

Elena Bacchelli

List of Publications by Year in descending order

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Version: 2024-02-01

44
papers

8,235
citations

185998

28
h-index

233125

45
g-index

45
all docs

45
docs citations

45
times ranked

12765
citing authors

#	ARTICLE	IF	CITATIONS
1	Functional impact of global rare copy number variation in autism spectrum disorders. <i>Nature</i> , 2010, 466, 368-372.	13.7	1,803
2	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. <i>Nature Genetics</i> , 2007, 39, 319-328.	9.4	1,272
3	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	6.0	1,085
4	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2014, 94, 677-694.	2.6	819
5	A genome-wide scan for common alleles affecting risk for autism. <i>Human Molecular Genetics</i> , 2010, 19, 4072-4082.	1.4	538
6	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. <i>Nature Genetics</i> , 2017, 49, 978-985.	9.4	401
7	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. <i>PLoS Genetics</i> , 2012, 8, e1002521.	1.5	358
8	Individual common variants exert weak effects on the risk for autism spectrum disorders. <i>Human Molecular Genetics</i> , 2012, 21, 4781-4792.	1.4	334
9	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. <i>Human Genetics</i> , 2012, 131, 565-579.	1.8	180
10	Characterization of a Family with Rare Deletions in CNTNAP5 and DOCK4 Suggests Novel Risk Loci for Autism and Dyslexia. <i>Biological Psychiatry</i> , 2010, 68, 320-328.	0.7	131
11	High-density SNP association study and copy number variation analysis of the AUTS1 and AUTS5 loci implicate the IMMP2L-DOCK4 gene region in autism susceptibility. <i>Molecular Psychiatry</i> , 2010, 15, 954-968.	4.1	126
12	Analysis of IMGSAC autism susceptibility loci: evidence for sex limited and parent of origin specific effects. <i>Journal of Medical Genetics</i> , 2005, 42, 132-137.	1.5	114
13	Screening of nine candidate genes for autism on chromosome 2q reveals rare nonsynonymous variants in the cAMP-GEFII gene. <i>Molecular Psychiatry</i> , 2003, 8, 916-924.	4.1	108
14	Absence of coding mutations in the X-linked genes neuroligin 3 and neuroligin 4 in individuals with autism from the IMGSAC collection. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2006, 141B, 220-221.	1.1	89
15	Mutation analysis of the coding sequence of the MECP2 gene in infantile autism. <i>Human Genetics</i> , 2002, 111, 305-309.	1.8	82
16	Linkage and candidate gene studies of autism spectrum disorders in European populations. <i>European Journal of Human Genetics</i> , 2010, 18, 1013-1019.	1.4	80
17	Mutation screening and association analysis of six candidate genes for autism on chromosome 7q. <i>European Journal of Human Genetics</i> , 2005, 13, 198-207.	1.4	74
18	Autism spectrum disorders: Molecular genetic advances. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2006, 142C, 13-23.	0.7	51

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19	SLC25A12 and CMYA3 gene variants are not associated with autism in the IMGSAC multiplex family sample. <i>European Journal of Human Genetics</i> , 2006, 14, 123-126.	1.4	44
20	Is ASMT a susceptibility gene for autism spectrum disorders? A replication study in European populations. <i>Molecular Psychiatry</i> , 2007, 12, 977-979.	4.1	42
21	Analysis of X chromosome inactivation in autism spectrum disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2008, 147B, 830-835.	1.1	42
22	Maternally inherited genetic variants of CADPS 2 are present in Autism Spectrum Disorders and Intellectual Disability patients. <i>EMBO Molecular Medicine</i> , 2014, 6, 795-809.	3.3	42
23	An integrated analysis of rare CNV and exome variation in Autism Spectrum Disorder using the Infinium PsychArray. <i>Scientific Reports</i> , 2020, 10, 3198.	1.6	42
24	Analysis of <i>CHRNA7</i> rare variants in autism spectrum disorder susceptibility. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 715-723.	0.7	41
25	Mutation screening and imprinting analysis of four candidate genes for autism in the 7q32 region. <i>Molecular Psychiatry</i> , 2002, 7, 289-301.	4.1	38
26	A genome-wide analysis in cluster headache points to neprilysin and PACAP receptor gene variants. <i>Journal of Headache and Pain</i> , 2016, 17, 114.	2.5	38
27	A CTNNA3 compound heterozygous deletion implicates a role for β -catenin in susceptibility to autism spectrum disorder. <i>Journal of Neurodevelopmental Disorders</i> , 2014, 6, 17.	1.5	37
28	Lack of replication of previous autism spectrum disorder GWAS hits in European populations. <i>Autism Research</i> , 2017, 10, 202-211.	2.1	34
29	Homozygous microdeletion of exon 5 in ZNF277 in a girl with specific language impairment. <i>European Journal of Human Genetics</i> , 2014, 22, 1165-1171.	1.4	27
30	Common and rare variants of microRNA genes in autism spectrum disorders. <i>World Journal of Biological Psychiatry</i> , 2015, 16, 376-386.	1.3	27
31	Mapping of partially overlapping de novo deletions across an autism susceptibility region (<i>AUTS5</i>) in two unrelated individuals affected by developmental delays with communication impairment. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 588-597.	0.7	21
32	Integrated DNA methylation analysis identifies topographical and tumoral biomarkers in pilocytic astrocytomas. <i>Oncotarget</i> , 2018, 9, 13807-13821.	0.8	18
33	Analysis of a Sardinian Multiplex Family with Autism Spectrum Disorder Points to Post-Synaptic Density Gene Variants and Identifies CAPG as a Functionally Relevant Candidate Gene. <i>Journal of Clinical Medicine</i> , 2019, 8, 212.	1.0	17
34	Contribution of ultrarare variants in mTOR pathway genes to sporadic focal epilepsies. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 475-485.	1.7	15
35	Genetic variation in <i>CHRNA7</i> and <i>CHRFAM7A</i> is associated with nicotine dependence and response to varenicline treatment. <i>European Journal of Human Genetics</i> , 2018, 26, 1824-1831.	1.4	13
36	ELMOD3- <i>SH2D6</i> gene fusion as a possible co-star actor in autism spectrum disorder scenario. <i>Journal of Cellular and Molecular Medicine</i> , 2020, 24, 2064-2069.	1.6	12

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37	Brain Magnetic Resonance Findings in 117 Children with Autism Spectrum Disorder under 5 Years Old. <i>Brain Sciences</i> , 2020, 10, 741.	1.1	10
38	Maternally inherited genetic variants of CADPS 2 are present in Autism Spectrum Disorders and Intellectual Disability patients. <i>EMBO Molecular Medicine</i> , 2014, 6, 1639-1639.	3.3	9
39	OO15. Evaluation of the genetic polymorphism of the $\hat{1}\pm 3$ (CHRNA3) and $\hat{1}\pm 5$ (CHRNA5) nicotinic receptor subunits, in patients with cluster headache. <i>Journal of Headache and Pain</i> , 2015, 16, A88.	2.5	4
40	Cytogenetic and molecular characterization of a recombinant X chromosome in a family with a severe neurologic phenotype and macular degeneration. <i>Molecular Cytogenetics</i> , 2015, 8, 58.	0.4	4
41	Contribution of CACNA1H Variants in Autism Spectrum Disorder Susceptibility. <i>Frontiers in Psychiatry</i> , 2022, 13, 858238.	1.3	4
42	An increased burden of rare exonic variants in NRXN1 microdeletion carriers is likely to enhance the penetrance for autism spectrum disorder. <i>Journal of Cellular and Molecular Medicine</i> , 2021, 25, 2459-2470.	1.6	3
43	Reply to Pembrey et al: $\hat{a}\hat{c}$ ZNF277 microdeletions, specific language impairment and the meiotic mismatch methylation (3M) hypothesis $\hat{a}\hat{c}$ ™. <i>European Journal of Human Genetics</i> , 2015, 23, 1113-1115.	1.4	2
44	The role of rare compound heterozygous events in autism spectrum disorder. <i>Translational Psychiatry</i> , 2020, 10, 204.	2.4	2