Catherine Boileau

List of Publications by Citations

182 papers	16,630 citations	61 h-index	127 g-index
198 ext. papers	19,866 ext. citations	7.1 avg, IF	5.61 L-index

#	Paper	IF	Citations
182	2014 ESC Guidelines on the diagnosis and treatment of aortic diseases: Document covering acute and chronic aortic diseases of the thoracic and abdominal aorta of the adult. The Task Force for the Diagnosis and Treatment of Aortic Diseases of the European Society of Cardiology (ESC). *European Heart Journal*, **2014**, 35, 2873-926	9.5	2334
181	Mutations in PCSK9 cause autosomal dominant hypercholesterolemia. *Nature Genetics*, **2003**, 34, 154-6	36.3	2025
180	Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease: consensus statement of the European Atherosclerosis Society. *European Heart Journal*, **2013**, 34, 3478-90a	9.5	1551
179	Homozygous familial hypercholesterolaemia: new insights and guidance for clinicians to improve detection and clinical management. A position paper from the Consensus Panel on Familial Hypercholesterolaemia of the European Atherosclerosis Society. *European Heart Journal*, **2014**, 35, 2146-57	9.5	614
178	Heterozygous TGFBR2 mutations in Marfan syndrome. *Nature Genetics*, **2004**, 36, 855-60	36.3	509
177	NARC-1/PCSK9 and its natural mutants: zymogen cleavage and effects on the low density lipoprotein (LDL) receptor and LDL cholesterol. *Journal of Biological Chemistry*, **2004**, 279, 48865-75	5.4	466
176	Familial hypercholesterolaemia in children and adolescents: gaining decades of life by optimizing detection and treatment. *European Heart Journal*, **2015**, 36, 2425-37	9.5	430
175	Mutations in STAT3 and IL12RB1 impair the development of human IL-17-producing T cells. *Journal of Experimental Medicine*, **2008**, 205, 1543-50	16.6	361
174	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
173	Update of the UMD-FBN1 mutation database and creation of an FBN1 polymorphism database. *Human Mutation*, **2003**, 22, 199-208	4.7	251
172	Genome-wide scan identifies TNIP1, PSORS1C1, and RHOB as novel risk loci for systemic sclerosis. *PLoS Genetics*, **2011**, 7, e1002091	6	176
171	Mutations and polymorphisms in the proprotein convertase subtilisin kexin 9 (PCSK9) gene in cholesterol metabolism and disease. *Human Mutation*, **2009**, 30, 520-9	4.7	175
170	MUC5B Promoter Variant and Rheumatoid Arthritis with Interstitial Lung Disease. *New England Journal of Medicine*, **2018**, 379, 2209-2219	59.2	173
169	Mutations in the TGF□binding-protein-like domain 5 of FBN1 are responsible for acromicric and geleophysic dysplasias. *American Journal of Human Genetics*, **2011**, 89, 7-14	11	171
168	C57BL/6J and A/J mice fed a high-fat diet delineate components of metabolic syndrome. *Obesity*, **2007**, 15, 1996-2005	8	169
167	Comparison of clinical presentations and outcomes between patients with TGFBR2 and FBN1 mutations in Marfan syndrome and related disorders. *Circulation*, **2009**, 120, 2541-9	16.7	160
166	UMD (Universal mutation database): a generic software to build and analyze locus-specific databases. *Human Mutation*, **2000**, 15, 86-94	4.7	159

165	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
164	Apolipoprotein B100 metabolism in autosomal-dominant hypercholesterolemia related to mutations in PCSK9. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2004**, 24, 1448-53	9.4	153
163	A second locus for Marfan syndrome maps to chromosome 3p24.2-p25. *Nature Genetics*, **1994**, 8, 264-8	36.3	149
162	Novel mutations of the PCSK9 gene cause variable phenotype of autosomal dominant hypercholesterolemia. *Human Mutation*, **2005**, 26, 497	4.7	142
161	Marfan Sartan: a randomized, double-blind, placebo-controlled trial. *European Heart Journal*, **2015**, 36, 2160-6	9.5	134
160	A third major locus for autosomal dominant hypercholesterolemia maps to 1p34.1-p32. *American Journal of Human Genetics*, **1999**, 64, 1378-87	11	126
159	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. *Circulation Research*, **2016**, 118, 928-34	15.7	122
158	Clinical and molecular study of 320 children with Marfan syndrome and related type I fibrillinopathies in a series of 1009 probands with pathogenic FBN1 mutations. *Pediatrics*, **2009**, 123, 391-8	7.4	120
157	Cohort profile of the CARTaGENE study: Quebec's population-based biobank for public health and personalized genomics. *International Journal of Epidemiology*, **2013**, 42, 1285-99	7.8	117
156	Quality, quantity and harmony: the DataSHaPER approach to integrating data across bioclinical studies. *International Journal of Epidemiology*, **2010**, 39, 1383-93	7.8	117
155	Aortic event rate in the Marfan population: a cohort study. *Circulation*, **2012**, 125, 226-32	16.7	117
154	In vivo evidence that furin from hepatocytes inactivates PCSK9. *Journal of Biological Chemistry*, **2011**, 286, 4257-63	5.4	110
153	Nomograms for aortic root diameters in children using two-dimensional echocardiography. *American Journal of Cardiology*, **2010**, 105, 888-94	3	110
152	International Registry of Patients Carrying TGFBR1 or TGFBR2 Mutations: Results of the MAC (Montalcino Aortic Consortium). *Circulation: Cardiovascular Genetics*, **2016**, 9, 548-558		105
151	Identification in daily practice of patients with Lynch syndrome (hereditary nonpolyposis colorectal cancer): revised Bethesda guidelines-based approach versus molecular screening. *American Journal of Gastroenterology*, **2008**, 103, 2825-35; quiz 2836	0.7	101
150	Cardiovascular manifestations in men and women carrying a FBN1 mutation. *European Heart Journal*, **2010**, 31, 2223-9	9.5	98
149	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
148	Heterozygous RTEL1 mutations are associated with familial pulmonary fibrosis. *European Respiratory Journal*, **2015**, 46, 474-85	13.6	96

147	Molecular genetics of Marfan syndrome. *Current Opinion in Cardiology*, **2005**, 20, 194-200	2.1	96
146	Parental origin and germline mosaicism of deletions and duplications of the dystrophin gene: a European study. *Human Genetics*, **1992**, 88, 249-57	6.3	92
145	UMD (Universal Mutation Database): 2005 update. *Human Mutation*, **2005**, 26, 184-91	4.7	91
144	Shared genetic predisposition in rheumatoid arthritis-interstitial lung disease and familial pulmonary fibrosis. *European Respiratory Journal*, **2017**, 49,	13.6	89
143	Identification and in silico analyses of novel TGFBR1 and TGFBR2 mutations in Marfan syndrome-related disorders. *Human Mutation*, **2006**, 27, 760-9	4.7	89
142	The UMD-LDLR database: additions to the software and 490 new entries to the database. *Human Mutation*, **2002**, 20, 81-7	4.7	89
141	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
140	Molecular spectrum of autosomal dominant hypercholesterolemia in France. *Human Mutation*, **2010** , 31, E1811-24	4.7	86
139	Description of a large family with autosomal dominant hypercholesterolemia associated with the APOE p.Leu167del mutation. *Human Mutation*, **2013**, 34, 83-7	4.7	84
138	Aortic Disease Presentation and Outcome Associated With ACTA2 Mutations. *Circulation: Cardiovascular Genetics*, **2015**, 8, 457-64		82
137	Molecular analysis and intestinal expression of SAR1 genes and proteins in Anderson's disease (Chylomicron retention disease). *Orphanet Journal of Rare Diseases*, **2011**, 6, 1	4.2	82
136	Maternal complication of pregnancy in Marfan syndrome. *International Journal of Cardiology*, **2009**, 136, 156-61	3.2	81
135	In-frame mutations in exon 1 of SKI cause dominant Shprintzen-Goldberg syndrome. *American Journal of Human Genetics*, **2012**, 91, 950-7	11	80
134	Characterization of Autosomal Dominant Hypercholesterolemia Caused by PCSK9 Gain of Function Mutations and Its Specific Treatment With Alirocumab, a PCSK9 Monoclonal Antibody. *Circulation: Cardiovascular Genetics*, **2015**, 8, 823-31		76
133	LTBP2 mutations cause Weill-Marchesani and Weill-Marchesani-like syndrome and affect disruptions in the extracellular matrix. *Human Mutation*, **2012**, 33, 1182-7	4.7	73
132	Living the PCSK9 adventure: from the identification of a new gene in familial hypercholesterolemia towards a potential new class of anticholesterol drugs. *Current Atherosclerosis Reports*, **2014**, 16, 439	6	72
131	Identification and characterization of new gain-of-function mutations in the PCSK9 gene responsible for autosomal dominant hypercholesterolemia. *Atherosclerosis*, **2012**, 223, 394-400	3.1	72
130	UMD-predictor, a new prediction tool for nucleotide substitution pathogenicity -- application to four genes: FBN1, FBN2, TGFBR1, and TGFBR2. *Human Mutation*, **2009**, 30, 952-9	4.7	72

129	The Proprotein Convertases in Hypercholesterolemia and Cardiovascular Diseases: Emphasis on Proprotein Convertase Subtilisin/Kexin 9. *Pharmacological Reviews*, **2017**, 69, 33-52	22.5	70
128	MAT2A mutations predispose individuals to thoracic aortic aneurysms. *American Journal of Human Genetics*, **2015**, 96, 170-7	11	68
127	Identification of 23 TGFBR2 and 6 TGFBR1 gene mutations and genotype-phenotype investigations in 457 patients with Marfan syndrome type I and II, Loeys-Dietz syndrome and related disorders. *Human Mutation*, **2008**, 29, E284-95	4.7	68
126	The molecular basis of familial hypercholesterolemia in Lebanon: spectrum of LDLR mutations and role of PCSK9 as a modifier gene. *Human Mutation*, **2009**, 30, E682-91	4.7	67
125	Identification of the minimal combination of clinical features in probands for efficient mutation detection in the FBN1 gene. *European Journal of Human Genetics*, **2009**, 17, 1121-8	5.3	67
124	Marfan syndrome in the third Millennium. *European Journal of Human Genetics*, **2002**, 10, 673-81	5.3	66
123	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. *Journal of Clinical Investigation*, **2016**, 126, 948-61	15.9	65
122	Dissection in Marfan syndrome: the importance of the descending aorta. *European Heart Journal*, **2011**, 32, 443-9	9.5	60
121	Rationale and design of a randomized clinical trial (Marfan Sartan) of angiotensin II receptor blocker therapy versus placebo in individuals with Marfan syndrome. *Archives of Cardiovascular Diseases*, **2010**, 103, 317-25	2.7	59
120	Resistance to high-fat diet in the female progeny of obese mice fed a control diet during the periconceptual, gestation, and lactation periods. *American Journal of Physiology - Endocrinology and Metabolism*, **2007**, 292, E1095-100	6	56
119	The translational science of Marfan syndrome. *Heart*, **2011**, 97, 1206-14	5.1	53
118	Evaluation and application of denaturing HPLC for mutation detection in Marfan syndrome: Identification of 20 novel mutations and two novel polymorphisms in the FBN1 gene. *Human Mutation*, **2002**, 19, 443-56	4.7	51
117	Brief report: candidate gene study in systemic sclerosis identifies a rare and functional variant of the TNFAIP3 locus as a risk factor for polyautoimmunity. *Arthritis and Rheumatism*, **2012**, 64, 2746-52		50
116	Phenotype-haplotype correlation of IRF5 in systemic sclerosis: role of 2 haplotypes in disease severity. *Journal of Rheumatology*, **2010**, 37, 987-92	4.1	50
115	Prognosis factors in probands with an FBN1 mutation diagnosed before the age of 1 year. *Pediatric Research*, **2011**, 69, 265-70	3.2	48
114	Angiogenic biomarkers predict the occurrence of digital ulcers in systemic sclerosis. *Annals of the Rheumatic Diseases*, **2012**, 71, 394-9	2.4	47
113	Genetic basis for systemic sclerosis. *Joint Bone Spine*, **2007**, 74, 577-83	2.9	46
112	Software and database for the analysis of mutations in the human LDL receptor gene. *Nucleic Acids Research*, **1997**, 25, 172-80	20.1	45

111	The Canadian Partnership for Tomorrow Project: a pan-Canadian platform for research on chronic disease prevention. *Cmaj*, **2018**, 190, E710-E717	3.5	44
110	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. *European Journal of Human Genetics*, **2018**, 26, 1759-1772	5.3	43
109	Independent replication establishes the CD247 gene as a genetic systemic sclerosis susceptibility factor. *Annals of the Rheumatic Diseases*, **2011**, 70, 1695-6	2.4	43
108	Plasma N-terminal pro-brain natriuretic peptide and brain natriuretic peptide in assessment of acute dyspnea. *Biomedicine and Pharmacotherapy*, **2005**, 59, 20-4	7.5	43
107	Calcium Signaling Pathway Genes RUNX2 and CACNA1C Are Associated With Calcific Aortic Valve Disease. *Circulation: Cardiovascular Genetics*, **2015**, 8, 812-22		42
106	Bone mineral density in Marfan syndrome. A large case-control study. *Joint Bone Spine*, **2006**, 73, 733-5	2.9	42
105	C8orf13-BLK is a genetic risk locus for systemic sclerosis and has additive effects with BANK1: results from a large french cohort and meta-analysis. *Arthritis and Rheumatism*, **2011**, 63, 2091-6		41
104	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
103	Mutations in human CPO gene predict clinical expression of either hepatic hereditary coproporphyria or erythropoietic harderoporphyria. *Human Molecular Genetics*, **2005**, 14, 3089-98	5.6	38
102	Study of phenotype evolution during childhood in Marfan syndrome to improve clinical recognition. *Genetics in Medicine*, **2014**, 16, 246-50	8.1	37
101	No evidence of somatic FGFR3 mutation in various types of carcinoma. *Oncogene*, **2001**, 20, 5059-61	9.2	37
100	Exome sequencing in suspected monogenic dyslipidemias. *Circulation: Cardiovascular Genetics*, **2015**, 8, 343-50		36
99	Enhanced late-outgrowth circulating endothelial progenitor cell levels in rheumatoid arthritis and correlation with disease activity. *Arthritis Research and Therapy*, **2010**, 12, R27	5.7	36
98	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, 605-12	4.9	35
97	The clinical presentation of Marfan syndrome is modulated by expression of wild-type FBN1 allele. *Human Molecular Genetics*, **2015**, 24, 2764-70	5.6	35
96	Genetics of thoracic aortic aneurysms. *Current Atherosclerosis Reports*, **2012**, 14, 219-26	6	35
95	Genetic background of systemic sclerosis: autoimmune genes take centre stage. *Rheumatology*, **2010**, 49, 203-10	3.9	35
94	Endothelial progenitor cells and rheumatic disorders. *Joint Bone Spine*, **2008**, 75, 131-7	2.9	34

93	Green space associations with mental health and cognitive function: Results from the Quebec CARTaGENE cohort. *Environmental Epidemiology*, **2019**, 3, e040	0.2	34
92	Methotrexate and rheumatoid arthritis associated interstitial lung disease. *European Respiratory Journal*, **2021**, 57,	13.6	34
91	Early-onset osteoarthritis, Charcot-Marie-Tooth like neuropathy, autoimmune features, multiple arterial aneurysms and dissections: an unrecognized and life threatening condition. *PLoS ONE*, **2014** , 9, e96387	3.7	33
90	Regulation of extrathymic T cell development and turnover by oncostatin M. *Journal of Immunology* , **2000**, 164, 5713-20	5.3	33
89	Independent replication and meta analysis of association studies establish TNFSF4 as a susceptibility gene preferentially associated with the subset of anticentromere-positive patients with systemic sclerosis. *Journal of Rheumatology*, **2012**, 39, 997-1003	4.1	32
88	A fourth locus for autosomal dominant hypercholesterolemia maps at 16q22.1. *European Journal of Human Genetics*, **2010**, 18, 1236-42	5.3	32
87	Demonstration of the recurrence of Marfan-like skeletal and cardiovascular manifestations due to germline mosaicism for an FBN1 mutation. *American Journal of Human Genetics*, **1999**, 65, 917-21	11	27
86	Clinical utility gene card for: Hereditary thoracic aortic aneurysm and dissection including next-generation sequencing-based approaches. *European Journal of Human Genetics*, **2016**, 24, e1-5	5.3	25
85	A regulatory variant in CCR6 is associated with susceptibility to antitopoisomerase-positive systemic sclerosis. *Arthritis and Rheumatism*, **2013**, 65, 3202-8		25
84	A new locus-specific database (LSDB) for mutations in the TGFBR2 gene: UMD-TGFBR2. *Human Mutation*, **2008**, 29, 33-8	4.7	25
83	Rationale, design, and methods for Canadian alliance for healthy hearts and minds cohort study (CAHHM) - a Pan Canadian cohort study. *BMC Public Health*, **2016**, 16, 650	4.1	24
82	Insights into the pathogenesis of systemic sclerosis based on the gene expression profile of progenitor-derived endothelial cells. *Arthritis and Rheumatism*, **2011**, 63, 3552-62		24
81	Strategies for proprotein convertase subtilisin kexin 9 modulation: a perspective on recent patents. *Expert Opinion on Therapeutic Patents*, **2010**, 20, 1547-71	6.8	24
80	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
79	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	8.1	23
78	R3531C mutation in the apolipoprotein B gene is not sufficient to cause hypercholesterolemia. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2000**, 20, E76-82	9.4	23
77	Regulator of telomere length 1 () mutations are associated with heterogeneous pulmonary and extra-pulmonary phenotypes. *European Respiratory Journal*, **2019**, 53,	13.6	23
76	Homozygous and compound heterozygous mutations in the FBN1 gene: unexpected findings in molecular diagnosis of Marfan syndrome. *Journal of Medical Genetics*, **2017**, 54, 100-103	5.8	21

75	A prognostic model for HIV seroconversion among injection drug users as a tool for stratification in clinical trials. *Journal of Acquired Immune Deficiency Syndromes (1999)*, **2005**, 39, 489-95	3.1	21
74	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. *Genetics in Medicine*, **2019**, 21, 144-151	8.1	20
73	Enhanced expression of ephrins and thrombospondins in the dermis of patients with early diffuse systemic sclerosis: potential contribution to perturbed angiogenesis and fibrosis. *Rheumatology*, **2011**, 50, 1494-504	3.9	20
72	Association study of ITGAM, ITGAX, and CD58 autoimmune risk loci in systemic sclerosis: results from 2 large European Caucasian cohorts. *Journal of Rheumatology*, **2011**, 38, 1033-8	4.1	20
71	PCSK9 Mutations in Familial Hypercholesterolemia: from a Groundbreaking Discovery to Anti-PCSK9 Therapies. *Current Atherosclerosis Reports*, **2017**, 19, 49	6	19
70	Effect of mutations in LDLR and PCSK9 genes on phenotypic variability in Tunisian familial hypercholesterolemia patients. *Atherosclerosis*, **2012**, 222, 158-66	3.1	19
69	Clinical utility gene card for: Marfan syndrome type 1 and related phenotypes [FBN1]. *European Journal of Human Genetics*, **2010**, 18,	5.3	19
68	Proprotein convertase subtilisin / kexin 9 (PCSK9) inhibitors and the future of dyslipidemia therapy: an updated patent review (2011-2015). *Expert Opinion on Therapeutic Patents*, **2016**, 26, 1377-1392	6.8	18
67	ELN gene triplication responsible for familial supravalvular aortic aneurysm. *Cardiology in the Young* , **2015**, 25, 712-7	1	17
66	Familial thoracic aortic aneurysms. *Current Opinion in Cardiology*, **2014**, 29, 492-8	2.1	17
65	Novel LRP5 gene mutation in a patient with osteoporosis-pseudoglioma syndrome. *Joint Bone Spine*, **2010**, 77, 151-3	2.9	17
64	Clinical relevance of genotype-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	8.1	17
63	Marfan syndrome. *Nature Reviews Disease Primers*, **2021**, 7, 64	51.1	17
62	Usefulness of the genetic risk score to identify phenocopies in families with familial hypercholesterolemia?. *European Journal of Human Genetics*, **2018**, 26, 570-578	5.3	16
61	Evaluation of predictive models in daily practice for the identification of patients with Lynch syndrome. *International Journal of Cancer*, **2012**, 130, 1367-77	7.5	16
60	Identification of secreted phosphoprotein 1 gene as a new rheumatoid arthritis susceptibility gene. *Annals of the Rheumatic Diseases*, **2015**, 74, e19	2.4	16
59	TGF receptor gene variants in systemic sclerosis-related pulmonary arterial hypertension: results from a multicentre EUSTAR study of European Caucasian patients. *Annals of the Rheumatic Diseases* , **2012**, 71, 1900-3	2.4	16
58	Updating the genetics of systemic sclerosis. *Current Opinion in Rheumatology*, **2010**, 22, 665-70	5.3	16

57	Changes in the lymph node microenvironment induced by oncostatin M. *Blood*, **2003**, 102, 1397-404	2.2	16
56	Expanding the skeletal phenotype of Loeys-Dietz syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1178-83	2.5	15
55	Familial hypercholesterolemia in Morocco: first report of mutations in the LDL receptor gene. *Journal of Human Genetics*, **2003**, 48, 199-203	4.3	15
54	Splicing mutation in the fibrillin-1 gene associated with neonatal Marfan syndrome and severe pulmonary emphysema with tracheobronchomalacia. *Pediatric Pulmonology*, **2005**, 39, 374-8	3.5	15
53	Polymorphic markers of the fibrillin-1 gene and systemic sclerosis in European Caucasian patients. *Journal of Rheumatology*, **2008**, 35, 643-9	4.1	15
52	A case-control study of cutaneous signs in adult patients with Marfan disease: diagnostic value of striae. *Journal of the American Academy of Dermatology*, **2011**, 64, 290-5	4.5	14
51	De novo 15q21.1q21.2 deletion identified through FBN1 MLPA and refined by 244K array-CGH in a female teenager with incomplete Marfan syndrome. *European Journal of Medical Genetics*, **2010**, 53, 208-12	2.6	14
50	Analytical correlation between plasma N-terminal pro-brain natriuretic peptide and brain natriuretic peptide in patients presenting with dyspnea. *Clinical Biochemistry*, **2004**, 37, 933-6	3.5	14
49	New Sequencing technologies help revealing unexpected mutations in Autosomal Dominant Hypercholesterolemia. *Scientific Reports*, **2018**, 8, 1943	4.9	13
48	Angiotensin-converting enzyme gene does not contribute to genetic susceptibility to systemic sclerosis in European Caucasians. *Journal of Rheumatology*, **2009**, 36, 337-40	4.1	13
47	In vivo corneal confocal microscopy in marfan syndrome. *Cornea*, **2007**, 26, 787-92	3.1	12
46	Pathogenic FBN1 Genetic Variation and Aortic Dissection in Patients With Marfan Syndrome. *Journal of the American College of Cardiology*, **2020**, 75, 843-853	15.1	10
45	Dermal tissue and cellular expression of fibrillin-1 in diffuse cutaneous systemic sclerosis. *Rheumatology*, **2010**, 49, 657-61	3.9	10
44	Association of metalloproteinase gene polymorphisms with systemic sclerosis in the European Caucasian population. *Journal of Rheumatology*, **2010**, 37, 599-602	4.1	10
43	APOE gene variants in primary dyslipidemia. *Atherosclerosis*, **2021**, 328, 11-22	3.1	10
42	Clinical Significance of Aortic Root Modification Associated With Bicuspid Aortic Valve in Marfan Syndrome. *Circulation: Cardiovascular Imaging*, **2019**, 12, e008129	3.9	9
41	Autosomal dominant type IIa hypercholesterolemia: evaluation of the respective contributions of LDLR and APOB gene defects as well as a third major group of defects. *European Journal of Human Genetics*, **2000**, 8, 621-30	5.3	9
40	Plasma proprotein-convertase-subtilisin/kexin type 9 (PCSK9) and cardiovascular events in type 2 diabetes. *Diabetes, Obesity and Metabolism*, **2018**, 20, 943-953	6.7	9

39	Systems pharmacology-based integration of human and mouse data for drug repurposing to treat thoracic aneurysms. *JCI Insight*, **2019**, 4,	9.9	8
38	Association study of serotonin transporter gene (SLC6A4) in systemic sclerosis in European Caucasian populations. *Journal of Rheumatology*, **2010**, 37, 1164-7	4.1	7
37	Fibulin-2: genetic mapping and exclusion as a candidate gene in Marfan syndrome type 2. *European Journal of Human Genetics*, **1996**, 4, 292-5	5.3	7
36	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
35	Unsuspected somatic mosaicism for FBN1 gene contributes to Marfan syndrome. *Genetics in Medicine*, **2021**, 23, 865-871	8.1	7
34	PCSK9 polymorphism in a Tunisian cohort: identification of a new allele, L8, and association of allele L10 with reduced coronary heart disease risk. *Molecular and Cellular Probes*, **2015**, 29, 1-6	3.3	6
33	Spinal imaging contributes to the diagnosis of Marfan syndrome. *Joint Bone Spine*, **2010**, 77, 445-50	2.9	6
32	Clinical and genetic data of 22 new patients with SMAD3 pathogenic variants and review of the literature. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1132	2.3	5
31	A new mutational hotspot in the SKI gene in the context of MFS/TAA molecular diagnosis. *Human Genetics*, **2020**, 139, 461-472	6.3	5
30	Actionable Genes, Core Databases, and Locus-Specific Databases. *Human Mutation*, **2016**, 37, 1299-1307	4.7	5
29	WES/WGS Reporting of Mutations from Cardiovascular "Actionable" Genes in Clinical Practice: A Key Role for UMD Knowledgebases in the Era of Big Databases. *Human Mutation*, **2016**, 37, 1308-1317	4.7	5
28	Reply to "The question of heterogeneity in Marfan syndrome". *Nature Genetics*, **1995**, 9, 230-231	36.3	5
27	First heterozygous mutation in familial pulmonary fibrosis. *European Respiratory Journal*, **2020**, 55,	13.6	5
26	Marfan Syndrome Variability: Investigation of the Roles of Sarcolipin and Calcium as Potential Transregulator of FBN1 Expression. *Genes*, **2018**, 9,	4.2	4
25	Surgical management of patients with Marfan syndrome: evolution throughout the years. *Archives of Cardiovascular Diseases*, **2012**, 105, 84-90	2.7	4
24	Marfanoid habitus, inguinal hernia, advanced bone age, and distinctive facial features: a new collagenopathy?. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1185-9	2.5	4
23	Reference Expression Profile of Three Transcript Isoforms and Their Association with Clinical Variability in Marfan Syndrome. *Genes*, **2019**, 10,	4.2	4
22	Identification of the first Tangier disease patient in Lebanon carrying a new pathogenic variant in ABCA1. *Journal of Clinical Lipidology*, **2018**, 12, 1374-1382	4.9	4

21	Neonatal Marfan Syndrome: Report of a Case with an Inherited Splicing Mutation outside the Neonatal Domain. *Molecular Syndromology*, **2016**, 6, 281-6	1.5	3
20	Genomic characterization of two deletions in the LDLR gene in Tunisian patients with familial hypercholesterolemia. *Clinica Chimica Acta*, **2012**, 414, 146-51	6.2	3
19	Quantifying the Genetic Basis of Marfan Syndrome Clinical Variability. *Genes*, **2020**, 11,	4.2	3
18	Molecular Genetics of the Fibrillinopathies **2016**, 1-13		3
17	Postprandial lipid absorption in seven heterozygous carriers of deleterious variants of MTTP in two abetalipoproteinemic families. *Journal of Clinical Lipidology*, **2019**, 13, 201-212	4.9	3
16	MUC5B promoter variant rs35705950 and rheumatoid arthritis associated interstitial lung disease survival and progression. *Seminars in Arthritis and Rheumatism*, **2021**, 51, 996-1004	5.3	3
15	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	5.8	2
14	Is there an agreement between self-reported medical diagnosis in the CARTaGENE cohort and the Québec administrative health databases?. *International Journal of Population Data Science*, **2020**, 5, 1155	1.4	2
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