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	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
2	Shared Genetic Predisposition in Peripartum and Dilated Cardiomyopathies. New England Journal of Medicine, 2016, 374, 233-241.	27.0	432
3	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
4	Titin-truncating variants affect heart function in disease cohorts and the general population. Nature Genetics, 2017, 49, 46-53.	21.4	255
5	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. Circulation, 2021, 144, 7-19.	1.6	213
6	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
7	Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. Circulation, 2020, 141, 387-398.	1.6	148
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.	8.2	90
9	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
10	Evaluation of gene validity for CPVT and short QT syndrome in sudden arrhythmic death. European Heart Journal, 2022, 43, 1500-1510.	2.2	57
11	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
12	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
13	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
14	Temporal Trend of Age at Diagnosis in Hypertrophic Cardiomyopathy. Circulation: Heart Failure, 2020, 13, e007230.	3.9	48
15	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
16	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
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	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

#	Article	IF	Citations
19	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
20	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. Genetics in Medicine, 2019, 21, 133-143.	2.4	25
21	Advantages and Perils of Clinical Whole-Exome and Whole-Genome Sequencing in Cardiomyopathy. Cardiovascular Drugs and Therapy, 2020, 34, 241-253.	2.6	21
22	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
23	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
24	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
25	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
26	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
27	Disease Progression of Hypertrophic Cardiomyopathy: Modeling Using Machine Learning. JMIR Medical Informatics, 2022, 10, e30483.	2.6	5
28	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
29	121â€...Re-evaluating the genetic contribution of monogenic dilated cardiomyopathy. , 2019, , .		1
30	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
31	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
32	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