Catherine Boileau

List of Publications by Year
in descending order

190 papers	22,299 citations	14655 66 h-index	8866 145 g-index
198 all docs	198 docs citations	198 times ranked	21398 citing authors

#	Article	IF	Citations
1	2014 ESC Guidelines on the diagnosis and treatment of aortic diseases. European Heart Journal, 2014, 35, 2873-2926.	2.2	3,549
2	Mutations in PCSK9 cause autosomal dominant hypercholesterolemia. Nature Genetics, 2003, 34, 154-156.	21.4	2,532
3	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, 3478-3490.	2.2	2,132
4	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, 2146-2157.	2.2	835
5	Familial hypercholesterolaemia in children and adolescents: gaining decades of life by optimizing detection and treatment. European Heart Journal, 2015, 36, 2425-2437.	2.2	644
6	Heterozygous TGFBR2 mutations in Marfan syndrome. Nature Genetics, 2004, 36, 855-860.	21.4	577
7	NARC-1/PCSK9 and Its Natural Mutants. Journal of Biological Chemistry, 2004, 279, 48865-48875.	3.4	544
8	Mutations in STAT3 and IL12RB1 impair the development of human IL-17â€“producing T cells. Journal of Experimental Medicine, 2008, 205, 1543-1550.	8.5	406
9	MUC5B Promoter Variant and Rheumatoid Arthritis with Interstitial Lung Disease. New England Journal of Medicine, 2018, 379, 2209-2219.	27.0	326
10	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. Nature Genetics, 2012, 44, 916-921.	21.4	319
11	Update of the UMD-FBN1mutation database and creation of anFBN1polymorphism database. Human Mutation, 2003, 22, 199-208.	2.5	299
12	Mutations and polymorphisms in the proprotein convertase subtilisin kexin 9 (PCSK9) gene in cholesterol metabolism and disease. Human Mutation, 2009, 30, 520-529.	2.5	211
13	Genome-Wide Scan Identifies TNIP1, PSORS1C1, and RHOB as Novel Risk Loci for Systemic Sclerosis. PLoS Genetics, 2011, 7, e1002091.	3.5	205
14	Comparison of Clinical Presentations and Outcomes Between Patients With TGFBR2 and FBN1 Mutations in Marfan Syndrome and Related Disorders. Circulation, 2009, 120, 2541-2549.	1.6	203
15	C57BL/6J and A/J Mice Fed a Highâ€Fat Diet Delineate Components of Metabolic Syndrome. Obesity, 2007, 15, 1996-2005.	3.0	200
16	Mutations in the TGFÎ² Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromicric and Geleophysic Dysplasias. American Journal of Human Genetics, 2011, 89, 7-14.	6.2	199
17	Recurrent Gain-of-Function Mutation in PRKG1 Causes Thoracic Aortic Aneurysms and Acute Aortic Dissections. American Journal of Human Genetics, 2013, 93, 398-404.	6.2	197
18	Clinical Validity of Genes for Heritable Thoracic Aortic Aneurysm and Dissection. Journal of the American College of Cardiology, 2018, 72, 605-615.	2.8	190

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19	UMD (Universal Mutation Database): A generic software to build and analyze locus-specific databases. Human Mutation, 2000, 15, 86-94.	2.5	184
20	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. Circulation Research, 2016, 118, 928-934.	4.5	180
21	Marfan Sartan: a randomized, double-blind, placebo-controlled trial. European Heart Journal, 2015, 36, 2160-2166.	2.2	179
22	Cohort profile of the CARTaGENE study: Quebecâ€™s population-based biobank for public health and personalized genomics. International Journal of Epidemiology, 2013, 42, 1285-1299.	1.9	172
23	Apolipoprotein B100 Metabolism in Autosomal-Dominant Hypercholesterolemia Related to Mutations in PCSK9. Arteriosclerosis, Thrombosis, and Vascular Biology, 2004, 24, 1448-1453.	2.4	171
24	A second locus for Marfan syndrome maps to chromosome 3p24.2â€“p25. Nature Genetics, 1994, 8, 264-268.	21.4	169
25	Novel mutations of thePCSK9 gene cause variable phenotype of autosomal dominant hypercholesterolemia. Human Mutation, 2005, 26, 497-497.	2.5	169
26	Aortic Event Rate in the Marfan Population. Circulation, 2012, 125, 226-232.	1.6	165
27	A Third Major Locus for Autosomal Dominant Hypercholesterolemia Maps to 1p34.1-p32. American Journal of Human Genetics, 1999, 64, 1378-1387.	6.2	154
28	Shared genetic predisposition in rheumatoid arthritis-interstitial lung disease and familial pulmonary fibrosis. European Respiratory Journal, 2017, 49, 1602314.	6.7	154
29	Quality, quantity and harmony: the DataSHaPER approach to integrating data across bioclinical studies. International Journal of Epidemiology, 2010, 39, 1383-1393.	1.9	148
30	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, 391-398.	2.1	146
31	International Registry of Patients Carrying TGFBR1 or TGFBR2 Mutations. Circulation: Cardiovascular Genetics, 2016, 9, 548-558.	5.1	145
32	Nomograms for Aortic Root Diameters in Children Using Two-Dimensional Echocardiography. American Journal of Cardiology, 2010, 105, 888-894.	1.6	140
33	HeterozygousRTEL1mutations are associated with familial pulmonary fibrosis. European Respiratory Journal, 2015, 46, 474-485.	6.7	135
34	Cardiovascular manifestations in men and women carrying a FBN1 mutation. European Heart Journal, 2010, 31, 2223-2229.	2.2	133
35	In Vivo Evidence That Furin from Hepatocytes Inactivates PCSK9. Journal of Biological Chemistry, 2011, 286, 4257-4263.	3.4	132
36	Identification in Daily Practice of Patients With Lynch Syndrome (Hereditary Nonpolyposis Colorectal) Tj ETQq0 0 0 rgBT /Overlock 10 Tf Journal of Gastroenterology, 2008, 103, 2825-2835.	0.4	118

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37	Molecular genetics of Marfan syndrome. Current Opinion in Cardiology, 2005, 20, 194-200.	1.8	117
38	Aortic Disease Presentation and Outcome Associated With ACTA2 Mutations. Circulation: Cardiovascular Genetics, 2015, 8, 457-464.	5.1	117
39	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.	2.7	116
40	Methotrexate and rheumatoid arthritis associated interstitial lung disease. European Respiratory Journal, 2021, 57, 2000337.	6.7	114
41	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, 736-743.	6.2	110
42	The UMD-LDLR database: additions to the software and 490 new entries to the database. Human Mutation, 2002, 20, 81-87.	2.5	105
43	Description of a Large Family with Autosomal Dominant Hypercholesterolemia Associated with theAPOEp.Leu167del Mutation. Human Mutation, 2013, 34, 83-87.	2.5	103
44	UMD (Universal Mutation Database): 2005 update. Human Mutation, 2005, 26, 184-191.	2.5	101
45	Parental origin and germline mosaicism of deletions and duplications of the dystrophin gene: a European study. Human Genetics, 1992, 88, 249-257.	3.8	100
46	Molecular Spectrum of Autosomal Dominant Hypercholesterolemia in France. Human Mutation, 2010, 31, E1811-E1824.	2.5	99
47	LTBP2 mutations cause Weill-Marchesani and Weill-Marchesani-like syndrome and affect disruptions in the extracellular matrix. Human Mutation, 2012, 33, 1182-1187.	2.5	99
48	Marfan syndrome. Nature Reviews Disease Primers, 2021, 7, 64.	30.5	99
49	Identification and in silico analyses of novelTGFBR1 andTGFBR2 mutations in Marfan syndrome-related disorders. Human Mutation, 2006, 27, 760-769.	2.5	98
50	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. American Journal of Human Genetics, 2012, 91, 950-957.	6.2	95
51	Maternal complication of pregnancy in Marfan syndrome. International Journal of Cardiology, 2009, 136, 156-161.	1.7	94
52	Identification and characterization of new gain-of-function mutations in the PCSK9 gene responsible for autosomal dominant hypercholesterolemia. Atherosclerosis, 2012, 223, 394-400.	0.8	92
53	MAT2A Mutations Predispose Individuals to Thoracic Aortic Aneurysms. American Journal of Human Genetics, 2015, 96, 170-177.	6.2	92
54	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, 823-831.	5.1	90

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55	The Proprotein Convertases in Hypercholesterolemia and Cardiovascular Diseases: Emphasis on Proprotein Convertase Subtilisin/Kexin 9. Pharmacological Reviews, 2017, 69, 33-52.	16.0	90
56	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.	4.8	87
57	Identification of 23TGFBR2and 6TGFBR1gene 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-E295.	2.5	86
58	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. Journal of Clinical Investigation, 2016, 126, 948-961.	8.2	84
59	The molecular basis of familial hypercholesterolemia in Lebanon: Spectrum ofLDLRmutations and role ofPCSK9as a modifier gene. Human Mutation, 2009, 30, E682-E691.	2.5	82
60	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, 1121-1128.	2.8	82
61	UMD-predictor, a new prediction tool for nucleotide substitution pathogenicity-application to four genes:FBN1,FBN2,TGFBR1, andTGFBR2. Human Mutation, 2009, 30, 952-959.	2.5	80
62	Marfan syndrome in the third Millennium. European Journal of Human Genetics, 2002, 10, 673-681.	2.8	79
63	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. European Journal of Human Genetics, 2018, 26, 1759-1772.	2.8	73
64	Dissection in Marfan syndrome: the importance of the descending aorta. European Heart Journal, 2011, 32, 443-449.	2.2	72
65	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-E1100.	3.5	71
66	The Canadian Partnership for Tomorrow Project: a pan-Canadian platform for research on chronic disease prevention. Cmaj, 2018, 190, E710-E717.	2.0	71
67	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, 317-325.	1.6	68
68	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, 2746-2752.	6.7	63
69	Clinical relevance of genotypeâ€“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.	2.4	63
70	The translational science of Marfan syndrome. Heart, 2011, 97, 1206-1214.	2.9	62
71	APOE gene variants in primary dyslipidemia. Atherosclerosis, 2021, 328, 11-22.	0.8	60
72	Prognosis Factors in Probands With an FBN1 Mutation Diagnosed Before the Age of 1 Year. Pediatric Research, 2011, 69, 265-270.	2.3	59

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73	Evaluation and application of denaturing HPLC for mutation detection in Marfan syndrome: Identification of 20 novel mutations and two novel polymorphisms in theFBN1gene. Human Mutation, 2002, 19, 443-456.	2.5	58
74	The clinical presentation of Marfan syndrome is modulated by expression of wild-type FBN1 allele. Human Molecular Genetics, 2015, 24, 2764-2770.	2.9	57
75	Genetic basis for systemic sclerosis. Joint Bone Spine, 2007, 74, 577-583.	1.6	56
76	Phenotype-Haplotype Correlation of IRF5 in Systemic Sclerosis: Role of 2 Haplotypes in Disease Severity. Journal of Rheumatology, 2010, 37, 987-992.	2.0	54
77	Green space associations with mental health and cognitive function. Environmental Epidemiology, 2019, 3, e040.	3.0	54
78	Angiogenic biomarkers predict the occurrence of digital ulcers in systemic sclerosis. Annals of the Rheumatic Diseases, 2012, 71, 394-399.	0.9	53
79	Plasma N-terminal pro-brain natriuretic peptide and brain natriuretic peptide in assessment of acute dyspnea. Biomedicine and Pharmacotherapy, 2005, 59, 20-24.	5.6	52
80	Calcium Signaling Pathway Genes RUNX2 and CACNA1C Are Associated With Calcific Aortic Valve Disease. Circulation: Cardiovascular Genetics, 2015, 8, 812-822.	5.1	51
81	Software and database for the analysis of mutations in the human LDL receptor gene. Nucleic Acids Research, 1997, 25, 172-180.	14.5	50
82	Bone mineral density inÂMarfan syndrome. A large case-control study. Joint Bone Spine, 2006, 73, 733-735.	1.6	50
83	TheFBN2gene: new mutations, locus-specific database (Universal Mutation DatabaseFBN2), and genotype-phenotype correlations. Human Mutation, 2009, 30, 181-190.	2.5	49
84	Independent replication establishes the CD247 gene as a genetic systemic sclerosis susceptibility factor. Annals of the Rheumatic Diseases, 2011, 70, 1695-1696.	0.9	46
85	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, 2091-2096.	6.7	45
86	Study of phenotype evolution during childhood in Marfan syndrome to improve clinical recognition. Genetics in Medicine, 2014, 16, 246-250.	2.4	45
87	Exome Sequencing in Suspected Monogenic Dyslipidemias. Circulation: Cardiovascular Genetics, 2015, 8, 343-350.	5.1	45
88	Regulator of telomere length 1 (RTEL1) mutations are associated with heterogeneous pulmonary and extra-pulmonary phenotypes. European Respiratory Journal, 2019, 53, 1800508.	6.7	45
89	Mutations in human CPO gene predict clinical expression of either hepatic hereditary coproporphyria or erythropoietic harderoporphyria. Human Molecular Genetics, 2005, 14, 3089-3098.	2.9	44
90	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.	2.7	44

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91	SMAD3pathogenic variants: risk for thoracic aortic disease and associated complications from the Montalcino Aortic Consortium. Journal of Medical Genetics, 2019, 56, 252-260.	3.2	43
92	Endothelial progenitor cells and rheumatic disorders. Joint Bone Spine, 2008, 75, 131-137.	1.6	42
93	Genetic background of systemic sclerosis: autoimmune genes take centre stage. Rheumatology, 2010, 49, 203-210.	1.9	42
94	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.	2.5	42
95	Enhanced late-outgrowth circulating endothelial progenitor cell levels in rheumatoid arthritis and correlation with disease activity. Arthritis Research and Therapy, 2010, 12, R27.	3.5	40
96	Genetics of Thoracic Aortic Aneurysms. Current Atherosclerosis Reports, 2012, 14, 219-226.	4.8	40
97	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.	2.4	39
98	No evidence of somatic FGFR3 mutation in various types of carcinoma. Oncogene, 2001, 20, 5059-5061.	5.9	38
99	A fourth locus for autosomal dominant hypercholesterolemia maps at 16q22.1. European Journal of Human Genetics, 2010, 18, 1236-1242.	2.8	38
100	Pathogenic FBN1 Genetic Variation and Aortic Dissection in Patients With MarfanÂSyndrome. Journal of the American College of Cardiology, 2020, 75, 843-853.	2.8	38
101	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. Genetics in Medicine, 2019, 21, 144-151.	2.4	36
102	Regulation of Extrathymic T Cell Development and Turnover by Oncostatin M. Journal of Immunology, 2000, 164, 5713-5720.	0.8	35
103	Independent Replication and Metaanalysis 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.	2.0	35
104	Rationale, design, and methods for Canadian alliance for healthy hearts and minds cohort study (CAHHM) â€“ a Pan Canadian cohort study. BMC Public Health, 2016, 16, 650.	2.9	31
105	PCSK9 Mutations in Familial Hypercholesterolemia: from a Groundbreaking Discovery to Anti-PCSK9 Therapies. Current Atherosclerosis Reports, 2017, 19, 49.	4.8	31
106	Homozygous and compound heterozygous mutations in theFBN1gene: unexpected findings in molecular diagnosis of Marfan syndrome. Journal of Medical Genetics, 2017, 54, 100-103.	3.2	30
107	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, 917-921.	6.2	29
108	R3531C Mutation in the Apolipoprotein B Gene Is Not Sufficient to Cause Hypercholesterolemia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2000, 20, E76-82.	2.4	28

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109	Strategies for proprotein convertase subtilisin kexin 9 modulation: a perspective on recent patents. Expert Opinion on Therapeutic Patents, 2010, 20, 1547-1571.	5.0	28
110	Clinical utility gene card for: Hereditary thoracic aortic aneurysm and dissection including next-generation sequencing-based approaches. European Journal of Human Genetics, 2016, 24, 146-150.	2.8	28
111	Familial hypercholesterolemia in Morocco: first report of mutations in the LDL receptor gene. Journal of Human Genetics, 2003, 48, 199-203.	2.3	27
112	A new locus-specific database (LSDB) for mutations in theTGFBR2gene: UMD-TGFBR2. Human Mutation, 2008, 29, 33-38.	2.5	27
113	Insights into the pathogenesis of systemic sclerosis based on the gene expression profile of progenitorâ€derived endothelial cells. Arthritis and Rheumatism, 2011, 63, 3552-3562.	6.7	26
114	Brief Report: A Regulatory Variant in CCR6 Is Associated With Susceptibility to Antitopoisomeraseâ€Positive Systemic Sclerosis. Arthritis and Rheumatism, 2013, 65, 3202-3208.	6.7	26
115	Clinical utility gene card for: Marfan syndrome type 1 and related phenotypes [FBN1]. European Journal of Human Genetics, 2010, 18, 1071-1071.	2.8	25
116	New Sequencing technologies help revealing unexpected mutations in Autosomal Dominant Hypercholesterolemia. Scientific Reports, 2018, 8, 1943.	3.3	25
117	Pathogenic variants in THSD4, encoding the ADAMTS-like 6 protein, predispose to inherited thoracic aortic aneurysm. Genetics in Medicine, 2021, 23, 111-122.	2.4	25
118	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, 290-295.	1.2	24
119	Identification of secreted phosphoprotein 1 gene as a new rheumatoid arthritis susceptibility gene. Annals of the Rheumatic Diseases, 2015, 74, e19-e19.	0.9	24
120	ELNgene triplication responsible for familial supravalvular aortic aneurysm. Cardiology in the Young, 2015, 25, 712-717.	0.8	23
121	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.	5.0	23
122	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, 1033-1038.	2.0	22
123	Effect of mutations in LDLR and PCSK9 genes on phenotypic variability in Tunisian familial hypercholesterolemia patients. Atherosclerosis, 2012, 222, 158-166.	0.8	22
124	Evaluation of predictive models in daily practice for the identification of patients with Lynch syndrome. International Journal of Cancer, 2012, 130, 1367-1377.	5.1	22
125	Familial thoracic aortic aneurysms. Current Opinion in Cardiology, 2014, 29, 492-498.	1.8	22
126	Usefulness of the genetic risk score to identify phenocopies in families with familial hypercholesterolemia?. European Journal of Human Genetics, 2018, 26, 570-578.	2.8	22

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127	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, 489-495.	2.1	21
128	Novel LRP5 gene mutation in a patient with osteoporosis-pseudoglioma syndrome. Joint Bone Spine, 2010, 77, 151-153.	1.6	21
129	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, 1494-1504.	1.9	21
130	Systems pharmacologyâ€“based integration of human and mouse data for drug repurposing to treat thoracic aneurysms. JCI Insight, 2019, 4, .	5.0	21
131	Splicing mutation in the fibrillin-1 gene associated with neonatal Marfan syndrome and severe pulmonary emphysema with tracheobronchomalacia. Pediatric Pulmonology, 2005, 39, 374-378.	2.0	19
132	Changes in the lymph node microenvironment induced by oncostatin M. Blood, 2003, 102, 1397-1404.	1.4	18
133	TGFÎ² 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, 1900-1903.	0.9	18
134	In Vivo Corneal Confocal Microscopy in Marfan Syndrome. Cornea, 2007, 26, 787-792.	1.7	17
135	Expanding the skeletal phenotype of Loeysâ€Dietz syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1178-1183.	1.2	17
136	MUC5B promoter variant rs35705950 and rheumatoid arthritis associated interstitial lung disease survival and progression. Seminars in Arthritis and Rheumatism, 2021, 51, 996-1004.	3.4	17
137	Plasma proproteinâ€convertaseâ€subtilisin/kexin type 9 (PCSK9) and cardiovascular events in type 2 diabetes. Diabetes, Obesity and Metabolism, 2018, 20, 943-953.	4.4	17
138	Updating the genetics of systemic sclerosis. Current Opinion in Rheumatology, 2010, 22, 665-670.	4.3	16
139	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, 208-212.	1.3	16
140	Is there an agreement between self-reported medical diagnosis in the CARTaGENE cohort and the QuÃ©bec administrative health databases?. International Journal of Population Data Science, 2020, 5, 1155.	0.1	16
141	Autosomal dominant typeÂIIa 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, 621-630.	2.8	15
142	Analytical correlation between plasma N-terminal pro-brain natriuretic peptide and brain natriuretic peptide in patients presenting with dyspnea. Clinical Biochemistry, 2004, 37, 933-936.	1.9	15
143	Angiotensin-Converting Enzyme Gene Does Not Contribute to Genetic Susceptibility to Systemic Sclerosis in European Caucasians. Journal of Rheumatology, 2009, 36, 337-340.	2.0	15
144	Clinical Significance of Aortic Root Modification Associated With Bicuspid Aortic Valve in Marfan Syndrome. Circulation: Cardiovascular Imaging, 2019, 12, e008129.	2.6	15

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145	Polymorphic markers of the fibrillin-1 gene and systemic sclerosis in European Caucasian patients. Journal of Rheumatology, 2008, 35, 643-9.	2.0	15
146	Unsuspected somatic mosaicism for FBN1 gene contributes to Marfan syndrome. Genetics in Medicine, 2021, 23, 865-871.	2.4	14
147	First heterozygous NOP10 mutation in familial pulmonary fibrosis. European Respiratory Journal, 2020, 55, 1902465.	6.7	13
148	Spinal imaging contributes to the diagnosis of Marfan syndrome. Joint Bone Spine, 2010, 77, 445-450.	1.6	12
149	Clinical and genetic data of 22 new patients with SMAD3 pathogenic variants and review of the literature. Molecular Genetics & Genomic Medicine, 2020, 8, e1132.	1.2	11
150	Quantifying the Genetic Basis of Marfan Syndrome Clinical Variability. Genes, 2020, 11, 574.	2.4	11
151	Determinants of survival after lung transplantation in telomerase-related gene mutation carriers: A retrospective cohort. American Journal of Transplantation, 2022, 22, 1236-1244.	4.7	11
152	Dermal tissue and cellular expression of fibrillin-1 in diffuse cutaneous systemic sclerosis. Rheumatology, 2010, 49, 657-661.	1.9	10
153	Association of Metalloproteinase Gene Polymorphisms with Systemic Sclerosis in the European Caucasian Population. Journal of Rheumatology, 2010, 37, 599-602.	2.0	10
154	Association Study of Serotonin Transporter Gene (SLC6A4) in Systemic Sclerosis in European Caucasian Populations. Journal of Rheumatology, 2010, 37, 1164-1167.	2.0	10
155	Fibulin-2: Genetic Mapping and Exclusion as a Candidate Gene in Marfan Syndrome Type 2. European Journal of Human Genetics, 1996, 4, 292-295.	2.8	9
156	Posttranscriptional Regulation of the Human LDL Receptor by the U2-Spliceosome. Circulation Research, 2022, 130, 80-95.	4.5	9
157	Reply to â€œThe question of heterogeneity in Marfan syndromeâ€• Nature Genetics, 1995, 9, 230-231.	21.4	8
158	Marfanoid habitus, inguinal hernia, advanced bone age, and distinctive facial features: A new collagenopathy?. American Journal of Medical Genetics, Part A, 2012, 158A, 1185-1189.	1.2	8
159	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.	2.1	8
160	A new mutational hotspot in the SKI gene in the context of MFS/TAA molecular diagnosis. Human Genetics, 2020, 139, 461-472.	3.8	8
161	Myelodysplastic syndromes and idiopathic pulmonary fibrosis: a dangerous liaison. Respiratory Research, 2019, 20, 182.	3.6	7
162	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.	3.2	7

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163	Actionable Genes, Core Databases, and Locus-Specific Databases. Human Mutation, 2016, 37, 1299-1307.	2.5	6
164	Identification of the first Tangier disease patient in Lebanon carrying a new pathogenic variant in ABCA1. Journal of Clinical Lipidology, 2018, 12, 1374-1382.	1.5	6
165	Reference Expression Profile of Three FBN1 Transcript Isoforms and Their Association with Clinical Variability in Marfan Syndrome. Genes, 2019, 10, 128.	2.4	6
166	Postprandial lipid absorption in seven heterozygous carriers of deleterious variants of MTTP in two abetalipoproteinemic families. Journal of Clinical Lipidology, 2019, 13, 201-212.	1.5	6
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