Claudio Toma

List of Publications by Year in descending order

47 papers	3,536 citations	236925 25 h-index	233421 45 g-index
51 all docs	51 docs citations	51 times ranked	5489 citing authors

#	Article	IF	Citations
1	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
2	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. Nature Genetics, 2021, 53, 817-829.	21.4	629
3	Paternally inherited cis-regulatory structural variants are associated with autism. Science, 2018, 360, 327-331.	12.6	174
4	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
5	Exome sequencing in multiplex autism families suggests a major role for heterozygous truncating mutations. Molecular Psychiatry, 2014, 19, 784-790.	7.9	110
6	MET and autism susceptibility: family and caseâ€“control studies. European Journal of Human Genetics, 2009, 17, 749-758.	2.8	86
7	Analysis of two language-related genes in autism. Psychiatric Genetics, 2013, 23, 82-85.	1.1	78
8	Copy number variation and association analysis of SHANK3 as a candidate gene for autism in the IMGSAC collection. European Journal of Human Genetics, 2009, 17, 1347-1353.	2.8	76
9	Chiari Malformation Type I: A Case-Control Association Study of 58 Developmental Genes. PLoS ONE, 2013, 8, e57241.	2.5	61
10	The dyslexia-associated gene KIAA0319 encodes highly N- and O-glycosylated plasma membrane and secreted isoforms. Human Molecular Genetics, 2008, 17, 859-871.	2.9	56
11	Cerebral Folate Deficiency Syndromes in Childhood. Archives of Neurology, 2011, 68, 615-21.	4.5	52
12	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
13	Progressive ataxia and myoclonic epilepsy in a patient with a homozygous mutation in the FOLR1 gene. Journal of Inherited Metabolic Disease, 2010, 33, 795-802.	3.6	43
14	Is ASMT a susceptibility gene for autism spectrum disorders? A replication study in European populations. Molecular Psychiatry, 2007, 12, 977-979.	7.9	42
15	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
16	Association study of six candidate genes asymmetrically expressed in the two cerebral hemispheres suggests the involvement of BAIAP2 in autism. Journal of Psychiatric Research, 2011, 45, 280-282.	3.1	40
17	Evaluation of single nucleotide polymorphisms in the miR-183â€“96â€“182 cluster in adulthood attention-deficit and hyperactivity disorder (ADHD) and substance use disorders (SUDs). European Neuropsychopharmacology, 2013, 23, 1463-1473.	0.7	38
18	Neurotransmitter systems and neurotrophic factors in autism: association study of 37 genes suggests involvement of DDC. World Journal of Biological Psychiatry, 2013, 14, 516-527.	2.6	36

#	Article	IF	Citations
19	Mutation Spectrum in the CACNA1A Gene in 49 Patients with Episodic Ataxia. Scientific Reports, 2017, 7, 2514.	3.3	36
20	Screening of <scp>CACNA1A</scp> and <scp>ATP1A2</scp> genes in hemiplegic migraine: clinical, genetic, and functional studies. Molecular Genetics & Genomic Medicine, 2013, 1, 206-222.	1.2	35
21	An examination of multiple classes of rare variants in extended families with bipolar disorder. Translational Psychiatry, 2018, 8, 65.	4.8	35
22	Lack of replication of previous autism spectrum disorder GWAS hits in European populations. Autism Research, 2017, 10, 202-211.	3.8	34
23	Contribution of common and rare variants of the PTCHD1 gene to autism spectrum disorders and intellectual disability. European Journal of Human Genetics, 2015, 23, 1694-1701.	2.8	31
24	Traumatic Stress Interacts With Bipolar Disorder Genetic Risk to Increase Risk for Suicide Attempts. Journal of the American Academy of Child and Adolescent Psychiatry, 2017, 56, 1073-1080.	0.5	31
25	Alternative splicing in the dyslexia-associated gene KIAA0319. Mammalian Genome, 2007, 18, 627-634.	2.2	30
26	Common and rare variants of microRNA genes in autism spectrum disorders. World Journal of Biological Psychiatry, 2015, 16, 376-386.	2.6	27
27	Comprehensive cross-disorder analyses of CNTNAP2 suggest it is unlikely to be a primary risk gene for psychiatric disorders. PLoS Genetics, 2018, 14, e1007535.	3.5	27
28	Truncating variant burden in high-functioning autism and pleiotropic effects of LRP1 across psychiatric phenotypes. Journal of Psychiatry and Neuroscience, 2019, 44, 350-359.	2.4	24
29	Tyrosine hydroxylase deficiency in three Greek patients with a common ancestral mutation. Movement Disorders, 2010, 25, 1086-1090.	3.9	22
30	Genetic Variation across Phenotypic Severity of Autism. Trends in Genetics, 2020, 36, 228-231.	6.7	21
31	Diverse phenotypic measurements of wellbeing: Heritability, temporal stability and the variance explained by polygenic scores. Genes, Brain and Behavior, 2020, 19, e12694.	2.2	19
32	Characterisation of two deletions involving NPC1 and flanking genes in Niemannâ€“Pick Type C disease patients. Molecular Genetics and Metabolism, 2012, 107, 716-720.	1.1	18
33	Exploration of experiences with and understanding of polygenic risk scores for bipolar disorder. Journal of Affective Disorders, 2020, 265, 342-350.	4.1	17
34	De Novo Gene Variants and Familial Bipolar Disorder. JAMA Network Open, 2020, 3, e203382.	5.9	15
35	Involvement of the 14-3-3 Gene Family in Autism Spectrum Disorder and Schizophrenia: Genetics, Transcriptomics and Functional Analyses. Journal of Clinical Medicine, 2020, 9, 1851.	2.4	14
36	Deletion in the tyrosine hydroxylase gene in a patient with a mild phenotype. Movement Disorders, 2011, 26, 1558-1560.	3.9	12

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37	Rare variants analysis of neurexin-1Î² in autism reveals a novel start codon mutation affecting protein levels at synapses. Psychiatric Genetics, 2013, 23, 262-266.	1.1	11
38	The involvement of serotonin polymorphisms in autistic spectrum symptomatology. Psychiatric Genetics, 2014, 24, 158-163.	1.1	8
39	Using linkage studies combined with wholeâ€exome sequencing to identify novel candidate genes for familial colorectal cancer. International Journal of Cancer, 2020, 146, 1568-1577.	5.1	8
40	Cortical mediation of relationships between dopamine receptor D2 and cognition is absent in youth at risk of bipolar disorder. Psychiatry Research - Neuroimaging, 2021, 309, 111258.	1.8	8
41	A linkage and exome study of multiplex families with bipolar disorder implicates rare coding variants of ANK3 and additional rare alleles at 10q11-q21. Journal of Psychiatry and Neuroscience, 2021, 46, E247-E257.	2.4	6
42	Identification of a Novel Candidate Gene for Serrated Polyposis Syndrome Germline Predisposition by Performing Linkage Analysis Combined With Whole-Exome Sequencing. Clinical and Translational Gastroenterology, 2019, 10, e00100.	2.5	5
43	Identifying Extreme Observations, Outliers and Noise in Clinical and Genetic Data. Current Bioinformatics, 2017, 12, 101-117.	1.5	5
44	Effects of polygenic risk for suicide attempt and risky behavior on brain structure in young people with familial risk of bipolar disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2021, 186, 485-507.	1.7	4
45	Screening of cacna1a and ATP1A2 genes in hemiplegic migraine: clinical, genetic and functional studies. Journal of Headache and Pain, 2013, 14, .	6.0	2
46	Extreme Observations in Biomedical Data. Trends in Mathematics, 2017, , 3-8.	0.1	0
47	Cover Image, Volume 186B, Number 8, December 2021. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2021, 186, .	1.7	0