# Donna M Muzny

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3858476/publications.pdf

Version: 2024-02-01

| 391 | 103,188 | 701 | 121 | 230 | 306 |
|---|---|---|---|---|---|
| papers | citations | | h-index | | g-index |

| 406 | 406 | 406 | 111811 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. Journal of Allergy and Clinical Immunology, 2022, 149, 758-766. | 2.9 | 6 |
| 2 | 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. | 0.4 | 2 |
| 3 | Rare coding variants in 35 genes associate with circulating lipid levelsâ€"A multi-ancestry analysis of 170,000 exomes. American Journal of Human Genetics, 2022, 109, 81-96. | 6.2 | 24 |
| 4 | Fully resolved assembly of Cryptosporidium parvum. GigaScience, 2022, 11, . | 6.4 | 8 |
| 5 | Abstract PD15-03: Overlapping molecular features (proliferation, immune signatures) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50 5 Cancer Research, 2022, 82, PD15-03-PD15-03. | 0.9 | 0 |
| 6 | 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. | 8.2 | 27 |
| 7 | Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918. | 2.5 | 8 |
| 8 | Implementation of preemptive DNA sequenceâ€"based pharmacogenomics testing across a large academic medical center: The Mayo-Baylor RIGHT 10K Study. Genetics in Medicine, 2022, 24, 1062-1072. | 2.4 | 28 |
| 9 | OP011: Physician recommendations after germline sequencing in pediatric cancer patients: Texas KidsCanSeq study. Genetics in Medicine, 2022, 24, S344. | 2.4 | 0 |
| 10 | Polygenic transcriptome risk scores for COPD and lung function improve cross-ethnic portability of prediction in the NHLBI TOPMed program. American Journal of Human Genetics, 2022, 109, 857-870. | 6.2 | 7 |
| 11 | Whole-exome sequencing of 14â€‰389 individuals from the ESP and CHARGE consortia identifies novel rare variation associated with hemostatic factors. Human Molecular Genetics, 2022, 31, 3120-3132. | 2.9 | 3 |
| 12 | Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600. | 1.2 | 16 |
| 13 | Molecular Features of Cancers Exhibiting Exceptional Responses to Treatment. Cancer Cell, 2021, 39, 38-53.e7. | 16.8 | 65 |
| 14 | Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014. | 1.7 | 10 |
| 15 | DNA methylation patterns identify subgroups of pancreatic neuroendocrine tumors with clinical association. Communications Biology, 2021, 4, 155. | 4.4 | 26 |
| 16 | Cross-oncopanel study reveals high sensitivity and accuracy with overall analytical performance depending on genomic regions. Genome Biology, 2021, 22, 109. | 8.8 | 20 |
| 17 | Chromosome Xq23 is associated with lower atherogenic lipid concentrations and favorable cardiometabolic indices. Nature Communications, 2021, 12, 2182. | 12.8 | 17 |
| 18 | Comparative genomic analysis of sifakas ( Propithecus ) reveals selection for folivory and high heterozygosity despite endangered status. Science Advances, 2021, 7, . | 10.3 | 14 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846. | 2.4 | 3 |
| 20 | Sequencing of a central nervous system tumor demonstrates cancer transmission in an organ transplant. Life Science Alliance, 2021, 4, e202000941. | 2.8 | 1 |
| 21 | Durable Response to Larotrectinib in a Child With Histologic Diagnosis of Recurrent Disseminated Ependymoma Discovered to Harbor an NTRK2 Fusion: The Impact of Integrated Genomic Profiling. JCO Precision Oncology, 2021, 5, 1221-1227. | 3.0 | 5 |
| 22 | Exome sequencing in children with clinically suspected <scp>maturityâ€onset</scp> diabetes of the young. Pediatric Diabetes, 2021, 22, 960-968. | 2.9 | 6 |
| 23 | Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals. PLoS ONE, 2021, 16, e0244468. | 2.5 | 20 |
| 24 | Genetic testing in ambulatory cardiology clinics reveals high rate of findings with clinical management implications. Genetics in Medicine, 2021, 23, 2404-2414. | 2.4 | 14 |
| 25 | COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724. | 6.2 | 18 |
| 26 | High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005. | 6.2 | 38 |
| 27 | Transmission event of SARS-CoV-2 delta variant reveals multiple vaccine breakthrough infections. BMC Medicine, 2021, 19, 255. | 5.5 | 137 |
| 28 | Germline Cancer Predisposition Variants inâ€, Pediatric Rhabdomyosarcoma: A Report From the Childrenâ€™s Oncology Group. Journal of the National Cancer Institute, 2021, 113, 875-883. | 6.3 | 55 |
| 29 | 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. | 7.9 | 191 |
| 30 | Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in RBM20. Acta Cardiologica, 2020, 75, 748-753. | 0.9 | 8 |
| 31 | 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. | 1.9 | 34 |
| 32 | A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501. | 2.5 | 58 |
| 33 | 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. | 2.7 | 1 |
| 34 | Whole genome sequence analysis of pulmonary function and COPD in 19,996 multi-ethnic participants. Nature Communications, 2020, 11, 5182. | 12.8 | 32 |
| 35 | 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. | 2.4 | 30 |
| 36 | PCM1 is necessary for focal ciliary integrity and is a candidate for severe schizophrenia. Nature Communications, 2020, 11, 5903. | 12.8 | 13 |

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|---|---|---|---|
| 37 | Loss of the Polyketide Synthase StlB Results in Stalk Cell Overproduction in Polysphondylium violaceum. Genome Biology and Evolution, 2020, 12, 674-683. | 2.5 | 8 |
| 38 | Next Generation Sequencing of 134 Children with Autism Spectrum Disorder and Regression. Genes, 2020, 11, 853. | 2.4 | 16 |
| 39 | Genome-enabled insights into the biology of thrips as crop pests. BMC Biology, 2020, 18, 142. | 3.8 | 54 |
| 40 | High-depth African genomes inform human migration and health. Nature, 2020, 586, 741-748. | 27.8 | 197 |
| 41 | Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104. | 2.5 | 8 |
| 42 | Communityâ€based recruitment and exome sequencing indicates high diagnostic yield in adults with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1439. | 1.2 | 6 |
| 43 | Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498. | 1.9 | 11 |
| 44 | Sequence analysis in Bos taurus reveals pervasiveness of Xâ€“Y arms races in mammalian lineages. Genome Research, 2020, 30, 1716-1726. | 5.5 | 29 |
| 45 | 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. | 5.5 | 48 |
| 46 | Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477. | 2.4 | 61 |
| 47 | CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641. | 2.4 | 36 |
| 48 | Mapping and characterization of structural variation in 17,795 human genomes. Nature, 2020, 583, 83-89. | 27.8 | 194 |
| 49 | Brown marmorated stink bug, Halyomorpha halys (StÃ¥l), genome: putative underpinnings of polyphagy, insecticide resistance potential and biology of a top worldwide pest. BMC Genomics, 2020, 21, 227. | 2.8 | 60 |
| 50 | <scp>Wolffâ€“Parkinsonâ€“White</scp> 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. | 1.2 | 14 |
| 51 | HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. Science, 2020, 369, 202-207. | 12.6 | 65 |
| 52 | Gene content evolution in the arthropods. Genome Biology, 2020, 21, 15. | 8.8 | 150 |
| 53 | Allelic Heterogeneity at the CRP Locus Identified by Whole-Genome Sequencing in Multi-ancestry Cohorts. American Journal of Human Genetics, 2020, 106, 112-120. | 6.2 | 9 |
| 54 | Genetics of schizophrenia in the South African Xhosa. Science, 2020, 367, 569-573. | 12.6 | 93 |

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|---|---|---|---|
| 55 | Sawfly Genomes Reveal Evolutionary Acquisitions That Fostered the Mega-Radiation of Parasitoid and Eusocial Hymenoptera. Genome Biology and Evolution, 2020, 12, 1099-1188. | 2.5 | 17 |
| 56 | Disease-associated CTNNBL1 mutation impairs somatic hypermutation by decreasing nuclear AID. Journal of Clinical Investigation, 2020, 130, 4411-4422. | 8.2 | 11 |
| 57 | Human NK cell deficiency as a result of biallelic mutations in MCM10. Journal of Clinical Investigation, 2020, 130, 5272-5286. | 8.2 | 44 |
| 58 | Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. PLoS Biology, 2020, 18, e3000954. | 5.6 | 73 |
| 59 | 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. | 1.2 | 0 |
| 60 | PATH-27. MUTATION DETECTION USING PLASMA CELL-FREE DNA IN CHILDREN WITH CENTRAL NERVOUS SYSTEM TUMORS. Neuro-Oncology, 2020, 22, iii430-iii430. | 1.2 | 0 |
| 61 | PATH-29. HIGH FREQUENCY OF CLINICALLY-RELEVANT TUMOR VARIANTS DETECTED BY MOLECULAR TESTING OF HIGH-RISK PEDIATRIC CNS TUMORS â€“ PRELIMINARY FINDINGS FROM THE TEXAS KidsCanSeq STUDY. Neuro-Oncology, 2020, 22, iii430-iii430. | 1.2 | 0 |
| 62 | Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954. | | 0 |
| 63 | Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954. | | 0 |
| 64 | Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954. | | 0 |
| 65 | Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954. | | 0 |
| 66 | Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954. | | 0 |
| 67 | Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954. | | 0 |
| 68 | A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172. | 2.4 | 60 |
| 69 | Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19. | 3.8 | 163 |
| 70 | MHC genotyping from rhesus macaque exome sequences. Immunogenetics, 2019, 71, 531-544. | 2.4 | 16 |
| 71 | The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150. | 6.2 | 74 |
| 72 | Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316. | 6.2 | 56 |

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|---|---|---|---|
| 73 | A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799. | 8.5 | 132 |
| 74 | A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986. | 6.2 | 30 |
| 75 | 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. | 7.1 | 122 |
| 76 | Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. American Journal of Human Genetics, 2019, 105, 588-605. | 6.2 | 99 |
| 77 | Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. Hepatology, 2019, 70, 899-910. | 7.3 | 58 |
| 78 | The comparative genomics and complex population history of Papio baboons. Science Advances, 2019, 5, eaau6947. | 10.3 | 115 |
| 79 | 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. | 3.6 | 53 |
| 80 | Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480. | 27.0 | 205 |
| 81 | 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. | 8.2 | 42 |
| 82 | Leveraging Human Microbiome Features to Diagnose and Stratify Children with Irritable Bowel Syndrome. Journal of Molecular Diagnostics, 2019, 21, 449-461. | 2.8 | 59 |
| 83 | Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25. | 8.2 | 22 |
| 84 | IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686. | 1.2 | 8 |
| 85 | Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. Genetics in Medicine, 2019, 21, 2135-2144. | 2.4 | 19 |
| 86 | De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12. | 8.2 | 23 |
| 87 | 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. | 3.3 | 9 |
| 88 | Molecular evolutionary trends and feeding ecology diversification in the Hemiptera, anchored by the milkweed bug genome. Genome Biology, 2019, 20, 64. | 8.8 | 114 |
| 89 | Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438. | 6.2 | 27 |
| 90 | 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. | 6.2 | 59 |

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|---|---|---|---|
| 91 | Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812. | 2.4 | 161 |
| 92 | Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573. | 2.8 | 44 |
| 93 | Genomic Characterization of a Pediatric Cohort with Non-Malignant Lymphoproliferative Disorders. Blood, 2019, 134, 83-83. | 1.4 | 0 |
| 94 | 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. | 1.2 | 11 |
| 95 | Sheep genome functional annotation reveals proximal regulatory elements contributed to the evolution of modern breeds. Nature Communications, 2018, 9, 859. | 12.8 | 126 |
| 96 | Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504. | 6.2 | 59 |
| 97 | Chemistry-First Approach for Nomination of Personalized Treatment in Lung Cancer. Cell, 2018, 173, 864-878.e29. | 28.9 | 102 |
| 98 | The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075. | 2.9 | 16 |
| 99 | The Toxicogenome of Hyalella azteca: A Model for Sediment Ecotoxicology and Evolutionary Toxicology. Environmental Science & Technology, 2018, 52, 6009-6022. | 10.0 | 79 |
| 100 | Sequence-Based Analysis of Lipid-Related Metabolites in a Multiethnic Study. Genetics, 2018, 209, 607-616. | 2.9 | 8 |
| 101 | SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493. | 8.1 | 265 |
| 102 | Hemimetabolous genomes reveal molecular basis of termite eusociality. Nature Ecology and Evolution, 2018, 2, 557-566. | 7.8 | 223 |
| 103 | Mutations in PI3K110Î´ cause impaired natural killer cell function partially rescued by rapamycin treatment. Journal of Allergy and Clinical Immunology, 2018, 142, 605-617.e7. | 2.9 | 36 |
| 104 | Sooty mangabey genome sequence provides insight into AIDS resistance in a natural SIV host. Nature, 2018, 553, 77-81. | 27.8 | 81 |
| 105 | Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537. | 2.4 | 104 |
| 106 | Phenotype expansion and development in Kosaki overgrowth syndrome. Clinical Genetics, 2018, 93, 919-924. | 2.0 | 17 |
| 107 | Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. Genetics in Medicine, 2018, 20, 855-866. | 2.4 | 22 |
| 108 | P1â€149: THE ALZHEIMER'S DISEASE SEQUENCING PROJECT (ADSP) DATA UPDATE 2018. Alzheimer's and Dementia, 2018, 14, P333. | 0.8 | 0 |

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|---|---|---|---|
| 109 | Spontaneous Spongiform Brainstem Degeneration in a Young Mouse Lemur (Microcebus murinus) with Conspicuous Behavioral, Motor, Growth, and Ocular Pathologies. Comparative Medicine, 2018, 68, 489-495. | 1.0 | 2 |
| 110 | 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. | 2.8 | 47 |
| 111 | Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74. | 8.2 | 105 |
| 112 | Reproductive Longevity Predicts Mutation Rates in Primates. Current Biology, 2018, 28, 3193-3197.e5. | 3.9 | 94 |
| 113 | Temporal development of the gut microbiome in early childhood from the TEDDY study. Nature, 2018, 562, 583-588. | 27.8 | 1,220 |
| 114 | Phenotypic expansion in <scp>DDX</scp>3X â€“ a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285. | 3.7 | 66 |
| 115 | Common Coding Variants in SCN10A Are Associated With the Nav1.8 Late Current and Cardiac Conduction. Circulation Genomic and Precision Medicine, 2018, 11, e001663. | 3.6 | 26 |
| 116 | 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. | 6.2 | 128 |
| 117 | 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, 07, 164-173. | 0.7 | 15 |
| 118 | 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. | 3.8 | 24 |
| 119 | IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260. | 6.2 | 69 |
| 120 | Comparative genomics of the miniature wasp and pest control agent Trichogramma pretiosum. BMC Biology, 2018, 16, 54. | 3.8 | 57 |
| 121 | Elucidating the molecular pathogenesis of glioma: integrated germline and somatic profiling of a familial glioma case series. Neuro-Oncology, 2018, 20, 1625-1633. | 1.2 | 12 |
| 122 | Mismatch repair gene mutations lead to lynch syndrome colorectal cancer in rhesus macaques. Genes and Cancer, 2018, 9, 142-152. | 1.9 | 18 |
| 123 | Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237. | 14.5 | 98 |
| 124 | Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genetics in Medicine, 2017, 19, 13-19. | 2.4 | 74 |
| 125 | The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192. | 6.2 | 142 |
| 126 | Practical Approaches for Whole-Genome Sequence Analysis of Heart- and Blood-Related Traits. American Journal of Human Genetics, 2017, 100, 205-215. | 6.2 | 50 |

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|---|---|---|---|
| 127 | 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. | 6.2 | 35 |
| 128 | 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. | 2.4 | 70 |
| 129 | An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7. | 28.9 | 66 |
| 130 | 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. | 6.2 | 181 |
| 131 | Evolutionary History of Chemosensory-Related Gene Families across the Arthropoda. Molecular Biology and Evolution, 2017, 34, 1838-1862. | 8.9 | 157 |
| 132 | Exome sequencing reveals novel genetic loci influencing obesityâ€related traits in Hispanic children. Obesity, 2017, 25, 1270-1276. | 3.0 | 10 |
| 133 | An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a TUBB3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984. | 1.2 | 18 |
| 134 | Extremely low-coverage whole genome sequencing in South Asians captures population genomics information. BMC Genomics, 2017, 18, 396. | 2.8 | 26 |
| 135 | Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26. | 8.2 | 184 |
| 136 | Improved full-length killer cell immunoglobulin-like receptor transcript discovery in Mauritian cynomolgus macaques. Immunogenetics, 2017, 69, 325-339. | 2.4 | 25 |
| 137 | Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617. | 21.4 | 40 |
| 138 | Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31. | 27.0 | 565 |
| 139 | A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137. | 6.2 | 96 |
| 140 | Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131. | 8.1 | 95 |
| 141 | Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438. | 6.2 | 348 |
| 142 | Neonatal fractures as a presenting feature of LMOD3â€associated congenital myopathy. American Journal of Medical Genetics, Part A, 2017, 173, 2789-2794. | 1.2 | 17 |
| 143 | Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252. | 2.8 | 34 |
| 144 | Whole-genome sequencing study of serum peptide levels: the Atherosclerosis Risk in Communities study. Human Molecular Genetics, 2017, 26, 3442-3450. | 2.9 | 25 |

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|---|---|---|---|
| 145 | REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. American Journal of Human Genetics, 2017, 101, 149-156. | 6.2 | 44 |
| 146 | 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. | 2.4 | 73 |
| 147 | Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245. | 2.9 | 261 |
| 148 | [P3â€“090]: THE ALZHEIMER's DISEASE SEQUENCING PROJECT (ADSP) DATA UPDATE 2017. Alzheimer's and Dementia, 2017, 13, P968. | 0.8 | 0 |
| 149 | SVachra: a tool to identify genomic structural variation in mate pair sequencing data containing inward and outward facing reads. BMC Genomics, 2017, 18, 691. | 2.8 | 7 |
| 150 | Whole exome sequencing in the Framingham Heart Study identifies rare variation in HYAL2 that influences platelet aggregation. Thrombosis and Haemostasis, 2017, 117, 1083-1092. | 3.4 | 11 |
| 151 | Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. Genome Medicine, 2017, 9, 95. | 8.2 | 37 |
| 152 | The house spider genome reveals an ancient whole-genome duplication during arachnid evolution. BMC Biology, 2017, 15, 62. | 3.8 | 286 |
| 153 | Hybrid de novo genome assembly and centromere characterization of the gray mouse lemur (Microcebus murinus). BMC Biology, 2017, 15, 110. | 3.8 | 53 |
| 154 | The gut mycobiome of the Human Microbiome Project healthy cohort. Microbiome, 2017, 5, 153. | 11.1 | 609 |
| 155 | Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016, 126, 762-778. | 8.2 | 82 |
| 156 | Whole exome sequencing identifies the first STRADA point mutation in a patient with polyhydramnios, megalencephaly, and symptomatic epilepsy syndrome (PMSE). American Journal of Medical Genetics, Part A, 2016, 170, 2181-2185. | 1.2 | 23 |
| 157 | A t(5;16) translocation is the likely driver of a syndrome with ambiguous genitalia, facial dysmorphism, intellectual disability, and speech delay. Journal of Physical Education and Sports Management, 2016, 2, a000703. | 1.2 | 1 |
| 158 | A potential founder variant inCARMIL2/RLTPRin three Norwegian families with warts, molluscum contagiosum, and T-cell dysfunction. Molecular Genetics & Genomic Medicine, 2016, 4, 604-616. | 1.2 | 59 |
| 159 | MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106. | 8.2 | 43 |
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