Josephina A N Meester

List of Publications by Year
in descending order

27 papers	1,046 citations	759233 12 h-index	552781 26 g-index
29 all docs	29 docs citations	29 times ranked	1932 citing authors

#	Article	IF	Citations
1	Molecular characterization and investigation of the role of genetic variation in phenotypic variability and response to treatment in a large pediatric Marfan syndrome cohort. Genetics in Medicine, 2022, 24, 1045-1053.	2.4	13
2	The fibrillinopathies: New insights with focus on the paradigm of opposing phenotypes for both FBN1 and FBN2. Human Mutation, 2022, 43, 815-831.	2.5	7
3	Genome-Wide Epistasis for Cardiovascular Severity in Marfan Study Design: Patient Organization Driven Research. Aorta, 2022, , .	0.5	0
4	Isolated aneurysmal disease as an underestimated finding in individuals with JAG1 pathogenic variants. Human Mutation, 2022, 43, 1824-1828.	2.5	3
5	A human importin-Î²-related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. American Journal of Human Genetics, 2021, 108, 1115-1125.	6.2	10
6	Novel LOX Variants in Five Families with Aortic/Arterial Aneurysm and Dissection with Variable Connective Tissue Findings. International Journal of Molecular Sciences, 2021, 22, 7111.	4.1	7
7	Two novel presentations of KCNMA1â€related pathologyâ€“â€“Expanding the clinical phenotype of a rare channelopathy. Molecular Genetics & Genomic Medicine, 2021, 9, e1797.	1.2	6
8	Clinically relevant variants in a large cohort of Indian patients with Marfan syndrome and related disorders identified by next-generation sequencing. Scientific Reports, 2021, 11, 764.	3.3	7
9	Meester-Loeys Syndrome. Advances in Experimental Medicine and Biology, 2021, 1348, 265-272.	1.6	4
10	The Role of Genetics in Risk Stratification of Thoracic Aortic Aneurysm Dissection. Hearts, 2020, 1, 50-61.	0.9	0
11	Chondrodysplasias and Aneurysmal Thoracic Aortopathy: An Emerging Tale of Molecular Intersection. Trends in Molecular Medicine, 2020, 26, 783-795.	6.7	2
12	Enrichment of Rare Variants in Loeysâ€“Dietz Syndrome Genes in Spontaneous Coronary Artery Dissection but Not in Severe Fibromuscular Dysplasia. Circulation, 2020, 142, 1021-1024.	1.6	30
13	Variants in ADRB1 and CYP2C9: Association with Response to Atenolol and Losartan in Marfan Syndrome. Journal of Pediatrics, 2020, 222, 213-220.e5.	1.8	8
14	Biglycan in the Skeleton. Journal of Histochemistry and Cytochemistry, 2020, 68, 747-762.	2.5	30
15	Overlapping but distinct roles for NOTCH receptors in human cardiovascular disease. Clinical Genetics, 2019, 95, 85-94.	2.0	44
16	Adamsâ€“Oliver syndrome caused by mutations of the EOGT gene. American Journal of Medical Genetics, Part A, 2019, 179, 2246-2251.	1.2	17
17	Confirmation of the role of pathogenic SMAD6 variants in bicuspid aortic valve-related aortopathy. European Journal of Human Genetics, 2019, 27, 1044-1053.	2.8	32
18	Cardiogeneticsbank@UZA: A Collection of DNA, Tissues, and Cell Lines as a Translational Tool. Frontiers in Medicine, 2019, 6, 198.	2.6	1

#	Article	IF	Citations
19	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	2.5	116
20	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31
21	Loss-of-function mutations in the X-linked biglycan gene cause a severe syndromic form of thoracic aortic aneurysms and dissections. Genetics in Medicine, 2017, 19, 386-395.	2.4	94
22	Candidate Gene Resequencing in a Large Bicuspid Aortic Valve-Associated Thoracic Aortic Aneurysm Cohort: SMAD6 as an Important Contributor. Frontiers in Physiology, 2017, 8, 400.	2.8	85
23	Differences in manifestations of Marfan syndrome, Ehlers-Danlos syndrome, and Loeys-Dietz syndrome. Annals of Cardiothoracic Surgery, 2017, 6, 582-594.	1.7	192
24	Inborn errors in RNA polymerase III underlie severe varicella zoster virus infections. Journal of Clinical Investigation, 2017, 127, 3543-3556.	8.2	125
25	Identification of FBN1 gene mutations in Ukrainian Marfan syndrome patients. Genetical Research, 2016, 98, e13.	0.9	2
26	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. Human Mutation, 2015, 36, 808-814.	2.5	97
27	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	6.2	73