Sisa-Pufacol Study Group

List of Publications by Year
in descending order

76 papers	2,597 citations	185998 28 h-index	205818 48 g-index
76 all docs	76 docs citations	76 times ranked	3114 citing authors

#	ARTICLE	IF	CITATIONS
1	Inherited Apolipoprotein A-V Deficiency in Severe Hypertriglyceridemia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2005, 25, 411-417.	1.1	177
2	The Molecular Basis of Lecithin:Cholesterol Acyltransferase Deficiency Syndromes. Arteriosclerosis, Thrombosis, and Vascular Biology, 2005, 25, 1972-1978.	1.1	158
3	Spectrum of mutations and phenotypic expression in patients with autosomal dominant hypercholesterolemia identified in Italy. Atherosclerosis, 2013, 227, 342-348.	0.4	128
4	Characterization of Three Kindreds With Familial Combined Hypolipidemia Caused by Loss-of-Function Mutations of ANGPTL3. Circulation: Cardiovascular Genetics, 2012, 5, 42-50.	5.1	115
5	Toward an international consensusâ€"Integrating lipoprotein apheresis and new lipid-lowering drugs. Journal of Clinical Lipidology, 2017, 11, 858-871.e3.	0.6	105
6	Additive effect of mutations in LDLR and PCSK9 genes on the phenotype of familial hypercholesterolemia. Atherosclerosis, 2006, 186, 433-440.	0.4	97
7	Autosomal recessive hypercholesterolemia (ARH) and homozygous familial hypercholesterolemia (FH): A phenotypic comparison. Atherosclerosis, 2006, 188, 398-405.	0.4	84
8	Clinical characteristics, management and in-hospital mortality of patients with coronavirus disease 2019 in Genoa, Italy. Clinical Microbiology and Infection, 2020, 26, 1537-1544.	2.8	84
9	Genetic polymorphisms affecting the phenotypic expression of familial hypercholesterolemia. Atherosclerosis, 2004, 174, 57-65.	0.4	77
10	Clinical characteristics and plasma lipids in subjects with familial combined hypolipidemia: a pooled analysis. Journal of Lipid Research, 2013, 54, 3481-3490.	2.0	76
11	Lysosomal lipase deficiency: Molecular characterization of eleven patients with Wolman or cholesteryl ester storage disease. Molecular Genetics and Metabolism, 2012, 105, 450-456.	0.5	71
12	Effect of ezetimibe coadministered with statins in genotype-confirmed heterozygous FH patients. Atherosclerosis, 2007, 194, e116-e122.	0.4	68
13	Nutraceutical pill containing berberine versus ezetimibe on plasma lipid pattern in hypercholesterolemic subjects and its additive effect in patients with familial hypercholesterolemia on stable cholesterol-lowering treatment. Lipids in Health and Disease, 2012, 11, 123.	1.2	68
14	Spectrum of mutations in Italian patients with familial hypercholesterolemia: New results from the LIPIGEN study. Atherosclerosis Supplements, 2017, 29, 17-24.	1.2	65
15	The Type of LDLR Gene Mutation Predicts Cardiovascular Risk in Children with Familial Hypercholesterolemia. Journal of Pediatrics, 2009, 155, 199-204.e2.	0.9	62
16	Cholesteryl Ester Storage Disease (CESD) due to novel mutations in the LIPA gene. Molecular Genetics and Metabolism, 2009, 97, 143-148.	0.5	59
17	Spectrum of mutations of the LPL gene identified in Italy in patients with severe hypertriglyceridemia. Atherosclerosis, 2015, 241, 79-86.	0.4	55
18	Traditional and non traditional risk factors in accelerated atherosclerosis in Systemic Lupus Erythematosus: Role of vascular endothelial growth factor (VEGATS Study). Autoimmunity Reviews, 2009, 8, 309-315.	2.5	54

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19	Familial hypercholesterolemia: The Italian Atherosclerosis Society Network (LIPIGEN). Atherosclerosis Supplements, 2017, 29, 11-16.	1.2	53
20	Evaluation of the performance of Dutch Lipid Clinic Network score in an Italian FH population: The LIPIGEN study. Atherosclerosis, 2018, 277, 413-418.	0.4	48
21	Familial HDL deficiency due to ABCA1 gene mutations with or without other genetic lipoprotein disorders. Atherosclerosis, 2004, 172, 309-320.	0.4	47
22	A novel sequence variant in APOA5 gene found in patients with severe hypertriglyceridemia. Atherosclerosis, 2006, 188, 215-217.	0.4	47
23	PCSK9 inhibitors for treating hypercholesterolemia. Expert Opinion on Pharmacotherapy, 2020, 21, 353-363.	0.9	47
24	Recurrent mutations of the apolipoprotein A-I gene in three kindreds with severe HDL deficiency. Atherosclerosis, 2003, 167, 335-345.	0.4	34
25	Effect of statins on LDL particle size in patients with familial combined hyperlipidemia: a comparison between atorvastatin and pravastatin. Nutrition, Metabolism and Cardiovascular Diseases, 2005, 15, 47-55.	1.1	33
26	Abnormal splicing of ABCA1 pre-mRNA in Tangier disease due to a IVS2 +5G>C mutation in ABCA1 gene. Journal of Lipid Research, 2003, 44, 254-264.	2.0	29
27	Influence of Î²0-Thalassemia on the Phenotypic Expression of Heterozygous Familial Hypercholesterolemia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2000, 20, 236-243.	1.1	28
28	Novel mutations of ABCA1 transporter in patients with Tangier disease and familial HDL deficiency. Molecular Genetics and Metabolism, 2012, 107, 534-541.	0.5	28
29	Î²-Thalassemia Is a Modifying Factor of the Clinical Expression of Familial Hypercholesterolemia. Seminars in Vascular Medicine, 2004, 4, 271-278.	2.1	25
30	A three month-old infant with severe hyperchylomicronemia: Molecular diagnosis and extracorporeal treatment. Atherosclerosis Supplements, 2013, 14, 73-76.	1.2	25
31	Homozygous familial hypercholesterolemia in Italy: Clinical and molecular features. Atherosclerosis, 2020, 312, 72-78.	0.4	25
32	Serum homocysteine, methylenetetrahydrofolate reductase gene polymorphism and cardiovascular disease in heterozygous familial hypercholesterolemia. Atherosclerosis, 2005, 179, 333-338.	0.4	24
33	Baseline hsâ€CRP predicts hypertension remission in metabolic syndrome. European Journal of Clinical Investigation, 2019, 49, e13128.	1.7	24
34	Proprotein Convertase Subtilisin Kexin Type 9 Inhibition for Autosomal Recessive Hypercholesterolemiaâ€"Brief Report. Arteriosclerosis, Thrombosis, and Vascular Biology, 2016, 36, 1647-1650.	1.1	23
35	Molecular and clinical characterization of a series of patients with childhood-onset lysosomal acid lipase deficiency. Retrospective investigations, follow-up and detection of two novel LIPA pathogenic variants. Atherosclerosis, 2017, 265, 124-132.	0.4	19
36	Combined monogenic hypercholesterolemia and hypoalphalipoproteinemia caused by mutations in LDL-R and LCAT genes. Atherosclerosis, 2005, 182, 153-159.	0.4	18

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37	Two novel rare variants of APOA5 gene found in subjects with severe hypertriglyceridemia. Clinica Chimica Acta, 2011, 412, 2194-2198.	0.5	17
38	Testing the Short-Term Efficacy of a Lipid-Lowering Nutraceutical in the Setting of Clinical Practice: A Multicenter Study. Journal of Medicinal Food, 2015, 18, 1270-1273.	0.8	17
39	Lipoprotein(a) concentration, genetic variants, apo(a) isoform size, and cellular cholesterol efflux in patients with elevated Lp(a) and coronary heart disease submitted or not to lipoprotein apheresis: An Italian case-control multicenter study on Lp(a). Journal of Clinical Lipidology, 2020, 14, 487-497.e1.	0.6	17
40	Lipoproteins, Stroke and Statins. Current Vascular Pharmacology, 2015, 13, 202-208.	0.8	17
41	Evaluation of RNA messengers involved in lipid trafficking of human intestinal cells by reverse-transcription polymerase chain reaction with competimer technology and microchip electrophoresis. Electrophoresis, 2003, 24, 3748-3754.	1.3	16
42	Denaturing high-performance liquid chromatography in the detection of ABCA1 gene mutations in familial HDL deficiency. Journal of Lipid Research, 2005, 46, 817-822.	2.0	16
43	Timely diagnosis of sitosterolemia by next generation sequencing in two children with severe hypercholesterolemia. Atherosclerosis, 2017, 262, 71-77.	0.4	16
44	The study of familial hypercholesterolemia in Italy: A narrative review. Atherosclerosis Supplements, 2017, 29, 1-10.	1.2	16
45	Clinical efficacy of eucaloric ketogenic nutrition in the COVID-19 cytokine storm: A retrospective analysis of mortality and intensive care unit admission. Nutrition, 2021, 89, 111236.	1.1	16
46	Multiple abnormally spliced ABCA1 mRNAs caused by a novel splice site mutation of ABCA1 gene in a patient with Tangier disease. Clinica Chimica Acta, 2010, 411, 524-530.	0.5	15
47	Pharmacological treatment of a Sardinian patient affected by Autosomal Recessive Hypercholesterolemia (ARH). Journal of Clinical Lipidology, 2015, 9, 103-106.	0.6	15
48	Plasma PCSK9 levels and lipoprotein distribution are preserved in carriers of genetic HDL disorders. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2018, 1863, 991-997.	1.2	14
49	Serum lipoprotein (a) predicts acute coronary syndromes in patients with severe carotid stenosis. European Journal of Clinical Investigation, 2018, 48, e12888.	1.7	13
50	Molecular characterization of two patients with severe LCAT deficiency. Nephrology Dialysis Transplantation, 2007, 22, 2379-2382.	0.4	12
51	Pseudoxanthoma elasticum and familial hypercholesterolemia: A deleterious combination of cardiovascular risk factors. Atherosclerosis, 2010, 210, 173-176.	0.4	12
52	Lipoprotein Glomerulopathy Associated with a Mutation in Apolipoprotein E. Clinical Medicine Insights: Case Reports, 2013, 6, CCRep.S12209.	0.3	12
53	Long term substrate reduction therapy with ezetimibe alone or associated with statins in three adult patients with lysosomal acid lipase deficiency. Orphanet Journal of Rare Diseases, 2018, 13, 24.	1.2	12
54	Efficacy of Nutraceutical Combination of Monacolin K, Berberine, and Silymarin on Lipid Profile and PCSK9 Plasma Level in a Cohort of Hypercholesterolemic Patients. Journal of Medicinal Food, 2020, 23, 658-666.	0.8	12

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55	LIPA gene mutations affect the composition of lipoproteins: Enrichment in ACAT-derived cholesteryl esters. Atherosclerosis, 2020, 297, 8-15.	0.4	12
56	Long-term efficacy of lipoprotein apheresis and lomitapide in the treatment of homozygous familial hypercholesterolemia (HoFH): a cross-national retrospective survey. Orphanet Journal of Rare Diseases, 2021, 16, 381.	1.2	12
57	A â€œde novoâ€•mutation of the LDL-receptor gene as the cause of familial hypercholesterolemia. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1587, 7-11.	1.8	11
58	A silent mutation of Niemann-Pick C1-like 1 and apolipoprotein E4 modulate cholesterol absorption in primary hyperlipidemias. Journal of Clinical Lipidology, 2013, 7, 147-152.	0.6	11
59	Effects of a Mediterranean Diet, Dairy, and Meat Products on Different Phenotypes of Dyslipidemia: A Preliminary Retrospective Analysis. Nutrients, 2021, 13, 1161.	1.7	10
60	An integrated approach to the evaluation of patients with asymptomatic or minimally symptomatic <scp>hyperCKemia</scp>. Muscle and Nerve, 2022, 65, 96-104.	1.0	10
61	A 33-year-old man with nephrotic syndrome and lecithin-cholesterol acyltransferase (LCAT) deficiency. Description of two new mutations in the LCAT gene. Nephrology Dialysis Transplantation, 2004, 19, 1622-1624.	0.4	9
62	A novel mutation of the apolipoprotein A-I gene in a family with familial combined hyperlipidemia. Atherosclerosis, 2008, 198, 145-151.	0.4	9
63	Serum osteopontin predicts glycaemic profile improvement in metabolic syndrome: A pilot study. European Journal of Clinical Investigation, 2021, 51, e13403.	1.7	8
64	An apparent inconsistency in parent to offspring transmission of point mutations of LDLR gene in Familial Hypercholesterolemia. Clinica Chimica Acta, 2009, 406, 75-80.	0.5	7
65	A successful term pregnancy with severe hypertriglyceridaemia and acute pancreatitis. Clinical management and review of the literature. Atherosclerosis Supplements, 2019, 40, 117-121.	1.2	7
66	A complex phenotype in a child with familial HDL deficiency due to a novel frameshift mutation in APOA1 gene (apoA-I Guastalla). Journal of Clinical Lipidology, 2015, 9, 837-846.	0.6	6
67	Short-term effect of rosuvastatin treatment on arterial stiffness in individuals with newly-diagnosed heterozygous familial hypercholesterolemia. International Journal of Cardiology, 2018, 255, 215-220.	0.8	6
68	Effect of a common missense variant in LIPA gene on fatty liver disease and lipid phenotype: New perspectives from a singleâ€center observational study. Pharmacology Research and Perspectives, 2021, 9, e00820.	1.1	6
69	Long term follow-up of genetically confirmed patients with familial hypercholesterolemia treated with first and second-generation statins and then with PCSK9 monoclonal antibodies. Atherosclerosis, 2020, 308, 6-14.	0.4	5
70	Mediterranean Diet Adherence in a Sample of Italian Adolescents Attending Secondary Schoolâ€”The â€œ#facciamoComunicAzioneâ€•Project. Nutrients, 2021, 13, 2806.	1.7	5
71	Cholesterol Lowering Therapy: Treat to Target or Reduce the Global Risk? The Unresolved Problem of Residual Risk. Current Pharmaceutical Design, 2016, 22, 5676-5686.	0.9	4
72	Quantitative polymerase chain reaction and microchip electrophoresis to detect major rearrangements of the low-density lipoprotein receptor gene causing familial hypercholesterolemia. Electrophoresis, 2004, 25, 3882-3889.	1.3	3

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73	InÂvitro functional characterization of splicing variants of the APOB gene found in familial hypobetalipoproteinemia. Journal of Clinical Lipidology, 2019, 13, 960-969.	0.6	3
74	Diet and Nutraceutical Supplementation in Dyslipidemic Patients: First Results of an Italian Single Center Real-World Retrospective Analysis. Nutrients, 2020, 12, 2056.	1.7	3
75	Hepatic Elastometry and Glissonian Line in the Assessment of Liver Fibrosis. Ultrasound in Medicine and Biology, 2021, 47, 947-959.	0.7	3
76	Homozygous familial hypercholesterolaemia in childhood â€“ The first case report in Southeast Europe. Atherosclerosis Supplements, 2019, 40, 122-124.	1.2	2