Mathilde Varret

List of Publications by Year
in descending order

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75 papers	6,649 citations	212478 28 h-index	107981 68 g-index
82 all docs	82 docs citations	82 times ranked	7184 citing authors

#	Article	IF	Citations
1	Mutations in PCSK9 cause autosomal dominant hypercholesterolemia. Nature Genetics, 2003, 34, 154-156.	9.4	2,532
2	Heterozygous TGFBR2 mutations in Marfan syndrome. Nature Genetics, 2004, 36, 855-860.	9.4	577
3	NARC-1/PCSK9 and Its Natural Mutants. Journal of Biological Chemistry, 2004, 279, 48865-48875.	1.6	544
4	TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. Nature Genetics, 2012, 44, 916-921.	9.4	319
5	Mutations and polymorphisms in the proprotein convertase subtilisin kexin 9 (PCSK9) gene in cholesterol metabolism and disease. Human Mutation, 2009, 30, 520-529.	1.1	211
6	Apolipoprotein B100 Metabolism in Autosomal-Dominant Hypercholesterolemia Related to Mutations in PCSK9. Arteriosclerosis, Thrombosis, and Vascular Biology, 2004, 24, 1448-1453.	1.1	171
7	Novel mutations of thePCSK9 gene cause variable phenotype of autosomal dominant hypercholesterolemia. Human Mutation, 2005, 26, 497-497.	1.1	169
8	Genetic heterogeneity of autosomal dominant hypercholesterolemia. Clinical Genetics, 2008, 73, 1-13.	1.0	160
9	A Third Major Locus for Autosomal Dominant Hypercholesterolemia Maps to 1p34.1-p32. American Journal of Human Genetics, 1999, 64, 1378-1387.	2.6	154
10	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.	1.2	116
11	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.	2.6	110
12	The UMD-LDLR database: additions to the software and 490 new entries to the database. Human Mutation, 2002, 20, 81-87.	1.1	105
13	Description of a Large Family with Autosomal Dominant Hypercholesterolemia Associated with theAPOEp.Leu167del Mutation. Human Mutation, 2013, 34, 83-87.	1.1	103
14	Molecular Spectrum of Autosomal Dominant Hypercholesterolemia in France. Human Mutation, 2010, 31, E1811-E1824.	1.1	99
15	Identification and characterization of new gain-of-function mutations in the PCSK9 gene responsible for autosomal dominant hypercholesterolemia. Atherosclerosis, 2012, 223, 394-400.	0.4	92
16	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.	2.0	87
17	The molecular basis of familial hypercholesterolemia in Lebanon: Spectrum ofLDLRmutations and role ofPCSK9as a modifier gene. Human Mutation, 2009, 30, E682-E691.	1.1	82
18	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.	6.5	77

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19	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. Human Mutation, 2012, 33, 1175-1181.	1.1	74
20	APOE gene variants in primary dyslipidemia. Atherosclerosis, 2021, 328, 11-22.	0.4	60
21	Software and database for the analysis of mutations in the human LDL receptor gene. Nucleic Acids Research, 1997, 25, 172-180.	6.5	50
22	Exome Sequencing in Suspected Monogenic Dyslipidemias. Circulation: Cardiovascular Genetics, 2015, 8, 343-350.	5.1	45
23	A PCSK9 variant and familial combined hyperlipidaemia. Journal of Medical Genetics, 2008, 45, 780-786.	1.5	39
24	A fourth locus for autosomal dominant hypercholesterolemia maps at 16q22.1. European Journal of Human Genetics, 2010, 18, 1236-1242.	1.4	38
25	High burden of recurrent cardiovascular events in heterozygous familial hypercholesterolemia: The French Familial Hypercholesterolemia Registry. Atherosclerosis, 2018, 277, 334-340.	0.4	33
26	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.	1.1	31
27	PCSK9 Mutations in Familial Hypercholesterolemia: from a Groundbreaking Discovery to Anti-PCSK9 Therapies. Current Atherosclerosis Reports, 2017, 19, 49.	2.0	31
28	New Insights into How Adipocytes Sense their Triglyceride Stores. Is Cholesterol a Signal?. Hormone and Metabolic Research, 2003, 35, 204-210.	0.7	30
29	R3531C Mutation in the Apolipoprotein B Gene Is Not Sufficient to Cause Hypercholesterolemia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2000, 20, E76-82.	1.1	28
30	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
31	Familial hypercholesterolemia in Morocco: first report of mutations in the LDL receptor gene. Journal of Human Genetics, 2003, 48, 199-203.	1.1	27
32	New Sequencing technologies help revealing unexpected mutations in Autosomal Dominant Hypercholesterolemia. Scientific Reports, 2018, 8, 1943.	1.6	25
33	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
34	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
35	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.	1.2	22
36	Effect of mutations in LDLR and PCSK9 genes on phenotypic variability in Tunisian familial hypercholesterolemia patients. Atherosclerosis, 2012, 222, 158-166.	0.4	22

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37	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
38	Mutational heterogeneity in low-density lipoprotein receptor gene related to familial hypercholesterolemia in Morocco. Clinica Chimica Acta, 2006, 373, 62-69.	0.5	21
39	Novel LRP5 gene mutation in a patient with osteoporosis-pseudoglioma syndrome. Joint Bone Spine, 2010, 77, 151-153.	0.8	21
40	Genetics of NIDDM in France: studies with 19 candidate genes in affected sib pairs. Diabetes, 1997, 46, 1062-1068.	0.3	21
41	Limited mutational heterogeneity in the LDLR gene in familial hypercholesterolemia in Tunisia. Atherosclerosis, 2009, 203, 449-453.	0.4	17
42	Moderate phenotypic expression of familial hypercholesterolemia in Tunisia. Clinica Chimica Acta, 2010, 411, 735-738.	0.5	17
43	Plasma proproteinâ€convertaseâ€subtilisin/kexin type 9 (PCSK9) and cardiovascular events in type 2 diabetes. Diabetes, Obesity and Metabolism, 2018, 20, 943-953.	2.2	17
44	A novel splice site mutation of the LDL receptor gene in a Tunisian hypercholesterolemic family. Clinica Chimica Acta, 2008, 392, 25-29.	0.5	16
45	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.	1.4	15
46	Angiotensin-Converting Enzyme Gene Does Not Contribute to Genetic Susceptibility to Systemic Sclerosis in European Caucasians. Journal of Rheumatology, 2009, 36, 337-340.	1.0	15
47	Identification of the first Lebanese mutation in the LPL gene and description of a rapid detection method. Clinical Genetics, 2004, 65, 158-161.	1.0	12
48	Dermal tissue and cellular expression of fibrillin-1 in diffuse cutaneous systemic sclerosis. Rheumatology, 2010, 49, 657-661.	0.9	10
49	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.	0.7	9
50	Posttranscriptional Regulation of the Human LDL Receptor by the U2-Spliceosome. Circulation Research, 2022, 130, 80-95.	2.0	9
51	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.	0.9	8
52	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.	0.9	7
53	Missense Mutation in the LDLR Gene: A Wide Spectrum in the Severity of Familial Hypercholesterolemia. , 0, , .		7
54	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.	0.8	6

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55	Identification of the first Tangier disease patient in Lebanon carrying a new pathogenic variant in ABCA1. Journal of Clinical Lipidology, 2018, 12, 1374-1382.	0.6	6
56	Postprandial lipid absorption in seven heterozygous carriers of deleterious variants of MTTP in two abetalipoproteinemic families. Journal of Clinical Lipidology, 2019, 13, 201-212.	0.6	6
57	Genomic characterization of two deletions in the LDLR gene in Tunisian patients with familial hypercholesterolemia. Clinica Chimica Acta, 2012, 414, 146-151.	0.5	5
58	Identification of a Variant in APOB Gene as a Major Cause of Hypobetalipoproteinemia in Lebanese Families. Metabolites, 2021, 11, 564.	1.3	5
59	APOE Molecular Spectrum in a French Cohort with Primary Dyslipidemia. International Journal of Molecular Sciences, 2022, 23, 5792.	1.8	4
60	Analysis of the 525 point mutations in the human LDL receptor gene database. Atherosclerosis, 1999, 144, 182-183.	0.4	3
61	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.	1.0	3
62	L'hypercholestÃ©rolÃ©mie familiale 25 ans aprÃ¨s.I- DÃ©fauts du rÃ©cepteur des LDL. Medecine/Sciences, 1997, 13, 1399.	0.0	3
63	Autosomal Dominant Hypercholesterolemia: Needs for Early Diagnosis and Cascade Screening in the Tunisian Population. Current Genomics, 2013, 14, 25-32.	0.7	1
64	Plasma PCSK9 and cardiovascular events in type 2 diabetes. Atherosclerosis, 2017, 263, e81.	0.4	1
65	L'hypercholestÃ©rolÃ©mie familiale 25 ans aprÃ¨s.II- Formes non-liÃ©es au rÃ©cepteur des LDL. Medecine/Sciences, 1997, 13, 1409.	0.0	1
66	Whole Exome/Genome Sequencing Joint Analysis of a Family with Oligogenic Familial Hypercholesterolemia. Metabolites, 2022, 12, 262.	1.3	1
67	Circulating PCSK9 Linked to Dyslipidemia in Lebanese Schoolchildren. Metabolites, 2022, 12, 504.	1.3	1
68	1.P.275 Results of the molecular analysis of the 220 point mutations in the human LDL receptor gene database. Atherosclerosis, 1997, 134, 74.	0.4	0
69	PC9, A New Actor in Autosomal Dominant Hypercholesterolemia. Current Genomics, 2005, 6, 535-543.	0.7	0
70	LOCALISATION OF A FIFTH GENE INVOLVED IN AUTOSOMAL DOMINANT HYPERCHOLESTEROLEMIA. Atherosclerosis Supplements, 2008, 9, 33.	1.2	0
71	PCSK9 FROM GENE AND VARIANTS TO PROTEIN AND PHENOTYPE. Atherosclerosis Supplements, 2008, 9, 101.	1.2	0
72	Identification of a new mutation in the N-terminal region of the apolipoprotein B gene in familial hypercholesterolemia. Atherosclerosis, 2016, 252, e34.	0.4	0

#	Article	IF	Citations
73	Effect of the p.Arg357His mutation of PCSK9 on basal and postprandial lipoprotein metabolism. Atherosclerosis, 2017, 263, e2.	0.4	0
74	Usefulness of the genetic risk score to identify phenocopies in families with autosomal dominant hypercholesterolemia?. Atherosclerosis, 2017, 263, e83.	0.4	0
75	Ephrin-B2 PB-mononuclear cells reduce early post-stroke deficit in diabetic mice but not long-term memory impairment. Experimental Neurology, 2021, 346, 113864.	2.0	0