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Kidney gene panel

Genetic Conditions and Genes

385 genes associated with monogenic disorders linked to kidney disease

Condition Name	Genes
17-Alpha-Hydroxylase 17/20 -Lyase Deficiency	*CYP17A1*
5-Oxoprolinase Deficiency	*OPLAH*
Acroosteolysis, Dominant	*NOTCH2*
Acro-Renal-Ocular Syndrome	*SALL4*
Adenine Phosphoribosyltransferase Deficiency	*APRT*
Alagille Syndrome, Type 1, 2	*JAG1* *NOTCH2*
Alkaptonuria	*HGD*
Alport Syndrome	*COL4A3* *COL4A4* *COL4A5*
Alstrom Syndrome	*ALMS1*
Amelogenesis Imperfecta, Type 1G, 2A3	*FAM20A* *WDR72*
Amyloidosis	*APOA1* *APOC2* *B2M* *GSN* *LYZ* *TTR*
Antley-Bixler Syndrome	*FGFR2*
Apert Syndrome	*FGFR2*
Apparent Mineralocorticoid Excess	*HSD11B2*
Arthrogryposis, Cleft Palate, Craniosynostosis, and Impaired Intellectual Development	*PPP3CA*
Atypical hemolytic uremic syndrome (aHUS)	*CFH* *CFHR5* *CFI* *THBD* *DGKE* *C3* *PLG*
Autoinflammation, Antibody Deficiency, and Immune Dysregulation Syndrome	*PLCG2*
Axenfeld-Rieger Syndrome, Type 3	*FOXC1*
Baraitser-Winter Syndrome, Type 1	*ACTB*
Bardet-Biedl Syndromes, Type 1-12, 14-18, 21	*ARL6* *BBIP1* *BBS1* *BBS10* *BBS12* *BBS2* *BBS4* *BBS5* *BBS7* *BBS9* *C8ORF37* *CEP290* *LZTFL1* *MKKS* *SDCCAG8* *TRIM32* *TTC8* *WDPCP*
Bartter Syndrome, Type 1, 2, 3/4B, 4a	*BSND* *CLCNKB* *KCNJ1* *SLC12A1*
Beckwith-Wiedemann Syndrome	*CDKN1C* *NSD1*
Beta-Hemoglobinopathies	*HBB*
Bifid Nose with or without Anorectal and Renal Anomalies	*FREM1*
Birt-Hogg-Dube Syndrome	*FLCN*
Bladder Dysfunction, Autonomic, with Impaired Pupillary Reflex and Congenital Anomalies of the Kidney and Urinary Tract (CAKUT)	*CHRNA3*
Branchiooculofacial Syndrome	*TFAP2A*
Branchio-Oto-Renal Syndrome, Type 1, 2	*SIX1* *EYA1* *SIX5*
Burn-McKeown Syndrome	*TXNL4A*
C3 Glomerulopathy	*C3* *CFHR5* *CFI* *CFH* *DGKE*
Congenital Anomalies of the Kidney and Urinary Tract (CAKUT)	*BMP4* *BMP7* *CHD1L* *CRKL* *GDNF* *GREM1* *ROBO2*
Cardiofaciocutaneous Syndrome	*KRAS*
Carnitine Palmitoyltransferase 2 Deficiency	*CPT2*
Cataract, Juvenile, with Microcornia and Glucosuria	*SLC16A12*
Cenani-Lenz Syndactyly Syndrome	*LRP4*
Cerebral Creatine Deficiency Syndrome 3	*GATM*
Pulmonary Venoocclusive Disease 1	*INF2*
CHARGE Syndrome	*CHD7* *SEMA3E*
CHILD Syndrome	*NSDHL*
Chondrodysplasia Punctata	*EBP*
Ciliopathies, RPGRIP1L-Related	*RPGRIP1L*
COACH Syndrome	*TMEM67*
Coenzyme Q10 Deficiency, Primary, Type 1, 2, 3, 6	*COQ2* *COQ6* *PDSS1* *PDSS2*

Condition Name	Genes
Combined Oxidative Phosphorylation Deficiency, Type 11	*RMND1*
Complement Component 5 Deficiency	*C5*
Complement Factor H, I Deficiency	*CFH* *CFI*
Congenital Adrenal Hyperplasia due to 3-Beta-Hydroxysteroid Dehydrogenase Deficiency and 11-Beta-Hydroxylase Deficiency	*CYP11B1* *HSD3B2*
Congenital Adrenal Hypoplasia with Hypogonadotropic Hypogonadism	*NR0B1*
Congenital Anomalies of the Kidney and Urinary Tract Syndrome with or without Hearing Loss, Abnormal Ears, or Developmental Delay (CAKUTHED)	*PBX1*
Congenital Disorder of Glycosylation, Type 1A, 1H, 1K, 1L	*ALG1* *ALG8* *ALG9* *PMM2*
Congenital Hyperinsulinism	*KCNJ11*
Cornelia de Lange Syndrome	*SMC1A*
Corticosterone Methyloxidase Deficiency	*CYP11B2*
Cranioectodermal Dysplasia, Type 1, 3	*IFT122* *IFT43* *WDR19*
Currarino Syndrome	*MNX1*
CYP11A1 Deficiency Syndrome	*CYP11A1*
Cystinosis	*CTNS*
Cystinuria	*SLC3A1* *SLC7A9*
Dent Disease	*CLCN5* *OCRL*
Denys-Drash Syndrome	*WT1*
Diabetes Insipidus, Nephrogenic	*AQP2* *AVPR2*
Diabetes insipidus, Neurohypophyseal Diabetes Mellitus	*AVP* *GCK* *HNF1A* *PAX4*
Diabetes Mellitus, Juvenile-Onset	*PCBD1*
Diabetes Mellitus, Neonatal, with Congenital Hypothyroidism	*GLIS3*
Diabetes Mellitus, Noninsulin-Dependent	*SLC2A2*
Diamond-Blackfan Anemia, Type 1, 3-11	*RPL11* *RPL26* *RPL35A* *RPL5* *RPS10* *RPS17* *RPS19* *RPS24* *RPS26* *RPS7*
Donnai-Barrow Syndrome	*LRP2*
Duane-Radial Ray Syndrome	*SALL4*
Eagle-Barrett Syndrome	*CHRM3*
Ectrodactyly, Ectodermal Dysplasia, and Cleft Lip/Palate Syndrome 3	*TP63*
Encephalocraniocutaneous Lipomatosis	*FGFR1*
Epilepsy, Progressive Myoclonic, 4 with or without Renal Failure	*SCARB2*
Epstein Syndrome	*MYH9*
Fabry Disease	*GLA*
Familial Cold-Induced Inflammatory Syndrome, Type 1, 3	*NLRP3* *PLCG2*
Familial Dysautonomia, Hereditary Sensory and Autonomic Neuropathy Type 3	*ELP1*
Familial Mediterranean Fever	*MEFV*
Fanconi Anemia, Group A, B, C, D2, E, F, G, I, L, M, N, O, P	*FANCA* *FANCB* *FANCC* *FANCD2* *FANCE* *FANCF* *FANCG* *FANCI* *FANCL* *FANCM* *PALB2* *RAD51C* *SLX4*
Fanconi Renotubular Syndrome, Type 1, 2, 4	*GATM* *HNF4A* *SLC34A1*
Fanconi-Bickel Syndrome	*SLC2A2*
Fechtner Syndrome	*MYH9*
Feingold Syndrome	*MYCN*
Floating-Harbor Syndrome	*SRCAP*
Focal Segmental Glomerulosclerosis, Type 2, 4, 5, 6, 7	*ACTN4* *ALG13* *APOL1* *CD2AP* *INF2* *MYO1E* *PAX2* *PMM2* *SYNPO* *TRPC6*
Fraser Syndrome	*FRAS1* *FREM2* *GRIP1* *WT1*
Galloway-Mowat Syndrome	*TP53RK* *WDR73*
Genitopatellar Syndrome	*KAT6B*
Gillessen Kaesbach Nishimura Syndrome (GIKANIS)	*ALG9*
Gitelman Syndrome	*SLC12A3*
Glomerulocystic Kidney Disease	*HNF1B* *REN* *UMOD*
Glomerulopathy with Fibronectin Deposits 2	*FN1*
Glucocorticoid Resistance, Generalized	*NR3C1*
Glycogen Storage Disease, Type 1A, 1B/1C, 11	*G6PC* *LDHA* *SLC37A4*
Golabi-Behmel Syndrome, Type 2	*OFD1*
Hajdu-Cheney Syndrome	*NOTCH2*
Hand-Foot-Uterus Syndrome	*HOXA13*
Hartnup Disorder	*SLC6A19*
Hereditary Angiopathy with Nephropathy, Aneurysms and Muscle Cramps (HANAC)	*COL4A1*
Hereditary Renal Amyloidosis	*FGA*
Hermansky-Pudlak Syndrome 1	*HPS1*
Hyperaldosteronism, Familial, Type 1, 2, 3, 4	*CACNA1H* *CLCN2* *CYP11B1* *CYP11B2* *KCNJ5*
Hypercalcemia Infantile, Type 1, 2	*CYP24A1* *SLC34A1*
Hypercalciuria, Hypophosphatemic Rickets	*ADCY10* *SLC34A3*
Hyperglycinuria	*SLC6A19* *SLC36A2* *SLC6A20*
Hyperinsulinemic Hypoglycemia, Diabetes Mellitus	*ABCC8*
Hyperparathyroidism 2	*CDC73*
Hyperphenylalaninemia, BH4-Deficient	*PCBD1*
Hyperphosphatemic Familial Tumoral Calcinosis	*GALNT3*
Hyperphosphatemic Tumoral Calcinosis	*KL*
Hyperprolinemia, Type 1	*PRODH*
Hyperuricemic Nephropathy	*REN* *SARS2* *UMOD*
Hypocalciuric Hypercalcemia, Familial	*AP2S1* *CASR* *GNA11*
Hypocalcemia	*CASR*
Hypogonadotropic hypogonadism 1	*ANOS1*
Hypokalemic Periodic Paralysis, Type 1, 2	*CACNA1S* *SCN4A*

Condition Name	Genes
Hypomagnesemia Type 1-6	*CLDN16* *CLDN19* *CNNM2* *CNNM2* *EGF* *FXYD* *KCNA1* *TRPM6*
Hypoparathyroidism, Familial Isolated	*GCM2*
Hypoparathyroidism, Sensorineural Deafness, and Renal Dysplasia	*GATA3*
Hypophosphatasia	*ALPL*
Hypophosphatemic Rickets	*CLCN5* *DMP1* *ENPP1* *FGF23* *PHEX* *VDR*
Hypotrichosis-Lymphedema-Telangiectasia-Renal Defect Syndrome	*SOX18*
Hypouricemia, Renal, Type 1, 2	*SLC22A12* *SLC2A9*
IMAGE Syndrome	*CDKN1C*
Interstitial Lung Disease with Nephrotic Syndrome and Epidermolysis Bullosa	*ITGA3*
Interstitial Nephritis, Karyomegalic	*FAN1*
IPEX Syndrome	*FOXP3*
Isolated Renal Hypoplasia	*PAX2*
IVIC Syndrome	*SALL4*
Joubert Syndrome Type 3, 4, 5, 6, 10 with Oculoreneal Defect	*AHI1* *CEP290* *NPHP1* *OFD1* *TMEM67*
Junctional Epidermolysis Bullosa-Pyloric Atresia Syndrome	*ITGA6* *ITGB4*
Kallmann Syndrome	*ANOS1* *FGFR1* *PROKR2*
Kelley-Seegmiller Syndrome	*HPRT1*
Koolen-De Vries Syndrome	*KANSL1*
LADD Syndrome	*FGF10* *FGFR2*
Lesch-Nyhan Syndrome	*HPRT1*
Liddle Syndrome, Type 1, 2	*SCNN1B* *SCNN1G*
Lipodystrophy Congenital Generalized, Type 1, 2, 3	*AGPAT2* *BSCL2* *CAV1*
Lipodystrophy, Familial Partial, Type 7	*CAV1*
Lipoid Adrenal Hyperplasia	*STAR*
Lipoprotein Glomerulopathy	*APOE*

Condition Name	Genes
LMNA-Related Disorders	*LMNA*
Lowe Syndrome	*OCRL*
LRP5-Related Disorders	*LRP5*
Lymphedema-Distichiasis Syndrome with Renal Disease and Diabetes Mellitus	*FOXC2*
Lysinuric Protein Intolerance	*SLC7A7*
Mandibulfacial Dysostosis with Alopecia	*EDNRA*
Maturity Onset Diabetes of the Young (MODY), Type 2-4, 6-9, 11	*BLK* *CEL* *GCK* *HNF1A* *KLF11* *NEUROD1* *PAX4* *PDX1*
Meckel Syndrome, Type 3,4,7	*CEP290* *NPHP3* *TMEM67*
Medullary Cystic Kidney Disease	*UMOD*
Megaloblastic Anemia 1	*CUBN* *AMN*
Metaphyseal Chondrodysplasia, Murk Jansen Type	*PTH1R*
Methylmalonic Acidemia, Type mut0	*MUT*
Methylmalonic Aciduria and Homocystinuria, Type cblC	*MMACHC*
Mevalonic Aciduria	*MVK*
Microphthalmia, syndromic 6	*BMP4*
Mitochondrial Complex 3 Deficiency	*BCS1L* *UQCC2*
Mitochondrial Complex 4 Deficiency	*APOPT1 (COA8)* *COX10* *COX14* *COX20* *COX6B1* *COX8A* *FASTKD2* *PET100* *SCO1* *TACO1*
Mitochondrial DNA Depletion Syndrome 8A	*RRM2B*
Muckle-Wells Syndrome	*NLRP3*
Mullerian Aplasia and Hyperandrogenism	*WNT4*
Multicentric Carpotarsal Osteolysis with or without Nephropathy	*MAFB*
Myoglobinuria, Acute Recurrent	*LPIN1*
Nail-Patella Syndrome	*LMX1B*
Nephrolithiasis	*CLCN5* *FGF23* *SLC26A1*

Condition Name	Genes
Nephrolithiasis/Osteoporosis, Hypophosphatemic 1 and 2	*SLC34A1* *SLC9A3R1*
Nephronophthisis 1–4, 7, 9, 11–13, 15, 16, 19	*ANKS6* *CEP164* *DCDC2* *GLIS2* *INVS* *NEK8* *NPHP1* *NPHP3* *NPHP4* *SLC41A1* *TMEM67* *TTC21B* *WDR19* *XPNPEP3*
Nephropathy due to CFHR5 Deficiency	*CFHR5*
Nephropathy with Pretibial Epidermolysis Bullosa and Deafness	*CD151*
Nephrotic Syndrome	*DLC1* *ITSN2* *KANK1* *TNS2*
Nephrotic Syndrome Type 2-7, 15, Steroid sensitive, Congenital	*DGKE* *LAMB2* *MAGI2* *NPHS1* *NPHS2* *PLCE1* *PLCG2* *PTPRO* *WT1*
Neurofibromatosis, Type 1	*NF1*
Noonan Syndrome	*BRAF* *PTPN11*
Norum Disease	*LCAT*
Obesity	*MC4R* *UCP3*
Ochoa Syndrome	*HPSE2*
Orofaciodigital Syndrome 6, I	*CPLANE1* *OFD1*
Pallister-Hall Syndrome	*GLI3*
Papillorenal Syndrome	*PAX2*
Permanent Neonatal Diabetes Mellitus	*INS* *KCNJ11*
Pfeiffer Syndrome Type 3	*FGFR2*
Phosphoglycerate Kinase 1 Deficiency	*PGK1*
Phosphoribosylpyrophosphate Synthetase Superactivity	*PRPS1*
Pierson Syndrome	*LAMB2*
Plasminogen Deficiency, Type 1	*PLG*
Polycystic Kidney and/or Polycystic Liver Disease	*ALG9* *GANAB*

Condition Name	Genes
Polycystic Kidney Disease (PKD), 1 and 2, Autosomal Dominant	*PKD1* *PKD2*
Polycystic Kidney Disease, Autosomal Recessive	*PKHD1*
Polycystic Liver Disease (PLD), 1–3	*ALG8* *PRKCSH* *SEC63*
Primary Hyperoxaluria Type 1, 2, 3	*AGXT* *GRHPR* *HOGA1*
Prune Belly Syndrome	*CHRM3*
Pseudohypoaldosteronism Type I, Autosomal Dominant Hypertension, Early-Onset	*NR3C2*
Pseudohypoaldosteronism, Type 1, 1B, 2B, 2C, 2D, 2E	*CUL3* *KLHL3* *SCNN1A* *SCNN1B* *SCNN1G* *WNK1* *WNK4*
Pseudohypoparathyroidism Type 1B	*GNAS* *STX16*
Pseudoxanthoma Elasticum Arterial Calcification, Generalized, of Infancy, 2	*ABCC6*
Pulmonary Hypertension, Familial Primary with or without Hereditary Hemorrhagic Telangiectasia	*BMPR2*
Pulmonary Hypertension, Primary 2, 3, 4	*CAV1* *KCNK3* *SMAD9*
Pulmonary venoocclusive disease 1	*BMPR2*
Renal Agenesis	*RET*
Renal Cysts and Diabetes Syndrome	*HNF1B*
Renal Dysplasia, Cystic	*BICC1*
Renal Glucosuria	*SLC5A1* *SLC5A2*
Renal Hypertension	*SLC12A2*
Renal Hypodysplasia	*SIX2* *UPK3A*
Renal Tubular Acidosis	*ATP6V0A4* *ATP6V1B1* *CA2* *FOXI1* *SLC4A1* *SLC4A4* *WDR72*
Renal Tubular Disease	*NEDD4L*
Renal Tubular Dysgenesis	*ACE* *AGT* *AGTR1* *REN*
Renal-Hepatic-Pancreatic Dysplasia 2	*NEK8*
Rickets due to Defect in Vitamin D 25-hydroxylation	*CYP2R1*
Robinow Syndrome	*ROR2* *WNT5A*
Rubinstein-Taybi Syndrome, Type 1	*CREBBP*
Scalp-Ear-Nipple Syndrome	*KCTD1*
Schimke Immunoosseous Dysplasia	*SMARCAL1*
Seizures, Sensorineural Deafness, Ataxia, Mental Retardation, and SeSAME Syndrome	*KCNJ10*
Senior-Loken Syndrome, Type 4, 5, 6, 7	*CEP290* *INVS* *IQCB1* *NPHP1* *NPHP3* *NPHP4* *SDCCAG8* *WDR19*
SERKAL Syndrome	*WNT4*
Short Stature, Microcephaly, and Endocrine Dysfunction	*XRCC4*
Short-Rib Thoracic Dysplasia 5, 9, 10	*IFT140* *IFT172* *WDR19*
Simpson-Golabi-Behmel Syndrome, Type 1	*GPC3*
Smith-Lemli-Opitz Syndrome	*DHCR7*
Sneddon Syndrome	*ADA2*
Sotos Syndrome 1	*NSD1*
Steroid-Resistant Nephrotic Syndrome	*ALG13* *SYNPO*
Steroid-Resistant Nephrotic Syndrome with Focal Segmental Hyalinosis	*ARHGAP24*
Sucrase-Isomaltase Deficiency	*SI*
Susceptibility to End-Stage Renal Disease	*APOL1*
Susceptibility to Gout	*ABCG2*
Susceptibility to Hypertension	*STK39*
Systemic Lupus Erythematosus 16	*DNASE1L3*
Thrombophilia due to Thrombomodulin Defect	*THBD*
Thrombotic Thrombocytopenic Purpura, Familial	*ADAMTS13*
Townes-Brocks Syndrome 1	*SALL1*
Tuberous Sclerosis 1, 2	*TSC1* *TSC2*
Tubulointerstitial Kidney Disease, Autosomal Dominant	*HNF1B* *REN* *UMOD*
Tumoral Calcinosis, Hyperphosphatemic	*FGF23*
Thyrotoxic periodic paralysis, susceptibility to, Type 2	*KCNJ18*
Type 1 Diabetes	*FOXP3* *INS*
Urofacial Syndrome 1	*HPSE2*
VACTERL association with hydrocephalus	*HOXD13*
Vasculitis, Autoinflammation, Immunodeficiency, and Hematologic Defects Syndrome	*ADA2*
Vesicoureteral Reflux 3	*SOX17*
Vitamin D-Dependent Rickets, Type 1A	*CYP27B1*
Von Hippel-Lindau Syndrome	*VHL*
Wilson Disease	*ATP7B*
Wiskott-Aldrich Syndrome	*WAS*
Wolcott-Rallison Syndrome	*EIF2AK3*
Wolfram Syndrome Type 1, 2	*CISD2* *WFS1*
Xanthinuria, Type 1, 2	*MOCOS* *XDH*

13011 McCallen Pass, Building A Suite 100 | Austin, TX 78753 | natera.com

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