Donna M Muzny

List of Publications by Year in Descending Order

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	Distinct somatic DICER1 hotspot mutations in three metachronous ovarian Sertoli-Leydig cell tumors in a patient with DICER1 syndrome.. *Cancer Genetics*, **2022**, 262-263, 53-56	2.3	1
384	Abstract PD15-03: Overlapping molecular features (proliferation, immune signatures andTP53mutations) associated with palbociclib resistance inER+HER2- primary breast cancer. *Cancer Research*, **2022**, 82, PD15-03-PD15-03	10.1	
383	Whole-genome sequencing as an investigational device for return of hereditary disease risk and pharmacogenomic results as part of the All of Us Research Program.. *Genome Medicine*, **2022**, 14, 34	14.4	0
382	Germline Cancer Predisposition Variants in Pediatric Rhabdomyosarcoma: A Report From the Children's Oncology Group. *Journal of the National Cancer Institute*, **2021**, 113, 875-883	9.7	13
381	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3593-3600	2.5	10
380	Cross-oncopanel study reveals high sensitivity and accuracy with overall analytical performance depending on genomic regions. *Genome Biology*, **2021**, 22, 109	18.3	6
379	Chromosome Xq23 is associated with lower atherogenic lipid concentrations and favorable cardiometabolic indices. *Nature Communications*, **2021**, 12, 2182	17.4	5
378	Comparative genomic analysis of sifakas () reveals selection for folivory and high heterozygosity despite endangered status. *Science Advances*, **2021**, 7,	14.3	3
377	Neptune: an environment for the delivery of genomic medicine. *Genetics in Medicine*, **2021**, 23, 1838-1848	8.1	1
376	Molecular Features of Cancers Exhibiting Exceptional Responses to Treatment. *Cancer Cell*, **2021**, 39, 38-53.e7	24.3	18
375	Germline mutation in : a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. *Human Genetics and Genomics Advances*, **2021**, 2, 100014-100014	0.8	3
374	DNA methylation patterns identify subgroups of pancreatic neuroendocrine tumors with clinical association. *Communications Biology*, **2021**, 4, 155	6.7	11
373	Transmission event of SARS-CoV-2 Delta variant reveals multiple vaccine breakthrough infections **2021**,		29
372	Durable Response to Larotrectinib in a Child With Histologic Diagnosis of Recurrent Disseminated Ependymoma Discovered to Harbor an Fusion: The Impact of Integrated Genomic Profiling. *JCO Precision Oncology*, **2021**, 5,	3.6	2
371	Exome sequencing in children with clinically suspected maturity-onset diabetes of the young. *Pediatric Diabetes*, **2021**, 22, 960-968	3.6	2
370	Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals. *PLoS ONE*, **2021**, 16, e0244468	3.7	8
369	Genetic testing in ambulatory cardiology clinics reveals high rate of findings with clinical management implications. *Genetics in Medicine*, **2021**, 23, 2404-2414	8.1	3

368	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. *American Journal of Human Genetics*, **2021**, 108, 1710-1724	11	2
367	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. *American Journal of Human Genetics*, **2021**, 108, 1981-2005	11	4
366	Transmission event of SARS-CoV-2 delta variant reveals multiple vaccine breakthrough infections. *BMC Medicine*, **2021**, 19, 255	11.4	68
365	Paternal age in rhesus macaques is positively associated with germline mutation accumulation but not with measures of offspring sociability. *Genome Research*, **2020**, 30, 826-834	9.7	19
364	Frequency of genomic secondary findings among 21,915 eMERGE network participants. *Genetics in Medicine*, **2020**, 22, 1470-1477	8.1	23
363	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. *Genetics in Medicine*, **2020**, 22, 1633-1641	8.1	18
362	Mapping and characterization of structural variation in 17,795 human genomes. *Nature*, **2020**, 583, 83-89	50.4	84
361	Brown marmorated stink bug, Halyomorpha halys (Stl), genome: putative underpinnings of polyphagy, insecticide resistance potential and biology of a top worldwide pest. *BMC Genomics*, **2020**, 21, 227	4.5	28
360	Wolff-Parkinson-White syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1387-1399	2.5	5
359	HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. *Science*, **2020**, 369, 202-207	33.3	36
358	Gene content evolution in the arthropods. *Genome Biology*, **2020**, 21, 15	18.3	63
357	Allelic Heterogeneity at the CRP Locus Identified by Whole-Genome Sequencing in Multi-ancestry Cohorts. *American Journal of Human Genetics*, **2020**, 106, 112-120	11	2
356	Genetics of schizophrenia in the South African Xhosa. *Science*, **2020**, 367, 569-573	33.3	44
355	Disease-associated CTNNBL1 mutation impairs somatic hypermutation by decreasing nuclear AID. *Journal of Clinical Investigation*, **2020**, 130, 4411-4422	15.9	5
354	Human NK cell deficiency as a result of biallelic mutations in MCM10. *Journal of Clinical Investigation*, **2020**, 130, 5272-5286	15.9	15
353	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. *PLoS Biology*, **2020**, 18, e3000954	9.7	24
352	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
351	PATH-27. MUTATION DETECTION USING PLASMA CELL-FREE DNA IN CHILDREN WITH CENTRAL NERVOUS SYSTEM TUMORS. *Neuro-Oncology*, **2020**, 22, iii430-iii430	1	78

350	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
349	Sawfly Genomes Reveal Evolutionary Acquisitions That Fostered the Mega-Radiation of Parasitoid and Eusocial Hymenoptera. *Genome Biology and Evolution*, **2020**, 12, 1099-1188	3.9	7
348	Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals **2020**,		8
347	Oligonucleotide Capture Sequencing of the SARS-CoV-2 Genome and Subgenomic Fragments from COVID-19 Individuals **2020**,		7
346	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. *Human Mutation*, **2020**, 41, 487-501	4.7	24
345	Genetic Burden Contributing to Extremely Low or High Bone Mineral Density in a Senior Male Population From the Osteoporotic Fractures in Men Study (MrOS). *JBMR Plus*, **2020**, 4, e10335	3.9	1
344	Whole genome sequence analysis of pulmonary function and COPD in 19,996 multi-ethnic participants. *Nature Communications*, **2020**, 11, 5182	17.4	6
343	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. *Genetics in Medicine*, **2020**, 22, 1768-1776	8.1	11
342	PCM1 is necessary for focal ciliary integrity and is a candidate for severe schizophrenia. *Nature Communications*, **2020**, 11, 5903	17.4	4
341	Loss of the Polyketide Synthase StlB Results in Stalk Cell Overproduction in Polysphondylium violaceum. *Genome Biology and Evolution*, **2020**, 12, 674-683	3.9	2
340	Next Generation Sequencing of 134 Children with Autism Spectrum Disorder and Regression. *Genes* , **2020**, 11,	4.2	7
339	Genome-enabled insights into the biology of thrips as crop pests. *BMC Biology*, **2020**, 18, 142	7.3	17
338	High-depth African genomes inform human migration and health. *Nature*, **2020**, 586, 741-748	50.4	75
337	Phenotypic expansion in KIF1A-related dominant disorders: A description of novel variants and review of published cases. *Human Mutation*, **2020**, 41, 2094-2104	4.7	1
336	Community-based recruitment and exome sequencing indicates high diagnostic yield in adults with intellectual disability. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1439	2.3	1
335	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. *Neurology: Genetics*, **2020**, 6, e498	3.8	5
334	Sequence analysis in reveals pervasiveness of X-Y arms races in mammalian lineages. *Genome Research*, **2020**, 30, 1716-1726	9.7	6
333	Whole exome sequencing study identifies novel rare and common Alzheimer-Associated variants involved in immune response and transcriptional regulation. *Molecular Psychiatry*, **2020**, 25, 1859-1875	15.1	106

332	Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in. *Acta Cardiologica*, **2020**, 75, 748-753	0.9	5
331	Cohort Profile: The Right Drug, Right Dose, Right Time: Using Genomic Data to Individualize Treatment Protocol (RIGHT Protocol). *International Journal of Epidemiology*, **2020**, 49, 23-24k	7.8	19
330	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression **2020**, 18, e3000954		
329	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression **2020**, 18, e3000954		
328	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression **2020**, 18, e3000954		
327	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression **2020**, 18, e3000954		
326	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression **2020**, 18, e3000954		
325	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression **2020**, 18, e3000954		
324	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	11.5	49
323	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *American Journal of Human Genetics*, **2019**, 105, 588-605	11	63
322	Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. *Hepatology*, **2019**, 70, 899-910	11.2	35
321	The comparative genomics and complex population history of baboons. *Science Advances*, **2019**, 5, eaau6947	12.3	69
320	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 3049-3067	5.6	30
319	Reanalysis of Clinical Exome Sequencing Data. *New England Journal of Medicine*, **2019**, 380, 2478-2480	59.2	109
318	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. *Genome Medicine*, **2019**, 11, 30	14.4	27
317	Leveraging Human Microbiome Features to Diagnose and Stratify Children with Irritable Bowel Syndrome. *Journal of Molecular Diagnostics*, **2019**, 21, 449-461	5.1	33
316	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. *Genome Medicine*, **2019**, 11, 25	14.4	14
315	IgG4-related disease: Association with a rare gene variant expressed in cytotoxic T cells. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e686	2.3	6

314	Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. *Genetics in Medicine*, **2019**, 21, 2135-2144	8.1	13
313	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smith-Magenis syndrome. *Genome Medicine*, **2019**, 11, 12	14.4	7
312	Rare variants in SLC5A10 are associated with serum 1,5-anhydroglucitol (1,5-AG) in the Atherosclerosis Risk in Communities (ARIC) Study. *Scientific Reports*, **2019**, 9, 5941	4.9	3
311	Molecular evolutionary trends and feeding ecology diversification in the Hemiptera, anchored by the milkweed bug genome. *Genome Biology*, **2019**, 20, 64	18.3	60
310	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. *Genetics in Medicine*, **2019**, 21, 161-172	8.1	36
309	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. *Npj Genomic Medicine*, **2019**, 4, 19	6.2	84
308	MHC genotyping from rhesus macaque exome sequences. *Immunogenetics*, **2019**, 71, 531-544	3.2	9
307	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. *American Journal of Human Genetics*, **2019**, 105, 132-150	11	50
306	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. *American Journal of Human Genetics*, **2019**, 105, 302-316	11	19
305	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. *Journal of Experimental Medicine*, **2019**, 216, 2778-2799	16.6	71
304	A Genocentric Approach to Discovery of Mendelian Disorders. *American Journal of Human Genetics*, **2019**, 105, 974-986	11	18
303	Genomic Characterization of a Pediatric Cohort with Non-Malignant Lymphoproliferative Disorders. *Blood*, **2019**, 134, 83-83	2.2	
302	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. *American Journal of Human Genetics*, **2019**, 104, 422-438	11	10
301	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. *American Journal of Human Genetics*, **2019**, 104, 164-178	11	27
300	Insights into genetics, human biology and disease gleaned from family based genomic studies. *Genetics in Medicine*, **2019**, 21, 798-812	8.1	100
299	Genetic architecture of laterality defects revealed by whole exome sequencing. *European Journal of Human Genetics*, **2019**, 27, 563-573	5.3	24
298	A biallelic ANTXR1 variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1015-1022	2.5	9
297	Sheep genome functional annotation reveals proximal regulatory elements contributed to the evolution of modern breeds. *Nature Communications*, **2018**, 9, 859	17.4	61

296	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. *American Journal of Human Genetics*, **2018**, 102, 494-504	11	44
295	Chemistry-First Approach for Nomination of Personalized Treatment in Lung Cancer. *Cell*, **2018**, 173, 864-878.e29	56.2	58
294	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. *Human Molecular Genetics*, **2018**, 27, 2064-2075	5.6	9
293	The Toxicogenome of Hyalella azteca: A Model for Sediment Ecotoxicology and Evolutionary Toxicology. *Environmental Science & Technology*, **2018**, 52, 6009-6022	10.3	54
292	Sequence-Based Analysis of Lipid-Related Metabolites in a Multiethnic Study. *Genetics*, **2018**, 209, 607-616	4	4
291	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. *Neuron*, **2018**, 97, 488-493	13.9	112
290	Hemimetabolous genomes reveal molecular basis of termite eusociality. *Nature Ecology and Evolution*, **2018**, 2, 557-566	12.3	120
289	Mutations in PI3K110□cause impaired natural killer cell function partially rescued by rapamycin treatment. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 605-617.e7	11.5	28
288	Sooty mangabey genome sequence provides insight into AIDS resistance in a natural SIV host. *Nature*, **2018**, 553, 77-81	50.4	57
287	Phenotypic expansion illuminates multilocus pathogenic variation. *Genetics in Medicine*, **2018**, 20, 1528-1537	8.1	71
286	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. *Journal of Pediatric Genetics*, **2018**, 7, 164-173	0.7	7
285	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. *Human Genetics*, **2018**, 137, 689-703	6.3	13
284	IRF2BPL Is Associated with Neurological Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 245-260	11	39
283	Comparative genomics of the miniature wasp and pest control agent Trichogramma pretiosum. *BMC Biology*, **2018**, 16, 54	7.3	33
282	Elucidating the molecular pathogenesis of glioma: integrated germline and somatic profiling of a familial glioma case series. *Neuro-Oncology*, **2018**, 20, 1625-1633	1	6
281	Mismatch repair gene mutations lead to lynch syndrome colorectal cancer in rhesus macaques. *Genes and Cancer*, **2018**, 9, 142-152	2.9	10
280	Phenotype expansion and development in Kosaki overgrowth syndrome. *Clinical Genetics*, **2018**, 93, 919-924	4	17
279	Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. *Genetics in Medicine*, **2018**, 20, 855-866	8.1	16

278	P1-149: THE ALZHEIMER'S DISEASE SEQUENCING PROJECT (ADSP) DATA UPDATE 2018 **2018**, 14, P333-P334		
277	Spontaneous Spongiform Brainstem Degeneration in a Young Mouse Lemur () with Conspicuous Behavioral, Motor, Growth, and Ocular Pathologies. *Comparative Medicine*, **2018**, 68, 489-495	1.6	1
276	The genome of the water strider Gerris buenoi reveals expansions of gene repertoires associated with adaptations to life on the water. *BMC Genomics*, **2018**, 19, 832	4.5	32
275	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. *Genome Medicine*, **2018**, 10, 74	14.4	65
274	Reproductive Longevity Predicts Mutation Rates in Primates. *Current Biology*, **2018**, 28, 3193-3197.e5	6.3	48
273	Temporal development of the gut microbiome in early childhood from the TEDDY study. *Nature*, **2018**, 562, 583-588	50.4	619
272	Phenotypic expansion in - a common cause of intellectual disability in females. *Annals of Clinical and Translational Neurology*, **2018**, 5, 1277-1285	5.3	37
271	Common Coding Variants in Are Associated With the Nav1.8 Late Current and Cardiac Conduction. *Circulation Genomic and Precision Medicine*, **2018**, 11, e001663	5.2	14
270	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
269	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
268	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. *American Journal of Human Genetics*, **2017**, 100, 185-192	11	102
267	Practical Approaches for Whole-Genome Sequence Analysis of Heart- and Blood-Related Traits. *American Journal of Human Genetics*, **2017**, 100, 205-215	11	29
266	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. *American Journal of Human Genetics*, **2017**, 100, 343-351	11	23
265	The next generation of population-based spinal muscular atrophy carrier screening: comprehensive pan-ethnic SMN1 copy-number and sequence variant analysis by massively parallel sequencing. *Genetics in Medicine*, **2017**, 19, 936-944	8.1	36
264	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. *Cell*, **2017**, 168, 830-842.e7	36.2	53
263	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
262	Evolutionary History of Chemosensory-Related Gene Families across the Arthropoda. *Molecular Biology and Evolution*, **2017**, 34, 1838-1862	8.3	94
261	Exome sequencing reveals novel genetic loci influencing obesity-related traits in Hispanic children. *Obesity*, **2017**, 25, 1270-1276	8	9

260	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a mutation. *Journal of Physical Education and Sports Management*, **2017**, 3, a000984	2.8	12
259	Extremely low-coverage whole genome sequencing in South Asians captures population genomics information. *BMC Genomics*, **2017**, 18, 396	4.5	18
258	Lessons learned from additional research analyses of unsolved clinical exome cases. *Genome Medicine*, **2017**, 9, 26	14.4	125
257	Improved full-length killer cell immunoglobulin-like receptor transcript discovery in Mauritian cynomolgus macaques. *Immunogenetics*, **2017**, 69, 325-339	3.2	18
256	Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. *Nature Genetics*, **2017**, 49, 613-617	36.3	29
255	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. *New England Journal of Medicine*, **2017**, 376, 21-31	59.2	391
254	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. *American Journal of Human Genetics*, **2017**, 100, 128-137	11	65
253	Loss of Nardilysin, a Mitochondrial Co-chaperone for □Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. *Neuron*, **2017**, 93, 115-131	13.9	65
252	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
251	Whole exome sequencing in the Framingham Heart Study identifies rare variation in HYAL2 that influences platelet aggregation. *Thrombosis and Haemostasis*, **2017**, 117, 1083-1092	7	9
250	Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. *Genome Medicine*, **2017**, 9, 95	14.4	21
249	The house spider genome reveals an ancient whole-genome duplication during arachnid evolution. *BMC Biology*, **2017**, 15, 62	7.3	182
248	Hybrid de novo genome assembly and centromere characterization of the gray mouse lemur (Microcebus murinus). *BMC Biology*, **2017**, 15, 110	7.3	40
247	The gut mycobiome of the Human Microbiome Project healthy cohort. *Microbiome*, **2017**, 5, 153	16.6	324
246	Neonatal fractures as a presenting feature of LMOD3-associated congenital myopathy. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2789-2794	2.5	12
245	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. *European Journal of Human Genetics*, **2017**, 25, 1246-1252	5.3	27
244	Whole-genome sequencing study of serum peptide levels: the Atherosclerosis Risk in Communities study. *Human Molecular Genetics*, **2017**, 26, 3442-3450	5.6	16
243	REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. *American Journal of Human Genetics*, **2017**, 101, 149-156	11	25

242	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
241	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 232-245	11.5	164
240	[P3090]: THE ALZHEIMER'S DISEASE SEQUENCING PROJECT (ADSP) DATA UPDATE 2017 **2017**, 13, P968-P968		
239	SVachra: a tool to identify genomic structural variation in mate pair sequencing data containing inward and outward facing reads. *BMC Genomics*, **2017**, 18, 691	4.5	3
238	Biallelic mutations in IRF8 impair human NK cell maturation and function. *Journal of Clinical Investigation*, **2017**, 127, 306-320	15.9	53
237	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. *Nucleic Acids Research*, **2017**, 45, 1633-1648	20.1	70
236	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. *Nature Communications*, **2016**, 7, 10713	17.4	174
235	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. *American Journal of Human Genetics*, **2016**, 99, 886-893	11	42
234	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
233	Targeted sequencing of genome wide significant loci associated with bone mineral density (BMD) reveals significant novel and rare variants: the Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) targeted sequencing study. *Human Molecular Genetics*, **2016**, 25, 5234-5243	5.6	6
232	Postmortem genetic screening for the identification, verification, and reporting of genetic variants contributing to the sudden death of the young. *Genome Research*, **2016**, 26, 1170-7	9.7	17
231	The population genomics of rhesus macaques (Macaca mulatta) based on whole-genome sequences. *Genome Research*, **2016**, 26, 1651-1662	9.7	76
230	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
229	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. *Genome Medicine*, **2016**, 8, 105	14.4	16
228	Loss-of-function variants influence the human serum metabolome. *Science Advances*, **2016**, 2, e1600800	14.3	27
227	An open access pilot freely sharing cancer genomic data from participants in Texas. *Scientific Data*, **2016**, 3, 160010	8.2	12
226	SV-STAT accurately detects structural variation via alignment to reference-based assemblies. *Source Code for Biology and Medicine*, **2016**, 11, 8	1.9	3
225	Multilevel Genomics-Based Taxonomy of Renal Cell Carcinoma. *Cell Reports*, **2016**, 14, 2476-89	10.6	228

224	PEHO Syndrome May Represent Phenotypic Expansion at the Severe End of the Early-Onset Encephalopathies. *Pediatric Neurology*, **2016**, 60, 83-7	2.9	22
223	POGZ truncating alleles cause syndromic intellectual disability. *Genome Medicine*, **2016**, 8, 3	14.4	49
222	Two male sibs with severe micrognathia and a missense variant in MED12. *European Journal of Medical Genetics*, **2016**, 59, 367-72	2.6	9
221	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. *Human Mutation*, **2016**, 37, 804-11	4.7	51
220	Rare Exome Sequence Variants in CLCN6 Reduce Blood Pressure Levels and Hypertension Risk. *Circulation: Cardiovascular Genetics*, **2016**, 9, 64-70		35
219	Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors. *JAMA Oncology*, **2016**, 2, 616-624	13.4	276
218	Molecular diagnostic experience of whole-exome sequencing in adult patients. *Genetics in Medicine*, **2016**, 18, 678-85	8.1	149
217	Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. *Human Mutation*, **2016**, 37, 231-234	4.7	14
216	Ampullary Cancers Harbor ELF3 Tumor Suppressor Gene Mutations and Exhibit Frequent WNT Dysregulation. *Cell Reports*, **2016**, 14, 907-919	10.6	75
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	Unique features of a global human ectoparasite identified through sequencing of the bed bug genome. *Nature Communications*, **2016**, 7, 10165	17.4	137
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4	Molecular evolutionary trends and feeding ecology diversification in the Hemiptera, anchored by the milkweed bug genome		11
3	Genome-enabled insights into the biology of thrips as crop pests		1
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