Supplemental Figure 2. List of inherited metabolic disorders presenting with kidney phenotypes, laboratory findings, OMIM references and hyperlinked IEMbase IDs.

Name (n=190)	Gene	OMIM#	IEMcode #	Inheritance	Tubulopathy	Glomerular diseases	Kidney cysts	Kidney stones	Malformations/anomalies	Others (multiple)	Laboratory investigations
INTERMEDIARY METABOLISM: NUTRIENTS (n=31)											
Urea cycle disorders and hyperammonemias											
Ornithine transcarbamylase deficiency	*OTC*	311250	IEM0058	XL						Kidney insufficiency	Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)
Argininosuccinate synthetase deficiency	*ASS*	215700	IEM0059	AR						Kidney insufficiency	Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)
Argininosuccinate lyase deficiency	*ASL*	207900	IEM0060	AR						Kidney insufficiency	Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)
Disorders of amino acid transport											
Nephropathic cystinosis	*CTNS*	219800 219900 219750	IEM0873	AR	Renal Fanconi Syndrome			Nephrocalcinosis Nephrolithiasis		Polyuria Kidney failure Kidney failure, chronic Kidney osteodystrophy	Amino acids (P), Cystine (WBC, PMN)
Cystinuria type A	*SLC3A1*	220100	IEM0068	AR				Urolithiasis Urolithiasis, cystine stones		Obstructive uropathy Kidney failure Kidney failure, chronic Urinary Infections Hematuria	Amino acids (P), Cystine (WBC, PMN)
Cystinuria type B	*SLC7A9*	220100	IEM0069	AD, AR				Urolithiasis Urolithiasis, cystine stones		Obstructive uropathy Kidney failure Kidney failure, chronic Urinary Infections Hematuria	Amino acids (P), Cystine (WBC, PMN)
Lysinuric protein intolerance	*SLC7A7*	222700	IEM0070	AR	Nephritis Kidney failure, end stage	Glomerulonephritis				Kidney failure	Amino acids (P, U), Ammonia (P), Ferritin (S), Lipid panel (S), Orotic acid (U)
Dicarboxylic aminoaciduria	*SLC1A1*	222730	IEM0071	AR				Urolithiasis			Amino acids (U)
Blue diaper syndrome	*PCSK1?*	211000	IEM1521	AR				Nephrocalcinosis			Calcium (P)
Organic acidurias											
Propionic acidemia due to propionyl-CoA carboxylase subunit alpha deficiency	*PCCA*	232000	IEM0124	AR						Kidney failure Kidney failure, chronic Temporary impairment of Kidney function	Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Propionic acidemia due to propionyl-CoA carboxylase subunit beta deficiency	*PCCB*	232000	IEM0125	AR						Kidney failure Kidney failure, chronic Temporary impairment of Kidney function	Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Glutaryl-CoA dehydrogenase deficiency	*GCDH*	231670	IEM0134	AR						Kidney failure, acute	ASAT/ALAT (P); Organic acids (U); Acylcarnitines (U, P, DBS)
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	*MMUT*	251000	IEM0127	AR	Kidney tubulopathy Tubulointerstitial nephritis Nephritis	Reduced glomerular filtration rate				Progressive Kidney impairment	Amino acids (P); Organic acids (U); Acylcarnitines (U, P, DBS); Anion gap
Disorders of tyrosine metabolism											
Homogentisic acid oxidase deficiency Alkaptonuria	*HGD*	203500	IEM0094	AR				Kidney stones		Kidney failure	Homogentisate (U)
Fumarylacetoacetase deficiency Tyrosinemia type 1	*FAH*	276700	IEM0096	AR	Kidney tubulopathy			Nephrocalcinosis		Kidney enlargement Kidney failure Kidney failure, chronic	Amino acids (P); Organic acids (U); Succinylacetone (DBS)
Disorders of the metabolism of sulfur-containing amino acids and hydrogen sulfide											
MIT sulfur dioxygenase deficiency Ethylmalonic encephalopathy	*ETHE1*	602473 608451	IEM0106	AR		Hematuria					Organic acid (U), acylcarnitines (P), thiosulphate (P), lactate (B)
Disorders of ornithine, proline and hydroxyproline metabolism											
MIT 4-hydroxy-2-oxoglutarate aldolase 1 deficiency deficiency Primary hyperoxaluria type 3	*HOGA1*	613616	IEM0906	AR				Nephrocalcinosis Nephrolithiasis		Kidney colic Kidney failure Kidney failure, chronic Urinary Infections Hematuria	Oxalic acid (P,U), Organic acids (U)
Disorders of lysine, hydroxylysine, and tryptophan metabolism											
3-Hydroxykynureninase deficiency	*KYNU*	605197	IEM0162	AR					Kidney hypoplasia		3-Hydroxykynurenine (P, U), NAD+ (P), Kynurenine (U), Xanthurenic acid (U)
3-hydroxyanthranilic acid 3,4-dioxygenase deficiency	*HAAO*	604521	IEM0163	AR					Kidney hypoplasia		3-Hydroxyanthranilic acid (P)
Disorders of glutathione metabolism											
5-Oxoprolinase deficiency	*OPLAH*	260005	IEM0052	AR				Urolithiasis		Kidney colic	5-Oxoproline (U)
Other disorders of peptide metabolism											

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Name (n=190)	Gene	OMIM#	IEMcode #	Inheritance	Tubulopathy	Glomerular diseases	Kidney cysts	Kidney stones	Malformations/anomalies	Others (multiple)	Laboratory investigations
X-prolyl aminopeptidase 3 deficiency	*XPNPEP3*	613159	IEM0145	AR				Nephrolithiasis		Kidney insufficiency	DNA
Disorders of galactose and fructose metabolism											
Aldolase B deficiency Hereditary fructose intolerance	*ALDOB*	229600	IEM0326	AR	Kidney tubulopathy						Glycerol (U), Glucose (P), Coagulation factors (P)
Disorders of gluconeogenesis											
Pyruvate carboxylase deficiency	*PC*	266150	IEM0372	AR	Kidney tubular acidosis						Amino acids (P), Lactate (P), Glucose (P), 3-OH-Butyrate/Acetoacetate (P)
Disorders of glycogen metabolism											
Glucose-6-phosphatase deficiency GSD1a	*G6PC*	232200	IEM0370	AR		Glomerulosclerosis Hyperfiltration				Kidney enlargement	ASAT/ALAT (P), Lactate (P), Glucose (S), Glycogen (L), Biotinidase (P)
Glucose-6-phosphate transporter deficiency GSD 1b	*SLC37A4*	232220	IEM0355	AR		Glomerulosclerosis Hyperfiltration					ASAT/ALAT (P), CK (P), Lactate (P), Glucose (P), Biotinidase (P)
Disorders of pentose metabolism											
Transaldolase deficiency	*TALDO1*	606003	IEM0329	AR	Kidney tubulopathy			Nephrocalcinosis		Kidney failure Kidney failure, chronic	ASAT/ALAT (P), ALP (S), GGT (S), Glucose (S), Total/direct bilirubin (S), Ferritin (S), Polyol (U)
Disorders of carbohydrate transmembrane transport and absorption											
Glucose transporter 2 deficiency Fanconi-Bickel syndrome	*SLC2A2*	227810	IEM0316	AR	Kidney tubular acidosis Kidney tubulopathy Kidney tubulopathy, generalized	Hyperfiltration			Nephromegaly	Kidney failure	AST/ALT (P), Glucose (S), Amino acids (U), Urinalysis, Lipid panel (S), Oligosaccharide (U), Galactose (RBC)
Intestinal sodium-glucose cotransporter 1 deficiency	*SLC5A1*	606824	IEM0317	AR				Urolithiasis			Glucose (U), Reducing sugars (stool)
Sucrase-isomaltase deficiency	*SI*	222900	IEM0318	AR				Urolithiasis			DNA
Lactase deficiency	*LCT*	223000	IEM0320	AR				Urolithiasis			Reducing sugars (stool)
INTERMEDIARY METABOLISM: ENERGY (n=46)											
Disorders of carnitine metabolism											
Carnitine palmitoyltransferase 1A deficiency	*CPT1A*	255120	IEM0628	AR	Kidney tubular acidosis						Organic acids (U), Acylcarnitines (DBS, P)
Carnitine palmitoyltransferase 2 deficiency	*CPT2*	255110	IEM0629	AR					Malformations (kidney)		Organic acids (U), Acylcarnitines (DBS, P)
Disorders of MIT fatty acid oxidation											
Electron transfer flavoprotein α subunit deficiency	*ETFA*	231680	IEM0238	AR			Kidney cysts		Congenital kidney anomalies		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)
Electron transfer flavoprotein β subunit deficiency	*ETFB*	231680	IEM0239	AR			Kidney cysts		Congenital kidney anomalies		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)
Multiple acyl-CoA dehydrogenase deficiency	*ETFDH*	231675	IEM0240	AR			Kidney cysts		Congenital kidney anomalies		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)
Disorders of creatine metabolism											
Arginine:glycine amidinotransferase aggregation syndrome Fanconi renotubular syndrome type 1	*GATM*	134600	IEM0043	AD	Kidney tubular acidosis					Kidney failure	Creatinine (P, U), Guanidino compounds (P, U)
Disorders of mtDNA-encoded oxidative phosphorylation proteins											
NADH dehydrogenase core subunit 5 deficiency	*MT-ND5*	252010	IEM0435	MIT						Kidney failure	Lactate (P)
Disorders of mtDNA-encoded tRNA and rRNA											
MIT tRNA(Leu) 1 deficiency	*MT-TL1*	590050	IEM0540	MIT						Kidney insufficiency	Lactate (P)
MIT tRNA(Leu) 2 deficiency	*MT-TL2*	590055	IEM0541	MIT						Kidney failure Kidney failure, acute	Lactate (P)
MIT tRNA(Phe) deficiency	*MT-TF*	590070	IEM0544	MIT						Kidney insufficiency	Lactate (P)
Disorders of complex I subunits and assembly factors											
Complex I assembly factor 2 deficiency	*NDUFAF2*	618233	IEM0438	AR	Kidney tubular acidosis						Lactate (P)
Transmembrane protein 126B deficiency	*TMEM126B*	618250	IEM0446	AR	Kidney tubular acidosis						Lactate (P)
Disorders of complex III subunits and assembly factors											
BCS1L deficiency GRACILE syndrome	*BCS1L*	603358	IEM0458	AR	Kidney tubulopathy Kidney tubulopathy, proximal Kidney Fanconi Syndrome						DNA

Supplemental Figure 2. List of inherited metabolic disorders presenting with kidney phenotypes, laboratory findings, OMIM references and hyperlinked IEMbase IDs.

Name (n=190)	Gene	OMIM#	IEMcode #	Inheritance	Tubulopathy	Glomerular diseases	Kidney cysts	Kidney stones	Malformations/anomalies	Others (multiple)	Laboratory investigations
UQCC2 deficiency	*UQCC2*	615824	IEM1077	AR	Kidney tubulopathy						Lactate (P)
Disorders of complex IV subunits and assembly factors											
TACO1 deficiency	*TACO1*	220110	IEM0477	AR	Kidney tubulopathy						Lactate (P)
Cytochrome c oxidase assembly factor 10 deficiency	*COX10*	220110 256000	IEM0470	AR	Proximal tubulopathy						Lactate (P), Hemoglobin (B)
Cytochrome c oxidase subunit NDUFA4 (COXFA4) deficiency	*NDUFA4*		IEM1149	AR	Kidney tubular acidosis						Lactate (P)
Disorders of complex V subunits and assembly factors											
Transmembrane protein 70 deficiency	*TMEM70*	614052	IEM0486	AR	Kidney tubulopathy						CK (P), Lactate (P), Ammonia (P), 3-Methylglutaconic acid (U)
MIT ATP synthase F1 assembly factor 2 deficiency	*ATPAF2*	604273	IEM1151	AR					Kidney hypoplasia		Lactate (P), Organic acids (U)
Disorders of coenzyme Q10 biosynthesis											
Prenyl diphosphate synthase subunit 1 deficiency	*PDSS1*	607429 607426	IEM0618	AR		Nephrotic syndrome					DNA
Prenyl diphosphate synthase subunit 2 deficiency	*PDSS2*	614652	IEM0619	AR		Nephrotic syndrome					Lactate (P), CoQ10 (M, P, WBC)
Coenzyme Q2 polyprenyltranferase deficiency	*COQ2*	609825 607426	IEM0620	AR		Nephrotic syndrome					Lactate (P), CoQ10 (M, P, WBC)
Coenzyme Q6 monooxygenase deficiency	*COQ6*	614650	IEM0622	AR		Nephrotic syndrome					DNA
Coenzyme Q7 hydroxylase deficiency	*COQ7*	616733	IEM0623	AR					Kidney dysplasia	Kidney dysfunction	Lactate (P), Organic acids (U)
Coenzyme Q8B (ADCK4) deficiency	*COQ8B*	615573	IEM0625	AR		Nephrotic syndrome Proteinuria				Kidney failure Kidney failure, chronic Kidney failure, end stage	Albumin (S), Proteins, total (U)
Coenzyme 9 deficiency	*COQ9*	614654	IEM0626	AR	Kidney tubulopathy						Lactate (P), CoQ10 (M, P, WBC)
Disorders of lipoic acid and iron-sulfur metabolism											
NFS1 deficiency	*NFS1*	603485	IEM1123	AR						Kidney failure	Lactate (P)
Disorders of MIT nucleotide pool maintenance											
MPV17 deficiency	*MPV17*	256810	IEM0494	AR	Kidney tubulopathy						Glucose (P); GGT (P); Activity respiratory chain complexes (I, III, IV, and V)
Mitochondrial thymidine kinase 2 deficiency	*TK2*	609560 188250	IEM0496	AR						Kidney failure	CK (P), Lactate (P)
MIT ribonucelotide reductase subunit 2 deficiency	*RRM2B*	604712	IEM0497	AD, AR	Kidney tubulopathy						Lactate (P), Amino acids (P)
Disorders of mtDNA replication and maintenance											
Single-stranded DNA-binding protein 1 deficiency	*SSBP1*	165510	IEM1533	AD						Nephropathy	DNA
tRNA methyltransferase 5 deficiency	*TRMT5*	616539	IEM0513	AR	Kidney tubulopathy						Lactate (P)
MIT RNA polymerase deficiency	*POLRMT*	601778	IEM1534		Renal Fanconi Syndrome						Lactate (P), 5-Methyl-THF (CSF)
TWINKLE mitochondrial DNA helicase deficiency	*TWNK*	271245 616138	IEM0495	AR	Proximal tubulopathy						DNA
Disorders of MIT transcript processing and modification											
Mitochondrial transcription factor A deficiency	*TFAM*	617156	IEM1153	AR	Kidney tubulopathy						ASAT/ALAT (P), Albumin (S), Bilirubin, conjugated (P)
Disorders of MIT aminoacyl-tRNA synthetases											
MIT leucyl-tRNA synthetase deficiency	*LARS2*	615300	IEM0560	AR						Kidney dysfunction	Lactate (P)
MIT seryl-tRNA synthetase deficiency	*SARS2*	613845	IEM0563	AR	Kidney salt loss					Polyuria Kidney failure	Lactate (P), Uric acid (S)
MIT prolyl-tRNA synthetase deficiency	*PARS2*	612036	IEM1078	AR						Progressive Kidney impairment	Lactate (P)
Disorders of the mitoribosome											
MIT ribosomal large subunit 3 deficiency	*MRPL3*	614582	IEM0516	AR	Tubulointerstitial nephritis Nephritis						Lactate (P), Ammonia (P)
RMND1 deficiency	*RMND1*	614922	IEM0524	AR						Kidney failure	Lactate (P)

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Name (n=190)	Gene	OMIM#	IEMcode #	Inheritance	Tubulopathy	Glomerular diseases	Kidney cysts	Kidney stones	Malformations/anomalies	Others (multiple)	Laboratory investigations
MIT ribosomal large subunit 44 deficiency	*MRPL44*	615395	IEM0517	AR						Kidney insufficiency	Lactate (P)
Combined Oxidative Phosphorylation Defect 5	*MRPS22*	611719	IEM0520	AR	Kidney tubulopathy						Lactate (P)
MIT ribosomal small subunit 7 deficiency	*MRPS7*	617872	IEM1156	AR						Kidney dysfunction	Lactate (P)
Disorders of MIT protein quality control											
HSPA9 deficiency	*HSPA9*	182170	IEM0597	AR					Hypoplastic kidney		Bone marrow stain; DNA
FBXL4 deficiency	*FBXL4*	615471	IEM0502	AR	Kidney tubular acidosis						Lactate (P)
Miscellaneous disorders associated with MIT dysfunction											
Maternally Inherited Deafness and Diabetes Ballinger-Wallace Syndrome	*MT-TL1, MT-TE, MT-TK*	520000	IEM1504	MIT		Focal Segmental Glomerulosclerosis Glomerulosclerosis					Glucose (P)
INTERMEDIARY METABOLISM: OTHERS (n=5)											
Disorders of glyoxylate and oxalate metabolism											
Glyoxylate reductase/hydroxypyruvate reductase deficiency Primary hyperoxaluria type 2	*GRHPR*	260000	IEM0905	AR				Nephrocalcinosis Nephrolithiasis		Kidney colic Kidney failure Kidney failure, chronic Urinary Infections Hematuria	Oxalic acid (U, P), Glyceric acid (U), Creatinine (P), Urea (P)
Hydroxyacid oxidase 1 deficiency	*HAO1*	605023	IEM0904	AR				Nephrolithiasis			Oxalic acid (P,U), Glycolic acid (P,U)
Oxalate transporter deficiency	*SLC26A1*	167030	IEM0907	AR				Nephrolithiasis		Kidney failure Kidney failure, acute	Oxalic acid (U)
Alanine-glyoxylate aminotransferase deficiency (peroxisomal) Primary hyperoxaluria type 1	*AGXT*	259900	IEM0903	AR				Nephrocalcinosis Nephrolithiasis		Kidney colic Kidney failure Kidney failure, chronic Urinary Infections Hematuria	Oxalic acid (U, P), Glycolic acid (P, U), Creatinine (P), Urea (P)
SLC26A6 deficiency	*SLC26A6*	610068	IEM1911	AD				Urolithiasis		Kidney colic	Oxalic acid (U), Citric acid (U)
LIPID METABOLISM AND TRANSPORT (n=21)											
Disorders of peroxisomal fatty acid oxidation											
Peroxisomal straight-chain acyl-CoA oxidase deficiency	*ACOX1*	264470	IEM0884	AR			Kidney cysts				VLCFA (P), Plasmalogens (P)
D-bifunctional protein deficiency Pseudo-Zellweger syndrome	*HSD17B4*	261515	IEM0885	AR			Kidney cysts				VLCFA (P), Plasmalogens (P), Organic acids (U)
L-bifunctional protein deficiency Fanconi renotubular syndrome type 3	*EHHADH*	615605	IEM0886	AR	Kidney tubulopathy Kidney tubulopathy, proximal Kidney Fanconi Syndrome						Amino acids (U), Glucose (U), Proteins, total (P)
Phytanoyl-CoA hydroxylase deficiency Refsum disease	*PHYH*	266500	IEM0888	AR		Haematuria					Pipecolic acid (P, U), Phytanic acid (S, U), Protein (CSF)
Disorders of glycerolipid metabolism											
Lipin 1 deficiency	*LPIN1*	268200	IEM0657	AR						Kidney failure, acute	CK (P), Myoglobin (U)
Disorders of glycerophospholipid metabolism											
Phosphatidylinositol 4,5-bisphosphate-5-phosphatase deficiency Lowe syndrome	*OCRL*	309000	IEM0692	XL		Proteinuria				Kidney failure Kidney failure, acute	Amino acids (U); Cholesterol (S); Phosphate (U)
Phosphatidylinositol 4,5-bisphosphate-5-phosphatase deficiency Dent disease type 2	*OCRL*	300555	IEM0692	XL	Kidney tubulopathy Kidney tubulopathy, proximal			Nephrocalcinosis		Kidney insufficiency	Amino acids (U); Cholesterol (S); Phosphate (U)
Peroxin 5 deficiency Zellweger	*PEX5*	214110	IEM0892	AR			Kidney cysts				ASAT/ALAT (P), VLCFA (P), Pipecolic acid (P, U)
Diacylglycerol kinase ε deficiency	*DGKE*	615008	IEM0675	AR		Glomerulopathy Hemolytic uremic syndrome (atypical)				Progressive Kidney impairment	DNA
Phosphatidylinositol 4,5-bisphosphate phospholipase C ε1 deficiency	*PLCE1*	610725	IEM0711	AR		Diffuse mesangial sclerosis Focal Segmental Glomerulosclerosis Glomerulosclerosis Nephrotic syndrome Proteinuria					Albumin (S), Proteins, total (P)
Phosphatidylinositol-4-phosphate 3-kinase catalytic subunit type 2 alpha deficiency	*PIK3C2A*	618440	IEM1265	AR				Nephrocalcinosis			Calcium (P), Mucopolysaccharides (U)
Disorders of sphingolipid synthesis and recycling											
Sphingosine-1-phosphate lyase deficiency	*SGPL1*	617575	IEM0682	AR		Nephrotic syndrome				Kidney insufficiency	ACTH (P); Glucose (P); Triglycerides (S); Albumin (U)
Disorders of sterol biosynthesis											

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CHILD syndrome, dominant	*NSDHL*	308050	IEM0747	XL					Kidney hypoplasia Kidney agenesis		Sterols (P)
Chondrodysplasia punctata 2, dominat Conradi-Hünermann syndrome	*EBP*	302960	IEM0749	XL					Kidney anomalies Kidney hypoplasia		Sterols (P)
Sterol C5-desaturase deficiency Lathosterolosis	*SC5D*	607330	IEM0751	AR							Sterols (P)
3β-hydroxysterol-delta24-reductase deficiency Desmosterol reductase deficiency	*DHCR24*	602398	IEM0752	AR					Kidney hypoplasia Kidney agenesis		Sterols (P)
7-dehydrocholesterol reductase deficiency Smith-Lemli-Opitz syndrome	*DHCR7*	270400	IEM0753	AR					Kidney hypoplasia Kidney agenesis		ASAT/ALAT (P), Lipid panel (S), 7/8-Dehydrocholesterol (P)
Disorders of bile acid metabolism											
Sterol 27-hydroxylase deficiency Cerebrotendinous Xanthomatosis	*CYP27A1*	213700	IEM0782	AR						Kidney insufficiency	Lipid panel (S); Sterols (P); Cholestane pentol glucuronide (U); 25-Hydroxy-Vitamin D (P)
Mixed hyperlipidemias											
Lipoprotein glomerulopathy	*APOE*	611771	IEM0727	AD		Glomerulopathy				Kidney failure	Lipid panel (S)
Disorders of high-density lipoprotein (HDL) metabolism											
Lecithin cholesterol acyl transferase deficiency (LCAT)	*LCAT*	606967 245900	IEM0734	AR		Proteinuria				Kidney failure	LDL/HDL cholesterol (P), Triglycerides (P), Apolipoprotein A-I
Hereditary apolipoprotein A1-related amyloidosis	*APOA1*	105200	IEM0737	AD		Hematuria					Lipid panel (S), Apolipoprotein A-I
METABOLISM OF HETEROCYCLIC COMPOUNDS (n=27)											
Disorders of pyrimidine metabolism											
Dihydroorotate dehydrogenase deficiency	*DHODH*	263750 126064	IEM0002	AR					Kidney anomalies		N-Carbamyl asparate (U), Purines and pyrimidines (U),
Uridine monophosphate synthase deficiency	*UMPS*	258900	IEM0003	AR				Urolithiasis		Haematuria	Purines and pyrimidines (U)
Disorders of purine metabolism											
Phosphoribosyl pyrophosphate synthetase 1 superactivity	*PRPS1*	300661	IEM0007	XL				Urolithiasis			Lactate (P), DNA
Xanthine oxidase deficiency	*XDH*	278300 607633	IEM0016	AR				Urolithiasis Urolithiasis, xanthine stones		Kidney failure Kidney failure, acute	Purines and pyrimidines (U, P), Uric acid (U)
Hypoxanthine guanine phosphoribosyltransferase deficiency Lesch-Nyhan syndrome	*HPRT1*	300322 308000	IEM0017	XL				Urolithiasis		Kidney failure Kidney failure, acute Urinary Infections Haematuria	Uric acid (U, P), Purines (U)
Adenine phosphoribosyl transferase deficiency	*APRT*	102600	IEM0018	AR				Urolithiasis		Kidney colic Kidney failure Kidney failure, acute Kidney failure, chronic Hematuria	Purines and pyrimidines (U, P)
Urate transporter 1 deficiency	*SLC22A12*	220150	IEM0024	AR	Exercise-induced acute Kidney failure with acute tubular necrosis			Urolithiasis		Kidney failure	Uric acid (P, U)
Urate voltage-driven efflux transporter 1 deficiency	*SLC2A9*	612076	IEM0025	AR	Exercise-induced acute Kidney failure with acute tubular necrosis					Kidney failure	Uric acid (P)
Disorders of ectonucleotide and nucleic acid metabolism											
3' Repair exonuclease 1 deficiency	*TREX1*	192315	IEM0026	AD						Nephropathy	ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 lysophosphatidylcholine
ABCC6 deficiency, severe	*ABCC6*	614473	IEM0036	AR				Nephrocalcinosis		Kidney artery calcification	Pyrophosphate (S), Lactate (P), DNA
Ectonucleotide pyrophosphatase-phosphodiesterase 1 deficiency	*ENPP1*	208000, 613312	IEM0037	AR	Hyperphosphaturia					Kidney artery calcification	ALP (P); Calcium (P); Phosphate (P)
Disorders of non-MIT tRNA processing and aminoacyl-tRNA synthetases											
Galloway-Mowat syndrome, YRDC type	*YRDC*	612276	IEM1293	AR		Nephrotic syndrome					DNA
Galloway-Mowat syndrome, GON7 type	*GON7*	617436	IEM1294	AR		Nephrotic syndrome					DNA
Galloway-Mowat syndrome type 2	*LAGE3*	301006	IEM1295	XL		Nephrotic syndrome					DNA
Galloway-Mowat syndrome type 3	*OSGEP*	617729	IEM1296	AR		Nephrotic syndrome					DNA
Galloway-Mowat syndrome type 4	*TP53RK*	617730	IEM1297	AR		Nephrotic syndrome Proteinuria					DNA
Galloway-Mowat syndrome type 5	*TPRKB*	617731	IEM1298	AR		Nephrotic syndrome Proteinuria					DNA
Galloway-Mowat syndrome type 6	*WDR4*	605924	IEM1300	AR		Nephrotic syndrome					DNA

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Phenylalanyl-tRNA synthetase subunit beta deficiency	*FARSB*	613658	IEM1314	AR		Proteinuria					DNA
Nephrotic syndrome, type 18 (NUP133)	*NUP133*	618177	IEM1903	AR		Nephrotic syndrome Proteinuria					Albumin (S), Proteins, total (U)
Nephrotic syndrome, type 11 (NUP107)	*NUP107*	618177 618348	IEM1904	AR		Nephrotic syndrome					DNA
Disorders of ribosomal biogenesis											
RBM28 deficiency	*RBM28*	612079	IEM1679	AR						Kidney insufficiency	IGF1 (P), Sterpoids (P)
Disorders of heme synthesis and degradation											
Coproporphyrinogen oxidase deficiency Hereditary coproporphyria	*CPOX*	121300	IEM0792	AD						Kidney failure	Urobilinogen (U), Porphyrines (U), Delta-ALA (U)
Delta-aminolevulinate dehydratase deficiency Doss porphyria	*ALAD*	125270	IEM0788	AR						Kidney failure	Porphyrines (U), Delta-ALA (U)
Protoporphyrinogen oxidase deficiency Porphyria variegata	*PPOX*	176200	IEM0794	AD						Kidney failure	PBG (U); Porphyrins (U, stools); Fluorescence scanning (P)
Porphobilinogen deaminase deficiency Acute intermittent porphyria	*HMBS*	176000	IEM0789	AD/AR						Kidney failure	Porphyrins (U); Porphobilinogen (U), Delta-ALA (U); DNA
Disorders of heme degradation and bilirubin metabolism											
Heme oxygenase 1 deficiency	*HMOX1*	614034	IEM0800	AR		Hematuria Proteinuria					Bilirubin (P), Haptoglobin (S), Proteins, total (P)
COMPLEX MOLECULE AND ORGANELLE METABOLISM (n=36)											
Disorders of N-linked protein glycosylation											
PMM2-CDG Phosphomannomutase 2 deficiency	*PMM2*	601785	IEM0908	AR		Proteinuria	Kidney cysts			Kidney enlargement	ASAT/ALAT (P), Lipid panel (S), Sialotransferrins (S), Albumin (S), Factor XI (B),
ALG1-CDG Mannosyltransferase 1 deficiency	*ALG1*	608540	IEM0914	AR		Nephrotic syndrome					Sialotransferrins (S), IGG (P), B cells, circulating (blood), Protein C (S)
ALG9-CDG Mannosyltransferase 7-9 deficiency	*ALG9*	608776	IEM0919	AR			Kidney cysts				Lipid panel (S), Sialotransferrins (S), Albumin (S), Factor XI (B)
ALG8-CDG Glucosyltransferase 2 deficiency	*ALG8*	608104	IEM0922	AD, AR	Kidney tubulopathy		Kidney cysts				CK (P), Sialotransferrins (S), Albumin (S), Factor IX and XII (B), Antithrombin III (P)
GANAB-CDG Alpha glucosidase II deficiency	*GANAB*	600666	IEM0928	AD			Polycystic Kidney				Sialotransferrins (S)
PRKCSH-CDG	*PRKCSH*	174050	IEM0929	AD			Polycystic Kidney				Sialotransferrins (S)
FUT8-CDG α-1,6-fucosyltransferase deficiency	*FUT8*	618005	IEM0932	AR				Nephrocalcinosis			Glucose (P); Blood count
ALG5-CDG	*ALG5*	604565	IEM1933	AD			Polycystic Kidney		Athropic Kidney	Kidney failure	DNA
B3GALTL-CDG O-Fucose-specific beta-1,3-N-glucosyltransferase deficiency	*B3GLCT*	261540	IEM0970	AR						Hydronephrosis	DNA
B4GAT1-CDG β-1,4-glucuronyltransferase 1 deficiency	*B4GAT1*	615287	IEM0943	AR			Kidney cysts		Kidney dysplasia	Hydronephrosis	CK (P), DNA
Disorders of lipid glycosylation											
Hyperuricemic nephropathy, familial juvenile 1	*UMOD*	162000	IEM1499	AD	Chronic interstitial nephritis Tubular atrophy Nephritis		Small medullary cysts			Nephropathy Kidney failure	Uric acid (P)
Disorders of multiple glycosylation pathways											
DHDDS-CDG Dehydrodolichyl diphosphate synthase deficiency	*DHDDS*	613861;60817 2	IEM0991	AR						Kidney failure Kidney failure, acute	DNA
Other disorders of glycan metabolism											
N-glycanase 1 deficiency	*NGLY1*	615273	IEM1032	AR						Kidney insufficiency	Oligosaccharides (U)
Peroxisomal biogenesis disorders											
Peroxin 1 deficiency Zellweger	*PEX1*	234580 214100 601539	IEM0889	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 2 deficiency Zellweger	*PEX2*	614866 614867	IEM0890	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 3 deficiency Zellweger	*PEX3*	617370 614882	IEM0891	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 6 deficiency Zellweger	*PEX6*	614862 614863 616617	IEM0893	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 10 deficiency Zellweger	*PEX10*	614870 614871	IEM0894	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 14B deficiency Zellweger	*PEX11B*	614920	IEM0895	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 12 deficiency Zellweger	*PEX12*	614859 266510	IEM0896	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)

Supplemental Figure 2. List of inherited metabolic disorders presenting with kidney phenotypes, laboratory findings, OMIM references and hyperlinked IEMbase IDs.

Name (n=190)	Gene	OMIM#	IEMcode #	Inheritance	Tubulopathy	Glomerular diseases	Kidney cysts	Kidney stones	Malformations/anomalies	Others (multiple)	Laboratory investigations
Peroxin 13 deficiency Zellweger	*PEX13*	614883 614885	IEM0897	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 14 deficiency Zellweger	*PEX14*	614887	IEM0898	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 16 deficiency Zellweger	*PEX16*	614876 614877	IEM0899	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 19 deficiency Zellweger	*PEX19*	614886	IEM0900	AR			Kidney cysts				VLCFA (P), Pipecolic acid (P)
Peroxin 26 deficiency Zellweger	*PEX26*	614872 614873	IEM0901	AR			Kidney cysts				VLCFA (P), Pristanic acid (S), Phytanic acid (S), AST/ALT (P), Plasmalogens (RBC), Pipecolic acid (S, U)
Disorders of lysosome-related organelle biogenesis											
Arthrogryposis-renal dysfunction-cholestasis syndrome type 1	*VPS33B*	208085	IEM1381	AR				Nephrocalcinosis		Kidney failure	DNA
Mucopolysaccharidosis-plus syndrome	*VPS33A*	617303	IEM0869	AR		Proteinuria					Mucopolysaccharides (U), Oligosaccharides (U)
Disorders of vesicular trafficking											
COG7-CDG Conserved oligomeric Golgi complex subunit 7 deficiency	*COG7*	608779	IEM1017	AR	Kidney tubulopathy					Obstructive uropathy	ASAT/ALAT (P), CK (P), Glucose (S), Total/direct bilirubin (S), Sialotransferins (S)
Arthrogryposis-renal dysfunction-cholestasis syndrome type 2	*VIPAS39*	613404	IEM1414	AR						Progressive Kidney impairment	DNA
Carpenter syndrome	*RAB23*	201000	IEM1436	AR						Hydronephrosis	DNA
STX5-CDG Syntaxin-5 deficiency	*STX5*	603189	IEM1674						Agenesis of left Kidney	Hydronephrosis Unilateral hydronephrosis	Alkaline phosphatase (P), IGF1 (P), Insulin (P), Coagulation factors (P), Ammonia (P)
Disorders of sphingolipid degradation											
Alpha-galactosidase A deficiency Fabry disease	*GLA*	301500	IEM0844	XL		Proteinuria				Kidney failure Kidney failure, chronic	Globotriaosylceramide (P); Globotriaosylsphingosine (P)
Alpha-neuraminidase deficiency Sialidosis	*NEU1*	256550	IEM0848	AR						Kidney failure Kidney failure, chronic	Oligosaccharides (U), Lysosomal enzymes (DBS)
Cathepsin A deficiency Galactosialidosis	*CTSA*	256540	IEM0849	AR		Proteinuria				Kidney failure	Oligosaccharides (U), Lysosomal enzymes (DBS)
Disorders of autophagy											
EPG5 deficiency Vici syndrome	*EPG5*	242840	IEM0811	AR	Kidney tubular acidosis						DNA
Other disorders of complex molecule degradation											
Glucocerebrosidase receptor deficiency	*SCARB2*	254900	IEM0875	AR						Kidney failure	Lysosomal enzymes (DBS)
COFACTOR AND MINERAL METABOLISM (n=21)											
Disorders of molybdenum cofactor metabolism											
Cyclic pyranopterin monophosphate synthase deficiency MoCo A	*MOCS1*	603707	IEM0275	AR				Nephrolithiasis			Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)
Molybdopterin synthase deficiency MoCo B	*MOCS2*	603708	IEM0276	AR				Nephrolithiasis			Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)
Gephyrin deficiency MoCo C	*GPHN*	603930	IEM0277	AR				Nephrolithiasis			Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)
Molybdenum cofactor sulfurase deficiency	*MOCOS*	603592	IEM0278	AR				Urolithiasis Urolithiasis, xanthine stones		Kidney failure Kidney failure, acute	Purines and pyrimidines (U, P), Uric acid (U)
Other disorders of vitamin metabolism											
Vitamin A receptor deficiency	*STRA6*	601186	IEM0256	AR					Hypoplastic kidney Horseshoe kidney Pelvic kidney	Hydronephrosis	DNA
Vitamin D 24-hydroxylase deficiency	*CYP24A1*	143880	IEM0269	AR				Nephrocalcinosis Nephrolithiasis		Polyuria	Calcium (U)
Disorders of niacin and NAD metabolism											
NAD synthetase 1 deficiency	*NADSYN1*	618845	IEM1559	AR					Hypoplastic or absent Kidneys Kidney anomalies		DNA
Disorders of folate metabolism											
5,10-Methylene-tetrahydrofolate dehydrogenase deficiency	*MTHFD1*	172460	IEM0224	AR		Hemolytic uremic syndrome (atypical)					Amino acids (P); Homocysteine (P); 5-Methyltetrahydrofolate (CSF), Folate (S)
Disorders of cobalamin metabolism											
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	277380	IEM0211	AR		Haematuria Haemolytic uraemic syndrome					Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P), Blood count
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*	614857	IEM0212	AR		Haematuria Haemolytic uraemic syndrome					Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P)
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	277400	IEM0213	AR		Haematuria Haemolytic uraemic syndrome					Homocysteine, total (P), B12 (S), Blood count, Organic acids (U), Acylcarnitines (DBS, P), Amino acids (P)

Supplemental Figure 2. List of inherited metabolic disorders presenting with kidney phenotypes, laboratory findings, OMIM references and hyperlinked IEMbase IDs.

Name (n=190)	Gene	OMIM#	IEMcode #	Inheritance	Tubulopathy	Glomerular diseases	Kidney cysts	Kidney stones	Malformations/anomalies	Others (multiple)	Laboratory investigations
Epi-cblC	*MMACHC* *PRDX1*	609831 176763	IEM0214	Digenic		Haematuria Haemolytic uraemic syndrome					Homocysteine, total (P), B12 (S), Blood count, Organic acids (U), Acylcarnitines (DBS, P), Amino acids (P)
Methylmalonic aciduria, cblDv2 type	*MMADHC*	277410	IEM0215	AR						Kidney failure Kidney failure, chronic	SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	277410	IEM0215	AR		Haematuria Haemolytic uraemic syndrome					SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Methylmalonic aciduria, cblA type	*MMAA*	251100	IEM0218	AR						Kidney failure Kidney failure, chronic	Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)
Methylmalonic aciduria, cblB type	*MMAB*	251110	IEM0219	AR						Kidney failure Kidney failure, chronic	Amino acids (P); Organic acids (U); Acylcarnitines (U, P, DBS); Anion gap
Disorders of copper metabolism											
Copper-transporting ATPase subunit beta deficiency Wilson disease	*ATP7B*	277900	IEM0279	AR	Kidney tubular acidosis						ASAT/ALAT (P); Copper (S, U); Ceruloplasmin (S)
Copper-transporting ATPase subunit alpha deficiency Menkes syndrome/Occipital horn syndrome	*ATP7A*	304150	IEM0280	XL						Urinary Infections Bladder diverticula	Copper (S, U), Ceruloplasmin (S)
Disorders of magnesium metabolism											
Sodium-potassium ATPase γ subunit deficiency	*FXYD2*	154020	IEM0307	AD						Kidney failure	Calcium (P, U, Magnesium (S, U)
KCNJ10 deficiency	*KCNJ10*	600791 612780	IEM0313	AR						Polydipsia Polyuria	Steroids (P), Calcium (P), Magnesium (P), Potassium (P)
Disorders of zinc metabolism											
Birk-Landau-Perez syndrome	*SLC30A9*	617595	IEM0300	AR	Tubulointerstitial nephritis Nephritis						Potassium (P)
OTHER (n=4)											
Monoamine neurotransmission											
Cytochrome b561 deficiency	*CYB561*	618182	IEM1049	AR	Kidney tubulopathy Kidney tubulopathy, generalized						Norepinephrine (P, U), Serotonin (S), Dopamine (P)
Disorders of insulin metabolism											
Kabuki syndrome	*KMT2D*	147920	IEM1492	AD					Congenital kidney anomalies		DNA
Disorders of steroid metabolism											
Cytochrome P450 oxidoreductase deficincy	*POR*	201750	IEM0760	AR					Kidney anomalies	Hydronephrosis Kidney insufficiency	Steroids (P, U)
Disorders of palmitoylation											
Porcupine palmitoyltransferase deficiency	*PORCN*	305600	IEM0688	XL					Horseshoe kidney	Hydronephrosis	DNA