

# Edgar A Otto

## List of Publications by Year in descending order

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137  
papers

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12597

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148  
docs citations

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times ranked

15891  
citing authors

#	ARTICLE	IF	CITATIONS
1	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. <i>Kidney International</i> , 2022, 101, 1116-1125.	2.6	11
2	Glomerular endothelial cell-podocyte stresses and crosstalk in structurally normal kidney transplants. <i>Kidney International</i> , 2022, 101, 779-792.	2.6	11
3	A reference tissue atlas for the human kidney. <i>Science Advances</i> , 2022, 8, .	4.7	67
4	PKD2 founder mutation is the most common mutation of polycystic kidney disease in Taiwan. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	4
5	A multimodal and integrated approach to interrogate human kidney biopsies with rigor and reproducibility: guidelines from the Kidney Precision Medicine Project. <i>Physiological Genomics</i> , 2021, 53, 1-11.	1.0	59
6	Patient perspectives and involvement in precision medicine research. <i>Kidney International</i> , 2021, 99, 511-514.	2.6	5
7	Rationale and design of the Kidney Precision Medicine Project. <i>Kidney International</i> , 2021, 99, 498-510.	2.6	94
8	Endoplasmic reticulum-associated degradation is required for nephrin maturation and kidney glomerular filtration function. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	21
9	Cadherin-11, Sparc-related modular calcium binding protein-2, and Pigment epithelium-derived factor are promising non-invasive biomarkers of kidney fibrosis. <i>Kidney International</i> , 2021, 100, 672-683.	2.6	21
10	SARS-CoV-2 receptor networks in diabetic and COVID-19-associated kidney disease. <i>Kidney International</i> , 2020, 98, 1502-1518.	2.6	64
11	Hypertension induces glomerulosclerosis in phospholipase C- $\mu$ 1 deficiency. <i>American Journal of Physiology - Renal Physiology</i> , 2020, 318, F1177-F1187.	1.3	9
12	Single cell transcriptomics identifies focal segmental glomerulosclerosis remission endothelial biomarker. <i>JCI Insight</i> , 2020, 5, .	2.3	108
13	Organoid single cell profiling identifies a transcriptional signature of glomerular disease. <i>JCI Insight</i> , 2019, 4, .	2.3	73
14	Glycine Amidinotransferase (GATM), Renal Fanconi Syndrome, and Kidney Failure. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 1849-1858.	3.0	53
15	Is ciliary Hedgehog signalling dispensable in the kidneys?. <i>Nature Reviews Nephrology</i> , 2018, 14, 415-416.	4.1	2
16	Single-cell analysis of progenitor cell dynamics and lineage specification in the human fetal kidney. <i>Development (Cambridge)</i> , 2018, 145, .	1.2	130
17	High-Throughput Screening Enhances Kidney Organoid Differentiation from Human Pluripotent Stem Cells and Enables Automated Multidimensional Phenotyping. <i>Cell Stem Cell</i> , 2018, 22, 929-940.e4.	5.2	328
18	An eQTL Landscape of Kidney Tissue in Human Nephrotic Syndrome. <i>American Journal of Human Genetics</i> , 2018, 103, 232-244.	2.6	147

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19	Biallelic variants in the ciliary gene TMEM67 cause RHYNS syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 1266-1271.	1.4	12
20	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. <i>New England Journal of Medicine</i> , 2017, 376, 742-754.	13.9	120
21	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
22	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. <i>American Journal of Human Genetics</i> , 2017, 101, 789-802.	2.6	63
23	A Case of Hyperphosphatemia and Elevated Fibroblast Growth Factor 23: A Brief Review of Hyperphosphatemia and Fibroblast Growth Factor 23 Pathway. <i>Kidney International Reports</i> , 2017, 2, 1238-1242.	0.4	0
24	Evaluating Mendelian nephrotic syndrome genes for evidence for risk alleles or oligogenicity that explain heritability. <i>Pediatric Nephrology</i> , 2017, 32, 467-476.	0.9	9
25	A Familial Infantile Renal Failure. <i>Kidney International Reports</i> , 2017, 2, 130-133.	0.4	1
26	tarSVM: Improving the accuracy of variant calls derived from microfluidic PCR-based targeted next generation sequencing using a support vector machine. <i>BMC Bioinformatics</i> , 2016, 17, 233.	1.2	2
27	FAT1 mutations cause a glomerulotubular nephropathy. <i>Nature Communications</i> , 2016, 7, 10822.	5.8	99
28	Using Population Genetics to Interrogate the Monogenic Nephrotic Syndrome Diagnosis in a Case Cohort. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 1970-1983.	3.0	41
29	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
30	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. <i>Journal of Medical Genetics</i> , 2016, 53, 62-72.	1.5	48
31	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
32	Integrative Genomics Identifies Novel Associations with APOL1 Risk Genotypes in Black NEPTUNE Subjects. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 814-823.	3.0	110
33	Whole Exome Sequencing Reveals Novel PHEX Splice Site Mutations in Patients with Hypophosphatemic Rickets. <i>PLoS ONE</i> , 2015, 10, e0130729.	1.1	32
34	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
35	A boy with proteinuria and focal global glomerulosclerosis: Answers. <i>Pediatric Nephrology</i> , 2015, 30, 1947-1949.	0.9	0
36	A boy with proteinuria and focal global glomerulosclerosis: Question. <i>Pediatric Nephrology</i> , 2015, 30, 1945-1946.	0.9	2

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37	TMEM231, mutated in orofacioidigital and Meckel syndromes, organizes the ciliary transition zone. <i>Journal of Cell Biology</i> , 2015, 209, 129-142.	2.3	95
38	Novel compound heterozygous mutations in AMN cause Imerslund-GrÅsbeck syndrome in two half-sisters: a case report. <i>BMC Medical Genetics</i> , 2015, 16, 35.	2.1	15
39	The kinetochore protein, <i>CENPF</i> , is mutated in human ciliopathy and microcephaly phenotypes. <i>Journal of Medical Genetics</i> , 2015, 52, 147-156.	1.5	75
40	<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
41	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2015, 125, 2375-2384.	3.9	159
42	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
43	Mutations in <i>RSPH1</i> Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2014, 189, 707-717.	2.5	191
44	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. <i>Kidney International</i> , 2014, 85, 880-887.	2.6	67
45	Mutations in EMP2 Cause Childhood-Onset Nephrotic Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 884-890.	2.6	101
46	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 905-914.	2.6	90
47	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
48	ANKS6 is a central component of a nephronophthisis module linking NEK8 to INVS and NPHP3. <i>Nature Genetics</i> , 2013, 45, 951-956.	9.4	183
49	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. <i>American Journal of Human Genetics</i> , 2013, 93, 336-345.	2.6	183
50	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 93, 672-686.	2.6	184
51	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. <i>American Journal of Human Genetics</i> , 2013, 93, 711-720.	2.6	135
52	<i>WDR19</i> : An ancient, retrograde, intraflagellar ciliary protein is mutated in autosomal recessive retinitis pigmentosa and in Senior-Løken syndrome. <i>Clinical Genetics</i> , 2013, 84, 150-159.	1.0	63
53	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. <i>American Journal of Human Genetics</i> , 2013, 93, 915-925.	2.6	196
54	Renal-Hepatic-Pancreatic dysplasia: A sibship with skeletal and central nervous system anomalies and <i>NPHP3</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1743-1749.	0.7	7

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55	Mutation of the Mg <sup>2+</sup> Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 967-977.	3.0	63
56	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. <i>Journal of Clinical Investigation</i> , 2013, 123, 5179-5189.	3.9	275
57	ARHGDI1 mutations cause nephrotic syndrome via defective RHO GTPase signaling. <i>Journal of Clinical Investigation</i> , 2013, 123, 3243-3253.	3.9	196
58	Identification of two novel CAKUT-causing genes by massively parallel exon resequencing of candidate genes in patients with unilateral renal agenesis. <i>Kidney International</i> , 2012, 81, 196-200.	2.6	75
59	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. <i>Cell</i> , 2012, 150, 533-548.	13.5	347
60	High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. <i>Journal of Medical Genetics</i> , 2012, 49, 756-767.	1.5	109
61	FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney failure to defective DNA damage repair. <i>Nature Genetics</i> , 2012, 44, 910-915.	9.4	205
62	Clinical characterization and NPHP1 mutations in nephronophthisis and associated ciliopathies: A single center experience. <i>Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia</i> , 2012, 23, 1090.	0.4	18
63	Genotype-phenotype correlation in 440 patients with NPHP-related ciliopathies. <i>Kidney International</i> , 2011, 80, 1239-1245.	2.6	99
64	Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. <i>Journal of Medical Genetics</i> , 2011, 48, 105-116.	1.5	123
65	A transition zone complex regulates mammalian ciliogenesis and ciliary membrane composition. <i>Nature Genetics</i> , 2011, 43, 776-784.	9.4	556
66	Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. <i>Cell</i> , 2011, 145, 513-528.	13.5	531
67	Exome capture and massively parallel sequencing identifies a novel HPSE2 mutation in a Saudi Arabian child with Ochoa (urofacial) syndrome. <i>Journal of Pediatric Urology</i> , 2011, 7, 569-573.	0.6	23
68	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. <i>Nature Genetics</i> , 2011, 43, 189-196.	9.4	326
69	Pseudodominant inheritance of nephronophthisis caused by a homozygous NPHP1 deletion. <i>Pediatric Nephrology</i> , 2011, 26, 967-971.	0.9	26
70	Mutation analysis in Bardet-Biedl syndrome by DNA pooling and massively parallel resequencing in 105 individuals. <i>Human Genetics</i> , 2011, 129, 79-90.	1.8	80
71	<i>MYO1E</i> Mutations and Childhood Familial Focal Segmental Glomerulosclerosis. <i>New England Journal of Medicine</i> , 2011, 365, 295-306.	13.9	221
72	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

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73	Homozygous NPHP1 deletions in Egyptian children with nephronophthisis including an infantile onset patient. <i>Pediatric Nephrology</i> , 2010, 25, 2193-2194.	0.9	7
74	Polycystic kidney and hepatic disease with mental retardation is nephronophthisis 11 caused by MKS3/TMEM67 mutations. <i>Pediatric Nephrology</i> , 2010, 25, 2375-2376.	0.9	1
75	AHL1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. <i>Nature Genetics</i> , 2010, 42, 175-180.	9.4	171
76	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. <i>Nature Genetics</i> , 2010, 42, 619-625.	9.4	261
77	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. <i>Nature Genetics</i> , 2010, 42, 840-850.	9.4	295
78	Identification of 11 novel mutations in eight BBS genes by high-resolution homozygosity mapping. <i>Journal of Medical Genetics</i> , 2010, 47, 262-267.	1.5	67
79	Mapping of a new locus for congenital anomalies of the kidney and urinary tract on chromosome 8q24. <i>Nephrology Dialysis Transplantation</i> , 2010, 25, 1496-1501.	0.4	17
80	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. <i>Journal of Clinical Investigation</i> , 2010, 120, 791-802.	3.9	102
81	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. <i>Journal of Clinical Investigation</i> , 2010, 120, 1362-1362.	3.9	0
82	A Systematic Approach to Mapping Recessive Disease Genes in Individuals from Outbred Populations. <i>PLoS Genetics</i> , 2009, 5, e1000353.	1.5	144
83	Hypomorphic mutations in meckelin (MKS3/TMEM67) cause nephronophthisis with liver fibrosis (NPHP11). <i>Journal of Medical Genetics</i> , 2009, 46, 663-670.	1.5	121
84	A novel chromosome 19p13.12 deletion in a child with multiple congenital anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 396-402.	0.7	31
85	Mutation analysis of the Uromodulin gene in 96 individuals with urinary tract anomalies (CAKUT). <i>Pediatric Nephrology</i> , 2009, 24, 55-60.	0.9	14
86	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. <i>Nature Genetics</i> , 2009, 41, 739-745.	9.4	255
87	Nephronophthisis. <i>Journal of the American Society of Nephrology: JASN</i> , 2009, 20, 23-35.	3.0	332
88	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
89	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. <i>American Journal of Human Genetics</i> , 2008, 83, 170-179.	2.6	352
90	CC2D2A Is Mutated in Joubert Syndrome and Interacts with the Ciliopathy-Associated Basal Body Protein CEP290. <i>American Journal of Human Genetics</i> , 2008, 83, 559-571.	2.6	202

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91	Mutations in <i>FN1</i> 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
92	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
93	Genetic and physical interaction between the NPHP5 and NPHP6 gene products. Human Molecular Genetics, 2008, 17, 3655-3662.	1.4	72
94	Evidence of Oligogenic Inheritance in Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2007, 18, 2789-2795.	3.0	141
95	Mutational analysis of the RPGRIP1L gene in patients with Joubert syndrome and nephronophthisis. Kidney International, 2007, 72, 1520-1526.	2.6	88
96	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
97	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
98	Transcription Factor SIX5 Is Mutated in Patients with Branchio-Oto-Renal Syndrome. American Journal of Human Genetics, 2007, 80, 800-804.	2.6	164
99	Loss of GLIS2 causes nephronophthisis in humans and mice by increased apoptosis and fibrosis. Nature Genetics, 2007, 39, 1018-1024.	9.4	221
100	Mutational analysis in 119 families with nephronophthisis. Pediatric Nephrology, 2007, 22, 366-370.	0.9	13
101	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature Genetics, 2006, 38, 674-681.	9.4	535
102	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
103	Identification of the first AH11 gene mutations in nephronophthisis-associated Joubert syndrome. Pediatric Nephrology, 2006, 21, 32-35.	0.9	87
104	Medullary cystic kidney disease type 1: mutational analysis in 37 genes based on haplotype sharing. Human Genetics, 2006, 119, 649-658.	1.8	34
105	Retinitis pigmentosa and renal failure in a patient with mutations in INVS. Nephrology Dialysis Transplantation, 2006, 21, 1989-1991.	0.4	28
106	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
107	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
108	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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109	Mutational analysis of the NPHP4 gene in 250 patients with nephronophthisis. <i>Human Mutation</i> , 2005, 25, 411-411.	1.1	60
110	Expression and Phenotype Analysis of the Nephrocystin-1 and Nephrocystin-4 Homologs in <i>Caenorhabditis elegans</i> . <i>Journal of the American Society of Nephrology: JASN</i> , 2005, 16, 676-687.	3.0	45
111	Mapping a new suggestive gene locus for autosomal dominant nephrolithiasis to chromosome 9q33.2-q34.2 by total genome search for linkage. <i>Nephrology Dialysis Transplantation</i> , 2005, 20, 909-914.	0.4	26
112	SIX1 mutations cause branchio-oto-renal syndrome by disruption of EYA1-SIX1-DNA complexes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 8090-8095.	3.3	374
113	Telomeric refinement of the MCKD1 locus on chromosome 1q21**See Editorial by Bichet and Fujiwara, p. 864.. <i>Kidney International</i> , 2004, 66, 580-585.	2.6	20
114	Clinical and histological presentation of 3 siblings with mutations in the NPHP4 gene. <i>American Journal of Kidney Diseases</i> , 2004, 43, 358-364.	2.1	10
115	Confirmation of the ATP6B1 gene as responsible for distal renal tubular acidosis. <i>Pediatric Nephrology</i> , 2003, 18, 105-109.	0.9	51
116	Refinement of the critical region for MCKD1 by detection of transcontinental haplotype sharing. <i>Kidney International</i> , 2003, 64, 788-792.	2.6	24
117	Mutations of the Uromodulin gene in MCKD type 2 patients cluster in exon 4, which encodes three EGF-like domains. <i>Kidney International</i> , 2003, 64, 1580-1587.	2.6	87
118	Mutations in a novel gene, NPHP3, cause adolescent nephronophthisis, tapeto-retinal degeneration and hepatic fibrosis. <i>Nature Genetics</i> , 2003, 34, 455-459.	9.4	345
119	Mutations in INVS encoding inversin cause nephronophthisis type 2, linking renal cystic disease to the function of primary cilia and left-right axis determination. <i>Nature Genetics</i> , 2003, 34, 413-420.	9.4	582
120	Identification of the First Gene Locus (SSNS1) for Steroid-Sensitive Nephrotic Syndrome on Chromosome 2p. <i>Journal of the American Society of Nephrology: JASN</i> , 2003, 14, 1897-1900.	3.0	42
121	Mapping of Gene Loci for Nephronophthisis Type 4 and Senior-LÅken Syndrome, to Chromosome 1p36. <i>American Journal of Human Genetics</i> , 2002, 70, 1240-1246.	2.6	56
122	A Gene Mutated in Nephronophthisis and Retinitis Pigmentosa Encodes a Novel Protein, Nephroretinin, Conserved in Evolution. <i>American Journal of Human Genetics</i> , 2002, 71, 1161-1167.	2.6	193
123	Identification of a Gene Locus for Senior-LÅken Syndrome in the Region of the Nephronophthisis Type 3 Gene. <i>Journal of the American Society of Nephrology: JASN</i> , 2002, 13, 75-79.	3.0	32
124	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. <i>Genomics</i> , 2001, 72, 278-284.	1.3	29
125	Improved strategy for molecular genetic diagnostics in juvenile nephronophthisis. <i>American Journal of Kidney Diseases</i> , 2001, 37, 1131-1139.	2.1	17
126	Establishing an algorithm for molecular genetic diagnostics in 127 families with juvenile nephronophthisis. <i>Kidney International</i> , 2001, 59, 434-445.	2.6	53



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127	Mutation of BSND causes Bartter syndrome with sensorineural deafness and kidney failure. <i>Nature Genetics</i> , 2001, 29, 310-314.	9.4	510
128	Barttin is a Cl <sup>-</sup> channel $\beta$ -subunit crucial for renal Cl <sup>-</sup> reabsorption and inner ear K <sup>+</sup> secretion. <i>Nature</i> , 2001, 414, 558-561.	13.7	538
129	A deletion distinct from the classical homologous recombination of juvenile nephronophthisis type 1 (NPH1) allows exact molecular definition of deletion breakpoints. <i>Human Mutation</i> , 2000, 16, 211-223.	1.1	27
130	Molecular Cloning of the Critical Region for Glomerulopathy with Fibronectin Deposits (GFND) and Evaluation of Candidate Genes. <i>Genomics</i> , 2000, 68, 127-135.	1.3	14
131	Children with ocular motor apraxia type Cogan carry deletions in the gene (NPHP1) for juvenile nephronophthisis. <i>Journal of Pediatrics</i> , 2000, 136, 828-831.	0.9	75
132	Children with ocular motor apraxia type Cogan carry deletions in the gene ( ) for juvenile nephronophthisis. <i>Journal of Pediatrics</i> , 2000, 136, 0828-0831.	0.9	30
133	Nephrocystin. <i>Journal of the American Society of Nephrology: JASN</i> , 2000, 11, 270-282.	3.0	49
134	Molecular Genetics of Nephronophthisis and Medullary Cystic Kidney Disease. <i>Journal of the American Society of Nephrology: JASN</i> , 2000, 11, 1753-1761.	3.0	88
135	Analysis of orbital T cells in thyroid-associated ophthalmopathy. <i>Clinical and Experimental Immunology</i> , 1998, 112, 427-434.	1.1	73
136	A novel gene encoding an SH3 domain protein is mutated in nephronophthisis type 1. <i>Nature Genetics</i> , 1997, 17, 149-153.	9.4	327
137	Orbital tissue-derived T lymphocytes from patients with Graves' ophthalmopathy recognize autologous orbital antigens. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1996, 81, 3045-3050.	1.8	36