

# Nicholas Katsanis

## List of Publications by Year in descending order

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Version: 2024-02-01

286  
papers

36,169  
citations

2802

94  
h-index

4015

176  
g-index

330  
all docs

330  
docs citations

330  
times ranked

37523  
citing authors

#	ARTICLE	IF	CITATIONS
1	Multidisciplinary approaches for elucidating genetics and molecular pathogenesis of urinary tract malformations. <i>Kidney International</i> , 2022, 101, 473-484.	5.2	16
2	A <i>BBS1</i> SVA F retrotransposon insertion is a frequent cause of Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2021, 99, 318-324.	2.0	21
3	Acoustofluidic rotational tweezing enables high-speed contactless morphological phenotyping of zebrafish larvae. <i>Nature Communications</i> , 2021, 12, 1118.	12.8	49
4	Biallelic variants in <i>LIG3</i> cause a novel mitochondrial neurogastrointestinal encephalomyopathy. <i>Brain</i> , 2021, 144, 1451-1466.	7.6	28
5	Haploinsufficiency of the Sin3/HDAC corepressor complex member <i>SIN3B</i> causes a syndromic intellectual disability/autism spectrum disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 929-941.	6.2	15
6	Dissecting the complexity of CNV pathogenicity: insights from <i>Drosophila</i> and zebrafish models. <i>Current Opinion in Genetics and Development</i> , 2021, 68, 79-87.	3.3	4
7	A recessive variant in <i>TFAM</i> causes mtDNA depletion associated with primary ovarian insufficiency, seizures, intellectual disability and hearing loss. <i>Human Genetics</i> , 2021, 140, 1733-1751.	3.8	15
8	<i>CSGALNACT1</i> congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. <i>Human Mutation</i> , 2020, 41, 655-667.	2.5	15
9	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. <i>Nature Genetics</i> , 2020, 52, 1145-1150.	21.4	22
10	Regulation of autism-relevant behaviors by cerebellar prefrontal cortical circuits. <i>Nature Neuroscience</i> , 2020, 23, 1102-1110.	14.8	149
11	<i>PCM1</i> is necessary for focal ciliary integrity and is a candidate for severe schizophrenia. <i>Nature Communications</i> , 2020, 11, 5903.	12.8	13
12	Mutations in <i>FAM50A</i> suggest that Armfield XLID syndrome is a spliceosomopathy. <i>Nature Communications</i> , 2020, 11, 3698.	12.8	38
13	Loss of <i>CBY1</i> results in a ciliopathy characterized by features of Joubert syndrome. <i>Human Mutation</i> , 2020, 41, 2179-2194.	2.5	16
14	<i>CFAP45</i> deficiency causes situs abnormalities and asthenospermia by disrupting an axonemal adenine nucleotide homeostasis module. <i>Nature Communications</i> , 2020, 11, 5520.	12.8	36
15	Mutations in the Kinesin-2 Motor <i>KIF3B</i> Cause an Autosomal-Dominant Ciliopathy. <i>American Journal of Human Genetics</i> , 2020, 106, 893-904.	6.2	29
16	<i>TCF12</i> haploinsufficiency causes autosomal dominant Kallmann syndrome and reveals network-level interactions between causal loci. <i>Human Molecular Genetics</i> , 2020, 29, 2435-2450.	2.9	10
17	Loss of function mutations in <i>CCDC32</i> cause a congenital syndrome characterized by craniofacial, cardiac and neurodevelopmental anomalies. <i>Human Molecular Genetics</i> , 2020, 29, 1489-1497.	2.9	6
18	Performance of computational methods for the evaluation of pericentriolar material 1 missense variants in <i>CAGL</i> . <i>Human Mutation</i> , 2019, 40, 1474-1485.	2.5	8

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19	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	30
20	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
21	Oligogenic Effects of 16p11.2 Copy-Number Variation on Craniofacial Development. Cell Reports, 2019, 28, 3320-3328.e4.	6.4	34
22	Polyketide Synthase Plays a Conserved Role in Otolith Formation. Zebrafish, 2019, 16, 363-369.	1.1	3
23	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	6.2	35
24	Bi-allelic Variants in DYNC112 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. American Journal of Human Genetics, 2019, 104, 1073-1087.	6.2	19
25	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
26	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
27	Genetics and functions of the retinoic acid pathway, with special emphasis on the eye. Human Genomics, 2019, 13, 61.	2.9	23
28	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
29	Kctd13-deficient mice display short-term memory impairment and sex-dependent genetic interactions. Human Molecular Genetics, 2019, 28, 1474-1486.	2.9	32
30	Loss-of-function mutations in PTPRJ cause a new form of inherited thrombocytopenia. Blood, 2019, 133, 1346-1357.	1.4	40
31	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
32	Genome-wide suppressor screen identifies USP35/USP38 as therapeutic candidates for ciliopathies. JCI Insight, 2019, 4, .	5.0	9
33	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
34	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
35	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
36	2017 Curt Stern Award: The Complexity of Simple Genetics. American Journal of Human Genetics, 2018, 102, 355-358.	6.2	0

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37	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights. <i>Nature Genetics</i> , 2018, 50, 538-548.	21.4	406
38	Loss-of-Function Mutations in UNC45A Cause a Syndrome Associating Cholestasis, Diarrhea, Impaired Hearing, and Bone Fragility. <i>American Journal of Human Genetics</i> , 2018, 102, 364-374.	6.2	40
39	A hypomorphic inherited pathogenic variant in DDX3X causes male intellectual disability with additional neurodevelopmental and neurodegenerative features. <i>Human Genomics</i> , 2018, 12, 11.	2.9	53
40	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018, 102, 744-759.	6.2	51
41	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	6.2	87
42	Survey of Human Chromosome 21 Gene Expression Effects on Early Development in <i>Danio rerio</i> . <i>G3: Genes, Genomes, Genetics</i> , 2018, 8, 2215-2223.	1.8	37
43	Biallelic <i>SQSTM1</i> mutations in early-onset, variably progressive neurodegeneration. <i>Neurology</i> , 2018, 91, e319-e330.	1.1	35
44	Endoglin interacts with VEGFR2 to promote angiogenesis. <i>FASEB Journal</i> , 2018, 32, 2934-2949.	0.5	56
45	Small molecule inhibition of RAS/MAPK signaling ameliorates developmental pathologies of Kabuki Syndrome. <i>Scientific Reports</i> , 2018, 8, 10779.	3.3	50
46	The complexity of the cilium: spatiotemporal diversity of an ancient organelle. <i>Current Opinion in Cell Biology</i> , 2018, 55, 139-149.	5.4	21
47	Bardet-Biedl Syndrome. , 2018, , 27-50.		0
48	Participant-Partners in Genetic Research: An Exome Study with Families of Children with Unexplained Medical Conditions. <i>Journal of Participatory Medicine</i> , 2018, 10, e2.	1.3	2
49	Putative digenic inheritance of heterozygous <i>RP1L1</i> and <i>C2orf71</i> null mutations in syndromic retinal dystrophy. <i>Ophthalmic Genetics</i> , 2017, 38, 127-132.	1.2	22
50	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. <i>Nature Genetics</i> , 2017, 49, 238-248.	21.4	131
51	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 100, 352-363.	6.2	86
52	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. <i>New England Journal of Medicine</i> , 2017, 376, 742-754.	27.0	120
53	ZNHIT3 is defective in PEHO syndrome, a severe encephalopathy with cerebellar granule neuron loss. <i>Brain</i> , 2017, 140, 1267-1279.	7.6	23
54	A truncating mutation in CEP55 is the likely cause of MARCH, a novel syndrome affecting neuronal mitosis. <i>Journal of Medical Genetics</i> , 2017, 54, 490-501.	3.2	45

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55	Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. American Journal of Human Genetics, 2017, 100, 666-675.	6.2	22
56	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	6.2	63
57	Temperature-activated ion channels in neural crest cells confer maternal fever-associated birth defects. Science Signaling, 2017, 10, .	3.6	51
58	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
59	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
60	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
61	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	6.2	119
62	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
63	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
64	Zebrafish: A Model System to Study the Architecture of Human Genetic Disease. , 2017, , 651-670.		2
65	Systematic Functional Testing of Rare Variants: Contributions of <i>CFI</i> to Age-Related Macular Degeneration. , 2017, 58, 1570.		13
66	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
67	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
68	AMD and the alternative complement pathway: genetics and functional implications. Human Genomics, 2016, 10, 23.	2.9	61
69	The continuum of causality in human genetic disorders. Genome Biology, 2016, 17, 233.	8.8	114
70	Mitochondrial Copy Number as a Biomarker for Autism?. Pediatrics, 2016, 137, e20160049.	2.1	3
71	The Genetic Basis of Hydrocephalus. Annual Review of Neuroscience, 2016, 39, 409-435.	10.7	93
72	Gene expression elucidates functional impact of polygenic risk for schizophrenia. Nature Neuroscience, 2016, 19, 1442-1453.	14.8	952

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73	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. <i>American Journal of Human Genetics</i> , 2016, 99, 318-336.	6.2	112
74	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. <i>Nature Communications</i> , 2016, 7, 11491.	12.8	207
75	The potential of DISC1 protein as a therapeutic target for mental illness. <i>Expert Opinion on Therapeutic Targets</i> , 2016, 20, 641-643.	3.4	6
76	Targeted resequencing identifies <i>PTCH1</i> as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. <i>Genome Research</i> , 2016, 26, 474-485.	5.5	37
77	DNAH11 Localization in the Proximal Region of Respiratory Cilia Defines Distinct Outer Dynein Arm Complexes. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2016, 55, 213-224.	2.9	107
78	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. <i>Nature Genetics</i> , 2016, 48, 134-143.	21.4	1,167
79	Unique among ciliopathies: primary ciliary dyskinesia, a motile cilia disorder. <i>F1000prime Reports</i> , 2015, 7, 36.	5.9	58
80	In vivo Modeling Implicates APOL1 in Nephropathy: Evidence for Dominant Negative Effects and Epistasis under Anemic Stress. <i>PLoS Genetics</i> , 2015, 11, e1005349.	3.5	45
81	Transient laminin beta 1a Induction Defines the Wound Epidermis during Zebrafish Fin Regeneration. <i>PLoS Genetics</i> , 2015, 11, e1005437.	3.5	43
82	Phosphorylation of Threonine 794 on Tie1 by Rac1/PAK1 Reveals a Novel Angiogenesis Regulatory Pathway. <i>PLoS ONE</i> , 2015, 10, e0139614.	2.5	8
83	Expansion of <i>CTG18.1</i> Trinucleotide Repeat in <i>TCF4</i> Is a Potent Driver of Fuchs' Corneal Dystrophy. , 2015, 56, 4531.		48
84	Recessive Mutations in the $\pm 3$ (VI) Collagen Gene COL6A3 Cause Early-Onset Isolated Dystonia. <i>American Journal of Human Genetics</i> , 2015, 96, 883-893.	6.2	79
85	Loss of Function Mutations in <i>NNT</i> Are Associated With Left Ventricular Noncompaction. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 544-552.	5.1	48
86	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 904-913.	6.2	65
87	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. <i>American Journal of Human Genetics</i> , 2015, 97, 790-800.	6.2	63
88	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015, 97, 922-932.	6.2	101
89	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. <i>Cell Reports</i> , 2015, 12, 1169-1183.	6.4	211
90	Metabolic Regulation and Energy Homeostasis through the Primary Cilium. <i>Cell Metabolism</i> , 2015, 21, 21-31.	16.2	67

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91	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. <i>American Journal of Human Genetics</i> , 2015, 96, 245-257.	6.2	111
92	<i>BRF1</i> mutations alter RNA polymerase III-dependent transcription and cause neurodevelopmental anomalies. <i>Genome Research</i> , 2015, 25, 155-166.	5.5	85
93	Neuroanatomical and behavioral deficits in mice haploinsufficient for Pericentriolar material 1 ( <i>Pcm1</i> ). <i>Neuroscience Research</i> , 2015, 98, 45-49.	1.9	17
94	Epigenetic control of intestinal barrier function and inflammation in zebrafish. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 2770-2775.	7.1	163
95	Mutations in <i>DDX3X</i> Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	6.2	230
96	Missense mutations in <i>TENM4</i> , a regulator of axon guidance and central myelination, cause essential tremor. <i>Human Molecular Genetics</i> , 2015, 24, 5677-5686.	2.9	134
97	Identification of cis-suppression of human disease mutations by comparative genomics. <i>Nature</i> , 2015, 524, 225-229.	27.8	106
98	Mutations in <i>RAD21</i> Disrupt Regulation of <i>APOB</i> in Patients With Chronic Intestinal Pseudo-Obstruction. <i>Gastroenterology</i> , 2015, 148, 771-782.e11.	1.3	71
99	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. <i>American Journal of Human Genetics</i> , 2015, 96, 816-825.	6.2	102
100	Loss of $\beta$ -catenin function in severe autism. <i>Nature</i> , 2015, 520, 51-56.	27.8	145
101	<i>Rbm8a</i> Haploinsufficiency Disrupts Embryonic Cortical Development Resulting in Microcephaly. <i>Journal of Neuroscience</i> , 2015, 35, 7003-7018.	3.6	75
102	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. <i>American Journal of Human Genetics</i> , 2015, 96, 784-796.	6.2	53
103	A Novel Missense Mutation of Wilms' Tumor 1 Causes Autosomal Dominant FSGS. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 831-843.	6.1	45
104	<i>TMEM231</i> , mutated in orofacioidigital and Meckel syndromes, organizes the ciliary transition zone. <i>Journal of Cell Biology</i> , 2015, 209, 129-142.	5.2	95
105	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. <i>American Journal of Human Genetics</i> , 2015, 96, 519-531.	6.2	47
106	A human laterality disorder caused by a homozygous deleterious mutation in <i>MMP21</i> . <i>Journal of Medical Genetics</i> , 2015, 52, 840-847.	3.2	46
107	Newborn screening and the era of medical genomics. <i>Seminars in Perinatology</i> , 2015, 39, 617-622.	2.5	17
108	Joubert Syndrome in French Canadians and Identification of Mutations in <i>CEP104</i> . <i>American Journal of Human Genetics</i> , 2015, 97, 744-753.	6.2	56

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109	Genetic Modifiers and Oligogenic Inheritance. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017145-a017145.	6.2	84
110	The kinetochore protein, <i>CENPF</i> , is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
111	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	8.2	69
112	Discovery and Functional Annotation of SIX6 Variants in Primary Open-Angle Glaucoma. PLoS Genetics, 2014, 10, e1004372.	3.5	78
113	Managing Incidental Genomic Findings in Clinical Trials: Fulfillment of the Principle of Justice. PLoS Medicine, 2014, 11, e1001584.	8.4	14
114	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
115	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
116	Functionally compromised CHD7 alleles 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
117	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
118	Whole Exome Sequencing of a Dominant Retinitis Pigmentosa Family Identifies a Novel Deletion in <i>PRPF31</i> . , 2014, 55, 2121.		26
119	Activating mutations in <i>STIM1</i> and <i>ORAI1</i> 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
120	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
121	Novel bone morphogenetic protein signaling through Smad2 and Smad3 to regulate cancer progression and development. FASEB Journal, 2014, 28, 1248-1267.	0.5	80
122	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
123	Dissecting Intraflagellar Transport, One Molecule at a Time. Developmental Cell, 2014, 31, 263-264.	7.0	3
124	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
125	Targeted Resequencing and Systematic In Vivo 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
126	Elution Profile Analysis of SDS-induced Subcomplexes by Quantitative Mass Spectrometry. Molecular and Cellular Proteomics, 2014, 13, 1382-1391.	3.8	28



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127	<i>CHD8</i> regulates neurodevelopmental pathways associated with autism spectrum disorder in neural progenitors. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, E4468-77.	7.1	297
128	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. <i>Cell</i> , 2014, 158, 263-276.	28.9	637
129	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 745-754.	6.2	80
130	Interpreting human genetic variation with in vivo zebrafish assays. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2014, 1842, 1960-1970.	3.8	63
131	Whole exome sequencing and functional studies identify an intronic mutation in <i>TRAPPC2</i> that causes <i>SEDT</i> . <i>Clinical Genetics</i> , 2014, 85, 359-364.	2.0	9
132	Ciliopathy proteins regulate paracrine signaling by modulating proteasomal degradation of mediators. <i>Journal of Clinical Investigation</i> , 2014, 124, 2059-2070.	8.2	79
133	Genetic architecture of reciprocal CNVs. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 240-248.	3.3	51
134	Renal cystic disease: from mechanisms to drug development. <i>Drug Discovery Today Disease Mechanisms</i> , 2013, 10, e125-e133.	0.8	1
135	The Bardet-Biedl syndrome-related protein CCDC28B modulates mTORC2 function and interacts with SIN1 to control cilia length independently of the mTOR complex. <i>Human Molecular Genetics</i> , 2013, 22, 4031-4042.	2.9	31
136	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. <i>American Journal of Human Genetics</i> , 2013, 93, 357-367.	6.2	150
137	Mutations in <i>AGBL1</i> Cause Dominant Late-Onset Fuchs Corneal Dystrophy and Alter Protein-Protein Interaction with <i>TCF4</i> . <i>American Journal of Human Genetics</i> , 2013, 93, 758-764.	6.2	86
138	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. <i>American Journal of Human Genetics</i> , 2013, 93, 197-210.	6.2	43
139	SCRIB and PUF60 Are Primary Drivers of the Multisystemic Phenotypes of the 8q24.3 Copy-Number Variant. <i>American Journal of Human Genetics</i> , 2013, 93, 798-811.	6.2	82
140	Defects in the IFT-B Component <i>IFT172</i> Cause Jeune and Mainzer-Saldino Syndromes in Humans. <i>American Journal of Human Genetics</i> , 2013, 93, 915-925.	6.2	196
141	Seven new loci associated with age-related macular degeneration. <i>Nature Genetics</i> , 2013, 45, 433-439.	21.4	687
142	Context-Dependent Regulation of Wnt Signaling through the Primary Cilium. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 10-18.	6.1	66
143	Molecular genetic testing and the future of clinical genomics. <i>Nature Reviews Genetics</i> , 2013, 14, 415-426.	16.3	334
144	A functional variant in the <i>CFI</i> gene confers a high risk of age-related macular degeneration. <i>Nature Genetics</i> , 2013, 45, 813-817.	21.4	162

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145	Ataxia, Dementia, and Hypogonadotropism Caused by Disordered Ubiquitination. <i>New England Journal of Medicine</i> , 2013, 368, 1992-2003.	27.0	208
146	Exonic Deletions in <i>AUTS2</i> Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. <i>American Journal of Human Genetics</i> , 2013, 92, 210-220.	6.2	135
147	&em&gt;In Vivo&lt;/em&gt; Modeling of the Morbid Human Genome using &lt;em&gt;Danio rerio&lt;/em&gt;. <i>Journal of Visualized Experiments</i> , 2013, , e50338.	0.3	49
148	Mutations in <i>LRRC50</i> Predispose Zebrafish and Humans to Seminomas. <i>PLoS Genetics</i> , 2013, 9, e1003384.	3.5	38
149	Rapid identification of kidney cyst mutations by whole exome sequencing in zebrafish. <i>Development (Cambridge)</i> , 2013, 140, 4445-4451.	2.5	43
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