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	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. Nature, 2012, 485, 246-250.	27.8	1,960
2	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
3	Excess of rare, inherited truncating mutations in autism. Nature Genetics, 2015, 47, 582-588.	21.4	531
4	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
5	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
6	Neurodevelopmental disease genes implicated by de novo mutation and copy number variation morbidity. Nature Genetics, 2019, 51, 106-116.	21.4	231
7	Progress in Understanding and Treating SCN2A-Mediated Disorders. Trends in Neurosciences, 2018, 41, 442-456.	8.6	210
8	The Cognitive and Behavioral Phenotype of the 16p11.2 Deletion in a Clinically Ascertained Population. Biological Psychiatry, 2015, 77, 785-793.	1.3	198
9	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
10	denovo-db: a compendium of humande novovariants. Nucleic Acids Research, 2017, 45, D804-D811.	14.5	173
11	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
12	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
13	Clinical phenotype of the recurrent 1q21.1 copy-number variant. Genetics in Medicine, 2016, 18, 341-349.	2.4	134
14	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
15	Inherited and multiple de novo mutations in autism/developmental delay risk genes suggest a multifactorial model. Molecular Autism, 2018, 9, 64.	4.9	114
16	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
17	De novo TBR1 mutations in sporadic autism disrupt protein functions. Nature Communications, 2014, 5, 4954.	12.8	109
18	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108

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19	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
20	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
21	Psychiatric disorders in children with 16p11.2 deletion and duplication. Translational Psychiatry, 2019, 9, 8.	4.8	93
22	Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. Genetics in Medicine, 2019, 21, 1611-1620.	2.4	88
23	Sex-Based Analysis of De Novo Variants in Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 1274-1285.	6.2	84
24	A framework for an evidence-based gene list relevant to autism spectrum disorder. Nature Reviews Genetics, 2020, 21, 367-376.	16.3	83
25	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
26	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
27	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	21.4	68
28	Prospective investigation of FOXP1 syndrome. Molecular Autism, 2017, 8, 57.	4.9	65
29	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
30	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
31	Clinical phenotype of ASD-associated DYRK1A haploinsufficiency. Molecular Autism, 2017, 8, 54.	4.9	55
32	Early enhanced processing and delayed habituation to deviance sounds in autism spectrum disorder. Brain and Cognition, 2018, 123, 110-119.	1.8	53
33	The autism spectrum phenotype in ADNP syndrome. Autism Research, 2018, 11, 1300-1310.	3.8	49
34	Suppression and facilitation of human neural responses. ELife, 2018, 7, .	6.0	48
35	Autism Spectrum Disorder, Developmental and Psychiatric Features in 16p11.2 Duplication. Journal of Autism and Developmental Disorders, 2016, 46, 2734-2748.	2.7	47
36	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

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37	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
38	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
39	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
40	Children with Autism Show Altered Autonomic Adaptation to Novel and Familiar Social Partners. Autism Research, 2016, 9, 579-591.	3.8	41
41	Reduced auditory cortical adaptation in autism spectrum disorder. ELife, 2018, 7, .	6.0	41
42	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
43	Concentrations of Cortical <scp>GABA</scp> and Glutamate in Young Adults With Autism Spectrum Disorder. Autism Research, 2020, 13, 1111-1129.	3.8	38
44	Sex Differences in Visual Motion Processing. Current Biology, 2018, 28, 2794-2799.e3.	3.9	35
45	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
46	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
47	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. American Journal of Human Genetics, 2020, 107, 555-563.	6.2	32
48	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
49	Imaging-genetics of sex differences in ASD: distinct effects of OXTR variants on brain connectivity. Translational Psychiatry, 2020, 10, 82.	4.8	31
50	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
51	Weaker neural suppression in autism. Nature Communications, 2020, 11, 2675.	12.8	28
52	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
53	Gene Disrupting Mutations Associated with Regression in Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2017, 47, 3600-3607.	2.7	26
54	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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55	A neurogenetic analysis of female autism. Brain, 2021, 144, 1911-1926.	7.6	24
56	The CHD8/CHD7/Kismet family links blood-brain barrier glia and serotonin to ASD-associated sleep defects. Science Advances, 2021, 7, .	10.3	24
57	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
58	The gap between IQ and adaptive functioning in autism spectrum disorder: Disentangling diagnostic and sex differences. Autism, 2021, 25, 1565-1579.	4.1	23
59	Electrodermal Response to Reward and Nonâ€Reward Among Children With Autism. Autism Research, 2015, 8, 357-370.	3.8	22
60	Clinical Phenotypes of Carriers of Mutations in CHD8 or Its Conserved Target Genes. Biological Psychiatry, 2020, 87, 123-131.	1.3	22
61	Neural responsivity to social rewards in autistic female youth. Translational Psychiatry, 2020, 10, 178.	4.8	22
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	Child and family characteristics moderate agreement between caregiver and clinician report of autism symptoms. Autism Research, 2018, 11, 476-487.	3.8	19
64	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
65	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
66	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
67	Glutamatergic facilitation of neural responses in MT enhances motion perception in humans. NeuroImage, 2019, 184, 925-931.	4.2	16
68	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
69	Coding and noncoding variants in EBF3 are involved in HADDS and simplex autism. Human Genomics, 2021, 15, 44.	2.9	16
70	Response Dissociation in Hierarchical Cortical Circuits: a Unique Feature of Autism Spectrum Disorder. Journal of Neuroscience, 2020, 40, 2269-2281.	3.6	13
71	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
72	Clinical delineation of SETBP1 haploinsufficiency disorder. European Journal of Human Genetics, 2021, 29, 1198-1205.	2.8	12

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73	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
74	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
75	Infantsâ€™ grip strength predicts mu rhythm attenuation during observation of lifting actions with weighted blocks. Developmental Science, 2016, 19, 195-207.	2.4	10
76	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
77	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
78	Impact of autism genetic risk on brain connectivity: a mechanism for the female protective effect. Brain, 2022, 145, 378-387.	7.6	9
79	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
80	Longitudinal report of child with de novo 16p11.2 triplication. Clinical Case Reports (discontinued), 2018, 6, 147-154.	0.5	5
81	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
82	Late fMRI Response Components Are Altered in Autism Spectrum Disorder. Frontiers in Human Neuroscience, 2020, 14, 241.	2.0	5
83	Associations between Familial Rates of Psychiatric Disorders and De Novo Genetic Mutations in Autism. Autism Research & Treatment, 2017, 2017, 1-9.	0.5	4
84	Co-occurring medical conditions among individuals with ASD-associated disruptive mutations. Children's Health Care, 2020, 49, 361-384.	0.9	3
85	Sleep Problems in Children with ASD and Gene Disrupting Mutations. Journal of Genetic Psychology, 2021, 182, 317-334.	1.2	3