Stanley F Nelson

List of Publications by Citations

292 papers	35,415 citations	94 h-index	185 g-index
316 ext. papers	40,160 ext. citations	10.6 avg, IF	6.72 L-index

#	Paper	IF	Citations
292	Shotgun bisulphite sequencing of the Arabidopsis genome reveals DNA methylation patterning. *Nature*, **2008**, 452, 215-9	50.4	1704
291	Melanomas acquire resistance to B-RAF(V600E) inhibition by RTK or N-RAS upregulation. *Nature*, **2010**, 468, 973-7	50.4	1678
290	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
289	Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **2010**, 466, 368-72	50.4	1499
288	Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia. *Science*, **2008**, 320, 539-43	33.3	1443
287	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*, **2007**, 39, 319-28	36.3	1083
286	High-throughput oncogene mutation profiling in human cancer. *Nature Genetics*, **2007**, 39, 347-51	36.3	847
285	Assessing the significance of chromosomal aberrations in cancer: methodology and application to glioma. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 20007-12	11.5	812
284	Resolving individuals contributing trace amounts of DNA to highly complex mixtures using high-density SNP genotyping microarrays. *PLoS Genetics*, **2008**, 4, e1000167	6	686
283	DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. *Genetics in Medicine*, **2011**, 13, 913-20	8.1	670
282	Clinical exome sequencing for genetic identification of rare Mendelian disorders. *JAMA - Journal of the American Medical Association*, **2014**, 312, 1880-7	27.4	640
281	Linkage, association, and gene-expression analyses identify CNTNAP2 as an autism-susceptibility gene. *American Journal of Human Genetics*, **2008**, 82, 150-9	11	623
280	Gene expression profiling of gliomas strongly predicts survival. *Cancer Research*, **2004**, 64, 6503-10	10.1	573
279	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. *Nature Neuroscience*, **2015**, 18, 199-209	25.5	572
278	Analysis of oncogenic signaling networks in glioblastoma identifies ASPM as a molecular target. *Proceedings of the National Academy of Sciences of the United States of America*, **2006**, 103, 17402-7	11.5	503
277	Melanoma whole-exome sequencing identifies (V600E)B-RAF amplification-mediated acquired B-RAF inhibitor resistance. *Nature Communications*, **2012**, 3, 724	17.4	500
276	A genome-wide scan for common alleles affecting risk for autism. *Human Molecular Genetics*, **2010**, 19, 4072-82	5.6	443

275	DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13 as well as Down syndrome: an international collaborative study. *Genetics in Medicine*, **2012**, 14, 296-305	8.1	407
274	BFAST: an alignment tool for large scale genome resequencing. *PLoS ONE*, **2009**, 4, e7767	3.7	394
273	Genome sequencing highlights the dynamic early history of dogs. *PLoS Genetics*, **2014**, 10, e1004016	6	372
272	Targeted therapy resistance mediated by dynamic regulation of extrachromosomal mutant EGFR DNA. *Science*, **2014**, 343, 72-6	33.3	316
271	Gene expression profile correlates with T-cell infiltration and relative survival in glioblastoma patients vaccinated with dendritic cell immunotherapy. *Clinical Cancer Research*, **2011**, 17, 1603-15	12.9	315
270	C-terminal truncations in human 3R5RDNA exonuclease TREX1 cause autosomal dominant retinal vasculopathy with cerebral leukodystrophy. *Nature Genetics*, **2007**, 39, 1068-70	36.3	307
269	Exome sequencing-based copy-number variation and loss of heterozygosity detection: ExomeCNV. *Bioinformatics*, **2011**, 27, 2648-54	7.2	305
268	Mutations in a human ROBO gene disrupt hindbrain axon pathway crossing and morphogenesis. *Science*, **2004**, 304, 1509-13	33.3	304
267	Gain-of-function mutations in TRPV4 cause autosomal dominant brachyolmia. *Nature Genetics*, **2008**, 40, 999-1003	36.3	295
266	Cold urticaria, immunodeficiency, and autoimmunity related to PLCG2 deletions. *New England Journal of Medicine*, **2012**, 366, 330-8	59.2	288
265	Genome-wide copy number variation study associates metabotropic glutamate receptor gene networks with attention deficit hyperactivity disorder. *Nature Genetics*, **2011**, 44, 78-84	36.3	279
264	Individual common variants exert weak effects on the risk for autism spectrum disorders. *Human Molecular Genetics*, **2012**, 21, 4781-92	5.6	279
263	Identification of inflammatory gene modules based on variations of human endothelial cell responses to oxidized lipids. *Proceedings of the National Academy of Sciences of the United States of America*, **2006**, 103, 12741-6	11.5	262
262	A genomewide scan for loci involved in attention-deficit/hyperactivity disorder. *American Journal of Human Genetics*, **2002**, 70, 1183-96	11	262
261	Gene connectivity, function, and sequence conservation: predictions from modular yeast co-expression networks. *BMC Genomics*, **2006**, 7, 40	4.5	258
260	Epidermal growth factor receptor activation in glioblastoma through novel missense mutations in the extracellular domain. *PLoS Medicine*, **2006**, 3, e485	11.6	242
259	Gene expression profiling identifies molecular subtypes of gliomas. *Oncogene*, **2003**, 22, 4918-23	9.2	241
258	Whole genome amplification using a degenerate oligonucleotide primer allows hundreds of genotypes to be performed on less than one nanogram of genomic DNA. *Proceedings of the National Academy of Sciences of the United States of America*, **1996**, 93, 14676-9	11.5	236

257	A Single CRISPR-Cas9 Deletion Strategy that Targets the Majority of DMD Patients Restores Dystrophin Function in hiPSC-Derived Muscle Cells. *Cell Stem Cell*, **2016**, 18, 533-40	18	233
256	Identification of molecular subtypes of glioblastoma by gene expression profiling. *Oncogene*, **2003**, 22, 2361-73	9.2	226
255	A genetic analysis of neural progenitor differentiation. *Neuron*, **2001**, 29, 325-39	13.9	226
254	Oxysterols are novel activators of the hedgehog signaling pathway in pluripotent mesenchymal cells. *Journal of Biological Chemistry*, **2007**, 282, 8959-68	5.4	221
253	U87MG decoded: the genomic sequence of a cytogenetically aberrant human cancer cell line. *PLoS Genetics*, **2010**, 6, e1000832	6	195
252	Genome-wide analysis of copy number variants in attention deficit hyperactivity disorder: the role of rare variants and duplications at 15q13.3. *American Journal of Psychiatry*, **2012**, 169, 195-204	11.9	195
251	Primary glioblastomas express mesenchymal stem-like properties. *Molecular Cancer Research*, **2006**, 4, 607-19	6.6	185
250	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. *Nature Genetics*, **2012**, 44, 575-80	36.3	183
249	Evidence that the dopamine D4 receptor is a susceptibility gene in attention deficit hyperactivity disorder. *Molecular Psychiatry*, **1998**, 3, 427-30	15.1	180
248	Mutations in PYCR1 cause cutis laxa with progeroid features. *Nature Genetics*, **2009**, 41, 1016-21	36.3	178
247	Genetic linkage of attention-deficit/hyperactivity disorder on chromosome 16p13, in a region implicated in autism. *American Journal of Human Genetics*, **2002**, 71, 959-63	11	176
246	A genomewide scan for attention-deficit/hyperactivity disorder in an extended sample: suggestive linkage on 17p11. *American Journal of Human Genetics*, **2003**, 72, 1268-79	11	174
245	Exome sequencing in the clinical diagnosis of sporadic or familial cerebellar ataxia. *JAMA Neurology* , **2014**, 71, 1237-46	17.2	171
244	Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. *Nature Genetics*, **2012**, 44, 704-8	36.3	170
243	Progressive ataxia due to a missense mutation in a calcium-channel gene. *American Journal of Human Genetics*, **1997**, 61, 1078-87	11	167
242	DNA-microarray analysis of brain cancer: molecular classification for therapy. *Nature Reviews Neuroscience*, **2004**, 5, 782-92	13.5	166
241	SGK196 is a glycosylation-specific O-mannose kinase required for dystroglycan function. *Science*, **2013**, 341, 896-9	33.3	162
240	Distinct transcription profiles of primary and secondary glioblastoma subgroups. *Cancer Research*, **2006**, 66, 159-67	10.1	158

239	Identification of EpCAM as the gene for congenital tufting enteropathy. *Gastroenterology*, **2008**, 135, 429-37	13.3	157
238	Evidence for linkage of a tandem duplication polymorphism upstream of the dopamine D4 receptor gene (DRD4) with attention deficit hyperactivity disorder (ADHD). *Molecular Psychiatry*, **2000**, 5, 531-6	15.1	156
237	Detection of differentially expressed genes in primary tumor tissues using representational differences analysis coupled to microarray hybridization. *Nucleic Acids Research*, **1998**, 26, 3059-65	20.1	154
236	Gene expression analysis of glioblastomas identifies the major molecular basis for the prognostic benefit of younger age. *BMC Medical Genomics*, **2008**, 1, 52	3.7	153
235	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. *Human Genetics*, **2012**, 131, 565-79	6.3	150
234	Insulin growth factor-binding protein 2 is a candidate biomarker for PTEN status and PI3K/Akt pathway activation in glioblastoma and prostate cancer. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 5563-8	11.5	150
233	Joint analysis of the DRD5 marker concludes association with attention-deficit/hyperactivity disorder confined to the predominantly inattentive and combined subtypes. *American Journal of Human Genetics*, **2004**, 74, 348-56	11	148
232	Evidence for sex-specific risk alleles in autism spectrum disorder. *American Journal of Human Genetics*, **2004**, 75, 1117-23	11	146
231	Mutations in the PCNA-binding domain of CDKN1C cause IMAGe syndrome. *Nature Genetics*, **2012**, 44, 788-92	36.3	140
230	Relationship between gene expression and enhancement in glioblastoma multiforme: exploratory DNA microarray analysis. *Radiology*, **2008**, 249, 268-77	20.5	133
229	Exome sequencing for the diagnosis of 46,XY disorders of sex development. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E333-44	5.6	132
228	Maternal embryonic leucine zipper kinase is a key regulator of the proliferation of malignant brain tumors, including brain tumor stem cells. *Journal of Neuroscience Research*, **2008**, 86, 48-60	4.4	132
227	Ciliary abnormalities due to defects in the retrograde transport protein DYNC2H1 in short-rib polydactyly syndrome. *American Journal of Human Genetics*, **2009**, 84, 542-9	11	131
226	Meta-analysis of genome-wide linkage scans of attention deficit hyperactivity disorder. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 1392-8	3.5	131
225	Familial episodic ataxia: clinical heterogeneity in four families linked to chromosome 19p. *Annals of Neurology*, **1997**, 41, 8-16	9.4	130
224	FXR regulates organic solute transporters alpha and beta in the adrenal gland, kidney, and intestine. *Journal of Lipid Research*, **2006**, 47, 201-14	6.3	130
223	Genome-wide analysis of single-nucleotide polymorphisms in human expressed sequences. *Nature Genetics*, **2000**, 26, 233-6	36.3	130
222	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. *New England Journal of Medicine*, **2018**, 379, 2131-2139	59.2	129

221	Prevalence and psychiatric comorbidity of attention-deficit/hyperactivity disorder in an adolescent Finnish population. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2007**, 46, 1575-83	7.2	124
220	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. *Nature Genetics*, **2012**, 44, 193-9	36.3	123
219	The apolipoprotein E epsilon4 allele is not a significant risk factor for frontotemporal dementia. *Annals of Neurology*, **1998**, 44, 134-8	9.4	122
218	Replication of autism linkage: fine-mapping peak at 17q21. *American Journal of Human Genetics*, **2005**, 76, 1050-6	11	122
217	Maternal embryonic leucine zipper kinase (MELK) regulates multipotent neural progenitor proliferation. *Journal of Cell Biology*, **2005**, 170, 413-27	7.3	122
216	Fyn and SRC are effectors of oncogenic epidermal growth factor receptor signaling in glioblastoma patients. *Cancer Research*, **2009**, 69, 6889-98	10.1	120
215	Multiple self-healing squamous epithelioma is caused by a disease-specific spectrum of mutations in TGFBR1. *Nature Genetics*, **2011**, 43, 365-9	36.3	119
214	Genomic mismatch scanning: a new approach to genetic linkage mapping. *Nature Genetics*, **1993**, 4, 11-8	36.3	116
213	Exome sequencing identifies PDE4D mutations in acrodysostosis. *American Journal of Human Genetics*, **2012**, 90, 746-51	11	114
212	High-throughput profiling of influenza A virus hemagglutinin gene at single-nucleotide resolution. *Scientific Reports*, **2014**, 4, 4942	4.9	113
211	Attention deficit hyperactivity disorder: fine mapping supports linkage to 5p13, 6q12, 16p13, and 17p11. *American Journal of Human Genetics*, **2004**, 75, 661-8	11	112
210	Biomarkers to predict antidepressant response. *Current Psychiatry Reports*, **2010**, 12, 553-62	9.1	111
209	Family-based genome-wide association scan of attention-deficit/hyperactivity disorder. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 898-905.e3	7.2	109
208	EGFR mutation-induced alternative splicing of Max contributes to growth of glycolytic tumors in brain cancer. *Cell Metabolism*, **2013**, 17, 1000-1008	24.6	105
207	Lethal skeletal dysplasia in mice and humans lacking the golgin GMAP-210. *New England Journal of Medicine*, **2010**, 362, 206-16	59.2	105
206	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. *American Journal of Human Genetics*, **2017**, 100, 185-192	11	102
205	ERBB3 and NGFR mark a distinct skeletal muscle progenitor cell in human development and hPSCs. *Nature Cell Biology*, **2018**, 20, 46-57	23.4	102
204	Assessing the necessity of confirmatory testing for exome-sequencing results in a clinical molecular diagnostic laboratory. *Genetics in Medicine*, **2014**, 16, 510-5	8.1	102

203	Biased paternal transmission of SNAP-25 risk alleles in attention-deficit hyperactivity disorder. *Molecular Psychiatry*, **2003**, 8, 309-15	15.1	100
202	Differential induction of glioblastoma migration and growth by two forms of pleiotrophin. *Journal of Biological Chemistry*, **2005**, 280, 26953-64	5.4	100
201	Molecular properties of CD133+ glioblastoma stem cells derived from treatment-refractory recurrent brain tumors. *Journal of Neuro-Oncology*, **2009**, 94, 1-19	4.8	98
200	Genomic landscape of meningiomas. *Brain Pathology*, **2010**, 20, 751-62	6	97
199	A recessive skeletal dysplasia, SEMD aggrecan type, results from a missense mutation affecting the C-type lectin domain of aggrecan. *American Journal of Human Genetics*, **2009**, 84, 72-9	11	94
198	Relationship between survival and edema in malignant gliomas: role of vascular endothelial growth factor and neuronal pentraxin 2. *Clinical Cancer Research*, **2007**, 13, 2592-8	12.9	92
197	Transmission disequilibrium testing of dopamine-related candidate gene polymorphisms in ADHD: confirmation of association of ADHD with DRD4 and DRD5. *Molecular Psychiatry*, **2004**, 9, 711-7	15.1	92
196	Joint mouse-human phenome-wide association to test gene function and disease risk. *Nature Communications*, **2016**, 7, 10464	17.4	91
195	Annexin A6 modifies muscular dystrophy by mediating sarcolemmal repair. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 6004-9	11.5	90
194	Disruption of POF1B binding to nonmuscle actin filaments is associated with premature ovarian failure. *American Journal of Human Genetics*, **2006**, 79, 113-9	11	89
193	Temperament and character profiles and the dopamine D4 receptor gene in ADHD. *American Journal of Psychiatry*, **2005**, 162, 906-13	11.9	82
192	De novo nonsense mutations in KAT6A, a lysine acetyl-transferase gene, cause a syndrome including microcephaly and global developmental delay. *American Journal of Human Genetics*, **2015**, 96, 498-506	11	79
191	PDE5 inhibition alleviates functional muscle ischemia in boys with Duchenne muscular dystrophy. *Neurology*, **2014**, 82, 2085-91	6.5	79
190	Association of the cannabinoid receptor gene (CNR1) with ADHD and post-traumatic stress disorder. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 1488-94	3.5	78
189	Loss of CHSY1, a secreted FRINGE enzyme, causes syndromic brachydactyly in humans via increased NOTCH signaling. *American Journal of Human Genetics*, **2010**, 87, 768-78	11	75
188	Accuracy of phenotyping of autistic children based on Internet implemented parent report. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1119-26	3.5	73
187	The functional O-mannose glycan on α-dystroglycan contains a phospho-ribitol primed for matriglycan addition. *ELife*, **2016**, 5,	8.9	73
186	Stem cell associated gene expression in glioblastoma multiforme: relationship to survival and the subventricular zone. *Journal of Neuro-Oncology*, **2010**, 96, 359-67	4.8	72

185	SeqWare Query Engine: storing and searching sequence data in the cloud. *BMC Bioinformatics*, **2010** , 11 Suppl 12, S2	3.6	72
184	Detecting tissue-specific regulation of alternative splicing as a qualitative change in microarray data. *Nucleic Acids Research*, **2004**, 32, e180	20.1	71
183	Collaborative analysis of DRD4 and DAT genotypes in population-defined ADHD subtypes. *Journal of Child Psychology and Psychiatry and Allied Disciplines*, **2005**, 46, 1067-73	7.9	70
182	Linkage-disequilibrium mapping without genotyping. *Nature Genetics*, **1998**, 18, 225-30	36.3	69
181	Sequence variant in the laminin gamma1 (LAMC1) gene associated with familial pelvic organ prolapse. *Human Genetics*, **2007**, 120, 847-56	6.3	69
180	Dantrolene enhances antisense-mediated exon skipping in human and mouse models of Duchenne muscular dystrophy. *Science Translational Medicine*, **2012**, 4, 164ra160	17.5	67
179	Klinefelter syndrome as a model of anomalous cerebral laterality: testing gene dosage in the X chromosome pseudoautosomal region using a DNA microarray. *Genesis*, **1998**, 23, 215-29		66
178	Familial migraine with vertigo: no mutations found in CACNA1A. *American Journal of Medical Genetics Part A*, **1998**, 79, 148-51		66
177	Loss of Nardilysin, a Mitochondrial Co-chaperone for α-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. *Neuron*, **2017**, 93, 115-131	13.9	65
176	Exome sequencing identifies de novo gain of function missense mutation in KCND2 in identical twins with autism and seizures that slows potassium channel inactivation. *Human Molecular Genetics*, **2014**, 23, 3481-9	5.6	65
175	Pontocerebellar hypoplasia type 1: clinical spectrum and relevance of EXOSC3 mutations. *Neurology*, **2013**, 80, 438-46	6.5	65
174	Genome-wide scan of reading ability in affected sibling pairs with attention-deficit/hyperactivity disorder: unique and shared genetic effects. *Molecular Psychiatry*, **2004**, 9, 485-93	15.1	64
173	Proteinase and growth factor alterations revealed by gene microarray analysis of human diabetic corneas. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 3604-15		64
172	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. *European Journal of Human Genetics*, **2015**, 23, 1473-81	5.3	63
171	De novo mutation in CACNA1A caused acetazolamide-responsive episodic ataxia. *American Journal of Medical Genetics Part A*, **1998**, 77, 298-301		61
170	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. *Genetics in Medicine*, **2020**, 22, 490-499	8.1	60
169	Targeted massively parallel sequencing provides comprehensive genetic diagnosis for patients with disorders of sex development. *Clinical Genetics*, **2013**, 83, 35-43	4	59
168	Loss of annexin A1 expression in human breast cancer detected by multiple high-throughput analyses. *Biochemical and Biophysical Research Communications*, **2005**, 326, 218-27	3.4	59

167	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. *Brain*, **2016**, 139, 2877-2890	11.2	58
166	Improved variant discovery through local re-alignment of short-read next-generation sequencing data using SRMA. *Genome Biology*, **2010**, 11, R99	18.3	57
165	Differential gene expression in glioblastoma defined by ADC histogram analysis: relationship to extracellular matrix molecules and survival. *American Journal of Neuroradiology*, **2012**, 33, 1059-64	4.4	56
164	Demographically-Based Evaluation of Genomic Regions under Selection in Domestic Dogs. *PLoS Genetics*, **2016**, 12, e1005851	6	56
163	Mutations in DYNC2LI1 disrupt cilia function and cause short rib polydactyly syndrome. *Nature Communications*, **2015**, 6, 7092	17.4	55
162	Emerging genetic therapies to treat Duchenne muscular dystrophy. *Current Opinion in Neurology*, **2009**, 22, 532-8	7.1	55
161	CBCL pediatric bipolar disorder profile and ADHD: comorbidity and quantitative trait loci analysis. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2008**, 47, 1151-7	7.2	55
160	Detection of a microRNA signal in an in vivo expression set of mRNAs. *PLoS ONE*, **2007**, 2, e804	3.7	55
159	Celsius: a community resource for Affymetrix microarray data. *Genome Biology*, **2007**, 8, R112	18.3	54
158	Identification of allele-specific alternative mRNA processing via transcriptome sequencing. *Nucleic Acids Research*, **2012**, 40, e104	20.1	53
157	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. *Nature Genetics*, **2012**, 44, 709-13	36.3	52
156	A new episodic ataxia syndrome with linkage to chromosome 19q13. *Archives of Neurology*, **2007**, 64, 749-52		52
155	Molecular analysis of glioblastoma: pathway profiling and its implications for patient therapy. *Cancer Biology and Therapy*, **2003**, 2, 242-7	4.6	51
154	High density SNP association study of a major autism linkage region on chromosome 17. *Human Molecular Genetics*, **2007**, 16, 704-15	5.6	50
153	Next-generation mapping: a novel approach for detection of pathogenic structural variants with a potential utility in clinical diagnosis. *Genome Medicine*, **2017**, 9, 90	14.4	49
152	Expanding the phenotype of mutations in DICER1: mosaic missense mutations in the RNase IIIb domain of DICER1 cause GLOW syndrome. *Journal of Medical Genetics*, **2014**, 51, 294-302	5.8	48
151	High-density SNP association study of the 17q21 chromosomal region linked to autism identifies CACNA1G as a novel candidate gene. *Molecular Psychiatry*, **2010**, 15, 996-1005	15.1	48
150	Pooled genome-wide linkage data on 424 ADHD ASPs suggests genetic heterogeneity and a common risk locus at 5p13. *Molecular Psychiatry*, **2006**, 11, 5-8	15.1	48

149	Characterization of transformation related genes in oral cancer cells. *Oncogene*, **1998**, 16, 1921-30	9.2	47
148	A genome-wide linkage scan of familial benign recurrent vertigo: linkage to 22q12 with evidence of heterogeneity. *Human Molecular Genetics*, **2006**, 15, 251-8	5.6	46
147	Identification of retinol binding protein 1 promoter hypermethylation in isocitrate dehydrogenase 1 and 2 mutant gliomas. *Journal of the National Cancer Institute*, **2012**, 104, 1458-69	9.7	45
146	Protective properties of radio-chemoresistant glioblastoma stem cell clones are associated with metabolic adaptation to reduced glucose dependence. *PLoS ONE*, **2013**, 8, e80397	3.7	45
145	Pleiotropic Mechanisms Indicated for Sex Differences in Autism. *PLoS Genetics*, **2016**, 12, e1006425	6	45
144	Expanding the mutational spectrum of LZTR1 in schwannomatosis. *European Journal of Human Genetics*, **2015**, 23, 963-8	5.3	44
143	A comparison of gene expression profiles produced by SAGE, long SAGE, and oligonucleotide chips. *Genomics*, **2004**, 84, 631-6	4.3	44
142	De Novo variants in the KMT2A (MLL) gene causing atypical Wiedemann-Steiner syndrome in two unrelated individuals identified by clinical exome sequencing. *BMC Medical Genetics*, **2014**, 15, 49	2.1	43
141	Repetitive sequence environment distinguishes housekeeping genes. *Gene*, **2007**, 390, 153-65	3.8	43
140	ADHD candidate gene study in a population-based birth cohort: association with DBH and DRD2. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2007**, 46, 1614-21	7.2	42
139	Association of Sex With Recurrence of Autism Spectrum Disorder Among Siblings. *JAMA Pediatrics*, **2017**, 171, 1107-1112	8.3	41
138	IRF2BPL Is Associated with Neurological Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 245-260	11	39
137	DMD genotype correlations from the Duchenne Registry: Endogenous exon skipping is a factor in prolonged ambulation for individuals with a defined mutation subtype. *Human Mutation*, **2018**, 39, 1193-1202	4.7	39
136	Distinct gene expression profiles in adult mouse heart following targeted MAP kinase activation. *Physiological Genomics*, **2006**, 25, 50-9	3.6	37
135	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. *Genetics in Medicine*, **2019**, 21, 161-172	8.1	36
134	Phenotypic and genetic analysis of a large family with migraine-associated vertigo. *Headache*, **2008**, 48, 1460-7	4.2	35
133	A novel mutation in KCNA1 causes episodic ataxia without myokymia. *Human Mutation*, **2004**, 24, 536	4.7	35
132	Homozygous mutation in leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome. *Journal of Medical Genetics*, **2017**, 54, 399-403	5.8	34

131	Familial clustering and DRD4 effects on electroencephalogram measures in multiplex families with attention deficit/hyperactivity disorder. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 368-77	7.2	34
130	Rich annotation of DNA sequencing variants by leveraging the Ensembl Variant Effect Predictor with plugins. *Briefings in Bioinformatics*, **2015**, 16, 255-64	13.4	33
129	Ribosomal Proteins RPS11 and RPS20, Two Stress-Response Markers of Glioblastoma Stem Cells, Are Novel Predictors of Poor Prognosis in Glioblastoma Patients. *PLoS ONE*, **2015**, 10, e0141334	3.7	33
128	Cognitive functioning in affected sibling pairs with ADHD: familial clustering and dopamine genes. *Journal of Child Psychology and Psychiatry and Allied Disciplines*, **2008**, 49, 950-7	7.9	33
127	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants. *Genetics in Medicine*, **2019**, 21, 850-860	8.1	33
126	Decitabine immunosensitizes human gliomas to NY-ESO-1 specific T lymphocyte targeting through the Fas/Fas ligand pathway. *Journal of Translational Medicine*, **2011**, 9, 192	8.5	32
125	Improving the efficiency of genomic loci capture using oligonucleotide arrays for high throughput resequencing. *BMC Genomics*, **2009**, 10, 646	4.5	32
124	Autocrine endothelin-3/endothelin receptor B signaling maintains cellular and molecular properties of glioblastoma stem cells. *Molecular Cancer Research*, **2011**, 9, 1668-85	6.6	31
123	BMPER mutation in diaphanospondylodysostosis identified by ancestral autozygosity mapping and targeted high-throughput sequencing. *American Journal of Human Genetics*, **2010**, 87, 532-7	11	31
122	Association of progesterone receptor with migraine-associated vertigo. *Neurogenetics*, **2007**, 8, 195-200	3	31
121	Local alignment of two-base encoded DNA sequence. *BMC Bioinformatics*, **2009**, 10, 175	3.6	30
120	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. *Cilia*, **2016**, 5, 8	5.5	29
119	Bone morphogenetic protein 7 sensitizes O6-methylguanine methyltransferase expressing-glioblastoma stem cells to clinically relevant dose of temozolomide. *Molecular Cancer*, **2015**, 14, 189	42.1	29
118	Familial Clustering and DRD4 Effects on Electroencephalogram Measures in Multiplex Families With Attention Deficit/Hyperactivity Disorder. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 368-377	7.2	29
117	Osteogenic oxysterol, 20(S)-hydroxycholesterol, induces notch target gene expression in bone marrow stromal cells. *Journal of Bone and Mineral Research*, **2010**, 25, 782-95	6.3	29
116	Rare Genomic Variants Link Bipolar Disorder with Anxiety Disorders to CREB-Regulated Intracellular Signaling Pathways. *Frontiers in Psychiatry*, **2013**, 4, 154	5	28
115	Genomic mismatch scanning identifies human genomic DNA shared identical by descent. *Genomics*, **1998**, 47, 1-6	4.3	28
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