James F Gusella

List of Publications by Year
in descending order

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210 papers	26,492 citations	8755 75 h-index	6471 157 g-index
240 all docs	240 docs citations	240 times ranked	20618 citing authors

#	Article	IF	Citations
1	A polymorphic DNA marker genetically linked to Huntington's disease. Nature, 1983, 306, 234-238.	27.8	2,289
2	Protease inhibitor domain encoded by an amyloid protein precursor mRNA associated with Alzheimer's disease. Nature, 1988, 331, 528-530.	27.8	1,105
3	The early-onset torsion dystonia gene (DYT1) encodes an ATP-binding protein. Nature Genetics, 1997, 17, 40-48.	21.4	1,051
4	Huntington disease. Nature Reviews Disease Primers, 2015, 1, 15005.	30.5	1,031
5	Relationship between trinucleotide repeat expansion and phenotypic variation in Huntington's disease. Nature Genetics, 1993, 4, 393-397.	21.4	672
6	Venezuelan kindreds reveal that genetic and environmental factors modulate Huntington's disease age of onset. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 3498-3503.	7.1	666
7	CAG repeat number governs the development rate of pathology in Huntington's disease. Annals of Neurology, 1997, 41, 689-692.	5.3	605
8	Tissue-Specific Expression of a Splicing Mutation in the Gene Causes Familial Dysautonomia. American Journal of Human Genetics, 2001, 68, 598-605.	6.2	558
9	Isolation of a novel gene underlying batten disease, CLN3. Cell, 1995, 82, 949-957.	28.9	554
10	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. Cell, 2012, 149, 525-537.	28.9	534
11	Huntingtons Disease. New England Journal of Medicine, 1986, 315, 1267-1276.	27.0	532
12	The FERM domain: a unique module involved in the linkage of cytoplasmic proteins to the membrane. Trends in Biochemical Sciences, 1998, 23, 281-282.	7.5	494
13	Loss of genes on chromosome 22 in tumorigenesis of human acoustic neuroma. Nature, 1986, 322, 644-647.	27.8	490
14	Genetic linkage of bilateral acoustic neurofibromatosis to a DNA marker on chromosome 22. Nature, 1987, 329, 246-248.	27.8	478
15	Huntingtin is required for neurogenesis and is not impaired by the Huntington's disease CAG expansion. Nature Genetics, 1997, 17, 404-410.	21.4	472
16	Molecular genetics: Unmasking polyglutamine triggers in neurodegenerative disease. Nature Reviews Neuroscience, 2000, 1, 109-115.	10.2	383
17	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. Nature, 2012, 485, 363-367.	27.8	363
18	Use of cyclosporin a in establishing epstein-barr virus-transformed human lymphoblastoid cell lines. In Vitro, 1984, 20, 856-858.	1.2	344

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19	Hyperkalemic periodic paralysis and the adult muscle sodium channel alpha-subunit gene. Science, 1990, 250, 1000-1002.	12.6	332
20	Familial dysautonomia. Current Opinion in Genetics and Development, 2002, 12, 307-311.	3.3	332
21	Homozygotes for Huntington's disease. Nature, 1987, 326, 194-197.	27.8	331
22	CAG Repeat Not Polyglutamine Length Determines Timing of Huntingtonâ€™s Disease Onset. Cell, 2019, 178, 887-900.e14.	28.9	301
23	CHD8 regulates neurodevelopmental pathways associated with autism spectrum disorder in neural progenitors. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4468-77.	7.1	297
24	The genetic defect in familial Alzheimer's disease is not tightly linked to the amyloid Î²-protein gene. Nature, 1987, 329, 156-157.	27.8	275
25	HD CAG repeat implicates a dominant property of huntingtin in mitochondrial energy metabolism. Human Molecular Genetics, 2005, 14, 2871-2880.	2.9	274
26	NF2/Merlin Is a Novel Negative Regulator of mTOR Complex 1, and Activation of mTORC1 Is Associated with Meningioma and Schwannoma Growth. Molecular and Cellular Biology, 2009, 29, 4250-4261.	2.3	264
27	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
28	Specific progressive cAMP reduction implicates energy deficit in presymptomatic Huntington's disease knock-in mice. Human Molecular Genetics, 2003, 12, 497-508.	2.9	250
29	Amyloid Formation by Mutant Huntingtin: Threshold, Progressivity and Recruitment of Normal Polyglutamine Proteins. Somatic Cell and Molecular Genetics, 1998, 24, 217-233.	0.7	249
30	Human gene for torsion dystonia located on chromosome 9q32-q34. Neuron, 1989, 2, 1427-1434.	8.1	246
31	Structure and expression of the Huntington's disease gene: Evidence against simple inactivation due to an expanded CAG repeat. Somatic Cell and Molecular Genetics, 1994, 20, 27-38.	0.7	246
32	Mutations in transcript isoforms of the neurofibromatosis 2 gene in multiple human tumour types. Nature Genetics, 1994, 6, 185-192.	21.4	236
33	NHE-RF, a Regulatory Cofactor for Na+-H+Exchange, Is a Common Interactor for Merlin and ERM (MERM) Proteins. Journal of Biological Chemistry, 1998, 273, 1273-1276.	3.4	229
34	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nature Genetics, 2012, 44, 390-397.	21.4	229
35	Huntington's disease gene: Regional and cellular expression in brain of normal and affected individuals. Annals of Neurology, 1995, 37, 218-230.	5.3	206
36	Exon scanning for mutation of the NF2 gene in schwannomas. Human Molecular Genetics, 1994, 3, 413-419.	2.9	200

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37	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. American Journal of Human Genetics, 2011, 89, 551-563.	6.2	195
38	Permanent inactivation of Huntington's disease mutation by personalized allele-specific CRISPR/Cas9. Human Molecular Genetics, 2016, 25, ddw286.	2.9	195
39	Biotin-Responsive Basal Ganglia Disease Maps to 2q36.3 and Is Due to Mutations in SLC19A3. American Journal of Human Genetics, 2005, 77, 16-26.	6.2	178
40	Clinical Diagnosis by Whole-Genome Sequencing of a Prenatal Sample. New England Journal of Medicine, 2012, 367, 2226-2232.	27.0	174
41	Huntingtin facilitates polycomb repressive complex 2. Human Molecular Genetics, 2010, 19, 573-583.	2.9	169
42	_{Predictive Testing for Huntingtons Disease with Use of a Linked DNA Marker}. New England Journal of Medicine, 1988, 318, 535-542.	27.0	167
43	Quantitative neuropathological changes in presymptomatic Huntington's disease. Annals of Neurology, 2001, 49, 29-34.	5.3	163
44	Early phenotypes that presage late-onset neurodegenerative disease allow testing of modifiers in Hdh CAG knock-in mice. Human Molecular Genetics, 2002, 11, 633-640.	2.9	162
45	The Huntington's disease candidate region exhibits many different haplotypes. Nature Genetics, 1992, 1, 99-103.	21.4	157
46	Neurofibromatosis type 1 gene mutations in neuroblastoma. Nature Genetics, 1993, 3, 62-66.	21.4	157
47	Next-Generation Sequencing Strategies Enable Routine Detection of Balanced Chromosome Rearrangements for Clinical Diagnostics and Genetic Research. American Journal of Human Genetics, 2011, 88, 469-481.	6.2	154
48	RNA Sequence Analysis of Human Huntington Disease Brain Reveals an Extensive Increase in Inflammatory and Developmental Gene Expression. PLoS ONE, 2015, 10, e0143563.	2.5	150
49	Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. Nature Genetics, 1993, 4, 160-164.	21.4	149
50	A Genome Scan for Modifiers of Age at Onset in Huntington Disease: The HD MAPS Study. American Journal of Human Genetics, 2003, 73, 682-687.	6.2	148
51	Heterogeneous Topographic and Cellular Distribution of Huntingtin Expression in the Normal Human Neostriatum. Journal of Neuroscience, 1997, 17, 3052-3063.	3.6	143
52	Dystonia gene in Ashkenazi Jewish population is located on chromosome 9q32-34. Annals of Neurology, 1990, 27, 114-120.	5.3	141
53	Rescue of a human mRNA splicing defect by the plant cytokinin kinetin. Human Molecular Genetics, 2003, 13, 429-436.	2.9	139
54	Frequency and distribution ofNF2 mutations in schwannomas. Genes Chromosomes and Cancer, 1996, 17, 45-55.	2.8	134

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55	Reversal of a full-length mutant huntingtin neuronal cell phenotype by chemical inhibitors of polyglutamine-mediated aggregation. BMC Neuroscience, 2005, 6, 1.	1.9	133
56	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
57	The predominantly HEAT-like motif structure of huntingtin and its association and coincident nuclear entry with dorsal, an NF-kB/Rel/dorsal family transcription factor. BMC Neuroscience, 2002, 3, 15.	1.9	130
58	The HTT CAG-Expansion Mutation Determines Age at Death but Not Disease Duration in Huntington Disease. American Journal of Human Genetics, 2016, 98, 287-298.	6.2	129
59	Huntington's disease: seeing the pathogenic process through a genetic lens. Trends in Biochemical Sciences, 2006, 31, 533-540.	7.5	128
60	The gene for achondroplasia maps to the telomeric region of chromosome 4p. Nature Genetics, 1994, 6, 314-317.	21.4	116
61	Genetic modifiers of Huntington's disease. Movement Disorders, 2014, 29, 1359-1365.	3.9	116
62	Transcriptional Consequences of 16p11.2 Deletion and Duplication in Mouse Cortex and Multiplex Autism Families. American Journal of Human Genetics, 2014, 94, 870-883.	6.2	116
63	Deletion of Huntington's disease-linked G8 (D4S10) locus in Wolfâ€“Hirschhorn syndrome. Nature, 1985, 318, 75-78.	27.8	114
64	miR-10b-5p expression in Huntingtonâ€™s disease brain relates to age of onset and the extent of striatal involvement. BMC Medical Genomics, 2015, 8, 10.	1.5	114
65	A comparison of neurological, metabolic, structural, and genetic evaluations in persons at risk for Huntington's disease. Annals of Neurology, 1990, 28, 614-621.	5.3	110
66	Frequent loss of chromosome 14 in atypical and malignant meningioma: identification of a putative `tumor progression' locus. Oncogene, 1997, 14, 611-616.	5.9	109
67	Huntington's disease: the case for genetic modifiers. Genome Medicine, 2009, 1, 80.	8.2	104
68	A cosmid contig and high resolution restriction map of the 2 megabase region containing the Huntington's disease gene. Nature Genetics, 1993, 4, 181-186.	21.4	102
69	Normal and Expanded Huntingtonâ€™s Disease Gene Alleles Produce Distinguishable Proteins Due to Translation Across the CAG Repeat. Molecular Medicine, 1995, 1, 374-383.	4.4	97
70	MicroRNAs Located in the Hox Gene Clusters Are Implicated in Huntington's Disease Pathogenesis. PLoS Genetics, 2014, 10, e1004188.	3.5	97
71	Regulation of mTOR Complex 2 Signaling in Neurofibromatosis 2â€“Deficient Target Cell Types. Molecular Cancer Research, 2012, 10, 649-659.	3.4	96
72	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. American Journal of Human Genetics, 2008, 82, 712-722.	6.2	95

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73	A modifier of Huntington's disease onset at the MLH1 locus. Human Molecular Genetics, 2017, 26, 3859-3867.	2.9	88
74	Mapping of the Mucolipidosis Type IV Gene to Chromosome 19p and Definition of Founder Haplotypes. American Journal of Human Genetics, 1999, 65, 773-778.	6.2	87
75	High resolution localization of recombination hot spots using sperm typing. Nature Genetics, 1994, 7, 420-424.	21.4	80
76	Huntington's Disease. NeuroMolecular Medicine, 2003, 4, 7-20.	3.4	79
77	Brain urea increase is an early Huntingtonâ€™s disease pathogenic event observed in a prodromal transgenic sheep model and HD cases. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E11293-E11302.	7.1	78
78	Huntington's disease CAG trinucleotide repeats in pathologically confirmed post-mortem brains. Neurobiology of Disease, 1994, 1, 159-166.	4.4	77
79	Differential Expression of Normal and Mutant Huntington's Disease Gene Alleles. Neurobiology of Disease, 1996, 3, 183-190.	4.4	77
80	Huntington's disease: two families with differing clinical features show linkage to the G8 probe. Science, 1985, 229, 776-779.	12.6	75
81	Functionally compromisedCHD7alleles in patients with isolated GnRH deficiency. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 17953-17958.	7.1	74
82	Isolation of polymorphic DNA segments from human chromosome 21. Nucleic Acids Research, 1985, 13, 6075-6088.	14.5	73
83	TRINUCLEOTIDE INSTABILITY: A Repeating Theme in Human Inherited Disorders. Annual Review of Medicine, 1996, 47, 201-209.	12.2	73
84	Genome-wide significance for a modifier of age at neurological onset in Huntington's Disease at 6q23-24: the HD MAPS study. BMC Medical Genetics, 2006, 7, 71.	2.1	72
85	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. Nature Neuroscience, 2016, 19, 517-522.	14.8	72
86	Huntingtin: a single bait hooks many species. Current Opinion in Neurobiology, 1998, 8, 425-430.	4.2	70
87	Precise Genetic Mapping and Haplotype Analysis of the Familial Dysautonomia Gene on Human Chromosome 9q31. American Journal of Human Genetics, 1999, 64, 1110-1118.	6.2	69
88	Evidence for a modifier of onset age in Huntington disease linked to the HD gene in 4p16. Neurogenetics, 2004, 5, 109-114.	1.4	67
89	HD CAG-correlated gene expression changes support a simple dominant gain of function. Human Molecular Genetics, 2011, 20, 2846-2860.	2.9	67
90	Magicin, a novel cytoskeletal protein associates with the NF2 tumor suppressor merlin and Grb2. Oncogene, 2004, 23, 8815-8825.	5.9	66

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91	Common SNP-Based Haplotype Analysis of the 4p16.3 Huntington Disease Gene Region. American Journal of Human Genetics, 2012, 90, 434-444.	6.2	60
92	Mutant Huntingtin Forms in Vivo Complexes with Distinct Context-Dependent Conformations of the Polyglutamine Segment. Neurobiology of Disease, 1999, 6, 364-375.	4.4	57
93	The Genetic Defect Causing Huntingtonâ€™s Disease: Repeated in Other Contexts?. Molecular Medicine, 1997, 3, 238-246.	4.4	56
94	Clonal Analysis of a Case of Multiple Meningiomas Using Multiple Molecular Genetic Approaches: Pathology Case Report. Neurosurgery, 1999, 45, 409-416.	1.1	56
95	Htt CAG repeat expansion confers pleiotropic gains of mutant huntingtin function in chromatin regulation. Human Molecular Genetics, 2015, 24, 2442-2457.	2.9	53
96	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.	6.2	53
97	CSF1Rmosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. Brain, 2016, 139, 1666-1672.	7.6	53
98	Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. American Journal of Human Genetics, 2016, 99, 1015-1033.	6.2	53
99	Timing and Impact of Psychiatric, Cognitive, and Motor Abnormalities in Huntington Disease. Neurology, 2021, 96, e2395-e2406.	1.1	53
100	Identification of a presymptomatic molecular phenotype in Hdh CAG knock-in mice. Human Molecular Genetics, 2002, 11, 2233-2241.	2.9	52
101	Deficiency of Huntingtin Has Pleiotropic Effects in the Social Amoeba Dictyostelium discoideum. PLoS Genetics, 2011, 7, e1002052.	3.5	52
102	Assessment of cortical and striatal involvement in 523 Huntington disease brains. Neurology, 2012, 79, 1708-1715.	1.1	52
103	Metabolic disruption identified in the Huntingtonâ€™s disease transgenic sheep model. Scientific Reports, 2016, 6, 20681.	3.3	52
104	Huntingtonâ€™s Disease Pathogenesis: Two Sequential Components. Journal of Huntington's Disease, 2021, 10, 35-51.	1.9	49
105	Promotion of somatic CAG repeat expansion by Fan1 knock-out in Huntingtonâ€™s disease knock-in mice is blocked by Mlh1 knock-out. Human Molecular Genetics, 2020, 29, 3044-3053.	2.9	48
106	Further Molecular Characterisation of the OVT73 Transgenic Sheep Model of Huntington's Disease Identifies Cortical Aggregates. Journal of Huntington's Disease, 2013, 2, 279-295.	1.9	47
107	Large-scale phenome analysis defines a behavioral signature for Huntington's disease genotype in mice. Nature Biotechnology, 2016, 34, 838-844.	17.5	46
108	A high-throughput kinome screen reveals serum/glucocorticoid-regulated kinase 1 as a therapeutic target for NF2-deficient meningiomas. Oncotarget, 2015, 6, 16981-16997.	1.8	46

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109	Merlin: the neurofibromatosis 2 tumor suppressor. Biochimica Et Biophysica Acta: Reviews on Cancer, 1999, 1423, M29-M36.	7.4	45
110	Meclizine is neuroprotective in models of Huntington's disease. Human Molecular Genetics, 2011, 20, 294-300.	2.9	45
111	Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. American Journal of Human Genetics, 2014, 95, 454-461.	6.2	45
112	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. American Journal of Human Genetics, 2015, 97, 170-176.	6.2	45
113	A novel neurodevelopmental disorder associated with compound heterozygous variants in the huntingtin gene. European Journal of Human Genetics, 2016, 24, 1826-1827.	2.8	45
114	Genetic and Functional Analyses Point to FAN1 as the Source of Multiple Huntington Disease Modifier Effects. American Journal of Human Genetics, 2020, 107, 96-110.	6.2	45
115	Modeling NF2 with human arachnoidal and meningioma cell culture systems: NF2 silencing reflects the benign character of tumor growth. Neurobiology of Disease, 2008, 29, 278-292.	4.4	42
116	Describing Sequencing Results of Structural Chromosome Rearrangements with a Suggested Next-Generation Cytogenetic Nomenclature. American Journal of Human Genetics, 2014, 94, 695-709.	6.2	42
117	Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. European Journal of Human Genetics, 2014, 22, 57-63.	2.8	42
118	Monozygotic twins discordant for neurofibromatosis 1. American Journal of Medical Genetics, Part A, 2010, 152A, 601-606.	1.2	40
119	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	1.2	40
120	Irradiation-reduced human chromosome 21 hybrids. Somatic Cell and Molecular Genetics, 1988, 14, 233-242.	0.7	38
121	Clinical-Genetic Associations in the Prospective Huntington at Risk Observational Study (PHAROS). JAMA Neurology, 2016, 73, 102.	9.0	38
122	High resolution time-course mapping of early transcriptomic, molecular and cellular phenotypes in Huntingtonâ€™s disease CAG knock-in mice across multiple genetic backgrounds. Human Molecular Genetics, 2017, 26, 913-922.	2.9	37
123	16p11.2 deletion is associated with hyperactivation of human iPSC-derived dopaminergic neuron networks and is rescued by RHOA inhibition in vitro. Nature Communications, 2021, 12, 2897.	12.8	35
124	Evidence for Subarachnoid Spread in the Development of Multiple Meningiomas. Brain Pathology, 1995, 5, 11-14.	4.1	33
125	De novo variants in the Helicase-C domain of CHD8 are associated with severe phenotypes including autism, language disability and overgrowth. Human Genetics, 2020, 139, 499-512.	3.8	32
126	Brain-derived neurotrophic factor does not influence age at neurologic onset of Huntingtonâ€™s disease. Neurobiology of Disease, 2006, 24, 280-285.	4.4	31

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127	Exome sequencing of individuals with Huntingtonâ€™s disease implicates FAN1 nuclease activity in slowing CAG expansion and disease onset. Nature Neuroscience, 2022, 25, 446-457.	14.8	31
128	The Genetic Modifiers of Motor OnsetAgeÂ(GeM MOA) Website: Genome-wide Association Analysis for Genetic Modifiers of Huntingtonâ€™s Disease. Journal of Huntington's Disease, 2015, 4, 279-284.	1.9	30
129	Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis. Human Molecular Genetics, 2016, 25, 1255-1270.	2.9	30
130	Genetic Modification of Huntington Disease Acts Early in the Prediagnosis Phase. American Journal of Human Genetics, 2018, 103, 349-357.	6.2	30
131	Rare DeleteriousPARD3Variants in the aPKC-Binding Region are Implicated in the Pathogenesis of Human Cranial Neural Tube Defects Via Disrupting Apical Tight Junction Formation. Human Mutation, 2017, 38, 378-389.	2.5	29
132	Genetic Risk Underlying Psychiatric and Cognitive Symptoms in Huntingtonâ€™s Disease. Biological Psychiatry, 2020, 87, 857-865.	1.3	29
133	Genetic modifiers of Huntington disease differentially influence motor and cognitive domains. American Journal of Human Genetics, 2022, 109, 885-899.	6.2	29
134	A Balanced Translocation in Kallmann Syndrome Implicates a Long Noncoding RNA, RMST, as a GnRH Neuronal Regulator. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e231-e244.	3.6	28
135	No post-genetics era in human disease research. Nature Reviews Genetics, 2002, 3, 72-79.	16.3	27
136	Population-specific genetic modification of Huntington's disease in Venezuela. PLoS Genetics, 2018, 14, e1007274.	3.5	27
137	Population stratification may bias analysis of PGC-1Î± as a modifier of age at Huntington disease motor onset. Human Genetics, 2012, 131, 1833-1840.	3.8	26
138	Allelic expression of the NF2 gene in neurofibromatosis 2 and schwannomatosis. Neurogenetics, 1999, 2, 101-108.	1.4	25
139	Linkage Analysis in a Family with Dominantly Inherited Torsion Dystonia: Exclusion of the Pro-Opiomelanocortin and Glutamic Acid Decarboxylase Genes and Other Chromosomal Regions Using DNA Polymorphisms. Journal of Neurogenetics, 1986, 3, 159-175.	1.4	24
140	Cloning, Characterization, and Genomic Structure of the Mouse Ikbkap Gene. DNA and Cell Biology, 2001, 20, 579-586.	1.9	24
141	Haplotype-based stratification of Huntington's disease. European Journal of Human Genetics, 2017, 25, 1202-1209.	2.8	24
142	Mutations causing Lopes-Maciel-Rodan syndrome are huntingtin hypomorphs. Human Molecular Genetics, 2021, 30, 135-148.	2.9	24
143	Hunting for Huntington's Disease. , 1993, 3, 139-158.		24
144	Differential effects of the Huntington's disease CAG mutation in striatum and cerebellum are quantitative not qualitative. Human Molecular Genetics, 2011, 20, 4258-4267.	2.9	23

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145	Expanding the Spectrum of Founder Mutations Causing Isolated Gonadotropin-Releasing Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1378-E1385.	3.6	22
146	Sequence-Level Analysis of the Major European Huntington Disease Haplotype. American Journal of Human Genetics, 2015, 97, 435-444.	6.2	22
147	EPH receptor signaling as a novel therapeutic target in NF2-deficient meningioma. Neuro-Oncology, 2018, 20, 1185-1196.	1.2	22
148	A rare exonic NRXN3 deletion segregating with neurodevelopmental and neuropsychiatric conditions in a threeâ€generation Chinese family. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 589-595.	1.7	22
149	Location cloning strategy for characterizing genetic defects in Huntington's disease and Alzheimer's disease. FASEB Journal, 1989, 3, 2036-2041.	0.5	21
150	Opposing Tumor-Promoting and -Suppressive Functions of Rictor/mTORC2 Signaling in Adult Glioma and Pediatric SHH Medulloblastoma. Cell Reports, 2018, 24, 463-478.e5.	6.4	21
151	TAA repeat variation in the GRIK2 gene does not influence age at onset in Huntingtonâ€™s disease. Biochemical and Biophysical Research Communications, 2012, 424, 404-408.	2.1	20
152	Prenatal diagnosis of familial dysautonomia by analysis of linked CA-repeat polymorphisms on chromosome 9q31-q33. American Journal of Medical Genetics Part A, 1995, 59, 349-355.	2.4	19
153	WNT/Î²-Catenin Pathway and Epigenetic Mechanisms Regulate the Pitt-Hopkins Syndrome and Schizophrenia Risk Gene TCF4. Molecular Neuropsychiatry, 2017, 3, 53-71.	2.9	19
154	Brigatinib causes tumor shrinkage in both NF2-deficient meningioma and schwannoma through inhibition of multiple tyrosine kinases but not ALK. PLoS ONE, 2021, 16, e0252048.	2.5	19
155	Mediator Subunit Med28 Is Essential for Mouse Peri-Implantation Development and Pluripotency. PLoS ONE, 2015, 10, e0140192.	2.5	19
156	Characterization of a duplication in the terminal band of 4p by molecular cytogenetics. American Journal of Medical Genetics Part A, 1993, 46, 72-76.	2.4	18
157	Genetic Variation in the 3â€² Untranslated Region of the Neurofibromatosis 1 Gene: Application to Unequal Allelic Expression. Somatic Cell and Molecular Genetics, 1998, 24, 107-119.	0.7	18
158	Molecular detection of a 4p deletion using PCR-based polymorphisms: A technique for the rapid detection of the Wolf-Hirschhorn syndrome. American Journal of Medical Genetics Part A, 1992, 44, 449-454.	2.4	17
159	Candidate loci for Zimmermannâ€“Laband syndrome at 3p14.3. American Journal of Medical Genetics, Part A, 2007, 143A, 107-111.	1.2	17
160	Traditional and systems biology based drug discovery for the rare tumor syndrome neurofibromatosis type 2. PLoS ONE, 2018, 13, e0197350.	2.5	17
161	CNV profiles of Chinese pediatric patients with developmental disorders. Genetics in Medicine, 2021, 23, 669-678.	2.4	17
162	Modified Single-Stranded Oligonucleotides Inhibit Aggregate Formation and Toxicity Induced by Expanded Polyglutamine. Journal of Molecular Neuroscience, 2004, 24, 257-268.	2.3	16

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163	Genetic analysis of the GRIK2modifier effect in Huntington's disease. BMC Neuroscience, 2006, 7, 62.	1.9	16
164	Probes in Huntington's chorea (reply). Nature, 1986, 320, 21-22.	27.8	15
165	Huntingtonâ€™s disease: nearly four decades of human molecular genetics. Human Molecular Genetics, 2021, 30, R254-R263.	2.9	15
166	Potential molecular consequences of transgene integration: The R6/2 mouse example. Scientific Reports, 2017, 7, 41120.	3.3	14
167	Novel allele-specific quantification methods reveal no effects of adult onset CAG repeats on HTT mRNA and protein levels. Human Molecular Genetics, 2017, 26, 1258-1267.	2.9	14
168	Hypomorphic mutation of the mouse Huntingtonâ€™s disease gene orthologue. PLoS Genetics, 2019, 15, e1007765.	3.5	13
169	Dystonia-specific mutations in THAP1 alter transcription of genes associated with neurodevelopment and myelin. American Journal of Human Genetics, 2021, 108, 2145-2158.	6.2	13
170	No Association between ?1-Antichymotrypsin and Familial Alzheimer's Diseases. Annals of the New York Academy of Sciences, 1996, 802, 35-41.	3.8	12
171	Genetic criteria for Huntington's disease pathogenesis. Brain Research Bulletin, 2007, 72, 78-82.	3.0	12
172	Extensive molecular genetic analysis of the 3p14.3 region in patients with Zimmermannâ€“Laband syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 2668-2674.	1.2	12
173	Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. European Journal of Human Genetics, 2016, 24, 1622-1626.	2.8	12
174	Characterization of an unusual and complex chromosome 21 rearrangement using somatic cell genetics and cloned DNA probes. American Journal of Medical Genetics Part A, 1989, 33, 369-375.	2.4	11
175	Prenatal diagnostic testing for familial dysautonomia using linked genetic markers. Prenatal Diagnosis, 1995, 15, 817-826.	2.3	11
176	Changing Models of Biomedical Research. Science Translational Medicine, 2009, 1, 1cm1.	12.4	11
177	Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. Molecular Autism, 2020, 11, 45.	4.9	11
178	Genetic Linkage of the Huntingtonâ€™s Disease Gene to a DNA Marker. Canadian Journal of Neurological Sciences, 1984, 11, 421-425.	0.5	10
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