomegaly, biventricular hypertrophy | NPHP-RC; \*609884 | Comp het (AR) | *TMEM67* | c.551G>A p.(Cys184Tyr)(m) | [1] | LP (PM2, PM3, PP2, PP3, PP5) | Identified a specific diagnosis |
| c.1055\_1065del p.(Gly352Alafs\*24)(p) | [1] | P (PVS1, PM2, PM3, PP3, PP5) |
| P163 | M | fam cons | NPHP-RC | 14 y | Biopsy: interstitial fibrosis and tubular atrophy, prot, (18mg/m2/h), HBP. CKD G5 at 16 y | No at 14 y | Maculopathy | Nephronophthisis 12; #613820 | Homo (AR) | *TTC21B* | c.626C>T p.(Pro209Leu)(p), c.626C>T p.(Pro209Leu)(m) | [38] | LP (PM3, PP3, PP4, PP5, BP1) | Confirmed the clinical diagnosis |
| P164 | M | sp | NPHP-RC | 1 y | Hypokalemia. ,CKD G5 at 1 y | No at 1 y | Trigonocephaly, kyphoscoliosis, CD, psychomotor delay, unilateral nystagmus, night blindness | Nephronophthisis 13; #614377 Senior-Loken syndrome 8; #616307 | Comp het (AR) | *WDR19* | c.1442A>G p.(His481Arg)(m) | [1] | LP (PM2, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| c.2741C>A p.(Ala914Asp)(p) | [36] | LP (PM2, PM3, PP3, PP5) |
| P165 | M | sp | UPKD | 9 y | Bilateral RC, HBP. Normal Cr at 12 y | No at 12 y | HC, CD, pancreatic cysts, | Nephronophthisis 13; #614377 Senior-Loken syndrome 8; #616307 | Comp het (AR) | *WDR19* | c.2333C>G p.(Ser778\*)(p) | [1] | P (PVS1, PM2, PM3, PP3, PP5) | Identified a specific diagnosis |
| c.3703G>A p.(Glu1235Lys) (m) | [36] | LP (PM2, PM3, PP3, PP5) |
| P166 | F | sp | UPKD | Prenatal (TOP) | Enlarged, echogenic kidneys, OH. Necropsy: multicystic-dysplastic kidneys | TOP | Pancreatic dysplasia, pancreatic cysts | Renal-hepatic-pancreatic dysplasia 1; #208540 | Homo (AR) | *NPHP3* | c.434\_437del p.(Glu145Valfs\*3)(m), c.434\_437del p.(Glu145Valfs\*3)(de novo) | [1] | P (PVS1, PM2, PM3, PP3, PP5) | Identified a specific diagnosis |
| P167 | F | sp | UPKD | Prenatal (TOP) | Enlarged kidneys, bilateral RC, AH | TOP |  | Renal-hepatic-pancreatic dysplasia 1; #208540 | Comp het (AR) | *NPHP3* | c.1084\_1087del p.(Val363Phefs\*6) | [1] | P (PVS1, PM2, PM3, PP3, PP5) | Identified a specific diagnosis |
| c.2694-2\_2694-1del p.(?) | [1] | P (PVS1, PP3, PP5) |
| P168 | U | fam cons | NPHP-RC | Prenatal (TOP) | Bilateral ureterohydronephrosis with cystic dilation of some renal tubules | TOP | Small thorax, pancreatic fibrosis, DPM, shortened femurs, trident pelvis | Short-rib thoracic dysplasia 3 with or without polydactyly; #613091 + Polycystic kidney disease 4 with or without hepatic disease; #263200 | CIP | *DYNC2H1* | c.5971A>T p.(Met1991Leu)(p), c.5971A>T p.(Met1991Leu)  (m) | [41] | P (PS1, PM1, PM2, PM3, PM5, PP2, PP5, BP4) | Reclassified the diagnosis |
| *PKHD1* | c.8312T>C p.(Val2771Ala)(p), c.8312T>C p.(Val2771Ala)  (m) | [30] | LP (PM1, PM2, PM3, PP2, PP3, PP5) |
| P169 | F | U | TSC | 23 y | AML, oncocytoma (bilateral radical nephrectomy (32 y)) | 25 y | Partial epilepsy, astrocytoma | Tuberous sclerosis-1; #191100 | Het (AD) | *TSC1* | c.278T>A p.(Leu93Gln) | [42] | LP (PM2, PM6, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| P170 | F | sp | TSC | 6 mo | AML (30 y). Normal Cr at 32 y | No at 32 y | West syndrome (6 m), epilepsy (28 y), hepatic hemangiomas (30 y), facial angiofibromas | Tuberous sclerosis-1; #191100 | Het (AD) | *TSC1* | c.2268\_2271del p.(Lys756Asnfs\*16) | [1] | P (PVS1, PM1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P171 | F | sp | TSC | 23 y | AML. Normal Cr at 37 y | No at 37 y | Cortical tubers, shagreen patch, ungual fibromas, hypomelanotic macules, facial angiofibromas | Tuberous sclerosis-1; #191100 | MOSAIC 9% reads | *TSC1* | c.1904\_1905del p.(Thr635=/Thr635Argfs\*52) | [43] | P (PVS1, PM6, PM1, PM2, PP5) | Confirmed the clinical diagnosis |
| P172 | F | sp | TSC | Birth | AML (20 y). Normal Cr at 35 y | No at 35 y | Seizures, mental retardation, developmental delay, lymphangioleiomyomatosis, hypomelanic macules, facial angiofibromas | Tuberous sclerosis-2; #613254 | Het (AD) | *TSC2* | c.(1716+1\_1717-1)\_(2545+1\_2546-1)del p.(?) | [1] | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P173 | F | sp | TSC | 8 y | AML. Normal Cr at 32 y | No at 32 y | Shagreen patch, lymphangioleiomyomatosis, cardiac rhabdomyoma, facial angiofibromas | Tuberous sclerosis-2; #613254 | Het (AD) | *TSC2* | c.2824G>T p.(Glu942\*) | [44] | P (PVS1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P174 | M | sp | TSC | 3 mo | RC, AML, left kidney nephrectomy (5 y). Normal Cr at 22 y | No at 22 y | Epilepsy (3 m), West syndrome (9 m), developmental delay (9 m), astrocytoma (12 y), ungual fibromas, hypomelanotic macules, angiofibromas, retinal phakoma, lesions in liver, lungs, and suprarenal gland | Tuberous sclerosis-2; #613254 | Het (AD) | *TSC2* | c.5228G>A p.(Arg1743Gln) | [45] | LP (PM1, PM2, PM5, PM6, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P175 | F | fam | TSC | 4 y | AML, prot. CKD G1 | No at 31 y | Cortical tubers, cardiac rhabdomyoma, subependymal nodule, retinal hamartomas, shagreen patch, ungual fibroma, hypomelanotic macules, facial angiofibromas | Tuberous sclerosis-2; #613254 | Het (AD) | *TSC2* | c.4515C>G p.(Tyr1505\*) | [46] | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P176 | M | sp | TSC | 14 y | AML. Normal Cr at 23 y | No at 23 y | Epilepsy, mental retardation, astrocytoma, cortical tubers, hepatic hemangiomas, cardiac rhabdomyoma, ungual fibromas, hypomelanotic macules | Tuberous sclerosis-2; #613254 | Het (AD) | *TSC2* | c.3251del p.(Asp1084Alafs\*19) | [1] | P (PVS1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P177 | M | sp | TSC | 10 mo | RC, AML (12 y) | U | Partial epilepsy (10 m), autism, cortical tubers (11 y), skin lesions, Klippel-Trenaunay syndrome | Tuberous sclerosis-2; #613254 | Het (AD) | *TSC2* | c.5161-1G>C p.(?) | [47] | P (PVS1, PM6, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P178 | F | sp | TSC | Childhood | Enlarged kidneys, AML, bilateral RC | No at 46 y | Epilepsy, mental retardation, psychomotor delay, obesity, hypothyroidism, left ocular atrophy, skin lesions, ungual fibromas | Tuberous sclerosis-2; #613254 | Het (AD) | *TSC2* | c.1257+1G>A p.(?) | [48] | P (PVS1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P179 | M | sp | UPKD | 19y | Slightly enlarged left kidney, normal-sized right kidney. Bilateral echogenic kidneys, loss of corticomedullary differentiation, RC (left kidney) | No at 21 y | – | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) (de novo) | [5] | P (PVS1, PM2, PP3, PP5) | Identified a specific diagnosis |
| P180 | F | fam | UPKD | 17 mo | Renal microcysts, echogenic kidneys, CKD (17 mo), prot (13 y). KFRT at 16 y | 16 y | – | Papillorenal syndrome; #120330 | Het (AD) | *PAX2* | c.70G>T p.(Gly24Trp) | [1] | LP (PM1, PM2, PM5, PP2, PP3, PP5) | Identified a specific diagnosis |
| P181 | F | fam | UPKD | Prenatal | Enlarged, echogenic kidneys, bilateral RC (prenatal). Normal Cr at 1.5 y | No at 1.5 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.9454C>G p.(Arg3152Gly) (p) | [1] | LP (PM1, PM2, PP1, PP3, PP4, PP5, BP1) | Identified a specific diagnosis |
| P182 | M | fam | UPKD | 12 y | Enlarged kidneys, RC, loss of corticomedullary differentiation, RL, HBP (18 y). KFRT at 30 y | 30 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.9365T>G p.(Ile3122Ser) (m) | [1] | LP (PM1, PM2, PP1, PP3, PP5, BP1) | Identified a specific diagnosis |
| P183 | M | sp | UPKD | 16 y | Bilateral RC, loss of corticomedullary differentiation (right kidney), HBP. Normal Cr at 16 y | No at 16 y |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.4306C>T p.(Arg1436\*) | [21] | P (PVS1, PM2, PM6, PP3, PP5) | Identified a specific diagnosis |
| P184 | F | fam | UPKD | 11 y | Bilateral RC. Normal Cr at 11 y | No at 11 y | – | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.7703G>A p.(Arg2568Lys)  (m) | Novel | LP (PM1, PM2, PP1, PP4, BP4) | Identified a specific diagnosis |
| P185 | M | sp | UPKD | 2 mo | Glomerulocystic kidney disease (2 mo). KFRT at 31 y | 31 y | Hypertransaminasemia, intrahepatic cholestasis | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.490A>C p.(Lys164Gln) (de novo) | [6] | P (PS2, PM1, PM2, PM6, PP2, PP3, PP5) | Identified a specific diagnosis |
| P186 | F | sp | UPKD | 16 y | Normal-sized kidneys, bilateral RC, RH. CKD G4 at 37 y | No at 37 y | Stroke | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.810\_1045del p.(Ala271Serfs\*10) | [49] | P (PVS1, PM2, PP3, PP5) | Identified a specific diagnosis |
| P187 | F | sp | UPKD | Birth | Enlarged kidneys (birth), RC, UTI, hypercalcemia. KFRT at 23 y | 23 y | T1DM, uterine fibroid | Congenital disorder of glycosylation, type Ia; #212065 | Comp het (AR) | *PMM2* | c.-167G>T p.(?) | [50] | P (PS3, PM3, PP3, PP4, PP5) | Identified a specific diagnosis |
| c.710C>G p.(Thr237Arg) | [51] | LP (PM1, PM3, PM5, PP3, PP3, PP5) |
| P188 | F | fam cons | UPKD | Birth | Bilateral RH, loss of corticomedullary differentiation, RC in the corticomedullary junction. CKD G3 at 5 y | No at 5 y | Respiratory distress, bilateral cortical atrophy in frontotemporal lobes | Renal hypodysplasia/aplasia 1; #191830 | Homo (AR) | *ITGA8* | c.2982+2T>C p.(?)(p), c.2982+2T>C p.(?)(m) | [52] | P (PVS1, PM2, PM3, PP3, PP5) | Identified a specific diagnosis |
| P189 | M | fam | UPKD | 29 y | Bilateral RC. CKD G3 at 34 y | No |  | Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps #611773 | Het (AD) | *COL4A1* | c.1793G>T p.(Gly598Val) | Novel | LP (PM1, PM2, PP2, PP3) | Identified a specific diagnosis |
| P190 | F | sp | AS | 11 y | Mh, prot. Normal Cr at 11 y | No at 11 y | Congenital cataracts, microcornea | Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps; #611773 | Het (AD) | *COL4A1* | c.3112G>A p.(Gly1038Ser)(*de novo*) | [1] | P (PS2, PM1, PM2, PP2, PP3, PP5) | Reclassified the diagnosis |
| P191 | F | fam | AS | 11 y | Mh | No at 11 y | – | Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps; #611773 | Het (AD) | *COL4A1* | c.4151G>A p.(Gly1384Asp)(m) | Novel | LP (PM1, PM2, PP1, PP2, PP3) | Reclassified the diagnosis |
| P192 | F | sp | AS | 28 y | Mh, prot. Biopsy: FSGS (LM). KFRT at 40 y | 40 y |  | Alport syndrome 3; #104200 | Het (AD) | *COL4A3* | c.2275G>A p.(Gly759Arg) | [1] | LP (PM1, PM2, PM6, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P193 | M | fam | AS | 7 y | Macroh (7 y), Mh, prot (11 y), intermittent hypercalciuria | No at 11 y |  | Alport syndrome 3; #104200 | Het (AD) | *COL4A3* | c.2126G>A p.(Gly709Glu)(m) | [1] | LP (PM1, PM2, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P194 | M | fam | AS | 8 y | Intermittent Mh, macroh, prot | U | – | AS digenic | CIP | *COL4A3* | c.3829G>A p.(Gly1277Ser)(p) | [53] | LP (PM1, PM2, PP1, PP2, PP3,  PP4 PP5) | Confirmed the clinical diagnosis |
| *COL4A4* | c.193G>A p.(Gly65Ser)(m) | Novel | LP (PM1, PM2, PP1, PP2, PP3) |
| P195 | M | fam | AS | 19 y | Mh, prot, HBP (19 y). Biopsy: mesangial GN (LM). CKD G5 at 50 y | No at 50 y | Hearing loss (50 y) | Alport syndrome 3; #104200 | Het (AD) | *COL4A3* | c.2074G>A p.(Gly692Ser) | Novel | LP (PM1, PM2, PP2, PP3) | Confirmed the clinical diagnosis |
| P196 | F | sp | AS | 10 y | Mh, NR prot | U 40 y |  | Alport syndrome 2; #203780 | Comp het (AR) | *COL4A3* | c.2153G>C p.(Gly718Ala) | Novel | LP (PM1, PM2, PP2, PP3) | Confirmed the clinical diagnosis |
| c.4732T>C, p.(Trp1578Arg) | Novel | LP (PM1, PM2, PP2, PP3) |
| P197 | F | fam | AS | 13 y | Clinical features compatible with AS | 21 y | Bilateral sensorineural hearing loss (5 y) | Alport syndrome 2; #203780 | Comp het (AR) | *COL4A3* | c.725G>A p.(Gly242Glu) | Novel | LP (PM1, PM2, PP2, PP4, PP3) | Confirmed the clinical diagnosis |
| c.4981C>T p.(Arg1661Cys) | [53] | LP (PM1, PP2, PP3, PP4, PP5) |
| P198 | M | fam | AS | 15 y | Mh, macroh with prot in infection context. Normal Cr at 8 y | No at 18 y | – | Alport syndrome 3; #104200 | Het (AD) | *COL4A3* | c.3829G>A p.(Gly1277Ser)  (m) | [53] | P (PS1, PM1, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P199 | F | fam | UGLO | 4 y | Mh, albuminuria, prot. Biopsy (16 y): proliferative mesangial GN (LM). Biopsy (34 y): FSGS (LM). CKD G3 at 35 y | No at 35 y | – | Alport syndrome 2; #203780 | Comp het (AR) | *COL4A3* | c.4045G>A p.(Gly1349Ser) | Novel | LP (PM1, PM2, PP2, PP3) | Identified a specific diagnosis |
| c.4649T>G p.(Val1550Gly) | Novel | LP (PM1, PM2, PP2, PP3) |
| P200 | F | fam cons | AS | 5 y | Mh, macroh, mild prot. Normal Cr at 11 y | No at 11 y | – | Alport syndrome 2; #203780 | Homo (AR) | *COL4A3* | c.547G>T p.(Gly183Cys)(p), c.547G>T p.(Gly183Cys)(m) | Novel | LP (PM1, PM2, PM3, PP1, PP2, PP3) | Confirmed the clinical diagnosis |
| P201 | M | fam | AS | 11 y | Mh. Normal Cr at 15 y | No at 15 y | – | Alport syndrome 3; #104200 | Het (AD) | *COL4A3* | c.2987\_2993del p.(Arg996Ilefs\*156)(m) | Novel | P (PVS1, PM1, PM2, PP1, PP3) | Confirmed the clinical diagnosis |
| P202 | F | sp | AS | 14 y | Mh, NR prot. Biopsy (14 y): membranous glomerulonephritis. Biopsy (24 y): FSGS | No at 25 y | Mild sensorineural hearing loss | Alport syndrome 2; #203780 | Homo (AR) | *COL4A3* | c.2371C>T p.(Arg791\*)(p), c.2371C>T p.(Arg791\*)(m) | Novel | P (PVS1, PM1, PM2, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| P203 | M | fam | AS | 3 y | Mh. Normal Cr at 6 y | No at 6 y | – | Alport syndrome 3; #104200 | Het (AD) | *COL4A3* | c.3044G>A p.(Gly1015Glu) (m) | [54] | LP (PM1, PM2, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P204 | F | sp | AS+RC | 24 y | Clinical features and skin biopsy compatible with AS. CKD G3 at 30 y | No at 30 y | Bilateral coloboma | Alport syndrome 3; #104200 + Papillorenal syndrome; #120330 | CIP | *COL4A3* | c.4996A>G p.(Met1666Val) | Novel | LP (PM1, PM2, PM6, PP2, PP3) | Reclassified the diagnosis |
| *PAX2* | c.343C>T p.(Arg115\*) | [55] | P (PVS1, PM1, PM2, PM6, PP3, PP5) |
| P205 | F | fam | AS | U | Mh, prot. Biopsy: FSGS (LM). CKD G3 at 27 y | No at 27 y | Myopia (14 y), bilateral high-tone sensorineural hearing loss (22 y), astigmatism (22 y) and occasional edemas | AS digenic | CIP | *COL4A3* | c.4826G>A p.(Arg1609Gln)  (m) | Novel | LP (PM1, PM2, PP1, PP2, PP3) | Confirmed the clinical diagnosis |
| *COL4A4* | c.2908C>T p.(Gln970\*)(p) | Novel | P (PVS1, PM1, PM2, PM3, PP1, PP3) |
| c.4351G>A p.(Gly1451Arg)  (m) | Novel | LP (PM1, PM2, PM3, PP1, PP2, PP3) |
| P206 | M | fam | AS | 17 y | Mh (17 y), prot (30 y), HBP. Biopsy: Proliferative mesangial GN (LM). CKD G3 at 57 y | No at 57 y | High-tone hearing loss | Alport syndrome 2; #203780 | Comp het (AR) | *COL4A4* | c.2320G>C p.(Gly774Arg)(m) | [56] | LP (PM1, PM2, PM3, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| c.4508del p.(His1503Profs\*49)(p) | [1] | P (PVS1, PM1, PM2, PM3, PP1, PP3, PP5) |
| P207 | M | fam | AS | 25 y | Mh. Normal Cr at 66 y | No at 66 y | Bilateral high-tone sensorineural hearing loss | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.3688G>T p.(Gly1230Cys)(p) | [1] | LP (PM1, PM2, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P208 | M | fam | AS | U | Mh, NR prot. CKD G3 at 30 y | No at 30 y | – | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.3205G>C p.(Gly1069Arg) | [1] | P (PS1, PM1, PM2, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P209 | F | fam | AS | 5 y | Mh, prot (5y). Biopsy: Mesangial GN (LM) abnormal α3 and α5 collagen. Normal Cr at 15 y | No at 15 y | – | Alport syndrome 2; #203780 | Comp het (AR) | *COL4A4* | c.755G>A p.(Gly252Asp)(m) | Novel | LP (PM1, PM2, PM3, PP1, PP2, PP3) | Confirmed the clinical diagnosis |
| c.4426C>T p.(Gln1476\*)(p) | Novel | P (PVS1, PM1, PM2, PM3, PP1, PP3) |
| P210 | F | fam | AS | 20 y | Mh, prot (20 y), HBP (32 y). Biopsy: MCD GN. KFRT at 43 y | 43 y |  | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.735+2T>C p.(?) | Novel | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P211 | M | fam | AS | 4 y | Mh, macroh (4 y) prot (5 y). Biopsy: Proliferative mesangial GN. Normal Cr at 12 | No at 12 y | Strabismus, sensorineural hearing loss (12 y) | Alport syndrome 2; #203780 | Homo (AR) | *COL4A4* | c.4334-23A>G p.(?)(p), c.4334-23A>G p.(?)(m) | [57] | LP (PM2, PM3, PP1, PP4, PP5) | Confirmed the clinical diagnosis |
| P212 | M | fam | AS | U | Clinical features compatible with AS. KFRT at 14 y | 14 y | Hearing loss | Alport syndrome 2; #203780 | Comp het (AR) | *COL4A4* | c.1045C>T p.(Arg349\*) | Novel | P (PVS1, PM1, PM2, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| c.1324G>T, p.(Gly442Cys) | Novel | P (PM1, PM2, PM3, PP2, PP3) |
| P213 | M | fam | AS | 14 y | Mh (14 y), prot (29 y), macroh (35 y). Biopsy (36 y): Proliferative mesangial GN (LM). Normal Cr at 42 y | No at 42 y | Mild bilateral hearing loss | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.1952G>T p.(Gly651Val) | Novel | LP (PM1, PM2, PP2, PP3) | Confirmed the clinical diagnosis |
| P214 | M | fam | AS | 10 y | Mh (10 y). Normal Cr at 14 y | No at 14 y |  | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.491G>A p.(Gly164Asp)(m) | Novel | LP (PM1, PM2, PP1, PP2, PP3) | Confirmed the clinical diagnosis |
| P215 | M | fam | AS | <30 y | Hemat, prot, HBP. CKD G4 at 37 y | No at 37 y |  | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.3559G>A p.(Gly1187Arg) | Novel | LP (PM1, PM2, PP2, PP3) | Confirmed the clinical diagnosis |
| P216 | F | fam | UGLO | 25 y | Hemat, prot (25 y), RC. CKD G3 at 55 y | No at 55 y |  | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.558+1G>A p.(?) | Novel | P (PVS1, PM2, PP3) | Identified a specific diagnosis |
| P217 | M | fam | AS | 7 y | Hemat, prot (7 y). Normal Cr at 7 y | No at 7 y | Ophthalmological findings compatible with AS | Alport syndrome 2; #203780 | Comp het (AR) | *COL4A4* | c.559-2A>C p.(?)(m) | Novel | P (PVS1, PM2, PM3, PP1, PP3) | Confirmed the clinical diagnosis |
| c.4334-23A>G p.(?)(p) | [57] | LP (PM2, PM3, PP1, PP4, PP5) |
| P218 | M | fam | AS | <3 y | Mh, prot. Biopsy (4 y): MCD (LM), diffuse effacement of the podocyte foot processes (EM) | No at 4 y |  | Alport syndrome 2; #203780 + Nephrotic syndrome type 2 #600995 | CIP | *COL4A4* | c.3619G>A p.(Gly1207Arg)(p) | Novel | LP (PM1, PM2, PP1, PP2, PP3) | Reclassified the diagnosis |
| *NPHS2* | c.412C>T p.(Arg138\*)(u) | [58] | P (PVS1, PM2, PP3, PP5) |
| c.686G>A p.(Arg229Gln)(p) | [59] | P (PS1, PS3, PM1, PM3, PP2, PP3, PP4, PP5) |
| P219 | M | fam cons | AS | U | Clinical features compatible with AS. KFRT at 31 y | 31 y | Bilateral sensorineural hearing loss (18 y) | Alport syndrome 2; #203780 | Homo (AR) | *COL4A4* | c.1323\_1340del p.(Pro444\_Leu449)(p), c.1323\_1340del p.(Pro444\_Leu449)(m) | [57] | LP (PM1, PM2, PM3, PM4, PP1, BP4) | Confirmed the clinical diagnosis |
| P220 | M | fam | AS | 12 y | Clinical features compatible with AS. Biopsy: Inconclusive. CKD G5 at 12 y | No at 12 y | Hearing loss, myopia, astigmatism | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.834del (p.Gly279Valfs\*67)(m) | [1] | LP (PVS1, PM1, PM2, PP1, PP5) | Confirmed the clinical diagnosis |
| P221 | M | fam | AS | 2 y | Mh, prot. Normal Cr at 4 y | No at 4 y |  | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.3902del p.(Leu1301Cysfs\*4)(m) | [1] | P (PVS1, PM1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P222 | F | fam | AS | 3 y | Mh, prot. Biopsy: MCD (LM). Normal Cr at 4 y | No at 4y | – | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.3659G>A p.(Gly1220Asp) (m) | [60] | LP (PM1, PM2, PM5, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P223 | F | fam | AS | 30 y | Clinical features compatible with AS. CKD at 31y | No at 31 y |  | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.3373G>A p.(Gly1125Arg) (m) | [1] | LP (PM1, PM2, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P224 | F | fam | AS | 2 y | Macroh (2 y), prot (3 y). Normal Cr at 4 y | No at 4 y |  | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.(3373+1\_3374-1)\_(3790+1\_3791-1)dup p.(?)(m) | [1] | LP (PM2, PM4, PP1, PP4) | Confirmed the clinical diagnosis |
| P225 | M | fam | AS | 2 y | Mh, macroh (2 y), NR prot (5 y), NS (6 y), HBP (7 y). Biopsy: proliferative mesangial GN (LM), IgA nephropathy (IF). Normal Cr at 15 y | No at 15 y | High-tone sensorineural hearing loss (8 y) | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.4907\_4968del p.(Ile1636Asnfs\*10)(m) | [1] | P (PVS1, PM1, PM2, PP1, PP3, PP5) | Confirmed the clinical diagnosis |
| P226 | F | fam | AS | 12 y | Mh, UTI. Normal Cr at 12 y | No at 12 y |  | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.3373G>A, p.(Gly1125Arg) (m) | [1] | LP (PM1, PM2, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P227 | F | fam | AS | Childhood | Mh (childhood), mild prot (30 y). Normal Cr at 30 y | No at 30 y |  | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.3373G>A p.(Gly1125Arg) (m) | [1] | LP (PM1, PM2, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P228 | M | fam | AS | 15 y | Mh, prot (15 y). Biopsy: FSGS (LM). KFRT at 29 y | 29 y |  | AS digenic | CIP | *COL4A5* | c.1643G>A p.(Gly548Asp) | [61] | LP (PM1, PM2, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| *COL4A3* | c.4649T>G p.(Val1550Gly) | Novel | LP (PM1, PM2, PP2, PP3) |
| P229 | M | sp | AS | 11 y | Hemat, prot (11 y). KFRT at 18 y | 18 y | Bilateral high-tone sensorineural hearing loss, possible lenticonus | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.1933C>T p.(Gln645\*)(de novo) | Novel | P (PVS1, PS2, PM1, PM2, PP3) | Confirmed the clinical diagnosis |
| P230 | M | fam | AS | Childhood | Clinical features compatible with AS. Echogenic RC in left kidney (26 y). KFRT at 23 y | 23 y | Bilateral sensorineural hearing loss, myopia, astigmatism, strabismus | AS digenic | CIP | *COL4A5* | c.465+2T>G p.(?)(m) | Novel | P (PVS1, PM2, PP1, PP3) | Confirmed the clinical diagnosis |
| *COL4A3* | c.2390C>T p.(Pro797Leu) | LOVD db | LP (PM1, PM2, PP2, PP3) |
| P231 | M | fam | AS | 4 y | Macroh, mh, prot | U | – | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.1826G>C p.(Gly609Ala)(m) | Novel | LP (PM1, PM2, PM5, PP1, PP2, PP3) | Confirmed the clinical diagnosis |
| P232 | F | fam | AS | 29 y | Mh, prot. Normal Cr at 29 y | No at 29 y |  | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.731A>C p.(Asp244Ala) | Novel | LP (PM1, PM2, PP2, PP3) | Confirmed the clinical diagnosis |
| P233 | M | fam | AS | 18 mo | Hemat, prot (18 mo). Hyperuricemia (27 y). Biopsy: diffuse and global mesangial hypercellularity, IgG-IgA immunodeficiency. Normal Cr at 27 y | No at 27 y |  | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.1481G>A p.(Gly494Asp)(m) | [62] | LP (PM1, PM2, PM5, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P234 | M | fam | AS | 13 y | Mh, NR prot. Normal Cr at 13 y | No at 13 y | Bilateral medium-tone sensorineural hearing loss | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.3119\_3120delinsA p.(Val1040Glufs\*112)(m) | Novel | P (PVS1, PM1, PM2, PP1) | Confirmed the clinical diagnosis |
| P235 | M | sp | AS | < 20 y | Clinical features compatible with AS. KFRT at 20 y | 20 y | Hearing loss, Café au lait spot, thalamic hematoma, ophthalmological findings not specified. | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.1234G>C p.(Gly412Arg) | Novel | LP (PM1, PM2, PM5, PM6, PP2, PP3) | Confirmed the clinical diagnosis |
| P236 | M | fam | AS | 8 y | Mh (8 y), prot (9 y ). Biopsy: proliferative mesangial GN (LM). Normal Cr at 12 y | No at 12 y | – | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.546+2T>G p.(?)(m) | Novel | P (PVS1, PM2, PM6, PP3) | Confirmed the clinical diagnosis |
| P237 | M | fam | AS | 2 y | Macroh (2 y), MAU (6 y), prot (17 y). Biopsy (16 y): FSGS(LM). Normal Cr at 17 y | No at 17 y |  | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.2677G>A p.(?) | [63] | LP (PM1, PM2, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P238 | F | fam | NS | 15 y | NR prot. Biopsy (15 y): FSGS (LM). KFRT at 24 y | 24 y |  | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.983G>A p.(Gly328Asp) | Novel | LP (PM1, PM2, PP2, PP3) | Reclassified the diagnosis |
| P239 | F | sp | AS | 9 y | Mh, prot (9 y). Biopsy (9 y): Irregular thickening of the GBM (LM) | No at 26 y | – | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.3511C>T p.(Gln1171\*) | Novel | P (PVS1, PS2, PM1, PM2, PP3) | Confirmed the clinical diagnosis |
| P240 | F | sp | AS | <17 y | HBP. Biopsy (17 y): abnormal α3 and α5 collagen. KFRT at 17 y | 17 y | Hyperparathyroidism, hypertensive retinopathy | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.466-12G>A p.(?) | [64] | LP (PM2, PM6, PP5, BP4) | Confirmed the clinical diagnosis |
| P241 | F | fam | AS | 2 y | Macroh, prot. Normal Cr at 4 y | No at 4 y | – | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.2668G>A p.(Gly890Arg) | Novel | LP (PM1, PM2, PP2, PP3) | Confirmed the clinical diagnosis |
| P242 | F | fam | AS | Childhood | Mh, NR prot, UTI. Normal Cr at 32 y | No at 32 y |  | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.1912G>A p.(Gly638Ser) | [60] | P (PS1, PM1, PM2, PM5, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P243 | F | fam | AS | Childhood | Mh | U | – | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.465+1G>A p.(?) | Novel | P (PVS1, PM2, PP3) | Confirmed the clinical diagnosis |
| P244 | M | fam | NPHP-RC | 15 y | Echogenic kidneys, microcysts, loss of corticomedullary differentiation. CKD G4 at 15 y | No at 15 y | Hyperparathyroidism | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.232-2A>G p.(?)(m) | Novel | P (PVS1, PM2, PP1, PP3) | Reclassified the diagnosis |
| P245 | M | fam | AS | U | Clinical features compatible with AS. KFRT at 17 y | 17 y | Hearing loss, macular pseudohole | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.1768A>T p.(Lys590\*) | Novel | P (PVS1, PM1, PM2, PP3) | Confirmed the clinical diagnosis |
| P246 | F | fam | AS | 5 y | Macroh, mh, prot. Normal Cr at 7 y | No at 7 y | – | Alport syndrome X-linked; #301050 | Het (XL) | *COL4A5* | c.4298-1G>T p.(?)(p) | Novel | P (PVS1, PM2, PP1, PP3) | Confirmed the clinical diagnosis |
| P247 | M | fam | AS | 4 y | Macroh (4 y). Biopsy (30 y, post-transplant): proliferative mesangial GN (LM), IgM and IgA deposits (IF). KFRT at 29 y | 29 y | Bilateral hearing loss, keratoconus, myopia, astigmatism | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.917del p.(Asn306Metfs\*40) | Novel | P (PVS1, PM1, PM2) | Confirmed the clinical diagnosis |
| P248 | M | sp | AS | U | Clinical features compatible with AS, HBP. KFRT at 23 y | 23 y | Hearing loss (14 y) | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.2039del p.(Pro680Glnfs\*56) | LOVD db | P (PVS1, PM1, PM2, PM6, PP3, PP5) | Confirmed the clinical diagnosis |
| P249 | M | fam | AS | 2 y | Prot, left kidney pyelectasis (2 y). Biopsy (30 y): Interstitial fibrosis and irregular thickening of the GBM (LM) | < 36 y |  | Alport syndrome X-linked; #301050 | Hemi (XL) | *COL4A5* | c.141+1G>A p.(?) | [65] | P (PVS1, PM2, PP3, PP5) | Confirmed the clinical diagnosis |
| P250 | M | fam | AS | 10 y | Mh. Normal Cr at 10 y | No at 10 y |  | Alport syndrome 2; #203780 | Het (AD) | *COL4A4* | c.2312dup p.(Arg773Glufs\*15) | Novel | P (PVS1, PM1, PM2) | Confirmed the clinical diagnosis |
| P251 | M | fam | AS | Childhood | NR prot, mh, hyperpotassemia. Biopsy: compatible with AS. CKD G4 at 25 y | No at 25 y | Bilateral hearing loss | Glomerulosclerosis, focal segmental, 2, #603965 | Het (AD) | *TRPC6* | c.2643dup p.(Trp783Metfs\*4) | Novel | P (PVS1, PM1, PP3) | Reclassified the diagnosis |
| P252 | M | fam cons | NS | 3 y | NR prot, hypoalbuminemia, hypertriglyceridemia, hypercholesterolemia. Biopsy: MCD (LM). Normal Cr at 4 y | No at 4 y |  | Megaloblastic anemia-1, Finnish type; #261100 | Homo (AR) | *CUBN* | c.5840C>A p.(Ser1947Tyr)(p), c.5840C>A p.(Ser1947Tyr) (m) | [66] | LP (PM3, PP1, PP3, PP4, PP5, BP1) | Confirmed the clinical diagnosis |
| P253 | F | sp | UGLO | 2 y | NR prot, intermittent mh, intermittent hypoalbuminemia, hypercholesterolemia. Normal Cr at 7 y | No at 7 y |  | Nail-patella syndrome; #161200 | Het (AD) | *LMX1B* | c.737G>A p.(R246Q)(*de novo*) | [67] | P (PS2, PM1, PM2, PP2, PP3, PP5) | Identified a specific diagnosis |
| P254 | M | sp | NS | 8 y | SRNS. CKD at 9 y | No at 9 y |  | Nephrotic syndrome, type 12; #616892 | Homo (AR) | *NUP93* | c.1772G>T p.(Gly591Val)(p), c.1772G>T p.(Gly591Val)(m) | [68] | LP (PM3, PP1, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| P255 | F | sp | NS | < 30 y | NR prot. CKD G5 at 31 y | No at 31 y |  | Nephrotic syndrome, type 2; #600995 | Comp het (AR) | *NPHS2* | c.686G>A p.(Arg229Gln)(p) | [59] | P (PS1, PS3, PM1, PM3, PP2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| c.953\_955del p.(Ala318del)(m) | Novel | LP (PM2, PM3, PM4, BP4) |
| P256 | M | sp | NS | 1.5 y | Prot. Biopsy: FSGS | No at 1.5 y |  | Glomerulosclerosis, focal segmental, 6; #614131 | Comp het (AR) | *MYO1E* | c.2481-12A>G p.(?)(p) | Novel | LP (PM2, PM3, PP3, PP4) | Confirmed the clinical diagnosis |
| c.2908C>T p.(Gln970\*)(m) | Novel | P (PVS1, PM2, PP3) |
| P257 | M | sp | NPHP-RC | <6 y | NR prot, hemat, small echogenic kidneys, hypoalbuminemia, hypertriglyceridemia, hypercholesterolemia hypoproteinemia, hypokalemia, hyperphosphatemia. Biopsy: Global glomerulosclerosis (6 glomeruli), FSGS (1 glomeruli), tubular atrophy (6 y) and irregularly thickened basal membrane | 6 y | Retinitis pigmentosa, weakness, developmental delay, seizures | Nephrotic syndrome type 9; #615573 | Comp het (AR) | *COQ8B* | c.439T>C p.(Cys147Arg) | Novel | LP (PM2, PM6, PP3, PP4) | Reclassified the diagnosis |
| c.1035+2T>C p.(?) | Novel | P (PVS1, PM2, PM3, PP3) |
| P258 | M | sp | NS | 8 y | NR prot. Biopsy: FSGS. Normal Cr at 11y | No at 11 y | – | Megaloblastic anemia-1, Finnish type; #261100 | Comp het (AR) | *CUBN* | c.4689\_4690delinsAT p.(Cys1563\*)(p) | Novel | P (PVS1, PM2, PM3, PP4, PP5) | Confirmed the clinical diagnosis |
| c.9053A>C p.(Tyr3018Ser) (m) | [66] | LP (PM3, PP3, PP4, PP5) |
| P259 | M | fam cons | UGLO | U | Prot. Normal Cr at 7 y | No at 7 y |  | Megaloblastic anemia-1, Finnish type; #261100 | Homo (AR) | *CUBN* | c.5840C>A p.(Ser1947Tyr)(p), c.5840C>A p.(Ser1947Tyr) (m) | [66] | LP (PM3, PP3, PP4, PP5, BP1) | Identified a specific diagnosis |
| P260 | M | sp | NS | 4 mo | NR prot | 3 y | Schimke immunoosseous dysplasia | Schimke immunoosseous dysplasia; #242900 | Comp het (AR) | *SMARCAL1* | c.1736C>T p.(Ser579Leu) | [69] | LP (PM1, PM2, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| c.(1147+1\_1148-1)\_(1334+1\_1335-1)del p.(?) | Novel | P (PVS1, PM3, PP3, PP5) |
| P261 | M | sp | NS | 4 y | NR prot, hypoalbuminemia, hypercholesterolemia. Biopsy: FSGS. Normal Cr at 4 y | No at 4 y |  | Focal segmental glomerulosclerosis 9; #616220 | Comp het (AR) | *CRB2* | c.1827C>A p.(Cys609\*)(p) | Novel | P (PVS1, PM1, PM2, PM3, PP4, PP3) | Confirmed the clinical diagnosis |
| c.3214C>T p.(Arg1072Cys)(m) | Novel | LP (PM2, PM3, PP3, PP4, BP1) |
| P262 | M | fam cons | NS | 45 days (5 mo) | NR prot, Poor corticomedullary differentiation, HBP, hyperkalemia, hyponatremia, hypomagnesemia. Biopsy: DMS, tubular atrophy, primitive glomeruli. | Exitus 5 mo | Hypothyroidism, microcephaly, brain atrophy, poor myelination, severe psychomotor delay for his age, axial hypotonia | Galloway-Mowat syndrome 3; #617729 | Homo (AR) | *OSGEP* | c.81C>G (p.Asn27Lys)(p), c.81C>G (p.Asn27Lys)(m) | [70] | LP (PM2, PM3, PP1, PP2, PP3, PP4) | Confirmed the clinical diagnosis |
| P263 | F | fam cons | NS | 75 days (3 mo) | NR prot, Cortical hyperechogenicity, hypoproteinemia, severe hypoalbuminemia, hypertriglyceridemia, hypercholesterolemia. Biopsy: Increased glomerular mesangial matrix | Exitus 3 mo | Dysmorphic features (wide nasal bridge, aquiline nose and retrognathia, low set ears, and arachnodactyly), microcephaly, brain atrophy, axial hypotonia, poor eye contact, developmental delay, epileptiform activity, seizures. | Galloway-Mowat syndrome 3; #617729 | Homo (AR) | *OSGEP* | c.157A>T p.(Ile53Phe)(p), c.157A>T p.(Ile53Phe)(m) | [70] | LP (PM2, PM3, PP1, PP2, PP4) | Confirmed the clinical diagnosis |
| P264 | M | sp | NS | U | NR prot. Biopsy: FSGS. CKD G3 at 4 y | No at 8 y |  | Glomerulosclerosis, focal segmental, 2; #603965 | Het (AD) | *TRPC6* | c.2641G>T p.(Glu881\*)(de novo) | Novel | LP (PS2, PM2, PP3) | Confirmed the clinical diagnosis |
| P265 | F | fam | UGLO | 15 y | Hemat. Normal Cr at 15 y | No at 15 y | – | Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps; #611773 | Het (AD) | *COL4A1* | c.1937del p.(Gly646Alafs\*156)(m) | Novel | P (PVS1, PM1, PM2, PP1, PP3) | Identified a specific diagnosis |
| P266 | F | sp | UGLO | Childhood | Prot, macroh, modified morphological red cells. Biopsy: abnormal expression of the alpha chains of collagen IV with a segmental distribution of the chain 5. | No |  | Alport syndrome 1, X-linked #301050 | Het (XL) | *COL4A5* | c.4298-20T>A p.(?) (de novo) | Novel | LP (PS2, PM2, BP4) | Identified a specific diagnosis |
| P267 | F | fam | UGLO | 27 y | Prot with no hemat (27 y). Biopsy: glomerulosclerosis | 28 y | Hearing loss (24 y) | Alport syndrome 2; #203780 | Homo (AR) | *COL4A4* | c.1205\_1369del p.(Gly402\_Ser456del)(p), c.1205\_1369del p.(Gly402\_Ser456del)(m) | Novel | P (PVS1, PM1, PM2, PM3, PP1) | Identified a specific diagnosis |
| P268 | M | sp | UGLO | 7 y | NR prot, NC. Biopsy: FSGS. Normal Cr at 8 y | No at 8 y | – | Nephrotic syndrome type 9; #615573 | Comp het (AR) | *COQ8B* | c.737G>A p.(Ser246Asn) (*in trans*) | Novel | LP (PM2, PM3, PP3, PP5) | Identified a specific diagnosis |
| c.748G>C p.(Asp250His) (*in trans*) | Novel | LP (PM2, PM3, PP3, PP4, BP1) |
| P269 | F | fam | UGLO | 5 y | Prot. CKD G3 at 27 y | No at 27 y | Cystic adenomatoid malformation of the lung (3 y), familial macrothrombocytopenia (5 y), iron-deficiency anemia and menorrhagia (19 y), bilateral sensorineural hearing loss (20 y) | Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss; #155100 | Het (AD) | *MYH9* | c.287C>T p.(Ser96Leu)(m) | [71] | LP (PM1, PM2, PP1, PP2, PP3, PP5) | Identified a specific diagnosis |
| P270 | M | sp | UGLO | 4 y | NR prot, hypercalciuria, glysosuria, slightly small kidneys, increased renal echogenicity, loss of corticomedullary differentiation. Hypercholesterolemia (4 y). | 8 y | Bilateral optic nerve dysplasia and anomalies of the optic disc (renal coloboma syndrome) (8 y) | Papillorenal syndrome; #120330 | Het (AD) | *PAX2* | c.493\_495delATTinsTCTTCCCTGA p.(Ile165Serfs\*18) (de novo) | [1] | P (PVS1, PS2, PM2, PP3, PP5) | Identified a specific diagnosis |
| P271 | M | sp | Nephrogenic diabetes insipidus | 19 y | Clinical features compatibles with Nephrogenic diabetes insipidus. Polyuria, polydipsia. | No at 19 y | – | Diabetes insipidus, nephrogenic; #304800 | Hemi (XL) | *AVPR2* | c.770dupG p.(Glu258\*) | Novel | P (PVS1, PM2, PM6) | Confirmed the clinical diagnosis |
| P272 | M | fam | Dent disease | Childhood | Clinical features compatible with Dent disease | U at 17 y |  | Dent disease 1; #300009 | Hemi (XL) | *CLCN5* | c.1546C>T p.(Arg516Trp)(m) | [72] | LP (PM1, PM2, PP1, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P273 | M | sp | Bartter syndrome | U | Clinical features compatible with Bartter syndrome | U at 31 y |  | Bartter syndrome type 3; #607364 | Comp het (AR) | *CLCNKB* | c.610G>A p.(Arg204Thr) | [73] | LP (PM2, PM3, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| c.(?\_-1)\_(\*1\_?)del p.(?) | [73] | P (PVS1, PM3, PP3, PP5) |
| P274 | F | U | Bartter syndrome | 1 y | Clinical features compatible with Bartter syndrome, hypokalemia. | U at 1 y |  | Bartter syndrome type 3; #607364 | Homo (AR) | *CLCNKB* | c.(?\_-1)\_(\*1\_?)del p.(?) | [73] | P (PVS1, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| P275 | M | U | Gitelman syndrome | 28 y | Clinical features compatible with Gitelman syndrome: Hypocalciuria, hyperuricemia, metabolic alkalosis, hypokalemia. CKD G3 at 33 y | No at 33 y |  | Bartter syndrome type 3; #607364 | Homo (AR) | *CLCNKB* | c.1270G>A p.(Gly424Arg)(p), c.1270G>A p.(Gly424Arg)(m) | [74] | LP (PM2, PM3, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P276 | F | fam | UTUB | 15 y | MSK, bilateral RL, UTI | No at 45 y | Hyperparathyroidism | Renal hypomagnesemia 3; #248250 | Homo (AR) | *CLDN16* | c.668A>G  p.(Asn223Ser)(p),  c.668A>G  p.(Asn223Ser)(m) | [75] | LP (PM2, PM3, PP2, PP3, PP5) | Identified a specific diagnosis |
| P277 | M | fam cons | Primary hyperoxaluria | 27 y | Bilateral nephrocalcinosis, high urinary oxalate, HBP. KFRT at 29 y | 29 y | Anemia, headache, neurological deterioration, chronic pelvic pain | Renal hypomagnesemia 5 with ocular involvement; #248190 | Homo (AR) | *CLDN19* | c.530T>G p.(Leu177Arg)(p), c.530T>G, p.(Leu177Arg)(m) | Novel | LP (PM2, PM3, PP1, PP3) | Reclassified the diagnosis |
| P278 | F | sp | UTUB | <18 y | Echogenic renal cortex, NC hypercalciuria, mh | No at 19 y |  | Renal hypomagnesemia 5 with ocular involvement; #248190 | Homo (AR) | *CLDN19* | c.59G>A p.(Gly20Asp)(p), c.59G>A p.(Gly20Asp)(m) | [76] | LP (PM2, PM3, PP3, PP5) | Identified a specific diagnosis |
| P279 | F | sp | Liddle syndrome | 6 mo | Polyuria, polydipsia, nephrocalcinosis, HBP, metabolic alkalosis | No at 15 y |  | Apparent mineralocorticoid excess; #218030 | Comp het (AR) | *HSD11B2* | c.983C>T p.(Ala328Val) | [77] | LP (PM2, PM3, PP3, PP5) | Reclassified the diagnosis |
| c.1020del p.(Gly341Alafs\*55) | Novel | P (PVS1, PM2, PM3) |
| P280 | M | fam | UTUB | 20 y | MSK, RL, tubular dysplasia, right tubular atrophy, right nephrectomy. CKD G2 at 49 y | No at 49 y | Epilepsy | Hyperuricemic nephropathy, familial juvenile 2; #613092 | Het (AD) | *REN* | c.145C>T p.(Arg49\*) | [78] | P (PVS1, PP3, PP4, PP5) | Identified a specific diagnosis |
| P281 | F | fam | UTUB | 22 y | MKS, UTI | No at 38 y | Infertility, gestational diabetes | Renal cysts and diabetes syndrome; #137920 | Het (AD) | *HNF1B* | c.(?\_-1)\_(\*1\_?)del p.(?) | [5] | P (PVS1, PM2, PP3, PP5) | Identified a specific diagnosis |
| P282 | M | fam | Primary renal tubular acidosis | 13 y | Renal tubular acidosis, NC (13 y), bilateral RL, bilateral RC. CKD G4 at 48 y | No at 48 y | – | Distal renal tubular acidosis; #179800 | Het (AD) | *SLC4A1* | c.1765C>T p.(Arg589Cys) | [79] | LP (PM1, PM2, PM5, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P283 | F | sp | Primary renal tubular acidosis | 3 y | Renal tubular acidosis, NC (3 y) HBP. Normal Cr at 29 y | No at 29 y | – | Distal renal tubular acidosis; #179800 | Het (AD) | *SLC4A1* | c.1765C>T p.(Arg589Cys) | [79] | LP (PM1, PM2, PM5, PP3, PP5, BP1) | Confirmed the clinical diagnosis |
| P284 | M | sp | Bartter syndrome | 8 y | Clinical features compatible with Bartter syndrome, metabolic alkalosis, NC. Normal Cr at 34 y | No at 34 y |  | Bartter syndrome, type 1; #601678 | Homo (AR) | *SLC12A1* | c.347G>A p.(Arg116His)(p), c.347G>A p.(Arg116His)(m) | [80] | LP (PM3, PP1, PP3, PP4, PP5, BS2) | Confirmed the clinical diagnosis |
| P285 | F | sp | Gitelman syndrome | Childhood | Clinical features compatible with Gitelman syndrome, hypokalemia, hypomagnesemia. Normal Cr at 32 y | No at 32 y |  | Gitelman syndrome; #263800 | Comp het (AR) | *SLC12A3* | c.1315G>A p.(Gly439Ser) | Novel | P (PS1, PS2, PM3, PP2, PP3) | Confirmed the clinical diagnosis |
| c.1367delT p.(Leu456Argfs\*36) | [74] | P (PVS1, PM2, PM3, PP3, PP5) |
| P286 | M | sp | Gitelman syndrome | 28 y | Clinical features compatible with Gitelman syndrome, metabolic alkalosis. Normal Cr at 32 y | No at 32 y |  | Gitelman syndrome; #263800 | Comp het (AR) | *SLC12A3* | c.1963C>T p.(Arg655Cys) | [81] | LP (PM2, PM3, PM5, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| c.1964G>A p.(Arg655His) | [82] | LP (PM3, PM5, PP2, PP3, PP5) |
| P287 | F | fam cons | Gitelman syndrome | 18 y | Clinical features compatible with Gitelman syndrome, hypocalciuria, hypokalemia, metabolic alkalosis, normocalcemia | No at 19 y |  | Gitelman syndrome; #263800 | Homo (AR) | *SLC12A3* | c.1180+1G>T p.(?)(p), c.1180+1G>T p.(?)(m) | [83] | P (PVS1, PS3, PM2, PM3, PP3, PP5) | Confirmed the clinical diagnosis |
| P288 | M | fam cons | UTUB | 1 y | NC, polyuria, polydipsia, hypercalciuria with normal blood calcium levels. Normal Cr at 24 y | No at 24 y |  | Hypophosphatemic rickets with hypercalciuria; #241530 | Homo (AR) | *SLC34A3* | c.1561dup p.(Leu521Profs\*72)(p), c.1561dup p.(Leu521Profs\*72)(m) | Novel | P (PVS1, PM2, PM3, PP5) | Identified a specific diagnosis |
| P289 | M | U | Cystinuria | 8 y | Bilateral RL, increased urinary excretion of cystine. Normal Cr at 24 y | No at 24 y |  | Cystinuria; #220100 | Comp het (AR) | *SLC3A1* | c.647C>T p.(Thr216Met) | [23] | LP (PM2, PP2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| c.1400T>C p.(Met467Thr) | [84] | LP (PM5, PP2, PP3, PP4, PP5, BS2) |
| P290 | M | sp | Cystinuria | 8 y | Bilateral RL, increased urinary excretion of cystine. Normal Cr at 24 y | No at 24 y |  | Cystinuria; #220100 | Homo (AR) | *SLC3A1* | c.647C>T p.(Thr216Met)(p), c.647C>T p.(Thr216Met) (m) | [23] | LP (PM2, PM3, PP2, PP3, PP5) | Confirmed the clinical diagnosis |
| P291 | M | sp | Cystinuria | U | Bilateral RL, increased urinary excretion of cystine, two RC | No at 20 y |  | Cystinuria; #220100 | Comp het (AR) | *SLC3A1* | c.647C>T p.(Thr216Met) | [23] | LP (PM2, PP2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| c.1400T>C p.(Met467Thr) | [84] | LP (PM5, PP2, PP3, PP4, PP5, BS2) |
| P292 | M | U | Cystinuria | 3 y | Bilateral NC, increased urinary excretion of cystine | No at 3 y |  | Cystinuria; #220100 | Homo (AR) | *SLC7A9* | c.997C>T p.(Arg333Trp)(p), c.997C>T p.(Arg333Trp)(m) | [85] | LP (PM2, PM3, PM5, PP3, PP5) | Confirmed the clinical diagnosis |
| P293 | M | fam | Cystinuria | 25 y | RL, increased urinary excretion of cystine, atrophic right kidney. CKD G1 at 29 y | No at 29 y |  | Cystinuria; #220100 | Comp het (AR) | *SLC7A9* | c.368C>T p.(Thr123Met) | [85] | LP (PM3, PP2, PP3, PP4, PP5) | Confirmed the clinical diagnosis |
| c.1397C>A p.(Ser466\*) | Novel | P (PVS1, PM2, PM3, PP3, PP4) |
| P294 | F | fam | Cystinosis | U | Metabolic acidosis, intra-lysosomal cystine accumulation, polyuria, bilateral RL. Normal Cr at 22 y | No at 22 y | Ocular involvement | Cystinosis; #219800 | Comp het (AR) | *CTNS* | c.18\_21del p.(Thr7Phefs\*7) | Novel | P (PVS1, PP5) | Confirmed the clinical diagnosis |
| c.519\_520del p.(Tyr173\*) | Novel | P (PVS1, PM1, PM2, PP5) |
| P295 | F | sp | Nephrogenic diabetes insipidus | Childhood | Clinical features compatibles with Nephrogenic diabetes insipidus. Polyuria, polydipsia | No at 36 y |  | Nephrogenic diabetes insipidus; #125800 | Het (AD) | *AQP2* | c.797\_\*17del p.(Pro266\_Ala271delins(56))(de novo) | Novel | P (PVS1, PS2, PM2, PP3) | Confirmed the clinical diagnosis |
| P296 | F | fam | ADTKD | 2 y | Echogenic kidneys, loss of corticomedullary differentiation, renal cortex thinning, hyperuricemia. CKD G3 at 14 y | No at 14 y |  | Hyperuricemic nephropathy, familial juvenile 2; #613092 | Het (AD) | *REN* | c.58T>C p.(Cys20Arg)(m) | [86] | LP (PM2, PP1, PP3, PP4, PP5, BP1) | Confirmed the clinical diagnosis |
| P297 | M | U | AS | U | KFRT at 31 y | 31 y | Hearing loss (15 y) | Hyperuricemic nephropathy, familial juvenile 1; #162000 | Het (AD) | *UMOD* | c.189C>G p.(Cys63Trp) | [HGMD](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=24577984&dopt=Abstract) db | LP (PM2, PP2, PP3, PP4, PP5) | Reclassified the diagnosis |
| P298 | F | fam | ADTKD | 14 y | Hyperuricemia. Normal Cr at 14 y | No at 14 y |  | Medullary cystic kidney disease 1, #174000 | Het (AD) | *MUC1* | c.428dup p.(Ala144Serf\*86) | [87] | P (PVS1, PP4, PP5) | Confirmed the clinical diagnosis |
| P299 | M | fam | ADTKD | 18 y | Biopsy: unspecific. KFRT at 19 y | 19 y |  | Medullary cystic kidney disease 1, #174000 | Het (AD) | *MUC1* | c.428dup p.(Ala144Serf\*86)(m) | [87] | P (PVS1, PP1, PP4, PP5) | Confirmed the clinical diagnosis |
| P300 | F | fam | ADTKD | 26 y | Small, echogenic, asymmetric kidneys (26 y). Biopsy: 10% Interstitial fibrosis and tubular atrophy. CKD G2 at 37 y | No at 37 y | – | Medullary cystic kidney disease 1, #174000 | Het (AD) | *MUC1* | c.428dup p.(Ala144Serf\*86) | [87] | P (PVS1, PP4, PP5) | Confirmed the clinical diagnosis |

Abbreviations: AD, autosomal dominant; ADPKD, autosomal dominant polycystic kidney disease; AH, anhydramnios; AML, angiomyolipoma; AR, autosomal recessive; ARPKD, autosomal recessive polycystic kidney disease; AS, Alport syndrome; CD, Caroli disease; CHF, congenital hepatic fibrosis; CIP, complex inheritance patterns; CKD, chronic kidney disease; cons, consanguineous; Cr, creatinine; db, database; DPM, ductal plate malformation; EM, electron microscopyKFRT; F, female; FA, fatty acids; fam, familial; FSGS, focal segmental glomerulosclerosis; GBM, glomerular basement membrane; GGT, gamma-glutamyltransferase; GN, glomerulonephritis; HBP, high blood pressure; HC, hepatic cysts; hemat, hematuria; het, heterozygosis; homo, homozygosis; IF, immunofluorescence; KFRT, kidney failure with replacement therapy; LM, light microscopy; LP, likely pathogenic; M, male; m, maternal; macroh, macrohematuria; MAU, microalbuminuria; MCD, minimal change disease; MCDK, multicystic dysplastic kidney; Mh, microhematuria; mo, months; MRI, magnetic resonance imaging; MSK, medullary sponge kidney; NC, nephrocalcinosis; NL, nephrolithiasis; NPHP-RC, nephronophthisis-related ciliopathies; NR, nephrotic range; NS, nephrotic syndrome; OH, oligohydramnios; P, pathogenic; p, paternal; prot, proteinuria; RA, renal agenesis; RC, renal cysts; RD, renal dysplasia; Ref, reference; RH, renal hypoplasia; RHD, renal hypodysplasia; RL, renal lithiasis; sp, sporadic; T1DM, type 1 diabetes mellitus; T2DM, type 2 diabetes mellitus; TOP, termination of pregnancy; TSC, tuberous sclerosis; U, unknown; UGLO, unspecified glomerulopathy; UPKD, unspecified polycystic disease; UTI, urinary tract infections; UTUB, unspecified tubulopathy; VUR, vesicoureteral reflux; VUS, variant of unknown significance; XL, X-linked; y, years.

Supplementary Table 3. Clinical and genetic data of patients in whom VUS were identified.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient** | **Sex** | **fam/sp** | **Clinical diagnosis** | **Age at CKD onset (death)** | **Renal presentation (onset, y)** | **Age at KFRTKFRT onset (y)** | **Extrarenal alterations** | **Potential genetic diagnosis #OMIM** | **Zygosity (inheritance)** | **Gene** | **Disease-causing variant/s (origin)** | **Ref** | **ACMG (HGMD)** |
| P301 | M | sp | CAKUT | Prenatal | Bilateral MCDK (prenatal), RH. CKD G3 at 2 y | No at 2 y |  | Renal cysts and diabetes syndrome, #137920 | Het (AD) | *HNF1B* | c.1501A>G p.(Met501Val)(p) | Novel | VUS (PM2, PP2, PP3) |
| P302 | F | sp | CAKUT | Birth | Unilateral RA, RC | No | Rokitansky syndrome | Orofaciodigital syndrome I, #311200 | Het (AD) | *OFD1* | c.2927A>C p.(Lys976Thr)(m) | Novel | VUS (PM1, PM2, PP3, BP1) |
| P303 | M | fam | CAKUT | >18 y | RH, unilateral RC | U |  | Alagille syndrome 1, #118450 | Het (AD) | *JAG1* | c.2429C>T p.(Pro810Leu) | [88] | VUS (PM2, PP2, PP3) |
| P304 | F | fam | CAKUT | Prenatal | Bilateral MCDK, Mh, polyuria, polydipsia. CKD G3 at 13 y | No at 13 y |  | \* 604994 | Het (AD) | *SIX2* | c.316G>A p.(Val106Met) | Novel | VUS (PM2, PP3) |
| P305 | M | sp | CAKUT | Birth | Bilateral urinary tract dilation, right RC | No at 3 y |  | \* 618281 | Comp het (AR) | *VWA2* | c.1096G>A p.(Val366Met)(m) | Novel | VUS (PP3, BS1) |
| c.1161C>A p.(Tyr387\*)(p) | Novel | VUS (PM2, PP3) |
| P306 | M | fam | CAKUT | Prenatal | Bilateral urinary tract dilation, RC | No at 9 y |  | Congenital anomalies of kidney and urinary tract 2, #143400 | Het (AD) | *TBX18* | c.60C>G p.(Phe20Leu)(p) | Novel | VUS (PM2, PP3) |
| P307 | M | sp | UPKD | Prenatal | Bilateral RC, prot, hemat, polyuria, HBP. CKD G3 at 55 y | No at 55 y | Polydactyly, growth retardation, left hearing loss, skeletal syndrome, hypospadia, clear cell renal cell carcinoma | \* 602430 | Het (AD) | *ROBO1* | c.3724C>G p.(Pro1242Ala) | Novel | VUS (PM6, PP3) |
| P308 | M | sp | CAKUT | Birth | Bilateral VUR (birth), left RC. NS post-kidney transplant | 9 y |  | \* 164761 | Het (AD) | *RET* | c.1529C>T p.(Ala510Val) | -[89] | VUS (PM6, PP2, PP3, PP5, BS1) |
| P309 | M | fam | ADPKD | Childhood | Renal ultrasound compatible with ADPKD | No |  | Polycystic kidney disease 1; #173900 | Het (AD) | *PKD1* | c.2139C>G p.(Asp713Glu) | Novel | VUS (PM2, PP3, PP4) |
| P310 | M | fam cons | NPHP-RC | Childhood | Left ectopic kidney, RC | U at 2 y | Polydactyly, hypospadia, butterfly vertebra | Alagille syndrome 1, #118450 | Het (AD) | *JAG1* | c.3638G>A p.(Arg1213Gln) | [90] | VUS (PP2, PP3, PP5, BS2) |
| P311 | F | fam | UPKD | Prenatal | Bilateral RC. Echogenic kidneys | No |  | \* 164761 | Het (AD) | *RET* | c.2601G>T p.(Glu867Asp) (p) | Novel | VUS (PM1, PP2, PP3, BS1) |
| P312 | M | sp | NPHP-RC | Childhood | Medullary cystic disease. Biopsy (19 y): cortical atrophy, moderate interstitial tubule injury and sclerosis in 30-40% of glomeruli. CKD G2 at 30 y. | No |  | Hyperuricemic nephropathy, familial juvenile 2, #613092 | Het (AD) | *REN* | c.49T>C p.(Trp17Arg) | Novel | VUS (PM2, PM6, PP3, BP1) |
| P313 | F | fam | AS | >18 y | Mh, MAU. Biopsy: slight non-specific changes. CKD G2I | No |  | Nephropathy due to CFHR5 deficiency, #614809 | Het (AD) | *CFHR5* | c.486dupA p.(Glu163Argfs\*35) | [91] | VUS (PP5, BS1) |
| P314 | M | fam | NS | 15 y | NR prot, bilateral RH, HBP. CKD at 15 y | No | Congenital hypothyroidism | Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, #218700 | Homo(AD) | *PAX8* | c.601+1G>T p.(?) | Novel | VUS (PM2, PP3) |
| P315 | M | sp | NS | 9 y | Prot, Mh. Biopsy: FSGS | No |  | Glomerulosclerosis, focal segmental, 2 #603965 | Het (AD) | *TRPC6* | c.2392G>C p.(Asp798His) | Novel | VUS (PM6, PP3, BP1) |
| P316 | F | U | NS | U | NS prot. Biopsy: FSGS. CKD at 13 y | No at 13 y | Hearing loss, retinal arterial alterations | Glomerulosclerosis, focal segmental, 2 #603965/Glomerulosclerosis, focal segmental, 1 #603278 | Het (AD) | *TRPC6* | c.1309C>T p.(Arg437Cys) | Novel | VUS (PP3, BP1) |
| Het (AD) | *ACTN4* | c.499\_543del p.(Glu167\_Lys181del) | Novel | VUS (PM2, PM4, BP4) |
| P317 | M | sp | NS | Childhood | FSGS (childhood). CKD at 47 y | No |  | \* 610586 | Het (AD) | *ARHGAP24* | c.1421C>T p.(Thr474Met) | Novel | VUS (PM6, PP3, BS1) |
| P318 | F | sp | UGLO | 5 y | Prot, hemat. Normal kidney biopsy. Normal Cr at 42 y | No at 42 y | Hearing loss (28 y), pain in the joints and muscles | Alport syndrome 1, X-linked #301050/Megaloblastic anemia-1, Finnish type #261100 | Het (XL) | *COL4A5* | c.4264C>T p.(Arg1422Cys) | [92] | VUS (PM1, PP2, PP3, PP5, BS2) |
| Homo (AR) | *CUBN* | c.7968\_7969delGCinsTGTTATACCTTATATAA p.(Leu2656\_Pro2657delinsPheValIleProTyrIleThr) | Novel | VUS (PM2, PM4, BP4) |
| P319 | M | fam | UGLO | 10 y | Macroh, prot, MAU | No |  | Complement factor H deficiency #609814 | Het (AD) | *CFH* | c.197G>A p.(Cys66Tyr) | [93] | VUS (PM2, PP3, PP5, BP1) |
| P320 |  | sp | ARPKD | Prenatal | Enlarged echogenic kidneys, RC, HBP. | No | Echogenic liver | Polycystic kidney disease 4 with or without hepatic disease; #263200 | Het (AD) | *PKHD1* | c.5410C>T p.(Arg1804Cys)(m) | [31] | VUS (PP2, PP5, BP4) |
| c.9830-8A>G p.(?) | Novel | VUS (PM2, BP4) |
| P321 | F | fam | ADPKD | 26 y | Normal-sized kidneys, bilateral RC, mild prot. Normal Cr at 28 y | No at 28 y |  | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.1796G>A p.(Gly599Asp) | Novel | VUS (PM2, PP3, PP4) |
| P322 | F | fam | ADPKD | 28 y | Bilateral RC. Normal Cr at 29 y | No at 29 y | – | Polycystic kidney disease 2; #613095 | Het (AD) | *PKD2* | c.1639\_1647dup p.(Gly547\_Pro549dup) | Novel | VUS (PM2, PM4, PP4, BP4) |

Abbreviations: AD, autosomal dominant; ADPKD, autosomal dominant polycystic kidney disease; AR, autosomal recessive; ARPKD, autosomal recessive polycystic kidney disease; AS, Alport syndrome; CKD, chronic kidney disease; cons, consanguineous; KFRT, end-stage kidney disease; F, female; fam, familial; FSGS, focal segmental glomerulosclerosis; HBP, high blood pressure; hemat, hematuria; het, heterozygosis; homo, homozygosis; IKD, inherited kidney diseases; KFRT, kidney failure with replacement therapy; M, male; m, maternal; macroh, macrohematuria; MAU, microalbuminuria; MCDK, multicystic dysplastic kidney; Mh, microhematuria; NPHP-RC, nephronophthisis-related ciliopathies; NS, nephrotic syndrome; p, paternal; prot, proteinuria; RA, renal agenesis; RC, renal cysts; Ref, reference; RH, renal hypoplasia; sp, sporadic; TOP, termination of pregnancy; U, unknown; UGLO, unspecified clinical diagnosis of glomerular IKD; UPKD, unspecified clinical diagnosis of cystic IKD; VUR, vesicoureteral reflux; VUS, variant of unknown significance; y, years.

**Supplementary Material and Methods**

**Bioinformatics Analysis**

All the bioinformatics tools used in this study were run using default settings unless otherwise stated. Reads were aligned to the human reference genome hg19 using the Burrows-Wheeler Aligner [94]. Alignments were converted to BAM format and sorted using samtools [95]. Local realignment around potential insertions/deletions and SNP clusters and base-quality recalibration was performed using the GATK4 software [96]. GATK4 HaplotypeCaller [97] was used for variant calling. The resulting variants were subsequently quality filtered using GATK VariantFiltration (parameters MQ < 0.0 || QUAL <25.0 || QD <0.0 || DP <5 || GQ <15). CoNVaDING (Copy Number Variation Detection In Next-generation sequencing Gene panels) was used to identify large indels and structural variants by read-depth analysis [98].

**Supplementary Data**

**Logistic regression model of predictive clinical features**

Logistic regression predicting DX\_positive : YES vs NO

adj. OR(95%CI) P(Wald's test) P(LR-test)

Disease\_group: ref.=CAKUT < 0.001

PKD 7.31 (3.93,13.63) < 0.001

GLO 3.54 (1.86,6.73) < 0.001

TUB 6.62 (2.48,17.69) < 0.001

ADTKD 3.32 (0.56,19.76) 0.188

Familiar: FAM vs NO\_FAM 2.38 (1.51,3.75) < 0.001 < 0.001

Extrarenal: EXTRA vs NO\_EXTRA 1.67 (1.04,2.7) 0.036 0.034

Log-likelihood = -240.2733

No. of observations = 418

AIC value = 494.5467

Model accuracy (proportion of correctly classified observations) was estimated at 74.16% based on training data.

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