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	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
2	Unsuspected somatic mosaicism for FBN1 gene contributes to Marfan syndrome. Genetics in Medicine, 2021, 23, 865-871.	1.1	14
3	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
4	The natural history of a family with aortic dissection associated with a novel ACTA2 variant. Annals of Vascular Surgery, 2021, , .	0.4	3
5	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
6	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
7	Cooperative Mechanism of ADAMTS/ ADAMTSL and Fibrillin-1 in the Marfan Syndrome and Acromelic Dysplasias. Frontiers in Genetics, 2021, 12, 734718.	1.1	2
8	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
9	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
10	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
11	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
12	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
13	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
14	Quantifying the Genetic Basis of Marfan Syndrome Clinical Variability. Genes, 2020, 11, 574.	1.0	11
15	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. Genetics in Medicine, 2019, 21, 144-151.	1.1	36
16	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
17	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
18	Reference Expression Profile of Three FBN1 Transcript Isoforms and Their Association with Clinical Variability in Marfan Syndrome. Genes, 2019, 10, 128.	1.0	6

#	Article	IF	Citations
19	Systems pharmacologyâ€“based integration of human and mouse data for drug repurposing to treat thoracic aneurysms. JCI Insight, 2019, 4, .	2.3	21
20	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
21	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
22	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. European Journal of Human Genetics, 2018, 26, 1759-1772.	1.4	73
23	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
24	Marfan Syndrome Variability: Investigation of the Roles of Sarcolipin and Calcium as Potential Transregulator of FBN1 Expression. Genes, 2018, 9, 421.	1.0	4
25	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
26	Association Between Mutation Size and Cardiac Involvement in Myotonic Dystrophy Type 1. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	39
27	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
28	Survival in myotonic dystrophy type 1 predicted by the new DM1 survival risk score. Neuromuscular Disorders, 2017, 27, S179.	0.3	0
29	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. Circulation Research, 2016, 118, 928-934.	2.0	180
30	Actionable Genes, Core Databases, and Locus-Specific Databases. Human Mutation, 2016, 37, 1299-1307.	1.1	6
31	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
32	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