Sebastien Jacquemont

List of Publications by Year
in descending order

151 papers	11,938 citations	34016 52 h-index	29081 104 g-index
175 all docs	175 docs citations	175 times ranked	12786 citing authors

#	Article	IF	Citations
1	Effects of copy number variations on brain structure and risk for psychiatric illness: Largeâ€scale studies from the<scp>ENIGMA</scp>working groups on<scp>CNVs</scp>. Human Brain Mapping, 2022, 43, 300-328.	1.9	30
2	Deletion of Loss-of-Functionâ€“Intolerant Genes and Risk of 5 Psychiatric Disorders. JAMA Psychiatry, 2022, 79, 78.	6.0	8
3	Genes To Mental Health (G2MH): A Framework to Map the Combined Effects of Rare and Common Variants on Dimensions of Cognition and Psychopathology. American Journal of Psychiatry, 2022, 179, 189-203.	4.0	29
4	Copy Number Variant Risk Scores Associated With Cognition, Psychopathology, and Brain Structure in Youths in the Philadelphia Neurodevelopmental Cohort. JAMA Psychiatry, 2022, 79, 699.	6.0	8
5	Possible association of 16p11.2 copy number variation with altered lymphocyte and neutrophil counts. Npj Genomic Medicine, 2022, 7, .	1.7	3
6	Convergence and Divergence of Rare Genetic Disorders on Brain Phenotypes. JAMA Psychiatry, 2022, 79, 818.	6.0	12
7	Effect Sizes of Deletions and Duplications on Autism Risk Across the Genome. American Journal of Psychiatry, 2021, 178, 87-98.	4.0	50
8	A Genetics-First Approach to Dissecting the Heterogeneity of Autism: Phenotypic Comparison of Autism Risk Copy Number Variants. American Journal of Psychiatry, 2021, 178, 77-86.	4.0	62
9	The psychiatric phenotypes of 1q21 distal deletion and duplication. Translational Psychiatry, 2021, 11, 105.	2.4	6
10	Conditional canonical correlation estimation based on covariates with random forests. Bioinformatics, 2021, 37, 2714-2721.	1.8	7
11	1q21.1 distal copy number variants are associated with cerebral and cognitive alterations in humans. Translational Psychiatry, 2021, 11, 182.	2.4	24
12	Dissecting autism and schizophrenia through neuroimaging genomics. Brain, 2021, 144, 1943-1957.	3.7	37
13	Bayonet-shaped language development in autism with regression: a retrospective study. Molecular Autism, 2021, 12, 35.	2.6	12
14	Omnigenic Impact of Copy Number Variants on Cognition and Psychopathology in the Philadelphia Neurodevelopmental Cohort. Biological Psychiatry, 2021, 89, S320.	0.7	0
15	Analysis of Genomic Copy Number Variation Across Psychiatric Disorders. Biological Psychiatry, 2021, 89, S106-S107.	0.7	0
16	Assessing the Effects of Rare Copy Number Variants on Psychiatric Symptoms and Cognitive Ability in 1M Individuals. Biological Psychiatry, 2021, 89, S46.	0.7	0
17	The General Impact of Haploinsufficiency on Brain Connectivity Underlies the Pleiotropic Effect of Neuropsychiatric CNVS. Biological Psychiatry, 2021, 89, S40.	0.7	0
18	White Matter Diffusion MRI Findings in Carriers of 16p11.2 Copy Number Variants. Biological Psychiatry, 2021, 89, S40.	0.7	1

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19	ENIGMA-CNV and Other Initiatives to Understand the Impact of Rare Copy Number Variants on Brain Structure and Other Measures. Biological Psychiatry, 2021, 89, S41.	0.7	0
20	Lessons Learned From Neuroimaging Studies of Copy Number Variants: A Systematic Review. Biological Psychiatry, 2021, 90, 596-610.	0.7	22
21	Structural and functional brain alterations revealed by neuroimaging in CNV carriers. Current Opinion in Genetics and Development, 2021, 68, 88-98.	1.5	14
22	Effects of eight neuropsychiatric copy number variants on human brain structure. Translational Psychiatry, 2021, 11, 399.	2.4	18
23	Genome-wide analysis of gene dosage in 24,092 individuals estimates that 10,000 genes modulate cognitive ability. Molecular Psychiatry, 2021, 26, 2663-2676.	4.1	33
24	EEG Signal Complexity Is Reduced During Resting-State in Fragile X Syndrome. Frontiers in Psychiatry, 2021, 12, 716707.	1.3	5
25	Dose response of the 16p11.2 distal copy number variant on intracranial volume and basal ganglia. Molecular Psychiatry, 2020, 25, 584-602.	4.1	49
26	Cardiovascular risk scoring and magnetic resonance imaging detected subclinical cerebrovascular disease. European Heart Journal Cardiovascular Imaging, 2020, 21, 692-700.	0.5	11
27	Association of Copy Number Variation of the 15q11.2 BP1-BP2 Region With Cortical and Subcortical Morphology and Cognition. JAMA Psychiatry, 2020, 77, 420.	6.0	54
28	Mutations associated with neuropsychiatric conditions delineate functional brain connectivity dimensions contributing to autism and schizophrenia. Nature Communications, 2020, 11, 5272.	5.8	35
29	Genotypeâ€“phenotype correlation at codon 1740 of <scp>SETD2</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2037-2048.	0.7	14
30	Measuring and Estimating the Effect Size of Rare Non-Recurrent Deletions and Duplications on General Intelligence. Biological Psychiatry, 2020, 87, S196.	0.7	1
31	Estimating the effects of copyâ€number variants on intelligence using hierarchical Bayesian models. Genetic Epidemiology, 2020, 44, 825-840.	0.6	1
32	The effects of sex on prevalence and mechanisms underlying neurodevelopmental disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2020, 173, 327-339.	1.0	7
33	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	2.6	110
34	Functional Connectivity Analyses Suggest Shared Molecular Mechanisms Across 12 Neuropsychiatric Mutations, Autism and Schizophrenia. Biological Psychiatry, 2020, 87, S395.	0.7	2
35	Reduced Cognitive Assessment Scores Among Individuals With Magnetic Resonance Imagingâ€“Detected Vascular Brain Injury. Stroke, 2020, 51, 1158-1165.	1.0	9
36	Estimating the Effect Size of CNVs on Risk for Major Psychopathology, the CAMP Project. Biological Psychiatry, 2020, 87, S392.	0.7	0

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37	Spatial control of nucleoporin condensation by fragile Xâ€related proteins. EMBO Journal, 2020, 39, e104467.	3.5	21
38	QUANTIFYING THE EFFECT OF COPY-NUMBER VARIANTS ON GENERAL INTELLIGENCE IN UNSELECTED POPULATIONS. European Neuropsychopharmacology, 2019, 29, S848.	0.3	0
39	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. NeuroImage, 2019, 203, 116155.	2.1	9
40	5 DIFFERENCES IN THE GENETIC BACKGROUND CONTRIBUTE TO RISK AND RESILIENCE TO AUTISM. European Neuropsychopharmacology, 2019, 29, S61.	0.3	0
41	250. Gene Dosage Effects on Neurobehavioral PhenotypesÂand Development: Relevance to Idiopathic Neuropsychiatric Disorders. Biological Psychiatry, 2019, 85, S104.	0.7	1
42	40 MAPPING THE EFFECT-SIZE OF GENE DOSAGE ON GENERAL INTELLIGENCE ACROSS THE GENOME. European Neuropsychopharmacology, 2019, 29, S81.	0.3	0
43	A framework for the investigation of rare genetic disorders in neuropsychiatry. Nature Medicine, 2019, 25, 1477-1487.	15.2	90
44	Psychiatric disorders in children with 16p11.2 deletion and duplication. Translational Psychiatry, 2019, 9, 8.	2.4	93
45	191. Mirror Effects of 4 Neurodevelopmental CNVs on Functional Connectivity and Implication for Idiopathic Autism. Biological Psychiatry, 2019, 85, S79.	0.7	0
46	Deleterious de novo variants of Xâ€linked ZC4H2 in females cause a variable phenotype with neurogenic arthrogryposis multiplex congenita. Human Mutation, 2019, 40, 2270-2285.	1.1	29
47	Tinkering with the vasopressin pathway in autism. Science Translational Medicine, 2019, 11, .	5.8	2
48	A GENETIC FIRST APPROACH TO DISSECTING THE HETEROGENEITY OF AUTISM: PHENOTYPIC COMPARISON OF AUTISM RISK COPY NUMBER VARIANTS. European Neuropsychopharmacology, 2019, 29, S783-S784.	0.3	6
49	Poster Withdrawn: QUANTIFYING THE EFFECTS OF 16P11.2 CNVs ON BRAIN STRUCTURE, A MULTI-SITE â€˜GENETIC-FIRSTâ€™MRI STUDY. European Neuropsychopharmacology, 2019, 29, S859-S860.	0.3	1
50	Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710.	1.5	43
51	251. Measuring and Estimating the Effects of Rare Variants, Genome-Wide, on Cognition. Biological Psychiatry, 2019, 85, S104.	0.7	0
52	M59 SHARED FUNCTIONAL CONNECTIVITY ALTERATIONS ACROSS NEURODEVELOPMENTAL MUTATIONS, AUTISM AND SCHIZOPHRENIA. European Neuropsychopharmacology, 2019, 29, S197-S198.	0.3	0
53	Duplication of 10q24 locus: broadening the clinical and radiological spectrum. European Journal of Human Genetics, 2019, 27, 525-534.	1.4	13
54	Clinimetric Properties of the Fragile Xâ€associated Tremor Ataxia Syndrome Rating Scale. Movement Disorders Clinical Practice, 2019, 6, 120-124.	0.8	12

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55	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	0.7	56
56	Protein synthesis levels are increased in a subset of individuals with fragile X syndrome. Human Molecular Genetics, 2018, 27, 2039-2051.	1.4	51
57	Measuring and Estimating the Effect Sizes of Copy Number Variants on General Intelligence in Community-Based Samples. JAMA Psychiatry, 2018, 75, 447.	6.0	77
58	Drug development for neurodevelopmental disorders: lessons learned from fragile X syndrome. Nature Reviews Drug Discovery, 2018, 17, 280-299.	21.5	247
59	Mavoglurant in Fragile X Syndrome: Results of two open-label, extension trials in adults and adolescents. Scientific Reports, 2018, 8, 16970.	1.6	33
60	Auditory repetition suppression alterations in relation to cognitive functioning in fragile X syndrome: a combined EEG and machine learning approach. Journal of Neurodevelopmental Disorders, 2018, 10, 4.	1.5	24
61	The Canadian Partnership for Tomorrow Project: a pan-Canadian platform for research on chronic disease prevention. Cmaj, 2018, 190, E710-E717.	0.9	71
62	In-vivo probabilistic atlas of human thalamic nuclei based on diffusion- weighted magnetic resonance imaging. Scientific Data, 2018, 5, 180270.	2.4	67
63	Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes. Molecular Psychiatry, 2017, 22, 836-849.	4.1	68
64	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
65	Sex chromosome aneuploidies and copy-number variants: a further explanation for neurodevelopmental prognosis variability?. European Journal of Human Genetics, 2017, 25, 930-934.	1.4	19
66	Altered visual repetition suppression in Fragile X Syndrome: New evidence from ERPs and oscillatory activity. International Journal of Developmental Neuroscience, 2017, 59, 52-59.	0.7	23
67	Robust thalamic nuclei segmentation method based on local diffusion magnetic resonance properties. Brain Structure and Function, 2017, 222, 2203-2216.	1.2	58
68	CNV-association meta-analysis in 191,161 European adults reveals new loci associated with anthropometric traits. Nature Communications, 2017, 8, 744.	5.8	64
69	The Immune Signaling Adaptor LAT Contributes to the Neuroanatomical Phenotype of 16p11.2 BP2-BP3 CNVs. American Journal of Human Genetics, 2017, 101, 564-577.	2.6	30
70	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	0.7	40
71	Reflections on Clinical Trials in Fragile X Syndrome. , 2017, , 419-441.		1
72	Complete Maxillo-Mandibular Syngnathia in a Newborn with Multiple Congenital Malformations. Pediatrics and Neonatology, 2016, 57, 65-68.	0.3	19

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73	Reciprocal changes in DNA methylation and hydroxymethylation and a broad repressive epigenetic switch characterize FMR1 transcriptional silencing in fragile X syndrome. Clinical Epigenetics, 2016, 8, 15.	1.8	18
74	Rationale, design, and methods for Canadian alliance for healthy hearts and minds cohort study (CAHHM) â€“ a Pan Canadian cohort study. BMC Public Health, 2016, 16, 650.	1.2	31
75	Reporting incidental findings of genomic disorder-associated copy number variants to unselected biobank participants. Personalized Medicine, 2016, 13, 303-314.	0.8	12
76	The Number of Genomic Copies at the 16p11.2 Locus Modulates Language, Verbal Memory, and Inhibition. Biological Psychiatry, 2016, 80, 129-139.	0.7	78
77	Mavoglurant in fragile X syndrome: Results of two randomized, double-blind, placebo-controlled trials. Science Translational Medicine, 2016, 8, 321ra5.	5.8	210
78	16p11.2 Locus modulates response to satiety before the onset of obesity. International Journal of Obesity, 2016, 40, 870-876.	1.6	31
79	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	6.0	195
80	Dysregulated ADAM10-Mediated Processing of APP during a Critical Time Window Leads to SynapticDeficits in Fragile X Syndrome. Neuron, 2015, 87, 908.	3.8	0
81	Earlyâ€onset encephalopathy with epilepsy associated with a novel splice site mutation in SMC1A. American Journal of Medical Genetics, Part A, 2015, 167, 3076-3081.	0.7	22
82	Copy Number Variations and Cognitive Phenotypes in Unselected Populations. Obstetrical and Gynecological Survey, 2015, 70, 559-560.	0.2	2
83	Copy Number Variations and Cognitive Phenotypes in Unselected Populations. JAMA - Journal of the American Medical Association, 2015, 313, 2044.	3.8	143
84	The 16p11.2 locus modulates brain structures common to autism, schizophrenia and obesity. Molecular Psychiatry, 2015, 20, 140-147.	4.1	160
85	Sex differences in brain plasticity: a new hypothesis for sex ratio bias in autism. Molecular Autism, 2015, 6, 33.	2.6	70
86	Dysregulated ADAM10-Mediated Processing of APP during a Critical Time Window Leads to Synaptic Deficits in Fragile X Syndrome. Neuron, 2015, 87, 382-398.	3.8	59
87	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	2.6	53
88	Development of mavoglurant and its potential for the treatment of fragile X syndrome. Expert Opinion on Investigational Drugs, 2014, 23, 125-134.	1.9	24
89	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
90	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	1.4	38

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91	The challenges of clinical trials in fragile X syndrome. Psychopharmacology, 2014, 231, 1237-1250.	1.5	98
92	A Higher Mutational Burden in Females Supports a â€œFemale Protective Modelâ€•in Neurodevelopmental Disorders. American Journal of Human Genetics, 2014, 94, 415-425.	2.6	457
93	Investigation of memory, executive functions, and anatomic correlates in asymptomatic FMR1 premutation carriers. Neurobiology of Aging, 2014, 35, 1939-1946.	1.5	20
94	Molecular characterization of 39 de novo sSMC : contribution to prognosis and genetic counselling, a prospective study. Clinical Genetics, 2014, 85, 233-244.	1.0	24
95	Translating Molecular Advances in Fragile X Syndrome Into Therapy. Journal of Clinical Psychiatry, 2014, 75, e294-e307.	1.1	46
96	Transmission Disequilibrium of Small CNVs in Simplex Autism. American Journal of Human Genetics, 2013, 93, 595-606.	2.6	87
97	SCRIB and PUF60 Are Primary Drivers of the Multisystemic Phenotypes of the 8q24.3 Copy-Number Variant. American Journal of Human Genetics, 2013, 93, 798-811.	2.6	82
98	Brain structure in asymptomatic FMR1 premutation carriers at risk for fragile X-associated tremor/ataxia syndrome. Neurobiology of Aging, 2013, 34, 1700-1707.	1.5	52
99	The Growing Importance of CNVs: New Insights for Detection and Clinical Interpretation. Frontiers in Genetics, 2013, 4, 92.	1.1	49
100	Rare Genomic Structural Variants in Complex Disease: Lessons from the Replication of Associations with Obesity. PLoS ONE, 2013, 8, e58048.	1.1	33
101	A 600â€…kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. Journal of Medical Genetics, 2012, 49, 660-668.	1.5	251
102	Mutations at a single codon in Mad homology 2 domain of SMAD4 cause Myhre syndrome. Nature Genetics, 2012, 44, 85-88.	9.4	125
103	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. Nature, 2012, 485, 363-367.	13.7	363
104	Carriers of the fragile X mental retardation 1 (FMR1) premutation allele present with increased levels of cytokine IL-10. Journal of Neuroinflammation, 2012, 9, 238.	3.1	20
105	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	9.4	234
106	Neuropathological, clinical and molecular pathology in female fragile X premutation carriers with and without FXTAS. Genes, Brain and Behavior, 2012, 11, 577-585.	1.1	117
107	Disease-associated mutations in the actin-binding domain of filamin B cause cytoplasmic focal accumulations correlating with disease severity. Human Mutation, 2012, 33, 665-673.	1.1	31
108	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394

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109	Epigenetic Modification of the FMR1 Gene in Fragile X Syndrome Is Associated with Differential Response to the mGluR5 Antagonist AFQ056. Science Translational Medicine, 2011, 3, 64ra1.	5.8	344
110	Eosinophilic infiltration related to CAPN3 mutations: a pathophysiological component of primary calpainopathy?. Clinical Genetics, 2011, 80, 398-402.	1.0	22
111	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. Nature Genetics, 2011, 43, 306-308.	9.4	181
112	Mutations in CEP57 cause mosaic variegated aneuploidy syndrome. Nature Genetics, 2011, 43, 527-529.	9.4	117
113	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	1.4	63
114	Clinical utility gene card for: fragile X mental retardation syndrome, fragile X-associated tremor/ataxia syndrome and fragile X-associated primary ovarian insufficiency. European Journal of Human Genetics, 2011, 19, 1017-1017.	1.4	12
115	Familial aplasia of the trapezius muscle: clinical and MRI findings. Acta Paediatrica, International Journal of Paediatrics, 2011, 100, 464-466.	0.7	4
116	A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. Nature, 2010, 463, 671-675.	13.7	476
117	A Comparative Phenotypic Study of Kallmann Syndrome Patients Carrying Monoallelic and Biallelic Mutations in the Prokineticin 2 or Prokineticin Receptor 2 Genes. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 659-669.	1.8	124
118	Mitochondrial tRNA^{Leu(UUR)} mutation m.3302Aâ€‰>â€‰G presenting as childhoodâ€onset severe myopathy: threshold determination through segregation study. Journal of Inherited Metabolic Disease, 2010, 33, 219-226.	1.7	10
119	Phenotypic and molecular characterization of a novel case of dyssegmental dysplasia, Silverman-Handmaker type. European Journal of Medical Genetics, 2010, 53, 294-298.	0.7	10
120	Preimplantation genetic diagnosis (PGD) for HLA typing: bases for setting up an open international collaboration when PGD is not available. Fertility and Sterility, 2010, 94, 1129-1131.	0.5	3
121	The Epidemiology of FXTAS. , 2010, , 17-30.		6
122	The Efficacy of Melatonin for Sleep Problems in Children with Autism, Fragile X Syndrome, or Autism and Fragile X Syndrome. Journal of Clinical Sleep Medicine, 2009, 05, 145-150.	1.4	149
123	Penetrance of marked cognitive impairment in older male carriers of the FMR1 gene premutation. Journal of Medical Genetics, 2009, 46, 818-824.	1.5	61
124	Early neurological impairment and severe anemia in a newborn with Pearson syndrome. European Journal of Pediatrics, 2009, 168, 311-315.	1.3	14
125	Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. European Journal of Medical Genetics, 2009, 52, 94-100.	0.7	157
126	Lifespan changes in working memory in fragile X premutation males. Brain and Cognition, 2009, 69, 551-558.	0.8	93

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127	The efficacy of melatonin for sleep problems in children with autism, fragile X syndrome, or autism and fragile X syndrome. Journal of Clinical Sleep Medicine, 2009, 5, 145-50.	1.4	82
128	Biallelic mutations in the prokineticin-2 gene in two sporadic cases of Kallmann syndrome. European Journal of Human Genetics, 2008, 16, 865-868.	1.4	44
129	Age-dependent cognitive changes in carriers of the fragile X syndrome. Cortex, 2008, 44, 628-636.	1.1	133
130	Progression of tremor and ataxia in male carriers of theFMR1 premutation. Movement Disorders, 2007, 22, 203-206.	2.2	134
131	Impairment of executive cognitive functioning in males with fragile X-associated tremor/ataxia syndrome. Movement Disorders, 2007, 22, 645-650.	2.2	84
132	Fragile Xâ€associated tremor/ataxia syndrome: Clinical features, genetics, and testing guidelines. Movement Disorders, 2007, 22, 2018-2030.	2.2	306
133	Fragile-X syndrome and fragile X-associated tremor/ataxia syndrome: two faces of FMR1. Lancet Neurology, The, 2007, 6, 45-55.	4.9	212
134	Impairment in the cognitive functioning of men with fragile X-associated tremor/ataxia syndrome (FXTAS). Journal of the Neurological Sciences, 2006, 248, 227-233.	0.3	126
135	Cutaneous lipoma in children: 5 cases with Bannayan-Riley-Ruvalcaba syndrome. Journal of Pediatric Surgery, 2006, 41, 1601-1603.	0.8	32
136	Cognitive Impairment in a 65-year-old Male With the Fragile X-associated Tremor-Ataxia Syndrome (FXTAS). Cognitive and Behavioral Neurology, 2006, 19, 165-171.	0.5	43
137	Clinical and molecular genetic features of ARC syndrome. Human Genetics, 2006, 120, 396-409.	1.8	118
138	Prevalence of FMR1 Repeat Expansions in Movement Disorders. Neuroepidemiology, 2006, 26, 151-155.	1.1	37
139	Size bias of fragile X premutation alleles in late-onset movement disorders. Journal of Medical Genetics, 2006, 43, 804-809.	1.5	99
140	Screening for FXTAS. European Journal of Human Genetics, 2005, 13, 2-3.	1.4	15
141	Detection of genomic imbalances by array based comparative genomic hybridisation in fetuses with multiple malformations. Journal of Medical Genetics, 2005, 42, 121-128.	1.5	174
142	Spastic paraparesis, cerebellar ataxia, and intention tremor: a severe variant of FXTAS?. Journal of Medical Genetics, 2005, 42, e14-e14.	1.5	55
143	Penetrance of the Fragile Xâ€“Associated Tremor/Ataxia Syndrome in a Premutation Carrier Population. JAMA - Journal of the American Medical Association, 2004, 291, 460.	3.8	571
144	Inherited ring chromosome 8Âwithout loss of subtelomeric sequences. Annales De GÃ©nÃ©tique, 2004, 47, 289-296.	0.4	25

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145	Screen for expandedFMR1 alleles in patients with essential tremor. Movement Disorders, 2004, 19, 930-933.	2.2	63
146	Fragile-Xâ€“Associated Tremor/Ataxia Syndrome (FXTAS) in Females with the FMR1 Premutation. American Journal of Human Genetics, 2004, 74, 1051-1056.	2.6	320
147	Fragile X Premutation Tremor/Ataxia Syndrome: Molecular, Clinical, and Neuroimaging Correlates. American Journal of Human Genetics, 2003, 72, 869-878.	2.6	720
148	The Fragile X Premutation Presenting as Essential Tremor. Archives of Neurology, 2003, 60, 117.	4.9	162
149	Neuronal intranuclear inclusions in a new cerebellar tremor/ataxia syndrome among fragile X carriers. Brain, 2002, 125, 1760-1771.	3.7	474
150	Deletion of the SIM1 gene (6q16.2) in a patient with a Prader-Willi-like phenotype. Journal of Medical Genetics, 2002, 39, 594-596.	1.5	105
151	Fragile X premutation carriers: characteristic MR imaging findings of adult male patients with progressive cerebellar and cognitive dysfunction. American Journal of Neuroradiology, 2002, 23, 1757-66.	1.2	272