Richard P Lifton

List of Publications by Year
in descending order

326 papers	69,393 citations	668 122 h-index	718 252 g-index
334 all docs	334 docs citations	334 times ranked	67842 citing authors

#	Article	IF	Citations
1	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
2	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,983
3	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. Nature, 2012, 485, 237-241.	13.7	1,863
4	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	6.0	1,749
5	Molecular basis of human hypertension: Role of angiotensinogen. Cell, 1992, 71, 169-180.	13.5	1,747
6	Molecular Mechanisms of Human Hypertension. Cell, 2001, 104, 545-556.	13.5	1,519
7	High Bone Density Due to a Mutation in LDL-Receptorâ€“Related Protein 5. New England Journal of Medicine, 2002, 346, 1513-1521.	13.9	1,498
8	Human Hypertension Caused by Mutations in WNK Kinases. Science, 2001, 293, 1107-1112.	6.0	1,344
9	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
10	Liddle's syndrome: Heritable human hypertension caused by mutations in the Î² subunit of the epithelial sodium channel. Cell, 1994, 79, 407-414.	13.5	1,230
11	Genetic diagnosis by whole exome capture and massively parallel DNA sequencing. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 19096-19101.	3.3	1,167
12	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
13	A chimaeric llÎ²-hydroxylase/aldosterone synthase gene causes glucocorticoid-remediable aldosteronism and human hypertension. Nature, 1992, 355, 262-265.	13.7	1,137
14	Gitelman's variant of Barter's syndrome, inherited hypokalaemic alkalosis, is caused by mutations in the thiazide-sensitive Naâ€“Cl cotransporter. Nature Genetics, 1996, 12, 24-30.	9.4	1,116
15	Exome sequencing identifies recurrent somatic RAC1 mutations in melanoma. Nature Genetics, 2012, 44, 1006-1014.	9.4	1,052
16	Paracellin-1, a Renal Tight Junction Protein Required for Paracellular Mg2+ Resorption. Science, 1999, 285, 103-106.	6.0	1,042
17	Sequence Variants in SLITRK1 Are Associated with Tourette's Syndrome. Science, 2005, 310, 317-320.	6.0	878
18	K ⁺ Channel Mutations in Adrenal Aldosterone-Producing Adenomas and Hereditary Hypertension. Science, 2011, 331, 768-772.	6.0	866

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19	Bartter's syndrome, hypokalaemic alkalosis with hypercalciuria, is caused by mutations in the Naâ€“Kâ€“2Cl cotransporter NKCC2. Nature Genetics, 1996, 13, 183-188.	9.4	838
20	Mutations in the chloride channel gene, CLCNKB, cause Bartter's syndrome type III. Nature Genetics, 1997, 17, 171-178.	9.4	812
21	De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223.	13.7	798
22	Genetic heterogeneity of Barter's syndrome revealed by mutations in the K+ channel, ROMK. Nature Genetics, 1996, 14, 152-156.	9.4	764
23	Mutations in subunits of the epithelial sodium channel cause salt wasting with hyperkalaemic acidosis, pseudohypoaldosteronism type 1. Nature Genetics, 1996, 12, 248-253.	9.4	752
24	Rare independent mutations in renal salt handling genes contribute to blood pressure variation. Nature Genetics, 2008, 40, 592-599.	9.4	728
25	Hypertension caused by a truncated epithelial sodium channel Î³ subunit: genetic heterogeneity of Liddle syndrome. Nature Genetics, 1995, 11, 76-82.	9.4	725
26	Genomic Analysis of Non- NF2 Meningiomas Reveals Mutations in TRAF7 , KLF4 , AKT1 , and SMO. Science, 2013, 339, 1077-1080.	6.0	714
27	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	6.0	646
28	Activating Mineralocorticoid Receptor Mutation in Hypertension Exacerbated by Pregnancy. Science, 2000, 289, 119-123.	6.0	635
29	Mutations in the gene encoding B1 subunit of H+-ATPase cause renal tubular acidosis with sensorineural deafness. Nature Genetics, 1999, 21, 84-90.	9.4	633
30	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	9.4	624
31	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
32	LRP6 Mutation in a Family with Early Coronary Disease and Metabolic Risk Factors. Science, 2007, 315, 1278-1282.	6.0	567
33	Mutations in kelch-like 3 and cullin 3 cause hypertension and electrolyte abnormalities. Nature, 2012, 482, 98-102.	13.7	560
34	Evidence for a Gene Influencing Blood Pressure on Chromosome 17. Hypertension, 2000, 36, 477-483.	1.3	534
35	Genome-wide association study identifies susceptibility loci for IgA nephropathy. Nature Genetics, 2011, 43, 321-327.	9.4	528
36	Somatic and germline CACNA1D calcium channel mutations in aldosterone-producing adenomas and primary aldosteronism. Nature Genetics, 2013, 45, 1050-1054.	9.4	519

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37	Discovery of new risk loci for IgA nephropathy implicates genes involved in immunity against intestinal pathogens. Nature Genetics, 2014, 46, 1187-1196.	9.4	505
38	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.	3.0	499
39	Molecular Cytogenetic Analysis and Resequencing of Contactin Associated Protein-Like 2 in Autism Spectrum Disorders. American Journal of Human Genetics, 2008, 82, 165-173.	2.6	494
40	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. Nature, 2010, 467, 207-210.	13.7	457
41	Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SeSAME) Tj ETQq1 1 0.784314 rgBT /Overloc the United States of America, 2009, 106, 5842-5847.	3.3	433
42	Recessive mutations in DGKE cause atypical hemolytic-uremic syndrome. Nature Genetics, 2013, 45, 531-536.	9.4	419
43	Mutation of NLRC4 causes a syndrome of enterocolitis and autoinflammation. Nature Genetics, 2014, 46, 1135-1139.	9.4	417
44	Exome sequencing links mutations in PARN and RTEL1 with familial pulmonary fibrosis and telomere shortening. Nature Genetics, 2015, 47, 512-517.	9.4	385
45	Mutations in the mineralocorticoid receptor gene cause autosomal dominant pseudohypoaldosteronism type I. Nature Genetics, 1998, 19, 279-281.	9.4	377
46	Absence of linkage between the angiotensin converting enzyme locus and human essential hypertension. Nature Genetics, 1992, 1, 72-75.	9.4	376
47	Molecular pathogenesis of inherited hypertension with hyperkalemia: The Na-Cl cotransporter is inhibited by wild-type but not mutant WNK4. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 680-684.	3.3	375
48	Mutations in ATP6N1B, encoding a new kidney vacuolar proton pump 116-kD subunit, cause recessive distal renal tubular acidosis with preserved hearing. Nature Genetics, 2000, 26, 71-75.	9.4	368
49	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. Science Immunology, 2021, 6, .	5.6	357
50	Roles of the cationâ€“chloride cotransporters in neurological disease. Nature Clinical Practice Neurology, 2008, 4, 490-503.	2.7	354
51	Exome sequencing identifies recurrent mutations in NF1 and RASopathy genes in sun-exposed melanomas. Nature Genetics, 2015, 47, 996-1002.	9.4	348
52	WNK4 regulates the balance between renal NaCl reabsorption and K+ secretion. Nature Genetics, 2003, 35, 372-376.	9.4	347
53	Genomic landscape of cutaneous T cell lymphoma. Nature Genetics, 2015, 47, 1011-1019.	9.4	347
54	Wnk4 controls blood pressure and potassium homeostasis via regulation of mass and activity of the distal convoluted tubule. Nature Genetics, 2006, 38, 1124-1132.	9.4	333

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55	Multilocus linkage identifies two new loci for a mendelian form of stroke, cerebral cavernous malformation, at 7p15-13 and 3q25.2-27. Human Molecular Genetics, 1998, 7, 1851-1858.	1.4	331
56	A Cluster of Metabolic Defects Caused by Mutation in a Mitochondrial tRNA. Science, 2004, 306, 1190-1194.	6.0	328
57	Hereditary hypertension caused by chimaeric gene duplications and ectopic expression of aldosterone synthase. Nature Genetics, 1992, 2, 66-74.	9.4	325
58	Mutational landscape of MCPyV-positive and MCPyV-negative Merkel cell carcinomas with implications for immunotherapy. Oncotarget, 2016, 7, 3403-3415.	0.8	306
59	L-Histidine Decarboxylase and Tourette's Syndrome. New England Journal of Medicine, 2010, 362, 1901-1908.	13.9	304
60	Geographic Differences in Genetic Susceptibility to IgA Nephropathy: GWAS Replication Study and Geospatial Risk Analysis. PLoS Genetics, 2012, 8, e1002765.	1.5	301
61	IgA nephropathy, the most common cause of glomerulonephritis, is linked to 6q22â€“23. Nature Genetics, 2000, 26, 354-357.	9.4	291
62	Characterization of the mutational landscape of anaplastic thyroid cancer via whole-exome sequencing. Human Molecular Genetics, 2015, 24, 2318-2329.	1.4	290
63	Gitelman's syndrome revisited: An evaluation of symptoms and health-related quality of life. Kidney International, 2001, 59, 710-717.	2.6	279
64	Landscape of somatic single-nucleotide and copy-number mutations in uterine serous carcinoma. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 2916-2921.	3.3	275
65	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. Journal of Clinical Investigation, 2013, 123, 5179-5189.	3.9	275
66	Recurrent gain of function mutation in calcium channel CACNA1H causes early-onset hypertension with primary aldosteronism. ELife, 2015, 4, e06315.	2.8	271
67	Sites of Regulated Phosphorylation that Control K-Cl Cotransporter Activity. Cell, 2009, 138, 525-536.	13.5	269
68	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	5.6	267
69	Mutations in SEC63 cause autosomal dominant polycystic liver disease. Nature Genetics, 2004, 36, 575-577.	9.4	263
70	Genome-wide association study of intracranial aneurysm identifies three new risk loci. Nature Genetics, 2010, 42, 420-425.	9.4	262
71	Hypertension with or without adrenal hyperplasia due to different inherited mutations in the potassium channel KCNJ5. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 2533-2538.	3.3	261
72	A Founder Mutation as a Cause of Cerebral Cavernous Malformation in Hispanic Americans. New England Journal of Medicine, 1996, 334, 946-951.	13.9	257

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73	Skint1, the prototype of a newly identified immunoglobulin superfamily gene cluster, positively selects epidermal Î³Î´ T cells. Nature Genetics, 2008, 40, 656-662.	9.4	257
74	A Novel Form of Human Mendelian Hypertension Featuring Nonglucocorticoid-Remediable Aldosteronism. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3117-3123.	1.8	256
75	The Activity of the Epithelial Sodium Channel Is Regulated by Clathrin-mediated Endocytosis. Journal of Biological Chemistry, 1997, 272, 25537-25541.	1.6	247
76	Susceptibility loci for intracranial aneurysm in European and Japanese populations. Nature Genetics, 2008, 40, 1472-1477.	9.4	247
77	Nonvalidation of Reported Genetic Risk Factors for Acute Coronary Syndrome in a Large-Scale Replication Study. JAMA - Journal of the American Medical Association, 2007, 297, 1551.	3.8	235
78	Aberrant IgA1 Glycosylation Is Inherited in Familial and Sporadic IgA Nephropathy. Journal of the American Society of Nephrology: JASN, 2008, 19, 1008-1014.	3.0	227
79	Rare copy number variations in congenital heart disease patients identify unique genes in left-right patterning. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 2915-2920.	3.3	226
80	Pioneering a Global Cure for Chronic Hepatitis C Virus Infection. Cell, 2016, 167, 12-15.	13.5	222
81	A translocation causing increased Î±-Klotho level results in hypophosphatemic rickets and hyperparathyroidism. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 3455-3460.	3.3	221
82	Angiotensin II signaling increases activity of the renal Na-Cl cotransporter through a WNK4-SPAK-dependent pathway. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 4384-4389.	3.3	215
83	Multilocus linkage of familial hyperkalaemia and hypertension, pseudohypoaldosteronism type II, to chromosomes 1q31-42 and 17p11-q21. Nature Genetics, 1997, 16, 202-205.	9.4	211
84	Recurrent activating mutation in PRKACA in cortisol-producing adrenal tumors. Nature Genetics, 2014, 46, 613-617.	9.4	211
85	Kelch-like 3 and Cullin 3 regulate electrolyte homeostasis via ubiquitination and degradation of WNK4. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 7838-7843.	3.3	209
86	Molecular Physiology of the WNK Kinases. Annual Review of Physiology, 2008, 70, 329-355.	5.6	202
87	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997.	2.6	201
88	Regulation of NKCC2 by a chloride-sensing mechanism involving the WNK3 and SPAK kinases. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 8458-8463.	3.3	199
89	Exome Sequencing Identifies Biallelic MSH3 Germline Mutations as a Recessive Subtype of Colorectal Adenomatous Polyposis. American Journal of Human Genetics, 2016, 99, 337-351.	2.6	198
90	WNK3 modulates transport of Cl- in and out of cells: Implications for control of cell volume and neuronal excitability. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 16783-16788.	3.3	195

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91	Molecular and cellular reorganization of neural circuits in the human lineage. Science, 2017, 358, 1027-1032.	6.0	192
92	CLCN2 chloride channel mutations in familial hyperaldosteronism type II. Nature Genetics, 2018, 50, 349-354.	9.4	188
93	Mutational landscape of uterine and ovarian carcinosarcomas implicates histone genes in epithelialâ€“mesenchymal transition. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 12238-12243.	3.3	181
94	Monogenic causes of chronic kidney disease in adults. Kidney International, 2019, 95, 914-928.	2.6	174
95	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2018, 13, 53-62.	2.2	170
96	Intracranial Aneurysm and Hemorrhagic Stroke in Glucocorticoid-remediable Aldosteronism. Hypertension, 1998, 31, 445-450.	1.3	169
97	Two locus inheritance of non-syndromic midline craniosynostosis via rare SMAD6 and common BMP2 alleles. ELife, 2016, 5, .	2.8	168
98	WNK3 kinase is a positive regulator of NKCC2 and NCC, renal cation-Cl- cotransporters required for normal blood pressure homeostasis. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 16777-16782.	3.3	167
99	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	9.4	164
100	Disruption of Contactin 4 (CNTN4) Results in Developmental Delay and Other Features of 3p Deletion Syndrome. American Journal of Human Genetics, 2004, 74, 1286-1293.	2.6	162
101	Mutations in the Na-Cl Cotransporter Reduce Blood Pressure in Humans. Hypertension, 2001, 37, 1458-1464.	1.3	161
102	WNK4 regulates apical and basolateral Clâ€“flux in extrarenal epithelia. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 2064-2069.	3.3	161
103	Phosphoregulation of the Naâ€“Kâ€“2Cl and Kâ€“Cl cotransporters by the WNK kinases. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 1150-1158.	1.8	161
104	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
105	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. Journal of Clinical Investigation, 2017, 127, 912-928.	3.9	160
106	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. Journal of Clinical Investigation, 2015, 125, 2375-2384.	3.9	159
107	Early and multiple origins of metastatic lineages within primary tumors. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 2140-2145.	3.3	157
108	Genetic landscape of metastatic and recurrent head and neck squamous cell carcinoma. Journal of Clinical Investigation, 2015, 126, 169-180.	3.9	156

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109	Comprehensive Re-Sequencing of Adrenal Aldosterone Producing Lesions Reveal Three Somatic Mutations near the KCNJ5 Potassium Channel Selectivity Filter. PLoS ONE, 2012, 7, e41926.	1.1	154
110	Paracellular Cl- permeability is regulated by WNK4 kinase: Insight into normal physiology and hypertension. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 14877-14882.	3.3	152
111	Mineralocorticoid Receptor Phosphorylation Regulates Ligand Binding and Renal Response to Volume Depletion and Hyperkalemia. Cell Metabolism, 2013, 18, 660-671.	7.2	152
112	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. Nature Genetics, 2016, 48, 457-465.	9.4	149
113	An SGK1 site in WNK4 regulates Na+ channel and K+ channel activity and has implications for aldosterone signaling and K+ homeostasis. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 4025-4029.	3.3	147
114	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.	3.0	147
115	Regression of Chemotherapy-Resistant Polymerase Ïµ (POLE) Ultra-Mutated and MSH6 Hyper-Mutated Endometrial Tumors with Nivolumab. Clinical Cancer Research, 2016, 22, 5682-5687.	3.2	145
116	Recessive loss of function of the neuronal ubiquitin hydrolase UCHL1 leads to early-onset progressive neurodegeneration. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 3489-3494.	3.3	144
117	Isolated polycystic liver disease genes define effectors of polycystin-1 function. Journal of Clinical Investigation, 2017, 127, 1772-1785.	3.9	137
118	KCNJ10 determines the expression of the apical Na-Cl cotransporter (NCC) in the early distal convoluted tubule (DCT1). Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 11864-11869.	3.3	136
119	Identification of Somatic Mutations in Parathyroid Tumors Using Whole-Exome Sequencing. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1774-E1781.	1.8	135
120	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213.	2.6	133
121	Skint-1 is a highly specific, unique selecting component for epidermal T cells. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 3330-3335.	3.3	132
122	Whole-Exome Sequencing Characterizes the Landscape of Somatic Mutations and Copy Number Alterations in Adrenocortical Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E493-E502.	1.8	131
123	WNK1, a kinase mutated in inherited hypertension with hyperkalemia, localizes to diverse Cl--transporting epithelia. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 663-668.	3.3	129
124	The B1-subunit of the H+ ATPase is required for maximal urinary acidification. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 13616-13621.	3.3	126
125	WNK4 regulates activity of the epithelial Na+ channel in vitro and in vivo. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 4020-4024.	3.3	121
126	Frequency and phenotypic spectrum of germline mutations in <scp>POLE</scp> and seven other polymerase genes in 266 patients with colorectal adenomas and carcinomas. International Journal of Cancer, 2015, 137, 320-331.	2.3	121

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127	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	13.9	120
128	Mutations in DSTYK and Dominant Urinary Tract Malformations. New England Journal of Medicine, 2013, 369, 621-629.	13.9	119
129	Autosomal Dominant Pseudohypoaldosteronism Type 1: Mechanisms, Evidence for Neonatal Lethality, and Phenotypic Expression in Adults. Journal of the American Society of Nephrology: JASN, 2006, 17, 1429-1436.	3.0	118
130	A Form of the Metabolic Syndrome Associated with Mutations inDYRK1B. New England Journal of Medicine, 2014, 370, 1909-1919.	13.9	116
131	Novel somatic mutations in primary hyperaldosteronism are related to the clinical, radiological and pathological phenotype. Clinical Endocrinology, 2015, 83, 779-789.	1.2	115
132	Multilineage somatic activating mutations in HRAS and NRAS cause mosaic cutaneous and skeletal lesions, elevated FGF23 and hypophosphatemia. Human Molecular Genetics, 2014, 23, 397-407.	1.4	112
133	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	3.8	112
134	The Centers for Mendelian Genomics: A new largeâ€scale initiative to identify the genes underlying rare Mendelian conditions. American Journal of Medical Genetics, Part A, 2012, 158A, 1523-1525.	0.7	110
135	Whole-Exome Sequencing Enables a Precision Medicine Approach for Kidney Transplant Recipients. Journal of the American Society of Nephrology: JASN, 2019, 30, 201-215.	3.0	110
136	KRIT1, a gene mutated in cerebral cavernous malformation, encodes a microtubule-associated protein. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 10677-10682.	3.3	108
137	The Psychiatric Cell Map Initiative: A Convergent Systems Biological Approach to Illuminating Key Molecular Pathways in Neuropsychiatric Disorders. Cell, 2018, 174, 505-520.	13.5	108
138	WNK3 bypasses the tonicity requirement for K-Cl cotransporter activation via a phosphatase-dependent pathway. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 1976-1981.	3.3	106
139	WNK Protein Kinases Modulate Cellular Clâ^' Flux by Altering the Phosphorylation State of the Na-K-Cl and K-Cl Cotransporters. Physiology, 2006, 21, 326-335.	1.6	105
140	Recessive LAMC3 mutations cause malformations of occipital cortical development. Nature Genetics, 2011, 43, 590-594.	9.4	102
141	Genomic characterization of sarcomatoid transformation in clear cell renal cell carcinoma. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 2170-2175.	3.3	102
142	Common variant near the endothelin receptor type A (EDNRA) gene is associated with intracranial aneurysm risk. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 19707-19712.	3.3	100
143	FAT1 mutations cause a glomerulotubular nephropathy. Nature Communications, 2016, 7, 10822.	5.8	99
144	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. American Journal of Human Genetics, 2015, 96, 81-92.	2.6	98

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145	Localization of a Gene for Autosomal Recessive Distal Renal Tubular Acidosis with Normal Hearing (rdRTA2) to 7q33-34. American Journal of Human Genetics, 1999, 65, 1656-1665.	2.6	96
146	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	9.4	96
147	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239.	3.8	95
148	[14] Isolation of specific RNAs using DNA covalently linked to diazobenzyloxymethyl cellulose or paper. Methods in Enzymology, 1979, 68, 206-220.	0.4	94
149	Molecular Cloning and Characterization of Atp6n1b. Journal of Biological Chemistry, 2001, 276, 42382-42388.	1.6	93
150	Decreased ENaC expression compensates the increased NCC activity following inactivation of the kidney-specific isoform of WNK1 and prevents hypertension. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 18109-18114.	3.3	93
151	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. PLoS Genetics, 2016, 12, e1005963.	1.5	92
152	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. Nature Communications, 2018, 9, 1960.	5.8	90
153	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	3.9	89
154	Syndromic patent ductus arteriosus: Evidence for haploinsufficient TFAP2B mutations and identification of a linked sleep disorder. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 2975-2979.	3.3	85
155	Finding genetic contributions to sporadic disease: A recessive locus at 12q24 commonly contributes to patent ductus arteriosus. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 15054-15059.	3.3	84
156	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	15.2	84
157	CARD14-associated papulosquamous eruption: A spectrum including features of psoriasis and pityriasis rubra pilaris. Journal of the American Academy of Dermatology, 2018, 79, 487-494.	0.6	82
158	The Congenital Heart Disease Genetic Network Study: Cohort description. PLoS ONE, 2018, 13, e0191319.	1.1	82
159	Localization and Regulation of the ATP6V0A4 (a4) Vacuolar H+-ATPase Subunit Defective in an Inherited Form of Distal Renal Tubular Acidosis. Journal of the American Society of Nephrology: JASN, 2003, 14, 3027-3038.	3.0	80
160	Mapping a Mendelian Form of Intracranial Aneurysm to 1p34.3-p36.13. American Journal of Human Genetics, 2005, 76, 172-179.	2.6	80
161	Whole-Exome Sequencing Reveals Somatic Mutations in HRAS and KRAS , which Cause Nevus Sebaceus. Journal of Investigative Dermatology, 2013, 133, 827-830.	0.3	79
162	Evaluation of the Dexamethasone Suppression Test for the Diagnosis of Glucocorticoid-Remediable Aldosteronism1. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 3570-3573.	1.8	78

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163	Angiotensin II signaling via protein kinase C phosphorylates Kelch-like 3, preventing WNK4 degradation. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 15556-15561.	3.3	77
164	Glucocorticoid-remediable aldosteronism (GRA): Diagnosis, variability of phenotype and regulation of potassium homeostasis. Steroids, 1995, 60, 48-51.	0.8	76
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