

Performance comparison of exome DNA sequencing technologies

Nature Biotechnology

29, 908-914

DOI: [10.1038/nbt.1975](https://doi.org/10.1038/nbt.1975)

Citation Report

#	ARTICLE	IF	CITATIONS
1	The best way to capture exons. <i>Nature Methods</i> , 2011, 8, 897-897.	9.0	0
3	Exome Sequencing Identifies 2 Rare Variants for Low High-Density Lipoprotein Cholesterol in an Extended Family. <i>Circulation: Cardiovascular Genetics</i> , 2012, 5, 538-546.	5.1	17
5	Use of whole exome and genome sequencing in the identification of genetic causes of primary immunodeficiencies. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2012, 12, 623-628.	1.1	65
6	The Next Generation of Complex Lung Genetic Studies. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2012, 186, 1087-1094.	2.5	18
7	Discovery of Rare Homozygous Mutations from Studies of Consanguineous Pedigrees. <i>Current Protocols in Human Genetics</i> , 2012, 75, Unit6.12.	3.5	87
8	Next-generation sequencing in hematologic malignancies: what will be the dividends?. <i>Therapeutic Advances in Hematology</i> , 2012, 3, 333-339.	1.1	19
9	Emerging Applications of Single-Cell Diagnostics. <i>Topics in Current Chemistry</i> , 2012, 336, 99-116.	4.0	7
10	Diagnostic applications of next generation sequencing: working towards quality standards/Diagnostische Anwendung von Next Generation Sequencing: Auf dem Weg zu Qualitätsstandards. <i>Laboratoriums Medizin</i> , 2012, 36, .	0.1	4
11	Clinical utility of sequence-based genotype compared with that derivable from genotyping arrays. <i>Journal of the American Medical Informatics Association: JAMIA</i> , 2012, 19, e21-e27.	2.2	4
12	Making the Most of Pathological Specimens: Molecular Diagnosis in Formalin-Fixed, Paraffin Embedded Tissue. <i>Current Drug Targets</i> , 2012, 13, 1475-1487.	1.0	35
13	Whole-genome and whole-exome sequencing in neurological diseases. <i>Nature Reviews Neurology</i> , 2012, 8, 508-517.	4.9	99
14	Systems biology: personalized medicine for the future?. <i>Current Opinion in Pharmacology</i> , 2012, 12, 623-628.	1.7	90
15	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. <i>Neurology</i> , 2012, 79, 127-131.	1.5	35
17	De novo mutations in neurological and psychiatric disorders: effects, diagnosis and prevention. <i>Genome Medicine</i> , 2012, 4, 71.	3.6	14
18	Exome sequencing: a transient technology for molecular diagnostics?. <i>Expert Review of Molecular Diagnostics</i> , 2012, 12, 211-214.	1.5	7
19	Future possibilities in migraine genetics. <i>Journal of Headache and Pain</i> , 2012, 13, 505-511.	2.5	15
20	Exome versus transcriptome sequencing in identifying coding region variants. <i>Expert Review of Molecular Diagnostics</i> , 2012, 12, 241-251.	1.5	43
21	Population-Based Variation in Cardiomyopathy Genes. <i>Circulation: Cardiovascular Genetics</i> , 2012, 5, 391-399.	5.1	126

#	ARTICLE	IF	CITATIONS
22	Rapid Whole-Genome Sequencing for Genetic Disease Diagnosis in Neonatal Intensive Care Units. <i>Science Translational Medicine</i> , 2012, 4, 154ra135.	5.8	534
23	Applications of targeted gene capture and next-generation sequencing technologies in studies of human deafness and other genetic disabilities. <i>Hearing Research</i> , 2012, 288, 67-76.	0.9	101
24	Personal Omics Profiling Reveals Dynamic Molecular and Medical Phenotypes. <i>Cell</i> , 2012, 148, 1293-1307.	13.5	1,134
25	Molecular diagnosis of putative Stargardt disease probands by exome sequencing. <i>BMC Medical Genetics</i> , 2012, 13, 67.	2.1	30
26	Solution-based targeted genomic enrichment for precious DNA samples. <i>BMC Biotechnology</i> , 2012, 12, 20.	1.7	19
27	Mitochondrial genomes gleaned from human whole-exome sequencing. <i>Nature Methods</i> , 2012, 9, 523-524.	9.0	102
28	Genetics of neuromuscular disorders. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 2012, 49, 33-48.	2.7	65
29	Gene discovery in familial cancer syndromes by exome sequencing: prospects for the elucidation of familial colorectal cancer type X. <i>Modern Pathology</i> , 2012, 25, 1055-1068.	2.9	35
30	Technological advances in DNA sequence enrichment and sequencing for germline genetic diagnosis. <i>Expert Review of Molecular Diagnostics</i> , 2012, 12, 159-173.	1.5	16
31	Discovery and Statistical Genotyping of Copy-Number Variation from Whole-Exome Sequencing Depth. <i>American Journal of Human Genetics</i> , 2012, 91, 597-607.	2.6	513
32	Systems Biology Approach for New Target and Biomarker Identification. <i>Current Topics in Microbiology and Immunology</i> , 2012, 363, 169-199.	0.7	17
33	Huvario: a web server resource of whole genome next-generation sequencing allelic frequencies to aid in pathological candidate gene selection. <i>Journal of Clinical Bioinformatics</i> , 2012, 2, 19.	1.2	20
34	Maize (<i>Zea mays</i> L.) Genome Diversity as Revealed by RNA-Sequencing. <i>PLoS ONE</i> , 2012, 7, e33071.	1.1	153
35	FastUniq: A Fast De Novo Duplicates Removal Tool for Paired Short Reads. <i>PLoS ONE</i> , 2012, 7, e52249.	1.1	446
36	Genotyping of Fanconi Anemia Patients by Whole Exome Sequencing: Advantages and Challenges. <i>PLoS ONE</i> , 2012, 7, e52648.	1.1	32
37	Performance comparison of whole-genome sequencing platforms. <i>Nature Biotechnology</i> , 2012, 30, 78-82.	9.4	281
38	Cost-effective, high-throughput DNA sequencing libraries for multiplexed target capture. <i>Genome Research</i> , 2012, 22, 939-946.	2.4	976
39	Application of Next Generation Sequencing to Molecular Diagnosis of Inherited Diseases. <i>Topics in Current Chemistry</i> , 2012, 336, 19-45.	4.0	43

#	ARTICLE	IF	CITATIONS
40	Non-invasive prenatal measurement of the fetal genome. <i>Nature</i> , 2012, 487, 320-324.	13.7	342
41	Whole genome sequencing for quantifying germline mutation frequency in humans and model species: Cautious optimism. <i>Mutation Research - Reviews in Mutation Research</i> , 2012, 750, 96-106.	2.4	25
42	Assessing the Enrichment Performance in Targeted Resequencing Experiments. <i>Human Mutation</i> , 2012, 33, 635-641.	1.1	27
43	Exome sequencing: Dual role as a discovery and diagnostic tool. <i>Annals of Neurology</i> , 2012, 71, 5-14.	2.8	157
44	Multiplex target capture with double-stranded DNA probes. <i>Genome Medicine</i> , 2013, 5, 50.	3.6	18
45	Low concordance of multiple variant-calling pipelines: practical implications for exome and genome sequencing. <i>Genome Medicine</i> , 2013, 5, 28.	3.6	381
46	Clinical application of targeted and genome-wide technologies: can we predict treatment responses in chronic lymphocytic leukemia?. <i>Personalized Medicine</i> , 2013, 10, 361-376.	0.8	9
47	Quantifying single nucleotide variant detection sensitivity in exome sequencing. <i>BMC Bioinformatics</i> , 2013, 14, 195.	1.2	74
48	Assessment of clinical analytical sensitivity and specificity of next-generation sequencing for detection of simple and complex mutations. <i>BMC Genetics</i> , 2013, 14, 6.	2.7	71
49	Variant discovery in targeted resequencing using whole genome amplified DNA. <i>BMC Genomics</i> , 2013, 14, 468.	1.2	7
50	ACMG clinical laboratory standards for next-generation sequencing. <i>Genetics in Medicine</i> , 2013, 15, 733-747.	1.1	794
51	Personal genomes, quantitative dynamic omics and personalized medicine. <i>Quantitative Biology</i> , 2013, 1, 71-90.	0.3	29
52	A glimpse into past, present, and future DNA sequencing. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 3-24.	0.5	146
53	Impact of the next-generation sequencing data depth on various biological result inferences. <i>Science China Life Sciences</i> , 2013, 56, 104-109.	2.3	11
54	The Role of New Sequencing Technology in Identifying Rare Mutations in New Susceptibility Genes for Cancer. <i>Current Genetic Medicine Reports</i> , 2013, 1, 175-181.	1.9	1
55	Whole-exome sequencing identifies tetratricopeptide repeat domain 7A (TTC7A) mutations for combined immunodeficiency with intestinal atresias. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 656-664.e17.	1.5	140
56	Impacts of Variation in the Human Genome on Gene Regulation. <i>Journal of Molecular Biology</i> , 2013, 425, 3970-3977.	2.0	125
57	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. <i>Human Mutation</i> , 2013, 34, 1721-1726.	1.1	303

#	ARTICLE	IF	CITATIONS
58	Detection of structural DNA variation from next generation sequencing data: a review of informatic approaches. <i>Cancer Genetics</i> , 2013, 206, 432-440.	0.2	131
59	Whole-exome sequencing links caspase recruitment domain 11 (CARD11) inactivation to severe combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1376-1383.e3.	1.5	127
60	Next Generation Sequencing. , 2013, , .		10
61	Bioinformatic perspectives in the neuronal ceroid lipofuscinoses. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2013, 1832, 1831-1841.	1.8	10
62	Oral Cavity and Oropharyngeal Squamous Cell Carcinoma Genomics. <i>Otolaryngologic Clinics of North America</i> , 2013, 46, 545-566.	0.5	14
63	Estimating exome genotyping accuracy by comparing to data from large scale sequencing projects. <i>Genome Medicine</i> , 2013, 5, 69.	3.6	23
64	Next generation sequencing and rare genetic variants: From human population studies to medical genetics. <i>Environmental and Molecular Mutagenesis</i> , 2013, 54, 518-532.	0.9	10
65	Detection of Clinically Relevant Genetic Variants in Autism Spectrum Disorder by Whole-Genome Sequencing. <i>American Journal of Human Genetics</i> , 2013, 93, 249-263.	2.6	429
66	Next-generation sequencing in the clinic: Promises and challenges. <i>Cancer Letters</i> , 2013, 340, 284-295.	3.2	272
67	Impacts of massively parallel sequencing for genetic diagnosis of neuromuscular disorders. <i>Acta Neuropathologica</i> , 2013, 125, 173-185.	3.9	37
68	Approaches to homozygosity mapping and exome sequencing for the identification of novel types of CDG. <i>Glycoconjugate Journal</i> , 2013, 30, 67-76.	1.4	16
69	From the periphery to centre stage: de novo single nucleotide variants play a key role in human genetic disease. <i>Journal of Medical Genetics</i> , 2013, 50, 203-211.	1.5	33
70	Promise of personalized omics to precision medicine. <i>Wiley Interdisciplinary Reviews: Systems Biology and Medicine</i> , 2013, 5, 73-82.	6.6	245
71	Molecular diagnosis of congenital muscular dystrophies with defective glycosylation of alpha-dystroglycan using next-generation sequencing technology. <i>Neuromuscular Disorders</i> , 2013, 23, 337-344.	0.3	13
72	Diagnostic Applications of Next Generation Sequencing in Immunogenetics and Molecular Oncology. <i>Transfusion Medicine and Hemotherapy</i> , 2013, 40, 196-206.	0.7	21
73	Next Generation Diagnostics in Inherited Arrhythmia Syndromes. <i>Journal of Cardiovascular Translational Research</i> , 2013, 6, 94-103.	1.1	31
74	High-throughput sequencing for biology and medicine. <i>Molecular Systems Biology</i> , 2013, 9, 640.	3.2	251
75	Integrated genomic analyses identify ARID1A and ARID1B alterations in the childhood cancer neuroblastoma. <i>Nature Genetics</i> , 2013, 45, 12-17.	9.4	374

#	ARTICLE	IF	CITATIONS
76	Next-Generation Sequencing: From Understanding Biology to Personalized Medicine. <i>Biology</i> , 2013, 2, 378-398.	1.3	35
77	Existing and Emerging Technologies for Tumor Genomic Profiling. <i>Journal of Clinical Oncology</i> , 2013, 31, 1815-1824.	0.8	129
78	A new paradigm emerges from the study of de novo mutations in the context of neurodevelopmental disease. <i>Molecular Psychiatry</i> , 2013, 18, 141-153.	4.1	85
79	Human Genome Variation Discovery via Exome and Whole-Genome Sequencing. , 2013, , 94-101.		0
80	Exome Sequencing by Targeted Enrichment. <i>Current Protocols in Molecular Biology</i> , 2013, 102, Unit7.12.	2.9	3
81	The new sequencer on the block: comparison of Life Technology's Proton sequencer to an Illumina HiSeq for whole-exome sequencing. <i>Human Genetics</i> , 2013, 132, 1153-1163.	1.8	75
82	Developing Genome and Exome Sequencing for Candidate Gene Identification in Inherited Disorders: An Integrated Technical and Bioinformatics Approach. <i>Archives of Pathology and Laboratory Medicine</i> , 2013, 137, 415-433.	1.2	45
83	Guidelines and Approaches to Compliance with Regulatory and Clinical Standards: Quality Control Procedures and Quality Assurance. , 2013, , 255-273.		4
84	Next Generation Sequencing Technologies in Medical Genetics. <i>SpringerBriefs in Genetics</i> , 2013, , .	0.1	3
85	Mitochondrial genetics. <i>British Medical Bulletin</i> , 2013, 106, 135-159.	2.7	275
86	Detecting Alu insertions from high-throughput sequencing data. <i>Nucleic Acids Research</i> , 2013, 41, e169-e169.	6.5	20
87	A Review of DNA Enrichment Technologies. <i>SpringerBriefs in Genetics</i> , 2013, , 25-32.	0.1	1
88	Clinical application of massively parallel sequencing in the molecular diagnosis of glycogen storage diseases of genetically heterogeneous origin. <i>Genetics in Medicine</i> , 2013, 15, 106-114.	1.1	65
89	Applications and data analysis of next-generation sequencing. <i>Laboratoriums Medizin</i> , 2013, 37, .	0.1	3
90	Whole-Exome Sequencing. <i>Journal of Bone and Joint Surgery - Series A</i> , 2013, 95, e185.	1.4	6
91	Implementing Genomic Medicine in Pathology. <i>Advances in Anatomic Pathology</i> , 2013, 20, 238-244.	2.4	20
92	MiST: A new approach to variant detection in deep sequencing datasets. <i>Nucleic Acids Research</i> , 2013, 41, e154-e154.	6.5	6
93	Short Read (Next-Generation) Sequencing. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 427-434.	5.1	23

#	ARTICLE	IF	CITATIONS
94	Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. <i>Genome Medicine</i> , 2013, 5, 57.	3.6	143
95	Next-Generation Sequence Analysis of Genes Associated with Obesity and Nonalcoholic Fatty Liver Disease-Related Cirrhosis in Extreme Obesity. <i>Human Heredity</i> , 2013, 75, 144-151.	0.4	22
96	Mutations in Extracellular Matrix Genes <i>NID1</i> and <i>LAMC1</i> Cause Autosomal Dominant Dandy-Walker Malformation and Occipital Cephaloceles. <i>Human Mutation</i> , 2013, 34, 1075-1079.	1.1	38
97	Ethical issues in neurogenetic disorders. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 118, 265-276.	1.0	5
98	Mutations in <i>AP2S1</i> cause familial hypocalciuric hypercalcemia type 3. <i>Nature Genetics</i> , 2013, 45, 93-97.	9.4	242
99	A commentary on The diagnostic utility of exome sequencing in Joubert syndrome and related disorders. <i>Journal of Human Genetics</i> , 2013, 58, 57-57.	1.1	2
100	EXCAVATOR: detecting copy number variants from whole-exome sequencing data. <i>Genome Biology</i> , 2013, 14, R120.	13.9	213
101	Massively parallel sequencing for diagnosing clinically and genetically heterogeneous disorders. <i>Personalized Medicine</i> , 2013, 10, 613-619.	0.8	0
102	DNA sequencing methods in human genetics and disease research. <i>F1000prime Reports</i> , 2013, 5, 34.	5.9	6
103	Computational methods for detecting copy number variations in cancer genome using next generation sequencing: principles and challenges. <i>Oncotarget</i> , 2013, 4, 1868-1881.	0.8	77
104	Exome sequencing: what clinicians need to know. <i>Advances in Genomics and Genetics</i> , 0, , 15.	0.8	6
105	±1-A680T Variant in <i>GLCY1A3</i> as a Candidate Conferring Protection From Pulmonary Hypertension Among Kyrgyz Highlanders. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 920-929.	5.1	23
106	Next Generation Sequencing for Disorders of Sex Development. <i>Endocrine Development</i> , 2014, 27, 53-62.	1.3	14
107	Next-generation genebanking: plant genetic resources management and utilization in the sequencing era. <i>Plant Genetic Resources: Characterisation and Utilisation</i> , 2014, 12, 298-307.	0.4	38
108	A commentary on the promise of whole-exome sequencing in medical genetics. <i>Journal of Human Genetics</i> , 2014, 59, 117-118.	1.1	9
109	CANOES: detecting rare copy number variants from whole exome sequencing data. <i>Nucleic Acids Research</i> , 2014, 42, e97-e97.	6.5	123
110	Genodermatoses. Part II: Other Hereditary Dermatologic Disease. , 2014, , 253-312.		0
111	Next-Generation Sequencing in Clinical Oncology: Next Steps Towards Clinical Validation. <i>Cancers</i> , 2014, 6, 2296-2312.	1.7	48

#	ARTICLE	IF	CITATIONS
112	Estimating genotype error rates from high-coverage next-generation sequence data. <i>Genome Research</i> , 2014, 24, 1734-1739.	2.4	121
113	Newborn screening: the genomic challenge. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 81-84.	0.6	19
114	Medical genomics: The intricate path from genetic variant identification to clinical interpretation. <i>Applied & Translational Genomics</i> , 2014, 3, 60-67.	2.1	32
115	Exome sequencing and genome-wide copy number variant mapping reveal novel associations with sensorineural hereditary hearing loss. <i>BMC Genomics</i> , 2014, 15, 1155.	1.2	27
116	Reducing INDEL calling errors in whole genome and exome sequencing data. <i>Genome Medicine</i> , 2014, 6, 89.	3.6	144
117	Performance comparison of SNP detection tools with illumina exome sequencing data – an assessment using both family pedigree information and sample-matched SNP array data. <i>Nucleic Acids Research</i> , 2014, 42, e101-e101.	6.5	50
118	A programmable method for massively parallel targeted sequencing. <i>Nucleic Acids Research</i> , 2014, 42, e88-e88.	6.5	13
119	Improved Variant Calling Accuracy by Merging Replicates in Whole-Exome Sequencing Studies. <i>BioMed Research International</i> , 2014, 2014, 1-7.	0.9	12
120	Whole exome sequencing for cancer – is there evidence of clinical utility?. <i>Advances in Genomics and Genetics</i> , 2014, , 115.	0.8	2
121	Targeted sequencing for gene discovery and quantification using RNA CaptureSeq. <i>Nature Protocols</i> , 2014, 9, 989-1009.	5.5	171
122	Ancestry estimation and control of population stratification for sequence-based association studies. <i>Nature Genetics</i> , 2014, 46, 409-415.	9.4	136
123	Exome sequencing greatly expedites the progressive research of Mendelian diseases. <i>Frontiers of Medicine</i> , 2014, 8, 42-57.	1.5	48
124	Whole exome sequencing detects homozygosity for ABCA4 p.Arg602Trp missense mutation in a pediatric patient with rapidly progressive retinal dystrophy. <i>BMC Medical Genetics</i> , 2014, 15, 11.	2.1	9
125	Genomic tools in acute myeloid leukemia: From the bench to the bedside. <i>Cancer</i> , 2014, 120, 1134-1144.	2.0	21
126	Performance of Common Analysis Methods for Detecting Low-Frequency Single Nucleotide Variants in Targeted Next-Generation Sequence Data. <i>Journal of Molecular Diagnostics</i> , 2014, 16, 75-88.	1.2	124
127	Gene hunting in the genomic era: Approaches to diagnostic dilemmas in patients with primary immunodeficiencies. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 262-268.	1.5	34
128	Sequencing depth and coverage: key considerations in genomic analyses. <i>Nature Reviews Genetics</i> , 2014, 15, 121-132.	7.7	1,116
129	Anchored multiplex PCR for targeted next-generation sequencing. <i>Nature Medicine</i> , 2014, 20, 1479-1484.	15.2	705

#	ARTICLE	IF	CITATIONS
130	Human leukocyte antigen haplotype phasing by allele-specific enrichment with peptide nucleic acid probes. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 245-253.	0.6	2
131	Chemical Diagnostics. <i>Topics in Current Chemistry</i> , 2014, , .	4.0	2
132	Comprehensive analysis of common coding sequence variants in Taiwanese Han population. <i>Biomarkers and Genomic Medicine</i> , 2014, 6, 133-143.	0.2	1
133	Targeted Analysis of Whole Genome Sequence Data to Diagnose Genetic Cardiomyopathy. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 751-759.	5.1	53
134	Resolving the genetic heterogeneity of prelingual hearing loss within one family: Performance comparison and application of two targeted next generation sequencing approaches. <i>Journal of Human Genetics</i> , 2014, 59, 599-607.	1.1	16
135	Molecular Diagnostics for Dermatology. , 2014, , .		2
136	Clinical exome sequencing in daily practice: 1,000 patients and beyond. <i>Genome Medicine</i> , 2014, 6, 2.	3.6	23
137	Targeted gene panel sequencing in children with very early onset inflammatory bowel disease—evaluation and prospective analysis. <i>Journal of Medical Genetics</i> , 2014, 51, 748-755.	1.5	91
138	Variant detection sensitivity and biases in whole genome and exome sequencing. <i>BMC Bioinformatics</i> , 2014, 15, 247.	1.2	197
139	Performance comparison of four exome capture systems for deep sequencing. <i>BMC Genomics</i> , 2014, 15, 449.	1.2	152
140	Validation of multiple single nucleotide variation calls by additional exome analysis with a semiconductor sequencer to supplement data of whole-genome sequencing of a human population. <i>BMC Genomics</i> , 2014, 15, 673.	1.2	10
141	Whole-genome sequencing of clarithromycin resistant <i>Helicobacter pylori</i> characterizes unidentified variants of multidrug resistant efflux pump genes. <i>Gut Pathogens</i> , 2014, 6, 27.	1.6	41
142	SeqControl: process control for DNA sequencing. <i>Nature Methods</i> , 2014, 11, 1071-1075.	9.0	10
143	Section II: Hematolymphoid malignancies. <i>Current Problems in Cancer</i> , 2014, 38, 159-174.	1.0	3
144	Analysis of off-target effects of CRISPR/Cas-derived RNA-guided endonucleases and nickases. <i>Genome Research</i> , 2014, 24, 132-141.	2.4	1,195
145	Exome capture from saliva produces high quality genomic and metagenomic data. <i>BMC Genomics</i> , 2014, 15, 262.	1.2	34
146	Analytical validation of whole exome and whole genome sequencing for clinical applications. <i>BMC Medical Genomics</i> , 2014, 7, 20.	0.7	100
147	Opening the genetic toolbox of niche model organisms with high throughput techniques: Novel proteins in regeneration as a case study. <i>BioEssays</i> , 2014, 36, 407-418.	1.2	7

#	ARTICLE	IF	CITATIONS
148	Toward better understanding of artifacts in variant calling from high-coverage samples. <i>Bioinformatics</i> , 2014, 30, 2843-2851.	1.8	790
149	Pseudogenes. <i>Methods in Molecular Biology</i> , 2014, 1167, v.	0.4	5
150	Extended Blood Group Molecular Typing and Next-Generation Sequencing. <i>Transfusion Medicine Reviews</i> , 2014, 28, 177-186.	0.9	41
151	Genomics in Newborn Screening. <i>Journal of Pediatrics</i> , 2014, 164, 14-19.	0.9	61
152	Successes and challenges of using whole exome sequencing to identify novel genes underlying an inherited predisposition for thoracic aortic aneurysms and acute aortic dissections. <i>Trends in Cardiovascular Medicine</i> , 2014, 24, 53-60.	2.3	35
153	Fine-mapping QTLs in advanced intercross lines and other outbred populations. <i>Mammalian Genome</i> , 2014, 25, 271-292.	1.0	25
154	ngsCAT: a tool to assess the efficiency of targeted enrichment sequencing. <i>Bioinformatics</i> , 2014, 30, 1767-1768.	1.8	9
155	Discovery of single-gene inborn errors of immunity by next generation sequencing. <i>Current Opinion in Immunology</i> , 2014, 30, 17-23.	2.4	83
156	Development and performance of a targeted whole exome sequencing enrichment kit for the dog (<i>Canis Familiaris</i> Build 3.1). <i>Scientific Reports</i> , 2014, 4, 5597.	1.6	23
157	Application of Massively Parallel Sequencing in the Clinical Diagnostic Testing of Inherited Cardiac Conditions. <i>Medical Sciences (Basel, Switzerland)</i> , 2014, 2, 98-126.	1.3	2
158	Identification of 2R-ohnologue gene families displaying the same mutation-load skew in multiple cancers. <i>Open Biology</i> , 2014, 4, 140029.	1.5	17
159	Genetic variant in folate homeostasis is associated with lower warfarin dose in African Americans. <i>Blood</i> , 2014, 124, 2298-2305.	0.6	57
160	DNA Testing for Malignant Hyperthermia. <i>Anesthesia and Analgesia</i> , 2014, 118, 397-406.	1.1	45
161	The long tail and rare disease research: the impact of next-generation sequencing for rare Mendelian disorders. <i>Genetical Research</i> , 2015, 97, e15.	0.3	21
162	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Protein-coding Regions. <i>Human Mutation</i> , 2015, 36, 815-822.	1.1	156
163	Evaluation of Hybridization Capture Versus Amplicon-based Methods for Whole-exome Sequencing. <i>Human Mutation</i> , 2015, 36, 903-914.	1.1	206
164	Tempo and mode of genomic mutations unveil human evolutionary history. <i>Genes and Genetic Systems</i> , 2015, 90, 123-131.	0.2	0
165	Improved canine exome designs, featuring ncRNAs and increased coverage of protein coding genes. <i>Scientific Reports</i> , 2015, 5, 12810.	1.6	31

#	ARTICLE	IF	CITATIONS
166	A combination of targeted enrichment methodologies for whole-exome sequencing reveals novel pathogenic mutations. <i>Scientific Reports</i> , 2015, 5, 9331.	1.6	14
167	Replicate exome-sequencing in a multiple-generation family: improved interpretation of next-generation sequencing data. <i>BMC Genomics</i> , 2015, 16, 998.	1.2	6
168	Overview of Target Enrichment Strategies. <i>Current Protocols in Molecular Biology</i> , 2015, 112, 7.21.1-7.21.23.	2.9	70
169	microRNAs in avian influenza virus H9N2-infected and non-infected chicken embryo fibroblasts. <i>Genetics and Molecular Research</i> , 2015, 14, 9081-9091.	0.3	23
170	Lessons Learned from Whole Exome Sequencing in Multiplex Families Affected by a Complex Genetic Disorder, Intracranial Aneurysm. <i>PLoS ONE</i> , 2015, 10, e0121104.	1.1	32
171	A Robust Protocol to Increase NimbleGen SeqCap EZ Multiplexing Capacity to 96 Samples. <i>PLoS ONE</i> , 2015, 10, e0123872.	1.1	13
172	Low-Frequency IL23R Coding Variant Associated with Crohn's Disease Susceptibility in Japanese Subjects Identified by Personal Genomics Analysis. <i>PLoS ONE</i> , 2015, 10, e0137801.	1.1	8
173	Exome Capture with Heterologous Enrichment in Pig (<i>Sus scrofa</i>). <i>PLoS ONE</i> , 2015, 10, e0139328.	1.1	1
174	Reliably Detecting Clinically Important Variants Requires Both Combined Variant Calls and Optimized Filtering Strategies. <i>PLoS ONE</i> , 2015, 10, e0143199.	1.1	38
175	Structural variation discovery in the cancer genome using next generation sequencing: Computational solutions and perspectives. <i>Oncotarget</i> , 2015, 6, 5477-5489.	0.8	33
176	Whole-Exome Enrichment with the Agilent SureSelect Human All Exon Platform. <i>Cold Spring Harbor Protocols</i> , 2015, 2015, pdb.prot083659.	0.2	38
177	Whole-Exome Enrichment with the Roche NimbleGen SeqCap EZ Exome Library SR Platform. <i>Cold Spring Harbor Protocols</i> , 2015, 2015, pdb.prot084855.	0.2	13
178	Whole-Exome Enrichment with the Illumina TruSeq Exome Enrichment Platform. <i>Cold Spring Harbor Protocols</i> , 2015, 2015, pdb.prot084863.	0.2	11
179	Microfluidic droplet enrichment for targeted sequencing. <i>Nucleic Acids Research</i> , 2015, 43, e86-e86.	6.5	32
180	Flexible, Scalable, and Efficient Targeted Resequencing on a Benchtop Sequencer for Variant Detection in Clinical Practice. <i>Human Mutation</i> , 2015, 36, 379-387.	1.1	43
181	The Impact of DNA Input Amount and DNA Source on the Performance of Whole-Exome Sequencing in Cancer Epidemiology. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2015, 24, 1207-1213.	1.1	26
182	Carrier frequency of guanidinoacetate methyltransferase deficiency in the general population by functional characterization of missense variants in the GAMT gene. <i>Molecular Genetics and Genomics</i> , 2015, 290, 2163-2171.	1.0	23
183	Exome Capture and Capturing Technologies in Cancer Research. , 2015, , 279-302.		0

#	ARTICLE	IF	CITATIONS
184	Exome Sequencing: Current and Future Perspectives. <i>G3: Genes, Genomes, Genetics</i> , 2015, 5, 1543-1550.	0.8	165
185	Somatic Diseases (Cancer). , 2015, , 343-360.		0
186	Insertions and Deletions (Indels). , 2015, , 129-150.		12
187	Targeted Hybrid Capture for Inherited Disease Panels. , 2015, , 251-269.		0
188	Targeted Hybrid Capture Methods. , 2015, , 37-55.		1
189	Comparison of whole-genome (13X) and capture (87X) resequencing methods for <i>SNP</i> and genotype callings. <i>Animal Genetics</i> , 2015, 46, 82-86.	0.6	2
190	Performance comparison of four commercial human whole-exome capture platforms. <i>Scientific Reports</i> , 2015, 5, 12742.	1.6	68
191	Genotyping of common <i>SIRPB1</i> copy number variant using Paralogue Ratio Test coupled to MALDI-MS quantification. <i>Molecular and Cellular Probes</i> , 2015, 29, 517-521.	0.9	2
192	New insights into the performance of human whole-exome capture platforms. <i>Nucleic Acids Research</i> , 2015, 43, e76-e76.	6.5	103
193	Understanding next generation sequencing in oncology: A guide for oncologists. <i>Critical Reviews in Oncology/Hematology</i> , 2015, 96, 463-474.	2.0	38
194	Targeted single molecule sequencing methodology for ovarian hyperstimulation syndrome. <i>BMC Genomics</i> , 2015, 16, 264.	1.2	8
195	Next Generation Sequencing in Cancer Research, Volume 2. , 2015, , .		4
196	Whole-genome sequencing is more powerful than whole-exome sequencing for detecting exome variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 5473-5478.	3.3	475
197	Cancer whole-genome sequencing: present and future. <i>Oncogene</i> , 2015, 34, 5943-5950.	2.6	87
198	Comparison and evaluation of two exome capture kits and sequencing platforms for variant calling. <i>BMC Genomics</i> , 2015, 16, 581.	1.2	17
199	Newborn screening and the era of medical genomics. <i>Seminars in Perinatology</i> , 2015, 39, 617-622.	1.1	17
200	Continuously tunable nucleic acid hybridization probes. <i>Nature Methods</i> , 2015, 12, 1191-1196.	9.0	48
201	Hidden Genetic Variation in LCA9-associated Congenital Blindness Explained by 5'UTR Mutations and Copy Number Variations of <i>NMNAT1</i> . <i>Human Mutation</i> , 2015, 36, 1188-1196.	1.1	30

#	ARTICLE	IF	CITATIONS
202	BRAF Targets in Melanoma. <i>Cancer Drug Discovery and Development</i> , 2015, , .	0.2	2
203	Comparison of Custom Capture for Targeted Next-Generation DNA Sequencing. <i>Journal of Molecular Diagnostics</i> , 2015, 17, 64-75.	1.2	65
204	Clinical Exome Performance for Reporting Secondary Genetic Findings. <i>Clinical Chemistry</i> , 2015, 61, 213-220.	1.5	34
205	Constitutional Disorders. , 2015, , 271-296.		0
207	Genomic Applications in Pathology. , 2015, , .		1
208	Enhanced whole exome sequencing by higher DNA insert lengths. <i>BMC Genomics</i> , 2016, 17, 399.	1.2	8
209	Challenges of Identifying Clinically Actionable Genetic Variants for Precision Medicine. <i>Journal of Healthcare Engineering</i> , 2016, 2016, 1-14.	1.1	34
210	Diagnostics of Primary Immunodeficiencies through Next-Generation Sequencing. <i>Frontiers in Immunology</i> , 2016, 7, 466.	2.2	80
211	Clinical Genomics: Challenges and Opportunities. <i>Critical Reviews in Eukaryotic Gene Expression</i> , 2016, 26, 97-113.	0.4	12
212	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. <i>Annals of the New York Academy of Sciences</i> , 2016, 1366, 49-60.	1.8	23
213	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. <i>Genome Biology</i> , 2016, 17, 243.	3.8	241
214	Integrating next-generation sequencing into clinical oncology: strategies, promises and pitfalls. <i>ESMO Open</i> , 2016, 1, e000094.	2.0	126
215	Single molecule targeted sequencing for cancer gene mutation detection. <i>Scientific Reports</i> , 2016, 6, 26110.	1.6	7
216	A systematic comparison of two new releases of exome sequencing products: the aim of use determines the choice of product. <i>Biological Chemistry</i> , 2016, 397, 791-801.	1.2	15
217	Targeted capture in evolutionary and ecological genomics. <i>Molecular Ecology</i> , 2016, 25, 185-202.	2.0	295
218	Precision Medicine in Gastrointestinal Pathology. <i>Archives of Pathology and Laboratory Medicine</i> , 2016, 140, 449-460.	1.2	3
219	Medical implications of technical accuracy in genome sequencing. <i>Genome Medicine</i> , 2016, 8, 24.	3.6	123
220	Genetics of Common Endocrine Disease: The Present and the Future. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 787-794.	1.8	12

#	ARTICLE	IF	CITATIONS
221	Exome and genome sequencing for inborn errors of immunity. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 957-969.	1.5	187
222	The Role of Quality Control in Targeted Next-generation Sequencing Library Preparation. <i>Genomics, Proteomics and Bioinformatics</i> , 2016, 14, 200-206.	3.0	9
223	Translating cancer genomes and transcriptomes for precision oncology. <i>Ca-A Cancer Journal for Clinicians</i> , 2016, 66, 75-88.	157.7	133
224	A highly robust and optimized sequence-based approach for genetic polymorphism discovery and genotyping in large plant populations. <i>Theoretical and Applied Genetics</i> , 2016, 129, 1739-1757.	1.8	12
225	Enhanced copy number variants detection from whole-exome sequencing data using EXCAVATOR2. <i>Nucleic Acids Research</i> , 2016, 44, gkw695.	6.5	75
226	Design of efficient simplified genomic DNA and bisulfite sequencing in large plant populations. <i>Quantitative Biology</i> , 2016, 4, 226-239.	0.3	0
227	Assessment of the latest NGS enrichment capture methods in clinical context. <i>Scientific Reports</i> , 2016, 6, 20948.	1.6	76
228	Performance comparison of two commercial human whole-exome capture systems on formalin-fixed paraffin-embedded lung adenocarcinoma samples. <i>BMC Cancer</i> , 2016, 16, 692.	1.1	27
229	The Rise and Rise of Exome Sequencing. <i>Public Health Genomics</i> , 2016, 19, 315-324.	0.6	15
230	Exome sequencing explained: a practical guide to its clinical application. <i>Briefings in Functional Genomics</i> , 2016, 15, 374-384.	1.3	58
231	Depletion of Abundant Sequences by Hybridization (DASH): using Cas9 to remove unwanted high-abundance species in sequencing libraries and molecular counting applications. <i>Genome Biology</i> , 2016, 17, 41.	3.8	248
232	Next Generation Sequencing Data Analysis in Primary Immunodeficiency Disorders – Future Directions. <i>Journal of Clinical Immunology</i> , 2016, 36, 68-75.	2.0	63
233	Next-Generation Sequencing: Principles for Clinical Application. , 2016, , 889-909.		0
234	Next-Generation Sequencing for the Analysis of Cancer Specimens. , 2016, , 911-931.		0
235	Metagenomes reveal microbial structures, functional potentials, and biofouling-related genes in a membrane bioreactor. <i>Applied Microbiology and Biotechnology</i> , 2016, 100, 5109-5121.	1.7	46
236	Advances in clinical next-generation sequencing: target enrichment and sequencing technologies. <i>Expert Review of Molecular Diagnostics</i> , 2016, 16, 357-372.	1.5	63
237	Lessons learned from gene identification studies in Mendelian epilepsy disorders. <i>European Journal of Human Genetics</i> , 2016, 24, 961-967.	1.4	20
238	Generating Exome Enriched Sequencing Libraries from Formalin-Fixed, Paraffin-Embedded Tissue DNA for Next-Generation Sequencing. <i>Current Protocols in Human Genetics</i> , 2017, 92, 18.10.1-18.10.25.	3.5	3

#	ARTICLE	IF	CITATIONS
239	Next Generation Sequencing Based Clinical Molecular Diagnosis of Human Genetic Disorders. , 2017, , .		2
240	Targeted sequencing of both DNA strands barcoded and captured individually by RNA probes to identify genome-wide ultra-rare mutations. Scientific Reports, 2017, 7, 3356.	1.6	10
241	The Applications and Challenges of Next-Generation Sequencing in Diagnosing Neuromuscular Disorders. , 2017, , 177-200.		2
242	Pathways in bacterial and archaeal communities dictated by ammonium stress in a high solid anaerobic digester with dewatered sludge. Bioresource Technology, 2017, 241, 95-102.	4.8	52
243	A genetic risk factor for thrombophilia in a Han Chinese family. Molecular Medicine Reports, 2017, 15, 1668-1672.	1.1	3
244	Guidelines for Validation of Next-Generation Sequencingâ€‘Based Oncology Panels. Journal of Molecular Diagnostics, 2017, 19, 341-365.	1.2	524
245	Genomic sequencing and precision medicine in head and neck cancers. European Journal of Surgical Oncology, 2017, 43, 884-892.	0.5	12
246	Promises and pitfalls of Illumina sequencing for HIV resistance genotyping. Virus Research, 2017, 239, 97-105.	1.1	27
247	Melting temperature and heat of fusion of cytosine revealed from fast scanning calorimetry. Thermochimica Acta, 2017, 657, 47-55.	1.2	46
248	The clinical implications of molecular monitoring and analyses of inherited retinal diseases. Expert Review of Molecular Diagnostics, 2017, 17, 1009-1021.	1.5	7
249	Newborn Screening in the Era of Precision Medicine. Advances in Experimental Medicine and Biology, 2017, 1005, 47-61.	0.8	12
250	Discovery of Variants Underlying Host Susceptibility to Virus Infection Using Whole-Exome Sequencing. Methods in Molecular Biology, 2017, 1656, 209-227.	0.4	0
251	Modular probes for enriching and detecting complex nucleic acid sequences. Nature Chemistry, 2017, 9, 1222-1228.	6.6	32
252	Novel metrics to measure coverage in whole exome sequencing datasets reveal local and global non-uniformity. Scientific Reports, 2017, 7, 885.	1.6	43
254	Human RECQ Helicase Pathogenic Variants, Population Variation and â€‘Missingâ€‘ Diseases. Human Mutation, 2017, 38, 193-203.	1.1	24
255	Relative performance of two DNA extraction and library preparation methods on archaeological human teeth samples. Science and Technology of Archaeological Research, 2017, 3, 80-88.	2.4	6
256	dCATCH-Seq: improved sequencing of large continuous genomic targets with double-hybridization. BMC Genomics, 2017, 18, 811.	1.2	4
257	Big Data Analytics for Genomic Medicine. International Journal of Molecular Sciences, 2017, 18, 412.	1.8	121

#	ARTICLE	IF	CITATIONS
258	Evaluation of Quality Assessment Protocols for High Throughput Genome Resequencing Data. <i>Frontiers in Genetics</i> , 2017, 8, 94.	1.1	10
259	Characterization of background noise in capture-based targeted sequencing data. <i>Genome Biology</i> , 2017, 18, 136.	3.8	50
260	Assessment of Capture and Amplicon-Based Approaches for the Development of a Targeted Next-Generation Sequencing Pipeline to Personalize Lymphoma Management. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 203-214.	1.2	58
261	Dense infraspecific sampling reveals rapid and independent trajectories of plastome degradation in a heterotrophic orchid complex. <i>New Phytologist</i> , 2018, 218, 1192-1204.	3.5	56
262	Clinical pharmacogenomics testing in the era of next generation sequencing: challenges and opportunities for precision medicine. <i>Expert Review of Molecular Diagnostics</i> , 2018, 18, 411-421.	1.5	23
263	Measuring coverage and accuracy of whole-exome sequencing in clinical context. <i>Genetics in Medicine</i> , 2018, 20, 1617-1626.	1.1	50
264	A homozygous variant disrupting the <i>PIGH</i> start-codon is associated with developmental delay, epilepsy, and microcephaly. <i>Human Mutation</i> , 2018, 39, 822-826.	1.1	18
265	Comprehensive analysis of the mutation spectrum in 301 German ALS families. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 817-827.	0.9	80
266	Target capture enrichment of nuclear SNP markers for massively parallel sequencing of degraded and mixed samples. <i>Forensic Science International: Genetics</i> , 2018, 34, 186-196.	1.6	45
267	Metagenomic analysis of microbial community and function involved in cd-contaminated soil. <i>BMC Microbiology</i> , 2018, 18, 11.	1.3	148
268	Targeting legume loci: A comparison of three methods for target enrichment bait design in Leguminosae phylogenomics. <i>Applications in Plant Sciences</i> , 2018, 6, e1036.	0.8	64
269	The molecular complexity of primary ovarian insufficiency aetiology and the use of massively parallel sequencing. <i>Molecular and Cellular Endocrinology</i> , 2018, 460, 170-180.	1.6	27
270	Analysis of functional genomes from metagenomes: Revealing the accelerated electron transfer in microbial fuel cell with rhamnolipid addition. <i>Bioelectrochemistry</i> , 2018, 119, 59-67.	2.4	28
271	Genetic architecture: the shape of the genetic contribution to human traits and disease. <i>Nature Reviews Genetics</i> , 2018, 19, 110-124.	7.7	335
272	Predicting DNA hybridization kinetics from sequence. <i>Nature Chemistry</i> , 2018, 10, 91-98.	6.6	146
273	The Role of Copy Number Variants in Disorders of Sex Development. <i>Sexual Development</i> , 2018, 12, 19-29.	1.1	37
274	New insights into the enhanced performance of high solid anaerobic digestion with dewatered sludge by thermal hydrolysis: Organic matter degradation and methanogenic pathways. <i>Journal of Hazardous Materials</i> , 2018, 342, 1-9.	6.5	115
275	Reliability of Whole-Exome Sequencing for Assessing Intratumor Genetic Heterogeneity. <i>Cell Reports</i> , 2018, 25, 1446-1457.	2.9	76

#	ARTICLE	IF	CITATIONS
276	Local mutational diversity drives intratumoral immune heterogeneity in non-small cell lung cancer. <i>Nature Communications</i> , 2018, 9, 5361.	5.8	294
277	Bioinformatics: Sequences, Structures, Phylogeny. , 2018, , .		0
278	Understanding Genomic Variations in the Context of Health and Disease: Annotation, Interpretation, and Challenges. , 2018, , 71-95.		0
279	Detection of copy number variants and loss of heterozygosity from impure tumor samples using whole exome sequencing data. <i>Oncology Letters</i> , 2018, 16, 4713-4720.	0.8	2
280	Metabolic Interactions of a Chain Elongation Microbiome. <i>Applied and Environmental Microbiology</i> , 2018, 84, .	1.4	93
281	Contributions of rare coding variants in hypotension syndrome genes to population blood pressure variation. <i>Medicine (United States)</i> , 2018, 97, e11865.	0.4	6
282	Expressed exome capture sequencing: A method for cost-effective exome sequencing for all organisms. <i>Molecular Ecology Resources</i> , 2018, 18, 1209-1222.	2.2	28
283	Computational analysis of next generation sequencing data and its applications in clinical oncology. <i>Informatics in Medicine Unlocked</i> , 2018, 11, 75-82.	1.9	36
284	CGT-seq: epigenome-guided de novo assembly of the core genome for divergent populations with large genome. <i>Nucleic Acids Research</i> , 2018, 46, e107-e107.	6.5	6
285	Applications of Probe Capture Enrichment Next Generation Sequencing for Whole Mitochondrial Genome and 426 Nuclear SNPs for Forensically Challenging Samples. <i>Genes</i> , 2018, 9, 49.	1.0	42
286	Simultaneous and stoichiometric purification of hundreds of oligonucleotides. <i>Nature Communications</i> , 2018, 9, 2467.	5.8	22
287	Analytical "bake-off" of whole genome sequencing quality for the Genome Russia project using a small cohort for autoimmune hepatitis. <i>PLoS ONE</i> , 2018, 13, e0200423.	1.1	7
288	Target enrichment sequencing of 307 germplasm accessions identified ancestry of ancient and modern hybrids and signatures of adaptation and selection in sugarcane (<i>Saccharum</i> spp.), a "sweet" crop with "bitter" genomes. <i>Plant Biotechnology Journal</i> , 2019, 17, 488-498.	4.1	33
289	Exome Sequencing Data Analysis. , 2019, , 164-175.		1
290	Detecting and Annotating Rare Variants. , 2019, , 388-399.		4
291	Bioinformatic Methods and Bridging of Assay Results for Reliable Tumor Mutational Burden Assessment in Non-Small-Cell Lung Cancer. <i>Molecular Diagnosis and Therapy</i> , 2019, 23, 507-520.	1.6	39
292	Investigation on polyphosphate accumulation in the sulfur transformation-centric EBPR (SEBPR) process for treatment of high-temperature saline wastewater. <i>Water Research</i> , 2019, 167, 115138.	5.3	8
293	Integrated Proteogenomic Characterization of HBV-Related Hepatocellular Carcinoma. <i>Cell</i> , 2019, 179, 561-577.e22.	13.5	629

#	ARTICLE	IF	CITATIONS
294	Evaluation of the Performance of AmpliSeq and SureSelect Exome Sequencing Libraries for Ion Proton. <i>Frontiers in Genetics</i> , 2019, 10, 856.	1.1	8
296	Use of synthetic DNA spike-in controls (sequins) for human genome sequencing. <i>Nature Protocols</i> , 2019, 14, 2119-2151.	5.5	22
297	PKD1 Duplicated regions limit clinical Utility of Whole Exome Sequencing for Genetic Diagnosis of Autosomal Dominant Polycystic Kidney Disease. <i>Scientific Reports</i> , 2019, 9, 4141.	1.6	44
298	Similarities and differences between variants called with human reference genome HG19 or HG38. <i>BMC Bioinformatics</i> , 2019, 20, 101.	1.2	33
299	Chiral DNA sequences as commutable controls for clinical genomics. <i>Nature Communications</i> , 2019, 10, 1342.	5.8	11
300	Capturing the Resistome: a Targeted Capture Method To Reveal Antibiotic Resistance Determinants in Metagenomes. <i>Antimicrobial Agents and Chemotherapy</i> , 2019, 64, .	1.4	63
302	ForestQC: Quality control on genetic variants from next-generation sequencing data using random forest. <i>PLoS Computational Biology</i> , 2019, 15, e1007556.	1.5	17
303	1. Genomic Technology/ Next-Generation Sequencing. , 2019, , 1-35.		0
304	Preprodynorphin gene mutation causes progressive cardiac conduction disease: A whole-exome analysis of a pedigree. <i>Life Sciences</i> , 2019, 219, 74-81.	2.0	1
305	A roadmap for high-throughput sequencing studies of wild animal populations using noninvasive samples and hybridization capture. <i>Molecular Ecology Resources</i> , 2019, 19, 609-622.	2.2	24
306	Clinical Implementation of Next-Generation Sequencing (NGS) Assays. , 2019, , 113-118.		0
307	C-TALE, a new cost-effective method for targeted enrichment of Hi-C/3C-seq libraries. <i>Methods</i> , 2020, 170, 48-60.	1.9	13
308	Whole-exome sequencing and whole-genome sequencing. , 2020, , 27-39.		0
309	Considerations for whole exome sequencing unique to prenatal care. <i>Human Genetics</i> , 2020, 139, 1149-1159.	1.8	18
310	Population-level inferences from environmental DNA—Current status and future perspectives. <i>Evolutionary Applications</i> , 2020, 13, 245-262.	1.5	105
311	A comparative study of single nucleotide variant detection performance using three massively parallel sequencing methods. <i>PLoS ONE</i> , 2020, 15, e0239850.	1.1	8
312	Recessive MYH3 variants cause “Contractures, pterygia, and variable skeletal fusions syndrome 1B” mimicking Escobar variant multiple pterygium syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2605-2610.	0.7	5
313	Acclimatization of anaerobic sludge with cow manure and realization of high-rate food waste digestion for biogas production. <i>Bioresource Technology</i> , 2020, 315, 123830.	4.8	19

#	ARTICLE	IF	CITATIONS
314	A Pilot Study for the Feasibility of Exome-Sequencing in Circulating Tumor Cells Versus Single Metastatic Biopsies in Breast Cancer. <i>International Journal of Molecular Sciences</i> , 2020, 21, 4826.	1.8	7
315	Evaluation of Whole-Exome Enrichment Solutions: Lessons from the High-End of the Short-Read Sequencing Scale. <i>Journal of Clinical Medicine</i> , 2020, 9, 3656.	1.0	11
316	STROBE-metagenomics: a STROBE extension statement to guide the reporting of metagenomics studies. <i>Lancet Infectious Diseases</i> , The, 2020, 20, e251-e260.	4.6	40
317	Methods for the identification of mitochondrial DNA variants. , 2020, , 243-275.		0
318	Identification of Genomic Alterations in Sporadic Pancreatic Neurogenic Tumors. <i>Pancreas</i> , 2020, 49, 1393-1397.	0.5	1
319	Retrospective evaluation of whole exome and genome mutation calls in 746 cancer samples. <i>Nature Communications</i> , 2020, 11, 4748.	5.8	27
320	Metagenomic Analysis Reveals the Mechanism for the Observed Increase in Antibacterial Activity of Penicillin against Uncultured Bacteria <i>Candidatus Liberibacter asiaticus</i> Relative to Oxytetracycline in <i>Planta</i> . <i>Antibiotics</i> , 2020, 9, 874.	1.5	4
321	Origins and characterization of variants shared between databases of somatic and germline human mutations. <i>BMC Bioinformatics</i> , 2020, 21, 227.	1.2	14
322	New genetic variations discovered in KRAS wild-type cetuximab resistant chinese colorectal cancer patients. <i>Molecular Carcinogenesis</i> , 2020, 59, 478-491.	1.3	10
323	Computational Prediction and Validation of Tumor-Associated Neoantigens. <i>Frontiers in Immunology</i> , 2020, 11, 27.	2.2	86
324	Shedding light on dark genes: enhanced targeted resequencing by optimizing the combination of enrichment technology and DNA fragment length. <i>Scientific Reports</i> , 2020, 10, 9424.	1.6	5
325	Deficiency of Adenosine Deaminase 2 (DADA2): Hidden Variants, Reduced Penetrance, and Unusual Inheritance. <i>Journal of Clinical Immunology</i> , 2020, 40, 917-926.	2.0	32
326	Systematic dissection of biases in whole-exome and whole-genome sequencing reveals major determinants of coding sequence coverage. <i>Scientific Reports</i> , 2020, 10, 2057.	1.6	72
327	Novel Approaches for Identifying the Molecular Background of Schizophrenia. <i>Cells</i> , 2020, 9, 246.	1.8	13
328	Detecting trisomy in products of conception from first-trimester spontaneous miscarriages by next-generation sequencing (NGS). <i>Medicine (United States)</i> , 2020, 99, e18731.	0.4	5
329	RLP/K enrichment sequencing; a novel method to identify receptor-like protein (RLP) and receptor-like kinase (RLK) genes. <i>New Phytologist</i> , 2020, 227, 1264-1276.	3.5	32
331	Association analysis of potentially functional variants within 8p12 with schizophrenia in the Han Chinese population. <i>World Journal of Biological Psychiatry</i> , 2021, 22, 27-33.	1.3	0
332	Next Generation Sequencing Methods: Pushing the Boundaries. , 2021, , 19-46.		0

#	ARTICLE	IF	CITATIONS
333	Comprehensive fundamental somatic variant calling and quality management strategies for human cancer genomes. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	10
334	The current landscape of molecular profiling in the treatment of epithelial ovarian cancer. <i>Gynecologic Oncology</i> , 2021, 160, 333-345.	0.6	40
335	Dealing with Pseudogenes in Molecular Diagnostics in the Next Generation Sequencing Era. <i>Methods in Molecular Biology</i> , 2021, 2324, 363-381.	0.4	1
336	Genomic and transcriptional alterations in first-line chemotherapy exert a potentially unfavorable influence on subsequent immunotherapy in NSCLC. <i>Theranostics</i> , 2021, 11, 7092-7109.	4.6	11
337	New tools for "ZEBRA-FISHING"™. <i>Briefings in Functional Genomics</i> , 2021, , .	1.3	0
341	Performance comparison of four types of target enrichment baits for exome DNA sequencing. <i>Hereditas</i> , 2021, 158, 10.	0.5	16
342	Whole-genome characterization of lung adenocarcinomas lacking alterations in the RTK/RAS/RAF pathway. <i>Cell Reports</i> , 2021, 34, 108707.	2.9	16
343	Performance comparison: exome sequencing as a single test replacing Sanger sequencing. <i>Molecular Genetics and Genomics</i> , 2021, 296, 653-663.	1.0	7
344	Side channel attack on a partially encrypted MPEG-G file. <i>Multimedia Tools and Applications</i> , 2021, 80, 20599-20618.	2.6	0
345	Next-generation sequencing for constitutional variants in the clinical laboratory, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2021, 23, 1399-1415.	1.1	64
346	Estudio molecular del síndrome de plaqueta pegajosa mediante secuenciación de exoma. <i>Revista Facultad De Medicina</i> , 2021, 69, e76806.	0.0	0
347	Application of basalt fibers in a biological contact oxidation reactor for the treatment of landfill leachate. <i>Journal of Cleaner Production</i> , 2021, 297, 126648.	4.6	11
348	Cell-Free Total Nucleic Acid-Based Genotyping of Aggressive Lymphoma: Comprehensive Analysis of Gene Fusions and Nucleotide Variants by Next-Generation Sequencing. <i>Cancers</i> , 2021, 13, 3032.	1.7	5
349	Application of Target Enrichment Sequencing for Population Genetic Analyses of the Obligate Plant Pathogens <i>Pseudoperonospora cubensis</i> and <i>P. humuli</i> in Michigan. <i>Molecular Plant-Microbe Interactions</i> , 2021, 34, 1103-1118.	1.4	3
350	Analytical Performance of NGS-Based Molecular Genetic Tests Used in the Diagnostic Workflow of Pheochromocytoma/Paraganglioma. <i>Cancers</i> , 2021, 13, 4219.	1.7	3
351	Whole-Genome Profiles of Malay Colorectal Cancer Patients with Intact MMR Proteins. <i>Genes</i> , 2021, 12, 1448.	1.0	5
352	Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders. , 2022, , 390-404.		0
353	Hyperthermophilic pretreatment composting to produce high quality sludge compost with superior humification degree and nitrogen retention. <i>Chemical Engineering Journal</i> , 2022, 429, 132247.	6.6	36

#	ARTICLE	IF	CITATIONS
354	Computational cancer genomics. , 2021, , 329-359.		0
355	Genomics in clinical care through precision medicine and personalized treatments. , 2021, , 49-58.		1
356	Review disorders of sex development: The evolving role of genomics in diagnosis and gene discovery. Birth Defects Research Part C: Embryo Today Reviews, 2016, 108, 337-350.	3.6	24
357	Dealing with Pseudogenes in Molecular Diagnostics in the Next-Generation Sequencing Era. Methods in Molecular Biology, 2014, 1167, 303-315.	0.4	29
358	Next-Generation Sequencing Technology in the Genetics of Cardiovascular Disease. Methods in Molecular Biology, 2015, 1299, 51-64.	0.4	1
359	Exon Sequencing of G Protein-Coupled Receptor Genes and Perspectives for Disease Treatment. Methods in Pharmacology and Toxicology, 2014, , 313-332.	0.1	5
360	Cancer Genomics in Precision Oncology: Applications, Challenges, and Prospects. , 2020, , 453-499.		9
370	Whole exome HBV DNA integration is independent of the intrahepatic HBV reservoir in HBeAg-negative chronic hepatitis B. Gut, 2021, 70, 2337-2348.	6.1	36
372	High-Resolution Mutational Profiling Suggests the Genetic Validity of Glioblastoma Patient-Derived Pre-Clinical Models. PLoS ONE, 2013, 8, e56185.	1.1	25
373	Identification of Chromosomal Errors in Human Preimplantation Embryos with Oligonucleotide DNA Microarray. PLoS ONE, 2013, 8, e61838.	1.1	21
374	Identification and Characterization of Cancer Mutations in Japanese Lung Adenocarcinoma without Sequencing of Normal Tissue Counterparts. PLoS ONE, 2013, 8, e73484.	1.1	41
375	Whole-Exome Sequencing to Identify a Novel LMNA Gene Mutation Associated with Inherited Cardiac Conduction Disease. PLoS ONE, 2013, 8, e83322.	1.1	12
376	Comprehensive Analysis to Improve the Validation Rate for Single Nucleotide Variants Detected by Next-Generation Sequencing. PLoS ONE, 2014, 9, e86664.	1.1	20
377	OTG-snpcaller: An Optimized Pipeline Based on TMAP and GATK for SNP Calling from Ion Torrent Data. PLoS ONE, 2014, 9, e97507.	1.1	32
378	Genome at Juncture of Early Human Migration: A Systematic Analysis of Two Whole Genomes and Thirteen Exomes from Kuwaiti Population Subgroup of Inferred Saudi Arabian Tribe Ancestry. PLoS ONE, 2014, 9, e99069.	1.1	41
379	Targeted Sequencing of Large Genomic Regions with CATCH-Seq. PLoS ONE, 2014, 9, e111756.	1.1	16
380	Profiling of Exome Mutations Associated with Progression of HBV-Related Hepatocellular Carcinoma. PLoS ONE, 2014, 9, e115152.	1.1	16
381	The Use of Non-Variant Sites to Improve the Clinical Assessment of Whole-Genome Sequence Data. PLoS ONE, 2015, 10, e0132180.	1.1	15

#	ARTICLE	IF	CITATIONS
382	Impact of Pre-Analytical Variables on Cancer Targeted Gene Sequencing Efficiency. PLoS ONE, 2015, 10, e0143092.	1.1	13
383	High-Quality Exome Sequencing of Whole-Genome Amplified Neonatal Dried Blood Spot DNA. PLoS ONE, 2016, 11, e0153253.	1.1	38
384	Exome sequencing covers >98% of mutations identified on targeted next generation sequencing panels. PLoS ONE, 2017, 12, e0170843.	1.1	94
385	MTTE: an innovative strategy for the evaluation of targeted/exome enrichment efficiency. Oncotarget, 2016, 7, 67266-67276.	0.8	4
386	Elucidating the cancer-specific genetic alteration spectrum of glioblastoma derived cell lines from whole exome and RNA sequencing. Oncotarget, 2015, 6, 43452-43471.	0.8	62
387	Reliability of Whole-Exome Sequencing for Assessing Intratumor Genetic Heterogeneity. SSRN Electronic Journal, 0, , .	0.4	2
388	Genetic tests by next-generation sequencing in children with developmental delay and/or intellectual disability. Clinical and Experimental Pediatrics, 2020, 63, 195-202.	0.9	29
389	An improved understanding of cancer genomics through massively parallel sequencing. Translational Cancer Research, 2014, 3, 243-259.	0.4	10
390	Personal Diagnostics Using DNA-Sequencing. Advances in Medical Technologies and Clinical Practice Book Series, 0, , 202-217.	0.3	1
391	Systematic Identification and Definition of Consistently Well-Characterized Protein-Coding Exons Using Next Generation Sequencing Technology. Journal of Genomes and Exomes, 0, 2, 1-18.	0.0	1
392	Structured Genome-Scale Variant and Clinical Data Reporting for Meta-Analysis in an Era of Genomic Medicine. Journal of Genomes and Exomes, 0, 2, 31-42.	0.0	2
393	Application of Whole Exome Sequencing to Identify Disease-Causing Variants in Inherited Human Diseases. Genomics and Informatics, 2012, 10, 214.	0.4	56
394	Homologous Recombination Deficiency Assays in Epithelial Ovarian Cancer: Current Status and Future Direction. Frontiers in Oncology, 2021, 11, 675972.	1.3	21
395	A Survey of Next-Generation Sequencing Technologies. SpringerBriefs in Genetics, 2013, , 13-24.	0.1	0
396	VARIANT PRIORIZATION AND ANALYSIS INCORPORATING PROBLEMATIC REGIONS OF THE GENOME. , 2013, , .		0
397	Maize (Zea Mays L.) Genome Diversity as Revealed by RNA-Sequencing. , 2014, , 299-325.		0
398	Current Massively Parallel Sequencing Technologies: Platforms and Reporting Considerations. , 2015, , 3-17.		0
399	Molecular Diagnostics and Tumor Mutational Analysis. Cancer Drug Discovery and Development, 2015, , 47-65.	0.2	2

#	ARTICLE	IF	CITATIONS
400	Analysis of AmpliSeq RNA-Sequencing Enrichment Panels. Lecture Notes in Computer Science, 2015, , 495-500.	1.0	0
401	Next-Generation Sequencing Applications in Head and Neck Oncology. , 2015, , 401-422.		0
402	Novel Trends in the Molecular Genetics of Hearing Loss. Journal of Hearing Science, 2015, 5, 9-15.	0.1	2
404	REPURPOSING GERMLINE EXOMES OF THE CANCER GENOME ATLAS DEMANDS A CAUTIOUS APPROACH AND SAMPLE-SPECIFIC VARIANT FILTERING. , 2016, , .		10
406	Application of Next-Generation Sequencing to Hearing Loss. , 2017, , 71-87.		0
408	Existing and Emerging Molecular Technologies in Myeloid Neoplasms. Molecular Pathology Library, 2018, , 369-412.	0.1	0
415	Rhizosphere effect alters the soil microbiome composition and C, N transformation in an arid ecosystem. Applied Soil Ecology, 2022, 170, 104296.	2.1	25
416	Genomic Tools Used in Molecular Clinical Aging Research. , 2020, , 87-109.		0
417	Methods for Target Enrichment Sequencing via Probe Capture in Legumes. Methods in Molecular Biology, 2020, 2107, 199-231.	0.4	2
421	Automation of molecular-based analyses: a primer on massively parallel sequencing. Clinical Biochemist Reviews, 2014, 35, 169-76.	3.3	6
422	REPURPOSING GERMLINE EXOMES OF THE CANCER GENOME ATLAS DEMANDS A CAUTIOUS APPROACH AND SAMPLE-SPECIFIC VARIANT FILTERING. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2016, 21, 207-18.	0.7	13
423	Validation of the EuroClonality-NGS DNA capture panel as an integrated genomic tool for lymphoproliferative disorders. Blood Advances, 2021, 5, 3188-3198.	2.5	2
425	Detecting Liquid Remnants of Solid Tumors: Circulating Tumor DNA Minimal Residual Disease. Cancer Discovery, 2021, 11, 2968-2986.	7.7	116
427	System analysis of the sequencing quality of human whole exome samples on BGI NGS platform. Scientific Reports, 2022, 12, 609.	1.6	8
430	Insights into National Laboratory Newborn Screening and Future Prospects. Medicina (Lithuania), 2022, 58, 272.	0.8	3
431	Artificial Intelligence and Cardiovascular Genetics. Life, 2022, 12, 279.	1.1	13
432	Fishing for DNA? Designing baits for population genetics in target enrichment experiments: Guidelines, considerations and the new tool superBaits. Molecular Ecology Resources, 2022, 22, 2105-2119.	2.2	5
433	Identification of missing persons through kinship analysis by microhaplotype sequencing of single-source DNA and two-person DNA mixtures. Forensic Science International: Genetics, 2022, 58, 102689.	1.6	8

#	ARTICLE	IF	CITATIONS
434	Unlocking the Complexity of Mitochondrial DNA: A Key to Understanding Neurodegenerative Disease Caused by Injury. <i>Cells</i> , 2021, 10, 3460.	1.8	5
435	Targeted Genotyping of a Whole-Gene Repertoire by an Ultrahigh-Multiplex and Flexible HD-Marker Approach. <i>Engineering</i> , 2021, , .	3.2	0
437	A rare genetic variant in the cleavage site of prepro-orexin is associated with idiopathic hypersomnia. <i>Npj Genomic Medicine</i> , 2022, 7, 29.	1.7	5
438	Genome-wide investigations reveal the population structure and selection signatures of Nigerian cattle adaptation in the sub-Saharan tropics. <i>BMC Genomics</i> , 2022, 23, 306.	1.2	4
444	Performance characterization of PCR-free whole genome sequencing for clinical diagnosis. <i>Medicine (United States)</i> , 2022, 101, e28972.	0.4	5
445	Application of modern approaches in the screening and early diagnosis programs for the orphan diseases. <i>Zhurnal Nevrologii I Psikiatrii Imeni S S Korsakova</i> , 2022, 122, 30.	0.1	1
447	Integrated proteogenomic characterization of medullary thyroid carcinoma. <i>Cell Discovery</i> , 2022, 8, .	3.1	18
448	Machine Learning Aided Interpretable Approach for Single Nucleotide-Based DNA Sequencing using a Model Nanopore. <i>Journal of Physical Chemistry Letters</i> , 2022, 13, 11818-11830.	2.1	5
449	High efficacy of azacitidine combined with homoharringtonine, idarubicin, and cytarabine in newly diagnosed patients with AML: A single arm, phase 2 trial. <i>Frontiers in Oncology</i> , 0, 12, .	1.3	1
450	Rare variant analyses in large-scale cohorts identified SLC13A1 associated with chronic pain. <i>Pain</i> , 2023, 164, 1841-1851.	2.0	3
456	DNA Fragment Enrichment for High-Throughput Sequencing. <i>Molecular Biology</i> , 2023, 57, 424-439.	0.4	1