Chad A Shaw

List of Publications by Year
in descending order

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223	22,108	9428	11282
papers	citations	76 h-index	141 g-index

234	234	234	37303
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical exome sequencing data reveal high diagnostic yields for congenital diaphragmatic hernia plus (CDH+) and new phenotypic expansions involving CDH. Journal of Medical Genetics, 2022, 59, 270-278.	1.5	27
2	Genome Sequencing in the Parkinson Disease Clinic. Neurology: Genetics, 2022, 8, .	0.9	7
3	Abstract PS5-29: Insights into the molecular underpinnings of the mevalonate pathway-YAP/TAZ-driven anti-HER2 therapy resistance in HER2+ breast cancer (BC). , 2021, , .		0
4	Selection for or against escape from nonsense mediated decay is a novel signature for the detection of cancer genes. Cancer Genetics, 2021, 258-259, 80-84.	0.2	1
5	Specificity and Heterogeneity of Trained Immunity in Hematopoietic Stem and Progenitor Cells. Blood, 2021, 138, 2149-2149.	0.6	1
6	Increased Moraxella and Streptococcus species abundance after severe bronchiolitis is associated with recurrent wheezing. Journal of Allergy and Clinical Immunology, 2020, 145, 518-527.e8.	1.5	50
7	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genetics in Medicine, 2020, 22, 1768-1776.	1.1	30
8	Bayesian modelling of high-throughput sequencing assays with malacoda. PLoS Computational Biology, 2020, 16, e1007504.	1.5	1
9	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498.	0.9	11
10	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	1.1	36
11	A joint modeling approach for longitudinal microbiome data improves ability to detect microbiome associations with disease. PLoS Computational Biology, 2020, 16, e1008473.	1.5	7
12	Title is missing!. , 2020, 16, e1008473.		0
13	Title is missing!. , 2020, 16, e1008473.		0
14	Title is missing!. , 2020, 16, e1008473.		0
15	Title is missing!. , 2020, 16, e1008473.		0
16	Title is missing!. , 2020, 16, e1008473.		0
17	Title is missing!. , 2020, 16, e1008473.		0
18	Bayesian modelling of high-throughput sequencing assays with malacoda. , 2020, 16, e1007504.		0

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19	Bayesian modelling of high-throughput sequencing assays with malacoda. , 2020, 16, e1007504.		0
20	Bayesian modelling of high-throughput sequencing assays with malacoda. , 2020, 16, e1007504.		0
21	Bayesian modelling of high-throughput sequencing assays with malacoda. , 2020, 16, e1007504.		0
22	Bayesian modelling of high-throughput sequencing assays with malacoda. , 2020, 16, e1007504.		0
23	Bayesian modelling of high-throughput sequencing assays with malacoda. , 2020, 16, e1007504.		0
24	Bayesian modelling of high-throughput sequencing assays with malacoda. , 2020, 16, e1007504.		0
25	Bayesian modelling of high-throughput sequencing assays with malacoda. , 2020, 16, e1007504.		0
26	Functionalization of CD36 cardiovascular disease and expression associated variants by interdisciplinary high throughput analysis. PLoS Genetics, 2019, 15, e1008287.	1.5	9
27	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	3.6	55
28	Targeting the Mevalonate Pathway to Overcome Acquired Anti-HER2 Treatment Resistance in Breast Cancer. Molecular Cancer Research, 2019, 17, 2318-2330.	1.5	41
29	Non-invasive prenatal sequencing for multiple Mendelian monogenic disorders using circulating cell-free fetal DNA. Nature Medicine, 2019, 25, 439-447.	15.2	160
30	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
31	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	3.6	42
32	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	3.6	22
33	Identification of the Regulatory Elements and Target Genes of Megakaryopoietic Transcription Factor MEF2C. Thrombosis and Haemostasis, 2019, 119, 716-725.	1.8	5
34	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10.	13.5	73
35	Validation Studies for Single Circulating Trophoblast Genetic Testing as a Form of Noninvasive Prenatal Diagnosis. American Journal of Human Genetics, 2019, 105, 1262-1273.	2.6	47
36	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52

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37	Combinatorial inhibition of PTPN12-regulated receptors leads to a broadly effective therapeutic strategy in triple-negative breast cancer. Nature Medicine, 2018, 24, 505-511.	15.2	47
38	Design tools for MPRA experiments. Bioinformatics, 2018, 34, 2682-2683.	1.8	7
39	Reliable detection of subchromosomal deletions and duplications using cellâ€based noninvasive prenatal testing. Prenatal Diagnosis, 2018, 38, 1069-1078.	1.1	42
40	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. Journal of Pediatric Genetics, 2018, 07, 164-173.	0.3	15
41	Effects of genetic variation in protease activated receptor 4 after an acute coronary syndrome: Analysis from the TRACER trial. Blood Cells, Molecules, and Diseases, 2018, 72, 37-43.	0.6	10
42	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187.	2.6	160
43	The association between anterior nares and nasopharyngeal microbiota in infants hospitalized for bronchiolitis. Microbiome, 2018, 6, 2.	4.9	56
44	Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Research, 2018, 28, 1228-1242.	2.4	74
45	Abstract 015: PAR4 Ala120Thr Variant Alters PAR4 Desensitization, Sensitivity to Platelet Antagonists and Risk of Large Vessel Stroke. Arteriosclerosis, Thrombosis, and Vascular Biology, 2018, 38, .	1.1	0
46	Identification of the Genetic Variant Responsible for Variable Platelet CD36 Expression By Massively Parallel Reporter Assay. Blood, 2018, 132, 520-520.	0.6	0
47	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	6.5	98
48	A paradox of transcriptional and functional innate interferon responses of human intestinal enteroids to enteric virus infection. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E570-E579.	3.3	112
49	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	2.6	86
50	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	13.5	66
51	HER2 Reactivation through Acquisition of the HER2 L755S Mutation as a Mechanism of Acquired Resistance to HER2-targeted Therapy in HER2+ Breast Cancer. Clinical Cancer Research, 2017, 23, 5123-5134.	3.2	85
52	EMT cells increase breast cancer metastasis via paracrine GLI activation in neighbouring tumour cells. Nature Communications, 2017, 8, 15773.	5.8	126
53	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	13.9	565
54	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348

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55	Sequence variability of the respiratory syncytial virus (RSV) fusion gene among contemporary and historical genotypes of RSV/A and RSV/B. PLoS ONE, 2017, 12, e0175792.	1.1	51
56	Sustained endocrine profiles of a girl with WAGR syndrome. BMC Medical Genetics, 2017, 18, 117.	2.1	4
57	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	3.6	50
58	Identification of a functional genetic variant driving racially dimorphic platelet gene expression of the thrombin receptor regulator, PCTP. Thrombosis and Haemostasis, 2017, 117, 962-970.	1.8	5
59	Identifying diagnosticallyâ€relevant resting state brain functional connectivity in the ventral posterior complex via genetic data mining in autism spectrum disorder. Autism Research, 2016, 9, 553-562.	2.1	10
60	Respiratory syncytial virus and rhinovirus severe bronchiolitis are associated with distinct nasopharyngeal microbiota. Journal of Allergy and Clinical Immunology, 2016, 137, 1909-1913.e4.	1.5	82
61	Integrative Multi-omic Analysis of Human Platelet eQTLs Reveals Alternative Start Site in Mitofusin 2. American Journal of Human Genetics, 2016, 98, 883-897.	2.6	27
62	De Novo Truncating Mutation of TRIM8 Causes Earlyâ€Onset Epileptic Encephalopathy. Annals of Human Genetics, 2016, 80, 235-240.	0.3	24
63	Copy number variation as a genetic basis for heterotaxy and heterotaxy-spectrum congenital heart defects. Philosophical Transactions of the Royal Society B: Biological Sciences, 2016, 371, 20150406.	1.8	29
64	CELF1 is a central node in post-transcriptional regulatory programmes underlying EMT. Nature Communications, 2016, 7, 13362.	5.8	53
65	Evidence for feasibility of fetal trophoblastic cellâ€based noninvasive prenatal testing. Prenatal Diagnosis, 2016, 36, 1009-1019.	1.1	78
66	Comparison of three whole genome amplification methods for detection of genomic aberrations in single cells. Prenatal Diagnosis, 2016, 36, 823-830.	1.1	22
67	Clinical bioinformatics: emergence of a new laboratory discipline. Expert Review of Molecular Diagnostics, 2016, 16, 1139-1141.	1.5	7
68	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. Genome Medicine, 2016, 8, 105.	3.6	20
69	Genome-wide copy number analysis on DNA from fetal cells isolated from the blood of pregnant women. Prenatal Diagnosis, 2016, 36, 1127-1134.	1.1	68
70	Association of nasopharyngeal microbiota profiles with bronchiolitis severity in infants hospitalised for bronchiolitis. European Respiratory Journal, 2016, 48, 1329-1339.	3.1	144
71	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. Genome Medicine, 2016, 8, 13.	3.6	37
72	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	1.1	186

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73	Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. Human Mutation, 2016, 37, 231-234.	1.1	18
74	Antiâ€“miR-148a regulates platelet FcÎ³RIIA signaling and decreases thrombosis in vivo in mice. Blood, 2015, 126, 2871-2881.	0.6	49
75	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. Human Molecular Genetics, 2015, 24, 4061-4077.	1.4	83
76	Genome-wide analyses of LINEâ€“LINE-mediated nonallelic homologous recombination. Nucleic Acids Research, 2015, 43, 2188-2198.	6.5	79
77	Circulating and disseminated tumor cells from breast cancer patient-derived xenograft-bearing mice as a novel model to study metastasis. Breast Cancer Research, 2015, 17, 3.	2.2	48
78	Somatic mosaicism: implications for disease and transmission genetics. Trends in Genetics, 2015, 31, 382-392.	2.9	234
79	Analysis of 13 cell types reveals evidence for the expression of numerous novel primate- and tissue-specific microRNAs. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E1106-15.	3.3	376
80	Variant interpretation through Bayesian fusion of frequency and genomic knowledge. Genome Medicine, 2015, 7, 4.	3.6	1
81	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564.	2.6	45
82	MeCP2 binds to non-CG methylated DNA as neurons mature, influencing transcription and the timing of onset for Rett syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 5509-5514.	3.3	256
83	An essential role for GÎ± _{i2} in Smoothened-stimulated epithelial cell proliferation in the mammary gland. Science Signaling, 2015, 8, ra92.	1.6	17
84	Genome-wide association study of platelet aggregation in African Americans. BMC Genetics, 2015, 16, 58.	2.7	50
85	Mus81 and converging forks limit the mutagenicity of replication fork breakage. Science, 2015, 349, 742-747.	6.0	162
86	The spliceosome is a therapeutic vulnerability in MYC-driven cancer. Nature, 2015, 525, 384-388.	13.7	392
87	Mutations in the transcriptional repressor REST predispose to Wilms tumor. Nature Genetics, 2015, 47, 1471-1474.	9.4	54
88	Host Transcriptional Response to Influenza and Other Acute Respiratory Viral Infections â€“ A Prospective Cohort Study. PLoS Pathogens, 2015, 11, e1004869.	2.1	147
89	Identification of the Genetic Mechanism Responsible for Racially-Dimorphic Expression of the Thrombin-Receptor Regulator, Pctp. Blood, 2015, 126, 415-415.	0.6	7
90	Gene Sequence Variability of the Three Surface Proteins of Human Respiratory Syncytial Virus (HRSV) in Texas. PLoS ONE, 2014, 9, e90786.	1.1	54

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91	MicroRNA Expression Differences in Human Hematopoietic Cell Lineages Enable Regulated Transgene Expression. PLoS ONE, 2014, 9, e102259.	1.1	77
92	Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. BMC Biology, 2014, 12, 74.	1.7	60
93	The Oncogenic STP Axis Promotes Triple-Negative Breast Cancer via Degradation of the REST Tumor Suppressor. Cell Reports, 2014, 9, 1318-1332.	2.9	24
94	An efficient algorithm for accurate computation of the Dirichlet-multinomial log-likelihood function. Bioinformatics, 2014, 30, 1547-1554.	1.8	22
95	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	1.4	112
96	The human platelet: strong transcriptome correlations among individuals associate weakly with the platelet proteome. Biology Direct, 2014, 9, 3.	1.9	77
97	Application of array comparative genomic hybridization in 256 patients with developmental delay or intellectual disability. Journal of Applied Genetics, 2014, 55, 125-144.	1.0	37
98	NetComm: a network analysis tool based on communicability. Bioinformatics, 2014, 30, 3387-3389.	1.8	2
99	Parent of Origin, Mosaicism, and Recurrence Risk: Probabilistic Modeling Explains the Broken Symmetry of Transmission Genetics. American Journal of Human Genetics, 2014, 95, 345-359.	2.6	103
100	Pathway-Centric Integrative Analysis Identifies RRM2 as a Prognostic Marker in Breast Cancer Associated with Poor Survival and Tamoxifen Resistance. Neoplasia, 2014, 16, 390-402.	2.3	66
101	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	2.6	219
102	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€‰362 consecutive cases. European Journal of Human Genetics, 2014, 22, 969-978.	1.4	51
103	Testicular sex cord-stromal tumor in a boy with 2q37 deletion syndrome. BMC Medical Genomics, 2014, 7, 19.	0.7	2
104	Human platelet microRNA-mRNA networks associated with age and gender revealed by integrated plateletomics. Blood, 2014, 123, e37-e45.	0.6	199
105	Common variants in the human platelet PAR4 thrombin receptor alter platelet function and differ by race. Blood, 2014, 124, 3450-3458.	0.6	107
106	Identification of a Racially Dimorphic Variant in the Human Platelet PAR4 Thrombin Receptor Altering Platelet Function and Pharmacologic Inhibition. Blood, 2014, 124, 1434-1434.	0.6	11
107	Mir-148a and Mir-25 Target TULA-2 and Regulate Human Platelet FcÎ³RIIA Signaling. Blood, 2014, 124, 337-337.	0.6	0
108	Platelet FcÎ³RIIA Signaling Results in Ubiquitination and Cellular Translocation of Activated Syk. Blood, 2014, 124, 2761-2761.	0.6	0

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109	Racial differences in human platelet PAR4 reactivity reflect expression of PCTP and miR-376c. Nature Medicine, 2013, 19, 1609-1616.	15.2	190
110	Rare DNA copy number variants in cardiovascular malformations with extracardiac abnormalities. European Journal of Human Genetics, 2013, 21, 173-181.	1.4	49
111	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	2.6	43
112	Neuroendocrine phenotypes in a boy with 5q14 deletion syndrome implicate the regulatory roles of myocyte-specific enhancer factor 2C in the postnatal hypothalamus. European Journal of Medical Genetics, 2013, 56, 475-483.	0.7	6
113	RASâ€“MAPKâ€“MSK1 pathway modulates ataxin 1 protein levels and toxicity in SCA1. Nature, 2013, 498, 325-331.	13.7	119
114	Comparison of chromosome analysis and chromosomal microarray analysis: what is the value of chromosome analysis in todayâ€™s genomic array era?. Genetics in Medicine, 2013, 15, 450-457.	1.1	63
115	Application of custom-designed oligonucleotide array CGH in 145 patients with autistic spectrum disorders. European Journal of Human Genetics, 2013, 21, 620-625.	1.4	37
116	Fusion of Large-Scale Genomic Knowledge and Frequency Data Computationally Prioritizes Variants in Epilepsy. PLoS Genetics, 2013, 9, e1003797.	1.5	22
117	Ataxin1L Is a Regulator of HSC Function Highlighting the Utility of Cross-Tissue Comparisons for Gene Discovery. PLoS Genetics, 2013, 9, e1003359.	1.5	5
118	Confounding by Repetitive Elements and CpG Islands Does Not Explain the Association between Hypomethylation and Genomic Instability. PLoS Genetics, 2013, 9, e1003333.	1.5	3
119	Screening and familial characterization of copyâ€number variations in NR5A1 in 46,XY disorders of sex development and premature ovarian failure. American Journal of Medical Genetics, Part A, 2013, 161, 2487-2494.	0.7	12
120	Incidental copy-number variants identified by routine genome testing in a clinical population. Genetics in Medicine, 2013, 15, 45-54.	1.1	37
121	A Renewable Tissue Resource of Phenotypically Stable, Biologically and Ethnically Diverse, Patient-Derived Human Breast Cancer Xenograft Models. Cancer Research, 2013, 73, 4885-4897.	0.4	394
122	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	2.4	120
123	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394.	2.4	62
124	Global Gene Expression Profiling in Infants With Acute Respiratory Syncytial Virus Bronchiolitis Demonstrates Systemic Activation of Interferon Signaling Networks. Pediatric Infectious Disease Journal, 2013, 32, e68-e76.	1.1	31
125	Integrative genomic analysis of the human immune response to influenza vaccination. ELife, 2013, 2, e00299.	2.8	126
126	Racial Differences In Thrombin-Induced Human Platelet PAR4 Reactivity. Blood, 2013, 122, 1054-1054.	0.6	0

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127	Effect Of Age and Gender On Human Platelet mRNA and Micro-RNA Levels. Blood, 2013, 122, 3518-3518.	0.6	0
128	Genomic Hypomethylation in the Human Germline Associates with Selective Structural Mutability in the Human Genome. PLoS Genetics, 2012, 8, e1002692.	1.5	80
129	The Hematopoietic Expression Viewer: expanding mobile apps as a scientific tool. Bioinformatics, 2012, 28, 1941-1942.	1.8	7
130	A SUMOylation-Dependent Transcriptional Subprogram Is Required for Myc-Driven Tumorigenesis. Science, 2012, 335, 348-353.	6.0	374
131	Penetrance of biallelic SMARCAL1 mutations is associated with environmental and genetic disturbances of gene expression. Human Molecular Genetics, 2012, 21, 2572-2587.	1.4	57
132	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	3.3	118
133	Crh and Oprm1 mediate anxiety-related behavior and social approach in a mouse model of MECP2 duplication syndrome. Nature Genetics, 2012, 44, 206-211.	9.4	146
134	Human Genome-Wide Association and Mouse Knockout Approaches Identify Platelet Supervillin as an Inhibitor of Thrombus Formation Under Shear Stress. Circulation, 2012, 125, 2762-2771.	1.6	25
135	Detection of â‰¥1â€‰Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. Prenatal Diagnosis, 2012, 32, 10-20.	1.1	29
136	Prenatal chromosomal microarray analysis in a diagnostic laboratory; experience with >1000 cases and review of the literature. Prenatal Diagnosis, 2012, 32, 351-361.	1.1	103
137	Application of array comparative genomic hybridization in 102 patients with epilepsy and additional neurodevelopmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 760-771.	1.1	48
138	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. Human Mutation, 2012, 33, 165-179.	1.1	45
139	Activation of Multiple Proto-oncogenic Tyrosine Kinases in Breast Cancer via Loss of the PTPN12 Phosphatase. Cell, 2011, 144, 703-718.	13.5	246
140	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
141	Platelet microRNA-mRNA coexpression profiles correlate with platelet reactivity. Blood, 2011, 117, 5189-5197.	0.6	305
142	High frequency of known copy number abnormalities and maternal duplication 15q11-q13 in patients with combined schizophrenia and epilepsy. BMC Medical Genetics, 2011, 12, 154.	2.1	57
143	Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. Genome Research, 2011, 21, 33-46.	2.4	72
144	Exercise and Genetic Rescue of SCA1 via the Transcriptional Repressor Capicua. Science, 2011, 334, 690-693.	6.0	144

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145	Protein Interactome Reveals Converging Molecular Pathways Among Autism Disorders. Science Translational Medicine, 2011, 3, 86ra49.	5.8	201
146	Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. Human Molecular Genetics, 2011, 20, 4360-4370.	1.4	101
147	Comparison of an expanded ataxia interactome with patient medical records reveals a relationship between macular degeneration and ataxia. Human Molecular Genetics, 2011, 20, 510-527.	1.4	45
148	Early Patterns of Gene Expression Correlate With the Humoral Immune Response to Influenza Vaccination in Humans. Journal of Infectious Diseases, 2011, 203, 921-929.	1.9	208
149	Recurrent Distal 7q11.23 Deletion Including HIP1 and YWHAG Identified in Patients with Intellectual Disabilities, Epilepsy, and Neurobehavioral Problems. American Journal of Human Genetics, 2010, 87, 857-865.	2.6	58
150	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	1.1	225
151	Insertional translocation detected using FISH confirmation of arrayâ€comparative genomic hybridization (aCGH) results. American Journal of Medical Genetics, Part A, 2010, 152A, 1111-1126.	0.7	85
152	VAMP8/endobrevin is overexpressed in hyperreactive human platelets: suggested role for platelet microRNA. Journal of Thrombosis and Haemostasis, 2010, 8, 369-378.	1.9	177
153	Identification of De Novo Copy Number Variants Associated with Human Disorders of Sexual Development. PLoS ONE, 2010, 5, e15392.	1.1	131
154	Proinflammatory Role for let-7 MicroRNAS in Experimental Asthma. Journal of Biological Chemistry, 2010, 285, 30139-30149.	1.6	222
155	Partial Loss of Ataxin-1 Function Contributes to Transcriptional Dysregulation in Spinocerebellar Ataxia Type 1 Pathogenesis. PLoS Genetics, 2010, 6, e1001021.	1.5	113
156	Array Comparative Genomic Hybridization Detects Chromosomal Abnormalities in Hematological Cancers That Are Not Detected by Conventional Cytogenetics. Journal of Molecular Diagnostics, 2010, 12, 670-679.	1.2	25
157	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203.	1.4	165
158	Gene expression in Barrett's esophagus: Laser capture versus whole tissue. Scandinavian Journal of Gastroenterology, 2009, 44, 787-795.	0.6	32
159	Activin Signaling: Effects on Body Composition and Mitochondrial Energy Metabolism. Endocrinology, 2009, 150, 3521-3529.	1.4	43
160	Mouse models of MeCP2 disorders share gene expression changes in the cerebellum and hypothalamus. Human Molecular Genetics, 2009, 18, 2431-2442.	1.4	228
161	Clinical use of array comparative genomic hybridization (aCGH) for prenatal diagnosis in 300 cases. Prenatal Diagnosis, 2009, 29, 29-39.	1.1	180
162	Dominant versus recessive traits conveyed by allelic mutations â€“ to what extent is nonsenseâ€mediated decay involved?. Clinical Genetics, 2009, 75, 394-400.	1.0	28

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163	Application of dual-genome oligonucleotidearray-based comparative genomic hybridization to the molecular diagnosis of mitochondrial DNA deletion and depletion syndromes. Genetics in Medicine, 2009, 11, 518-526.	1.1	51
164	Microarray-Based Comparative Genomic Hybridization Using Sex-Matched Reference DNA Provides Greater Sensitivity for Detection of Sex Chromosome Imbalances than Array-Comparative Genomic Hybridization with Sex-Mismatched Reference DNA. Journal of Molecular Diagnostics, 2009, 11, 226-237.	1.2	11
165	Brachyâ€“syndactyly caused by loss of Sfrp2 function. Journal of Cellular Physiology, 2008, 217, 127-137.	2.0	61
166	Validation of a targeted DNA microarray for the clinical evaluation of recurrent abnormalities in chronic lymphocytic leukemia. American Journal of Hematology, 2008, 83, 540-546.	2.0	54
167	Lowâ€level mosaicism of trisomy 14: Phenotypic and molecular characterization. American Journal of Medical Genetics, Part A, 2008, 146A, 1395-1405.	0.7	34
168	Identification of chromosome abnormalities in subtelomeric regions by microarray analysis: A study of 5,380 cases. American Journal of Medical Genetics, Part A, 2008, 146A, 2242-2251.	0.7	113
169	22q11.2 Distal Deletion: A Recurrent Genomic Disorder Distinct from DiGeorge Syndrome and Velocardiofacial Syndrome. American Journal of Human Genetics, 2008, 82, 214-221.	2.6	182
170	MeCP2, a Key Contributor to Neurological Disease, Activates and Represses Transcription. Science, 2008, 320, 1224-1229.	6.0	1,582
171	Genomic Imbalances in Neonates With Birth Defects: High Detection Rates by Using Chromosomal Microarray Analysis. Pediatrics, 2008, 122, 1310-1318.	1.0	137
172	Disruption of the circadian clock within the cardiomyocyte influences myocardial contractile function, metabolism, and gene expression. American Journal of Physiology - Heart and Circulatory Physiology, 2008, 294, H1036-H1047.	1.5	310
173	Mouse let-7 miRNA populations exhibit RNA editing that is constrained in the 5'-seed/ cleavage/anchor regions and stabilize predicted mmu-let-7a:mRNA duplexes. Genome Research, 2008, 18, 1571-1581.	2.4	87
174	The insulin-like growth factor pathway is altered in spinocerebellar ataxia type 1 and type 7. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 1291-1296.	3.3	85
175	Bacterial artificial chromosome-emulation oligonucleotide arrays for targeted clinical array-comparative genomic hybridization analyses. Genetics in Medicine, 2008, 10, 278-289.	1.1	90
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