Sean V Tavtigian

List of Publications by Year
in descending order

59	7,271	87888 38	133252 59
papers	citations	h-index	g-index

69	69	69	9527
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results. Human Mutation, 2008, 29, 1282-1291.	2.5	782
2	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	27.0	764
3	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.	6.2	416
4	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
5	Modeling the ACMG/AMP variant classification guidelines as a Bayesian classification framework. Genetics in Medicine, 2018, 20, 1054-1060.	2.4	366
6	Integrated Evaluation of DNA Sequence Variants of Unknown Clinical Significance: Application to BRCA1 and BRCA2. American Journal of Human Genetics, 2004, 75, 535-544.	6.2	351
7	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. Genome Medicine, 2020, 12, 3.	8.2	312
8	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.	14.5	295
9	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.	2.5	269
10	Classification of rare missense substitutions, using risk surfaces, with genetic- and molecular-epidemiology applications. Human Mutation, 2008, 29, 1342-1354.	2.5	209
11	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.	2.5	190
12	In silico analysis of missense substitutions using sequence-alignment based methods. Human Mutation, 2008, 29, 1327-1336.	2.5	181
13	Rare, Evolutionarily Unlikely Missense Substitutions in ATM Confer Increased Risk of Breast Cancer. American Journal of Human Genetics, 2009, 85, 427-446.	6.2	165
14	Genetic and Histopathologic Evaluation ofBRCA1andBRCA2DNA Sequence Variants of Unknown Clinical Significance. Cancer Research, 2006, 66, 2019-2027.	0.9	153
15	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	3.5	148
16	Genetic predisposition to colorectal cancer: syndromes, genes, classification of genetic variants and implications for precision medicine. Journal of Pathology, 2019, 247, 574-588.	4.5	131
17	Functional Assays for Classification ofBRCA2Variants of Uncertain Significance. Cancer Research, 2008, 68, 3523-3531.	0.9	108
18	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.	2.9	106

#	Article	IF	Citations
19	Fitting a naturally scaled point system to the ACMG/AMP variant classification guidelines. Human Mutation, 2020, 41, 1734-1737.	2.5	105
20	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.	2.5	102
21	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.	5.0	99
22	Functional evaluation and cancer risk assessment of BRCA2 unclassified variants. Cancer Research, 2005, 65, 417-26.	0.9	97
23	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.	3.2	94
24	A unified test of linkage analysis and rare-variant association for analysis of pedigree sequence data. Nature Biotechnology, 2014, 32, 663-669.	17.5	93
25	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
26	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
27	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	2.5	80
28	Assessing pathogenicity: overview of results from the IARC Unclassified Genetic Variants Working Group. Human Mutation, 2008, 29, 1261-1264.	2.5	79
29	BRCA1/2 Sequence Variants of Uncertain Significance: A Primer for Providers to Assist in Discussions and in Medical Management. Oncologist, 2013, 18, 518-524.	3.7	76
30	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.	5.0	74
31	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
32	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.	1.6	72
33	Classification of missense substitutions in the BRCA genes: A database dedicated to Ex-UVs. Human Mutation, 2012, 33, 22-28.	2.5	65
34	ClassifyingMLH1andMSH2variants using bioinformatic prediction, splicing assays, segregation, and tumor characteristics. Human Mutation, 2009, 30, 757-770.	2.5	60
35	Pathological assessment of mismatch repair gene variants in Lynch syndrome: Past, present, and future. Human Mutation, 2012, 33, 1617-1625.	2.5	60
36	Identification of BRCA1 missense substitutions that confer partial functional activity: potential moderate risk variants?. Breast Cancer Research, 2007, 9, R82.	5.0	58

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37	Adding In Silico Assessment of Potential Splice Aberration to the Integrated Evaluation of BRCA Gene Unclassified Variants. Human Mutation, 2016, 37, 627-639.	2.5	52
38	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.	2.8	42
39	A functional assayâ€"based procedure to classify mismatch repair gene variants in Lynch syndrome. Genetics in Medicine, 2019, 21, 1486-1496.	2.4	36
40	BRCA1 Circos: a visualisation resource for functional analysis of missense variants. Journal of Medical Genetics, 2015, 52, 224-230.	3.2	32
41	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357.	3.2	32
42	Improved, ACMG-compliant, in silico prediction of pathogenicity for missense substitutions encoded by TP53 variants. Human Mutation, 2018, 39, 1061-1069.	2.5	29
43	Pancreatic cancer as a sentinel for hereditary cancer predisposition. BMC Cancer, 2018, 18, 697.	2.6	29
44	Growing recognition of the role forÂrare missense substitutions in breast cancer susceptibility. Biomarkers in Medicine, 2014, 8, 589-603.	1.4	24
45	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.	1.3	21
46	A quantitative model to predict pathogenicity of missense variants in the TP53 gene. Human Mutation, 2019, 40, 788-800.	2.5	21
47	Contribution of mRNA Splicing to Mismatch Repair Gene Sequence Variant Interpretation. Frontiers in Genetics, 2020, 11, 798.	2.3	19
48	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
49	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
50	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
51	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.	2.5	8
52	An updated quantitative model to classify missense variants in the TP53 gene: A novel multifactorial strategy. Human Mutation, 2021, 42, 1351-1361.	2.5	7
53	Comprehensive evaluation and efficient classification of BRCA1 RING domain missense substitutions. American Journal of Human Genetics, 2022, 109, 1153-1174.	6.2	6
54	Targeted germline sequencing of patients with three or more primary melanomas reveals high rate of pathogenic variants. Melanoma Research, 2020, 30, 247-251.	1.2	5

#	Article	IF	Citations
55	FANCM c5791C>T stopgain mutation (rs144567652) is a familial colorectal cancer risk factor. Molecular Genetics & Genomic Medicine, 2020, 8, e1532.	1.2	5
56	Mobile element insertions and associated structural variants in longitudinal breast cancer samples. Scientific Reports, 2021, 11, 13020.	3.3	3
57	Comparison of Programs for in silico Assessment of Missense Substitutions. Human Mutation, 2011, 32, v-v.	2.5	2
58	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.	1.6	0
59	Integrated evaluation of CHD7 missense substitutions for CHARGE syndrome clinical genetics. Human Mutation, 2012, 33, v-v.	2.5	0