Marzia De Bortoli

List of Publications by Year
in descending order

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26	1,160	516710 16	580821 25
papers	citations	h-index	g-index

26	26	26	1696
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	Circulating miR-185-5p as a Potential Biomarker for Arrhythmogenic Right Ventricular Cardiomyopathy. Cells, 2021, 10, 2578.	4.1	5
3	Genetic and Metabolic Determinants of Atrial Fibrillation in a General Population Sample: The CHRIS Study. Biomolecules, 2021, 11, 1663.	4.0	5
4	Novel Missense Variant in MYL2 Gene Associated With Hypertrophic Cardiomyopathy Showing High Incidence of Restrictive Physiology. Circulation Genomic and Precision Medicine, 2020, 13, e002824.	3.6	6
5	A targeted next-generation gene panel reveals a novel heterozygous nonsense variant in the TP63 gene in patients with arrhythmogenic cardiomyopathy. Heart Rhythm, 2019, 16, 773-780.	0.7	15
6	KCND3 potassium channel gene variant confers susceptibility to electrocardiographic early repolarization pattern. JCI Insight, 2019, 4, .	5.0	15
7	New <scp>FIG</scp>4 gene mutations causing aggressive <scp>ALS</scp>. European Journal of Neurology, 2018, 25, e41-e42.	3.3	14
8	Inherited Cardiomyopathies: From Genotype to Phenotype. Journal of Clinical & Medical Genomics, 2018, 06, .	0.1	0
9	Whole-Exome Sequencing Identifies Pathogenic Variants in TJP1 Gene Associated With Arrhythmogenic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2018, 11, e002123.	3.6	38
10	Large Genomic Rearrangements of Desmosomal Genes in Italian Arrhythmogenic Cardiomyopathy Patients. Circulation: Arrhythmia and Electrophysiology, 2017, 10, .	4.8	35
11	Co-inheritance of mutations associated with arrhythmogenic cardiomyopathy and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2017, 25, 1165-1169.	2.8	10
12	Mutations in NEBL encoding the cardiac Z-disk protein nebulette are associated with various cardiomyopathies. Archives of Medical Science, 2016, 2, 263-278.	0.9	26
13	Arrhythmogenic right-ventricular cardiomyopathy. Journal of Cardiovascular Medicine, 2016, 17, 399-407.	1.5	16
14	A founderMYBPC3mutation results in HCM with a high risk of sudden death after the fourth decade of life. Journal of Medical Genetics, 2015, 52, 338-347.	3.2	41
15	The novel S59P mutation in the TNFRSF1A gene identified in an adult onset TNF receptor associated periodic syndrome (TRAPS) constitutively activates NF-Î²B pathway. Arthritis Research and Therapy, 2015, 17, 93.	3.5	43
16	Autosomal dominant lateral temporal epilepsy (ADLTE): Novel structural and single-nucleotide LGI1 mutations in families with predominant visual auras. Epilepsy Research, 2015, 110, 132-138.	1.6	17
17	Arrhythmogenic cardiomyopathy: a disease of intercalated discs. Cell and Tissue Research, 2015, 360, 491-500.	2.9	41
18	Homozygous Desmocollin-2 Mutations and Arrhythmogenic Cardiomyopathy. American Journal of Cardiology, 2015, 116, 1245-1251.	1.6	38

#	Article	IF	Citations
19	Desmin Mutations and Arrhythmogenic Right Ventricular Cardiomyopathy. American Journal of Cardiology, 2013, 111, 400-405.	1.6	62
20	Identification of a PKP2 gene deletion in a family with arrhythmogenic right ventricular cardiomyopathy. European Journal of Human Genetics, 2013, 21, 1226-1231.	2.8	39
21	Compound and Digenic Heterozygosity Predicts Lifetime Arrhythmic Outcome and Sudden Cardiac Death in Desmosomal Geneâ€“Related Arrhythmogenic Right Ventricular Cardiomyopathy. Circulation: Cardiovascular Genetics, 2013, 6, 533-542.	5.1	209
22	Mutations in the area composita protein Î±T-catenin are associated with arrhythmogenic right ventricular cardiomyopathy. European Heart Journal, 2013, 34, 201-210.	2.2	175
23	Clinical phenotype and diagnosis of arrhythmogenic right ventricular cardiomyopathy in pediatric patients carrying desmosomal gene mutations. Heart Rhythm, 2011, 8, 1686-1695.	0.7	66
24	The p.A897KfsX4 frameshift variation in desmocollin-2 is not a causative mutation in arrhythmogenic right ventricular cardiomyopathy. European Journal of Human Genetics, 2010, 18, 776-782.	2.8	19
25	Multiple mutations in desmosomal proteins encoding genes in arrhythmogenic right ventricular cardiomyopathy/dysplasia. Heart Rhythm, 2010, 7, 22-29.	0.7	161
26	Missense mutations in Desmocollin-2 N-terminus, associated with arrhythmogenic right ventricular cardiomyopathy, affect intracellular localization of desmocollin-2 in vitro. BMC Medical Genetics, 2007, 8, 65.	2.1	61