Catherine Boileau

List of Publications by Year in Descending Order

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	APOE Molecular Spectrum in a French Cohort with Primary Dyslipidemia. *International Journal of Molecular Sciences*, **2022**, 23, 5792	6.3	0
181	Circulating PCSK9 Linked to Dyslipidemia in Lebanese Schoolchildren. *Metabolites*, **2022**, 12, 504	5.6	0
180	Cooperative Mechanism of ADAMTS/ ADAMTSL and Fibrillin-1 in the Marfan Syndrome and Acromelic Dysplasias.. *Frontiers in Genetics*, **2021**, 12, 734718	4.5	1
179	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
178	Methotrexate and rheumatoid arthritis associated interstitial lung disease. *European Respiratory Journal*, **2021**, 57,	13.6	34
177	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
176	Unsuspected somatic mosaicism for FBN1 gene contributes to Marfan syndrome. *Genetics in Medicine*, **2021**, 23, 865-871	8.1	7
175	Agreement in the CARTaGENE cohort between self-reported medication use and claim data. *Chronic Illness*, **2021**, 1742395320985913	1.4	1
174	APOE gene variants in primary dyslipidemia. *Atherosclerosis*, **2021**, 328, 11-22	3.1	10
173	Marfan syndrome. *Nature Reviews Disease Primers*, **2021**, 7, 64	51.1	17
172	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
171	A Case of Trisomy 13 Mosaicism Presenting with a Severe Aortic Root Dilatation and Marfanoid Habitus due to an Unpredictable Cytogenetic Mechanism. *Cytogenetic and Genome Research*, **2020**, 160, 72-79	1.9	0
170	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
169	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
168	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
167	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
166	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

165	Quantifying the Genetic Basis of Marfan Syndrome Clinical Variability. *Genes*, **2020**, 11,	4.2	3
164	First heterozygous mutation in familial pulmonary fibrosis. *European Respiratory Journal*, **2020**, 55,	13.6	5
163	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
162	Clinical Significance of Aortic Root Modification Associated With Bicuspid Aortic Valve in Marfan Syndrome. *Circulation: Cardiovascular Imaging*, **2019**, 12, e008129	3.9	9
161	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
160	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. *Genetics in Medicine*, **2019**, 21, 144-151	8.1	20
159	Myelodysplastic syndromes and idiopathic pulmonary fibrosis: a dangerous liaison. *Respiratory Research*, **2019**, 20, 182	7.3	1
158	Systems pharmacology-based integration of human and mouse data for drug repurposing to treat thoracic aneurysms. *JCI Insight*, **2019**, 4,	9.9	8
157	Reference Expression Profile of Three Transcript Isoforms and Their Association with Clinical Variability in Marfan Syndrome. *Genes*, **2019**, 10,	4.2	4
156	Green space associations with mental health and cognitive function: Results from the Quebec CARTaGENE cohort. *Environmental Epidemiology*, **2019**, 3, e040	0.2	34
155	Genetic testing for aortopathies: primer for the nongeneticist. *Current Opinion in Cardiology*, **2019**, 34, 585-593	2.1	1
154	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
153	Regulator of telomere length 1 () mutations are associated with heterogeneous pulmonary and extra-pulmonary phenotypes. *European Respiratory Journal*, **2019**, 53,	13.6	23
152	New Sequencing technologies help revealing unexpected mutations in Autosomal Dominant Hypercholesterolemia. *Scientific Reports*, **2018**, 8, 1943	4.9	13
151	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
150	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. *European Journal of Human Genetics*, **2018**, 26, 1759-1772	5.3	43
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	Marfan Syndrome Variability: Investigation of the Roles of Sarcolipin and Calcium as Potential Transregulator of FBN1 Expression. *Genes*, **2018**, 9,	4.2	4

147	The Canadian Partnership for Tomorrow Project: a pan-Canadian platform for research on chronic disease prevention. *Cmaj*, **2018**, 190, E710-E717	3.5	44
146	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
145	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
144	MUC5B Promoter Variant and Rheumatoid Arthritis with Interstitial Lung Disease. *New England Journal of Medicine*, **2018**, 379, 2209-2219	59.2	173
143	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
142	Shared genetic predisposition in rheumatoid arthritis-interstitial lung disease and familial pulmonary fibrosis. *European Respiratory Journal*, **2017**, 49,	13.6	89
141	The Proprotein Convertases in Hypercholesterolemia and Cardiovascular Diseases: Emphasis on Proprotein Convertase Subtilisin/Kexin 9. *Pharmacological Reviews*, **2017**, 69, 33-52	22.5	70
140	PCSK9 Mutations in Familial Hypercholesterolemia: from a Groundbreaking Discovery to Anti-PCSK9 Therapies. *Current Atherosclerosis Reports*, **2017**, 19, 49	6	19
139	International Registry of Patients Carrying TGFBR1 or TGFBR2 Mutations: Results of the MAC (Montalcino Aortic Consortium). *Circulation: Cardiovascular Genetics*, **2016**, 9, 548-558		105
138	Actionable Genes, Core Databases, and Locus-Specific Databases. *Human Mutation*, **2016**, 37, 1299-1307	4.7	5
137	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
136	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
135	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
134	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
133	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. *Journal of Clinical Investigation*, **2016**, 126, 948-61	15.9	65
132	Molecular Genetics of the Fibrillinopathies **2016**, 1-13		3
131	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
130	AndroSSL: A Platform to Test Android Applications Connection Security. *Lecture Notes in Computer Science*, **2016**, 294-302	0.9	1

129	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. *Circulation Research*, **2016**, 118, 928-34	15.7	122
128	Aortic Disease Presentation and Outcome Associated With ACTA2 Mutations. *Circulation: Cardiovascular Genetics*, **2015**, 8, 457-64		82
127	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
126	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
125	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
124	Heterozygous RTEL1 mutations are associated with familial pulmonary fibrosis. *European Respiratory Journal*, **2015**, 46, 474-85	13.6	96
123	Calcium Signaling Pathway Genes RUNX2 and CACNA1C Are Associated With Calcific Aortic Valve Disease. *Circulation: Cardiovascular Genetics*, **2015**, 8, 812-22		42
122	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
121	ELN gene triplication responsible for familial supravalvular aortic aneurysm. *Cardiology in the Young* , **2015**, 25, 712-7	1	17
120	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
119	Marfan Sartan: a randomized, double-blind, placebo-controlled trial. *European Heart Journal*, **2015**, 36, 2160-6	9.5	134
118	Identification of secreted phosphoprotein 1 gene as a new rheumatoid arthritis susceptibility gene. *Annals of the Rheumatic Diseases*, **2015**, 74, e19	2.4	16
117	Exome sequencing in suspected monogenic dyslipidemias. *Circulation: Cardiovascular Genetics*, **2015**, 8, 343-50		36
116	MAT2A mutations predispose individuals to thoracic aortic aneurysms. *American Journal of Human Genetics*, **2015**, 96, 170-7	11	68
115	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
114	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
113	Study of phenotype evolution during childhood in Marfan syndrome to improve clinical recognition. *Genetics in Medicine*, **2014**, 16, 246-50	8.1	37
112	Association study of CRP gene in systemic sclerosis in European Caucasian population. *Rheumatology International*, **2014**, 34, 389-92	3.6	1

111	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
110	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
109	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
108	Familial thoracic aortic aneurysms. *Current Opinion in Cardiology*, **2014**, 29, 492-8	2.1	17
107	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
106	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
105	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
104	A regulatory variant in CCR6 is associated with susceptibility to antitopoisomerase-positive systemic sclerosis. *Arthritis and Rheumatism*, **2013**, 65, 3202-8		25
103	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
102	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
101	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
100	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
99	Effect of mutations in LDLR and PCSK9 genes on phenotypic variability in Tunisian familial hypercholesterolemia patients. *Atherosclerosis*, **2012**, 222, 158-66	3.1	19
98	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
97	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
96	Surgical management of patients with Marfan syndrome: evolution throughout the years. *Archives of Cardiovascular Diseases*, **2012**, 105, 84-90	2.7	4
95	In-frame mutations in exon 1 of SKI cause dominant Shprintzen-Goldberg syndrome. *American Journal of Human Genetics*, **2012**, 91, 950-7	11	80
94	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

93	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
92	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
91	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
90	Genetics of thoracic aortic aneurysms. *Current Atherosclerosis Reports*, **2012**, 14, 219-26	6	35
89	Angiogenic biomarkers predict the occurrence of digital ulcers in systemic sclerosis. *Annals of the Rheumatic Diseases*, **2012**, 71, 394-9	2.4	47
88	Aortic event rate in the Marfan population: a cohort study. *Circulation*, **2012**, 125, 226-32	16.7	117
87	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
86	G□□ique et physiopathologie de la scl□odermie syst□mique. *Bulletin De L'Academie Nationale De Medecine*, **2011**, 195, 55-67	0.1	
85	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
84	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
83	Expanding the skeletal phenotype of Loeys-Dietz syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1178-83	2.5	15
82	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
81	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
80	Prognosis factors in probands with an FBN1 mutation diagnosed before the age of 1 year. *Pediatric Research*, **2011**, 69, 265-70	3.2	48
79	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
78	In vivo evidence that furin from hepatocytes inactivates PCSK9. *Journal of Biological Chemistry*, **2011**, 286, 4257-63	5.4	110
77	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
76	Dissection in Marfan syndrome: the importance of the descending aorta. *European Heart Journal*, **2011**, 32, 443-9	9.5	60

75	The translational science of Marfan syndrome. *Heart*, **2011**, 97, 1206-14	5.1	53
74	Genome-wide scan identifies TNIP1, PSORS1C1, and RHOB as novel risk loci for systemic sclerosis. *PLoS Genetics*, **2011**, 7, e1002091	6	176
73	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
72	Clinical utility gene card for: Marfan syndrome type 1 and related phenotypes [FBN1]. *European Journal of Human Genetics*, **2010**, 18,	5.3	19
71	A fourth locus for autosomal dominant hypercholesterolemia maps at 16q22.1. *European Journal of Human Genetics*, **2010**, 18, 1236-42	5.3	32
70	Quality, quantity and harmony: the DataSHaPER approach to integrating data across bioclinical studies. *International Journal of Epidemiology*, **2010**, 39, 1383-93	7.8	117
69	Dermal tissue and cellular expression of fibrillin-1 in diffuse cutaneous systemic sclerosis. *Rheumatology*, **2010**, 49, 657-61	3.9	10
68	Association of metalloproteinase gene polymorphisms with systemic sclerosis in the European Caucasian population. *Journal of Rheumatology*, **2010**, 37, 599-602	4.1	10
67	Genetic background of systemic sclerosis: autoimmune genes take centre stage. *Rheumatology*, **2010**, 49, 203-10	3.9	35
66	Cardiovascular manifestations in men and women carrying a FBN1 mutation. *European Heart Journal*, **2010**, 31, 2223-9	9.5	98
65	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
64	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
63	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
62	Association study of serotonin transporter gene (SLC6A4) in systemic sclerosis in European Caucasian populations. *Journal of Rheumatology*, **2010**, 37, 1164-7	4.1	7
61	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
60	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
59	Updating the genetics of systemic sclerosis. *Current Opinion in Rheumatology*, **2010**, 22, 665-70	5.3	16
58	Nomograms for aortic root diameters in children using two-dimensional echocardiography. *American Journal of Cardiology*, **2010**, 105, 888-94	3	110

57	Novel LRP5 gene mutation in a patient with osteoporosis-pseudoglioma syndrome. *Joint Bone Spine*, **2010**, 77, 151-3	2.9	17
56	Spinal imaging contributes to the diagnosis of Marfan syndrome. *Joint Bone Spine*, **2010**, 77, 445-50	2.9	6
55	Molecular spectrum of autosomal dominant hypercholesterolemia in France. *Human Mutation*, **2010** , 31, E1811-24	4.7	86
54	L□magerie du rachis contribue au diagnostic de syndrome de Marfan. *Revue Du Rhumatisme (Edition Francaise)*, **2010**, 77, 495-500	0.1	
53	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
52	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
51	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
50	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
49	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
48	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
47	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
46	Maternal complication of pregnancy in Marfan syndrome. *International Journal of Cardiology*, **2009**, 136, 156-61	3.2	81
45	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
44	Endothelial progenitor cells and rheumatic disorders. *Joint Bone Spine*, **2008**, 75, 131-7	2.9	34
43	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
42	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
41	A new locus-specific database (LSDB) for mutations in the TGFBR2 gene: UMD-TGFBR2. *Human Mutation*, **2008**, 29, 33-8	4.7	25
40	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

39	Polymorphic markers of the fibrillin-1 gene and systemic sclerosis in European Caucasian patients. *Journal of Rheumatology*, **2008**, 35, 643-9	4.1	15
38	C57BL/6J and A/J mice fed a high-fat diet delineate components of metabolic syndrome. *Obesity*, **2007**, 15, 1996-2005	8	169
37	Genetic basis for systemic sclerosis. *Joint Bone Spine*, **2007**, 74, 577-83	2.9	46
36	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
35	In vivo corneal confocal microscopy in marfan syndrome. *Cornea*, **2007**, 26, 787-92	3.1	12
34	Bone mineral density in Marfan syndrome. A large case-control study. *Joint Bone Spine*, **2006**, 73, 733-5	2.9	42
33	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
32	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
31	Molecular genetics of Marfan syndrome. *Current Opinion in Cardiology*, **2005**, 20, 194-200	2.1	96
30	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
29	UMD (Universal Mutation Database): 2005 update. *Human Mutation*, **2005**, 26, 184-91	4.7	91
28	Novel mutations of the PCSK9 gene cause variable phenotype of autosomal dominant hypercholesterolemia. *Human Mutation*, **2005**, 26, 497	4.7	142
27	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
26	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
25	Apolipoprotein B100 metabolism in autosomal-dominant hypercholesterolemia related to mutations in PCSK9. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2004**, 24, 1448-53	9.4	153
24	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
23	Heterozygous TGFBR2 mutations in Marfan syndrome. *Nature Genetics*, **2004**, 36, 855-60	36.3	509
22	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

21	The Marfan Mutation Database **2004**, 101-112		
20	Marfan Syndrome and Related Fibrillinopathies **2004**, 761-765		
19	Changes in the lymph node microenvironment induced by oncostatin M. *Blood*, **2003**, 102, 1397-404	2.2	16
18	Familial hypercholesterolemia in Morocco: first report of mutations in the LDL receptor gene. *Journal of Human Genetics*, **2003**, 48, 199-203	4.3	15
17	Update of the UMD-FBN1 mutation database and creation of an FBN1 polymorphism database. *Human Mutation*, **2003**, 22, 199-208	4.7	251
16	Mutations in PCSK9 cause autosomal dominant hypercholesterolemia. *Nature Genetics*, **2003**, 34, 154-6	36.3	2025
15	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
14	The UMD-LDLR database: additions to the software and 490 new entries to the database. *Human Mutation*, **2002**, 20, 81-7	4.7	89
13	Marfan syndrome in the third Millennium. *European Journal of Human Genetics*, **2002**, 10, 673-81	5.3	66
12	No evidence of somatic FGFR3 mutation in various types of carcinoma. *Oncogene*, **2001**, 20, 5059-61	9.2	37
11	UMD (Universal mutation database): a generic software to build and analyze locus-specific databases. *Human Mutation*, **2000**, 15, 86-94	4.7	159
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