

Elena Bacchelli

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/1821256/publications.pdf>

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	Contribution of CACNA1H Variants in Autism Spectrum Disorder Susceptibility. <i>Frontiers in Psychiatry</i> , 2022, 13, 858238.	1.3	4
2	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
3	ELMOD3 \leftrightarrow SH2D6 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
4	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
5	The role of rare compound heterozygous events in autism spectrum disorder. <i>Translational Psychiatry</i> , 2020, 10, 204.	2.4	2
6	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
7	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
8	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
9	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	6.0	1,085
10	Genetic variation in CHRNA7 and CHRFAM7A is associated with nicotine dependence and response to varenicline treatment. <i>European Journal of Human Genetics</i> , 2018, 26, 1824-1831.	1.4	13
11	Integrated DNA methylation analysis identifies topographical and tumoral biomarkers in pilocytic astrocytomas. <i>Oncotarget</i> , 2018, 9, 13807-13821.	0.8	18
12	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
13	Lack of replication of previous autism spectrum disorder GWAS hits in European populations. <i>Autism Research</i> , 2017, 10, 202-211.	2.1	34
14	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
15	OO15. Evaluation of the genetic polymorphism of the α 3 (CHRNA3) and α 5 (CHRNA5) nicotinic receptor subunits, in patients with cluster headache. <i>Journal of Headache and Pain</i> , 2015, 16, A88.	2.5	4
16	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
17	Reply to Pembrey et al: α ZNF277 microdeletions, specific language impairment and the meiotic mismatch methylation (3M) hypothesis TM . <i>European Journal of Human Genetics</i> , 2015, 23, 1113-1115.	1.4	2
18	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

#	ARTICLE	IF	CITATIONS
19	Analysis of <i>CHRNA7</i> rare variants in autism spectrum disorder susceptibility. American Journal of Medical Genetics, Part A, 2015, 167, 715-723.	0.7	41
20	Homozygous microdeletion of exon 5 in ZNF277 in a girl with specific language impairment. European Journal of Human Genetics, 2014, 22, 1165-1171.	1.4	27
21	Maternally inherited genetic variants of CADPS 2 are present in Autism Spectrum Disorders and Intellectual Disability patients. EMBO Molecular Medicine, 2014, 6, 795-809.	3.3	42
22	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	2.6	819
23	A CTNNA3 compound heterozygous deletion implicates a role for β -catenin in susceptibility to autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2014, 6, 17.	1.5	37
24	Maternally inherited genetic variants of CADPS 2 are present in Autism Spectrum Disorders and Intellectual Disability patients. EMBO Molecular Medicine, 2014, 6, 1639-1639.	3.3	9
25	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. PLoS Genetics, 2012, 8, e1002521.	1.5	358
26	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	1.4	334
27	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	1.8	180
28	Linkage and candidate gene studies of autism spectrum disorders in European populations. European Journal of Human Genetics, 2010, 18, 1013-1019.	1.4	80
29	High-density SNP association study and copy number variation analysis of the AUTS1 and AUTS5 loci implicate the <i>IMMP2L</i> - <i>DOCK4</i> gene region in autism susceptibility. Molecular Psychiatry, 2010, 15, 954-968.	4.1	126
30	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	13.7	1,803
31	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	1.4	538
32	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.	0.7	131
33	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. American Journal of Medical Genetics, Part A, 2009, 149A, 588-597.	0.7	21
34	Analysis of X chromosome inactivation in autism spectrum disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 830-835.	1.1	42
35	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	9.4	1,272
36	Is ASMT a susceptibility gene for autism spectrum disorders? A replication study in European populations. Molecular Psychiatry, 2007, 12, 977-979.	4.1	42

#	ARTICLE	IF	CITATIONS
37	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
38	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
39	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
40	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
41	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
42	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
43	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
44	Mutation analysis of the coding sequence of the MECP2 gene in infantile autism. <i>Human Genetics</i> , 2002, 111, 305-309.	1.8	82