

James F Gusella

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	A polymorphic DNA marker genetically linked to Huntington's disease. <i>Nature</i> , 1983 , 306, 234-8	50.4	1977
209	Protease inhibitor domain encoded by an amyloid protein precursor mRNA associated with Alzheimer's disease. <i>Nature</i> , 1988 , 331, 528-30	50.4	1045
208	The early-onset torsion dystonia gene (DYT1) encodes an ATP-binding protein. <i>Nature Genetics</i> , 1997 , 17, 40-8	36.3	915
207	Huntington disease. <i>Nature Reviews Disease Primers</i> , 2015 , 1, 15005	51.1	672
206	Relationship between trinucleotide repeat expansion and phenotypic variation in Huntington's disease. <i>Nature Genetics</i> , 1993 , 4, 393-7	36.3	606
205	Venezuelan kindreds reveal that genetic and environmental factors modulate Huntington's disease age of onset. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004 , 101, 3498-503	11.5	572
204	CAG repeat number governs the development rate of pathology in Huntington's disease. <i>Annals of Neurology</i> , 1997 , 41, 689-92	9.4	512
203	Isolation of a novel gene underlying Batten disease, CLN3. The International Batten Disease Consortium. <i>Cell</i> , 1995 , 82, 949-57	56.2	484
202	Tissue-specific expression of a splicing mutation in the IKBKAP gene causes familial dysautonomia. <i>American Journal of Human Genetics</i> , 2001 , 68, 598-605	11	477
201	Huntington's disease. Pathogenesis and management. <i>New England Journal of Medicine</i> , 1986 , 315, 1267-76	59.6	472
200	Loss of genes on chromosome 22 in tumorigenesis of human acoustic neuroma. <i>Nature</i> , 1986 , 322, 644-7	50.4	443
199	Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. <i>Cell</i> , 2012 , 149, 525-37	56.2	441
198	The FERM domain: a unique module involved in the linkage of cytoplasmic proteins to the membrane. <i>Trends in Biochemical Sciences</i> , 1998 , 23, 281-2	10.3	440
197	Huntingtin is required for neurogenesis and is not impaired by the Huntington's disease CAG expansion. <i>Nature Genetics</i> , 1997 , 17, 404-10	36.3	416
196	Genetic linkage of bilateral acoustic neurofibromatosis to a DNA marker on chromosome 22. <i>Nature</i> , 1987 , 329, 246-8	50.4	413
195	Use of cyclosporin A in establishing Epstein-Barr virus-transformed human lymphoblastoid cell lines. <i>In Vitro</i> , 1984 , 20, 856-8		323
194	Molecular genetics: unmasking polyglutamine triggers in neurodegenerative disease. <i>Nature Reviews Neuroscience</i> , 2000 , 1, 109-15	13.5	322

193	Homozygotes for Huntington's disease. <i>Nature</i> , 1987 , 326, 194-7	50.4	298
192	Hyperkalemic periodic paralysis and the adult muscle sodium channel alpha-subunit gene. <i>Science</i> , 1990 , 250, 1000-2	33.3	289
191	KCTD13 is a major driver of mirrored neuroanatomical phenotypes of the 16p11.2 copy number variant. <i>Nature</i> , 2012 , 485, 363-7	50.4	281
190	Familial dysautonomia. <i>Current Opinion in Genetics and Development</i> , 2002 , 12, 307-11	4.9	274
189	The genetic defect in familial Alzheimer's disease is not tightly linked to the amyloid beta-protein gene. <i>Nature</i> , 1987 , 329, 156-7	50.4	253
188	HD CAG repeat implicates a dominant property of huntingtin in mitochondrial energy metabolism. <i>Human Molecular Genetics</i> , 2005 , 14, 2871-80	5.6	246
187	Specific progressive cAMP reduction implicates energy deficit in presymptomatic Huntington's disease knock-in mice. <i>Human Molecular Genetics</i> , 2003 , 12, 497-508	5.6	218
186	Mutations in transcript isoforms of the neurofibromatosis 2 gene in multiple human tumour types. <i>Nature Genetics</i> , 1994 , 6, 185-92	36.3	215
185	CHD8 regulates neurodevelopmental pathways associated with autism spectrum disorder in neural progenitors. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014 , 111, E4468-77	11.5	210
184	Structure and expression of the Huntington's disease gene: evidence against simple inactivation due to an expanded CAG repeat. <i>Somatic Cell and Molecular Genetics</i> , 1994 , 20, 27-38		210
183	Amyloid formation by mutant huntingtin: threshold, progressivity and recruitment of normal polyglutamine proteins. <i>Somatic Cell and Molecular Genetics</i> , 1998 , 24, 217-33		207
182	Human gene for torsion dystonia located on chromosome 9q32-q34. <i>Neuron</i> , 1989 , 2, 1427-34	13.9	207
181	NHE-RF, a regulatory cofactor for Na(+)-H+ exchange, is a common interactor for merlin and ERM (MERM) proteins. <i>Journal of Biological Chemistry</i> , 1998 , 273, 1273-6	5.4	204
180	NF2/merlin is a novel negative regulator of mTOR complex 1, and activation of mTORC1 is associated with meningioma and schwannoma growth. <i>Molecular and Cellular Biology</i> , 2009 , 29, 4250-61 ^{4.8}		202
179	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. <i>Nature Genetics</i> , 2012 , 44, 390-7, S1	36.3	190
178	Exon scanning for mutation of the NF2 gene in schwannomas. <i>Human Molecular Genetics</i> , 1994 , 3, 413-9	5.6	179
177	Huntington's disease gene: regional and cellular expression in brain of normal and affected individuals. <i>Annals of Neurology</i> , 1995 , 37, 218-30	9.4	178
176	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017 , 49, 36-45	36.3	172

175	Assessment of 2q23.1 microdeletion syndrome implicates MBD5 as a single causal locus of intellectual disability, epilepsy, and autism spectrum disorder. <i>American Journal of Human Genetics</i> , 2011 , 89, 551-63	11	166
174	Biotin-responsive basal ganglia disease maps to 2q36.3 and is due to mutations in SLC19A3. <i>American Journal of Human Genetics</i> , 2005 , 77, 16-26	11	158
173	CAG Repeat Not Polyglutamine Length Determines Timing of Huntington's Disease Onset. <i>Cell</i> , 2019 , 178, 887-900.e14	56.2	155
172	Predictive testing for Huntington's disease with use of a linked DNA marker. <i>New England Journal of Medicine</i> , 1988 , 318, 535-42	59.2	151
171	Permanent inactivation of Huntington's disease mutation by personalized allele-specific CRISPR/Cas9. <i>Human Molecular Genetics</i> , 2016 , 25, 4566-4576	5.6	150
170	Clinical diagnosis by whole-genome sequencing of a prenatal sample. <i>New England Journal of Medicine</i> , 2012 , 367, 2226-32	59.2	144
169	The Huntington's disease candidate region exhibits many different haplotypes. <i>Nature Genetics</i> , 1992 , 1, 99-103	36.3	142
168	Quantitative neuropathological changes in presymptomatic Huntington's disease. <i>Annals of Neurology</i> , 2001 , 49, 29-34	9.4	141
167	Neurofibromatosis type 1 gene mutations in neuroblastoma. <i>Nature Genetics</i> , 1993 , 3, 62-6	36.3	141
166	Early phenotypes that presage late-onset neurodegenerative disease allow testing of modifiers in Hdh CAG knock-in mice. <i>Human Molecular Genetics</i> , 2002 , 11, 633-40	5.6	134
165	Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. <i>Nature Genetics</i> , 1993 , 4, 160-4	36.3	133
164	Next-generation sequencing strategies enable routine detection of balanced chromosome rearrangements for clinical diagnostics and genetic research. <i>American Journal of Human Genetics</i> , 2011 , 88, 469-81	11	132
163	Heterogeneous topographic and cellular distribution of huntingtin expression in the normal human neostriatum. <i>Journal of Neuroscience</i> , 1997 , 17, 3052-63	6.6	131
162	A genome scan for modifiers of age at onset in Huntington disease: The HD MAPS study. <i>American Journal of Human Genetics</i> , 2003 , 73, 682-7	11	131
161	Huntingtin facilitates polycomb repressive complex 2. <i>Human Molecular Genetics</i> , 2010 , 19, 573-83	5.6	128
160	Dystonia gene in Ashkenazi Jewish population is located on chromosome 9q32-34. <i>Annals of Neurology</i> , 1990 , 27, 114-20	9.4	117
159	Rescue of a human mRNA splicing defect by the plant cytokinin kinetin. <i>Human Molecular Genetics</i> , 2004 , 13, 429-36	5.6	114
158	The predominantly HEAT-like motif structure of huntingtin and its association and coincident nuclear entry with dorsal, an NF- κ B/Rel/dorsal family transcription factor. <i>BMC Neuroscience</i> , 2002 , 3, 15	3.2	109

157	Huntington's disease: seeing the pathogenic process through a genetic lens. <i>Trends in Biochemical Sciences</i> , 2006 , 31, 533-40	10.3	107
156	Deletion of Huntington's disease-linked G8 (D4S10) locus in Wolf-Hirschhorn syndrome. <i>Nature</i> , 1985 , 318, 75-8	50.4	106
155	Frequency and distribution of NF2 mutations in schwannomas. <i>Genes Chromosomes and Cancer</i> , 1996 , 17, 45-55	5	105
154	Reversal of a full-length mutant huntingtin neuronal cell phenotype by chemical inhibitors of polyglutamine-mediated aggregation. <i>BMC Neuroscience</i> , 2005 , 6, 1	3.2	102
153	A comparison of neurological, metabolic, structural, and genetic evaluations in persons at risk for Huntington's disease. <i>Annals of Neurology</i> , 1990 , 28, 614-21	9.4	100
152	RNA Sequence Analysis of Human Huntington Disease Brain Reveals an Extensive Increase in Inflammatory and Developmental Gene Expression. <i>PLoS ONE</i> , 2015 , 10, e0143563	3.7	99
151	Frequent loss of chromosome 14 in atypical and malignant meningioma: identification of a putative 'tumor progression' locus. <i>Oncogene</i> , 1997 , 14, 611-6	9.2	96
150	A cosmid contig and high resolution restriction map of the 2 megabase region containing the Huntington's disease gene. <i>Nature Genetics</i> , 1993 , 4, 181-6	36.3	93
149	The HTT CAG-Expansion Mutation Determines Age at Death but Not Disease Duration in Huntington Disease. <i>American Journal of Human Genetics</i> , 2016 , 98, 287-98	11	92
148	The gene for achondroplasia maps to the telomeric region of chromosome 4p. <i>Nature Genetics</i> , 1994 , 6, 314-7	36.3	91
147	Normal and Expanded Huntingtin Disease Gene Alleles Produce Distinguishable Proteins Due to Translation Across the CAG Repeat. <i>Molecular Medicine</i> , 1995 , 1, 374-383	6.2	90
146	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. <i>Nature Genetics</i> , 2017 , 49, 238-248	36.3	88
145	Huntington's disease: the case for genetic modifiers. <i>Genome Medicine</i> , 2009 , 1, 80	14.4	87
144	Genetic modifiers of Huntington's disease. <i>Movement Disorders</i> , 2014 , 29, 1359-65	7	84
143	Characterization of apparently balanced chromosomal rearrangements from the developmental genome anatomy project. <i>American Journal of Human Genetics</i> , 2008 , 82, 712-22	11	84
142	Mapping of the mucopolidosis type IV gene to chromosome 19p and definition of founder haplotypes. <i>American Journal of Human Genetics</i> , 1999 , 65, 773-8	11	83
141	Transcriptional consequences of 16p11.2 deletion and duplication in mouse cortex and multiplex autism families. <i>American Journal of Human Genetics</i> , 2014 , 94, 870-83	11	78
140	CSIG-42. HIGH THROUGHPUT KINOME AND TRANSCRIPTOME ANALYSES REVEAL NOVEL THERAPEUTIC TARGETS IN NF2-DEFICIENT MENINGIOMA. <i>Neuro-Oncology</i> , 2018 , 20, vi52-vi52	1	78

139	miR-10b-5p expression in Huntington's disease brain relates to age of onset and the extent of striatal involvement. <i>BMC Medical Genomics</i> , 2015 , 8, 10	3.7	76
138	Regulation of mTOR complex 2 signaling in neurofibromatosis 2-deficient target cell types. <i>Molecular Cancer Research</i> , 2012 , 10, 649-59	6.6	74
137	High resolution localization of recombination hot spots using sperm typing. <i>Nature Genetics</i> , 1994 , 7, 420-4	36.3	74
136	MicroRNAs located in the Hox gene clusters are implicated in huntington's disease pathogenesis. <i>PLoS Genetics</i> , 2014 , 10, e1004188	6	73
135	Isolation of polymorphic DNA segments from human chromosome 21. <i>Nucleic Acids Research</i> , 1985 , 13, 6075-88	20.1	73
134	Huntington's disease: two families with differing clinical features show linkage to the G8 probe. <i>Science</i> , 1985 , 229, 776-9	33.3	71
133	Huntington's disease CAG trinucleotide repeats in pathologically confirmed post-mortem brains. <i>Neurobiology of Disease</i> , 1994 , 1, 159-66	7.5	70
132	Differential expression of normal and mutant Huntington's disease gene alleles. <i>Neurobiology of Disease</i> , 1996 , 3, 183-90	7.5	68
131	Trinucleotide instability: a repeating theme in human inherited disorders. <i>Annual Review of Medicine</i> , 1996 , 47, 201-9	17.4	66
130	Huntingtin: a single bait hooks many species. <i>Current Opinion in Neurobiology</i> , 1998 , 8, 425-30	7.6	64
129	Evidence for a modifier of onset age in Huntington disease linked to the HD gene in 4p16. <i>Neurogenetics</i> , 2004 , 5, 109-14	3	63
128	Genome-wide significance for a modifier of age at neurological onset in Huntington's disease at 6q23-24: the HD MAPS study. <i>BMC Medical Genomics</i> , 2006 , 7, 71	2.1	62
127	Huntington's disease. <i>NeuroMolecular Medicine</i> , 2003 , 4, 7-20	4.6	61
126	A modifier of Huntington's disease onset at the MLH1 locus. <i>Human Molecular Genetics</i> , 2017 , 26, 3859-3867	3.67	59
125	HD CAG-correlated gene expression changes support a simple dominant gain of function. <i>Human Molecular Genetics</i> , 2011 , 20, 2846-60	5.6	59
124	Precise genetic mapping and haplotype analysis of the familial dysautonomia gene on human chromosome 9q31. <i>American Journal of Human Genetics</i> , 1999 , 64, 1110-8	11	59
123	Magicin, a novel cytoskeletal protein associates with the NF2 tumor suppressor merlin and Grb2. <i>Oncogene</i> , 2004 , 23, 8815-25	9.2	56
122	The Genetic Defect Causing Huntington's Disease: Repeated in Other Contexts?. <i>Molecular Medicine</i> , 1997 , 3, 238-246	6.2	54

121	Mutant huntingtin forms in vivo complexes with distinct context-dependent conformations of the polyglutamine segment. <i>Neurobiology of Disease</i> , 1999 , 6, 364-75	7.5	53
120	Functionally compromised CHD7 alleles in patients with isolated GnRH deficiency. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014 , 111, 17953-8	11.5	51
119	Identification of a presymptomatic molecular phenotype in Hdh CAG knock-in mice. <i>Human Molecular Genetics</i> , 2002 , 11, 2233-41	5.6	49
118	Assessment of cortical and striatal involvement in 523 Huntington disease brains. <i>Neurology</i> , 2012 , 79, 1708-15	6.5	48
117	Common SNP-based haplotype analysis of the 4p16.3 Huntington disease gene region. <i>American Journal of Human Genetics</i> , 2012 , 90, 434-44	11	48
116	Clonal analysis of a case of multiple meningiomas using multiple molecular genetic approaches: pathology case report. <i>Neurosurgery</i> , 1999 , 45, 409-16	3.2	48
115	Deficiency of huntingtin has pleiotropic effects in the social amoeba <i>Dictyostelium discoideum</i> . <i>PLoS Genetics</i> , 2011 , 7, e1002052	6	44
114	Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. <i>American Journal of Human Genetics</i> , 2016 , 99, 1015-1033	11.33	43
113	Brain urea increase is an early Huntington's disease pathogenic event observed in a prodromal transgenic sheep model and HD cases. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017 , 114, E11293-E11302	11.5	43
112	Describing sequencing results of structural chromosome rearrangements with a suggested next-generation cytogenetic nomenclature. <i>American Journal of Human Genetics</i> , 2014 , 94, 695-709	11	40
111	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. <i>Nature Neuroscience</i> , 2016 , 19, 517-22	25.5	39
110	Htt CAG repeat expansion confers pleiotropic gains of mutant huntingtin function in chromatin regulation. <i>Human Molecular Genetics</i> , 2015 , 24, 2442-57	5.6	38
109	Further molecular characterisation of the OVT73 transgenic sheep model of Huntington's disease identifies cortical aggregates. <i>Journal of Huntingtons Disease</i> , 2013 , 2, 279-95	1.9	38
108	CSF1R mosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. <i>Brain</i> , 2016 , 139, 1666-72	11.2	38
107	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. <i>American Journal of Human Genetics</i> , 2015 , 97, 170-6	11	37
106	Cryptic and complex chromosomal aberrations in early-onset neuropsychiatric disorders. <i>American Journal of Human Genetics</i> , 2014 , 95, 454-61	11	37
105	Meclizine is neuroprotective in models of Huntington's disease. <i>Human Molecular Genetics</i> , 2011 , 20, 294-300	5.6	36
104	Irradiation-reduced human chromosome 21 hybrids. <i>Somatic Cell and Molecular Genetics</i> , 1988 , 14, 233-42		36

103	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. <i>American Journal of Human Genetics</i> , 2015 , 96, 784-96	11	35
102	Metabolic disruption identified in the Huntington's disease transgenic sheep model. <i>Scientific Reports</i> , 2016 , 6, 20681	4.9	35
101	Monozygotic twins discordant for neurofibromatosis 1. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 601-6	2.5	35
100	Modeling NF2 with human arachnoidal and meningioma cell culture systems: NF2 silencing reflects the benign character of tumor growth. <i>Neurobiology of Disease</i> , 2008 , 29, 278-92	7.5	35
99	Merlin: the neurofibromatosis 2 tumor suppressor. <i>Biochimica Et Biophysica Acta: Reviews on Cancer</i> , 1999 , 1423, M29-36	11.2	35
98	A high-throughput kinome screen reveals serum/glucocorticoid-regulated kinase 1 as a therapeutic target for NF2-deficient meningiomas. <i>Oncotarget</i> , 2015 , 6, 16981-97	3.3	35
97	Large-scale phenome analysis defines a behavioral signature for Huntington's disease genotype in mice. <i>Nature Biotechnology</i> , 2016 , 34, 838-44	44.5	30
96	Clinical-Genetic Associations in the Prospective Huntington at Risk Observational Study (PHAROS): Implications for Clinical Trials. <i>JAMA Neurology</i> , 2016 , 73, 102-10	17.2	29
95	Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. <i>European Journal of Human Genetics</i> , 2014 , 22, 57-63	5.3	29
94	Brain-derived neurotrophic factor does not influence age at neurologic onset of Huntington's disease. <i>Neurobiology of Disease</i> , 2006 , 24, 280-5	7.5	29
93	Evidence for subarachnoid spread in the development of multiple meningiomas. <i>Brain Pathology</i> , 1995 , 5, 11-4	6	28
92	High resolution time-course mapping of early transcriptomic, molecular and cellular phenotypes in Huntington's disease CAG knock-in mice across multiple genetic backgrounds. <i>Human Molecular Genetics</i> , 2017 , 26, 913-922	5.6	27
91	Population stratification may bias analysis of PGC-1 α as a modifier of age at Huntington disease motor onset. <i>Human Genetics</i> , 2012 , 131, 1833-40	6.3	25
90	No post-genetics era in human disease research. <i>Nature Reviews Genetics</i> , 2002 , 3, 72-9	30.1	25
89	A novel neurodevelopmental disorder associated with compound heterozygous variants in the huntingtin gene. <i>European Journal of Human Genetics</i> , 2016 , 24, 1826-1827	5.3	23
88	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. <i>Journal of Neurogenetics</i> , 1986 , 3, 159-75	1.6	23
87	Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis. <i>Human Molecular Genetics</i> , 2016 , 25, 1255-70	5.6	22
86	Genetic Modification of Huntington Disease Acts Early in the Prediagnosis Phase. <i>American Journal of Human Genetics</i> , 2018 , 103, 349-357	11	22

85	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 395-406	2.5	22
84	Cloning, characterization, and genomic structure of the mouse Ikbkap gene. <i>DNA and Cell Biology</i> , 2001 , 20, 579-86	3.6	22
83	Promotion of somatic CAG repeat expansion by Fan1 knock-out in Huntington's disease knock-in mice is blocked by Mlh1 knock-out. <i>Human Molecular Genetics</i> , 2020 , 29, 3044-3053	5.6	22
82	Genetic and Functional Analyses Point to FAN1 as the Source of Multiple Huntington Disease Modifier Effects. <i>American Journal of Human Genetics</i> , 2020 , 107, 96-110	11	21
81	Hunting for Huntington's disease. <i>Molecular Genetic Medicine</i> , 1993 , 3, 139-58		21
80	The Genetic Modifiers of Motor OnsetAge[(GeM MOA) Website: Genome-wide Association Analysis for Genetic Modifiers of Huntington's Disease. <i>Journal of Huntingtons Disease</i> , 2015 , 4, 279-84	1.9	20
79	Differential effects of the Huntington's disease CAG mutation in striatum and cerebellum are quantitative not qualitative. <i>Human Molecular Genetics</i> , 2011 , 20, 4258-67	5.6	20
78	Location cloning strategy for characterizing genetic defects in Huntington's disease and Alzheimer's disease. <i>FASEB Journal</i> , 1989 , 3, 2036-41	0.9	19
77	Population-specific genetic modification of Huntington's disease in Venezuela. <i>PLoS Genetics</i> , 2018 , 14, e1007274	6	18
76	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. <i>Human Mutation</i> , 2017 , 38, 378-389	4.7	17
75	De novo variants in the Helicase-C domain of CHD8 are associated with severe phenotypes including autism, language disability and overgrowth. <i>Human Genetics</i> , 2020 , 139, 499-512	6.3	17
74	TAA repeat variation in the GRIK2 gene does not influence age at onset in Huntington's disease. <i>Biochemical and Biophysical Research Communications</i> , 2012 , 424, 404-8	3.4	17
73	Allelic expression of the NF2 gene in neurofibromatosis 2 and schwannomatosis. <i>Neurogenetics</i> , 1999 , 2, 101-8	3	17
72	Molecular detection of a 4p deletion using PCR-based polymorphisms: a technique for the rapid detection of the Wolf-Hirschhorn syndrome. <i>American Journal of Medical Genetics Part A</i> , 1992 , 44, 449-54		17
71	Expanding the Spectrum of Founder Mutations Causing Isolated Gonadotropin-Releasing Hormone Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015 , 100, E1378-85	5.6	16
70	Prenatal diagnosis of familial dysautonomia by analysis of linked CA-repeat polymorphisms on chromosome 9q31-q33. <i>American Journal of Medical Genetics Part A</i> , 1995 , 59, 349-55		16
69	Huntington's Disease Pathogenesis: Two Sequential Components. <i>Journal of Huntingtons Disease</i> , 2021 , 10, 35-51	1.9	16
68	Candidate loci for Zimmermann-Laband syndrome at 3p14.3. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 107-11	2.5	15

67	Genetic analysis of the GRIK2 modifier effect in Huntington's disease. <i>BMC Neuroscience</i> , 2006 , 7, 62	3.2	15
66	Characterization of a duplication in the terminal band of 4p by molecular cytogenetics. <i>American Journal of Medical Genetics Part A</i> , 1993 , 46, 72-6		15
65	Probes in Huntington's chorea (reply). <i>Nature</i> , 1986 , 320, 21-22	50.4	15
64	Sequence-Level Analysis of the Major European Huntington Disease Haplotype. <i>American Journal of Human Genetics</i> , 2015 , 97, 435-44	11	14
63	EPH receptor signaling as a novel therapeutic target in NF2-deficient meningioma. <i>Neuro-Oncology</i> , 2018 , 20, 1185-1196	1	14
62	Haplotype-based stratification of Huntington's disease. <i>European Journal of Human Genetics</i> , 2017 , 25, 1202-1209	5.3	14
61	WNT/ β -Catenin Pathway and Epigenetic Mechanisms Regulate the Pitt-Hopkins Syndrome and Schizophrenia Risk Gene. <i>Molecular Neuropsychiatry</i> , 2017 , 3, 53-71	4.9	14
60	Genetic Risk Underlying Psychiatric and Cognitive Symptoms in Huntington's Disease. <i>Biological Psychiatry</i> , 2020 , 87, 857-865	7.9	13
59	Genetic variation in the 3' untranslated region of the neurofibromatosis 1 gene: application to unequal allelic expression. <i>Somatic Cell and Molecular Genetics</i> , 1998 , 24, 107-19		12
58	Modified single-stranded oligonucleotides inhibit aggregate formation and toxicity induced by expanded polyglutamine. <i>Journal of Molecular Neuroscience</i> , 2004 , 24, 257-67	3.3	12
57	Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. <i>European Journal of Human Genetics</i> , 2016 , 24, 1622-1626	5.3	12
56	Opposing Tumor-Promoting and -Suppressive Functions of Rictor/mTORC2 Signaling in Adult Glioma and Pediatric SHH Medulloblastoma. <i>Cell Reports</i> , 2018 , 24, 463-478.e5	10.6	11
55	A rare exonic NRXN3 deletion segregating with neurodevelopmental and neuropsychiatric conditions in a three-generation Chinese family. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018 , 177, 589-595	3.5	11
54	Extensive molecular genetic analysis of the 3p14.3 region in patients with Zimmermann-Laband syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 2668-74	2.5	11
53	Genetic criteria for Huntington's disease pathogenesis. <i>Brain Research Bulletin</i> , 2007 , 72, 78-82	3.9	11
52	Prenatal diagnostic testing for familial dysautonomia using linked genetic markers. <i>Prenatal Diagnosis</i> , 1995 , 15, 817-26	3.2	11
51	Characterization of an unusual and complex chromosome 21 rearrangement using somatic cell genetics and cloned DNA probes. <i>American Journal of Medical Genetics Part A</i> , 1989 , 33, 369-75		11
50	Timing and Impact of Psychiatric, Cognitive, and Motor Abnormalities in Huntington Disease. <i>Neurology</i> , 2021 , 96, e2395-e2406	6.5	11

49	A Balanced Translocation in Kallmann Syndrome Implicates a Long Noncoding RNA, RMST, as a GnRH Neuronal Regulator. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020 , 105,	5.6	11
48	Novel allele-specific quantification methods reveal no effects of adult onset CAG repeats on HTT mRNA and protein levels. <i>Human Molecular Genetics</i> , 2017 , 26, 1258-1267	5.6	10
47	Traditional and systems biology based drug discovery for the rare tumor syndrome neurofibromatosis type 2. <i>PLoS ONE</i> , 2018 , 13, e0197350	3.7	10
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