Nicholas Katsanis

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3583068/publications.pdf

Version: 2024-02-01

286	36,169	2802	4015
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

#	Article	IF	Citations
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

#	Article	IF	Citations
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

#	Article	IF	Citations
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

#	Article	IF	Citations
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

#	Article	IF	Citations
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

#	Article	IF	Citations
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

#	Article	IF	Citations
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

#	Article	IF	Citations
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

#	Article	IF	Citations
163	Targeted Resequencing and Systematic InÂVivo Functional Testing Identifies Rare Variants in MEIS1 as Significant Contributors to Restless Legs Syndrome. American Journal of Human Genetics, 2014, 95, 85-95.	6.2	52
164	Epistasis between RET and BBS mutations modulates enteric innervation and causes syndromic Hirschsprung disease. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 13921-13926.	7.1	51
165	Molecular Analysis of Bardet-Biedl Syndrome Families: Report of 21 Novel Mutations in 10 Genes. , 2011, 52, 5317.		51
166	Genetic architecture of reciprocal CNVs. Current Opinion in Genetics and Development, 2013, 23, 240-248.	3.3	51
167	Temperature-activated ion channels in neural crest cells confer maternal feverâ€“associated birth defects. Science Signaling, 2017, 10, .	3.6	51
168	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
169	Small molecule inhibition of RAS/MAPK signaling ameliorates developmental pathologies of Kabuki Syndrome. Scientific Reports, 2018, 8, 10779.	3.3	50
170	In Vivo Modeling of the Morbid Human Genome using Danio rerio. Journal of Visualized Experiments, 2013, , e50338.	0.3	49
171	Acoustofluidic rotational tweezing enables high-speed contactless morphological phenotyping of zebrafish larvae. Nature Communications, 2021, 12, 1118.	12.8	49
172	An Essential Role for DYF-11/MIP-T3 in Assembling Functional Intraflagellar Transport Complexes. PLoS Genetics, 2008, 4, e1000044.	3.5	48
173	Expansion ofCTG18.1Trinucleotide Repeat inTCF4Is a Potent Driver of Fuchs' Corneal Dystrophy. , 2015, 56, 4531.		48
174	Loss of Function Mutations in NNT Are Associated With Left Ventricular Noncompaction. Circulation: Cardiovascular Genetics, 2015, 8, 544-552.	5.1	48
175	Delineation of the Critical Interval of Bardet-Biedl Syndrome 1 (BBS1) to a Small Region of 11q13, through Linkage and Haplotype Analysis of 91 Pedigrees. American Journal of Human Genetics, 1999, 65, 1672-1679.	6.2	47
176	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	6.2	47
177	A human laterality disorder caused by a homozygous deleterious mutation inMMP21. Journal of Medical Genetics, 2015, 52, 840-847.	3.2	46
178	In vivo Modeling Implicates APOL1 in Nephropathy: Evidence for Dominant Negative Effects and Epistasis under Anemic Stress. PLoS Genetics, 2015, 11, e1005349.	3.5	45
179	A Novel Missense Mutation of Wilmsâ€™ Tumor 1 Causes Autosomal Dominant FSGS. Journal of the American Society of Nephrology: JASN, 2015, 26, 831-843.	6.1	45
180	A truncating mutation in CEP55 is the likely cause of MARCH, a novel syndrome affecting neuronal mitosis. Journal of Medical Genetics, 2017, 54, 490-501.	3.2	45

#	Article	IF	Citations
181	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
182	Rapid identification of kidney cyst mutations by whole exome sequencing in zebrafish. Development (Cambridge), 2013, 140, 4445-4451.	2.5	43
183	Transient laminin beta 1a Induction Defines the Wound Epidermis during Zebrafish Fin Regeneration. PLoS Genetics, 2015, 11, e1005437.	3.5	43
184	Endoglin regulates PI3-kinase/Akt trafficking and signaling to alter endothelial capillary stability during angiogenesis. Molecular Biology of the Cell, 2012, 23, 2412-2423.	2.1	41
185	Dosage Changes of a Segment at 17p13.1 Lead to Intellectual Disability and Microcephaly as a Result of Complex Genetic Interaction of Multiple Genes. American Journal of Human Genetics, 2014, 95, 565-578.	6.2	40
186	Loss-of-Function Mutations in UNC45A Cause a Syndrome Associating Cholestasis, Diarrhea, Impaired Hearing, and Bone Fragility. American Journal of Human Genetics, 2018, 102, 364-374.	6.2	40
187	Loss-of-function mutations in PTPRJ cause a new form of inherited thrombocytopenia. Blood, 2019, 133, 1346-1357.	1.4	40
188	Arrayed primer extension technology simplifies mutation detection in Bardetâ€“Biedl and AlstrÃ¶m syndrome. European Journal of Human Genetics, 2011, 19, 485-488.	2.8	38
189	Mutations in LRRC50 Predispose Zebrafish and Humans to Seminomas. PLoS Genetics, 2013, 9, e1003384.	3.5	38
190	Mutations in ATP13A2 (PARK9) are associated with an amyotrophic lateral sclerosis-like phenotype, implicating this locus in further phenotypic expansion. Human Genomics, 2019, 13, 19.	2.9	38
191	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. Nature Communications, 2020, 11, 3698.	12.8	38
192	New Polymorphic Short Tandem Repeats for PCR-based Charcot-Marie-Tooth Disease Type 1A Duplication Diagnosis. Clinical Chemistry, 2001, 47, 838-843.	3.2	37
193	Targeted resequencing identifies PTCH1 as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. Genome Research, 2016, 26, 474-485.	5.5	37
194	Survey of Human Chromosome 21 Gene Expression Effects on Early Development in Danio rerio. G3: Genes, Genomes, Genetics, 2018, 8, 2215-2223.	1.8	37
195	The Meckel syndrome- associated protein MKS1 functionally interacts with components of the BBSome and IFT complexes to mediate ciliary trafficking and hedgehog signaling. PLoS ONE, 2017, 12, e0173399.	2.5	36
196	CFAP45 deficiency causes situs abnormalities and asthenospermia by disrupting an axonemal adenine nucleotide homeostasis module. Nature Communications, 2020, 11, 5520.	12.8	36
197	Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology, 2018, 91, e319-e330.	1.1	35
198	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	6.2	35

#	Article	IF	Citations
199	Ectopic Overexpression of Sonic Hedgehog (Shh) Induces Stromal Expansion and Metaplasia in the Adult Murine Pancreas. Neoplasia, 2011, 13, 923-IN18.	5.3	34
200	Prevalence and Severity of Fuchs Corneal Dystrophy in Tangier Island. American Journal of Ophthalmology, 2012, 153, 1067-1072.	3.3	34
201	Oligogenic Effects of 16p11.2 Copy-Number Variation on Craniofacial Development. Cell Reports, 2019, 28, 3320-3328.e4.	6.4	34
202	A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype. Human Molecular Genetics, 2014, 23, 3607-3617.	2.9	33
203	An evaluation of the draft human genome sequence. Nature Genetics, 2001, 29, 88-91.	21.4	32
204	Kctd13-deficient mice display short-term memory impairment and sex-dependent genetic interactions. Human Molecular Genetics, 2019, 28, 1474-1486.	2.9	32
205	A computational/functional genomics approach for the enrichment of the retinal transcriptome and the identification of positional candidate retinopathy genes. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 14326-14331.	7.1	31
206	The Bardetâ€“Biedl syndrome-related protein CCDC28B modulates mTORC2 function and interacts with SIN1 to control cilia length independently of the mTOR complex. Human Molecular Genetics, 2013, 22, 4031-4042.	2.9	31
207	The Immune Signaling Adaptor LAT Contributes to the Neuroanatomical Phenotype of 16p11.2 BP2-BP3 CNVs. American Journal of Human Genetics, 2017, 101, 564-577.	6.2	30
208	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	30
209	A Novel Test for Recessive Contributions to Complex Diseases Implicates Bardet-Biedl Syndrome Gene BBS10 in Idiopathic Type 2 Diabetes and Obesity. American Journal of Human Genetics, 2014, 95, 509-520.	6.2	29
210	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. American Journal of Human Genetics, 2020, 106, 893-904.	6.2	29
211	Small molecule intervention in microtubule-associated human disease. Human Molecular Genetics, 2005, 14, R291-R300.	2.9	28
212	Elution Profile Analysis of SDS-induced Subcomplexes by Quantitative Mass Spectrometry. Molecular and Cellular Proteomics, 2014, 13, 1382-1391.	3.8	28
213	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2021, 144, 1451-1466.	7.6	28
214	Identification, Expression, and Chromosomal Localization of Ubiquitin Conjugating Enzyme 7 (UBE2G2), a Human Homologue of theSaccharomyces cerevisiae Ubc7Gene. Genomics, 1998, 51, 128-131.	2.9	27
215	Mutations in NCAPG2 Cause a Severe Neurodevelopmental Syndrome that Expands the Phenotypic Spectrum of Condensinopathies. American Journal of Human Genetics, 2019, 104, 94-111.	6.2	27
216	Whole Exome Sequencing of a Dominant Retinitis Pigmentosa Family Identifies a Novel Deletion inPRPF31. , 2014, 55, 2121.		26

#	Article	IF	Citations
217	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. Molecular Psychiatry, 2019, 24, 1748-1768.	7.9	26
218	ZNHIT3 is defective in PEHO syndrome, a severe encephalopathy with cerebellar granule neuron loss. Brain, 2017, 140, 1267-1279.	7.6	23
219	Genetics and functions of the retinoic acid pathway, with special emphasis on the eye. Human Genomics, 2019, 13, 61.	2.9	23
220	Progression of Fuchs Corneal Dystrophy in a Family Linked to theFCD1Locus. , 2009, 50, 5662.		22
221	Putative digenic inheritance of heterozygousRP1L1andC2orf71null mutations in syndromic retinal dystrophy. Ophthalmic Genetics, 2017, 38, 127-132.	1.2	22
222	Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. American Journal of Human Genetics, 2017, 100, 666-675.	6.2	22
223	Partial uniparental isodisomy of chromosome 16 unmasks a deleterious biallelic mutation in IFT140 that causes Mainzer-Saldino syndrome. Human Genomics, 2017, 11, 16.	2.9	22
224	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nature Genetics, 2020, 52, 1145-1150.	21.4	22
225	The complexity of the cilium: spatiotemporal diversity of an ancient organelle. Current Opinion in Cell Biology, 2018, 55, 139-149.	5.4	21
226	A <scp>BBS1 SVA</scp> F retrotransposon insertion is a frequent cause of <scp>Bardetâ€Biedl</scp> syndrome. Clinical Genetics, 2021, 99, 318-324.	2.0	21
227	Ciliary proteins and exencephaly. Nature Genetics, 2006, 38, 135-136.	21.4	20
228	BBS4 regulates the expression and secretion of FSTL1, a protein that participates in ciliogenesis and the differentiation of 3T3-L1. Scientific Reports, 2017, 7, 9765.	3.3	20
229	Bi-allelic Variants in DYNC1I2 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. American Journal of Human Genetics, 2019, 104, 1073-1087.	6.2	19
230	Zebrafish Assays of Ciliopathies. Methods in Cell Biology, 2011, 105, 257-272.	1.1	18
231	Human guanylate kinase (GUK1): cDNA sequence, expression and chromosomal localisation. FEBS Letters, 1996, 385, 185-188.	2.8	17
232	Thermosensory and mechanosensory perception in human genetic disease. Human Molecular Genetics, 2009, 18, R146-R155.	2.9	17
233	Neuroanatomical and behavioral deficits in mice haploinsufficient for Pericentriolar material 1 (Pcm1). Neuroscience Research, 2015, 98, 45-49.	1.9	17
234	Newborn screening and the era of medical genomics. Seminars in Perinatology, 2015, 39, 617-622.	2.5	17

#	Article	IF	Citations
235	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. Genetics in Medicine, 2019, 21, 2532-2542.	2.4	17
236	The gene encoding the p60 subunit of chromatin assembly factor I (CAF1P60) maps to human chromosome 21q22.2, a region associated with some of the major features of Down syndrome. Human Genetics, 1996, 98, 497-499.	3.8	16
237	A manually curated functional annotation of the human X chromosome. Nature Genetics, 2005, 37, 331-332.	21.4	16
238	Life without Centrioles: Cilia in the Spotlight. Cell, 2006, 125, 1228-1230.	28.9	16
239	Analysis of 30 Genes (355 SNPS) Related to Energy Homeostasis for Association with Adiposity in European-American and Yupâ€™ik Eskimo Populations. Human Heredity, 2009, 67, 193-205.	0.8	16
240	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. Human Mutation, 2020, 41, 2179-2194.	2.5	16
241	Multidisciplinary approaches for elucidating genetics and molecular pathogenesis of urinary tract malformations. Kidney International, 2022, 101, 473-484.	5.2	16
242	CSGALNACT1â€congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. Human Mutation, 2020, 41, 655-667.	2.5	15
243	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
244	A recessive variant in TFAM causes mtDNA depletion associated with primary ovarian insufficiency, seizures, intellectual disability and hearing loss. Human Genetics, 2021, 140, 1733-1751.	3.8	15
245	Localisation of receptor interacting protein140 (RIP140) within 100kb of D21S13 on 21q11, a gene-poor region of the human genome. Human Genetics, 1998, 102, 221-223.	3.8	14
246	Managing Incidental Genomic Findings in Clinical Trials: Fulfillment of the Principle of Justice. PLoS Medicine, 2014, 11, e1001584.	8.4	14
247	Systematic Functional Testing of Rare Variants: Contributions of CFI to Age-Related Macular Degeneration. , 2017, 58, 1570.		13
248	PCM1 is necessary for focal ciliary integrity and is a candidate for severe schizophrenia. Nature Communications, 2020, 11, 5903.	12.8	13
249	Age-Severity Relationships in Families Linked toFCD2with Retroillumination Photography. , 2010, 51, 6298.		12
250	TCF12 haploinsufficiency causes autosomal dominant Kallmann syndrome and reveals network-level interactions between causal loci. Human Molecular Genetics, 2020, 29, 2435-2450.	2.9	10
251	High-resolution comparative physical mapping of mouse Chromosome 10 in the region of homology with human Chromosome 21. Mammalian Genome, 1999, 10, 229-234.	2.2	9
252	Oligogenic Disease. , 2010, , 243-262.		9

#	Article	IF	Citations
253	Whole exome sequencing and functional studies identify an intronic mutation in <scp>TRAPPC2</scp> that causes <scp>SEDT</scp>. Clinical Genetics, 2014, 85, 359-364.	2.0	9
254	Genome-wide suppressor screen identifies USP35/USP38 as therapeutic candidates for ciliopathies. JCI Insight, 2019, 4, .	5.0	9
255	Cell Polarization Defects in Early Heart Development. Circulation Research, 2007, 101, 122-124.	4.5	8
256	Phosphorylation of Threonine 794 on Tie1 by Rac1/PAK1 Reveals a Novel Angiogenesis Regulatory Pathway. PLoS ONE, 2015, 10, e0139614.	2.5	8
257	Performance of computational methods for the evaluation of pericentriolar material 1 missense variants in CAGIâ€5. Human Mutation, 2019, 40, 1474-1485.	2.5	8
258	Analysis of Single Nucleotide Variants in CRISPR-Cas9 Edited Zebrafish Exomes Shows No Evidence of Off-Target Inflation. Frontiers in Genetics, 2019, 10, 949.	2.3	7
259	Bardet-Biedl Syndrome in an African-American Patient: Should the Diagnostic Criteria Be Expanded to Include Hydrometrocolpos?. Ophthalmic Genetics, 2007, 28, 95-99.	1.2	6
260	From association to causality: the new frontier for complex traits. Genome Medicine, 2009, 1, 23.	8.2	6
261	The potential of DISC1 protein as a therapeutic target for mental illness. Expert Opinion on Therapeutic Targets, 2016, 20, 641-643.	3.4	6
262	Loss of function mutations in CCDC32 cause a congenital syndrome characterized by craniofacial, cardiac and neurodevelopmental anomalies. Human Molecular Genetics, 2020, 29, 1489-1497.	2.9	6
263	Identification of novel genes expressed during metanephric induction through single-cell library screening. Kidney International, 2000, 57, 2221-2228.	5.2	5
264	Exome and genome sequencing of neonates with neurodevelopmental disorders. Future Neurology, 2012, 7, 655-658.	0.5	4
265	Dissecting the complexity of CNV pathogenicity: insights from Drosophila and zebrafish models. Current Opinion in Genetics and Development, 2021, 68, 79-87.	3.3	4
266	Understanding cargo specificity in intraflagellar transport. EMBO Journal, 2011, 30, 2518-2519.	7.8	3
267	Dissecting Intraflagellar Transport, One Molecule at a Time. Developmental Cell, 2014, 31, 263-264.	7.0	3
268	Mitochondrial Copy Number as a Biomarker for Autism?. Pediatrics, 2016, 137, e20160049.	2.1	3
269	Polyketide Synthase Plays a Conserved Role in Otolith Formation. Zebrafish, 2019, 16, 363-369.	1.1	3
270	Disruption of a Ciliary B9 Protein Complex Causes Meckel Syndrome. American Journal of Human Genetics, 2011, 89, 589.	6.2	2

#	Article	IF	Citations
271	Zebrafish: A Model System to Study the Architecture of Human Genetic Disease. , 2017, , 651-670.		2
272	Point: Treating Human Genetic Disease One Base Pair at a Time: The Benefits of Gene Editing. Clinical Chemistry, 2018, 64, 486-488.	3.2	2
273	Next-Generation Sequencing of the Human Olfactory Receptors. Methods in Molecular Biology, 2013, 1003, 133-147.	0.9	2
274	Participant-Partners in Genetic Research: An Exome Study with Families of Children with Unexplained Medical Conditions. Journal of Participatory Medicine, 2018, 10, e2.	1.3	2
275	Mapping of a novel SH3 domain protein and two proteins of unknown function to human chromosome 21. Human Genetics, 1997, 100, 477-480.	3.8	1
276	An Age-Old Problem. PLoS Genetics, 2007, 3, e37.	3.5	1
277	Imprinting in the brain. Nature, 2011, 475, 299-300.	27.8	1
278	Renal cystic disease: from mechanisms to drug development. Drug Discovery Today Disease Mechanisms, 2013, 10, e125-e133.	0.8	1
279	A t(5;16) translocation is the likely driver of a syndrome with ambiguous genitalia, facial dysmorphism, intellectual disability, and speech delay. Journal of Physical Education and Sports Management, 2016, 2, a000703.	1.2	1
280	Oligogenic Effects of 16p11.2 Copy Number Variation on Craniofacial Development. SSRN Electronic Journal, 0, , .	0.4	1
281	An improved protocol for the analysis of SODI gene mutations, and a new mutation in exon 4. Human Molecular Genetics, 1995, 4, 1474-1474.	2.9	0
282	2017 Curt Stern Award: The Complexity of Simple Genetics. American Journal of Human Genetics, 2018, 102, 355-358.	6.2	0
283	Bardet-Biedl Syndrome. , 2018, , 27-50.		0
284	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 1362-1362.	8.2	0
285	Ciliary Syndromes and Obesity. Growth Hormone, 2011, , 71-93.	0.2	0
286	SAT-LB071 Loss of Function (LoF) mutations in TCF12 Cause Autosomal Dominant Kallmann Syndrome and Reveal Network-level Interactions Between Causal Loci. Journal of the Endocrine Society, 2019, 3, .	0.2	0