

# Amelie van der Ven

## List of Publications by Year in descending order

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162  
papers

12,976  
citations

22099

59  
h-index

26548

107  
g-index

166  
all docs

166  
docs citations

166  
times ranked

13429  
citing authors

#	ARTICLE	IF	CITATIONS
1	Ciliopathies. <i>New England Journal of Medicine</i> , 2011, 364, 1533-1543.	13.9	1,227
2	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. <i>Nature Genetics</i> , 2006, 38, 674-681.	9.4	535
3	A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 1279-1289.	3.0	499
4	A Dynamic Protein Interaction Landscape of the Human Centrosome-Cilium Interface. <i>Cell</i> , 2015, 163, 1484-1499.	13.5	446
5	Patients with Mutations in NPHS2 (Podocin) Do Not Respond to Standard Steroid Treatment of Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2004, 15, 722-732.	3.0	370
6	Nephronophthisis-Associated Ciliopathies. <i>Journal of the American Society of Nephrology: JASN</i> , 2007, 18, 1855-1871.	3.0	354
7	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. <i>Journal of Clinical Investigation</i> , 2011, 121, 2013-2024.	3.9	343
8	Nephronophthisis. <i>Journal of the American Society of Nephrology: JASN</i> , 2009, 20, 23-35.	3.0	332
9	HomozygosityMapper—an interactive approach to homozygosity mapping. <i>Nucleic Acids Research</i> , 2009, 37, W593-W599.	6.5	331
10	Ciliopathies. <i>Cold Spring Harbor Perspectives in Biology</i> , 2017, 9, a028191.	2.3	325
11	Exploring the genetic basis of early-onset chronic kidney disease. <i>Nature Reviews Nephrology</i> , 2016, 12, 133-146.	4.1	276
12	Podocytopathies. <i>Nature Reviews Disease Primers</i> , 2020, 6, 68.	18.1	237
13	Mutations in 12 known dominant disease-causing genes clarify many congenital anomalies of the kidney and urinary tract. <i>Kidney International</i> , 2014, 85, 1429-1433.	2.6	203
14	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. <i>Human Genetics</i> , 2013, 132, 865-884.	1.8	199
15	Single-gene causes of congenital anomalies of the kidney and urinary tract (CAKUT) in humans. <i>Pediatric Nephrology</i> , 2014, 29, 695-704.	0.9	178
16	Monogenic causes of chronic kidney disease in adults. <i>Kidney International</i> , 2019, 95, 914-928.	2.6	174
17	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2018, 13, 53-62.	2.2	170
18	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. <i>Nature Genetics</i> , 2017, 49, 1529-1538.	9.4	164

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19	Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 543-551.	3.0	163
20	Genetic testing in steroid-resistant nephrotic syndrome: when and how?. <i>Nephrology Dialysis Transplantation</i> , 2016, 31, 1802-1813.	0.4	159
21	Immunosuppression and Renal Outcome in Congenital and Pediatric Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2010, 5, 2075-2084.	2.2	153
22	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. <i>Nature Genetics</i> , 2016, 48, 457-465.	9.4	149
23	Mutations in DZIP1L, which encodes a ciliary-transition-zone protein, cause autosomal recessive polycystic kidney disease. <i>Nature Genetics</i> , 2017, 49, 1025-1034.	9.4	148
24	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2348-2361.	3.0	147
25	A Systematic Approach to Mapping Recessive Disease Genes in Individuals from Outbred Populations. <i>PLoS Genetics</i> , 2009, 5, e1000353.	1.5	144
26	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. <i>Nature Genetics</i> , 2019, 51, 117-127.	9.4	144
27	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and nephrocalcinosis. <i>Kidney International</i> , 2018, 93, 204-213.	2.6	133
28	Mutations in <i>FN1</i> cause glomerulopathy with fibronectin deposits. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 2538-2543.	3.3	125
29	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. <i>Nature Genetics</i> , 2016, 48, 648-656.	9.4	119
30	Whole-Exome Sequencing Enables a Precision Medicine Approach for Kidney Transplant Recipients. <i>Journal of the American Society of Nephrology: JASN</i> , 2019, 30, 201-215.	3.0	110
31	Novel Insights into the Pathogenesis of Monogenic Congenital Anomalies of the Kidney and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 36-50.	3.0	108
32	Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association. <i>Kidney International</i> , 2014, 85, 1310-1317.	2.6	106
33	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2016, 11, 664-672.	2.2	105
34	Clinical Features and Histology of Apolipoprotein L1-Associated Nephropathy in the FSGS Clinical Trial. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 1443-1448.	3.0	104
35	Mutations in EMP2 Cause Childhood-Onset Nephrotic Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 884-890.	2.6	101
36	Genotype-phenotype correlation in 440 patients with NPHP-related ciliopathies. <i>Kidney International</i> , 2011, 80, 1239-1245.	2.6	99

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37	FAT1 mutations cause a glomerulotubular nephropathy. <i>Nature Communications</i> , 2016, 7, 10822.	5.8	99
38	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 96, 81-92.	2.6	98
39	Mild Recessive Mutations in Six Fraser Syndrome-Related Genes Cause Isolated Congenital Anomalies of the Kidney and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , 2014, 25, 1917-1922.	3.0	97
40	Exome Sequencing Reveals Cubilin Mutation as a Single-Gene Cause of Proteinuria. <i>Journal of the American Society of Nephrology: JASN</i> , 2011, 22, 1815-1820.	3.0	90
41	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 905-914.	2.6	90
42	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. <i>Nature Communications</i> , 2018, 9, 1960.	5.8	90
43	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2018, 128, 4313-4328.	3.9	89
44	Impaired complex I repair causes recessive Leber's hereditary optic neuropathy. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	89
45	Defects of CRB2 Cause Steroid-Resistant Nephrotic Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 153-161.	2.6	88
46	Genotype/Phenotype Correlation in Nephrotic Syndrome Caused by WT1 Mutations. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2010, 5, 1655-1662.	2.2	87
47	Spectrum of mutations in Chinese children with steroid-resistant nephrotic syndrome. <i>Pediatric Nephrology</i> , 2017, 32, 1181-1192.	0.9	81
48	A Multi-layered Quantitative In Vivo Expression Atlas of the Podocyte Unravels Kidney Disease Candidate Genes. <i>Cell Reports</i> , 2018, 23, 2495-2508.	2.9	81
49	Exome Sequencing Discerns Syndromes in Patients from Consanguineous Families with Congenital Anomalies of the Kidneys and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 69-75.	3.0	79
50	Mutation analysis in nephronophthisis using a combined approach of homozygosity mapping, CEL I endonuclease cleavage, and direct sequencing. <i>Human Mutation</i> , 2008, 29, 418-426.	1.1	76
51	Mutations in CSPP1 Lead to Classical Joubert Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 80-86.	2.6	75
52	Thirteen novel NPHS1 mutations in a large cohort of children with congenital nephrotic syndrome. <i>Nephrology Dialysis Transplantation</i> , 2008, 23, 3527-3533.	0.4	74
53	Rapid Detection of Monogenic Causes of Childhood-Onset Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2014, 9, 1109-1116.	2.2	74
54	Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. <i>Kidney International</i> , 2016, 89, 468-475.	2.6	74

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55	Nephronophthisis-Associated CEP164 Regulates Cell Cycle Progression, Apoptosis and Epithelial-to-Mesenchymal Transition. <i>PLoS Genetics</i> , 2014, 10, e1004594.	1.5	73
56	Mutations in TBX18 Cause Dominant Urinary Tract Malformations via Transcriptional Dysregulation of Ureter Development. <i>American Journal of Human Genetics</i> , 2015, 97, 291-301.	2.6	72
57	Modeling Monogenic Human Nephrotic Syndrome in the <i>Drosophila</i> Garland Cell Nephrocyte. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 1521-1533.	3.0	70
58	Nineteen novel NPHS1 mutations in a worldwide cohort of patients with congenital nephrotic syndrome (CNS). <i>Nephrology Dialysis Transplantation</i> , 2010, 25, 2970-2976.	0.4	69
59	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. <i>Kidney International</i> , 2014, 85, 880-887.	2.6	67
60	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. <i>Nature Communications</i> , 2019, 10, 3967.	5.8	66
61	Healthcare recommendations for Joubert syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 229-249.	0.7	66
62	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. <i>ELife</i> , 2015, 4, e06602.	2.8	64
63	Renal-Retinal Ciliopathy Gene <i>Sdccag8</i> Regulates DNA Damage Response Signaling. <i>Journal of the American Society of Nephrology: JASN</i> , 2014, 25, 2573-2583.	3.0	63
64	Exome-wide Association Study Identifies <i>GREB1L</i> Mutations in Congenital Kidney Malformations. <i>American Journal of Human Genetics</i> , 2017, 101, 789-802.	2.6	63
65	Recessive nephrocerebellar syndrome on the Galloway-Mowat syndrome spectrum is caused by homozygous protein-truncating mutations of <i>WDR73</i> . <i>Brain</i> , 2015, 138, 2173-2190.	3.7	60
66	Personalized medicine in chronic kidney disease by detection of monogenic mutations. <i>Nephrology Dialysis Transplantation</i> , 2020, 35, 390-397.	0.4	58
67	Mutations in <i>WDR4</i> as a new cause of Galloway-Mowat syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2460-2465.	0.7	56
68	Paralog Studies Augment Gene Discovery: <i>DDX</i> and <i>DHX</i> Genes. <i>American Journal of Human Genetics</i> , 2019, 105, 302-316.	2.6	56
69	<i>SDCCAG8</i> Regulates Pericentriolar Material Recruitment and Neuronal Migration in the Developing Cortex. <i>Neuron</i> , 2014, 83, 805-822.	3.8	52
70	Secreted metalloproteases <i>ADAMTS9</i> and <i>ADAMTS20</i> have a non-canonical role in ciliary vesicle growth during ciliogenesis. <i>Nature Communications</i> , 2019, 10, 953.	5.8	51
71	<i>MKS1</i> regulates ciliary <i>INPP5E</i> levels in Joubert syndrome. <i>Journal of Medical Genetics</i> , 2016, 53, 62-72.	1.5	48
72	Rare Variants in <i>BNC2</i> Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. <i>American Journal of Human Genetics</i> , 2019, 104, 994-1006.	2.6	47

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73	Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies Implicate <i>ZIC3</i> and <i>FOXF1</i> in Human VATER/VACTERL Association. <i>Human Mutation</i> , 2015, 36, 1150-1154.	1.1	46
74	Mutations in <i>COQ8B</i> ( <i>ADCK4</i> ) found in patients with steroid-resistant nephrotic syndrome alter <i>COQ8B</i> function. <i>Human Mutation</i> , 2018, 39, 406-414.	1.1	43
75	<i>WDR73</i> Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. <i>Human Mutation</i> , 2015, 36, 1021-1028.	1.1	42
76	<i>GAPVD1</i> and <i>ANKFY1</i> Mutations Implicate <i>RAB5</i> Regulation in Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2123-2138.	3.0	42
77	Whole exome sequencing identified <i>ATP6V1C2</i> as a novel candidate gene for recessive distal renal tubular acidosis. <i>Kidney International</i> , 2020, 97, 567-579.	2.6	42
78	Exome sequencing in Jewish and Arab patients with rhabdomyolysis reveals single-gene etiology in 43% of cases. <i>Pediatric Nephrology</i> , 2017, 32, 2273-2282.	0.9	40
79	Multisystem inflammation and susceptibility to viral infections in human <i>ZNFX1</i> deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 381-393.	1.5	40
80	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. <i>Journal of Medical Genetics</i> , 2016, 53, 208-214.	1.5	39
81	Advillin acts upstream of phospholipase C $\mu 1$ in steroid-resistant nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2017, 127, 4257-4269.	3.9	39
82	<i>ADCK4</i> Deficiency Destabilizes the Coenzyme Q Complex, Which Is Rescued by 2,4-Dihydroxybenzoic Acid Treatment. <i>Journal of the American Society of Nephrology: JASN</i> , 2020, 31, 1191-1211.	3.0	38
83	<i>ZMYND10</i> stabilizes intermediate chain proteins in the cytoplasmic pre-assembly of dynein arms. <i>PLoS Genetics</i> , 2018, 14, e1007316.	1.5	37
84	Treatment with 2,4-Dihydroxybenzoic Acid Prevents FSGS Progression and Renal Fibrosis in Podocyte-Specific <i>Coq6</i> Knockout Mice. <i>Journal of the American Society of Nephrology: JASN</i> , 2019, 30, 393-405.	3.0	36
85	Whole exome sequencing in childhood-onset lupus frequently detects single gene etiologies. <i>Pediatric Rheumatology</i> , 2019, 17, 52.	0.9	34
86	Mutation of Growth Arrest Specific 8 Reveals a Role in Motile Cilia Function and Human Disease. <i>PLoS Genetics</i> , 2016, 12, e1006220.	1.5	33
87	<i>IFT81</i> , encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. <i>Journal of Medical Genetics</i> , 2015, 52, 657-665.	1.5	32
88	Clinical, biochemical, and pathophysiological analysis of <i>SLC34A1</i> mutations. <i>Physiological Reports</i> , 2018, 6, e13715.	0.7	32
89	Dominant <i>PAX2</i> mutations may cause steroid-resistant nephrotic syndrome and FSGS in children. <i>Pediatric Nephrology</i> , 2019, 34, 1607-1613.	0.9	31
90	Analysis of 24 genes reveals a monogenic cause in 11.1% of cases with steroid-resistant nephrotic syndrome at a single center. <i>Pediatric Nephrology</i> , 2018, 33, 305-314.	0.9	30

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91	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. <i>American Journal of Human Genetics</i> , 2017, 100, 323-333.	2.6	29
92	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. <i>American Journal of Human Genetics</i> , 2019, 104, 45-54.	2.6	29
93	Homozygous frameshift mutations in FAT1 cause a syndrome characterized by colobomatous-microphthalmia, ptosis, nephropathy and syndactyly. <i>Nature Communications</i> , 2019, 10, 1180.	5.8	27
94	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. <i>Human Genetics</i> , 2019, 138, 211-219.	1.8	26
95	TBC1D8B Mutations Implicate RAB11-Dependent Vesicular Trafficking in the Pathogenesis of Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2019, 30, 2338-2353.	3.0	25
96	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. <i>American Journal of Human Genetics</i> , 2020, 107, 727-742.	2.6	25
97	A FANCD2/FANCI-Associated Nuclease 1-Knockout Model Develops Karyomegalic Interstitial Nephritis. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 3552-3559.	3.0	24
98	PLCE1 regulates the migration, proliferation, and differentiation of podocytes. <i>Experimental and Molecular Medicine</i> , 2020, 52, 594-603.	3.2	24
99	Mutations in KIRREL1, a slit diaphragm component, cause steroid-resistant nephrotic syndrome. <i>Kidney International</i> , 2019, 96, 883-889.	2.6	23
100	Whole Exome Sequencing Reveals a Monogenic Cause of Disease in ~43% of 35 Families With Midaortic Syndrome. <i>Hypertension</i> , 2018, 71, 691-699.	1.3	22
101	Human urine-derived renal epithelial cells provide insights into kidney-specific alternate splicing variants. <i>European Journal of Human Genetics</i> , 2018, 26, 1791-1796.	1.4	22
102	Genetic variants in the LAMA5 gene in pediatric nephrotic syndrome. <i>Nephrology Dialysis Transplantation</i> , 2019, 34, 485-493.	0.4	22
103	Mouse genetics reveals Barttin as a genetic modifier of Joubert syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 1113-1118.	3.3	22
104	Responsiveness of sphingosine phosphate lyase insufficiency syndrome to vitamin B6 cofactor supplementation. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1131-1142.	1.7	21
105	Recessive <i>NOS1AP</i> variants impair actin remodeling and cause glomerulopathy in humans and mice. <i>Science Advances</i> , 2021, 7, .	4.7	21
106	Osteoclast stimulation factor 1 ( <i>Ostf1</i> ) KNOCKOUT increases trabecular bone mass in mice. <i>Mammalian Genome</i> , 2017, 28, 498-514.	1.0	19
107	SDCCAG8 Interacts with RAB Effector Proteins RABEP2 and ERC1 and Is Required for Hedgehog Signaling. <i>PLoS ONE</i> , 2016, 11, e0156081.	1.1	19
108	Acute multi-sgRNA knockdown of KEOPS complex genes reproduces the microcephaly phenotype of the stable knockout zebrafish model. <i>PLoS ONE</i> , 2018, 13, e0191503.	1.1	18



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109	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. <i>American Journal of Human Genetics</i> , 2019, 105, 1286-1293.	2.6	18
110	Copy Number Variant Analysis and Genome-wide Association Study Identify Loci with Large Effect for Vesicoureteral Reflux. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 805-820.	3.0	17
111	Galloway-Mowat syndrome in Taiwan: OSGEP mutation and unique clinical phenotype. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 226.	1.2	16
112	Effects of Diet and Social Housing on Reproductive Success in Adult Zebrafish, <i>&lt;i&gt;Danio rerio&lt;/i&gt;</i> . <i>Zebrafish</i> , 2018, 15, 445-453.	0.5	16
113	Novel homozygous <i>&lt;i&gt;ENPP1&lt;/i&gt;</i> mutation causes generalized arterial calcifications of infancy, thrombocytopenia, and cardiovascular and central nervous system syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2112-2118.	0.7	16
114	Whole-Exome Sequencing in Critically Ill Neonates and Infants: Diagnostic Yield and Predictability of Monogenic Diagnosis. <i>Neonatology</i> , 2021, 118, 454-461.	0.9	16
115	Deep learning is widely applicable to phenotyping embryonic development and disease. <i>Development (Cambridge)</i> , 2021, 148, .	1.2	16
116	Targeted sequencing of 96 renal developmental microRNAs in 1213 individuals from 980 families with congenital anomalies of the kidney and urinary tract. <i>Nephrology Dialysis Transplantation</i> , 2016, 31, 1280-1283.	0.4	15
117	<i>&lt;i&gt;HSPA6&lt;/i&gt;</i> : A new autosomal recessive candidate gene for the VATER/VACTERL malformation spectrum. <i>Birth Defects Research</i> , 2019, 111, 591-597.	0.8	15
118	Utility of Genomic Testing after Renal Biopsy. <i>American Journal of Nephrology</i> , 2020, 51, 43-53.	1.4	15
119	Mutations in PRDM15 Are a Novel Cause of Galloway-Mowat Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 580-596.	3.0	15
120	Inhibition of endoplasmic reticulum stress signaling rescues cytotoxicity of human apolipoprotein-L1 risk variants in <i>Drosophila</i> . <i>Kidney International</i> , 2022, 101, 1216-1231.	2.6	15
121	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. <i>American Journal of Human Genetics</i> , 2021, 108, 357-367.	2.6	14
122	The utility of a genetic kidney disease clinic employing a broad range of genomic testing platforms: experience of the Irish Kidney Gene Project. <i>Journal of Nephrology</i> , 2022, 35, 1655-1665.	0.9	14
123	The nucleoside-diphosphate kinase NME3 associates with nephronophthisis proteins and is required for ciliary function during renal development. <i>Journal of Biological Chemistry</i> , 2018, 293, 15243-15255.	1.6	13
124	Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. <i>Nephrology Dialysis Transplantation</i> , 2019, 34, 474-485.	0.4	13
125	COL4A1 mutations as a potential novel cause of autosomal dominant CAKUT in humans. <i>Human Genetics</i> , 2019, 138, 1105-1115.	1.8	13
126	Disruption of MAGI2-RapGEF2-Rap1 signaling contributes to podocyte dysfunction in congenital nephrotic syndrome caused by mutations in MAGI2. <i>Kidney International</i> , 2019, 96, 642-655.	2.6	13



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127	Beyond the tubule: pathological variants of <i>LRP2</i> , encoding the megalin receptor, result in glomerular loss and early progressive chronic kidney disease. <i>American Journal of Physiology - Renal Physiology</i> , 2020, 319, F988-F999.	1.3	13
128	PRDM15 loss of function links NOTCH and WNT/PCP signaling to patterning defects in holoprosencephaly. <i>Science Advances</i> , 2020, 6, eaax9852.	4.7	13
129	A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. <i>Genetics in Medicine</i> , 2021, 23, 1158-1162.	1.1	13
130	Cystin genetic variants cause autosomal recessive polycystic kidney disease associated with altered Myc expression. <i>Scientific Reports</i> , 2021, 11, 18274.	1.6	13
131	Reverse phenotyping facilitates disease allele calling in exome sequencing of patients with CAKUT. <i>Genetics in Medicine</i> , 2022, 24, 307-318.	1.1	13
132	Genetic dissection of kidney disorders. <i>Nature Reviews Nephrology</i> , 2015, 11, 635-636.	4.1	12
133	Roscovitine blocks collecting duct cyst growth in Cep164-deficient kidneys. <i>Kidney International</i> , 2019, 96, 320-326.	2.6	12
134	DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. <i>American Journal of Human Genetics</i> , 2020, 107, 1113-1128.	2.6	12
135	A discarded synonymous variant in <i>NPHP3</i> explains nephronophthisis and congenital hepatic fibrosis in several families. <i>Human Mutation</i> , 2021, 42, 1221-1228.	1.1	12
136	Cystic kidneys in fetal Walker-Warburg syndrome with <i>POMT2</i> mutation: Intrafamilial phenotypic variability in four siblings and review of literature. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2697-2702.	0.7	11
137	Progressive Pseudorheumatoid Dysplasia resolved by whole exome sequencing: a novel mutation in WISP3 and review of the literature. <i>BMC Medical Genetics</i> , 2019, 20, 53.	2.1	11
138	Loss of <i>Anks6</i> leads to YAP deficiency and liver abnormalities. <i>Human Molecular Genetics</i> , 2020, 29, 3064-3080.	1.4	11
139	Immunological Impact of a Gluten-Free Dairy-Free Diet in Children With Kidney Disease: A Feasibility Study. <i>Frontiers in Immunology</i> , 2021, 12, 624821.	2.2	11
140	A small molecule screening to detect potential therapeutic targets in human podocytes. <i>American Journal of Physiology - Renal Physiology</i> , 2017, 312, F157-F171.	1.3	10
141	Corticosteroid treatment exacerbates nephrotic syndrome in a zebrafish model of <i>magi2a</i> knockout. <i>Kidney International</i> , 2019, 95, 1079-1090.	2.6	9
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