Pauline Arnaud

List of Publications by Year
in descending order

32	953	623574 14	477173 29
papers	citations	h-index	g-index

34	34	34	1455
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical Validity of Genes for Heritable Thoracic Aortic Aneurysm and Dissection. Journal of the American College of Cardiology, 2018, 72, 605-615.	1.2	190
2	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. Circulation Research, 2016, 118, 928-934.	2.0	180
3	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. European Journal of Human Genetics, 2018, 26, 1759-1772.	1.4	73
4	Clinical relevance of genotypeâ€“phenotype correlations beyond vascular events in a cohort study of 1500 Marfan syndrome patients with FBN1 pathogenic variants. Genetics in Medicine, 2021, 23, 1296-1304.	1.1	63
5	SMAD3pathogenic variants: risk for thoracic aortic disease and associated complications from the Montalcino Aortic Consortium. Journal of Medical Genetics, 2019, 56, 252-260.	1.5	43
6	Association Between Mutation Size and Cardiac Involvement in Myotonic Dystrophy Type 1. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	39
7	Genetic diversity and pathogenic variants as possible predictors of severity in a French sample of nonsyndromic heritable thoracic aortic aneurysms and dissections (nshTAAD). Genetics in Medicine, 2019, 21, 2015-2024.	1.1	39
8	Pathogenic FBN1 Genetic Variation and Aortic Dissection in Patients With MarfanÂSyndrome. Journal of the American College of Cardiology, 2020, 75, 843-853.	1.2	38
9	Unusual association of a unique CAG interruption in 5â€² of DM1 CTG repeats with intergenerational contractions and low somatic mosaicism. Human Mutation, 2018, 39, 970-982.	1.1	37
10	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. Genetics in Medicine, 2019, 21, 144-151.	1.1	36
11	Development and Validation of a New Scoring System to Predict Survival in Patients With Myotonic Dystrophy Type 1. JAMA Neurology, 2018, 75, 573.	4.5	32
12	Homozygous and compound heterozygous mutations in theFBN1gene: unexpected findings in molecular diagnosis of Marfan syndrome. Journal of Medical Genetics, 2017, 54, 100-103.	1.5	30
13	Pathogenic variants in THSD4, encoding the ADAMTS-like 6 protein, predispose to inherited thoracic aortic aneurysm. Genetics in Medicine, 2021, 23, 111-122.	1.1	25
14	Systems pharmacologyâ€“based integration of human and mouse data for drug repurposing to treat thoracic aneurysms. JCI Insight, 2019, 4, .	2.3	21
15	Phenotypic spectrum of TGFB3 diseaseâ€causing variants in a Dutchâ€French cohort and first report of a homozygous patient. Clinical Genetics, 2020, 97, 723-730.	1.0	15
16	Unsuspected somatic mosaicism for FBN1 gene contributes to Marfan syndrome. Genetics in Medicine, 2021, 23, 865-871.	1.1	14
17	Clinical and genetic data of 22 new patients with SMAD3 pathogenic variants and review of the literature. Molecular Genetics & Genomic Medicine, 2020, 8, e1132.	0.6	11
18	Quantifying the Genetic Basis of Marfan Syndrome Clinical Variability. Genes, 2020, 11, 574.	1.0	11

#	Article	IF	Citations
19	A new mutational hotspot in the SKI gene in the context of MFS/TAA molecular diagnosis. Human Genetics, 2020, 139, 461-472.	1.8	8
20	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. Journal of Medical Genetics, 2020, 57, 466-474.	1.5	7
21	Actionable Genes, Core Databases, and Locus-Specific Databases. Human Mutation, 2016, 37, 1299-1307.	1.1	6
22	Reference Expression Profile of Three FBN1 Transcript Isoforms and Their Association with Clinical Variability in Marfan Syndrome. Genes, 2019, 10, 128.	1.0	6
23	Neonatal Marfan Syndrome: Report of a Case with an Inherited Splicing Mutation outside the Neonatal Domain. Molecular Syndromology, 2015, 6, 281-286.	0.3	5
24	WES/WGS Reporting of Mutations from Cardiovascular â€œActionableâ€•Genes in Clinical Practice: A Key Role for UMD Knowledgebases in the Era of Big Databases. Human Mutation, 2016, 37, 1308-1317.	1.1	5
25	Marfan Syndrome Variability: Investigation of the Roles of Sarcolipin and Calcium as Potential Transregulator of FBN1 Expression. Genes, 2018, 9, 421.	1.0	4
26	Inhibition of HIPK2 Alleviates Thoracic Aortic Disease in Mice With Progressively Severe Marfan Syndrome. Arteriosclerosis, Thrombosis, and Vascular Biology, 2021, 41, 2483-2493.	1.1	4
27	The natural history of a family with aortic dissection associated with a novel ACTA2 variant. Annals of Vascular Surgery, 2021, , .	0.4	3
28	Cooperative Mechanism of ADAMTS/ ADAMTSL and Fibrillin-1 in the Marfan Syndrome and Acromelic Dysplasias. Frontiers in Genetics, 2021, 12, 734718.	1.1	2
29	A Case of Trisomy 13 Mosaicism Presenting with a Severe Aortic Root Dilatation and Marfanoid Habitus due to an Unpredictable Cytogenetic Mechanism. Cytogenetic and Genome Research, 2020, 160, 72-79.	0.6	1
30	Association between mutation size and cardiac involvement in myotonic dystrophy type 1: an analysis of the DM1-heart registry. Neuromuscular Disorders, 2017, 27, S178-S179.	0.3	0
31	Survival in myotonic dystrophy type 1 predicted by the new DM1 survival risk score. Neuromuscular Disorders, 2017, 27, S179.	0.3	0
32	A giant abdominal aortic aneurysm revealing a Marfan syndrome with a new FBN1 mutation. Canadian Journal of Cardiology, 2021, 37, 1870-1872.	0.8	0