Detection of Clinically Relevant Genetic Variants in Aut Whole-Genome Sequencing

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Citation Report

#	Article	IF	Citations
1	Identification of candidate intergenic risk loci in autism spectrum disorder. BMC Genomics, 2013, 14, 499.	1.2	51
2	Small study yields big results for whole genome sequencing in autism diagnosis: Geneticists optimistic that findings could aid with early identification of disorder. American Journal of Medical Genetics, Part A, 2013, 161, vii.	0.7	1
3	Transcriptomic analysis of genetically defined autism candidate genes reveals common mechanisms of action. Molecular Autism, 2013, 4, 45.	2.6	43
4	Transcriptional Dysregulation of Neocortical Circuit Assembly in ASD. International Review of Neurobiology, 2013, 113, 167-205.	0.9	31
5	High-throughput sequencing of autism spectrum disorders comes of age. Genetical Research, 2013, 95, 121-129.	0.3	4
7	Architects of the genome: CHD dysfunction in cancer, developmental disorders and neurological syndromes. Epigenomics, 2014, 6, 381-395.	1.0	40
8	Genomics in Neurological Disorders. Genomics, Proteomics and Bioinformatics, 2014, 12, 156-163.	3.0	23
9	Progressive increase in mtDNA 3243A>G heteroplasmy causes abrupt transcriptional reprogramming. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4033-42.	3.3	251
10	GWATCH: a web platform for automated gene association discovery analysis. GigaScience, 2014, 3, 18.	3.3	5
11	Towards a molecular characterization of autism spectrum disorders: an exome sequencing and systems approach. Translational Psychiatry, 2014, 4, e394-e394.	2.4	57
12	Finding Novel Molecular Connections between Developmental Processes and Disease. PLoS Computational Biology, 2014, 10, e1003578.	1.5	10
13	Contribution of highâ€throughput DNA sequencing to the study of primary immunodeficiencies. European Journal of Immunology, 2014, 44, 2854-2861.	1.6	56
14	The intriguing relationship between cerebral palsy and autism. Developmental Medicine and Child Neurology, 2014, 56, 7-8.	1.1	7
15	Cellular evidence for selfish spermatogonial selection in aged human testes. Andrology, 2014, 2, 304-314.	1.9	60
16	Exon resequencing of H3K9 methyltransferase complex genes, EHMT1, EHTM2 and WIZ, in Japanese autism subjects. Molecular Autism, 2014, 5, 49.	2.6	26
17	Clinical Application of Whole-Genome Sequencing. JAMA - Journal of the American Medical Association, 2014, 311, 1017.	3.8	54
18	Intellectual disability and autism spectrum disorders: Causal genes and molecular mechanisms. Neuroscience and Biobehavioral Reviews, 2014, 46, 161-174.	2.9	181
19	Exome sequencing identifies de novo gain of function missense mutation in KCND2 in identical twins with autism and seizures that slows potassium channel inactivation. Human Molecular Genetics, 2014, 23, 3481-3489.	1.4	90

#	Article	IF	Citations
20	Glutamatergic candidate genes in autism spectrum disorder: an overview. Journal of Neural Transmission, 2014, 121, 1081-1106.	1.4	23
21	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	2.6	819
22	Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. Nature Genetics, 2014, 46, 742-747.	9.4	149
23	Prioritization of neurodevelopmental disease genes by discovery of new mutations. Nature Neuroscience, 2014, 17, 764-772.	7.1	148
24	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Human Molecular Genetics, 2014, 23, 2752-2768.	1.4	140
25	Genetics of recessive cognitive disorders. Trends in Genetics, 2014, 30, 32-39.	2.9	120
26	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
27	Reducing False-Positive Incidental Findings with Ensemble Genotyping and Logistic Regression Based Variant Filtering Methods. Human Mutation, 2014, 35, 936-944.	1.1	10
28	Evolutionary and Molecular Facts Link the WWC Protein Family to Hippo Signaling. Molecular Biology and Evolution, 2014, 31, 1710-1723.	3.5	57
29	Using familial information for variant filtering in high-throughput sequencing studies. Human Genetics, 2014, 133, 1331-1341.	1.8	10
30	Promoter Decommissioning by the NuRD Chromatin Remodeling Complex Triggers Synaptic Connectivity in the Mammalian Brain. Neuron, 2014, 83, 122-134.	3.8	92
31	Next Generation Sequencing and the Future of Genetic Diagnosis. Neurotherapeutics, 2014, 11, 699-707.	2.1	126
32	Advances in Genetic Discovery and Implications for Counseling of Patients and Families with Autism Spectrum Disorders. Current Genetic Medicine Reports, 2014, 2, 124-134.	1.9	7
33	Copy number variation in Han Chinese individuals with autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2014, 6, 34.	1.5	55
34	De novoand rare inherited mutations implicate the transcriptional coregulator TCF20/SPBP in autism spectrum disorder. Journal of Medical Genetics, 2014, 51, 737-747.	1.5	31
35	Etiology of Autism Spectrum Disorder: A Genomics Perspective. Current Psychiatry Reports, 2014, 16, 501.	2.1	12
36	A Twin Study of Heritable and Shared Environmental Contributions to Autism. Journal of Autism and Developmental Disorders, 2014, 44, 2013-2025.	1.7	95
37	Transcriptional and functional complexity of Shank3 provides a molecular framework to understand the phenotypic heterogeneity of SHANK3 causing autism and Shank3 mutant mice. Molecular Autism, 2014, 5, 30.	2.6	137

#	Article	IF	Citations
38	The Genome Clinic: A Multidisciplinary Approach to Assessing the Opportunities and Challenges of Integrating Genomic Analysis into Clinical Care. Human Mutation, 2014, 35, 513-519.	1.1	31
39	Exome Sequencing: New Insights into Lipoprotein Disorders. Current Cardiology Reports, 2014, 16, 507.	1.3	17
40	Advances in Genetic Diagnosis of Autism Spectrum Disorders. Current Pediatrics Reports, 2014, 2, 71-81.	1.7	3
41	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	13.5	637
42	Research Review: Polygenic methods and their application to psychiatric traits. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2014, 55, 1068-1087.	3.1	578
43	Determinants of Mutation Rate Variation in the Human Germline. Annual Review of Genomics and Human Genetics, 2014, 15, 47-70.	2.5	295
44	Autism spectrum disorder: advances in evidence-based practice. Cmaj, 2014, 186, 509-519.	0.9	158
45	Genetic diagnosis of autism spectrum disorders: The opportunity and challenge in the genomics era. Critical Reviews in Clinical Laboratory Sciences, 2014, 51, 249-262.	2.7	38
46	Genetic targeting of the amphetamine and methylphenidate-sensitive dopamine transporter: On the path to an animal model of attention-deficit hyperactivity disorder. Neurochemistry International, 2014, 73, 56-70.	1.9	24
47	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	13.7	996
48	From genomes to societies: a holistic view of determinants of human health. Current Opinion in Biotechnology, 2014, 28, 134-142.	3.3	7
49	Current concepts and clinical applications of stroke genetics. Lancet Neurology, The, 2014, 13, 405-418.	4.9	86
50	A CGG-Repeat Expansion Mutation inZNF713Causes FRA7A: Association with Autistic Spectrum Disorder in two Families. Human Mutation, 2014, 35, n/a-n/a.	1.1	28
51	Whole genome sequencing of 35 individuals provides insights into the genetic architecture of Korean population. BMC Bioinformatics, 2014, 15, S6.	1.2	34
52	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Proteinâ€Coding Regions. Human Mutation, 2015, 36, 815-822.	1.1	156
53	CRISPR/Cas9-mediated heterozygous knockout of the autism gene CHD8 and characterization of its transcriptional networks in neurodevelopment. Molecular Autism, 2015, 6, 55.	2.6	135
54	Investigation of sex differences in the expression of RORA and its transcriptional targets in the brain as a potential contributor to the sex bias in autism. Molecular Autism, 2015, 6, 7.	2.6	68
55	Whole-exome analysis of foetal autopsy tissue reveals a frameshift mutation in OBSL1, consistent with a diagnosis of 3-M Syndrome. BMC Genomics, 2015, 16, S12.	1.2	9

#	Article	IF	Citations
56	Performance of case-control rare copy number variation annotation in classification of autism. BMC Medical Genomics, 2015, 8, S7.	0.7	15
57	Genetics of Venous Thrombosis: update in 2015. Thrombosis and Haemostasis, 2015, 114, 910-919.	1.8	81
58	Identification of Distinct Tumor Subpopulations in Lung Adenocarcinoma via Single-Cell RNA-seq. PLoS ONE, 2015, 10, e0135817.	1.1	54
59	The Impact of Neuroimmune Alterations in Autism Spectrum Disorder. Frontiers in Psychiatry, 2015, 6, 121.	1.3	124
60	Chromodomain Helicase DNA-Binding Proteins in Stem Cells and Human Developmental Diseases. Stem Cells and Development, 2015, 24, 917-926.	1.1	38
61	The human gut microbiota with reference to autism spectrum disorder: considering the whole as more than a sum of its parts. Microbial Ecology in Health and Disease, 2015, 26, 26309.	3.8	32
62	Language-related abilities in â€˜unaffectedâ€™ school-aged siblings of children with ASD. Research in Autism Spectrum Disorders, 2015, 18, 83-96.	0.8	12
63	Next-Generation Sequencing in Intellectual Disability. Journal of Pediatric Genetics, 2015, 04, 128-135.	0.3	30
64	Leveraging Distant Relatedness to Quantify Human Mutation and Gene-Conversion Rates. American Journal of Human Genetics, 2015, 97, 775-789.	2.6	77
65	Genomic Perspectives of Transcriptional Regulation in Forebrain Development. Neuron, 2015, 85, 27-47.	3.8	136
66	Advancing parental age and autism: multifactorial pathways. Trends in Molecular Medicine, 2015, 21, 118-125.	3.5	38
67	â€˜Neonatalâ€™ Nav1.2 reduces neuronal excitability and affects seizure susceptibility and behaviour. Human Molecular Genetics, 2015, 24, 1457-1468.	1.4	66
68	Whole-genome sequencing of quartet families with autism spectrum disorder. Nature Medicine, 2015, 21, 185-191.	15.2	457
69	From De Novo Mutations to Personalized Therapeutic Interventions in Autism. Annual Review of Medicine, 2015, 66, 487-507.	5.0	41
70	mirTrios: an integrated pipeline for detection of de novo and rare inherited mutations from trios-based next-generation sequencing. Journal of Medical Genetics, 2015, 52, 275-281.	1.5	35
71	Epigenetic Developmental Disorders: CHARGE Syndrome, a Case Study. Current Genetic Medicine Reports, 2015, 3, 1-7.	1.9	19
72	Integrated Genomics Identifies Convergence of Ankylosing Spondylitis with Global Immune Mediated Disease Pathways. Scientific Reports, 2015, 5, 10314.	1.6	20
73	Autism and Medical Comorbidities. Key Issues in Mental Health, 2015, , 20-33.	0.6	2

#	Article	IF	Citations
74	High-Resolution Chromosome Ideogram Representation of Currently Recognized Genes for Autism Spectrum Disorders. International Journal of Molecular Sciences, 2015, 16, 6464-6495.	1.8	51
75	Discovery of Rare Mutations in Autism: Elucidating Neurodevelopmental Mechanisms. Neurotherapeutics, 2015, 12, 553-571.	2.1	21
76	Characteristics of de novo structural changes in the human genome. Genome Research, 2015, 25, 792-801.	2.4	115
77	Integrated analysis of whole-exome sequencing and transcriptome profiling in males with autism spectrum disorders. Molecular Autism, 2015, 6, 21.	2.6	106
78	Characterization of 26 deletion CNVs reveals the frequent occurrence of micro-mutations within the breakpoint-flanking regions and frequent repair of double-strand breaks by templated insertions derived from remote genomic regions. Human Genetics, 2015, 134, 589-603.	1.8	25
79	The Use of Induced Pluripotent Stem Cell Technology to Advance Autism Research and Treatment. Neurotherapeutics, 2015, 12, 534-545.	2.1	24
80	Epilepsies in Children with 2q24.3 Deletion/Duplication. Journal of Pediatric Epilepsy, 2015, 04, 008-016.	0.1	1
81	Early Identification and Interventions for Autism Spectrum Disorder: Executive Summary. Pediatrics, 2015, 136, S1-S9.	1.0	87
82	The origins, determinants, and consequences of human mutations. Science, 2015, 349, 1478-1483.	6.0	143
83	Somatic mutation in cancer and normal cells. Science, 2015, 349, 1483-1489.	6.0	996
84	Current Tools for Interpretation of Genomic Copy Number Variants. Current Genetic Medicine Reports, 2015, 3, 202-208.	1.9	0
85	Genetic architecture, epigenetic influence and environment exposure in the pathogenesis of Autism. Science China Life Sciences, 2015, 58, 958-967.	2.3	24
86	Penetrance of pathogenic mutations in haploinsufficient genes for intellectual disability and related disorders. European Journal of Medical Genetics, 2015, 58, 715-718.	0.7	31
87	Future of whole genome sequencing. Journal of Paediatrics and Child Health, 2015, 51, 251-254.	0.4	10
88	Early Screening of Autism Spectrum Disorder: Recommendations for Practice and Research. Pediatrics, 2015, 136, S41-S59.	1.0	201
89	From the genetic architecture to synaptic plasticity in autism spectrum disorder. Nature Reviews Neuroscience, 2015, 16, 551-563.	4.9	764
90	Molecular Diagnostic Yield of Chromosomal Microarray Analysis and Whole-Exome Sequencing in Children With Autism Spectrum Disorder. JAMA - Journal of the American Medical Association, 2015, 314, 895.	3.8	352
91	Genetic disruption of voltage-gated calcium channels in psychiatric and neurological disorders. Progress in Neurobiology, 2015, 134, 36-54.	2.8	187

#	Article	IF	Citations
92	The human splicing code reveals new insights into the genetic determinants of disease. Science, 2015, 347, 1254806.	6.0	1,053
93	Identification of a novel MSH6 germline variant in a family with multiple gastro-intestinal malignancies by next generation sequencing. Familial Cancer, 2015, 14, 69-75.	0.9	1
94	Copy Number Variable MicroRNAs in Schizophrenia and Their Neurodevelopmental Gene Targets. Biological Psychiatry, 2015, 77, 158-166.	0.7	58
95	MBD5 haploinsufficiency is associated with sleep disturbance and disrupts circadian pathways common to Smithâ€“Magenis and fragile X syndromes. European Journal of Human Genetics, 2015, 23, 781-789.	1.4	29
97	Syngap1 Haploinsufficiency Damages a Postnatal Critical Period of Pyramidal Cell Structural Maturation Linked to Cortical Circuit Assembly. Biological Psychiatry, 2015, 77, 805-815.	0.7	102
98	Bioinformatics Workflow for Clinical Whole Genome Sequencing at Partners HealthCare Personalized Medicine. Journal of Personalized Medicine, 2016, 6, 12.	1.1	22
99	Mutations of Voltage-Gated Sodium Channel Genes SCN1A and SCN2A in Epilepsy, Intellectual Disability, and Autism. , 2016, , 233-251.		12
100	Experimental Tools for the Identification ofÂSpecific Genes in Autism Spectrum DisordersÂand Intellectual Disability. , 2016, , 3-12.		1
101	Mutation Detection in an Antibody-Producing Chinese Hamster Ovary Cell Line by Targeted RNA Sequencing. BioMed Research International, 2016, 2016, 1-8.	0.9	3
102	Genomic Technologies in Medicine and Health. , 2016, , 15-28.		8
103	A Subset of Autism-Associated Genes Regulate the Structural Stability of Neurons. Frontiers in Cellular Neuroscience, 2016, 10, 263.	1.8	84
104	Language Impairments in ASD Resulting from a Failed Domestication of the Human Brain. Frontiers in Neuroscience, 2016, 10, 373.	1.4	52
105	Genetic Causes of Autism Spectrum Disorders. , 2016, , 13-24.		6
106	Clinical Genetic Aspects of Autism Spectrum Disorders. International Journal of Molecular Sciences, 2016, 17, 180.	1.8	55
107	Human Germline Mutation and the Erratic Evolutionary Clock. PLoS Biology, 2016, 14, e2000744.	2.6	70
108	Genetics of X-Linked Intellectual Disability. , 2016, , 25-41.		2
109	Are physicians prepared for whole genome sequencing? a qualitative analysis. Clinical Genetics, 2016, 89, 228-234.	1.0	108
110	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	1.7	295

#	Article	IF	Citations
111	CHD7 promotes proliferation of neural stem cells mediated by MIF. Molecular Brain, 2016, 9, 96.	1.3	28
112	Mutations inHECW2are associated with intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 697-704.	1.5	55
113	Comprehensive behavioral analysis of RNG105 (Caprin1) heterozygous mice: Reduced social interaction and attenuated response to novelty. Scientific Reports, 2016, 6, 20775.	1.6	33
115	Quo Vadis clinical genomics of ASD?. Autism, 2016, 20, 259-261.	2.4	5
116	Gene expression elucidates functional impact of polygenic risk for schizophrenia. Nature Neuroscience, 2016, 19, 1442-1453.	7.1	952
117	Increased burden of ultra-rare protein-altering variants among 4,877 individuals with schizophrenia. Nature Neuroscience, 2016, 19, 1433-1441.	7.1	427
118	Activity-Dependent Changes in Gene Expression in Schizophrenia Human-Induced Pluripotent Stem Cell Neurons. JAMA Psychiatry, 2016, 73, 1180.	6.0	40
119	The whole genome sequences and experimentally phased haplotypes of over 100 personal genomes. GigaScience, 2016, 5, 42.	3.3	21
120	Exploiting the potential of next-generation sequencing in genomic medicine. Expert Review of Molecular Diagnostics, 2016, 16, 1037-1047.	1.5	5
121	ASD Validity. Review Journal of Autism and Developmental Disorders, 2016, 3, 302-329.	2.2	69
122	Î± 5GABA _A receptor deficiency causes autismâ€like behaviors. Annals of Clinical and Translational Neurology, 2016, 3, 392-398.	1.7	43
123	Genome Tools and Methods. , 2016, , 63-72.		0
124	Genome-wide characteristics of de novo mutations in autism. Npj Genomic Medicine, 2016, 1, 160271-1602710.	1.7	200
125	Uncovering obsessive-compulsive disorder risk genes in a pediatric cohort by high-resolution analysis of copy number variation. Journal of Neurodevelopmental Disorders, 2016, 8, 36.	1.5	55
126	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	1.6	35
127	Cognitive analysis of schizophrenia risk genes that function as epigenetic regulators of gene expression. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1170-1179.	1.1	43
128	Genomic medicine goes mainstream. Npj Genomic Medicine, 2016, 1, 15001.	1.7	1
129	Whole-exome sequencing identifies a novel heterozygous missense variant of the EN2 gene in two unrelated patients with autism spectrum disorder. Psychiatric Genetics, 2016, 26, 297-301.	0.6	13

#	Article	IF	Citations
130	Genetic heterogeneity in autism: From single gene to a pathway perspective. Neuroscience and Biobehavioral Reviews, 2016, 68, 442-453.	2.9	46
131	The Genetics of Autism Spectrum Disorders. Research and Perspectives in Endocrine Interactions, 2016, , 101-129.	0.2	47
132	Molecular subtyping and improved treatment of neurodevelopmental disease. Genome Medicine, 2016, 8, 22.	3.6	17
133	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	4.1	243
134	A common molecular signature in ASD gene expression: following Root 66 to autism. Translational Psychiatry, 2016, 6, e705-e705.	2.4	40
135	The importance of de novo mutations for pediatric neurological diseaseâ€"It is not all in utero or birth trauma. Mutation Research - Reviews in Mutation Research, 2016, 767, 42-58.	2.4	7
136	Autism spectrum disorders: Integration of the genome, transcriptome and the environment. Journal of the Neurological Sciences, 2016, 364, 167-176.	0.3	49
137	Frequency and Complexity of De Novo Structural Mutation in Autism. American Journal of Human Genetics, 2016, 98, 667-679.	2.6	88
138	Atypical phenotypes associated with pathogenic CHD7 variants and a proposal for broadening CHARGE syndrome clinical diagnostic criteria. American Journal of Medical Genetics, Part A, 2016, 170, 344-354.	0.7	122
139	Early Identification of Autism Spectrum Disorder: Current Challenges and Future Global Directions. Current Developmental Disorders Reports, 2016, 3, 67-74.	0.9	40
140	Next Generation Sequencing in Neurology and Psychiatry. , 2016, , 97-136.		0
141	Improving treatment of neurodevelopmental disorders: recommendations based on preclinical studies. Expert Opinion on Drug Discovery, 2016, 11, 11-25.	2.5	16
142	Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 2016, 17, 9-18.	7.7	614
143	Genes with de novo mutations are shared by four neuropsychiatric disorders discovered from NPdenovo database. Molecular Psychiatry, 2016, 21, 290-297.	4.1	167
144	Modeling autism spectrum disorders with human neurons. Brain Research, 2017, 1656, 49-54.	1.1	17
145	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genetics in Medicine, 2017, 19, 13-19.	1.1	74
146	Effects of Multiple Genetic Loci on Age atÂOnset in Frontotemporal Dementia. Journal of Alzheimer's Disease, 2017, 56, 1271-1278.	1.2	4
147	The Impact of Next-Generation Sequencing on the Diagnosis and Treatment of Epilepsy in Paediatric Patients. Molecular Diagnosis and Therapy, 2017, 21, 357-373.	1.6	49

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148	A novel de novo frameshift deletion in <scp>EHMT</scp>1 in a patient with Kleefstra Syndrome results in decreased H3K9 dimethylation. Molecular Genetics & Genomic Medicine, 2017, 5, 141-146.	0.6	8
149	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	7.1	691
150	Vitamin Dâ€related genes are subjected to significant de novo mutation burdens in autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 568-577.	1.1	20
151	Genetic Approaches to Understanding Psychiatric Disease. Neurotherapeutics, 2017, 14, 564-581.	2.1	6
152	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
153	Genetic variants in the transcription regulatory region of MEGF10 are associated with autism in Chinese Han population. Scientific Reports, 2017, 7, 2292.	1.6	7
154	Genetic diagnosis of a Chinese multiple endocrine neoplasia type 2A family through whole genome sequencing. Journal of Biosciences, 2017, 42, 209-218.	0.5	5
155	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
156	Whole-genome sequencing of monozygotic twins discordant for schizophrenia indicates multiple genetic risk factors for schizophrenia. Journal of Genetics and Genomics, 2017, 44, 295-306.	1.7	36
157	Global developmental delay and intellectual disability associated with a de novo TOP2B mutation. Clinica Chimica Acta, 2017, 469, 63-68.	0.5	24
158	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	3.7	426
159	Genetics implicate common mechanisms in autism and schizophrenia: synaptic activity and immunity. Journal of Medical Genetics, 2017, 54, 511.2-520.	1.5	22
160	Genomic strategies to understand causes of keratoconus. Molecular Genetics and Genomics, 2017, 292, 251-269.	1.0	40
161	denovo-db: a compendium of humande novovariants. Nucleic Acids Research, 2017, 45, D804-D811.	6.5	173
162	Direct estimate of the spontaneous germ line mutation rate in African green monkeys. Evolution; International Journal of Organic Evolution, 2017, 71, 2858-2870.	1.1	40
163	ADNP Plays a Key Role in Autophagy: From Autism to Schizophrenia and Alzheimer's Disease. BioEssays, 2017, 39, 1700054.	1.2	41
164	Genomic Patterns of De Novo Mutation in Simplex Autism. Cell, 2017, 171, 710-722.e12.	13.5	308
165	Arc Requires PSD95 for Assembly into Postsynaptic Complexes Involved with Neural Dysfunction and Intelligence. Cell Reports, 2017, 21, 679-691.	2.9	79

#	Article	IF	Citations
166	Targeted sequencing and functional analysis reveal brain-size-related genes and their networks in autism spectrum disorders. Molecular Psychiatry, 2017, 22, 1282-1290.	4.1	95
167	Novel metrics to measure coverage in whole exome sequencing datasets reveal local and global non-uniformity. Scientific Reports, 2017, 7, 885.	1.6	43
168	Whole exome sequencing reveals inherited and de novo variants in autism spectrum disorder: a trio study from Saudi families. Scientific Reports, 2017, 7, 5679.	1.6	70
169	Human genomics projects and precision medicine. Gene Therapy, 2017, 24, 551-561.	2.3	109
170	10 Years of GWAS Discovery: Biology, Function, and Translation. American Journal of Human Genetics, 2017, 101, 5-22.	2.6	2,793
171	Three novel recessive mutations in LAMA2, SYNE1, and TTN are identified in a single case with congenital muscular dystrophy. Neuromuscular Disorders, 2017, 27, 1018-1022.	0.3	6
172	Genotype-phenotype evaluation of MED13L defects in the light of a novel truncating and a recurrent missense mutation. European Journal of Medical Genetics, 2017, 60, 451-464.	0.7	34
173	Cross-disorder comparative analysis of comorbid conditions reveals novel autism candidate genes. BMC Genomics, 2017, 18, 315.	1.2	24
174	A Cell Type-Specific Expression Signature Predicts Haploinsufficient Autism-Susceptibility Genes. Human Mutation, 2017, 38, 204-215.	1.1	38
175	mirVAFC: A Web Server for Prioritizations of Pathogenic Sequence Variants from Exome Sequencing Data via Classifications. Human Mutation, 2017, 38, 25-33.	1.1	3
176	Modeling of Autism Using Organoid Technology. Molecular Neurobiology, 2017, 54, 7789-7795.	1.9	17
177	Atypical autism in a boy with double duplication of 22q11.2: implications of increasing dosage. Npj Genomic Medicine, 2017, 2, 28.	1.7	3
179	DNA Methylation and Adult Neurogenesis. Brain Plasticity, 2017, 3, 5-26.	1.9	56
180	When loss-of-function is loss of function: assessing mutational signatures and impact of loss-of-function genetic variants. Bioinformatics, 2017, 33, i389-i398.	1.8	53
181	RNG105/caprin1, an RNA granule protein for dendritic mRNA localization, is essential for long-term memory formation. ELife, 2017, 6, .	2.8	45
182	Advanced Personal Genome Sequencing as the Ultimate Diagnostic Test. , 2017, , 155-172.		0
183	Individual Clinically Diagnosed with CHARGE Syndrome but with a Mutation in KMT2D, a Gene Associated with Kabuki Syndrome: A Case Report. Frontiers in Genetics, 2017, 8, 210.	1.1	18
184	High resolution measurement of DUF1220 domain copy number from whole genome sequence data. BMC Genomics, 2017, 18, 614.	1.2	17

#	Article	IF	Citations
185	Strength of functional signature correlates with effect size in autism. Genome Medicine, 2017, 9, 64.	3.6	12
186	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. Cmaj, 2018, 190, E126-E136.	0.9	57
187	Progress in the genetics of autism spectrum disorder. Developmental Medicine and Child Neurology, 2018, 60, 445-451.	1.1	116
188	Systematic reconstruction of autism biology from massive genetic mutation profiles. Science Advances, 2018, 4, e1701799.	4.7	43
189	Cost-effectiveness of Genome and Exome Sequencing in Children Diagnosed with Autism Spectrum Disorder. Applied Health Economics and Health Policy, 2018, 16, 481-493.	1.0	20
190	The DNA double-strand â€œbreakomeâ€•of mouse spermatids. Cellular and Molecular Life Sciences, 2018, 75, 2859-2872.	2.4	14
191	Brain Transcriptome Databases: A User's Guide. Journal of Neuroscience, 2018, 38, 2399-2412.	1.7	68
192	ZNF462 and KLF12 are disrupted by a de novo translocation in a patient with syndromic intellectual disability and autism spectrum disorder. European Journal of Medical Genetics, 2018, 61, 376-383.	0.7	13
193	Identification of Disease Susceptibility Alleles in the Next Generation Sequencing Era. Methods in Molecular Biology, 2018, 1706, 3-16.	0.4	4
194	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. Pharmacological Reviews, 2018, 70, 142-173.	7.1	215
195	Copy Number Variation Analysis of 100 Twin Pairs Enriched for Neurodevelopmental Disorders. Twin Research and Human Genetics, 2018, 21, 1-11.	0.3	27
196	A Comprehensive Workflow for Read Depth-Based Identification of Copy-Number Variation from Whole-Genome Sequence Data. American Journal of Human Genetics, 2018, 102, 142-155.	2.6	156
197	Whole exome sequencing in three families segregating a pediatric case of sarcoidosis. BMC Medical Genomics, 2018, 11, 23.	0.7	26
198	Advanced Whole-Genome Sequencing and Analysis of Fetal Genomes from Amniotic Fluid. Clinical Chemistry, 2018, 64, 715-725.	1.5	10
199	Identification of De Novo DNMT3A Mutations That Cause West Syndrome by Using Whole-Exome Sequencing. Molecular Neurobiology, 2018, 55, 2483-2493.	1.9	8
200	A Schizophrenia-Related Deletion Leads to KCNQ2-Dependent Abnormal Dopaminergic Modulation of Prefrontal Cortical Interneuron Activity. Cerebral Cortex, 2018, 28, 2175-2191.	1.6	19
201	Macrophage migration inhibitory factor: A multifaceted cytokine implicated in multiple neurological diseases. Experimental Neurology, 2018, 301, 83-91.	2.0	59
202	Quantitative mapping of genetic similarity in human heritable diseases by shared mutations. Human Mutation, 2018, 39, 292-301.	1.1	8

#	Article	IF	Citations
203	A rare ANOS1 variant in siblings with Kallmann syndrome identified by whole exome sequencing. Andrology, 2018, 6, 53-57.	1.9	7
204	Understanding Neurodevelopmental Disorders: The Promise of Regulatory Variation in the 3â€²UTRome. Biological Psychiatry, 2018, 83, 548-557.	0.7	48
205	Identification of de novo germline mutations and causal genes for sporadic diseases using trioâ€based wholeâ€exome/genome sequencing. Biological Reviews, 2018, 93, 1014-1031.	4.7	35
206	Communicating complex genomic information: A counselling approach derived from research experience with Autism Spectrum Disorder. Patient Education and Counseling, 2018, 101, 352-361.	1.0	27
207	Regulation of the Expression of the Psychiatric Risk Gene <i>Cacna1c</i> during Associative Learning. Molecular Neuropsychiatry, 2018, 4, 149-157.	3.0	8
208	Complete Disruption of Autism-Susceptibility Genes by Gene Editing Predominantly Reduces Functional Connectivity of Isogenic Human Neurons. Stem Cell Reports, 2018, 11, 1211-1225.	2.3	111
209	Autism-associated CHD8 deficiency impairs axon development and migration of cortical neurons. Molecular Autism, 2018, 9, 65.	2.6	58
210	Whole Genome Sequencing of a Vietnamese Family from a Dioxin Contamination Hotspot Reveals Novel Variants in the Son with Undiagnosed Intellectual Disability. International Journal of Environmental Research and Public Health, 2018, 15, 2629.	1.2	4
211	Synaptosomes. Neuromethods, 2018, , .	0.2	3
212	The Use of Synaptosomes in Studying Autism Spectrum Disorder and Other Neurodevelopmental Disorders. Neuromethods, 2018, , 287-296.	0.2	0
213	Prioritization of Variants Detected by Next Generation Sequencing According to the Mutation Tolerance and Mutational Architecture of the Corresponding Genes. International Journal of Molecular Sciences, 2018, 19, 1584.	1.8	16
214	Genomic landscapes of Chinese sporadic autism spectrum disorders revealed by whole-genome sequencing. Journal of Genetics and Genomics, 2018, 45, 527-538.	1.7	33
215	AutismKB 2.0: a knowledgebase for the genetic evidence of autism spectrum disorder. Database: the Journal of Biological Databases and Curation, 2018, 2018, .	1.4	37
216	NGL-2 Deletion Leads to Autistic-like Behaviors Responsive to NMDAR Modulation. Cell Reports, 2018, 23, 3839-3851.	2.9	41
217	Whole genome sequencing and mutation rate analysis of trios with paternal dioxin exposure. Human Mutation, 2018, 39, 1384-1392.	1.1	14
218	Association analysis of two synapse-related gene mutations with autism spectrum disorder in a Chinese population. Research in Autism Spectrum Disorders, 2018, 53, 67-72.	0.8	0
219	NGS Technologies as a Turning Point in Rare Disease Research , Diagnosis and Treatment. Current Medicinal Chemistry, 2018, 25, 404-432.	1.2	111
220	Next Generation Sequencing Methods for Diagnosis of Epilepsy Syndromes. Frontiers in Genetics, 2018, 9, 20.	1.1	102

#	Article	IF	Citations
221	Nav1.2 haplodeficiency in excitatory neurons causes absence-like seizures in mice. Communications Biology, 2018, 1, 96.	2.0	75
222	Autism spectrum disorder: prospects for treatment using gene therapy. Molecular Autism, 2018, 9, 39.	2.6	42
223	A Statistical Framework for Mapping Risk Genes from De Novo Mutations in Whole-Genome-Sequencing Studies. American Journal of Human Genetics, 2018, 102, 1031-1047.	2.6	26
224	CACNA1C: Association With Psychiatric Disorders, Behavior, and Neurogenesis. Schizophrenia Bulletin, 2018, 44, 958-965.	2.3	119
225	A comparative study of the genetic components of three subcategories of autism spectrum disorder. Molecular Psychiatry, 2019, 24, 1720-1731.	4.1	22
226	Information needs in genetic testing: A needs assessment survey among Taiwanese parents of children with autism spectrum disorders. Autism, 2019, 23, 902-909.	2.4	7
227	Family-Based Quantitative Trait Meta-Analysis Implicates Rare Noncoding Variants in DENND1A in Polycystic Ovary Syndrome. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3835-3850.	1.8	51
228	Scn2a Haploinsufficiency in Mice Suppresses Hippocampal Neuronal Excitability, Excitatory Synaptic Drive, and Long-Term Potentiation, and Spatial Learning and Memory. Frontiers in Molecular Neuroscience, 2019, 12, 145.	1.4	39
229	Detection of Clinically Relevant Genetic Variants in Chinese Patients With Nanophthalmos by Trio-Based Whole-Genome Sequencing Study. , 2019, 60, 2904.		25
230	Hypogonadotrophic hypogonadism, delayed puberty and risk for neurodevelopmental disorders. Journal of Neuroendocrinology, 2019, 31, e12803.	1.2	13
231	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	1.7	118
232	De novo Mutations From Whole Exome Sequencing in Neurodevelopmental and Psychiatric Disorders: From Discovery to Application. Frontiers in Genetics, 2019, 10, 258.	1.1	49
233	Identification of Copy Number Variation by Array-CGH in Portuguese Children and Adolescents Diagnosed with Autism Spectrum Disorders. Neuropediatrics, 2019, 50, 367-377.	0.3	6
234	Detection of de novo genetic variants in Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser syndrome by whole genome sequencing. European Journal of Obstetrics and Gynecology and Reproductive Biology: X, 2019, 4, 100089.	0.6	12
235	NaV1.2 haploinsufficiency in Scn2a knock-out mice causes an autistic-like phenotype attenuated with age. Scientific Reports, 2019, 9, 12886.	1.6	25
236	Association between distress and knowledge among parents of autistic children. PLoS ONE, 2019, 14, e0223119.	1.1	5
237	Early life exposures, neurodevelopmental disorders, and transposable elements. Neurobiology of Stress, 2019, 11, 100174.	1.9	27
238	Whole-genome deep-learning analysis identifies contribution of noncoding mutations to autism risk. Nature Genetics, 2019, 51, 973-980.	9.4	216

#	Article	IF	Citations
239	Biallelic SCN2A Gene Mutation Causing Early Infantile Epileptic Encephalopathy: Case Report and Review. Journal of Central Nervous System Disease, 2019, 11, 117957351984993.	0.7	10
240	Genetic insights and neurobiological implications from NRXN1 in neuropsychiatric disorders. Molecular Psychiatry, 2019, 24, 1400-1414.	4.1	58
241	Scn2a haploinsufficient mice display a spectrum of phenotypes affecting anxiety, sociability, memory flexibility and ampakine CX516 rescues their hyperactivity. Molecular Autism, 2019, 10, 15.	2.6	56
242	Approach to the Genetic Diagnosis of Epileptic Encephalopathies and Developmental Encephalopathies with Epilepsy of Early Childhood. , 2019, , 60-68.		0
243	Altered Urinary Amino Acids in Children With Autism Spectrum Disorders. Frontiers in Cellular Neuroscience, 2019, 13, 7.	1.8	40
244	The Genetics of Autism Spectrum Disorders. , 2019, , 112-128.		14
245	The Neurobiology of Autism. , 2019, , 129-157.		3
246	Artificial intelligence for precision medicine in neurodevelopmental disorders. Npj Digital Medicine, 2019, 2, 112.	5.7	121
247	An autism-causing calcium channel variant functions with selective autophagy to alter axon targeting and behavior. PLoS Genetics, 2019, 15, e1008488.	1.5	21
248	Phenotypic spectrum and genetics of <scp>SCN</scp>2Aâ€related disorders, treatment options, and outcomes in epilepsy and beyond. Epilepsia, 2019, 60, S59-S67.	2.6	49
249	Identifying Genomic Variations in Monozygotic Twins Discordant for Autism Spectrum Disorder Using Whole-Genome Sequencing. Molecular Therapy - Nucleic Acids, 2019, 14, 204-211.	2.3	40
250	Increased serum levels of macrophage migration inhibitory factor in autism spectrum disorders. NeuroToxicology, 2019, 71, 1-5.	1.4	20
251	Altered TAOK2 activity causes autism-related neurodevelopmental and cognitive abnormalities through RhoA signaling. Molecular Psychiatry, 2019, 24, 1329-1350.	4.1	128
252	Nonrandom occurrence of multiple de novo coding variants in a proband indicates the existence of an oligogenic model in autism. Genetics in Medicine, 2020, 22, 170-180.	1.1	23
253	Genomics of autism spectrum disorders. , 2020, , 161-171.		0
254	Phenotypic Spectrum of Idiopathic Hypogonadotropic Hypogonadism Patients With CHD7 Variants From a Large Chinese Cohort. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1515-1526.	1.8	12
255	Just Expect It: Compound Heterozygous Variants of POMT1 in a Consanguineous Familyâ€"The Role of Next Generation Sequencing in Neuromuscular Disorders. Neuropediatrics, 2020, 51, 072-075.	0.3	1
256	Identification, Evaluation, and Management of Children With Autism Spectrum Disorder. Pediatrics, 2020, 145, .	1.0	621

#	Article	IF	Citations
257	Exome sequencing identifies de novo splicing variant in XRCC6 in sporadic case of autism. Journal of Human Genetics, 2020, 65, 287-296.	1.1	8
258	CUL3 Deficiency Causes Social Deficits and Anxiety-like Behaviors by Impairing Excitation-Inhibition Balance through the Promotion of Cap-Dependent Translation. Neuron, 2020, 105, 475-490.e6.	3.8	70
259	Precision Therapies in Neurodevelopmental Disorders. Advances in Molecular Pathology, 2020, 3, 21-27.	0.2	0
260	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	5.8	305
261	Using Zebrafish to Model Autism Spectrum Disorder: A Comparison of ASD Risk Genes Between Zebrafish and Their Mammalian Counterparts. Frontiers in Molecular Neuroscience, 2020, 13, 575575.	1.4	32
262	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2020, 107, 683-697.	2.6	23
263	Circulating Cell-Free Nucleic Acids as Epigenetic Biomarkers in Precision Medicine. Frontiers in Genetics, 2020, 11, 844.	1.1	32
264	Synaptic dysregulation in autism spectrum disorders. Journal of Neuroscience Research, 2020, 98, 2111-2114.	1.3	1
265	The Role of Kv7.2 in Neurodevelopment: Insights and Gaps in Our Understanding. Frontiers in Physiology, 2020, 11, 570588.	1.3	35
266	Neuroligins and Neurodevelopmental Disorders: X-Linked Genetics. Frontiers in Synaptic Neuroscience, 2020, 12, 33.	1.3	33
267	Heteromeric Kv7.2 current changes caused by loss-of-function of KCNQ2 mutations are correlated with long-term neurodevelopmental outcomes. Scientific Reports, 2020, 10, 13375.	1.6	8
268	Identifying Genes Associated With Autism Spectrum Disorders by Random Walk Method With Significance Tests. IEEE Access, 2020, 8, 156686-156694.	2.6	6
269	Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature, 2020, 586, 80-86.	13.7	155
270	Whole genome investigation of an atypical autism case identifies a novel ANOS1 mutation with subsequent diagnosis of Kallmann syndrome. Molecular Genetics and Metabolism Reports, 2020, 23, 100593.	0.4	2
271	Transcriptome analysis of neural progenitor cells derived from Lowe syndrome induced pluripotent stem cells: identification of candidate genes for the neurodevelopmental and eye manifestations. Journal of Neurodevelopmental Disorders, 2020, 12, 14.	1.5	12
272	Genetic associations between voltage-gated calcium channels and autism spectrum disorder: a systematic review. Molecular Brain, 2020, 13, 96.	1.3	42
273	Linking risk factors and outcomes in autism spectrum disorder: is there evidence for resilience?. BMJ, The, 2020, 368, l6880.	3.0	45
274	A missense variant, p.(Ile269Asn), in MC4R as a secondary finding in a child with BCL11A-related intellectual disability. European Journal of Medical Genetics, 2020, 63, 103969.	0.7	1

#	Article	IF	Citations
275	Determination of genetic changes in etiology of autism spectrum disorder in twins by whole-exome sequencing. Gene Reports, 2020, 19, 100618.	0.4	2
276	Perceived utility of biological testing for autism spectrum disorder is associated with child and family functioning. Research in Developmental Disabilities, 2020, 100, 103605.	1.2	7
277	Leveraging large genomic datasets to illuminate the pathobiology of autism spectrum disorders. Neuropsychopharmacology, 2021, 46, 55-69.	2.8	31
278	Adaptation and validation of the Genetic Counseling Outcome Scale for autism spectrum disorders and related conditions. Journal of Genetic Counseling, 2021, 30, 305-318.	0.9	7
279	Dysregulated Translation in Autism Spectrum Disorder. , 0, , 451-486.		0
280	Effect Sizes of Deletions and Duplications on Autism Risk Across the Genome. American Journal of Psychiatry, 2021, 178, 87-98.	4.0	50
281	Role of Munc18-1 in the biological functions and pathogenesis of neurological disorders (Review). Molecular Medicine Reports, 2021, 23, .	1.1	5
282	Coinheritance of OLFM2 and SIX6 variants in a Chinese family with juvenile-onset primary open-angle glaucoma: A case report. World Journal of Clinical Cases, 2021, 9, 697-706.	0.3	3
283	Testosterone and the Brain: From Cognition to Autism. Physiological Research, 0, , S403-S419.	0.4	7
284	A Review of Genetic and Physiological Disease Mechanisms Associated With Cav1 Channels: Implications for Incomplete Congenital Stationary Night Blindness Treatment. Frontiers in Genetics, 2021, 12, 637780.	1.1	2
285	Neural Mechanisms Underlying Repetitive Behaviors in Rodent Models of Autism Spectrum Disorders. Frontiers in Cellular Neuroscience, 2020, 14, 592710.	1.8	40
286	MTSplice predicts effects of genetic variants on tissue-specific splicing. Genome Biology, 2021, 22, 94.	3.8	23
287	<scp>PLXNA2</scp> and <scp>LRRC40</scp> as candidate genes in autism spectrum disorder. Autism Research, 2021, 14, 1088-1100.	2.1	5
288	Targeted sequencing and integrative analysis of 3,195 Chinese patients with neurodevelopmental disorders prioritized 26 novel candidate genes. Journal of Genetics and Genomics, 2021, 48, 312-323.	1.7	11
289	Differential Metabolites in Chinese Autistic Children: A Multi-Center Study Based on Urinary 1H-NMR Metabolomics Analysis. Frontiers in Psychiatry, 2021, 12, 624767.	1.3	9
290	Environmental exposures associated with elevated risk for autism spectrum disorder may augment the burden of deleterious de novo mutations among probands. Molecular Psychiatry, 2022, 27, 710-730.	4.1	36
292	Potassium channels and autism spectrum disorder: An overview. International Journal of Developmental Neuroscience, 2021, 81, 479-491.	0.7	16
293	LRRC4 functions as a neuron-protective role in experimental autoimmune encephalomyelitis. Molecular Medicine, 2021, 27, 44.	1.9	9

#	Article	IF	Citations
294	An Intronic Variant of CHD7 Identified in Autism Patients Interferes with Neuronal Differentiation and Development. Neuroscience Bulletin, 2021, 37, 1091-1106.	1.5	9
296	Interaction hot spots for phase separation revealed by NMR studies of a CAPRIN1 condensed phase. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	40
297	Advances in clinical genetics and genomics. Intelligent Medicine, 2021, 1, 128-133.	1.6	4
298	Automated Detection Approaches to Autism Spectrum Disorder Based on Human Activity Analysis: A Review. Cognitive Computation, 2022, 14, 1773-1800.	3.6	6
301	Major brain malformations: corpus callosum dysgenesis, agenesis of septum pellucidum and polymicrogyria in patients with BCORL1-related disorders. Journal of Human Genetics, 2022, 67, 95-101.	1.1	2
302	Technological Improvements in the Genetic Diagnosis of Rett Syndrome Spectrum Disorders. International Journal of Molecular Sciences, 2021, 22, 10375.	1.8	3
304	Prioritizing de novo autism risk variants with calibrated gene- and variant-scoring models. Human Genetics, 2021, , 1.	1.8	1
306	Rare and low frequency genomic variants impacting neuronal functions modify the Dup7q11.23 phenotype. Orphanet Journal of Rare Diseases, 2021, 16, 6.	1.2	4
307	Genome sequencing identifies rare tandem repeat expansions and copy number variants in Lennoxâ€“Gastaut syndrome. Brain Communications, 2021, 3, fcab207.	1.5	4
308	Sleep phenotype of individuals with autism spectrum disorder bearing mutations in the <scp>PER2</scp> circadian rhythm gene. American Journal of Medical Genetics, Part A, 2021, 185, 1120-1130.	0.7	17
309	Ca²⁺â€Ðependent Hyperpolarization Pathways in Sleep Homeostasis and Mental Disorders. BioEssays, 2018, 40, 1700105.	1.2	19
310	Genetics of Psychiatric Disorders. , 2016, , 553-600.		1
311	Emerging Technologies for Gene Identification in Rare Diseases. Advances in Predictive, Preventive and Personalised Medicine, 2015, , 33-45.	0.6	1
312	A microcosting and costâ€“consequence analysis of clinical genomic testing strategies in autism spectrum disorder. Genetics in Medicine, 2017, 19, 1268-1275.	1.1	62
326	Analytical and Clinical Validity Study of FirstStepDx PLUS: A Chromosomal Microarray Optimized for Patients with Neurodevelopmental Conditions. PLOS Currents, 2017, 9, .	1.4	9
327	Multi-nucleotide de novo Mutations in Humans. PLoS Genetics, 2016, 12, e1006315.	1.5	111
328	Inference of Candidate Germline Mutator Loci in Humans from Genome-Wide Haplotype Data. PLoS Genetics, 2017, 13, e1006549.	1.5	22
329	Ultra High-Resolution Gene Centric Genomic Structural Analysis of a Non-Syndromic Congenital Heart Defect, Tetralogy of Fallot. PLoS ONE, 2014, 9, e87472.	1.1	16

#	Article	IF	Citations
330	Comorbid Analysis of Genes Associated with Autism Spectrum Disorders Reveals Differential Evolutionary Constraints. PLoS ONE, 2016, 11, e0157937.	1.1	24
331	The Dynamic Exome: acquired variants as individuals age. Aging, 2014, 6, 511-521.	1.4	12
332	Nicotine and oxidative stress induced exomic variations are concordant and overrepresented in cancer-associated genes. Oncotarget, 2014, 5, 4788-4798.	0.8	28
333	Syndromic autism spectrum disorders: moving from a clinically defined to a molecularly defined approach. Dialogues in Clinical Neuroscience, 2017, 19, 353-371.	1.8	105
334	Genetic tests by next-generation sequencing in children with developmental delay and/or intellectual disability. Clinical and Experimental Pediatrics, 2020, 63, 195-202.	0.9	29
335	Expressional Subpopulation of Cancers Determined by G64, a Co-regulated Module. Genomics and Informatics, 2015, 13, 132.	0.4	2
336	CNTN5-/+or EHMT2-/+human iPSC-derived neurons from individuals with autism develop hyperactive neuronal networks. ELife, 2019, 8, .	2.8	72
337	Proteins in DNA methylation and their role in neural stem cell proliferation and differentiation. Cell Regeneration, 2021, 10, 7.	1.1	8
338	Aortic root dilatation and dilated cardiomyopathy in an adult with <scp>Tattonâ€Brownâ€Rahman</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 628-634.	0.7	4
339	The Role of Ion Channel-Related Genes in Autism Spectrum Disorder: A Study Using Next-Generation Sequencing. Frontiers in Genetics, 2021, 12, 595934.	1.1	4
340	Neuroligin-3: A Circuit-Specific Synapse Organizer That Shapes Normal Function and Autism Spectrum Disorder-Associated Dysfunction. Frontiers in Molecular Neuroscience, 2021, 14, 749164.	1.4	28
341	Future Directions in Psychopharmacology of Autism Spectrum Disorder. , 2014, , 473-495.		0
342	Whole Genome Sequencing in Autism: Clinical Translation. , 2014, , 69-97.		0
343	Phenotype Definition: A Cornerstone of Autism Research, Diagnosis and Therapy. , 2014, , 3-22.		0
344	Cognitive Deficits and Behavioral Disorders in Children: A Comprehensive Multidisciplinary Approach to Management. , 2015, 01, .		0
345	Genetics of Autism Spectrum Disorders: The Opportunity and Challenge in the Genetics Clinic. , 2015, , 33-66.		0
349	Epilepsy Associated with ASD and Intellectual Disability. , 2016, , 365-373.		0
350	22q11.2 Deletion Syndrome: A Paradigmatic Copy-Number-Variant (CNV) Disorder. , 2016, , 723-730.		0

#	Article	IF	Citations
351	PACAP, VIP, and ADNP: Autism and Schizophrenia. Current Topics in Neurotoxicity, 2016, , 781-792.	0.4	0
352	Deciphering the Risk Factors of Autism: Are We There Yet?. Journal of Biosciences and Medicines, 2016, 04, 65-76.	0.1	0
360	Whole exome and genome sequencing for Mendelian immune disorders: from molecular diagnostics to new disease variant and gene discovery. LymphoSign Journal, 0, , .	0.1	1
364	Autism Spectrum Disorder in the Child with Cerebral Palsy. , 2019, , 1-10.		0
365	Psychotic Disorders and Best Models of Care. , 2019, , 113-133.		0
366	Expected future developments in child neurology. Journal of International Child Neurology Association, 0, , .	0.0	0
369	Autism Spectrum Disorder in the Child with Cerebral Palsy. , 2020, , 505-514.		1
370	Significance of exome sequencing for diagnosis of epilepsy in children. Epilepsy and Paroxysmal Conditions, 2020, 11, 379-387.	0.2	1
371	Functional and Neuropathological Evidence for a Role of the Brainstem in Autism. Frontiers in Integrative Neuroscience, 2021, 15, 748977.	1.0	7
372	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. Npj Genomic Medicine, 2021, 6, 91.	1.7	9
373	Next Generation Sequencing and Health Technology Assessment in Autism Spectrum Disorder. Journal of the Canadian Academy of Child and Adolescent Psychiatry, 2015, 24, 123-7.	0.7	7
374	A Low Rank Model for Phenotype Imputation in Autism Spectrum Disorder. AMIA Summits on Translational Science Proceedings, 2018, 2017, 178-187.	0.4	3
375	The benefit of diagnostic whole genome sequencing in schizophrenia and other psychotic disorders. Molecular Psychiatry, 2022, 27, 1435-1447.	4.1	12
376	Altered Metabolic Characteristics in Plasma of Young Boys with Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2022, 52, 4897-4907.	1.7	9
377	Diagnosis of Genetic and Metabolic Conditions. , 2021, , .		0
379	Testosterone and the brain: from cognition to autism. Physiological Research, 2020, 69, S403-S419.	0.4	9
380	KCNQ2 Selectivity Filter Mutations Cause Kv7.2 M-Current Dysfunction and Configuration Changes Manifesting as Epileptic Encephalopathies and Autistic Spectrum Disorders. Cells, 2022, 11, 894.	1.8	2
381	AFF2 Is Associated With X-Linked Partial (Focal) Epilepsy With Antecedent Febrile Seizures. Frontiers in Molecular Neuroscience, 2022, 15, 795840.	1.4	9

#	Article	IF	Citations
383	A Next Generation Sequencing-Based Protocol for Screening of Variants of Concern in Autism Spectrum Disorder. Cells, 2022, 11, 10.	1.8	16
384	Epigenetic Epidemiology of Autism and Other Neurodevelopmental Disorders. , 2022, , 405-426.		1
393	TT-Mars: structural variants assessment based on haplotype-resolved assemblies. Genome Biology, 2022, 23, 110.	3.8	9
394	Regionally defined proteomic profiles of human cerebral tissue and organoids reveal conserved molecular modules of neurodevelopment. Cell Reports, 2022, 39, 110846.	2.9	7
395	Identifying Rare Genetic Variants of Immune Mediators as Risk Factors for Autism Spectrum Disorder. Genes, 2022, 13, 1098.	1.0	6
396	Genetic and Epigenetic Regulation of Brain Organoids. Frontiers in Cell and Developmental Biology, 0, 10, .	1.8	3
397	Complex Diagnostics of Non-Specific Intellectual Developmental Disorder. International Journal of Molecular Sciences, 2022, 23, 7764.	1.8	3
398	Prediction of the Effects of Missense Mutations on Human Myeloperoxidase Protein Stability Using In Silico Saturation Mutagenesis. Genes, 2022, 13, 1412.	1.0	2
399	Screening for Fragile X Syndrome Among Filipino Children with Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 0, , .	1.7	0
400	De novo variants in genes regulating stress granule assembly associate with neurodevelopmental disorders. Science Advances, 2022, 8, .	4.7	16
401	Synaptic genes and neurodevelopmental disorders: From molecular mechanisms to developmental strategies of behavioral testing. Neurobiology of Disease, 2022, 173, 105856.	2.1	12
402	Mapping the per-residue surface electrostatic potential of CAPRIN1 along its phase-separation trajectory. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	19
403	CAPRIN1P512L causes aberrant protein aggregation and associates with early-onset ataxia. Cellular and Molecular Life Sciences, 2022, 79, .	2.4	6
404	SCIP: software for efficient clinical interpretation of copy number variants detected by whole-genome sequencing. Human Genetics, 2023, 142, 201-216.	1.8	1
405	Trends and features of autism spectrum disorder research using artificial intelligence techniques: a bibliometric approach. Current Psychology, 2023, 42, 31317-31332.	1.7	1
406	The Role of Zinc and NMDA Receptors in Autism Spectrum Disorders. Pharmaceuticals, 2023, 16, 1.	1.7	4
407	Surface electrostatics dictate RNA-binding protein CAPRIN1 condensate concentration and hydrodynamic properties. Journal of Biological Chemistry, 2023, 299, 102776.	1.6	5
408	Dendritic Spine in Autism Genetics: Whole-Exome Sequencing Identifying De Novo Variant of CTTNBP2 in a Quad Family Affected by Autism Spectrum Disorder. Children, 2023, 10, 80.	0.6	3

#	Article	IF	Citations
409	A biallelic variant of DCAF13 implicated in a neuromuscular disorder in humans. European Journal of Human Genetics, 2023, 31, 629-637.	1.4	3
410	Genetics and Epigenetics of ASD. , 2023, , 293-307.		0
411	Identification of de novo Mutations in the Chinese Autism Spectrum Disorder Cohort via Whole-Exome Sequencing Unveils Brain Regions Implicated in Autism. Neuroscience Bulletin, 2023, 39, 1469-1480.	1.5	4
412	Genetic diagnostic yields of 354 Chinese ASD children with rare mutations by a pipeline of genomic tests. Frontiers in Genetics, 0, 14, .	1.1	2
413	Opportunities and achievements of using massive parallel sequencing in the diagnosis of neurodevelopmental diseases. Epilepsy and Paroxysmal Conditions, 2023, 15, 44-52.	0.2	1
414	CHARGE syndrome-associated CHD7 acts at ISL1-regulated enhancers to modulate second heart field gene expression. Cardiovascular Research, 2023, 119, 2089-2105.	1.8	5
421	Molecular testing in autism spectrum disorder. , 2024, , 291-301.		0
422	Rare genetic brain disorders with overlapping neurological and psychiatric phenotypes. Nature Reviews Neurology, 2024, 20, 7-21.	4.9	1
427	Autism Spectrum Disorder Classification via Local and Global Feature Representation of Facial Image. , 2023, , .		0