Richard P Lifton

List of Publications by Citations

318 papers	54,103 citations	109 h-index	230 g-index
334 ext. papers	63,116 ext. citations	16 avg, IF	6.69 L-index

#	Paper	IF	Citations
318	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
317	Molecular basis of human hypertension: role of angiotensinogen. *Cell*, **1992**, 71, 169-80	56.2	1569
316	De novo mutations revealed by whole-exome sequencing are strongly associated with autism. *Nature*, **2012**, 485, 237-41	50.4	1470
315	High bone density due to a mutation in LDL-receptor-related protein 5. *New England Journal of Medicine*, **2002**, 346, 1513-21	59.2	1357
314	Molecular mechanisms of human hypertension. *Cell*, **2001**, 104, 545-56	56.2	1323
313	Human hypertension caused by mutations in WNK kinases. *Science*, **2001**, 293, 1107-12	33.3	1160
312	Liddle's syndrome: heritable human hypertension caused by mutations in the beta subunit of the epithelial sodium channel. *Cell*, **1994**, 79, 407-14	56.2	1093
311	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	1090
310	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
309	A chimaeric 11 beta-hydroxylase/aldosterone synthase gene causes glucocorticoid-remediable aldosteronism and human hypertension. *Nature*, **1992**, 355, 262-5	50.4	992
308	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, 19096-101	11.5	971
307	Gitelman's variant of Bartter's syndrome, inherited hypokalaemic alkalosis, is caused by mutations in the thiazide-sensitive Na-Cl cotransporter. *Nature Genetics*, **1996**, 12, 24-30	36.3	960
306	Paracellin-1, a renal tight junction protein required for paracellular Mg2+ resorption. *Science*, **1999**, 285, 103-6	33.3	941
305	Multiple recurrent de novo CNVs, including duplications of the 7q11.23 Williams syndrome region, are strongly associated with autism. *Neuron*, **2011**, 70, 863-85	13.9	932
304	New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196	50.4	920
303	Exome sequencing identifies recurrent somatic RAC1 mutations in melanoma. *Nature Genetics*, **2012**, 44, 1006-14	36.3	887
302	Sequence variants in SLITRK1 are associated with Tourette's syndrome. *Science*, **2005**, 310, 317-20	33.3	776

301	Bartter's syndrome, hypokalaemic alkalosis with hypercalciuria, is caused by mutations in the Na-K-2Cl cotransporter NKCC2. *Nature Genetics*, **1996**, 13, 183-8	36.3	734
300	Mutations in the chloride channel gene, CLCNKB, cause Bartter's syndrome type III. *Nature Genetics*, **1997**, 17, 171-8	36.3	718
299	K+ channel mutations in adrenal aldosterone-producing adenomas and hereditary hypertension. *Science*, **2011**, 331, 768-72	33.3	707
298	Genetic heterogeneity of Bartter's syndrome revealed by mutations in the K+ channel, ROMK. *Nature Genetics*, **1996**, 14, 152-6	36.3	670
297	Mutations in subunits of the epithelial sodium channel cause salt wasting with hyperkalaemic acidosis, pseudohypoaldosteronism type 1. *Nature Genetics*, **1996**, 12, 248-53	36.3	667
296	Hypertension caused by a truncated epithelial sodium channel gamma subunit: genetic heterogeneity of Liddle syndrome. *Nature Genetics*, **1995**, 11, 76-82	36.3	648
295	Rare independent mutations in renal salt handling genes contribute to blood pressure variation. *Nature Genetics*, **2008**, 40, 592-599	36.3	639
294	De novo mutations in histone-modifying genes in congenital heart disease. *Nature*, **2013**, 498, 220-3	50.4	591
293	Mutations in the gene encoding B1 subunit of H+-ATPase cause renal tubular acidosis with sensorineural deafness. *Nature Genetics*, **1999**, 21, 84-90	36.3	551
292	Activating mineralocorticoid receptor mutation in hypertension exacerbated by pregnancy. *Science*, **2000**, 289, 119-23	33.3	521
291	Genomic analysis of non-NF2 meningiomas reveals mutations in TRAF7, KLF4, AKT1, and SMO. *Science*, **2013**, 339, 1077-80	33.3	508
290	Evidence for a gene influencing blood pressure on chromosome 17. Genome scan linkage results for longitudinal blood pressure phenotypes in subjects from the framingham heart study. *Hypertension*, **2000**, 36, 477-83	8.5	483
289	LRP6 mutation in a family with early coronary disease and metabolic risk factors. *Science*, **2007**, 315, 1278-82	33.3	465
288	Mutations in kelch-like 3 and cullin 3 cause hypertension and electrolyte abnormalities. *Nature*, **2012**, 482, 98-102	50.4	440
287	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics*, **2015**, 97, 199-215	11	432
286	Molecular cytogenetic analysis and resequencing of contactin associated protein-like 2 in autism spectrum disorders. *American Journal of Human Genetics*, **2008**, 82, 165-73	11	427
285	Genome-wide association study identifies susceptibility loci for IgA nephropathy. *Nature Genetics*, **2011**, 43, 321-7	36.3	414
284	Somatic and germline CACNA1D calcium channel mutations in aldosterone-producing adenomas and primary aldosteronism. *Nature Genetics*, **2013**, 45, 1050-4	36.3	410

283	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. *Science*, **2015**, 350, 1262-6	33.3	406
282	Whole-exome sequencing identifies recessive WDR62 mutations in severe brain malformations. *Nature*, **2010**, 467, 207-10	50.4	395
281	A single-gene cause in 29.5% of cases of steroid-resistant nephrotic syndrome. *Journal of the American Society of Nephrology: JASN*, **2015**, 26, 1279-89	12.7	378
280	Recessive mutations in DGKE cause atypical hemolytic-uremic syndrome. *Nature Genetics*, **2013**, 45, 531-6	36.3	357
279	Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SeSAME syndrome) caused by mutations in KCNJ10. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 5842-7	11.5	356
278	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. *Nature Genetics*, **2017**, 49, 1593-1601	36.3	348
277	Absence of linkage between the angiotensin converting enzyme locus and human essential hypertension. *Nature Genetics*, **1992**, 1, 72-5	36.3	343
276	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, 680-4	11.5	342
275	Mutation of NLRC4 causes a syndrome of enterocolitis and autoinflammation. *Nature Genetics*, **2014**, 46, 1135-1139	36.3	337
274	Mutations in the mineralocorticoid receptor gene cause autosomal dominant pseudohypoaldosteronism type I. *Nature Genetics*, **1998**, 19, 279-81	36.3	335
273	Discovery of new risk loci for IgA nephropathy implicates genes involved in immunity against intestinal pathogens. *Nature Genetics*, **2014**, 46, 1187-96	36.3	325
272	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, 71-5	36.3	319
271	WNK4 regulates the balance between renal NaCl reabsorption and K+ secretion. *Nature Genetics*, **2003**, 35, 372-6	36.3	318
270	Wnk4 controls blood pressure and potassium homeostasis via regulation of mass and activity of the distal convoluted tubule. *Nature Genetics*, **2006**, 38, 1124-32	36.3	298
269	Roles of the cation-chloride cotransporters in neurological disease. *Nature Clinical Practice Neurology*, **2008**, 4, 490-503		294
268	Hereditary hypertension caused by chimaeric gene duplications and ectopic expression of aldosterone synthase. *Nature Genetics*, **1992**, 2, 66-74	36.3	288
267	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, 1851-8	5.6	287
266	A cluster of metabolic defects caused by mutation in a mitochondrial tRNA. *Science*, **2004**, 306, 1190-4	33.3	282

265	Exome sequencing links mutations in PARN and RTEL1 with familial pulmonary fibrosis and telomere shortening. *Nature Genetics*, **2015**, 47, 512-7	36.3	279
264	Exome sequencing identifies recurrent mutations in NF1 and RASopathy genes in sun-exposed melanomas. *Nature Genetics*, **2015**, 47, 996-1002	36.3	261
263	IgA nephropathy, the most common cause of glomerulonephritis, is linked to 6q22-23. *Nature Genetics*, **2000**, 26, 354-7	36.3	250
262	Genomic landscape of cutaneous T cell lymphoma. *Nature Genetics*, **2015**, 47, 1011-9	36.3	247
261	L-histidine decarboxylase and Tourette's syndrome. *New England Journal of Medicine*, **2010**, 362, 1901-8	59.2	242
260	The activity of the epithelial sodium channel is regulated by clathrin-mediated endocytosis. *Journal of Biological Chemistry*, **1997**, 272, 25537-41	5.4	235
259	Genome-wide association study of intracranial aneurysm identifies three new risk loci. *Nature Genetics*, **2010**, 42, 420-5	36.3	234
258	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. *Journal of Clinical Investigation*, **2013**, 123, 5179-89	15.9	231
257	Geographic differences in genetic susceptibility to IgA nephropathy: GWAS replication study and geospatial risk analysis. *PLoS Genetics*, **2012**, 8, e1002765	6	231
256	Mutational landscape of MCPyV-positive and MCPyV-negative Merkel cell carcinomas with implications for immunotherapy. *Oncotarget*, **2016**, 7, 3403-15	3.3	229
255	Characterization of the mutational landscape of anaplastic thyroid cancer via whole-exome sequencing. *Human Molecular Genetics*, **2015**, 24, 2318-29	5.6	225
254	Mutations in SEC63 cause autosomal dominant polycystic liver disease. *Nature Genetics*, **2004**, 36, 575-7	36.3	224
253	Gitelman's syndrome revisited: an evaluation of symptoms and health-related quality of life. *Kidney International*, **2001**, 59, 710-7	9.9	224
252	Susceptibility loci for intracranial aneurysm in European and Japanese populations. *Nature Genetics*, **2008**, 40, 1472-7	36.3	222
251	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, 2916-21	11.5	221
250	A founder mutation as a cause of cerebral cavernous malformation in Hispanic Americans. *New England Journal of Medicine*, **1996**, 334, 946-51	59.2	219
249	Skint1, the prototype of a newly identified immunoglobulin superfamily gene cluster, positively selects epidermal gammadelta T cells. *Nature Genetics*, **2008**, 40, 656-62	36.3	216
248	Sites of regulated phosphorylation that control K-Cl cotransporter activity. *Cell*, **2009**, 138, 525-36	56.2	215

247	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, 2533-8	11.5	215
246	A novel form of human mendelian hypertension featuring nonglucocorticoid-remediable aldosteronism. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 3117-23	5.6	211
245	Recurrent gain of function mutation in calcium channel CACNA1H causes early-onset hypertension with primary aldosteronism. *ELife*, **2015**, 4, e06315	8.9	203
244	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, 4384-9	11.5	200
243	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, 1551-61	27.4	199
242	Multilocus linkage of familial hyperkalaemia and hypertension, pseudohypoaldosteronism type II, to chromosomes 1q31-42 and 17p11-q21. *Nature Genetics*, **1997**, 16, 202-5	36.3	197
241	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, 2915-20	11.5	192
240	A translocation causing increased alpha-klotho level results in hypophosphatemic rickets and hyperparathyroidism. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 3455-60	11.5	188
239	Molecular physiology of the WNK kinases. *Annual Review of Physiology*, **2008**, 70, 329-55	23.1	184
238	Aberrant IgA1 glycosylation is inherited in familial and sporadic IgA nephropathy. *Journal of the American Society of Nephrology: JASN*, **2008**, 19, 1008-14	12.7	183
237	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, 16783-8	11.5	178
236	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, 8458-63	11.5	174
235	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, 7838-43	11.5	167
234	Recurrent activating mutation in PRKACA in cortisol-producing adrenal tumors. *Nature Genetics*, **2014**, 46, 613-7	36.3	165
233	Copy-number disorders are a common cause of congenital kidney malformations. *American Journal of Human Genetics*, **2012**, 91, 987-97	11	161
232	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, 16777-82	11.5	156
231	WNK4 regulates apical and basolateral Cl- flux in extrarenal epithelia. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 2064-9	11.5	151
230	Intracranial aneurysm and hemorrhagic stroke in glucocorticoid-remediable aldosteronism. *Hypertension*, **1998**, 31, 445-50	8.5	144

229	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, 14877-82	11.5	143
228	Mutations in the Na-Cl cotransporter reduce blood pressure in humans. *Hypertension*, **2001**, 37, 1458-64	8.5	142
227	Exome Sequencing Identifies Biallelic MSH3 Germline Mutations as a Recessive Subtype of Colorectal Adenomatous Polyposis. *American Journal of Human Genetics*, **2016**, 99, 337-51	11	139
226	Disruption of contactin 4 (CNTN4) results in developmental delay and other features of 3p deletion syndrome. *American Journal of Human Genetics*, **2004**, 74, 1286-93	11	135
225	Phosphoregulation of the Na-K-2Cl and K-Cl cotransporters by the WNK kinases. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2010**, 1802, 1150-8	6.9	134
224	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. *Journal of Clinical Investigation*, **2015**, 125, 2375-84	15.9	133
223	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, 4025-9	11.5	131
222	Comprehensive re-sequencing of adrenal aldosterone producing lesions reveal three somatic mutations near the KCNJ5 potassium channel selectivity filter. *PLoS ONE*, **2012**, 7, e41926	3.7	128
221	Molecular and cellular reorganization of neural circuits in the human lineage. *Science*, **2017**, 358, 1027-1032	33.3	127
220	Mutational landscape of uterine and ovarian carcinosarcomas implicates histone genes in epithelial-mesenchymal transition. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, 12238-12243	11.5	123
219	Mineralocorticoid receptor phosphorylation regulates ligand binding and renal response to volume depletion and hyperkalemia. *Cell Metabolism*, **2013**, 18, 660-71	24.6	122
218	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, 663-8	11.5	120
217	CLCN2 chloride channel mutations in familial hyperaldosteronism type II. *Nature Genetics*, **2018**, 50, 349-354	38.3	117
216	Two locus inheritance of non-syndromic midline craniosynostosis via rare and common alleles. *ELife* , **2016**, 5,	8.9	115
215	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, 13616-21	11.5	113
214	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. *Journal of Clinical Investigation*, **2017**, 127, 912-928	15.9	112
213	Identification of somatic mutations in parathyroid tumors using whole-exome sequencing. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E1774-81	5.6	112
212	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, 3489-94	11.5	111

211	Whole-exome sequencing characterizes the landscape of somatic mutations and copy number alterations in adrenocortical carcinoma. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E493-502	5.6	110
210	Genetic landscape of metastatic and recurrent head and neck squamous cell carcinoma. *Journal of Clinical Investigation*, **2016**, 126, 169-80	15.9	110
209	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. *Nature Genetics*, **2016**, 48, 457-65	36.3	109
208	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, 3330-5	11.5	109
207	Regression of Chemotherapy-Resistant Polymerase (POLE) Ultra-Mutated and MSH6 Hyper-Mutated Endometrial Tumors with Nivolumab. *Clinical Cancer Research*, **2016**, 22, 5682-5687	12.9	109
206	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, 11864-9	11.5	107
205	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, 4020-4	11.5	106
204	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. *Nature Genetics*, **2017**, 49, 1529-1538	36.3	105
203	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, 10677-82	11.5	104
202	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. *Clinical Journal of the American Society of Nephrology: CJASN*, **2018**, 13, 53-62	6.9	103
201	Frequency and phenotypic spectrum of germline mutations in POLE and seven other polymerase genes in 266 patients with colorectal adenomas and carcinomas. *International Journal of Cancer*, **2015**, 137, 320-31	7.5	102
200	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, 1429-36	12.7	101
199	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, 1976-81	11.5	100
198	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
197	WNK protein kinases modulate cellular Cl- flux by altering the phosphorylation state of the Na-K-Cl and K-Cl cotransporters. *Physiology*, **2006**, 21, 326-35	9.8	98
196	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, 2140-5	11.5	95
195	Mutations in DSTYK and dominant urinary tract malformations. *New England Journal of Medicine*, **2013**, 369, 621-9	59.2	95
194	The Centers for Mendelian Genomics: a new large-scale initiative to identify the genes underlying rare Mendelian conditions. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1523-5	2.5	92

193	Monogenic causes of chronic kidney disease in adults. *Kidney International*, **2019**, 95, 914-928	9.9	92
192	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,	28	91
191	Novel somatic mutations in primary hyperaldosteronism are related to the clinical, radiological and pathological phenotype. *Clinical Endocrinology*, **2015**, 83, 779-89	3.4	88
190	A form of the metabolic syndrome associated with mutations in DYRK1B. *New England Journal of Medicine*, **2014**, 370, 1909-1919	59.2	86
189	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	5.6	85
188	Recessive LAMC3 mutations cause malformations of occipital cortical development. *Nature Genetics*, **2011**, 43, 590-4	36.3	85
187	Molecular cloning and characterization of Atp6n1b: a novel fourth murine vacuolar H+-ATPase a-subunit gene. *Journal of Biological Chemistry*, **2001**, 276, 42382-8	5.4	84
186	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. *New England Journal of Medicine*, **2017**, 376, 742-754	59.2	83
185	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, 2170-5	11.5	83
184	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, 19707-12	11.5	82
183	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, 1656-65	11	80
182	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, 18109-14	11.5	79
181	Isolation of specific RNAs using DNA covalently linked to diazobenzyloxymethyl cellulose or paper. *Methods in Enzymology*, **1979**, 68, 206-20	1.7	78
180	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and nephrocalcinosis. *Kidney International*, **2018**, 93, 204-213	9.9	77
179	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	12.7	75
178	Isolated polycystic liver disease genes define effectors of polycystin-1 function. *Journal of Clinical Investigation*, **2017**, 127, 1772-1785	15.9	74
177	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, 15054-9	11.5	73
176	Evaluation of the dexamethasone suppression test for the diagnosis of glucocorticoid-remediable aldosteronism. *Journal of Clinical Endocrinology and Metabolism*, **1997**, 82, 3570-3	5.6	70

175	FAT1 mutations cause a glomerulotubular nephropathy. *Nature Communications*, **2016**, 7, 10822	17.4	69
174	The Psychiatric Cell Map Initiative: A Convergent Systems Biological Approach to Illuminating Key Molecular Pathways in Neuropsychiatric Disorders. *Cell*, **2018**, 174, 505-520	56.2	69
173	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, 3027-38	12.7	69
172	Mapping a Mendelian form of intracranial aneurysm to 1p34.3-p36.13. *American Journal of Human Genetics*, **2005**, 76, 172-9	11	68
171	Glucocorticoid-remediable aldosteronism (GRA): diagnosis, variability of phenotype and regulation of potassium homeostasis. *Steroids*, **1995**, 60, 48-51	2.8	68
170	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. *Nature Communications*, **2018**, 9, 1960	17.4	68
169	Mutations in KATNB1 cause complex cerebral malformations by disrupting asymmetrically dividing neural progenitors. *Neuron*, **2014**, 84, 1226-39	13.9	67
168	WNK2 kinase is a novel regulator of essential neuronal cation-chloride cotransporters. *Journal of Biological Chemistry*, **2011**, 286, 30171-80	5.4	67
167	Epigenetic abnormalities associated with a chromosome 18(q21-q22) inversion and a Gilles de la Tourette syndrome phenotype. *Proceedings of the National Academy of Sciences of the United States of America*, **2003**, 100, 4684-9	11.5	67
166	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. *PLoS Genetics*, **2016**, 12, e1005963	6	67
165	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. *Science Immunology*, **2021**, 6,	28	67
164	DCDC2 mutations cause a renal-hepatic ciliopathy by disrupting Wnt signaling. *American Journal of Human Genetics*, **2015**, 96, 81-92	11	66
163	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	9.9	60
162	Regulation of diverse ion transport pathways by WNK4 kinase: a novel molecular switch. *Trends in Endocrinology and Metabolism*, **2005**, 16, 98-103	8.8	60
161	Mutations in TBX18 Cause Dominant Urinary Tract Malformations via Transcriptional Dysregulation of Ureter Development. *American Journal of Human Genetics*, **2015**, 97, 291-301	11	59
160	Comprehensive DNA methylation analysis of benign and malignant adrenocortical tumors. *Genes Chromosomes and Cancer*, **2012**, 51, 949-60	5	59
159	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, 2975-9	11.5	59
158	Whole-exome sequencing reveals somatic mutations in HRAS and KRAS, which cause nevus sebaceus. *Journal of Investigative Dermatology*, **2013**, 133, 827-830	4.3	58

157	Low density lipoprotein (LDL) receptor-related protein 6 (LRP6) regulates body fat and glucose homeostasis by modulating nutrient sensing pathways and mitochondrial energy expenditure. *Journal of Biological Chemistry*, **2012**, 287, 7213-23	5.4	58
156	WNK kinases: molecular regulators of integrated epithelial ion transport. *Current Opinion in Nephrology and Hypertension*, **2004**, 13, 557-62	3.5	57
155	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, 15556-61	11.5	56
154	Wild-type LRP6 inhibits, whereas atherosclerosis-linked LRP6R611C increases PDGF-dependent vascular smooth muscle cell proliferation. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 1914-8	11.5	55
153	KRIT1/cerebral cavernous malformation 1 protein localizes to vascular endothelium, astrocytes, and pyramidal cells of the adult human cerebral cortex. *Neurosurgery*, **2004**, 54, 943-9; discussion 949	3.2	55
152	Glucocorticoid-remediable aldosteronism. *Journal of Clinical Endocrinology and Metabolism*, **1999**, 84, 4341-4	5.6	55
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