

Laura Muino Mosquera

List of Publications by Year in descending order

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Version: 2024-02-01

33
papers

769
citations

623574

14
h-index

526166

27
g-index

39
all docs

39
docs citations

39
times ranked

1348
citing authors

#	ARTICLE	IF	CITATIONS
1	International Registry of Patients Carrying <i>TGFBR1</i> or <i>TGFBR2</i> Mutations. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 548-558.	5.1	145
2	Arterial tortuosity syndrome: 40 new families and literature review. <i>Genetics in Medicine</i> , 2018, 20, 1236-1245.	1.1	66
3	Gene panel sequencing in heritable thoracic aortic disorders and related entities – results of comprehensive testing in a cohort of 264 patients. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 9.	1.2	62
4	Features of Marfan syndrome not listed in the Ghent nosology – the dark side of the disease. <i>Expert Review of Cardiovascular Therapy</i> , 2019, 17, 883-915.	0.6	46
5	Intrinsic cardiomyopathy in Marfan syndrome: results from in-vivo and ex-vivo studies of the <i>Fbn1</i> C1039G/+ model and longitudinal findings in humans. <i>Pediatric Research</i> , 2015, 78, 256-263.	1.1	45
6	<i>SMAD3</i> pathogenic variants: risk for thoracic aortic disease and associated complications from the Montalcino Aortic Consortium. <i>Journal of Medical Genetics</i> , 2019, 56, 252-260.	1.5	43
7	Sex, pregnancy and aortic disease in Marfan syndrome. <i>PLoS ONE</i> , 2017, 12, e0181166.	1.1	40
8	Efficacy of losartan as add-on therapy to prevent aortic growth and ventricular dysfunction in patients with Marfan syndrome: a randomized, double-blind clinical trial. <i>Acta Cardiologica</i> , 2017, 72, 616-624.	0.3	36
9	Aortic disease in Marfan syndrome is caused by overactivation of sGC-PRKG signaling by NO. <i>Nature Communications</i> , 2021, 12, 2628.	5.8	28
10	Tailoring the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines for the Interpretation of Sequenced Variants in the <i>FBN1</i> Gene for Marfan Syndrome. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002039.	1.6	20
11	More than meets the eye: Expanding and reviewing the clinical and mutational spectrum of brittle cornea syndrome. <i>Human Mutation</i> , 2021, 42, 711-730.	1.1	19
12	Association of Mitral Annular Disjunction With Cardiovascular Outcomes Among Patients With Marfan Syndrome. <i>JAMA Cardiology</i> , 2021, 6, 1177.	3.0	19
13	Myocardial Function, Heart Failure and Arrhythmia in Marfan Syndrome: A Systematic Literature Review. <i>Diagnostics</i> , 2020, 10, 751.	1.3	19
14	Case-matched Comparison of Cardiovascular Outcome in Loeys-Dietz Syndrome versus Marfan Syndrome. <i>Journal of Clinical Medicine</i> , 2019, 8, 2079.	1.0	17
15	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. <i>Genetics in Medicine</i> , 2021, 23, 94-102.	1.1	16
16	PGM1 deficiency diagnosed during an endocrine work-up of low IGF-1 mediated growth failure. <i>Acta Clinica Belgica</i> , 2016, 71, 435-437.	0.5	15
17	Myocardial disease and ventricular arrhythmia in Marfan syndrome: a prospective study. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 300.	1.2	14
18	Marfan Syndrome and Related Heritable Thoracic Aortic Aneurysms and Dissections. <i>Current Pharmaceutical Design</i> , 2015, 21, 4061-4075.	0.9	13

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19	Mitral valve prolapse syndrome and MASS phenotype: Stability of aortic dilatation but progression of mitral valve prolapse. <i>IJC Heart and Vasculature</i> , 2016, 10, 39-46.	0.6	12
20	Frequency of Ventricular Arrhythmias and Other Rhythm Abnormalities in Children and Young Adults With the Marfan Syndrome. <i>American Journal of Cardiology</i> , 2018, 122, 1429-1436.	0.7	12
21	Looking for the Missing Links. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e002185.	1.6	11
22	Sleep apnea and the impact on cardiovascular risk in patients with Marfan syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e805.	0.6	11
23	Megaconial muscular dystrophy caused by mitochondrial membrane homeostasis defect, new insights from skeletal and heart muscle analyses. <i>Mitochondrion</i> , 2016, 27, 32-38.	1.6	9
24	Managing aortic aneurysms and dissections during pregnancy. <i>Expert Review of Cardiovascular Therapy</i> , 2015, 13, 703-714.	0.6	8
25	Pregnancy in Women With SMAD3 Mutation. <i>Journal of the American College of Cardiology</i> , 2017, 69, 1356-1358.	1.2	8
26	A Reassessment of Copy Number Variations in Congenital Heart Defects: Picturing the Whole Genome. <i>Genes</i> , 2021, 12, 1048.	1.0	6
27	Angiotensin-II receptor blockade in Marfan syndrome. <i>Lancet, The</i> , 2019, 394, 2206-2207.	6.3	4
28	Analysis of the recovery phase after maximal exercise in children with repaired tetralogy of Fallot and the relationship with ventricular function. <i>PLoS ONE</i> , 2020, 15, e0244312.	1.1	4
29	Arrhythmia and impaired myocardial function in heritable thoracic aortic disease: An international retrospective cohort study. <i>European Journal of Medical Genetics</i> , 2022, 65, 104503.	0.7	4
30	Exploring the Mutational Landscape of Isolated Congenital Heart Defects: An Exome Sequencing Study Using Cardiac DNA. <i>Genes</i> , 2022, 13, 1214.	1.0	4
31	Genetics in congenital heart disease. Are we ready for it?. <i>Revista Espanola De Cardiologia (English Ed)</i> Tj ETQq1 1 0,784314 rgBT /Ov 0,4 2		
32	Cardiomyopathy in Genetic Aortic Diseases. <i>Frontiers in Pediatrics</i> , 2021, 9, 682390.	0.9	2
33	Marfan Syndrome. , 2019, , .		0