

Santhosh D Girirajan

List of Publications by Year in descending order

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94
papers

10,795
citations

94433

37
h-index

51608

86
g-index

133
all docs

133
docs citations

133
times ranked

15354
citing authors

#	ARTICLE	IF	CITATIONS
1	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. <i>Nature</i> , 2012, 485, 246-250.	27.8	1,960
2	A copy number variation morbidity map of developmental delay. <i>Nature Genetics</i> , 2011, 43, 838-846.	21.4	1,141
3	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. <i>Nature Genetics</i> , 2011, 43, 585-589.	21.4	1,080
4	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. <i>Nature Genetics</i> , 2010, 42, 203-209.	21.4	539
5	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. <i>American Journal of Human Genetics</i> , 2009, 84, 148-161.	6.2	530
6	Phenotypic Heterogeneity of Genomic Disorders and Rare Copy-Number Variants. <i>New England Journal of Medicine</i> , 2012, 367, 1321-1331.	27.0	519
7	De Novo Pathogenic SCN8A Mutation Identified by Whole-Genome Sequencing of a Family Quartet Affected by Infantile Epileptic Encephalopathy and SUDEP. <i>American Journal of Human Genetics</i> , 2012, 90, 502-510.	6.2	365
8	Human Copy Number Variation and Complex Genetic Disease. <i>Annual Review of Genetics</i> , 2011, 45, 203-226.	7.6	344
9	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. <i>PLoS Genetics</i> , 2011, 7, e1002334.	3.5	293
10	Refinement and Discovery of New Hotspots of Copy-Number Variation Associated with Autism Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2013, 92, 221-237.	6.2	279
11	Phenotypic variability and genetic susceptibility to genomic disorders. <i>Human Molecular Genetics</i> , 2010, 19, R176-R187.	2.9	234
12	A burst of segmental duplications in the genome of the African great ape ancestor. <i>Nature</i> , 2009, 457, 877-881.	27.8	222
13	Smith's Magenis syndrome. <i>European Journal of Human Genetics</i> , 2008, 16, 412-421.	2.8	195
14	Speech delays and behavioral problems are the predominant features in individuals with developmental delays and 16p11.2 microdeletions and microduplications. <i>Journal of Neurodevelopmental Disorders</i> , 2010, 2, 26-38.	3.1	147
15	Genotype-phenotype correlation in Smith-Magenis syndrome: Evidence that multiple genes in 17p11.2 contribute to the clinical spectrum. <i>Genetics in Medicine</i> , 2006, 8, 417-427.	2.4	146
16	The origins and impact of primate segmental duplications. <i>Trends in Genetics</i> , 2009, 25, 443-454.	6.7	137
17	Exonic Deletions in AUTS2 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
18	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. <i>Genetics in Medicine</i> , 2019, 21, 816-825.	2.4	127

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19	R47H Variant of <i>TREM2</i> Associated With Alzheimer Disease in a Large Late-Onset Family. <i>JAMA Neurology</i> , 2015, 72, 920.	9.0	122
20	Support for the N-Methyl-D-Aspartate Receptor Hypofunction Hypothesis of Schizophrenia From Exome Sequencing in Multiplex Families. <i>JAMA Psychiatry</i> , 2013, 70, 582.	11.0	119
21	Autosomal Dominant Familial Dyskinesia and Facial Myokymia. <i>Archives of Neurology</i> , 2012, 69, 630.	4.5	109
22	The genetic variability and commonality of neurodevelopmental disease. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2012, 160C, 118-129.	1.6	101
23	<i>Rai1</i> haploinsufficiency causes reduced <i>Bdnf</i> expression resulting in hyperphagia, obesity and altered fat distribution in mice and humans with no evidence of metabolic syndrome. <i>Human Molecular Genetics</i> , 2010, 19, 4026-4042.	2.9	95
24	Comorbidity of intellectual disability confounds ascertainment of autism: implications for genetic diagnosis. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2015, 168, 600-608.	1.7	92
25	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. <i>PLoS Genetics</i> , 2016, 12, e1005963.	3.5	92
26	An assessment of sex bias in neurodevelopmental disorders. <i>Genome Medicine</i> , 2015, 7, 94.	8.2	90
27	A large and complex structural polymorphism at 16p12.1 underlies microdeletion disease risk. <i>Nature Genetics</i> , 2010, 42, 745-750.	21.4	89
28	Haploinsufficiency of <i>SOX5</i> at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. <i>Human Mutation</i> , 2012, 33, 728-740.	2.5	85
29	Epigenetics of Autism-related Impairment. <i>Journal of Developmental and Behavioral Pediatrics</i> , 2015, 36, 61-67.	1.1	83
30	Quantitative Assessment of Eye Phenotypes for Functional Genetic Studies Using <i>Drosophila melanogaster</i> . <i>G3: Genes, Genomes, Genetics</i> , 2016, 6, 1427-1437.	1.8	67
31	Rare copy number variation in cerebral palsy. <i>European Journal of Human Genetics</i> , 2014, 22, 40-45.	2.8	65
32	Lessons from Model Organisms: Phenotypic Robustness and Missing Heritability in Complex Disease. <i>PLoS Genetics</i> , 2012, 8, e1003041.	3.5	64
33	Global increases in both common and rare copy number load associated with autism. <i>Human Molecular Genetics</i> , 2013, 22, 2870-2880.	2.9	56
34	Pervasive genetic interactions modulate neurodevelopmental defects of the autism-associated 16p11.2 deletion in <i>Drosophila melanogaster</i> . <i>Nature Communications</i> , 2018, 9, 2548.	12.8	56
35	Parental Origin of Interstitial Duplications at 15q11.2-q13.3 in Schizophrenia and Neurodevelopmental Disorders. <i>PLoS Genetics</i> , 2016, 12, e1005993.	3.5	51
36	How much is too much? Phenotypic consequences of <i>Rai1</i> overexpression in mice. <i>European Journal of Human Genetics</i> , 2008, 16, 941-954.	2.8	47

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37	A genetic model for neurodevelopmental disease. <i>Current Opinion in Neurobiology</i> , 2012, 22, 829-836.	4.2	47
38	RAI1 point mutations, CAG repeat variation, and SNP analysis in non-deletion Smith-Magenis syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2454-2463.	1.2	46
39	Novel metrics to measure coverage in whole exome sequencing datasets reveal local and global non-uniformity. <i>Scientific Reports</i> , 2017, 7, 885.	3.3	43
40	A machine-learning approach for accurate detection of copy number variants from exome sequencing. <i>Genome Research</i> , 2019, 29, 1134-1143.	5.5	41
41	Implication of <i>LRRC4C</i> and <i>DPP6</i> in neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 395-406.	1.2	40
42	An interaction-based model for neuropsychiatric features of copy-number variants. <i>PLoS Genetics</i> , 2019, 15, e1007879.	3.5	39
43	The joint effect of air pollution exposure and copy number variation on risk for autism. <i>Autism Research</i> , 2017, 10, 1470-1480.	3.8	38
44	Rare copy number variants in isolated sporadic and syndromic atrioventricular septal defects. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1279-1284.	1.2	37
45	A Duplication CNV That Conveys Traits Reciprocal to Metabolic Syndrome and Protects against Diet-Induced Obesity in Mice and Men. <i>PLoS Genetics</i> , 2012, 8, e1002713.	3.5	36
46	Dissecting the genetic basis of comorbid epilepsy phenotypes in neurodevelopmental disorders. <i>Genome Medicine</i> , 2019, 11, 65.	8.2	35
47	Mapping a shared genetic basis for neurodevelopmental disorders. <i>Genome Medicine</i> , 2017, 9, 109.	8.2	31
48	NCBP2 modulates neurodevelopmental defects of the 3q29 deletion in <i>Drosophila</i> and <i>Xenopus laevis</i> models. <i>PLoS Genetics</i> , 2020, 16, e1008590.	3.5	30
49	Sequencing human-gibbon breakpoints of synteny reveals mosaic new insertions at rearrangement sites. <i>Genome Research</i> , 2009, 19, 178-190.	5.5	29
50	Evidence for involvement of <i>GNB1L</i> in autism. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 61-71.	1.7	28
51	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. <i>Journal of Medical Genetics</i> , 2010, 47, 223-229.	3.2	26
52	Gene discovery and functional assessment of rare copy-number variants in neurodevelopmental disorders. <i>Briefings in Functional Genomics</i> , 2015, 14, 315-328.	2.7	24
53	Abnormal maternal behavior, altered sociability, and impaired serotonin metabolism in <i>Rai1</i> -transgenic mice. <i>Mammalian Genome</i> , 2009, 20, 247-255.	2.2	22
54	<i>Tom1l2</i> hypomorphic mice exhibit increased incidence of infections and tumors and abnormal immunologic response. <i>Mammalian Genome</i> , 2008, 19, 246-262.	2.2	21

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55	Phenobarbital use and neurological problems in FMR1 premutation carriers. <i>NeuroToxicology</i> , 2016, 53, 141-147.	3.0	20
56	Smithâ€™Magenis syndrome and moyamoya disease in a patient with del(17)(p11.2p13.1). <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 999-1008.	1.2	19
57	An evolutionary driver of interspersed segmental duplications in primates. <i>Genome Biology</i> , 2020, 21, 202.	8.8	19
58	A novel copy number variants kernel association test with application to autism spectrum disorders studies. <i>Bioinformatics</i> , 2016, 32, 3603-3610.	4.1	17
59	A higher rare CNV burden in the genetic background potentially contributes to intellectual disability phenotypes in 22q11.2 deletion syndrome. <i>European Journal of Medical Genetics</i> , 2018, 61, 209-212.	1.3	17
60	Diagnosing Smithâ€™Magenis Syndrome and Duplication 17p11.2 Syndrome byRAI1Gene Copy Number Variation Using Quantitative Real-Time PCR. <i>Genetic Testing and Molecular Biomarkers</i> , 2008, 12, 67-73.	1.7	14
61	Parental-age effects in Down syndrome. <i>Journal of Genetics</i> , 2009, 88, 1-7.	0.7	13
62	De Novo CNVs in Bipolar Disorder: Recurrent Themes or New Directions?. <i>Neuron</i> , 2011, 72, 885-887.	8.1	12
63	Gene discoveries in autism are biased towards comorbidity with intellectual disability. <i>Journal of Medical Genetics</i> , 2020, 57, 647-652.	3.2	12
64	Functional assessment of the â€œtwo-hitâ€•model for neurodevelopmental defects in <i>Drosophila</i> and <i>X. laevis</i> . <i>PLoS Genetics</i> , 2021, 17, e1009112.	3.5	12
65	Genomic architecture of aggression: Rare copy number variants in intermittent explosive disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2011, 156, 808-816.	1.7	11
66	Genetic studies in <i>Drosophila</i> and humans support a model for the concerted function of <i>CISD2</i> , <i>PPT1</i> and <i>CLN3</i> in disease. <i>Biology Open</i> , 2014, 3, 342-352.	1.2	10
67	Distorted Mendelian transmission as a function of genetic background in <i>Rai1</i> -haploinsufficient mice. <i>European Journal of Medical Genetics</i> , 2009, 52, 224-228.	1.3	9
68	<i>Drosophila</i> models of pathogenic copy-number variant genes show global and non-neuronal defects during development. <i>PLoS Genetics</i> , 2020, 16, e1008792.	3.5	9
69	The chromatin remodeler ISWI acts during <i>Drosophila</i> development to regulate adult sleep. <i>Science Advances</i> , 2021, 7, .	10.3	9
70	<sc>Birtâ€™Hoggâ€™DubÃ©</sc> symptoms in <sc>Smithâ€™Magenis</sc> syndrome include pediatricâ€™onset pneumothorax. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1922-1924.	1.2	7
71	A general framework for identifying oligogenic combinations of rare variants in complex disorders. <i>Genome Research</i> , 2022, , .	5.5	7
72	Genomic disorders: complexity at multiple levels. <i>Genome Medicine</i> , 2013, 5, 43.	8.2	6

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73	Improving the Power of Structural Variation Detection by Augmenting the Reference. PLoS ONE, 2015, 10, e0136771.	2.5	6
74	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
75	Macrophage Selenoproteins Restrict Intracellular Replication of Francisella tularensis and Are Essential for Host Immunity. Frontiers in Immunology, 2021, 12, 701341.	4.8	5
76	Clinical utility gene card for: 16p12.2 microdeletion. European Journal of Human Genetics, 2017, 25, 271-271.	2.8	4
77	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
78	Brachydactyly A1: New relatives for old families?. Journal of Genetics, 2005, 84, 95-98.	0.7	3
79	UNCOVERING THE ETIOLOGY OF AUTISM SPECTRUM DISORDERS: GENOMICS, BIOINFORMATICS, ENVIRONMENT, DATA COLLECTION AND EXPLORATION, AND FUTURE POSSIBILITIES. , 2013, , .		3
80	Counting Chromosomes to Exons: Advances in Copy Number Detection. Current Genetic Medicine Reports, 2013, 1, 71-80.	1.9	1
81	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
82	Molecular basis for phenotypic similarity of genetic disorders. Genome Medicine, 2019, 11, 24.	8.2	1
83	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
84	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
85	Genetic subtypes, allelic effects, and convergent neurodevelopmental mechanisms. Genome Medicine, 2021, 13, 99.	8.2	0
86	Statistical Considerations in the Analysis of Rare Variants. , 2014, , 405-422.		0
87	Title is missing!. , 2020, 16, e1008792.		0
88	Title is missing!. , 2020, 16, e1008792.		0
89	Title is missing!. , 2020, 16, e1008792.		0
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91	Title is missing!. , 2020, 16, e1008590.		0
92	Title is missing!. , 2020, 16, e1008590.		0
93	Title is missing!.. , 2020, 16, e1008590.		0
94	Title is missing!.. , 2020, 16, e1008590.		0