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List of Publications by Year
in descending order

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51 papers	8,984 citations	145106 33 h-index	242451 47 g-index
51 all docs	51 docs citations	51 times ranked	15576 citing authors

#	Article	IF	Citations
1	Sex Differences in Anemia and Red Blood Cell Transfusions Among General Internal Medicine Inpatients. Blood, 2021, 138, 1074-1074.	0.6	0
2	Improved diagnostic yield compared with targeted gene sequencing panels suggests a role for whole-genome sequencing as a first-tier genetic test. Genetics in Medicine, 2018, 20, 435-443.	1.1	404
3	Complete Disruption of Autism-Susceptibility Genes by Gene Editing Predominantly Reduces Functional Connectivity of Isogenic Human Neurons. Stem Cell Reports, 2018, 11, 1211-1225.	2.3	111
4	Molecular characterization of NRXN1 deletions from 19,263 clinical microarray cases identifies exons important for neurodevelopmental disease expression. Genetics in Medicine, 2017, 19, 53-61.	1.1	70
5	De novo pathogenic variant in TUBB2A presenting with arthrogryposis multiplex congenita, brain abnormalities, and severe developmental delay. , 2017, 173, 2725-2730.		15
6	Impact of IQ on the diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. Genome Medicine, 2017, 9, 105.	3.6	30
7	MED23â€associated refractory epilepsy successfully treated with the ketogenic diet. American Journal of Medical Genetics, Part A, 2016, 170, 2421-2425.	0.7	21
8	Copy-number variations are enriched for neurodevelopmental genes in children with developmental coordination disorder. Journal of Medical Genetics, 2016, 53, 812-819.	1.5	40
9	Genome-wide rare copy number variations contribute to genetic risk for transposition of the great arteries. International Journal of Cardiology, 2016, 204, 115-121.	0.8	26
10	15q11.2 Duplication Encompassing Only theUBE3AGene Is Associated with Developmental Delay and Neuropsychiatric Phenotypes. Human Mutation, 2015, 36, 689-693.	1.1	67
11	Performance of case-control rare copy number variation annotation in classification of autism. BMC Medical Genomics, 2015, 8, S7.	0.7	15
12	MG-123â€...Exonic and intronic NRXN1 deletions: Novel genotype-phenotype correlations. Journal of Medical Genetics, 2015, 52, A9.1-A9.	1.5	0
13	Single cell-derived clonal analysis of human glioblastoma links functional and genomic heterogeneity. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 851-856.	3.3	321
14	A high-resolution copy-number variation resource for clinical and population genetics. Genetics in Medicine, 2015, 17, 747-752.	1.1	73
15	Molecular Diagnostic Yield of Chromosomal Microarray Analysis and Whole-Exome Sequencing in Children With Autism Spectrum Disorder. JAMA - Journal of the American Medical Association, 2015, 314, 895.	3.8	352
16	ISDN2014_0253: High resolution genomic analyses of a clinically defined autism spectrum disorder cohort. International Journal of Developmental Neuroscience, 2015, 47, 76-76.	0.7	2
17	Using extended pedigrees to identify novel autism spectrum disorder (ASD) candidate genes. Human Genetics, 2015, 134, 191-201.	1.8	20
18	Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature. Genetics in Medicine, 2015, 17, 149-157.	1.1	103

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19	Novel Population Specific Autosomal Copy Number Variation and Its Functional Analysis amongst Negritos from Peninsular Malaysia. PLoS ONE, 2014, 9, e100371.	1.1	6
20	Copy number variant study of bipolar disorder in Canadian and UK populations implicates synaptic genes. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 303-313.	1.1	76
21	Identification of risk genes for autism spectrum disorder through copy number variation analysis in Austrian families. Neurogenetics, 2014, 15, 117-127.	0.7	98
22	Outfoxed by RBFOX1â€A caution about ascertainment bias. American Journal of Medical Genetics, Part A, 2014, 164, 1411-1418.	0.7	9
23	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Human Molecular Genetics, 2014, 23, 2752-2768.	1.4	140
24	Adult neuropsychiatric expression and familial segregation of 2q13 duplications. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 337-344.	1.1	23
25	Copy number variation in Han Chinese individuals with autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2014, 6, 34.	1.5	55
26	CHD2 haploinsufficiency is associated with developmental delay, intellectual disability, epilepsy and neurobehavioural problems. Journal of Neurodevelopmental Disorders, 2014, 6, 9.	1.5	71
27	Adult expression of a 3q13.31 microdeletion. Molecular Cytogenetics, 2014, 7, 23.	0.4	10
28	Rare exonic deletions implicate the synaptic organizer Gephyrin (GPHN) in risk for autism, schizophrenia and seizures. Human Molecular Genetics, 2013, 22, 2055-2066.	1.4	139
29	Pathogenic rare copy number variants in community-based schizophrenia suggest a potential role for clinical microarrays. Human Molecular Genetics, 2013, 22, 4485-4501.	1.4	120
30	Copy Number Variation in Autism Spectrum Disorders. , 2013, , 145-154.		1
31	1q21.1 Microduplication expression in adults. Genetics in Medicine, 2013, 15, 282-289.	1.1	91
32	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.	2.6	135
33	Network Topologies and Convergent Aetiologies Arising from Deletions and Duplications Observed in Individuals with Autism. PLoS Genetics, 2013, 9, e1003523.	1.5	51
34	Deletions in 16q24.2 are associated with autism spectrum disorder, intellectual disability and congenital renal malformation. Journal of Medical Genetics, 2013, 50, 163-173.	1.5	36
35	A Discovery Resource of Rare Copy Number Variations in Individuals with Autism Spectrum Disorder. G3: Genes, Genomes, Genetics, 2012, 2, 1665-1685.	0.8	175
36	Rare Copy Number Variations in Adults with Tetralogy of Fallot Implicate Novel Risk Gene Pathways. PLoS Genetics, 2012, 8, e1002843.	1.5	149

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37	Severe intellectual disability and autistic features associated with microduplication 2q23.1. European Journal of Human Genetics, 2012, 20, 398-403.	1.4	31
38	Genome-Wide Analysis of Copy Number Variants in Attention Deficit Hyperactivity Disorder: The Role of Rare Variants and Duplications at 15q13.3. American Journal of Psychiatry, 2012, 169, 195-204.	4.0	242
39	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	3.3	118
40	Identification of germline genomic copy number variation in familial pancreatic cancer. Human Genetics, 2012, 131, 1481-1494.	1.8	25
41	Rare Deletions at the Neurexin 3 Locus in Autism Spectrum Disorder. American Journal of Human Genetics, 2012, 90, 133-141.	2.6	182
42	SHANK1 Deletions in Males with Autism Spectrum Disorder. American Journal of Human Genetics, 2012, 90, 879-887.	2.6	292
43	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. Nature Biotechnology, 2011, 29, 512-520.	9.4	384
44	A genotype resource for postmortem brain samples from the Autism Tissue Program. Autism Research, 2011, 4, 89-97.	2.1	23
45	Human PTCHD3 nulls: rare copy number and sequence variants suggest a non-essential gene. BMC Medical Genetics, 2011, 12, 45.	2.1	13
46	Rare Copy Number Variation Discovery and Cross-Disorder Comparisons Identify Risk Genes for ADHD. Science Translational Medicine, 2011, 3, 95ra75.	5.8	304
47	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	13.7	1,803
48	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	1.4	538
49	Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. Science Translational Medicine, 2010, 2, 49ra68.	5.8	178
50	Structural Variation of Chromosomes in Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 477-488.	2.6	1,641
51	Copy number variations and risk for schizophrenia in 22q11.2 deletion syndrome. Human Molecular Genetics, 2008, 17, 4045-4053.	1.4	155