

Catherine Boileau

List of Publications by Year in descending order

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Version: 2024-02-01

190
papers

22,299
citations

14614

66
h-index

8835

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. <i>European Heart Journal</i> , 2014, 35, 2873-2926.	1.0	3,549
2	Mutations in PCSK9 cause autosomal dominant hypercholesterolemia. <i>Nature Genetics</i> , 2003, 34, 154-156.	9.4	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. <i>European Heart Journal</i> , 2013, 34, 3478-3490.	1.0	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. <i>European Heart Journal</i> , 2014, 35, 2146-2157.	1.0	835
5	Familial hypercholesterolaemia in children and adolescents: gaining decades of life by optimizing detection and treatment. <i>European Heart Journal</i> , 2015, 36, 2425-2437.	1.0	644
6	Heterozygous TGFBR2 mutations in Marfan syndrome. <i>Nature Genetics</i> , 2004, 36, 855-860.	9.4	577
7	NARC-1/PCSK9 and Its Natural Mutants. <i>Journal of Biological Chemistry</i> , 2004, 279, 48865-48875.	1.6	544
8	Mutations in <i>STAT3</i> and <i>IL12RB1</i> impair the development of human IL-17-producing T cells. <i>Journal of Experimental Medicine</i> , 2008, 205, 1543-1550.	4.2	406
9	<i>MUC5B</i> Promoter Variant and Rheumatoid Arthritis with Interstitial Lung Disease. <i>New England Journal of Medicine</i> , 2018, 379, 2209-2219.	13.9	326
10	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. <i>Nature Genetics</i> , 2012, 44, 916-921.	9.4	319
11	Update of the UMD-FBN1 mutation database and creation of an FBN1 polymorphism database. <i>Human Mutation</i> , 2003, 22, 199-208.	1.1	299
12	Mutations and polymorphisms in the proprotein convertase subtilisin kexin 9 (<i>PCSK9</i>) gene in cholesterol metabolism and disease. <i>Human Mutation</i> , 2009, 30, 520-529.	1.1	211
13	Genome-Wide Scan Identifies TNIP1, PSORS1C1, and RHOB as Novel Risk Loci for Systemic Sclerosis. <i>PLoS Genetics</i> , 2011, 7, e1002091.	1.5	205
14	Comparison of Clinical Presentations and Outcomes Between Patients With <i>TGFBR2</i> and <i>FBN1</i> Mutations in Marfan Syndrome and Related Disorders. <i>Circulation</i> , 2009, 120, 2541-2549.	1.6	203
15	C57BL/6J and A/J Mice Fed a High-Fat Diet Delineate Components of Metabolic Syndrome. <i>Obesity</i> , 2007, 15, 1996-2005.	1.5	200
16	Mutations in the TGF β 2 Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromiocric and Geleophysic Dysplasias. <i>American Journal of Human Genetics</i> , 2011, 89, 7-14.	2.6	199
17	Recurrent Gain-of-Function Mutation in PRKG1 Causes Thoracic Aortic Aneurysms and Acute Aortic Dissections. <i>American Journal of Human Genetics</i> , 2013, 93, 398-404.	2.6	197
18	Clinical Validity of Genes for Heritable Thoracic Aortic Aneurysm and Dissection. <i>Journal of the American College of Cardiology</i> , 2018, 72, 605-615.	1.2	190

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19	UMD (Universal Mutation Database): A generic software to build and analyze locus-specific databases. <i>Human Mutation</i> , 2000, 15, 86-94.	1.1	184
20	<i>LOX</i> Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. <i>Circulation Research</i> , 2016, 118, 928-934.	2.0	180
21	Marfan Sartan: a randomized, double-blind, placebo-controlled trial. <i>European Heart Journal</i> , 2015, 36, 2160-2166.	1.0	179
22	Cohort profile of the CARTaGENE study: Québec's population-based biobank for public health and personalized genomics. <i>International Journal of Epidemiology</i> , 2013, 42, 1285-1299.	0.9	172
23	Apolipoprotein B100 Metabolism in Autosomal-Dominant Hypercholesterolemia Related to Mutations in PCSK9. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2004, 24, 1448-1453.	1.1	171
24	A second locus for Marfan syndrome maps to chromosome 3p24.2â€“p25. <i>Nature Genetics</i> , 1994, 8, 264-268.	9.4	169
25	Novel mutations of the PCSK9 gene cause variable phenotype of autosomal dominant hypercholesterolemia. <i>Human Mutation</i> , 2005, 26, 497-497.	1.1	169
26	Aortic Event Rate in the Marfan Population. <i>Circulation</i> , 2012, 125, 226-232.	1.6	165
27	A Third Major Locus for Autosomal Dominant Hypercholesterolemia Maps to 1p34.1-p32. <i>American Journal of Human Genetics</i> , 1999, 64, 1378-1387.	2.6	154
28	Shared genetic predisposition in rheumatoid arthritis-interstitial lung disease and familial pulmonary fibrosis. <i>European Respiratory Journal</i> , 2017, 49, 1602314.	3.1	154
29	Quality, quantity and harmony: the DataSHaPER approach to integrating data across bioclinical studies. <i>International Journal of Epidemiology</i> , 2010, 39, 1383-1393.	0.9	148
30	Clinical and Molecular Study of 320 Children With Marfan Syndrome and Related Type I Fibrillinopathies in a Series of 1009 Proband With Pathogenic <i>FBN1</i> Mutations. <i>Pediatrics</i> , 2009, 123, 391-398.	1.0	146
31	International Registry of Patients Carrying <i>TGFBR1</i> or <i>TGFBR2</i> Mutations. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 548-558.	5.1	145
32	Nomograms for Aortic Root Diameters in Children Using Two-Dimensional Echocardiography. <i>American Journal of Cardiology</i> , 2010, 105, 888-894.	0.7	140
33	Heterozygous <i>RTEL1</i> mutations are associated with familial pulmonary fibrosis. <i>European Respiratory Journal</i> , 2015, 46, 474-485.	3.1	135
34	Cardiovascular manifestations in men and women carrying a <i>FBN1</i> mutation. <i>European Heart Journal</i> , 2010, 31, 2223-2229.	1.0	133
35	In Vivo Evidence That Furin from Hepatocytes Inactivates PCSK9. <i>Journal of Biological Chemistry</i> , 2011, 286, 4257-4263.	1.6	132
36	Identification in Daily Practice of Patients With Lynch Syndrome (Hereditary Nonpolyposis Colorectal) Tj ETQq0 0 0 rgBT /Overlock 10 TF <i>Journal of Gastroenterology</i> , 2008, 103, 2825-2835.	0.2	118

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37	Molecular genetics of Marfan syndrome. <i>Current Opinion in Cardiology</i> , 2005, 20, 194-200.	0.8	117
38	Aortic Disease Presentation and Outcome Associated With <i>ACTA2</i> Mutations. <i>Circulation: Cardiovascular Genetics</i> , 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). <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 1.	1.2	116
40	Methotrexate and rheumatoid arthritis associated interstitial lung disease. <i>European Respiratory Journal</i> , 2021, 57, 2000337.	3.1	114
41	MFAP5 Loss-of-Function Mutations Underscore the Involvement of Matrix Alteration in the Pathogenesis of Familial Thoracic Aortic Aneurysms and Dissections. <i>American Journal of Human Genetics</i> , 2014, 95, 736-743.	2.6	110
42	The UMD-LDLR database: additions to the software and 490 new entries to the database. <i>Human Mutation</i> , 2002, 20, 81-87.	1.1	105
43	Description of a Large Family with Autosomal Dominant Hypercholesterolemia Associated with the <i>APOE</i> p.Leu167del Mutation. <i>Human Mutation</i> , 2013, 34, 83-87.	1.1	103
44	UMD (Universal Mutation Database): 2005 update. <i>Human Mutation</i> , 2005, 26, 184-191.	1.1	101
45	Parental origin and germline mosaicism of deletions and duplications of the dystrophin gene: a European study. <i>Human Genetics</i> , 1992, 88, 249-257.	1.8	100
46	Molecular Spectrum of Autosomal Dominant Hypercholesterolemia in France. <i>Human Mutation</i> , 2010, 31, E1811-E1824.	1.1	99
47	LTBP2 mutations cause Weill-Marchesani and Weill-Marchesani-like syndrome and affect disruptions in the extracellular matrix. <i>Human Mutation</i> , 2012, 33, 1182-1187.	1.1	99
48	Marfan syndrome. <i>Nature Reviews Disease Primers</i> , 2021, 7, 64.	18.1	99
49	Identification and in silico analyses of novel <i>TGFBR1</i> and <i>TGFBR2</i> mutations in Marfan syndrome-related disorders. <i>Human Mutation</i> , 2006, 27, 760-769.	1.1	98
50	In-Frame Mutations in Exon 1 of <i>SKI</i> Cause Dominant Shprintzen-Goldberg Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 950-957.	2.6	95
51	Maternal complication of pregnancy in Marfan syndrome. <i>International Journal of Cardiology</i> , 2009, 136, 156-161.	0.8	94
52	Identification and characterization of new gain-of-function mutations in the <i>PCSK9</i> gene responsible for autosomal dominant hypercholesterolemia. <i>Atherosclerosis</i> , 2012, 223, 394-400.	0.4	92
53	<i>MAT2A</i> Mutations Predispose Individuals to Thoracic Aortic Aneurysms. <i>American Journal of Human Genetics</i> , 2015, 96, 170-177.	2.6	92
54	Characterization of Autosomal Dominant Hypercholesterolemia Caused by <i>PCSK9</i> Gain of Function Mutations and Its Specific Treatment With Alirocumab, a <i>PCSK9</i> Monoclonal Antibody. <i>Circulation: Cardiovascular Genetics</i> , 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. <i>Pharmacological Reviews</i> , 2017, 69, 33-52.	7.1	90
56	Living the PCSK9 Adventure: from the Identification of a New Gene in Familial Hypercholesterolemia Towards a Potential New Class of Anticholesterol Drugs. <i>Current Atherosclerosis Reports</i> , 2014, 16, 439.	2.0	87
57	Identification of 23TGFB2 and 6TGFB1 gene mutations and genotype-phenotype investigations in 457 patients with Marfan syndrome type I and II, Loeys-Dietz syndrome and related disorders. <i>Human Mutation</i> , 2008, 29, E284-E295.	1.1	86
58	FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. <i>Journal of Clinical Investigation</i> , 2016, 126, 948-961.	3.9	84
59	The molecular basis of familial hypercholesterolemia in Lebanon: Spectrum of LDLR mutations and role of PCSK9 as a modifier gene. <i>Human Mutation</i> , 2009, 30, E682-E691.	1.1	82
60	Identification of the minimal combination of clinical features in probands for efficient mutation detection in the FBN1 gene. <i>European Journal of Human Genetics</i> , 2009, 17, 1121-1128.	1.4	82
61	UMD-predictor, a new prediction tool for nucleotide substitution pathogenicity-application to four genes: FBN1, FBN2, TGFB1, and TGFB2. <i>Human Mutation</i> , 2009, 30, 952-959.	1.1	80
62	Marfan syndrome in the third Millennium. <i>European Journal of Human Genetics</i> , 2002, 10, 673-681.	1.4	79
63	Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. <i>European Journal of Human Genetics</i> , 2018, 26, 1759-1772.	1.4	73
64	Dissection in Marfan syndrome: the importance of the descending aorta. <i>European Heart Journal</i> , 2011, 32, 443-449.	1.0	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. <i>American Journal of Physiology - Endocrinology and Metabolism</i> , 2007, 292, E1095-E1100.	1.8	71
66	The Canadian Partnership for Tomorrow Project: a pan-Canadian platform for research on chronic disease prevention. <i>Cmaj</i> , 2018, 190, E710-E717.	0.9	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. <i>Archives of Cardiovascular Diseases</i> , 2010, 103, 317-325.	0.7	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. <i>Arthritis and Rheumatism</i> , 2012, 64, 2746-2752.	6.7	63
69	Clinical relevance of genotype-phenotype correlations beyond vascular events in a cohort study of 1500 Marfan syndrome patients with FBN1 pathogenic variants. <i>Genetics in Medicine</i> , 2021, 23, 1296-1304.	1.1	63
70	The translational science of Marfan syndrome. <i>Heart</i> , 2011, 97, 1206-1214.	1.2	62
71	APOE gene variants in primary dyslipidemia. <i>Atherosclerosis</i> , 2021, 328, 11-22.	0.4	60
72	Prognosis Factors in Proband With an FBN1 Mutation Diagnosed Before the Age of 1 Year. <i>Pediatric Research</i> , 2011, 69, 265-270.	1.1	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 the FBN1 gene. <i>Human Mutation</i> , 2002, 19, 443-456.	1.1	58
74	The clinical presentation of Marfan syndrome is modulated by expression of wild-type FBN1 allele. <i>Human Molecular Genetics</i> , 2015, 24, 2764-2770.	1.4	57
75	Genetic basis for systemic sclerosis. <i>Joint Bone Spine</i> , 2007, 74, 577-583.	0.8	56
76	Phenotype-Haplotype Correlation of <i>IRF5</i> in Systemic Sclerosis: Role of 2 Haplotypes in Disease Severity. <i>Journal of Rheumatology</i> , 2010, 37, 987-992.	1.0	54
77	Green space associations with mental health and cognitive function. <i>Environmental Epidemiology</i> , 2019, 3, e040.	1.4	54
78	Angiogenic biomarkers predict the occurrence of digital ulcers in systemic sclerosis. <i>Annals of the Rheumatic Diseases</i> , 2012, 71, 394-399.	0.5	53
79	Plasma N-terminal pro-brain natriuretic peptide and brain natriuretic peptide in assessment of acute dyspnea. <i>Biomedicine and Pharmacotherapy</i> , 2005, 59, 20-24.	2.5	52
80	Calcium Signaling Pathway Genes <i>RUNX2</i> and <i>CACNA1C</i> Are Associated With Calcific Aortic Valve Disease. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 812-822.	5.1	51
81	Software and database for the analysis of mutations in the human LDL receptor gene. <i>Nucleic Acids Research</i> , 1997, 25, 172-180.	6.5	50
82	Bone mineral density in Marfan syndrome. A large case-control study. <i>Joint Bone Spine</i> , 2006, 73, 733-735.	0.8	50
83	The <i>FBN2</i> gene: new mutations, locus-specific database (Universal Mutation Database <i>FBN2</i>), and genotype-phenotype correlations. <i>Human Mutation</i> , 2009, 30, 181-190.	1.1	49
84	Independent replication establishes the CD247 gene as a genetic systemic sclerosis susceptibility factor. <i>Annals of the Rheumatic Diseases</i> , 2011, 70, 1695-1696.	0.5	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. <i>Arthritis and Rheumatism</i> , 2011, 63, 2091-2096.	6.7	45
86	Study of phenotype evolution during childhood in Marfan syndrome to improve clinical recognition. <i>Genetics in Medicine</i> , 2014, 16, 246-250.	1.1	45
87	Exome Sequencing in Suspected Monogenic Dyslipidemias. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 343-350.	5.1	45
88	Regulator of telomere length 1 (<i>RTEL1</i>) mutations are associated with heterogeneous pulmonary and extra-pulmonary phenotypes. <i>European Respiratory Journal</i> , 2019, 53, 1800508.	3.1	45
89	Mutations in human CPO gene predict clinical expression of either hepatic hereditary coproporphyrin or erythropoietic harderoporphyria. <i>Human Molecular Genetics</i> , 2005, 14, 3089-3098.	1.4	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. <i>American Heart Journal</i> , 2015, 169, 605-612.	1.2	44

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91	<i>SMAD3</i> pathogenic variants: risk for thoracic aortic disease and associated complications from the Montalcino Aortic Consortium. <i>Journal of Medical Genetics</i> , 2019, 56, 252-260.	1.5	43
92	Endothelial progenitor cells and rheumatic disorders. <i>Joint Bone Spine</i> , 2008, 75, 131-137.	0.8	42
93	Genetic background of systemic sclerosis: autoimmune genes take centre stage. <i>Rheumatology</i> , 2010, 49, 203-210.	0.9	42
94	Early-Onset Osteoarthritis, Charcot-Marie-Tooth Like Neuropathy, Autoimmune Features, Multiple Arterial Aneurysms and Dissections: An Unrecognized and Life Threatening Condition. <i>PLoS ONE</i> , 2014, 9, e96387.	1.1	42
95	Enhanced late-outgrowth circulating endothelial progenitor cell levels in rheumatoid arthritis and correlation with disease activity. <i>Arthritis Research and Therapy</i> , 2010, 12, R27.	1.6	40
96	Genetics of Thoracic Aortic Aneurysms. <i>Current Atherosclerosis Reports</i> , 2012, 14, 219-226.	2.0	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). <i>Genetics in Medicine</i> , 2019, 21, 2015-2024.	1.1	39
98	No evidence of somatic <i>FGFR3</i> mutation in various types of carcinoma. <i>Oncogene</i> , 2001, 20, 5059-5061.	2.6	38
99	A fourth locus for autosomal dominant hypercholesterolemia maps at 16q22.1. <i>European Journal of Human Genetics</i> , 2010, 18, 1236-1242.	1.4	38
100	Pathogenic <i>FBN1</i> Genetic Variation and Aortic Dissection in Patients With Marfan Syndrome. <i>Journal of the American College of Cardiology</i> , 2020, 75, 843-853.	1.2	38
101	<i>MYLK</i> pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. <i>Genetics in Medicine</i> , 2019, 21, 144-151.	1.1	36
102	Regulation of Extrathymic T Cell Development and Turnover by Oncostatin M. <i>Journal of Immunology</i> , 2000, 164, 5713-5720.	0.4	35
103	Independent Replication and Metaanalysis of Association Studies Establish <i>TNFSF4</i> as a Susceptibility Gene Preferentially Associated with the Subset of Anticentromere-positive Patients with Systemic Sclerosis. <i>Journal of Rheumatology</i> , 2012, 39, 997-1003.	1.0	35
104	Rationale, design, and methods for Canadian alliance for healthy hearts and minds cohort study (CAHHM) – a Pan Canadian cohort study. <i>BMC Public Health</i> , 2016, 16, 650.	1.2	31
105	<i>PCSK9</i> Mutations in Familial Hypercholesterolemia: from a Groundbreaking Discovery to Anti- <i>PCSK9</i> Therapies. <i>Current Atherosclerosis Reports</i> , 2017, 19, 49.	2.0	31
106	Homozygous and compound heterozygous mutations in the <i>FBN1</i> gene: unexpected findings in molecular diagnosis of Marfan syndrome. <i>Journal of Medical Genetics</i> , 2017, 54, 100-103.	1.5	30
107	Demonstration of the Recurrence of Marfan-like Skeletal and Cardiovascular Manifestations Due to Germline Mosaicism for an <i>FBN1</i> Mutation. <i>American Journal of Human Genetics</i> , 1999, 65, 917-921.	2.6	29
108	R3531C Mutation in the Apolipoprotein B Gene Is Not Sufficient to Cause Hypercholesterolemia. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2000, 20, E76-82.	1.1	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.	2.4	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.	1.4	28
111	Familial hypercholesterolemia in Morocco: first report of mutations in the LDL receptor gene. Journal of Human Genetics, 2003, 48, 199-203.	1.1	27
112	A new locus-specific database (LSDB) for mutations in the <i>TGFBR2</i> gene: UMD- <i>TGFBR2</i> . Human Mutation, 2008, 29, 33-38.	1.1	27
113	Insights into the pathogenesis of systemic sclerosis based on the gene expression profile of progenitor-derived endothelial cells. Arthritis and Rheumatism, 2011, 63, 3552-3562.	6.7	26
114	Brief Report: A Regulatory Variant in <i>CCR6</i> Is Associated With Susceptibility to Antitopoisomerase-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.	1.4	25
116	New Sequencing technologies help revealing unexpected mutations in Autosomal Dominant Hypercholesterolemia. Scientific Reports, 2018, 8, 1943.	1.6	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.	1.1	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.	0.6	24
119	Identification of secreted phosphoprotein 1 gene as a new rheumatoid arthritis susceptibility gene. Annals of the Rheumatic Diseases, 2015, 74, e19-e19.	0.5	24
120	<i>ELN</i> gene triplication responsible for familial supraaortic aneurysm. Cardiology in the Young, 2015, 25, 712-717.	0.4	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.	2.4	23
122	Association Study of <i>ITGAM</i> , <i>ITGAX</i> , and <i>CD58</i> Autoimmune Risk Loci in Systemic Sclerosis: Results from 2 Large European Caucasian Cohorts. Journal of Rheumatology, 2011, 38, 1033-1038.	1.0	22
123	Effect of mutations in LDLR and PCSK9 genes on phenotypic variability in Tunisian familial hypercholesterolemia patients. Atherosclerosis, 2012, 222, 158-166.	0.4	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.	2.3	22
125	Familial thoracic aortic aneurysms. Current Opinion in Cardiology, 2014, 29, 492-498.	0.8	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.	1.4	22

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127	A Prognostic Model for HIV Seroconversion Among Injection Drug Users as a Tool for Stratification in Clinical Trials. <i>Journal of Acquired Immune Deficiency Syndromes</i> (1999), 2005, 39, 489-495.	0.9	21
128	Novel LRP5 gene mutation in a patient with osteoporosis-pseudoglioma syndrome. <i>Joint Bone Spine</i> , 2010, 77, 151-153.	0.8	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. <i>Rheumatology</i> , 2011, 50, 1494-1504.	0.9	21
130	Systems pharmacology-based integration of human and mouse data for drug repurposing to treat thoracic aneurysms. <i>JCI Insight</i> , 2019, 4, .	2.3	21
131	Splicing mutation in the fibrillin-1 gene associated with neonatal Marfan syndrome and severe pulmonary emphysema with tracheobronchomalacia. <i>Pediatric Pulmonology</i> , 2005, 39, 374-378.	1.0	19
132	Changes in the lymph node microenvironment induced by oncostatin M. <i>Blood</i> , 2003, 102, 1397-1404.	0.6	18
133	TGF β 2 receptor gene variants in systemic sclerosis-related pulmonary arterial hypertension: results from a multicentre EUSTAR study of European Caucasian patients. <i>Annals of the Rheumatic Diseases</i> , 2012, 71, 1900-1903.	0.5	18
134	In Vivo Corneal Confocal Microscopy in Marfan Syndrome. <i>Cornea</i> , 2007, 26, 787-792.	0.9	17
135	Expanding the skeletal phenotype of Loeys-Dietz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1178-1183.	0.7	17
136	MUC5B promoter variant rs35705950 and rheumatoid arthritis associated interstitial lung disease survival and progression. <i>Seminars in Arthritis and Rheumatism</i> , 2021, 51, 996-1004.	1.6	17
137	Plasma proproteinase-convertase-subtilisin/kexin type 9 (PCSK9) and cardiovascular events in type 2 diabetes. <i>Diabetes, Obesity and Metabolism</i> , 2018, 20, 943-953.	2.2	17
138	Updating the genetics of systemic sclerosis. <i>Current Opinion in Rheumatology</i> , 2010, 22, 665-670.	2.0	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. <i>European Journal of Medical Genetics</i> , 2010, 53, 208-212.	0.7	16
140	Is there an agreement between self-reported medical diagnosis in the CARTaGENE cohort and the Quebec administrative health databases?. <i>International Journal of Population Data Science</i> , 2020, 5, 1155.	0.1	16
141	Autosomal dominant type IIIa hypercholesterolemia: evaluation of the respective contributions of LDLR and APOB gene defects as well as a third major group of defects. <i>European Journal of Human Genetics</i> , 2000, 8, 621-630.	1.4	15
142	Analytical correlation between plasma N-terminal pro-brain natriuretic peptide and brain natriuretic peptide in patients presenting with dyspnea. <i>Clinical Biochemistry</i> , 2004, 37, 933-936.	0.8	15
143	Angiotensin-Converting Enzyme Gene Does Not Contribute to Genetic Susceptibility to Systemic Sclerosis in European Caucasians. <i>Journal of Rheumatology</i> , 2009, 36, 337-340.	1.0	15
144	Clinical Significance of Aortic Root Modification Associated With Bicuspid Aortic Valve in Marfan Syndrome. <i>Circulation: Cardiovascular Imaging</i> , 2019, 12, e008129.	1.3	15

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166	Postprandial lipid absorption in seven heterozygous carriers of deleterious variants of MTTP in two abetalipoproteinemic families. <i>Journal of Clinical Lipidology</i> , 2019, 13, 201-212.	0.6	6
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