Orphanet classification of rare renal diseases (ORPHA:93626)

ORPHAcode	Classification Level	
ORPHA:93626	Group of disorders	**Rare renal disease**
ORPHA:93545	Group of disorders	**Renal or urinary tract malformation**
ORPHA:93546	Group of disorders	**Non-syndromic renal or urinary tract malformation**
ORPHA:1851	Disorder	Multicystic dysplastic kidney
ORPHA:97363	Subtype of disorder	Unilateral multicystic dysplastic kidney
ORPHA:97364	Subtype of disorder	Bilateral multicystic dysplastic kidney
ORPHA:322	Disorder	Exstrophy-epispadias complex
ORPHA:93928	Subtype of disorder	Isolated epispadias
ORPHA:93929	Subtype of disorder	Cloacal exstrophy
ORPHA:93930	Subtype of disorder	Classic bladder exstrophy
ORPHA:3033	Disorder	Renal tubular dysgenesis
ORPHA:97367	Subtype of disorder	Renal tubular dysgenesis due to twin-twin transfusion
ORPHA:97368	Subtype of disorder	Drug-related renal tubular dysgenesis
ORPHA:97369	Subtype of disorder	Renal tubular dysgenesis of genetic origin
ORPHA:1309	Disorder	Medullary sponge kidney
ORPHA:2260	Disorder	Oligomeganephronia
ORPHA:237	Disorder	Duplication of urethra
ORPHA:617	Disorder	Congenital primary megaureter
ORPHA:238642	Subtype of disorder	Primary megaureter, adult-onset form
ORPHA:238646	Subtype of disorder	Congenital primary megaureter, obstructed form
ORPHA:238650	Subtype of disorder	Congenital primary megaureter, refluxing form
ORPHA:238654	Subtype of disorder	Congenital primary megaureter, nonrefluxing and unobstructed form
ORPHA:544578	Subtype of disorder	Congenital primary megaureter, refluxing and obstructed form
ORPHA:93101	Disorder	Renal hypoplasia
ORPHA:97361	Subtype of disorder	Renal hypoplasia, unilateral
ORPHA:97362	Subtype of disorder	Renal hypoplasia, bilateral
ORPHA:93108	Disorder	Renal dysplasia
ORPHA:93172	Subtype of disorder	Renal dysplasia, unilateral
ORPHA:93173	Subtype of disorder	Renal dysplasia, bilateral

ORPHA:93109	Disorder	Congenital megacalycosis
ORPHA:93176	Subtype of disorder	Unilateral congenital megacalycosis
ORPHA:93177	Subtype of disorder	Congenital bilateral megacalycosis
ORPHA:238637	Disorder	Megacystis-megaureter syndrome
ORPHA:289365	Disorder	Familial vesicoureteral reflux
ORPHA:411709	Disorder	Renal agenesis
ORPHA:1848	Subtype of disorder	Renal agenesis, bilateral
ORPHA:93100	Subtype of disorder	Renal agenesis, unilateral
ORPHA:435365	Group of disorders	**Fetal lower urinary tract obstruction**
ORPHA:2970	Disorder	Prune belly syndrome
ORPHA:105	Disorder	Atresia of urethra
ORPHA:93110	Disorder	Posterior urethral valve
ORPHA:435372	Disorder	Anterior urethral valve
ORPHA:435743	Group of disorders	**Congenital urachal anomaly**
ORPHA:488	Disorder	Urachal cyst
ORPHA:431341	Disorder	Patent urachus
ORPHA:431344	Disorder	Urachal sinus
ORPHA:431347	Disorder	Urachal diverticulum
ORPHA:652528	Disorder	Non-syndromic supernumerary kidneys
ORPHA:93547	Group of disorders	**Syndromic renal or urinary tract malformation**
ORPHA:881	Disorder	Turner syndrome
ORPHA:99226	Subtype of disorder	Monosomy X syndrome
ORPHA:99228	Subtype of disorder	Mosaic monosomy X syndrome
ORPHA:99413	Subtype of disorder	Turner syndrome due to structural X chromosome anomalies
ORPHA:138	Disorder	CHARGE syndrome
ORPHA:567	Disorder	22q11.2 deletion syndrome
ORPHA:783	Disorder	Rubinstein-Taybi syndrome
ORPHA:353277	Subtype of disorder	Rubinstein-Taybi syndrome due to CREBBP mutations
ORPHA:353281	Subtype of disorder	Rubinstein-Taybi syndrome due to 16p13.3 microdeletion
ORPHA:353284	Subtype of disorder	Rubinstein-Taybi syndrome due to EP300 haploinsufficiency
ORPHA:648	Disorder	Noonan syndrome
ORPHA:893	Disorder	WAGR syndrome
ORPHA:107	Disorder	BOR syndrome

ORPHA:195	Disorder	Cat-eye syndrome
ORPHA:52	Disorder	Alagille syndrome
ORPHA:261600	Subtype of disorder	Alagille syndrome due to 20p12 microdeletion
ORPHA:261619	Subtype of disorder	Alagille syndrome due to a JAG1 point mutation
ORPHA:261629	Subtype of disorder	Alagille syndrome due to a NOTCH2 point mutation
ORPHA:116	Disorder	Beckwith-Wiedemann syndrome
ORPHA:96076	Subtype of disorder	Beckwith-Wiedemann syndrome due to 11p15 microduplication
ORPHA:96193	Subtype of disorder	Beckwith-Wiedemann syndrome due to paternal uniparental disomy of chromosome 11
ORPHA:231117	Subtype of disorder	Beckwith-Wiedemann syndrome due to imprinting defect of 11p15
ORPHA:231120	Subtype of disorder	Beckwith-Wiedemann syndrome due to CDKN1C mutation
ORPHA:231127	Subtype of disorder	Beckwith-Wiedemann syndrome due to 11p15 microdeletion
ORPHA:231130	Subtype of disorder	Beckwith-Wiedemann syndrome due to 11p15 translocation/inversion
ORPHA:238613	Subtype of disorder	Beckwith-Wiedemann syndrome due to NSD1 mutation
ORPHA:564	Disorder	Meckel syndrome
ORPHA:289	Disorder	Ellis Van Creveld syndrome
ORPHA:3378	Disorder	Trisomy 13 syndrome
ORPHA:3380	Disorder	Trisomy 18 syndrome
ORPHA:887	Disorder	VACTERL/VATER association
ORPHA:373	Disorder	Simpson-Golabi-Behmel syndrome
ORPHA:3027	Disorder	Caudal regression syndrome
ORPHA:2052	Disorder	Fraser syndrome
ORPHA:955	Disorder	Hajdu-Cheney syndrome
ORPHA:971	Disorder	Acrorenal syndrome
ORPHA:1064	Disorder	Aniridia-renal agenesis-psychomotor retardation syndrome
ORPHA:1133	Disorder	AREDYLD syndrome
ORPHA:1756	Disorder	Caudal duplication
ORPHA:1834	Disorder	Axial mesodermal dysplasia spectrum
ORPHA:1896	Disorder	EEC syndrome
ORPHA:1973	Disorder	Faciocardiorenal syndrome
ORPHA:2111	Disorder	Cystic hamartoma of lung and kidney
ORPHA:2186	Disorder	Hydrocephalus-blue sclerae-nephropathy syndrome
ORPHA:2237	Disorder	Hypoparathyroidism-sensorineural deafness-renal disease syndrome
ORPHA:2241	Disorder	Megacystis-microcolon-intestinal hypoperistalsis syndrome

ORPHA:2256	Disorder	Fibulo-ulnar hypoplasia-renal anomalies syndrome
ORPHA:672	Disorder	Pallister-Hall syndrome
ORPHA:2278	Disorder	Ichthyosis-intellectual disability-dwarfism-renal impairment syndrome
ORPHA:2669	Disorder	Nephrosis-deafness-urinary tract-digital malformations syndrome
ORPHA:1475	Disorder	Renal coloboma syndrome
ORPHA:2673	Disorder	Neurofaciodigitorenal syndrome
ORPHA:2697	Disorder	Arthrogryposis-renal dysfunction-cholestasis syndrome
ORPHA:2704	Disorder	Urofacial syndrome
ORPHA:2750	Disorder	Orofaciodigital syndrome type 1
ORPHA:2774	Disorder	Multicentric carpo-tarsal osteolysis with or without nephropathy
ORPHA:2820	Disorder	Spastic paraplegia-nephritis-deafness syndrome
ORPHA:2838	Disorder	Renal caliceal diverticuli-deafness syndrome
ORPHA:3015	Disorder	Radio-renal syndrome
ORPHA:3032	Disorder	NPHP3-related Meckel-like syndrome
ORPHA:3109	Disorder	Mayer-Rokitansky-Küster-Hauser syndrome
ORPHA:2578	Subtype of disorder	Mayer-Rokitansky-Küster-Hauser syndrome type 2
ORPHA:247775	Subtype of disorder	Mayer-Rokitansky-Küster-Hauser syndrome type 1
ORPHA:798	Disorder	Schinzel-Giedion syndrome
ORPHA:3186	Disorder	Holoprosencephaly-radial heart renal anomalies syndrome
ORPHA:3316	Disorder	Thomas syndrome
ORPHA:3326	Disorder	Thymic-renal-anal-lung dysplasia
ORPHA:3327	Disorder	Thyrocerebrorenal syndrome
ORPHA:3404	Disorder	Ulbright-Hodes syndrome
ORPHA:1192	Disorder	Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome
ORPHA:3411	Disorder	Double uterus-hemivagina-renal agenesis syndrome
ORPHA:818	Disorder	Smith-Lemli-Opitz syndrome
ORPHA:71273	Disorder	Renal nutcracker syndrome
ORPHA:93111	Subtype of disorder	HNF1B-related autosomal dominant tubulointerstitial kidney disease
ORPHA:217266	Disorder	BNAR syndrome
ORPHA:439897	Disorder	Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome
ORPHA:444069	Disorder	Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome
ORPHA:500095	Disorder	Tall stature-intellectual disability-renal anomalies syndrome
ORPHA:500135	Disorder	Multinucleated neurons-anhydramnios-renal dysplasia-cerebellar hypoplasia-hydranencephaly syndrome

ORPHA:508488	Disorder	8q24.3 microdeletion syndrome
ORPHA:521438	Disorder	Congenital vertebral-cardiac-renal anomalies syndrome
ORPHA:592574	Disorder	Menke-Hennekam syndrome
ORPHA:597743	Disorder	SETD2-related microcephaly-severe intellectual disability-multiple congenital anomalies syndrome
ORPHA:656130	Disorder	PBX1-related congenital anomalies of kidney-urinary tract syndrome
ORPHA:689822	Disorder	Structural heart defects-renal anomalies syndrome
ORPHA:93548	Group of disorders	**Glomerular disease**
ORPHA:34145	Disorder	Immunoglobulin A nephropathy
ORPHA:54370	Disorder	Primary membranoproliferative glomerulonephritis
ORPHA:329903	Subtype of disorder	Immunoglobulin-mediated membranoproliferative glomerulonephritis
ORPHA:329918	Subtype of disorder	C3 glomerulopathy
ORPHA:93571	Subtype of disorder	Dense deposit disease
ORPHA:329931	Subtype of disorder	C3 glomerulonephritis
ORPHA:84087	Disorder	Collagen type III glomerulopathy
ORPHA:84090	Disorder	Fibronectin glomerulopathy
ORPHA:91137	Group of disorders	**Immunotactoid or fibrillary glomerulopathy**
ORPHA:97566	Disorder	Non-amyloid fibrillary glomerulopathy
ORPHA:97567	Disorder	Immunotactoid glomerulopathy
ORPHA:97560	Disorder	Primary membranous glomerulonephritis
ORPHA:329481	Disorder	Lipoprotein glomerulopathy
ORPHA:544590	Group of disorders	**Collagen-related glomerular basement membrane disease**
ORPHA:63	Disorder	Alport syndrome
ORPHA:1018	Subtype of disorder	X-linked Alport syndrome-diffuse leiomyomatosis
ORPHA:88917	Subtype of disorder	X-linked Alport syndrome
ORPHA:88918	Subtype of disorder	Autosomal dominant Alport syndrome
ORPHA:88919	Subtype of disorder	Autosomal recessive Alport syndrome
ORPHA:653722	Subtype of disorder	Digenic Alport syndrome
ORPHA:73229	Disorder	HANAC syndrome
ORPHA:567544	Disorder	Idiopathic non-lupus full-house nephropathy
ORPHA:567554	Group of disorders	**Systemic disease with glomerulopathy as a major feature**
ORPHA:86818	Disorder	Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome
ORPHA:567556	Group of disorders	**Genetic systemic disease with glomerulopathy as a major feature**
ORPHA:342	Disorder	Familial Mediterranean fever

ORPHA:575	Disorder	Muckle-Wells syndrome
ORPHA:36412	Disorder	Hypocomplementemic urticarial vasculitis
ORPHA:85450	Disorder	Hereditary amyloidosis with primary renal involvement
ORPHA:93560	Subtype of disorder	AApoAI amyloidosis
ORPHA:93561	Subtype of disorder	ALys amyloidosis
ORPHA:93562	Subtype of disorder	AFib amyloidosis
ORPHA:238269	Subtype of disorder	AApoAII amyloidosis
ORPHA:444092	Disorder	Autoimmune interstitial lung disease-arthritis syndrome
ORPHA:567558	Group of disorders	**Non-genetic systemic disease with glomerulopathy as a major feature**
ORPHA:732	Disorder	Polymyositis
ORPHA:221	Disorder	Dermatomyositis
ORPHA:645613	Subtype of disorder	Classical dermatomyositis
ORPHA:645617	Subtype of disorder	Amyopathic dermatomyositis
ORPHA:645626	Subtype of disorder	Adermatopathic dermatomyositis
ORPHA:117	Disorder	Behçet disease
ORPHA:797	Disorder	Sarcoidosis
ORPHA:809	Disorder	Mixed connective tissue disease
ORPHA:829	Disorder	Adult-onset Still disease
ORPHA:779	Disorder	Reynolds syndrome
ORPHA:85443	Disorder	AL amyloidosis
ORPHA:314701	Subtype of disorder	Primary systemic amyloidosis
ORPHA:314709	Subtype of disorder	Primary localized amyloidosis
ORPHA:85445	Disorder	AA amyloidosis
ORPHA:86861	Disorder	Non-amyloid monoclonal immunoglobulin deposition disease
ORPHA:93556	Subtype of disorder	Heavy chain deposition disease
ORPHA:93557	Subtype of disorder	Light and heavy chain deposition disease
ORPHA:93558	Subtype of disorder	Light chain deposition disease
ORPHA:90291	Disorder	Systemic sclerosis
ORPHA:220393	Subtype of disorder	Diffuse cutaneous systemic sclerosis
ORPHA:220402	Subtype of disorder	Limited cutaneous systemic sclerosis
ORPHA:220407	Subtype of disorder	Limited systemic sclerosis
ORPHA:93568	Disorder	Juvenile polymyositis
ORPHA:93672	Disorder	Juvenile dermatomyositis

ORPHA:439232	Disorder	AApoAIV amyloidosis
ORPHA:442582	Disorder	AH amyloidosis
ORPHA:449395	Subtype of disorder	IgG4-related kidney disease
ORPHA:567560	Group of disorders	**Systemic vasculitis associated with glomerulopathy**
ORPHA:536	Disorder	Systemic lupus erythematosus
ORPHA:183	Disorder	Eosinophilic granulomatosis with polyangiitis
ORPHA:375	Disorder	Anti-glomerular basement membrane disease
ORPHA:761	Disorder	Immunoglobulin A vasculitis
ORPHA:727	Disorder	Microscopic polyangiitis
ORPHA:900	Disorder	Granulomatosis with polyangiitis
ORPHA:3287	Disorder	Takayasu arteritis
ORPHA:397	Disorder	Giant cell arteritis
ORPHA:767	Disorder	Polyarteritis nodosa
ORPHA:439737	Subtype of disorder	Primary polyarteritis nodosa
ORPHA:439729	Subtype of disorder	Cutaneous polyarteritis nodosa
ORPHA:439755	Subtype of disorder	Single-organ polyarteritis nodosa
ORPHA:439762	Subtype of disorder	Systemic polyarteritis nodosa
ORPHA:439746	Subtype of disorder	Secondary polyarteritis nodosa
ORPHA:728	Disorder	Relapsing polychondritis
ORPHA:36258	Disorder	Buerger disease
ORPHA:91138	Disorder	Cryoglobulinemic vasculitis
ORPHA:93554	Subtype of disorder	Mixed cryoglobulinemia type II
ORPHA:93555	Subtype of disorder	Mixed cryoglobulinemia type III
ORPHA:93126	Disorder	Pauci-immune glomerulonephritis
ORPHA:97563	Subtype of disorder	Pauci-immune glomerulonephritis with ANCA
ORPHA:97564	Subtype of disorder	Pauci-immune glomerulonephritis without ANCA
ORPHA:93552	Disorder	Pediatric systemic lupus erythematosus
ORPHA:567562	Group of disorders	**Disorder with multisystemic involvement and glomerulopathy**
ORPHA:2614	Disorder	Nail-patella syndrome
ORPHA:1830	Disorder	Schimke immuno-osseous dysplasia
ORPHA:2065	Disorder	Galloway-Mowat syndrome
ORPHA:2613	Disorder	Nail-patella-like renal disease
ORPHA:2670	Disorder	Pierson syndrome

ORPHA:2715	Disorder	Severe oculo-renal-cerebellar syndrome
ORPHA:220	Disorder	Denys-Drash syndrome
ORPHA:347	Disorder	Frasier syndrome
ORPHA:69063	Disorder	Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization
ORPHA:69735	Disorder	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome
ORPHA:93114	Disorder	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E
ORPHA:163696	Disorder	Action myoclonus-renal failure syndrome
ORPHA:182050	Disorder	MYH9-related syndromic thrombocytopenia
ORPHA:280406	Disorder	Familial steroid-resistant nephrotic syndrome with sensorineural deafness
ORPHA:300333	Disorder	Nephrotic syndrome-epidermolysis bullosa-sensorineural deafness syndrome
ORPHA:306504	Disorder	Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome
ORPHA:506334	Disorder	Familial steroid-resistant nephrotic syndrome with adrenal insufficiency
ORPHA:567564	Group of disorders	**Nephrotic syndrome without extrarenal manifestations**
ORPHA:357502	Group of disorders	**Idiopathic nephrotic syndrome**
ORPHA:69061	Disorder	Idiopathic steroid-sensitive nephrotic syndrome
ORPHA:567546	Disorder	Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance
ORPHA:567548	Disorder	Idiopathic steroid-resistant nephrotic syndrome
ORPHA:567550	Subtype of disorder	Idiopathic multidrug-resistant nephrotic syndrome
ORPHA:567552	Subtype of disorder	Idiopathic steroid-resistant nephrotic syndrome with sensitivity to second-line immunosuppressive therapy
ORPHA:564127	Group of disorders	Genetic nephrotic syndrome
ORPHA:656	Disorder	Hereditary steroid-resistant nephrotic syndrome
ORPHA:839	Disorder	Congenital nephrotic syndrome, Finnish type
ORPHA:93573	Group of disorders	**Thrombotic microangiopathy**
ORPHA:536	Disorder	Systemic lupus erythematosus
ORPHA:54057	Disorder	Thrombotic thrombocytopenic purpura
ORPHA:93583	Subtype of disorder	Congenital thrombotic thrombocytopenic purpura
ORPHA:93585	Subtype of disorder	Immune-mediated thrombotic thrombocytopenic purpura
ORPHA:93552	Disorder	Pediatric systemic lupus erythematosus
ORPHA:244275	Disorder	De novo thrombotic microangiopathy after kidney transplantation
ORPHA:544458	Group of disorders	**Hemolytic uremic syndrome**
ORPHA:2134	Disorder	Atypical hemolytic uremic syndrome
ORPHA:93581	Subtype of disorder	Atypical hemolytic uremic syndrome with anti-factor H antibodies
ORPHA:544472	Subtype of disorder	Atypical hemolytic uremic syndrome with complement gene abnormality

ORPHA:79282	Subtype of disorder	Methylmalonic acidemia with homocystinuria, type cblC
ORPHA:357008	Disorder	Hemolytic uremic syndrome with DGKE deficiency
ORPHA:544482	Disorder	Infection-related hemolytic uremic syndrome
ORPHA:90038	Subtype of disorder	Shiga toxin-associated hemolytic uremic syndrome
ORPHA:544493	Subtype of disorder	Streptococcus pneumoniae-associated hemolytic uremic syndrome
ORPHA:93587	Group of disorders	**Genetic cystic renal disease**
ORPHA:731	Disorder	Autosomal recessive polycystic kidney disease
ORPHA:892	Disorder	Von Hippel-Lindau disease
ORPHA:564	Disorder	Meckel syndrome
ORPHA:289	Disorder	Ellis Van Creveld syndrome
ORPHA:2318	Disorder	Joubert syndrome with oculorenal defect
ORPHA:805	Disorder	Tuberous sclerosis complex
ORPHA:730	Disorder	Autosomal dominant polycystic kidney disease
ORPHA:1515	Disorder	Cranioectodermal dysplasia
ORPHA:2031	Disorder	Hepatic fibrosis-renal cysts-intellectual disability syndrome
ORPHA:3156	Disorder	Senior-Loken syndrome
ORPHA:2666	Disorder	Adult familial nephronophthisis-spastic quadriparesia syndrome
ORPHA:110	Disorder	Bardet-Biedl syndrome
ORPHA:655	Disorder	Nephronophthisis
ORPHA:93589	Subtype of disorder	Late-onset nephronophthisis
ORPHA:93591	Subtype of disorder	Infantile nephronophthisis
ORPHA:93592	Subtype of disorder	Juvenile nephronophthisis
ORPHA:34149	Disorder	Autosomal dominant tubulointerstitial kidney disease
ORPHA:88949	Subtype of disorder	MUC1-related autosomal dominant tubulointerstitial kidney disease
ORPHA:88950	Subtype of disorder	UMOD-related autosomal dominant tubulointerstitial kidney disease
ORPHA:93111	Subtype of disorder	HNF1B-related autosomal dominant tubulointerstitial kidney disease
ORPHA:217330	Subtype of disorder	REN-related autosomal dominant tubulointerstitial kidney disease
ORPHA:79118	Disorder	Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome
ORPHA:84081	Disorder	Senior-Boichis syndrome
ORPHA:88924	Disorder	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis
ORPHA:140969	Disorder	Saldino-Mainzer syndrome
ORPHA:140976	Disorder	RHYNS syndrome
ORPHA:220497	Disorder	Joubert syndrome with renal defect

ORPHA:294415	Disorder	Renal-hepatic-pancreatic dysplasia
ORPHA:401996	Disorder	Karyomegalic interstitial nephritis
ORPHA:443988	Disorder	Ventriculomegaly-cystic kidney disease
ORPHA:93593	Group of disorders	**Nephropathy secondary to a storage or other metabolic disease**
ORPHA:324	Disorder	Fabry disease
ORPHA:905	Disorder	Wilson disease
ORPHA:60	Disorder	Alpha-1-antitrypsin deficiency
ORPHA:912	Disorder	Zellweger syndrome
ORPHA:352	Group of disorders	**Galactosemia**
ORPHA:79237	Disorder	Galactokinase deficiency
ORPHA:79238	Disorder	Galactose epimerase deficiency
ORPHA:308473	Subtype of disorder	Erythrocyte galactose epimerase deficiency
ORPHA:308487	Subtype of disorder	Generalized galactose epimerase deficiency
ORPHA:79239	Disorder	Classic galactosemia
ORPHA:570422	Disorder	Galactose mutarotase deficiency
ORPHA:2116	Disorder	Hartnup disease
ORPHA:469	Disorder	Hereditary fructose intolerance
ORPHA:364	Disorder	Glycogen storage disease due to glucose-6-phosphatase deficiency
ORPHA:79258	Subtype of disorder	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia
ORPHA:79259	Subtype of disorder	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib
ORPHA:738	Group of disorders	**Porphyria**
ORPHA:659681	Group of disorders	**Erythropoietic porphyria**
ORPHA:79277	Disorder	Congenital erythropoietic porphyria
ORPHA:79278	Disorder	Autosomal erythropoietic protoporphyria
ORPHA:95159	Disorder	Hepatoerythropoietic porphyria
ORPHA:280379	Disorder	Erythropoietic uroporphyria associated with myeloid malignancy
ORPHA:443197	Disorder	X-linked erythropoietic protoporphyria
ORPHA:659672	Disorder	Harderoporphyria
ORPHA:659694	Group of disorders	**Hepatic porphyria**
ORPHA:95157	Group of disorders	**Acute hepatic porphyria**
ORPHA:79273	Disorder	Hereditary coproporphyria
ORPHA:79276	Disorder	Acute intermittent porphyria
ORPHA:79473	Disorder	Variegate porphyria

ORPHA:100924	Disorder	Porphyria due to ALA dehydratase deficiency
ORPHA:659698	Group of disorders	**Hepatic cutaneous porphyria**
ORPHA:101330	Disorder	Porphyria cutanea tarda
ORPHA:443057	Subtype of disorder	Sporadic porphyria cutanea tarda
ORPHA:443062	Subtype of disorder	Familial porphyria cutanea tarda
ORPHA:3467	Disorder	Hereditary xanthinuria
ORPHA:93601	Subtype of disorder	Xanthinuria type I
ORPHA:93602	Subtype of disorder	Xanthinuria type II
ORPHA:976	Disorder	Adenine phosphoribosyltransferase deficiency
ORPHA:650	Disorder	LCAT deficiency
ORPHA:79292	Subtype of disorder	Fish-eye disease
ORPHA:79293	Subtype of disorder	Familial LCAT deficiency
ORPHA:27	Disorder	Vitamin B12-unresponsive methylmalonic acidemia
ORPHA:79312	Subtype of disorder	Vitamin B12-unresponsive methylmalonic acidemia type mut-
ORPHA:289916	Subtype of disorder	Vitamin B12-unresponsive methylmalonic acidemia type mut0
ORPHA:1031	Disorder	Enamel-renal syndrome
ORPHA:3222	Disorder	Phosphoribosylpyrophosphate synthetase superactivity
ORPHA:411536	Subtype of disorder	Mild phosphoribosylpyrophosphate synthetase superactivity
ORPHA:411543	Subtype of disorder	Severe phosphoribosylpyrophosphate synthetase superactivity
ORPHA:28	Disorder	Vitamin B12-responsive methylmalonic acidemia
ORPHA:79310	Subtype of disorder	Vitamin B12-responsive methylmalonic acidemia type cblA
ORPHA:79311	Subtype of disorder	Vitamin B12-responsive methylmalonic acidemia type cblB
ORPHA:308442	Subtype of disorder	Vitamin B12-responsive methylmalonic acidemia, type cblDv2
ORPHA:882	Disorder	Tyrosinemia type 1
ORPHA:2088	Disorder	Fanconi-Bickel syndrome
ORPHA:416	Disorder	Primary hyperoxaluria
ORPHA:93598	Subtype of disorder	Primary hyperoxaluria type 1
ORPHA:93599	Subtype of disorder	Primary hyperoxaluria type 2
ORPHA:93600	Subtype of disorder	Primary hyperoxaluria type 3
ORPHA:35858	Disorder	Imerslund-Gräsbeck syndrome
ORPHA:69076	Disorder	Familial renal glucosuria
ORPHA:87876	Disorder	Sialidosis type 2
ORPHA:93399	Subtype of disorder	Juvenile sialidosis type 2

ORPHA:93400	Subtype of disorder	Congenital sialidosis type 2
ORPHA:206428	Group of disorders	Hypoxanthine-guanine phosphoribosyltransferase deficiency
ORPHA:510	Disorder	Lesch-Nyhan syndrome
ORPHA:79233	Disorder	Hypoxanthine guanine phosphoribosyltransferase partial deficiency
ORPHA:247794	Disorder	Juvenile cataract-microcornea-renal glucosuria syndrome
ORPHA:300547	Disorder	Autosomal recessive infantile hypercalcemia
ORPHA:324525	Disorder	Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation
ORPHA:371207	Group of disorders	**Congenital disorder of glycosylation with nephropathy as a major feature**
ORPHA:2953	Disorder	Musculocontractural Ehlers-Danlos syndrome
ORPHA:79325	Disorder	ALG8-CDG
ORPHA:238459	Disorder	SLC35A1-CDG
ORPHA:411629	Subtype of disorder	Infantile nephropathic cystinosis
ORPHA:411634	Subtype of disorder	Juvenile nephropathic cystinosis
ORPHA:620371	Disorder	Gitelman-like kidney tubulopathy due to mitochondrial DNA mutation
ORPHA:93603	Group of disorders	**Rare renal tubular disease**
ORPHA:534	Disorder	Oculocerebrorenal syndrome of Lowe
ORPHA:437	Group of disorders	**Hypophosphatemic rickets**
ORPHA:213	Disorder	Cystinosis
ORPHA:411629	Subtype of disorder	Infantile nephropathic cystinosis
ORPHA:411634	Subtype of disorder	Juvenile nephropathic cystinosis
ORPHA:411641	Subtype of disorder	Ocular cystinosis
ORPHA:1652	Disorder	Dent disease
ORPHA:93622	Subtype of disorder	Dent disease type 1
ORPHA:93623	Subtype of disorder	Dent disease type 2
ORPHA:89936	Disorder	X-linked hypophosphatemia
ORPHA:89937	Disorder	Autosomal dominant hypophosphatemic rickets
ORPHA:157215	Disorder	Hereditary hypophosphatemic rickets with hypercalciuria
ORPHA:244305	Disorder	Dominant hypophosphatemia with nephrolithiasis or osteoporosis
ORPHA:289176	Disorder	Autosomal recessive hypophosphatemic rickets
ORPHA:214	Disorder	Cystinuria
ORPHA:93612	Subtype of disorder	Cystinuria type A
ORPHA:93613	Subtype of disorder	Cystinuria type B
ORPHA:112	Disorder	Bartter syndrome

ORPHA:89938	Subtype of disorder	Bartter syndrome type 4
ORPHA:93605	Subtype of disorder	Bartter syndrome type 3
ORPHA:570371	Subtype of disorder	Bartter syndrome type 5
ORPHA:620217	Subtype of disorder	Bartter syndrome type 1
ORPHA:620220	Subtype of disorder	Bartter syndrome type 2
ORPHA:474	Disorder	Jeune syndrome
ORPHA:358	Disorder	Gitelman syndrome
ORPHA:64	Disorder	Alström syndrome
ORPHA:1380	Disorder	Cataract-nephropathy-encephalopathy syndrome
ORPHA:3145	Disorder	Arginine vasopressin resistance-intracranial calcification-short stature-facial dysmorphism syndrome
ORPHA:2197	Disorder	Idiopathic hypercalciuria
ORPHA:223	Disorder	Arginine vasopressin resistance
ORPHA:3337	Disorder	Primary Fanconi renotubular syndrome
ORPHA:30924	Disorder	Primary hypomagnesemia with secondary hypocalcemia
ORPHA:34528	Disorder	Autosomal dominant primary hypomagnesemia with hypocalciuria
ORPHA:73224	Disorder	Kidney tubulopathy-dilated cardiomyopathy syndrome
ORPHA:91136	Disorder	Acquired monoclonal Ig light chain-associated Fanconi syndrome
ORPHA:91500	Disorder	Tubulointerstitial nephritis and uveitis syndrome
ORPHA:93606	Disorder	Nephrogenic syndrome of inappropriate antidiuresis
ORPHA:94088	Disorder	Hereditary renal hypouricemia
ORPHA:97593	Group of disorders	**Pseudohypoparathyroidism**
ORPHA:457059	Group of disorders	**Pseudohypoparathyroidism with Albright hereditary osteodystrophy**
ORPHA:79443	Disorder	Pseudohypoparathyroidism type 1A
ORPHA:79444	Disorder	Pseudohypoparathyroidism type 1C
ORPHA:79445	Disorder	Pseudopseudohypoparathyroidism
ORPHA:457062	Group of disorders	**Pseudohypoparathyroidism without Albright hereditary osteodystrophy**
ORPHA:94089	Disorder	Pseudohypoparathyroidism type 1B
ORPHA:94090	Disorder	Pseudohypoparathyroidism type 2
ORPHA:199326	Disorder	Isolated autosomal dominant hypomagnesemia, Glaudemans type
ORPHA:199343	Disorder	EAST syndrome
ORPHA:238517	Group of disorders	**Hypotonia-cystinuria type 1 syndrome**
ORPHA:163690	Disorder	Hypotonia-cystinuria syndrome
ORPHA:163693	Disorder	2p21 microdeletion syndrome

ORPHA:238523	Disorder	Atypical hypotonia-cystinuria syndrome
ORPHA:306516	Disorder	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis
ORPHA:2196	Subtype of disorder	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement
ORPHA:31043	Subtype of disorder	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement
ORPHA:314822	Group of disorders	Primary renal tubular acidosis
ORPHA:2785	Disorder	Osteopetrosis with renal tubular acidosis
ORPHA:3240	Disorder	Early-onset progressive leukoencephalopathy-central nervous system calcification-deafness-visual impairment syndrome
ORPHA:18	Disorder	Distal renal tubular acidosis
ORPHA:93608	Subtype of disorder	Autosomal dominant distal renal tubular acidosis
ORPHA:93610	Subtype of disorder	Distal renal tubular acidosis with anemia
ORPHA:402041	Subtype of disorder	Autosomal recessive distal renal tubular acidosis
ORPHA:47159	Disorder	Proximal renal tubular acidosis
ORPHA:93607	Subtype of disorder	Autosomal recessive proximal renal tubular acidosis
ORPHA:314889	Subtype of disorder	Autosomal dominant proximal renal tubular acidosis
ORPHA:324525	Disorder	Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation
ORPHA:352540	Disorder	Oncogenic osteomalacia
ORPHA:363534	Disorder	Mitochondrial DNA depletion syndrome, hepatocerebrorenal form
ORPHA:363694	Disorder	Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome
ORPHA:444916	Group of disorders	**Pseudohypoaldosteronism**
ORPHA:757	Disorder	Pseudohypoaldosteronism type 2
ORPHA:88938	Subtype of disorder	Pseudohypoaldosteronism type 2A
ORPHA:88939	Subtype of disorder	Pseudohypoaldosteronism type 2B
ORPHA:88940	Subtype of disorder	Pseudohypoaldosteronism type 2C
ORPHA:300525	Subtype of disorder	Pseudohypoaldosteronism type 2D
ORPHA:300530	Subtype of disorder	Pseudohypoaldosteronism type 2E
ORPHA:756	Disorder	Pseudohypoaldosteronism type 1
ORPHA:171871	Subtype of disorder	Renal pseudohypoaldosteronism type 1
ORPHA:171876	Subtype of disorder	Generalized pseudohypoaldosteronism type 1
ORPHA:93164	Disorder	Transient pseudohypoaldosteronism
ORPHA:505242	Disorder	Psychomotor regression-oculomotor apraxia-movement disorder-nephropathy syndrome
ORPHA:528105	Disorder	Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome
ORPHA:544628	Disorder	Atypical Fanconi syndrome-neonatal hyperinsulinism syndrome
ORPHA:564178	Disorder	Primary hypomagnesemia-refractory seizures-intellectual disability syndrome

ORPHA:620363	Disorder	Primary hypomagnesemia-generalized seizures-intellectual disability-obesity syndrome
ORPHA:620368	Disorder	EGF-related primary hypomagnesemia with intellectual disability
ORPHA:620371	Disorder	Gitelman-like kidney tubulopathy due to mitochondrial DNA mutation
ORPHA:688581	Disorder	Midface hypoplasia-hearing impairment-elliptocytosis-nephrocalcinosis syndrome
ORPHA:93614	Group of disorders	**Hematological disorder with renal involvement**
ORPHA:848	Disorder	Beta-thalassemia
ORPHA:231214	Subtype of disorder	Beta-thalassemia major
ORPHA:231222	Subtype of disorder	Beta-thalassemia intermedia
ORPHA:231226	Subtype of disorder	Dominant beta-thalassemia
ORPHA:232	Disorder	Sickle cell anemia
ORPHA:84	Disorder	Fanconi anemia
ORPHA:93618	Group of disorders	**Rare cause of hypertension**
ORPHA:97598	Disorder	Congenital renal artery stenosis
ORPHA:156629	Group of disorders	**Rare genetic cause of hypertension**
ORPHA:904	Disorder	Williams syndrome
ORPHA:636	Disorder	Neurofibromatosis type 1
ORPHA:97685	Subtype of disorder	17q11 microdeletion syndrome
ORPHA:363700	Subtype of disorder	Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion
ORPHA:526	Disorder	Liddle syndrome
ORPHA:1276	Disorder	Brachydactyly-arterial hypertension syndrome
ORPHA:424	Disorder	Familial hyperthyroidism due to mutations in TSH receptor
ORPHA:757	Disorder	Pseudohypoaldosteronism type 2
ORPHA:88938	Subtype of disorder	Pseudohypoaldosteronism type 2A
ORPHA:88939	Subtype of disorder	Pseudohypoaldosteronism type 2B
ORPHA:88940	Subtype of disorder	Pseudohypoaldosteronism type 2C
ORPHA:300525	Subtype of disorder	Pseudohypoaldosteronism type 2D
ORPHA:300530	Subtype of disorder	Pseudohypoaldosteronism type 2E
ORPHA:758	Disorder	Pseudoxanthoma elasticum
ORPHA:320	Disorder	Apparent mineralocorticoid excess
ORPHA:88659	Disorder	Autosomal dominant progressive nephropathy with hypertension
ORPHA:88660	Disorder	Hypertension due to gain-of-function mutations in the mineralocorticoid receptor
ORPHA:235936	Group of disorders	**Familial hyperaldosteronism**
ORPHA:403	Disorder	Familial hyperaldosteronism type I

ORPHA:404	Disorder	Familial hyperaldosteronism type II
ORPHA:251274	Disorder	Familial hyperaldosteronism type III
ORPHA:642671	Disorder	Familial hyperaldosteronism type IV
ORPHA:688649	Disorder	Isolated adrenal medullary hyperplasia
ORPHA:693839	Disorder	Renal arteriovenous malformation
ORPHA:93619	Group of disorders	**Rare renal tumor**
ORPHA:2665	Disorder	Congenital mesoblastic nephroma
ORPHA:654	Disorder	Nephroblastoma
ORPHA:2111	Disorder	Cystic hamartoma of lung and kidney
ORPHA:97366	Disorder	Multiloculated renal cyst
ORPHA:217071	Group of disorders	**Renal cell carcinoma**
ORPHA:247203	Disorder	Collecting duct carcinoma
ORPHA:319276	Disorder	Clear cell renal carcinoma
ORPHA:319287	Subtype of disorder	Multilocular cystic renal neoplasm of low malignant potential
ORPHA:404511	Subtype of disorder	Clear cell papillary renal cell carcinoma
ORPHA:319298	Disorder	Papillary renal cell carcinoma
ORPHA:319303	Disorder	Chromophobe renal cell carcinoma
ORPHA:319308	Disorder	MiT family translocation renal cell carcinoma
ORPHA:319319	Disorder	Renal medullary carcinoma
ORPHA:319322	Disorder	Mucinous tubular and spindle cell renal carcinoma
ORPHA:319325	Disorder	Tubulocystic renal cell carcinoma
ORPHA:404514	Disorder	Acquired cystic disease-associated renal cell carcinoma
ORPHA:457246	Disorder	Clear cell sarcoma of kidney
ORPHA:464359	Disorder	Benign metanephric tumor
ORPHA:268316	Disorder	Complication in hemodialysis
ORPHA:319328	Group of disorders	**Inherited renal cancer-predisposing syndrome**
ORPHA:892	Disorder	Von Hippel-Lindau disease
ORPHA:893	Disorder	WAGR syndrome
ORPHA:116	Disorder	Beckwith-Wiedemann syndrome
ORPHA:96076	Subtype of disorder	Beckwith-Wiedemann syndrome due to 11p15 microduplication
ORPHA:96193	Subtype of disorder	Beckwith-Wiedemann syndrome due to paternal uniparental disomy of chromosome 11
ORPHA:231117	Subtype of disorder	Beckwith-Wiedemann syndrome due to imprinting defect of 11p15
ORPHA:231120	Subtype of disorder	Beckwith-Wiedemann syndrome due to CDKN1C mutation

ORPHA:231127	Subtype of disorder	Beckwith-Wiedemann syndrome due to 11p15 microdeletion
ORPHA:231130	Subtype of disorder	Beckwith-Wiedemann syndrome due to 11p15 translocation/inversion
ORPHA:238613	Subtype of disorder	Beckwith-Wiedemann syndrome due to NSD1 mutation
ORPHA:805	Disorder	Tuberous sclerosis complex
ORPHA:2849	Disorder	Perlman syndrome
ORPHA:122	Disorder	Birt-Hogg-Dubé syndrome
ORPHA:523	Disorder	Hereditary leiomyomatosis and renal cell cancer
ORPHA:47044	Disorder	Hereditary papillary renal cell carcinoma
ORPHA:97290	Disorder	Familial papillary thyroid carcinoma with renal papillary neoplasia
ORPHA:99880	Disorder	Hyperparathyroidism-jaw tumor syndrome
ORPHA:319462	Disorder	Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations
ORPHA:404476	Disorder	Global developmental delay-lung cysts-overgrowth-Wilms tumor syndrome
ORPHA:422526	Disorder	Hereditary clear cell renal cell carcinoma
ORPHA:664912	Disorder	Neonatal renal venous thrombosis