Rachel J Buchan

List of Publications by Year
in descending order

26 papers	1,476 citations	840119 11 h-index	839053 18 g-index
30 all docs	30 docs citations	30 times ranked	3387 citing authors

#	Article	IF	Citations
1	Direct and indirect effect of the COVID-19 pandemic on patients with cardiomyopathy. Open Heart, 2022, 9, e001918.	0.9	3
2	New Variant With a Previously Unrecognized Mechanism of Pathogenicity in Hypertrophic Cardiomyopathy. Circulation, 2021, 144, 754-757.	1.6	4
3	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
4	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
5	The Egyptian Collaborative Cardiac Genomics (ECCO-GEN) Project: defining a healthy volunteer cohort. Npj Genomic Medicine, 2020, 5, 46.	1.7	5
6	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
7	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	13.7	338
8	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
9	121â€…Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , .		1
10	CardioClassifier: disease- and gene-specific computational decision support for clinical genome interpretation. Genetics in Medicine, 2018, 20, 1246-1254.	1.1	75
11	Three-dimensional cardiovascular imaging-genetics: a mass univariate framework. Bioinformatics, 2018, 34, 97-103.	1.8	34
12	5â€…Defining the effects of genetic variation using machine learning analysis of CMRS: a study in hypertrophic cardiomyopathy and in a healthy population. , 2018, , .		0
13	Defining the genetic architecture of hypertrophic cardiomyopathy: re-evaluating the role of non-sarcomeric genes. European Heart Journal, 2017, 38, ehw603.	1.0	142
14	Phenotype and Clinical Outcomes of TitinÂCardiomyopathy. Journal of the American College of Cardiology, 2017, 70, 2264-2274.	1.2	86
15	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
16	209â€…Whole Exome Sequencing Identifies Genetic Cause of Histiocytoid Cardiomyopathy. Heart, 2016, 102, A138.2-A139.	1.2	0
17	P35â€…Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: Phenotypes linked by truncating variants inNDUFB11. Heart, 2016, 102, A18.2-A18.	1.2	0
18	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

#	Article	IF	Citations
19	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
20	Development of a Comprehensive Sequencing Assay for Inherited Cardiac Condition Genes. Journal of Cardiovascular Translational Research, 2016, 9, 3-11.	1.1	80
21	175â€…Aortopathy-causing mutations increase aortic stiffness in healthy individuals. Heart, 2015, 101, A99.1-A99.	1.2	1
22	171â€…The genetic signature in ischaemic heart disease with myocardial infarction (MI) and significant left ventricular (LV) dysfunction. Heart, 2015, 101, A97-A98.	1.2	0
23	163â€…Integrated allelic, transcriptional, and phenotypic dissection of the cardiac effects of titin variation in health and disease. Heart, 2015, 101, A93.1-A93.	1.2	0
24	76â€…Comprehensive Assessment of Rare Genetic Variation in Dilated Cardiomyopathy Genes in Patients and Controls: Abstract 76 Table 1. Heart, 2015, 101, A41.2-A42.	1.2	0
25	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. Science Translational Medicine, 2015, 7, 270ra6.	5.8	375
26	95â€…Identification Of Likely Pathogenic Variants In Patients With Bicuspid Aortic Valve: Correlation Of Complex Genotype With A More Severe Aortic Phenotype. Heart, 2014, 100, A55-A56.	1.2	4