Dianna M Milewicz

List of Publications by Year in Descending Order

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	Proteomic analysis of descending thoracic aorta identifies unique and universal signatures of aneurysm and dissection.. *JVS Vascular Science*, **2022**, 3, 85-181	1.3	0
213	Aortic dissection in pregnancy and the postpartum period.. *Seminars in Vascular Surgery*, **2022**, 35, 60-68	1.2	2
212	Current state and future directions of genomic medicine in aortic dissection: A path to prevention and personalized care.. *Seminars in Vascular Surgery*, **2022**, 35, 51-59	1.2	2
211	A mixed method approach to understanding the impact of COVID-19 on patients with or at risk for aortic dissection.. *Seminars in Vascular Surgery*, **2022**, 35, 100-109	1.2	1
210	Patient-Specific Modeling Could Predict Occurrence of Pediatric Stroke.. *Frontiers in Physiology*, **2022**, 13, 846404	4.6	0
209	The Secrets of the Frogs Heart.. *Pediatric Cardiology*, **2022**, 1	2.1	1
208	The annual ASHG dinner.. *American Journal of Human Genetics*, **2022**, 109, 377-378	11	1
207	Extracellular matrix genetics of thoracic and abdominal aortic diseases **2022**, 351-359		
206	Cerebrovascular Disease Progression in Patients With Arg179 Pathogenic Variants. *Neurology*, **2021**, 96, e538-e552	6.5	5
205	Aortic root dilatation and dilated cardiomyopathy in an adult with Tatton-Brown-Rahman syndrome. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	0
204	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
203	Update on the genetic risk for thoracic aortic aneurysms and acute aortic dissections: implications for clinical care. *Journal of Cardiovascular Surgery*, **2021**, 62, 203-210	0.7	1
202	Risk of sudden cardiac death in EXOSC5-related disease. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2532-2540	2.5	1
201	Germline variants in HEY2 functional domains lead to congenital heart defects and thoracic aortic aneurysms. *Genetics in Medicine*, **2021**, 23, 103-110	8.1	3
200	Pathogenic variants in THSD4, encoding the ADAMTS-like 6 protein, predispose to inherited thoracic aortic aneurysm. *Genetics in Medicine*, **2021**, 23, 111-122	8.1	7
199	Association of De Novo Variants With Childhood Onset Moyamoya Disease and Diffuse Occlusive Vasculopathy. *Neurology*, **2021**, 96, e1783-e1791	6.5	5
198	Image-based patient-specific flow simulations are consistent with stroke in pediatric cerebrovascular disease. *Biomechanics and Modeling in Mechanobiology*, **2021**, 20, 2071-2084	3.8	1

197	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. *PLoS Genetics*, **2021**, 17, e1009679	6	1
196	Summary: international consensus statement on nomenclature and classification of the congenital bicuspid aortic valve and its aortopathy, for clinical, surgical, interventional and research purposes. *European Journal of Cardio-thoracic Surgery*, **2021**, 60, 481-496	3	1
195	International consensus statement on nomenclature and classification of the congenital bicuspid aortic valve and its aortopathy, for clinical, surgical, interventional and research purposes. *European Journal of Cardio-thoracic Surgery*, **2021**, 60, 448-476	3	5
194	International Consensus Statement on Nomenclature and Classification of the Congenital Bicuspid Aortic Valve and Its Aortopathy, for Clinical, Surgical, Interventional and Research Purposes. *Radiology: Cardiothoracic Imaging*, **2021**, 3, e200496	8.3	2
193	Human SMAD4 Genomic Variants Identified in Individuals with Heritable and Early-Onset Thoracic Aortic Disease. *Neurology International*, **2021**, 11, 132-138	0	
192	DIAPH1 Variants in Non-East Asian Patients With Sporadic Moyamoya Disease. *JAMA Neurology*, **2021**, 78, 993-1003	17.2	7
191	International Consensus Statement on Nomenclature and Classification of the Congenital Bicuspid Aortic Valve and Its Aortopathy, for Clinical, Surgical, Interventional and Research Purposes. *Annals of Thoracic Surgery*, **2021**, 112, e203-e235	2.7	3
190	International consensus statement on nomenclature and classification of the congenital bicuspid aortic valve and its aortopathy, for clinical, surgical, interventional and research purposes. *Journal of Thoracic and Cardiovascular Surgery*, **2021**, 162, e383-e414	1.5	9
189	Regulatory variants in TCF7L2 are associated with thoracic aortic aneurysm. *American Journal of Human Genetics*, **2021**, 108, 1578-1589	11	2
188	Summary: International consensus statement on nomenclature and classification of the congenital bicuspid aortic valve and its aortopathy, for clinical, surgical, interventional, and research purposes. *Journal of Thoracic and Cardiovascular Surgery*, **2021**, 162, 781-797	1.5	0
187	Marfan syndrome. *Nature Reviews Disease Primers*, **2021**, 7, 64	51.1	17
186	Midterm outcomes of aortic root surgery in patients with Marfan syndrome: A prospective, multicenter, comparative study. *Journal of Thoracic and Cardiovascular Surgery*, **2021**,	1.5	3
185	Summary: International Consensus Statement on Nomenclature and Classification of the Congenital Bicuspid Aortic Valve and Its Aortopathy, for Clinical, Surgical, Interventional and Research Purposes. *Annals of Thoracic Surgery*, **2021**, 112, 1005-1022	2.7	0
184	Resistance of Acta2 mice to aortic disease is associated with defective release of mutant smooth muscle β-actin from the chaperonin-containing TCP1 folding complex. *Journal of Biological Chemistry*, **2021**, 297, 101228	5.4	1
183	Actin R256 Mono-methylation Is a Conserved Post-translational Modification Involved in Transcription. *Cell Reports*, **2020**, 32, 108172	10.6	4
182	TGFBR1 Rare Variant Associated With Thoracic Aortic Aneurysm, Double Chamber Left Ventricle, Coronary Anomaly, and Inducible Ventricular Tachycardia. *Circulation: Cardiovascular Imaging*, **2020**, 13, e010084	3.9	0
181	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
180	Heritable Thoracic Aortic Disease: Single Gene Disorders Predisposing to Thoracic Aortic Aneurysms and Acute Aortic Dissections **2020**, 175-195		1

179	Critical Role of Cytosolic DNA and Its Sensing Adaptor STING in Aortic Degeneration, Dissection, and Rupture. *Circulation*, **2020**, 141, 42-66	16.7	45
178	Type B Aortic Dissection in Young Individuals With Confirmed and Presumed Heritable Thoracic Aortic Disease. *Annals of Thoracic Surgery*, **2020**, 109, 534-540	2.7	6
177	Epac1 (Exchange Protein Directly Activated by cAMP 1) Upregulates LOX-1 (Oxidized Low-Density Lipoprotein Receptor 1) to Promote Foam Cell Formation and Atherosclerosis Development. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2020**, 40, e322-e335	9.4	10
176	Rare deleterious variants of NOTCH1, GATA4, SMAD6, and ROBO4 are enriched in BAV with early onset complications but not in BAV with heritable thoracic aortic disease. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1406	2.3	5
175	A multi-institutional experience in vascular Ehlers-Danlos syndrome diagnosis. *Journal of Vascular Surgery*, **2020**, 71, 149-157	3.5	9
174	The pleiotropy associated with de novo variants in CHD4, CNOT3, and SETD5 extends to moyamoya angiopathy. *Genetics in Medicine*, **2020**, 22, 427-431	8.1	10
173	Therapies for Thoracic Aortic Aneurysms and Acute Aortic Dissections. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2019**, 39, 126-136	9.4	48
172	Reply: More Genes for Thoracic Aortic Aneurysms and Dissections. *Journal of the American College of Cardiology*, **2019**, 73, 529-530	15.1	
171	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
170	Clinical Implications of Identifying Pathogenic Variants in Individuals With Thoracic Aortic Dissection. *Circulation Genomic and Precision Medicine*, **2019**, 12, e002476	5.2	27
169	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
168	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
167	The natural history of type B aortic dissection in patients with PRKG1 mutation c.530G>A (p.Arg177Gln). *Journal of Vascular Surgery*, **2019**, 70, 718-723	3.5	7
166	Phosphatidic acid generated by PLD2 promotes the plasma membrane recruitment of IQGAP1 and neointima formation. *FASEB Journal*, **2019**, 33, 6713-6725	0.9	6
165	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
164	Genetics of Thoracic and Abdominal Aortic Diseases. *Circulation Research*, **2019**, 124, 588-606	15.7	112
163	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. *Genetics in Medicine*, **2019**, 21, 144-151	8.1	20
162	Aortic pathology from protein kinase G activation is prevented by an antioxidant vitamin B analog. *Nature Communications*, **2019**, 10, 3533	17.4	15

161	Grange syndrome due to homozygous YY1AP1 missense rare variants. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2500-2505	2.5	2
160	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
159	Do Variants Predispose to Thoracic Aortic Aneurysms and Dissections?. *Circulation Genomic and Precision Medicine*, **2019**, 12, e002626	5.2	4
158	Reversal of Aortic Enlargement Induced by Increased Biomechanical Forces Requires AT1R Inhibition in Conjunction With AT2R Activation. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2019**, 39, 459-466	9.4	8
157	Genome-wide analysis yields new loci associating with aortic valve stenosis. *Nature Communications* , **2018**, 9, 987	17.4	56
156	Ari-1 Regulates Myonuclear Organization Together with Parkin and Is Associated with Aortic Aneurysms. *Developmental Cell*, **2018**, 45, 226-244.e8	10.2	33
155	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
154	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
153	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	9.4	151
152	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
151	Endovascular thoracic aortic repair in confirmed or suspected genetically triggered thoracic aortic dissection. *Journal of Vascular Surgery*, **2018**, 68, 364-371	3.5	21
150	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
149	Genetic approaches to identify pathological limitations in aortic smooth muscle contraction. *PLoS ONE*, **2018**, 13, e0193769	3.7	4
148	LTBP3 Pathogenic Variants Predispose Individuals to Thoracic Aortic Aneurysms and Dissections. *American Journal of Human Genetics*, **2018**, 102, 706-712	11	34
147	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
146	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
145	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
144	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

143	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
142	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
141	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
140	Turner syndrome-specific and general population Z-scores are equivalent for most adults with Turner syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1094-1096	2.5	3
139	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
138	Critical Role of ADAMTS-4 in the Development of Sporadic Aortic Aneurysm and Dissection in Mice. *Scientific Reports*, **2017**, 7, 12351	4.9	43
137	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
136	Heritable Thoracic Aortic Disease Genes in Sporadic Aortic Dissection. *Journal of the American College of Cardiology*, **2017**, 70, 2728-2730	15.1	24
135	Asprosin is a centrally acting orexigenic hormone. *Nature Medicine*, **2017**, 23, 1444-1453	50.5	130
134	Vascular disease-causing mutation, smooth muscle actin R258C, dominantly suppresses functions of actin in human patient fibroblasts. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E5569-E5578	11.5	6
133	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
132	Genetic Variants in LRP1 and ULK4 Are Associated with Acute Aortic Dissections. *American Journal of Human Genetics*, **2016**, 99, 762-769	11	44
131	International Registry of Patients Carrying TGFBR1 or TGFBR2 Mutations: Results of the MAC (Montalcino Aortic Consortium). *Circulation: Cardiovascular Genetics*, **2016**, 9, 548-558		105
130	Shared Genetic Risk Factors of Intracranial, Abdominal, and Thoracic Aneurysms. *Journal of the American Heart Association*, **2016**, 5,	6	34
129	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
128	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. *Journal of Clinical Investigation*, **2016**, 126, 948-61	15.9	65
127	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
126	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

125	Whole-exome sequencing reveals an inherited R566X mutation of the epithelial sodium channel β subunit in a case of early-onset phenotype of Liddle syndrome. *Journal of Physical Education and Sports Management*, **2016**, 2, a001255	2.8	5
124	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
123	Asprosin, a Fasting-Induced Glucogenic Protein Hormone. *Cell*, **2016**, 165, 566-79	56.2	187
122	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. *Circulation Research*, **2016**, 118, 928-34	15.7	122
121	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
120	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
119	Childhood-Onset Essential Hypertension and the Family Structure. *Journal of Clinical Hypertension*, **2016**, 18, 431-8	2.3	6
118	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
117	Thoracic aortic dissection and rupture in conotruncal cardiac defects: A population-based study. *International Journal of Cardiology*, **2015**, 184, 521-527	3.2	26
116	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
115	Deficiency of MMP17/MT4-MMP proteolytic activity predisposes to aortic aneurysm in mice. *Circulation Research*, **2015**, 117, e13-26	15.7	36
114	Aortic Disease Presentation and Outcome Associated With ACTA2 Mutations. *Circulation: Cardiovascular Genetics*, **2015**, 8, 457-64		82
113	Role of mechanotransduction in vascular biology: focus on thoracic aortic aneurysms and dissections. *Circulation Research*, **2015**, 116, 1448-61	15.7	216
112	Bicuspid aortic valve aortopathy in adults: Incidence, etiology, and clinical significance. *International Journal of Cardiology*, **2015**, 201, 400-7	3.2	89
111	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
110	Molecular Genetics of Aortic Aneurysms and Aortic Dissections **2015**, 1-8		2
109	The defining pathology of the new clinical and histopathologic entity ACTA2-related cerebrovascular disease. *Acta Neuropathologica Communications*, **2015**, 3, 81	7.3	24
108	SMAD2 Mutations Are Associated with Arterial Aneurysms and Dissections. *Human Mutation*, **2015**, 36, 1145-9	4.7	65

107	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
106	MAT2A mutations predispose individuals to thoracic aortic aneurysms. *American Journal of Human Genetics*, **2015**, 96, 170-7	11	68
105	Rescuing the physician-scientist workforce: the time for action is now. *Journal of Clinical Investigation*, **2015**, 125, 3742-7	15.9	106
104	Cell biology. Dysfunctional mechanosensing in aneurysms. *Science*, **2014**, 344, 477-9	33.3	107
103	Advanced atherosclerosis is associated with increased medial degeneration in sporadic ascending aortic aneurysms. *Atherosclerosis*, **2014**, 232, 361-8	3.1	11
102	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
101	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
100	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
99	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
98	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
97	RNF213 rare variants in an ethnically diverse population with Moyamoya disease. *Stroke*, **2014**, 45, 3200-7	6.7	103
96	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
95	Acute aortic dissections with pregnancy in women with ACTA2 mutations. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 106-12	2.5	24
94	Clinical and biochemical profiles suggest fibromuscular dysplasia is a systemic disease with altered TGF-β expression and connective tissue features. *FASEB Journal*, **2014**, 28, 3313-24	0.9	51
93	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
92	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
91	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
90	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

89	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
88	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
87	Coagulation factor V(A2440G) causes east Texas bleeding disorder via TFPI *Journal of Clinical Investigation*, **2013**, 123, 3777-87	15.9	81
86	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
85	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
84	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
83	Genetic Variants in Smooth Muscle Contraction and Adhesion Genes Cause Thoracic Aortic Aneurysms and Dissections and Other Vascular Diseases **2012**, 1291-1300		
82	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
81	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
80	Eye features in three Danish patients with multisystemic smooth muscle dysfunction syndrome. *British Journal of Ophthalmology*, **2012**, 96, 1227-31	5.5	29
79	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
78	A novel distinctive cerebrovascular phenotype is associated with heterozygous Arg179 ACTA2 mutations. *Brain*, **2012**, 135, 2506-14	11.2	88
77	MicroRNAs, fibrotic remodeling, and aortic aneurysms. *Journal of Clinical Investigation*, **2012**, 122, 490-3	15.9	27
76	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
75	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
74	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
73	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
72	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

71	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
70	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
69	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
68	Recurrent chromosome 16p13.1 duplications are a risk factor for aortic dissections. *PLoS Genetics*, **2011**, 7, e1002118	6	67
67	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
66	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
65	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, [illegible] *Circulation*, **2010**, 121, e266-369	16.7	1569
64	TGFBR2 mutations alter smooth muscle cell phenotype and predispose to thoracic aortic aneurysms and dissections. *Cardiovascular Research*, **2010**, 88, 520-9	9.9	95
63	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
62	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
61	Genetic testing in aortic aneurysm disease: PRO. *Cardiology Clinics*, **2010**, 28, 191-7	2.5	23
60	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
59	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, [illegible]	15.1	980
58	Paucity of skeletal manifestations in Hispanic families with FBN1 mutations. *European Journal of Medical Genetics*, **2010**, 53, 80-4 [illegible] Interventional Radiology, Society of Thora. *Journal of the American College of Cardiology*, **2010**, 55, e27-e129	2.6	11
57	The revised Ghent nosology for the Marfan syndrome. *Journal of Medical Genetics*, **2010**, 47, 476-85	5.8	1282
56	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, [illegible]	3.9	89
55	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
54	Mutations in myosin light chain kinase cause familial aortic dissections. *American Journal of Human Genetics*, **2010**, 87, 701-7	11	228

53	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
52	Large deletions and uniparental disomy detected by SNP arrays in adults with thoracic aortic aneurysms and dissections. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2399-405	2.5	6
51	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
50	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
49	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 *Interventions*, **2010**, 76, E43-86	1.5	52
48	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
47	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
46	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
45	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
44	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
43	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
42	Mutations in smooth muscle alpha-actin (ACTA2) lead to thoracic aortic aneurysms and dissections. *Nature Genetics*, **2007**, 39, 1488-93	36.3	623
41	Severe aortic and arterial aneurysms associated with a TGFBR2 mutation. *Nature Clinical Practice Cardiovascular Medicine*, **2007**, 4, 167-71		55
40	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	5.6	210
39	Inherited Disorders of Connective Tissue **2007**, 2557-2565		1
38	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
37	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
36	Aneurysm syndromes caused by mutations in the TGF-beta receptor. *New England Journal of Medicine*, **2006**, 355, 788-98	59.2	1243

35	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
34	Long-term survival in a child with severe congenital contractural arachnodactyly, autism and severe intellectual disability. *Clinical Dysmorphology*, **2006**, 15, 95-9	0.9	3
33	Pathogenesis of thoracic and abdominal aortic aneurysms. *Annals of the New York Academy of Sciences*, **2006**, 1085, 339-52	6.5	171
32	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
31	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
30	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
29	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
28	Mutations in transforming growth factor-beta receptor type II cause familial thoracic aortic aneurysms and dissections. *Circulation*, **2005**, 112, 513-20	16.7	296
27	Treatment of aortic disease in patients with Marfan syndrome. *Circulation*, **2005**, 111, e150-7	16.7	273
26	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
25	Familial Thoracic Aortic Aneurysms and Dissections **2004**, 113-122		
24	Pyrosequencing **2004**, 1104-1107		
23	Methodology for using a universal primer to label amplified DNA segments for molecular analysis. *Biotechnology Letters*, **2003**, 25, 2079-83	3	29
22	Profibrillin-1 maturation by human dermal fibroblasts: proteolytic processing and molecular chaperones. *Journal of Cellular Biochemistry*, **2003**, 90, 641-52	4.7	38
21	Mapping a locus for familial thoracic aortic aneurysms and dissections (TAAD2) to 3p24-25. *Circulation*, **2003**, 107, 3184-90	16.7	124
20	Ten novel FBN2 mutations in congenital contractural arachnodactyly: delineation of the molecular pathogenesis and clinical phenotype. *Human Mutation*, **2002**, 19, 39-48	4.7	89
19	Genetic basis of thoracic aortic aneurysms and dissections. *Current Opinion in Cardiology*, **2002**, 17, 677-83	2.1	49
18	FBN1 exon 2 splicing error in a patient with Marfan syndrome. *American Journal of Medical Genetics Part A*, **2001**, 101, 130-4		16

17	Association of fibrillin 1 single-nucleotide polymorphism haplotypes with systemic sclerosis in Choctaw and Japanese populations. *Arthritis and Rheumatism*, **2001**, 44, 893-901		105
16	Systemic sclerosis (scleroderma): specific autoantigen genes are selectively overexpressed in scleroderma fibroblasts. *Journal of Immunology*, **2001**, 167, 7126-33	5.3	62
15	Familial thoracic aortic aneurysms and dissections: genetic heterogeneity with a major locus mapping to 5q13-14. *Circulation*, **2001**, 103, 2461-8	16.7	177
14	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
13	Genetics of Cardiovascular Disease. *Circulation*, **2000**, 102,	16.7	3
12	Processing of the fibrillin-1 carboxyl-terminal domain. *Journal of Biological Chemistry*, **1999**, 274, 8933-40	5.4	62
11	Reduced penetrance and variable expressivity of familial thoracic aortic aneurysms/dissections. *American Journal of Cardiology*, **1998**, 82, 474-9	3	125
10	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
9	Familial thoracic aortic dilatations and dissections: a case control study. *Journal of Vascular Surgery*, **1997**, 25, 506-11	3.5	173
8	A dimorphic Alu Sb-like insertion in COL3A1 is ethnic-specific. *Journal of Molecular Evolution*, **1996**, 42, 117-23	3.1	12
7	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
6	Fibrillin-1 (FBN1) mutations in patients with thoracic aortic aneurysms. *Circulation*, **1996**, 94, 2708-11	16.7	139
5	Fibrillin-2 (FBN2) mutations result in the Marfan-like disorder, congenital contractural arachnodactyly. *Nature Genetics*, **1995**, 11, 456-8	36.3	235
4	Distinct skeletal abnormalities in four girls with Shprintzen-Goldberg syndrome. *American Journal of Medical Genetics Part A*, **1995**, 57, 565-72		30
3	Marfan syndrome. Long-term survival and complications after aortic aneurysm repair. *Circulation*, **1995**, 91, 728-33	16.7	216
2	Dolichonychia in a patient with the Marfan syndrome. *Journal of Dermatology*, **1993**, 20, 779-82	1.6	5
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