

# Elaine R Mardis

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5215952/publications.pdf>

Version: 2024-02-01

244  
papers

107,533  
citations

2802

94  
h-index

1316

224  
g-index

276  
all docs

276  
docs citations

276  
times ranked

115215  
citing authors

| #  | ARTICLE  | IF   | CITATIONS |
|----|--|------|-----------|
| 1  | Initial sequencing and analysis of the human genome. <i>Nature</i> , 2001, 409, 860-921.   | 27.8 | 21,074    |
| 2  | An obesity-associated gut microbiome with increased capacity for energy harvest. <i>Nature</i> , 2006, 444, 1027-1031.   | 27.8 | 10,136    |
| 3  | Initial sequencing and comparative analysis of the mouse genome. <i>Nature</i> , 2002, 420, 520-562.   | 27.8 | 6,319     |
| 4  | EGF receptor gene mutations are common in lung cancers from "never smokers" and are associated with sensitivity of tumors to gefitinib and erlotinib. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 13306-13311. | 7.1  | 4,106     |
| 5  | VarScan 2: Somatic mutation and copy number alteration discovery in cancer by exome sequencing. <i>Genome Research</i> , 2012, 22, 568-576.  | 5.5  | 4,086     |
| 6  | Integrated genomic characterization of endometrial carcinoma. <i>Nature</i> , 2013, 497, 67-73.  | 27.8 | 4,075     |
| 7  | Supervised Risk Predictor of Breast Cancer Based on Intrinsic Subtypes. <i>Journal of Clinical Oncology</i> , 2009, 27, 1160-1167.   | 1.6  | 3,730     |
| 8  | Somatic mutations affect key pathways in lung adenocarcinoma. <i>Nature</i> , 2008, 455, 1069-1075.  | 27.8 | 2,694     |
| 9  | Recurring Mutations Found by Sequencing an Acute Myeloid Leukemia Genome. <i>New England Journal of Medicine</i> , 2009, 361, 1058-1066.   | 27.0 | 2,009     |
| 10 | Clonal evolution in relapsed acute myeloid leukaemia revealed by whole-genome sequencing. <i>Nature</i> , 2012, 481, 506-510.  | 27.8 | 1,795     |
| 11 | Next-Generation DNA Sequencing Methods. <i>Annual Review of Genomics and Human Genetics</i> , 2008, 9, 387-402.  | 6.2  | 1,788     |
| 12 | <i>DNMT3A</i> Mutations in Acute Myeloid Leukemia. <i>New England Journal of Medicine</i> , 2010, 363, 2424-2433.  | 27.0 | 1,777     |
| 13 | Checkpoint blockade cancer immunotherapy targets tumour-specific mutant antigens. <i>Nature</i> , 2014, 515, 577-581.  | 27.8 | 1,705     |
| 14 | Age-related mutations associated with clonal hematopoietic expansion and malignancies. <i>Nature Medicine</i> , 2014, 20, 1472-1478.   | 30.7 | 1,533     |
| 15 | The Origin and Evolution of Mutations in Acute Myeloid Leukemia. <i>Cell</i> , 2012, 150, 264-278.   | 28.9 | 1,365     |
| 16 | BreakDancer: an algorithm for high-resolution mapping of genomic structural variation. <i>Nature Methods</i> , 2009, 6, 677-681.   | 19.0 | 1,322     |
| 17 | Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. <i>Science</i> , 2007, 316, 222-234.  | 12.6 | 1,283     |
| 18 | DNA sequencing of a cytogenetically normal acute myeloid leukaemia genome. <i>Nature</i> , 2008, 456, 66-72.   | 27.8 | 1,275     |

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|----|---|------|-----------|
| 19 | VarScan: variant detection in massively parallel sequencing of individual and pooled samples. <i>Bioinformatics</i> , 2009, 25, 2283-2285.  | 4.1  | 1,193     |
| 20 | Targetable Kinase-Activating Lesions in Ph-like Acute Lymphoblastic Leukemia. <i>New England Journal of Medicine</i> , 2014, 371, 1005-1015.  | 27.0 | 1,161     |
| 21 | A dendritic cell vaccine increases the breadth and diversity of melanoma neoantigen-specific T cells. <i>Science</i> , 2015, 348, 803-808.  | 12.6 | 1,139     |
| 22 | Genome remodelling in a basal-like breast cancer metastasis and xenograft. <i>Nature</i> , 2010, 464, 999-1005.   | 27.8 | 1,077     |
| 23 | Cancer exome analysis reveals a T-cell-dependent mechanism of cancer immunoediting. <i>Nature</i> , 2012, 482, 400-404.   | 27.8 | 1,075     |
| 24 | Characterizing the cancer genome in lung adenocarcinoma. <i>Nature</i> , 2007, 450, 893-898.  | 27.8 | 1,020     |
| 25 | Germline Mutations in Predisposition Genes in Pediatric Cancer. <i>New England Journal of Medicine</i> , 2015, 373, 2336-2346.  | 27.0 | 949       |
| 26 | Whole-genome analysis informs breast cancer response to aromatase inhibition. <i>Nature</i> , 2012, 486, 353-360.   | 27.8 | 922       |
| 27 | The Next-Generation Sequencing Revolution and Its Impact on Genomics. <i>Cell</i> , 2013, 155, 27-38.   | 28.9 | 856       |
| 28 | A decade's perspective on DNA sequencing technology. <i>Nature</i> , 2011, 470, 198-203.  | 27.8 | 731       |
| 29 | Whole-genome sequencing identifies genetic alterations in pediatric low-grade gliomas. <i>Nature Genetics</i> , 2013, 45, 602-612.  | 21.4 | 704       |
| 30 | Activating HER2 Mutations in HER2 Gene Amplification Negative Breast Cancer. <i>Cancer Discovery</i> , 2013, 3, 224-237.  | 9.4  | 697       |
| 31 | Clonal Architecture of Secondary Acute Myeloid Leukemia. <i>New England Journal of Medicine</i> , 2012, 366, 1090-1098.   | 27.0 | 688       |
| 32 | Role of TP53 mutations in the origin and evolution of therapy-related acute myeloid leukaemia. <i>Nature</i> , 2015, 518, 552-555.  | 27.8 | 685       |
| 33 | A Comparison of PAM50 Intrinsic Subtyping with Immunohistochemistry and Clinical Prognostic Factors in Tamoxifen-Treated Estrogen Receptor-Positive Breast Cancer. <i>Clinical Cancer Research</i> , 2010, 16, 5222-5232. | 7.0  | 676       |
| 34 | Genome analysis of the platypus reveals unique signatures of evolution. <i>Nature</i> , 2008, 453, 175-183.   | 27.8 | 657       |
| 35 | The genomic landscape of hypodiploid acute lymphoblastic leukemia. <i>Nature Genetics</i> , 2013, 45, 242-252.  | 21.4 | 588       |
| 36 | MuSiC: Identifying mutational significance in cancer genomes. <i>Genome Research</i> , 2012, 22, 1589-1598.   | 5.5  | 586       |

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|----|---|------|-----------|
| 37 | Recurrent Somatic Structural Variations Contribute to Tumorigenesis in Pediatric Osteosarcoma. <i>Cell Reports</i> , 2014, 7, 104-112.  | 6.4  | 583       |
| 38 | SomaticSniper: identification of somatic point mutations in whole genome sequencing data. <i>Bioinformatics</i> , 2012, 28, 311-317.  | 4.1  | 566       |
| 39 | C11orf95-RELA fusions drive oncogenic NF- $\kappa$ B signalling in ependymoma. <i>Nature</i> , 2014, 506, 451-455.  | 27.8 | 559       |
| 40 | Endocrine-Therapy-Resistant ESR1 Variants Revealed by Genomic Characterization of Breast-Cancer-Derived Xenografts. <i>Cell Reports</i> , 2013, 4, 1116-1130.                 | 6.4  | 539       |
| 41 | Next-Generation Sequencing Platforms. <i>Annual Review of Analytical Chemistry</i> , 2013, 6, 287-303.  | 5.4  | 519       |
| 42 | Tumor neoantigens: building a framework for personalized cancer immunotherapy. <i>Journal of Clinical Investigation</i> , 2015, 125, 3413-3421.                               | 8.2  | 502       |
| 43 | Convergent loss of PTEN leads to clinical resistance to a PI(3)K inhibitor. <i>Nature</i> , 2015, 518, 240-244.   | 27.8 | 486       |
| 44 | CIViC is a community knowledgebase for expert crowdsourcing the clinical interpretation of variants in cancer. <i>Nature Genetics</i> , 2017, 49, 170-174.                    | 21.4 | 460       |
| 45 | CREST maps somatic structural variation in cancer genomes with base-pair resolution. <i>Nature Methods</i> , 2011, 8, 652-654.  | 19.0 | 451       |
| 46 | DGIdb: mining the druggable genome. <i>Nature Methods</i> , 2013, 10, 1209-1210.  | 19.0 | 443       |
| 47 | A novel retinoblastoma therapy from genomic and epigenetic analyses. <i>Nature</i> , 2012, 481, 329-334.  | 27.8 | 442       |
| 48 | Genomic Landscape of Ewing Sarcoma Defines an Aggressive Subtype with Co-Association of <i>STAG2</i> and <i>TP53</i> Mutations. <i>Cancer Discovery</i> , 2014, 4, 1342-1353. | 9.4  | 418       |
| 49 | SciClone: Inferring Clonal Architecture and Tracking the Spatial and Temporal Patterns of Tumor Evolution. <i>PLoS Computational Biology</i> , 2014, 10, e1003665.            | 3.2  | 400       |
| 50 | Whole-genome sequencing and variant discovery in <i>C. elegans</i> . <i>Nature Methods</i> , 2008, 5, 183-188.  | 19.0 | 380       |
| 51 | Association of Age at Diagnosis and Genetic Mutations in Patients With Neuroblastoma. <i>JAMA - Journal of the American Medical Association</i> , 2012, 307, 1062.            | 7.4  | 379       |
| 52 | DGIdb 2.0: mining clinically relevant drug-gene interactions. <i>Nucleic Acids Research</i> , 2016, 44, D1036-D1044.  | 14.5 | 359       |
| 53 | Development and verification of the PAM50-based Prosigna breast cancer gene signature assay. <i>BMC Medical Genomics</i> , 2015, 8, 54.                                       | 1.5  | 352       |
| 54 | The emerging clinical relevance of genomics in cancer medicine. <i>Nature Reviews Clinical Oncology</i> , 2018, 15, 353-365.  | 27.6 | 351       |

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|----|---|------|-----------|
| 55 | pVAC-Seq: A genome-guided in silico approach to identifying tumor neoantigens. <i>Genome Medicine</i> , 2016, 8, 11.  | 8.2  | 350       |
| 56 | The landscape of somatic mutations in epigenetic regulators across 1,000 paediatric cancer genomes. <i>Nature Communications</i> , 2014, 5, 3630.   | 12.8 | 342       |
| 57 | Functional Heterogeneity of Genetically Defined Subclones in Acute Myeloid Leukemia. <i>Cancer Cell</i> , 2014, 25, 379-392.  | 16.8 | 330       |
| 58 | The Pediatric Cancer Genome Project. <i>Nature Genetics</i> , 2012, 44, 619-622.  | 21.4 | 315       |
| 59 | Association Between Mutation Clearance After Induction Therapy and Outcomes in Acute Myeloid Leukemia. <i>JAMA - Journal of the American Medical Association</i> , 2015, 314, 811.                      | 7.4  | 302       |
| 60 | Genetic alterations in uncommon low-grade neuroepithelial tumors: BRAF, FGFR1, and MYB mutations occur at high frequency and align with morphology. <i>Acta Neuropathologica</i> , 2016, 131, 833-845.  | 7.7  | 288       |
| 61 | DNA sequencing technologies: 2006–2016. <i>Nature Protocols</i> , 2017, 12, 213-218.  | 12.0 | 266       |
| 62 | A 50-Gene Intrinsic Subtype Classifier for Prognosis and Prediction of Benefit from Adjuvant Tamoxifen. <i>Clinical Cancer Research</i> , 2012, 18, 4465-4472.  | 7.0  | 258       |
| 63 | Contribution of systemic and somatic factors to clinical response and resistance to PD-L1 blockade in urothelial cancer: An exploratory multi-omic analysis. <i>PLoS Medicine</i> , 2017, 14, e1002309. | 8.4  | 256       |
| 64 | Integrated analysis of germline and somatic variants in ovarian cancer. <i>Nature Communications</i> , 2014, 5, 3156.   | 12.8 | 253       |
| 65 | Targeting Oxidative Stress in Embryonal Rhabdomyosarcoma. <i>Cancer Cell</i> , 2013, 24, 710-724.   | 16.8 | 252       |
| 66 | Immunogenomics of Hypermutated Glioblastoma: A Patient with Germline <i>POLE</i> Deficiency Treated with Checkpoint Blockade Immunotherapy. <i>Cancer Discovery</i> , 2016, 6, 1230-1236.               | 9.4  | 242       |
| 67 | Temporally Distinct PD-L1 Expression by Tumor and Host Cells Contributes to Immune Escape. <i>Cancer Immunology Research</i> , 2017, 5, 106-117.  | 3.4  | 236       |
| 68 | Use of Whole-Genome Sequencing to Diagnose a Cryptic Fusion Oncogene. <i>JAMA - Journal of the American Medical Association</i> , 2011, 305, 1577.  | 7.4  | 233       |
| 69 | Deregulation of DUX4 and ERG in acute lymphoblastic leukemia. <i>Nature Genetics</i> , 2016, 48, 1481-1489.   | 21.4 | 231       |
| 70 | The \$1,000 genome, the \$100,000 analysis?. <i>Genome Medicine</i> , 2010, 2, 84.  | 8.2  | 229       |
| 71 | Orthotopic patient-derived xenografts of paediatric solid tumours. <i>Nature</i> , 2017, 549, 96-100.   | 27.8 | 223       |
| 72 | The Dynamic Epigenetic Landscape of the Retina During Development, Reprogramming, and Tumorigenesis. <i>Neuron</i> , 2017, 94, 550-568.e10.   | 8.1  | 222       |

| #  | ARTICLE  | IF   | CITATIONS |
|----|--|------|-----------|
| 73 | Applications of Immunogenomics to Cancer. <i>Cell</i> , 2017, 168, 600-612.  | 28.9 | 198       |
| 74 | Cancer genome sequencing: a review. <i>Human Molecular Genetics</i> , 2009, 18, R163-R168.   | 2.9  | 185       |
| 75 | Clinical Significance of CTNNB1 Mutation and Wnt Pathway Activation in Endometrioid Endometrial Carcinoma. <i>Journal of the National Cancer Institute</i> , 2014, 106, .  | 6.3  | 182       |
| 76 | Optimizing Cancer Genome Sequencing and Analysis. <i>Cell Systems</i> , 2015, 1, 210-223.  | 6.2  | 174       |
| 77 | Analysis of the prostate cancer cell line LNCaP transcriptome using a sequencing-by-synthesis approach. <i>BMC Genomics</i> , 2006, 7, 246.  | 2.8  | 173       |
| 78 | Genomic landscape of paediatric adrenocortical tumours. <i>Nature Communications</i> , 2015, 6, 6302.  | 12.8 | 166       |
| 79 | Germline genetic variation in ETV6 and risk of childhood acute lymphoblastic leukaemia: a systematic genetic study. <i>Lancet Oncology</i> , The, 2015, 16, 1659-1666.   | 10.7 | 161       |
| 80 | Anticipating the 1,000 dollar genome. <i>Genome Biology</i> , 2006, 7, 112.  | 9.6  | 154       |
| 81 | The Genomic Landscape of Childhood and Adolescent Melanoma. <i>Journal of Investigative Dermatology</i> , 2015, 135, 816-823.  | 0.7  | 148       |
| 82 | Next-generation sequencing identifies the natural killer cell microRNA transcriptome. <i>Genome Research</i> , 2010, 20, 1590-1604.  | 5.5  | 144       |
| 83 | Next-Generation Sequencing Technologies. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2019, 9, a036798.  | 6.2  | 143       |
| 84 | Germline Genetic IKZF1 Variation and Predisposition to Childhood Acute Lymphoblastic Leukemia. <i>Cancer Cell</i> , 2018, 33, 937-948.e8.  | 16.8 | 142       |
| 85 | Ancestry estimation and control of population stratification for sequence-based association studies. <i>Nature Genetics</i> , 2014, 46, 409-415.   | 21.4 | 136       |
| 86 | pVACtools: A Computational Toolkit to Identify and Visualize Cancer Neoantigens. <i>Cancer Immunology Research</i> , 2020, 8, 409-420.   | 3.4  | 132       |
| 87 | Visualizing tumor evolution with the fishplot package for R. <i>BMC Genomics</i> , 2016, 17, 880.  | 2.8  | 131       |
| 88 | Integrated RNA and DNA sequencing reveals early drivers of metastatic breast cancer. <i>Journal of Clinical Investigation</i> , 2018, 128, 1371-1383.  | 8.2  | 126       |
| 89 | Comprehensive gene expression meta-analysis identifies signature genes that distinguish microglia from peripheral monocytes/macrophages in health and glioma. <i>Acta Neuropathologica Communications</i> , 2019, 7, 20. | 5.2  | 124       |
| 90 | Analysis of next-generation genomic data in cancer: accomplishments and challenges. <i>Human Molecular Genetics</i> , 2010, 19, R188-R196.   | 2.9  | 122       |

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|-----|--|------|-----------|
| 91  | TYK2 Protein-Coding Variants Protect against Rheumatoid Arthritis and Autoimmunity, with No Evidence of Major Pleiotropic Effects on Non-Autoimmune Complex Traits. <i>PLoS ONE</i> , 2015, 10, e0122271.                    | 2.5  | 120       |
| 92  | Clonal Architecture of Secondary Acute Myeloid Leukemia Defined by Single-Cell Sequencing. <i>PLoS Genetics</i> , 2014, 10, e1004462.  | 3.5  | 115       |
| 93  | Recurrent WNT pathway alterations are frequent in relapsed small cell lung cancer. <i>Nature Communications</i> , 2018, 9, 3787.   | 12.8 | 112       |
| 94  | Identification of Therapeutic Targets in Rhabdomyosarcoma through Integrated Genomic, Epigenomic, and Proteomic Analyses. <i>Cancer Cell</i> , 2018, 34, 411-426.e19.  | 16.8 | 106       |
| 95  | INTEGRATE-neo: a pipeline for personalized gene fusion neoantigen discovery. <i>Bioinformatics</i> , 2017, 33, 555-557.  | 4.1  | 105       |
| 96  | DoCM: a database of curated mutations in cancer. <i>Nature Methods</i> , 2016, 13, 806-807.  | 19.0 | 96        |
| 97  | Rapid expansion of preexisting nonleukemic hematopoietic clones frequently follows induction therapy for de novo AML. <i>Blood</i> , 2016, 127, 893-897.   | 1.4  | 94        |
| 98  | Sequencing a mouse acute promyelocytic leukemia genome reveals genetic events relevant for disease progression. <i>Journal of Clinical Investigation</i> , 2011, 121, 1445-1455.   | 8.2  | 91        |
| 99  | The prognostic effects of somatic mutations in ER-positive breast cancer. <i>Nature Communications</i> , 2018, 9, 3476.  | 12.8 | 89        |
| 100 | Tumor Evolution in Two Patients with Basal-like Breast Cancer: A Retrospective Genomics Study of Multiple Metastases. <i>PLoS Medicine</i> , 2016, 13, e1002174.   | 8.4  | 86        |
| 101 | A Phase I Trial of BKM120 (Buparlisib) in Combination with Fulvestrant in Postmenopausal Women with Estrogen Receptor-Positive Metastatic Breast Cancer. <i>Clinical Cancer Research</i> , 2016, 22, 1583-1591.              | 7.0  | 86        |
| 102 | A Surprising Cross-Species Conservation in the Genomic Landscape of Mouse and Human Oral Cancer Identifies a Transcriptional Signature Predicting Metastatic Disease. <i>Clinical Cancer Research</i> , 2014, 20, 2873-2884. | 7.0  | 84        |
| 103 | <i>Drosophila</i> Muller F Elements Maintain a Distinct Set of Genomic Properties Over 40 Million Years of Evolution. <i>G3: Genes, Genomes, Genetics</i> , 2015, 5, 719-740.  | 1.8  | 84        |
| 104 | Genome Modeling System: A Knowledge Management Platform for Genomics. <i>PLoS Computational Biology</i> , 2015, 11, e1004274.  | 3.2  | 83        |
| 105 | Long non-coding RNA RAMS11 promotes metastatic colorectal cancer progression. <i>Nature Communications</i> , 2020, 11, 2156.   | 12.8 | 83        |
| 106 | Neoantigens and genome instability: impact on immunogenomic phenotypes and immunotherapy response. <i>Genome Medicine</i> , 2019, 11, 71.  | 8.2  | 78        |
| 107 | New strategies and emerging technologies for massively parallel sequencing: applications in medical research. <i>Genome Medicine</i> , 2009, 1, 40.  | 8.2  | 75        |
| 108 | RNA Sequencing of Tumor-Associated Microglia Reveals Ccl5 as a Stromal Chemokine Critical for Neurofibromatosis-1 Glioma Growth. <i>Neoplasia</i> , 2015, 17, 776-788.   | 5.3  | 75        |

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|-----|--|------|-----------|
| 109 | Breast Cancer Neoantigens Can Induce CD8+ T-Cell Responses and Antitumor Immunity. <i>Cancer Immunology Research</i> , 2017, 5, 516-523.   | 3.4  | 74        |
| 110 | Integrated Analysis of RNA and DNA from the Phase III Trial CALGB 40601 Identifies Predictors of Response to Trastuzumab-Based Neoadjuvant Chemotherapy in HER2-Positive Breast Cancer. <i>Clinical Cancer Research</i> , 2018, 24, 5292-5304. | 7.0  | 73        |
| 111 | Inherited coding variants at the CDKN2A locus influence susceptibility to acute lymphoblastic leukaemia in children. <i>Nature Communications</i> , 2015, 6, 7553.   | 12.8 | 72        |
| 112 | Aromatase inhibition remodels the clonal architecture of estrogen-receptor-positive breast cancers. <i>Nature Communications</i> , 2016, 7, 12498.   | 12.8 | 69        |
| 113 | Quantitative Chromatographic Estimation of $\hat{\pm}$ -Amino-Acids. <i>Nature</i> , 1948, 161, 763-763.   | 27.8 | 66        |
| 114 | A Phase II Trial of Neoadjuvant MK-2206, an AKT Inhibitor, with Anastrozole in Clinical Stage II or III <i>PIK3CA</i> -Mutant ER-Positive and HER2-Negative Breast Cancer. <i>Clinical Cancer Research</i> , 2017, 23, 6823-6832.              | 7.0  | 66        |
| 115 | MYCN amplification and ATRX mutations are incompatible in neuroblastoma. <i>Nature Communications</i> , 2020, 11, 913.   | 12.8 | 66        |
| 116 | Genome sequencing and cancer. <i>Current Opinion in Genetics and Development</i> , 2012, 22, 245-250.  | 3.3  | 62        |
| 117 | A deep learning approach to automate refinement of somatic variant calling from cancer sequencing data. <i>Nature Genetics</i> , 2018, 50, 1735-1743.  | 21.4 | 62        |
| 118 | CMDS: a population-based method for identifying recurrent DNA copy number aberrations in cancer from high-resolution data. <i>Bioinformatics</i> , 2010, 26, 464-469.  | 4.1  | 59        |
| 119 | What is Finished, and Why Does it Matter. <i>Genome Research</i> , 2002, 12, 669-671.  | 5.5  | 57        |
| 120 | The Dynamic Genome and Transcriptome of the Human Fungal Pathogen <i>Blastomyces</i> and Close Relative <i>Emmonsia</i> . <i>PLoS Genetics</i> , 2015, 11, e1005493.   | 3.5  | 57        |
| 121 | Genetic risk factors for the development of osteonecrosis in children under age 10 treated for acute lymphoblastic leukemia. <i>Blood</i> , 2016, 127, 558-564.  | 1.4  | 56        |
| 122 | A pilot study of high-throughput, sequence-based mutational profiling of primary human acute myeloid leukemia cell genomes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003, 100, 14275-14280.   | 7.1  | 55        |
| 123 | Landscape of somatic single nucleotide variants and indels in colorectal cancer and impact on survival. <i>Nature Communications</i> , 2020, 11, 3644.   | 12.8 | 55        |
| 124 | Exome sequencing of case-unaffected-parents trios reveals recessive and de novo genetic variants in sporadic ALS. <i>Scientific Reports</i> , 2015, 5, 9124.   | 3.3  | 53        |
| 125 | Research-based PAM50 signature and long-term breast cancer survival. <i>Breast Cancer Research and Treatment</i> , 2020, 179, 197-206.   | 2.5  | 53        |
| 126 | Body Mass Index, PAM50 Subtype, and Outcomes in Node-Positive Breast Cancer: CALGB 9741 (Alliance). <i>Journal of the National Cancer Institute</i> , 2015, 107, .   | 6.3  | 52        |

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|-----|--|------|-----------|
| 127 | Detection of neoantigen-specific T cells following a personalized vaccine in a patient with glioblastoma. <i>Oncolmmunology</i> , 2019, 8, e1561106.   | 4.6  | 50        |
| 128 | RNA-seq reveals oligodendrocyte and neuronal transcripts in microglia relevant to central nervous system disease. <i>Glia</i> , 2015, 63, 531-548.   | 4.9  | 44        |
| 129 | Comprehensive genomic analysis reveals FLT3 activation and a therapeutic strategy for a patient with relapsed adult B-lymphoblastic leukemia. <i>Experimental Hematology</i> , 2016, 44, 603-613.  | 0.4  | 44        |
| 130 | The Impact of Next-Generation Sequencing on Cancer Genomics: From Discovery to Clinic. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2019, 9, a036269.  | 6.2  | 43        |
| 131 | Accounting for proximal variants improves neoantigen prediction. <i>Nature Genetics</i> , 2019, 51, 175-179.   | 21.4 | 43        |
| 132 | Applying next-generation sequencing to pancreatic cancer treatment. <i>Nature Reviews Gastroenterology and Hepatology</i> , 2012, 9, 477-486.  | 17.8 | 41        |
| 133 | A multiple myeloma-specific capture sequencing platform discovers novel translocations and frequent, risk-associated point mutations in IGLL5. <i>Blood Cancer Journal</i> , 2018, 8, 35.  | 6.2  | 41        |
| 134 | The clonal evolution of metastatic colorectal cancer. <i>Science Advances</i> , 2020, 6, eaay9691.   | 10.3 | 41        |
| 135 | cDNA Hybrid Capture Improves Transcriptome Analysis on Low-Input and Archived Samples. <i>Journal of Molecular Diagnostics</i> , 2014, 16, 440-451.  | 2.8  | 40        |
| 136 | Immunological ignorance is an enabling feature of the oligo-clonal T cell response to melanoma neoantigens. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 23662-23670.                               | 7.1  | 40        |
| 137 | Frontiers in cancer immunotherapy—a symposium report. <i>Annals of the New York Academy of Sciences</i> , 2021, 1489, 30-47.   | 3.8  | 39        |
| 138 | Melorheostosis: Exome sequencing of an associated dermatosis implicates postzygotic mosaicism of mutated KRAS. <i>Bone</i> , 2017, 101, 145-155.   | 2.9  | 37        |
| 139 | Integration of Sequence Data from a Consanguineous Family with Genetic Data from an Outbred Population Identifies PLB1 as a Candidate Rheumatoid Arthritis Risk Gene. <i>PLoS ONE</i> , 2014, 9, e87645.   | 2.5  | 34        |
| 140 | Impact of mutational profiles on response of primary oestrogen receptor-positive breast cancers to oestrogen deprivation. <i>Nature Communications</i> , 2016, 7, 13294.   | 12.8 | 34        |
| 141 | Characterization of the Genomic and Immunologic Diversity of Malignant Brain Tumors through Multisector Analysis. <i>Cancer Discovery</i> , 2022, 12, 154-171.   | 9.4  | 34        |
| 142 | A vertebrate case study of the quality of assemblies derived from next-generation sequences. <i>Genome Biology</i> , 2011, 12, R31.  | 9.6  | 32        |
| 143 | Whole genome analyses reveal no pathogenetic single nucleotide or structural differences between monozygotic twins discordant for amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2015, 16, 385-392. | 1.7  | 27        |
| 144 | Oral Cavity Squamous Cell Carcinoma Xenografts Retain Complex Genotypes and Intertumor Molecular Heterogeneity. <i>Cell Reports</i> , 2018, 24, 2167-2178.   | 6.4  | 26        |

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|-----|--|------|-----------|
| 145 | Immunotherapeutic Challenges for Pediatric Cancers. <i>Molecular Therapy - Oncolytics</i> , 2019, 15, 38-48.   | 4.4  | 26        |
| 146 | Somatic SLC35A2 mosaicism correlates with clinical findings in epilepsy brain tissue. <i>Neurology: Genetics</i> , 2020, 6, e460.  | 1.9  | 26        |
| 147 | <i>Caenorhabditis elegans glp-4</i> Encodes a Valyl Aminoacyl tRNA Synthetase. <i>G3: Genes, Genomes, Genetics</i> , 2015, 5, 2719-2728.   | 1.8  | 25        |
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