Harry C Dietz

List of Publications by Year
in descending order

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115 papers	21,213 citations	32410 55 h-index	28425 109 g-index
120 all docs	120 docs citations	120 times ranked	15321 citing authors

#	Article	IF	Citations
1	Midterm outcomes of aortic root surgery in patients with Marfan syndrome: A prospective, multicenter, comparative study. Journal of Thoracic and Cardiovascular Surgery, 2023, 165, 1790-1799.e12.	0.4	14
2	Predictors of low bone density and fracture risk in Loeysâ€“Dietz syndrome. Genetics in Medicine, 2022, 24, 419-429.	1.1	3
3	Accurate assignment of disease liability to genetic variants using only population data. Genetics in Medicine, 2022, 24, 87-99.	1.1	4
4	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.	1.1	13
5	Assessment of pleural pressure during sleep in Marfan syndrome. Journal of Clinical Sleep Medicine, 2022, 18, 1583-1592.	1.4	4
6	Massive ductal aneurysm in an asymptomatic child with Loeys-Dietz syndrome. Annals of Pediatric Cardiology, 2021, 14, 113.	0.2	1
7	Toward precision medicine in vascular connective tissue disorders. American Journal of Medical Genetics, Part A, 2021, 185, 3340-3349.	0.7	2
8	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	2.6	3
9	Association of sleep apnoea risk and aortic enlargement in Marfan syndrome. BMJ Open Respiratory Research, 2021, 8, e000942.	1.2	5
10	Loeys-Dietz Syndrome. Advances in Experimental Medicine and Biology, 2021, 1348, 251-264.	0.8	16
11	Management of the aortic arch in patients with Loeysâ€“Dietz syndrome. Journal of Thoracic and Cardiovascular Surgery, 2020, 160, 1166-1175.	0.4	27
12	A seX(X/Y) Article on Marfan Syndrome. Journal of the American Heart Association, 2020, 9, e018814.	1.6	9
13	A positively selected FBN1 missense variant reduces height in Peruvian individuals. Nature, 2020, 582, 234-239.	13.7	39
14	Safety and outcome of gastrostomy tube placement in patients with Loeys-Dietz syndrome. BMC Gastroenterology, 2020, 20, 71.	0.8	1
15	Hereditary connective tissue disorders. , 2020, , 127-145.		1
16	Calpain 9 as a therapeutic target in TGFÎ²-induced mesenchymal transition and fibrosis. Science Translational Medicine, 2019, 11, .	5.8	30
17	Epigenetic activation and memory at a TGFB2 enhancer in systemic sclerosis. Science Translational Medicine, 2019, 11, .	5.8	47
18	Regenerative and durable small-diameter graft as an arterial conduit. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 12710-12719.	3.3	52

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19	Oxytocin antagonism prevents pregnancy-associated aortic dissection in a mouse model of Marfan syndrome. Science Translational Medicine, 2019, 11, .	5.8	42
20	Targeting of dermal myofibroblasts through death receptor 5 arrests fibrosis in mouse models of scleroderma. Nature Communications, 2019, 10, 1128.	5.8	28
21	Confirmation of the role of pathogenic SMAD6 variants in bicuspid aortic valve-related aortopathy. European Journal of Human Genetics, 2019, 27, 1044-1053.	1.4	32
22	Copy number variation analysis in bicuspid aortic valve-related aortopathy identifies TBX20 as a contributing gene. European Journal of Human Genetics, 2019, 27, 1033-1043.	1.4	24
23	Valve-sparing aortic root replacement in children: Outcomes from 100 consecutive cases. Journal of Thoracic and Cardiovascular Surgery, 2019, 157, 1100-1109.	0.4	23
24	Chronic mucocutaneous candidiasis and connective tissue disorder in humans with impaired JNK1-dependent responses to IL-17A/F and TGF-Î². Science Immunology, 2019, 4, .	5.6	45
25	ROBO4 variants predispose individuals to bicuspid aortic valve and thoracic aortic aneurysm. Nature Genetics, 2019, 51, 42-50.	9.4	101
26	Lineage-specific events underlie aortic root aneurysm pathogenesis in Loeys-Dietz syndrome. Journal of Clinical Investigation, 2019, 129, 659-675.	3.9	81
27	Targetable cellular signaling events mediate vascular pathology in vascular Ehlers-Danlos syndrome. Journal of Clinical Investigation, 2019, 130, 686-698.	3.9	40
28	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	1.1	66
29	FEVR findings in patients with Loeys-Dietz syndrome type II. Ophthalmic Genetics, 2018, 39, 754-758.	0.5	6
30	Pregnancy after Aortic Root Replacement in Marfan's Syndrome: A Case Series and Review of the Literature. AJP Reports, 2018, 08, e234-e240.	0.4	12
31	First evidence of maternally inherited mosaicism in TGFBR1 and subtle primary myocardial changes in Loeys-Dietz syndrome: a case report. BMC Medical Genetics, 2018, 19, 170.	2.1	4
32	Decreased mitochondrial respiration in aneurysmal aortas of Fibulin-4 mutant mice is linked to PGC1A regulation. Cardiovascular Research, 2018, 114, 1776-1793.	1.8	47
33	Proteomics reveals Rictor as a noncanonical TGF-Î² signaling target during aneurysm progression in Marfan mice. American Journal of Physiology - Heart and Circulatory Physiology, 2018, 315, H1112-H1126.	1.5	20
34	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
35	Carotid Artery Tortuosity Index Is Associated With the Need for Early Aortic Root Replacement in Patients With Loeys-Dietz Syndrome. Journal of Computer Assisted Tomography, 2018, 42, 747-753.	0.5	13
36	Proteomics Reveals Contextâ€Ðependent Activation of Rictor Signaling by TGFÎ² in Vascular Smooth Muscle Cells. FASEB Journal, 2018, 32, .	0.2	0

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37	TGF-Î² Family Signaling in Connective Tissue and Skeletal Diseases. Cold Spring Harbor Perspectives in Biology, 2017, 9, a022269.	2.3	86
38	2016 Presidential Address: Letâ€™s Make Human Genetics Great (Again): The Importance of Beauty in Science 1. American Journal of Human Genetics, 2017, 100, 379-384.	2.6	0
39	Ectopic calcification in pseudoxanthoma elasticum responds to inhibition of tissue-nonspecific alkaline phosphatase. Science Translational Medicine, 2017, 9, .	5.8	83
40	Aortic Root Replacement for Children With Loeys-Dietz Syndrome. Annals of Thoracic Surgery, 2017, 103, 1513-1518.	0.7	31
41	Cardiovascular operations for Loeys-Dietz syndrome: Intermediate-term results. Journal of Thoracic and Cardiovascular Surgery, 2017, 153, 406-412.	0.4	51
42	Simplified mitral valve repair in pediatric patients with connective tissue disorders. Journal of Thoracic and Cardiovascular Surgery, 2017, 153, 399-403.	0.4	11
43	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.	1.1	94
44	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.	1.3	85
45	Nonmyocyte ERK1/2 signaling contributes to load-induced cardiomyopathy in Marfan mice. JCI Insight, 2017, 2, .	2.3	44
46	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, .	1.6	71
47	Mesenchymal state of intimal cells may explain higher propensity to ascending aortic aneurysm in bicuspid aortic valves. Scientific Reports, 2016, 6, 35712.	1.6	36
48	Increased Prevalence of Inflammatory Bowel Disease in Patients with Mutations in Genes Encoding the Receptor Subunits for TGFÎ². Inflammatory Bowel Diseases, 2016, 22, 2058-2062.	0.9	15
49	Aortic Dissection in Patients With Genetically Mediated Aneurysms. Journal of the American College of Cardiology, 2016, 67, 2744-2754.	1.2	84
50	Long-term outcomes of aortic root operations for Marfan syndrome: A comparison of Bentall versus aortic valve-sparing procedures. Journal of Thoracic and Cardiovascular Surgery, 2016, 151, 330-338.	0.4	111
51	Dysregulated TGFâ€Î² signaling alters bone microstructure in a mouse model of Loeysâ€Dietz syndrome. Journal of Orthopaedic Research, 2015, 33, 1447-1454.	1.2	11
52	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. Human Mutation, 2015, 36, 808-814.	1.1	97
53	A deleterious gene-by-environment interaction imposed by calcium channel blockers in Marfan syndrome. ELife, 2015, 4, .	2.8	87
54	One integrin to rule them all?. Science Translational Medicine, 2015, 7, 288fs21.	5.8	11

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55	Potential Phenotypeâ€“Genotype Correlation in Marfan Syndrome. Circulation: Cardiovascular Genetics, 2015, 8, 256-260.	5.1	20
56	Multimodality Imaging of Diseases of the Thoracic Aorta in Adults: From the American Society of Echocardiography and the European Association of Cardiovascular Imaging. Journal of the American Society of Echocardiography, 2015, 28, 119-182.	1.2	500
57	Determinants of Aortic Root Dilatation and Reference Values Among Young Adults Over a 20-Year Period. Hypertension, 2015, 66, 23-29.	1.3	35
58	Mutations in a TGF-Î² Ligand, TGFB3, CauseÂSyndromic Aortic Aneurysms andÂDissections. Journal of the American College of Cardiology, 2015, 65, 1324-1336.	1.2	238
59	Design and rationale of a prospective, collaborative meta-analysis of all randomized controlled trials of angiotensin receptor antagonists in Marfan syndrome, based on individual patient data: A report from the Marfan Treatment Trialists' Collaboration. American Heart Journal, 2015, 169, 605-612.	1.2	44
60	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	1.4	48
61	Absence of Cardiovascular Manifestations in a Haploinsufficient Tgfbr1 Mouse Model. PLoS ONE, 2014, 9, e89749.	1.1	9
62	Tgfbr2 disruption in postnatal smooth muscle impairs aortic wall homeostasis. Journal of Clinical Investigation, 2014, 124, 755-767.	3.9	223
63	Angiotensin IIâ€“dependent TGF-Î² signaling contributes to Loeys-Dietz syndrome vascular pathogenesis. Journal of Clinical Investigation, 2014, 124, 448-460.	3.9	214
64	Response to Pyeritz et al.. Genetics in Medicine, 2014, 16, 642-644.	1.1	3
65	Atenolol versus Losartan in Children and Young Adults with Marfan's Syndrome. New England Journal of Medicine, 2014, 371, 2061-2071.	13.9	457
66	Clinical and biochemical profiles suggest fibromuscular dysplasia is a systemic disease with altered TGFâ€Î² expression and connective tissue features. FASEB Journal, 2014, 28, 3313-3324.	0.2	68
67	Histone deacetylase inhibition rescues structural and functional brain deficits in a mouse model of Kabuki syndrome. Science Translational Medicine, 2014, 6, 256ra135.	5.8	142
68	Loeysâ€“Dietz syndrome: a primer for diagnosis and management. Genetics in Medicine, 2014, 16, 576-587.	1.1	435
69	The Genetic Basis of Aortic Aneurysm. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a015909-a015909.	2.9	61
70	Mutations in SGOL1 cause a novel cohesinopathy affecting heart and gut rhythm. Nature Genetics, 2014, 46, 1245-1249.	9.4	98
71	Abstract 490: Vertebral Artery Tortuosity Index is a Novel Biomarker of Surgery and Aortic Dissection or Rupture in Children and Young Adults: Findings From the National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions. Arteriosclerosis, Thrombosis, and Vascular Biology, 2014, 34, .	1.1	1
72	Integrin-modulating therapy prevents fibrosis and autoimmunity in mouse models of scleroderma. Nature, 2013, 503, 126-130.	13.7	159

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73	TGFÎ² Receptor Mutations Impose a Strong Predisposition for Human Allergic Disease. Science Translational Medicine, 2013, 5, 195ra94.	5.8	165
74	Increased fracture risk and low bone mineral density in patients with loeysâ€“dietz syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1910-1914.	0.7	24
75	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. Nature Genetics, 2012, 44, 922-927.	9.4	391
76	Phenotypic spectrum of the SMAD3-related aneurysmsâ€“osteoarthritis syndrome. Journal of Medical Genetics, 2012, 49, 47-57.	1.5	221
77	Mutations in the TGF-Î² repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm. Nature Genetics, 2012, 44, 1249-1254.	9.4	237
78	Multi-Scale Biomechanical Remodeling in Aging and Genetic Mutant Murine Mitral Valve Leaflets: Insights into Marfan Syndrome. PLoS ONE, 2012, 7, e44639.	1.1	18
79	Lessons on the pathogenesis of aneurysm from heritable conditions. Nature, 2011, 473, 308-316.	13.7	411
80	Valve-Sparing Aortic Root Replacement in Loeys-Dietz Syndrome. Annals of Thoracic Surgery, 2011, 92, 556-561.	0.7	60
81	Noncanonical TGFÎ² Signaling Contributes to Aortic Aneurysm Progression in Marfan Syndrome Mice. Science, 2011, 332, 358-361.	6.0	422
82	Angiotensin II Type 2 Receptor Signaling Attenuates Aortic Aneurysm in Mice Through ERK Antagonism. Science, 2011, 332, 361-365.	6.0	414
83	New Therapeutic Approaches to Mendelian Disorders. New England Journal of Medicine, 2010, 363, 852-863.	13.9	84
84	TGF-Î² in the pathogenesis and prevention of disease: a matter of aneurysmic proportions. Journal of Clinical Investigation, 2010, 120, 403-406.	3.9	102
85	Aortic Root Replacement in 372 Marfan Patients: Evolution of Operative Repair Over 30 Years. Annals of Thoracic Surgery, 2009, 87, 1344-1350.	0.7	179
86	Histopathologic Findings in Ascending Aortas From Individuals With Loeys-Dietz Syndrome (LDS). American Journal of Surgical Pathology, 2009, 33, 194-201.	2.1	109
87	Angiotensin II Blockade and Aortic-Root Dilation in Marfan's Syndrome. New England Journal of Medicine, 2008, 358, 2787-2795.	13.9	767
88	Early Surgical Experience With Loeys-Dietz: A New Syndrome of Aggressive Thoracic Aortic Aneurysm Disease. Annals of Thoracic Surgery, 2007, 83, S757-S763.	0.7	254
89	Marfan Syndrome: From Molecules to Medicines*. American Journal of Human Genetics, 2007, 81, 662-667.	2.6	13
90	Familial thoracic aortic dilation and bicommissural aortic valve: A prospective analysis of natural history and inheritance. American Journal of Medical Genetics, Part A, 2007, 143A, 1960-1967.	0.7	176

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91	Angiotensin II type 1 receptor blockade attenuates TGF-Î²â€“induced failure of muscle regeneration in multiple myopathic states. Nature Medicine, 2007, 13, 204-210.	15.2	603
92	Losartan, an AT1 Antagonist, Prevents Aortic Aneurysm in a Mouse Model of Marfan Syndrome. Science, 2006, 312, 117-121.	6.0	1,591
93	Aneurysm Syndromes Caused by Mutations in the TGF-Î² Receptor. New England Journal of Medicine, 2006, 355, 788-798.	13.9	1,490
94	A syndrome of altered cardiovascular, craniofacial, neurocognitive and skeletal development caused by mutations in TGFBR1 or TGFBR2. Nature Genetics, 2005, 37, 275-281.	9.4	1,543
95	Recent progress towards a molecular understanding of Marfan syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 139C, 4-9.	0.7	176
96	Evidence for a critical contribution of haploinsufficiency in the complex pathogenesis of Marfan syndrome. Journal of Clinical Investigation, 2004, 114, 172-181.	3.9	319
97	TGF-Î²â€“dependent pathogenesis of mitral valve prolapse in a mouse model of Marfan syndrome. Journal of Clinical Investigation, 2004, 114, 1586-1592.	3.9	467
98	Evidence for a critical contribution of haploinsufficiency in the complex pathogenesis of Marfan syndrome. Journal of Clinical Investigation, 2004, 114, 172-181.	3.9	225
99	Dysregulation of TGF-Î² activation contributes to pathogenesis in Marfan syndrome. Nature Genetics, 2003, 33, 407-411.	9.4	1,298
100	Characterization of microsatellite markers flankingFBN1: Utility in the diagnostic evaluation for Marfan syndrome. American Journal of Medical Genetics Part A, 2001, 99, 39-47.	2.4	12
101	A strategy for disease gene identification through nonsense-mediated mRNA decay inhibition. Nature Biotechnology, 2001, 19, 434-439.	9.4	171
102	Enzymatic mutation detection (EMD?) of novel mutations (R565X and R1523X) in the FBN1 gene of patients with Marfan syndrome using T4 endonuclease VII. Human Mutation, 2000, 16, 92-93.	1.1	18
103	Targetting of the gene encoding fibrillinâ€“1 recapitulates the vascular aspect of Marfan syndrome. Nature Genetics, 1997, 17, 218-222.	9.4	366
104	Revised diagnostic criteria for the Marfan syndrome. , 1996, 62, 417-426.		1,335
105	Mutation in fibrillin-1 and the Marfanoid-craniosynostosis (Shprintzen-Goldberg) syndrome. Nature Genetics, 1996, 12, 209-211.	9.4	179
106	Nonstop treatment of cystic fibrosis. Nature Medicine, 1996, 2, 608-608.	15.2	4
107	Revised diagnostic criteria for the Marfan syndrome. American Journal of Medical Genetics Part A, 1996, 62, 417-426.	2.4	48
108	The question of heterogeneity in Marfan syndrome. Nature Genetics, 1995, 9, 228-229.	9.4	51

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109	A missense mutation in type VII collagen in two affected siblings with recessive dystrophic epidermolysis bullosa. Nature Genetics, 1993, 4, 62-66.	9.4	200
110	Fabrillin (FBN1) mutations in Marfan syndrome. Human Mutation, 1992, 1, 79-79.	1.1	14
111	Clustering of fibrillin (FBN1) missense mutations in Marfan syndrome patients at cysteine residues in EGF-like domains. Human Mutation, 1992, 1, 366-374.	1.1	131
112	The Marfan syndrome locus: Confirmation of assignment to chromosome 15 and identification of tightly linked markers at 15q15-q21.3. Genomics, 1991, 9, 355-361.	1.3	185
113	Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. Nature, 1991, 352, 337-339.	13.7	1,901
114	Marfan Syndrome and Other Microfibrillar Disorders. , 0, , 585-626.		20
115	Connective Tissue Disorders. , 0, , 537-546.		0