Stanley F Nelson

List of Publications by Year
in descending order

298	43,282	1893	2385
papers	citations	102 h-index	198 g-index

316	316	316	57838
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	21.4	2,067
2	Shotgun bisulphite sequencing of the Arabidopsis genome reveals DNA methylation patterning. Nature, 2008, 452, 215-219.	27.8	2,039
3	Melanomas acquire resistance to B-RAF(V600E) inhibition by RTK or N-RAS upregulation. Nature, 2010, 468, 973-977.	27.8	1,944
4	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
5	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. Science, 2008, 320, 539-543.	12.6	1,654
6	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
7	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, 20007-20012.	7.1	927
8	High-throughput oncogene mutation profiling in human cancer. Nature Genetics, 2007, 39, 347-351.	21.4	927
9	Resolving Individuals Contributing Trace Amounts of DNA to Highly Complex Mixtures Using High-Density SNP Genotyping Microarrays. PLoS Genetics, 2008, 4, e1000167.	3.5	892
10	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
11	DNA sequencing of maternal plasma to detect Down syndrome: An international clinical validation study. Genetics in Medicine, 2011, 13, 913-920.	2.4	809
12	Linkage, Association, and Gene-Expression Analyses Identify CNTNAP2 as an Autism-Susceptibility Gene. American Journal of Human Genetics, 2008, 82, 150-159.	6.2	738
13	Psychiatric genome-wide association study analyses implicate neuronal, immune and histone pathways. Nature Neuroscience, 2015, 18, 199-209.	14.8	701
14	Gene Expression Profiling of Gliomas Strongly Predicts Survival. Cancer Research, 2004, 64, 6503-6510.	0.9	659
15	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, 17402-17407.	7.1	606
16	Melanoma whole-exome sequencing identifies V600EB-RAF amplification-mediated acquired B-RAF inhibitor resistance. Nature Communications, 2012, 3, 724.	12.8	567
17	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	2.9	538
18	Genome Sequencing Highlights the Dynamic Early History of Dogs. PLoS Genetics, 2014, 10, e1004016.	3.5	481

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19	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.	2.4	471
20	Targeted Therapy Resistance Mediated by Dynamic Regulation of Extrachromosomal Mutant EGFR DNA. Science, 2014, 343, 72-76.	12.6	460
21	BFAST: An Alignment Tool for Large Scale Genome Resequencing. PLoS ONE, 2009, 4, e7767.	2.5	444
22	Cold Urticaria, Immunodeficiency, and Autoimmunity Related toPLCG2Deletions. New England Journal of Medicine, 2012, 366, 330-338.	27.0	391
23	Gene Expression Profile Correlates with T-Cell Infiltration and Relative Survival in Glioblastoma Patients Vaccinated with Dendritic Cell Immunotherapy. Clinical Cancer Research, 2011, 17, 1603-1615.	7.0	378
24	C-terminal truncations in human 3â€²-5â€² DNA exonuclease TREX1 cause autosomal dominant retinal vasculopathy with cerebral leukodystrophy. Nature Genetics, 2007, 39, 1068-1070.	21.4	366
25	Mutations in a Human ROBO Gene Disrupt Hindbrain Axon Pathway Crossing and Morphogenesis. Science, 2004, 304, 1509-1513.	12.6	361
26	Exome sequencing-based copy-number variation and loss of heterozygosity detection: ExomeCNV. Bioinformatics, 2011, 27, 2648-2654.	4.1	357
27	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	2.9	334
28	Genome-wide copy number variation study associates metabotropic glutamate receptor gene networks with attention deficit hyperactivity disorder. Nature Genetics, 2012, 44, 78-84.	21.4	334
29	Gene connectivity, function, and sequence conservation: predictions from modular yeast co-expression networks. BMC Genomics, 2006, 7, 40.	2.8	327
30	Gain-of-function mutations in TRPV4 cause autosomal dominant brachyolmia. Nature Genetics, 2008, 40, 999-1003.	21.4	320
31	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, 533-540.	11.1	307
32	A Genomewide Scan for Loci Involved in Attention-Deficit/Hyperactivity Disorder. American Journal of Human Genetics, 2002, 70, 1183-1196.	6.2	304
33	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, 12741-12746.	7.1	303
34	Epidermal Growth Factor Receptor Activation in Glioblastoma through Novel Missense Mutations in the Extracellular Domain. PLoS Medicine, 2006, 3, e485.	8.4	298
35	Gene expression profiling identifies molecular subtypes of gliomas. Oncogene, 2003, 22, 4918-4923.	5.9	264
36	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	27.0	261

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37	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, 14676-14679.	7.1	256
38	Oxysterols Are Novel Activators of the Hedgehog Signaling Pathway in Pluripotent Mesenchymal Cells. Journal of Biological Chemistry, 2007, 282, 8959-8968.	3.4	254
39	Identification of molecular subtypes of glioblastoma by gene expression profiling. Oncogene, 2003, 22, 2361-2373.	5.9	247
40	A Genetic Analysis of Neural Progenitor Differentiation. Neuron, 2001, 29, 325-339.	8.1	243
41	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.	7.2	242
42	U87MG Decoded: The Genomic Sequence of a Cytogenetically Aberrant Human Cancer Cell Line. PLoS Genetics, 2010, 6, e1000832.	3.5	229
43	Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. Nature Genetics, 2012, 44, 704-708.	21.4	216
44	Primary Glioblastomas Express Mesenchymal Stem-Like Properties. Molecular Cancer Research, 2006, 4, 607-619.	3.4	215
45	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nature Genetics, 2012, 44, 575-580.	21.4	212
46	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211
47	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. JAMA Neurology, 2014, 71, 1237.	9.0	211
48	Genetic Linkage of Attention-Deficit/Hyperactivity Disorder on Chromosome 16p13, in a Region Implicated in Autism. American Journal of Human Genetics, 2002, 71, 959-963.	6.2	210
49	A Genomewide Scan for Attention-Deficit/Hyperactivity Disorder in an Extended Sample: Suggestive Linkage on 17p11. American Journal of Human Genetics, 2003, 72, 1268-1279.	6.2	206
50	Evidence that the dopamine D4 receptor is a susceptibility gene in attention deficit hyperactivity disorder. Molecular Psychiatry, 1998, 3, 427-430.	7.9	198
51	SGK196 Is a Glycosylation-Specific O -Mannose Kinase Required for Dystroglycan Function. Science, 2013, 341, 896-899.	12.6	197
52	Progressive Ataxia Due to a Missense Mutation in a Calcium-Channel Gene. American Journal of Human Genetics, 1997, 61, 1078-1087.	6.2	191
53	Joint mouseâ€“human phenome-wide association to test gene function and disease risk. Nature Communications, 2016, 7, 10464.	12.8	190
54	DNA-microarray analysis of brain cancer: molecular classification for therapy. Nature Reviews Neuroscience, 2004, 5, 782-792.	10.2	189

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55	Identification of EpCAM as the Gene for Congenital Tufting Enteropathy. Gastroenterology, 2008, 135, 429-437.	1.3	185
56	Distinct Transcription Profiles of Primary and Secondary Glioblastoma Subgroups. Cancer Research, 2006, 66, 159-167.	0.9	182
57	Gene expression analysis of glioblastomas identifies the major molecular basis for the prognostic benefit of younger age. BMC Medical Genomics, 2008, 1, 52.	1.5	181
58	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	3.8	180
59	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, 5563-5568.	7.1	173
60	Exome Sequencing for the Diagnosis of 46,XY Disorders of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E333-E344.	3.6	172
61	Detection of differentially expressed genes in primary tumor tissues using representational differences analysis coupled to microarray hybridization. Nucleic Acids Research, 1998, 26, 3059-3065.	14.5	169
62	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, 531-536.	7.9	169
63	Mutations in the PCNA-binding domain of CDKN1C cause IMAGe syndrome. Nature Genetics, 2012, 44, 788-792.	21.4	169
64	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, 348-356.	6.2	168
65	Evidence for Sex-Specific Risk Alleles in Autism Spectrum Disorder. American Journal of Human Genetics, 2004, 75, 1117-1123.	6.2	165
66	Metaâ€analysis of genomeâ€wide linkage scans of attention deficit hyperactivity disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1392-1398.	1.7	160
67	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	21.4	157
68	FXR regulates organic solute transporters Î± and Î² in the adrenal gland, kidney, and intestine. Journal of Lipid Research, 2006, 47, 201-214.	4.2	153
69	ERBB3 and NGFR mark a distinct skeletal muscle progenitor cell in human development and hPSCs. Nature Cell Biology, 2018, 20, 46-57.	10.3	151
70	Ciliary Abnormalities Due to Defects in the Retrograde Transport Protein DYNC2H1 in Short-Rib Polydactyly Syndrome. American Journal of Human Genetics, 2009, 84, 542-549.	6.2	149
71	Genome-wide analysis of single-nucleotide polymorphisms in human expressed sequences. Nature Genetics, 2000, 26, 233-236.	21.4	147
72	Multiple self-healing squamous epithelioma is caused by a disease-specific spectrum of mutations in TGFBR1. Nature Genetics, 2011, 43, 365-369.	21.4	147

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73	High-throughput profiling of influenza A virus hemagglutinin gene at single-nucleotide resolution. Scientific Reports, 2014, 4, 4942.	3.3	147
74	Relationship between Gene Expression and Enhancement in Glioblastoma Multiforme: Exploratory DNA Microarray Analysis. Radiology, 2008, 249, 268-277.	7.3	146
75	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.	2.9	144
76	Familial episodic ataxia: Clinical heterogeneity in four families linked to chromosome 19p. Annals of Neurology, 1997, 41, 8-16.	5.3	143
77	Replication of Autism Linkage: Fine-Mapping Peak at 17q21. American Journal of Human Genetics, 2005, 76, 1050-1056.	6.2	142
78	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
79	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, 1575-1583.	0.5	141
80	The apolipoprotein E Îµ4 allele is not a significant risk factor for frontotemporal dementia. Annals of Neurology, 1998, 44, 134-138.	5.3	136
81	Maternal embryonic leucine zipper kinase (MELK) regulates multipotent neural progenitor proliferation. Journal of Cell Biology, 2005, 170, 413-427.	5.2	136
82	Fyn and Src Are Effectors of Oncogenic Epidermal Growth Factor Receptor Signaling in Glioblastoma Patients. Cancer Research, 2009, 69, 6889-6898.	0.9	136
83	Biomarkers to Predict Antidepressant Response. Current Psychiatry Reports, 2010, 12, 553-562.	4.5	136
84	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genetics in Medicine, 2020, 22, 490-499.	2.4	136
85	Genomic mismatch scanning: a new approach to genetic linkage mapping. Nature Genetics, 1993, 4, 11-18.	21.4	134
86	EGFR Mutation-Induced Alternative Splicing of Max Contributes to Growth of Glycolytic Tumors in Brain Cancer. Cell Metabolism, 2013, 17, 1000-1008.	16.2	130
87	Exome Sequencing Identifies PDE4D Mutations in Acrodysostosis. American Journal of Human Genetics, 2012, 90, 746-751.	6.2	128
88	Genomic Landscape of Meningiomas. Brain Pathology, 2010, 20, 751-762.	4.1	124
89	Lethal Skeletal Dysplasia in Mice and Humans Lacking the Golgin GMAP-210. New England Journal of Medicine, 2010, 362, 206-216.	27.0	122
90	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.	0.5	122

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91	Attention Deficit Hyperactivity Disorder: Fine Mapping Supports Linkage to 5p13, 6q12, 16p13, and 17p11. American Journal of Human Genetics, 2004, 75, 661-668.	6.2	121
92	Assessing the necessity of confirmatory testing for exome-sequencing results in a clinical molecular diagnostic laboratory. Genetics in Medicine, 2014, 16, 510-515.	2.4	121
93	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, 72-79.	6.2	120
94	Accuracy of phenotyping of autistic children based on internet implemented parent report. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1119-1126.	1.7	120
95	Annexin A6 modifies muscular dystrophy by mediating sarcolemmal repair. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 6004-6009.	7.1	117
96	Disruption of POF1B Binding to Nonmuscle Actin Filaments Is Associated with Premature Ovarian Failure. American Journal of Human Genetics, 2006, 79, 113-119.	6.2	116
97	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.	6.2	115
98	Differential Induction of Glioblastoma Migration and Growth by Two Forms of Pleiotrophin. Journal of Biological Chemistry, 2005, 280, 26953-26964.	3.4	112
99	Molecular properties of CD133+ glioblastoma stem cells derived from treatment-refractory recurrent brain tumors. Journal of Neuro-Oncology, 2009, 94, 1-19.	2.9	111
100	Relationship between Survival and Edema in Malignant Gliomas: Role of Vascular Endothelial Growth Factor and Neuronal Pentraxin 2. Clinical Cancer Research, 2007, 13, 2592-2598.	7.0	108
101	Biased paternal transmission of SNAP-25 risk alleles in attention-deficit hyperactivity disorder. Molecular Psychiatry, 2003, 8, 309-315.	7.9	107
102	Transmission disequilibrium testing of dopamine-related candidate gene polymorphisms in ADHD: confirmation of association of ADHD with DRD4 and DRD5. Molecular Psychiatry, 2004, 9, 711-717.	7.9	105
103	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. European Journal of Human Genetics, 2015, 23, 1473-1481.	2.8	101
104	Association of the cannabinoid receptor gene (CNR1) with ADHD and postâ€traumatic stress disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1488-1494.	1.7	99
105	The functional O-mannose glycan on Î±-dystroglycan contains a phospho-ribitol primed for matriglycan addition. ELife, 2016, 5, .	6.0	98
106	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	8.1	95
107	Temperament and Character Profiles and the Dopamine D4 Receptor Gene in ADHD. American Journal of Psychiatry, 2005, 162, 906-913.	7.2	94
108	PDE5 inhibition alleviates functional muscle ischemia in boys with Duchenne muscular dystrophy. Neurology, 2014, 82, 2085-2091.	1.1	94

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109	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, 3481-3489.	2.9	90
110	Detecting tissue-specific regulation of alternative splicing as a qualitative change in microarray data. Nucleic Acids Research, 2004, 32, e180-e180.	14.5	87
111	Stem cell associated gene expression in glioblastoma multiforme: relationship to survival and the subventricular zone. Journal of Neuro-Oncology, 2010, 96, 359-367.	2.9	86
112	Next-generation mapping: a novel approach for detection of pathogenic structural variants with a potential utility in clinical diagnosis. Genome Medicine, 2017, 9, 90.	8.2	86
113	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.1	84
114	Linkage-disequilibrium mapping without genotyping. Nature Genetics, 1998, 18, 225-230.	21.4	82
115	Loss of CHSY1, a Secreted FRINGE Enzyme, Causes Syndromic Brachydactyly in Humans via Increased NOTCH Signaling. American Journal of Human Genetics, 2010, 87, 768-778.	6.2	82
116	Collaborative analysis of DRD4 and DAT genotypes in population-defined ADHD subtypes. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2005, 46, 1067-1073.	5.2	79
117	Mutations in DYNC2LI1 disrupt cilia function and cause short rib polydactyly syndrome. Nature Communications, 2015, 6, 7092.	12.8	79
118	Familial migraine with vertigo: No mutations found in CACNA1A. American Journal of Medical Genetics Part A, 1998, 79, 148-151.	2.4	78
119	SeqWare Query Engine: storing and searching sequence data in the cloud. BMC Bioinformatics, 2010, 11, S2.	2.6	78
120	Klinefelter's syndrome as a model of anomalous cerebral laterality: Testing gene dosage in the X chromosome pseudoautosomal region using a DNA microarray. Genesis, 1998, 23, 215-229.	2.1	77
121	Dantrolene Enhances Antisense-Mediated Exon Skipping in Human and Mouse Models of Duchenne Muscular Dystrophy. Science Translational Medicine, 2012, 4, 164ra160.	12.4	77
122	Demographically-Based Evaluation of Genomic Regions under Selection in Domestic Dogs. PLoS Genetics, 2016, 12, e1005851.	3.5	77
123	Sequence variant in the laminin Î³1 (LAMC1) gene associated with familial pelvic organ prolapse. Human Genetics, 2007, 120, 847-856.	3.8	76
124	Proteinase and Growth Factor Alterations Revealed by Gene Microarray Analysis of Human Diabetic Corneas. , 2005, 46, 3604.		75
125	Identification of allele-specific alternative mRNA processing via transcriptome sequencing. Nucleic Acids Research, 2012, 40, e104-e104.	14.5	74
126	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. Brain, 2016, 139, 2877-2890.	7.6	74

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127	De novo mutation in CACNA1A caused acetazolamide-responsive episodic ataxia. , 1998, 77, 298-301.		73
128	Genome-wide scan of reading ability in affected sibling pairs with attention-deficit/hyperactivity disorder: unique and shared genetic effects. Molecular Psychiatry, 2004, 9, 485-493.	7.9	72
129	Targeted massively parallel sequencing provides comprehensive genetic diagnosis for patients with disorders of sex development. Clinical Genetics, 2013, 83, 35-43.	2.0	72
130	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
131	Loss of annexin A1 expression in human breast cancer detected by multiple high-throughput analyses. Biochemical and Biophysical Research Communications, 2004, 326, 218-227.	2.1	68
132	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713.	21.4	68
133	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	2.4	68
134	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.	7.9	67
135	Association of Sex With Recurrence of Autism Spectrum Disorder Among Siblings. JAMA Pediatrics, 2017, 171, 1107.	6.2	66
136	A New Episodic Ataxia Syndrome With Linkage to Chromosome 19q13. Archives of Neurology, 2007, 64, 749.	4.5	65
137	Expanding the phenotype of mutations in DICER1: mosaic missense mutations in the RNase IIIb domain ofDICER1cause GLOW syndrome. Journal of Medical Genetics, 2014, 51, 294-302.	3.2	65
138	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.	2.5	65
139	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, 1151-1157.	0.5	64
140	Differential Gene Expression in Glioblastoma Defined by ADC Histogram Analysis: Relationship to Extracellular Matrix Molecules and Survival. American Journal of Neuroradiology, 2012, 33, 1059-1064.	2.4	64
141	Pleiotropic Mechanisms Indicated for Sex Differences in Autism. PLoS Genetics, 2016, 12, e1006425.	3.5	64
142	Emerging genetic therapies to treat Duchenne muscular dystrophy. Current Opinion in Neurology, 2009, 22, 532-538.	3.6	63
143	Homozygous mutation inNUP107leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome. Journal of Medical Genetics, 2017, 54, 399-403.	3.2	62
144	Improved variant discovery through local re-alignment of short-read next-generation sequencing data using SRMA. Genome Biology, 2010, 11, R99.	8.8	61

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145	Detection of a MicroRNA Signal in an In Vivo Expression Set of mRNAs. PLoS ONE, 2007, 2, e804.	2.5	61
146	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
147	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.	6.2	59
148	Expanding the mutational spectrum of LZTR1 in schwannomatosis. European Journal of Human Genetics, 2015, 23, 963-968.	2.8	58
149	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	2.5	58
150	Molecular Analysis of Glioblastoma: Pathway Profiling and Its Implications for Patient Therapy. Cancer Biology and Therapy, 2003, 2, 242-247.	3.4	57
151	High density SNP association study of a major autism linkage region on chromosome 17. Human Molecular Genetics, 2007, 16, 704-715.	2.9	57
152	Celsius: a community resource for Affymetrix microarray data. Genome Biology, 2007, 8, R112.	9.6	57
153	A genome-wide linkage scan of familial benign recurrent vertigo: linkage to 22q12 with evidence of heterogeneity. Human Molecular Genetics, 2006, 15, 251-258.	2.9	56
154	Identification of Retinol Binding Protein 1 Promoter Hypermethylation in Isocitrate Dehydrogenase 1 and 2 Mutant Gliomas. Journal of the National Cancer Institute, 2012, 104, 1458-1469.	6.3	56
155	Characterization of transformation related genes in oral cancer cells. Oncogene, 1998, 16, 1921-1930.	5.9	55
156	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.	7.9	55
157	Rare Genomic Variants Link Bipolar Disorder with Anxiety Disorders to CREB-Regulated Intracellular Signaling Pathways. Frontiers in Psychiatry, 2013, 4, 154.	2.6	54
158	A comparison of gene expression profiles produced by SAGE, long SAGE, and oligonucleotide chips. Genomics, 2004, 84, 631-636.	2.9	53
159	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. ELife, 2018, 7, .	6.0	53
160	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphateâ€“binding region. Epilepsia, 2019, 60, 406-418.	5.1	53
161	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.	2.5	52
162	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, 1614-1621.	0.5	50

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163	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	49
164	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.	0.5	48
165	Protective Properties of Radio-Chemoresistant Glioblastoma Stem Cell Clones Are Associated with Metabolic Adaptation to Reduced Glucose Dependence. PLoS ONE, 2013, 8, e80397.	2.5	48
166	A novel mutation in KCNA1 causes episodic ataxia without myokymia. Human Mutation, 2004, 24, 536-536.	2.5	47
167	Repetitive sequence environment distinguishes housekeeping genes. Gene, 2007, 390, 153-165.	2.2	47
168	Phenotypic and Genetic Analysis of a Large Family With Migraineâ€Associated Vertigo. Headache, 2008, 48, 1460-1467.	3.9	46
169	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
170	Distinct gene expression profiles in adult mouse heart following targeted MAP kinase activation. Physiological Genomics, 2006, 25, 50-59.	2.3	41
171	Rich annotation of DNA sequencing variants by leveraging the Ensembl Variant Effect Predictor with plugins. Briefings in Bioinformatics, 2015, 16, 255-264.	6.5	41
172	Missense-depleted regions in population exomes implicate ras superfamily nucleotide-binding protein alteration in patients with brain malformation. Npj Genomic Medicine, 2016, 1, .	3.8	41
173	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
174	Osteogenic oxysterol, 20(S)-hydroxycholesterol, induces notch target gene expression in bone marrow stromal cells. Journal of Bone and Mineral Research, 2010, 25, 782-795.	2.8	38
175	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.	4.4	38
176	Autocrine Endothelin-3/Endothelin Receptor B Signaling Maintains Cellular and Molecular Properties of Glioblastoma Stem Cells. Molecular Cancer Research, 2011, 9, 1668-1685.	3.4	38
177	Bone morphogenetic protein 7 sensitizes O6-methylguanine methyltransferase expressing-glioblastoma stem cells to clinically relevant dose of temozolomide. Molecular Cancer, 2015, 14, 189.	19.2	38
178	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. Cilia, 2016, 5, 8.	1.8	37
179	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.	0.5	37
180	Cognitive functioning in affected sibling pairs with ADHD: familial clustering and dopamine genes. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2008, 49, 950-957.	5.2	36

#	Article	IF	Citations
181	Therapeutic Potential of HSP90 Inhibition for Neurofibromatosis Type 2. Clinical Cancer Research, 2013, 19, 3856-3870.	7.0	36
182	Disseminated Coccidioidomycosis Treated with Interferon-Î³ and Dupilumab. New England Journal of Medicine, 2020, 382, 2337-2343.	27.0	36
183	Association of progesterone receptor with migraine-associated vertigo. Neurogenetics, 2007, 8, 195-200.	1.4	35
184	BMPER Mutation in Diaphanospondylodysostosis Identified by Ancestral Autozygosity Mapping and Targeted High-Throughput Sequencing. American Journal of Human Genetics, 2010, 87, 532-537.	6.2	35
185	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.	6.2	35
186	Improving the efficiency of genomic loci capture using oligonucleotide arrays for high throughput resequencing. BMC Genomics, 2009, 10, 646.	2.8	34
187	De novo truncating variants in the intronless IRF2BPL are responsible for developmental epileptic encephalopathy. Genetics in Medicine, 2019, 21, 1008-1014.	2.4	34
188	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
189	Local alignment of two-base encoded DNA sequence. BMC Bioinformatics, 2009, 10, 175.	2.6	32
190	Genomic Mismatch Scanning Identifies Human Genomic DNA Shared Identical by Descent. Genomics, 1998, 47, 1-6.	2.9	31
191	Robustness of gene expression profiling in glioma specimen samplings and derived cell lines. Molecular Brain Research, 2005, 136, 99-103.	2.3	31
192	Loss of the scavenger mRNA decapping enzyme DCPS causes syndromic intellectual disability with neuromuscular defects. Human Molecular Genetics, 2015, 24, 3163-3171.	2.9	31
193	Loss of ADAM17 is associated with severe multiorgan dysfunction. Human Pathology, 2015, 46, 923-928.	2.0	31
194	Molecular diagnosis of putative Stargardt disease probands by exome sequencing. BMC Medical Genetics, 2012, 13, 67.	2.1	30
195	Reciprocal Regulation of the Cardiac Epigenome by Chromatin Structural Proteins Hmgb and Ctcf. Journal of Biological Chemistry, 2016, 291, 15428-15446.	3.4	30
196	Disease Gene Characterization through Large-Scale Co-Expression Analysis. PLoS ONE, 2009, 4, e8491.	2.5	30
197	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, 759-767.	1.8	29
198	Evaluation of techniques using amplified nucleic acid probes for gene expression profiling. New Biotechnology, 2003, 20, 97-106.	2.7	28

#	Article	IF	Citations
199	Genetically defined EWS/FLI1 model system suggests mesenchymal origin of Ewing's family tumors. Laboratory Investigation, 2008, 88, 1291-1302.	3.7	28
200	Rare deleterious mutations are associated with disease in bipolar disorder families. Molecular Psychiatry, 2017, 22, 1009-1014.	7.9	28
201	Ppp1r1b-lncRNA inhibits PRC2 at myogenic regulatory genes to promote cardiac and skeletal muscle development in mouse and human. Rna, 2020, 26, 481-491.	3.5	28
202	Deletion mapping in Xp21 for patients with complex glycerol kinase deficiency using SNP mapping arrays. Human Mutation, 2007, 28, 235-242.	2.5	27
203	CREB regulates Meis1 expression in normal and malignant hematopoietic cells. Leukemia, 2008, 22, 665-667.	7.2	26
204	Clinical aggressiveness of malignant gliomas is linked to augmented metabolism of amino acids. Journal of Neuro-Oncology, 2016, 128, 57-66.	2.9	26
205	Genomic Identification of Significant Targets in Ciliochoroidal Melanoma. , 2011, 52, 3018.		25
206	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, 164, 164-169.	1.2	25
207	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.	2.9	25
208	Online Self-Report Data for Duchenne Muscular Dystrophy Confirms Natural History and Can Be Used to Assess for Therapeutic Benefits. PLOS Currents, 2014, 6, .	1.4	25
209	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.	0.5	24
210	Identification of somatic and germline mutations using whole exome sequencing of congenital acute lymphoblastic leukemia. BMC Cancer, 2013, 13, 55.	2.6	24
211	Whole exome sequencing of pediatric gastric adenocarcinoma reveals an atypical presentation of Liâ€Fraumeni syndrome. Pediatric Blood and Cancer, 2013, 60, 570-574.	1.5	24
212	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.	2.5	24
213	Cardiac MRI biomarkers for Duchenne muscular dystrophy. Biomarkers in Medicine, 2018, 12, 1271-1289.	1.4	24
214	Evidence-Based Consensus and Systematic Review on Reducing the Time to Diagnosis of Duchenne Muscular Dystrophy. Journal of Pediatrics, 2019, 204, 305-313.e14.	1.8	24
215	COVIDâ€19 drug repurposing: Summary statistics on current clinical trials and promising untested candidates. Transboundary and Emerging Diseases, 2021, 68, 313-317.	3.0	24
216	Functional Consequences of a Novel Variant of PCSK1. PLoS ONE, 2013, 8, e55065.	2.5	24

#	Article	IF	Citations
217	Segmental uniparental isodisomy on 5q32-qter in a patient with childhood-onset schizophrenia. Journal of Medical Genetics, 2006, 43, 887-892.	3.2	23
218	Sex-specific influence of DRD 2 on ADHD-type temperament in a large population-based birth cohort. Psychiatric Genetics, 2012, 22, 197-201.	1.1	23
219	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. Annals of the New York Academy of Sciences, 2016, 1366, 49-60.	3.8	23
220	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	2.4	22
221	A biotinylated MutS fusion protein and its use in a rapid mutation screening technique. Genetic Analysis, Techniques and Applications, 1996, 13, 105-111.	1.5	21
222	High-throughput sequencing of the DBA/2J mouse genome. BMC Bioinformatics, 2010, 11, .	2.6	21
223	Identification of Candidate Tumor Oncogenes by Integrative Molecular Analysis of Choroidal Melanoma Fine-Needle Aspiration Biopsy Specimens. JAMA Ophthalmology, 2010, 128, 1170.	2.4	20
224	A wellâ€tolerated core needle muscle biopsy process suitable for children and adults. Muscle and Nerve, 2020, 62, 688-698.	2.2	20
225	Phenotype Sequencing: Identifying the Genes That Cause a Phenotype Directly from Pooled Sequencing of Independent Mutants. PLoS ONE, 2011, 6, e16517.	2.5	20
226	Cartilage-selective genes identified in genome-scale analysis of non-cartilage and cartilage gene expression. BMC Genomics, 2007, 8, 165.	2.8	19
227	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	2.5	19
228	Multimarker analysis and imputation of multiple platform pooling-based genome-wide association studies. Bioinformatics, 2008, 24, 1896-1902.	4.1	18
229	Truncating mutations in APP cause a distinct neurological phenotype. Annals of Neurology, 2016, 80, 456-460.	5.3	18
230	FDA Approval of Eteplirsen for Muscular Dystrophy. JAMA - Journal of the American Medical Association, 2017, 317, 1480.	7.4	18
231	Calpain 3 and CaMKIIÎ² signaling are required to induce HSP70 necessary for adaptive muscle growth after atrophy. Human Molecular Genetics, 2018, 27, 1642-1653.	2.9	18
232	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.	6.2	18
233	Efficient discovery of single-nucleotide polymorphisms in coding regions of human genes. Pharmacogenomics Journal, 2002, 2, 236-242.	2.0	17
234	Genomic predictors of remission to antidepressant treatment in geriatric depression using genomeâ€wide expression analyses: a pilot study. International Journal of Geriatric Psychiatry, 2016, 31, 510-517.	2.7	17

#	Article	IF	Citations
235	ZBED4, a BED-Type Zinc-Finger Protein in the Cones of the Human Retina. , 2009, 50, 3580.		16
236	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	2.4	16
237	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	8.2	16
238	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, 22759-22770.	3.4	15
239	The case for eteplirsen: Paving the way for precision medicine. Molecular Genetics and Metabolism, 2016, 118, 70-71.	1.1	15
240	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.	5.1	15
241	Identification of novel candidate genes for 46,XY disorders of sex development (DSD) using a C57BL/6J-Y POS mouse model. Biology of Sex Differences, 2018, 9, 8.	4.1	14
242	Sex chromosome anomalies in childhood onset schizophrenia: an update. Molecular Psychiatry, 2008, 13, 910-911.	7.9	13
243	Calculating the statistical significance of rare variants causal for Mendelian and complex disorders. BMC Medical Genomics, 2018, 11, 53.	1.5	13
244	Quantitative immuno-mass spectrometry imaging of skeletal muscle dystrophin. Scientific Reports, 2021, 11, 1128.	3.3	13
245	A Path to Implement Precision Child Health Cardiovascular Medicine. Frontiers in Cardiovascular Medicine, 2017, 4, 36.	2.4	12
246	Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling. Science Advances, 2022, 8, eabl5613.	10.3	12
247	Genomic Organization, 5â€² Flanking Enhancer Region, and Chromosomal Assignment of the Cell Cycle Gene, p55Cdc. Molecular Genetics and Metabolism, 1998, 64, 52-57.	1.1	11
248	Predicting the severity of Duchenne muscular dystrophy. Neurology, 2011, 76, 208-209.	1.1	11
249	A toolkit for genetics providers in followâ€up of patients with nonâ€diagnostic exome sequencing. Journal of Genetic Counseling, 2019, 28, 213-228.	1.6	11
250	Gene-environment regulatory circuits of right ventricular pathology in tetralogy of fallot. Journal of Molecular Medicine, 2019, 97, 1711-1722.	3.9	11
251	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.	5.1	10
252	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an RFX6 mutation. Development (Cambridge), 2020, 147, .	2.5	10

#	Article	IF	Citations
253	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	2.4	10
254	Genomic mismatch scanning: Current progress and potential applications. Electrophoresis, 1995, 16, 279-285.	2.4	9
255	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	9
256	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, 130-135.	5.1	8
257	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	1.2	8
258	Congenital myasthenic syndrome caused by a frameshift insertion mutation in GFPT1. Neurology: Genetics, 2020, 6, e468.	1.9	8
259	Representational oligonucleotide microarray analysis (ROMA) and comparison of binning and change-point methods of analysis: application to detection of del22q11.2 (DiGeorge) Syndrome. Human Mutation, 2008, 29, 176-181.	2.5	7
260	Expansion of <scp>NEUROD2</scp> phenotypes to include developmental delay without seizures. American Journal of Medical Genetics, Part A, 2021, 185, 1076-1080.	1.2	7
261	Characterization of three cell lines derived from fine needle biopsy of choroidal melanoma with metastatic outcome. Molecular Vision, 2011, 17, 607-15.	1.1	7
262	What can Duchenne Connect teach us about treating Duchenne muscular dystrophy?. Current Opinion in Neurology, 2015, 28, 535-541.	3.6	6
263	Dusp6 is a genetic modifier of growth through enhanced ERK activity. Human Molecular Genetics, 2018, 28, 279-289.	2.9	6
264	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
265	Large in-frame 5â€² deletions in DMD associated with mild Duchenne muscular dystrophy: Two case reports and a review of the literature. Neuromuscular Disorders, 2019, 29, 863-873.	0.6	6
266	The frontiers of sequencing in undiagnosed neurodevelopmental diseases. Current Opinion in Genetics and Development, 2020, 65, 76-83.	3.3	6
267	Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing. Neuropathology, 2020, 40, 302-307.	1.2	6
268	Mosaic de novo SNRPN gene variant associated with Prader-Willi syndrome. Journal of Medical Genetics, 2022, 59, 719-722.	3.2	6
269	Novel NUDT2 variant causes intellectual disability and polyneuropathy. Annals of Clinical and Translational Neurology, 2020, 7, 2320-2325.	3.7	5
270	MPEG1/Perforin-2 Haploinsufficiency Associated Polymicrobial Skin Infections and Considerations for Interferon-Î³ Therapy. Frontiers in Immunology, 2020, 11, 601584.	4.8	5

#	Article	IF	Citations
271	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	2.7	5
272	Rethinking clinical practice: clinical implementation of exome sequencing. Personalized Medicine, 2012, 9, 785-787.	1.5	4
273	Validation and Detection of Exon Skipping Boosters in DMD Patient Cell Models and mdx Mouse. Methods in Molecular Biology, 2018, 1828, 309-326.	0.9	4
274	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.	1.1	4
275	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.	3.9	4
276	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.	3.7	4
277	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.	0.6	4
278	DNA Sequencing of Maternal Plasma to Detect Down Syndrome. Obstetrical and Gynecological Survey, 2012, 67, 86-88.	0.4	3
279	Leveraging ancestry to improve causal variant identification in exome sequencing for monogenic disorders. European Journal of Human Genetics, 2016, 24, 113-119.	2.8	3
280	In silico enhanced restriction enzyme based methylation analysis of the human glioblastoma genome using Agilent 244K CpG Island microarrays. Frontiers in Neuroscience, 2009, 3, 57.	2.8	2
281	Kcnd2 Mutation Associated with Autism and Epilepsy Impairs Inactivation Gating in Kv4.2 K+ Channels. Biophysical Journal, 2014, 106, 741a.	0.5	2
282	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.	0.5	2
283	Downregulation of SATB1 by miRNAs reduces megakaryocyte/erythroid progenitor expansion in preclinical models of Diamondâ€“Blackfan anemia. Experimental Hematology, 2022, 111, 66-78.	0.4	2
284	Selective surface treatment of micro printing pin and its performance. Applied Physics Letters, 2006, 89, 083901.	3.3	1
285	Local alignment of generalized k-base encoded DNA sequence. BMC Bioinformatics, 2010, 11, 347.	2.6	1
286	Interpreting Whole-Genome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 296.	7.4	1
287	Case Report: Whole Exome Sequencing Identifies Compound Heterozygous Variants in TSFM Gene Causing Juvenile Hypertrophic Cardiomyopathy. Frontiers in Cardiovascular Medicine, 2021, 8, 798985.	2.4	1
288	The dopamine receptor DRD4 gene. Trends in Pharmacological Sciences, 2001, 22, 56.	8.7	0

#	Article	IF	Citations
289	735. Functional Restoration of Dystrophin Protein in HiPSC-Derived Skeletal Myotubes and Cardiomyocytes After CRISPR/Cas9-Mediated Deletion of 530-725kb of DMD. Molecular Therapy, 2016, 24, S290.	8.2	0
290	Gene-environment regulation of chamber-specific maturation during hypoxemic perinatal circulatory transition. Journal of Molecular Medicine, 2020, 98, 1009-1020.	3.9	0
291	Identification of Meis1 as a Target of CREB in Myeloid Leukemogenesis.. Blood, 2006, 108, 1945-1945.	1.4	0
292	Requirement of CREB in Normal and Malignant Hematopoiesis.. Blood, 2006, 108, 1168-1168.	1.4	0
293	Abstract 4749: Genetic signature of malignant glioma is associated with responsiveness to autologous tumor lysate-pulsed dendritic cell vaccination + TLR agonists. , 2010, , .		0
294	Abstract LB-245: Loss of function of the TGFÎ²RI receptor leads to the spontaneously regressing squamous carcinoma condition, multiple self-healing squamous epithelioma (Ferguson-Smith disease). , 2011, , .		0
295	Abstract 447: The IDH1 mutation in human glioblastoma and its effects on epigenetic modification and cell fate selection. , 2014, , .		0
296	Abstract 08: Clinical exome sequencing in the diagnosis of pediatric cancer predisposition. , 2014, , .		0
297	Role of Mir-34 Upregulation in Disruption of c-Myc, c-Myb and NOTCH Signaling in Diamond-Blackfan Anemia. Blood, 2016, 128, 3895-3895.	1.4	0
298	RABENOSYN separation-of-function mutations uncouple endosomal recycling from lysosomal degradation, causing a distinct Mendelian Disorder. Human Molecular Genetics, 0, , .	2.9	0