Stanley F Nelson

List of Publications by Year in Descending Order

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	Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling.. *Science Advances*, **2022**, 8, eabl5613	14.3	1
291	Modeling Patient-Specific Muscular Dystrophy Phenotypes and Therapeutic Responses in Reprogrammed Myotubes Engineered on Micromolded Gelatin Hydrogels.. *Frontiers in Cell and Developmental Biology*, **2022**, 10, 830415	5.7	1
290	Case Report: Whole Exome Sequencing Identifies Compound Heterozygous Variants in Gene Causing Juvenile Hypertrophic Cardiomyopathy.. *Frontiers in Cardiovascular Medicine*, **2021**, 8, 798985	5.4	0
289	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1649-1665	2.5	0
288	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. *Orphanet Journal of Rare Diseases*, **2021**, 16, 136	4.2	1
287	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. *Genetics in Medicine*, **2021**, 23, 1465-1473	8.1	1
286	Genotype-phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 90	14.4	2
285	Mosaic de novo gene variant associated with Prader-Willi syndrome. *Journal of Medical Genetics*, **2021**,	5.8	1
284	COVID-19 drug repurposing: Summary statistics on current clinical trials and promising untested candidates. *Transboundary and Emerging Diseases*, **2021**, 68, 313-317	4.2	16
283	Quantitative immuno-mass spectrometry imaging of skeletal muscle dystrophin. *Scientific Reports*, **2021**, 11, 1128	4.9	4
282	Expansion of NEUROD2 phenotypes to include developmental delay without seizures. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1076-1080	2.5	2
281	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. *Genetics in Medicine*, **2021**, 23, 1075-1085	8.1	2
280	Recessive ciliopathy mutations in primary endocardial fibroelastosis: a rare neonatal cardiomyopathy in a case of Alstrom syndrome. *Journal of Molecular Medicine*, **2021**, 99, 1623-1638	5.5	0
279	Novel NUDT2 variant causes intellectual disability and polyneuropathy. *Annals of Clinical and Translational Neurology*, **2020**, 7, 2320-2325	5.3	0
278	MPEG1/Perforin-2 Haploinsufficiency Associated Polymicrobial Skin Infections and Considerations for Interferon-□Therapy. *Frontiers in Immunology*, **2020**, 11, 601584	8.4	3
277	Genetic characterization and long-term management of severely affected siblings with intellectual developmental disorder with cardiac arrhythmia syndrome. *Molecular Genetics and Metabolism Reports*, **2020**, 23, 100582	1.8	2
276	Gene-environment regulation of chamber-specific maturation during hypoxemic perinatal circulatory transition. *Journal of Molecular Medicine*, **2020**, 98, 1009-1020	5.5	

275	Disseminated Coccidioidomycosis Treated with Interferon-and Dupilumab. *New England Journal of Medicine*, **2020**, 382, 2337-2343	59.2	10
274	The frontiers of sequencing in undiagnosed neurodevelopmental diseases. *Current Opinion in Genetics and Development*, **2020**, 65, 76-83	4.9	1
273	Ppp1r1b-lncRNA inhibits PRC2 at myogenic regulatory genes to promote cardiac and skeletal muscle development in mouse and human. *Rna*, **2020**, 26, 481-491	5.8	14
272	Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing. *Neuropathology*, **2020**, 40, 302-307	2	3
271	GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. *Genetics in Medicine*, **2020**, 22, 878-888	8.1	9
270	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. *Human Mutation*, **2020**, 41, 487-501	4.7	24
269	Congenital myasthenic syndrome caused by a frameshift insertion mutation in. *Neurology: Genetics*, **2020**, 6, e468	3.8	3
268	Mitchell-Riley syndrome iPSCs exhibit reduced pancreatic endoderm differentiation due to a mutation in. *Development (Cambridge)*, **2020**, 147,	6.6	3
267	A well-tolerated core needle muscle biopsy process suitable for children and adults. *Muscle and Nerve*, **2020**, 62, 688-698	3.4	5
266	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. *Genetics in Medicine*, **2020**, 22, 490-499	8.1	60
265	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Signaling. *Biological Psychiatry*, **2020**, 87, 100-112	7.9	19
264	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region. *Epilepsia*, **2019**, 60, 406-418	6.4	26
263	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. *Human Mutation*, **2019**, 40, 1115-1126	4.7	7
262	IgG4-related disease: Association with a rare gene variant expressed in cytotoxic T cells. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e686	2.3	6
261	A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing. *Journal of Genetic Counseling*, **2019**, 28, 213-228	2.5	4
260	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. *Genetics in Medicine*, **2019**, 21, 161-172	8.1	36
259	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. *Clinical Imaging*, **2019**, 58, 108-113	2.7	4
258	Targeting RyR Activity Boosts Antisense Exon 44 and 45 Skipping in Human DMD Skeletal or Cardiac Muscle Culture Models. *Molecular Therapy - Nucleic Acids*, **2019**, 18, 580-589	10.7	7

257	SLC35A2-CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. *Human Mutation*, **2019**, 40, 908-925	4.7	23
256	Large in-frame 5R deletions in DMD associated with mild Duchenne muscular dystrophy: Two case reports and a review of the literature. *Neuromuscular Disorders*, **2019**, 29, 863-873	2.9	4
255	Gene-environment regulatory circuits of right ventricular pathology in tetralogy of fallot. *Journal of Molecular Medicine*, **2019**, 97, 1711-1722	5.5	3
254	De novo truncating variants in the intronless IRF2BPL are responsible for developmental epileptic encephalopathy. *Genetics in Medicine*, **2019**, 21, 1008-1014	8.1	18
253	Evidence-Based Consensus and Systematic Review on Reducing the Time to Diagnosis of Duchenne Muscular Dystrophy. *Journal of Pediatrics*, **2019**, 204, 305-313.e14	3.6	17
252	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. *American Journal of Human Genetics*, **2019**, 104, 164-178	11	27
251	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants. *Genetics in Medicine*, **2019**, 21, 850-860	8.1	33
250	Dusp6 is a genetic modifier of growth through enhanced ERK activity. *Human Molecular Genetics*, **2019**, 28, 279-289	5.6	3
249	Variant in human POFUT1 reduces enzymatic activity and likely causes a recessive microcephaly, global developmental delay with cardiac and vascular features. *Glycobiology*, **2018**, 28, 276-283	5.8	16
248	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
247	Calpain 3 and CaMKII signaling are required to induce HSP70 necessary for adaptive muscle growth after atrophy. *Human Molecular Genetics*, **2018**, 27, 1642-1653	5.6	15
246	A homozygous loss-of-function mutation causes growth delay, frequent seizures and severe intellectual disability. *ELife*, **2018**, 7,	8.9	24
245	IRF2BPL Is Associated with Neurological Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 245-260	11	39
244	Identification of novel candidate genes for 46,XY disorders of sex development (DSD) using a C57BL/6J-Y mouse model. *Biology of Sex Differences*, **2018**, 9, 8	9.3	7
243	Repurposing Dantrolene for Long-Term Combination Therapy to Potentiate Antisense-Mediated DMD Exon Skipping in the mdx Mouse. *Molecular Therapy - Nucleic Acids*, **2018**, 11, 180-191	10.7	5
242	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
241	Calculating the statistical significance of rare variants causal for Mendelian and complex disorders. *BMC Medical Genomics*, **2018**, 11, 53	3.7	9
240	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. *American Journal of Human Genetics*, **2018**, 103, 948-967	11	8

239	Cardiac MRI biomarkers for Duchenne muscular dystrophy. *Biomarkers in Medicine*, **2018**, 12, 1271-1289	2.3	11
238	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. *New England Journal of Medicine*, **2018**, 379, 2131-2139	59.2	129
237	Validation and Detection of Exon Skipping Boosters in DMD Patient Cell Models and mdx Mouse. *Methods in Molecular Biology*, **2018**, 1828, 309-326	1.4	4
236	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. *American Journal of Human Genetics*, **2017**, 100, 185-192	11	102
235	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. *American Journal of Human Genetics*, **2017**, 100, 343-351	11	23
234	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
233	FDA Approval of Eteplirsen for Muscular Dystrophy. *JAMA - Journal of the American Medical Association*, **2017**, 317, 1480	27.4	13
232	Loss of Nardilysin, a Mitochondrial Co-chaperone for -Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. *Neuron*, **2017**, 93, 115-131	13.9	65
231	Association of Sex With Recurrence of Autism Spectrum Disorder Among Siblings. *JAMA Pediatrics*, **2017**, 171, 1107-1112	8.3	41
230	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
229	Rare deleterious mutations are associated with disease in bipolar disorder families. *Molecular Psychiatry*, **2017**, 22, 1009-1014	15.1	23
228	A Path to Implement Precision Child Health Cardiovascular Medicine. *Frontiers in Cardiovascular Medicine*, **2017**, 4, 36	5.4	7
227	Leveraging ancestry to improve causal variant identification in exome sequencing for monogenic disorders. *European Journal of Human Genetics*, **2016**, 24, 113-9	5.3	0
226	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. *Brain*, **2016**, 139, 2877-2890	11.2	58
225	Truncating mutations in APP cause a distinct neurological phenotype. *Annals of Neurology*, **2016**, 80, 456-60	9.4	8
224	Joint mouse-human phenome-wide association to test gene function and disease risk. *Nature Communications*, **2016**, 7, 10464	17.4	91
223	Missense-depleted regions in population exomes implicate ras superfamily nucleotide-binding protein alteration in patients with brain malformation. *Npj Genomic Medicine*, **2016**, 1,	6.2	26
222	Reciprocal Regulation of the Cardiac Epigenome by Chromatin Structural Proteins Hmgb and Ctcf: IMPLICATIONS FOR TRANSCRIPTIONAL REGULATION. *Journal of Biological Chemistry*, **2016**, 291, 15428-46	5.4	18

221	The case for eteplirsen: Paving the way for precision medicine. *Molecular Genetics and Metabolism*, **2016**, 118, 70-1	3.7	10
220	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. *Cilia*, **2016**, 5, 8	5.5	29
219	Failure to up-regulate transcription of genes necessary for muscle adaptation underlies limb girdle muscular dystrophy 2A (calpainopathy). *Human Molecular Genetics*, **2016**, 25, 2194-2207	5.6	21
218	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
217	Clinical aggressiveness of malignant gliomas is linked to augmented metabolism of amino acids. *Journal of Neuro-Oncology*, **2016**, 128, 57-66	4.8	17
216	Exome Sequencing Identified a Splice Site Mutation in FHL1 that Causes Uruguay Syndrome, an X-Linked Disorder With Skeletal Muscle Hypertrophy and Premature Cardiac Death. *Circulation: Cardiovascular Genetics*, **2016**, 9, 130-5		7
215	Demographically-Based Evaluation of Genomic Regions under Selection in Domestic Dogs. *PLoS Genetics*, **2016**, 12, e1005851	6	56
214	Pleiotropic Mechanisms Indicated for Sex Differences in Autism. *PLoS Genetics*, **2016**, 12, e1006425	6	45
213	The functional O-mannose glycan on □dystroglycan contains a phospho-ribitol primed for matriglycan addition. *ELife*, **2016**, 5,	8.9	73
212	Role of Mir-34 Upregulation in Disruption of c-Myc, c-Myb and NOTCH Signaling in Diamond-Blackfan Anemia. *Blood*, **2016**, 128, 3895-3895	2.2	
211	Genomic predictors of remission to antidepressant treatment in geriatric depression using genome-wide expression analyses: a pilot study. *International Journal of Geriatric Psychiatry*, **2016**, 31, 510-7	3.9	14
210	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. *Annals of the New York Academy of Sciences*, **2016**, 1366, 49-60	6.5	20
209	Mutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior Change. *Archives of Iranian Medicine*, **2016**, 19, 87-91	2.4	4
208	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. *Nature Neuroscience*, **2015**, 18, 199-209	25.5	572
207	Mutations in DYNC2LI1 disrupt cilia function and cause short rib polydactyly syndrome. *Nature Communications*, **2015**, 6, 7092	17.4	55
206	Loss of ADAM17 is associated with severe multiorgan dysfunction. *Human Pathology*, **2015**, 46, 923-8	3.7	23
205	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
204	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

203	Expanding the mutational spectrum of LZTR1 in schwannomatosis. *European Journal of Human Genetics*, **2015**, 23, 963-8	5.3	44
202	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
201	What can Duchenne Connect teach us about treating Duchenne muscular dystrophy?. *Current Opinion in Neurology*, **2015**, 28, 535-41	7.1	6
200	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
199	Loss of the scavenger mRNA decapping enzyme DCPS causes syndromic intellectual disability with neuromuscular defects. *Human Molecular Genetics*, **2015**, 24, 3163-71	5.6	20
198	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
197	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
196	Whole exome sequencing detects homozygosity for ABCA4 p.Arg602Trp missense mutation in a pediatric patient with rapidly progressive retinal dystrophy. *BMC Medical Genetics*, **2014**, 15, 11	2.1	8
195	Targeted therapy resistance mediated by dynamic regulation of extrachromosomal mutant EGFR DNA. *Science*, **2014**, 343, 72-6	33.3	316
194	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
193	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
192	High-throughput profiling of influenza A virus hemagglutinin gene at single-nucleotide resolution. *Scientific Reports*, **2014**, 4, 4942	4.9	113
191	Genome sequencing highlights the dynamic early history of dogs. *PLoS Genetics*, **2014**, 10, e1004016	6	372
190	PDE5 inhibition alleviates functional muscle ischemia in boys with Duchenne muscular dystrophy. *Neurology*, **2014**, 82, 2085-91	6.5	79
189	X chromosome exome sequencing reveals a novel ALG13 mutation in a nonsyndromic intellectual disability family with multiple affected male siblings. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 164-9	2.5	19
188	Interpreting whole-genome sequencing. *JAMA - Journal of the American Medical Association*, **2014**, 312, 296	27.4	1
187	Exome sequencing in the clinical diagnosis of sporadic or familial cerebellar ataxia. *JAMA Neurology* , **2014**, 71, 1237-46	17.2	171
186	Clinical exome sequencing for genetic identification of rare Mendelian disorders. *JAMA - Journal of the American Medical Association*, **2014**, 312, 1880-7	27.4	640

185	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
184	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
183	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
182	Online self-report data for duchenne muscular dystrophy confirms natural history and can be used to assess for therapeutic benefits. *PLOS Currents*, **2014**, 6,		22
181	Identification of somatic and germline mutations using whole exome sequencing of congenital acute lymphoblastic leukemia. *BMC Cancer*, **2013**, 13, 55	4.8	18
180	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
179	Targeted massively parallel sequencing provides comprehensive genetic diagnosis for patients with disorders of sex development. *Clinical Genetics*, **2013**, 83, 35-43	4	59
178	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
177	Whole exome sequencing of pediatric gastric adenocarcinoma reveals an atypical presentation of Li-Fraumeni syndrome. *Pediatric Blood and Cancer*, **2013**, 60, 570-4	3	18
176	Therapeutic potential of HSP90 inhibition for neurofibromatosis type 2. *Clinical Cancer Research*, **2013**, 19, 3856-70	12.9	27
175	Pontocerebellar hypoplasia type 1: clinical spectrum and relevance of EXOSC3 mutations. *Neurology*, **2013**, 80, 438-46	6.5	65
174	Exome sequencing finds a novel PCSK1 mutation in a child with generalized malabsorptive diarrhea and diabetes insipidus. *Journal of Pediatric Gastroenterology and Nutrition*, **2013**, 57, 759-67	2.8	25
173	SGK196 is a glycosylation-specific O-mannose kinase required for dystroglycan function. *Science*, **2013**, 341, 896-9	33.3	162
172	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
171	Rare Genomic Variants Link Bipolar Disorder with Anxiety Disorders to CREB-Regulated Intracellular Signaling Pathways. *Frontiers in Psychiatry*, **2013**, 4, 154	5	28
170	Functional consequences of a novel variant of PCSK1. *PLoS ONE*, **2013**, 8, e55065	3.7	18
169	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
168	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

167	Dantrolene enhances antisense-mediated exon skipping in human and mouse models of Duchenne muscular dystrophy. *Science Translational Medicine*, **2012**, 4, 164ra160	17.5	67
166	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. *Human Genetics*, **2012**, 131, 565-79	6.3	150
165	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. *Nature Genetics*, **2012**, 44, 193-9	36.3	123
164	Cold urticaria, immunodeficiency, and autoimmunity related to PLCG2 deletions. *New England Journal of Medicine*, **2012**, 366, 330-8	59.2	288
163	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. *Nature Genetics*, **2012**, 44, 709-13	36.3	52
162	Molecular diagnosis of putative Stargardt Disease probands by exome sequencing. *BMC Medical Genetics*, **2012**, 13, 67	2.1	24
161	Genome-wide association study of intelligence: additive effects of novel brain expressed genes. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2012**, 51, 432-440.e2	7.2	15
160	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
159	Melanoma whole-exome sequencing identifies (V600E)B-RAF amplification-mediated acquired B-RAF inhibitor resistance. *Nature Communications*, **2012**, 3, 724	17.4	500
158	Exome sequencing identifies PDE4D mutations in acrodysostosis. *American Journal of Human Genetics*, **2012**, 90, 746-51	11	114
157	Sex-specific influence of DRD2 on ADHD-type temperament in a large population-based birth cohort. *Psychiatric Genetics*, **2012**, 22, 197-201	2.9	19
156	Identification of allele-specific alternative mRNA processing via transcriptome sequencing. *Nucleic Acids Research*, **2012**, 40, e104	20.1	53
155	High throughput screening for compounds that alter muscle cell glycosylation identifies new role for N-glycans in regulating sarcolemmal protein abundance and laminin binding. *Journal of Biological Chemistry*, **2012**, 287, 22759-70	5.4	13
154	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
153	Mutations in the PCNA-binding domain of CDKN1C cause IMAGe syndrome. *Nature Genetics*, **2012**, 44, 788-92	36.3	140
152	Individual common variants exert weak effects on the risk for autism spectrum disorders. *Human Molecular Genetics*, **2012**, 21, 4781-92	5.6	279
151	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
150	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. *Nature Genetics*, **2012**, 44, 575-80	36.3	183

149	DNA Sequencing of Maternal Plasma to Detect Down Syndrome. *Obstetrical and Gynecological Survey*, **2012**, 67, 86-88	2.4	2
148	Rethinking clinical practice: clinical implementation of exome sequencing. *Personalized Medicine*, **2012**, 9, 785-787	2.2	4
147	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
146	Predicting the severity of Duchenne muscular dystrophy: implications for treatment. *Neurology*, **2011**, 76, 208-9	6.5	6
145	DNA sequencing of maternal plasma to detect Down syndrome: an international clinical validation study. *Genetics in Medicine*, **2011**, 13, 913-20	8.1	670
144	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
143	Exome sequencing-based copy-number variation and loss of heterozygosity detection: ExomeCNV. *Bioinformatics*, **2011**, 27, 2648-54	7.2	305
142	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
141	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
140	Genomic identification of significant targets in ciliochoroidal melanoma **2011**, 52, 3018-22		20
139	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
138	Phenotype sequencing: identifying the genes that cause a phenotype directly from pooled sequencing of independent mutants. *PLoS ONE*, **2011**, 6, e16517	3.7	18
137	Characterization of three cell lines derived from fine needle biopsy of choroidal melanoma with metastatic outcome. *Molecular Vision*, **2011**, 17, 607-15	2.3	6
136	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
135	Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **2010**, 466, 368-72	50.4	1499
134	Melanomas acquire resistance to B-RAF(V600E) inhibition by RTK or N-RAS upregulation. *Nature*, **2010**, 468, 973-7	50.4	1678
133	A genome-wide scan for common alleles affecting risk for autism. *Human Molecular Genetics*, **2010**, 19, 4072-82	5.6	443
132	Lethal skeletal dysplasia in mice and humans lacking the golgin GMAP-210. *New England Journal of Medicine*, **2010**, 362, 206-16	59.2	105

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, 368-377	7.2	29
130	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
129	U87MG decoded: the genomic sequence of a cytogenetically aberrant human cancer cell line. *PLoS Genetics*, **2010**, 6, e1000832	6	195
128	Improved variant discovery through local re-alignment of short-read next-generation sequencing data using SRMA. *Genome Biology*, **2010**, 11, R99	18.3	57
127	Genomic landscape of meningiomas. *Brain Pathology*, **2010**, 20, 751-62	6	97
126	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
125	Identification of candidate tumor oncogenes by integrative molecular analysis of choroidal melanoma fine-needle aspiration biopsy specimens. *JAMA Ophthalmology*, **2010**, 128, 1170-7		15
124	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
123	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
122	Biomarkers to predict antidepressant response. *Current Psychiatry Reports*, **2010**, 12, 553-62	9.1	111
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