Sean V Tavtigian

List of Publications by Year
in descending order

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59 papers	7,271 citations	100601 38 h-index	150775 59 g-index
69 all docs	69 docs citations	69 times ranked	10227 citing authors

#	Article	IF	Citations
1	Comprehensive evaluation and efficient classification of BRCA1 RING domain missense substitutions. American Journal of Human Genetics, 2022, 109, 1153-1174.	2.6	6
2	Mobile element insertions and associated structural variants in longitudinal breast cancer samples. Scientific Reports, 2021, 11, 13020.	1.6	3
3	An updated quantitative model to classify missense variants in the TP53 gene: A novel multifactorial strategy. Human Mutation, 2021, 42, 1351-1361.	1.1	7
4	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. Genome Medicine, 2020, 12, 3.	3.6	312
5	Targeted germline sequencing of patients with three or more primary melanomas reveals high rate of pathogenic variants. Melanoma Research, 2020, 30, 247-251.	0.6	5
6	Fitting a naturally scaled point system to the ACMG/AMP variant classification guidelines. Human Mutation, 2020, 41, 1734-1737.	1.1	105
7	Contribution of mRNA Splicing to Mismatch Repair Gene Sequence Variant Interpretation. Frontiers in Genetics, 2020, 11, 798.	1.1	19
8	FANCM c5791C>T stopgain mutation (rs144567652) is a familial colorectal cancer risk factor. Molecular Genetics & Genomic Medicine, 2020, 8, e1532.	0.6	5
9	A novel ribosomal protein <scp>S20</scp> variant in a family with unexplained colorectal cancer and polyposis. Clinical Genetics, 2020, 97, 943-944.	1.0	14
10	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.	1.1	16
11	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	1.1	102
12	Assessing the performance of in silico methods for predicting the pathogenicity of variants in the gene CHEK2, among Hispanic females with breast cancer. Human Mutation, 2019, 40, 1612-1622.	1.1	8
13	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357.	1.5	32
14	A quantitative model to predict pathogenicity of missense variants in the TP53 gene. Human Mutation, 2019, 40, 788-800.	1.1	21
15	Genetic predisposition to colorectal cancer: syndromes, genes, classification of genetic variants and implications for precision medicine. Journal of Pathology, 2019, 247, 574-588.	2.1	131
16	A functional assayâ€“based procedure to classify mismatch repair gene variants in Lynch syndrome. Genetics in Medicine, 2019, 21, 1486-1496.	1.1	36
17	Modeling the ACMG/AMP variant classification guidelines as a Bayesian classification framework. Genetics in Medicine, 2018, 20, 1054-1060.	1.1	366
18	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	1.5	148

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19	Improved, ACMG-compliant, in silico prediction of pathogenicity for missense substitutions encoded by TP53 variants. Human Mutation, 2018, 39, 1061-1069.	1.1	29
20	Pancreatic cancer as a sentinel for hereditary cancer predisposition. BMC Cancer, 2018, 18, 697.	1.1	29
21	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.	1.1	16
22	Screening for germline BRCA1, BRCA2, TP53 and CHEK2 mutations in families at-risk for hereditary breast cancer identified in a population-based study from Southern Brazil. Genetics and Molecular Biology, 2016, 39, 210-222.	0.6	21
23	Adding In Silico Assessment of Potential Splice Aberration to the Integrated Evaluation of BRCA Gene Unclassified Variants. Human Mutation, 2016, 37, 627-639.	1.1	52
24	Combined genetic and splicing analysis of BRCA1 c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. Human Molecular Genetics, 2016, 25, 2256-2268.	1.4	106
25	No evidence that protein truncating variants inBRIP1are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	1.5	94
26	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	13.9	764
27	BRCA1 Circos: a visualisation resource for functional analysis of missense variants. Journal of Medical Genetics, 2015, 52, 224-230.	1.5	32
28	Growing recognition of the role forÂrare missense substitutions in breast cancer susceptibility. Biomarkers in Medicine, 2014, 8, 589-603.	0.6	24
29	A unified test of linkage analysis and rare-variant association for analysis of pedigree sequence data. Nature Biotechnology, 2014, 32, 663-669.	9.4	93
30	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.	9.4	410
31	Rare key functional domain missense substitutions in MRE11A, RAD50, and NBNcontribute to breast cancer susceptibility: results from a Breast Cancer Family Registry case-control mutation-screening study. Breast Cancer Research, 2014, 16, R58.	2.2	99
32	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.	1.1	81
33	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	1.1	80
34	BRCA1/2 Sequence Variants of Uncertain Significance: A Primer for Providers to Assist in Discussions and in Medical Management. Oncologist, 2013, 18, 518-524.	1.9	76
35	Pathological assessment of mismatch repair gene variants in Lynch syndrome: Past, present, and future. Human Mutation, 2012, 33, 1617-1625.	1.1	60
36	Integrated evaluation of CHD7 missense substitutions for CHARGE syndrome clinical genetics. Human Mutation, 2012, 33, v-v.	1.1	0

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37	A review of a multifactorial probability-based model for classification of BRCA1 and BRCA2 variants of uncertain significance (VUS). Human Mutation, 2012, 33, 8-21.	1.1	190
38	ENIGMA-Evidence-based network for the interpretation of germline mutant alleles: An international initiative to evaluate risk and clinical significance associated with sequence variation in BRCA1 and BRCA2 genes. Human Mutation, 2012, 33, 2-7.	1.1	269
39	Classification of missense substitutions in the BRCA genes: A database dedicated to Ex-UVs. Human Mutation, 2012, 33, 22-28.	1.1	65
40	Rare, evolutionarily unlikely missense substitutions in CHEK2contribute to breast cancer susceptibility: results from a breast cancer family registry case-control mutation-screening study. Breast Cancer Research, 2011, 13, R6.	2.2	74
41	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.	1.1	74
42	Comparison of Programs for in silico Assessment of Missense Substitutions. Human Mutation, 2011, 32, v-v.	1.1	2
43	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.	1.1	86
44	71: The Breast Cancer Genes IARC Database: a tool to improve evaluation of BRCA1 and BRCA2 uncertain sequence variants. Bulletin Du Cancer, 2010, 97, S61.	0.6	0
45	ClassifyingMLH1andMSH2variants using bioinformatic prediction, splicing assays, segregation, and tumor characteristics. Human Mutation, 2009, 30, 757-770.	1.1	60
46	Rare, Evolutionarily Unlikely Missense Substitutions in ATM Confer Increased Risk of Breast Cancer. American Journal of Human Genetics, 2009, 85, 427-446.	2.6	165
47	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results. Human Mutation, 2008, 29, 1282-1291.	1.1	782
48	In silico analysis of missense substitutions using sequence-alignment based methods. Human Mutation, 2008, 29, 1327-1336.	1.1	181
49	Classification of rare missense substitutions, using risk surfaces, with genetic- and molecular-epidemiology applications. Human Mutation, 2008, 29, 1342-1354.	1.1	209
50	Assessing pathogenicity: overview of results from the IARC Unclassified Genetic Variants Working Group. Human Mutation, 2008, 29, 1261-1264.	1.1	79
51	Pathogenicity of the BRCA1 missense variant M1775K is determined by the disruption of the BRCT phosphopeptide-binding pocket: a multi-modal approach. European Journal of Human Genetics, 2008, 16, 820-832.	1.4	42
52	Clinical Classification ofBRCA1andBRCA2DNA Sequence Variants: The Value of Cytokeratin Profiles and Evolutionary Analysisâ€"A Report From the kConFab Investigators. Journal of Clinical Oncology, 2008, 26, 1657-1663.	0.8	72
53	Functional Assays for Classification ofBRCA2Variants of Uncertain Significance. Cancer Research, 2008, 68, 3523-3531.	0.4	108
54	A Systematic Genetic Assessment of 1,433 Sequence Variants of Unknown Clinical Significance in the BRCA1 and BRCA2 Breast Cancerâ€"Predisposition Genes. American Journal of Human Genetics, 2007, 81, 873-883.	2.6	416

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55	Identification of BRCA1 missense substitutions that confer partial functional activity: potential moderate risk variants?. Breast Cancer Research, 2007, 9, R82.	2.2	58
56	Computational approaches for predicting the biological effect of p53 missense mutations: a comparison of three sequence analysis based methods. Nucleic Acids Research, 2006, 34, 1317-1325.	6.5	295
57	Genetic and Histopathologic Evaluation of BRCA1 and BRCA2 DNA Sequence Variants of Unknown Clinical Significance. Cancer Research, 2006, 66, 2019-2027.	0.4	153
58	Functional evaluation and cancer risk assessment of BRCA2 unclassified variants. Cancer Research, 2005, 65, 417-26.	0.4	97
59	Integrated Evaluation of DNA Sequence Variants of Unknown Clinical Significance: Application to BRCA1 and BRCA2. American Journal of Human Genetics, 2004, 75, 535-544.	2.6	351