Francesco Mazzarotto

List of Publications by Year
in descending order

32 papers	2,882 citations	361413 20 h-index	501196 28 g-index
38 all docs	38 docs citations	38 times ranked	4191 citing authors

#	Article	IF	Citations
1	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. European Heart Journal, 2022, 43, 1500-1510.	2.2	57
2	Quantifying evidence toward pathogenicity for rare phenotypes: The case of succinate dehydrogenase genes, SDHB and SDHD. Genetics in Medicine, 2022, 24, 41-50.	2.4	5
3	Disease Progression of Hypertrophic Cardiomyopathy: Modeling Using Machine Learning. JMIR Medical Informatics, 2022, 10, e30483.	2.6	5
4	Enhancing rare variant interpretation in inherited arrhythmias through quantitative analysis of consortium disease cohorts and population controls. Genetics in Medicine, 2021, 23, 47-58.	2.4	57
5	Computational prediction of protein subdomain stability in MYBPC3 enables clinical risk stratification in hypertrophic cardiomyopathy and enhances variant interpretation. Genetics in Medicine, 2021, 23, 1281-1287.	2.4	11
6	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. Circulation, 2021, 144, 7-19.	1.6	213
7	Arrhythmogenic potential of myocardial disarray in hypertrophic cardiomyopathy: genetic basis, functional consequences and relation to sudden cardiac death. Europace, 2021, 23, 985-995.	1.7	11
8	A machine learning-based risk stratification model for ventricular tachycardia and heart failure in hypertrophic cardiomyopathy. Computers in Biology and Medicine, 2021, 135, 104648.	7.0	27
9	Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardiomyopathy. Journal of the American College of Cardiology, 2021, 78, 1097-1110.	2.8	55
10	Shared genetic pathways contribute to risk of hypertrophic and dilated cardiomyopathies with opposite directions of effect. Nature Genetics, 2021, 53, 128-134.	21.4	155
11	Systematic large-scale assessment of the genetic architecture of left ventricular noncompaction reveals diverse etiologies. Genetics in Medicine, 2021, 23, 856-864.	2.4	45
12	Sex-related differences in exercise performance and outcome of patients with hypertrophic cardiomyopathy. European Journal of Preventive Cardiology, 2020, 27, 1821-1831.	1.8	15
13	Temporal Trend of Age at Diagnosis in Hypertrophic Cardiomyopathy. Circulation: Heart Failure, 2020, 13, e007230.	3.9	48
14	Spatial and Functional Distribution of MYBPC3 Pathogenic Variants and Clinical Outcomes in Patients With Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, 396-405.	3.6	47
15	Advantages and Perils of Clinical Whole-Exome and Whole-Genome Sequencing in Cardiomyopathy. Cardiovascular Drugs and Therapy, 2020, 34, 241-253.	2.6	21
16	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398.	1.6	148
17	Contemporary Insights Into the Genetics of Hypertrophic Cardiomyopathy: Toward a New Era in Clinical Testing?. Journal of the American Heart Association, 2020, 9, e015473.	3.7	42
18	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. Genetics in Medicine, 2019, 21, 133-143.	2.4	25

#	Article	IF	Citations
19	Defining the diagnostic effectiveness of genes for inclusion in panels: the experience of two decades of genetic testing for hypertrophic cardiomyopathy at a single center. Genetics in Medicine, 2019, 21, 284-292.	2.4	54
20	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.	8.2	90
21	121â€...Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , .		1
22	Comparison of longâ€term outcome in anthracyclineâ€related versus idiopathic dilated cardiomyopathy: a single centre experience. European Journal of Heart Failure, 2018, 20, 898-906.	7.1	54
23	Reassessment of Mendelian gene pathogenicity using 7,855 cardiomyopathy cases and 60,706 reference samples. Genetics in Medicine, 2017, 19, 192-203.	2.4	585
24	Titin-truncating variants affect heart function in disease cohorts and the general population. Nature Genetics, 2017, 49, 46-53.	21.4	255
25	142â€...Effects of Truncating Variants in Titin on Cardiac Phenotype and Left Ventricular Remodelling in Dilated Cardiomyopathy. Heart, 2016, 102, A102-A103.	2.9	0
26	143â€...Clinical and Genetic Characteristics of Familial Dilated Cardiomyopathy in a Large UK Prospective Cohort: Abstract 143 Table 1. Heart, 2016, 102, A103-A104.	2.9	4
27	Shared Genetic Predisposition in Peripartum and Dilated Cardiomyopathies. New England Journal of Medicine, 2016, 374, 233-241.	27.0	432
28	163â€...Integrated allelic, transcriptional, and phenotypic dissection of the cardiac effects of titin variation in health and disease. Heart, 2015, 101, A93.1-A93.	2.9	0
29	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.	2.9	0
30	ZBTB17 (MIZ1) Is Important for the Cardiac Stress Response and a Novel Candidate Gene for Cardiomyopathy and Heart Failure. Circulation: Cardiovascular Genetics, 2015, 8, 643-652.	5.1	12
31	Integrated allelic, transcriptional, and phenomic dissection of the cardiac effects of titin truncations in health and disease. Science Translational Medicine, 2015, 7, 270ra6.	12.4	375
32	FineSplice, enhanced splice junction detection and quantification: a novel pipeline based on the assessment of diverse RNA-Seq alignment solutions. Nucleic Acids Research, 2014, 42, e71-e71.	14.5	30