Santhosh D Girirajan

List of Publications by Year
in descending order

94	10,795	94433 37	51608 86
papers	citations	h-index	g-index

133	133	133	15354
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A general framework for identifying oligogenic combinations of rare variants in complex disorders. Genome Research, 2022, , .	5.5	7
2	16p12.1 Deletion Orthologs are Expressed in Motile Neural Crest Cells and are Important for Regulating Craniofacial Development in Xenopus laevis. Frontiers in Genetics, 2022, 13, 833083.	2.3	1
3	The chromatin remodeler ISWI acts during Drosophila development to regulate adult sleep. Science Advances, 2021, 7, .	10.3	9
4	<scp>Birtâ€Hoggâ€DubÃ©</scp> symptoms in <scp>Smithâ€Magenis</scp> syndrome include pediatricâ€onset pneumothorax. American Journal of Medical Genetics, Part A, 2021, 185, 1922-1924.	1.2	7
5	Functional assessment of the â€œtwo-hitâ€•model for neurodevelopmental defects in Drosophila and X. laevis. PLoS Genetics, 2021, 17, e1009112.	3.5	12
6	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
7	Genetic subtypes, allelic effects, and convergent neurodevelopmental mechanisms. Genome Medicine, 2021, 13, 99.	8.2	0
8	Combinatorial patterns of gene expression changes contribute to variable expressivity of the developmental delay-associated 16p12.1 deletion. Genome Medicine, 2021, 13, 163.	8.2	5
9	Macrophage Selenoproteins Restrict Intracellular Replication of Francisella tularensis and Are Essential for Host Immunity. Frontiers in Immunology, 2021, 12, 701341.	4.8	5
10	An evolutionary driver of interspersed segmental duplications in primates. Genome Biology, 2020, 21, 202.	8.8	19
11	Gene discoveries in autism are biased towards comorbidity with intellectual disability. Journal of Medical Genetics, 2020, 57, 647-652.	3.2	12
12	Drosophila models of pathogenic copy-number variant genes show global and non-neuronal defects during development. PLoS Genetics, 2020, 16, e1008792.	3.5	9
13	NCBP2 modulates neurodevelopmental defects of the 3q29 deletion in Drosophila and Xenopus laevis models. PLoS Genetics, 2020, 16, e1008590.	3.5	30
14	Title is missing!. , 2020, 16, e1008792.		0
15	Title is missing!. , 2020, 16, e1008792.		0
16	Title is missing!. , 2020, 16, e1008792.		0
17	Title is missing!. , 2020, 16, e1008792.		0
18	Title is missing!. , 2020, 16, e1008590.		0

#	Article	IF	Citations
19	Title is missing!. , 2020, 16, e1008590.		0
20	Title is missing!. , 2020, 16, e1008590.		0
21	Title is missing!. , 2020, 16, e1008590.		0
22	Dissecting the genetic basis of comorbid epilepsy phenotypes in neurodevelopmental disorders. Genome Medicine, 2019, 11, 65.	8.2	35
23	An interaction-based model for neuropsychiatric features of copy-number variants. PLoS Genetics, 2019, 15, e1007879.	3.5	39
24	A machine-learning approach for accurate detection of copy number variants from exome sequencing. Genome Research, 2019, 29, 1134-1143.	5.5	41
25	Molecular basis for phenotypic similarity of genetic disorders. Genome Medicine, 2019, 11, 24.	8.2	1
26	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	2.4	127
27	A higher rare CNV burden in the genetic background potentially contributes to intellectual disability phenotypes in 22q11.2 deletion syndrome. European Journal of Medical Genetics, 2018, 61, 209-212.	1.3	17
28	Pervasive genetic interactions modulate neurodevelopmental defects of the autism-associated 16p11.2 deletion in Drosophila melanogaster. Nature Communications, 2018, 9, 2548.	12.8	56
29	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
30	The joint effect of air pollution exposure and copy number variation on risk for autism. Autism Research, 2017, 10, 1470-1480.	3.8	38
31	Clinical utility gene card for: 16p12.2 microdeletion. European Journal of Human Genetics, 2017, 25, 271-271.	2.8	4
32	Novel metrics to measure coverage in whole exome sequencing datasets reveal local and global non-uniformity. Scientific Reports, 2017, 7, 885.	3.3	43
33	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	1.2	40
34	Mapping a shared genetic basis for neurodevelopmental disorders. Genome Medicine, 2017, 9, 109.	8.2	31
35	Parental Origin of Interstitial Duplications at 15q11.2-q13.3 in Schizophrenia and Neurodevelopmental Disorders. PLoS Genetics, 2016, 12, e1005993.	3.5	51
36	A need for precision medicine to enable tailored special education. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 300-300.	1.7	1

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37	Quantitative Assessment of Eye Phenotypes for Functional Genetic Studies UsingDrosophila melanogaster. G3: Genes, Genomes, Genetics, 2016, 6, 1427-1437.	1.8	67
38	A novel copy number variants kernel association test with application to autism spectrum disorders studies. Bioinformatics, 2016, 32, 3603-3610.	4.1	17
39	Phenobarbital use and neurological problems in FMR1 premutation carriers. NeuroToxicology, 2016, 53, 141-147.	3.0	20
40	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. PLoS Genetics, 2016, 12, e1005963.	3.5	92
41	An assessment of sex bias in neurodevelopmental disorders. Genome Medicine, 2015, 7, 94.	8.2	90
42	Comorbidity of intellectual disability confounds ascertainment of autism: implications for genetic diagnosis. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2015, 168, 600-608.	1.7	92
43	R47H Variant ofTREM2Associated With Alzheimer Disease in a Large Late-Onset Family. JAMA Neurology, 2015, 72, 920.	9.0	122
44	Gene discovery and functional assessment of rare copy-number variants in neurodevelopmental disorders. Briefings in Functional Genomics, 2015, 14, 315-328.	2.7	24
45	Epigenetics of Autism-related Impairment. Journal of Developmental and Behavioral Pediatrics, 2015, 36, 61-67.	1.1	83
46	Improving the Power of Structural Variation Detection by Augmenting the Reference. PLoS ONE, 2015, 10, e0136771.	2.5	6
47	Genetic studies in Drosophila and humans support a model for the concerted function of CISD2, PPT1 and CLN3 in disease. Biology Open, 2014, 3, 342-352.	1.2	10
48	Rare copy number variation in cerebral palsy. European Journal of Human Genetics, 2014, 22, 40-45.	2.8	65
49	Statistical Considerations in the Analysis of Rare Variants. , 2014, , 405-422.		0
50	Genomic disorders: complexity at multiple levels. Genome Medicine, 2013, 5, 43.	8.2	6
51	Support for the N -Methyl-D-Aspartate Receptor Hypofunction Hypothesis of Schizophrenia From Exome Sequencing in Multiplex Families. JAMA Psychiatry, 2013, 70, 582.	11.0	119
52	Counting Chromosomes to Exons: Advances in Copy Number Detection. Current Genetic Medicine Reports, 2013, 1, 71-80.	1.9	1
53	Refinement and Discovery of New Hotspots of Copy-Number Variation Associated with Autism Spectrum Disorder. American Journal of Human Genetics, 2013, 92, 221-237.	6.2	279
54	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

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55	Global increases in both common and rare copy number load associated with autism. Human Molecular Genetics, 2013, 22, 2870-2880.	2.9	56
56	UNCOVERING THE ETIOLOGY OF AUTISM SPECTRUM DISORDERS: GENOMICS, BIOINFORMATICS, ENVIRONMENT, DATA COLLECTION AND EXPLORATION, AND FUTURE POSSIBILITIES. , 2013, , .		3
57	Lessons from Model Organisms: Phenotypic Robustness and Missing Heritability in Complex Disease. PLoS Genetics, 2012, 8, e1003041.	3.5	64
58	A Duplication CNV That Conveys Traits Reciprocal to Metabolic Syndrome and Protects against Diet-Induced Obesity in Mice and Men. PLoS Genetics, 2012, 8, e1002713.	3.5	36
59	Phenotypic Heterogeneity of Genomic Disorders and Rare Copy-Number Variants. New England Journal of Medicine, 2012, 367, 1321-1331.	27.0	519
60	Autosomal Dominant Familial Dyskinesia and Facial Myokymia. Archives of Neurology, 2012, 69, 630.	4.5	109
61	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. Nature, 2012, 485, 246-250.	27.8	1,960
62	A genetic model for neurodevelopmental disease. Current Opinion in Neurobiology, 2012, 22, 829-836.	4.2	47
63	Rare copy number variants in isolated sporadic and syndromic atrioventricular septal defects. American Journal of Medical Genetics, Part A, 2012, 158A, 1279-1284.	1.2	37
64	The genetic variability and commonality of neurodevelopmental disease. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 118-129.	1.6	101
65	De Novo Pathogenic SCN8A Mutation Identified by Whole-Genome Sequencing of a Family Quartet Affected by Infantile Epileptic Encephalopathy and SUDEP. American Journal of Human Genetics, 2012, 90, 502-510.	6.2	365
66	Haploinsufficiency ofSOX5at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. Human Mutation, 2012, 33, 728-740.	2.5	85
67	Evidence for involvement of GNB1L in autism. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 61-71.	1.7	28
68	A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846.	21.4	1,141
69	De Novo CNVs in Bipolar Disorder: Recurrent Themes or New Directions?. Neuron, 2011, 72, 885-887.	8.1	12
70	Human Copy Number Variation and Complex Genetic Disease. Annual Review of Genetics, 2011, 45, 203-226.	7.6	344
71	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. Nature Genetics, 2011, 43, 585-589.	21.4	1,080
72	Genomic architecture of aggression: Rare copy number variants in intermittent explosive disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 808-816.	1.7	11

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73	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. PLoS Genetics, 2011, 7, e1002334.	3.5	293
74	Speech delays and behavioral problems are the predominant features in individuals with developmental delays and 16p11.2 microdeletions and microduplications. Journal of Neurodevelopmental Disorders, 2010, 2, 26-38.	3.1	147
75	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	21.4	539
76	A large and complex structural polymorphism at 16p12.1 underlies microdeletion disease risk. Nature Genetics, 2010, 42, 745-750.	21.4	89
77	Array comparative genomic hybridisation of 52 subjects with a Smith-Magenis-like phenotype: identification of dosage sensitive loci also associated with schizophrenia, autism, and developmental delay. Journal of Medical Genetics, 2010, 47, 223-229.	3.2	26
78	Rai1 haploinsufficiency causes reduced Bdnf expression resulting in hyperphagia, obesity and altered fat distribution in mice and humans with no evidence of metabolic syndrome. Human Molecular Genetics, 2010, 19, 4026-4042.	2.9	95
79	Phenotypic variability and genetic susceptibility to genomic disorders. Human Molecular Genetics, 2010, 19, R176-R187.	2.9	234
80	Sequencing humanâ€“gibbon breakpoints of synteny reveals mosaic new insertions at rearrangement sites. Genome Research, 2009, 19, 178-190.	5.5	29
81	The origins and impact of primate segmental duplications. Trends in Genetics, 2009, 25, 443-454.	6.7	137
82	Parental-age effects in Down syndrome. Journal of Genetics, 2009, 88, 1-7.	0.7	13
83	Abnormal maternal behavior, altered sociability, and impaired serotonin metabolism in Rai1-transgenic mice. Mammalian Genome, 2009, 20, 247-255.	2.2	22
84	A burst of segmental duplications in the genome of the African great ape ancestor. Nature, 2009, 457, 877-881.	27.8	222
85	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. American Journal of Human Genetics, 2009, 84, 148-161.	6.2	530
86	Distorted Mendelian transmission as a function of genetic background in Rai1-haploinsufficient mice. European Journal of Medical Genetics, 2009, 52, 224-228.	1.3	9
87	Tom1l2 hypomorphic mice exhibit increased incidence of infections and tumors and abnormal immunologic response. Mammalian Genome, 2008, 19, 246-262.	2.2	21
88	How much is too much? Phenotypic consequences of Rai1 overexpression in mice. European Journal of Human Genetics, 2008, 16, 941-954.	2.8	47
89	Smithâ€“Magenis syndrome. European Journal of Human Genetics, 2008, 16, 412-421.	2.8	195
90	Diagnosing Smithâ€“Magenis Syndrome and Duplication 17p11.2 Syndrome byRAI1Gene Copy Number Variation Using Quantitative Real-Time PCR. Genetic Testing and Molecular Biomarkers, 2008, 12, 67-73.	1.7	14

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91	Smithâ€“Magenis syndrome and moyamoya disease in a patient with del(17)(p11.2p13.1). American Journal of Medical Genetics, Part A, 2007, 143A, 999-1008.	1.2	19
92	Genotypeâ€“phenotype correlation in Smith-Magenis syndrome: Evidence that multiple genes in 17p11.2 contribute to the clinical spectrum. Genetics in Medicine, 2006, 8, 417-427.	2.4	146
93	RAI1 point mutations, CAG repeat variation, and SNP analysis in non-deletion Smithâ€“Magenis syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2454-2463.	1.2	46
94	Brachydactyly A1: New relatives for old families?. Journal of Genetics, 2005, 84, 95-98.	0.7	3