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List of Publications by Year
in descending order

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63 papers	5,593 citations	101543 36 h-index	98798 67 g-index
75 all docs	75 docs citations	75 times ranked	6840 citing authors

#	Article	IF	Citations
1	Multidisciplinary approaches for elucidating genetics and molecular pathogenesis of urinary tract malformations. Kidney International, 2022, 101, 473-484.	5.2	16
2	GWAS in Mice Maps Susceptibility to HIV-Associated Nephropathy to the Ssbp2 Locus. Journal of the American Society of Nephrology: JASN, 2022, 33, 108-120.	6.1	3
3	Genetics in chronic kidney disease: conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney International, 2022, 101, 1126-1141.	5.2	46
4	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
5	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
6	Integrative analysis of rare copy number variants and gene expression data in alopecia areata implicates an aetiological role for autophagy. Experimental Dermatology, 2020, 29, 243-253.	2.9	21
7	Longitudinal Changes in Health-Related Quality of Life in Primary Glomerular Disease: Results From the CureGN Study. Kidney International Reports, 2020, 5, 1679-1689.	0.8	17
8	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
9	Type IV Collagen Mutations in Familial IgA Nephropathy. Kidney International Reports, 2020, 5, 1075-1078.	0.8	26
10	Pilot Study of Return of Genetic Results to Patients in Adult Nephrology. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 651-664.	4.5	28
11	The genetic architecture of membranous nephropathy and its potential to improve non-invasive diagnosis. Nature Communications, 2020, 11, 1600.	12.8	120
12	Persistent Disease Activity in Patients With Long-Standing Glomerular Disease. Kidney International Reports, 2020, 5, 860-871.	0.8	2
13	Human and mouse studies establish TBX6 in Mendelian CAKUT and as a potential driver of kidney defects associated with the 16p11.2 microdeletion syndrome. Kidney International, 2020, 98, 1020-1030.	5.2	17
14	Phenocopies, Phenotypic Expansion, and Coincidental Diagnoses: Time to Abandon Targeted Gene Panels?. American Journal of Kidney Diseases, 2020, 76, 451-453.	1.9	7
15	Exome-Based Rare-Variant Analyses in CKD. Journal of the American Society of Nephrology: JASN, 2019, 30, 1109-1122.	6.1	40
16	Immunohistochemical expression pattern of RIP5, FGFR1, FGFR2 and HIP2 in the normal human kidney development. Acta Histochemica, 2019, 121, 531-538.	1.8	14
17	Health-related quality of life in glomerular disease. Kidney International, 2019, 95, 1209-1224.	5.2	38
18	Diagnostic Utility of Exome Sequencing for Kidney Disease. New England Journal of Medicine, 2019, 380, 142-151.	27.0	456

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19	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127.	21.4	144
20	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11.	3.9	60
21	CureGN Study Rationale, Design, and Methods: Establishing a Large Prospective Observational Study of Glomerular Disease. American Journal of Kidney Diseases, 2019, 73, 218-229.	1.9	68
22	A noncoding variant in GANAB explains isolated polycystic liver disease (PCLD) in a large family. Human Mutation, 2018, 39, 378-382.	2.5	21
23	Clinical Characteristics and Treatment Patterns of Children and Adults With IgA Nephropathy or IgA Vasculitis: Findings From the CureGN Study. Kidney International Reports, 2018, 3, 1373-1384.	0.8	39
24	Whole-Exome Sequencing in Adults With Chronic Kidney Disease. Annals of Internal Medicine, 2018, 168, 100.	3.9	154
25	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
26	Genetic basis of human congenital anomalies of the kidney and urinary tract. Journal of Clinical Investigation, 2018, 128, 4-15.	8.2	91
27	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	27.0	120
28	A Dominant Mutation in Nuclear Receptor Interacting Protein 1 Causes Urinary Tract Malformations via Dysregulation of Retinoic Acid Signaling. Journal of the American Society of Nephrology: JASN, 2017, 28, 2364-2376.	6.1	40
29	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	6.2	63
30	A Novel SUCLA2 Mutation Presenting as a Complex Childhood Movement Disorder. Journal of Child Neurology, 2017, 32, 246-250.	1.4	15
31	Isolated polycystic liver disease genes define effectors of polycystin-1 function. Journal of Clinical Investigation, 2017, 127, 1772-1785.	8.2	137
32	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.	2.6	2
33	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
34	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
35	Genomic imbalances in pediatric patients with chronic kidney disease. Journal of Clinical Investigation, 2015, 125, 2171-2178.	8.2	68
36	Copy number variation analysis identifies novel CAKUT candidate genes in children with a solitary functioning kidney. Kidney International, 2015, 88, 1402-1410.	5.2	65

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37	Recessive mutations in CAKUT and VACTERL association. Kidney International, 2014, 85, 1253-1255.	5.2	4
38	The emerging role of genomics in the diagnosis and workup of congenital urinary tract defects: a novel deletion syndrome on chromosome 3q13.31-22.1. Pediatric Nephrology, 2014, 29, 257-267.	1.7	15
39	Clinical Implications of the Solitary Functioning Kidney. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 978-986.	4.5	81
40	Phenotypic Expansion of DGKE-Associated Diseases. Journal of the American Society of Nephrology: JASN, 2014, 25, 1408-1414.	6.1	59
41	Discovery of new risk loci for IgA nephropathy implicates genes involved in immunity against intestinal pathogens. Nature Genetics, 2014, 46, 1187-1196.	21.4	505
42	Î±â€“Intercalated cells defend the urinary system from bacterial infection. Journal of Clinical Investigation, 2014, 124, 2963-2976.	8.2	127
43	Mutations in DSTYK and Dominant Urinary Tract Malformations. New England Journal of Medicine, 2013, 369, 621-629.	27.0	119
44	Geographic Differences in Genetic Susceptibility to IgA Nephropathy: GWAS Replication Study and Geospatial Risk Analysis. PLoS Genetics, 2012, 8, e1002765.	3.5	301
45	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997.	6.2	201
46	Infantile Encephaloneuromyopathy and Defective Mitochondrial Translation Are Due to a Homozygous RMND1 Mutation. American Journal of Human Genetics, 2012, 91, 729-736.	6.2	35
47	Exome sequencing identified MYO1E and NEIL1 as candidate genes for human autosomal recessive steroid-resistant nephrotic syndrome. Kidney International, 2011, 80, 389-396.	5.2	69
48	Genome-wide association study identifies susceptibility loci for IgA nephropathy. Nature Genetics, 2011, 43, 321-327.	21.4	528
49	HNF1B and PAX2 mutations are a common cause of renal hypodysplasia in the CKiD cohort. Pediatric Nephrology, 2011, 26, 897-903.	1.7	114
50	'Congenital solitary functioning kidneys: which ones warrant follow-up into adult life?'. Nephrology Dialysis Transplantation, 2011, 26, 1458-1460.	0.7	23
51	Familial forms of nephrotic syndrome. Pediatric Nephrology, 2010, 25, 241-252.	1.7	41
52	Urinary NGAL Marks Cystic Disease in HIV-Associated Nephropathy. Journal of the American Society of Nephrology: JASN, 2009, 20, 1687-1692.	6.1	47
53	A Recessive Gene for Primary Vesicoureteral Reflux Maps to Chromosome 12p11-q13. Journal of the American Society of Nephrology: JASN, 2009, 20, 1633-1640.	6.1	42
54	Renal outcome in patients with congenital anomalies of the kidney and urinary tract. Kidney International, 2009, 76, 528-533.	5.2	309

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55	Scara5 Is a Ferritin Receptor Mediating Non-Transferrin Iron Delivery. Developmental Cell, 2009, 16, 35-46.	7.0	264
56	Characterization of a large Lebanese family segregating IgA nephropathy. Nephrology Dialysis Transplantation, 2007, 22, 772-777.	0.7	39
57	Alterations of Type IV Collagen Î± Chains in Patients with Chronic Acquired Glomerulopathies: mRNA Levels, Protein Expression and Urinary Loss. American Journal of Nephrology, 2007, 27, 129-137.	3.1	7
58	Localization of a Gene for Nonsyndromic Renal Hypodysplasia to Chromosome 1p32-33. American Journal of Human Genetics, 2007, 80, 539-549.	6.2	33
59	Genetic approaches to human renal agenesis/hypoplasia and dysplasia. Pediatric Nephrology, 2007, 22, 1675-1684.	1.7	99
60	Familial Vesicoureteral Reflux: Testing Replication of Linkage in Seven New Multigenerational Kindreds. Journal of the American Society of Nephrology: JASN, 2005, 16, 1781-1787.	6.1	56
61	Cyclosporine in patients with steroid-resistant nephrotic syndrome: an open-label, nonrandomized, retrospective study. Clinical Therapeutics, 2004, 26, 1411-1418.	2.5	48
62	Recurrence of focal segmental glomerulosclerosis after renal transplantation in patients with mutations of podocin. American Journal of Kidney Diseases, 2003, 41, 1314-1321.	1.9	144
63	Apolipoprotein E in idiopathic nephrotic syndrome and focal segmental glomerulosclerosis. Kidney International, 2003, 63, 686-695.	5.2	23