The Genome Analysis Toolkit: A MapReduce framework
sequencing data

Genome Research

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Protein Residues That Control the Reaction Trajectory in <i>S-</i>Adenosylmethionine Radical Enzymes: Mutagenesis of Asparagine 153 and Aspartate 155 in <i>Escherichia coli</i> Biotin Synthase. Biochemistry, 2009, 48, 2448-2458.	1.2	21
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1919	Whole exome sequencing in a case of sporadic multiple meningioma reveals shared NF2, FAM109B, and TPRXL mutations, together with unique SMARCB1 alterations in a subset of tumor nodules. Cancer Genetics, 2015, 208, 327-332.	0.2	14
1920	Mutations in DYNC2LI1 disrupt cilia function and cause short rib polydactyly syndrome. Nature Communications, 2015, 6, 7092.	5.8	79
1921	Construction of a high-density integrated genetic linkage map of rubber tree (Hevea brasiliensis) using genotyping-by-sequencing (GBS). Frontiers in Plant Science, 2015, 6, 367.	1.7	102
1922	Whole-exome sequencing identifies OR2W3 mutation as a cause of autosomal dominant retinitis pigmentosa. Scientific Reports, 2015, 5, 9236.	1.6	25
1923	Genetics and genotypeâ€"phenotype correlations in Finnish patients with dilated cardiomyopathy. European Heart Journal, 2015, 36, 2327-2337.	1.0	130
1924	Mutation of ATF6 causes autosomal recessive achromatopsia. Human Genetics, 2015, 134, 941-950.	1.8	69
1925	Genome Analysis of Latin American Cervical Cancer: Frequent Activation of the PIK3CA Pathway. Clinical Cancer Research, 2015, 21, 5360-5370.	3.2	68
1926	SNP-Seek database of SNPs derived from 3000 rice genomes. Nucleic Acids Research, 2015, 43, D1023-D1027.	6.5	357
1927	Whole-exome sequencing reveals genetic variants associated with chronic kidney disease characterized by tubulointerstitial damages in North Central Region, Sri Lanka. Environmental Health and Preventive Medicine, 2015, 20, 354-359.	1.4	29
1928	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	1.4	25
1929	Novel<i>katG</i>mutations causing isoniazid resistance in clinical<i>M. tuberculosis</i>isolates. Emerging Microbes and Infections, 2015, 4, 1-9.	3.0	95
1930	multiSNV: a probabilistic approach for improving detection of somatic point mutations from multiple related tumour samples. Nucleic Acids Research, 2015, 43, e61-e61.	6.5	36
1931	Chimeric EWSR1-FLI1 regulates the Ewing sarcoma susceptibility gene EGR2 via a GGAA microsatellite. Nature Genetics, 2015, 47, 1073-1078.	9.4	157
1932	Physiology, Genomics, and Pathway Engineering of an Ethanol-Tolerant Strain of Clostridium phytofermentans. Applied and Environmental Microbiology, 2015, 81, 5440-5448.	1.4	20
1933	Anatomical distribution of Mycobacterium bovis genotypes in experimentally infected white-tailed deer. Veterinary Microbiology, 2015, 180, 75-81.	0.8	11
1934	Whole-genome sequencing for identification of Mendelian disorders in critically ill infants: a retrospective analysis of diagnostic and clinical findings. Lancet Respiratory Medicine,the, 2015, 3, 377-387.	5.2	322
1935	Transcriptomic analysis provides insight into high-altitude acclimation in domestic goats. Gene, 2015, 567, 208-216.	1.0	26

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1936	Severe dermatitis, multiple allergies, and metabolic wasting syndrome caused by a novel mutation in the N-terminal plakin domain of desmoplakin. Journal of Allergy and Clinical Immunology, 2015, 136, 1268-1276.	1.5	103
1937	Compound heterozygous NOTCH1 mutations underlie impaired cardiogenesis in a patient with hypoplastic left heart syndrome. Human Genetics, 2015, 134, 1003-1011.	1.8	71
1938	Loss of function of PGAP1 as a cause of severe encephalopathy identified by Whole Exome Sequencing: Lessons of the bioinformatics pipeline. Molecular and Cellular Probes, 2015, 29, 323-329.	0.9	24
1939	Performance comparison of four commercial human whole-exome capture platforms. Scientific Reports, 2015, 5, 12742.	1.6	68
1940	Diagnostic yield of targeted next generation sequencing in various cancer types: An information-theoretic approach. Cancer Genetics, 2015, 208, 441-447.	0.2	9
1941	Dynamic Provisioning of Data Intensive Computing Middleware Frameworks. , 2015, , .		1
1942	On the design and analysis of next-generation sequencing genotyping for a cohort with haplotype-informative reads. Methods, 2015, 79-80, 41-46.	1.9	3
1943	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. American Journal of Human Genetics, 2015, 96, 474-479.	2.6	127
1944	A Systematic Comparison of Traditional and Multigene Panel Testing for Hereditary Breast and Ovarian Cancer Genes in More Than 1000 Patients. Journal of Molecular Diagnostics, 2015, 17, 533-544.	1.2	167
1945	An ENU-induced mutation in the mouse Rnf212 gene is associated with male meiotic failure and infertility. Reproduction, 2015, 149, 67-74.	1.1	14
1946	Whole exome sequencing in extended families with autism spectrum disorder implicates four candidate genes. Human Genetics, 2015, 134, 1055-1068.	1.8	49
1947	The structure of the Cyberlindnera jadinii genome and its relation to Candida utilis analyzed by the occurrence of single nucleotide polymorphisms. Journal of Biotechnology, 2015, 211, 20-30.	1.9	10
1948	Genomics and drug profiling of fatal TCF3-HLFâ"positive acute lymphoblastic leukemia identifies recurrent mutation patterns and therapeutic options. Nature Genetics, 2015, 47, 1020-1029.	9.4	190
1949	Missense mutations in<i>TENM4</i>, a regulator of axon guidance and central myelination, cause essential tremor. Human Molecular Genetics, 2015, 24, 5677-5686.	1.4	134
1950	High-throughput genetic characterization of a cohort of Brugada syndrome patients. Human Molecular Genetics, 2015, 24, 5828-5835.	1.4	35
1951	Exome Sequencing of Bilateral Testicular Germ Cell Tumors Suggests Independent Development Lineages. Neoplasia, 2015, 17, 167-174.	2.3	17
1952	Next generation sequencing in endocrine practice. Molecular Genetics and Metabolism, 2015, 115, 61-71.	0.5	24
1953	Key features and clinical variability of COG6-CDG. Molecular Genetics and Metabolism, 2015, 116, 163-170.	0.5	49

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1954	Relapsed neuroblastomas show frequent RAS-MAPK pathway mutations. Nature Genetics, 2015, 47, 864-871.	9.4	451
1955	Germline mutation rates and the long-term phenotypic effects of mutation accumulation in wild-type laboratory mice and mutator mice. Genome Research, 2015, 25, 1125-1134.	2.4	155
1956	Impairment of immunity to <i>Candida</i> and <i>Mycobacterium</i> in humans with bi-allelic <i>RORC</i> mutations. Science, 2015, 349, 606-613.	6.0	366
1957	Concurrent exome-targeted next-generation sequencing and single nucleotide polymorphism array to identify the causative genetic aberrations of isolated Mayer-Rokitansky-Kuster-Hauser syndrome. Human Reproduction, 2015, 30, 1732-1742.	0.4	19
1958	Rare A2ML1 variants confer susceptibility to otitis media. Nature Genetics, 2015, 47, 917-920.	9.4	38
1959	Advanced Proteogenomic Analysis Reveals Multiple Peptide Mutations and Complex Immunoglobulin Peptides in Colon Cancer. Journal of Proteome Research, 2015, 14, 3555-3567.	1.8	36
1960	Allele-specific analysis of DNA replication origins in mammalian cells. Nature Communications, 2015, 6, 7051.	5.8	40
1961	Very small deletions within the NESP55 gene in pseudohypoparathyroidism type 1b. European Journal of Human Genetics, 2015, 23, 494-499.	1.4	20
1962	Upregulation of the microRNA cluster at the Dlk1-Dio3 locus in lung adenocarcinoma. Oncogene, 2015, 34, 94-103.	2.6	46
1963	Whole Exome Sequencing for a Patient with Rubinstein-Taybi Syndrome Reveals de Novo Variants besides an Overt CREBBP Mutation. International Journal of Molecular Sciences, 2015, 16, 5697-5713.	1.8	8
1964	Pseudomonas aeruginosa High-Level Resistance to Polymyxins and Other Antimicrobial Peptides Requires <i>cprA</i> , a Gene That Is Disrupted in the PAO1 Strain. Antimicrobial Agents and Chemotherapy, 2015, 59, 5377-5387.	1.4	23
1965	Loss-of-function variants in ATM confer risk of gastric cancer. Nature Genetics, 2015, 47, 906-910.	9.4	155
1966	The Genetic Architecture of the Genome-Wide Transcriptional Response to ER Stress in the Mouse. PLoS Genetics, 2015, 11, e1004924.	1.5	32
1967	Genomics of Divergence along a Continuum of Parapatric Population Differentiation. PLoS Genetics, 2015, 11, e1004966.	1.5	135
1968	Exome and Transcriptome Sequencing of Aedes aegypti Identifies a Locus That Confers Resistance to Brugia malayi and Alters the Immune Response. PLoS Pathogens, 2015, 11, e1004765.	2.1	37
1969	ConPADE: Genome Assembly Ploidy Estimation from Next-Generation Sequencing Data. PLoS Computational Biology, 2015, 11, e1004229.	1.5	41
1970	Whole-Genome Resequencing Analysis of Hanwoo and Yanbian Cattle to Identify Genome-Wide SNPs and Signatures of Selection. Molecules and Cells, 2015, 38, 466-473.	1.0	77
1971	Genomic landscape of carcinogen-induced and genetically induced mouse skin squamous cell carcinoma. Nature Medicine, 2015, 21, 946-954.	15.2	179

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1972	Consensus Genotyper for Exome Sequencing (CGES): improving the quality of exome variant genotypes. Bioinformatics, 2015, 31, 187-193.	1.8	18
1973	Whole-genome resequencing analyses of five pig breeds, including Korean wild and native, and three European origin breeds. DNA Research, 2015, 22, 259-267.	1.5	54
1974	Novel compound heterozygous DNA ligase IV mutations in an adolescent with a slowly-progressing radiosensitive-severe combined immunodeficiency. Clinical Immunology, 2015, 160, 255-260.	1.4	29
1975	Exome Sequencing Reveals Germline SMAD9 Mutation That Reduces Phosphatase and Tensin Homolog Expression and Is Associated With Hamartomatous Polyposis and Gastrointestinal Ganglioneuromas. Gastroenterology, 2015, 149, 886-889.e5.	0.6	24
1976	HLA-E coding and 3â€² untranslated region variability determined by next-generation sequencing in two West-African population samples. Human Immunology, 2015, 76, 945-953.	1.2	33
1977	Genetic evidence for two founding populations of the Americas. Nature, 2015, 525, 104-108.	13.7	348
1978	Exome Sequencing Analysis Reveals Variants in Primary Immunodeficiency Genes in Patients With Very Early Onset Inflammatory Bowel Disease. Gastroenterology, 2015, 149, 1415-1424.	0.6	99
1979	Germline mutations causing familial lung cancer. Journal of Human Genetics, 2015, 60, 597-603.	1.1	20
1980	PolySia-Specific Retargeting of Oncolytic Viruses Triggers Tumor-Specific Immune Responses and Facilitates Therapy of Disseminated Lung Cancer. Cancer Immunology Research, 2015, 3, 751-763.	1.6	20
1981	Parentâ€“progeny sequencing indicates higher mutation rates in heterozygotes. Nature, 2015, 523, 463-467.	13.7	157
1982	Sparse whole-genome sequencing identifies two loci for major depressive disorder. Nature, 2015, 523, 588-591.	13.7	777
1983	Evidence Suggesting That Discontinuous Dosing of ALK Kinase Inhibitors May Prolong Control of ALK+ Tumors. Cancer Research, 2015, 75, 2916-2927.	0.4	40
1984	Resolving Evolutionary Relationships in Closely Related Species with Whole-Genome Sequencing Data. Systematic Biology, 2015, 64, 1000-1017.	2.7	102
1985	Experimental Swap of Anopheles gambiae's Assortative Mating Preferences Demonstrates Key Role of X-Chromosome Divergence Island in Incipient Sympatric Speciation. PLoS Genetics, 2015, 11, e1005141.	1.5	34
1986	Polymerase Î¸ is a key driver of genome evolution and of CRISPR/Cas9-mediated mutagenesis. Nature Communications, 2015, 6, 7394.	5.8	87
1987	Integrated Genomic Analysis Suggests <i>MLL3</i> Is a Novel Candidate Susceptibility Gene for Familial Nasopharyngeal Carcinoma. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 1222-1228.	1.1	17
1988	VarSim: a high-fidelity simulation and validation framework for high-throughput genome sequencing with cancer applications. Bioinformatics, 2015, 31, 1469-1471.	1.8	59
1989	<i>De novo</i>point mutations in patients diagnosed with ataxic cerebral palsy. Brain, 2015, 138, 1817-1832.	3.7	129

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1990	Whole-Genome Sequencing of Six Mauritian Cynomolgus Macaques (Macaca fascicularis) Reveals a Genome-Wide Pattern of Polymorphisms under Extreme Population Bottleneck. Genome Biology and Evolution, 2015, 7, 821-830.	1.1	37
1991	Massively parallel quantification of the regulatory effects of noncoding genetic variation in a human cohort. Genome Research, 2015, 25, 1206-1214.	2.4	100
1992	Identification of the gene defect responsible for severe hypercholesterolaemia using whole-exome sequencing. Scientific Reports, 2015, 5, 11380.	1.6	9
1993	A Missense Change in the ATG4D Gene Links Aberrant Autophagy to a Neurodegenerative Vacuolar Storage Disease. PLoS Genetics, 2015, 11, e1005169.	1.5	48
1994	Zebrafish <i>foxc1a</i> drives appendage-specific neural circuit development. Development (Cambridge), 2015, 142, 753-762.	1.2	16
1995	Exome-level comparison of primary well-differentiated neuroendocrine tumors and their cell lines. Cancer Genetics, 2015, 208, 374-381.	0.2	63
1996	Next-generation re-sequencing as a tool for rapid bioinformatic screening of presence and absence of genes and accessory chromosomes across isolates of Zymoseptoria tritici. Fungal Genetics and Biology, 2015, 79, 71-75.	0.9	7
1997	Genomic determinants of coral heat tolerance across latitudes. Science, 2015, 348, 1460-1462.	6.0	473
1998	Viral Infection of Tumors Overcomes Resistance to PD-1-immunotherapy by Broadening Neoantigenome-directed T-cell Responses. Molecular Therapy, 2015, 23, 1630-1640.	3.7	165
1999	Assembly and diploid architecture of an individual human genome via single-molecule technologies. Nature Methods, 2015, 12, 780-786.	9.0	465
2000	SERPINB11 Frameshift Variant Associated with Novel Hoof Specific Phenotype in Connemara Ponies. PLoS Genetics, 2015, 11, e1005122.	1.5	21
2001	Inconsistency and features of single nucleotide variants detected in whole exome sequencing versus transcriptome sequencing: A case study in lung cancer. Methods, 2015, 83, 118-127.	1.9	33
2002	Germline RECQL mutations are associated with breast cancer susceptibility. Nature Genetics, 2015, 47, 643-646.	9.4	168
2003	Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. Genome Research, 2015, 25, 948-957.	2.4	54
2004	The chimerical genome of <scp>I</scp>sla del <scp>C</scp>oco feral pigs (<scp>C</scp>osta) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 18 Ecology, 2015, 24, 2364-2378.	2.0	15
2005	A wide spectrum of EGFR mutations in glioblastoma is detected by a single clinical oncology targeted next-generation sequencing panel. Experimental and Molecular Pathology, 2015, 98, 568-573.	0.9	14
2006	The coffee genome hub: a resource for coffee genomes. Nucleic Acids Research, 2015, 43, D1028-D1035.	6.5	59
2007	Genetic variability in SQSTM1 and risk of early-onset Alzheimer dementia: a European early-onset dementia consortium study. Neurobiology of Aging, 2015, 36, 2005.e15-2005.e22.	1.5	34

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2008	Adult-onset painful axonal polyneuropathy caused by a dominant<i>NAGLU</i>mutation. Brain, 2015, 138, 1477-1483.	3.7	24
2009	Whole exome sequencing combined with integrated variant annotation prediction identifies asymptomatic Tangier disease with compound heterozygous mutations in ABCA1 gene. Atherosclerosis, 2015, 240, 324-329.	0.4	16
2010	Novel mutations in TNFRSF7/CD27: Clinical, immunologic, and genetic characterization of human CD27 deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 703-712.e10.	1.5	109
2011	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660.	9.4	302
2012	Exome Sequencing to Detect Rare Variants Associated With General Cognitive Ability: A Pilot Study. Twin Research and Human Genetics, 2015, 18, 117-125.	0.3	7
2013	Homozygous/Compound Heterozygous Triadin Mutations Associated With Autosomal-Recessive Long-QT Syndrome and Pediatric Sudden Cardiac Arrest. Circulation, 2015, 131, 2051-2060.	1.6	92
2014	De Novo Mutation in X-Linked Hearing Lossâ€"Associated POU3F4 in a Sporadic Case of Congenital Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 169S-176S.	0.6	19
2015	ARID1A and TERT promoter mutations in dedifferentiated meningioma. Cancer Genetics, 2015, 208, 345-350.	0.2	73
2016	Intra-host viral variability in children clinically infected with H1N1 (2009) pandemic influenza. Infection, Genetics and Evolution, 2015, 33, 47-54.	1.0	8
2017	A novel LMX1B mutation in a family with end-stage renal disease of 'unknown cause'. CKJ: Clinical Kidney Journal, 2015, 8, 113-119.	1.4	30
2018	Novel <i>PTPRQ</i> Mutations Identified in Three Congenital Hearing Loss Patients With Various Types of Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 184S-192S.	0.6	19
2019	Mutations in <i>LOXHD1</i> Gene Cause Various Types and Severities of Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 135S-141S.	0.6	24
2020	SNV-PPILP: refined SNV calling for tumor data using perfect phylogenies and ILP. Bioinformatics, 2015, 31, 1133-1135.	1.8	42
2021	A Phylogenetic Analysis of 34 Chloroplast Genomes Elucidates the Relationships between Wild and Domestic Species within the Genus<i>Citrus</i>. Molecular Biology and Evolution, 2015, 32, 2015-2035.	3.5	272
2022	Comparative analysis of primary tumour and matched metastases in colorectal cancer patients: Evaluation of concordance between genomic and transcriptional profiles. European Journal of Cancer, 2015, 51, 791-799.	1.3	83
2023	Rapid and reliable identification of tomato fruit weight and locule number loci by QTL-seq. Theoretical and Applied Genetics, 2015, 128, 1329-1342.	1.8	153
2024	Immunomics of the koala (Phascolarctos cinereus). Immunogenetics, 2015, 67, 305-321.	1.2	19
2025	Mitochondrial genome sequences reveal evolutionary relationships of the Phytophthora 1c clade species. Current Genetics, 2015, 61, 567-577.	0.8	23

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2026	Spinocerebellar ataxia in the Italian Spinone dog is associated with an intronic GAA repeat expansion in ITPR1. Mammalian Genome, 2015, 26, 108-117.	1.0	23
2027	NEFL E396K mutation is associated with a novel dominant intermediate Charcotâ€“Marieâ€“Tooth disease phenotype. Journal of Neurology, 2015, 262, 1289-1300.	1.8	24
2028	Comparative transcriptome analysis of the petal degeneration mutant pdm in Chinese cabbage (Brassica campestris ssp. pekinensis) using RNA-Seq. Molecular Genetics and Genomics, 2015, 290, 1833-1847.	1.0	30
2029	Genomic analysis identifies candidate pathogenic variants in 9 of 18 patients with unexplained West syndrome. Human Genetics, 2015, 134, 649-658.	1.8	51
2030	Characterization and review of MTHFD1 deficiency: four new patients, cellular delineation and response to folic and folinic acid treatment. Journal of Inherited Metabolic Disease, 2015, 38, 863-872.	1.7	39
2031	A novel POLE mutation associated with cancers of colon, pancreas, ovaries and small intestine. Familial Cancer, 2015, 14, 437-448.	0.9	67
2032	Combined Immunodeficiency Due to MALT1 Mutations, Treated by Hematopoietic Cell Transplantation. Journal of Clinical Immunology, 2015, 35, 135-146.	2.0	94
2033	Caspase-8 Deficiency Presenting as Late-Onset Multi-Organ Lymphocytic Infiltration with Granulomas in two Adult Siblings. Journal of Clinical Immunology, 2015, 35, 348-355.	2.0	35
2034	Whole genome analysis of a Vietnamese trio. Journal of Biosciences, 2015, 40, 113-124.	0.5	4
2035	CRISPR/Cas9-mediated gene editing in human tripronuclear zygotes. Protein and Cell, 2015, 6, 363-372.	4.8	929
2036	Viral subpopulation diversity in influenza virus isolates compared to clinical specimens. Journal of Clinical Virology, 2015, 68, 16-23.	1.6	10
2037	Exome sequencing identifies ATP4A gene as responsible of an atypical familial type I gastric neuroendocrine tumour. Human Molecular Genetics, 2015, 24, 2914-2922.	1.4	60
2038	PEAT: an intelligent and efficient paired-end sequencing adapter trimming algorithm. BMC Bioinformatics, 2015, 16, S2.	1.2	49
2039	Underlying Data for Sequencing the Mitochondrial Genome with the Massively Parallel Sequencing Platform Ion Torrentâ„¢ PGMâ„¢. BMC Genomics, 2015, 16, S4.	1.2	43
2040	Comparing variant calling algorithms for target-exon sequencing in a large sample. BMC Bioinformatics, 2015, 16, 75.	1.2	6
2041	GBSX: a toolkit for experimental design and demultiplexing genotyping by sequencing experiments. BMC Bioinformatics, 2015, 16, 73.	1.2	102
2042	Genome-wide detection and characterization of positive selection in Korean Native Black Pig from Jeju Island. BMC Genetics, 2015, 16, 3.	2.7	18
2043	Cetaceans evolution: insights from the genome sequences of common minke whales. BMC Genomics, 2015, 16, 13.	1.2	28

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2044	Whole genome sequencing of an ethnic Pathan (Pakhtun) from the north-west of Pakistan. BMC Genomics, 2015, 16, 172.	1.2	16
2045	A genome-wide scan for signatures of directional selection in domesticated pigs. BMC Genomics, 2015, 16, 130.	1.2	67
2046	Genetic variants in root architecture-related genes in a Glycine soja accession, a potential resource to improve cultivated soybean. BMC Genomics, 2015, 16, 132.	1.2	67
2047	RNASeqBrowser: A genome browser for simultaneous visualization of raw strand specific RNAseq reads and UCSC genome browser custom tracks. BMC Genomics, 2015, 16, 145.	1.2	7
2048	Establishment of Anthoceros agrestis as a model species for studying the biology of hornworts. BMC Plant Biology, 2015, 15, 98.	1.6	53
2049	Targeted Next Generation Sequencing reveals previously unidentified TSC1 and TSC2 mutations. BMC Medical Genetics, 2015, 16, 10.	2.1	62
2050	Variation in DNAH1 may contribute to primary ciliary dyskinesia. BMC Medical Genetics, 2015, 16, 14.	2.1	54
2051	No significant viral transcription detected in whole breast cancer transcriptomes. BMC Cancer, 2015, 15, 147.	1.1	15
2052	The recently identified modifier of murine metastable epialleles, Rearranged L-Myc Fusion, is involved in maintaining epigenetic marks at CpG island shores and enhancers. BMC Biology, 2015, 13, 21.	1.7	16
2053	Epidermolysis bullosa in Danish Hereford calves is caused by a deletion in LAMC2 gene. BMC Veterinary Research, 2015, 11, 23.	0.7	13
2054	DNA-based diagnosis of rare diseases in veterinary medicine: a 4.4Âkb deletion of ITGB4 is associated with epidermolysis bullosa in Charolais cattle. BMC Veterinary Research, 2015, 11, 48.	0.7	14
2055	ASEQ: fast allele-specific studies from next-generation sequencing data. BMC Medical Genomics, 2015, 8, 9.	0.7	51
2056	A simple data-adaptive probabilistic variant calling model. Algorithms for Molecular Biology, 2015, 10, 10.	0.3	3
2057	Causes and consequences of crossing-over evidenced via a high-resolution recombinational landscape of the honey bee. Genome Biology, 2015, 16, 15.	3.8	67
2058	High-depth sequencing of over 750 genes supports linear progression of primary tumors and metastases in most patients with liver-limited metastatic colorectal cancer. Genome Biology, 2015, 16, 32.	3.8	42
2059	A haplotype map of allohexaploid wheat reveals distinct patterns of selection on homoeologous genomes. Genome Biology, 2015, 16, 48.	3.8	216
2060	SNES: single nucleus exome sequencing. Genome Biology, 2015, 16, 55.	13.9	83
2061	Fastq2vcf: a concise and transparent pipeline for whole-exome sequencing data analyses. BMC Research Notes, 2015, 8, 72.	0.6	14

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2062	USH2 Caused by <i>GPR98</i> Mutation Diagnosed by Massively Parallel Sequencing in Advance of the Occurrence of Visual Symptoms. Annals of Otology, Rhinology and Laryngology, 2015, 124, 123S-128S.	0.6	9
2063	Habitat Visualization and Genomic Analysis of â€œCandidatus Pantoea carbekii,â€•the Primary Symbiont of the Brown Marmorated Stink Bug. Genome Biology and Evolution, 2015, 7, 620-635.	1.1	50
2064	Association of a Novel<i>ACTA1</i>Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. JAMA Neurology, 2015, 72, 689.	4.5	35
2065	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. Nature Genetics, 2015, 47, 647-653.	9.4	146
2066	Whole-exome sequencing as a diagnostic tool: current challenges and future opportunities. Expert Review of Molecular Diagnostics, 2015, 15, 749-760.	1.5	62
2067	Whole-Genome Sequencing for National Surveillance of Shiga Toxinâ€"Producing<i>Escherichia coli</i>O157. Clinical Infectious Diseases, 2015, 61, 305-312.	2.9	181
2068	Case Report Whole-exome analysis of a child with polycystic kidney disease and ventriculomegaly. Genetics and Molecular Research, 2015, 14, 3618-3624.	0.3	5
2069	Next-Generation Sequencing of Stage IV Squamous Cell Lung Cancers Reveals an Association of PI3K Aberrations and Evidence of Clonal Heterogeneity in Patients with Brain Metastases. Cancer Discovery, 2015, 5, 610-621.	7.7	129
2070	Examining Phylogenetic Relationships Among Gibbon Genera Using Whole Genome Sequence Data Using an Approximate Bayesian Computation Approach. Genetics, 2015, 200, 295-308.	1.2	44
2071	Homozygosity mapping and sequencing identify two genes that might contribute to pointing behavior in hunting dogs. Canine Genetics and Epidemiology, 2015, 2, 5.	2.9	11
2072	Evaluating the performance of anchored hybrid enrichment at the tips of the tree of life: a phylogenetic analysis of Australian Eugongylus group scincid lizards. BMC Evolutionary Biology, 2015, 15, 62.	3.2	57
2073	Skeletal dysplasia in a consanguineous clan from the island of Nias/Indonesia is caused by a novel mutation in B3GAT3. Human Genetics, 2015, 134, 691-704.	1.8	27
2074	Integrated analysis of whole-exome sequencing and transcriptome profiling in males with autism spectrum disorders. Molecular Autism, 2015, 6, 21.	2.6	106
2075	Single-molecule sequencing reveals the molecular basis of multidrug-resistance in ST772 methicillin-resistant Staphylococcus aureus. BMC Genomics, 2015, 16, 388.	1.2	24
2076	BAP1 mutation is a frequent somatic event in peritoneal malignant mesothelioma. Journal of Translational Medicine, 2015, 13, 122.	1.8	69
2077	Genome-wide patterns and properties of de novo mutations in humans. Nature Genetics, 2015, 47, 822-826.	9.4	384
2078	Genome measures used for quality control are dependent on gene function and ancestry. Bioinformatics, 2015, 31, 318-323.	1.8	134
2079	Germline mutations in ETV6 are associated with thrombocytopenia, red cell macrocytosis and predisposition to lymphoblastic leukemia. Nature Genetics, 2015, 47, 535-538.	9.4	274

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2080	Loss of Î´-catenin function in severe autism. Nature, 2015, 520, 51-56.	13.7	145
2081	A Genome-wide Screen Identifies PAPP-AA-Mediated IGFR Signaling as a Novel Regulator of Habituation Learning. Neuron, 2015, 85, 1200-1211.	3.8	85
2082	Life-threatening influenza and impaired interferon amplification in human IRF7 deficiency. Science, 2015, 348, 448-453.	6.0	389
2083	Prevalence of PALB2 mutations in the Creighton University Breast Cancer Family Registry. Breast Cancer Research and Treatment, 2015, 150, 637-641.	1.1	0
2084	Next-Generation Diagnostics: Gene Panel, Exome, or Whole Genome?. Human Mutation, 2015, 36, 648-655.	1.1	124
2085	Using population data for assessing next-generation sequencing performance. Bioinformatics, 2015, 31, 56-61.	1.8	0
2086	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. Science Translational Medicine, 2015, 7, 270ra6.	5.8	375
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