Nicola Whiffin

List of Publications by Year
in descending order

59	12,033	147726 31	133188 59
papers	citations	h-index	g-index

85	85	85	24376
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
2	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614
3	Withdrawal of pharmacological treatment for heart failure in patients with recovered dilated cardiomyopathy (TRED-HF): an open-label, pilot, randomised trial. Lancet, The, 2019, 393, 61-73.	6.3	379
4	Using high-resolution variant frequencies to empower clinical genome interpretation. Genetics in Medicine, 2017, 19, 1151-1158.	1.1	355
5	Adaptation and validation of the ACMG/AMP variant classification framework for MYH7-associated inherited cardiomyopathies: recommendations by ClinGenâ€™s Inherited Cardiomyopathy Expert Panel. Genetics in Medicine, 2018, 20, 351-359.	1.1	283
6	Evaluating the Clinical Validity of Hypertrophic Cardiomyopathy Genes. Circulation Genomic and Precision Medicine, 2019, 12, e002460.	1.6	267
7	Common variation near CDKN1A, POLD3 and SHROOM2 influences colorectal cancer risk. Nature Genetics, 2012, 44, 770-776.	9.4	210
8	Genetic Variants Associated With Cancer Therapyâ€“Induced Cardiomyopathy. Circulation, 2019, 140, 31-41.	1.6	195
9	Multiple Common Susceptibility Variants near BMP Pathway Loci GREM1, BMP4, and BMP2 Explain Part of the Missing Heritability of Colorectal Cancer. PLoS Genetics, 2011, 7, e1002105.	1.5	188
10	Capture Hi-C identifies the chromatin interactome of colorectal cancer risk loci. Nature Communications, 2015, 6, 6178.	5.8	186
11	Genetic Etiology for Alcohol-Induced Cardiac Toxicity. Journal of the American College of Cardiology, 2018, 71, 2293-2302.	1.2	182
12	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. Nature Genetics, 2021, 53, 128-134.	9.4	155
13	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398.	1.6	148
14	Transcript expression-aware annotation improves rare variant interpretation. Nature, 2020, 581, 452-458.	13.7	142
15	Putative cis-regulatory drivers in colorectal cancer. Nature, 2014, 512, 87-90.	13.7	136
16	Identification of susceptibility loci for colorectal cancer in a genome-wide meta-analysis. Human Molecular Genetics, 2014, 23, 4729-4737.	1.4	128
17	Evaluating drug targets through human loss-of-function genetic variation. Nature, 2020, 581, 459-464.	13.7	115
18	A new GWAS and meta-analysis with 1000Genomes imputation identifies novel risk variants for colorectal cancer. Scientific Reports, 2015, 5, 10442.	1.6	109

#	Article	IF	Citations
19	Characterising the loss-of-function impact of 5â€™ untranslated region variants in 15,708 individuals. Nature Communications, 2020, 11, 2523.	5.8	99
20	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93
21	Quantitative approaches to variant classification increase the yield and precision of genetic testing in Mendelian diseases: the case of hypertrophic cardiomyopathy. Genome Medicine, 2019, 11, 5.	3.6	90
22	Phenotype and Clinical Outcomes of TitinÂCardiomyopathy. Journal of the American College of Cardiology, 2017, 70, 2264-2274.	1.2	86
23	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. Journal of Cardiovascular Translational Research, 2016, 9, 3-11.	1.1	80
24	The effect of LRRK2 loss-of-function variants in humans. Nature Medicine, 2020, 26, 869-877.	15.2	79
25	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. Genetics in Medicine, 2018, 20, 1246-1254.	1.1	75
26	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	3.6	65
27	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. Genetics in Medicine, 2021, 23, 47-58.	1.1	57
28	Truncating Variants in Titin Independently Predict Early Arrhythmias in Patients With Dilated Cardiomyopathy. Journal of the American College of Cardiology, 2017, 69, 2466-2468.	1.2	56
29	Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardiomyopathy. Journal of the American College of Cardiology, 2021, 78, 1097-1110.	1.2	55
30	The TERT variant rs2736100 is associated with colorectal cancer risk. British Journal of Cancer, 2012, 107, 1001-1008.	2.9	50
31	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
32	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	2.6	42
33	A high-resolution map of human RNA translation. Molecular Cell, 2022, 82, 2885-2899.e8.	4.5	37
34	Meta-analysis of genome-wide association studies identifies common susceptibility polymorphisms for colorectal and endometrial cancer near SH2B3 and TSHZ1. Scientific Reports, 2015, 5, 17369.	1.6	35
35	Three-dimensional cardiovascular imaging-genetics: a mass univariate framework. Bioinformatics, 2018, 34, 97-103.	1.8	34
36	Association of Titin-Truncating Genetic Variants With Life-threatening Cardiac Arrhythmias in Patients With Dilated Cardiomyopathy and Implanted Defibrillators. JAMA Network Open, 2019, 2, e196520.	2.8	33

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37	MLH1-93G > A is a risk factor for MSI colorectal cancer. Carcinogenesis, 2011, 32, 1157-1161.	1.3	32
38	Spatiotemporal organization of Aurora-B by APC/CCdh1 after mitosis coordinates cell spreading via FHOD1. Journal of Cell Science, 2013, 126, 2845-56.	1.2	32
39	Annotating high-impact 5â€²untranslated region variants with the UTRannotator. Bioinformatics, 2021, 37, 1171-1173.	1.8	27
40	Recurrent Coding Sequence Variation Explains Only A Small Fraction of the Genetic Architecture of Colorectal Cancer. Scientific Reports, 2015, 5, 16286.	1.6	24
41	Precision Phenotyping of Dilated Cardiomyopathy Using Multidimensional Data. Journal of the American College of Cardiology, 2022, 79, 2219-2232.	1.2	24
42	Relationship between 16 susceptibility loci and colorectal cancer phenotype in 3146 patients. Carcinogenesis, 2012, 33, 108-112.	1.3	22
43	Deciphering the genetic architecture of low-penetrance susceptibility to colorectal cancer. Human Molecular Genetics, 2013, 22, 5075-5082.	1.4	19
44	Genetic Studies of Hypertrophic Cardiomyopathy in Singaporeans Identify Variants in TNNI3 and TNNT2 That Are Common in Chinese Patients. Circulation Genomic and Precision Medicine, 2020, 13, 424-434.	1.6	18
45	Correspondence on â€œACMG SF v3.0 list for reporting of secondary findings in clinical exome and genome sequencing: a policy statement of the American College of Medical Genetics and Genomics (ACMG)â€•byÂMiller etÂal. Genetics in Medicine, 2022, 24, 744-746.	1.1	17
46	Using High-Resolution Variant Frequencies Empowers Clinical Genome Interpretation and Enables Investigation of Genetic Architecture. American Journal of Human Genetics, 2019, 104, 187-190.	2.6	15
47	Architecture of Inherited Susceptibility to Colorectal Cancer: A Voyage of Discovery. Genes, 2014, 5, 270-284.	1.0	13
48	MRSD: A quantitative approach for assessing suitability of RNA-seq in the investigation of mis-splicing in Mendelian disease. American Journal of Human Genetics, 2022, 109, 210-222.	2.6	12
49	A Retrospective Observational Study of the Relationship between Single Nucleotide Polymorphisms Associated with the Risk of Developing Colorectal Cancer and Survival. PLoS ONE, 2015, 10, e0117816.	1.1	10
50	Improving the Understanding of Genetic Variants in Rare Disease With Large-scale Reference Populations. JAMA - Journal of the American Medical Association, 2019, 322, 1305.	3.8	7
51	Moderate excess alcohol consumption and adverse cardiac remodelling in dilated cardiomyopathy. Heart, 2022, 108, 619-625.	1.2	6
52	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. Npj Genomic Medicine, 2020, 5, 46.	1.7	5
53	Quantifying evidence toward pathogenicity for rare phenotypes: The case of succinate dehydrogenase genes, SDHB and SDHD. Genetics in Medicine, 2022, 24, 41-50.	1.1	5
54	143â€…Clinical and Genetic Characteristics of Familial Dilated Cardiomyopathy in a Large UK Prospective Cohort: Abstract 143 Table 1. Heart, 2016, 102, A103-A104.	1.2	4

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55	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. Circulation, 2021, 144, 754-757.	1.6	4
56	125â€…Evaluation of titin cardiomyopathy in patients with dilated cardiomyopathy reveals a blunted hypertrophic response, an early arrhythmic risk and a significant interaction with alcohol. Heart, 2017, 103, A95.1-A95.	1.2	1
57	121â€…Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , .		1
58	142â€…Effects of Truncating Variants in Titin on Cardiac Phenotype and Left Ventricular Remodelling in Dilated Cardiomyopathy. Heart, 2016, 102, A102-A103.	1.2	0
59	Abstract 407: Targeted Hi-C and integrative analyses reveal functionality of colorectal cancer risk loci. , 2014, , .		0