Donna M Muzny

List of Publications by Citations

385 papers	81,575 citations	111 h-index	284 g-index
406 ext. papers	95,063 ext. citations	14.6 avg, IF	6.34 L-index

#	Paper	IF	Citations
385	Initial sequencing and analysis of the human genome. *Nature*, **2001**, 409, 860-921	50.4	17366
384	Initial sequencing and comparative analysis of the mouse genome. *Nature*, **2002**, 420, 520-62	50.4	5376
383	The genome sequence of Drosophila melanogaster. *Science*, **2000**, 287, 2185-95	33.3	4857
382	Somatic mutations affect key pathways in lung adenocarcinoma. *Nature*, **2008**, 455, 1069-75	50.4	2280
381	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. *Nature*, **2004**, 428, 493-521	50.4	1689
380	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, 16899-903	11.5	1457
379	Pancreatic cancer genomes reveal aberrations in axon guidance pathway genes. *Nature*, **2012**, 491, 399-405	50.4	1427
378	The complete genome of an individual by massively parallel DNA sequencing. *Nature*, **2008**, 452, 872-6	50.4	1424
377	Clinical whole-exome sequencing for the diagnosis of mendelian disorders. *New England Journal of Medicine*, **2013**, 369, 1502-11	59.2	1393
376	An integrated map of structural variation in 2,504 human genomes. *Nature*, **2015**, 526, 75-81	50.4	1368
375	Exome sequencing of head and neck squamous cell carcinoma reveals inactivating mutations in NOTCH1. *Science*, **2011**, 333, 1154-7	33.3	1331
374	Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature*, **2012**, 485, 242-5	50.4	1300
373	Evolutionary and biomedical insights from the rhesus macaque genome. *Science*, **2007**, 316, 222-34	33.3	1072
372	The genome of the model beetle and pest Tribolium castaneum. *Nature*, **2008**, 452, 949-55	50.4	1043
371	The genome of the social amoeba Dictyostelium discoideum. *Nature*, **2005**, 435, 43-57	50.4	1042
370	Molecular findings among patients referred for clinical whole-exome sequencing. *JAMA - Journal of the American Medical Association*, **2014**, 312, 1870-9	27.4	915
369	The genome of the sea urchin Strongylocentrotus purpuratus. *Science*, **2006**, 314, 941-52	33.3	886

368	The genome sequence of taurine cattle: a window to ruminant biology and evolution. *Science*, **2009**, 324, 522-8	33.3	863
367	The DNA sequence of the human X chromosome. *Nature*, **2005**, 434, 325-37	50.4	822
366	A high-resolution map of human evolutionary constraint using 29 mammals. *Nature*, **2011**, 478, 476-82	50.4	802
365	Functional and evolutionary insights from the genomes of three parasitoid Nasonia species. *Science* , **2010**, 327, 343-8	33.3	682
364	Mind the gap: upgrading genomes with Pacific Biosciences RS long-read sequencing technology. *PLoS ONE*, **2012**, 7, e47768	3.7	665
363	Temporal development of the gut microbiome in early childhood from the TEDDY study. *Nature*, **2018**, 562, 583-588	50.4	619
362	Whole-genome sequencing in a patient with Charcot-Marie-Tooth neuropathy. *New England Journal of Medicine*, **2010**, 362, 1181-91	59.2	613
361	Genome-wide survey of SNP variation uncovers the genetic structure of cattle breeds. *Science*, **2009**, 324, 528-32	33.3	612
360	Direct selection of human genomic loci by microarray hybridization. *Nature Methods*, **2007**, 4, 903-5	21.6	543
359	The somatic genomic landscape of chromophobe renal cell carcinoma. *Cancer Cell*, **2014**, 26, 319-330	24.3	521
358	A catalog of reference genomes from the human microbiome. *Science*, **2010**, 328, 994-9	33.3	508
357	Trans-ancestry mutational landscape of hepatocellular carcinoma genomes. *Nature Genetics*, **2014**, 46, 1267-73	36.3	491
356	Characterization of a murine gene expressed from the inactive X chromosome. *Nature*, **1991**, 351, 325-9	50.4	471
355	Demographic history and rare allele sharing among human populations. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 11983-8	11.5	455
354	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics*, **2015**, 97, 199-215	11	432
353	Comparative and demographic analysis of orang-utan genomes. *Nature*, **2011**, 469, 529-33	50.4	431
352	Comparative genome sequencing of Drosophila pseudoobscura: chromosomal, gene, and cis-element evolution. *Genome Research*, **2005**, 15, 1-18	9.7	410
351	Mammalian Y chromosomes retain widely expressed dosage-sensitive regulators. *Nature*, **2014**, 508, 494-9	50.4	406

350	The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Collection (MGC). *Genome Research*, **2004**, 14, 2121-7	9.7	404
349	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. *New England Journal of Medicine*, **2017**, 376, 21-31	59.2	391
348	Integrative genomic characterization of oral squamous cell carcinoma identifies frequent somatic drivers. *Cancer Discovery*, **2013**, 3, 770-81	24.4	391
347	Natural variation in genome architecture among 205 Drosophila melanogaster Genetic Reference Panel lines. *Genome Research*, **2014**, 24, 1193-208	9.7	372
346	Two independent mutational events in the loss of urate oxidase during hominoid evolution. *Journal of Molecular Evolution*, **1992**, 34, 78-84	3.1	367
345	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, 617-27	11	364
344	Mutational landscape of aggressive cutaneous squamous cell carcinoma. *Clinical Cancer Research*, **2014**, 20, 6582-92	12.9	362
343	Complete Khoisan and Bantu genomes from southern Africa. *Nature*, **2010**, 463, 943-7	50.4	342
342	Genomics. Genome project standards in a new era of sequencing. *Science*, **2009**, 326, 236-7	33.3	326
341	The gut mycobiome of the Human Microbiome Project healthy cohort. *Microbiome*, **2017**, 5, 153	16.6	324
340	Comprehensive Pan-Genomic Characterization of Adrenocortical Carcinoma. *Cancer Cell*, **2016**, 29, 723-736	24.3	324
339	Generation of cDNA probes directed by amino acid sequence: cloning of urate oxidase. *Science*, **1988**, 239, 1288-91	33.3	309
338	Inactivating mutations in NPC1L1 and protection from coronary heart disease. *New England Journal of Medicine*, **2014**, 371, 2072-82	59.2	307
337	The sheep genome illuminates biology of the rumen and lipid metabolism. *Science*, **2014**, 344, 1168-1173	33.3	294
336	Finding the missing honey bee genes: lessons learned from a genome upgrade. *BMC Genomics*, **2014**, 15, 86	4.5	284
335	Integrative annotation of variants from 1092 humans: application to cancer genomics. *Science*, **2013**, 342, 1235587	33.3	281
334	A recombination hotspot responsible for two inherited peripheral neuropathies is located near a mariner transposon-like element. *Nature Genetics*, **1996**, 12, 288-97	36.3	278
333	Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors. *JAMA Oncology*, **2016**, 2, 616-624	13.4	276

332	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. *Nature Genetics*, **2011**, 43, 189-96	36.3	271
331	Epistasis dominates the genetic architecture of Drosophila quantitative traits. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 15553-9	11.5	264
330	Convergent evolution of the genomes of marine mammals. *Nature Genetics*, **2015**, 47, 272-5	36.3	263
329	The complete genome sequence of Escherichia coli DH10B: insights into the biology of a laboratory workhorse. *Journal of Bacteriology*, **2008**, 190, 2597-606	3.5	248
328	Gibbon genome and the fast karyotype evolution of small apes. *Nature*, **2014**, 513, 195-201	50.4	241
327	Exome sequencing of ion channel genes reveals complex profiles confounding personal risk assessment in epilepsy. *Cell*, **2011**, 145, 1036-48	56.2	240
326	A drosophila genetic resource of mutants to study mechanisms underlying human genetic diseases. *Cell*, **2014**, 159, 200-214	56.2	239
325	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. *Nature Genetics*, **2009**, 41, 739-45	36.3	236
324	The genomes of two key bumblebee species with primitive eusocial organization. *Genome Biology*, **2015**, 16, 76	18.3	229
323	Multilevel Genomics-Based Taxonomy of Renal Cell Carcinoma. *Cell Reports*, **2016**, 14, 2476-89	10.6	228
322	Whole-genome sequencing for optimized patient management. *Science Translational Medicine*, **2011**, 3, 87re3	17.5	226
321	Use of Exome Sequencing for Infants in Intensive Care Units: Ascertainment of Severe Single-Gene Disorders and Effect on Medical Management. *JAMA Pediatrics*, **2017**, 171, e173438	8.3	215
320	Rise and fall of subclones from diagnosis to relapse in pediatric B-acute lymphoblastic leukaemia. *Nature Communications*, **2015**, 6, 6604	17.4	215
319	Activation of multiple proto-oncogenic tyrosine kinases in breast cancer via loss of the PTPN12 phosphatase. *Cell*, **2011**, 144, 703-18	56.2	214
318	Structure and function of the healthy pre-adolescent pediatric gut microbiome. *Microbiome*, **2015**, 3, 36	16.6	204
317	Genomic profiling of S□ary syndrome identifies alterations of key T cell signaling and differentiation genes. *Nature Genetics*, **2015**, 47, 1426-34	36.3	199
316	Complete genome sequence of Rickettsia typhi and comparison with sequences of other rickettsiae. *Journal of Bacteriology*, **2004**, 186, 5842-55	3.5	192
315	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190

314	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. *Nature Genetics*, **2015**, 47, 654-60	36.3	188
313	Exonuclease mutations in DNA polymerase epsilon reveal replication strand specific mutation patterns and human origins of replication. *Genome Research*, **2014**, 24, 1740-50	9.7	187
312	The house spider genome reveals an ancient whole-genome duplication during arachnid evolution. *BMC Biology*, **2017**, 15, 62	7.3	182
311	The first myriapod genome sequence reveals conservative arthropod gene content and genome organisation in the centipede Strigamia maritima. *PLoS Biology*, **2014**, 12, e1002005	9.7	182
310	Strict evolutionary conservation followed rapid gene loss on human and rhesus Y chromosomes. *Nature*, **2012**, 483, 82-6	50.4	181
309	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. *Nature Communications*, **2016**, 7, 10713	17.4	174
308	Expression of the murine Duchenne muscular dystrophy gene in muscle and brain. *Science*, **1988**, 239, 1416-8	33.3	171
307	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. *Neuron*, **2015**, 88, 499-513	13.9	170
306	Association of MTOR Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. *JAMA Neurology*, **2016**, 73, 836-845	17.2	166
305	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 232-245	11.5	164
304	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. *Cell Reports*, **2015**, 12, 1169-83	10.6	164
303	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	11.5	163
302	Analyses of deep mammalian sequence alignments and constraint predictions for 1% of the human genome. *Genome Research*, **2007**, 17, 760-74	9.7	163
301	Genome of the Asian longhorned beetle (Anoplophora glabripennis), a globally significant invasive species, reveals key functional and evolutionary innovations at the beetle-plant interface. *Genome Biology*, **2016**, 17, 227	18.3	161
300	Sequence and analysis of chromosome 2 of Dictyostelium discoideum. *Nature*, **2002**, 418, 79-85	50.4	158
299	Targeted enrichment beyond the consensus coding DNA sequence exome reveals exons with higher variant densities. *Genome Biology*, **2011**, 12, R68	18.3	157
298	Launching genomics into the cloud: deployment of Mercury, a next generation sequence analysis pipeline. *BMC Bioinformatics*, **2014**, 15, 30	3.6	154
297	Pulmonary alveolar proteinosis caused by deletion of the GM-CSFRalpha gene in the X chromosome pseudoautosomal region 1. *Journal of Experimental Medicine*, **2008**, 205, 2711-6	16.6	150

296	Molecular diagnostic experience of whole-exome sequencing in adult patients. *Genetics in Medicine*, **2016**, 18, 678-85	8.1	149
295	Human CLP1 mutations alter tRNA biogenesis, affecting both peripheral and central nervous system function. *Cell*, **2014**, 157, 636-50	56.2	147
294	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. *Nature Communications*, **2015**, 6, 5897	17.4	147
293	Demographic histories and patterns of linkage disequilibrium in Chinese and Indian rhesus macaques. *Science*, **2007**, 316, 240-3	33.3	146
292	Hemichordate genomes and deuterostome origins. *Nature*, **2015**, 527, 459-65	50.4	144
291	Unique features of a global human ectoparasite identified through sequencing of the bed bug genome. *Nature Communications*, **2016**, 7, 10165	17.4	137
290	Germline mutations in shelterin complex genes are associated with familial glioma. *Journal of the National Cancer Institute*, **2015**, 107, 384	9.7	133
289	Novel somatic and germline mutations in intracranial germ cell tumours. *Nature*, **2014**, 511, 241-5	50.4	131
288	Combined sequence-based and genetic mapping analysis of complex traits in outbred rats. *Nature Genetics*, **2013**, 45, 767-75	36.3	131
287	A massive expansion of effector genes underlies gall-formation in the wheat pest Mayetiola destructor. *Current Biology*, **2015**, 25, 613-20	6.3	126
286	Large-scale comparative sequence analysis of the human and murine Bruton tyrosine kinase loci reveals conserved regulatory domains. *Genome Research*, **1997**, 7, 315-29	9.7	126
285	Lessons learned from additional research analyses of unsolved clinical exome cases. *Genome Medicine*, **2017**, 9, 26	14.4	125
284	Hemimetabolous genomes reveal molecular basis of termite eusociality. *Nature Ecology and Evolution*, **2018**, 2, 557-566	12.3	120
283	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, 11473-8	11.5	120
282	Oligogenic heterozygosity in individuals with high-functioning autism spectrum disorders. *Human Molecular Genetics*, **2011**, 20, 3366-75	5.6	118
281	Assessing structural variation in a personal genome-towards a human reference diploid genome. *BMC Genomics*, **2015**, 16, 286	4.5	117
280	Whole-genome sequence-based analysis of high-density lipoprotein cholesterol. *Nature Genetics*, **2013**, 45, 899-901	36.3	117
279	PGM3 mutations cause a congenital disorder of glycosylation with severe immunodeficiency and skeletal dysplasia. *American Journal of Human Genetics*, **2014**, 95, 96-107	11	115

278	Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. *Genome Medicine*, **2013**, 5, 57	14.4	115
277	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	11	113
276	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. *Neuron*, **2018**, 97, 488-493	13.9	112
275	Multifaceted biological insights from a draft genome sequence of the tobacco hornworm moth, Manduca sexta. *Insect Biochemistry and Molecular Biology*, **2016**, 76, 118-147	4.5	112
274	Reanalysis of Clinical Exome Sequencing Data. *New England Journal of Medicine*, **2019**, 380, 2478-2480	59.2	109
273	Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls. *PLoS Genetics*, **2013**, 9, e1003443	6	108
272	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. *American Journal of Human Genetics*, **2016**, 98, 1051-1066	11	107
271	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. *Journal of Clinical Investigation*, **2015**, 125, 636-51	15.9	106
270	Whole exome sequencing study identifies novel rare and common Alzheimer's-Associated variants involved in immune response and transcriptional regulation. *Molecular Psychiatry*, **2020**, 25, 1859-1875	15.1	106
269	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	11	104
268	De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. *Genome Medicine*, **2013**, 5, 11	14.4	103
267	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. *American Journal of Human Genetics*, **2017**, 100, 185-192	11	102
266	Paradoxical DNA repair and peroxide resistance gene conservation in Bacillus pumilus SAFR-032. *PLoS ONE*, **2007**, 2, e928	3.7	100
265	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
264	The completion of the Mammalian Gene Collection (MGC). *Genome Research*, **2009**, 19, 2324-33	9.7	98
263	Comparative genomics of Gardnerella vaginalis strains reveals substantial differences in metabolic and virulence potential. *PLoS ONE*, **2010**, 5, e12411	3.7	98
262	Genomic Profiling of Pediatric Acute Myeloid Leukemia Reveals a Changing Mutational Landscape from Disease Diagnosis to Relapse. *Cancer Research*, **2016**, 76, 2197-205	10.1	95
261	Comparative validation of the D. melanogaster modENCODE transcriptome annotation. *Genome Research*, **2014**, 24, 1209-23	9.7	95

260	Heterozygous de novo and inherited mutations in the smooth muscle actin (ACTG2) gene underlie megacystis-microcolon-intestinal hypoperistalsis syndrome. *PLoS Genetics*, **2014**, 10, e1004258	6	95
259	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, 167-77	9.7	95
258	Evolutionary History of Chemosensory-Related Gene Families across the Arthropoda. *Molecular Biology and Evolution*, **2017**, 34, 1838-1862	8.3	94
257	BCOR-CCNB3 fusions are frequent in undifferentiated sarcomas of male children. *Modern Pathology*, **2015**, 28, 575-86	9.8	93
256	The Chinese hamster HPRT gene: restriction map, sequence analysis, and multiplex PCR deletion screen. *Genomics*, **1991**, 9, 247-56	4.3	90
255	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	18.3	90
254	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, 757-62	11.5	86
253	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, 275-81	11.2	86
252	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	4.8	86
251	Mapping and characterization of structural variation in 17,795 human genomes. *Nature*, **2020**, 583, 83-89	50.4	84
250	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. *Npj Genomic Medicine*, **2019**, 4, 19	6.2	84
249	DVL1 frameshift mutations clustering in the penultimate exon cause autosomal-dominant Robinow syndrome. *American Journal of Human Genetics*, **2015**, 96, 612-22	11	82
248	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, 1571-81	9.7	82
247	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. *Nature Medicine*, **2012**, 18, 1423-8	50.5	80
246	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	11	80
245	Candidate Loci Associated with AIDS Virus Replication Identified by Whole Genome Sequencing of SIV-Infected Macaques. *AIDS Research and Human Retroviruses*, **2014**, 30, A41-A41	1.6	78
244	EPEN-25. EXCEPTIONAL CLINICAL AND IMAGING RESPONSE TO TRK-INHIBITION IN A PATIENT WITH SUPRATENTORIAL EPENDYMOMA HARBORING NTRK2 GENE FUSION. *Neuro-Oncology*, **2020**, 22, iii312-iii313	1	78
243	PATH-27. MUTATION DETECTION USING PLASMA CELL-FREE DNA IN CHILDREN WITH CENTRAL NERVOUS SYSTEM TUMORS. *Neuro-Oncology*, **2020**, 22, iii430-iii430	1	78

242	PATH-29. HIGH FREQUENCY OF CLINICALLY-RELEVANT TUMOR VARIANTS DETECTED BY MOLECULAR TESTING OF HIGH-RISK PEDIATRIC CNS TUMORS PRELIMINARY FINDINGS FROM THE TEXAS KidsCanSeq STUDY. *Neuro-Oncology*, **2020**, 22, iii430-iii430	1	78
241	The population genomics of rhesus macaques (Macaca mulatta) based on whole-genome sequences. *Genome Research*, **2016**, 26, 1651-1662	9.7	76
240	Ampullary Cancers Harbor ELF3 Tumor Suppressor Gene Mutations and Exhibit Frequent WNT Dysregulation. *Cell Reports*, **2016**, 14, 907-919	10.6	75
239	High-depth African genomes inform human migration and health. *Nature*, **2020**, 586, 741-748	50.4	75
238	NR2F1 mutations cause optic atrophy with intellectual disability. *American Journal of Human Genetics*, **2014**, 94, 303-9	11	74
237	The genome sequence of Mannheimia haemolytica A1: insights into virulence, natural competence, and Pasteurellaceae phylogeny. *Journal of Bacteriology*, **2006**, 188, 7257-66	3.5	73
236	Phenotypic expansion illuminates multilocus pathogenic variation. *Genetics in Medicine*, **2018**, 20, 1528-1537	8.3	71
235	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. *Journal of Experimental Medicine*, **2019**, 216, 2778-2799	16.6	71
234	Molecular and phenotypic variation in patients with severe Hunter syndrome. *Human Molecular Genetics*, **1997**, 6, 479-86	5.6	71
233	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. *Nucleic Acids Research*, **2017**, 45, 1633-1648	20.1	70
232	The comparative genomics and complex population history of baboons. *Science Advances*, **2019**, 5, eaau6947	14.3	69
231	Transmission event of SARS-CoV-2 delta variant reveals multiple vaccine breakthrough infections. *BMC Medicine*, **2021**, 19, 255	11.4	68
230	Recurrent CNVs and SNVs at the NPHP1 locus contribute pathogenic alleles to Bardet-Biedl syndrome. *American Journal of Human Genetics*, **2014**, 94, 745-54	11	67
229	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. *American Journal of Human Genetics*, **2017**, 100, 128-137	11	65
228	Loss of Nardilysin, a Mitochondrial Co-chaperone for Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. *Neuron*, **2017**, 93, 115-131	13.9	65
227	Mutations in PURA cause profound neonatal hypotonia, seizures, and encephalopathy in 5q31.3 microdeletion syndrome. *American Journal of Human Genetics*, **2014**, 95, 579-83	11	65
226	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, 871-90	5.7	65
225	A collagen-binding adhesin, Acb, and ten other putative MSCRAMM and pilus family proteins of Streptococcus gallolyticus subsp. gallolyticus (Streptococcus bovis Group, biotype I). *Journal of Bacteriology*, **2009**, 191, 6643-53	3.5	65

224	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. *Genome Medicine*, **2018**, 10, 74	14.4	65
223	Monoallelic and biallelic mutations in MAB21L2 cause a spectrum of major eye malformations. *American Journal of Human Genetics*, **2014**, 94, 915-23	11	64
222	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *American Journal of Human Genetics*, **2019**, 105, 588-605	11	63
221	Gene content evolution in the arthropods. *Genome Biology*, **2020**, 21, 15	18.3	63
220	DVL3 Alleles Resulting in a -1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. *American Journal of Human Genetics*, **2016**, 98, 553-561	11	63
219	Sheep genome functional annotation reveals proximal regulatory elements contributed to the evolution of modern breeds. *Nature Communications*, **2018**, 9, 859	17.4	61
218	Molecular evolutionary trends and feeding ecology diversification in the Hemiptera, anchored by the milkweed bug genome. *Genome Biology*, **2019**, 20, 64	18.3	60
217	Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. *Journal of Clinical Investigation*, **2016**, 126, 762-78	15.9	59
216	Chemistry-First Approach for Nomination of Personalized Treatment in Lung Cancer. *Cell*, **2018**, 173, 864-878.e29	56.2	58
215	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. *American Journal of Human Genetics*, **2016**, 98, 347-57	11	58
214	Sooty mangabey genome sequence provides insight into AIDS resistance in a natural SIV host. *Nature*, **2018**, 553, 77-81	50.4	57
213	The Toxicogenome of Hyalella azteca: A Model for Sediment Ecotoxicology and Evolutionary Toxicology. *Environmental Science & Technology*, **2018**, 52, 6009-6022	10.3	54
212	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. *Cell*, **2017**, 168, 830-842.e7	56.2	53
211	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	8.1	53
210	Analysis of the quality and utility of random shotgun sequencing at low redundancies. *Genome Research*, **1998**, 8, 1074-84	9.7	53
209	Biallelic mutations in IRF8 impair human NK cell maturation and function. *Journal of Clinical Investigation*, **2017**, 127, 306-320	15.9	53
208	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. *Genetics in Medicine*, **2017**, 19, 13-19	8.1	52
207	Whole-Exome Sequencing in Familial Parkinson Disease. *JAMA Neurology*, **2016**, 73, 68-75	17.2	52

206	Deep sequencing of systematic combinatorial libraries reveals β-lactamase sequence constraints at high resolution. *Journal of Molecular Biology*, **2012**, 424, 150-67	6.5	52
205	Lucilia cuprina genome unlocks parasitic fly biology to underpin future interventions. *Nature Communications*, **2015**, 6, 7344	17.4	51
204	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. *Human Mutation*, **2016**, 37, 804-11	4.7	51
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