Thomas Bourgeron

List of Publications by Year
in descending order

91	13,698	66234	43802
papers	citations	42 h-index	91 g-index

111	111	111	15156
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.	13.7	1,803
2	Mutations of the X-linked genes encoding neuroligins NLGN3 and NLGN4 are associated with autism. Nature Genetics, 2003, 34, 27-29.	9.4	1,612
3	Mutations in the gene encoding the synaptic scaffolding protein SHANK3 are associated with autism spectrum disorders. Nature Genetics, 2007, 39, 25-27.	9.4	1,408
4	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	2.6	819
5	From the genetic architecture to synaptic plasticity in autism spectrum disorder. Nature Reviews Neuroscience, 2015, 16, 551-563.	4.9	764
6	A synaptic trek to autism. Current Opinion in Neurobiology, 2009, 19, 231-234.	2.0	596
7	Autistic-like behaviours and hyperactivity in mice lacking ProSAP1/Shank2. Nature, 2012, 486, 256-260.	13.7	570
8	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	1.5	501
9	Searching for ways out of the autism maze: genetic, epigenetic and environmental clues. Trends in Neurosciences, 2006, 29, 349-358.	4.2	498
10	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. PLoS Genetics, 2012, 8, e1002521.	1.5	358
11	The Genetic Landscapes of Autism Spectrum Disorders. Annual Review of Genomics and Human Genetics, 2013, 14, 191-213.	2.5	352
12	Key role for gene dosage and synaptic homeostasis in autism spectrum disorders. Trends in Genetics, 2010, 26, 363-372.	2.9	296
13	SHANK1 Deletions in Males with Autism Spectrum Disorder. American Journal of Human Genetics, 2012, 90, 879-887.	2.6	292
14	The EU-AIMS Longitudinal European Autism Project (LEAP): design and methodologies to identify and validate stratification biomarkers for autism spectrum disorders. Molecular Autism, 2017, 8, 24.	2.6	183
15	Progress toward treatments for synaptic defects in autism. Nature Medicine, 2013, 19, 685-694.	15.2	167
16	Adult Male Mice Emit Context-Specific Ultrasonic Vocalizations That Are Modulated by Prior Isolation or Group Rearing Environment. PLoS ONE, 2012, 7, e29401.	1.1	154
17	Gender differences in autism spectrum disorders: Divergence among specific core symptoms. Autism Research, 2017, 10, 680-689.	2.1	140
18	Behavioral profiles of mouse models for autism spectrum disorders. Autism Research, 2011, 4, 5-16.	2.1	133

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19	The EU-AIMS Longitudinal European Autism Project (LEAP): clinical characterisation. Molecular Autism, 2017, 8, 27.	2.6	126
20	Identification and validation of biomarkers for autism spectrum disorders. Nature Reviews Drug Discovery, 2016, 15, 70-70.	21.5	117
21	Real-time analysis of the behaviour of groups of mice via a depth-sensing camera and machine learning. Nature Biomedical Engineering, 2019, 3, 930-942.	11.6	112
22	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	5.8	111
23	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	2.6	110
24	Neuroanatomical Diversity of Corpus Callosum and Brain Volume in Autism: Meta-analysis, Analysis of the Autism Brain Imaging Data Exchange Project, and Simulation. Biological Psychiatry, 2015, 78, 126-134.	0.7	108
25	Current knowledge on the genetics of autism and propositions for future research. Comptes Rendus - Biologies, 2016, 339, 300-307.	0.1	97
26	Genome-wide analyses of self-reported empathy: correlations with autism, schizophrenia, and anorexia nervosa. Translational Psychiatry, 2018, 8, 35.	2.4	95
27	Disruption of melatonin synthesis is associated with impaired 14-3-3 and miR-451 levels in patients with autism spectrum disorders. Scientific Reports, 2017, 7, 2096.	1.6	83
28	Genome-wide association scan identifies new variants associated with a cognitive predictor of dyslexia. Translational Psychiatry, 2019, 9, 77.	2.4	82
29	Altered Connectivity Between Cerebellum, Visual, and Sensory-Motor Networks in Autism Spectrum Disorder: Results from the EU-AIMS Longitudinal European Autism Project. Biological Psychiatry: Cognitive Neuroscience and Neuroimaging, 2019, 4, 260-270.	1.1	82
30	Human Pluripotent Stem Cell-derived Cortical Neurons for High Throughput Medication Screening in Autism: A Proof of Concept Study in SHANK3 Haploinsufficiency Syndrome. EBioMedicine, 2016, 9, 293-305.	2.7	79
31	Measuring and Estimating the Effect Sizes of Copy Number Variants on General Intelligence in Community-Based Samples. JAMA Psychiatry, 2018, 75, 447.	6.0	77
32	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. European Journal of Human Genetics, 2018, 26, 1759-1772.	1.4	73
33	Cerebellar Volume in Autism: Literature Meta-analysis and Analysis of the Autism BrainÂImaging Data Exchange Cohort. Biological Psychiatry, 2018, 83, 579-588.	0.7	59
34	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	2.6	59
35	The role of cholesterol metabolism and various steroid abnormalities in autism spectrum disorders: A hypothesis paper. Autism Research, 2017, 10, 1022-1044.	2.1	58
36	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	1.7	58

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37	Social and non-social autism symptoms and trait domains are genetically dissociable. Communications Biology, 2019, 2, 328.	2.0	57
38	Zinc deficiency and low enterocyte zinc transporter expression in human patients with autism related mutations in SHANK3. Scientific Reports, 2017, 7, 45190.	1.6	56
39	Genome-wide association study reveals new insights into the heritability and genetic correlates of developmental dyslexia. Molecular Psychiatry, 2021, 26, 3004-3017.	4.1	56
40	Genetic and Environmental Influences on the Visual Word Form and Fusiform Face Areas. Cerebral Cortex, 2015, 25, 2478-2493.	1.6	54
41	Crystal structure and functional mapping of human ASMT, the last enzyme of the melatonin synthesis pathway. Journal of Pineal Research, 2013, 54, 46-57.	3.4	51
42	Altered spinogenesis in iPSC-derived cortical neurons from patients with autism carrying de novo SHANK3 mutations. Scientific Reports, 2019, 9, 94.	1.6	51
43	Operative list of genes associated with autism and neurodevelopmental disorders based on database review. Molecular and Cellular Neurosciences, 2021, 113, 103623.	1.0	51
44	Genetic correlates of phenotypic heterogeneity in autism. Nature Genetics, 2022, 54, 1293-1304.	9.4	51
45	Effect Sizes of Deletions and Duplications on Autism Risk Across the Genome. American Journal of Psychiatry, 2021, 178, 87-98.	4.0	50
46	Recording Mouse Ultrasonic Vocalizations to Evaluate Social Communication. Journal of Visualized Experiments, 2016, , .	0.2	47
47	HyPyP: a Hyperscanning Python Pipeline for inter-brain connectivity analysis. Social Cognitive and Affective Neuroscience, 2021, 16, 72-83.	1.5	46
48	Dynamics in enzymatic protein complexes offer a novel principle for the regulation of melatonin synthesis in the human pineal gland. Journal of Pineal Research, 2011, 51, 145-155.	3.4	45
49	A de novo microdeletion of SEMA5A in a boy with autism spectrum disorder and intellectual disability. European Journal of Human Genetics, 2016, 24, 838-843.	1.4	40
50	Fractionating autism based on neuroanatomical normative modeling. Translational Psychiatry, 2020, 10, 384.	2.4	40
51	Autism-associated SHANK3 mutations impair maturation of neuromuscular junctions and striated muscles. Science Translational Medicine, 2020, 12, .	5.8	38
52	Alpha Waves as a Neuromarker of Autism Spectrum Disorder: The Challenge of Reproducibility and Heterogeneity. Frontiers in Neuroscience, 2018, 12, 662.	1.4	37
53	Mutations associated with neuropsychiatric conditions delineate functional brain connectivity dimensions contributing to autism and schizophrenia. Nature Communications, 2020, 11, 5272.	5.8	35
54	Lack of replication of previous autism spectrum disorder GWAS hits in European populations. Autism Research, 2017, 10, 202-211.	2.1	34

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55	Genome-wide analysis of gene dosage in 24,092 individuals estimates that 10,000 genes modulate cognitive ability. Molecular Psychiatry, 2021, 26, 2663-2676.	4.1	33
56	Dissecting the phenotypic heterogeneity in sensory features in autism spectrum disorder: a factor mixture modelling approach. Molecular Autism, 2020, 11, 67.	2.6	32
57	Phelan-McDermid syndrome: a classification system after 30Âyears of experience. Orphanet Journal of Rare Diseases, 2022, 17, 27.	1.2	32
58	Heritability of the melatonin synthesis variability in autism spectrum disorders. Scientific Reports, 2017, 7, 17746.	1.6	28
59	Sequencing ASMT Identifies Rare Mutations in Chinese Han Patients with Autism. PLoS ONE, 2013, 8, e53727.	1.1	26
60	The meaning of significant mean group differences for biomarker discovery. PLoS Computational Biology, 2021, 17, e1009477.	1.5	26
61	11q24.2â€25 microâ€rearrangements in autism spectrum disorders: Relation to brain structures. American Journal of Medical Genetics, Part A, 2015, 167, 3019-3030.	0.7	25
62	Dlx5 and Dlx6 expression in GABAergic neurons controls behavior, metabolism, healthy aging and lifespan. Aging, 2019, 11, 6638-6656.	1.4	25
63	Systematic detection of brain protein-coding genes under positive selection during primate evolution and their roles in cognition. Genome Research, 2021, 31, 484-496.	2.4	25
64	Insights from an autism imaging biomarker challenge: Promises and threats to biomarker discovery. NeuroImage, 2022, 255, 119171.	2.1	24
65	mouseTube â€“ a database to collaboratively unravel mouse ultrasonic communication. F1000Research, 2016, 5, 2332.	0.8	23
66	Behavioural Phenotypes and Neural Circuit Dysfunctions in Mouse Models of Autism Spectrum Disorder. Advances in Anatomy, Embryology and Cell Biology, 2017, 224, 85-101.	1.0	21
67	Shank2 Mutant Mice Display Hyperactivity Insensitive to Methylphenidate and Reduced Flexibility in Social Motivation, but Normal Social Recognition. Frontiers in Molecular Neuroscience, 2018, 11, 365.	1.4	21
68	Morning Plasma Melatonin Differences in Autism: Beyond the Impact of Pineal Gland Volume. Frontiers in Psychiatry, 2019, 10, 11.	1.3	21
69	Increased risk of ADHD in families with ASD. European Child and Adolescent Psychiatry, 2019, 28, 281-288.	2.8	19
70	Imbalanced social-communicative and restricted repetitive behavior subtypes of autism spectrum disorder exhibit different neural circuitry. Communications Biology, 2021, 4, 574.	2.0	17
71	LMT USV Toolbox, a Novel Methodological Approach to Place Mouse Ultrasonic Vocalizations in Their Behavioral Contextsâ€"A Study in Female and Male C57BL/6J Mice and in Shank3 Mutant Females. Frontiers in Behavioral Neuroscience, 2021, 15, 735920.	1.0	17
72	The genetics and neurobiology of ESSENCE: The third Birgit Olsson lecture. Nordic Journal of Psychiatry, 2016, 70, 1-9.	0.7	16

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73	Polygenic Architecture of Human Neuroanatomical Diversity. Cerebral Cortex, 2020, 30, 2307-2320.	1.6	16
74	Social Communication in Mice â€“ Are There Optimal Cage Conditions?. PLoS ONE, 2015, 10, e0121802.	1.1	15
75	Heterogeneous Pattern of Selective Pressure for PRRT2 in Human Populations, but No Association with Autism Spectrum Disorders. PLoS ONE, 2014, 9, e88600.	1.1	14
76	Homozygous 2p11.2 deletion supports the implication of ELMOD3 in hearing loss and reveals the potential association of CAPG with ASD/ID etiology. Journal of Applied Genetics, 2019, 60, 49-56.	1.0	11
77	Decreased phenol sulfotransferase activities associated with hyperserotonemia in autism spectrum disorders. Translational Psychiatry, 2021, 11, 23.	2.4	11
78	Anatomy and Cell Biology of Autism Spectrum Disorder: Lessons from Human Genetics. Advances in Anatomy, Embryology and Cell Biology, 2017, 224, 1-25.	1.0	10
79	Editorial: Shankopathies: Shank Protein Deficiency-Induced Synaptic Diseases. Frontiers in Molecular Neuroscience, 2020, 13, 11.	1.4	9
80	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. Npj Genomic Medicine, 2021, 6, 91.	1.7	9
81	Testosterone Increases the Emission of Ultrasonic Vocalizations With Different Acoustic Characteristics in Mice. Frontiers in Psychology, 2021, 12, 680176.	1.1	8
82	Synesthesia & autistic features in a large family: Evidence for spatial imagery as a common factor. Behavioural Brain Research, 2019, 362, 266-272.	1.2	7
83	A chimeric mouse model to study human iPSC-derived neurons: the case of a truncating SHANK3 mutation. Scientific Reports, 2020, 10, 13315.	1.6	7
84	Interactive Psychometrics for Autism With the Human Dynamic Clamp: Interpersonal Synchrony From Sensorimotor to Sociocognitive Domains. Frontiers in Psychiatry, 2020, 11, 510366.	1.3	7
85	Three-dimensional Quantification of Dendritic Spines from Pyramidal Neurons Derived from Human Induced Pluripotent Stem Cells. Journal of Visualized Experiments, 2015, , .	0.2	6
86	The functional database of the ARCHI project: Potential and perspectives. NeuroImage, 2019, 197, 527-543.	2.1	6
87	Massâ€spectrometry analysis of the human pineal proteome during night and day and in autism. Journal of Pineal Research, 2021, 70, e12713.	3.4	4
88	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.	1.1	3
89	The role of rare compound heterozygous events in autism spectrum disorder. Translational Psychiatry, 2020, 10, 204.	2.4	2
90	Discriminant value of repetitive behaviors in families with autism spectrum disorder and obsessional compulsive disorder probands. Autism Research, 2021, 14, 2373-2382.	2.1	2

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91	The Synaptic Gene Study: Design and Methodology to Identify Neurocognitive Markers in Phelan-McDermid Syndrome and NRXN1 Deletions. Frontiers in Neuroscience, 2022, 16, 806990.	1.4	2