

Marzia De Bortoli

List of Publications by Year in descending order

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Version: 2024-02-01

26
papers

1,160
citations

516561

16
h-index

580701

25
g-index

26
all docs

26
docs citations

26
times ranked

1696
citing authors

#	ARTICLE	IF	CITATIONS
1	GCN5 contributes to intracellular lipid accumulation in human primary cardiac stromal cells from patients affected by Arrhythmogenic cardiomyopathy. <i>Journal of Cellular and Molecular Medicine</i> , 2022, 26, 3687-3701.	1.6	3
2	Circulating miR-185-5p as a Potential Biomarker for Arrhythmogenic Right Ventricular Cardiomyopathy. <i>Cells</i> , 2021, 10, 2578.	1.8	5
3	Genetic and Metabolic Determinants of Atrial Fibrillation in a General Population Sample: The CHRIS Study. <i>Biomolecules</i> , 2021, 11, 1663.	1.8	5
4	Novel Missense Variant in <i>MYL2</i> Gene Associated With Hypertrophic Cardiomyopathy Showing High Incidence of Restrictive Physiology. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e002824.	1.6	6
5	A targeted next-generation gene panel reveals a novel heterozygous nonsense variant in the TP63 gene in patients with arrhythmogenic cardiomyopathy. <i>Heart Rhythm</i> , 2019, 16, 773-780.	0.3	15
6	KCND3 potassium channel gene variant confers susceptibility to electrocardiographic early repolarization pattern. <i>JCI Insight</i> , 2019, 4, .	2.3	15
7	New <i>FIG4</i> gene mutations causing aggressive <i>ALS</i> . <i>European Journal of Neurology</i> , 2018, 25, e41-e42.	1.7	14
8	Inherited Cardiomyopathies: From Genotype to Phenotype. <i>Journal of Clinical & Medical Genomics</i> , 2018, 06, .	0.1	0
9	Whole-Exome Sequencing Identifies Pathogenic Variants in <i>TJP1</i> Gene Associated With Arrhythmogenic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002123.	1.6	38
10	Large Genomic Rearrangements of Desmosomal Genes in Italian Arrhythmogenic Cardiomyopathy Patients. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2017, 10, .	2.1	35
11	Co-inheritance of mutations associated with arrhythmogenic cardiomyopathy and hypertrophic cardiomyopathy. <i>European Journal of Human Genetics</i> , 2017, 25, 1165-1169.	1.4	10
12	Mutations in NEBL encoding the cardiac Z-disk protein nebulin are associated with various cardiomyopathies. <i>Archives of Medical Science</i> , 2016, 2, 263-278.	0.4	26
13	Arrhythmogenic right-ventricular cardiomyopathy. <i>Journal of Cardiovascular Medicine</i> , 2016, 17, 399-407.	0.6	16
14	A founder <i>MYBPC3</i> mutation results in HCM with a high risk of sudden death after the fourth decade of life. <i>Journal of Medical Genetics</i> , 2015, 52, 338-347.	1.5	41
15	The novel S59P mutation in the TNFRSF1A gene identified in an adult onset TNF receptor associated periodic syndrome (TRAPS) constitutively activates NF- κ B pathway. <i>Arthritis Research and Therapy</i> , 2015, 17, 93.	1.6	43
16	Autosomal dominant lateral temporal epilepsy (ADLTE): Novel structural and single-nucleotide <i>LGI1</i> mutations in families with predominant visual auras. <i>Epilepsy Research</i> , 2015, 110, 132-138.	0.8	17
17	Arrhythmogenic cardiomyopathy: a disease of intercalated discs. <i>Cell and Tissue Research</i> , 2015, 360, 491-500.	1.5	41
18	Homozygous Desmocollin-2 Mutations and Arrhythmogenic Cardiomyopathy. <i>American Journal of Cardiology</i> , 2015, 116, 1245-1251.	0.7	38

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19	Desmin Mutations and Arrhythmogenic Right Ventricular Cardiomyopathy. <i>American Journal of Cardiology</i> , 2013, 111, 400-405.	0.7	62
20	Identification of a PKP2 gene deletion in a family with arrhythmogenic right ventricular cardiomyopathy. <i>European Journal of Human Genetics</i> , 2013, 21, 1226-1231.	1.4	39
21	Compound and Digenic Heterozygosity Predicts Lifetime Arrhythmic Outcome and Sudden Cardiac Death in Desmosomal Gene-Related Arrhythmogenic Right Ventricular Cardiomyopathy. <i>Circulation: Cardiovascular Genetics</i> , 2013, 6, 533-542.	5.1	209
22	Mutations in the area composita protein β -catenin are associated with arrhythmogenic right ventricular cardiomyopathy. <i>European Heart Journal</i> , 2013, 34, 201-210.	1.0	175
23	Clinical phenotype and diagnosis of arrhythmogenic right ventricular cardiomyopathy in pediatric patients carrying desmosomal gene mutations. <i>Heart Rhythm</i> , 2011, 8, 1686-1695.	0.3	66
24	The p.A897KfsX4 frameshift variation in desmocollin-2 is not a causative mutation in arrhythmogenic right ventricular cardiomyopathy. <i>European Journal of Human Genetics</i> , 2010, 18, 776-782.	1.4	19
25	Multiple mutations in desmosomal proteins encoding genes in arrhythmogenic right ventricular cardiomyopathy/dysplasia. <i>Heart Rhythm</i> , 2010, 7, 22-29.	0.3	161
26	Missense mutations in Desmocollin-2 N-terminus, associated with arrhythmogenic right ventricular cardiomyopathy, affect intracellular localization of desmocollin-2 in vitro. <i>BMC Medical Genetics</i> , 2007, 8, 65.	2.1	61