Amelie van der Ven

List of Publications by Year
in descending order

162	12,976	22153	26613
papers	citations	59 h-index	107 g-index

166	166	166	13429
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Whole-exome sequencing identifiesFOXL2,FOXA2andFOXA3as candidate genes for monogenic congenital anomalies of the kidneys and urinary tract. Nephrology Dialysis Transplantation, 2022, 37, 1833-1843.	0.7	6
2	Reverse phenotyping facilitates disease allele calling in exome sequencing of patients with CAKUT. Genetics in Medicine, 2022, 24, 307-318.	2.4	13
3	Whole exome sequencing identifies potential candidate genes for spina bifida derived from mouse models. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
4	Proteomic analysis identifies ZMYM2 as endogenous binding partner of TBX18 protein in 293 and A549 cells. Biochemical Journal, 2022, 479, 91-109.	3.7	6
5	The utility of a genetic kidney disease clinic employing a broad range of genomic testing platforms: experience of the Irish Kidney Gene Project. Journal of Nephrology, 2022, 35, 1655-1665.	2.0	14
6	Inhibition of endoplasmic reticulum stress signaling rescues cytotoxicity of human apolipoprotein-L1 risk variants in Drosophila. Kidney International, 2022, 101, 1216-1231.	5.2	15
7	Expression of a Truncated Form of ODAD1 Associated with an Unusually Mild Primary Ciliary Dyskinesia Phenotype. International Journal of Molecular Sciences, 2022, 23, 1753.	4.1	6
8	A Novel form of Familial Vasopressin Deficient Diabetes Insipidus Transmitted in an X-linked Recessive manner. Journal of Clinical Endocrinology and Metabolism, 2022, , .	3.6	0
9	Recessive Mutations in SYNPO2 as a Candidate of Monogenic Nephrotic Syndrome. Kidney International Reports, 2021, 6, 472-483.	0.8	7
10	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471.	0.8	2
11	Mutations in transcription factor CP2-like 1 may cause a novel syndrome with distal renal tubulopathy in humans. Nephrology Dialysis Transplantation, 2021, 36, 237-246.	0.7	0
12	Recessive NOS1AP variants impair actin remodeling and cause glomerulopathy in humans and mice. Science Advances, 2021, 7, .	10.3	21
13	Mutations in PRDM15 Are a Novel Cause of Galloway-Mowat Syndrome. Journal of the American Society of Nephrology: JASN, 2021, 32, 580-596.	6.1	15
14	A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. Genetics in Medicine, 2021, 23, 1158-1162.	2.4	13
15	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. American Journal of Human Genetics, 2021, 108, 357-367.	6.2	14
16	Copy Number Variant Analysis and Genome-wide Association Study Identify Loci with Large Effect for Vesicoureteral Reflux. Journal of the American Society of Nephrology: JASN, 2021, 32, 805-820.	6.1	17
17	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89
18	Immunological Impact of a Gluten-Free Dairy-Free Diet in Children With Kidney Disease: A Feasibility Study. Frontiers in Immunology, 2021, 12, 624821.	4.8	11

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19	Homozygous <scp>WNT9B</scp> variants in two families with bilateral renal agenesis/hypoplasia/dysplasia. American Journal of Medical Genetics, Part A, 2021, 185, 3005-3011.	1.2	5
20	ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. Neurogenetics, 2021, 22, 263-269.	1.4	8
21	A discarded synonymous variant in NPHP3 explains nephronophthisis and congenital hepatic fibrosis in several families. Human Mutation, 2021, 42, 1221-1228.	2.5	12
22	Exome survey of individuals affected by VATER / VACTERL with renal phenotypes identifies phenocopies and novel candidate genes. American Journal of Medical Genetics, Part A, 2021, 185, 3784-3792.	1.2	6
23	Multisystem inflammation and susceptibility to viral infections in human ZNFX1 deficiency. Journal of Allergy and Clinical Immunology, 2021, 148, 381-393.	2.9	40
24	Ttc30a affects tubulin modifications in a model for ciliary chondrodysplasia with polycystic kidney disease. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	6
25	Cystin genetic variants cause autosomal recessive polycystic kidney disease associated with altered Myc expression. Scientific Reports, 2021, 11, 18274.	3.3	13
26	Prevalence and clinical prediction of mitochondrial disorders in a large neuropediatric cohort. Clinical Genetics, 2021, 100, 766-770.	2.0	5
27	A truncating NRIP1 variant in an Arabic family with congenital anomalies of the kidneys and urinary tract. American Journal of Medical Genetics, Part A, 2021, , .	1.2	2
28	Whole-Exome Sequencing in Critically Ill Neonates and Infants: Diagnostic Yield and Predictability of Monogenic Diagnosis. Neonatology, 2021, 118, 454-461.	2.0	16
29	Deep learning is widely applicable to phenotyping embryonic development and disease. Development (Cambridge), 2021, 148, .	2.5	16
30	Whole exome sequencing identifies monogenic forms of nephritis in a previously unsolved cohort of children with steroid-resistant nephrotic syndrome and hematuria. Pediatric Nephrology, 2021, , 1.	1.7	0
31	Sequencing the CaSR locus in Pakistani stone formers reveals a novel loss-of-function variant atypically associated with nephrolithiasis. BMC Medical Genomics, 2021, 14, 266.	1.5	1
32	Personalized medicine in chronic kidney disease by detection of monogenic mutations. Nephrology Dialysis Transplantation, 2020, 35, 390-397.	0.7	58
33	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
34	Whole exome sequencing identified ATP6V1C2 as a novel candidate gene for recessive distal renal tubular acidosis. Kidney International, 2020, 97, 567-579.	5.2	42
35	Utility of Genomic Testing after Renal Biopsy. American Journal of Nephrology, 2020, 51, 43-53.	3.1	15
36	DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. American Journal of Human Genetics, 2020, 107, 1113-1128.	6.2	12

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37	Podocytopathies. Nature Reviews Disease Primers, 2020, 6, 68.	30.5	237
38	Beyond the tubule: pathological variants of LRP2, encoding the megalin receptor, result in glomerular loss and early progressive chronic kidney disease. American Journal of Physiology - Renal Physiology, 2020, 319, F988-F999.	2.7	13
39	Loss ofAnks6leads to YAP deficiency and liver abnormalities. Human Molecular Genetics, 2020, 29, 3064-3080.	2.9	11
40	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
41	ADCK4 Deficiency Destabilizes the Coenzyme Q Complex, Which Is Rescued by 2,4-Dihydroxybenzoic Acid Treatment. Journal of the American Society of Nephrology: JASN, 2020, 31, 1191-1211.	6.1	38
42	Mouse genetics reveals Barttin as a genetic modifier of Joubert syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 1113-1118.	7.1	22
43	Novel nephronophthisis-associated variants reveal functional importance of MAPKBP1 dimerization for centriolar recruitment. Kidney International, 2020, 98, 958-969.	5.2	6
44	PRDM15 loss of function links NOTCH and WNT/PCP signaling to patterning defects in holoprosencephaly. Science Advances, 2020, 6, eaax9852.	10.3	13
45	PLCE1 regulates the migration, proliferation, and differentiation of podocytes. Experimental and Molecular Medicine, 2020, 52, 594-603.	7.7	24
46	Responsiveness of sphingosine phosphate lyase insufficiency syndrome to vitamin <scp>B6</scp> cofactor supplementation. Journal of Inherited Metabolic Disease, 2020, 43, 1131-1142.	3.6	21
47	Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. Nephrology Dialysis Transplantation, 2019, 34, 474-485.	0.7	13
48	Roscovitine blocks collecting duct cyst growth in Cep164-deficient kidneys. Kidney International, 2019, 96, 320-326.	5.2	12
49	Mutations in KIRREL1, a slit diaphragm component, cause steroid-resistant nephrotic syndrome. Kidney International, 2019, 96, 883-889.	5.2	23
50	Whole exome sequencing in childhood-onset lupus frequently detects single gene etiologies. Pediatric Rheumatology, 2019, 17, 52.	2.1	34
51	FO027DIAGNOSTIC UTILITY OF NEXT GENERATION SEQUENCING TECHNIQUES IN PATIENTS WITH FAMILIAL KIDNEY DISEASE WHO HAVE UNDERGONE PERCUTANEOUS NATIVE KIDNEY BIOPSY. Nephrology Dialysis Transplantation, 2019, 34, .	0.7	0
52	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	6.2	56
53	Novel homozygous ENPP1 mutation causes generalized arterial calcifications of infancy, thrombocytopenia, and cardiovascular and central nervous system syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2112-2118.	1.2	16
54	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. Nature Communications, 2019, 10, 3967.	12.8	66

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55	COL4A1 mutations as a potential novel cause of autosomal dominant CAKUT in humans. Human Genetics, 2019, 138, 1105-1115.	3.8	13
56	Dominant PAX2 mutations may cause steroid-resistant nephrotic syndrome and FSGS in children. Pediatric Nephrology, 2019, 34, 1607-1613.	1.7	31
57	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. American Journal of Human Genetics, 2019, 104, 994-1006.	6.2	47
58	Progressive Pseudorheumatoid Dysplasia resolved by whole exome sequencing: a novel mutation in WISP3 and review of the literature. BMC Medical Genetics, 2019, 20, 53.	2.1	11
59	HSPA6: A new autosomal recessive candidate gene for the VATER/VACTERL malformation spectrum. Birth Defects Research, 2019, 111, 591-597.	1.5	15
60	Corticosteroid treatment exacerbates nephrotic syndrome in a zebrafish model of magi2a knockout. Kidney International, 2019, 95, 1079-1090.	5.2	9
61	Homozygous frameshift mutations in FAT1 cause a syndrome characterized by colobomatous-microphthalmia, ptosis, nephropathy and syndactyly. Nature Communications, 2019, 10, 1180.	12.8	27
62	Disruption of MAGI2-RapGEF2-Rap1 signaling contributes to podocyte dysfunction in congenital nephrotic syndrome caused by mutations in MAGI2. Kidney International, 2019, 96, 642-655.	5.2	13
63	Treatment with 2,4-Dihydroxybenzoic Acid Prevents FSGS Progression and Renal Fibrosis in Podocyte-Specific Coq6 Knockout Mice. Journal of the American Society of Nephrology: JASN, 2019, 30, 393-405.	6.1	36
64	Secreted metalloproteases ADAMTS9 and ADAMTS20 have a non-canonical role in ciliary vesicle growth during ciliogenesis. Nature Communications, 2019, 10, 953.	12.8	51
65	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. Human Genetics, 2019, 138, 211-219.	3.8	26
66	Monogenic causes of chronic kidney disease in adults. Kidney International, 2019, 95, 914-928.	5.2	174
67	TBC1D8B Mutations Implicate RAB11-Dependent Vesicular Trafficking in the Pathogenesis of Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2019, 30, 2338-2353.	6.1	25
68	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. American Journal of Human Genetics, 2019, 105, 1286-1293.	6.2	18
69	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. American Journal of Human Genetics, 2019, 104, 45-54.	6.2	29
70	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127.	21.4	144
71	Whole-Exome Sequencing Enables a Precision Medicine Approach for Kidney Transplant Recipients. Journal of the American Society of Nephrology: JASN, 2019, 30, 201-215.	6.1	110
72	Genetic variants in the LAMA5 gene in pediatric nephrotic syndrome. Nephrology Dialysis Transplantation, 2019, 34, 485-493.	0.7	22

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73	Whole Exome Sequencing Reveals a Monogenic Cause of Disease in â‰ˆ43% of 35 Families With Midaortic Syndrome. Hypertension, 2018, 71, 691-699.	2.7	22
74	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213.	5.2	133
75	Novel Insights into the Pathogenesis of Monogenic Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 36-50.	6.1	108
76	Analysis of 24 genes reveals a monogenic cause in 11.1% of cases with steroid-resistant nephrotic syndrome at a single center. Pediatric Nephrology, 2018, 33, 305-314.	1.7	30
77	Mutations in COQ8B (ADCK4) found in patients with steroid-resistant nephrotic syndrome alter COQ8B function. Human Mutation, 2018, 39, 406-414.	2.5	43
78	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2018, 13, 53-62.	4.5	170
79	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	8.2	89
80	Galloway-Mowat syndrome in Taiwan: OSGEP mutation and unique clinical phenotype. Orphanet Journal of Rare Diseases, 2018, 13, 226.	2.7	16
81	A Multi-layered Quantitative InÂVivo Expression Atlas of the Podocyte Unravels Kidney Disease Candidate Genes. Cell Reports, 2018, 23, 2495-2508.	6.4	81
82	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. Nature Communications, 2018, 9, 1960.	12.8	90
83	Clinical, biochemical, and pathophysiological analysis of SLC34A1 mutations. Physiological Reports, 2018, 6, e13715.	1.7	32
84	GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2123-2138.	6.1	42
85	Mutations in WDR4 as a new cause of Gallowayâ€“Mowat syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2460-2465.	1.2	56
86	Human urine-derived renal epithelial cells provide insights into kidney-specific alternate splicing variants. European Journal of Human Genetics, 2018, 26, 1791-1796.	2.8	22
87	The nucleoside-diphosphate kinase NME3 associates with nephronophthisis proteins and is required for ciliary function during renal development. Journal of Biological Chemistry, 2018, 293, 15243-15255.	3.4	13
88	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 2348-2361.	6.1	147
89	Effects of Diet and Social Housing on Reproductive Success in Adult Zebrafish, Danio rerio. Zebrafish, 2018, 15, 445-453.	1.1	16
90	Acute multi-sgRNA knockdown of KEOPS complex genes reproduces the microcephaly phenotype of the stable knockout zebrafish model. PLoS ONE, 2018, 13, e0191503.	2.5	18

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91	ZMYND10 stabilizes intermediate chain proteins in the cytoplasmic pre-assembly of dynein arms. PLoS Genetics, 2018, 14, e1007316.	3.5	37
92	A homozygous missense variant in VWA2, encoding an interactor of the Fraser-complex, in a patient with vesicoureteral reflux. PLoS ONE, 2018, 13, e0191224.	2.5	5
93	Exome Sequencing Discerns Syndromes in Patients from Consanguineous Families with Congenital Anomalies of the Kidneys and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2017, 28, 69-75.	6.1	79
94	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. American Journal of Human Genetics, 2017, 100, 323-333.	6.2	29
95	Spectrum of mutations in Chinese children with steroid-resistant nephrotic syndrome. Pediatric Nephrology, 2017, 32, 1181-1192.	1.7	81
96	Mutations in DZIP1L, which encodes a ciliary-transition-zone protein, cause autosomal recessive polycystic kidney disease. Nature Genetics, 2017, 49, 1025-1034.	21.4	148
97	Modeling Monogenic Human Nephrotic Syndrome in the Drosophila Garland Cell Nephrocyte. Journal of the American Society of Nephrology: JASN, 2017, 28, 1521-1533.	6.1	70
98	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	6.2	63
99	Osteoclast stimulation factor 1 (Ostf1) KNOCKOUT increases trabecular bone mass in mice. Mammalian Genome, 2017, 28, 498-514.	2.2	19
100	Whole-Exome Sequencing Reveals FAT4 Mutations in a Clinically Unrecognizable Patient with Syndromic CAKUT: A Case Report. Molecular Syndromology, 2017, 8, 272-277.	0.8	7
101	Cystic kidneys in fetal Walkerâ€“Warburg syndrome with POMT2 mutation: Intrafamilial phenotypic variability in four siblings and review of literature. American Journal of Medical Genetics, Part A, 2017, 173, 2697-2702.	1.2	11
102	Exome sequencing in Jewish and Arab patients with rhabdomyolysis reveals single-gene etiology in 43% of cases. Pediatric Nephrology, 2017, 32, 2273-2282.	1.7	40
103	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164
104	Ciliopathies. Cold Spring Harbor Perspectives in Biology, 2017, 9, a028191.	5.5	325
105	A small molecule screening to detect potential therapeutic targets in human podocytes. American Journal of Physiology - Renal Physiology, 2017, 312, F157-F171.	2.7	10
106	Advillin acts upstream of phospholipase C Ïµ1 in steroid-resistant nephrotic syndrome. Journal of Clinical Investigation, 2017, 127, 4257-4269.	8.2	39
107	Targeted sequencing of 96 renal developmental microRNAs in 1213 individuals from 980 families with congenital anomalies of the kidney and urinary tract. Nephrology Dialysis Transplantation, 2016, 31, 1280-1283.	0.7	15
108	A FANCD2/FANCI-Associated Nuclease 1-Knockout Model Develops Karyomegalic Interstitial Nephritis. Journal of the American Society of Nephrology: JASN, 2016, 27, 3552-3559.	6.1	24

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109	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656.	21.4	119
110	FAT1 mutations cause a glomerulotubular nephropathy. Nature Communications, 2016, 7, 10822.	12.8	99
111	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 664-672.	4.5	105
112	Exploring the genetic basis of early-onset chronic kidney disease. Nature Reviews Nephrology, 2016, 12, 133-146.	9.6	276
113	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. Nature Genetics, 2016, 48, 457-465.	21.4	149
114	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214.	3.2	39
115	Genetic testing in steroid-resistant nephrotic syndrome: when and how?. Nephrology Dialysis Transplantation, 2016, 31, 1802-1813.	0.7	159
116	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. Journal of Medical Genetics, 2016, 53, 62-72.	3.2	48
117	Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. Kidney International, 2016, 89, 468-475.	5.2	74
118	Mutation of Growth Arrest Specific 8 Reveals a Role in Motile Cilia Function and Human Disease. PLoS Genetics, 2016, 12, e1006220.	3.5	33
119	SDCCAG8 Interacts with RAB Effector Proteins RABEP2 and ERC1 and Is Required for Hedgehog Signaling. PLoS ONE, 2016, 11, e0156081.	2.5	19
120	Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies ImplicateZIC3andFOXF1in Human VATER/VACTERL Association. Human Mutation, 2015, 36, 1150-1154.	2.5	46
121	WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028.	2.5	42
122	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	6.0	64
123	A Dynamic Protein Interaction Landscape of the Human Centrosome-Cilium Interface. Cell, 2015, 163, 1484-1499.	28.9	446
124	Defects of CRB2 Cause Steroid-Resistant Nephrotic Syndrome. American Journal of Human Genetics, 2015, 96, 153-161.	6.2	88
125	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. American Journal of Human Genetics, 2015, 96, 81-92.	6.2	98
126	Clinical Features and Histology of Apolipoprotein L1-Associated Nephropathy in the FSGS Clinical Trial. Journal of the American Society of Nephrology: JASN, 2015, 26, 1443-1448.	6.1	104

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127	Mutations in TBX18 Cause Dominant Urinary Tract Malformations via Transcriptional Dysregulation of Ureter Development. American Journal of Human Genetics, 2015, 97, 291-301.	6.2	72
128	Recessive nephrocerebellar syndrome on the Galloway-Mowat syndrome spectrum is caused by homozygous protein-truncating mutations ofWDR73. Brain, 2015, 138, 2173-2190.	7.6	60
129	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	3.2	32
130	Genetic dissection of kidney disorders. Nature Reviews Nephrology, 2015, 11, 635-636.	9.6	12
131	A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2015, 26, 1279-1289.	6.1	499
132	Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. Journal of the American Society of Nephrology: JASN, 2015, 26, 543-551.	6.1	163
133	Rapid Detection of Monogenic Causes of Childhood-Onset Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 1109-1116.	4.5	74
134	Nephronophthisis-Associated CEP164 Regulates Cell Cycle Progression, Apoptosis and Epithelial-to-Mesenchymal Transition. PLoS Genetics, 2014, 10, e1004594.	3.5	73
135	Mild Recessive Mutations in Six Fraser Syndromeâ€“Related Genes Cause Isolated Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2014, 25, 1917-1922.	6.1	97
136	Mutations in 12 known dominant disease-causing genes clarify many congenital anomalies of the kidney and urinary tract. Kidney International, 2014, 85, 1429-1433.	5.2	203
137	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
138	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	6.2	75
139	Mutations in EMP2 Cause Childhood-Onset Nephrotic Syndrome. American Journal of Human Genetics, 2014, 94, 884-890.	6.2	101
140	Single-gene causes of congenital anomalies of the kidney and urinary tract (CAKUT) in humans. Pediatric Nephrology, 2014, 29, 695-704.	1.7	178
141	Renal-Retinal Ciliopathy Gene Sdccag8 Regulates DNA Damage Response Signaling. Journal of the American Society of Nephrology: JASN, 2014, 25, 2573-2583.	6.1	63
142	Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association. Kidney International, 2014, 85, 1310-1317.	5.2	106
143	SDCCAG8 Regulates Pericentriolar Material Recruitment and Neuronal Migration in the Developing Cortex. Neuron, 2014, 83, 805-822.	8.1	52
144	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914.	6.2	90

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145	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. Human Genetics, 2013, 132, 865-884.	3.8	199
146	Karyomegalic interstitial nephritis. Lancet, The, 2013, 382, 2093.	13.7	2
147	Genotypeâ€“phenotype correlation in 440 patients with NPHP-related ciliopathies. Kidney International, 2011, 80, 1239-1245.	5.2	99
148	Ciliopathies. New England Journal of Medicine, 2011, 364, 1533-1543.	27.0	1,227
149	Exome Sequencing Reveals Cubilin Mutation as a Single-Gene Cause of Proteinuria. Journal of the American Society of Nephrology: JASN, 2011, 22, 1815-1820.	6.1	90
150	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024.	8.2	343
151	Genotype/Phenotype Correlation in Nephrotic Syndrome Caused by WT1 Mutations. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 1655-1662.	4.5	87
152	Immunosuppression and Renal Outcome in Congenital and Pediatric Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 2075-2084.	4.5	153
153	Nineteen novel NPHS1 mutations in a worldwide cohort of patients with congenital nephrotic syndrome (CNS). Nephrology Dialysis Transplantation, 2010, 25, 2970-2976.	0.7	69
154	A Systematic Approach to Mapping Recessive Disease Genes in Individuals from Outbred Populations. PLoS Genetics, 2009, 5, e1000353.	3.5	144
155	HomozygosityMapper--an interactive approach to homozygosity mapping. Nucleic Acids Research, 2009, 37, W593-W599.	14.5	331
156	Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2009, 20, 23-35.	6.1	332
157	Mutation analysis in nephronophthisis using a combined approach of homozygosity mapping, CEL I endonuclease cleavage, and direct sequencing. Human Mutation, 2008, 29, 418-426.	2.5	76
158	Mutations in FN1 cause glomerulopathy with fibronectin deposits. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 2538-2543.	7.1	125
159	Thirteen novel NPHS1 mutations in a large cohort of children with congenital nephrotic syndrome. Nephrology Dialysis Transplantation, 2008, 23, 3527-3533.	0.7	74
160	Nephronophthisis-Associated Ciliopathies. Journal of the American Society of Nephrology: JASN, 2007, 18, 1855-1871.	6.1	354
161	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature Genetics, 2006, 38, 674-681.	21.4	535
162	Patients with Mutations in NPHS2 (Podocin) Do Not Respond to Standard Steroid Treatment of Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2004, 15, 722-732.	6.1	370