

Michael T Parsons

List of Publications by Year in descending order

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Version: 2024-02-01

59
papers

3,818
citations

201674

27
h-index

138484

58
g-index

69
all docs

69
docs citations

69
times ranked

5996
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|---|------|-----------|
| 1 | Breast Cancer Risk Genes " Association Analysis in More than 113,000 Women. <i>New England Journal of Medicine</i> , 2021, 384, 428-439. | 27.0 | 532 |
| 2 | Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. <i>Nature Genetics</i> , 2020, 52, 572-581. | 21.4 | 265 |
| 3 | Correlation of tumour BRAF mutations and <i>MLH1</i> methylation with germline mismatch repair (MMR) gene mutation status: a literature review assessing utility of tumour features for MMR variant classification. <i>Journal of Medical Genetics</i> , 2012, 49, 151-157. | 3.2 | 253 |
| 4 | Mutational spectrum in a worldwide study of 29,700 families with <i>BRCA1</i> or <i>BRCA2</i> mutations. <i>Human Mutation</i> , 2018, 39, 593-620. | 2.5 | 224 |
| 5 | Tumor Mismatch Repair Immunohistochemistry and DNA <i>MLH1</i> Methylation Testing of Patients With Endometrial Cancer Diagnosed at Age Younger Than 60 Years Optimizes Triage for Population-Level Germline Mismatch Repair Gene Mutation Testing. <i>Journal of Clinical Oncology</i> , 2014, 32, 90-100. | 1.6 | 195 |
| 6 | Germline pathogenic variants of 11 breast cancer genes in 7,051 Japanese patients and 11,241 controls. <i>Nature Communications</i> , 2018, 9, 4083. | 12.8 | 179 |
| 7 | <i>BRCA1</i> and <i>BRCA2</i> genetic testing "pitfalls and recommendations for managing variants of uncertain clinical significance. <i>Annals of Oncology</i> , 2015, 26, 2057-2065. | 1.2 | 163 |
| 8 | BRCA Challenge: BRCA Exchange as a global resource for variants in <i>BRCA1</i> and <i>BRCA2</i> . <i>PLoS Genetics</i> , 2018, 14, e1007752. | 3.5 | 148 |
| 9 | Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73. | 21.4 | 120 |
| 10 | Combined genetic and splicing analysis of <i>BRCA1</i> c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. <i>Human Molecular Genetics</i> , 2016, 25, 2256-2268. | 2.9 | 106 |
| 11 | Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019, 40, 1557-1578. | 2.5 | 102 |
| 12 | Refined histopathological predictors of <i>BRCA1</i> and <i>BRCA2</i> mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. <i>Breast Cancer Research</i> , 2014, 16, 3419. | 5.0 | 97 |
| 13 | Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019, 10, 1741. | 12.8 | 90 |
| 14 | Cancer Risks Associated With <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>Journal of Clinical Oncology</i> , 2022, 40, 1529-1541. | 1.6 | 90 |
| 15 | Shared heritability and functional enrichment across six solid cancers. <i>Nature Communications</i> , 2019, 10, 431. | 12.8 | 88 |
| 16 | Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of <i>BRCA1</i> and <i>BRCA2</i> pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1653-1666. | 2.4 | 82 |
| 17 | A Multifactorial Likelihood Model for MMR Gene Variant Classification Incorporating Probabilities Based on Sequence Bioinformatics and Tumor Characteristics: A Report from the Colon Cancer Family Registry. <i>Human Mutation</i> , 2013, 34, 200-209. | 2.5 | 81 |
| 18 | Novel diagnostic tool for prediction of variant spliceogenicity derived from a set of 395 combined in silico/in vitro studies: an international collaborative effort. <i>Nucleic Acids Research</i> , 2018, 46, 7913-7923. | 14.5 | 71 |

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|----|--|-----|-----------|
| 19 | Expansion of Cancer Risk Profile for <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>JAMA Oncology</i> , 2022, 8, 871. | 7.1 | 70 |
| 20 | Naturally occurring <i>BRCA2</i> alternative mRNA splicing events in clinically relevant samples. <i>Journal of Medical Genetics</i> , 2016, 53, 548-558. | 3.2 | 69 |
| 21 | A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. <i>Cancer Research</i> , 2018, 78, 5419-5430. | 0.9 | 54 |
| 22 | Adding In Silico Assessment of Potential Splice Aberration to the Integrated Evaluation of <i>BRCA</i> Gene Unclassified Variants. <i>Human Mutation</i> , 2016, 37, 627-639. | 2.5 | 52 |
| 23 | A plugin for the Ensembl Variant Effect Predictor that uses MaxEntScan to predict variant spliceogenicity. <i>Bioinformatics</i> , 2019, 35, 2315-2317. | 4.1 | 52 |
| 24 | Characterization of the Cancer Spectrum in Men With Germline <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>JAMA Oncology</i> , 2020, 6, 1218. | 7.1 | 48 |
| 25 | Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020, 80, 624-638. | 0.9 | 39 |
| 26 | The spectrum of <i>BRCA1</i> and <i>BRCA2</i> pathogenic sequence variants in Middle Eastern, North African, and South European countries. <i>Human Mutation</i> , 2019, 40, e1-e23. | 2.5 | 34 |
| 27 | Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. <i>Journal of Medical Genetics</i> , 2019, 56, 347-357. | 3.2 | 32 |
| 28 | Transcriptome-wide association study of breast cancer risk by estrogen receptor status. <i>Genetic Epidemiology</i> , 2020, 44, 442-468. | 1.3 | 32 |
| 29 | The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019, 5, 38. | 5.2 | 28 |
| 30 | Classification of variants of uncertain significance in <i>BRCA1</i> and <i>BRCA2</i> using personal and family history of cancer from individuals in a large hereditary cancer multigene panel testing cohort. <i>Genetics in Medicine</i> , 2020, 22, 701-708. | 2.4 | 28 |
| 31 | Evaluation of copy-number variants as modifiers of breast and ovarian cancer risk for <i>BRCA1</i> pathogenic variant carriers. <i>European Journal of Human Genetics</i> , 2017, 25, 432-438. | 2.8 | 26 |
| 32 | <i>BRCA1</i> and <i>BRCA2</i> pathogenic sequence variants in women of African origin or ancestry. <i>Human Mutation</i> , 2019, 40, 1781-1796. | 2.5 | 26 |
| 33 | Polygenic risk modeling for prediction of epithelial ovarian cancer risk. <i>European Journal of Human Genetics</i> , 2022, 30, 349-362. | 2.8 | 23 |
| 34 | The <i>BRCA2</i> c.68-7T>A variant is not pathogenic: A model for clinical calibration of spliceogenicity. <i>Human Mutation</i> , 2018, 39, 729-741. | 2.5 | 19 |
| 35 | Assessment of blind predictions of the clinical significance of <i>BRCA1</i> and <i>BRCA2</i> variants. <i>Human Mutation</i> , 2019, 40, 1546-1556. | 2.5 | 19 |
| 36 | Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 <i>BRCA1</i> and <i>BRCA2</i> mutation carriers. <i>British Journal of Cancer</i> , 2019, 121, 180-192. | 6.4 | 19 |

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|----|--|------|-----------|
| 37 | Contribution of mRNA Splicing to Mismatch Repair Gene Sequence Variant Interpretation. <i>Frontiers in Genetics</i> , 2020, 11, 798. | 2.3 | 19 |
| 38 | A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. <i>Nature Communications</i> , 2021, 12, 1078. | 12.8 | 19 |
| 39 | Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122. | 6.3 | 19 |
| 40 | Breast cancer risks associated with missense variants in breast cancer susceptibility genes. <i>Genome Medicine</i> , 2022, 14, 51. | 8.2 | 19 |
| 41 | Multifactorial Likelihood Assessment of BRCA1 and BRCA2 Missense Variants Confirms That BRCA1:c.122A>G(p.His41Arg) Is a Pathogenic Mutation. <i>PLoS ONE</i> , 2014, 9, e86836. | 2.5 | 17 |
| 42 | The predictive ability of the 313 variant-based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. <i>Genetics in Medicine</i> , 2021, 23, 1726-1737. | 2.4 | 16 |
| 43 | <i>BRCA1</i> and <i>BRCA2</i> 5' noncoding region variants identified in breast cancer patients alter promoter activity and protein binding. <i>Human Mutation</i> , 2018, 39, 2025-2039. | 2.5 | 15 |
| 44 | Considerations in assessing germline variant pathogenicity using cosegregation analysis. <i>Genetics in Medicine</i> , 2020, 22, 2052-2059. | 2.4 | 15 |
| 45 | Consequences of germline variation disrupting the constitutional translational initiation codon start sites of <i>MLH1</i> and <i>BRCA2</i> : Use of potential alternative start sites and implications for predicting variant pathogenicity. <i>Molecular Carcinogenesis</i> , 2015, 54, 513-522. | 2.7 | 14 |
| 46 | DNA methylation profiling to assess pathogenicity of BRCA1 unclassified variants in breast cancer. <i>Epigenetics</i> , 2015, 10, 1121-1132. | 2.7 | 12 |
| 47 | Comprehensive Assessment of BARD1 Messenger Ribonucleic Acid Splicing With Implications for Variant Classification. <i>Frontiers in Genetics</i> , 2019, 10, 1139. | 2.3 | 10 |
| 48 | Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants. <i>Genetics in Medicine</i> , 2022, 24, 119-129. | 2.4 | 10 |
| 49 | The splicing effect of variants at branchpoint elements in cancer genes. <i>Genetics in Medicine</i> , 2022, 24, 398-409. | 2.4 | 9 |
| 50 | Considerations for using population frequency data in germline variant interpretation: Cancer syndrome genes as a model. <i>Human Mutation</i> , 2021, 42, 530-536. | 2.5 | 8 |
| 51 | Substantial evidence for the clinical significance of missense variant BRCA1 c.5309G>Tp.(Gly1770Val). <i>Breast Cancer Research and Treatment</i> , 2018, 172, 497-503. | 2.5 | 7 |
| 52 | Reply to J. Moline et al. <i>Journal of Clinical Oncology</i> , 2014, 32, 2278-2279. | 1.6 | 5 |
| 53 | Association of germline variation with the survival of women with BRCA1/2 pathogenic variants and breast cancer. <i>Npj Breast Cancer</i> , 2020, 6, 44. | 5.2 | 5 |
| 54 | GFP-Fragment Reassembly Screens for the Functional Characterization of Variants of Uncertain Significance in Protein Interaction Domains of the BRCA1 and BRCA2 Genes. <i>Cancers</i> , 2019, 11, 151. | 3.7 | 4 |

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|----|--|-----|-----------|
| 55 | TRACEBACK: Testing of Historical Tubo-Ovarian Cancer Patients for Hereditary Risk Genes as a Cancer Prevention Strategy in Family Members. <i>Journal of Clinical Oncology</i> , 2022, , JCO2102108. | 1.6 | 3 |
| 56 | Value of the loss of heterozygosity to BRCA1 variant classification. <i>Npj Breast Cancer</i> , 2022, 8, 9. | 5.2 | 2 |
| 57 | Federated analysis of BRCA1 and BRCA2 variation in a Japanese cohort. <i>Cell Genomics</i> , 2022, 2, 100109. | 6.5 | 1 |
| 58 | Under-ascertainment of breast cancer susceptibility gene carriers in a cohort of New Zealand female breast cancer patients. <i>Breast Cancer Research and Treatment</i> , 2021, 185, 583-590. | 2.5 | 0 |
| 59 | Classification of genetic variants in hereditary cancer genes. , 2021, , 349-387. | | 0 |