Dianna M Milewicz

List of Publications by Citations

214 papers	17,877 citations	67 h-index	131 g-index
230 ext. papers	21,023 ext. citations	8.4 avg, IF	6.28 L-index

#	Paper	IF	Citations
214	2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM guidelines for the diagnosis and management of patients with Thoracic Aortic Disease: a report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines, American Association for Thoracic Surgery, American College of Radiology, American Stroke Association, Society of Cardiovascular Anesthesiologists, Society for Cardiovascular Angiography and Interventions, Society of Interventional Radiology, Society of Thor. *Circulation*, **2010**, 121, e266-369	16.7	1569
213	The revised Ghent nosology for the Marfan syndrome. *Journal of Medical Genetics*, **2010**, 47, 476-85	5.8	1282
212	Aneurysm syndromes caused by mutations in the TGF-beta receptor. *New England Journal of Medicine*, **2006**, 355, 788-98	59.2	1243
211	2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM Guidelines for the diagnosis and management of patients with thoracic aortic disease. A Report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines, American Association for Thoracic Surgery, American College of Radiology,American Stroke Association, Society of Cardiovascular Anesthesiologists, Society for Cardiovascular Angiography and Interventional Radiology, Society of Thora. *Journal of the American College of Cardiology*, **2010**, 55, e27-e129	15.1	980
210	Mutations in smooth muscle alpha-actin (ACTA2) lead to thoracic aortic aneurysms and dissections. *Nature Genetics*, **2007**, 39, 1488-93	36.3	623
209	Mutations in smooth muscle alpha-actin (ACTA2) cause coronary artery disease, stroke, and Moyamoya disease, along with thoracic aortic disease. *American Journal of Human Genetics*, **2009**, 84, 617-27	11	364
208	An adventitial IL-6/MCP1 amplification loop accelerates macrophage-mediated vascular inflammation leading to aortic dissection in mice. *Journal of Clinical Investigation*, **2009**, 119, 3637-51	15.9	318
207	Genetic basis of thoracic aortic aneurysms and dissections: focus on smooth muscle cell contractile dysfunction. *Annual Review of Genomics and Human Genetics*, **2008**, 9, 283-302	9.7	304
206	Mutations in transforming growth factor-beta receptor type II cause familial thoracic aortic aneurysms and dissections. *Circulation*, **2005**, 112, 513-20	16.7	296
205	Treatment of aortic disease in patients with Marfan syndrome. *Circulation*, **2005**, 111, e150-7	16.7	273
204	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. *Nature Genetics*, **2012**, 44, 916-21	36.3	257
203	Fibrillin-2 (FBN2) mutations result in the Marfan-like disorder, congenital contractural arachnodactyly. *Nature Genetics*, **1995**, 11, 456-8	36.3	235
202	Mutations in myosin light chain kinase cause familial aortic dissections. *American Journal of Human Genetics*, **2010**, 87, 701-7	11	228
201	Bicuspid aortic valve: identifying knowledge gaps and rising to the challenge from the International Bicuspid Aortic Valve Consortium (BAVCon). *Circulation*, **2014**, 129, 2691-704	16.7	227
200	Characterization of the inflammatory and apoptotic cells in the aortas of patients with ascending thoracic aortic aneurysms and dissections. *Journal of Thoracic and Cardiovascular Surgery*, **2006**, 131, 671-8	1.5	225
199	Exome sequencing identifies SMAD3 mutations as a cause of familial thoracic aortic aneurysm and dissection with intracranial and other arterial aneurysms. *Circulation Research*, **2011**, 109, 680-6	15.7	221
198	Role of mechanotransduction in vascular biology: focus on thoracic aortic aneurysms and dissections. *Circulation Research*, **2015**, 116, 1448-61	15.7	216

197	Marfan syndrome. Long-term survival and complications after aortic aneurysm repair. *Circulation*, **1995**, 91, 728-33	16.7	216
196	2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM guidelines for the diagnosis and management of patients with thoracic aortic disease: executive summary. A report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines, American Association for Thoracic Surgery, American College of Radiology, American Stroke	2.7	210
195	MYH11 mutations result in a distinct vascular pathology driven by insulin-like growth factor 1 and angiotensin II. *Human Molecular Genetics*, **2007**, 16, 2453-62 *Interventions*, **2010**, 76, E43-86	5.6	210
194	Asprosin, a Fasting-Induced Glucogenic Protein Hormone. *Cell*, **2016**, 165, 566-79	56.2	187
193	Familial thoracic aortic aneurysms and dissections: genetic heterogeneity with a major locus mapping to 5q13-14. *Circulation*, **2001**, 103, 2461-8	16.7	177
192	De novo ACTA2 mutation causes a novel syndrome of multisystemic smooth muscle dysfunction. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2437-43	2.5	174
191	Familial thoracic aortic dilatations and dissections: a case control study. *Journal of Vascular Surgery*, **1997**, 25, 506-11	3.5	173
190	Pathogenesis of thoracic and abdominal aortic aneurysms. *Annals of the New York Academy of Sciences*, **2006**, 1085, 339-52	6.5	171
189	Recurrent gain-of-function mutation in PRKG1 causes thoracic aortic aneurysms and acute aortic dissections. *American Journal of Human Genetics*, **2013**, 93, 398-404	11	153
188	Consideration of Sex Differences in Design and Reporting of Experimental Arterial Pathology Studies-Statement From ATVB Council. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2018**, 38, 292-303	6.4	151
187	Identification of a chromosome 11q23.2-q24 locus for familial aortic aneurysm disease, a genetically heterogeneous disorder. *Circulation*, **2001**, 103, 2469-75	16.7	150
186	Genome-wide association study identifies a susceptibility locus for thoracic aortic aneurysms and aortic dissections spanning FBN1 at 15q21.1. *Nature Genetics*, **2011**, 43, 996-1000	36.3	148
185	Fibrillin-1 (FBN1) mutations in patients with thoracic aortic aneurysms. *Circulation*, **1996**, 94, 2708-11	16.7	139
184	Asprosin is a centrally acting orexigenic hormone. *Nature Medicine*, **2017**, 23, 1444-1453	50.5	130
183	Reduced penetrance and variable expressivity of familial thoracic aortic aneurysms/dissections. *American Journal of Cardiology*, **1998**, 82, 474-9	3	125
182	Mapping a locus for familial thoracic aortic aneurysms and dissections (TAAD2) to 3p24-25. *Circulation*, **2003**, 107, 3184-90	16.7	124
181	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. *Circulation Research*, **2016**, 118, 928-34	15.7	122
180	A roadmap to investigate the genetic basis of bicuspid aortic valve and its complications: insights from the International BAVCon (Bicuspid Aortic Valve Consortium). *Journal of the American College of Cardiology*, **2014**, 64, 832-9	15.1	119

179	2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM Guidelines for the Diagnosis and Management of Patients With Thoracic Aortic Disease: Executive Summary. *Journal of the American College of Cardiology*, **2010**, 55, 1509-1544	15.1	119
178	Altered Smooth Muscle Cell Force Generation as a Driver of Thoracic Aortic Aneurysms and Dissections. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2017**, 37, 26-34	9.4	113
177	Genetics of Thoracic and Abdominal Aortic Diseases. *Circulation Research*, **2019**, 124, 588-606	15.7	112
176	Cell biology. Dysfunctional mechanosensing in aneurysms. *Science*, **2014**, 344, 477-9	33.3	107
175	Spectrum of aortic operations in 300 patients with confirmed or suspected Marfan syndrome. *Annals of Thoracic Surgery*, **2006**, 81, 2063-78; discussion 2078	2.7	106
174	Rescuing the physician-scientist workforce: the time for action is now. *Journal of Clinical Investigation*, **2015**, 125, 3742-7	15.9	106
173	International Registry of Patients Carrying TGFBR1 or TGFBR2 Mutations: Results of the MAC (Montalcino Aortic Consortium). *Circulation: Cardiovascular Genetics*, **2016**, 9, 548-558		105
172	Association of fibrillin 1 single-nucleotide polymorphism haplotypes with systemic sclerosis in Choctaw and Japanese populations. *Arthritis and Rheumatism*, **2001**, 44, 893-901		105
171	RNF213 rare variants in an ethnically diverse population with Moyamoya disease. *Stroke*, **2014**, 45, 3200-7	6.7	103
170	Delineation of the Marfan phenotype associated with mutations in exons 23-32 of the FBN1 gene. *American Journal of Medical Genetics Part A*, **1996**, 62, 233-42		101
169	Clinical Validity of Genes for Heritable Thoracic Aortic Aneurysm and Dissection. *Journal of the American College of Cardiology*, **2018**, 72, 605-615	15.1	97
168	TGFBR2 mutations alter smooth muscle cell phenotype and predispose to thoracic aortic aneurysms and dissections. *Cardiovascular Research*, **2010**, 88, 520-9	9.9	95
167	Nitric oxide mediates aortic disease in mice deficient in the metalloprotease Adamts1 and in a mouse model of Marfan syndrome. *Nature Medicine*, **2017**, 23, 200-212	50.5	92
166	Characterization of the inflammatory cells in ascending thoracic aortic aneurysms in patients with Marfan syndrome, familial thoracic aortic aneurysms, and sporadic aneurysms. *Journal of Thoracic and Cardiovascular Surgery*, **2008**, 136, 922-9, 929.e1	1.5	92
165	Bicuspid aortic valve aortopathy in adults: Incidence, etiology, and clinical significance. *International Journal of Cardiology*, **2015**, 201, 400-7	3.2	89
164	2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM Guidelines for the diagnosis and management of patients with thoracic aortic disease: Executive summary: A report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines, American Association for Thoracic Surgery, American College of Radiology, American Stroke	3.9	89
163	Ten novel FBN2 mutations in congenital contractural arachnodactyly: delineation of the molecular pathogenesis and clinical phenotype. *Human Mutation*, **2002**, 19, 39-48 algesia, **2010**, 111, 279-315	4.7	89
162	MFAP5 loss-of-function mutations underscore the involvement of matrix alteration in the pathogenesis of familial thoracic aortic aneurysms and dissections. *American Journal of Human Genetics*, **2014**, 95, 736-43	11	88

161	A novel distinctive cerebrovascular phenotype is associated with heterozygous Arg179 ACTA2 mutations. *Brain*, **2012**, 135, 2506-14	11.2	88
160	Aortic Disease Presentation and Outcome Associated With ACTA2 Mutations. *Circulation: Cardiovascular Genetics*, **2015**, 8, 457-64		82
159	From genetics to response to injury: vascular smooth muscle cells in aneurysms and dissections of the ascending aorta. *Cardiovascular Research*, **2018**, 114, 578-589	9.9	82
158	Coagulation factor V(A2440G) causes east Texas bleeding disorder via TFPI *Journal of Clinical Investigation*, **2013**, 123, 3777-87	15.9	81
157	Structure of the Elastin-Contractile Units in the Thoracic Aorta and How Genes That Cause Thoracic Aortic Aneurysms and Dissections Disrupt This Structure. *Canadian Journal of Cardiology*, **2016**, 32, 26-34	3.8	78
156	Interleukin-6-signal transducer and activator of transcription-3 signaling mediates aortic dissections induced by angiotensin II via the T-helper lymphocyte 17-interleukin 17 axis in C57BL/6 mice. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2013**, 33, 1612-21	9.4	78
155	Therapeutics Targeting Drivers of Thoracic Aortic Aneurysms and Acute Aortic Dissections: Insights from Predisposing Genes and Mouse Models. *Annual Review of Medicine*, **2017**, 68, 51-67	17.4	74
154	Early and 1-year outcomes of aortic root surgery in patients with Marfan syndrome: a prospective, multicenter, comparative study. *Journal of Thoracic and Cardiovascular Surgery*, **2014**, 147, 1758-66, 1767.e1-4	1.5	70
153	Genetic variants promoting smooth muscle cell proliferation can result in diffuse and diverse vascular diseases: evidence for a hyperplastic vasculomyopathy. *Genetics in Medicine*, **2010**, 12, 196-203	8.1	70
152	Molecular diagnosis in vascular Ehlers-Danlos syndrome predicts pattern of arterial involvement and outcomes. *Journal of Vascular Surgery*, **2014**, 60, 160-9	3.5	69
151	MAT2A mutations predispose individuals to thoracic aortic aneurysms. *American Journal of Human Genetics*, **2015**, 96, 170-7	11	68
150	Recurrent chromosome 16p13.1 duplications are a risk factor for aortic dissections. *PLoS Genetics*, **2011**, 7, e1002118	6	67
149	Classification analysis of the transcriptosome of nonlesional cultured dermal fibroblasts from systemic sclerosis patients with early disease. *Arthritis and Rheumatism*, **2005**, 52, 865-76		67
148	SMAD2 Mutations Are Associated with Arterial Aneurysms and Dissections. *Human Mutation*, **2015**, 36, 1145-9	4.7	65
147	The genetics of colored sequence synesthesia: suggestive evidence of linkage to 16q and genetic heterogeneity for the condition. *Behavioural Brain Research*, **2011**, 223, 48-52	3.4	65
146	Autoantibodies to fibrillin-1 activate normal human fibroblasts in culture through the TGF-beta pathway to recapitulate the "scleroderma phenotype". *Journal of Immunology*, **2005**, 175, 4555-60	5.3	65
145	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. *Journal of Clinical Investigation*, **2016**, 126, 948-61	15.9	65
144	Rare copy number variants disrupt genes regulating vascular smooth muscle cell adhesion and contractility in sporadic thoracic aortic aneurysms and dissections. *American Journal of Human Genetics*, **2010**, 87, 743-56	11	64

143	Systemic sclerosis (scleroderma): specific autoantigen genes are selectively overexpressed in scleroderma fibroblasts. *Journal of Immunology*, **2001**, 167, 7126-33	5.3	62
142	Processing of the fibrillin-1 carboxyl-terminal domain. *Journal of Biological Chemistry*, **1999**, 274, 8933-40	5.4	62
141	Rare, nonsynonymous variant in the smooth muscle-specific isoform of myosin heavy chain, MYH11, R247C, alters force generation in the aorta and phenotype of smooth muscle cells. *Circulation Research*, **2012**, 110, 1411-22	15.7	61
140	Clustering of FBN2 mutations in patients with congenital contractural arachnodactyly indicates an important role of the domains encoded by exons 24 through 34 during human development **1998**, 78, 350-355		59
139	IL-6 regulates extracellular matrix remodeling associated with aortic dilation in a fibrillin-1 hypomorphic mgR/mgR mouse model of severe Marfan syndrome. *Journal of the American Heart Association*, **2014**, 3, e000476	6	58
138	Ultra High-Resolution In vivo Computed Tomography Imaging of Mouse Cerebrovasculature Using a Long Circulating Blood Pool Contrast Agent. *Scientific Reports*, **2015**, 5, 10178	4.9	56
137	Genome-wide analysis yields new loci associating with aortic valve stenosis. *Nature Communications* , **2018**, 9, 987	17.4	56
136	Severe aortic and arterial aneurysms associated with a TGFBR2 mutation. *Nature Clinical Practice Cardiovascular Medicine*, **2007**, 4, 167-71		55
135	Genetic basis of thoracic aortic aneurysms and aortic dissections. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2005**, 139C, 10-6	3.1	53
134	Valve-sparing and valve-replacing techniques for aortic root replacement in patients with Marfan syndrome: Analysis of early outcome. *Journal of Thoracic and Cardiovascular Surgery*, **2009**, 137, 1124-32	1.5	52
133	Diabetes and reduced risk for thoracic aortic aneurysms and dissections: a nationwide case-control study. *Journal of the American Heart Association*, **2012**, 1,	6	52
132	NLRP3 (Nucleotide Oligomerization Domain-Like Receptor Family, Pyrin Domain Containing 3)-Caspase-1 Inflammasome Degrades Contractile Proteins: Implications for Aortic Biomechanical Dysfunction and Aneurysm and Dissection Formation. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2017**, 37, 694-706	9.4	51
131	Clinical and biochemical profiles suggest fibromuscular dysplasia is a systemic disease with altered TGF-□expression and connective tissue features. *FASEB Journal*, **2014**, 28, 3313-24	0.9	51
130	Impact of image analysis methodology on diagnostic and surgical classification of patients with thoracic aortic aneurysms. *Annals of Thoracic Surgery*, **2011**, 92, 904-12	2.7	51
129	2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM Guidelines for the Diagnosis and Management of Patients With Thoracic Aortic Disease: Executive Summary. *Circulation*, **2010**, 121, 1544-1579	16.7	51
128	Genetic basis of thoracic aortic aneurysms and dissections. *Current Opinion in Cardiology*, **2002**, 17, 677-82	2.1	49
127	Therapies for Thoracic Aortic Aneurysms and Acute Aortic Dissections. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2019**, 39, 126-136	9.4	48
126	Aortic Complications Associated With Pregnancy in Marfan Syndrome: The NHLBI National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC). *Journal of the American Heart Association*, **2016**, 5,	6	46

125	Differentiation defect in neural crest-derived smooth muscle cells in patients with aortopathy associated with bicuspid aortic valves. *EBioMedicine*, **2016**, 10, 282-90	8.8	46
124	Critical Role of Cytosolic DNA and Its Sensing Adaptor STING in Aortic Degeneration, Dissection, and Rupture. *Circulation*, **2020**, 141, 42-66	16.7	45
123	Genetic Variants in LRP1 and ULK4 Are Associated with Acute Aortic Dissections. *American Journal of Human Genetics*, **2016**, 99, 762-769	11	44
122	Critical Role of ADAMTS-4 in the Development of Sporadic Aortic Aneurysm and Dissection in Mice. *Scientific Reports*, **2017**, 7, 12351	4.9	43
121	Myh11(R247C/R247C) mutations increase thoracic aorta vulnerability to intramural damage despite a general biomechanical adaptivity. *Journal of Biomechanics*, **2015**, 48, 113-21	2.9	39
120	The FBN2 gene: new mutations, locus-specific database (Universal Mutation Database FBN2), and genotype-phenotype correlations. *Human Mutation*, **2009**, 30, 181-90	4.7	39
119	Profibrillin-1 maturation by human dermal fibroblasts: proteolytic processing and molecular chaperones. *Journal of Cellular Biochemistry*, **2003**, 90, 641-52	4.7	38
118	Thoracic aortic disease in tuberous sclerosis complex: molecular pathogenesis and potential therapies in Tsc2+/- mice. *Human Molecular Genetics*, **2010**, 19, 1908-20	5.6	37
117	Genetic basis of thoracic aortic aneurysms and dissections: potential relevance to abdominal aortic aneurysms. *Annals of the New York Academy of Sciences*, **2006**, 1085, 242-55	6.5	37
116	Familial aggregation of both aortic and cerebral aneurysms: evidence for a common genetic basis in a subset of families. *Neurosurgery*, **2005**, 56, 655-61; discussion 655-61	3.2	37
115	Loss-of-Function Mutations in YY1AP1 Lead to Grange Syndrome and a Fibromuscular Dysplasia-Like Vascular Disease. *American Journal of Human Genetics*, **2017**, 100, 21-30	11	36
114	Deficiency of MMP17/MT4-MMP proteolytic activity predisposes to aortic aneurysm in mice. *Circulation Research*, **2015**, 117, e13-26	15.7	36
113	Long-term implications of emergency versus elective proximal aortic surgery in patients with Marfan syndrome in the Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions Consortium Registry. *Journal of Thoracic and Cardiovascular Surgery*, **2012**, 143, 282-6	1.5	36
112	Thoracic aortic disease in two patients with juvenile polyposis syndrome and SMAD4 mutations. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 185-91	2.5	36
111	Loss of Smooth Muscle Actin Leads to NF-B-Dependent Increased Sensitivity to Angiotensin II in Smooth Muscle Cells and Aortic Enlargement. *Circulation Research*, **2017**, 120, 1903-1915	15.7	35
110	Single-nucleotide polymorphism array genotyping is equivalent to metaphase cytogenetics for diagnosis of Turner syndrome. *Genetics in Medicine*, **2014**, 16, 53-9	8.1	35
109	Shared Genetic Risk Factors of Intracranial, Abdominal, and Thoracic Aneurysms. *Journal of the American Heart Association*, **2016**, 5,	6	34
108	Autosomal dominant inheritance of a predisposition to thoracic aortic aneurysms and dissections and intracranial saccular aneurysms. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2125-30	2.5	34

107	LTBP3 Pathogenic Variants Predispose Individuals to Thoracic Aortic Aneurysms and Dissections. *American Journal of Human Genetics*, **2018**, 102, 706-712	11	34
106	Associations of Age and Sex With Marfan Phenotype: The National Heart, Lung, and Blood Institute GenTAC (Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions) Registry. *Circulation: Cardiovascular Genetics*, **2017**, 10,		33
105	Ari-1 Regulates Myonuclear Organization Together with Parkin and Is Associated with Aortic Aneurysms. *Developmental Cell*, **2018**, 45, 226-244.e8	10.2	33
104	Use of genetics for personalized management of heritable thoracic aortic disease: how do we get there?. *Journal of Thoracic and Cardiovascular Surgery*, **2015**, 149, S3-5	1.5	32
103	Autosomal and X chromosome structural variants are associated with congenital heart defects in Turner syndrome: The NHLBI GenTAC registry. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3157-3164	2.5	32
102	Familial thoracic aortic aneurysms and dissections: identification of a novel locus for stable aneurysms with a low risk for progression to aortic dissection. *Circulation: Cardiovascular Genetics*, **2011**, 4, 36-42		31
101	Smooth muscle hyperplasia due to loss of smooth muscle □-actin is driven by activation of focal adhesion kinase, altered p53 localization and increased levels of platelet-derived growth factor receptor-□*Human Molecular Genetics*, **2013**, 22, 3123-37	5.6	30
100	Distinct skeletal abnormalities in four girls with Shprintzen-Goldberg syndrome. *American Journal of Medical Genetics Part A*, **1995**, 57, 565-72		30
99	Deletion of NF-B/RelA in Angiotensin II-Sensitive Mesenchymal Cells Blocks Aortic Vascular Inflammation and Abdominal Aortic Aneurysm Formation. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2017**, 37, 1881-1890	9.4	29
98	Eye features in three Danish patients with multisystemic smooth muscle dysfunction syndrome. *British Journal of Ophthalmology*, **2012**, 96, 1227-31	5.5	29
97	Methodology for using a universal primer to label amplified DNA segments for molecular analysis. *Biotechnology Letters*, **2003**, 25, 2079-83	3	29
96	Clinical Implications of Identifying Pathogenic Variants in Individuals With Thoracic Aortic Dissection. *Circulation Genomic and Precision Medicine*, **2019**, 12, e002476	5.2	27
95	Clinical history and management recommendations of the smooth muscle dysfunction syndrome due to ACTA2 arginine 179 alterations. *Genetics in Medicine*, **2018**, 20, 1206-1215	8.1	27
94	MicroRNAs, fibrotic remodeling, and aortic aneurysms. *Journal of Clinical Investigation*, **2012**, 122, 490-3	15.9	27
93	Update on Clinical Trials of Losartan With and Without -Blockers to Block Aneurysm Growth in Patients With Marfan Syndrome: A Review. *JAMA Cardiology*, **2019**, 4, 702-707	16.2	26
92	Thoracic aortic dissection and rupture in conotruncal cardiac defects: A population-based study. *International Journal of Cardiology*, **2015**, 184, 521-527	3.2	26
91	The National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC): results from phase I and scientific opportunities in phase II. *American Heart Journal*, **2011**, 162, 627-632.e1	4.9	26
90	THSD1 (Thrombospondin Type 1 Domain Containing Protein 1) Mutation in the Pathogenesis of Intracranial Aneurysm and Subarachnoid Hemorrhage. *Stroke*, **2016**, 47, 3005-3013	6.7	26

89	Successes and challenges of using whole exome sequencing to identify novel genes underlying an inherited predisposition for thoracic aortic aneurysms and acute aortic dissections. *Trends in Cardiovascular Medicine*, **2014**, 24, 53-60	6.9	25
88	Overexpression of smooth muscle myosin heavy chain leads to activation of the unfolded protein response and autophagic turnover of thick filament-associated proteins in vascular smooth muscle cells. *Journal of Biological Chemistry*, **2014**, 289, 14075-88	5.4	25
87	Missense mutations in FBN1 exons 41 and 42 cause Weill-Marchesani syndrome with thoracic aortic disease and Marfan syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2305-10	2.5	25
86	Heritable Thoracic Aortic Disease Genes in Sporadic Aortic Dissection. *Journal of the American College of Cardiology*, **2017**, 70, 2728-2730	15.1	24
85	The defining pathology of the new clinical and histopathologic entity ACTA2-related cerebrovascular disease. *Acta Neuropathologica Communications*, **2015**, 3, 81	7.3	24
84	Acute aortic dissections with pregnancy in women with ACTA2 mutations. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 106-12	2.5	24
83	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
82	Genetic testing in aortic aneurysm disease: PRO. *Cardiology Clinics*, **2010**, 28, 191-7	2.5	23
81	Endovascular thoracic aortic repair in confirmed or suspected genetically triggered thoracic aortic dissection. *Journal of Vascular Surgery*, **2018**, 68, 364-371	3.5	21
80	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. *Genetics in Medicine*, **2019**, 21, 144-151	8.1	20
79	Spontaneous pneumothorax and hemothorax frequently precede the arterial and intestinal complications of vascular Ehlers-Danlos syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 797-802	2.5	19
78	Treatment guidelines for thoracic aortic aneurysms and dissections based on the underlying causative gene. *Journal of Thoracic and Cardiovascular Surgery*, **2010**, 140, S2-4; discussion S45-51	1.5	18
77	Recurrent Rare Genomic Copy Number Variants and Bicuspid Aortic Valve Are Enriched in Early Onset Thoracic Aortic Aneurysms and Dissections. *PLoS ONE*, **2016**, 11, e0153543	3.7	18
76	Precision medical and surgical management for thoracic aortic aneurysms and acute aortic dissections based on the causative mutant gene. *Journal of Cardiovascular Surgery*, **2016**, 57, 172-7	0.7	18
75	Susceptibility to acute thoracic aortic dissections in patients dying outside the hospital: an autopsy study. *American Heart Journal*, **2011**, 162, 474-9	4.9	17
74	An FBN1 pseudoexon mutation in a patient with Marfan syndrome: confirmation of cryptic mutations leading to disease. *Journal of Human Genetics*, **2008**, 53, 1007-1011	4.3	17
73	Marfan syndrome. *Nature Reviews Disease Primers*, **2021**, 7, 64	51.1	17
72	Diffuse and uncontrolled vascular smooth muscle cell proliferation in rapidly progressing pediatric moyamoya disease. *Journal of Neurosurgery: Pediatrics*, **2010**, 6, 244-9	2.1	16

71	FBN1 exon 2 splicing error in a patient with Marfan syndrome. *American Journal of Medical Genetics Part A*, **2001**, 101, 130-4		16
70	Cholesterol-Induced Phenotypic Modulation of Smooth Muscle Cells to Macrophage/Fibroblast-like Cells Is Driven by an Unfolded Protein Response. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2021**, 41, 302-316	9.4	16
69	Variants of Unknown Significance in Genes Associated with Heritable Thoracic Aortic Disease Can Be Low Penetrant "Risk Variants". *American Journal of Human Genetics*, **2018**, 103, 138-143	11	16
68	In Vitro Lineage-Specific Differentiation of Vascular Smooth Muscle Cells in Response to SMAD3 Deficiency: Implications for SMAD3-Related Thoracic Aortic Aneurysm. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2020**, 40, 1651-1663	9.4	15
67	Aortic pathology from protein kinase G activation is prevented by an antioxidant vitamin B analog. *Nature Communications*, **2019**, 10, 3533	17.4	15
66	Familial thoracic aortic aneurysms and dissections: three families with early-onset ascending and descending aortic dissections in women. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1196-202	2.5	15
65	Chronobiology of Acute Aortic Dissection in the Marfan Syndrome (from the National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions and the International Registry of Acute Aortic Dissection). *American Journal of Cardiology*, **2017**, 119, 785-789	3	13
64	SMAD4 rare variants in individuals and families with thoracic aortic aneurysms and dissections. *European Journal of Human Genetics*, **2019**, 27, 1054-1060	5.3	13
63	A multi-institutional experience in the aortic and arterial pathology in individuals with genetically confirmed vascular Ehlers-Danlos syndrome. *Journal of Vascular Surgery*, **2019**, 70, 1543-1554	3.5	12
62	A dimorphic Alu Sb-like insertion in COL3A1 is ethnic-specific. *Journal of Molecular Evolution*, **1996**, 42, 117-23	3.1	12
61	Fatal thoracic aortic aneurysm and dissection in a large family with a novel MYLK gene mutation: delineation of the clinical phenotype. *Orphanet Journal of Rare Diseases*, **2018**, 13, 41	4.2	11
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