Frank R Kooy

List of Publications by Year
in descending order

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192 papers	13,499 citations	31949 53 h-index	28275 105 g-index
269 all docs	269 docs citations	269 times ranked	16215 citing authors

#	Article	IF	Citations
1	ZNF711 puts a spell on DNA. European Journal of Human Genetics, 2022, , .	1.4	0
2	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	1.5	13
3	Protein interaction network analysis reveals genetic enrichment of immune system genes in frontotemporal dementia. Neurobiology of Aging, 2022, 116, 67-79.	1.5	2
4	Expanding the Phenotype of B3GALNT2-Related Disorders. Genes, 2022, 13, 694.	1.0	3
5	Towards Kinase Inhibitor Therapies for Fragile X Syndrome: Tweaking Twists in the Autism Spectrum Kinase Signaling Network. Cells, 2022, 11, 1325.	1.8	13
6	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	1.1	13
7	Discovery of autism/intellectual disability somatic mutations in Alzheimer's brains: mutated ADNP cytoskeletal impairments and repair as a case study. Molecular Psychiatry, 2021, 26, 1619-1633.	4.1	60
8	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
9	The relevance of deep genomic analyses in families with variably expressive CNVs in the era of personalized medicine. Molecular Genetics and Metabolism, 2021, 132, S69.	0.5	0
10	Abundancy of polymorphic CGG repeats in the human genome suggest a broad involvement in neurological disease. Scientific Reports, 2021, 11, 2515.	1.6	25
11	Single-Cell and Neuronal Network Alterations in an In Vitro Model of Fragile X Syndrome. Cerebral Cortex, 2020, 30, 31-46.	1.6	8
12	A contemporary view on the molecular basis of neurodevelopmental disorders. , 2020, , 57-78.		0
13	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	1.8	40
14	GABAergic abnormalities in the fragile X syndrome. European Journal of Paediatric Neurology, 2020, 24, 100-104.	0.7	20
15	PUM1 haploinsufficiency is associated with syndromic neurodevelopmental delay and epilepsy. American Journal of Medical Genetics, Part A, 2020, 182, 591-594.	0.7	1
16	A Survey of Rare Epigenetic Variation in 23,116 Human Genomes Identifies Disease-Relevant Epivariations and CGG Expansions. American Journal of Human Genetics, 2020, 107, 654-669.	2.6	40
17	Tauopathy in the young autistic brain: novel biomarker and therapeutic target. Translational Psychiatry, 2020, 10, 228.	2.4	57
18	Overrepresentation of genetic variation in the AnkyrinG interactome is related to a range of neurodevelopmental disorders. European Journal of Human Genetics, 2020, 28, 1726-1733.	1.4	4

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19	High-throughput screening identifies histone deacetylase inhibitors that modulate GTF2I expression in 7q11.23 microduplication autism spectrum disorder patient-derived cortical neurons. Molecular Autism, 2020, 11, 88.	2.6	20
20	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
21	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
22	Reduced serum levels of pro-inflammatory chemokines in fragile X syndrome. BMC Neurology, 2020, 20, 138.	0.8	10
23	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
24	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508.	2.6	48
25	The translational regulator FMRP controls lipid and glucose metabolism in mice and humans. Molecular Metabolism, 2019, 21, 22-35.	3.0	39
26	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	1.4	32
27	Genetic variants in the KDM6B gene are associated with neurodevelopmental delays and dysmorphic features. American Journal of Medical Genetics, Part A, 2019, 179, 1276-1286.	0.7	38
28	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
29	Novel BRPF1 mutation in a boy with intellectual disability, coloboma, facial nerve palsy and hypoplasia of the corpus callosum. European Journal of Medical Genetics, 2019, 62, 103691.	0.7	15
30	Phenotypic and biochemical analysis of an international cohort of individuals with variants in NAA10 and NAA15. Human Molecular Genetics, 2019, 28, 2900-2919.	1.4	46
31	Gaps in Current Autism Research: The Thoughts of the Autism Research Editorial Board and Associate Editors. Autism Research, 2019, 12, 700-714.	2.1	28
32	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
33	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
34	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	1.1	127
35	A mouse model for intellectual disability caused by mutations in the X-linked 2â€²â€²Oâ€²methyltransferase Ftsj1 gene. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2083-2093.	1.8	17
36	Association of hereditary angioedema type 1 with developmental anomalies due to a large and unusual de novo pericentromeric rearrangement of chromosome 11 spanning the entire C1 inhibitor gene (SERPING1). Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 1352-1354.e3.	2.0	1

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37	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
38	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	2.6	59
39	A higher rare CNV burden in the genetic background potentially contributes to intellectual disability phenotypes in 22q11.2 deletion syndrome. European Journal of Medical Genetics, 2018, 61, 209-212.	0.7	17
40	Mutations in ADNP affect expression and subcellular localization of the protein. Cell Cycle, 2018, 17, 1068-1075.	1.3	21
41	Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes. Molecular Psychiatry, 2017, 22, 836-849.	4.1	68
42	Impaired GABAergic inhibition in the hippocampus of Fmr1 knockout mice. Neuropharmacology, 2017, 116, 71-81.	2.0	58
43	Premature primary tooth eruption in cognitive/motor-delayed ADNP-mutated children. Translational Psychiatry, 2017, 7, e1043-e1043.	2.4	55
44	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	9.4	443
45	Behavioural characterization of AnkyrinG deficient mice, a model for ANK3 related disorders. Behavioural Brain Research, 2017, 328, 218-226.	1.2	16
46	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
47	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	1.0	26
48	Fragile X syndrome. Nature Reviews Disease Primers, 2017, 3, 17065.	18.1	490
49	Dominant variants in the splicing factor PUF60 cause a recognizable syndrome with intellectual disability, heart defects and short stature. European Journal of Human Genetics, 2017, 25, 43-51.	1.4	44
50	The GABAergic System Contributions to the Fragile X Syndrome Phenotype. , 2017, , 205-215.		2
51	The Eight and a Half Year Journey of Undiagnosed AD: Gene Sequencing and Funding of Advanced Genetic Testing Has Led to Hope and New Beginnings. Frontiers in Endocrinology, 2017, 8, 107.	1.5	35
52	A randomized double-blind, placebo-controlled trial of ganaxolone in children and adolescents with fragile X syndrome. Journal of Neurodevelopmental Disorders, 2017, 9, 26.	1.5	67
53	Animal Models of Fragile X Syndrome. , 2017, , 123-147.		3
54	A detailed clinical analysis of 13 patients with AUTS2 syndrome further delineates the phenotypic spectrum and underscores the behavioural phenotype. Journal of Medical Genetics, 2016, 53, 523-532.	1.5	51

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55	PLCB1 epileptic encephalopathies; Review and expansion of the phenotypic spectrum. European Journal of Paediatric Neurology, 2016, 20, 474-479.	0.7	14
56	Whole genome sequencing of a dizygotic twin suggests a role for the serotonin receptor HTR7 in autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1049-1056.	1.1	8
57	Novel microdeletions on chromosome 14q32.2 suggest a potential role for non-coding RNAs in Kagami-Ogata syndrome. European Journal of Human Genetics, 2016, 24, 1724-1729.	1.4	27
58	Thoracic dimples and dysmorphic features associated with a partial duplication and triplication of chromosome 12q24. Clinical Dysmorphology, 2016, 25, 167-173.	0.1	0
59	Clinical delineation of the PACS1â€related syndromeâ€"Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	0.7	44
60	Multiplexed High Resolution Melting Assay for Versatile Sample Tracking in a Diagnostic and Research Setting. Journal of Molecular Diagnostics, 2016, 18, 32-38.	1.2	4
61	Severe congenital neutropenia with neurological impairment due to a homozygous VPS45 p.E238K mutation: A case report suggesting a genotypeâ€"phenotype correlation. American Journal of Medical Genetics, Part A, 2015, 167, 3214-3218.	0.7	17
62	A Robust Protocol to Increase NimbleGen SeqCap EZ Multiplexing Capacity to 96 Samples. PLoS ONE, 2015, 10, e0123872.	1.1	13
63	The GABAA Receptor as a Therapeutic Target for Neurodevelopmental Disorders. Neuron, 2015, 86, 1119-1130.	3.8	261
64	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	2.6	230
65	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
66	The GABA_A receptor is an FMRP target with therapeutic potential in fragile X syndrome. Cell Cycle, 2015, 14, 2985-2995.	1.3	87
67	The Compassionate Side of Neuroscience: Tony Sermoneâ€™s Undiagnosed Genetic Journeyâ€"ADNP Mutation. Journal of Molecular Neuroscience, 2015, 56, 751-757.	1.1	37
68	Five patients with a chromosome 1q21.1 triplication show macrocephaly, increased weight and facial similarities. European Journal of Medical Genetics, 2015, 58, 503-508.	0.7	15
69	Two male adults with pathogenic AUTS2 variants, including a two-base pair deletion, further delineate the AUTS2 syndrome. European Journal of Human Genetics, 2015, 23, 803-807.	1.4	28
70	Novel IL1RAPL1 mutations associated with intellectual disability impair synaptogenesis. Human Molecular Genetics, 2015, 24, 1106-1118.	1.4	31
71	Insights into GABAAergic system deficits in fragile X syndrome lead to clinical trials. Neuropharmacology, 2015, 88, 48-54.	2.0	71
72	Challenges and opportunities in the investigation of unexplained intellectual disability using familyâ€based wholeâ€exome sequencing. Clinical Genetics, 2015, 88, 140-148.	1.0	25

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73	Positron Emission Tomography (PET) Quantification of GABAA Receptors in the Brain of Fragile X Patients. PLoS ONE, 2015, 10, e0131486.	1.1	52
74	VariantDB: a flexible annotation and filtering portal for next generation sequencing data. Genome Medicine, 2014, 6, 74.	3.6	60
75	FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. PLoS Genetics, 2014, 10, e1004242.	1.5	41
76	The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 315-326.	0.7	68
77	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. European Journal of Medical Genetics, 2014, 57, 151-156.	0.7	91
78	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	9.4	293
79	Mouse models of the fragile X premutation and fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 25.	1.5	57
80	Fragile X syndrome neurobiology translates into rational therapy. Drug Discovery Today, 2014, 19, 510-519.	3.2	31
81	A CGG-Repeat Expansion Mutation inZNF713Causes FRA7A: Association with Autistic Spectrum Disorder in two Families. Human Mutation, 2014, 35, n/a-n/a.	1.1	28
82	Craniofacial characteristics of fragile X syndrome in mouse and man. European Journal of Human Genetics, 2013, 21, 816-823.	1.4	43
83	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	1.4	137
84	Detection and interpretation of genomic structural variation in health and disease. Expert Review of Molecular Diagnostics, 2013, 13, 61-82.	1.5	13
85	Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy. Human Molecular Genetics, 2013, 22, 2590-2602.	1.4	103
86	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
87	Pharmacological treatment of fragile X syndrome with GABAergic drugs in a knockout mouse model. Behavioural Brain Research, 2012, 229, 244-249.	1.2	109
88	Haploinsufficiency of <scp>CMIP</scp> in a Girl With Autism Spectrum Disorder and Developmental Delay due to a De Novo Deletion on Chromosome 16q23.2. Autism Research, 2012, 5, 277-281.	2.1	19
89	The Contribution of CLIP2 Haploinsufficiency to the Clinical Manifestations of the Williams-Beuren Syndrome. American Journal of Human Genetics, 2012, 90, 1071-1078.	2.6	41
90	A de novo balanced t(2;6)(p15;p22.3) in a patient with West Syndrome disrupts a lnc-RNA. Epilepsy Research, 2012, 99, 346-349.	0.8	9

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91	On the spot: very local chromosomal rearrangements. F1000 Biology Reports, 2012, 4, 22.	4.0	2
92	Metabonomics adds a new dimension to fragile Ã— syndrome. Genome Medicine, 2011, 3, 80.	3.6	2
93	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394
94	Fragile X syndrome: from gene discovery to therapy. Frontiers in Bioscience - Landmark, 2011, 16, 1211.	3.0	30
95	CNV-WebStore: Online CNV Analysis, Storage and Interpretation. BMC Bioinformatics, 2011, 12, 4.	1.2	54
96	Arrayâ€based MLPA to detect recurrent copy number variations in patients with idiopathic mental retardation. American Journal of Medical Genetics, Part A, 2011, 155, 343-348.	0.7	12
97	Advances in understanding fragile X syndrome and related disorders. Current Opinion in Pediatrics, 2011, 23, 601-606.	1.0	20
98	A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. Nature, 2010, 463, 671-675.	13.7	476
99	Involvement and Therapeutic Potential of the GABAergic System in the Fragile X Syndrome. Scientific World Journal, The, 2010, 10, 2198-2206.	0.8	17
100	Genetic Overlaps in Mental Retardation, Autism and Schizophrenia. Monographs in Human Genetics, 2010, , 126-136.	0.5	3
101	A boy with mental retardation, obesity and hypertrichosis caused by a microdeletion of 19p13.12. European Journal of Medical Genetics, 2010, 53, 291-293.	0.7	23
102	Further molecular and clinical delineation of co-locating 17p13.3 microdeletions and microduplications that show distinctive phenotypes. Journal of Medical Genetics, 2010, 47, 299-311.	1.5	137
103	Distinct disorders affecting the brain share common genetic origins. F1000 Biology Reports, 2010, 2, .	4.0	6
104	The complexity of the GABAA receptor shapes unique pharmacological profiles. Drug Discovery Today, 2009, 14, 866-875.	3.2	165
105	Expression of the GABAergic system in animal models for fragile X syndrome and fragile X associated tremor/ataxia syndrome (FXTAS). Brain Research, 2009, 1253, 176-183.	1.1	153
106	Balanced translocations in mental retardation. Human Genetics, 2009, 126, 133-147.	1.8	27
107	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
108	Fragile X syndrome: from molecular genetics to therapy. Journal of Medical Genetics, 2009, 46, 577-584.	1.5	83

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109	Germline mutation of microRNA-125a is associated with breast cancer. Journal of Medical Genetics, 2009, 46, 358-360.	1.5	94
110	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. Journal of Medical Genetics, 2009, 46, 511-523.	1.5	250
111	GABA neurotransmitter signaling in the developing mouse lens: Dynamic regulation of components and functionality. Developmental Dynamics, 2008, 237, 3830-3841.	0.8	15
112	Identification of non-recurrent submicroscopic genome imbalances: the advantage of genome-wide microarrays over targeted approaches. European Journal of Human Genetics, 2008, 16, 395-400.	1.4	14
113	Clinical and molecular characteristics of 1qter microdeletion syndrome: delineating a critical region for corpus callosum agenesis/hypogenesis. Journal of Medical Genetics, 2008, 45, 346-354.	1.5	87
114	Effect of genetic background on acoustic startle response in fragile X knockout mice. Genetical Research, 2008, 90, 341-345.	0.3	24
115	FRA18C: a new aphidicolin-inducible fragile site on chromosome 18q22, possibly associated with in vivo chromosome breakage. Journal of Medical Genetics, 2007, 44, 347-352.	1.5	24
116	Fragile sites and human disease. Human Molecular Genetics, 2007, 16, R150-R158.	1.4	94
117	A de novo subtelomeric monosomy 11q (11q24.2-qter) and trisomy 20q (20q13.3-qter) in a girl with findings compatible with Jacobsen syndrome: case report and review. Clinical Dysmorphology, 2007, 16, 231-239.	0.1	7
118	The molecular basis of the folate-sensitive fragile site FRA11A at 11q13. Cytogenetic and Genome Research, 2007, 119, 9-14.	0.6	25
119	The GABAA receptor: a novel target for treatment of fragile X?. Trends in Neurosciences, 2007, 30, 425-431.	4.2	157
120	CGG-Repeat Expansion in the DIP2B Gene Is Associated with the Fragile Site FRA12A on Chromosome 12q13.1. American Journal of Human Genetics, 2007, 80, 221-231.	2.6	92
121	The Reduced Expression of the HADH2 Protein Causes X-Linked Mental Retardation, Choreoathetosis, and Abnormal Behavior. American Journal of Human Genetics, 2007, 80, 372-377.	2.6	57
122	Dandy-Walker complex in a boy with a 5 Mb deletion of region 1q44 due to a paternal t(1;20)(q44;q13.33). American Journal of Medical Genetics, Part A, 2007, 143A, 1038-1044.	0.7	49
123	Beckwithâ€“Wiedemannâ€like macroglossia and 18q23 haploinsufficiency. American Journal of Medical Genetics, Part A, 2007, 143A, 2796-2803.	0.7	8
124	Diverse chromosome breakage mechanisms underlie subtelomeric rearrangements, a common cause of mental retardation. Human Mutation, 2007, 28, 177-182.	1.1	15
125	Subtelomeric imbalances in phenotypically normal individuals. Human Mutation, 2007, 28, 958-967.	1.1	72
126	FVB.129P2-Pde6b+Tyrc-ch/Ant, a sighted variant of the FVB/N mouse strain suitable for behavioral analysis. Genes, Brain and Behavior, 2007, 6, 552-557.	1.1	39

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127	A de novo subterminal trisomy 10p andÂmonosomy 18q inÂaÂgirl with MCA/MR: case report andÂreview. European Journal of Medical Genetics, 2006, 49, 402-413.	0.7	8
128	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	9.4	418
129	TBP as a candidate gene for mental retardation in patients with subtelomeric 6q deletions. European Journal of Human Genetics, 2006, 14, 1090-1096.	1.4	40
130	Decreased expression of the GABAA receptor in fragile X syndrome. Brain Research, 2006, 1121, 238-245.	1.1	297
131	Expression profiling suggests underexpression of the GABAA receptor subunit Î´ in the fragile X knockout mouse model. Neurobiology of Disease, 2006, 21, 346-357.	2.1	151
132	Multiplex ligation-dependent probe amplification to detect subtelomeric rearrangements in routine diagnostics. Clinical Genetics, 2005, 69, 58-64.	1.0	57
133	Subtelomeric rearrangements in the mentally retarded: A comparison of detection methods. Human Mutation, 2005, 25, 513-524.	1.1	46
134	Clinical report of a pure subtelomeric 1qter deletion in a boy with mental retardation and multiple anomalies adds further evidence for a specific phenotype. American Journal of Medical Genetics, Part A, 2005, 135A, 91-95.	0.7	55
135	A missense mutation in the coiled-coil motif of the HP1-interacting domain of ATR-X in a family with X-linked mental retardation. Neurogenetics, 2005, 6, 45-47.	0.7	2
136	Mice lacking Dfna5 show a diverging number of cochlear fourth row outer hair cells. Neurobiology of Disease, 2005, 19, 386-399.	2.1	51
137	Cognitive decline, neuromotor and behavioural disturbances in a mouse model for fragile-X-associated tremor/ataxia syndrome (FXTAS). Behavioural Brain Research, 2005, 162, 233-239.	1.2	117
138	A splice site mutation in the methyltransferase gene FTSJ1 in Xp11.23 is associated with non-syndromic mental retardation in a large Belgian family (MRX9). Journal of Medical Genetics, 2004, 41, 679-683.	1.5	60
139	Genetic modifiers in mice: the example of the fragile X mouse model. Cytogenetic and Genome Research, 2004, 105, 448-454.	0.6	18
140	Annotation: Deconstructing the attention deficit in fragile X syndrome: a developmental neuropsychological approach. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2004, 45, 1042-1053.	3.1	81
141	The Collaborative Cross, a community resource for the genetic analysis of complex traits. Nature Genetics, 2004, 36, 1133-1137.	9.4	1,034
142	Screening for subtelomeric rearrangements using genetic markers in 70Âpatients with unexplained mental retardation. Annales De GÃ©nÃ©tique, 2004, 47, 53-59.	0.4	10
143	TM4SF10 gene sequencing in XLMR patients identifies common polymorphisms but no disease-associated mutation. BMC Medical Genetics, 2004, 5, 22.	2.1	5
144	Fragile X syndrome phenotype with normalFMR1 gene studies. American Journal of Medical Genetics Part A, 2004, 129A, 326-328.	2.4	1

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145	Subtelomeric deletions detected in patients with idiopathic mental retardation using multiplex ligation-dependent probe amplification (MLPA). Human Mutation, 2004, 23, 17-21.	1.1	94
146	Novel Truncating Mutations in the Polyglutamine Tract Binding Protein 1 Gene (PQBP1) Cause Renpenning Syndrome and X-Linked Mental Retardation in Another Family with Microcephaly. American Journal of Human Genetics, 2004, 74, 777-780.	2.6	67
147	Of mice and the fragile X syndrome. Trends in Genetics, 2003, 19, 148-154.	2.9	120
148	Expansion of the Fragile X CGG Repeat in Females with Premutation or Intermediate Alleles. American Journal of Human Genetics, 2003, 72, 454-464.	2.6	345
149	FMR1 Gene Deletion/Reversion: A Pitfall of Fragile X Carrier Testing. Genetic Testing and Molecular Biomarkers, 2003, 7, 303-308.	1.7	9
150	Mental Retardation: A Review of the Genetic Causes. British Journal of Developmental Disabilities, 2003, 49, 29-44.	0.1	31
151	Abnormal expression of the KLF8 (ZNF741) gene in a female patient with an X;autosome translocation t(X;21)(p11.2;q22.3) and non-syndromic mental retardation. Journal of Medical Genetics, 2002, 39, 113-117.	1.5	27
152	A novel 2 bp deletion in the TM4SF2 gene is associated with MRX58. Journal of Medical Genetics, 2002, 39, 430-433.	1.5	37
153	Targeting fragile X. Genome Biology, 2002, 3, reviews1014.1.	13.9	8
154	Family MRX9 revisited: Further evidence for locus heterogeneity in MRX. American Journal of Medical Genetics Part A, 2002, 112, 17-22.	2.4	2
155	Identification of a family with nonspecific mental retardation (MRX79) with the A140V mutation in theMECP2 gene: Is there a need for routine screening?. Human Mutation, 2002, 20, 249-252.	1.1	34
156	Abnormal dendritic spine characteristics in the temporal and visual cortices of patients with fragile-X syndrome: A quantitative examination. American Journal of Medical Genetics Part A, 2001, 98, 161-167.	2.4	671
157	Brain studies of mouse models for neurogenetic disorders using in vivo magnetic resonance imaging (MRI). European Journal of Human Genetics, 2001, 9, 153-159.	1.4	24
158	Precise mapping of the fragile site FRA12A on chromosome 12q13.1. GeneScreen, 2001, 1, 131-137.	0.7	1
159	Abnormal dendritic spine characteristics in the temporal and visual cortices of patients with fragile-X syndrome: A quantitative examination. , 2001, 98, 161.		3
160	Introduction of aFMR1 transgene in the fragile X knockout mouse. Neuroscience Research Communications, 2000, 26, 265-277.	0.2	12
161	Fragile X syndrome at the turn of the century. Trends in Molecular Medicine, 2000, 6, 193-198.	2.6	73
162	Long-term potentiation in mice lacking the neural cell adhesion molecule L1. Current Biology, 2000, 10, 1607-1610.	1.8	48

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163	The SOX8 Gene Is Located within 700 kb of the Tip of Chromosome 16p and Is Deleted in a Patient with ATR-16 Syndrome. Genomics, 2000, 63, 108-116.	1.3	60
164	Spatial learning, contextual fear conditioning and conditioned emotional response in Fmr1 knockout mice. Behavioural Brain Research, 2000, 117, 127-136.	1.2	133
165	Familial Mental Retardation Syndrome ATR-16 Due to an Inherited Cryptic Subtelomeric Translocation, t(3;16)(q29;p13.3). American Journal of Human Genetics, 2000, 66, 16-25.	2.6	54
166	Neuroanatomy of the fragile X knockout mouse brain studied using in vivo high resolution magnetic resonance imaging. European Journal of Human Genetics, 1999, 7, 526-532.	1.4	61
167	Postmortem examination of two fragile X brothers with anFMR1 full mutation. , 1999, 84, 245-249.		62
168	CAG repeat contraction in the androgen receptor gene in three brothers with mental retardation. , 1999, 85, 209-213.		12
169	A New Neurological Syndrome with Mental Retardation, Choreoathetosis, and Abnormal Behavior Maps to Chromosome Xp11. American Journal of Human Genetics, 1999, 65, 1406-1412.	2.6	44
170	CAG repeat contraction in the androgen receptor gene in three brothers with mental retardation. American Journal of Medical Genetics Part A, 1999, 85, 209-213.	2.4	3
171	Incomplete Eco RI digestion may lead to false diagnosis of fragile X syndrome. Human Genetics, 1998, 102, 54-56.	1.8	6
172	L1 knockout mice show dilated ventricles, vermis hypoplasia and impaired exploration patterns. Human Molecular Genetics, 1998, 7, 999-1009.	1.4	228
173	<title>MRI as a tool to study brain structure from mouse models for mental retardation</title>. , 1998, , .		1
174	The Fragile X Syndrome and Other Fragile Site Disorders. Results and Problems in Cell Differentiation, 1998, 21, 1-46.	0.2	18
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