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List of Publications by Year
in descending order

137 papers	17,224 citations	10956 71 h-index	14702 127 g-index
148 all docs	148 docs citations	148 times ranked	14608 citing authors

#	Article	IF	Citations
1	Mutations in INVS encoding inversin cause nephronophthisis type 2, linking renal cystic disease to the function of primary cilia and left-right axis determination. Nature Genetics, 2003, 34, 413-420.	9.4	582
2	A transition zone complex regulates mammalian ciliogenesis and ciliary membrane composition. Nature Genetics, 2011, 43, 776-784.	9.4	556
3	Barttin is a Cl- channel Î²-subunit crucial for renal Cl- reabsorption and inner ear K+ secretion. Nature, 2001, 414, 558-561.	13.7	538
4	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature Genetics, 2006, 38, 674-681.	9.4	535
5	Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. Cell, 2011, 145, 513-528.	13.5	531
6	Mutation of BSND causes Bartter syndrome with sensorineural deafness and kidney failure. Nature Genetics, 2001, 29, 310-314.	9.4	510
7	Positional cloning uncovers mutations in PLCE1 responsible for a nephrotic syndrome variant that may be reversible. Nature Genetics, 2006, 38, 1397-1405.	9.4	510
8	SIX1 mutations cause branchio-oto-renal syndrome by disruption of EYA1-SIX1-DNA complexes. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 8090-8095.	3.3	374
9	Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. Nature Genetics, 2005, 37, 282-288.	9.4	367
10	In-frame deletion in a novel centrosomal/ciliary protein CEP290/NPHP6 perturbs its interaction with RPGR and results in early-onset retinal degeneration in the rd16 mouse. Human Molecular Genetics, 2006, 15, 1847-1857.	1.4	353
11	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	2.6	352
12	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	13.5	347
13	Mutations in a novel gene, NPHP3, cause adolescent nephronophthisis, tapeto-retinal degeneration and hepatic fibrosis. Nature Genetics, 2003, 34, 455-459.	9.4	345
14	Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2009, 20, 23-35.	3.0	332
15	High-Throughput Screening Enhances Kidney Organoid Differentiation from Human Pluripotent Stem Cells and Enables Automated Multidimensional Phenotyping. Cell Stem Cell, 2018, 22, 929-940.e4.	5.2	328
16	A novel gene encoding an SH3 domain protein is mutated in nephronophthisis type 1. Nature Genetics, 1997, 17, 149-153.	9.4	327
17	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	9.4	326
18	Cilia and centrosomes: a unifying pathogenic concept for cystic kidney disease?. Nature Reviews Genetics, 2005, 6, 928-940.	7.7	296

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19	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. Nature Genetics, 2010, 42, 840-850.	9.4	295
20	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. Journal of Clinical Investigation, 2013, 123, 5179-5189.	3.9	275
21	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	9.4	261
22	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	9.4	255
23	Loss of GLIS2 causes nephronophthisis in humans and mice by increased apoptosis and fibrosis. Nature Genetics, 2007, 39, 1018-1024.	9.4	221
24	MYO1EMutations and Childhood Familial Focal Segmental Glomerulosclerosis. New England Journal of Medicine, 2011, 365, 295-306.	13.9	221
25	FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney failure to defective DNA damage repair. Nature Genetics, 2012, 44, 910-915.	9.4	205
26	CC2D2A Is Mutated in Joubert Syndrome and Interacts with the Ciliopathy-Associated Basal Body Protein CEP290. American Journal of Human Genetics, 2008, 83, 559-571.	2.6	202
27	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. Human Genetics, 2013, 132, 865-884.	1.8	199
28	NEK8 Mutations Affect Ciliary and Centrosomal Localization and May Cause Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2008, 19, 587-592.	3.0	196
29	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	2.6	196
30	ARHGDIA mutations cause nephrotic syndrome via defective RHO GTPase signaling. Journal of Clinical Investigation, 2013, 123, 3243-3253.	3.9	196
31	A Gene Mutated in Nephronophthisis and Retinitis Pigmentosa Encodes a Novel Protein, Nephroretinin, Conserved in Evolution. American Journal of Human Genetics, 2002, 71, 1161-1167.	2.6	193
32	Mutations in RSPH1 Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. American Journal of Respiratory and Critical Care Medicine, 2014, 189, 707-717.	2.5	191
33	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 672-686.	2.6	184
34	ANKS6 is a central component of a nephronophthisis module linking NEK8 to INVS and NPHP3. Nature Genetics, 2013, 45, 951-956.	9.4	183
35	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. American Journal of Human Genetics, 2013, 93, 336-345.	2.6	183
36	AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. Nature Genetics, 2010, 42, 175-180.	9.4	171

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37	Transcription Factor SIX5 Is Mutated in Patients with Branchio-Oto-Renal Syndrome. American Journal of Human Genetics, 2007, 80, 800-804.	2.6	164
38	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. Journal of Clinical Investigation, 2015, 125, 2375-2384.	3.9	159
39	Mutations in DZIP1L, which encodes a ciliary-transition-zone protein, cause autosomal recessive polycystic kidney disease. Nature Genetics, 2017, 49, 1025-1034.	9.4	148
40	An eQTL Landscape of Kidney Tissue in Human Nephrotic Syndrome. American Journal of Human Genetics, 2018, 103, 232-244.	2.6	147
41	A Systematic Approach to Mapping Recessive Disease Genes in Individuals from Outbred Populations. PLoS Genetics, 2009, 5, e1000353.	1.5	144
42	Evidence of Oligogenic Inheritance in Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2007, 18, 2789-2795.	3.0	141
43	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	2.6	135
44	Single-cell analysis of progenitor cell dynamics and lineage specification in the human fetal kidney. Development (Cambridge), 2018, 145, .	1.2	130
45	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.	3.3	125
46	Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. Journal of Medical Genetics, 2011, 48, 105-116.	1.5	123
47	Hypomorphic mutations in meckelin (MKS3/TMEM67) cause nephronophthisis with liver fibrosis (NPHP11). Journal of Medical Genetics, 2009, 46, 663-670.	1.5	121
48	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	13.9	120
49	Integrative Genomics Identifies Novel Associations with APOL1 Risk Genotypes in Black NEPTUNE Subjects. Journal of the American Society of Nephrology: JASN, 2016, 27, 814-823.	3.0	110
50	High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. Journal of Medical Genetics, 2012, 49, 756-767.	1.5	109
51	Single cell transcriptomics identifies focal segmental glomerulosclerosis remission endothelial biomarker. JCI Insight, 2020, 5, .	2.3	108
52	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 791-802.	3.9	102
53	Mutations in EMP2 Cause Childhood-Onset Nephrotic Syndrome. American Journal of Human Genetics, 2014, 94, 884-890.	2.6	101
54	Genotypeâ€“phenotype correlation in 440 patients with NPHP-related ciliopathies. Kidney International, 2011, 80, 1239-1245.	2.6	99

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55	FAT1 mutations cause a glomerulotubular nephropathy. Nature Communications, 2016, 7, 10822.	5.8	99
56	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. American Journal of Human Genetics, 2015, 96, 81-92.	2.6	98
57	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	2.3	95
58	Rationale and design of the Kidney Precision Medicine Project. Kidney International, 2021, 99, 498-510.	2.6	94
59	Mutation analysis of NPHP6/CEP290 in patients with Joubert syndrome and Senior Loken syndrome. Journal of Medical Genetics, 2007, 44, 657-663.	1.5	93
60	Exome Sequencing Reveals Cubilin Mutation as a Single-Gene Cause of Proteinuria. Journal of the American Society of Nephrology: JASN, 2011, 22, 1815-1820.	3.0	90
61	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914.	2.6	90
62	Mutational analysis of the RPGRIP1L gene in patients with Joubert syndrome and nephronophthisis. Kidney International, 2007, 72, 1520-1526.	2.6	88
63	Molecular Genetics of Nephronophthisis and Medullary Cystic Kidney Disease. Journal of the American Society of Nephrology: JASN, 2000, 11, 1753-1761.	3.0	88
64	Mutations of the Uromodulin gene in MCKD type 2 patients cluster in exon 4, which encodes three EGF-like domains. Kidney International, 2003, 64, 1580-1587.	2.6	87
65	Identification of the first AHI1 gene mutations in nephronophthisis-associated Joubert syndrome. Pediatric Nephrology, 2006, 21, 32-35.	0.9	87
66	Mutation analysis in Bardetâ€“Biedl syndrome by DNA pooling and massively parallel resequencing in 105 individuals. Human Genetics, 2011, 129, 79-90.	1.8	80
67	Mutation analysis in nephronophthisis using a combined approach of homozygosity mapping, CEL I endonuclease cleavage, and direct sequencing. Human Mutation, 2008, 29, 418-426.	1.1	76
68	Chilren with ocular motor apraxia type Cogan carry deletions in the gene (NPHP1) for juvenile nephronopthisis. Journal of Pediatrics, 2000, 136, 828-831.	0.9	75
69	Identification of two novel CAKUT-causing genes by massively parallel exon resequencing of candidate genes in patients with unilateral renal agenesis. Kidney International, 2012, 81, 196-200.	2.6	75
70	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	1.5	75
71	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.	2.2	74
72	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.	2.6	74

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73	Analysis of orbital T cells in thyroid-associated ophthalmopathy. Clinical and Experimental Immunology, 1998, 112, 427-434.	1.1	73
74	Organoid single cell profiling identifies a transcriptional signature of glomerular disease. JCI Insight, 2019, 4, .	2.3	73
75	Genetic and physical interaction between the NPHP5 and NPHP6 gene products. Human Molecular Genetics, 2008, 17, 3655-3662.	1.4	72
76	Identification of 11 novel mutations in eight BBS genes by high-resolution homozygosity mapping. Journal of Medical Genetics, 2010, 47, 262-267.	1.5	67
77	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	2.6	67
78	A reference tissue atlas for the human kidney. Science Advances, 2022, 8, .	4.7	67
79	SARS-CoV-2 receptor networks in diabetic and COVID-19â€“associated kidney disease. Kidney International, 2020, 98, 1502-1518.	2.6	64
80	WDR19 : An ancient, retrograde, intraflagellar ciliary protein is mutated in autosomal recessive retinitis pigmentosa and in Seniorâ€Loken syndrome. Clinical Genetics, 2013, 84, 150-159.	1.0	63
81	Mutation of the Mg2+ Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype. Journal of the American Society of Nephrology: JASN, 2013, 24, 967-977.	3.0	63
82	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	2.6	63
83	Mutational analysis of theNPHP4 gene in 250 patients with nephronophthisis. Human Mutation, 2005, 25, 411-411.	1.1	60
84	A multimodal and integrated approach to interrogate human kidney biopsies with rigor and reproducibility: guidelines from the Kidney Precision Medicine Project. Physiological Genomics, 2021, 53, 1-11.	1.0	59
85	Mapping of Gene Loci for Nephronophthisis Type 4 and Senior-LÃ¸ken Syndrome, to Chromosome 1p36. American Journal of Human Genetics, 2002, 70, 1240-1246.	2.6	56
86	Establishing an algorithm for molecular genetic diagnostics in 127 families with juvenile nephronophthisis. Kidney International, 2001, 59, 434-445.	2.6	53
87	Glycine Amidinotransferase (GATM), Renal Fanconi Syndrome, and Kidney Failure. Journal of the American Society of Nephrology: JASN, 2018, 29, 1849-1858.	3.0	53
88	Confirmation of the ATP6B1 gene as responsible for distal renal tubular acidosis. Pediatric Nephrology, 2003, 18, 105-109.	0.9	51
89	Nephrocystin. Journal of the American Society of Nephrology: JASN, 2000, 11, 270-282.	3.0	49
90	The Uromodulin C744G mutation causes MCKD2 and FJHN in children and adults and may be due to a possible founder effect. Kidney International, 2007, 71, 574-581.	2.6	48

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91	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. Journal of Medical Genetics, 2016, 53, 62-72.	1.5	48
92	Expression and Phenotype Analysis of the Nephrocystin-1 and Nephrocystin-4 Homologs in Caenorhabditiselegans. Journal of the American Society of Nephrology: JASN, 2005, 16, 676-687.	3.0	45
93	Identification of the First Gene Locus (SSNS1) for Steroid-Sensitive Nephrotic Syndrome on Chromosome 2p. Journal of the American Society of Nephrology: JASN, 2003, 14, 1897-1900.	3.0	42
94	Using Population Genetics to Interrogate the Monogenic Nephrotic Syndrome Diagnosis in a Case Cohort. Journal of the American Society of Nephrology: JASN, 2016, 27, 1970-1983.	3.0	41
95	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214.	1.5	39
96	Orbital tissue-derived T lymphocytes from patients with Graves' ophthalmopathy recognize autologous orbital antigens. Journal of Clinical Endocrinology and Metabolism, 1996, 81, 3045-3050.	1.8	36
97	Medullary cystic kidney disease type 1: mutational analysis in 37 genes based on haplotype sharing. Human Genetics, 2006, 119, 649-658.	1.8	34
98	Whole Exome Sequencing Reveals Novel PHEX Splice Site Mutations in Patients with Hypophosphatemic Rickets. PLoS ONE, 2015, 10, e0130729.	1.1	32
99	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	1.5	32
100	Identification of a Gene Locus for Senior-LÃ¸ken Syndrome in the Region of the Nephronophthisis Type 3 Gene. Journal of the American Society of Nephrology: JASN, 2002, 13, 75-79.	3.0	32
101	A novel chromosome 19p13.12 deletion in a child with multiple congenital anomalies. American Journal of Medical Genetics, Part A, 2009, 149A, 396-402.	0.7	31
102	Children with ocular motor apraxia type Cogan carry deletions in the gene () for juvenile nephronophthisis. Journal of Pediatrics, 2000, 136, 0828-0831.	0.9	30
103	Refinement of the Gene Locus for Autosomal Dominant Medullary Cystic Kidney Disease Type 1 (MCKD1) and Construction of a Physical and Partial Transcriptional Map of the Region. Genomics, 2001, 72, 278-284.	1.3	29
104	Retinitis pigmentosa and renal failure in a patient with mutations in INVS. Nephrology Dialysis Transplantation, 2006, 21, 1989-1991.	0.4	28
105	A deletion distinct from the classical homologous recombination of juvenile nephronophthisis type 1 (NPH1) allows exact molecular definition of deletion breakpoints. Human Mutation, 2000, 16, 211-223.	1.1	27
106	Mapping a new suggestive gene locus for autosomal dominant nephrolithiasis to chromosome 9q33.2â€“q34.2 by total genome search for linkage. Nephrology Dialysis Transplantation, 2005, 20, 909-914.	0.4	26
107	Pseudodominant inheritance of nephronophthisis caused by a homozygous NPHP1 deletion. Pediatric Nephrology, 2011, 26, 967-971.	0.9	26
108	Refinement of the critical region for MCKD1 by detection of transcontinental haplotype sharing. Kidney International, 2003, 64, 788-792.	2.6	24

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109	Exome capture and massively parallel sequencing identifies a novel HPSE2 mutation in a Saudi Arabian child with Ochoa (urofacial) syndrome. Journal of Pediatric Urology, 2011, 7, 569-573.	0.6	23
110	Endoplasmic reticulumâ€“associated degradation is required for nephrin maturation and kidney glomerular filtration function. Journal of Clinical Investigation, 2021, 131, .	3.9	21
111	Cadherin-11, Sparc-related modular calcium binding protein-2, and Pigment epithelium-derived factor are promising non-invasive biomarkers of kidney fibrosis. Kidney International, 2021, 100, 672-683.	2.6	21
112	Telomeric refinement of the MCKD1 locuson chromosome 1q21**See Editorial by Bichet and Fujiwara, p. 864.. Kidney International, 2004, 66, 580-585.	2.6	20
113	Clinical characterization and NPHP1 mutations in nephronophthisis and associated ciliopathies: A single center experience. Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia, 2012, 23, 1090.	0.4	18
114	Improved strategy for molecular genetic diagnostics in juvenile nephronophthisis. American Journal of Kidney Diseases, 2001, 37, 1131-1139.	2.1	17
115	Mapping of a new locus for congenital anomalies of the kidney and urinary tract on chromosome 8q24. Nephrology Dialysis Transplantation, 2010, 25, 1496-1501.	0.4	17
116	Novel compound heterozygous mutations in AMN cause Imerslund-GrÃsbeck syndrome in two half-sisters: a case report. BMC Medical Genetics, 2015, 16, 35.	2.1	15
117	Molecular Cloning of the Critical Region for Glomerulopathy with Fibronectin Deposits (GFND) and Evaluation of Candidate Genes. Genomics, 2000, 68, 127-135.	1.3	14
118	Mutation analysis of the Uromodulin gene in 96 individuals with urinary tract anomalies (CAKUT). Pediatric Nephrology, 2009, 24, 55-60.	0.9	14
119	Mutational analysis in 119 families with nephronophthisis. Pediatric Nephrology, 2007, 22, 366-370.	0.9	13
120	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. European Journal of Human Genetics, 2018, 26, 1266-1271.	1.4	12
121	Integrated single-cell sequencing and histopathological analyses reveal diverse injury and repair responses in a participant with acute kidney injury: a clinical-molecular-pathologic correlation. Kidney International, 2022, 101, 1116-1125.	2.6	11
122	Glomerular endothelial cell-podocyte stresses and crosstalk in structurally normal kidney transplants. Kidney International, 2022, 101, 779-792.	2.6	11
123	Clinical and histological presentation of 3 siblings with mutations in the NPHP4 gene. American Journal of Kidney Diseases, 2004, 43, 358-364.	2.1	10
124	Evaluating Mendelian nephrotic syndrome genes for evidence for risk alleles or oligogenicity that explain heritability. Pediatric Nephrology, 2017, 32, 467-476.	0.9	9
125	Hypertension induces glomerulosclerosis in phospholipase C-Îµ1 deficiency. American Journal of Physiology - Renal Physiology, 2020, 318, F1177-F1187.	1.3	9
126	Homozygous NPHP1 deletions in Egyptian children with nephronophthisis including an infantile onset patient. Pediatric Nephrology, 2010, 25, 2193-2194.	0.9	7

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127	Renalâ€hepaticâ€pancreatic dysplasia: A sibship with skeletal and central nervous system anomalies and NPHP3 mutation. American Journal of Medical Genetics, Part A, 2013, 161, 1743-1749.	0.7	7
128	Patient perspectives and involvement in precision medicine research. Kidney International, 2021, 99, 511-514.	2.6	5
129	PKD2 founder mutation is the most common mutation of polycystic kidney disease in Taiwan. Npj Genomic Medicine, 2022, 7, .	1.7	4
130	A boy with proteinuria and focal global glomerulosclerosis: Question. Pediatric Nephrology, 2015, 30, 1945-1946.	0.9	2
131	tarSVM: Improving the accuracy of variant calls derived from microfluidic PCR-based targeted next generation sequencing using a support vector machine. BMC Bioinformatics, 2016, 17, 233.	1.2	2
132	Is ciliary Hedgehog signalling dispensable in the kidneys?. Nature Reviews Nephrology, 2018, 14, 415-416.	4.1	2
133	Polycystic kidney and hepatic disease with mental retardation is nephronophthisis 11 caused by MKS3/TMEM67 mutations. Pediatric Nephrology, 2010, 25, 2375-2376.	0.9	1
134	A Familial Infantile Renal Failure. Kidney International Reports, 2017, 2, 130-133.	0.4	1
135	A boy with proteinuria and focal global glomerulosclerosis: Answers. Pediatric Nephrology, 2015, 30, 1947-1949.	0.9	0
136	A Case of Hyperphosphatemia and Elevated Fibroblast Growth Factor 23: A Brief Review of Hyperphosphatemia and Fibroblast Growth Factor 23 Pathway. Kidney International Reports, 2017, 2, 1238-1242.	0.4	0
137	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 1362-1362.	3.9	0