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List of Publications by Year
in descending order

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75 papers	6,649 citations	186265 28 h-index	95266 68 g-index
82 all docs	82 docs citations	82 times ranked	6627 citing authors

#	Article	IF	Citations
1	Posttranscriptional Regulation of the Human LDL Receptor by the U2-Spliceosome. Circulation Research, 2022, 130, 80-95.	4.5	9
2	Whole Exome/Genome Sequencing Joint Analysis of a Family with Oligogenic Familial Hypercholesterolemia. Metabolites, 2022, 12, 262.	2.9	1
3	APOE Molecular Spectrum in a French Cohort with Primary Dyslipidemia. International Journal of Molecular Sciences, 2022, 23, 5792.	4.1	4
4	Circulating PCSK9 Linked to Dyslipidemia in Lebanese Schoolchildren. Metabolites, 2022, 12, 504.	2.9	1
5	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
6	Polymorphisms rs2745557 in PTGS2 and rs2075797 in PTGER2 are associated with the risk of chronic obstructive pulmonary disease development in a Tunisian cohort. Prostaglandins Leukotrienes and Essential Fatty Acids, 2021, 166, 102252.	2.2	3
7	APOE gene variants in primary dyslipidemia. Atherosclerosis, 2021, 328, 11-22.	0.8	60
8	Identification of a Variant in APOB Gene as a Major Cause of Hypobetalipoproteinemia in Lebanese Families. Metabolites, 2021, 11, 564.	2.9	5
9	Ephrin-B2 PB-mononuclear cells reduce early post-stroke deficit in diabetic mice but not long-term memory impairment. Experimental Neurology, 2021, 346, 113864.	4.1	0
10	Lipoprotein(a): Pathophysiology, measurement, indication and treatment in cardiovascular disease. A consensus statement from the Nouvelle SociÃ©tÃ© Francophone dâ€™AthÃ©rosclÃ©rose (NSFA). Archives of Cardiovascular Diseases, 2021, 114, 828-847.	1.6	9
11	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
12	New Sequencing technologies help revealing unexpected mutations in Autosomal Dominant Hypercholesterolemia. Scientific Reports, 2018, 8, 1943.	3.3	25
13	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
14	High burden of recurrent cardiovascular events in heterozygous familial hypercholesterolemia: The French Familial Hypercholesterolemia Registry. Atherosclerosis, 2018, 277, 334-340.	0.8	33
15	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
16	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
17	Effect of the p.Arg357His mutation of PCSK9 on basal and postprandial lipoprotein metabolism. Atherosclerosis, 2017, 263, e2.	0.8	0
18	PCSK9 Mutations in Familial Hypercholesterolemia: from a Groundbreaking Discovery to Anti-PCSK9 Therapies. Current Atherosclerosis Reports, 2017, 19, 49.	4.8	31

#	Article	IF	Citations
19	Usefulness of the genetic risk score to identify phenocopies in families with autosomal dominant hypercholesterolemia?. Atherosclerosis, 2017, 263, e83.	0.8	0
20	Plasma PCSK9 and cardiovascular events in type 2 diabetes. Atherosclerosis, 2017, 263, e81.	0.8	1
21	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
22	Identification of a new mutation in the N-terminal region of the apolipoprotein B gene in familial hypercholesterolemia. Atherosclerosis, 2016, 252, e34.	0.8	0
23	Exome Sequencing in Suspected Monogenic Dyslipidemias. Circulation: Cardiovascular Genetics, 2015, 8, 343-350.	5.1	45
24	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
25	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
26	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
27	Description of a Large Family with Autosomal Dominant Hypercholesterolemia Associated with theAPOEp.Leu167del Mutation. Human Mutation, 2013, 34, 83-87.	2.5	103
28	Autosomal Dominant Hypercholesterolemia: Needs for Early Diagnosis and Cascade Screening in the Tunisian Population. Current Genomics, 2013, 14, 25-32.	1.6	1
29	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
30	Effect of mutations in LDLR and PCSK9 genes on phenotypic variability in Tunisian familial hypercholesterolemia patients. Atherosclerosis, 2012, 222, 158-166.	0.8	22
31	Genomic characterization of two deletions in the LDLR gene in Tunisian patients with familial hypercholesterolemia. Clinica Chimica Acta, 2012, 414, 146-151.	1.1	5
32	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
33	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. Human Mutation, 2012, 33, 1175-1181.	2.5	74
34	Effect of a splice site mutation in LDLR gene and two variations in PCSK9 gene in Tunisian families with familial hypercholesterolaemia. Annals of Clinical Biochemistry, 2011, 48, 83-86.	1.6	6
35	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
36	Novel LRP5 gene mutation in a patient with osteoporosis-pseudoglioma syndrome. Joint Bone Spine, 2010, 77, 151-153.	1.6	21

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37	Molecular Spectrum of Autosomal Dominant Hypercholesterolemia in France. Human Mutation, 2010, 31, E1811-E1824.	2.5	99
38	A fourth locus for autosomal dominant hypercholesterolemia maps at 16q22.1. European Journal of Human Genetics, 2010, 18, 1236-1242.	2.8	38
39	Dermal tissue and cellular expression of fibrillin-1 in diffuse cutaneous systemic sclerosis. Rheumatology, 2010, 49, 657-661.	1.9	10
40	Moderate phenotypic expression of familial hypercholesterolemia in Tunisia. Clinica Chimica Acta, 2010, 411, 735-738.	1.1	17
41	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
42	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
43	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
44	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
45	Limited mutational heterogeneity in the LDLR gene in familial hypercholesterolemia in Tunisia. Atherosclerosis, 2009, 203, 449-453.	0.8	17
46	Genetic heterogeneity of autosomal dominant hypercholesterolemia. Clinical Genetics, 2008, 73, 1-13.	2.0	160
47	A novel splice site mutation of the LDL receptor gene in a Tunisian hypercholesterolemic family. Clinica Chimica Acta, 2008, 392, 25-29.	1.1	16
48	LOCALISATION OF A FIFTH GENE INVOLVED IN AUTOSOMAL DOMINANT HYPERCHOLESTEROLEMIA. Atherosclerosis Supplements, 2008, 9, 33.	1.2	0
49	PCSK9 FROM GENE AND VARIANTS TO PROTEIN AND PHENOTYPE. Atherosclerosis Supplements, 2008, 9, 101.	1.2	0
50	A PCSK9 variant and familial combined hyperlipidaemia. Journal of Medical Genetics, 2008, 45, 780-786.	3.2	39
51	Mutational heterogeneity in low-density lipoprotein receptor gene related to familial hypercholesterolemia in Morocco. Clinica Chimica Acta, 2006, 373, 62-69.	1.1	21
52	DnaJA4 is a SREBP-regulated chaperone involved in the cholesterol biosynthesis pathway. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2006, 1761, 1107-1113.	2.4	22
53	Novel mutations of thePCSK9 gene cause variable phenotype of autosomal dominant hypercholesterolemia. Human Mutation, 2005, 26, 497-497.	2.5	169
54	PC9, A New Actor in Autosomal Dominant Hypercholesterolemia. Current Genomics, 2005, 6, 535-543.	1.6	0

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55	Apolipoprotein B100 Metabolism in Autosomal-Dominant Hypercholesterolemia Related to Mutations in PCSK9. Arteriosclerosis, Thrombosis, and Vascular Biology, 2004, 24, 1448-1453.	2.4	171
56	NARC-1/PCSK9 and Its Natural Mutants. Journal of Biological Chemistry, 2004, 279, 48865-48875.	3.4	544
57	Identification of the first Lebanese mutation in the LPL gene and description of a rapid detection method. Clinical Genetics, 2004, 65, 158-161.	2.0	12
58	Heterozygous TGFBR2 mutations in Marfan syndrome. Nature Genetics, 2004, 36, 855-860.	21.4	577
59	Familial hypercholesterolemia in Morocco: first report of mutations in the LDL receptor gene. Journal of Human Genetics, 2003, 48, 199-203.	2.3	27
60	Mutations in PCSK9 cause autosomal dominant hypercholesterolemia. Nature Genetics, 2003, 34, 154-156.	21.4	2,532
61	New Insights into How Adipocytes Sense their Triglyceride Stores. Is Cholesterol a Signal?. Hormone and Metabolic Research, 2003, 35, 204-210.	1.5	30
62	The UMD-LDLR database: additions to the software and 490 new entries to the database. Human Mutation, 2002, 20, 81-87.	2.5	105
63	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
64	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
65	Mutation analysis in a small cohort of New Zealand patients originating from the United Kingdom demonstrates genetic heterogeneity in familial hypercholesterolemia. Molecular and Cellular Probes, 2000, 14, 299-304.	2.1	7
66	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
67	Analysis of the 525 point mutations in the human LDL receptor gene database. Atherosclerosis, 1999, 144, 182-183.	0.8	3
68	LDLR Database (second edition): new additions to the database and the software, and results of the first molecular analysis. Nucleic Acids Research, 1998, 26, 248-252.	14.5	77
69	Software and database for the analysis of mutations in the human LDL receptor gene. Nucleic Acids Research, 1997, 25, 172-180.	14.5	50
70	1.P.275 Results of the molecular analysis of the 220 point mutations in the human LDL receptor gene database. Atherosclerosis, 1997, 134, 74.	0.8	0
71	Familial ligand-defective apolipoprotein B-100: Simultaneous detection of the ARG3500â†'GLN and ARG3531â†'CYS mutations in a French population. Human Mutation, 1997, 10, 160-163.	2.5	31
72	Genetics of NIDDM in France: studies with 19 candidate genes in affected sib pairs. Diabetes, 1997, 46, 1062-1068.	0.6	21

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73	L'hypercholestÃ©rolÃ©mie familiale 25 ans aprÃ¨s.I- DÃ©fauts du rÃ©cepteur des LDL. Medecine/Sciences, 1997, 13, 1399.	0.2	3
74	L'hypercholestÃ©rolÃ©mie familiale 25 ans aprÃ¨s.II- Formes non-liÃ©es au rÃ©cepteur des LDL. Medecine/Sciences, 1997, 13, 1409.	0.2	1
75	Missense Mutation in the LDLR Gene: A Wide Spectrum in the Severity of Familial Hypercholesterolemia. , 0, , .		7