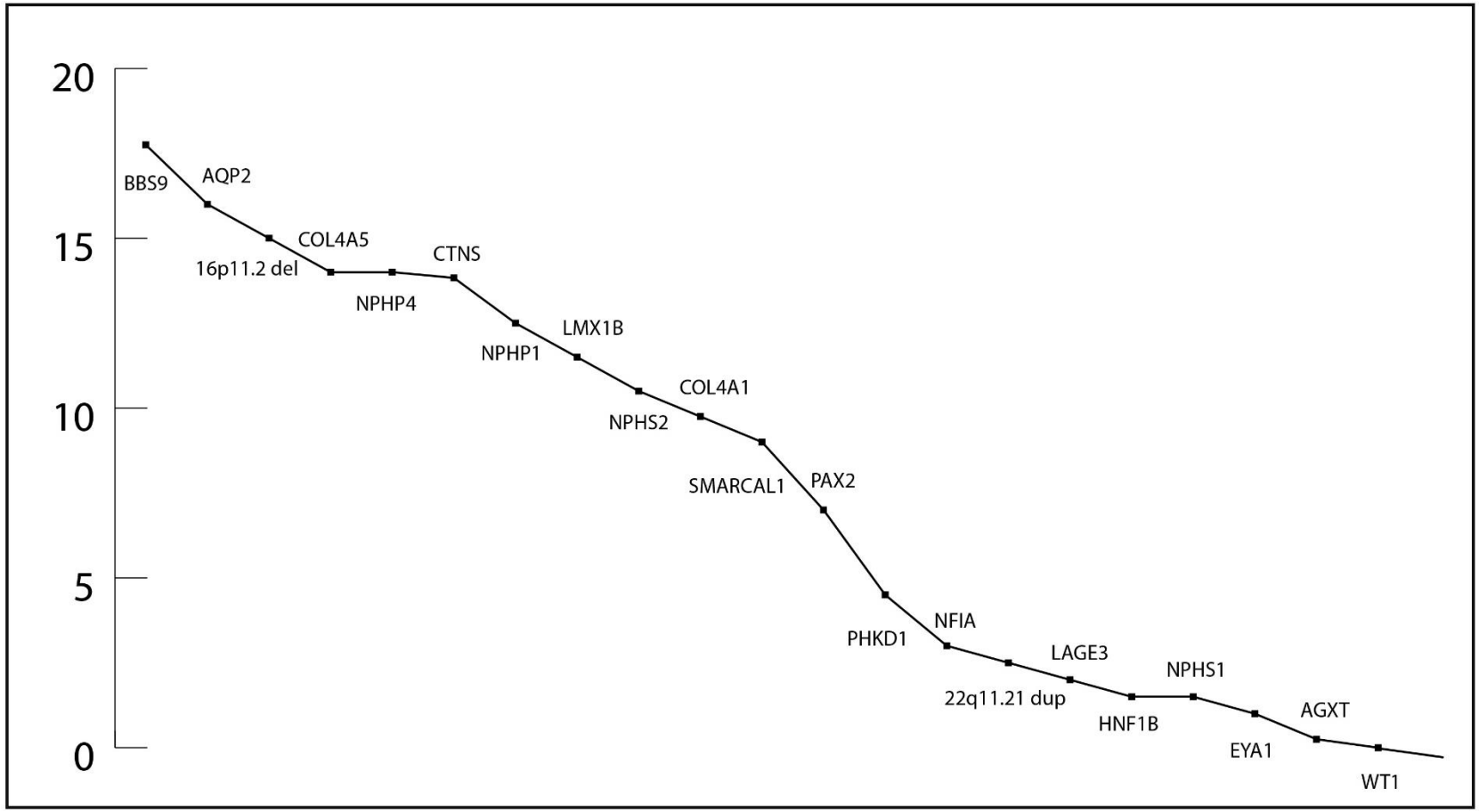
Supplementary Figure S1. Age of kidney failure for each monogenic etiology



Age of dialysis for each ESKD-causing gene found in the cohort. For genes that appear multiple times the mean age of ESKD is presented. ESKD End-stage kidney disease.

Supplementary Table S1. Monogenic CKD gene panel

CAKUT

Gene	Inheritance	Reference	Syndrome
BMP4	AD	1	
CHD1L	AD	1	
DSTYK	AD	1	CAKUT1
EYA1	AD	1	Branchiootorenal syndrome 1, with or without cataracts
GATA3	AD	1	Hypoparathyroidism, sensorineural deafness, and renal dysplasia
HNF1B	AD	1	Renal cysts and diabetes syndrome
MUC1	AD	1	Medullary cystic kidney disease 1
PAX2	AD	1	FSGS 7/ Papillorenal syndrome
ROBO2	AD	1	Vesicoureteral reflux 2
SALL1	AD	1	Townes-Brocks branchiootorenal-like syndrome
SIX1	AD	1	
SIX2	AD	1	
SIX5	AD	1	Branchiootorenal syndrome 2
SOX17	AD	1	Vesicoureteral reflux 3
SRGAP1	AD	1	
TBX18	AD	1	CAKUT2
TNXB	AD	1	VUR8
UMOD	AD	1	
UPK3A	AD	1	
WNT4	AD	1	Mullerian aplasia and hyperandrogenism
ACE	AR	1	Renal tubular dysgenesis
AGT	AR	1	Renal tubular dysgenesis
AGTR1	AR	1	Renal tubular dysgenesis

CHRM3	AR	1	
FGF20	AR	1	Renal hypodysplasia/aplasia 2
FRAS1	AR	1	Fraser syndrome 1
FREM1	AR	1	Bifid nose with or without anorectal and renal anomalies
FREM2	AR	1	Fraser syndrome 2
GRIP1	AR	1	Fraser syndrome 3
HPSE2	AR	1	Urofacial syndrome 1
ITGA8	AR	1	Renal hypodysplasia/aplasia 1
LRIG2	AR	1	Urofacial syndrome 2
REN	AR/AD	1	Renal tubular dysgenesis/ Hyperuricemic nephropathy, familial juvenile 2
TRAP1	AR	1	
KAL1/ ANOS1	XLR	3	Kallmann
PROKR2	AD	2	
TBX3	AD	2	
FBN1	AD	2	
TBX1	AD	2	
MAP2K2	AD	2	Cardiofaciocutaneous syndrome 4
WFS1	AD	2	
FANCI	AR	2	Fanconi anemia, complementation group I
ACTG2	AD	3	Visceral myopathy
COL11A1	AD	3	Stickler syndrome type II
FANCB	XLR	3	Fanconi anemia B
BRIP1	AR?	3	Fanconi anemia J
KANSL1	AD	3	Koolen-De Vries syndrome
MYCN	AD	3	Feingold syndrome
RERE	AD	3	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart
SMC1A	XLD	3	Cornelia de Lange syndrome 2

BBS10	AR	4	Bardet-Biedl syndrome 10
HDAC8	XLD	4	Cornelia De Lange syndrome 5
ACTB	AD	3	Baraitser-Winter syndrome 1
ARID1A	AD	3	Occipital horn syndrome- vesicoureteric obstruction
ARID1B	AD	3	COFFIN-SIRIS SYNDROME - horseshoe kidney, hypospadias
ARNT2	AR	3	Webb Dattani syndrome -anomalies as hydronephrosis
ARX	XL	3	Proud syn- renal dysplasia
ATP7A	XLR	3	Occipital horn syndrome- vesicoureteric obstruction
B3GLCT	AR	3	Peters-plus syndrome
B4GAT1	AR	3	Muscular dystrophy dystroglycanopathy
B9D1	AR	3	Meckel syndrome
BCOR	XLD	3	Microphthalmia
BRAF	AD	3	Noonan 7
CCBE1	AR	3	Hennekam Lymphangiectasia Lymphedema Syndrome
CCDC22	XLR	3	
CDC5L	?	3	CAKUT
CENPF	AR	3	Stromme syndrome
CHD7	AD	3	CHARGE
CHST14	AR	3	Ehlers-Danlos
CISD2	AR	3	Wolfram syndrome 2
COL18A1	AR	3	Knobloch syndrome- unilateral duplication of the renal collecting system
COL5A1	AD	3	Ehlers-Danlos
CRTAP	AR	3	Osteogenesis imperfecta type 7
DCHS1	AR	3	Van Maldergem syndrome 1- renal hypoplasia
DDX59	AR	3	Orofaciodigital syndrome V- fused kidneys
DHCR7	AR	3	Smith-Lemli-Opitz syndrome
DKC1	XLR	3	Dyskeratosis congenita- horseshoe kidney
TERC	AD	3	Dyskeratosis congenita

DLL3	AR	3	Spondylocostal dysostosis
DLL4	AD	3	Adams-Oliver syndrome 6- small kidney
DNA2	AR	3	Seckel syndrome 8- ectopic kidney
DPH1	AR	3	Developmental delay with short stature, dysmorphic features, and sparse hair- horseshoe kidney, dysplastic kidney
EBP	XLD	3	Chondrodysplasia punctata- hydronephrosis
EDNRA	AD	3	Mandibulfacial dysostosis with alopecia- megaureter
EIF2B4	AR	3	Leukoencephalopathy with Vanishing White Matter- renal hypoplasia
ERBB3	AD	3	Lethal congenital contracture syndrome 2- distended urinary bladder
ERCC4	AR	3	Fanconi anemia Q
ESCO2	AR	3	Roberts syndrome- horseshoe kidney, polcystic kidney
FAM20C	AR	3	Raine syndrome-hydroureter
FAM58A	XLD	3	STAR
FANCA	AR	3	Fanconi anemia
FANCC	AR	3	Fanconi anemia
FANCD2	AR	3	Fanconi anemia
FANCE	AR	3	Fanconi anemia
FANCF	AR	3	Fanconi anemia
FANCG	?	3	Fanconi anemia
FANCL	AR	3	Fanconi anemia
FANCM	AR	3	Fanconi anemia
FAT4	AR	3	Van Maldergem Syndrome 2
FGF10	AD	3	Lacrimoauriculodentodigital syndrome
FGFR3	AD	3	Thanatophoric dysplasia
FKBP14	AR	3	Ehlers-Danlos
FLNA	XLR/XLD	3	Congenital short bowel syndrome ,Melnick-Needles syndrome ,Otopalatodigital syndrome type 2,Frontometaphyseal dysplasia-hydrpnephrosis

FOXC1	AD	3	Axenfeld-Rieger syndrome,Type 3-urethral defect
FOXC2	AD	3	Lymphedema distichiasis syndrome with renal disease and diabetes mellitus
FOXF1	AD	3	Alveolar capillary dysplasia with misalignment of pulmonary veins
GATA6	AD	3	Pancreatic agenesis and congenital heart defects- ureteral duplication
GDNF	AD	3	
GPC3	XLR	3	Simpson golabi behmel syndrome type 1
HES7	AR	3	spondylocostal dysostosis
HOXA13	AD	3	Hand foot uterus syndrome
HOXD13	?	3	VACTREL association with hydrocephalus
HSD3B2	AR	3	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency- hypospadias
HSPA9	AR	3	Even plus- VUR, hypoplastic kidney
HYLS1	AR	3	Hydrolethalus syndrome- hydronephrosis
ICK	AR	3	Endocrine cerebroosteodysplasia
IFT27/ BBS19	AR	3	BBS19
IRF6	AD	3	Popliteal Pterygium Syndrome
ITGA6	AR	3	Junctional epidermolysis bullosa-pyloric atresia syndrome
JAM3	AR	3	Hemorrhagic destruction of the brain,subependymal calcification, and cataracts
KDM6A	XLD	3	KABUKI 2
LFNG	AR	3	Spondylocostal dysostosis 3/ Cenani-Lenz syndactyly syndrome
MAP2K1	AD	3	Cardiofaciocutaneous syndrome
MAPRE2	AD	3	Symmetric circumferential skin creases, congenital, 2
MBTPS2	XLR	3	IFAP Syndrome with or without Bresheck Syndrome
MESP2	AR	3	Spondylocostal dysostosis 2
MIR17HG	AD	3	Feingold syndrome 2
MNX1	AD	3	Currarino syndrome
NAA10	XL	3	Microphthalmia, syndromic 1

NBN	AR	3	Nijmegen breakage syndrome-Hydronephrosis
NOLA2	AR	3	Dyskeratosis congenita, autosomal recessive 2
NIPBL	AD	3	Cornelia De Lange syndrome 1
NSDHL	XLD	3	CHILD syndrome
OCLN	AR	3	Pseudo-TORCH syndrome
PALB2/ FANCN	AR?	3	Fanconi anemia N
PHGDH	AR	3	Neu-Laxova syndrome
PIGA	XLR	3	Multiple congenital anomalies-hypotonia-seizures syndrome 2
PIGL	AR	3	CHIME syndrome
PIGN	AR	3	Multiple congenital anomalies-hypotonia-seizures syndrome 1
PIK3R2	AD	3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome
PITX2	AD	3	Axenfeld-Rieger syndrome, type 1- hypospadias
POMT1	AR	3	Muscular dystrophy-dystroglycanopathy
POR	AR	3	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
PORCN	XLD	3	Focal dermal hypoplasia
PPP1R15B	AR	3	Microcephaly, short stature, and impaired glucose metabolism
PQBP1	XLR	3	Renpenning syndrome
PUF60	AD	3	Verheij syndrome
RAB23	AR	3	Carpenter syndrome - Hydronephrosis
RAB18	AR	3	Warburg micro syndrome
RAB3GAP1	AR	3	Warburg micro syndrome
RAB3GAP2	AR	3	Warburg micro syndrome
TBC1D20	AR	3	Warburg micro syndrome 4
RAI1	AD	3	Smith-Magenis syndrome
RAP1A	AD?	3	KABUKI
RAP1B	AD?	3	KABUKI
RBBP8	AR	3	Seckel syndrome 2- ECTOPIC KIDNEY

RBM10	XLR	3	TARP syndrome
RBM8A	AR	3	TAR syndrome
RECQL4	AR	3	Baller-Gerold syndrome
RET	AR?	3	Bilateral renal agenesis
RIN2	AR	3	Macrocephaly, alopecia, cutis laxa, and scoliosis- Urethral stenosis
RIPK4	AR	3	Popliteal pterygium syndrome, Bartsocas-Papas type
RIPPLY2	AR	3	Spondylocostal dysostosis 6
RMND1	AR	3	Combined oxidative phosphorylation deficiency 11
ROR2	AR	3	Robinow syndrome
RPL11	AD	3	Diamond-Blackfan anemia 7
RPL26	AD	3	Diamond-Blackfan anemia 11
RPL35A	AD	3	Diamond-Blackfan anemia 5
RPL5	AD	3	Diamond-Blackfan anemia 6
RPS10	AD	3	Diamond-Blackfan anemia 6
RPS17	AD	3	Diamond-Blackfan anemia 4
RPS19	AD	3	Diamond-Blackfan anemia 1
RPS24	AD	3	Diamond-Blackfan anemia 3
RPS26	AD	3	Diamond-Blackfan anemia 10
RPS28	AD	3	Diamond-Blackfan anemia 15
RPS29	AD	3	Diamond-Blackfan anemia 13
RPS7	AD	3	Diamond-Blackfan anemia 8
RTTN	AR	3	Microcephaly, short stature, and polymicrogyria with seizures
SALL4	AD	3	IVIC syndrome, Duane-radial ray syndrome
SC5D	AR	3	Lathosterolosis- pyeloctasis
SEMA3E	AD	3	CHARGE syndrome
SETBP1	AD	3	Schinzel-Giedion midface retraction syndrome
SF3B4	AD	3	Acrofacial dysostosis 1, Nager type
SHH	AD	3	Single median maxillary central incisor

SLIT2	?	3	
SLX4	AR	3	Fanconi anemia P
SMARCE1	AD	3	Coffin-Siris syndrome 5
SNRPB	AD	3	Cerebrocostomandibular syndrome- horseshoe kidney
SOX11	AD	3	Coffin-Siris syndrome
SOX9	AD	3	Campomelic dysplasia- hydronephrosis
SPECC1L	AD	3	Opitz GBBB syndrome, type II- urethral abnormalities
SPINT2	AR	3	Diarrhea 3, secretory sodium, congenital, syndromic
STRA6	AR	3	Microphthalmia, syndromic 9
TAPT1	AR	3	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type-hydronephrosis
TBC1D24	AR	3	DOORS syndrome
TCTN3	AR	3	Joubert syndrome
TFAP2A	AD	3	Branchiooculofacial syndrome
THOC6	AR	3	Beaulieu-Boycott-Innes syndrome
TMCO1	AR	3	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome
TP63	AD	3	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3
TRAIP	AR	3	Seckel syndrome 9
TSR2	XLR	3	Diamond-Blackfan anemia 14 with mandibulofacial dysostosis
TXNL4A	AR	3	Burn-McKeown syndrome
UBE2T	AR	3	Fanconi anemia T
UBR1	AR	3	Johanson-Blizzard syndrom
UPB1	AR	3	Beta-ureidopropionase deficiency
USP9X	XLD	3	Mental retardation, X-linked 99, syndromic, female-restricted
VANGL1	AD	3	Caudal regression syndrome
WNT3	AR	3	Tetra-amelia syndrome
WNT5A	AD	3	Robinow syndrome, autosomal dominant 1

WNT7A	AR	3	Ulna and fibula, absence of, with severe limb deficiency
XRCC4	AR	3	Short stature, microcephaly, and endocrine dysfunction
XYLT2	AR	3	Spondyloocular syndrome
ZIC3	XLR	3	VACTERL association
JAG1	AD	3	Alagille; renal dysplasia, RTA, VUR, urinary obstruction, RAS, CKD
NECTIN1/ PVRL1	AR	16	Cleft palate ectodermal dysplasia syndrome (CLPED1); orofacial cleft 7 (OFC7)- Renal dysplasia
USF2	?	16	Upstream stimulatory factor 2- Cystic renal dysplasia and hypodysplasia
DACH1	AR	17	Dachshund 1
ETV4	AR	17	ETS translocation variant 4, E1A enhancer binding protein
CRKL	AD	17	CRK Like Proto-Oncogene, adaptor protein
GREB1L	AD	17	Renal hypodysplasia/aplasia 3
NRIP1	AD	17	CAKUT3
PBX1	AD	17	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay
MYOG	?	19	MYOGENIC FACTOR 4
DLX5	AR	19	Split-hand/foot malformation 1 with sensorineural hearing loss
DLX6	?	19	
B3GALTL	AR	20	Peters-plus syndrome
CHRNG	AR	20	Escobar syndrome
CTU2	AR	20	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome
EMG1	AR	20	Bowen-Conradi syndrome
FOXP1	AD	20	Mental retardation with language impairment and with or without autistic features
IFT74/ BBS20/ CCDC22	AR	20	BBS20

LRP4	AR	20	Cenani-Lenz syndactyly syndrome
PROK2	AR/AD	20	Hypogonadotropic hypogonadism 4 with or without anosmia
SCARF2	AR	20	
PIGO	AR	20	Hyperphosphatasia with mental retardation syndrome 2
PIGV	AR	20	Hyperphosphatasia with mental retardation syndrome 1
PIGY	AR	20	Hyperphosphatasia with mental retardation syndrome 6
PTF1A	AR	20	Pancreatic and cerebellar agenesis
ACTG1	AD	20	Baraitser-Winter syndrome 2
AIFM3	AD	20	
BMP7	AD	20	
DACT1	AD	20	Townes-Brocks syndrome 2
EP300/ EID1	AD	20	Rubinstein-Taybi syndrome 2
ESRRG	AD	20	
FGF8	AD	20	Hypogonadotropic hypogonadism 6 with or without anosmia
FMN1	AD	20	
GDF3	AD	20	Klippel-Feil syndrome 3
GFRA1	AD	20	
GLI2	AD	20	Culler-Jones syndrome, Holoprosencephaly 9
KCNH2	AD	20	Scalp-ear-nipple syndrome
LPP	AD	20	
RAF1	AD	20	Noonan syndrome 5
SEMA3A	AD	20	Hypogonadotropic hypogonadism 16 with or without anosmia
SNAP29	AD/ AR	20	Di George syndrome
SOS1	AD	20	Noonan syndrome
TRPS1	AD	20	Trichorhinophalangeal syndrome
GDF6	AD/ AR	20	
GLI3	AD/ AR	20	
PCSK5	AD/ AR	20	

AXIN1		20	Caudal duplication anomaly
H19	AD	20	BWS
KCNQ1OT1	AD	20	BWS
MID1	XLR	20	Opitz GBBB syndrome, type I
UPF3B	XLR	20	Mental retardation, X-linked, syndromic 14
OSR1	?	20	
SH2B1	?	20	
IFT46	AR	21	
NFIX	AD	21	
PAX8	AD	21	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia
GDF11	?	21	
TTC30A	?	21	
ATP2A2	AD	22	Darier-white disease, DAR
SOX3	XL	22	Intellectual disability X linked with isolated growth hormone deficiency
TBC1D1	AD	23	RHD, obstructive uropathy, cysts, VUR
CREBBP	AD	3	Rubinstein-Taybi syndrome (CAKUT according to ref 21)
FGFR2	AD	3	Pfeiffer syndrome- hydronephrosis, pelvic kidneys
GREM1	AR	29	Fraser syndrome
KAT6B	AD	30	Genitopatellar syndrome
PARN	AD/AR	31	Renal fibrosis
CYP11A1	?	3	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
7q 11.23	AD	45	Williams-Beuren syndrome
VWA2	?	51	VUR
DLG1	AD	57	
KIF12	AD	57	
LAMA3	AR	59	Epidermolysis bullosa
NBS1	AR	60	Nijmegen breakage syndrome
TNNT3	AD	61	Arthrogryposis, distal, type 2B2, Freeman-Sheldon

TNNI2	AD	61	Arthrogryposis, distal, type 2B1, Freeman-Sheldon
MYH3	AD	62	Arthrogryposis, distal, type 2A, Freeman-Sheldon
COL2A1	AD	63	Spondyloepimetaphyseal dysplasia
LIS1	AD	66	Lissencephaly 1
MPI	AR	67	Congenital disorder of glycosylation, type Ib
FUZ	?	3	Caudal regression sequence
Chromosomal		68	Digeorge/VCF (Catch22)
chromosomal		68	Down syndrome
chromosomal		68	Edward syndrome
chromosomal		68	Patau syndrome
chromosomal		70	Miller-Dieker
chromosomal		69	Pallister-Killian

SRNS

Gene	Inheritance	Reference	Syndrome
ARHGDIA	AR	1	Nephrotic syndrome 8
CD2AP	AR	1	FSGS 3
CRB2	AR	1	FSGS 9
CUBN	AR	1	Megaloblastic anemia-1, Finnish type
DGKE	AR	1	Nephrotic syndrome 7
EMP2	AR	1	Nephrotic syndrome 10
FAT1	AR	1	
ITGA3	AR	1	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital
ITGB4	AR	1	
KANK1	AR	1	
KANK2	AR	1	Nephrotic syndrome, type 16

KANK4	AR	1	
LAMB2	AR	1	Nephrotic syndrome, type 5, with or without ocular abnormalities
MYO1E	AR	1	FSGS 6
NPHS1	AR	1	Nephrotic syndrome, type 1
NPHS2	AR	1	Nephrotic syndrome, type 2
PLCE1	AR	1	Nephrotic syndrome, type 3
PTPRO/ GLEPP1	AR	1	Nephrotic syndrome, type 6
SCARB2	AR	1	Epilepsy, progressive myoclonic 4, with or without renal failure
SMARCAL1	AR	1	Schimke immunoosseous dysplasia
WDR73	AR	1	Galloway-Mowat syndrome 1
XPO5	AR	1	
ACTN4	AD	1	FSGS 1
ANLN	AD	1	FSGS 8
ARHGAP24	AD	1	
INF2	AD	1	FSGS 5
LMX1B	AD	1	nail patella
MYH9	AD	1	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss
TRPC6	AD	1	FSGS 2
WT1	AD	1	Nephrotic syndrome, type 4
COQ8B/ ADCK4	AR	3	Nephrotic syndrome, type 9
CTC1	AR	3	Dyskeratosis congenita
ERCC6	AR	3	Cockayne syndrome
ERCC8	AR	4	Cockayne syndrome
FGA	AD	3	Amyloidosis, familial visceral
APOA1	AD	3	Amyloidosis

B2M	AD	3	Amyloidosis
G6PC	AR	3	Glycogen storage disease Ia
GBA	AR	3	Gaucher disease
GSN	AD	3	Amyloidosis, Finnish type
IKBKAP/ ELP1	AR	3	Hereditary sensory and autonomic neuropathy type 3- glomerulosclerosis
LAMB3	AR/AD	3	Amelogenesis imperfecta, type IA; Epidermolysis bullosa, junctional, Herlitz type; Epidermolysis bullosa, junctional, non-Herlitz type
LAMC2	AR	3	Epidermolysis bullosa, junctional, Herlitz and non- Herlitz type
LYZ	AD	3	Amyloidosis
MEFV	AR/AD	3	FMF
MVK	AR	3	Mevalonic aciduria- amyloidosis
NARS2	AR	3	Combined oxidative phosphorylation deficiency- FSGS, TUBALOPATHY
NEXMIF	XLD	3	Mental retardation, X-linked
NLRP3	AD	3	Muckle-Wells syndrome- Amyloidosis
NUP107	AR	3	Nephrotic syndrome,type 11
NUP205	AR	3	Nephrotic syndrome,type 13
NUP93	AR	3	Nephrotic syndrome,type 12
PMM2	AR	3	Congenital disorder of glycosylation
PRODH	AR	3	Hyperprolinemia, type I
TNFRSF1A	AD	3	Periodic fever, familial
TTR	AD	3	Amyloidosis, hereditary, transthyretin-related
ZAP70	AR	3	Autoimmune disease, multisystem, infantile-onset, 2
PSTPIP1	AD	16	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne- Proteinuria, possibly immune mediated
RPL15	AD	17	Diamond-Blackfan anemia 12
SGPL1	AR	17	Nephrotic syndrome, type 14
ALG1	AR	20	Congenital disorder of glycosylation type Ik
AVIL	AR	20	Nephrotic syndrome, type 21

LAGE3	XLR/ AD	20	Galloway-Mowat syndrome 2
MAGI2	AR	20	Nephrotic syndrome, type 15
NUP133	AR	20	Nephrotic syndrome, type 18
NUP85	AR	20	Nephrotic syndrome, type 17
TP53RK	AR	20	Galloway-Mowat syndrome 4
TPRKB	AR	20	Galloway-Mowat syndrome 5
PODXL	AD	20	
NXF5	XL	20	
APOE	XL	20	
GPC5	?	20	
SYNPO	?	20	
E2F3	AD/XL	34	FSGS
NUP160	AR	35	FSGS 19
ANKFY1	AR	37	
GAPVD1	AR	37	
PUS3	AR	14	Mental retardation, autosomal recessive 55- FSGS
LAMA5	AD/AR	36	FSGS
NEIL1	AR	51	
DLC1	AR	38	**pTSNS**
TNS2	AR	38	**pTSNS**
CDK20	AR	38	**pTSNS**
ITSN1	AR	38	**pTSNS**
ITSN2	AR	38	**pTSNS**
APOL1	AR	42	FSGS, HIVAN, SLE, Sickle cell nephropathy
PLA2R		30	Idiopathic Membranous Nephropathy

Glomerular disease

Gene	Inheritance	Reference	Syndrome
CFH	AD/AR	1	Complement factor H deficiency- MPGN, aHUS
CFI	AD/AR	1	Complement factor I deficiency- MPGN, aHUS
CFHR5	AD	1	Nephropathy due to CFHR5 deficiency, MPGN, aHUS
FN1	AD	1	GFND
COL4A3	AD/AR	1	Alport
COL4A4	AD/AR	1	Alport
COL4A5	XLD	1	Alport
COL4A6	XLR	1	Alport w LM
ACTA2	AD	3	Multisystemic smooth muscle dysfunction syndrome- glomerulosclerosis
AFF4	AD	3	CHOPS syndrome
APC2	AR	3	Sotos syndrome 3
CD151	AR	3	Nephropathy with pretibial epidermolysis bullosa and deafness
CD19	AR	3	CVID
CD81	AR	3	CVID
COPA	AD	3	Autoimmune interstitial lung, joint, and kidney disease
EFEMP2	AR	3	Cutis laxa
FBLN5	AD/AR	3	Cutis laxa, Neuropathy, hereditary, with or without age-related macular degeneration
GLB1	AR	3	GM1-gangliosidosis
LCAT	AR	3	Norum disease
LDHA	AR	3	Glycogen storage disease XI
LMNA	AD/AR	3	Atypical Werner syndrome
LTBP4	AR	3	Cutis laxa
MAFB	AD	3	Multicentric carpotarsal osteolysis syndrome
PGM3	AR	3	Immunodeficiency 23-membranoproliferative glomerulonephritis
SOX18	AD	3	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome
WAS	XLR	3	Wiskott-Aldrich syndrome

YAP1	AD	3	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation
ZMPSTE24	AR	3	Restrictive dermopathy, lethal
GLA	XL	2	Fabry
OSGEP	AR	17	Galloway-Mowat syndrome 3
CFB	AR/ AD	20	Complement factor B deficiency
NEU1	AR	16	Sialidosis
CD46/ MCP	AD/AR	1	
CFHR1	AD/AR	19	aHUS
CFHR3	AD/AR	19	aHUS
THBD	AD	21	aHUS
SPRY2	AD	21	

Nephrolithiasis/ Nephrocalcinosis

Gene	Inheritance	Reference	Syndrome
ATP6V0A4/ ATP6N1B	AR	1	distal RTA
APRT	AR	1	Adenine phosphoribosyltransferase deficiency
CASR	AD		Hypocalciuric hypercalcemia, type I
CLDN16	AR	1	Hypomagnesemia 3, renal
CLDN19	AR	1	Hypomagnesemia 5, renal, with ocular involvement
CYP24A1	AR	1	Hypercalcemia, infantile, 1
FAM20A	AR	1	Amelogenesis imperfecta, type IG (enamel-renal syndrome)
HNF4A	AD	1	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, MODY1
HPRT1	XLR	1	Lesch-Nyhan syndrome
OCRL	XLR	1	Dent disease 2, Lowe syndrome

SLC22A12	AD/AR	1	Hypouricemia, renal
SLC2A9	AD/AR	1	Hypouricemia, renal
SLC34A1	AD/AR	1	
SLC34A3	AR	1	Hypophosphatemic rickets with hypercalciuria
SLC3A1/ CSNU1	AD/AR	1	Cystinuria
SLC4A1	AD/AR	1	distal RTA
SLC7A9/ CSNU1	AD/AR	1	Cystinuria
SLC9A3R1/ NHERF1	AD	1	Nephrolithiasis/osteoporosis, hypophosphatemic, 2
VDR	AD	1	Rickets, vitamin D-resistant, type IIA
XDH	AR	1	Xanthinuria, type I
SLC26A1	AR	3	Nephrolithiasis, calcium oxalate
ABCC6	AD/AR	3	
AUH	AR	3	3-methylglutaconic aciduria, type I
BSCL2	AR	3	Lipodystrophy, congenital generalized, type 2
CDC73	AD	3	Hyperparathyroidism
CLCN5	XLR	3	Dent disease nephrolithiasis type 1 hypophosphatemic Ricket
DCDC2	AR	3	Nephronophthisis 19
ENPP1	AR	3	Hypophosphataemic rickets
GCM2	AD/AR	3	Hypoparathyroidism,familial isolated
GNA11	AD	3	Hypocalciuric hypercalcemia,type 2
GNAS	AD	3	pseudohypoparathyroidism
GNB1	AD	3	Mental retardation, autosomal dominant 42
STX16	AD	3	Pseudohypoparathyroidism, type IB
GRHPR	AR	3	Hyperoxaluria
HGD	AR	3	Alkaptonuria

INSR	AR	3	Rabson-Mendenhall syndrome- nephrocalcinosis
KCNH1	AD	3	Zimmermann-Laband Syndrome
KCTD1	AD	3	Scalp-ear-nipple syndrome
LRP2	AR	3	Donnai-barrow syndrome
MTM1	XLR	3	Myotubular myopathy
OPLAH	AD/AR	3	Oxoprolinase deficiency
PIGT	AR	3	Multiple congenital anomalies-hypotonia-seizures syndrome-nephrocalcinosis, renal cysts
PLG	AR	3	Plasminogen deficiency, type I
PRPS1	XLR	3	Phosphoribosylpyrophosphate synthetase superactivity
PTH	AD/AR	3	Hypoparathyroidism
PTH1R	AD	3	Metaphyseal chondrodysplasia, Murk Jansen type
SBDS	AR	3	Shwachman-Diamond syndrom
SERPINH1	AR	3	Osteogenesis imperfecta, type X
SI	AR	3	Sucrase-isomaltase deficiency, congenita
SLC1A1	AR	3	Dicarboxylic aminoaciduria
SLC36A2	AD	3	Hyperglycinuria
SLC37A4	AR	3	Glycogen storage disease Ib, IC
SLC6A20	AD	3	Hyperglycinuria
STRADA	AR	3	Polyhydramnios, megalencephaly, and symptomatic epilepsy
TRNT1	AR	3	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay
VIPAS39	AR	3	Arthrogryposis, renal dysfunction, and cholestasis 2
VPS33B	AR	3	Arthrogryposis, renal dysfunction, and cholestasis 1
ZNF687	AD	3	Paget disease of bone 6
ALDOB	AR	20	Fructose intolerance, hereditary- Hyperoxaluria, primary, type 1
ALPL	AR	3	Hypophosphatasia, infantile
ADCY10/ SAC	AD	1	Hypercalciuria

Cystic renal disease

Gene	Inheritance	Reference	Syndrome
NPHP1 (JBTS4)	AR	1	Joubert syndrome 4
INVS (NPHP2)	AR	1	Nephronophthisis 2, infantile
NPHP3	AR	1	Nephronophthisis 3
NPHP4	AR	1	Nephronophthisis 4
IQCB1 (NPHP5)	AR	1	Senior-Loken syndrome 5
CEP290 (NPHP6)	AR	1	
GLIS2 (NPHP7)	AR	1	Nephronophthisis 7
RPGRIP1L (NPHP8)	AR	1	
NEK8 (NPHP9)	AR	1	Nephronophthisis 9
SDCCAG8 (NPHP10)	AR	1	BBS 16
TMEM67/ MKS3	AR	1	NPHP11
TTC21B	AD/AR	1	NPHP12
WDR19	AR	1	NPHP13
ZNF423	AD/AR	1	NPHP14
CEP164	AR	1	NPHP15
ANKS6	AR	1	NPHP16
ATXN10	AR	1	AR according to paper, AD according to OMIM
FAN1	AR	1	Interstitial nephritis, karyomegalic
SLC41A1	AR	1	
CEP83/ CCDC41	AR	1	NPHP18
AGXT	AR	1	Hyperoxaluria, primary, type 1

PKHD1	AR	1	Polycystic kidney disease 4, with or without hepatic disease
INPP5E/ JBTS1	AR	1	Joubert syndrome 1
TMEM216/ JBTS2	AR	1	
AHI1/ JBTS3	AR	1	Joubert syndrome 3
ARL13B	AR	1	Joubert syndrome 8
CC2D2A	AR	1	
OFD1/ CXORF5	AR	1	
KIF7	AR	1	
TCTN1	AR	1	Joubert syndrome 13
TMEM237	AR	1	Joubert syndrome 14
CEP41/ TSGA14	AR	1	Joubert syndrome 15
TMEM138	AR	1	Joubert syndrome 16
C5orf42/ CPLANE1	AR	1	Joubert syndrome 17
TMEM231	AR	1	
CSPP1	AR	1	Joubert syndrome 21
PDE6D	AR	1	Joubert syndrome 22
TBC1D32	AR	1	
SCLT1	AR	1	
MKS1	AR	1	
TCTN2	AR	1	
BBS1	AR	1	
BBS2	AR	1	
BBS4	AR	1	
BBS5	AR	1	
MKKS/ BBS6	AR	1	

BBS7	AR	1	
TTC8/ BBS8	AR	1	
PTHB1/ BBS9	AR	1	
C21orf58	AR	1	
TRIM32/ BBS11	AR	1	
C4orf24/ BBS12	AR	1	
WDPCP/ BBS15	AR	1	
LZTFL1/ BBS17	AR	1	
ALMS1	AR	1	Alstrom syndrome
IFT122	AR	1	Cranioectodermal dysplasia 1
WDR35/ IFT121	AR	1	Cranioectodermal dysplasia 2
IFT140	AR	1	
C14ORF179/ IFT43	AR	1	Cranioectodermal dysplasia 3
DYNC2H1	AD/AR	1	Short-rib thoracic dysplasia 3 with or without polydactyly
WDR34	AR	1	Short-rib thoracic dysplasia 11 with or without polydactyly
WDR60	AR	1	Short-rib thoracic dysplasia 8 with or without polydactyly
IFT80	AR	1	Short-rib thoracic dysplasia 2 with or without polydactyly
IFT172	AR	1	
TRAF3IP1	AR	1	Senior-Loken syndrome 9
NEK1	AD/AR	1	Short-rib thoracic dysplasia 6 with or without polydactyly
POC1A	AR	1	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis
EVC	AR	1	Ellis-van Creveld syndrome
EVC2	AR	1	Ellis-van Creveld syndrome
PKD1	AD	3	Polycystic kidney disease 1
PKD2	AD	3	Polycystic kidney disease 2
SRCAP	AD	4	Floating-Harbor syndrome

ALG8	AD/AR	3	Polycystic liver disease 3 with or without kidney cysts, Congenital disorder of glycosylation, type Il
ALG9	AR	3	Gillessen-Kaesbach-Nishimura syndrome, Congenital disorder of glycosylation, type Il
AMER1	XLD	3	Osteopathia striata with cranial sclerosis
ARL6/ BBS3	AR	3	
B9D2	AR	3	Meckel syndrome type10
BBIP1/ BBS18	AR	3	Bardet-Biedl syndrome 18
BMPER	AR	3	Diaphanospondylodysostosis
BUB1B	AR	3	
CD96	AD	3	C syndrome
CEP104	AR	3	Joubert syndrome 25
CEP120/ SRTD13	AR	3	Short-rib thoracic-dysplasia
COL4A1	AD	3	HANAC
CPT2	AR	3	CPT II deficiency, lethal neonatal
DNAAF1	AR	3	PCD 13
FLT4	AD	3	cystic renal lymphangiectasia
GLIS3	AR	3	Diabetes Mellitus,Neonatal,With Congenital Hypothyroidism
HSD17B4	AR	3	D-bifunctional protein deficiency
NOTCH2	AD	3	Hajdu-Cheney syndrome, alagille
PAF1	?	3	Zellweger syndrome 3
PEX1	AR	3	Zellweger syndrome
PEX10	AR	3	Zellweger syndrome
PEX11B	AR	3	Zellweger syndrome
PEX12	AR	3	Zellweger syndrome
PEX13	AR	3	Zellweger syndrome
PEX14	AR	3	Zellweger syndrome

PEX16	AR	3	Zellweger syndrome
PEX19	AR	3	Zellweger syndrome
PEX2	AR	3	Zellweger syndrome
PEX3	AR	3	Zellweger syndrome
PEX26	AR	3	Zellweger syndrome
PEX5	AR	3	Zellweger syndrome
PEX6	AR	3	Zellweger syndrome
PIEZO2	AD	3	Marden-Walker syndrome
PNPLA6	AR	3	Laurence-Moon syndrome
RAD51C	AR	3	Fanconi anemia, complementation group O
RNU4ATAC	AR	3	Microcephalic osteodysplastic primordial dwarfism, type I
SCN4A	AD	3	Hypokalemic periodic paralysis
STK11	AD	3	Peutz-Jeghers syndrome
TTC37	AR	3	Trichohepatoenteric syndrome 1
VHL	AD	3	Von Hippel-Lindau syndrome
C2CD3/ OFD14	AR	20	Orofaciodigital syndrome
HOXA4	AD	20	
HOXB6	AD	20	
IFT52	AR	20	Short-rib thoracic dysplasia 16 with or without polydactyly
IFT57	AR	20	Orofaciodigital syndrome XVIII
IFT81	AR	20	Short-rib thoracic dysplasia 19 with or without polydactyly
KIAA0586	AR	20	Joubert syndrome 23
KIAA0556	AR	20	Joubert syndrome 26
KIAA0753	AR	20	Orofaciodigital syndrome XV
KIF14	AR	20	Meckel syndrome 12
MAPKBP1	AR	20	NPHP20
POC1B	AR	20	Cone-rod dystrophy 2
TMEM107	AR	20	Meckel syndrome 13, Joubert syndrome 29

USH2A	AR	20	Usher syndrome type 2A
PIK3R4	AR	21	
TXNDC15	AR	21	
MRE11A	AR	22	Ataxia-telangiectasia-like disorder- NPHP
SEC61A1	AD	3	Familial juvenile hyperuricemic (gouty) nephropathy
ARMC9	AR	41	Joubert syndrome 30
CFAP410	?		In a joubert panel, CKD panel (KidneySeq™ v4)
DZIP1L	AR	43	Polycystic kidney disease 5
GANAB	AD	44	Polycystic kidney disease 3
PRKCSH	AD	53	Polycystic liver disease 1
SEC63	AD	54	Polycystic liver disease 2
LRP5	AD	55	Polycystic liver disease 4 with or without kidney cysts
DNAJB11	AD	56	Polycystic kidney disease 6 with or without polycystic liver disease
C8orf37 (BBS21)	AR	58	
ADAMTS9	AR	65	NPHP
BICC1	AD	20	

SLE

Gene	Inheritance	Reference	Syndrome
C1QA	AR	5	
C1QB	AR	5	
C1QC	AR	5	
C1R	AR	5	
C1S	AR	5	
C2	AR	5	
C3	AR/ AD	5	AR- sle, AD- aHUS

C4A	AR	5	
C4B	AR	6	
DNASE1	AD	5	
TMEM173/ STING1	AD	5	STING-associated vasculopathy, infantile-onset
SAMHD1	AD/AR	5	Chilblain lupus 2- AD, AGS- AR
ADAR1	AD/AR	5	AGS
IFIH1	AD	5	AGS
RNASEH2B	AR	5	AGS
ACP5	AR	5	Spondyloenchondrodysplasia
TREX1	AD/AR	5	AGS, Chilblain lupus
DNASE1L3	AR	5	Systemic lupus erythematosus 16
RNASEH2A	AR	5	AGS
SHOC2	AD	5	Noonan
KRAS	AD	5	Noonan
PSMA3	AD	5	CANDLE
PSMB4	AR	5	Proteasome-associated autoinflammatory syndrome 3
PSMB8	AD	5	Proteasome-associated autoinflammatory syndrome 1
FAS/ TNFRSF6	AD	5	Autoimmune lymphoproliferative syndrome
FASLG	AD	5	Autoimmune lymphoproliferative syndrome, type IB
PRKCD	AR	5	Autoimmune lymphoproliferative syndrome, type III
RAG2	AD/AR	5	Combined cellular and humoral immune defects with granulomas, Omenn syndrome
CYBB	XL	5	Chronic granulomatous disease
NEIL3	AR	5	Autoimmune cytopenia, Chronic diarrhea, Recurrent Infections
PTEN	AD	5	
PEPD	AR	5	Prolidase deficiency
SLC7A7	AR	5	Lysinuric protein intolerance

MAN2B1/ LAMAN	AR	5	Mannosidosis, alpha, types I and II
ISG15	AR	6	Immunodeficiency 38 (OMIM), AGS (paper)
RNASEH2C	AR	6	AGS
CGAS	?	8	
RIGI/ DDX58	AD	8	
CECR1/ ADA2	AR	7	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
KMT2D/ MLL2	AD	8	KABUKI
NCF2	AR	9	CGD
TNFRSF13B	AD/AR	10	CVID
TCF3	AD	10	Agammaglobulinemia + SLE
PSEN1	AD	11	
TNFAIP3	AD	12	Autoinflammatory syndrome, familial, Behcet-like
SAT1	?	13	
PTPN11	AD	3	Noonan- also diabetic nephropathy
CASP10	AD	3	ALPS

Renal tubulopathy

Gene	Inheritance	Reference	Syndrome
AQP2	AD/ AR	2	Diabetes insipidus, nephrogenic
WNK4	AD	2	Pseudohypoaldosteronism
CLCNKB	AR	2	Bartter
SLC12A3	AR	3	Gitelman
KCNJ1/ ROMK	AR	1	Bartter
SLC12A1/ NKCC2	AR	1	Bartter

ATP6V1B1/ ATP6B1	AR	1	dRTA
AP2S1	AD	3	FHH
BSND	AR	3	Bartter 4A
CAD	AR	3	Epileptic Encephalopathy- RTA
CNNM2	AD/ AR	3	Hypomagnesemia type 6
CTNS	AR	3	Cystinosis
CUL3	AD	3	Pseudohypoaldosteronism
DMP1	AR	3	Hypophosphataemic rickets
EGF	AR	3	Hypomagnesemia 4, renal
EHHADH	AD	3	Fanconi renotubular syndrome 3
EIF2AK3	AR	3	Wolcott Rallison Syndrome
EPG5	AR	3	Vici syndrome- RTA
FAH	AR	3	Tyrosinemia Type 1- fanconi syn, nephrocalcinosis
FGF23	AD	3	Hypophosphatemic Rickets
FGFR1	AD	3	Kallmann, Encephalocraniocutaneous lipomatosis
FXYD2	AD	3	Hypomagnesemia 2, renal
GALNT3	AR	3	Tumoral calcinosis,hyperphosphatemic,familial
HSD11B2	AR	3	Apparent mineralocorticoid excess
INPPL1	AR	3	Opsismodysplasia
KCNJ10	AR	3	SESAME syndrome
KCNJ5	AD	3	Hyperaldosteronism, familial, type III
KL	?	3	Tumoral calcinosis, hyperphosphatemic, familial, 3
KLHL3	AD/ AR	3	Pseudohypoaldosteronism,type 2d
KYNU	AR	3	Hydroxykynureninuria, VCRL2
LARS1	AR	3	Infantile Liver Failure Syndrome 1
MAGED2	XLR	3	Bartter
NR3C2	AD	3	Pseudohypoaldosteronism type I, autosomal dominant

PC	AR	3	Pyruvate carboxylase deficiency
PHEX	XLD	3	Hypophosphatemic rickets, X-linked dominant
SCNN1A	AR	3	Pseudohypoaldosteronism, type I
SCNN1B	AD/ AR	3	Liddle syndrome
SCNN1G	AD/ AR	3	Pseudohypoaldosteronism, Liddle syndrome
SLC26A4	AR	3	Pendred syndrome
SLC2A2	AR	3	Fanconi-Bickel syndrome
SLC4A4	AR	3	Renal tubular acidosis, proximal, with ocular abnormalities
SLC5A2/ SGLT2	AD/ AR	3	Renal glucosuria
SLC6A19	AD/ AR	3	Hartnup disorder, Hyperglycinuria
TRMT5	AR	3	Combined oxidative phosphorylation deficiency
TRPM6	AR	3	Hypomagnesemia 1, intestinal
WNK1	AD	3	Pseudohypoaldosteronism, type IIC
CACNA1S	AD	3	Hypokalemic periodic paralysis
GIF	AR	16	Intrinsic factor deficiency- tubular proteinuria
GSS	AR	16	Glutathione synthetase deficiency, RTA
WDR72	AR	18	Amelogenesis imperfecta, type IIA3, RTA
ATP6V1C2	AR	18	
SLC5A1	AR	19	Glucose/galactose malabsorption- Renal glucosuria
LCORL/ MLR1	AR	19	Pseudohypoaldosteronism type 1
COG6	AR	20	Congenital disorder of glycosylation, type Iil
CYP17A1	AR	3	17-alpha-hydroxylase 17,20-lyase deficiency
CYP27B1	AR	20	Vitamin D-dependent rickets, type I
SLC4A5	AR	20	
VIPAR	AR	20	Arthrogryposis, renal dysfunction, and cholestasis 2
AVP	AD	3	Diabetes insipidus, neurohypophyseal
FXYD6-FXYD2	AD	20	

AVPR2	XLR	4	Nephrogenic DI
CA2	AR	22	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis
FRTS	AD	22	Fanconi syndrome
HNF1A	AD	3	MODY III
FOXI1	AR	32	dRTA
NEDD4L	?	33	Renal tubular disease, hypertension related
CYP11B1	AR	3	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
CLDN10	AR	40	Hypokalemic-alkalotic salt-losing nephropathy
GATM	AR	46	Fanconi syndrome

Tumor

Gene	Inheritance	Reference	Syndrome
CDKN1B	AD	3	MEN type 4- Renal angiolypoma
DIS3L2	AR	3	Perlman's syndrome-nephroblastomatosis
ASXL1	AD	3	Bohring Opitz syndrome- Wilms tumor, rec infections
CDKN1C/ LIT1	AD	3	BWS- Increased frequency of kidney malformations, renal medullary dysplasia, nephrocalcinosis, and nephrolithiasis
FLCN	AD	3	Birt-Hogg-Dubé syndrome
MLH1	AD	3	Muir-Torre syndrome- genitourinary malignancies
MSH2	AD/AR	3	Muir-Torre syndrome, lynch- RCC
MSH6	AD/AR	3	Muir-Torre syndrome (paper), Lynch (OMIM)
NSD1	AD	3	Sotos, BWS
PIK3CA	AD	3	Cowden syndrome
SDHB	AD	3	Paragangliomas 4, Cowden syndrome
SDHC	AD	3	Cowden syndrome (paper), Paraganglioma (OMIM)
SDHD	AD	3	Pheochromocytoma Cowden syndrome 3
TSC1	AD	4	Tuberous sclerosis

ROBO1	?	50	RCC
TSC2	AD	3	Tuberous sclerosis
MET	AD	19	Papillary RCC

Mitochondrial disease

Gene	Inheritance	Reference	Syndrome
PDSS2	AR	15	
COQ2	AR	15	Coenzyme Q10 deficiency, primary, 1
COQ6	AR	15	Coenzyme Q10 deficiency, primary, 6
COQ9	AR	15	Coenzyme Q10 deficiency, primary, 5
SARS2	AR	15	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
MRPS22	AR	15	Combined oxidative phosphorylation deficiency
TSFM	AR	15	Combined oxidative phosphorylation deficiency 3
XPNPEP3	AR	15	Nephronophthisis-like nephropathy 1
SURF1	AR, MI	15	Leigh syndrome
COX10	AR, MI	15	Mitochondrial complex IV deficiency, Leigh syndrome
TMEM70	AR	15	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2
MPV17	AR	15	
C10orf2	AD/AR	15	
SUCLA2	AR	15	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
SUCLG1	AR	15	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)
DGUOK	AR	15	

APOPT1/ COA8	AR, MI	3	Mitochondrial complex IV deficiency
BCS1L	AR	3	GRACILE syndrome, Mitochondria complex 3 deficiency,nuclear type 1, Leigh
COQ7	AR	3	Coenzyme Q10 deficiency, primary, 8
COX14	AR, MI	3	Mitochondrial complex IV deficiency
COX20	AR, MI	3	Mitochondrial complex IV deficiency
COX6B1	AR, MI	3	Mitochondrial complex IV deficiency
COX7B	XLD	3	Linear skin defects with multiple congenital anomalies 2
COX8A	AR, MI	3	Mitochondrial complex IV deficiency
CPT1A	AR	3	CPT deficiency, hepatic, type IA
ETFA	AR	3	Glutaric acidemia IIA
ETFB	AR	3	Glutaric acidemia IIB
ETFDH	AR	3	Glutaric acidemia IIC
FASTKD2	AR, MI	3	Mitochondrial complex IV deficiency
FBXL4	AR	3	Mitochondrial DNA depletion syndrome 13
LONP1	AR	3	CODAS syndrome
PDSS1	AR	3	Coenzyme Q10 deficiency, primary, 2
PET100	AR, MI	3	Mitochondrial complex IV deficiency
RRM2B	AR	3	Mitochondrial DNA depletion syndrome 8A
SCO1	AR, Mi	3	Mitochondrial complex IV deficiency
SLC25A1	AR	3	Combined D-2- and L-2-hydroxyglutaric aciduria
STAR	AR	3	Lipoid adrenal hyperplasia
TACO1	AR, Mi	3	Mitochondrial complex IV deficiency
UQCC2	AR	3	Mitochondrial complex III deficiency
UQCRB	AR	22	Mitochondrial complex III deficiency, nuclear type 3
UQCRQ	AR	22	Mitochondrial complex III deficiency, nuclear type 4
HOGA1	AR	1	Hyperoxaluria, primary, type III
MTTL1	AR	1	MELAS

POLG	AR?	25	
NDUFAF2	AR	25	Mitochondrial complex I deficiency, nuclear type 10
ETHE1	AR	25	Ethylmalonic encephalopathy
TK2	AR	25	
MTTK	?, Mi	25	MELAS
MTND5	?, Mi	25, 39	MELAS, Pearson- deletetion
MTTF	?, Mi	25	MELAS, **m.G586A, m.A608G, T616C?
MTTY	?, Mi	25	MELAS, FSGS, **m.A5843G
MTND4	?, Mi	39	Pearson- deletetion
MTND3	?, Mi	28, 39	KSS- deletetion, Leigh, pearson- deletetion
MTND4L	?, Mi	28, 39	KSS-deletetion, pearson- deletetion
MTTL2	?, Mi	28	KSS-deletetion
MTTV	?, Mi	26	Leigh
MTND6	?, Mi	26	MELAS
MTTC	?, Mi	26	MELAS
MTTQ	?, Mi	26	MELAS
MTTH	?, Mi	26	MELAS
MTTI	?, Mi	24	**m.A4269G
MTTN	?, Mi	47	Isolated ophthalmoplegia, mitochondrial Complex I deficiency, **m.A5728G
MTTW	?, Mi	48	**G5538A
A547T	?, Mi	49	TIN
MTTS1	?, Mi	26	MELAS
MTTS2	?, Mi	26	MELAS
LRPPRC	AR	26	Leigh syndrome, French-Canadian type
NDUFA9	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 26
NDUFA10	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 22
NDUFA12	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 23
NDUFV1	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 4

NDUFS3	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 8
NDUFS4	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 1
NDUFS7	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 3
NDUFS8	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 2
SDHA	AR, AR Mi	26	Leigh, Mitochondrial respiratory chain complex II deficiency
MTND2	Mi	26	Leigh
MTATP6	Mi	26	Leigh
C8orf38/ NDUFAF6	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 17
COX15	AR, AR Mi	26	Leigh
DLD	AR	26	Leigh, Dihydrolipoamide dehydrogenase deficiency
FOXRED1	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 19
PDHA1	XLD	26	Leigh, Pyruvate dehydrogenase E1-alpha deficiency
MTFMT	AR	26	Leigh, Mitochondrial complex I deficiency, nuclear type 27
MTCO1/ COX1	?, Mi	39	Pearson- deletetion
ATPase6	?, Mi	39	Pearson- deletetion
ATPase8	?, Mi	39	Pearson- deletetion
CYTB	?, Mi	39	Pearson- deletetion
MTCO3/ COX3	?, Mi	27	Pearson- deletetion
MTCO2/ COX2	?, Mi	27	Pearson- deletetion

Others

Gene	Inheritance	Reference	Syndrome
HBB	AR	3	sickle cell nephropathy
NF1	AD	3	Neurofibromatosis, type 1
MC4R	AD/AR	3	Obesity (BMIQ20)
SLC16A12	AD	3	Cataract 47 juvenile with microcornea- renal glucosuria

ABCD4	AR	3	Methylmalonic aciduria and homocystinuria, cblJ type
ACVRL1	AD	3	HHT
ENG	AD	3	HHT
GDF2	AD	3	HHT
ADAMTS13	AR	3	TTP
AGPAT2	AR	3	Lipodystrophy, congenital generalized, type 1
AMN	AR	3	Megaloblastic anemia-1, Norwegian type- proteinuria (unknown mechanism)
ATP7B	AR	3	Wilson
CYP21A2	AR	3	21-hydroxylase deficiency
DLG3	XLR	3	Mental retardation, x-linked 90
DLX4	AD	3	Orofacial cleft 15
DNMT3B	AR	3	Immunodeficiency centromeric instability
FLNB	AR	3	Spondylocarpotarsal synostosis syndrome
GCDH	AR	3	Glutaricaciduria
HRAS	AD	3	Costello syndrome
HSD17B3	AR	3	17-beta- hydroxysteroid dehydrogenase 3 deficiency (on OMIM Pseudohermaphroditism, male, with gynecomastia)
HSPG2	AR	3	Schwartz-Jampel syndrome
KCNA1	AD	3	Episodic ataxia/myokymia syndrome
KDM1A	AD	3	Cleft palate, psychomotor retardation, and distinctive facial features
LMBRD1	AR	3	Methylmalonic aciduria and homocystinuria
LPIN1	AR	3	Myoglobinuria, acute recurrent
MMACHC	AR	3	Methylmalonic aciduria and homocystinuria- TMA
NOTCH3	AD	3	Infantile myofibromatosis, Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, Lateral meningocele syndrome
NR0B1	XLR	3	Adrenal hypoplasia, congenital
PGK1	XLR	3	Phosphoglycerate kinase 1 deficiency- Due to myoglobin
PLOD1	AR	3	Ehlers-Danlos syndrome, kyphoscoliotic type - renal artery anueysm

POMC	AR	3	Obesity, adrenal insufficiency, and red hair due to POMC deficiency
PSAP	AR	3	Metachromatic leukodystrophy due to SAP-b deficiency
PYGM	AR	3	McArdle disease- Myoglobinuria
SLC2A10	AR	3	Arterial tortuosity syndrome- RAS
SMAD3	AD	3	Loeys-Dietz syndrome- renal artey aneurysm
STUB1	AR	3	Spinocerebellar ataxia 16
TBCE	AR	3	Kenny-Caffey syndrome
TWIST2	AR	3	Focal facial dermal dysplasia 3, Setleis type
UMPS	AR	3	Orotic aciduria- orotic acid urinary obstruction
SUGCT	AR	3	Glutaric aciduria III
MMUT/ MUT	AR	3	Methylmalonic aciduria, mut(0) type
LIFR	AR	?	Stuve-Wiedemann syndrome
TMEM260	AR	64	
TMEM127	AD	21	

STROBE Statement—checklist of items that should be included in reports of observational studies

	Item No	Recommendation
Title and abstract	1	(*a*) Indicate the study's design with a commonly used term in the title or the abstract
		(*b*) Provide in the abstract an informative and balanced summary of what was done and what was found
Introduction		
Background/rationale	2	Explain the scientific background and rationale for the investigation being reported
Objectives	3	State specific objectives, including any prespecified hypotheses
Methods		
Study design	4	Present key elements of study design early in the paper
Setting	5	Describe the setting, locations, and relevant dates, including periods of recruitment, exposure, follow-up, and data collection
Participants	6	(*a*) *Cohort study*—Give the eligibility criteria, and the sources and methods of selection of participants. Describe methods of follow-up *Case-control study*—Give the eligibility criteria, and the sources and methods of case ascertainment and control selection. Give the rationale for the choice of cases and controls *Cross-sectional study*—Give the eligibility criteria, and the sources and methods of selection of participants
		(*b*) *Cohort study*—For matched studies, give matching criteria and number of exposed and unexposed *Case-control study*—For matched studies, give matching criteria and the number of controls per case
Variables	7	Clearly define all outcomes, exposures, predictors, potential confounders, and effect modifiers. Give diagnostic criteria, if applicable
Data sources/ measurement	8*	For each variable of interest, give sources of data and details of methods of assessment (measurement). Describe comparability of assessment methods if there is more than one group
Bias	9	Describe any efforts to address potential sources of bias

Study size	10	Explain how the study size was arrived at
Quantitative variables	11	Explain how quantitative variables were handled in the analyses. If applicable, describe which groupings were chosen and why
Statistical methods	12	(*a*) Describe all statistical methods, including those used to control for confounding
		(*b*) Describe any methods used to examine subgroups and interactions
		(*c*) Explain how missing data were addressed
		(*d*) *Cohort study*—If applicable, explain how loss to follow-up was addressed *Case-control study*—If applicable, explain how matching of cases and controls was addressed *Cross-sectional study*—If applicable, describe analytical methods taking account of sampling strategy
		(*e*) Describe any sensitivity analyses

Continued on next page

Results		
Participants	13*	(a) Report numbers of individuals at each stage of study—eg numbers potentially eligible, examined for eligibility, confirmed eligible, included in the study, completing follow-up, and analysed
		(b) Give reasons for non-participation at each stage
		(c) Consider use of a flow diagram
Descriptive data	14*	(a) Give characteristics of study participants (eg demographic, clinical, social) and information on exposures and potential confounders
		(b) Indicate number of participants with missing data for each variable of interest
		(c) *Cohort study*—Summarise follow-up time (eg, average and total amount)
Outcome data	15*	*Cohort study*—Report numbers of outcome events or summary measures over time
		Case-control study—Report numbers in each exposure category, or summary measures of exposure
		Cross-sectional study—Report numbers of outcome events or summary measures
Main results	16	(*a*) Give unadjusted estimates and, if applicable, confounder-adjusted estimates and their precision (eg, 95% confidence interval). Make clear which confounders were adjusted for and why they were included
		(*b*) Report category boundaries when continuous variables were categorized
		(*c*) If relevant, consider translating estimates of relative risk into absolute risk for a meaningful time period
Other analyses	17	Report other analyses done—eg analyses of subgroups and interactions, and sensitivity analyses
Discussion		
Key results	18	Summarise key results with reference to study objectives
Limitations	19	Discuss limitations of the study, taking into account sources of potential bias or imprecision. Discuss both direction and magnitude of any potential bias
Interpretation	20	Give a cautious overall interpretation of results considering objectives, limitations, multiplicity of analyses, results from similar studies, and other relevant evidence

Generalisability	21	Discuss the generalisability (external validity) of the study results
Other information		
Funding	22	Give the source of funding and the role of the funders for the present study and, if applicable, for the original study on which the present article is based

*Give information separately for cases and controls in case-control studies and, if applicable, for exposed and unexposed groups in cohort and cross-sectional studies.

Note: An Explanation and Elaboration article discusses each checklist item and gives methodological background and published examples of transparent reporting. The STROBE checklist is best used in conjunction with this article (freely available on the Web sites of PLoS Medicine at http://www.plosmedicine.org/, Annals of Internal Medicine at http://www.annals.org/, and Epidemiology at http://www.epidem.com/). Information on the STROBE Initiative is available at www.strobe-statement.org.