

Marzia De Bortoli

List of Publications by Year in descending order

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Version: 2024-02-01

26
papers

1,160
citations

516710

16
h-index

580821

25
g-index

26
all docs

26
docs citations

26
times ranked

1696
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	Mutations in the area composita protein β -catenin are associated with arrhythmogenic right ventricular cardiomyopathy. <i>European Heart Journal</i> , 2013, 34, 201-210.	2.2	175
3	Multiple mutations in desmosomal proteins encoding genes in arrhythmogenic right ventricular cardiomyopathy/dysplasia. <i>Heart Rhythm</i> , 2010, 7, 22-29.	0.7	161
4	Clinical phenotype and diagnosis of arrhythmogenic right ventricular cardiomyopathy in pediatric patients carrying desmosomal gene mutations. <i>Heart Rhythm</i> , 2011, 8, 1686-1695.	0.7	66
5	Desmin Mutations and Arrhythmogenic Right Ventricular Cardiomyopathy. <i>American Journal of Cardiology</i> , 2013, 111, 400-405.	1.6	62
6	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
7	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.	3.5	43
8	A founder MYBPC3 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.	3.2	41
9	Arrhythmogenic cardiomyopathy: a disease of intercalated discs. <i>Cell and Tissue Research</i> , 2015, 360, 491-500.	2.9	41
10	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.	2.8	39
11	Homozygous Desmocollin-2 Mutations and Arrhythmogenic Cardiomyopathy. <i>American Journal of Cardiology</i> , 2015, 116, 1245-1251.	1.6	38
12	Whole-Exome Sequencing Identifies Pathogenic Variants in TJP1 Gene Associated With Arrhythmogenic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002123.	3.6	38
13	Large Genomic Rearrangements of Desmosomal Genes in Italian Arrhythmogenic Cardiomyopathy Patients. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2017, 10, .	4.8	35
14	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.	0.9	26
15	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.	2.8	19
16	Autosomal dominant lateral temporal epilepsy (ADLTE): Novel structural and single-nucleotide LGI1 mutations in families with predominant visual auras. <i>Epilepsy Research</i> , 2015, 110, 132-138.	1.6	17
17	Arrhythmogenic right-ventricular cardiomyopathy. <i>Journal of Cardiovascular Medicine</i> , 2016, 17, 399-407.	1.5	16
18	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.	0.7	15

#	ARTICLE	IF	CITATIONS
19	KCND3 potassium channel gene variant confers susceptibility to electrocardiographic early repolarization pattern. JCI Insight, 2019, 4, .	5.0	15
20	New <i>FIG4</i> gene mutations causing aggressive ALS. European Journal of Neurology, 2018, 25, e41-e42.	3.3	14
21	Co-inheritance of mutations associated with arrhythmogenic cardiomyopathy and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2017, 25, 1165-1169.	2.8	10
22	Novel Missense Variant in <i>MYL2</i> Gene Associated With Hypertrophic Cardiomyopathy Showing High Incidence of Restrictive Physiology. Circulation Genomic and Precision Medicine, 2020, 13, e002824.	3.6	6
23	Circulating miR-185-5p as a Potential Biomarker for Arrhythmogenic Right Ventricular Cardiomyopathy. Cells, 2021, 10, 2578.	4.1	5
24	Genetic and Metabolic Determinants of Atrial Fibrillation in a General Population Sample: The CHRIS Study. Biomolecules, 2021, 11, 1663.	4.0	5
25	GCN5 contributes to intracellular lipid accumulation in human primary cardiac stromal cells from patients affected by Arrhythmogenic cardiomyopathy. Journal of Cellular and Molecular Medicine, 2022, 26, 3687-3701.	3.6	3
26	Inherited Cardiomyopathies: From Genotype to Phenotype. Journal of Clinical & Medical Genomics, 2018, 06, .	0.1	0