Brian O'Roak

List of Publications by Year
in descending order

56 papers	22,907 citations	76031 42 h-index	169272 56 g-index
67 all docs	67 docs citations	67 times ranked	39483 citing authors

#	Article	IF	Citations
1	Validation of Autism Diagnosis and Clinical Data in the SPARK Cohort. Journal of Autism and Developmental Disorders, 2022, 52, 3383-3398.	1.7	41
2	Cas12a-Capture: A Novel, Low-Cost, and Scalable Method for Targeted Sequencing. CRISPR Journal, 2022, 5, 548-557.	1.4	0
3	Autism and attentionâ€deficit/hyperactivity disorders and symptoms in children with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2021, 63, 226-232.	1.1	17
4	Autism questionnaire scores do not only rise because of autism. Developmental Medicine and Child Neurology, 2021, 63, 235-236.	1.1	6
5	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	9.4	68
6	High-content single-cell combinatorial indexing. Nature Biotechnology, 2021, 39, 1574-1580.	9.4	39
7	Neutralization of SARS-CoV-2 variants by convalescent and BNT162b2 vaccinated serum. Nature Communications, 2021, 12, 5135.	5.8	107
8	Clinical Phenotypes of Carriers of Mutations in CHD8 or Its Conserved Target Genes. Biological Psychiatry, 2020, 87, 123-131.	0.7	22
9	Beliefs in vaccine as causes of autism among SPARK cohort caregivers. Vaccine, 2020, 38, 1794-1803.	1.7	12
10	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. Journal of Medical Genetics, 2020, 57, 466-474.	1.5	7
11	An Integrated Deep-Mutational-Scanning Approach Provides Clinical Insights on PTEN Genotype-Phenotype Relationships. American Journal of Human Genetics, 2020, 106, 818-829.	2.6	38
12	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
13	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	1.7	163
14	Highly scalable generation of DNA methylation profiles in single cells. Nature Biotechnology, 2018, 36, 428-431.	9.4	215
15	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493.	3.8	265
16	A Saturation Mutagenesis Approach to Understanding PTEN Lipid Phosphatase Activity and Genotype-Phenotype Relationships. American Journal of Human Genetics, 2018, 102, 943-955.	2.6	149
17	Exonic Mosaic Mutations Contribute Risk for Autism Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 369-390.	2.6	151
18	Current Perspectives in Autism Spectrum Disorder: From Genes to Therapy. Journal of Neuroscience, 2016, 36, 11402-11410.	1.7	44

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19	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214.	1.5	39
20	Mutations and Modeling of the Chromatin Remodeler CHD8 Define an Emerging Autism Etiology. Frontiers in Neuroscience, 2015, 9, 477.	1.4	90
21	Genome-wide association study and admixture mapping reveal new loci associated with total IgE levels in Latinos. Journal of Allergy and Clinical Immunology, 2015, 135, 1502-1510.	1.5	52
22	PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. Brain, 2015, 138, 1613-1628.	3.7	286
23	Joubert syndrome: a model for untangling recessive disorders with extreme genetic heterogeneity. Journal of Medical Genetics, 2015, 52, 514-522.	1.5	219
24	Whole-Genome Sequencing of Individuals from a Founder Population Identifies Candidate Genes for Asthma. PLoS ONE, 2014, 9, e104396.	1.1	42
25	Mutations in CSPP1 Cause Primary Cilia Abnormalities and Joubert Syndrome with or without Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2014, 94, 62-72.	2.6	104
26	Rare-Variant Extensions of the Transmission Disequilibrium Test: Application to Autism Exome Sequence Data. American Journal of Human Genetics, 2014, 94, 33-46.	2.6	69
27	A de novo convergence of autism genetics and molecular neuroscience. Trends in Neurosciences, 2014, 37, 95-105.	4.2	410
28	A general framework for estimating the relative pathogenicity of human genetic variants. Nature Genetics, 2014, 46, 310-315.	9.4	5,167
29	De novo TBR1 mutations in sporadic autism disrupt protein functions. Nature Communications, 2014, 5, 4954.	5.8	109
30	The contribution of de novo coding mutations to autism spectrum disorder. Nature, 2014, 515, 216-221.	13.7	2,188
31	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
32	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
33	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	13.5	637
34	MIPgen: optimized modeling and design of molecular inversion probes for targeted resequencing. Bioinformatics, 2014, 30, 2670-2672.	1.8	138
35	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	9.4	326
36	Transmission Disequilibrium of Small CNVs in Simplex Autism. American Journal of Human Genetics, 2013, 93, 595-606.	2.6	87

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37	Single molecule molecular inversion probes for targeted, high-accuracy detection of low-frequency variation. Genome Research, 2013, 23, 843-854.	2.4	292
38	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
39	Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. Nature Methods, 2013, 10, 903-909.	9.0	31
40	Detection of structural variants and indels within exome data. Nature Methods, 2012, 9, 176-178.	9.0	109
41	Copy number variation detection and genotyping from exome sequence data. Genome Research, 2012, 22, 1525-1532.	2.4	550
42	Multiplex Targeted Sequencing Identifies Recurrently Mutated Genes in Autism Spectrum Disorders. Science, 2012, 338, 1619-1622.	6.0	1,133
43	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. Nature, 2012, 485, 246-250.	13.7	1,960
44	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	9.4	237
45	Estimating the human mutation rate using autozygosity in a founder population. Nature Genetics, 2012, 44, 1277-1281.	9.4	202
46	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	9.4	621
47	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
48	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. Nature Genetics, 2011, 43, 585-589.	9.4	1,080
49	Additional support for the association of SLITRK1 var321 and Tourette syndrome. Molecular Psychiatry, 2010, 15, 447-450.	4.1	55
50	L-Histidine Decarboxylase and Tourette's Syndrome. New England Journal of Medicine, 2010, 362, 1901-1908.	13.9	304
51	Haplotype evolution of SLITRK1, a candidate gene for Gilles de la Tourette Syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 463-466.	1.1	16
52	Autism genetics: strategies, challenges, and opportunities. Autism Research, 2008, 1, 4-17.	2.1	123
53	Rare independent mutations in renal salt handling genes contribute to blood pressure variation. Nature Genetics, 2008, 40, 592-599.	9.4	728
54	Molecular Cytogenetic Analysis and Resequencing of Contactin Associated Protein-Like 2 in Autism Spectrum Disorders. American Journal of Human Genetics, 2008, 82, 165-173.	2.6	494

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55	Mutations in Apoptosis-related Gene, PDCD10, Cause Cerebral Cavernous Malformation 3. Neurosurgery, 2005, 57, 1008-1013.	0.6	91
56	Sequence Variants in SLITRK1 Are Associated with Tourette's Syndrome. Science, 2005, 310, 317-320.	6.0	878