Richard P Lifton

List of Publications by Year in Descending Order

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	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity.. *Circulation Genomic and Precision Medicine*, **2022**, CIRCGEN121003500	5.2	0
317	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
316	Genetic Influence on Neurodevelopment in Nonsyndromic Craniosynostosis.. *Plastic and Reconstructive Surgery*, **2022**,	2.7	3
315	Whole Exome Sequencing Reveals Damaging Gene Variants Associated with Hypoalphalipoproteinemia.. *Journal of Lipid Research*, **2022**, 100209	6.3	
314	Mutation spectrum of congenital heart disease in a consanguineous Turkish population.. *Molecular Genetics & Genomic Medicine*, **2022**, e1944	2.3	0
313	Integrated mutational landscape analysis of uterine leiomyosarcomas. *Proceedings of the National Academy of Sciences of the United States of America*, **2021**, 118,	11.5	11
312	Genetic Defects in Underlie Male Infertility With Multiple Morphological Abnormalities of the Sperm Flagella in Humans and Mice. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 662903	5.7	2
311	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. *Circulation Research* , **2021**, 128, 1156-1169	15.7	2
310	Enhanced Ca signaling, mild primary aldosteronism, and hypertension in a familial hyperaldosteronism mouse model ( ). *Proceedings of the National Academy of Sciences of the United States of America*, **2021**, 118,	11.5	4
309	Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child. *Nature Medicine*, **2021** , 27, 1646-1654	50.5	17
308	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. *JAMA Cardiology*, **2021**, 6, 457-462	16.2	12
307	Recessive Mutations in as a Candidate of Monogenic Nephrotic Syndrome. *Kidney International Reports*, **2021**, 6, 472-483	4.1	4
306	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. *Kidney International Reports*, **2021**, 6, 460-471	4.1	2
305	Recessive variants impair actin remodeling and cause glomerulopathy in humans and mice. *Science Advances*, **2021**, 7,	14.3	6
304	Mutations in Are a Novel Cause of Galloway-Mowat Syndrome. *Journal of the American Society of Nephrology: JASN*, **2021**, 32, 580-596	12.7	3
303	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	11	1
302	Exome survey of individuals affected by VATER/VACTERL with renal phenotypes identifies phenocopies and novel candidate genes. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3784-3792	2.5	1

301	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
300	DIAPH1 Variants in Non-East Asian Patients With Sporadic Moyamoya Disease. *JAMA Neurology*, **2021**, 78, 993-1003	17.2	7
299	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. *Science Immunology*, **2021**, 6,	28	67
298	Systems Analysis Implicates WAVE2 Complex in the Pathogenesis of Developmental Left-Sided Obstructive Heart Defects. *JACC Basic To Translational Science*, **2020**, 5, 376-386	8.7	2
297	Mutations in ASPRV1 Cause Dominantly Inherited Ichthyosis. *American Journal of Human Genetics*, **2020**, 107, 158-163	11	5
296	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. *Genome Medicine*, **2020**, 12, 42	14.4	8
295	Late-Onset Bartter Syndrome Type II Due to a Homozygous Mutation in KCNJ1 Gene: A Case Report and Literature Review. *American Journal of Case Reports*, **2020**, 21, e924527	1.3	5
294	Whole exome sequencing identified ATP6V1C2 as a novel candidate gene for recessive distal renal tubular acidosis. *Kidney International*, **2020**, 97, 567-579	9.9	26
293	Organic Solute Transporter Alpha Deficiency: A Disorder With Cholestasis, Liver Fibrosis, and Congenital Diarrhea. *Hepatology*, **2020**, 71, 1879-1882	11.2	10
292	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. *Nature Medicine*, **2020**, 26, 1754-1765	50.5	23
291	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. *Nature Genetics*, **2020**, 52, 1046-1056	30.9	38
290	Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia. *IScience*, **2020**, 23, 101552	6.1	10
289	DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. *American Journal of Human Genetics*, **2020**, 107, 1113-1128	11	5
288	A genome-wide case-only test for the detection of digenic inheritance in human exomes. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 19367-19375	11.5	5
287	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
286	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	1090
285	Recessive Mutations in AP1B1 Cause Ichthyosis, Deafness, and Photophobia. *American Journal of Human Genetics*, **2019**, 105, 1023-1029	11	12
284	COL4A1 mutations as a potential novel cause of autosomal dominant CAKUT in humans. *Human Genetics*, **2019**, 138, 1105-1115	6.3	8

283	Histone H2B monoubiquitination regulates heart development via epigenetic control of cilia motility. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 14049-14054	11.5	17
282	Calcineurin dephosphorylates Kelch-like 3, reversing phosphorylation by angiotensin II and regulating renal electrolyte handling. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 3155-3160	11.5	26
281	Identification of a dominant MYH11 causal variant in chronic intestinal pseudo-obstruction: Results of whole-exome sequencing. *Clinical Genetics*, **2019**, 96, 473-477	4	13
280	Contributions of Rare Gene Variants to Familial and Sporadic FSGS. *Journal of the American Society of Nephrology: JASN*, **2019**, 30, 1625-1640	12.7	22
279	CELA2A mutations predispose to early-onset atherosclerosis and metabolic syndrome and affect plasma insulin and platelet activation. *Nature Genetics*, **2019**, 51, 1233-1243	36.3	11
278	Whole-exome sequencing of cervical carcinomas identifies activating ERBB2 and PIK3CA mutations as targets for combination therapy. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 22730-22736	11.5	26
277	Developmentally regulated KCC2 phosphorylation is essential for dynamic GABA-mediated inhibition and survival. *Science Signaling*, **2019**, 12,	8.8	31
276	Recessive Inheritance of Congenital Hydrocephalus With Other Structural Brain Abnormalities Caused by Compound Heterozygous Mutations in. *Frontiers in Cellular Neuroscience*, **2019**, 13, 425	6.1	7
275	Monogenic causes of chronic kidney disease in adults. *Kidney International*, **2019**, 95, 914-928	9.9	92
274	Mutations in and previously unimplicated genes of the BMP, Wnt, and Hedgehog pathways in syndromic craniosynostosis. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 15116-15121	11.5	7
273	Mutations Implicate RAB11-Dependent Vesicular Trafficking in the Pathogenesis of Nephrotic Syndrome. *Journal of the American Society of Nephrology: JASN*, **2019**, 30, 2338-2353	12.7	11
272	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. *American Journal of Human Genetics*, **2019**, 105, 1286-1293	11	8
271	Mutational landscape of primary, metastatic, and recurrent ovarian cancer reveals c-MYC gains as potential target for BET inhibitors. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 619-624	11.5	28
270	Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. *Neuron* , **2019**, 101, 429-443.e4	13.9	33
269	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. *American Journal of Human Genetics*, **2019**, 104, 45-54	11	15
268	Mutations in PERP Cause Dominant and Recessive Keratoderma. *Journal of Investigative Dermatology*, **2019**, 139, 380-390	4.3	9
267	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
266	Whole-Exome Sequencing Enables a Precision Medicine Approach for Kidney Transplant Recipients. *Journal of the American Society of Nephrology: JASN*, **2019**, 30, 201-215	12.7	54

265	Hypokalemia Associated With a Claudin 10 Mutation: A Case Report. *American Journal of Kidney Diseases*, **2019**, 73, 425-428	7.4	12
264	Genetic variants in the LAMA5 gene in pediatric nephrotic syndrome. *Nephrology Dialysis Transplantation*, **2019**, 34, 485-493	4.3	17
263	Whole Exome Sequencing Reveals a Monogenic Cause of Disease in 43% of 35 Families With Midaortic Syndrome. *Hypertension*, **2018**, 71, 691-699	8.5	12
262	CLCN2 chloride channel mutations in familial hyperaldosteronism type II. *Nature Genetics*, **2018**, 50, 349-354	36.3	117
261	Robust identification of mosaic variants in congenital heart disease. *Human Genetics*, **2018**, 137, 183-193	6.3	26
260	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	4.5	43
259	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and nephrocalcinosis. *Kidney International*, **2018**, 93, 204-213	9.9	77
258	and Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 2123-2138	12.7	21
257	Co-occurrence of frameshift mutations in and in a child with complex craniosynostosis. *Human Genome Variation*, **2018**, 5, 14	1.8	11
256	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
255	Comprehensive Genetic Analysis of Follicular Thyroid Carcinoma Predicts Prognosis Independent of Histology. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 2640-2650	5.6	41
254	ULK1 Phosphorylates and Regulates Mineralocorticoid Receptor. *Cell Reports*, **2018**, 24, 569-576	10.6	15
253	Clonal evolution analysis of paired anaplastic and well-differentiated thyroid carcinomas reveals shared common ancestor. *Genes Chromosomes and Cancer*, **2018**, 57, 645-652	5	17
252	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
251	The Congenital Heart Disease Genetic Network Study: Cohort description. *PLoS ONE*, **2018**, 13, e0191319	3.7	43
250	A homozygous missense variant in VWA2, encoding an interactor of the Fraser-complex, in a patient with vesicoureteral reflux. *PLoS ONE*, **2018**, 13, e0191224	3.7	3
249	C-terminally truncated, kidney-specific variants of the WNK4 kinase lack several sites that regulate its activity. *Journal of Biological Chemistry*, **2018**, 293, 12209-12221	5.4	6
248	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

247	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. *Journal of Clinical Investigation*, **2018**, 128, 4313-4328	15.9	49
246	Human Genetics and Molecular Mechanisms of Congenital Hydrocephalus. *World Neurosurgery*, **2018**, 119, 441-443	2.1	10
245	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. *Nature Communications*, **2018**, 9, 1960	17.4	68
244	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. *Neuron*, **2018**, 99, 302-314.e4	13.9	53
243	Phosphorylation by PKC and PKA regulate the kinase activity and downstream signaling of WNK4. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E879-E886	11.5	28
242	Application of Whole Exome Sequencing in the Clinical Diagnosis and Management of Inherited Cardiovascular Diseases in Adults. *Circulation: Cardiovascular Genetics*, **2017**, 10,		35
241	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. *New England Journal of Medicine*, **2017**, 376, 742-754	59.2	83
240	Mutations in KDSR Cause Recessive Progressive Symmetric Erythrokeratoderma. *American Journal of Human Genetics*, **2017**, 100, 978-984	11	41
239	A Dominant Mutation in Nuclear Receptor Interacting Protein 1 Causes Urinary Tract Malformations Dysregulation of Retinoic Acid Signaling. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 2364-2376	12.7	31
238	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. *American Journal of Human Genetics*, **2017**, 101, 789-802	11	36
237	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. *Nature Genetics*, **2017**, 49, 1593-1601	36.3	348
236	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. *Journal of Clinical Investigation*, **2017**, 127, 912-928	15.9	112
235	Isolated polycystic liver disease genes define effectors of polycystin-1 function. *Journal of Clinical Investigation*, **2017**, 127, 1772-1785	15.9	74
234	De novo mutations in inhibitors of Wnt, BMP, and Ras/ERK signaling pathways in non-syndromic midline craniosynostosis. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E7341-E7347	11.5	50
233	Exome sequencing in Jewish and Arab patients with rhabdomyolysis reveals single-gene etiology in 43% of cases. *Pediatric Nephrology*, **2017**, 32, 2273-2282	3.2	20
232	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. *Nature Genetics*, **2017**, 49, 1529-1538	36.3	105
231	Molecular and cellular reorganization of neural circuits in the human lineage. *Science*, **2017**, 358, 1027-1032	33.3	127
230	A Genome-Wide Association Study to Identify Single-Nucleotide Polymorphisms for Acute Kidney Injury. *American Journal of Respiratory and Critical Care Medicine*, **2017**, 195, 482-490	10.2	22

229	Macrolides selectively inhibit mutant KCNJ5 potassium channels that cause aldosterone-producing adenoma. *Journal of Clinical Investigation*, **2017**, 127, 2739-2750	15.9	46
228	Advillin acts upstream of phospholipase C ?1 in steroid-resistant nephrotic syndrome. *Journal of Clinical Investigation*, **2017**, 127, 4257-4269	15.9	25
227	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
226	Pioneering a Global Cure for Chronic Hepatitis C Virus Infection. *Cell*, **2016**, 167, 12-15	56.2	8
225	ACOX2 deficiency: A disorder of bile acid synthesis with transaminase elevation, liver fibrosis, ataxia, and cognitive impairment. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, 11289-11293	11.5	53
224	FAT1 mutations cause a glomerulotubular nephropathy. *Nature Communications*, **2016**, 7, 10822	17.4	69
223	Loss of RNA expression and allele-specific expression associated with congenital heart disease. *Nature Communications*, **2016**, 7, 12824	17.4	33
222	Absence of KMT2D/MLL2 mutations in abdominal paraganglioma. *Clinical Endocrinology*, **2016**, 84, 632-4	3.4	3
221	Exome sequencing identifies potential novel candidate genes in patients with unexplained colorectal adenomatous polyposis. *Familial Cancer*, **2016**, 15, 281-8	3	36
220	Low-level APC mutational mosaicism is the underlying cause in a substantial fraction of unexplained colorectal adenomatous polyposis cases. *Journal of Medical Genetics*, **2016**, 53, 172-9	5.8	41
219	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. *Nature Genetics*, **2016**, 48, 457-65	36.3	109
218	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
217	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
216	Genetic landscape of metastatic and recurrent head and neck squamous cell carcinoma. *Journal of Clinical Investigation*, **2016**, 126, 169-80	15.9	110
215	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. *PLoS Genetics*, **2016**, 12, e1005963	6	67
214	Predictors of Chemosensitivity in Triple Negative Breast Cancer: An Integrated Genomic Analysis. *PLoS Medicine*, **2016**, 13, e1002193	11.6	48
213	Two locus inheritance of non-syndromic midline craniosynostosis via rare and common alleles. *ELife* , **2016**, 5,	8.9	115
212	Genomic characterization of sarcomatoid transformation in clear cell renal cell carcinoma.. *Journal of Clinical Oncology*, **2016**, 34, 509-509	2.2	

211	Mutational landscape of uterine and ovarian carcinosarcomas.. *Journal of Clinical Oncology*, **2016**, 34, 5589-5589	2.2	
210	Mutational landscape of MCPyV-positive and MCPyV-negative Merkel cell carcinomas with implications for immunotherapy. *Oncotarget*, **2016**, 7, 3403-15	3.3	229
209	Shifting patterns of genomic variation in the somatic evolution of papillary thyroid carcinoma. *BMC Cancer*, **2016**, 16, 646	4.8	5
208	Recurrent recessive mutation in deoxyguanosine kinase causes idiopathic noncirrhotic portal hypertension. *Hepatology*, **2016**, 63, 1977-86	11.2	25
207	Potassium depletion stimulates Na-Cl cotransporter phosphorylation and inactivation of the ubiquitin ligase Kelch-like 3. *Biochemical and Biophysical Research Communications*, **2016**, 480, 745-751	3.4	23
206	Digenic mutations of human paralogs in Dent disease type 2 associated with Chiari I malformation. *Human Genome Variation*, **2016**, 3, 16042	1.8	7
205	Mutations in the Histone Modifier PRDM6 Are Associated with Isolated Nonsyndromic Patent Ductus Arteriosus. *American Journal of Human Genetics*, **2016**, 98, 1082-1091	11	15
204	Identification of a gain-of-function STAT3 mutation (p.Y640F) in lymphocytic variant hypereosinophilic syndrome. *Blood*, **2016**, 127, 948-51	2.2	42
203	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
202	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
201	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
200	DCDC2 mutations cause a renal-hepatic ciliopathy by disrupting Wnt signaling. *American Journal of Human Genetics*, **2015**, 96, 81-92	11	66
199	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
198	Exome sequencing identifies recurrent mutations in NF1 and RASopathy genes in sun-exposed melanomas. *Nature Genetics*, **2015**, 47, 996-1002	36.3	261
197	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics*, **2015**, 97, 199-215	11	432
196	Genomic landscape of cutaneous T cell lymphoma. *Nature Genetics*, **2015**, 47, 1011-9	36.3	247
195	Src-family protein tyrosine kinase phosphorylates WNK4 and modulates its inhibitory effect on KCNJ1 (ROMK). *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 4495-500	11.5	14
194	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

193	Neomorphic effects of recurrent somatic mutations in Yin Yang 1 in insulin-producing adenomas. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 4062-7	11.5	48
192	Exome sequencing links mutations in PARN and RTEL1 with familial pulmonary fibrosis and telomere shortening. *Nature Genetics*, **2015**, 47, 512-7	36.3	279
191	Genes and environment in neonatal intraventricular hemorrhage. *Seminars in Perinatology*, **2015**, 39, 592-603	3.3	28
190	Mutations of the SLIT2-ROBO2 pathway genes SLIT2 and SRGAP1 confer risk for congenital anomalies of the kidney and urinary tract. *Human Genetics*, **2015**, 134, 905-16	6.3	48
189	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. *Journal of Medical Genetics*, **2015**, 52, 657-65	5.8	27
188	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
187	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
186	Whole-exome sequencing defines the mutational landscape of pheochromocytoma and identifies KMT2D as a recurrently mutated gene. *Genes Chromosomes and Cancer*, **2015**, 54, 542-54	5	50
185	Novel somatic mutations in primary hyperaldosteronism are related to the clinical, radiological and pathological phenotype. *Clinical Endocrinology*, **2015**, 83, 779-89	3.4	88
184	Recurrent gain of function mutation in calcium channel CACNA1H causes early-onset hypertension with primary aldosteronism. *ELife*, **2015**, 4, e06315	8.9	203
183	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. *Science*, **2015**, 350, 1262-6	33.3	406
182	New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196	50.4	920
181	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
180	Dominant De Novo Mutations in GJA1 Cause Erythrokeratodermia Variabilis et Progressiva, without Features of Oculodentodigital Dysplasia. *Journal of Investigative Dermatology*, **2015**, 135, 1540-1547	4.3	36
179	Characterization of the mutational landscape of anaplastic thyroid cancer via whole-exome sequencing. *Human Molecular Genetics*, **2015**, 24, 2318-29	5.6	225
178	Frequent somatic reversion of KRT1 mutations in ichthyosis with confetti. *Journal of Clinical Investigation*, **2015**, 125, 1703-7	15.9	45
177	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. *Journal of Clinical Investigation*, **2015**, 125, 2375-84	15.9	133
176	Recurrent activating mutation in PRKACA in cortisol-producing adrenal tumors. *Nature Genetics*, **2014**, 46, 613-7	36.3	165

175	A form of the metabolic syndrome associated with mutations in DYRK1B. *New England Journal of Medicine*, **2014**, 370, 1909-1919	59.2	86
174	Phenotypic expansion of DGKE-associated diseases. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 1408-14	12.7	50
173	Mutation of NLRC4 causes a syndrome of enterocolitis and autoinflammation. *Nature Genetics*, **2014**, 46, 1135-1139	36.3	337
172	Individual exome analysis in diagnosis and management of paediatric liver failure of indeterminate aetiology. *Journal of Hepatology*, **2014**, 61, 1056-63	13.4	40
171	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
170	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
169	Rare deleterious mutations of the gene EFR3A in autism spectrum disorders. *Molecular Autism*, **2014**, 5, 31	6.5	11
168	Maternal race, demography, and health care disparities impact risk for intraventricular hemorrhage in preterm neonates. *Journal of Pediatrics*, **2014**, 164, 1005-1011.e3	3.6	38
167	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
166	Gene-environment interactions in severe intraventricular hemorrhage of preterm neonates. *Pediatric Research*, **2014**, 75, 241-50	3.2	42
165	Somatic HRAS p.G12S mutation causes woolly hair and epidermal nevi. *Journal of Investigative Dermatology*, **2014**, 134, 1149-1152	4.3	26
164	Mutations in KATNB1 cause complex cerebral malformations by disrupting asymmetrically dividing neural progenitors. *Neuron*, **2014**, 84, 1226-39	13.9	67
163	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
162	Somatic and germline CACNA1D calcium channel mutations in aldosterone-producing adenomas and primary aldosteronism. *Nature Genetics*, **2013**, 45, 1050-4	36.3	410
161	Genomic analysis of non-NF2 meningiomas reveals mutations in TRAF7, KLF4, AKT1, and SMO. *Science*, **2013**, 339, 1077-80	33.3	508
160	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
159	Candidate gene analysis: severe intraventricular hemorrhage in inborn preterm neonates. *Journal of Pediatrics*, **2013**, 163, 1503-6.e1	3.6	13
158	Mineralocorticoid receptor phosphorylation regulates ligand binding and renal response to volume depletion and hyperkalemia. *Cell Metabolism*, **2013**, 18, 660-71	24.6	122

157	Inherited Disorders of Renal Salt Homeostasis **2013**, 1213-1240		6
156	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
155	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
154	Recessive mutations in DGKE cause atypical hemolytic-uremic syndrome. *Nature Genetics*, **2013**, 45, 531-6	36.3	357
153	De novo mutations in histone-modifying genes in congenital heart disease. *Nature*, **2013**, 498, 220-3	50.4	591
152	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
151	Mutations in DSTYK and dominant urinary tract malformations. *New England Journal of Medicine*, **2013**, 369, 621-9	59.2	95
150	New insights into aldosterone-producing adenomas and hereditary aldosteronism: mutations in the K+ channel KCNJ5. *Current Opinion in Nephrology and Hypertension*, **2013**, 22, 141-7	3.5	27
149	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. *Journal of Clinical Investigation*, **2013**, 123, 5179-89	15.9	231
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