Raphael A Bernier

List of Publications by Year
in descending order

85 papers	8,156 citations	101543 36 h-index	58581 82 g-index
90 all docs	90 docs citations	90 times ranked	12207 citing authors

#	Article	IF	Citations
1	Impact of autism genetic risk on brain connectivity: a mechanism for the female protective effect. Brain, 2022, 145, 378-387.	7.6	9
2	Brief Report: Associations Between Self-injurious Behaviors and Abdominal Pain Among Individuals with ASD-Associated Disruptive Mutations. Journal of Autism and Developmental Disorders, 2021, 51, 3365-3373.	2.7	5
3	Brief Report: Can a Composite Heart Rate Variability Biomarker Shed New Insights About Autism Spectrum Disorder in School-Aged Children?. Journal of Autism and Developmental Disorders, 2021, 51, 346-356.	2.7	11
4	The gap between IQ and adaptive functioning in autism spectrum disorder: Disentangling diagnostic and sex differences. Autism, 2021, 25, 1565-1579.	4.1	23
5	Clinical delineation of SETBP1 haploinsufficiency disorder. European Journal of Human Genetics, 2021, 29, 1198-1205.	2.8	12
6	A neurogenetic analysis of female autism. Brain, 2021, 144, 1911-1926.	7.6	24
7	Sleep Problems in Children with ASD and Gene Disrupting Mutations. Journal of Genetic Psychology, 2021, 182, 317-334.	1.2	3
8	Reflections on the genetics-first approach to advancements in molecular genetic and neurobiological research on neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2021, 13, 24.	3.1	12
9	The CHD8/CHD7/Kismet family links blood-brain barrier glia and serotonin to ASD-associated sleep defects. Science Advances, 2021, 7, .	10.3	24
10	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	21.4	68
11	Coding and noncoding variants in EBF3 are involved in HADDS and simplex autism. Human Genomics, 2021, 15, 44.	2.9	16
12	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
13	Clinical Phenotypes of Carriers of Mutations in CHD8 or Its Conserved Target Genes. Biological Psychiatry, 2020, 87, 123-131.	1.3	22
14	Language characterization in 16p11.2 deletion and duplication syndromes. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 380-391.	1.7	16
15	Late fMRI Response Components Are Altered in Autism Spectrum Disorder. Frontiers in Human Neuroscience, 2020, 14, 241.	2.0	5
16	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. American Journal of Human Genetics, 2020, 107, 555-563.	6.2	32
17	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976.	6.2	18
18	Neural responsivity to social rewards in autistic female youth. Translational Psychiatry, 2020, 10, 178.	4.8	22

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19	Weaker neural suppression in autism. Nature Communications, 2020, 11, 2675.	12.8	28
20	Imaging-genetics of sex differences in ASD: distinct effects of OXTR variants on brain connectivity. Translational Psychiatry, 2020, 10, 82.	4.8	31
21	Evaluating heterogeneity in <scp>ASD</scp> symptomatology, cognitive ability, and adaptive functioning among 16p11.2 <scp>CNV</scp> carriers. Autism Research, 2020, 13, 1300-1310.	3.8	23
22	Response Dissociation in Hierarchical Cortical Circuits: a Unique Feature of Autism Spectrum Disorder. Journal of Neuroscience, 2020, 40, 2269-2281.	3.6	13
23	Day-to-Day Test-Retest Reliability of EEG Profiles in Children With Autism Spectrum Disorder and Typical Development. Frontiers in Integrative Neuroscience, 2020, 14, 21.	2.1	32
24	Sex Differences in Functional Connectivity of the Salience, Default Mode, and Central Executive Networks in Youth with ASD. Cerebral Cortex, 2020, 30, 5107-5120.	2.9	46
25	Co-occurring medical conditions among individuals with ASD-associated disruptive mutations. Children's Health Care, 2020, 49, 361-384.	0.9	3
26	Concentrations of Cortical <scp>GABA</scp> and Glutamate in Young Adults With Autism Spectrum Disorder. Autism Research, 2020, 13, 1111-1129.	3.8	38
27	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	16.3	83
28	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
29	The state of research on the genetics of autism spectrum disorder: methodological, clinical and conceptual progress. Current Opinion in Psychology, 2019, 27, 1-5.	4.9	27
30	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
31	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
32	The Human-Specific BOLA2 Duplication Modifies Iron Homeostasis and Anemia Predisposition in Chromosome 16p11.2 Autism Individuals. American Journal of Human Genetics, 2019, 105, 947-958.	6.2	30
33	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
34	Psychiatric disorders in children with 16p11.2 deletion and duplication. Translational Psychiatry, 2019, 9, 8.	4.8	93
35	Î±7 Nicotinic Acetylcholine Receptor Signaling Modulates Ovine Fetal Brain Astrocytes Transcriptome in Response to Endotoxin. Frontiers in Immunology, 2019, 10, 1063.	4.8	18
36	Methodological considerations in the use of Noldus EthoVision XT video tracking of children with autism in multi-site studies. Biological Psychology, 2019, 146, 107712.	2.2	10

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37	Habituation Learning Is a Widely Affected Mechanism in Drosophila Models of Intellectual Disability and Autism Spectrum Disorders. Biological Psychiatry, 2019, 86, 294-305.	1.3	39
38	Sex-Based Analysis of De Novo Variants in Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 1274-1285.	6.2	84
39	Neurodevelopmental disease genes implicated by de novo mutation and copy number variation morbidity. Nature Genetics, 2019, 51, 106-116.	21.4	231
40	Glutamatergic facilitation of neural responses in MT enhances motion perception in humans. NeuroImage, 2019, 184, 925-931.	4.2	16
41	Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. Genetics in Medicine, 2019, 21, 1611-1620.	2.4	88
42	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
43	Progress in Understanding and Treating SCN2A-Mediated Disorders. Trends in Neurosciences, 2018, 41, 442-456.	8.6	210
44	Longitudinal report of child with de novo 16p11.2 triplication. Clinical Case Reports (discontinued), 2018, 6, 147-154.	0.5	5
45	Interdisciplinary Team Evaluation: An Effective Method for the Diagnostic Assessment of Autism Spectrum Disorder. Journal of Developmental and Behavioral Pediatrics, 2018, 39, 271-281.	1.1	18
46	Child and family characteristics moderate agreement between caregiver and clinician report of autism symptoms. Autism Research, 2018, 11, 476-487.	3.8	19
47	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
48	Early enhanced processing and delayed habituation to deviance sounds in autism spectrum disorder. Brain and Cognition, 2018, 123, 110-119.	1.8	53
49	Comorbid symptoms of inattention, autism, and executive cognition in youth with putative genetic risk. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2018, 59, 268-276.	5.2	8
50	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
51	Inherited and multiple de novo mutations in autism/developmental delay risk genes suggest a multifactorial model. Molecular Autism, 2018, 9, 64.	4.9	114
52	Auditory perception is associated with implicit language learning and receptive language ability in autism spectrum disorder. Brain and Language, 2018, 187, 1-8.	1.6	9
53	Suppression and facilitation of human neural responses. ELife, 2018, 7, .	6.0	48
54	Sex Differences in Visual Motion Processing. Current Biology, 2018, 28, 2794-2799.e3.	3.9	35

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55	The autism spectrum phenotype in ADNP syndrome. Autism Research, 2018, 11, 1300-1310.	3.8	49
56	Reduced auditory cortical adaptation in autism spectrum disorder. ELife, 2018, 7, .	6.0	41
57	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
58	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
59	Developmental trajectories for young children with 16p11.2 copy number variation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 367-380.	1.7	42
60	denovo-db: a compendium of humande novovariants. Nucleic Acids Research, 2017, 45, D804-D811.	14.5	173
61	Gene Disrupting Mutations Associated with Regression in Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2017, 47, 3600-3607.	2.7	26
62	Exploring the heterogeneity of neural social indices for genetically distinct etiologies of autism. Journal of Neurodevelopmental Disorders, 2017, 9, 24.	3.1	19
63	Associations between Familial Rates of Psychiatric Disorders and De Novo Genetic Mutations in Autism. Autism Research & Treatment, 2017, 2017, 1-9.	0.5	4
64	Prospective investigation of FOXP1 syndrome. Molecular Autism, 2017, 8, 57.	4.9	65
65	Clinical phenotype of ASD-associated DYRK1A haploinsufficiency. Molecular Autism, 2017, 8, 54.	4.9	55
66	Autism Spectrum Disorder, Developmental and Psychiatric Features in 16p11.2 Duplication. Journal of Autism and Developmental Disorders, 2016, 46, 2734-2748.	2.7	47
67	Children with Autism Show Altered Autonomic Adaptation to Novel and Familiar Social Partners. Autism Research, 2016, 9, 579-591.	3.8	41
68	Infantsâ€™ grip strength predicts mu rhythm attenuation during observation of lifting actions with weighted blocks. Developmental Science, 2016, 19, 195-207.	2.4	10
69	The Number of Genomic Copies at the 16p11.2 Locus Modulates Language, Verbal Memory, and Inhibition. Biological Psychiatry, 2016, 80, 129-139.	1.3	78
70	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
71	Maternal Modifiers and Parent-of-Origin Bias of the Autism-Associated 16p11.2 CNV. American Journal of Human Genetics, 2016, 98, 45-57.	6.2	55
72	Face processing among twins with and without autism: social correlates and twin concordance. Social Cognitive and Affective Neuroscience, 2016, 11, 44-54.	3.0	24

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73	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
74	Clinical phenotype of the recurrent 1q21.1 copy-number variant. Genetics in Medicine, 2016, 18, 341-349.	2.4	134
75	Modulation of mu attenuation to social stimuli in children and adults with 16p11.2 deletions and duplications. Journal of Neurodevelopmental Disorders, 2015, 7, 25.	3.1	12
76	Electrodermal Response to Reward and Nonâ€Reward Among Children With Autism. Autism Research, 2015, 8, 357-370.	3.8	22
77	The Role of Parental Cognitive, Behavioral, and Motor Profiles in Clinical Variability in Individuals With Chromosome 16p11.2 Deletions. JAMA Psychiatry, 2015, 72, 119.	11.0	112
78	Excess of rare, inherited truncating mutations in autism. Nature Genetics, 2015, 47, 582-588.	21.4	531
79	The Cognitive and Behavioral Phenotype of the 16p11.2 Deletion in a Clinically Ascertained Population. Biological Psychiatry, 2015, 77, 785-793.	1.3	198
80	The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 315-326.	1.6	68
81	De novo TBR1 mutations in sporadic autism disrupt protein functions. Nature Communications, 2014, 5, 4954.	12.8	109
82	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
83	A 600â€…kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. Journal of Medical Genetics, 2012, 49, 660-668.	3.2	251
84	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. Nature, 2012, 485, 246-250.	27.8	1,960
85	The Broader Autism Phenotype and Its Implications on the Etiology and Treatment of Autism Spectrum Disorders. Autism Research & Treatment, 2011, 2011, 1-19.	0.5	103