James F Gusella

List of Publications by Year in Descending Order

210 papers	21,951 citations	74 h-index	146 g-index
240 ext. papers	24,455 ext. citations	13.1 avg, IF	5.99 L-index

#	Paper	IF	Citations
210	Genetic modifiers of Huntington disease differentially influence motor and cognitive domains.. *American Journal of Human Genetics*, **2022**,	11	2
209	Exome sequencing of individuals with Huntington's disease implicates FAN1 nuclease activity in slowing CAG expansion and disease onset.. *Nature Neuroscience*, **2022**,	25.5	3
208	Dystonia-specific mutations in THAP1 alter transcription of genes associated with neurodevelopment and myelin. *American Journal of Human Genetics*, **2021**, 108, 2145-2158	11	1
207	Timing and Impact of Psychiatric, Cognitive, and Motor Abnormalities in Huntington Disease. *Neurology*, **2021**, 96, e2395-e2406	6.5	11
206	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	17.4	5
205	Association Analysis of Chromosome X to Identify Genetic Modifiers of Huntington's Disease. *Journal of Huntingtonks Disease*, **2021**, 10, 367-375	1.9	1
204	Huntington's disease: nearly four decades of human molecular genetics. *Human Molecular Genetics*, **2021**, 30, R254-R263	5.6	1
203	Mutations causing Lopes-Maciel-Rodan syndrome are huntingtin hypomorphs. *Human Molecular Genetics*, **2021**, 30, 135-148	5.6	2
202	Huntington's Disease Pathogenesis: Two Sequential Components. *Journal of Huntingtonks Disease*, **2021**, 10, 35-51	1.9	16
201	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	3.7	5
200	A Multi-Omic Huntington's Disease Transgenic Sheep-Model Database for Investigating Disease Pathogenesis. *Journal of Huntingtonks Disease*, **2021**, 10, 423-434	1.9	1
199	CNV profiles of Chinese pediatric patients with developmental disorders. *Genetics in Medicine*, **2021** , 23, 669-678	8.1	7
198	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	5.4	1
197	Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. *Molecular Autism*, **2020**, 11, 45	6.5	3
196	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	11	21
195	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	6.3	17
194	Genetic Risk Underlying Psychiatric and Cognitive Symptoms in Huntington's Disease. *Biological Psychiatry*, **2020**, 87, 857-865	7.9	13

193	Promotion of somatic CAG repeat expansion by Fan1 knock-out in Huntington's disease knock-in mice is blocked by Mlh1 knock-out. *Human Molecular Genetics*, **2020**, 29, 3044-3053	5.6	22
192	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,	5.6	11
191	Hypomorphic mutation of the mouse Huntington's disease gene orthologue. *PLoS Genetics*, **2019**, 15, e1007765	6	6
190	CAG Repeat Not Polyglutamine Length Determines Timing of Huntington's Disease Onset. *Cell*, **2019**, 178, 887-900.e14	56.2	155
189	Full sequence of mutant huntingtin 3'-untranslated region and modulation of its gene regulatory activity by endogenous microRNA. *Journal of Human Genetics*, **2019**, 64, 995-1004	4.3	3
188	A rare case of acquired immunodeficiency associated with myelodysplastic syndrome. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e923	2.3	1
187	Traditional and systems biology based drug discovery for the rare tumor syndrome neurofibromatosis type 2. *PLoS ONE*, **2018**, 13, e0197350	3.7	10
186	Opposing Tumor-Promoting and -Suppressive Functions of Rictor/mTORC2 Signaling in Adult Glioma and Pediatric SHH Medulloblastoma. *Cell Reports*, **2018**, 24, 463-478.e5	10.6	11
185	EPH receptor signaling as a novel therapeutic target in NF2-deficient meningioma. *Neuro-Oncology*, **2018**, 20, 1185-1196	1	14
184	A rare exonic NRXN3 deletion segregating with neurodevelopmental and neuropsychiatric conditions in a three-generation Chinese family. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 589-595	3.5	11
183	Genetic Modification of Huntington Disease Acts Early in the Prediagnosis Phase. *American Journal of Human Genetics*, **2018**, 103, 349-357	11	22
182	Population-specific genetic modification of Huntington's disease in Venezuela. *PLoS Genetics*, **2018**, 14, e1007274	6	18
181	CSIG-42. HIGH THROUGHPUT KINOME AND TRANSCRIPTOME ANALYSES REVEAL NOVEL THERAPEUTIC TARGETS IN NF2-DEFICIENT MENINGIOMA. *Neuro-Oncology*, **2018**, 20, vi52-vi52	1	78
180	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. *Nature Genetics*, **2017**, 49, 238-248	36.3	88
179	Potential molecular consequences of transgene integration: The R6/2 mouse example. *Scientific Reports*, **2017**, 7, 41120	4.9	8
178	High resolution time-course mapping of early transcriptomic, molecular and cellular phenotypes in Huntington's disease CAG knock-in mice across multiple genetic backgrounds. *Human Molecular Genetics*, **2017**, 26, 913-922	5.6	27
177	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	5.6	10
176	2016 William Allan Award: Human Disease Research: Genetic Cycling and Re-cycling. *American Journal of Human Genetics*, **2017**, 100, 387-394	11	

175	A complex intragenic rearrangement of ERCC8 in Chinese siblings with Cockayne syndrome. *Scientific Reports*, **2017**, 7, 44271	4.9	5
174	Rare Deleterious PARD3 Variants 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	4.7	17
173	Haplotype-based stratification of Huntington's disease. *European Journal of Human Genetics*, **2017**, 25, 1202-1209	5.3	14
172	A novel microduplication of ARID1B: Clinical, genetic, and proteomic findings. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2478-2484	2.5	5
171	WNT/β-Catenin Pathway and Epigenetic Mechanisms Regulate the Pitt-Hopkins Syndrome and Schizophrenia Risk Gene. *Molecular Neuropsychiatry*, **2017**, 3, 53-71	4.9	14
170	Brain urea increase is an early Huntington'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	11.5	43
169	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. *Nature Genetics*, **2017**, 49, 36-45	36.3	172
168	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. *American Journal of Medical Genetics, Part A*, **2017**, 173, 395-406	2.5	22
167	A modifier of Huntington's disease onset at the MLH1 locus. *Human Molecular Genetics*, **2017**, 26, 3859-3867	5.6	59
166	Permanent inactivation of Huntington's disease mutation by personalized allele-specific CRISPR/Cas9. *Human Molecular Genetics*, **2016**, 25, 4566-4576	5.6	150
165	Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. *American Journal of Human Genetics*, **2016**, 99, 1015-1033	11	43
164	Metabolic disruption identified in the Huntington's disease transgenic sheep model. *Scientific Reports*, **2016**, 6, 20681	4.9	35
163	A novel neurodevelopmental disorder associated with compound heterozygous variants in the huntingtin gene. *European Journal of Human Genetics*, **2016**, 24, 1826-1827	5.3	23
162	The HTT CAG-Expansion Mutation Determines Age at Death but Not Disease Duration in Huntington Disease. *American Journal of Human Genetics*, **2016**, 98, 287-98	11	92
161	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. *Nature Neuroscience*, **2016**, 19, 517-22	25.5	39
160	Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis *Human Molecular Genetics*, **2016**, 25, 1255-70	5.6	22
159	Clinical-Genetic Associations in the Prospective Huntington at Risk Observational Study (PHAROS): Implications for Clinical Trials. *JAMA Neurology*, **2016**, 73, 102-10	17.2	29
158	Large-scale phenome analysis defines a behavioral signature for Huntington's disease genotype in mice. *Nature Biotechnology*, **2016**, 34, 838-44	44.5	30

157	Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. *European Journal of Human Genetics*, **2016**, 24, 1622-1626	5.3	12
156	CSF1R mosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. *Brain*, **2016**, 139, 1666-72	11.2	38
155	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. *American Journal of Human Genetics*, **2015**, 97, 170-6	11	37
154	Heritability of Risk for Sudden Cardiac Arrest in ESRD. *Journal of the American Society of Nephrology: JASN*, **2015**, 26, 2815-20	12.7	7
153	miR-10b-5p expression in Huntington's disease brain relates to age of onset and the extent of striatal involvement. *BMC Medical Genomics*, **2015**, 8, 10	3.7	76
152	Huntington disease. *Nature Reviews Disease Primers*, **2015**, 1, 15005	51.1	672
151	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. *American Journal of Human Genetics*, **2015**, 96, 784-96	11	35
150	Expanding the Spectrum of Founder Mutations Causing Isolated Gonadotropin-Releasing Hormone Deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E1378-85	5.6	16
149	Sequence-Level Analysis of the Major European Huntington Disease Haplotype. *American Journal of Human Genetics*, **2015**, 97, 435-44	11	14
148	The Genetic Modifiers of Motor OnsetAge (GeM MOA) Website: Genome-wide Association Analysis for Genetic Modifiers of Huntington's Disease. *Journal of Huntingtonks Disease*, **2015**, 4, 279-84	1.9	20
147	Prevalence of Huntington's disease gene CAG trinucleotide repeat alleles in patients with bipolar disorder. *Bipolar Disorders*, **2015**, 17, 403-8	3.8	5
146	RNA Sequence Analysis of Human Huntington Disease Brain Reveals an Extensive Increase in Inflammatory and Developmental Gene Expression. *PLoS ONE*, **2015**, 10, e0143563	3.7	99
145	Htt CAG repeat expansion confers pleiotropic gains of mutant huntingtin function in chromatin regulation. *Human Molecular Genetics*, **2015**, 24, 2442-57	5.6	38
144	Mediator Subunit Med28 Is Essential for Mouse Peri-Implantation Development and Pluripotency. *PLoS ONE*, **2015**, 10, e0140192	3.7	6
143	A high-throughput kinome screen reveals serum/glucocorticoid-regulated kinase 1 as a therapeutic target for NF2-deficient meningiomas. *Oncotarget*, **2015**, 6, 16981-97	3.3	35
142	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	0.9	
141	Describing sequencing results of structural chromosome rearrangements with a suggested next-generation cytogenetic nomenclature. *American Journal of Human Genetics*, **2014**, 94, 695-709	11	40
140	Cryptic and complex chromosomal aberrations in early-onset neuropsychiatric disorders. *American Journal of Human Genetics*, **2014**, 95, 454-61	11	37

139	Genetic modifiers of Huntington's disease. *Movement Disorders*, **2014**, 29, 1359-65	7	84
138	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	11.5	210
137	Transcriptional consequences of 16p11.2 deletion and duplication in mouse cortex and multiplex autism families. *American Journal of Human Genetics*, **2014**, 94, 870-83	11	78
136	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	5.3	29
135	Huntingtin Supplies a csaA-Independent Function Essential for EDTA-Resistant Homotypic Cell Adhesion in Dictyostelium discoideum. *Journal of Huntingtonks Disease*, **2014**, 3, 261-71	1.9	6
134	MicroRNAs located in the Hox gene clusters are implicated in huntington's disease pathogenesis. *PLoS Genetics*, **2014**, 10, e1004188	6	73
133	Functionally compromised CHD7 alleles in patients with isolated GnRH deficiency. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 17953-8	11.5	51
132	HD CAGnome: a search tool for huntingtin CAG repeat length-correlated genes. *PLoS ONE*, **2014**, 9, e95556	3.7	1
131	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntington's disease motor onset. *Neurogenetics*, **2013**, 14, 173-9	3	9
130	Further molecular characterisation of the OVT73 transgenic sheep model of Huntington's disease identifies cortical aggregates. *Journal of Huntingtonks Disease*, **2013**, 2, 279-95	1.9	38
129	Clinical diagnosis by whole-genome sequencing of a prenatal sample. *New England Journal of Medicine*, **2012**, 367, 2226-32	59.2	144
128	Assessment of cortical and striatal involvement in 523 Huntington disease brains. *Neurology*, **2012**, 79, 1708-15	6.5	48
127	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. *Nature Genetics*, **2012**, 44, 390-7, S1	36.3	190
126	Population stratification may bias analysis of PGC-1 as a modifier of age at Huntington disease motor onset. *Human Genetics*, **2012**, 131, 1833-40	6.3	25
125	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. *Nature*, **2012**, 485, 363-7	50.4	281
124	TAA repeat variation in the GRIK2 gene does not influence age at onset in Huntington's disease. *Biochemical and Biophysical Research Communications*, **2012**, 424, 404-8	3.4	17
123	Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. *Cell*, **2012**, 149, 525-37	56.2	441
122	Common SNP-based haplotype analysis of the 4p16.3 Huntington disease gene region. *American Journal of Human Genetics*, **2012**, 90, 434-44	11	48

121	Regulation of mTOR complex 2 signaling in neurofibromatosis 2-deficient target cell types. *Molecular Cancer Research*, **2012**, 10, 649-59	6.6	74
120	Next-generation sequencing strategies enable routine detection of balanced chromosome rearrangements for clinical diagnostics and genetic research. *American Journal of Human Genetics*, **2011**, 88, 469-81	11	132
119	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, 551-63	11	166
118	HD CAG-correlated gene expression changes support a simple dominant gain of function. *Human Molecular Genetics*, **2011**, 20, 2846-60	5.6	59
117	Meclizine is neuroprotective in models of Huntington's disease. *Human Molecular Genetics*, **2011**, 20, 294-300	5.6	36
116	Differential effects of the Huntington's disease CAG mutation in striatum and cerebellum are quantitative not qualitative. *Human Molecular Genetics*, **2011**, 20, 4258-67	5.6	20
115	Deficiency of huntingtin has pleiotropic effects in the social amoeba Dictyostelium discoideum. *PLoS Genetics*, **2011**, 7, e1002052	6	44
114	Huntingtin facilitates polycomb repressive complex 2. *Human Molecular Genetics*, **2010**, 19, 573-83	5.6	128
113	Monozygotic twins discordant for neurofibromatosis 1. *American Journal of Medical Genetics, Part A* , **2010**, 152A, 601-6	2.5	35
112	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, 4250-61	4.8	202
111	Changing models of biomedical research. *Science Translational Medicine*, **2009**, 1, 1cm1	17.5	6
110	Huntington's disease: the case for genetic modifiers. *Genome Medicine*, **2009**, 1, 80	14.4	87
109	Modeling NF2 with human arachnoidal and meningioma cell culture systems: NF2 silencing reflects the benign character of tumor growth. *Neurobiology of Disease*, **2008**, 29, 278-92	7.5	35
108	Characterization of apparently balanced chromosomal rearrangements from the developmental genome anatomy project. *American Journal of Human Genetics*, **2008**, 82, 712-22	11	84
107	Expanding the notion of disease in Huntington's disease. *Biological Psychiatry*, **2007**, 62, 1340	7.9	4
106	Candidate loci for Zimmermann-Laband syndrome at 3p14.3. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 107-11	2.5	15
105	Extensive molecular genetic analysis of the 3p14.3 region in patients with Zimmermann-Laband syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2668-74	2.5	11
104	Genetic criteria for Huntington's disease pathogenesis. *Brain Research Bulletin*, **2007**, 72, 78-82	3.9	11

103	Huntington's disease: seeing the pathogenic process through a genetic lens. *Trends in Biochemical Sciences*, **2006**, 31, 533-40	10.3	107
102	Brain-derived neurotrophic factor does not influence age at neurologic onset of Huntington's disease. *Neurobiology of Disease*, **2006**, 24, 280-5	7.5	29
101	Genetic analysis of the GRIK2 modifier effect in Huntington's disease. *BMC Neuroscience*, **2006**, 7, 62	3.2	15
100	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	62
99	Understanding the role of the merlin interacting protein magicin as part of the mammalian Mediator complex. *FASEB Journal*, **2006**, 20, A79	0.9	
98	Magicin (MED28), a Potential Adaptor Protein. *FASEB Journal*, **2006**, 20, A103	0.9	1
97	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	11	158
96	Reversal of a full-length mutant huntingtin neuronal cell phenotype by chemical inhibitors of polyglutamine-mediated aggregation. *BMC Neuroscience*, **2005**, 6, 1	3.2	102
95	HD CAG repeat implicates a dominant property of huntingtin in mitochondrial energy metabolism. *Human Molecular Genetics*, **2005**, 14, 2871-80	5.6	246
94	Rescue of a human mRNA splicing defect by the plant cytokinin kinetin. *Human Molecular Genetics*, **2004**, 13, 429-36	5.6	114
93	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, 3498-503	11.5	572
92	Magicin, a novel cytoskeletal protein associates with the NF2 tumor suppressor merlin and Grb2. *Oncogene*, **2004**, 23, 8815-25	9.2	56
91	Modified single-stranded oligonucleotides inhibit aggregate formation and toxicity induced by expanded polyglutamine. *Journal of Molecular Neuroscience*, **2004**, 24, 257-67	3.3	12
90	Evidence for a modifier of onset age in Huntington disease linked to the HD gene in 4p16. *Neurogenetics*, **2004**, 5, 109-14	3	63
89	Specific progressive cAMP reduction implicates energy deficit in presymptomatic Huntington's disease knock-in mice. *Human Molecular Genetics*, **2003**, 12, 497-508	5.6	218
88	Huntington's disease. *NeuroMolecular Medicine*, **2003**, 4, 7-20	4.6	61
87	A genome scan for modifiers of age at onset in Huntington disease: The HD MAPS study. *American Journal of Human Genetics*, **2003**, 73, 682-7	11	131
86	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	3.2	109

85	No post-genetics era in human disease research. *Nature Reviews Genetics*, **2002**, 3, 72-9	30.1	25
84	Early phenotypes that presage late-onset neurodegenerative disease allow testing of modifiers in Hdh CAG knock-in mice. *Human Molecular Genetics*, **2002**, 11, 633-40	5.6	134
83	Identification of a presymptomatic molecular phenotype in Hdh CAG knock-in mice. *Human Molecular Genetics*, **2002**, 11, 2233-41	5.6	49
82	Familial dysautonomia. *Current Opinion in Genetics and Development*, **2002**, 12, 307-11	4.9	274
81	Quantitative neuropathological changes in presymptomatic Huntington's disease. *Annals of Neurology*, **2001**, 49, 29-34	9.4	141
80	Huntington Disease **2001**,		2
79	Cloning, characterization, and genomic structure of the mouse Ikbkap gene. *DNA and Cell Biology*, **2001**, 20, 579-86	3.6	22
78	Tissue-specific expression of a splicing mutation in the IKBKAP gene causes familial dysautonomia. *American Journal of Human Genetics*, **2001**, 68, 598-605	11	477
77	Molecular genetics: unmasking polyglutamine triggers in neurodegenerative disease. *Nature Reviews Neuroscience*, **2000**, 1, 109-15	13.5	322
76	Cloning, mapping, and expression of a novel brain-specific transcript in the familial dysautonomia candidate region on chromosome 9q31. *Mammalian Genome*, **2000**, 11, 81-3	3.2	5
75	Clonal analysis of a case of multiple meningiomas using multiple molecular genetic approaches: pathology case report. *Neurosurgery*, **1999**, 45, 409-16	3.2	48
74	Allelic expression of the NF2 gene in neurofibromatosis 2 and schwannomatosis. *Neurogenetics*, **1999**, 2, 101-8	3	17
73	Merlin: the neurofibromatosis 2 tumor suppressor. *Biochimica Et Biophysica Acta: Reviews on Cancer* , **1999**, 1423, M29-36	11.2	35
72	Precise genetic mapping and haplotype analysis of the familial dysautonomia gene on human chromosome 9q31. *American Journal of Human Genetics*, **1999**, 64, 1110-8	11	59
71	Mapping of the mucolipidosis type IV gene to chromosome 19p and definition of founder haplotypes. *American Journal of Human Genetics*, **1999**, 65, 773-8	11	83
70	Mutant huntingtin forms in vivo complexes with distinct context-dependent conformations of the polyglutamine segment. *Neurobiology of Disease*, **1999**, 6, 364-75	7.5	53
69	Genetic variation in the 3' untranslated region of the neurofibromatosis 1 gene: application to unequal allelic expression. *Somatic Cell and Molecular Genetics*, **1998**, 24, 107-19		12
68	Amyloid formation by mutant huntingtin: threshold, progressivity and recruitment of normal polyglutamine proteins. *Somatic Cell and Molecular Genetics*, **1998**, 24, 217-33		207

67	The FERM domain: a unique module involved in the linkage of cytoplasmic proteins to the membrane. *Trends in Biochemical Sciences*, **1998**, 23, 281-2	10.3	440
66	Huntingtin: a single bait hooks many species. *Current Opinion in Neurobiology*, **1998**, 8, 425-30	7.6	64
65	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, 1273-6	5.4	204
64	The Genetic Defect Causing Huntington's Disease: Repeated in Other Contexts?. *Molecular Medicine*, **1997**, 3, 238-246	6.2	54
63	Heterogeneous topographic and cellular distribution of huntingtin expression in the normal human neostriatum. *Journal of Neuroscience*, **1997**, 17, 3052-63	6.6	131
62	Frequent loss of chromosome 14 in atypical and malignant meningioma: identification of a putative 'tumor progression' locus. *Oncogene*, **1997**, 14, 611-6	9.2	96
61	The early-onset torsion dystonia gene (DYT1) encodes an ATP-binding protein. *Nature Genetics*, **1997**, 17, 40-8	36.3	915
60	Huntingtin is required for neurogenesis and is not impaired by the Huntington's disease CAG expansion. *Nature Genetics*, **1997**, 17, 404-10	36.3	416
59	Exon trapping and sequence-based methods of gene finding in transcript mapping of human 4p16.3. *Somatic Cell and Molecular Genetics*, **1997**, 23, 413-27		3
58	CAG repeat number governs the development rate of pathology in Huntington's disease. *Annals of Neurology*, **1997**, 41, 689-92	9.4	512
57	No association between alpha 1-antichymotrypsin and familial Alzheimer's disease. *Annals of the New York Academy of Sciences*, **1996**, 802, 35-41	6.5	8
56	Differential expression of normal and mutant Huntington's disease gene alleles. *Neurobiology of Disease*, **1996**, 3, 183-90	7.5	68
55	Frequency and distribution of NF2 mutations in schwannomas. *Genes Chromosomes and Cancer*, **1996**, 17, 45-55	5	105
54	Trinucleotide instability: a repeating theme in human inherited disorders. *Annual Review of Medicine*, **1996**, 47, 201-9	17.4	66
53	Prenatal diagnostic testing for familial dysautonomia using linked genetic markers. *Prenatal Diagnosis*, **1995**, 15, 817-26	3.2	11
52	Normal and Expanded Huntington's Disease Gene Alleles Produce Distinguishable Proteins Due to Translation Across the CAG Repeat. *Molecular Medicine*, **1995**, 1, 374-383	6.2	90
51	Isolation of a novel gene underlying Batten disease, CLN3. The International Batten Disease Consortium. *Cell*, **1995**, 82, 949-57	56.2	484
50	Evidence for subarachnoid spread in the development of multiple meningiomas. *Brain Pathology*, **1995**, 5, 11-4	6	28

49	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, 349-55		16
48	Huntington's disease gene: regional and cellular expression in brain of normal and affected individuals. *Annals of Neurology*, **1995**, 37, 218-30	9.4	178
47	Exon scanning for mutation of the NF2 gene in schwannomas. *Human Molecular Genetics*, **1994**, 3, 413-9	5.6	179
46	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		210
45	Mutations in transcript isoforms of the neurofibromatosis 2 gene in multiple human tumour types. *Nature Genetics*, **1994**, 6, 185-92	36.3	215
44	The gene for achondroplasia maps to the telomeric region of chromosome 4p. *Nature Genetics*, **1994**, 6, 314-7	36.3	91
43	High resolution localization of recombination hot spots using sperm typing. *Nature Genetics*, **1994**, 7, 420-4	36.3	74
42	Huntington's disease CAG trinucleotide repeats in pathologically confirmed post-mortem brains. *Neurobiology of Disease*, **1994**, 1, 159-66	7.5	70
41	Characterization of a duplication in the terminal band of 4p by molecular cytogenetics. *American Journal of Medical Genetics Part A*, **1993**, 46, 72-6		15
40	Neurofibromatosis type 1 gene mutations in neuroblastoma. *Nature Genetics*, **1993**, 3, 62-6	36.3	141
39	Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. *Nature Genetics*, **1993**, 4, 160-4	36.3	133
38	A cosmid contig and high resolution restriction map of the 2 megabase region containing the Huntington's disease gene. *Nature Genetics*, **1993**, 4, 181-6	36.3	93
37	Relationship between trinucleotide repeat expansion and phenotypic variation in Huntington's disease. *Nature Genetics*, **1993**, 4, 393-7	36.3	606
36	Discrepancy resolved. *Nature Genetics*, **1993**, 5, 215	36.3	5
35	A linkage study with DNA markers (D4S95, D4S115, and D4S111) in Japanese Huntington disease families. *Japanese Journal of Human Genetics*, **1993**, 38, 193-201		1
34	Hunting for Huntington's disease. *Molecular Genetic Medicine*, **1993**, 3, 139-58		21
33	The Huntington's disease candidate region exhibits many different haplotypes. *Nature Genetics*, **1992**, 1, 99-103	36.3	142
32	A 15-bp deletion in exon 5 of the ornithine aminotransferase (OAT) locus associated with gyrate atrophy. *Human Mutation*, **1992**, 1, 293-7	4.7	5

31	Linkage analysis in juvenile neuronal ceroid lipofuscinosis. *American Journal of Medical Genetics Part A*, **1992**, 42, 542-5		10
30	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, 449-54		17
29	Dystonia gene in Ashkenazi Jewish population is located on chromosome 9q32-34. *Annals of Neurology*, **1990**, 27, 114-20	9.4	117
28	A comparison of neurological, metabolic, structural, and genetic evaluations in persons at risk for Huntington's disease. *Annals of Neurology*, **1990**, 28, 614-21	9.4	100
27	Hyperkalemic periodic paralysis and the adult muscle sodium channel alpha-subunit gene. *Science*, **1990**, 250, 1000-2	33.3	289
26	Location cloning strategy for characterizing genetic defects in Huntington's disease and Alzheimer's disease. *FASEB Journal*, **1989**, 3, 2036-41	0.9	19
25	The role of mitochondrial DNA in Huntington's disease. *Journal of Molecular Neuroscience*, **1989**, 1, 129-136	3.3	9
24	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, 369-75		11
23	Human gene for torsion dystonia located on chromosome 9q32-q34. *Neuron*, **1989**, 2, 1427-34	13.9	207
22	Partial linkage map of chromosome 13q in the region of the Wilson disease and retinoblastoma genes. *Genetic Epidemiology*, **1988**, 5, 375-80	2.6	9
21	Protease inhibitor domain encoded by an amyloid protein precursor mRNA associated with Alzheimer's disease. *Nature*, **1988**, 331, 528-30	50.4	1045
20	Irradiation-reduced human chromosome 21 hybrids. *Somatic Cell and Molecular Genetics*, **1988**, 14, 233-42		36
19	Predictive testing for Huntington's disease with use of a linked DNA marker. *New England Journal of Medicine*, **1988**, 318, 535-42	59.2	151
18	Homozygotes for Huntington's disease. *Nature*, **1987**, 326, 194-7	50.4	298
17	The genetic defect in familial Alzheimer's disease is not tightly linked to the amyloid beta-protein gene. *Nature*, **1987**, 329, 156-7	50.4	253
16	Genetic linkage of bilateral acoustic neurofibromatosis to a DNA marker on chromosome 22. *Nature*, **1987**, 329, 246-8	50.4	413
15	Probes in Huntington's chorea (reply). *Nature*, **1986**, 320, 21-22	50.4	15
14	Loss of genes on chromosome 22 in tumorigenesis of human acoustic neuroma. *Nature*, **1986**, 322, 644-7	50.4	443

13	Accuracy of testing for Huntington's disease. *Nature*, **1986**, 323, 118	50.4	3
12	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, 159-75	1.6	23
11	Huntington's disease. Pathogenesis and management. *New England Journal of Medicine*, **1986**, 315, 1267-76	59.2	472
10	Deletion of Huntington's disease-linked G8 (D4S10) locus in Wolf-Hirschhorn syndrome. *Nature*, **1985**, 318, 75-8	50.4	106
9	Huntington's disease: two families with differing clinical features show linkage to the G8 probe. *Science*, **1985**, 229, 776-9	33.3	71
8	Isolation of polymorphic DNA segments from human chromosome 21. *Nucleic Acids Research*, **1985**, 13, 6075-88	20.1	73
7	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	6.5	5
6	Genetic linkage of the Huntington's disease gene to a DNA marker. *Canadian Journal of Neurological Sciences*, **1984**, 11, 421-5	1	8
5	Use of cyclosporin A in establishing Epstein-Barr virus-transformed human lymphoblastoid cell lines. *In Vitro*, **1984**, 20, 856-8		323
4	A polymorphic DNA marker genetically linked to Huntington's disease. *Nature*, **1983**, 306, 234-8	50.4	1977
3	Parallelized engineering of mutational models using piggyBac transposon delivery of CRISPR libraries		2
2	Huntington disease onset is determined by length of uninterrupted CAG, not encoded polyglutamine, and is modified by DNA maintenance mechanisms		2
1	A cross-disorder dosage sensitivity map of the human genome		5