Laura Muino Mosquera

List of Publications by Year in Descending Order

32 papers	467 citations	10 h-index	21 g-index
39 ext. papers	629 ext. citations	5.4 avg, IF	3.32 L-index

#	Paper	IF	Citations
32	Arrhythmia and impaired myocardial function in heritable thoracic aortic disease: An international retrospective cohort study.. *European Journal of Medical Genetics*, **2022**, 104503	2.6	0
31	More than meets the eye: Expanding and reviewing the clinical and mutational spectrum of brittle cornea syndrome. *Human Mutation*, **2021**, 42, 711-730	4.7	4
30	Aortic disease in Marfan syndrome is caused by overactivation of sGC-PRKG signaling by NO. *Nature Communications*, **2021**, 12, 2628	17.4	4
29	Cardiomyopathy in Genetic Aortic Diseases. *Frontiers in Pediatrics*, **2021**, 9, 682390	3.4	0
28	Association of Mitral Annular Disjunction With Cardiovascular Outcomes Among Patients With Marfan Syndrome. *JAMA Cardiology*, **2021**, 6, 1177-1186	16.2	4
27	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. *Genetics in Medicine*, **2021**, 23, 94-102	8.1	4
26	A Reassessment of Copy Number Variations in Congenital Heart Defects: Picturing the Whole Genome. *Genes*, **2021**, 12,	4.2	1
25	Gen□ica en la cardiopat□ cong□ita: ¿estamos preparados?. *Revista Espanola De Cardiologia*, **2020**, 73, 937-947	1.5	3
24	Genetics in congenital heart disease. Are we ready for it?. *Revista Espanola De Cardiologia (English Ed)*, **2020**, 73, 937-947	0.7	
23	Myocardial Function, Heart Failure and Arrhythmia in Marfan Syndrome: A Systematic Literature Review. *Diagnostics*, **2020**, 10,	3.8	7
22	Analysis of the recovery phase after maximal exercise in children with repaired tetralogy of Fallot and the relationship with ventricular function. *PLoS ONE*, **2020**, 15, e0244312	3.7	1
21	Myocardial disease and ventricular arrhythmia in Marfan syndrome: a prospective study. *Orphanet Journal of Rare Diseases*, **2020**, 15, 300	4.2	6
20	pathogenic variants: risk for thoracic aortic disease and associated complications from the Montalcino Aortic Consortium. *Journal of Medical Genetics*, **2019**, 56, 252-260	5.8	23
19	Marfan Syndrome **2019**,		
18	Sleep apnea and the impact on cardiovascular risk in patients with Marfan syndrome. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e805	2.3	3
17	Case-matched Comparison of Cardiovascular Outcome in Loeys-Dietz Syndrome versus Marfan Syndrome. *Journal of Clinical Medicine*, **2019**, 8,	5.1	9
16	Angiotensin-II receptor blockade in Marfan syndrome. *Lancet, The*, **2019**, 394, 2206-2207	40	2

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15	Features of Marfan syndrome not listed in the Ghent nosology - the dark side of the disease. *Expert Review of Cardiovascular Therapy*, **2019**, 17, 883-915	2.5	30
14	Arterial tortuosity syndrome: 40 new families and literature review. *Genetics in Medicine*, **2018**, 20, 1236-1245	8.1	40
13	Frequency of Ventricular Arrhythmias and Other Rhythm Abnormalities in Children and Young Adults With the Marfan Syndrome. *American Journal of Cardiology*, **2018**, 122, 1429-1436	3	9
12	Tailoring the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines for the Interpretation of Sequenced Variants in the Gene for Marfan Syndrome: Proposal for a Disease- and Gene-Specific Guideline. *Circulation Genomic and Precision Medicine*, **2018**, 11, e002039	5.2	15
11	Pregnancy in Women With SMAD3 Mutation. *Journal of the American College of Cardiology*, **2017**, 69, 1356-1358	15.1	4
10	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. *Acta Cardiologica*, **2017**, 72, 616-624	0.9	25
9	Sex, pregnancy and aortic disease in Marfan syndrome. *PLoS ONE*, **2017**, 12, e0181166	3.7	20
8	International Registry of Patients Carrying TGFBR1 or TGFBR2 Mutations: Results of the MAC (Montalcino Aortic Consortium). *Circulation: Cardiovascular Genetics*, **2016**, 9, 548-558		105
7	PGM1 deficiency diagnosed during an endocrine work-up of low IGF-1 mediated growth failure. *Acta Clinica Belgica*, **2016**, 71, 435-437	1.8	14
6	Megaconial muscular dystrophy caused by mitochondrial membrane homeostasis defect, new insights from skeletal and heart muscle analyses. *Mitochondrion*, **2016**, 27, 32-8	4.9	6
5	Mitral valve prolapse syndrome and MASS phenotype: Stability of aortic dilatation but progression of mitral valve prolapse. *IJC Heart and Vasculature*, **2016**, 10, 39-46	2.4	10
4	Gene panel sequencing in heritable thoracic aortic disorders and related entities - results of comprehensive testing in a cohort of 264 patients. *Orphanet Journal of Rare Diseases*, **2015**, 10, 9	4.2	57
3	Intrinsic cardiomyopathy in Marfan syndrome: results from in-vivo and ex-vivo studies of the Fbn1C1039G/+ model and longitudinal findings in humans. *Pediatric Research*, **2015**, 78, 256-63	3.2	34
2	Managing aortic aneurysms and dissections during pregnancy. *Expert Review of Cardiovascular Therapy*, **2015**, 13, 703-14	2.5	5
1	Marfan Syndrome and Related Heritable Thoracic Aortic Aneurysms and Dissections. *Current Pharmaceutical Design*, **2015**, 21, 4061-75	3.3	10