Ivan Macciocca

List of Publications by Year
in descending order

21	1,307	567281 15	713466 21
papers	citations	h-index	g-index

22	22	22	2970
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. Genetics in Medicine, 2016, 18, 1090-1096.	2.4	332
2	Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. JAMA Pediatrics, 2017, 171, 855.	6.2	252
3	Prospective comparison of the cost-effectiveness of clinical whole-exome sequencing with that of usual care overwhelmingly supports early use and reimbursement. Genetics in Medicine, 2017, 19, 867-874.	2.4	194
4	Cpipe: a shared variant detection pipeline designed for diagnostic settings. Genome Medicine, 2015, 7, 68.	8.2	78
5	A comprehensive evaluation of myocardial fibrosis in hypertrophic cardiomyopathy with cardiac magnetic resonance imaging: linking genotype with fibrotic phenotype. European Heart Journal Cardiovascular Imaging, 2014, 15, 1108-1116.	1.2	77
6	Diffuse Ventricular Fibrosis on Cardiac Magnetic Resonance Imaging Associates With Ventricular Tachycardia in Patients With Hypertrophic Cardiomyopathy. Journal of Cardiovascular Electrophysiology, 2016, 27, 571-580.	1.7	56
7	ALPK3-deficient cardiomyocytes generated from patient-derived induced pluripotent stem cells and mutant human embryonic stem cells display abnormal calcium handling and establish that ALPK3 deficiency underlies familial cardiomyopathy. European Heart Journal, 2016, 37, 2586-2590.	2.2	49
8	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 318-325.	3.7	36
9	Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution. American Journal of Human Genetics, 2021, 108, 1551-1557.	6.2	36
10	Genetic Testing in Inherited Heart Diseases. Heart Lung and Circulation, 2020, 29, 505-511.	0.4	34
11	Readiness of clinical genetic healthcare professionals to provide genomic medicine: An Australian census. Journal of Genetic Counseling, 2019, 28, 367-377.	1.6	29
12	Prospective Evaluation of the Utility of Whole Exome Sequencing in Dilated Cardiomyopathy. Journal of the American Heart Association, 2020, 9, e013346.	3.7	28
13	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119.	2.7	25
14	A clinically driven variant prioritization framework outperforms purely computational approaches for the diagnostic analysis of singleton WES data. European Journal of Human Genetics, 2017, 25, 1268-1272.	2.8	24
15	Genetic Counseling in the Era of Genomics: What's all the Fuss about?. Journal of Genetic Counseling, 2018, 27, 1010-1021.	1.6	18
16	A novel approach to offering additional genomic findingsâ€"A protocol to test a twoâ€step approach in the healthcare system. Journal of Genetic Counseling, 2019, 28, 388-397.	1.6	14
17	Clinical and laboratory reporting impact of ACMG-AMP and modified ClinGen variant classification frameworks in MYH7-related cardiomyopathy. Genetics in Medicine, 2021, 23, 1108-1115.	2.4	14
18	Epigenetics and DOHaD: how translation to predictive testing will require a better public understanding. Journal of Developmental Origins of Health and Disease, 2022, 13, 424-430.	1.4	4

#	Article	IF	Citations
19	Public knowledge and opinion of epigenetics and epigenetic concepts. Journal of Developmental Origins of Health and Disease, 2021, , 1-10.	1.4	3
20	Experience of Asian males communicating cardiac genetic risk within the family. Journal of Community Genetics, 2018, 9, 293-303.	1.2	2
21	Investigation of current models of care for genetic heart disease in Australia: A national clinical audit. International Journal of Cardiology, 2021, 330, 128-134.	1.7	2