Catalina Betancur

List of Publications by Year
in descending order

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124 papers	24,912 citations	30070 54 h-index	16183 124 g-index

130 all docs	130 docs citations	130 times ranked	25612 citing authors

#	Article	IF	Citations
1	Strong evidence for genotypeâ€“phenotype correlations in Phelan-McDermid syndrome: results from the developmental synaptopathies consortium. Human Molecular Genetics, 2022, 31, 625-637.	2.9	32
2	Clinical validity assessment of genes frequently tested on intellectual disability/autism sequencing panels. Genetics in Medicine, 2022, 24, 1899-1908.	2.4	9
3	Clinical and neurocognitive issues associated with Boschâ€Boonstraâ€Schaaf optic atrophy syndrome: A case study. American Journal of Medical Genetics, Part A, 2020, 182, 213-218.	1.2	11
4	Gene constraint and genotypeâ€“phenotype correlations in neurodevelopmental disorders. Current Opinion in Genetics and Development, 2020, 65, 69-75.	3.3	7
5	Psychiatric illness and regression in individuals with Phelan-McDermid syndrome. Journal of Neurodevelopmental Disorders, 2020, 12, 7.	3.1	51
6	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
7	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	16.3	83
8	Recessive gene disruptions in autism spectrum disorder. Nature Genetics, 2019, 51, 1092-1098.	21.4	109
9	Rigor in science and science reporting: updated guidelines for submissions to Molecular Autism. Molecular Autism, 2019, 10, 6.	4.9	4
10	Viral vectorâ€mediated Cre recombinase expression in substantia nigra induces lesions of the nigrostriatal pathway associated with perturbations of dopamineâ€related behaviors and hallmarks of programmed cell death. Journal of Neurochemistry, 2019, 150, 330-340.	3.9	32
11	Neuropsychiatric decompensation in adolescents and adults with Phelan-McDermid syndrome: a systematic review of the literature. Molecular Autism, 2019, 10, 50.	4.9	47
12	Delineation of the genetic and clinical spectrum of Phelan-McDermid syndrome caused by SHANK3 point mutations. Molecular Autism, 2018, 9, 31.	4.9	152
13	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
14	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
15	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. Nature Neuroscience, 2017, 20, 1217-1224.	14.8	212
16	Meta-analysis of GWAS of over 16,000 individuals with autism spectrum disorder highlights a novel locus at 10q24.32 and a significant overlap with schizophrenia. Molecular Autism, 2017, 8, 21.	4.9	495
17	The 22q11 PRODH/DGCR6 deletion is frequent in hyperprolinemic subjects but is not a strong risk factor for ASD. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 377-382.	1.7	3
18	Genetic and functional analyses demonstrate a role for abnormal glycinergic signaling in autism. Molecular Psychiatry, 2016, 21, 936-945.	7.9	85

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19	Joint Analysis of Psychiatric Disorders Increases Accuracy of Risk Prediction for Schizophrenia, Bipolar Disorder, and Major Depressive Disorder. American Journal of Human Genetics, 2015, 96, 283-294.	6.2	225
20	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. Nature Neuroscience, 2015, 18, 199-209.	14.8	701
21	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. Molecular Autism, 2015, 6, 19.	4.9	29
22	The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. Nature Communications, 2014, 5, 4074.	12.8	52
23	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
24	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	6.2	819
25	Prospective investigation of autism and genotype-phenotype correlations in 22q13 deletion syndrome and SHANK3 deficiency. Molecular Autism, 2013, 4, 18.	4.9	278
26	SHANK3 haploinsufficiency: a â€œcommonâ€•but underdiagnosed highly penetrant monogenic cause of autism spectrum disorders. Molecular Autism, 2013, 4, 17.	4.9	152
27	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	21.4	2,067
28	Heterozygous FA2H mutations in autism spectrum disorders. BMC Medical Genetics, 2013, 14, 124.	2.1	7
29	No evidence that common genetic risk variation is shared between schizophrenia and autism. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 55-60.	1.7	24
30	Etiological Heterogeneity in Autism Spectrum Disorders. , 2013, , 113-144.		10
31	Network Topologies and Convergent Aetiologies Arising from Deletions and Duplications Observed in Individuals with Autism. PLoS Genetics, 2013, 9, e1003523.	3.5	51
32	Characterization of SLITRK1 Variation in Obsessive-Compulsive Disorder. PLoS ONE, 2013, 8, e70376.	2.5	47
33	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. PLoS Genetics, 2012, 8, e1002521.	3.5	358
34	Autism multiplex family with 16p11.2p12.2 microduplication syndrome in monozygotic twins and distal 16p11.2 deletion in their brother. European Journal of Human Genetics, 2012, 20, 540-546.	2.8	38
35	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	2.9	334
36	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	7.1	118

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37	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	3.8	180
38	High-functioning autism spectrum disorder and fragile X syndrome: report of two affected sisters. Molecular Autism, 2012, 3, 5.	4.9	13
39	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	27.8	1,597
40	Networkâ€•and attributeâ€•based classifiers can prioritize genes and pathways for autism spectrum disorders and intellectual disability. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 130-142.	1.6	32
41	Optimizing the phenotyping of rodent ASD models: enrichment analysis of mouse and human neurobiological phenotypes associated with high-risk autism genes identifies morphological, electrophysiological, neurological, and behavioral features. Molecular Autism, 2012, 3, 1.	4.9	50
42	Clinical utility gene card for: Deletion 22q13 syndrome. European Journal of Human Genetics, 2011, 19, 492-492.	2.8	11
43	Gene-ontology enrichment analysis in two independent family-based samples highlights biologically plausible processes for autism spectrum disorders. European Journal of Human Genetics, 2011, 19, 1082-1089.	2.8	39
44	Etiological heterogeneity in autism spectrum disorders: More than 100 genetic and genomic disorders and still counting. Brain Research, 2011, 1380, 42-77.	2.2	788
45	Novel method for combined linkage and genome-wide association analysis finds evidence of distinct genetic architecture for two subtypes of autism. Journal of Neurodevelopmental Disorders, 2011, 3, 113-123.	3.1	22
46	A family with autism and rare copy number variants disrupting the Duchenne/Becker muscular dystrophy gene DMD and TRPM3. Journal of Neurodevelopmental Disorders, 2011, 3, 124-131.	3.1	35
47	A large-scale survey of the novel 15q24 microdeletion syndrome in autism spectrum disorders identifies an atypical deletion that narrows the critical region. Molecular Autism, 2010, 1, 5.	4.9	40
48	Molecular characterization of a de novo 6q24.2q25.3 duplication interrupting UTRN in a patient with arthrogryposis. American Journal of Medical Genetics, Part A, 2010, 152A, 1781-1788.	1.2	13
49	Paracentric inversion of chromosome 2 associated with cryptic duplication of 2q14 and deletion of 2q37 in a patient with autism. American Journal of Medical Genetics, Part A, 2010, 152A, 2346-2354.	1.2	17
50	Reduced 3â€Oâ€methylâ€dopa levels in OCD patients and their unaffected parents is associated with the low activity M158 COMT allele. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 542-548.	1.7	3
51	Search for copy number variants in chromosomes 15q11-q13 and 22q11.2 in obsessive compulsive disorder. BMC Medical Genetics, 2010, 11, 100.	2.1	14
52	Mutation screening of NOS1AP gene in a large sample of psychiatric patients and controls. BMC Medical Genetics, 2010, 11, 108.	2.1	31
53	Linkage and candidate gene studies of autism spectrum disorders in European populations. European Journal of Human Genetics, 2010, 18, 1013-1019.	2.8	80
54	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803

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55	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	2.9	538
56	Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. Science Translational Medicine, 2010, 2, 49ra68.	12.4	178
57	Possible association between the androgen receptor gene and autism spectrum disorder. Psychoneuroendocrinology, 2009, 34, 752-761.	2.7	58
58	An investigation of ribosomal protein L10 gene in autism spectrum disorders. BMC Medical Genetics, 2009, 10, 7.	2.1	25
59	A genome-wide linkage and association scan reveals novel loci for autism. Nature, 2009, 461, 802-808.	27.8	570
60	The emerging role of synaptic cell-adhesion pathways in the pathogenesis of autism spectrum disorders. Trends in Neurosciences, 2009, 32, 402-412.	8.6	271
61	Screening for Genomic Rearrangements and Methylation Abnormalities of the 15q11-q13 Region in Autism Spectrum Disorders. Biological Psychiatry, 2009, 66, 349-359.	1.3	133
62	Autism, language delay and mental retardation in a patient with 7q11 duplication. BMJ Case Reports, 2009, 2009, bcr0520091911-bcr0520091911.	0.5	3
63	Multiplex ligation-dependent probe amplification for genetic screening in autism spectrum disorders: Efficient identification of known microduplications and identification of a novel microduplication in ASMT. BMC Medical Genomics, 2008, 1, 50.	1.5	74
64	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
65	Abnormal melatonin synthesis in autism spectrum disorders. Molecular Psychiatry, 2008, 13, 90-98.	7.9	423
66	Genome-wide Linkage Analyses of Quantitative and Categorical Autism Subphenotypes. Biological Psychiatry, 2008, 64, 561-570.	1.3	80
67	Loss of VGLUT1 and VGLUT2 in the prefrontal cortex is correlated with cognitive decline in Alzheimer disease. Neurobiology of Aging, 2008, 29, 1619-1630.	3.1	136
68	Parallel Loss of Hippocampal LTD and Cognitive Flexibility in a Genetic Model of Hyperdopaminergia. Neuropsychopharmacology, 2007, 32, 2108-2116.	5.4	106
69	Autism, language delay and mental retardation in a patient with 7q11 duplication. Journal of Medical Genetics, 2007, 44, 452-458.	3.2	75
70	Altered expression of vesicular glutamate transporters VGLUT1 and VGLUT2 in Parkinson disease. Neurobiology of Aging, 2007, 28, 568-578.	3.1	109
71	Shared executive dysfunctions in unaffected relatives of patients with autism and obsessive-compulsive disorder. European Psychiatry, 2007, 22, 32-38.	0.2	106
72	Mutation screening of theARX gene in patients with autism. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 228-230.	1.7	24

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73	Mutation screening of the PTEN gene in patients with autism spectrum disorders and macrocephaly. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 484-491.	1.7	248
74	Mutation analysis of the NSD1 gene in patients with autism spectrum disorders and macrocephaly. BMC Medical Genetics, 2007, 8, 68.	2.1	20
75	Mutations in the gene encoding the synaptic scaffolding protein SHANK3 are associated with autism spectrum disorders. Nature Genetics, 2007, 39, 25-27.	21.4	1,408
76	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
77	Tryptophan hydroxylase 2 (TPH2) haplotypes predict levels of TPH2 mRNA expression in human pons. Molecular Psychiatry, 2007, 12, 491-501.	7.9	124
78	No Human Tryptophan Hydroxylase-2 Gene R441H Mutation in a Large Cohort of Psychiatric Patients and Control Subjects. Biological Psychiatry, 2006, 60, 202-203.	1.3	49
79	Exploratory analysis of obsessive compulsive symptom dimensions in children and adolescents: a Prospective follow-up study. BMC Psychiatry, 2006, 6, 1.	2.6	140
80	Expression and genetic variability ofPCDH11Y, a gene specific toHomo sapiens and candidate for susceptibility to psychiatric disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 67-70.	1.7	46
81	Constitutional Downregulation of SEMA5A Expression in Autism. Neuropsychobiology, 2006, 54, 64-69.	1.9	76
82	The reinforcing effects of chronic d-amphetamine and morphine are impaired in a line of memory-deficient mice overexpressing calcineurin. European Journal of Neuroscience, 2005, 21, 3089-3096.	2.6	35
83	Support for the association between the rare functional variant I425V of the serotonin transporter gene and susceptibility to obsessive compulsive disorder. Molecular Psychiatry, 2005, 10, 1059-1061.	7.9	46
84	Platelet Serotonergic Markers as Endophenotypes for Obsessive-Compulsive Disorder. Neuropsychopharmacology, 2005, 30, 1539-1547.	5.4	32
85	Altered neurotensin mrna expression in mice lacking the dopamine transporter. Neuroscience, 2004, 123, 537-546.	2.3	7
86	Analysis of transmission of novel polymorphisms in the somatostatin receptor 5 (SSTR5) gene in patients with autism. American Journal of Medical Genetics Part A, 2003, 121B, 100-104.	2.4	7
87	Mutations of the X-linked genes encoding neuroligins NLGN3 and NLGN4 are associated with autism. Nature Genetics, 2003, 34, 27-29.	21.4	1,612
88	Acute and Chronic Effects of Methamphetamine on Tele-Methylhistamine Levels in Mouse Brain: Selective Involvement of the D₂ and not D₃Receptor. Journal of Pharmacology and Experimental Therapeutics, 2002, 300, 621-628.	2.5	54
89	Y chromosome haplogroups in autistic subjects. Molecular Psychiatry, 2002, 7, 217-219.	7.9	44
90	Linkage and association of the glutamate receptor 6 gene with autism. Molecular Psychiatry, 2002, 7, 302-310.	7.9	279

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91	Increased Rate of Twins among Affected Sibling Pairs with Autism. American Journal of Human Genetics, 2002, 70, 1381-1383.	6.2	50
92	Response to Visscher. American Journal of Human Genetics, 2002, 71, 996-999.	6.2	7
93	Investigation of two variants in the DOPA decarboxylase gene in patients with autism. American Journal of Medical Genetics Part A, 2002, 114, 466-470.	2.4	12
94	Absence of association between a polymorphic GGC repeat in the 5â€² untranslated region of the reelin gene and autism. Molecular Psychiatry, 2002, 7, 801-804.	7.9	96
95	Serotonin transporter gene polymorphisms and hyperserotonemia in autistic disorder. Molecular Psychiatry, 2002, 7, 67-71.	7.9	55
96	Hypolocomotor effects of acute and daily d-amphetamine in mice lacking the dopamine transporter. Psychopharmacology, 2001, 159, 2-9.	3.1	88
97	A balanced reciprocal translocation t(5;7)(q14;q32) associated with autistic disorder: Molecular analysis of the chromosome 7 breakpoint. American Journal of Medical Genetics Part A, 2001, 105, 729-736.	2.4	17
98	Neurotensin Gene Expression and Behavioral Responses Following Administration of Psychostimulants and Antipsychotic Drugs in Dopamine D3 Receptor Deficient Mice. Neuropsychopharmacology, 2001, 24, 170-182.	5.4	45
99	Role of dopamine D3 receptors in thermoregulation. NeuroReport, 2000, 11, 221-225.	1.2	42
100	Behavioural disturbances associated with hyperdopaminergia in dopamine-transporter knockout mice. Behavioural Pharmacology, 2000, 11, 279-290.	1.7	210
101	Regulation of the neurotensin NT1 receptor in the developing rat brain following chronic treatment with the antagonist SR 48692. , 2000, 60, 362-369.		6
102	Increased rewarding properties of morphine in dopamine-transporter knockout mice. European Journal of Neuroscience, 2000, 12, 1827-1837.	2.6	75
103	Differential ontogenetic patterns of levocabastine-sensitive neurotensin NT2 receptors and of NT1 receptors in the rat brain revealed by in situ hybridization. Developmental Brain Research, 1999, 113, 115-131.	1.7	40
104	Tissue distribution and cellular localization of the levocabastine-sensitive neurotensin receptor mRNA in adult rat brain. Molecular Brain Research, 1998, 57, 193-200.	2.3	65
105	Characterization of binding sites of a new neurotensin receptor antagonist, []SR 142948A, in the rat brain. European Journal of Pharmacology, 1998, 343, 67-77.	3.5	51
106	Repeated Administration of the Neurotensin Receptor Antagonist SR 48692 Differentially Regulates Mesocortical and Mesolimbic Dopaminergic Systems. Journal of Neurochemistry, 1998, 71, 1158-1167.	3.9	32
107	Role of Endogenous Neurotensin in the Behavioral and Neuroendocrine Effects of Cocaine. Neuropsychopharmacology, 1998, 19, 322-332.	5.4	36
108	Nonpeptide antagonists of neuropeptide receptors: tools for research and therapy. Trends in Pharmacological Sciences, 1997, 18, 372-386.	8.7	47

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109	Chronic cocaine increases neurotensin gene expression in the shell of the nucleus accumbens and in discrete regions of the striatum. Molecular Brain Research, 1997, 44, 334-340.	2.3	36
110	Use of Nonpeptide Antagonists to Explore the Physiological Roles of Neurotensin.. Annals of the New York Academy of Sciences, 1997, 814, 125-141.	3.8	53
111	Endogenous Neurotensin Regulates Hypothalamicâ€Pituitaryâ€Adrenal Axis Activity and Peptidergic Neurons in the Rat Hypothalamic Paraventricular Nucleus. Journal of Neuroendocrinology, 1997, 9, 263-269.	2.6	40
112	Cytokine Regulation of Corticosteroid Receptors in the Rat Hippocampus: Effects of Interleukin-1, Interleukin-6, Tumor Necrosis Factor and Lipopolysaccharide. Neuroendocrinology, 1995, 62, 47-54.	2.5	24
113	Characterization and distribution of binding sites for a new neurotensin receptor antagonist ligand, [3H]SR 48692, in the guinea pig brain. Journal of Pharmacology and Experimental Therapeutics, 1995, 273, 1450-8.	2.5	18
114	Magnetic field effects on stress-induced analgesia in mice: modulation by light. Neuroscience Letters, 1994, 182, 147-150.	2.1	42
115	Corticosteroid Regulation of IL-1 Receptors in the Mouse Hippocampus: Effects of Glucocorticoid Treatment, Stress, and Adrenalectomy. Neuroendocrinology, 1994, 59, 120-128.	2.5	30
116	Activity of the hypothalamic-pituitary-adrenal axis in mice selected for left- or right-handedness. Brain Research, 1992, 589, 302-306.	2.2	10
117	Natural killer cell activity is associated with brain asymmetry in male mice. Brain, Behavior, and Immunity, 1991, 5, 162-169.	4.1	53
118	Sex-dependent association between immune function and paw preference in two substrains of C3H mice. Brain Research, 1991, 559, 347-351.	2.2	24
119	Functional brain asymmetry and lymphocyte proliferation in female mice: effects of right and left cortical ablation. Brain Research, 1991, 550, 125-128.	2.2	28
120	Strain and sex differences in the degree of paw preference in mice. Behavioural Brain Research, 1991, 45, 97-101.	2.2	50
121	Association between left-handedness and allergy: A reappraisal. Neuropsychologia, 1990, 28, 223-227.	1.6	38
122	Functional brain asymmetry and murine systemic lupus erythematosus. Brain Research, 1989, 498, 159-162.	2.2	43
123	Brain modulation of the immune system: association between lymphocyte responsiveness and paw preference in mice. Brain Research, 1988, 457, 392-394.	2.2	87
124	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. SSRN Electronic Journal, 0, , .	0.4	12