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	The Collaborative Cross, a community resource for the genetic analysis of complex traits. Nature Genetics, 2004, 36, 1133-1137.	9.4	1,034
2	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
3	Fragile X syndrome. Nature Reviews Disease Primers, 2017, 3, 17065.	18.1	490
4	A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. Nature, 2010, 463, 671-675.	13.7	476
5	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	9.4	443
6	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	9.4	418
7	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394
8	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
9	Decreased expression of the GABAA receptor in fragile X syndrome. Brain Research, 2006, 1121, 238-245.	1.1	297
10	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	9.4	293
11	The GABAA Receptor as a Therapeutic Target for Neurodevelopmental Disorders. Neuron, 2015, 86, 1119-1130.	3.8	261
12	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
13	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
14	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
15	L1 knockout mice show dilated ventricles, vermis hypoplasia and impaired exploration patterns. Human Molecular Genetics, 1998, 7, 999-1009.	1.4	228
16	Mildly impaired water maze performance in maleFmr1 knockout mice. Neuroscience, 1997, 76, 367-376.	1.1	189
17	The complexity of the GABAA receptor shapes unique pharmacological profiles. Drug Discovery Today, 2009, 14, 866-875.	3.2	165
18	The GABAA receptor: a novel target for treatment of fragile X?. Trends in Neurosciences, 2007, 30, 425-431.	4.2	157

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19	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
20	Transgenic mouse model for the fragile X syndrome. , 1996, 64, 241-245.		156
21	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
22	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
23	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
24	Long-term potentiation in the hippocampus of fragile X knockout mice. , 1996, 64, 246-251.		146
25	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
26	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	1.4	137
27	Spatial learning, contextual fear conditioning and conditioned emotional response in Fmr1 knockout mice. Behavioural Brain Research, 2000, 117, 127-136.	1.2	133
28	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
29	Of mice and the fragile X syndrome. Trends in Genetics, 2003, 19, 148-154.	2.9	120
30	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
31	Pharmacological treatment of fragile X syndrome with GABAergic drugs in a knockout mouse model. Behavioural Brain Research, 2012, 229, 244-249.	1.2	109
32	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
33	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
34	Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy. Human Molecular Genetics, 2013, 22, 2590-2602.	1.4	103
35	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
36	Fragile sites and human disease. Human Molecular Genetics, 2007, 16, R150-R158.	1.4	94

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37	Germline mutation of microRNA-125a is associated with breast cancer. Journal of Medical Genetics, 2009, 46, 358-360.	1.5	94
38	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
39	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
40	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
41	The GABA_A receptor is an FMRP target with therapeutic potential in fragile X syndrome. Cell Cycle, 2015, 14, 2985-2995.	1.3	87
42	Fragile X syndrome: from molecular genetics to therapy. Journal of Medical Genetics, 2009, 46, 577-584.	1.5	83
43	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
44	Fragile X syndrome at the turn of the century. Trends in Molecular Medicine, 2000, 6, 193-198.	2.6	73
45	Subtelomeric imbalances in phenotypically normal individuals. Human Mutation, 2007, 28, 958-967.	1.1	72
46	Insights into GABAAergic system deficits in fragile X syndrome lead to clinical trials. Neuropharmacology, 2015, 88, 48-54.	2.0	71
47	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
48	Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes. Molecular Psychiatry, 2017, 22, 836-849.	4.1	68
49	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
50	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
51	Postmortem examination of two fragile X brothers with anFMR1 full mutation. , 1999, 84, 245-249.		62
52	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
53	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
54	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

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55	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
56	VariantDB: a flexible annotation and filtering portal for next generation sequencing data. Genome Medicine, 2014, 6, 74.	3.6	60
57	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
58	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
59	Impaired GABAergic inhibition in the hippocampus of Fmr1 knockout mice. Neuropharmacology, 2017, 116, 71-81.	2.0	58
60	Multiplex ligation-dependent probe amplification to detect subtelomeric rearrangements in routine diagnostics. Clinical Genetics, 2005, 69, 58-64.	1.0	57
61	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
62	Mouse models of the fragile X premutation and fragile X-associated tremor/ataxia syndrome. Journal of Neurodevelopmental Disorders, 2014, 6, 25.	1.5	57
63	Tauopathy in the young autistic brain: novel biomarker and therapeutic target. Translational Psychiatry, 2020, 10, 228.	2.4	57
64	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
65	Premature primary tooth eruption in cognitive/motor-delayed ADNP-mutated children. Translational Psychiatry, 2017, 7, e1043-e1043.	2.4	55
66	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
67	CNV-WebStore: Online CNV Analysis, Storage and Interpretation. BMC Bioinformatics, 2011, 12, 4.	1.2	54
68	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
69	Positron Emission Tomography (PET) Quantification of GABAA Receptors in the Brain of Fragile X Patients. PLoS ONE, 2015, 10, e0131486.	1.1	52
70	Mice lacking Dfna5 show a diverging number of cochlear fourth row outer hair cells. Neurobiology of Disease, 2005, 19, 386-399.	2.1	51
71	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
72	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50

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73	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
74	Long-term potentiation in mice lacking the neural cell adhesion molecule L1. Current Biology, 2000, 10, 1607-1610.	1.8	48
75	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
76	Subtelomeric rearrangements in the mentally retarded: A comparison of detection methods. Human Mutation, 2005, 25, 513-524.	1.1	46
77	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
78	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
79	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
80	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
81	Craniofacial characteristics of fragile X syndrome in mouse and man. European Journal of Human Genetics, 2013, 21, 816-823.	1.4	43
82	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
83	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
84	FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. PLoS Genetics, 2014, 10, e1004242.	1.5	41
85	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
86	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	1.8	40
87	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
88	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
89	The translational regulator FMRP controls lipid and glucose metabolism in mice and humans. Molecular Metabolism, 2019, 21, 22-35.	3.0	39
90	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

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91	A novel 2 bp deletion in the TM4SF2 gene is associated with MRX58. Journal of Medical Genetics, 2002, 39, 430-433.	1.5	37
92	The Compassionate Side of Neuroscience: Tony Sermoneâ€™s Undiagnosed Genetic Journeyâ€"ADNP Mutation. Journal of Molecular Neuroscience, 2015, 56, 751-757.	1.1	37
93	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
94	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
95	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
96	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
97	Mental Retardation: A Review of the Genetic Causes. British Journal of Developmental Disabilities, 2003, 49, 29-44.	0.1	31
98	Fragile X syndrome neurobiology translates into rational therapy. Drug Discovery Today, 2014, 19, 510-519.	3.2	31
99	Novel IL1RAPL1 mutations associated with intellectual disability impair synaptogenesis. Human Molecular Genetics, 2015, 24, 1106-1118.	1.4	31
100	On the DNA content of Trypanosoma cruzi. Molecular and Biochemical Parasitology, 1989, 36, 73-76.	0.5	30
101	Fragile X syndrome: from gene discovery to therapy. Frontiers in Bioscience - Landmark, 2011, 16, 1211.	3.0	30
102	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
103	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
104	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
105	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
106	Balanced translocations in mental retardation. Human Genetics, 2009, 126, 133-147.	1.8	27
107	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
108	Evidence for diploidy in metacyclic forms of African trypanosomes.. Proceedings of the National Academy of Sciences of the United States of America, 1989, 86, 5469-5472.	3.3	26

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109	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	1.0	26
110	The molecular basis of the folate-sensitive fragile site FRA11A at 11q13. Cytogenetic and Genome Research, 2007, 119, 9-14.	0.6	25
111	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
112	Abundancy of polymorphic CGG repeats in the human genome suggest a broad involvement in neurological disease. Scientific Reports, 2021, 11, 2515.	1.6	25
113	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
114	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
115	Effect of genetic background on acoustic startle response in fragile X knockout mice. Genetical Research, 2008, 90, 341-345.	0.3	24
116	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
117	A Yeast Artificial Chromosome Contig That Spans the RB1-D13S31 Interval on Human Chromosome 13 and Encompasses the Frequently Deleted Region in B-cell Chronic Lymphocytic Leukemia. Genomics, 1995, 30, 425-430.	1.3	21
118	Mutations in ADNP affect expression and subcellular localization of the protein. Cell Cycle, 2018, 17, 1068-1075.	1.3	21
119	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
120	Advances in understanding fragile X syndrome and related disorders. Current Opinion in Pediatrics, 2011, 23, 601-606.	1.0	20
121	GABAergic abnormalities in the fragile X syndrome. European Journal of Paediatric Neurology, 2020, 24, 100-104.	0.7	20
122	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
123	TheTrypanosoma bruceiDNA polymerase Î± core subunit gene is developmentally regulated and linked to a constitutively expressed open reading frame. Nucleic Acids Research, 1991, 19, 6441-6447.	6.5	19
124	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
125	Genetic modifiers in mice: the example of the fragile X mouse model. Cytogenetic and Genome Research, 2004, 105, 448-454.	0.6	18
126	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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127	Involvement and Therapeutic Potential of the GABAergic System in the Fragile X Syndrome. Scientific World Journal, The, 2010, 10, 2198-2206.	0.8	17
128	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
129	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
130	A mouse model for intellectual disability caused by mutations in the X-linked 2â€²â€˜Oâ€˜methyltransferase Ftsj1 gene. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2083-2093.	1.8	17
131	Behavioural characterization of AnkyrinG deficient mice, a model for ANK3 related disorders. Behavioural Brain Research, 2017, 328, 218-226.	1.2	16
132	Diverse chromosome breakage mechanisms underlie subtelomeric rearrangements, a common cause of mental retardation. Human Mutation, 2007, 28, 177-182.	1.1	15
133	GABA neurotransmitter signaling in the developing mouse lens: Dynamic regulation of components and functionality. Developmental Dynamics, 2008, 237, 3830-3841.	0.8	15
134	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
135	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
136	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
137	PLCB1 epileptic encephalopathies; Review and expansion of the phenotypic spectrum. European Journal of Paediatric Neurology, 2016, 20, 474-479.	0.7	14
138	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
139	Detection and interpretation of genomic structural variation in health and disease. Expert Review of Molecular Diagnostics, 2013, 13, 61-82.	1.5	13
140	A Robust Protocol to Increase NimbleGen SeqCap EZ Multiplexing Capacity to 96 Samples. PLoS ONE, 2015, 10, e0123872.	1.1	13
141	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	1.1	13
142	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
143	Towards Kinase Inhibitor Therapies for Fragile X Syndrome: Tweaking Twists in the Autism Spectrum Kinase Signaling Network. Cells, 2022, 11, 1325.	1.8	13
144	CAG repeat contraction in the androgen receptor gene in three brothers with mental retardation. , 1999, 85, 209-213.		12

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145	Introduction of aFMR1 transgene in the fragile X knockout mouse. Neuroscience Research Communications, 2000, 26, 265-277.	0.2	12
146	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
147	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
148	Reduced serum levels of pro-inflammatory chemokines in fragile X syndrome. BMC Neurology, 2020, 20, 138.	0.8	10
149	Physical localisation of the chromosomal marker D13S31 places the Wilson disease locus at the junction of bands q14.3 and q21.1 of chromosome 13. Human Genetics, 1993, 91, 504-506.	1.8	9
150	FMR1 Gene Deletion/Reversion: A Pitfall of Fragile X Carrier Testing. Genetic Testing and Molecular Biomarkers, 2003, 7, 303-308.	1.7	9
151	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
152	European Gene Mapping Project (EUROGEM): Breakpoint panels for human chromosomes based on the CEPH reference families. Annals of Human Genetics, 1996, 60, 447-486.	0.3	8
153	Severe mental retardation and macroorchidism without mutation in the FMR1 gene. , 1996, 64, 408-412.		8
154	Targeting fragile X. Genome Biology, 2002, 3, reviews1014.1.	13.9	8
155	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
156	Beckwithâ€“Wiedemannâ€like macroglossia and 18q23 haploinsufficiency. American Journal of Medical Genetics, Part A, 2007, 143A, 2796-2803.	0.7	8
157	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
158	Single-Cell and Neuronal Network Alterations in an In Vitro Model of Fragile X Syndrome. Cerebral Cortex, 2020, 30, 31-46.	1.6	8
159	A Deletion Hybrid Breakpoint Map of the Chromosomal Region 13q14-q21 Orders 19 Genetic Markers in 10 Intervals. European Journal of Human Genetics, 1994, 2, 59-65.	1.4	8
160	Ordering of polymorphic markers in the chromosome region 3p21. Cytogenetic and Genome Research, 1996, 72, 225-228.	0.6	7
161	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
162	Incomplete Eco RI digestion may lead to false diagnosis of fragile X syndrome. Human Genetics, 1998, 102, 54-56.	1.8	6

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163	Distinct disorders affecting the brain share common genetic origins. F1000 Biology Reports, 2010, 2, .	4.0	6
164	TM4SF10 gene sequencing in XLMR patients identifies common polymorphisms but no disease-associated mutation. BMC Medical Genetics, 2004, 5, 22.	2.1	5
165	Influence of fever and flurbiprofen on trypanosome growth. Acta Tropica, 1990, 47, 115-123.	0.9	4
166	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
167	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
168	Long-term potentiation in the hippocampus of fragile X knockout mice. , 1996, 64, 246.		4
169	Genetic Overlaps in Mental Retardation, Autism and Schizophrenia. Monographs in Human Genetics, 2010, , 126-136.	0.5	3
170	Animal Models of Fragile X Syndrome. , 2017, , 123-147.		3
171	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
172	Abnormal dendritic spine characteristics in the temporal and visual cortices of patients with fragile-X syndrome: A quantitative examination. , 2001, 98, 161.		3
173	An Integrated Map of Human Chromosome 13 Allowing Regional Localization of Genetic Markers. European Journal of Human Genetics, 1995, 3, 180-187.	1.4	3
174	Expanding the Phenotype of B3GALNT2-Related Disorders. Genes, 2022, 13, 694.	1.0	3
175	Family MRX9 revisited: Further evidence for locus heterogeneity in MRX. American Journal of Medical Genetics Part A, 2002, 112, 17-22.	2.4	2
176	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
177	Metabonomics adds a new dimension to fragile Ã— syndrome. Genome Medicine, 2011, 3, 80.	3.6	2
178	The GABAergic System Contributions to the Fragile X Syndrome Phenotype. , 2017, , 205-215.		2
179	On the spot: very local chromosomal rearrangements. F1000 Biology Reports, 2012, 4, 22.	4.0	2
180	Therapeutic potential of GABAA receptor subunit expression abnormalities in fragile X syndrome. Expert Review of Precision Medicine and Drug Development, 0, , 1-16.	0.4	2

#	Article	IF	Citations
181	Protein interaction network analysis reveals genetic enrichment of immune system genes in frontotemporal dementia. Neurobiology of Aging, 2022, 116, 67-79.	1.5	2
182	Mean corpuscular hemoglobin is not increased in Fmr1 knockout mice. Human Genetics, 1996, 97, 49-50.	1.8	1
183	<title>MRI as a tool to study brain structure from mouse models for mental retardation</title>. , 1998, , .		1
184	Precise mapping of the fragile site FRA12A on chromosome 12q13.1. GeneScreen, 2001, 1, 131-137.	0.7	1
185	Fragile X syndrome phenotype with normalFMR1 gene studies. American Journal of Medical Genetics Part A, 2004, 129A, 326-328.	2.4	1
186	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
187	PUM1 haploinsufficiency is associated with syndromic neurodevelopmental delay and epilepsy. American Journal of Medical Genetics, Part A, 2020, 182, 591-594.	0.7	1
188	A highly informative dinucleotide repeat polymorphism at D13S201, between RB1 and WND. Human Genetics, 1995, 95, 589.	1.8	0
189	Thoracic dimples and dysmorphic features associated with a partial duplication and triplication of chromosome 12q24. Clinical Dysmorphology, 2016, 25, 167-173.	0.1	0
190	A contemporary view on the molecular basis of neurodevelopmental disorders. , 2020, , 57-78.		0
191	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
192	ZNF711 puts a spell on DNA. European Journal of Human Genetics, 2022, , .	1.4	0