Elena Bacchelli

List of Publications by Year
in descending order

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44	8,235	186265	233421
papers	citations	28 h-index	45 g-index

45	45	45	12765
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
2	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
3	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
4	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	6.2	819
5	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	2.9	538
6	Polygenic transmission disequilibrium confirms that common and rare variation act additively to create risk for autism spectrum disorders. Nature Genetics, 2017, 49, 978-985.	21.4	401
7	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. PLoS Genetics, 2012, 8, e1002521.	3.5	358
8	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	2.9	334
9	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	3.8	180
10	Characterization of a Family with Rare Deletions in CNTNAP5 and DOCK4 Suggests Novel Risk Loci for Autism and Dyslexia. Biological Psychiatry, 2010, 68, 320-328.	1.3	131
11	High-density SNP association study and copy number variation analysis of the AUTS1 and AUTS5 loci implicate the IMMP2Lâ€“DOCK4 gene region in autism susceptibility. Molecular Psychiatry, 2010, 15, 954-968.	7.9	126
12	Analysis of IMGSAC autism susceptibility loci: evidence for sex limited and parent of origin specific effects. Journal of Medical Genetics, 2005, 42, 132-137.	3.2	114
13	Screening of nine candidate genes for autism on chromosome 2q reveals rare nonsynonymous variants in the cAMP-GEFII gene. Molecular Psychiatry, 2003, 8, 916-924.	7.9	108
14	Absence of coding mutations in the X-linked genes neuroligin 3 and neuroligin 4 in individuals with autism from the IMGSAC collection. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 220-221.	1.7	89
15	Mutation analysis of the coding sequence of the MECP2 gene in infantile autism. Human Genetics, 2002, 111, 305-309.	3.8	82
16	Linkage and candidate gene studies of autism spectrum disorders in European populations. European Journal of Human Genetics, 2010, 18, 1013-1019.	2.8	80
17	Mutation screening and association analysis of six candidate genes for autism on chromosome 7q. European Journal of Human Genetics, 2005, 13, 198-207.	2.8	74
18	Autism spectrum disorders: Molecular genetic advances. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2006, 142C, 13-23.	1.6	51

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19	SLC25A12 and CMYA3 gene variants are not associated with autism in the IMGSAC multiplex family sample. European Journal of Human Genetics, 2006, 14, 123-126.	2.8	44
20	Is ASMT a susceptibility gene for autism spectrum disorders? A replication study in European populations. Molecular Psychiatry, 2007, 12, 977-979.	7.9	42
21	Analysis of X chromosome inactivation in autism spectrum disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 830-835.	1.7	42
22	Maternally inherited genetic variants of CADPS 2 are present in Autism Spectrum Disorders and Intellectual Disability patients. EMBO Molecular Medicine, 2014, 6, 795-809.	6.9	42
23	An integrated analysis of rare CNV and exome variation in Autism Spectrum Disorder using the Infinium PsychArray. Scientific Reports, 2020, 10, 3198.	3.3	42
24	Analysis of CHRNA7 rare variants in autism spectrum disorder susceptibility. American Journal of Medical Genetics, Part A, 2015, 167, 715-723.	1.2	41
25	Mutation screening and imprinting analysis of four candidate genes for autism in the 7q32 region. Molecular Psychiatry, 2002, 7, 289-301.	7.9	38
26	A genome-wide analysis in cluster headache points to neprilysin and PACAP receptor gene variants. Journal of Headache and Pain, 2016, 17, 114.	6.0	38
27	A CTNNA3 compound heterozygous deletion implicates a role for Î±T-catenin in susceptibility to autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2014, 6, 17.	3.1	37
28	Lack of replication of previous autism spectrum disorder GWAS hits in European populations. Autism Research, 2017, 10, 202-211.	3.8	34
29	Homozygous microdeletion of exon 5 in ZNF277 in a girl with specific language impairment. European Journal of Human Genetics, 2014, 22, 1165-1171.	2.8	27
30	Common and rare variants of microRNA genes in autism spectrum disorders. World Journal of Biological Psychiatry, 2015, 16, 376-386.	2.6	27
31	Mapping of partially overlapping de novo deletions across an autism susceptibility region (AUTS5) in two unrelated individuals affected by developmental delays with communication impairment. American Journal of Medical Genetics, Part A, 2009, 149A, 588-597.	1.2	21
32	Integrated DNA methylation analysis identifies topographical and tumoral biomarkers in pilocytic astrocytomas. Oncotarget, 2018, 9, 13807-13821.	1.8	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. Journal of Clinical Medicine, 2019, 8, 212.	2.4	17
34	Contribution of ultrarare variants in mTOR pathway genes to sporadic focal epilepsies. Annals of Clinical and Translational Neurology, 2019, 6, 475-485.	3.7	15
35	Genetic variation in CHRNA7 and CHRFAM7A is associated with nicotine dependence and response to varenicline treatment. European Journal of Human Genetics, 2018, 26, 1824-1831.	2.8	13
36	ELMOD3 â€•SH2D6 gene fusion as a possible coâ€€star actor in autism spectrum disorder scenario. Journal of Cellular and Molecular Medicine, 2020, 24, 2064-2069.	3.6	12

#	Article	IF	Citations
37	Brain Magnetic Resonance Findings in 117 Children with Autism Spectrum Disorder under 5 Years Old. Brain Sciences, 2020, 10, 741.	2.3	10
38	Maternally inherited genetic variants of CADPS 2 are present in Autism Spectrum Disorders and Intellectual Disability patients. EMBO Molecular Medicine, 2014, 6, 1639-1639.	6.9	9
39	O015. Evaluation of the genetic polymorphism of the Î±3 (CHRNA3) and Î±5 (CHRNA5) nicotinic receptor subunits, in patients with cluster headache. Journal of Headache and Pain, 2015, 16, A88.	6.0	4
40	Cytogenetic and molecular characterization of a recombinant X chromosome in a family with a severe neurologic phenotype and macular degeneration. Molecular Cytogenetics, 2015, 8, 58.	0.9	4
41	Contribution of CACNA1H Variants in Autism Spectrum Disorder Susceptibility. Frontiers in Psychiatry, 2022, 13, 858238.	2.6	4
42	An increased burden of rare exonic variants in NRXN1 microdeletion carriers is likely to enhance the penetrance for autism spectrum disorder. Journal of Cellular and Molecular Medicine, 2021, 25, 2459-2470.	3.6	3
43	Reply to Pembrey et al: â€˜ZNF277 microdeletions, specific language impairment and the meiotic mismatch methylation (3M) hypothesisâ€™. European Journal of Human Genetics, 2015, 23, 1113-1115.	2.8	2
44	The role of rare compound heterozygous events in autism spectrum disorder. Translational Psychiatry, 2020, 10, 204.	4.8	2