

A recurrent 15q13.3 microdeletion syndrome associated seizures

Nature Genetics

40, 322-328

DOI: [10.1038/ng.93](https://doi.org/10.1038/ng.93)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Novel microdeletion syndromes detected by chromosome microarrays. <i>Human Genetics</i> , 2008, 124, 1-17.	1.8	198
2	Molecular and clinical characterization of two patients with Prader-Willi syndrome and atypical deletions of proximal chromosome 15q. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1955-1962.	0.7	14
3	The copy number variant involving part of the 7 nicotinic receptor gene contains a polymorphic inversion. <i>European Journal of Human Genetics</i> , 2008, 16, 1364-1371.	1.4	38
4	Rare chromosomal deletions and duplications increase risk of schizophrenia. <i>Nature</i> , 2008, 455, 237-241.	13.7	1,387
5	Copy-number variations associated with neuropsychiatric conditions. <i>Nature</i> , 2008, 455, 919-923.	13.7	587
6	Incriminating genomic evidence. <i>Nature</i> , 2008, 455, 178-179.	13.7	46
8	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. <i>Nature Genetics</i> , 2008, 40, 1466-1471.	9.4	535
9	The array CGH and its clinical applications. <i>Drug Discovery Today</i> , 2008, 13, 760-770.	3.2	171
10	Fluorescence in situ hybridization techniques in medical diagnostics. <i>Expert Opinion on Medical Diagnostics</i> , 2008, 2, 1381-1390.	1.6	4
11	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. <i>New England Journal of Medicine</i> , 2008, 359, 1685-1699.	13.9	663
12	Detection of cryptic pathogenic copy number variations and constitutional loss of heterozygosity using high resolution SNP microarray analysis in 117 patients referred for cytogenetic analysis and impact on clinical practice. <i>Journal of Medical Genetics</i> , 2008, 46, 123-131.	1.5	61
13	Mechanisms for human genomic rearrangements. <i>PathoGenetics</i> , 2008, 1, 4.	5.7	523
14	CNV and nervous system diseases – what's new?. <i>Cytogenetic and Genome Research</i> , 2008, 123, 54-64.	0.6	35
15	Inducing Segmental Aneuploid Mosaicism in the Mouse Through Targeted Asymmetric Sister Chromatid Event of Recombination. <i>Genetics</i> , 2008, 180, 51-59.	1.2	17
16	<i>DupMasker</i> : A tool for annotating primate segmental duplications. <i>Genome Research</i> , 2008, 18, 1362-1368.	2.4	39
17	Sequencing human-gibbon breakpoints of synteny reveals mosaic new insertions at rearrangement sites. <i>Genome Research</i> , 2009, 19, 178-190.	2.4	29
18	Molecular Cytogenetics and Cytogenomics of Brain Diseases. <i>Current Genomics</i> , 2008, 9, 452-465.	0.7	75
19	Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. <i>Journal of Medical Genetics</i> , 2008, 45, 710-720.	1.5	191

#	ARTICLE	IF	CITATIONS
21	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. <i>Human Molecular Genetics</i> , 2009, 18, 3626-3631.	1.4	211
22	Copy Number Variation and Schizophrenia. <i>Schizophrenia Bulletin</i> , 2009, 35, 9-12.	2.3	93
23	Copy number variants, diseases and gene expression. <i>Human Molecular Genetics</i> , 2009, 18, R1-R8.	1.4	370
24	Aneuploidy: From a Physiological Mechanism of Variance to Down Syndrome. <i>Physiological Reviews</i> , 2009, 89, 887-920.	13.1	106
25	High-resolution mapping and analysis of copy number variations in the human genome: A data resource for clinical and research applications. <i>Genome Research</i> , 2009, 19, 1682-1690.	2.4	313
26	Characterization of six human disease-associated inversion polymorphisms. <i>Human Molecular Genetics</i> , 2009, 18, 2555-2566.	1.4	118
27	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. <i>Human Molecular Genetics</i> , 2009, 18, 3579-3593.	1.4	143
28	Recurrent Rearrangements in Synaptic and Neurodevelopmental Genes and Shared Biologic Pathways in Schizophrenia, Autism, and Mental Retardation. <i>Archives of General Psychiatry</i> , 2009, 66, 947.	13.8	374
30	Validation of the Agilent 244K Oligonucleotide Array-Based Comparative Genomic Hybridization Platform for Clinical Cytogenetic Diagnosis. <i>American Journal of Clinical Pathology</i> , 2009, 132, 349-360.	0.4	38
31	Genotype to phenotype discovery and characterization of novel genomic disorders in a genome-first era. <i>Genetics in Medicine</i> , 2009, 11, 836-842.	1.1	37
32	A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. <i>Genome Research</i> , 2009, 19, 1579-1585.	2.4	118
33	Genome-Wide Analyses of Exonic Copy Number Variants in a Family-Based Study Point to Novel Autism Susceptibility Genes. <i>PLoS Genetics</i> , 2009, 5, e1000536.	1.5	374
34	Dissecting the many genetic faces of schizophrenia. <i>Epidemiologia E Psichiatria Sociale</i> , 2009, 18, 91-95.	1.0	20
35	Segmental duplications mediate novel, clinically relevant chromosome rearrangements. <i>Human Molecular Genetics</i> , 2009, 18, 2957-2962.	1.4	63
36	Clinical Application of Microarray-Based Molecular Cytogenetics: An Emerging New Era of Genomic Medicine. <i>Journal of Pediatrics</i> , 2009, 155, 311-317.	0.9	35
37	The genetic landscape of intellectual disability arising from chromosome X. <i>Trends in Genetics</i> , 2009, 25, 308-316.	2.9	190
38	Complex human chromosomal and genomic rearrangements. <i>Trends in Genetics</i> , 2009, 25, 298-307.	2.9	239
39	Rare structural variants in schizophrenia: one disorder, multiple mutations; one mutation, multiple disorders. <i>Trends in Genetics</i> , 2009, 25, 528-535.	2.9	235

#	ARTICLE	IF	CITATIONS
40	A 2-base pair deletion polymorphism in the partial duplication of the 7 nicotinic acetylcholine gene (CHRFAM7A) on chromosome 15q14 is associated with schizophrenia. <i>Brain Research</i> , 2009, 1291, 1-11.	1.1	82
41	A 781 kb deletion of 13q12.3 in a patient with Peters plus syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1842-1845.	0.7	16
42	Molecular karyotyping of patients with unexplained mental retardation by SNP arrays: A multicenter study. <i>Human Mutation</i> , 2009, 30, 1082-1092.	1.1	65
43	Balanced translocations in mental retardation. <i>Human Genetics</i> , 2009, 126, 133-147.	1.8	27
44	Redefined genomic architecture in 15q24 directed by patient deletion/duplication breakpoint mapping. <i>Human Genetics</i> , 2009, 126, 589-602.	1.8	65
45	Chromosomale Ursachen der geistigen Behinderung. <i>Medizinische Genetik</i> , 2009, 21, 237-245.	0.1	0
46	Genetics of autism spectrum disorders. <i>Current Neurology and Neuroscience Reports</i> , 2009, 9, 188-197.	2.0	125
47	Molecular genetics of autism. <i>Current Psychiatry Reports</i> , 2009, 11, 137-142.	2.1	11
48	Current controversies in prenatal diagnosis 3: for prenatal diagnosis, should we offer less or more than metaphase karyotyping?. <i>Prenatal Diagnosis</i> , 2009, 29, 11-14.	1.1	37
49	Common inversion polymorphisms and rare microdeletions at 15q13.3. <i>European Journal of Human Genetics</i> , 2009, 17, 149-150.	1.4	12
50	A 15q13.3 microdeletion segregating with autism. <i>European Journal of Human Genetics</i> , 2009, 17, 687-692.	1.4	129
51	Finding the missing heritability of complex diseases. <i>Nature</i> , 2009, 461, 747-753.	13.7	7,490
52	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. <i>Nature Genetics</i> , 2009, 41, 160-162.	9.4	511
53	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. <i>Nature Genetics</i> , 2009, 41, 1269-1271.	9.4	171
54	BreakDancer: an algorithm for high-resolution mapping of genomic structural variation. <i>Nature Methods</i> , 2009, 6, 677-681.	9.0	1,322
55	An important risk factor in idiopathic generalized epilepsies. <i>Clinical Genetics</i> , 2009, 76, 21-23.	1.0	2
56	Clinical Utility of Array CGH for the Detection of Chromosomal Imbalances Associated with Mental Retardation and Multiple Congenital Anomalies. <i>Annals of the New York Academy of Sciences</i> , 2009, 1151, 157-166.	1.8	109
57	Copy number and sequence variants implicate <i>APBA2</i> as an autism candidate gene. <i>Autism Research</i> , 2009, 2, 359-364.	2.1	32

#	ARTICLE	IF	CITATIONS
58	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. <i>American Journal of Human Genetics</i> , 2009, 84, 148-161.	2.6	530
59	The 12q14 microdeletion syndrome: Additional patients and further evidence that HMGA2 is an important genetic determinant for human height. <i>European Journal of Medical Genetics</i> , 2009, 52, 101-107.	0.7	46
60	Duplication hotspots, rare genomic disorders, and common disease. <i>Current Opinion in Genetics and Development</i> , 2009, 19, 196-204.	1.5	191
61	Chromosomal Microarray Interpretation: What's a Child Neurologist to Do?. <i>Pediatric Neurology</i> , 2009, 41, 391-398.	1.0	24
62	Copy Number Variation in Human Health, Disease, and Evolution. <i>Annual Review of Genomics and Human Genetics</i> , 2009, 10, 451-481.	2.5	1,026
63	Imprinting regulates mammalian snoRNA-encoding chromatin decondensation and neuronal nucleolar size. <i>Human Molecular Genetics</i> , 2009, 18, 4227-4238.	1.4	67
64	The Role of DNA Copy Number Variation in Schizophrenia. <i>Biological Psychiatry</i> , 2009, 66, 1005-1012.	0.7	91
65	Microdeletion/duplication at 15q13.2q13.3 among individuals with features of autism and other neuropsychiatric disorders. <i>Journal of Medical Genetics</i> , 2009, 46, 242-248.	1.5	300
66	Genetic overlap between autism, schizophrenia and bipolar disorder. <i>Genome Medicine</i> , 2009, 1, 102.	3.6	259
67	Genomic disorders ten years on. <i>Genome Medicine</i> , 2009, 1, 42.	3.6	135
68	Chipping away at the common epilepsies with complex genetics: the 15q13.3 microdeletion shows the way. <i>Genome Medicine</i> , 2009, 1, 33.	3.6	17
69	Autism genetics: emerging data from genome-wide copy-number and single nucleotide polymorphism scans. <i>Expert Review of Molecular Diagnostics</i> , 2009, 9, 795-803.	1.5	76
70	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. <i>Journal of Medical Genetics</i> , 2009, 46, 511-523.	1.5	250
71	Microdeletion 15q13.3: a locus with incomplete penetrance for autism, mental retardation, and psychiatric disorders. <i>Journal of Medical Genetics</i> , 2009, 46, 382-388.	1.5	213
72	Genetics of antiepileptic drug resistance. <i>Current Opinion in Neurology</i> , 2009, 22, 150-156.	1.8	25
73	Rare Copy Number Variants_{title}>A Point of Rarity in Genetic Risk for Bipolar Disorder and Schizophrenia</sub>>. <i>Archives of General Psychiatry</i> , 2010, 67, 318.	13.8	173
74	Genome-Scale Technologies Foster Advances in Neurological and Behavioral Research. <i>Current Psychiatry Reviews</i> , 2010, 6, 74-81.	0.9	0
75	Retinoic Acid Induced 1, RAI1: A Dosage Sensitive Gene Related to Neurobehavioral Alterations Including Autistic Behavior. <i>Current Genomics</i> , 2010, 11, 607-617.	0.7	45

#	ARTICLE	IF	CITATIONS
76	A 223-kb De Novo Deletion of PAX9 in a Patient With Oligodontia. <i>Journal of Craniofacial Surgery</i> , 2010, 21, 837-839.	0.3	7
77	Genetic Strategies in Psychiatric Disorders. <i>Focus (American Psychiatric Publishing)</i> , 2010, 8, 307-315.	0.4	2
78	Disorders caused by chromosome abnormalities. <i>The Application of Clinical Genetics</i> , 2010, 3, 159.	1.4	43
79	Newly Recognized Mental Retardation Microdeletion/Duplication Syndromes. <i>Monographs in Human Genetics</i> , 2010, , 101-113.	0.5	0
80	Diversity of Human Copy Number Variation and Multicopy Genes. <i>Science</i> , 2010, 330, 641-646.	6.0	609
81	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2010, 86, 749-764.	2.6	2,325
82	Familial Isolated Clubfoot Is Associated with Recurrent Chromosome 17q23.1q23.2 Microduplications Containing TBX4. <i>American Journal of Human Genetics</i> , 2010, 87, 154-160.	2.6	73
83	Microdeletions of 3q29 Confer High Risk for Schizophrenia. <i>American Journal of Human Genetics</i> , 2010, 87, 229-236.	2.6	215
84	Recurrent Distal 7q11.23 Deletion Including HIP1 and YWHAG Identified in Patients with Intellectual Disabilities, Epilepsy, and Neurobehavioral Problems. <i>American Journal of Human Genetics</i> , 2010, 87, 857-865.	2.6	58
85	Structural Variation in the Human Genome and its Role in Disease. <i>Annual Review of Medicine</i> , 2010, 61, 437-455.	5.0	1,015
86	Copy number variants at Williams-Beuren syndrome 7q11.23 region. <i>Human Genetics</i> , 2010, 128, 3-26.	1.8	134
87	A de novo 11p12-p15.4 duplication in a patient with pharmaco-resistant epilepsy, mental retardation, and dysmorphisms. <i>Brain and Development</i> , 2010, 32, 248-252.	0.6	3
88	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. <i>Human Mutation</i> , 2010, 31, 840-850.	1.1	111
89	A de novo 8.8-Mb deletion of 21q21.1-q21.3 in an autistic male with a complex rearrangement involving chromosomes 6, 10, and 21. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 196-202.	0.7	27
90	Common recurrent microduplication syndromes: Diagnosis and management in clinical practice. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1066-1078.	0.7	32
91	An interstitial 15q11-q14 deletion: Expanded Prader-Willi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 404-408.	0.7	16
92	Challenges in clinical interpretation of microduplications detected by array CGH analysis. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1089-1100.	0.7	35
93	A 15q13.3 homozygous microdeletion associated with a severe neurodevelopmental disorder suggests putative functions of the <i>TRPM1</i> , <i>CHRNA7</i> , and other homozygously deleted genes. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1300-1304.	0.7	55

#	ARTICLE	IF	CITATIONS
94	Homozygous loss of <i>CHRNA7</i> on chromosome 15q13.3 causes severe encephalopathy with seizures and hypotonia. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2908-2911.	0.7	48
95	Epilepsy and chromosomal abnormalities. <i>Italian Journal of Pediatrics</i> , 2010, 36, 36.	1.0	23
96	Genetic Studies of Prader-Willi Patients Provide Evidence for Conservation of Genomic Architecture in Proximal Chromosome 15q. <i>Annals of Human Genetics</i> , 2010, 75, no-no.	0.3	2
97	Recurrent copy number changes in mentally retarded children harbour genes involved in cellular localization and the glutamate receptor complex. <i>European Journal of Human Genetics</i> , 2010, 18, 39-46.	1.4	40
98	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. <i>Nature Genetics</i> , 2010, 42, 203-209.	9.4	539
99	Genome destabilization by homologous recombination in the germ line. <i>Nature Reviews Molecular Cell Biology</i> , 2010, 11, 182-195.	16.1	211
100	Delineation of 15q13.3 microdeletions. <i>Clinical Genetics</i> , 2010, 78, 149-161.	1.0	103
101	Genetics of Cognition-What can Developmental Disorders Teach Us?. , 0, , .		0
102	Copy number variants—an unexpected risk factor for the idiopathic generalized epilepsies. <i>Brain</i> , 2010, 133, 7-8.	3.7	20
103	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. <i>Journal of Medical Genetics</i> , 2010, 47, 332-341.	1.5	447
104	A large replication study and meta-analysis in European samples provides further support for association of <i>AHI1</i> markers with schizophrenia. <i>Human Molecular Genetics</i> , 2010, 19, 1379-1386.	1.4	51
105	The clinical context of copy number variation in the human genome. <i>Expert Reviews in Molecular Medicine</i> , 2010, 12, e8.	1.6	157
106	Penetrance for copy number variants associated with schizophrenia. <i>Human Molecular Genetics</i> , 2010, 19, 3477-3481.	1.4	132
107	Deletion and duplication of 15q24: Molecular mechanisms and potential modification by additional copy number variants. <i>Genetics in Medicine</i> , 2010, 12, 573-586.	1.1	31
108	Classification of pathogenic or benign status of CNVs detected by microarray analysis. <i>Expert Review of Molecular Diagnostics</i> , 2010, 10, 717-721.	1.5	12
109	Diagnosis of Cryptic Chromosomal Syndromes by Fluorescence In Situ Hybridization (FISH). <i>Current Protocols in Human Genetics</i> , 2010, 67, Unit 8.10.1-20.	3.5	4
110	Copy Number Variations in Schizophrenia: Critical Review and New Perspectives on Concepts of Genetics and Disease. <i>American Journal of Psychiatry</i> , 2010, 167, 899-914.	4.0	180
111	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. <i>Brain</i> , 2010, 133, 23-32.	3.7	406

#	ARTICLE	IF	CITATIONS
112	Genomic rearrangements of the GREM1-FMN1 locus cause oligosyndactyly, radio-ulnar synostosis, hearing loss, renal defects syndrome and Cenani-Lenz-like non-syndromic oligosyndactyly. <i>Journal of Medical Genetics</i> , 2010, 47, 569-574.	1.5	38
113	Accurate Distinction of Pathogenic from Benign CNVs in Mental Retardation. <i>PLoS Computational Biology</i> , 2010, 6, e1000752.	1.5	46
114	Genome-Wide Copy Number Variation in Epilepsy: Novel Susceptibility Loci in Idiopathic Generalized and Focal Epilepsies. <i>PLoS Genetics</i> , 2010, 6, e1000962.	1.5	414
115	Genetic Overlaps in Mental Retardation, Autism and Schizophrenia. <i>Monographs in Human Genetics</i> , 2010, , 126-136.	0.5	3
116	A functional analysis of GABARAP on 17p13.1 by knockdown zebrafish. <i>Journal of Human Genetics</i> , 2010, 55, 155-162.	1.1	27
117	Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity. <i>Genetics in Medicine</i> , 2010, 12, 641-647.	1.1	178
118	MEF2C haploinsufficiency caused by either microdeletion of the 5q14.3 region or mutation is responsible for severe mental retardation with stereotypic movements, epilepsy and/or cerebral malformations. <i>Journal of Medical Genetics</i> , 2010, 47, 22-29.	1.5	195
119	The Role of Genetics in the Etiology of Schizophrenia. <i>Psychiatric Clinics of North America</i> , 2010, 33, 35-66.	0.7	212
120	Characterization of a Family with Rare Deletions in CNTNAP5 and DOCK4 Suggests Novel Risk Loci for Autism and Dyslexia. <i>Biological Psychiatry</i> , 2010, 68, 320-328.	0.7	131
121	Neurological Channelopathies. <i>Annual Review of Neuroscience</i> , 2010, 33, 151-172.	5.0	109
122	Detection of microchromosomal aberrations in refractory epilepsy: a pilot study. <i>Epileptic Disorders</i> , 2010, 12, 192-198.	0.7	14
123	Genomic microarrays in mental retardation: from copy number variation to gene, from research to diagnosis. <i>Journal of Medical Genetics</i> , 2010, 47, 289-297.	1.5	135
124	Genetics of Autism. , 2010, , 699-714.		4
125	Genome-wide approaches to schizophrenia. <i>Brain Research Bulletin</i> , 2010, 83, 93-102.	1.4	47
126	Identification of gene copy number variations in patients with mental retardation using array-CGH: Novel syndromes in a large French series. <i>European Journal of Medical Genetics</i> , 2010, 53, 66-75.	0.7	29
127	A de novo 15q13.2q13.3 deletion in a boy with an Angelman syndrome like phenotype. <i>European Journal of Medical Genetics</i> , 2010, 53, 221-224.	0.7	1
128	Autism spectrum disorders associated with chromosomal abnormalities. <i>Research in Autism Spectrum Disorders</i> , 2010, 4, 319-327.	0.8	11
129	Genetics of Early Onset Cognitive Impairment. <i>Annual Review of Genomics and Human Genetics</i> , 2010, 11, 161-187.	2.5	317

#	ARTICLE	IF	CITATIONS
130	Large, rare chromosomal deletions associated with severe early-onset obesity. <i>Nature</i> , 2010, 463, 666-670.	13.7	487
131	A recurrent 14q32.2 microdeletion mediated by expanded TGG repeats. <i>Human Molecular Genetics</i> , 2010, 19, 1967-1973.	1.4	36
132	Copy number variations associated with autism spectrum disorders contribute to a spectrum of neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2010, 12, 694-702.	1.1	116
133	Clinical Genetic Testing for Patients With Autism Spectrum Disorders. <i>Pediatrics</i> , 2010, 125, e727-e735.	1.0	339
134	Chromosomes. , 2010, , 55-138.		2
135	Copy-number variations on the X chromosome in Japanese patients with mental retardation detected by array-based comparative genomic hybridization analysis. <i>Journal of Human Genetics</i> , 2010, 55, 590-599.	1.1	55
136	A three-step workflow procedure for the interpretation of array-based comparative genome hybridization results in patients with idiopathic mental retardation and congenital anomalies. <i>Genetics in Medicine</i> , 2010, 12, 478-485.	1.1	20
137	Inversion variants in the human genome: role in disease and genome architecture. <i>Genome Medicine</i> , 2010, 2, 11.	3.6	60
138	Genetically complex epilepsies, copy number variants and syndrome constellations. <i>Genome Medicine</i> , 2010, 2, 71.	3.6	32
139	Further molecular and clinical delineation of co-locating 17p13.3 microdeletions and microduplications that show distinctive phenotypes. <i>Journal of Medical Genetics</i> , 2010, 47, 299-311.	1.5	137
140	The role of copy number variation in schizophrenia. <i>Expert Review of Neurotherapeutics</i> , 2010, 10, 25-32.	1.4	51
141	Phenotypic variability and genetic susceptibility to genomic disorders. <i>Human Molecular Genetics</i> , 2010, 19, R176-R187.	1.4	234
142	Visualization of Fine-Scale Genomic Structure by Oligonucleotide-Based High-Resolution FISH. <i>Cytogenetic and Genome Research</i> , 2011, 132, 248-254.	0.6	75
143	The Evolving Picture of Microdeletion/Microduplication Syndromes in the Age of Microarray Analysis: Variable Expressivity and Genomic Complexity. <i>Clinics in Laboratory Medicine</i> , 2011, 31, 543-564.	0.7	24
144	Autism spectrum disordersâ€”A genetics review. <i>Genetics in Medicine</i> , 2011, 13, 278-294.	1.1	466
145	Development of new postnatal diagnostic methods for chromosome disorders. <i>Seminars in Fetal and Neonatal Medicine</i> , 2011, 16, 114-118.	1.1	4
146	New cases and refinement of the critical region in the 1q41q42 microdeletion syndrome. <i>European Journal of Medical Genetics</i> , 2011, 54, 42-49.	0.7	36
147	Clinical and molecular characterization of 17q21.31 microdeletion syndrome in 14 French patients with mental retardation. <i>European Journal of Medical Genetics</i> , 2011, 54, 144-151.	0.7	48

#	ARTICLE	IF	CITATIONS
148	Novel deletion encompassing exons 5â€“12 of the UBE3A gene in a girl with Angelman syndrome. <i>European Journal of Medical Genetics</i> , 2011, 54, 348-350.	0.7	5
149	Homozygous deletion of chromosome 15q13.3 including CHRNA7 causes severe mental retardation, seizures, muscular hypotonia, and the loss of KLF13 and TRPM1 potentially cause macrocytosis and congenital retinal dysfunction in siblings. <i>European Journal of Medical Genetics</i> , 2011, 54, e441-e445.	0.7	47
150	Oligonucleotide microarrays in constitutional genetic diagnosis. <i>Expert Review of Molecular Diagnostics</i> , 2011, 11, 521-532.	1.5	15
151	Genome Arrays for the Detection of Copy Number Variations in Idiopathic Mental Retardation, Idiopathic Generalized Epilepsy and Neuropsychiatric Disorders: Lessons for Diagnostic Workflow and Research. <i>Cytogenetic and Genome Research</i> , 2011, 135, 174-202.	0.6	103
152	Human Copy Number Variation and Complex Genetic Disease. <i>Annual Review of Genetics</i> , 2011, 45, 203-226.	3.2	344
153	An evidence-based approach to establish the functional and clinical significance of copy number variants in intellectual and developmental disabilities. <i>Genetics in Medicine</i> , 2011, 13, 777-784.	1.1	371
154	SNP Array Analysis in Constitutional and Cancer Genome Diagnostics â€“ Copy Number Variants, Genotyping and Quality Control. <i>Cytogenetic and Genome Research</i> , 2011, 135, 212-221.	0.6	34
156	Identifying Variations Within Unstable Regions of the Genome Reveal Autism Associated Patterns. , 2011, , .		1
157	Genetic variations and associated pathophysiology in the management of epilepsy. <i>The Application of Clinical Genetics</i> , 2011, 4, 113.	1.4	4
158	Innovations in the Early Diagnosis of Chromosomal Disorders Associated with Intellectual Disability. <i>International Review of Research in Developmental Disabilities</i> , 2011, , 211-228.	0.6	0
159	Prioritization of Epilepsy Associated Candidate Genes by Convergent Analysis. <i>PLoS ONE</i> , 2011, 6, e17162.	1.1	24
160	Causes of learning disability and epilepsy: a review. <i>Current Opinion in Neurology</i> , 2011, 24, 154-158.	1.8	9
161	Genetic contribution to common epilepsies. <i>Current Opinion in Neurology</i> , 2011, 24, 140-145.	1.8	41
162	Chromosome microarray analysis in a clinical environment: new perspective and new challenge. <i>British Journal of Biomedical Science</i> , 2011, 68, 100-108.	1.2	5
164	Epilepsy and the new cytogenetics. <i>Epilepsia</i> , 2011, 52, 423-432.	2.6	56
165	Absence seizures with intellectual disability as a phenotype of the 15q13.3 microdeletion syndrome. <i>Epilepsia</i> , 2011, 52, e194-8.	2.6	29
166	The phenotype of recurrent 10q22q23 deletions and duplications. <i>European Journal of Human Genetics</i> , 2011, 19, 400-408.	1.4	63
167	Analysis of genetic deletions and duplications in the University College London bipolar disorder case control sample. <i>European Journal of Human Genetics</i> , 2011, 19, 588-592.	1.4	38

#	ARTICLE	IF	CITATIONS
168	Deletions flanked by breakpoints 3 and 4 on 15q13 may contribute to abnormal phenotypes. <i>European Journal of Human Genetics</i> , 2011, 19, 547-554.	1.4	18
169	Microdeletions and Microduplications in Patients with Congenital Heart Disease and Multiple Congenital Anomalies. <i>Congenital Heart Disease</i> , 2011, 6, 592-602.	0.0	82
170	Etiological heterogeneity in autism spectrum disorders: More than 100 genetic and genomic disorders and still counting. <i>Brain Research</i> , 2011, 1380, 42-77.	1.1	788
171	Genetics of Schizophrenia: New Findings and Challenges. <i>Annual Review of Genomics and Human Genetics</i> , 2011, 12, 121-144.	2.5	160
172	Microdeletion/microduplication of proximal 15q11.2 between BP1 and BP2: a susceptibility region for neurological dysfunction including developmental and language delay. <i>Human Genetics</i> , 2011, 130, 517-528.	1.8	219
174	The effects of aging vs. ± 7 nAChR subunit deficiency on the mouse brain transcriptome: aging beats the deficiency. <i>Age</i> , 2011, 33, 1-13.	3.0	6
175	Copy Number Variants: A New Molecular Frontier in Clinical Psychiatry. <i>Current Psychiatry Reports</i> , 2011, 13, 129-137.	2.1	11
176	Reversed clinical phenotype due to a microduplication of Sotos syndrome region detected by array CGH: Microcephaly, developmental delay and delayed bone age. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1374-1378.	0.7	25
177	Pharmaco-genetically guided treatment of recurrent rage outbursts in an adult male with 15q13.3 deletion syndrome. , 2011, 155, 805-810.		46
178	A novel 800â€‰kb microduplication of chromosome 16q22.1 resulting in learning disability and epilepsy may explain phenotypic variability in a family with 15q13 microdeletion. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1453-1457.	0.7	3
179	Phenotypic variability of distal 22q11.2 copy number abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1623-1633.	0.7	59
180	A de novo 2.1â€‰Mb deletion of 13q12.11 in a child with developmental delay and minor dysmorphic features. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2538-2542.	0.7	9
181	A small homozygous microdeletion of 15q13.3 including the <i>CHRNA7</i> gene in a girl with a spectrum of severe neurodevelopmental features. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2795-2800.	0.7	44
182	Rare copy number variants are an important cause of epileptic encephalopathies. <i>Annals of Neurology</i> , 2011, 70, 974-985.	2.8	222
183	On the sequence-directed nature of human gene mutation: The role of genomic architecture and the local DNA sequence environment in mediating gene mutations underlying human inherited disease. <i>Human Mutation</i> , 2011, 32, 1075-1099.	1.1	99
184	Challenges in studying genomic structural variant formation mechanisms: The short-read dilemma and beyond. <i>BioEssays</i> , 2011, 33, 840-850.	1.2	34
185	Schizophrenia Genes: On the Matter of Their Convergence. <i>Current Topics in Behavioral Neurosciences</i> , 2011, 12, 429-440.	0.8	5
186	Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. <i>Genome Research</i> , 2011, 21, 33-46.	2.4	72

#	ARTICLE	IF	CITATIONS
187	Common neurological co-morbidities in autism spectrum disorders. <i>Current Opinion in Pediatrics</i> , 2011, 23, 609-615.	1.0	83
188	Pharmacotherapeutic Implications of the Association Between Genomic Instability at Chromosome 15q13.3 and Autism Spectrum Disorders. <i>Clinical Neuropharmacology</i> , 2011, 34, 203-205.	0.2	20
189	Phenotype mining in CNV carriers from a population cohort â€“. <i>Human Molecular Genetics</i> , 2011, 20, 2686-2695.	1.4	13
190	15q11.2â€“13.3 chromatin analysis reveals epigenetic regulation of CHRNA7 with deficiencies in Rett and autism brain. <i>Human Molecular Genetics</i> , 2011, 20, 4311-4323.	1.4	93
191	Neurodevelopmental Disabilities. , 2011, , .		18
192	Ionising radiation and genetic risks. XVI. A genome-based framework for risk estimation in the light of recent advances in genome research. <i>International Journal of Radiation Biology</i> , 2011, 87, 161-178.	1.0	15
193	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. <i>PLoS Genetics</i> , 2012, 8, e1002521.	1.5	358
194	Unique and atypical deletions in Praderâ€“Willi syndrome reveal distinct phenotypes. <i>European Journal of Human Genetics</i> , 2012, 20, 283-290.	1.4	86
195	Small rare recurrent deletions and reciprocal duplications in 2q21.1, including brain-specific ARHGEF4 and GPR148. <i>Human Molecular Genetics</i> , 2012, 21, 3345-3355.	1.4	22
196	Multiple genes in the 15q13-q14 chromosomal region are associated with schizophrenia. <i>Psychiatric Genetics</i> , 2012, 22, 1-14.	0.6	30
197	Phenotypic Heterogeneity of Genomic Disorders and Rare Copy-Number Variants. <i>New England Journal of Medicine</i> , 2012, 367, 1321-1331.	13.9	519
198	Genetics and Epigenetics of Autism Spectrum Disorders. <i>Research and Perspectives in Neurosciences</i> , 2012, , 105-132.	0.4	4
199	Opportunities and Challenges for Genome Sequencing in the Clinic. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 65-83.	1.0	9
200	Genomic structural variation in psychiatric disorders. <i>Development and Psychopathology</i> , 2012, 24, 1335-1344.	1.4	14
201	Molekulare Karyotypisierung in der Routinediagnostik â€“ RÃ¼ckblick und Ausblick/Molecular karyotyping in routine diagnostics â€“ a view back and forth. <i>Laboratoriums Medizin</i> , 2012, 36, .	0.1	0
202	Atypical face shape and genomic structural variants in epilepsy. <i>Brain</i> , 2012, 135, 3101-3114.	3.7	77
203	Current Progress in the Genetic Research of Schizophrenia: Relevance for Drug Discovery?. <i>Current Pharmaceutical Biotechnology</i> , 2012, 13, 1614-1621.	0.9	4
204	Recurrent Transmission of a 17q12 Microdeletion and a Variable Clinical Spectrum. <i>Molecular Syndromology</i> , 2011, 2, 72-75.	0.3	24

#	ARTICLE	IF	CITATIONS
205	A genetic model for neurodevelopmental disease. <i>Current Opinion in Neurobiology</i> , 2012, 22, 829-836.	2.0	47
206	Clinical impact of copy number variation analysis using high-resolution microarray technologies: advantages, limitations and concerns. <i>Genome Medicine</i> , 2012, 4, 80.	3.6	63
207	The Challenges of Studying Complex and Dynamic Regions of the Human Genome. <i>Methods in Molecular Biology</i> , 2012, 838, 187-207.	0.4	13
208	CNVs: Harbingers of a Rare Variant Revolution in Psychiatric Genetics. <i>Cell</i> , 2012, 148, 1223-1241.	13.5	759
209	Homozygous deletion of a gene-free region of 4p15 in a child with multiple anomalies: Could biallelic loss of conserved, non-coding elements lead to a phenotype?. <i>European Journal of Medical Genetics</i> , 2012, 55, 63-66.	0.7	3
210	Schizophrenia genetics: progress, at last. <i>Current Opinion in Genetics and Development</i> , 2012, 22, 238-244.	1.5	46
211	Rare copy number variants in neuropsychiatric disorders: Specific phenotype or not?. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 812-822.	1.1	34
212	Genomics, Intellectual Disability, and Autism. <i>New England Journal of Medicine</i> , 2012, 366, 733-743.	13.9	276
213	Exome Sequencing Followed by Large-Scale Genotyping Fails to Identify Single Rare Variants of Large Effect in Idiopathic Generalized Epilepsy. <i>American Journal of Human Genetics</i> , 2012, 91, 293-302.	2.6	95
214	Acquired genomic copy number changes in CML patients with the Philadelphia chromosome (Ph+). <i>Cancer Genetics</i> , 2012, 205, 513-518.	0.2	6
215	Copy number variations in neurodevelopmental disorders. <i>Progress in Neurobiology</i> , 2012, 99, 81-91.	2.8	150
216	Independent estimation of the frequency of rare CNVs in the UK population confirms their role in schizophrenia. <i>Schizophrenia Research</i> , 2012, 135, 1-7.	1.1	73
217	Epigenetics, Brain and Behavior. <i>Research and Perspectives in Neurosciences</i> , 2012, , .	0.4	5
220	Microdeletions in 16p11.2 and 13q31.3 associated with developmental delay and generalized overgrowth. <i>Genetics and Molecular Research</i> , 2012, 11, 3133-3137.	0.3	1
221	Clinical applications of schizophrenia genetics: genetic diagnosis, risk, and counseling in the molecular era. <i>The Application of Clinical Genetics</i> , 2012, 5, 1.	1.4	35
223	Genetic counselling and ethical issues with chromosome microarray analysis in prenatal testing. <i>Prenatal Diagnosis</i> , 2012, 32, 389-395.	1.1	82
224	Microdeletion and Microduplication Syndromes. <i>Methods in Molecular Biology</i> , 2012, 838, 29-75.	0.4	58
225	The genetic variability and commonality of neurodevelopmental disease. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2012, 160C, 118-129.	0.7	101

#	ARTICLE	IF	CITATIONS
226	A novel X-linked disorder with developmental delay and autistic features. <i>Annals of Neurology</i> , 2012, 71, 498-508.	2.8	33
227	The Genetic Architecture of Schizophrenia: New Mutations and Emerging Paradigms. <i>Annual Review of Medicine</i> , 2012, 63, 63-80.	5.0	98
228	Structural Genomic Variation in Intellectual Disability. <i>Methods in Molecular Biology</i> , 2012, 838, 77-95.	0.4	10
229	Diagnostic Yield of Chromosomal Microarray Analysis in an Autism Primary Care Practice: Which Guidelines to Implement?. <i>Journal of Autism and Developmental Disorders</i> , 2012, 42, 1582-1591.	1.7	56
230	NIPA2 located in 15q11.2 is mutated in patients with childhood absence epilepsy. <i>Human Genetics</i> , 2012, 131, 1217-1224.	1.8	33
231	Resolving the Breakpoints of the 17q21.31 Microdeletion Syndrome with Next-Generation Sequencing. <i>American Journal of Human Genetics</i> , 2012, 90, 599-613.	2.6	22
232	Genome-wide survey implicates the influence of copy number variants (CNVs) in the development of early-onset bipolar disorder. <i>Molecular Psychiatry</i> , 2012, 17, 421-432.	4.1	76
233	High rates of de novo 15q11q13 inversions in human spermatozoa. <i>Molecular Cytogenetics</i> , 2012, 5, 11.	0.4	7
234	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. <i>Human Mutation</i> , 2012, 33, 165-179.	1.1	45
235	Genetic architecture of reciprocal CNVs. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 240-248.	1.5	51
236	Microdeletion syndromes. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 232-239.	1.5	42
237	The unexpected role of copy number variations in juvenile myoclonic epilepsy. <i>Epilepsy and Behavior</i> , 2013, 28, S66-S68.	0.9	16
238	The Genetic Landscapes of Autism Spectrum Disorders. <i>Annual Review of Genomics and Human Genetics</i> , 2013, 14, 191-213.	2.5	352
239	Deletions and Other Structural Abnormalities of the Autosomes. , 2013, , 1-37.		4
241	Array Comparative Genomic Hybridization. <i>Methods in Molecular Biology</i> , 2013, , .	0.4	1
242	Neurodevelopmental Genomics of Autism, Schizophrenia, and Related Disorders. , 2013, , 695-708.		0
243	Recent advances in the molecular genetics of epilepsy. <i>Journal of Medical Genetics</i> , 2013, 50, 271-279.	1.5	111
244	The emerging spectrum of allelic variation in schizophrenia: current evidence and strategies for the identification and functional characterization of common and rare variants. <i>Molecular Psychiatry</i> , 2013, 18, 38-52.	4.1	75

#	ARTICLE	IF	CITATIONS
245	Transcriptome study of differential expression in schizophrenia. <i>Human Molecular Genetics</i> , 2013, 22, 5001-5014.	1.4	73
246	Epigenetics and Complex Traits. , 2013, , .		1
247	Does epilepsy in multiplex autism pedigrees define a different subgroup in terms of clinical characteristics and genetic risk?. <i>Molecular Autism</i> , 2013, 4, 47.	2.6	43
248	Medizin, 2013, 36, --.	0.1	0
249	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. <i>Neurology</i> , 2013, 81, 1507-1514.	1.5	140
250	Copy number variation analysis implicates the cell polarity gene glypican 5 as a human spina bifida candidate gene. <i>Human Molecular Genetics</i> , 2013, 22, 1097-1111.	1.4	29
251	Expression of autism spectrum and schizophrenia in patients with a 22q11.2 deletion. <i>Schizophrenia Research</i> , 2013, 143, 55-59.	1.1	68
252	Epilepsy with cognitive deficit and autism spectrum disorders: Prospective diagnosis by array CGH. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2013, 162, 24-35.	1.1	20
253	Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis. <i>Human Mutation</i> , 2013, 34, 210-220.	1.1	48
254	A case of an atypically large proximal 15q deletion as cause for Prader-Willi syndrome arising from a de novo unbalanced translocation. <i>European Journal of Medical Genetics</i> , 2013, 56, 510-514.	0.7	3
255	Mosaic 15q13.3 deletion including CHRNA7 gene in monozygotic twins. <i>European Journal of Medical Genetics</i> , 2013, 56, 274-277.	0.7	9
256	Identification of single gene deletions at 15q13.3: further evidence that <i>CHRNA7</i> causes the 15q13.3 microdeletion syndrome phenotype. <i>Clinical Genetics</i> , 2013, 83, 345-351.	1.0	89
257	Distinguishing Somatic and Germline Copy Number Events in Cancer Patient DNA Hybridized to Whole-Genome SNP Genotyping Arrays. <i>Methods in Molecular Biology</i> , 2013, 973, 355-372.	0.4	6
258	Clinical application of 2.7M Cytogenetics array for CNV detection in subjects with idiopathic autism and/or intellectual disability. <i>Clinical Genetics</i> , 2013, 83, 145-154.	1.0	43
259	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. <i>Clinical Genetics</i> , 2013, 84, 507-521.	1.0	23
260	Distribution of Disease-Associated Copy Number Variants Across Distinct Disorders of Cognitive Development. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2013, 52, 414-430.e14.	0.3	28
261	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. <i>Nature Genetics</i> , 2013, 45, 546-551.	9.4	301
262	Developmental brain dysfunction: revival and expansion of old concepts based on new genetic evidence. <i>Lancet Neurology</i> , The, 2013, 12, 406-414.	4.9	268

#	ARTICLE	IF	CITATIONS
263	Etiological Heterogeneity in Autism Spectrum Disorders. , 2013, , 113-144.		10
264	Application of custom-designed oligonucleotide array CGH in 145 patients with autistic spectrum disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 620-625.	1.4	37
265	Allosteric alpha-7 nicotinic receptor modulation and P50 sensory gating in schizophrenia: A proof-of-mechanism study. <i>Neuropharmacology</i> , 2013, 64, 197-204.	2.0	59
266	Angelman syndrome in Denmark. Birth incidence, genetic findings, and age at diagnosis. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2197-2203.	0.7	76
267	Role of interactions in pharmacogenetic studies: leukotrienes in asthma. <i>Pharmacogenomics</i> , 2013, 14, 923-929.	0.6	4
268	Genetics of Epilepsy and Refractory Epilepsy. <i>Colloquium Series on the Genetic Basis of Human Disease</i> , 2013, 2, 1-119.	0.0	2
269	Familial cosegregation of rare genetic variants with disease in complex disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 444-450.	1.4	14
270	Rare CNVs and Tag SNPs at 15q11.2 Are Associated With Schizophrenia in the Han Chinese Population. <i>Schizophrenia Bulletin</i> , 2013, 39, 712-719.	2.3	52
271	Genetics of idiopathic generalized epilepsy: An overview. <i>Neurology India</i> , 2013, 61, 572.	0.2	12
272	Developmental Disabilities, Autism, and Schizophrenia at a Single Locus. , 2013, , 617-630.		1
273	Genome-wide gene expression in a patient with 15q13.3 homozygous microdeletion syndrome. <i>European Journal of Human Genetics</i> , 2013, 21, 1093-1099.	1.4	23
274	Different electroclinical picture of generalized epilepsy in two families with 15q13.3 microdeletion. <i>Epilepsia</i> , 2013, 54, e69-73.	2.6	14
275	Exonâ€disrupting deletions of <sc><i>NRXN1</i></sc> in idiopathic generalized epilepsy. <i>Epilepsia</i> , 2013, 54, 256-264.	2.6	59
276	BBCRE: brain and body genetic resource exchange. <i>Database: the Journal of Biological Databases and Curation</i> , 2013, 2013, bat067.	1.4	4
277	De novo 15q13.3 microdeletion with cryptogenic west syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2582-2587.	0.7	8
278	Expanded Praderâ€Willi syndrome due to chromosome 15q11.2â€14 deletion: Report and a review of literature. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1309-1318.	0.7	9
279	Association Study of the 2-bp Deletion Polymorphism in Exon 6 of the <i>CHRFAM7A</i> Gene with Idiopathic Generalized Epilepsy. <i>DNA and Cell Biology</i> , 2013, 32, 640-647.	0.9	17
280	An emerging role for Wnt and <sc>GSK3</sc> signaling pathways in schizophrenia. <i>Clinical Genetics</i> , 2013, 83, 511-517.	1.0	44

#	ARTICLE	IF	CITATIONS
281	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. <i>Genome Research</i> , 2013, 23, 1395-1409.	2.4	120
282	Genetic pathways to autism spectrum disorders. <i>Neuropsychiatry</i> , 2013, 3, 193-207.	0.4	4
283	Original article A transcript coding for a partially duplicated form of $\alpha 7$ nicotinic acetylcholine receptor is absent from the CD4 + T-lymphocytes of patients with autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE). <i>Folia Neuropathologica</i> , 2013, 1, 65-75.	0.5	12
284	Discovering the Genetics of Autism. , 0, , .		0
285	Investigation of 15q11-q13, 16p11.2 and 22q13 CNVs in Autism Spectrum Disorder Brazilian Individuals with and without Epilepsy. <i>PLoS ONE</i> , 2014, 9, e107705.	1.1	17
288	Ohnologs are overrepresented in pathogenic copy number mutations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 361-366.	3.3	57
289	SNP arrays: comparing diagnostic yields for four platforms in children with developmental delay. <i>BMC Medical Genomics</i> , 2014, 7, 70.	0.7	13
290	Neurodevelopmental and neuropsychiatric disorders represent an interconnected molecular system. <i>Molecular Psychiatry</i> , 2014, 19, 294-301.	4.1	188
291	Clinical utility gene card for: 15q13.3 microdeletion syndrome. <i>European Journal of Human Genetics</i> , 2014, 22, 1338-1338.	1.4	10
292	Family based genome-wide copy number scan identifies complex rearrangements at 17q21.31 in dyslexics. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2014, 165, 572-580.	1.1	16
293	Further delineation of eye manifestations in homozygous 15q13.3 microdeletions including <i>TRPM1</i> : A differential diagnosis of ceroid lipofuscinosis. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1537-1544.	0.7	22
294	Congenital Neuroblastoma in a Neonate With Isotretinoin Embryopathy. <i>Journal of Pediatric Hematology/Oncology</i> , 2014, 36, e75-e77.	0.3	4
295	Cognitive deficit and autism spectrum disorders: prospective diagnosis by array CGH. <i>Pathology</i> , 2014, 46, 41-45.	0.3	18
296	Genome-wide analysis associates familial colorectal cancer with increases in copy number variations and a rare structural variation at 12p12.3. <i>Carcinogenesis</i> , 2014, 35, 315-323.	1.3	31
297	Expanding the Clinical Phenotype Associated With <i>ELOVL4</i> Mutation. <i>JAMA Neurology</i> , 2014, 71, 470.	4.5	110
298	Biological Overlap of Attention-Deficit/Hyperactivity Disorder and Autism Spectrum Disorder: Evidence From Copy Number Variants. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2014, 53, 761-770.e26.	0.3	105
299	Cortical synaptic NMDA receptor deficits in $\alpha 7$ nicotinic acetylcholine receptor gene deletion models: Implications for neuropsychiatric diseases. <i>Neurobiology of Disease</i> , 2014, 63, 129-140.	2.1	55
300	Theory of mind and the social brain: implications for understanding the genetic basis of schizophrenia. <i>Genes, Brain and Behavior</i> , 2014, 13, 104-117.	1.1	39

#	ARTICLE	IF	CITATIONS
301	The Penetrance of Copy Number Variations for Schizophrenia and Developmental Delay. <i>Biological Psychiatry</i> , 2014, 75, 378-385.	0.7	321
302	A genome-wide association study and biological pathway analysis of epilepsy prognosis in a prospective cohort of newly treated epilepsy. <i>Human Molecular Genetics</i> , 2014, 23, 247-258.	1.4	33
303	Compound heterozygous microdeletion of chromosome 15q13.3 region in a child with hypotonia, impaired vision, and global developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1815-1820.	0.7	14
304	Nicotinic acetylcholine receptors in human genetic disease. <i>Genetics in Medicine</i> , 2014, 16, 649-656.	1.1	82
305	Of mice and men: molecular genetics of congenital heart disease. <i>Cellular and Molecular Life Sciences</i> , 2014, 71, 1327-1352.	2.4	159
306	CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. <i>European Journal of Human Genetics</i> , 2014, 22, 1071-1076.	1.4	37
307	Application of array comparative genomic hybridization in 256 patients with developmental delay or intellectual disability. <i>Journal of Applied Genetics</i> , 2014, 55, 125-144.	1.0	37
308	A de novo convergence of autism genetics and molecular neuroscience. <i>Trends in Neurosciences</i> , 2014, 37, 95-105.	4.2	410
309	Palindromic GOLGA8 core duplicons promote chromosome 15q13.3 microdeletion and evolutionary instability. <i>Nature Genetics</i> , 2014, 46, 1293-1302.	9.4	96
310	Remind me again what disease we are studying? A population genetics, genetic analysis, and real data perspective on why progress on identifying genetic influences on common epilepsies has been so slow. <i>Progress in Brain Research</i> , 2014, 213, 199-221.	0.9	8
311	CNVs in Epilepsy. <i>Current Genetic Medicine Reports</i> , 2014, 2, 162-167.	1.9	28
312	Advances in Genetic Discovery and Implications for Counseling of Patients and Families with Autism Spectrum Disorders. <i>Current Genetic Medicine Reports</i> , 2014, 2, 124-134.	1.9	7
313	New technologies in molecular genetics. <i>Progress in Brain Research</i> , 2014, 213, 253-278.	0.9	6
314	Congenital Hand Anomalies and Associated Syndromes. , 2014, , .		16
315	Genome-wide association tests of inversions with application to psoriasis. <i>Human Genetics</i> , 2014, 133, 967-974.	1.8	11
316	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. <i>European Journal of Medical Genetics</i> , 2014, 57, 163-168.	0.7	11
317	Cortical parvalbumin GABAergic deficits with 1±7 nicotinic acetylcholine receptor deletion: implications for schizophrenia. <i>Molecular and Cellular Neurosciences</i> , 2014, 61, 163-175.	1.0	55
318	The Genetics of Microdeletion and Microduplication Syndromes: An Update. <i>Annual Review of Genomics and Human Genetics</i> , 2014, 15, 215-244.	2.5	145

#	ARTICLE	IF	CITATIONS
319	A Mouse Model that Recapitulates Cardinal Features of the 15q13.3 Microdeletion Syndrome Including Schizophrenia- and Epilepsy-Related Alterations. <i>Biological Psychiatry</i> , 2014, 76, 128-137.	0.7	95
320	Reduced <i>Chrna7</i> expression in C3H mice is associated with increases in hippocampal parvalbumin and glutamate decarboxylase-67 (GAD67) as well as altered levels of GABAA receptor subunits. <i>Neuroscience</i> , 2014, 273, 52-64.	1.1	12
321	The role of nicotinic receptors in shaping and functioning of the glutamatergic system: A window into cognitive pathology. <i>Neuroscience and Biobehavioral Reviews</i> , 2014, 46, 315-325.	2.9	25
322	High rate of disease-related copy number variations in childhood onset schizophrenia. <i>Molecular Psychiatry</i> , 2014, 19, 568-572.	4.1	116
323	Structural genomic variants in pediatric seizure disorders. <i>Journal of Pediatric Epilepsy</i> , 2015, 01, 161-169.	0.1	0
325	Partial tetrasomy of the proximal long arm of chromosome 15 in two patients: the significance of the gene dosage in terms of phenotype. <i>Molecular Cytogenetics</i> , 2015, 8, 41.	0.4	3
326	An assessment of sex bias in neurodevelopmental disorders. <i>Genome Medicine</i> , 2015, 7, 94.	3.6	90
327	Pharmacological Characterisation of Nicotinic Acetylcholine Receptors Expressed in Human iPSC-Derived Neurons. <i>PLoS ONE</i> , 2015, 10, e0125116.	1.1	29
328	Functional Impact of 14 Single Nucleotide Polymorphisms Causing Missense Mutations of Human $\alpha 7$ Nicotinic Receptor. <i>PLoS ONE</i> , 2015, 10, e0137588.	1.1	5
329	Recent developments in the genetics of childhood epileptic encephalopathies: impact in clinical practice. <i>Arquivos De Neuro-Psiquiatria</i> , 2015, 73, 946-958.	0.3	11
330	Clinical Genetic Testing in Epilepsy. <i>Epilepsy Currents</i> , 2015, 15, 197-201.	0.4	22
331	Human inversions and their functional consequences. <i>Briefings in Functional Genomics</i> , 2015, 14, 369-379.	1.3	117
332	Clinical and Genetic Heterogeneity of the 15q13.3 Microdeletion Syndrome. <i>Molecular Syndromology</i> , 2015, 6, 222-228.	0.3	18
333	Mouse Model of Chromosome 15q13.3 Microdeletion Syndrome Demonstrates Features Related to Autism Spectrum Disorder. <i>Journal of Neuroscience</i> , 2015, 35, 16282-16294.	1.7	51
334	A case study of autism spectrum disorder (ASD) symptomatology in a child with 15q13.3 deletion and Williams syndrome. <i>Journal of Developmental and Physical Disabilities</i> , 2015, 27, 111-118.	1.0	0
335	The $\alpha 7$ nicotinic acetylcholine receptor: A mediator of pathogenesis and therapeutic target in autism spectrum disorders and Down syndrome. <i>Biochemical Pharmacology</i> , 2015, 97, 363-377.	2.0	22
336	Modulation of aggressive behavior in mice by nicotinic receptor subtypes. <i>Biochemical Pharmacology</i> , 2015, 97, 488-497.	2.0	27
337	Global diversity, population stratification, and selection of human copy-number variation. <i>Science</i> , 2015, 349, aab3761.	6.0	293

#	ARTICLE	IF	CITATIONS
338	Epilepsy genetics: The ongoing revolution. <i>Revue Neurologique</i> , 2015, 171, 539-557.	0.6	10
339	Long-lasting changes in neural networks to compensate for altered nicotinic input. <i>Biochemical Pharmacology</i> , 2015, 97, 418-424.	2.0	10
340	The human clinical phenotypes of altered CHRNA7 copy number. <i>Biochemical Pharmacology</i> , 2015, 97, 352-362.	2.0	97
341	CNVs in neuropsychiatric disorders. <i>Human Molecular Genetics</i> , 2015, 24, R45-R49.	1.4	134
342	New Genes for Focal Epilepsies with Speech and Language Disorders. <i>Current Neurology and Neuroscience Reports</i> , 2015, 15, 35.	2.0	56
343	Global Developmental Delay and Intellectual Disability. , 2015, , 151-161.		4
344	Analysis of <i>CHRNA7</i> rare variants in autism spectrum disorder susceptibility. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 715-723.	0.7	41
345	Copy number variants in attention-deficit hyperactive disorder. <i>Psychiatric Genetics</i> , 2015, 25, 59-70.	0.6	25
346	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. <i>Epilepsy Research</i> , 2015, 117, 70-73.	0.8	6
347	A roadmap for precision medicine in the epilepsies. <i>Lancet Neurology, The</i> , 2015, 14, 1219-1228.	4.9	160
348	Genetic variation and the de novo assembly of human genomes. <i>Nature Reviews Genetics</i> , 2015, 16, 627-640.	7.7	310
349	Therapeutic Potential of <i>CHRNA7</i> Nicotinic Acetylcholine Receptors. <i>Pharmacological Reviews</i> , 2015, 67, 1025-1073.	7.1	123
351	Copy Number Variants and Epilepsy: New Emerging Syndromes. , 2015, , 1-14.		0
352	Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature. <i>Genetics in Medicine</i> , 2015, 17, 149-157.	1.1	103
353	Multiple Biological Sequence Alignment: Scoring Functions, Algorithms and Applications. , 2016, , .		9
354	Major influence of repetitive elements on disease-associated copy number variants (CNVs). <i>Human Genomics</i> , 2016, 10, 30.	1.4	18
355	The mechanism of different diseases treated by the same way of acupuncture at Chǎngqiǎng (GV 1) for cognitive disorder. <i>World Journal of Acupuncture-moxibustion</i> , 2016, 26, 24-29.	0.1	0
357	Replication analyses of four chromosomal deletions with schizophrenia via independent large-scale meta-analyses. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2016, 171, 1161-1169.	1.1	7

#	ARTICLE	IF	CITATIONS
358	15q13.3 duplication in two patients with childhood-onset schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 777-783.	1.1	27
359	Characterizing polymorphic inversions in human genomes by single-cell sequencing. Genome Research, 2016, 26, 1575-1587.	2.4	67
360	Human adaptation and evolution by segmental duplication. Current Opinion in Genetics and Development, 2016, 41, 44-52.	1.5	157
361	15q13.3 homozygous knockout mouse model display epilepsy-, autism- and schizophrenia-related phenotypes. Translational Psychiatry, 2016, 6, e860-e860.	2.4	45
362	Identifying CNVs in 15q11q13 and 16p11.2 of Patients with Seizures Increases the Rates of Detecting Pathogenic Changes. Molecular Syndromology, 2016, 7, 329-336.	0.3	0
364	Genomic architecture of human chromosomal diseases. Russian Journal of Genetics, 2016, 52, 447-462.	0.2	2
365	Low-pass whole-genome sequencing in clinical cytogenetics: a validated approach. Genetics in Medicine, 2016, 18, 940-948.	1.1	138
366	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. Nature Neuroscience, 2016, 19, 517-522.	7.1	72
367	The complex behavioral phenotype of 15q13.3 microdeletion syndrome. Genetics in Medicine, 2016, 18, 1111-1118.	1.1	45
368	The 15q13.3 deletion syndrome: Deficient $\alpha 7$ -containing nicotinic acetylcholine receptor-mediated neurotransmission in the pathogenesis of neurodevelopmental disorders. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2016, 64, 109-117.	2.5	36
369	Recurrent copy number variations as risk factors for neurodevelopmental disorders: critical overview and analysis of clinical implications. Journal of Medical Genetics, 2016, 53, 73-90.	1.5	90
370	Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 2016, 17, 9-18.	7.7	614
371	A CNV Catalogue. , 2017, , 235-417.		1
372	A clear bias in parental origin of de novo pathogenic CNVs related to intellectual disability, developmental delay and multiple congenital anomalies. Scientific Reports, 2017, 7, 44446.	1.6	19
373	A de novo nonsense mutation in <i>ZBTB18</i> plus a de novo 15q13.3 microdeletion in a 6-year-old female. American Journal of Medical Genetics, Part A, 2017, 173, 1251-1256.	0.7	7
374	Oxidative stress-driven parvalbumin interneuron impairment as a common mechanism in models of schizophrenia. Molecular Psychiatry, 2017, 22, 936-943.	4.1	280
375	Chrna7 deficient mice manifest no consistent neuropsychiatric and behavioral phenotypes. Scientific Reports, 2017, 7, 39941.	1.6	43
376	Clinical interpretation of copy number variants in the human genome. Journal of Applied Genetics, 2017, 58, 449-457.	1.0	99

#	ARTICLE	IF	CITATIONS
377	MLPA analysis in a cohort of patients with autism. <i>Molecular Cytogenetics</i> , 2017, 10, 2.	0.4	1
378	Allosteric modulation of nicotinic and GABA A receptor subtypes differentially modify autism-like behaviors in the BTBR mouse model. <i>Neuropharmacology</i> , 2017, 126, 38-47.	2.0	24
379	Nicotine Elicits Convulsive Seizures by Activating Amygdalar Neurons. <i>Frontiers in Pharmacology</i> , 2017, 8, 57.	1.6	25
380	Modulating Neuroinflammation to Treat Neuropsychiatric Disorders. <i>BioMed Research International</i> , 2017, 2017, 1-21.	0.9	51
381	Recurrent de novo mutations in neurodevelopmental disorders: properties and clinical implications. <i>Genome Medicine</i> , 2017, 9, 101.	3.6	112
382	Informed Decision-Making in the Context of Prenatal Chromosomal Microarray. <i>Journal of Genetic Counseling</i> , 2018, 27, 1130-1147.	0.9	7
383	An estimation of the prevalence of genomic disorders using chromosomal microarray data. <i>Journal of Human Genetics</i> , 2018, 63, 795-801.	1.1	49
384	Otud7a Knockout Mice Recapitulate Many Neurological Features of 15q13.3 Microdeletion Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 296-308.	2.6	65
385	OTUD7A Regulates Neurodevelopmental Phenotypes in the 15q13.3 Microdeletion Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 278-295.	2.6	81
386	CNV biology in neurodevelopmental disorders. <i>Current Opinion in Neurobiology</i> , 2018, 48, 183-192.	2.0	89
387	An Exploratory Trial of Transdermal Nicotine for Aggression and Irritability in Adults with Autism Spectrum Disorder. <i>Journal of Autism and Developmental Disorders</i> , 2018, 48, 2748-2757.	1.7	20
388	Bidirectional Regulation of Aggression in Mice by Hippocampal Alpha-7 Nicotinic Acetylcholine Receptors. <i>Neuropsychopharmacology</i> , 2018, 43, 1267-1275.	2.8	27
389	Predicting Local Inversions Using Rectangle Clustering and Representative Rectangle Prediction. , 2018, , .		3
390	Taurodeoxycholate Increases the Number of Myeloid-Derived Suppressor Cells That Ameliorate Sepsis in Mice. <i>Frontiers in Immunology</i> , 2018, 9, 1984.	2.2	38
391	Recent Advances in the Genetics of Schizophrenia. <i>Molecular Neuropsychiatry</i> , 2018, 4, 35-51.	3.0	81
392	BAMSI: a multi-cloud service for scalable distributed filtering of massive genome data. <i>BMC Bioinformatics</i> , 2018, 19, 240.	1.2	4
393	Identification of novel genomic imbalances in Saudi patients with congenital heart disease. <i>Molecular Cytogenetics</i> , 2018, 11, 9.	0.4	4
394	Genetic generalized epilepsies. <i>Epilepsia</i> , 2018, 59, 1148-1153.	2.6	72

#	ARTICLE	IF	CITATIONS
395	Copy number variation related disease genes. Quantitative Biology, 2018, 6, 99-112.	0.3	4
396	Association study and mutation sequencing of genes on chromosome 15q11-q13 identified GABRG3 as a susceptibility gene for autism in Chinese Han population. Translational Psychiatry, 2018, 8, 152.	2.4	13
397	Chromosome Disorders. , 2018, , 211-223.e2.		1
398	New avenues in molecular genetics for the diagnosis and application of therapeutics to the epilepsies. Epilepsy and Behavior, 2021, 121, 106428.	0.9	6
399	Predicting Local Inversions Using Rectangle Clustering and Representative Rectangle Prediction. IEEE Transactions on Nanobioscience, 2019, 18, 316-323.	2.2	2
400	Atypical Prader-Willi and 15q13.3 Microdeletion Syndromes in a Patient with an Unbalanced Translocation. Cytogenetic and Genome Research, 2019, 158, 192-198.	0.6	0
401	Chromosomal Abnormalities and Cortical Malformations. , 2019, , 547-585.		0
402	Modeling and Predicting Developmental Trajectories of Neuropsychiatric Dimensions Associated With Copy Number Variations. International Journal of Neuropsychopharmacology, 2019, 22, 488-500.	1.0	19
403	First report of two successive deletions on chromosome 15q13 cytogenetic bands in a boy and girl: additional data to 15q13.3 syndrome with a report of high IQ patient. Molecular Cytogenetics, 2019, 12, 21.	0.4	2
404	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	5.8	636
405	Ion Channel Contributions to Wing Development in <i>Drosophila melanogaster</i> . G3: Genes, Genomes, Genetics, 2019, 9, 999-1008.	0.8	38
406	Homozygous 15q13.3 microdeletion in a child with hypotonia and impaired vision: A new report and review of the literature. Clinical Case Reports (discontinued), 2019, 7, 2311-2315.	0.2	7
407	Interactive effects between hemizygous 15q13.3 microdeletion and peripubertal stress on adult behavioral functions. Neuropsychopharmacology, 2019, 44, 703-710.	2.8	8
408	Central and peripheral immune responses to low-dose lipopolysaccharide in a mouse model of the 15q13.3 microdeletion. Cytokine, 2020, 126, 154879.	1.4	4
409	Regulation of aggressive behaviors by nicotinic acetylcholine receptors: Animal models, human genetics, and clinical studies. Neuropharmacology, 2020, 167, 107929.	2.0	14
410	Chromosome 15q13.3 microduplications are associated with treatment refractory major depressive disorder. Genes, Brain and Behavior, 2020, 19, e12628.	1.1	1
411	Rare copy number variants in individuals at clinical high risk for psychosis: Enrichment of synaptic/brain-related functional pathways. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 140-151.	1.1	0
412	Global developmental delay and intellectual disability. , 2020, , 269-281.		0

#	ARTICLE	IF	CITATIONS
413	Molecular, physiological and behavioral characterization of the heterozygous Df[h15q13]/+ mouse model associated with the human 15q13.3 microdeletion syndrome. <i>Brain Research</i> , 2020, 1746, 147024.	1.1	5
414	Genetic Landscape of Common Epilepsies: Advancing towards Precision in Treatment. <i>International Journal of Molecular Sciences</i> , 2020, 21, 7784.	1.8	51
415	Safety and tolerability of antipsychotic agents in neurodevelopmental disorders: a systematic review. <i>Expert Opinion on Drug Safety</i> , 2020, 19, 1419-1444.	1.0	19
416	Clinical and experimental insight into pathophysiology, comorbidity and therapy of absence seizures. <i>Brain</i> , 2020, 143, 2341-2368.	3.7	118
417	Common variants in FAN1, located in 15q13.3, confer risk for schizophrenia and bipolar disorder in Han Chinese. <i>Progress in Neuro-Psychopharmacology and Biological Psychiatry</i> , 2020, 103, 109973.	2.5	5
418	Population-Specific Genetic and Expression Differentiation in Europeans. <i>Genome Biology and Evolution</i> , 2020, 12, 358-369.	1.1	4
419	The sociability spectrum: evidence from reciprocal genetic copy number variations. <i>Molecular Autism</i> , 2020, 11, 50.	2.6	10
420	A Link between Genetic Disorders and Cellular Impairment, Using Human Induced Pluripotent Stem Cells to Reveal the Functional Consequences of Copy Number Variations in the Central Nervous System—A Close Look at Chromosome 15. <i>International Journal of Molecular Sciences</i> , 2020, 21, 1860.	1.8	4
421	Developmental disabilities, autism, and schizophrenia at a single locus. , 2020, , 201-221.		1
422	Chromosome 15q BP3 to BP5 deletion is a likely locus for speech delay and language impairment: Report on a four-member family and an unrelated boy. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1109.	0.6	3
423	An integrated analysis of rare CNV and exome variation in Autism Spectrum Disorder using the Infinium PsychArray. <i>Scientific Reports</i> , 2020, 10, 3198.	1.6	42
424	Regulation of NMDA glutamate receptor functions by the GluN2 subunits. <i>Journal of Neurochemistry</i> , 2020, 154, 121-143.	2.1	90
425	Report of the first patient with a homozygous <i>OTUD7A</i> variant responsible for epileptic encephalopathy and related proteasome dysfunction. <i>Clinical Genetics</i> , 2020, 97, 567-575.	1.0	18
426	Pharmacological Characterization of the Novel and Selective $\alpha 7$ Nicotinic Acetylcholine Receptor-Positive Allosteric Modulator BNC375. <i>Journal of Pharmacology and Experimental Therapeutics</i> , 2020, 373, 311-324.	1.3	9
427	Population prevalence and inheritance pattern of recurrent CNVs associated with neurodevelopmental disorders in 12,252 newborns and their parents. <i>European Journal of Human Genetics</i> , 2021, 29, 205-215.	1.4	40
428	Network Effects of the 15q13.3 Microdeletion on the Transcriptome and Epigenome in Human-Induced Neurons. <i>Biological Psychiatry</i> , 2021, 89, 497-509.	0.7	17
429	Df(h15q13)/+ Mouse Model Reveals Loss of Astrocytes and Synaptic-Related Changes of the Excitatory and Inhibitory Circuits in the Medial Prefrontal Cortex. <i>Cerebral Cortex</i> , 2021, 31, 1609-1621.	1.6	6
430	Mice with mutations in <i>Trpm1</i> , a gene in the locus of 15q13.3 microdeletion syndrome, display pronounced hyperactivity and decreased anxiety-like behavior. <i>Molecular Brain</i> , 2021, 14, 61.	1.3	4

#	ARTICLE	IF	CITATIONS
432	Spread of X-chromosome inactivation into autosomal regions in patients with unbalanced X-autosome translocations and its phenotypic effects. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2295-2305.	0.7	2
433	Human-Specific Genes, Cortical Progenitor Cells, and Microcephaly. <i>Cells</i> , 2021, 10, 1209.	1.8	23
434	Clinical evaluation of patients with a neuropsychiatric risk copy number variant. <i>Current Opinion in Genetics and Development</i> , 2021, 68, 26-34.	1.5	12
435	Sex-specific recombination patterns predict parent of origin for recurrent genomic disorders. <i>BMC Medical Genomics</i> , 2021, 14, 154.	0.7	2
437	The Phenotypic Spectrum of 15q13.3 Region Duplications: Report of 5 Patients. <i>Genes</i> , 2021, 12, 1025.	1.0	12
438	Detection of Morphological Abnormalities in Schizophrenia: An Important Step to Identify Associated Genetic Disorders or Etiologic Subtypes. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9464.	1.8	6
439	The spectrum of epilepsy in children with 15q13.3 microdeletion syndrome. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 92, 221-229.	0.9	9
440	Genomic regions associated with microdeletion/microduplication syndromes exhibit extreme diversity of structural variation. <i>Genetics</i> , 2021, 217, .	1.2	13
441	Consideration of the haplotype diversity at nonallelic homologous recombination hotspots improves the precision of rearrangement breakpoint identification. <i>Human Mutation</i> , 2017, 38, 1711-1722.	1.1	9
442	Nicotinic Acetylcholine Receptors in Autism Spectrum Disorders: Therapeutic Implications. , 2014, , 755-777.		5
443	Global Developmental Delay and Mental Retardation/Intellectual Disability. , 2012, , 554-574.		1
444	Deubiquitylases in developmental ubiquitin signaling and congenital diseases. <i>Cell Death and Differentiation</i> , 2021, 28, 538-556.	5.0	27
447	The genetics of intellectual disability: advancing technology and gene editing. <i>F1000Research</i> , 2020, 9, 22.	0.8	56
448	Genetic Variation Stimulated by Epigenetic Modification. <i>PLoS ONE</i> , 2008, 3, e4075.	1.1	9
449	De Novo Unbalanced Translocations in Prader-Willi and Angelman Syndrome Might Be the Reciprocal Product of inv dup(15)s. <i>PLoS ONE</i> , 2012, 7, e39180.	1.1	5
450	Parental Imbalances Involving Chromosomes 15q and 22q May Predispose to the Formation of De Novo Pathogenic Microdeletions and Microduplications in the Offspring. <i>PLoS ONE</i> , 2013, 8, e57910.	1.1	7
451	Altered Social Behaviours in Neurexin 1 α Knockout Mice Resemble Core Symptoms in Neurodevelopmental Disorders. <i>PLoS ONE</i> , 2013, 8, e67114.	1.1	149
452	Copy Number Variation Screen Identifies a Rare De Novo Deletion at Chromosome 15q13.1-13.3 in a Child with Language Impairment. <i>PLoS ONE</i> , 2015, 10, e0134997.	1.1	22

#	ARTICLE	IF	CITATIONS
453	Chromosome 15q BP4-BP5 Deletion in a Girl with Nocturnal Frontal Lobe Epilepsy, Migraine, Circumscribed Hypertrichosis, and Language Impairment. <i>Journal of Epilepsy Research</i> , 2020, 10, 84-91.	0.1	2
454	Genetics of bipolar disorder. <i>The Application of Clinical Genetics</i> , 2014, 7, 33.	1.4	92
455	Copy Number Variations in Adult-onset Neuropsychiatric Diseases. <i>Current Genomics</i> , 2018, 19, 420-430.	0.7	29
456	Alpha-7 Nicotinic Receptors in Nervous System Disorders: From Function to Therapeutic Perspectives. <i>Central Nervous System Agents in Medicinal Chemistry</i> , 2017, 17, 100-108.	0.5	29
457	Whole genome association studies in complex diseases: where do we stand?. <i>Dialogues in Clinical Neuroscience</i> , 2010, 12, 37-46.	1.8	33
458	Neuropsychiatric genomics in precision medicine: diagnostics, gene discovery, and translation. <i>Dialogues in Clinical Neuroscience</i> , 2016, 18, 237-252.	1.8	6
459	A case of isodicentric chromosome 15 presented with epilepsy and developmental delay. <i>Korean Journal of Pediatrics</i> , 2012, 55, 487.	1.9	2
460	Distinct disorders affecting the brain share common genetic origins. <i>F1000 Biology Reports</i> , 2010, 2, .	4.0	6
461	Advances in the genetics of schizophrenia: will high-risk copy number variants be useful in clinical genetics or diagnostics?. <i>F1000 Medicine Reports</i> , 2009, 1, .	2.9	3
463	The Importance of Genome Architecture in Mental Retardation. <i>Monographs in Human Genetics</i> , 2010, , 43-56.	0.5	1
466	Genetics of Sudden Death in Epilepsy. , 2010, , .		2
467	Genetic Evaluation in Developmental Disabilities. , 2011, , 69-78.		0
469	Array Comparative Genomic Hybridization in Cytogenetics and Molecular Genetics. , 2012, , 21-35.		0
470	Cytogenetic Testing and Chromosomal Disorders. , 2012, , 39-59.		0
471	Array-CGH and SNP-Arrays, the New Karyotype. , 2012, , 39-52.		0
472	Specific Chromosome Disorders in Newborns. , 2012, , 196-208.		1
473	Recurrent CNVs in the Etiology of Epigenetic Neurodevelopmental Disorders. , 2013, , 147-178.		0
477	Chromosomal Microarray in the New High-Throughput Technological and Bioinformatic Era. , 2019, , 61-84.		0

#	ARTICLE	IF	CITATIONS
478	Genomic Architecture of ASD. , 2019, , 23-34.		0
481	Array-CGH and SNP-Arrays, the New Karyotype. , 2012, , 39-52.		0
482	Functional outcomes of copy number variations of Chrna7 gene. , 2022, , 269-306.		0
485	Oral-Facial-Digital Syndrome Type 1: A Case Report and Review. Annals of Dermatology, 2022, 34, 132.	0.3	0
486	Genetics and Clinical Neuroscience in Intellectual Disability. Brain Sciences, 2022, 12, 338.	1.1	4
489	Bringing machine learning to research on intellectual and developmental disabilities: taking inspiration from neurological diseases. Journal of Neurodevelopmental Disorders, 2022, 14, 28.	1.5	9
490	Loss of function of OTUD7A in the schizophrenia- associated 15q13.3 deletion impairs synapse development and function in human neurons. American Journal of Human Genetics, 2022, 109, 1500-1519.	2.6	5
491	Peripheral immune challenges elicit differential up-regulation of hippocampal cytokine and chemokine mRNA expression in a mouse model of the 15q13.3 microdeletion syndrome. Cytokine, 2022, 159, 156005.	1.4	0
493	Gene editing hPSCs for modeling neurological disorders. , 2023, , 289-311.		0
495	Neurodevelopmental disordersâ€™high-resolution rethinking of disease modeling. Molecular Psychiatry, 2023, 28, 34-43.	4.1	9
496	Heterozygous Deletion of Chromosome 15q13.3 in a Boy with Developmental Regression, Global Developmental Delay, Hypotonia, and Short Stature. Pediatric Reports, 2022, 14, 528-532.	0.5	0
497	Impaired OTUD7A-dependent Ankyrin regulation mediates neuronal dysfunction in mouse and human models of the 15q13.3 microdeletion syndrome. Molecular Psychiatry, 0, , .	4.1	3
498	Deepening the understanding of CNVs on chromosome 15q11â€™13 by using hiPSCs: An overview. Frontiers in Cell and Developmental Biology, 0, 10, .	1.8	1
499	Evaluation of Individuals with Non-Syndromic Global Developmental Delay and Intellectual Disability. Children, 2023, 10, 414.	0.6	0
500	Nicotinic acetylcholine receptors and epilepsy. Pharmacological Research, 2023, 189, 106698.	3.1	7
502	Chromosome Disorders. , 2024, , 347-361.e2.		0