Donna M Muzny

List of Publications by Year
in descending order

391	103,188	831 121	267 306
papers	citations	h-index	g-index

406	406	406	122883
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	13.7	21,074
2	Initial sequencing and comparative analysis of the mouse genome. Nature, 2002, 420, 520-562.	13.7	6,319
3	The Genome Sequence of Drosophila melanogaster. Science, 2000, 287, 2185-2195.	6.0	5,566
4	Somatic mutations affect key pathways in lung adenocarcinoma. Nature, 2008, 455, 1069-1075.	13.7	2,694
5	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	13.7	1,994
6	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	13.7	1,943
7	Pancreatic cancer genomes reveal aberrations in axon guidance pathway genes. Nature, 2012, 491, 399-405.	13.7	1,741
8	Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine, 2013, 369, 1502-1511.	13.9	1,717
9	The complete genome of an individual by massively parallel DNA sequencing. Nature, 2008, 452, 872-876.	13.7	1,635
10	Generation and initial analysis of more than 15,000 full-length human and mouse cDNA sequences. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 16899-16903.	3.3	1,610
11	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	13.7	1,597
12	Exome Sequencing of Head and Neck Squamous Cell Carcinoma Reveals Inactivating Mutations in NOTCH1. Science, 2011, 333, 1154-1157.	6.0	1,568
13	Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. Science, 2007, 316, 222-234.	6.0	1,283
14	The genome of the model beetle and pest Tribolium castaneum. Nature, 2008, 452, 949-955.	13.7	1,255
15	Temporal development of the gut microbiome in early childhood from the TEDDY study. Nature, 2018, 562, 583-588.	13.7	1,220
16	The genome of the social amoeba Dictyostelium discoideum. Nature, 2005, 435, 43-57.	13.7	1,179
17	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
18	The Genome Sequence of Taurine Cattle: A Window to Ruminant Biology and Evolution. Science, 2009, 324, 522-528.	6.0	1,038

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19	The Genome of the Sea Urchin Strongylocentrotus purpuratus. Science, 2006, 314, 941-952.	6.0	1,018
20	A high-resolution map of human evolutionary constraint using 29 mammals. Nature, 2011, 478, 476-482.	13.7	1,016
21	The DNA sequence of the human X chromosome. Nature, 2005, 434, 325-337.	13.7	985
22	Mind the Gap: Upgrading Genomes with Pacific Biosciences RS Long-Read Sequencing Technology. PLoS ONE, 2012, 7, e47768.	1.1	896
23	Functional and Evolutionary Insights from the Genomes of Three Parasitoid Nasonia Species. Science, 2010, 327, 343-348.	6.0	808
24	Genome-Wide Survey of SNP Variation Uncovers the Genetic Structure of Cattle Breeds. Science, 2009, 324, 528-532.	6.0	746
25	Whole-Genome Sequencing in a Patient with Charcotâ€“Marieâ€“Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191.	13.9	698
26	The Somatic Genomic Landscape of Chromophobe Renal Cell Carcinoma. Cancer Cell, 2014, 26, 319-330.	7.7	665
27	Trans-ancestry mutational landscape of hepatocellular carcinoma genomes. Nature Genetics, 2014, 46, 1267-1273.	9.4	655
28	A Catalog of Reference Genomes from the Human Microbiome. Science, 2010, 328, 994-999.	6.0	621
29	Direct selection of human genomic loci by microarray hybridization. Nature Methods, 2007, 4, 903-905.	9.0	617
30	The gut mycobiome of the Human Microbiome Project healthy cohort. Microbiome, 2017, 5, 153.	4.9	609
31	Demographic history and rare allele sharing among human populations. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11983-11988.	3.3	589
32	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
33	Natural variation in genome architecture among 205 Drosophila melanogaster Genetic Reference Panel lines. Genome Research, 2014, 24, 1193-1208.	2.4	565
34	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	13.9	565
35	Mammalian Y chromosomes retain widely expressed dosage-sensitive regulators. Nature, 2014, 508, 494-499.	13.7	546
36	Comparative and demographic analysis of orang-utan genomes. Nature, 2011, 469, 529-533.	13.7	541

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37	Characterization of a murine gene expressed from the inactive X chromosome. Nature, 1991, 351, 325-329.	13.7	527
38	Mutational Landscape of Aggressive Cutaneous Squamous Cell Carcinoma. Clinical Cancer Research, 2014, 20, 6582-6592.	3.2	493
39	The Status, Quality, and Expansion of the NIH Full-Length cDNA Project: The Mammalian Gene Collection (MGC). Genome Research, 2004, 14, 2121-2127.	2.4	486
40	Integrative Genomic Characterization of Oral Squamous Cell Carcinoma Identifies Frequent Somatic Drivers. Cancer Discovery, 2013, 3, 770-781.	7.7	484
41	Comprehensive Pan-Genomic Characterization of Adrenocortical Carcinoma. Cancer Cell, 2016, 29, 723-736.	7.7	482
42	Mutations in Smooth Muscle Alpha-Actin (ACTA2) Cause Coronary Artery Disease, Stroke, and Moyamoya Disease, Along with Thoracic Aortic Disease. American Journal of Human Genetics, 2009, 84, 617-627.	2.6	466
43	Comparative genome sequencing of Drosophila pseudoobscura: Chromosomal, gene, and cis-element evolution. Genome Research, 2005, 15, 1-18.	2.4	453
44	Two independent mutational events in the loss of urate oxidase during hominoid evolution. Journal of Molecular Evolution, 1992, 34, 78-84.	0.8	440
45	The sheep genome illuminates biology of the rumen and lipid metabolism. Science, 2014, 344, 1168-1173.	6.0	436
46	Complete Khoisan and Bantu genomes from southern Africa. Nature, 2010, 463, 943-947.	13.7	400
47	Convergent evolution of the genomes of marine mammals. Nature Genetics, 2015, 47, 272-275.	9.4	392
48	Inactivating Mutations in NPC1L1 and Protection from Coronary Heart Disease. New England Journal of Medicine, 2014, 371, 2072-2082.	13.9	386
49	Genome Project Standards in a New Era of Sequencing. Science, 2009, 326, 236-237.	6.0	382
50	Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors. JAMA Oncology, 2016, 2, 616.	3.4	378
51	Finding the missing honey bee genes: lessons learned from a genome upgrade. BMC Genomics, 2014, 15, 86.	1.2	375
52	Epistasis dominates the genetic architecture of Drosophila quantitative traits. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 15553-15559.	3.3	348
53	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
54	Integrative Annotation of Variants from 1092 Humans: Application to Cancer Genomics. Science, 2013, 342, 1235587.	6.0	341

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55	Generation of cDNA probes directed by amino acid sequence: cloning of urate oxidase. Science, 1988, 239, 1288-1291.	6.0	336
56	The Complete Genome Sequence ofEscherichia coliDH10B: Insights into the Biology of a Laboratory Workhorse. Journal of Bacteriology, 2008, 190, 2597-2606.	1.0	331
57	The genomes of two key bumblebee species with primitive eusocial organization. Genome Biology, 2015, 16, 76.	3.8	330
58	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	9.4	326
59	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	13.5	322
60	Gibbon genome and the fast karyotype evolution of small apes. Nature, 2014, 513, 195-201.	13.7	320
61	A recombination hotspot responsible for two inherited peripheral neuropathies is located near a mariner transposon-like element. Nature Genetics, 1996, 12, 288-297.	9.4	304
62	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660.	9.4	302
63	Multilevel Genomics-Based Taxonomy of Renal Cell Carcinoma. Cell Reports, 2016, 14, 2476-2489.	2.9	298
64	The house spider genome reveals an ancient whole-genome duplication during arachnid evolution. BMC Biology, 2017, 15, 62.	1.7	286
65	Structure and function of the healthy pre-adolescent pediatric gut microbiome. Microbiome, 2015, 3, 36.	4.9	283
66	Rise and fall of subclones from diagnosis to relapse in pediatric B-acute lymphoblastic leukaemia. Nature Communications, 2015, 6, 6604.	5.8	281
67	Genomic profiling of SÃ©zary syndrome identifies alterations of key T cell signaling and differentiation genes. Nature Genetics, 2015, 47, 1426-1434.	9.4	276
68	Exome Sequencing of Ion Channel Genes Reveals Complex Profiles Confounding Personal Risk Assessment in Epilepsy. Cell, 2011, 145, 1036-1048.	13.5	274
69	Whole-Genome Sequencing for Optimized Patient Management. Science Translational Medicine, 2011, 03, 87re3.	5.8	272
70	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493.	3.8	265
71	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	1.5	261
72	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	3.8	258

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73	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	9.4	255
74	Activation of Multiple Proto-oncogenic Tyrosine Kinases in Breast Cancer via Loss of the PTPN12 Phosphatase. Cell, 2011, 144, 703-718.	13.5	246
75	Strict evolutionary conservation followed rapid gene loss on human and rhesus Y chromosomes. Nature, 2012, 483, 82-86.	13.7	245
76	Exonuclease mutations in DNA polymerase epsilon reveal replication strand specific mutation patterns and human origins of replication. Genome Research, 2014, 24, 1740-1750.	2.4	244
77	Genome of the Asian longhorned beetle (Anoplophora glabripennis), a globally significant invasive species, reveals key functional and evolutionary innovations at the beetleâ€“plant interface. Genome Biology, 2016, 17, 227.	3.8	244
78	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	3.8	242
79	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	4.5	234
80	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	5.8	227
81	Complete Genome Sequence of Rickettsia typhi and Comparison with Sequences of Other Rickettsiae. Journal of Bacteriology, 2004, 186, 5842-5855.	1.0	223
82	Hemimetabolous genomes reveal molecular basis of termite eusociality. Nature Ecology and Evolution, 2018, 2, 557-566.	3.4	223
83	The First Myriapod Genome Sequence Reveals Conservative Arthropod Gene Content and Genome Organisation in the Centipede Strigamia maritima. PLoS Biology, 2014, 12, e1002005.	2.6	221
84	Hemichordate genomes and deuterostome origins. Nature, 2015, 527, 459-465.	13.7	217
85	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	2.9	211
86	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
87	Copy number variation detection in whole-genome sequencing data using the Bayesian information criterion. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, E1128-36.	3.3	200
88	Launching genomics into the cloud: deployment of Mercury, a next generation sequence analysis pipeline. BMC Bioinformatics, 2014, 15, 30.	1.2	199
89	High-depth African genomes inform human migration and health. Nature, 2020, 586, 741-748.	13.7	197
90	Mapping and characterization of structural variation in 17,795 human genomes. Nature, 2020, 583, 83-89.	13.7	194

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91	Targeted enrichment beyond the consensus coding DNA sequence exome reveals exons with higher variant densities. Genome Biology, 2011, 12, R68.	13.9	192
92	Whole exome sequencing study identifies novel rare and common Alzheimerâ€™s-Associated variants involved in immune response and transcriptional regulation. Molecular Psychiatry, 2020, 25, 1859-1875.	4.1	191
93	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	13.5	189
94	Expression of the murine Duchenne muscular dystrophy gene in muscle and brain. Science, 1988, 239, 1416-1418.	6.0	188
95	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	1.1	186
96	Analyses of deep mammalian sequence alignments and constraint predictions for 1% of the human genome. Genome Research, 2007, 17, 760-774.	2.4	184
97	Unique features of a global human ectoparasite identified through sequencing of the bed bug genome. Nature Communications, 2016, 7, 10165.	5.8	184
98	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
99	Novel somatic and germline mutations in intracranial germ cell tumours. Nature, 2014, 511, 241-245.	13.7	181
100	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	2.6	181
101	Sequence and analysis of chromosome 2 of Dictyostelium discoideum. Nature, 2002, 418, 79-85.	13.7	176
102	Combined sequence-based and genetic mapping analysis of complex traits in outbred rats. Nature Genetics, 2013, 45, 767-775.	9.4	176
103	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. Nature Communications, 2015, 6, 5897.	5.8	173
104	Germline Mutations in Shelterin Complex Genes Are Associated With Familial Glioma. Journal of the National Cancer Institute, 2015, 107, 384.	3.0	172
105	Pulmonary alveolar proteinosis caused by deletion of the GM-CSFRÎ± gene in the X chromosome pseudoautosomal region 1. Journal of Experimental Medicine, 2008, 205, 2711-2716.	4.2	171
106	A Massive Expansion of Effector Genes Underlies Gall-Formation in the Wheat Pest Mayetiola destructor. Current Biology, 2015, 25, 613-620.	1.8	171
107	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	1.7	163
108	Demographic Histories and Patterns of Linkage Disequilibrium in Chinese and Indian Rhesus Macaques. Science, 2007, 316, 240-243.	6.0	161

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109	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
110	Next-generation sequencing identifies rare variants associated with Noonan syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 11473-11478.	3.3	158
111	Evolutionary History of Chemosensory-Related Gene Families across the Arthropoda. Molecular Biology and Evolution, 2017, 34, 1838-1862.	3.5	157
112	Multifaceted biological insights from a draft genome sequence of the tobacco hornworm moth, Manduca sexta. Insect Biochemistry and Molecular Biology, 2016, 76, 118-147.	1.2	154
113	Assessing structural variation in a personal genomeâ€"towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	1.2	153
114	Gene content evolution in the arthropods. Genome Biology, 2020, 21, 15.	3.8	150
115	Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. Human Molecular Genetics, 2011, 20, 3366-3375.	1.4	149
116	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	2.6	148
117	Comparative validation of the D. melanogaster modENCODE transcriptome annotation. Genome Research, 2014, 24, 1209-1223.	2.4	147
118	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	2.6	146
119	Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. Genome Medicine, 2013, 5, 57.	3.6	143
120	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	2.6	142
121	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	2.6	137
122	Transmission event of SARS-CoV-2 delta variant reveals multiple vaccine breakthrough infections. BMC Medicine, 2021, 19, 255.	2.3	137
123	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651.	3.9	136
124	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. PLoS Genetics, 2013, 9, e1003443.	1.5	133
125	Genomic Profiling of Pediatric Acute Myeloid Leukemia Reveals a Changing Mutational Landscape from Disease Diagnosis to Relapse. Cancer Research, 2016, 76, 2197-2205.	0.4	133
126	Whole-genome sequenceâ€"based analysis of high-density lipoprotein cholesterol. Nature Genetics, 2013, 45, 899-901.	9.4	132

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127	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	4.2	132
128	Large-Scale Comparative Sequence Analysis of the Human and Murine Brutonâ€™s Tyrosine Kinase Loci Reveals Conserved Regulatory Domains. Genome Research, 1997, 7, 315-329.	2.4	131
129	The whole genome sequence of the Mediterranean fruit fly, Ceratitis capitata (Wiedemann), reveals insights into the biology and adaptive evolution of a highly invasive pest species. Genome Biology, 2016, 17, 192.	3.8	130
130	De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. Genome Medicine, 2013, 5, 11.	3.6	128
131	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	2.6	128
132	Sheep genome functional annotation reveals proximal regulatory elements contributed to the evolution of modern breeds. Nature Communications, 2018, 9, 859.	5.8	126
133	The completion of the Mammalian Gene Collection (MGC). Genome Research, 2009, 19, 2324-2333.	2.4	125
134	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	2.6	125
135	Comparative Genomics of Gardnerella vaginalis Strains Reveals Substantial Differences in Metabolic and Virulence Potential. PLoS ONE, 2010, 5, e12411.	1.1	124
136	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. PLoS Genetics, 2014, 10, e1004258.	1.5	122
137	BCORâ€“CCNB3 fusions are frequent in undifferentiated sarcomas of male children. Modern Pathology, 2015, 28, 575-586.	2.9	122
138	Molecular profiling predicts meningioma recurrence and reveals loss of DREAM complex repression in aggressive tumors. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 21715-21726.	3.3	122
139	Paradoxical DNA Repair and Peroxide Resistance Gene Conservation in Bacillus pumilus SAFR-032. PLoS ONE, 2007, 2, e928.	1.1	118
140	The comparative genomics and complex population history of Papio baboons. Science Advances, 2019, 5, eaau6947.	4.7	115
141	Molecular evolutionary trends and feeding ecology diversification in the Hemiptera, anchored by the milkweed bug genome. Genome Biology, 2019, 20, 64.	3.8	114
142	A sequence-level map of chromosomal breakpoints in the MCF-7 breast cancer cell line yields insights into the evolution of a cancer genome. Genome Research, 2009, 19, 167-177.	2.4	111
143	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	2.6	110
144	Ampullary Cancers Harbor ELF3 Tumor Suppressor Gene Mutations and Exhibit Frequent WNT Dysregulation. Cell Reports, 2016, 14, 907-919.	2.9	107

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145	Whole Genome Sequences of Three Treponema pallidum ssp. pertenue Strains: Yaws and Syphilis Treponemes Differ in Less than 0.2% of the Genome Sequence. PLoS Neglected Tropical Diseases, 2012, 6, e1471.	1.3	106
146	Neutral genomic regions refine models of recent rapid human population growth. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 757-762.	3.3	106
147	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	3.6	105
148	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	1.1	104
149	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	15.2	103
150	Chemistry-First Approach for Nomination of Personalized Treatment in Lung Cancer. Cell, 2018, 173, 864-878.e29.	13.5	102
151	The population genomics of rhesus macaques (Macaca mulatta) based on whole-genome sequences. Genome Research, 2016, 26, 1651-1662.	2.4	101
152	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605.	2.6	99
153	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	6.5	98
154	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	2.6	98
155	Single nucleotide polymorphism-mediated translational suppression of endoplasmic reticulum mannosidase I modifies the onset of end-stage liver disease in alpha1-antitrypsin deficiency. Hepatology, 2009, 50, 275-281.	3.6	96
156	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	2.6	96
157	The Chinese hamster HPRT gene: Restriction map, sequence analysis, and multiplex PCR deletion screen. Genomics, 1991, 9, 247-256.	1.3	95
158	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	3.8	95
159	The Genome Sequence of Mannheimia haemolytica A1: Insights into Virulence, Natural Competence, and Pasteurellaceae Phylogeny. Journal of Bacteriology, 2006, 188, 7257-7266.	1.0	94
160	Reproductive Longevity Predicts Mutation Rates in Primates. Current Biology, 2018, 28, 3193-3197.e5.	1.8	94
161	Genetics of schizophrenia in the South African Xhosa. Science, 2020, 367, 569-573.	6.0	93
162	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	2.6	92

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163	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	1.1	92
164	DVL3 Alleles Resulting in a â˜’1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2016, 98, 553-561.	2.6	88
165	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
166	Molecular and phenotypic variation in patients with severe Hunter syndrome. Human Molecular Genetics, 1997, 6, 479-486.	1.4	82
167	Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016, 126, 762-778.	3.9	82
168	Sooty mangabey genome sequence provides insight into AIDS resistance in a natural SIV host. Nature, 2018, 553, 77-81.	13.7	81
169	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	2.6	80
170	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. American Journal of Human Genetics, 2014, 94, 915-923.	2.6	79
171	The Toxicogenome of Hyalella azteca: A Model for Sediment Ecotoxicology and Evolutionary Toxicology. Environmental Science & Technology, 2018, 52, 6009-6022.	4.6	79
172	Compound Heterozygous CORO1A Mutations in Siblings with a Mucocutaneous-Immunodeficiency Syndrome of Epidermodysplasia Verruciformis-HPV, Molluscum Contagiosum and Granulomatous Tuberculoid Leprosy. Journal of Clinical Immunology, 2014, 34, 871-890.	2.0	78
173	POGZ truncating alleles cause syndromic intellectual disability. Genome Medicine, 2016, 8, 3.	3.6	78
174	A Collagen-Binding Adhesin, Acb, and Ten Other Putative MSCRAMM and Pilus Family Proteins of Streptococcus gallolyticus subsp. gallolyticus (Streptococcus bovis Group,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 2	1.0	78
175	Deep Sequencing of Systematic Combinatorial Libraries Reveals Î²-Lactamase Sequence Constraints at High Resolution. Journal of Molecular Biology, 2012, 424, 150-167.	2.0	76
176	Biallelic mutations in IRF8 impair human NK cell maturation and function. Journal of Clinical Investigation, 2016, 127, 306-320.	3.9	76
177	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genetics in Medicine, 2017, 19, 13-19.	1.1	74
178	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150.	2.6	74
179	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. Genetics in Medicine, 2017, 19, 412-420.	1.1	73
180	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. PLoS Biology, 2020, 18, e3000954.	2.6	73

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181	Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68.	4.5	71
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