

# Thomas Bourgeron

## List of Publications by Year in descending order

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Version: 2024-02-01

91  
papers

13,698  
citations

66343

42  
h-index

43889

91  
g-index

111  
all docs

111  
docs citations

111  
times ranked

15156  
citing authors

#	ARTICLE	IF	CITATIONS
1	Functional impact of global rare copy number variation in autism spectrum disorders. <i>Nature</i> , 2010, 466, 368-372.	27.8	1,803
2	Mutations of the X-linked genes encoding neuroligins NLGN3 and NLGN4 are associated with autism. <i>Nature Genetics</i> , 2003, 34, 27-29.	21.4	1,612
3	Mutations in the gene encoding the synaptic scaffolding protein SHANK3 are associated with autism spectrum disorders. <i>Nature Genetics</i> , 2007, 39, 25-27.	21.4	1,408
4	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2014, 94, 677-694.	6.2	819
5	From the genetic architecture to synaptic plasticity in autism spectrum disorder. <i>Nature Reviews Neuroscience</i> , 2015, 16, 551-563.	10.2	764
6	A synaptic trek to autism. <i>Current Opinion in Neurobiology</i> , 2009, 19, 231-234.	4.2	596
7	Autistic-like behaviours and hyperactivity in mice lacking ProSAP1/Shank2. <i>Nature</i> , 2012, 486, 256-260.	27.8	570
8	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. <i>PLoS Genetics</i> , 2014, 10, e1004580.	3.5	501
9	Searching for ways out of the autism maze: genetic, epigenetic and environmental clues. <i>Trends in Neurosciences</i> , 2006, 29, 349-358.	8.6	498
10	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. <i>PLoS Genetics</i> , 2012, 8, e1002521.	3.5	358
11	The Genetic Landscapes of Autism Spectrum Disorders. <i>Annual Review of Genomics and Human Genetics</i> , 2013, 14, 191-213.	6.2	352
12	Key role for gene dosage and synaptic homeostasis in autism spectrum disorders. <i>Trends in Genetics</i> , 2010, 26, 363-372.	6.7	296
13	SHANK1 Deletions in Males with Autism Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2012, 90, 879-887.	6.2	292
14	The EU-AIMS Longitudinal European Autism Project (LEAP): design and methodologies to identify and validate stratification biomarkers for autism spectrum disorders. <i>Molecular Autism</i> , 2017, 8, 24.	4.9	183
15	Progress toward treatments for synaptic defects in autism. <i>Nature Medicine</i> , 2013, 19, 685-694.	30.7	167
16	Adult Male Mice Emit Context-Specific Ultrasonic Vocalizations That Are Modulated by Prior Isolation or Group Rearing Environment. <i>PLoS ONE</i> , 2012, 7, e29401.	2.5	154
17	Gender differences in autism spectrum disorders: Divergence among specific core symptoms. <i>Autism Research</i> , 2017, 10, 680-689.	3.8	140
18	Behavioral profiles of mouse models for autism spectrum disorders. <i>Autism Research</i> , 2011, 4, 5-16.	3.8	133

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19	The EU-AIMS Longitudinal European Autism Project (LEAP): clinical characterisation. <i>Molecular Autism</i> , 2017, 8, 27.	4.9	126
20	Identification and validation of biomarkers for autism spectrum disorders. <i>Nature Reviews Drug Discovery</i> , 2016, 15, 70-70.	46.4	117
21	Real-time analysis of the behaviour of groups of mice via a depth-sensing camera and machine learning. <i>Nature Biomedical Engineering</i> , 2019, 3, 930-942.	22.5	112
22	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. <i>Nature Communications</i> , 2019, 10, 4919.	12.8	111
23	Insufficient Evidence for "Autism-Specific" Genes. <i>American Journal of Human Genetics</i> , 2020, 106, 587-595.	6.2	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. <i>Biological Psychiatry</i> , 2015, 78, 126-134.	1.3	108
25	Current knowledge on the genetics of autism and propositions for future research. <i>Comptes Rendus - Biologies</i> , 2016, 339, 300-307.	0.2	97
26	Genome-wide analyses of self-reported empathy: correlations with autism, schizophrenia, and anorexia nervosa. <i>Translational Psychiatry</i> , 2018, 8, 35.	4.8	95
27	Disruption of melatonin synthesis is associated with impaired 14-3-3 and miR-451 levels in patients with autism spectrum disorders. <i>Scientific Reports</i> , 2017, 7, 2096.	3.3	83
28	Genome-wide association scan identifies new variants associated with a cognitive predictor of dyslexia. <i>Translational Psychiatry</i> , 2019, 9, 77.	4.8	82
29	Altered Connectivity Between Cerebellum, Visual, and Sensory-Motor Networks in Autism Spectrum Disorder: Results from the EU-AIMS Longitudinal European Autism Project. <i>Biological Psychiatry: Cognitive Neuroscience and Neuroimaging</i> , 2019, 4, 260-270.	1.5	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. <i>EBioMedicine</i> , 2016, 9, 293-305.	6.1	79
31	Measuring and Estimating the Effect Sizes of Copy Number Variants on General Intelligence in Community-Based Samples. <i>JAMA Psychiatry</i> , 2018, 75, 447.	11.0	77
32	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. <i>European Journal of Human Genetics</i> , 2018, 26, 1759-1772.	2.8	73
33	Cerebellar Volume in Autism: Literature Meta-analysis and Analysis of the Autism Brain Imaging Data Exchange Cohort. <i>Biological Psychiatry</i> , 2018, 83, 579-588.	1.3	59
34	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. <i>American Journal of Human Genetics</i> , 2019, 104, 815-834.	6.2	59
35	The role of cholesterol metabolism and various steroid abnormalities in autism spectrum disorders: A hypothesis paper. <i>Autism Research</i> , 2017, 10, 1022-1044.	3.8	58
36	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. <i>Npj Genomic Medicine</i> , 2017, 2, 32.	3.8	58

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37	Social and non-social autism symptoms and trait domains are genetically dissociable. <i>Communications Biology</i> , 2019, 2, 328.	4.4	57
38	Zinc deficiency and low enterocyte zinc transporter expression in human patients with autism related mutations in SHANK3. <i>Scientific Reports</i> , 2017, 7, 45190.	3.3	56
39	Genome-wide association study reveals new insights into the heritability and genetic correlates of developmental dyslexia. <i>Molecular Psychiatry</i> , 2021, 26, 3004-3017.	7.9	56
40	Genetic and Environmental Influences on the Visual Word Form and Fusiform Face Areas. <i>Cerebral Cortex</i> , 2015, 25, 2478-2493.	2.9	54
41	Crystal structure and functional mapping of human ASMT, the last enzyme of the melatonin synthesis pathway. <i>Journal of Pineal Research</i> , 2013, 54, 46-57.	7.4	51
42	Altered spinogenesis in iPSC-derived cortical neurons from patients with autism carrying de novo SHANK3 mutations. <i>Scientific Reports</i> , 2019, 9, 94.	3.3	51
43	Operative list of genes associated with autism and neurodevelopmental disorders based on database review. <i>Molecular and Cellular Neurosciences</i> , 2021, 113, 103623.	2.2	51
44	Genetic correlates of phenotypic heterogeneity in autism. <i>Nature Genetics</i> , 2022, 54, 1293-1304.	21.4	51
45	Effect Sizes of Deletions and Duplications on Autism Risk Across the Genome. <i>American Journal of Psychiatry</i> , 2021, 178, 87-98.	7.2	50
46	Recording Mouse Ultrasonic Vocalizations to Evaluate Social Communication. <i>Journal of Visualized Experiments</i> , 2016, , .	0.3	47
47	HyPyP: a Hyperscanning Python Pipeline for inter-brain connectivity analysis. <i>Social Cognitive and Affective Neuroscience</i> , 2021, 16, 72-83.	3.0	46
48	Dynamics in enzymatic protein complexes offer a novel principle for the regulation of melatonin synthesis in the human pineal gland. <i>Journal of Pineal Research</i> , 2011, 51, 145-155.	7.4	45
49	A de novo microdeletion of SEMA5A in a boy with autism spectrum disorder and intellectual disability. <i>European Journal of Human Genetics</i> , 2016, 24, 838-843.	2.8	40
50	Fractionating autism based on neuroanatomical normative modeling. <i>Translational Psychiatry</i> , 2020, 10, 384.	4.8	40
51	Autism-associated SHANK3 mutations impair maturation of neuromuscular junctions and striated muscles. <i>Science Translational Medicine</i> , 2020, 12, .	12.4	38
52	Alpha Waves as a Neuromarker of Autism Spectrum Disorder: The Challenge of Reproducibility and Heterogeneity. <i>Frontiers in Neuroscience</i> , 2018, 12, 662.	2.8	37
53	Mutations associated with neuropsychiatric conditions delineate functional brain connectivity dimensions contributing to autism and schizophrenia. <i>Nature Communications</i> , 2020, 11, 5272.	12.8	35
54	Lack of replication of previous autism spectrum disorder GWAS hits in European populations. <i>Autism Research</i> , 2017, 10, 202-211.	3.8	34

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55	Genome-wide analysis of gene dosage in 24,092 individuals estimates that 10,000 genes modulate cognitive ability. <i>Molecular Psychiatry</i> , 2021, 26, 2663-2676.	7.9	33
56	Dissecting the phenotypic heterogeneity in sensory features in autism spectrum disorder: a factor mixture modelling approach. <i>Molecular Autism</i> , 2020, 11, 67.	4.9	32
57	Phelan-McDermid syndrome: a classification system after 30 years of experience. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 27.	2.7	32
58	Heritability of the melatonin synthesis variability in autism spectrum disorders. <i>Scientific Reports</i> , 2017, 7, 17746.	3.3	28
59	Sequencing ASMT Identifies Rare Mutations in Chinese Han Patients with Autism. <i>PLoS ONE</i> , 2013, 8, e53727.	2.5	26
60	The meaning of significant mean group differences for biomarker discovery. <i>PLoS Computational Biology</i> , 2021, 17, e1009477.	3.2	26
61	11q24.2-25 microrearrangements in autism spectrum disorders: Relation to brain structures. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3019-3030.	1.2	25
62	Dlx5 and Dlx6 expression in GABAergic neurons controls behavior, metabolism, healthy aging and lifespan. <i>Aging</i> , 2019, 11, 6638-6656.	3.1	25
63	Systematic detection of brain protein-coding genes under positive selection during primate evolution and their roles in cognition. <i>Genome Research</i> , 2021, 31, 484-496.	5.5	25
64	Insights from an autism imaging biomarker challenge: Promises and threats to biomarker discovery. <i>NeuroImage</i> , 2022, 255, 119171.	4.2	24
65	mouseTube – a database to collaboratively unravel mouse ultrasonic communication. <i>F1000Research</i> , 2016, 5, 2332.	1.6	23
66	Behavioural Phenotypes and Neural Circuit Dysfunctions in Mouse Models of Autism Spectrum Disorder. <i>Advances in Anatomy, Embryology and Cell Biology</i> , 2017, 224, 85-101.	1.6	21
67	Shank2 Mutant Mice Display Hyperactivity Insensitive to Methylphenidate and Reduced Flexibility in Social Motivation, but Normal Social Recognition. <i>Frontiers in Molecular Neuroscience</i> , 2018, 11, 365.	2.9	21
68	Morning Plasma Melatonin Differences in Autism: Beyond the Impact of Pineal Gland Volume. <i>Frontiers in Psychiatry</i> , 2019, 10, 11.	2.6	21
69	Increased risk of ADHD in families with ASD. <i>European Child and Adolescent Psychiatry</i> , 2019, 28, 281-288.	4.7	19
70	Imbalanced social-communicative and restricted repetitive behavior subtypes of autism spectrum disorder exhibit different neural circuitry. <i>Communications Biology</i> , 2021, 4, 574.	4.4	17
71	LMT USV Toolbox, a Novel Methodological Approach to Place Mouse Ultrasonic Vocalizations in Their Behavioral Contexts – A Study in Female and Male C57BL/6J Mice and in Shank3 Mutant Females. <i>Frontiers in Behavioral Neuroscience</i> , 2021, 15, 735920.	2.0	17
72	The genetics and neurobiology of ESSENCE: The third Birgit Olsson lecture. <i>Nordic Journal of Psychiatry</i> , 2016, 70, 1-9.	1.3	16

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73	Polygenic Architecture of Human Neuroanatomical Diversity. <i>Cerebral Cortex</i> , 2020, 30, 2307-2320.	2.9	16
74	Social Communication in Mice – Are There Optimal Cage Conditions?. <i>PLoS ONE</i> , 2015, 10, e0121802.	2.5	15
75	Heterogeneous Pattern of Selective Pressure for PRRT2 in Human Populations, but No Association with Autism Spectrum Disorders. <i>PLoS ONE</i> , 2014, 9, e88600.	2.5	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. <i>Journal of Applied Genetics</i> , 2019, 60, 49-56.	1.9	11
77	Decreased phenol sulfotransferase activities associated with hyperserotonemia in autism spectrum disorders. <i>Translational Psychiatry</i> , 2021, 11, 23.	4.8	11
78	Anatomy and Cell Biology of Autism Spectrum Disorder: Lessons from Human Genetics. <i>Advances in Anatomy, Embryology and Cell Biology</i> , 2017, 224, 1-25.	1.6	10
79	Editorial: Shankopathies: Shank Protein Deficiency-Induced Synaptic Diseases. <i>Frontiers in Molecular Neuroscience</i> , 2020, 13, 11.	2.9	9
80	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. <i>Npj Genomic Medicine</i> , 2021, 6, 91.	3.8	9
81	Testosterone Increases the Emission of Ultrasonic Vocalizations With Different Acoustic Characteristics in Mice. <i>Frontiers in Psychology</i> , 2021, 12, 680176.	2.1	8
82	Synesthesia & autistic features in a large family: Evidence for spatial imagery as a common factor. <i>Behavioural Brain Research</i> , 2019, 362, 266-272.	2.2	7
83	A chimeric mouse model to study human iPSC-derived neurons: the case of a truncating SHANK3 mutation. <i>Scientific Reports</i> , 2020, 10, 13315.	3.3	7
84	Interactive Psychometrics for Autism With the Human Dynamic Clamp: Interpersonal Synchrony From Sensorimotor to Sociocognitive Domains. <i>Frontiers in Psychiatry</i> , 2020, 11, 510366.	2.6	7
85	Three-dimensional Quantification of Dendritic Spines from Pyramidal Neurons Derived from Human Induced Pluripotent Stem Cells. <i>Journal of Visualized Experiments</i> , 2015, , .	0.3	6
86	The functional database of the ARCH1 project: Potential and perspectives. <i>NeuroImage</i> , 2019, 197, 527-543.	4.2	6
87	Mass spectrometry analysis of the human pineal proteome during night and day and in autism. <i>Journal of Pineal Research</i> , 2021, 70, e12713.	7.4	4
88	Reduced 3-methoxydopa levels in OCD patients and their unaffected parents is associated with the low activity M158 COMT allele. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 542-548.	1.7	3
89	The role of rare compound heterozygous events in autism spectrum disorder. <i>Translational Psychiatry</i> , 2020, 10, 204.	4.8	2
90	Discriminant value of repetitive behaviors in families with autism spectrum disorder and obsessional compulsive disorder probands. <i>Autism Research</i> , 2021, 14, 2373-2382.	3.8	2

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91	The Synaptic Gene Study: Design and Methodology to Identify Neurocognitive Markers in Phelan-McDermid Syndrome and NRXN1 Deletions. <i>Frontiers in Neuroscience</i> , 2022, 16, 806990.	2.8	2