

Claudio Toma

List of Publications by Year in descending order

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Version: 2024-02-01

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	Cortical mediation of relationships between dopamine receptor D2 and cognition is absent in youth at risk of bipolar disorder. <i>Psychiatry Research - Neuroimaging</i> , 2021, 309, 111258.	1.8	8
2	A linkage and exome study of multiplex families with bipolar disorder implicates rare coding variants of ANK3 and additional rare alleles at 10q11-q21. <i>Journal of Psychiatry and Neuroscience</i> , 2021, 46, E247-E257.	2.4	6
3	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. <i>Nature Genetics</i> , 2021, 53, 817-829.	21.4	629
4	Effects of polygenic risk for suicide attempt and risky behavior on brain structure in young people with familial risk of bipolar disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2021, 186, 485-507.	1.7	4
5	Cover Image, Volume 186B, Number 8, December 2021. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2021, 186, .	1.7	0
6	Using linkage studies combined with whole-exome sequencing to identify novel candidate genes for familial colorectal cancer. <i>International Journal of Cancer</i> , 2020, 146, 1568-1577.	5.1	8
7	Diverse phenotypic measurements of wellbeing: Heritability, temporal stability and the variance explained by polygenic scores. <i>Genes, Brain and Behavior</i> , 2020, 19, e12694.	2.2	19
8	De Novo Gene Variants and Familial Bipolar Disorder. <i>JAMA Network Open</i> , 2020, 3, e203382.	5.9	15
9	Involvement of the 14-3-3 Gene Family in Autism Spectrum Disorder and Schizophrenia: Genetics, Transcriptomics and Functional Analyses. <i>Journal of Clinical Medicine</i> , 2020, 9, 1851.	2.4	14
10	Genetic Variation across Phenotypic Severity of Autism. <i>Trends in Genetics</i> , 2020, 36, 228-231.	6.7	21
11	Exploration of experiences with and understanding of polygenic risk scores for bipolar disorder. <i>Journal of Affective Disorders</i> , 2020, 265, 342-350.	4.1	17
12	Identification of a Novel Candidate Gene for Serrated Polyposis Syndrome Germline Predisposition by Performing Linkage Analysis Combined With Whole-Exome Sequencing. <i>Clinical and Translational Gastroenterology</i> , 2019, 10, e00100.	2.5	5
13	Truncating variant burden in high-functioning autism and pleiotropic effects of <i>LRP1</i> across psychiatric phenotypes. <i>Journal of Psychiatry and Neuroscience</i> , 2019, 44, 350-359.	2.4	24
14	Paternally inherited cis-regulatory structural variants are associated with autism. <i>Science</i> , 2018, 360, 327-331.	12.6	174
15	An examination of multiple classes of rare variants in extended families with bipolar disorder. <i>Translational Psychiatry</i> , 2018, 8, 65.	4.8	35
16	Comprehensive cross-disorder analyses of CNTNAP2 suggest it is unlikely to be a primary risk gene for psychiatric disorders. <i>PLoS Genetics</i> , 2018, 14, e1007535.	3.5	27
17	Mutation Spectrum in the CACNA1A Gene in 49 Patients with Episodic Ataxia. <i>Scientific Reports</i> , 2017, 7, 2514.	3.3	36
18	Traumatic Stress Interacts With Bipolar Disorder Genetic Risk to Increase Risk for Suicide Attempts. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2017, 56, 1073-1080.	0.5	31

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19	Lack of replication of previous autism spectrum disorder GWAS hits in European populations. <i>Autism Research</i> , 2017, 10, 202-211.	3.8	34
20	Identifying Extreme Observations, Outliers and Noise in Clinical and Genetic Data. <i>Current Bioinformatics</i> , 2017, 12, 101-117.	1.5	5
21	Extreme Observations in Biomedical Data. <i>Trends in Mathematics</i> , 2017, , 3-8.	0.1	0
22	Contribution of common and rare variants of the PTCHD1 gene to autism spectrum disorders and intellectual disability. <i>European Journal of Human Genetics</i> , 2015, 23, 1694-1701.	2.8	31
23	Common and rare variants of microRNA genes in autism spectrum disorders. <i>World Journal of Biological Psychiatry</i> , 2015, 16, 376-386.	2.6	27
24	The involvement of serotonin polymorphisms in autistic spectrum symptomatology. <i>Psychiatric Genetics</i> , 2014, 24, 158-163.	1.1	8
25	Exome sequencing in multiplex autism families suggests a major role for heterozygous truncating mutations. <i>Molecular Psychiatry</i> , 2014, 19, 784-790.	7.9	110
26	Screening of cacna1a and ATP1A2 genes in hemiplegic migraine: clinical, genetic and functional studies. <i>Journal of Headache and Pain</i> , 2013, 14, .	6.0	2
27	Evaluation of single nucleotide polymorphisms in the miR-183-96-182 cluster in adulthood attention-deficit and hyperactivity disorder (ADHD) and substance use disorders (SUDs). <i>European Neuropsychopharmacology</i> , 2013, 23, 1463-1473.	0.7	38
28	Analysis of two language-related genes in autism. <i>Psychiatric Genetics</i> , 2013, 23, 82-85.	1.1	78
29	Neurotransmitter systems and neurotrophic factors in autism: association study of 37 genes suggests involvement of DDC. <i>World Journal of Biological Psychiatry</i> , 2013, 14, 516-527.	2.6	36
30	Rare variants analysis of neurexin-1 ² in autism reveals a novel start codon mutation affecting protein levels at synapses. <i>Psychiatric Genetics</i> , 2013, 23, 262-266.	1.1	11
31	Screening of <i>CACNA1A</i> and <i>ATP1A2</i> genes in hemiplegic migraine: clinical, genetic, and functional studies. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 206-222.	1.2	35
32	Chiari Malformation Type I: A Case-Control Association Study of 58 Developmental Genes. <i>PLoS ONE</i> , 2013, 8, e57241.	2.5	61
33	Characterisation of two deletions involving NPC1 and flanking genes in Niemann-Pick Type C disease patients. <i>Molecular Genetics and Metabolism</i> , 2012, 107, 716-720.	1.1	18
34	Association study of six candidate genes asymmetrically expressed in the two cerebral hemispheres suggests the involvement of BAIAP2 in autism. <i>Journal of Psychiatric Research</i> , 2011, 45, 280-282.	3.1	40
35	Deletion in the tyrosine hydroxylase gene in a patient with a mild phenotype. <i>Movement Disorders</i> , 2011, 26, 1558-1560.	3.9	12
36	Cerebral Folate Deficiency Syndromes in Childhood. <i>Archives of Neurology</i> , 2011, 68, 615-21.	4.5	52

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37	Progressive ataxia and myoclonic epilepsy in a patient with a homozygous mutation in the <i>FOLR1</i> gene. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 795-802.	3.6	43
38	Tyrosine hydroxylase deficiency in three Greek patients with a common ancestral mutation. <i>Movement Disorders</i> , 2010, 25, 1086-1090.	3.9	22
39	High-density SNP association study and copy number variation analysis of the <i>AUTS1</i> and <i>AUTS5</i> loci implicate the <i>IMMP2L</i> – <i>DOCK4</i> gene region in autism susceptibility. <i>Molecular Psychiatry</i> , 2010, 15, 954-968.	7.9	126
40	<i>MET</i> and autism susceptibility: family and case–control studies. <i>European Journal of Human Genetics</i> , 2009, 17, 749-758.	2.8	86
41	Copy number variation and association analysis of <i>SHANK3</i> as a candidate gene for autism in the IMGSAC collection. <i>European Journal of Human Genetics</i> , 2009, 17, 1347-1353.	2.8	76
42	Analysis of X chromosome inactivation in autism spectrum disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2008, 147B, 830-835.	1.7	42
43	The dyslexia-associated gene <i>KIAA0319</i> encodes highly N- and O-glycosylated plasma membrane and secreted isoforms. <i>Human Molecular Genetics</i> , 2008, 17, 859-871.	2.9	56
44	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. <i>Nature Genetics</i> , 2007, 39, 319-328.	21.4	1,272
45	Is <i>ASMT</i> a susceptibility gene for autism spectrum disorders? A replication study in European populations. <i>Molecular Psychiatry</i> , 2007, 12, 977-979.	7.9	42
46	Alternative splicing in the dyslexia-associated gene <i>KIAA0319</i> . <i>Mammalian Genome</i> , 2007, 18, 627-634.	2.2	30
47	<i>SLC25A12</i> and <i>CMYA3</i> gene variants are not associated with autism in the IMGSAC multiplex family sample. <i>European Journal of Human Genetics</i> , 2006, 14, 123-126.	2.8	44