Nicholas Katsanis

List of Publications by Year
in descending order

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286	36,169	2802	4014
papers	citations	94 h-index	176 g-index

330	330	330	37523
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A syndrome of altered cardiovascular, craniofacial, neurocognitive and skeletal development caused by mutations in TGFBR1 or TGFBR2. Nature Genetics, 2005, 37, 275-281.	21.4	1,543
2	Ciliopathies. New England Journal of Medicine, 2011, 364, 1533-1543.	27.0	1,227
3	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nature Genetics, 2016, 48, 134-143.	21.4	1,167
4	The Ciliopathies: An Emerging Class of Human Genetic Disorders. Annual Review of Genomics and Human Genetics, 2006, 7, 125-148.	6.2	996
5	Gene expression elucidates functional impact of polygenic risk for schizophrenia. Nature Neuroscience, 2016, 19, 1442-1453.	14.8	952
6	Comparative Genomics Identifies a Flagellar and Basal Body Proteome that Includes the BBS5 Human Disease Gene. Cell, 2004, 117, 541-552.	28.9	721
7	Seven new loci associated with age-related macular degeneration. Nature Genetics, 2013, 45, 433-439.	21.4	687
8	The Vertebrate Primary Cilium in Development, Homeostasis, and Disease. Cell, 2009, 137, 32-45.	28.9	653
9	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
10	Basal body dysfunction is a likely cause of pleiotropic Bardetâ€“Biedl syndrome. Nature, 2003, 425, 628-633.	27.8	607
11	A transition zone complex regulates mammalian ciliogenesis and ciliary membrane composition. Nature Genetics, 2011, 43, 776-784.	21.4	556
12	Disruption of Bardet-Biedl syndrome ciliary proteins perturbs planar cell polarity in vertebrates. Nature Genetics, 2005, 37, 1135-1140.	21.4	536
13	Genetic variants near TIMP3 and high-density lipoproteinâ€“associated loci influence susceptibility to age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7401-7406.	7.1	475
14	Genome-wide association study of advanced age-related macular degeneration identifies a role of the hepatic lipase gene (LIPC). Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7395-7400.	7.1	406
15	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights. Nature Genetics, 2018, 50, 538-548.	21.4	406
16	The Bardet-Biedl protein BBS4 targets cargo to the pericentriolar region and is required for microtubule anchoring and cell cycle progression. Nature Genetics, 2004, 36, 462-470.	21.4	372
17	Hypomorphic mutations in syndromic encephalocele genes are associated with Bardet-Biedl syndrome. Nature Genetics, 2008, 40, 443-448.	21.4	367
18	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. Nature, 2012, 485, 363-367.	27.8	363

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19	Disruption of the basal body compromises proteasomal function and perturbs intracellular Wnt response. Nature Genetics, 2007, 39, 1350-1360.	21.4	361
20	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
21	Molecular genetic testing and the future of clinical genomics. Nature Reviews Genetics, 2013, 14, 415-426.	16.3	334
22	Loss of BBS proteins causes anosmia in humans and defects in olfactory cilia structure and function in the mouse. Nature Genetics, 2004, 36, 994-998.	21.4	329
23	Mechanistic insights into Bardet-Biedl syndrome, a model ciliopathy. Journal of Clinical Investigation, 2009, 119, 428-437.	8.2	328
24	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
25	Beyond Mendel: an evolving view of human genetic disease transmission. Nature Reviews Genetics, 2002, 3, 779-789.	16.3	325
26	Loss of C. elegans BBS-7 and BBS-8 protein function results in cilia defects and compromised intraflagellar transport. Genes and Development, 2004, 18, 1630-1642.	5.9	318
27	Mutations in a member of the Ras superfamily of small GTP-binding proteins causes Bardet-Biedl syndrome. Nature Genetics, 2004, 36, 989-993.	21.4	313
28	Mutations in MKKS cause obesity, retinal dystrophy and renal malformations associated with Bardet-Biedl syndrome. Nature Genetics, 2000, 26, 67-70.	21.4	311
29	Rare variants in CFI, C3 and C9 are associated with high risk of advanced age-related macular degeneration. Nature Genetics, 2013, 45, 1366-1370.	21.4	311
30	Planar Cell Polarity Acts Through Septins to Control Collective Cell Movement and Ciliogenesis. Science, 2010, 329, 1337-1340.	12.6	309
31	CCDC39 is required for assembly of inner dynein arms and the dynein regulatory complex and for normal ciliary motility in humans and dogs. Nature Genetics, 2011, 43, 72-78.	21.4	302
32	CHD8 regulates neurodevelopmental pathways associated with autism spectrum disorder in neural progenitors. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4468-77.	7.1	297
33	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. Nature Genetics, 2010, 42, 840-850.	21.4	295
34	A rare penetrant mutation in CFH confers high risk of age-related macular degeneration. Nature Genetics, 2011, 43, 1232-1236.	21.4	291
35	The ciliary proteome database: an integrated community resource for the genetic and functional dissection of cilia. Nature Genetics, 2006, 38, 961-962.	21.4	265
36	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261

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37	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. Nature Genetics, 2006, 38, 521-524.	21.4	259
38	Dissection of epistasis in oligogenic Bardetâ€“Biedl syndrome. Nature, 2006, 439, 326-330.	27.8	255
39	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	21.4	255
40	Clinical and genetic epidemiology of Bardet-Biedl syndrome in Newfoundland: A 22-year prospective, population-based, cohort study. American Journal of Medical Genetics, Part A, 2005, 132A, 352-360.	1.2	249
41	Genetic Interaction of BBS1 Mutations with Alleles at Other BBS Loci Can Result in Non-Mendelian Bardet-Biedl Syndrome. American Journal of Human Genetics, 2003, 72, 1187-1199.	6.2	246
42	The Meckelâ€“Gruber Syndrome proteins MKS1 and meckelin interact and are required for primary cilium formation. Human Molecular Genetics, 2007, 16, 173-186.	2.9	245
43	Common variants near FRK/COL10A1 and VEGFA are associated with advanced age-related macular degeneration. Human Molecular Genetics, 2011, 20, 3699-3709.	2.9	232
44	Nde1-mediated inhibition of ciliogenesis affects cell cycle re-entry. Nature Cell Biology, 2011, 13, 351-360.	10.3	230
45	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
46	Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. Nature Genetics, 2012, 44, 450-455.	21.4	226
47	The centrosome in human genetic disease. Nature Reviews Genetics, 2005, 6, 194-205.	16.3	225
48	Identification of a Novel BBS Gene (BBS12) Highlights the Major Role of a Vertebrate-Specific Branch of Chaperonin-Related Proteins in Bardet-Biedl Syndrome. American Journal of Human Genetics, 2007, 80, 1-11.	6.2	219
49	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	6.4	211
50	Toll-like Receptor 3 and Geographic Atrophy in Age-Related Macular Degeneration. New England Journal of Medicine, 2008, 359, 1456-1463.	27.0	209
51	Ataxia, Dementia, and Hypogonadotropism Caused by Disordered Ubiquitination. New England Journal of Medicine, 2013, 368, 1992-2003.	27.0	208
52	Identification of a Novel Bardet-Biedl Syndrome Protein, BBS7, That Shares Structural Features with BBS1 and BBS2. American Journal of Human Genetics, 2003, 72, 650-658.	6.2	207
53	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
54	Activating mutations in STIM1 and ORAI1 cause overlapping syndromes of tubular myopathy and congenital miosis. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 4197-4202.	7.1	205

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55	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	21.4	203
56	CC2D2A Is Mutated in Joubert Syndrome and Interacts with the Ciliopathy-Associated Basal Body Protein CEP290. American Journal of Human Genetics, 2008, 83, 559-571.	6.2	202
57	The oligogenic properties of Bardet-Biedl syndrome. Human Molecular Genetics, 2004, 13, 65R-71.	2.9	197
58	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
59	Heterozygous mutations in BBS1, BBS2 and BBS6 have a potential epistatic effect on Bardet-Biedl patients with two mutations at a second BBS locus. Human Molecular Genetics, 2003, 12, 1651-1659.	2.9	194
60	DISC1-dependent switch from progenitor proliferation to migration in the developing cortex. Nature, 2011, 473, 92-96.	27.8	181
61	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. American Journal of Human Genetics, 2011, 89, 713-730.	6.2	178
62	Missense Mutations in TCF8 Cause Late-Onset Fuchs Corneal Dystrophy and Interact with FCD4 on Chromosome 9p. American Journal of Human Genetics, 2010, 86, 45-53.	6.2	167
63	MKKS/BBS6, a divergent chaperonin-like protein linked to the obesity disorder Bardet-Biedl syndrome, is a novel centrosomal component required for cytokinesis. Journal of Cell Science, 2005, 118, 1007-1020.	2.0	166
64	Epigenetic control of intestinal barrier function and inflammation in zebrafish. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 2770-2775.	7.1	163
65	A functional variant in the CFI gene confers a high risk of age-related macular degeneration. Nature Genetics, 2013, 45, 813-817.	21.4	162
66	Paralogy Mapping: Identification of a Region in the Human MHC Triplicated onto Human Chromosomes 1 and 9 Allows the Prediction and Isolation of NovelPBXandNOTCHLoci. Genomics, 1996, 35, 101-108.	2.9	161
67	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	6.2	150
68	Regulation of autism-relevant behaviors by cerebellarâ€“prefrontal cortical circuits. Nature Neuroscience, 2020, 23, 1102-1110.	14.8	149
69	Loss of Î´-catenin function in severe autism. Nature, 2015, 520, 51-56.	27.8	145
70	Cilia in vertebrate development and disease. Development (Cambridge), 2012, 139, 443-448.	2.5	144
71	Mutations in LOXHD1, a Recessive-Deafness Locus, Cause Dominant Late-Onset Fuchs Corneal Dystrophy. American Journal of Human Genetics, 2012, 90, 533-539.	6.2	141
72	The ciliopathies: a transitional model into systems biology of human genetic disease. Current Opinion in Genetics and Development, 2012, 22, 290-303.	3.3	137

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73	Disruption of a Ciliary B9 Protein Complex Causes Meckel Syndrome. American Journal of Human Genetics, 2011, 89, 94-110.	6.2	136
74	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. American Journal of Human Genetics, 2013, 92, 210-220.	6.2	135
75	Exploring the molecular basis of Bardet-Biedl syndrome. Human Molecular Genetics, 2001, 10, 2293-2299.	2.9	134
76	Missense mutations inTENM4, a regulator of axon guidance and central myelination, cause essential tremor. Human Molecular Genetics, 2015, 24, 5677-5686.	2.9	134
77	Pitchfork Regulates Primary Cilia Disassembly and Left-Right Asymmetry. Developmental Cell, 2010, 19, 66-77.	7.0	133
78	The Emerging Complexity of the Vertebrate Cilium: New Functional Roles for an Ancient Organelle. Developmental Cell, 2006, 11, 9-19.	7.0	131
79	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
80	The 1.4-Mb CMT1A Duplication/HNPP Deletion Genomic Region Reveals Unique Genome Architectural Features and Provides Insights into the Recent Evolution of New Genes. Genome Research, 2001, 11, 1018-1033.	5.5	129
81	Recruitment of PCM1 to the Centrosome by the Cooperative Action of DISC1 and BBS4. Archives of General Psychiatry, 2008, 65, 996.	12.3	124
82	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. American Journal of Human Genetics, 2016, 99, 174-187.	6.2	124
83	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	27.0	120
84	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	6.2	119
85	Missense mutations in the sodium borate cotransporter SLC4A11 cause late-onset Fuchs corneal dystrophya. Human Mutation, 2010, 31, 1261-1268.	2.5	117
86	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and NEK2 as a new disease gene. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 16139-16144.	7.1	115
87	The continuum of causality in human genetic disorders. Genome Biology, 2016, 17, 233.	8.8	114
88	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	6.2	112
89	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	6.2	111
90	BBS4 Is a Minor Contributor to Bardet-Biedl Syndrome and May Also Participate in Triallelic Inheritance. American Journal of Human Genetics, 2002, 71, 22-29.	6.2	110

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91	Functional analyses of variants reveal a significant role for dominant negative and common alleles in oligogenic Bardetâ€“Biedl syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 10602-10607.	7.1	110
92	A Splice-Site Mutation in a Retina-Specific Exon of BBS8 Causes Nonsyndromic Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 86, 805-812.	6.2	109
93	DNAH11 Localization in the Proximal Region of Respiratory Cilia Defines Distinct Outer Dynein Arm Complexes. American Journal of Respiratory Cell and Molecular Biology, 2016, 55, 213-224.	2.9	107
94	Impaired photoreceptor protein transport and synaptic transmission in a mouse model of Bardetâ€“Biedl syndrome. Vision Research, 2007, 47, 3394-3407.	1.4	106
95	Identification of cis-suppression of human disease mutations by comparative genomics. Nature, 2015, 524, 225-229.	27.8	106
96	Phenotypic characterization of Bbs4 null mice reveals age-dependent penetrance and variable expressivity. Human Genetics, 2006, 120, 211-226.	3.8	104
97	Loss of Bardetâ€“Biedl syndrome protein-8 (BBS8) perturbs olfactory function, protein localization, and axon targeting. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 10320-10325.	7.1	103
98	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	30.7	103
99	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. American Journal of Human Genetics, 2015, 96, 816-825.	6.2	102
100	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 791-802.	8.2	102
101	Genetic and Functional Dissection of HTRA1 and LOC387715 in Age-Related Macular Degeneration. PLoS Genetics, 2010, 6, e1000836.	3.5	101
102	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
103	Bardet-Biedl Syndrome-associated Small GTPase ARL6 (BBS3) Functions at or near the Ciliary Gate and Modulates Wnt Signaling. Journal of Biological Chemistry, 2010, 285, 16218-16230.	3.4	100
104	Recurrent De Novo Mutations in PACS1 Cause Defective Cranial-Neural-Crest Migration and Define a Recognizable Intellectual-Disability Syndrome. American Journal of Human Genetics, 2012, 91, 1122-1127.	6.2	96
105	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
106	The Genetic Basis of Hydrocephalus. Annual Review of Neuroscience, 2016, 39, 409-435.	10.7	93
107	A Novel Ribosomopathy Caused by Dysfunction of RPL10 Disrupts Neurodevelopment and Causes X-Linked Microcephaly in Humans. Genetics, 2014, 198, 723-733.	2.9	92
108	Newfoundland Rod-Cone Dystrophy, an Early-Onset Retinal Dystrophy, Is Caused by Splice-Junction Mutations in RLBP1. American Journal of Human Genetics, 2002, 70, 955-964.	6.2	91

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109	Functional modules, mutational load and human genetic disease. Trends in Genetics, 2010, 26, 168-176.	6.7	89
110	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
111	Mutations in AGBL1 Cause Dominant Late-Onset Fuchs Corneal Dystrophy and Alter Protein-Protein Interaction with TCF4. American Journal of Human Genetics, 2013, 93, 758-764.	6.2	86
112	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
113	BRF1 mutations alter RNA polymerase IIIâ€“dependent transcription and cause neurodevelopmental anomalies. Genome Research, 2015, 25, 155-166.	5.5	85
114	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84
115	Genetic Modifiers and Oligogenic Inheritance. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017145-a017145.	6.2	84
116	SCRIB and PUF60 Are Primary Drivers of the Multisystemic Phenotypes of the 8q24.3 Copy-Number Variant. American Journal of Human Genetics, 2013, 93, 798-811.	6.2	82
117	Genetic and Mutational Analyses of a Large Multiethnic Bardet-Biedl Cohort Reveal a Minor Involvement of BBS6 and Delineate the Critical Intervals of Other Loci. American Journal of Human Genetics, 2001, 68, 606-616.	6.2	80
118	Linkage of a Mild Late-Onset Phenotype of Fuchs Corneal Dystrophy to a Novel Locus at 5q33.1-q35.2. , 2009, 50, 5667.		80
119	Mutation analysis in Bardetâ€“Biedl syndrome by DNA pooling and massively parallel resequencing in 105 individuals. Human Genetics, 2011, 129, 79-90.	3.8	80
120	Novel bone morphogenetic protein signaling through Smad2 and Smad3 to regulate cancer progression and development. FASEB Journal, 2014, 28, 1248-1267.	0.5	80
121	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	6.2	80
122	Recessive Mutations in the Î±3 (VI) Collagen Gene COL6A3 Cause Early-Onset Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 883-893.	6.2	79
123	Ciliopathy proteins regulate paracrine signaling by modulating proteasomal degradation of mediators. Journal of Clinical Investigation, 2014, 124, 2059-2070.	8.2	79
124	Discovery and Functional Annotation of SIX6 Variants in Primary Open-Angle Glaucoma. PLoS Genetics, 2014, 10, e1004372.	3.5	78
125	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79.	6.2	77
126	Rbm8aHaploinsufficiency Disrupts Embryonic Cortical Development Resulting in Microcephaly. Journal of Neuroscience, 2015, 35, 7003-7018.	3.6	75

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127	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
128	Functionally compromisedCHD7alleles in patients with isolated GnRH deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 17953-17958.	7.1	74
129	Heritability and Genome-Wide Association Study to Assess Genetic Differences between Advanced Age-related Macular Degeneration Subtypes. Ophthalmology, 2012, 119, 1874-1885.	5.2	73
130	Endoglin mediates fibronectin/Î±5Î²1 integrin and TGF-Î² pathway crosstalk in endothelial cells. EMBO Journal, 2012, 31, 3885-3900.	7.8	73
131	Loss of Bardetâ€“Biedl syndrome proteins causes defects in peripheral sensory innervation and function. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17524-17529.	7.1	71
132	Functional interactions between the ciliopathy-associated Meckel syndrome 1 (MKS1) protein and two novel MKS1-related (MKSR) proteins. Journal of Cell Science, 2009, 122, 611-624.	2.0	71
133	Mutations in RAD21 Disrupt Regulation of APOB in Patients With Chronic Intestinal Pseudo-Obstruction. Gastroenterology, 2015, 148, 771-782.e11.	1.3	71
134	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	8.2	69
135	Identification and mapping of a novel human gene, HRMT1L1, homologous to the rat protein arginine N-methyltransferase 1 (PRMT1) gene. Mammalian Genome, 1997, 8, 526-529.	2.2	68
136	A Mutation in SLC24A1 Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2010, 87, 523-531.	6.2	67
137	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
138	Metabolic Regulation and Energy Homeostasis through the Primary Cilium. Cell Metabolism, 2015, 21, 21-31.	16.2	67
139	Replication of TCF4 through Association and Linkage Studies in Late-Onset Fuchs Endothelial Corneal Dystrophy. PLoS ONE, 2011, 6, e18044.	2.5	66
140	Context-Dependent Regulation of Wnt Signaling through the Primary Cilium. Journal of the American Society of Nephrology: JASN, 2013, 24, 10-18.	6.1	66
141	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	6.2	65
142	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
143	Chapter 7 Ciliary Function and Wnt Signal Modulation. Current Topics in Developmental Biology, 2008, 85, 175-195.	2.2	64
144	Interpreting human genetic variation with in vivo zebrafish assays. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1960-1970.	3.8	63

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145	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. American Journal of Human Genetics, 2015, 97, 790-800.	6.2	63
146	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	6.2	63
147	An improved protocol for the analysis of SOD1 gene mutations, and a new mutation in exon 4. Human Molecular Genetics, 1995, 4, 1101-1104.	2.9	62
148	A Novel C-Terminal Binding Protein (CTBP2) Is Closely Related toCTBP1,an Adenovirus E1A-Binding Protein, and Maps to Human Chromosome 21q21.3. Genomics, 1998, 47, 294-299.	2.9	61
149	Replication of theTCF4Intronic Variant in Late-Onset Fuchs Corneal Dystrophy and Evidence of Independence from theFCD2Locus. , 2011, 52, 2825.		61
150	AMD and the alternative complement pathway: genetics and functional implications. Human Genomics, 2016, 10, 23.	2.9	61
151	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
152	Triallelic inheritance: a bridge between Mendelian and multifactorial traits. Annals of Medicine, 2004, 36, 262-272.	3.8	58
153	OTX2mutations contribute to the otocephaly-dysgnathia complex. Journal of Medical Genetics, 2012, 49, 373-379.	3.2	58
154	Unique among ciliopathies: primary ciliary dyskinesia, a motile cilia disorder. F1000prime Reports, 2015, 7, 36.	5.9	58
155	Cystic diseases of the kidney: ciliary dysfunction and cystogenic mechanisms. Pediatric Nephrology, 2011, 26, 1181-1195.	1.7	56
156	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	6.2	56
157	Endoglin interacts with VEGFR2 to promote angiogenesis. FASEB Journal, 2018, 32, 2934-2949.	0.5	56
158	Population Bottlenecks as a Potential Major Shaping Force of Human Genome Architecture. PLoS Genetics, 2007, 3, e119.	3.5	55
159	Direct role of Bardetâ€“Biedl syndrome proteins in transcriptional regulation. Journal of Cell Science, 2012, 125, 362-375.	2.0	53
160	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	6.2	53
161	A hypomorphic inherited pathogenic variant in DDX3X causes male intellectual disability with additional neurodevelopmental and neurodegenerative features. Human Genomics, 2018, 12, 11.	2.9	53
162	Association of Whirlin with Ca_v1.3 (Î±_{1D}) Channels in Photoreceptors, Defining a Novel Member of the Usher Protein Network. , 2010, 51, 2338.		52

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