James F Gusella

List of Publications by Year
in descending order

210 papers	26,492 citations	10070 75 h-index	7427 157 g-index
240 all docs	240 docs citations	240 times ranked	22650 citing authors

#	Article	IF	Citations
1	Genetic modifiers of Huntington disease differentially influence motor and cognitive domains. American Journal of Human Genetics, 2022, 109, 885-899.	2.6	29
2	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.	7.1	31
3	Inherited HTT CAG repeat length does not have a major impact on Huntington disease duration. American Journal of Human Genetics, 2022, 109, 1338-1340.	2.6	3
4	Mutations causing Lopes-Maciel-Rodan syndrome are huntingtin hypomorphs. Human Molecular Genetics, 2021, 30, 135-148.	1.4	24
5	Huntingtonâ€™s Disease Pathogenesis: Two Sequential Components. Journal of Huntington's Disease, 2021, 10, 35-51.	0.9	49
6	Timing and Impact of Psychiatric, Cognitive, and Motor Abnormalities in Huntington Disease. Neurology, 2021, 96, e2395-e2406.	1.5	53
7	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.	5.8	35
8	Association Analysis of Chromosome X to Identify Genetic Modifiers of Huntingtonâ€™s Disease. Journal of Huntington's Disease, 2021, 10, 367-375.	0.9	5
9	Huntingtonâ€™s disease: nearly four decades of human molecular genetics. Human Molecular Genetics, 2021, 30, R254-R263.	1.4	15
10	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.	1.1	19
11	A Multi-Omic Huntingtonâ€™s Disease Transgenic Sheep-Model Database for Investigating Disease Pathogenesis. Journal of Huntington's Disease, 2021, 10, 423-434.	0.9	6
12	C04â€...Protein coding tandem repeat in TCERG1 modifies huntingtonâ€™s disease onset. , 2021, , .		0
13	CNV profiles of Chinese pediatric patients with developmental disorders. Genetics in Medicine, 2021, 23, 669-678.	1.1	17
14	mTOR kinase inhibition disrupts neuregulin 1-ERBB3 autocrine signaling and sensitizes NF2-deficient meningioma cellular models to IGF1R inhibition. Journal of Biological Chemistry, 2021, 296, 100157.	1.6	8
15	Dystonia-specific mutations in THAP1 alter transcription of genes associated with neurodevelopment and myelin. American Journal of Human Genetics, 2021, 108, 2145-2158.	2.6	13
16	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.	1.8	28
17	Genetic Risk Underlying Psychiatric and Cognitive Symptoms in Huntingtonâ€™s Disease. Biological Psychiatry, 2020, 87, 857-865.	0.7	29
18	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.	1.4	48

#	Article	IF	Citations
19	Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. Molecular Autism, 2020, 11, 45.	2.6	11
20	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.	2.6	45
21	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.	1.8	32
22	CAG Repeat Not Polyglutamine Length Determines Timing of Huntingtonâ€™s Disease Onset. Cell, 2019, 178, 887-900.e14.	13.5	301
23	Full sequence of mutant huntingtin 3â€²-untranslated region and modulation of its gene regulatory activity by endogenous microRNA. Journal of Human Genetics, 2019, 64, 995-1004.	1.1	8
24	A rare case of acquired immunodeficiency associated with myelodysplastic syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e923.	0.6	1
25	Hypomorphic mutation of the mouse Huntingtonâ€™s disease gene orthologue. PLoS Genetics, 2019, 15, e1007765.	1.5	13
26	C10â€…Shared genetic liability between neuropsychiatric disorders and psychiatric symptoms in hd. , 2018, , .		0
27	CSIG-42. HIGH THROUGHPUT KINOME AND TRANSCRIPTOME ANALYSES REVEAL NOVEL THERAPEUTIC TARGETS IN NF2-DEFICIENT MENINGIOMA. Neuro-Oncology, 2018, 20, vi52-vi52.	0.6	0
28	Traditional and systems biology based drug discovery for the rare tumor syndrome neurofibromatosis type 2. PLoS ONE, 2018, 13, e0197350.	1.1	17
29	Opposing Tumor-Promoting and -Suppressive Functions of Rictor/mTORC2 Signaling in Adult Glioma and Pediatric SHH Medulloblastoma. Cell Reports, 2018, 24, 463-478.e5.	2.9	21
30	EPH receptor signaling as a novel therapeutic target in NF2-deficient meningioma. Neuro-Oncology, 2018, 20, 1185-1196.	0.6	22
31	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.	1.1	22
32	Genetic Modification of Huntington Disease Acts Early in the Prediagnosis Phase. American Journal of Human Genetics, 2018, 103, 349-357.	2.6	30
33	Population-specific genetic modification of Huntington's disease in Venezuela. PLoS Genetics, 2018, 14, e1007274.	1.5	27
34	Cover Image, Volume 173A, Number 2, February 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
35	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
36	Potential molecular consequences of transgene integration: The R6/2 mouse example. Scientific Reports, 2017, 7, 41120.	1.6	14

#	Article	IF	Citations
37	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.	1.4	37
38	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.	1.4	14
39	2016 William Allan Award: Human Disease Research: Genetic Cycling and Re-cycling 1. American Journal of Human Genetics, 2017, 100, 387-394.	2.6	0
40	A complex intragenic rearrangement of ERCC8 in Chinese siblings with Cockayne syndrome. Scientific Reports, 2017, 7, 44271.	1.6	7
41	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.	1.1	29
42	Haplotype-based stratification of Huntington's disease. European Journal of Human Genetics, 2017, 25, 1202-1209.	1.4	24
43	A novel microduplication of ARID1B: Clinical, genetic, and proteomic findings. American Journal of Medical Genetics, Part A, 2017, 173, 2478-2484.	0.7	7
44	WNT/Î²-Catenin Pathway and Epigenetic Mechanisms Regulate the Pitt-Hopkins Syndrome and Schizophrenia Risk Gene TCF4. Molecular Neuropsychiatry, 2017, 3, 53-71.	3.0	19
45	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.	3.3	78
46	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	9.4	251
47	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	0.7	40
48	A modifier of Huntington's disease onset at the MLH1 locus. Human Molecular Genetics, 2017, 26, 3859-3867.	1.4	88
49	Large-scale phenome analysis defines a behavioral signature for Huntington's disease genotype in mice. Nature Biotechnology, 2016, 34, 838-844.	9.4	46
50	Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. European Journal of Human Genetics, 2016, 24, 1622-1626.	1.4	12
51	CSF1Rmosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. Brain, 2016, 139, 1666-1672.	3.7	53
52	Permanent inactivation of Huntington's disease mutation by personalized allele-specific CRISPR/Cas9. Human Molecular Genetics, 2016, 25, ddw286.	1.4	195
53	Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. American Journal of Human Genetics, 2016, 99, 1015-1033.	2.6	53
54	Metabolic disruption identified in the Huntingtonâ€™s disease transgenic sheep model. Scientific Reports, 2016, 6, 20681.	1.6	52

#	Article	IF	Citations
55	A novel neurodevelopmental disorder associated with compound heterozygous variants in the huntingtin gene. European Journal of Human Genetics, 2016, 24, 1826-1827.	1.4	45
56	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.	2.6	129
57	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. Nature Neuroscience, 2016, 19, 517-522.	7.1	72
58	Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis. Human Molecular Genetics, 2016, 25, 1255-1270.	1.4	30
59	Clinical-Genetic Associations in the Prospective Huntington at Risk Observational Study (PHAROS). JAMA Neurology, 2016, 73, 102.	4.5	38
60	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.	0.9	30
61	Prevalence of Huntington's disease gene CAG trinucleotide repeat alleles in patients with bipolar disorder. Bipolar Disorders, 2015, 17, 403-408.	1.1	6
62	RNA Sequence Analysis of Human Huntington Disease Brain Reveals an Extensive Increase in Inflammatory and Developmental Gene Expression. PLoS ONE, 2015, 10, e0143563.	1.1	150
63	Htt CAG repeat expansion confers pleiotropic gains of mutant huntingtin function in chromatin regulation. Human Molecular Genetics, 2015, 24, 2442-2457.	1.4	53
64	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. American Journal of Human Genetics, 2015, 97, 170-176.	2.6	45
65	Heritability of Risk for Sudden Cardiac Arrest in ESRD. Journal of the American Society of Nephrology: JASN, 2015, 26, 2815-2820.	3.0	9
66	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.	0.7	114
67	Huntington disease. Nature Reviews Disease Primers, 2015, 1, 15005.	18.1	1,031
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	Expanding the Spectrum of Founder Mutations Causing Isolated Gonadotropin-Releasing Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1378-E1385.	1.8	22
70	Sequence-Level Analysis of the Major European Huntington Disease Haplotype. American Journal of Human Genetics, 2015, 97, 435-444.	2.6	22
71	Mediator Subunit Med28 Is Essential for Mouse Peri-Implantation Development and Pluripotency. PLoS ONE, 2015, 10, e0140192.	1.1	19
72	A high-throughput kinome screen reveals serum/glucocorticoid-regulated kinase 1 as a therapeutic target for NF2-deficient meningiomas. Oncotarget, 2015, 6, 16981-16997.	0.8	46

#	Article	IF	Citations
73	Kinome Screen Reveals SGK1 as a Therapeutic Target for NF2: Inhibition of mTORC1/2 is More Effective than Rapamycin. FASEB Journal, 2015, 29, 889.4.	0.2	0
74	MicroRNAs Located in the Hox Gene Clusters Are Implicated in Huntington's Disease Pathogenesis. PLoS Genetics, 2014, 10, e1004188.	1.5	97
75	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.	3.3	74
76	Describing Sequencing Results of Structural Chromosome Rearrangements with a Suggested Next-Generation Cytogenetic Nomenclature. American Journal of Human Genetics, 2014, 94, 695-709.	2.6	42
77	Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. American Journal of Human Genetics, 2014, 95, 454-461.	2.6	45
78	Genetic modifiers of Huntington's disease. Movement Disorders, 2014, 29, 1359-1365.	2.2	116
79	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.	3.3	297
80	Transcriptional Consequences of 16p11.2 Deletion and Duplication in Mouse Cortex and Multiplex Autism Families. American Journal of Human Genetics, 2014, 94, 870-883.	2.6	116
81	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.	1.4	42
82	Huntingtin Supplies a csaA-Independent Function Essential for EDTA-Resistant Homotypic Cell Adhesion in Dictyostelium discoideum. Journal of Huntington's Disease, 2014, 3, 261-271.	0.9	6
83	HD CAGnome: A Search Tool for Huntingtin CAG Repeat Length-Correlated Genes. PLoS ONE, 2014, 9, e95556.	1.1	3
84	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntingtonâ€™s disease motor onset. Neurogenetics, 2013, 14, 173-179.	0.7	10
85	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.	0.9	47
86	Regulation of mTOR Complex 2 Signaling in Neurofibromatosis 2â€“Deficient Target Cell Types. Molecular Cancer Research, 2012, 10, 649-659.	1.5	96
87	Clinical Diagnosis by Whole-Genome Sequencing of a Prenatal Sample. New England Journal of Medicine, 2012, 367, 2226-2232.	13.9	174
88	Assessment of cortical and striatal involvement in 523 Huntington disease brains. Neurology, 2012, 79, 1708-1715.	1.5	52
89	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nature Genetics, 2012, 44, 390-397.	9.4	229
90	Population stratification may bias analysis of PGC-1Î± as a modifier of age at Huntington disease motor onset. Human Genetics, 2012, 131, 1833-1840.	1.8	26

#	Article	IF	Citations
91	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. Nature, 2012, 485, 363-367.	13.7	363
92	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.	1.0	20
93	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. Cell, 2012, 149, 525-537.	13.5	534
94	Common SNP-Based Haplotype Analysis of the 4p16.3 Huntington Disease Gene Region. American Journal of Human Genetics, 2012, 90, 434-444.	2.6	60
95	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.	2.6	154
96	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.	2.6	195
97	HD CAG-correlated gene expression changes support a simple dominant gain of function. Human Molecular Genetics, 2011, 20, 2846-2860.	1.4	67
98	Meclizine is neuroprotective in models of Huntington's disease. Human Molecular Genetics, 2011, 20, 294-300.	1.4	45
99	Differential effects of the Huntington's disease CAG mutation in striatum and cerebellum are quantitative not qualitative. Human Molecular Genetics, 2011, 20, 4258-4267.	1.4	23
100	Deficiency of Huntingtin Has Pleiotropic Effects in the Social Amoeba Dictyostelium discoideum. PLoS Genetics, 2011, 7, e1002052.	1.5	52
101	Monozygotic twins discordant for neurofibromatosis 1. American Journal of Medical Genetics, Part A, 2010, 152A, 601-606.	0.7	40
102	Huntingtin facilitates polycomb repressive complex 2. Human Molecular Genetics, 2010, 19, 573-583.	1.4	169
103	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.	1.1	264
104	Changing Models of Biomedical Research. Science Translational Medicine, 2009, 1, 1cm1.	5.8	11
105	Huntington's disease: the case for genetic modifiers. Genome Medicine, 2009, 1, 80.	3.6	104
106	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. American Journal of Human Genetics, 2008, 82, 712-722.	2.6	95
107	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.	2.1	42
108	Genetic criteria for Huntington's disease pathogenesis. Brain Research Bulletin, 2007, 72, 78-82.	1.4	12

#	Article	IF	Citations
109	Expanding the Notion of Disease in Huntingtonâ€™s Disease. Biological Psychiatry, 2007, 62, 1340.	0.7	4
110	Candidate loci for Zimmermannâ€“Laband syndrome at 3p14.3. American Journal of Medical Genetics, Part A, 2007, 143A, 107-111.	0.7	17
111	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.	0.7	12
112	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
113	Huntington's disease: seeing the pathogenic process through a genetic lens. Trends in Biochemical Sciences, 2006, 31, 533-540.	3.7	128
114	Brain-derived neurotrophic factor does not influence age at neurologic onset of Huntingtonâ€™s disease. Neurobiology of Disease, 2006, 24, 280-285.	2.1	31
115	Genetic analysis of the GRIK2 modifier effect in Huntington's disease. BMC Neuroscience, 2006, 7, 62.	0.8	16
116	Understanding the role of the merlin interacting protein magicin as part of the mammalian Mediator complex. FASEB Journal, 2006, 20, A79.	0.2	0
117	Magicin (MED28), a Potential Adaptor Protein. FASEB Journal, 2006, 20, A103.	0.2	1
118	Reversal of a full-length mutant huntingtin neuronal cell phenotype by chemical inhibitors of polyglutamine-mediated aggregation. BMC Neuroscience, 2005, 6, 1.	0.8	133
119	HD CAG repeat implicates a dominant property of huntingtin in mitochondrial energy metabolism. Human Molecular Genetics, 2005, 14, 2871-2880.	1.4	274
120	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.	2.6	178
121	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.	3.3	666
122	Magicin, a novel cytoskeletal protein associates with the NF2 tumor suppressor merlin and Grb2. Oncogene, 2004, 23, 8815-8825.	2.6	66
123	Modified Single-Stranded Oligonucleotides Inhibit Aggregate Formation and Toxicity Induced by Expanded Polyglutamine. Journal of Molecular Neuroscience, 2004, 24, 257-268.	1.1	16
124	Evidence for a modifier of onset age in Huntington disease linked to the HD gene in 4p16. Neurogenetics, 2004, 5, 109-114.	0.7	67
125	Huntington's Disease. NeuroMolecular Medicine, 2003, 4, 7-20.	1.8	79
126	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.	2.6	148

#	Article	IF	Citations
127	Specific progressive cAMP reduction implicates energy deficit in presymptomatic Huntington's disease knock-in mice. Human Molecular Genetics, 2003, 12, 497-508.	1.4	250
128	Rescue of a human mRNA splicing defect by the plant cytokinin kinetin. Human Molecular Genetics, 2003, 13, 429-436.	1.4	139
129	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.	1.4	162
130	Identification of a presymptomatic molecular phenotype in Hdh CAG knock-in mice. Human Molecular Genetics, 2002, 11, 2233-2241.	1.4	52
131	Familial dysautonomia. Current Opinion in Genetics and Development, 2002, 12, 307-311.	1.5	332
132	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.	0.8	130
133	No post-genetics era in human disease research. Nature Reviews Genetics, 2002, 3, 72-79.	7.7	27
134	Tissue-Specific Expression of a Splicing Mutation in the Gene Causes Familial Dysautonomia. American Journal of Human Genetics, 2001, 68, 598-605.	2.6	558
135	Quantitative neuropathological changes in presymptomatic Huntington's disease. Annals of Neurology, 2001, 49, 29-34.	2.8	163
136	Cloning, Characterization, and Genomic Structure of the Mouse Ikbkap Gene. DNA and Cell Biology, 2001, 20, 579-586.	0.9	24
137	Molecular genetics: Unmasking polyglutamine triggers in neurodegenerative disease. Nature Reviews Neuroscience, 2000, 1, 109-115.	4.9	383
138	Cloning, mapping, and expression of a novel brain-specific transcript in the Familial Dysautonomia candidate region on Chromosome 9q31. Mammalian Genome, 2000, 11, 81-83.	1.0	5
139	Clonal Analysis of a Case of Multiple Meningiomas Using Multiple Molecular Genetic Approaches: Pathology Case Report. Neurosurgery, 1999, 45, 409-416.	0.6	56
140	Allelic expression of the NF2 gene in neurofibromatosis 2 and schwannomatosis. Neurogenetics, 1999, 2, 101-108.	0.7	25
141	Merlin: the neurofibromatosis 2 tumor suppressor. Biochimica Et Biophysica Acta: Reviews on Cancer, 1999, 1423, M29-M36.	3.3	45
142	Precise Genetic Mapping and Haplotype Analysis of the Familial Dysautonomia Gene on Human Chromosome 9q31. American Journal of Human Genetics, 1999, 64, 1110-1118.	2.6	69
143	Mapping of the Mucolipidosis Type IV Gene to Chromosome 19p and Definition of Founder Haplotypes. American Journal of Human Genetics, 1999, 65, 773-778.	2.6	87
144	Mutant Huntingtin Forms in Vivo Complexes with Distinct Context-Dependent Conformations of the Polyglutamine Segment. Neurobiology of Disease, 1999, 6, 364-375.	2.1	57

#	Article	IF	Citations
145	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
146	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
147	The FERM domain: a unique module involved in the linkage of cytoplasmic proteins to the membrane. Trends in Biochemical Sciences, 1998, 23, 281-282.	3.7	494
148	Huntingtin: a single bait hooks many species. Current Opinion in Neurobiology, 1998, 8, 425-430.	2.0	70
149	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.	1.6	229
150	The Genetic Defect Causing Huntingtonâ€™s Disease: Repeated in Other Contexts?. Molecular Medicine, 1997, 3, 238-246.	1.9	56
151	Heterogeneous Topographic and Cellular Distribution of Huntingtin Expression in the Normal Human Neostriatum. Journal of Neuroscience, 1997, 17, 3052-3063.	1.7	143
152	Frequent loss of chromosome 14 in atypical and malignant meningioma: identification of a putative `tumor progression' locus. Oncogene, 1997, 14, 611-616.	2.6	109
153	The early-onset torsion dystonia gene (DYT1) encodes an ATP-binding protein. Nature Genetics, 1997, 17, 40-48.	9.4	1,051
154	Huntingtin is required for neurogenesis and is not impaired by the Huntington's disease CAG expansion. Nature Genetics, 1997, 17, 404-410.	9.4	472
155	Exon trapping and sequence-based methods of gene finding in transcript mapping of human 4p 16.3. Somatic Cell and Molecular Genetics, 1997, 23, 413-427.	0.7	4
156	CAG repeat number governs the development rate of pathology in Huntington's disease. Annals of Neurology, 1997, 41, 689-692.	2.8	605
157	No Association between ?1-Antichymotrypsin and Familial Alzheimer's Diseases. Annals of the New York Academy of Sciences, 1996, 802, 35-41.	1.8	12
158	Differential Expression of Normal and Mutant Huntington's Disease Gene Alleles. Neurobiology of Disease, 1996, 3, 183-190.	2.1	77
159	Frequency and distribution ofNF2 mutations in schwannomas. , 1996, 17, 45-55.		134
160	TRINUCLEOTIDE INSTABILITY: A Repeating Theme in Human Inherited Disorders. Annual Review of Medicine, 1996, 47, 201-209.	5.0	73
161	Evidence for Subarachnoid Spread in the Development of Multiple Meningiomas. Brain Pathology, 1995, 5, 11-14.	2.1	33
162	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

#	Article	IF	Citations
163	Huntington's disease gene: Regional and cellular expression in brain of normal and affected individuals. Annals of Neurology, 1995, 37, 218-230.	2.8	206
164	Prenatal diagnostic testing for familial dysautonomia using linked genetic markers. Prenatal Diagnosis, 1995, 15, 817-826.	1.1	11
165	Normal and Expanded Huntingtonâ€™s Disease Gene Alleles Produce Distinguishable Proteins Due to Translation Across the CAG Repeat. Molecular Medicine, 1995, 1, 374-383.	1.9	97
166	Isolation of a novel gene underlying batten disease, CLN3. Cell, 1995, 82, 949-957.	13.5	554
167	Exon scanning for mutation of the NF2 gene in schwannomas. Human Molecular Genetics, 1994, 3, 413-419.	1.4	200
168	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
169	Mutations in transcript isoforms of the neurofibromatosis 2 gene in multiple human tumour types. Nature Genetics, 1994, 6, 185-192.	9.4	236
170	The gene for achondroplasia maps to the telomeric region of chromosome 4p. Nature Genetics, 1994, 6, 314-317.	9.4	116
171	High resolution localization of recombination hot spots using sperm typing. Nature Genetics, 1994, 7, 420-424.	9.4	80
172	Huntington's disease CAG trinucleotide repeats in pathologically confirmed post-mortem brains. Neurobiology of Disease, 1994, 1, 159-166.	2.1	77
173	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
174	Neurofibromatosis type 1 gene mutations in neuroblastoma. Nature Genetics, 1993, 3, 62-66.	9.4	157
175	Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. Nature Genetics, 1993, 4, 160-164.	9.4	149
176	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.	9.4	102
177	Relationship between trinucleotide repeat expansion and phenotypic variation in Huntington's disease. Nature Genetics, 1993, 4, 393-397.	9.4	672
178	Discrepancy resolved. Nature Genetics, 1993, 5, 215-215.	9.4	5
179	A linkage study with DNA markers (D4S95, D4S115, and D4S111) in Japanese Huntington disease families. Japanese Journal of Human Genetics, 1993, 38, 193-201.	0.8	1
180	Hunting for Huntington's Disease. , 1993, 3, 139-158.		24

#	Article	IF	Citations
181	The Huntington's disease candidate region exhibits many different haplotypes. Nature Genetics, 1992, 1, 99-103.	9.4	157
182	A 15-bp deletion in exon 5 of the ornithine aminotransferase (OAT) locus associated with gyrate atrophy. Human Mutation, 1992, 1, 293-297.	1.1	6
183	Linkage analysis in juvenile neuronal ceroid lipofuscinosis. American Journal of Medical Genetics Part A, 1992, 42, 542-545.	2.4	10
184	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
185	Dystonia gene in Ashkenazi Jewish population is located on chromosome 9q32-34. Annals of Neurology, 1990, 27, 114-120.	2.8	141
186	A comparison of neurological, metabolic, structural, and genetic evaluations in persons at risk for Huntington's disease. Annals of Neurology, 1990, 28, 614-621.	2.8	110
187	Hyperkalemic periodic paralysis and the adult muscle sodium channel alpha-subunit gene. Science, 1990, 250, 1000-1002.	6.0	332
188	Location cloning strategy for characterizing genetic defects in Huntington's disease and Alzheimer's disease. FASEB Journal, 1989, 3, 2036-2041.	0.2	21
189	The role of mitochondrial DNA in Huntington's disease. Journal of Molecular Neuroscience, 1989, 1, 129-136.	1.1	9
190	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
191	Human gene for torsion dystonia located on chromosome 9q32-q34. Neuron, 1989, 2, 1427-1434.	3.8	246
192	Partial linkage map of chromosome 13q in the region of the Wilson disease and retinoblastoma genes. Genetic Epidemiology, 1988, 5, 375-380.	0.6	10
193	Protease inhibitor domain encoded by an amyloid protein precursor mRNA associated with Alzheimer's disease. Nature, 1988, 331, 528-530.	13.7	1,105
194	Irradiation-reduced human chromosome 21 hybrids. Somatic Cell and Molecular Genetics, 1988, 14, 233-242.	0.7	38
195	_{Predictive Testing for Huntingtons Disease with Use of a Linked DNA Marker}. New England Journal of Medicine, 1988, 318, 535-542.	13.9	167
196	Homozygotes for Huntington's disease. Nature, 1987, 326, 194-197.	13.7	331
197	The genetic defect in familial Alzheimer's disease is not tightly linked to the amyloid Î²-protein gene. Nature, 1987, 329, 156-157.	13.7	275
198	Genetic linkage of bilateral acoustic neurofibromatosis to a DNA marker on chromosome 22. Nature, 1987, 329, 246-248.	13.7	478

#	Article	IF	Citations
199	Probes in Huntington's chorea (reply). Nature, 1986, 320, 21-22.	13.7	15
200	Loss of genes on chromosome 22 in tumorigenesis of human acoustic neuroma. Nature, 1986, 322, 644-647.	13.7	490
201	Accuracy of testing for Huntington's disease. Nature, 1986, 323, 118-118.	13.7	3
202	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.	0.6	24
203	Huntingtons Disease. New England Journal of Medicine, 1986, 315, 1267-1276.	13.9	532
204	Deletion of Huntington's disease-linked G8 (D4S10) locus in Wolfâ€“Hirschhorn syndrome. Nature, 1985, 318, 75-78.	13.7	114
205	Huntington's disease: two families with differing clinical features show linkage to the G8 probe. Science, 1985, 229, 776-779.	6.0	75
206	Isolation of polymorphic DNA segments from human chromosome 21. Nucleic Acids Research, 1985, 13, 6075-6088.	6.5	73
207	Linkage Map on Chromosome 21q and the Association of a DNA Haplotype with a Propensity to Nondisjunction and Trisomy 21. Annals of the New York Academy of Sciences, 1985, 450, 95-107.	1.8	5
208	Genetic Linkage of the Huntingtonâ€™s Disease Gene to a DNA Marker. Canadian Journal of Neurological Sciences, 1984, 11, 421-425.	0.3	10
209	Use of cyclosporin a in establishing epstein-barr virus-transformed human lymphoblastoid cell lines. In Vitro, 1984, 20, 856-858.	1.2	344
210	A polymorphic DNA marker genetically linked to Huntington's disease. Nature, 1983, 306, 234-238.	13.7	2,289