Bryony A Thompson

List of Publications by Year
in descending order

50	2,013	394421 19	254184 43
papers	citations	h-index	g-index

56	56	56	3543
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A novel candidate gene in autosomal dominant facial pruritus. Clinical and Experimental Dermatology, 2022, 47, 184-186.	1.3	2
2	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
3	Arrhythmic Phenotypes Are a Defining Feature of Dilated Cardiomyopathy-Associated SCN5A Variants: A Systematic Review. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003432.	3.6	13
4	TRACEBACK: Testing of Historical Tubo-Ovarian Cancer Patients for Hereditary Risk Genes as a Cancer Prevention Strategy in Family Members. Journal of Clinical Oncology, 2022, , JCO2102108.	1.6	3
5	Real world outcomes and implementation pathways of exome sequencing in an adult genetic department. Genetics in Medicine, 2022, , .	2.4	4
6	Predictive functional assayâ€based classification of PMS2 variants in Lynch syndrome. Human Mutation, 2022, , .	2.5	1
7	Rivaroxaban in the treatment of <scp>TEK</scp> â€related venous malformation. Australasian Journal of Dermatology, 2022, , .	0.7	2
8	Comprehensive evaluation and efficient classification of BRCA1 RING domain missense substitutions. American Journal of Human Genetics, 2022, 109, 1153-1174.	6.2	6
9	Quantitative modeling. , 2021, , 41-58.		1
10	A homozygous truncating variant in GDF9 in siblings with primary ovarian insufficiency. Journal of Assisted Reproduction and Genetics, 2021, 38, 1539-1543.	2.5	3
11	Genetic Dominant Variants in STUB1, Segregating in Families with SCA48, Display In Vitro Functional Impairments Indistinctive from Recessive Variants Associated with SCAR16. International Journal of Molecular Sciences, 2021, 22, 5870.	4.1	10
12	Genetic variants associated with inherited cardiovascular disorders among 13,131 asymptomatic older adults of European descent. Npj Genomic Medicine, 2021, 6, 51.	3.8	11
13	Genomic Risk Prediction for Breast Cancer in Older Women. Cancers, 2021, 13, 3533.	3.7	6
14	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. American Journal of Human Genetics, 2021, 108, 1551-1557.	6.2	36
15	Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. Cancers, 2020, 12, 1799.	3.7	15
16	Contribution of mRNA Splicing to Mismatch Repair Gene Sequence Variant Interpretation. Frontiers in Genetics, 2020, 11, 798.	2.3	19
17	FANCM c5791C>T stopgain mutation (rs144567652) is a familial colorectal cancer risk factor. Molecular Genetics & Genomic Medicine, 2020, 8, e1532.	1.2	5
18	A novel ribosomal protein <scp>S20</scp> variant in a family with unexplained colorectal cancer and polyposis. Clinical Genetics, 2020, 97, 943-944.	2.0	14

#	Article	IF	Citations
19	Medically actionable pathogenic variants in a population of 13,131 healthy elderly individuals. Genetics in Medicine, 2020, 22, 1883-1886.	2.4	20
20	A novel AFG3L2 mutation close to AAA domain leads to aberrant OMA1 and OPA1 processing in a family with optic atrophy. Acta Neuropathologica Communications, 2020, 8, 93.	5.2	10
21	Two integrated and highly predictive functional analysis-based procedures for the classification of MSH6 variants in Lynch syndrome. Genetics in Medicine, 2020, 22, 847-856.	2.4	16
22	Tumour characteristics provide evidence for germline mismatch repair missense variant pathogenicity. Journal of Medical Genetics, 2020, 57, 62-69.	3.2	11
23	A plugin for the Ensembl Variant Effect Predictor that uses MaxEntScan to predict variant spliceogenicity. Bioinformatics, 2019, 35, 2315-2317.	4.1	52
24	Determining the clinical validity of hereditary colorectal cancer and polyposis susceptibility genes using the Clinical Genome Resource Clinical Validity Framework. Genetics in Medicine, 2019, 21, 1507-1516.	2.4	19
25	A functional assayâ€“based procedure to classify mismatch repair gene variants in Lynch syndrome. Genetics in Medicine, 2019, 21, 1486-1496.	2.4	36
26	Pancreatic cancer as a sentinel for hereditary cancer predisposition. BMC Cancer, 2018, 18, 697.	2.6	29
27	Elucidating the clinical significance of two PMS2 missense variants coexisting in a family fulfilling hereditary cancer criteria. Familial Cancer, 2017, 16, 501-507.	1.9	3
28	Elucidating the molecular basis of MSH2â€deficient tumors by combined germline and somatic analysis. International Journal of Cancer, 2017, 141, 1365-1380.	5.1	26
29	Panel sequencing of 264 candidate susceptibility genes and segregation analysis in a cohort of non-BRCA1, non-BRCA2 breast cancer families. Breast Cancer Research and Treatment, 2017, 166, 937-949.	2.5	16
30	Assessment of the InSiGHT Interpretation Criteria for the Clinical Classification of 24MLH1andMSH2Gene Variants. Human Mutation, 2017, 38, 64-77.	2.5	29
31	Understanding the Pathogenicity of Noncoding Mismatch Repair Gene Promoter Variants in Lynch Syndrome. Human Mutation, 2016, 37, 417-426.	2.5	10
32	Evaluation of CADD Scores in Curated Mismatch Repair Gene Variants Yields a Model for Clinical Validation and Prioritization. Human Mutation, 2015, 36, 712-719.	2.5	39
33	Microsatellite Instability Use in Mismatch Repair Gene Sequence Variant Classification. Genes, 2015, 6, 150-162.	2.4	7
34	Detailed characterization of <scp>MLH1</scp> p.<scp>D41H</scp> and p.<scp>N710D</scp> variants coexisting in a Lynch syndrome family with conserved <scp>MLH1</scp> expression tumors. Clinical Genetics, 2015, 87, 543-548.	2.0	6
35	Consequences of germline variation disrupting the constitutional translational initiation codon start sites of MLH1 and BRCA2 : Use of potential alternative start sites and implications for predicting variant pathogenicity. Molecular Carcinogenesis, 2015, 54, 513-522.	2.7	14
36	A review of mismatch repair gene transcripts: issues for interpretation of <scp>mRNA</scp> splicing assays. Clinical Genetics, 2015, 87, 100-108.	2.0	27

#	Article	IF	Citations
37	Nucleosome positioning is unaltered at MLH1 splice site mutations in cells derived from Lynch syndrome patients. Clinical Epigenetics, 2014, 6, 32.	4.1	0
38	Reply to J. Moline et al. Journal of Clinical Oncology, 2014, 32, 2278-2279.	1.6	5
39	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. Nature Genetics, 2014, 46, 107-115.	21.4	410
40	Tumor Mismatch Repair Immunohistochemistry and DNA MLH1 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. Journal of Clinical Oncology, 2014, 32, 90-100.	1.6	195
41	The InSiGHT database: utilizing 100Âyears of insights into Lynch Syndrome. Familial Cancer, 2013, 12, 175-180.	1.9	100
42	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. Human Mutation, 2013, 34, 200-209.	2.5	81
43	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	2.5	80
44	BRCA1 R1699Q variant displaying ambiguous functional abrogation confers intermediate breast and ovarian cancer risk. Journal of Medical Genetics, 2012, 49, 525-532.	3.2	97
45	Correlation of tumour BRAF mutations andMLH1methylation with germline mismatch repair (MMR) gene mutation status: a literature review assessing utility of tumour features for MMR variant classification. Journal of Medical Genetics, 2012, 49, 151-157.	3.2	253
46	Pancreatic Cancer and a Novel MSH2 Germline Alteration. Pancreas, 2011, 40, 1138-1140.	1.1	13
47	Mutation deep within an intron of MSH2 causes Lynch syndrome. Familial Cancer, 2011, 10, 297-301.	1.9	43
48	Splicing and multifactorial analysis of intronic BRCA1 and BRCA2 sequence variants identifies clinically significant splicing aberrations up to 12 nucleotides from the intron/exon boundary. Human Mutation, 2011, 32, 678-687.	2.5	74
49	Detection of splicing aberrations caused by BRCA1 and BRCA2 sequence variants encoding missense substitutions: implications for prediction of pathogenicity. Human Mutation, 2010, 31, E1484-E1505.	2.5	86
50	Use of DNAâ€“Damaging Agents and RNA Pooling to Assess Expression Profiles Associated with BRCA1 and BRCA2 Mutation Status in Familial Breast Cancer Patients. PLoS Genetics, 2010, 6, e1000850.	3.5	9