

# Sisa-Pufacol Study Group

## List of Publications by Citations

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75  
papers

2,018  
citations

26  
h-index

42  
g-index

76  
ext. papers

2,328  
ext. citations

3.5  
avg, IF

3.98  
L-index

#	Paper	IF	Citations
75	Inherited apolipoprotein A-V deficiency in severe hypertriglyceridemia. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2005</b> , 25, 411-7	9.4	154
74	The molecular basis of lecithin:cholesterol acyltransferase deficiency syndromes: a comprehensive study of molecular and biochemical findings in 13 unrelated Italian families. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2005</b> , 25, 1972-8	9.4	136
73	Spectrum of mutations and phenotypic expression in patients with autosomal dominant hypercholesterolemia identified in Italy. <i>Atherosclerosis</i> , <b>2013</b> , 227, 342-8	3.1	106
72	Characterization of three kindreds with familial combined hypolipidemia caused by loss-of-function mutations of ANGPTL3. <i>Circulation: Cardiovascular Genetics</i> , <b>2012</b> , 5, 42-50		89
71	Additive effect of mutations in LDLR and PCSK9 genes on the phenotype of familial hypercholesterolemia. <i>Atherosclerosis</i> , <b>2006</b> , 186, 433-40	3.1	82
70	Toward an international consensus-Integrating lipoprotein apheresis and new lipid-lowering drugs. <i>Journal of Clinical Lipidology</i> , <b>2017</b> , 11, 858-871.e3	4.9	80
69	Autosomal recessive hypercholesterolemia (ARH) and homozygous familial hypercholesterolemia (FH): a phenotypic comparison. <i>Atherosclerosis</i> , <b>2006</b> , 188, 398-405	3.1	72
68	Genetic polymorphisms affecting the phenotypic expression of familial hypercholesterolemia. <i>Atherosclerosis</i> , <b>2004</b> , 174, 57-65	3.1	68
67	Effect of ezetimibe coadministered with statins in genotype-confirmed heterozygous FH patients. <i>Atherosclerosis</i> , <b>2007</b> , 194, e116-22	3.1	62
66	Lysosomal lipase deficiency: molecular characterization of eleven patients with Wolman or cholesteryl ester storage disease. <i>Molecular Genetics and Metabolism</i> , <b>2012</b> , 105, 450-6	3.7	60
65	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. <i>Lipids in Health and Disease</i> , <b>2012</b> , 11, 123	4.4	60
64	Cholesteryl Ester Storage Disease (CESD) due to novel mutations in the LIPA gene. <i>Molecular Genetics and Metabolism</i> , <b>2009</b> , 97, 143-8	3.7	56
63	Clinical characteristics and plasma lipids in subjects with familial combined hypolipidemia: a pooled analysis. <i>Journal of Lipid Research</i> , <b>2013</b> , 54, 3481-90	6.3	55
62	The type of LDLR gene mutation predicts cardiovascular risk in children with familial hypercholesterolemia. <i>Journal of Pediatrics</i> , <b>2009</b> , 155, 199-204.e2	3.6	55
61	Clinical characteristics, management and in-hospital mortality of patients with coronavirus disease 2019 in Genoa, Italy. <i>Clinical Microbiology and Infection</i> , <b>2020</b> , 26, 1537-1544	9.5	50
60	Spectrum of mutations in Italian patients with familial hypercholesterolemia: New results from the LIPIGEN study. <i>Atherosclerosis Supplements</i> , <b>2017</b> , 29, 17-24	1.7	45
59	Traditional and non traditional risk factors in accelerated atherosclerosis in systemic lupus erythematosus: role of vascular endothelial growth factor (VEGATS Study). <i>Autoimmunity Reviews</i> , <b>2009</b> , 8, 309-15	13.6	44

58	Spectrum of mutations of the LPL gene identified in Italy in patients with severe hypertriglyceridemia. <i>Atherosclerosis</i> , <b>2015</b> , 241, 79-86	3.1	43
57	A novel sequence variant in APOA5 gene found in patients with severe hypertriglyceridemia. <i>Atherosclerosis</i> , <b>2006</b> , 188, 215-7	3.1	40
56	Familial hypercholesterolemia: The Italian Atherosclerosis Society Network (LIPIGEN). <i>Atherosclerosis Supplements</i> , <b>2017</b> , 29, 11-16	1.7	38
55	Evaluation of the performance of Dutch Lipid Clinic Network score in an Italian FH population: The LIPIGEN study. <i>Atherosclerosis</i> , <b>2018</b> , 277, 413-418	3.1	35
54	Familial HDL deficiency due to ABCA1 gene mutations with or without other genetic lipoprotein disorders. <i>Atherosclerosis</i> , <b>2004</b> , 172, 309-20	3.1	33
53	Effect of statins on LDL particle size in patients with familial combined hyperlipidemia: a comparison between atorvastatin and pravastatin. <i>Nutrition, Metabolism and Cardiovascular Diseases</i> , <b>2005</b> , 15, 47-55	4.5	30
52	Recurrent mutations of the apolipoprotein A-I gene in three kindreds with severe HDL deficiency. <i>Atherosclerosis</i> , <b>2003</b> , 167, 335-45	3.1	29
51	Abnormal splicing of ABCA1 pre-mRNA in Tangier disease due to a IVS2 +5G>C mutation in ABCA1 gene. <i>Journal of Lipid Research</i> , <b>2003</b> , 44, 254-64	6.3	27
50	Influence of beta(0)-thalassemia on the phenotypic expression of heterozygous familial hypercholesterolemia : a study of patients with familial hypercholesterolemia from Sardinia. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2000</b> , 20, 236-43	9.4	26
49	Novel mutations of ABCA1 transporter in patients with Tangier disease and familial HDL deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2012</b> , 107, 534-41	3.7	25
48	A three month-old infant with severe hyperchylomicronemia: molecular diagnosis and extracorporeal treatment. <i>Atherosclerosis Supplements</i> , <b>2013</b> , 14, 73-6	1.7	21
47	Beta-thalassemia is a modifying factor of the clinical expression of familial hypercholesterolemia. <i>Seminars in Vascular Medicine</i> , <b>2004</b> , 4, 271-8		21
46	Serum homocysteine, methylenetetrahydrofolate reductase gene polymorphism and cardiovascular disease in heterozygous familial hypercholesterolemia. <i>Atherosclerosis</i> , <b>2005</b> , 179, 333-8	3.1	20
45	Proprotein Convertase Subtilisin Kexin Type 9 Inhibition for Autosomal Recessive Hypercholesterolemia-Brief Report. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2016</b> , 36, 1647-50	9.4	19
44	Two novel rare variants of APOA5 gene found in subjects with severe hypertriglyceridemia. <i>Clinica Chimica Acta</i> , <b>2011</b> , 412, 2194-8	6.2	16
43	Evaluation of RNA messengers involved in lipid trafficking of human intestinal cells by reverse-transcription polymerase chain reaction with competitor technology and microchip electrophoresis. <i>Electrophoresis</i> , <b>2003</b> , 24, 3748-54	3.6	16
42	PCSK9 inhibitors for treating hypercholesterolemia. <i>Expert Opinion on Pharmacotherapy</i> , <b>2020</b> , 21, 353-363	3.1	16
41	Combined monogenic hypercholesterolemia and hypoalphalipoproteinemia caused by mutations in LDL-R and LCAT genes. <i>Atherosclerosis</i> , <b>2005</b> , 182, 153-9	3.1	15

40	Lipoproteins, stroke and statins. <i>Current Vascular Pharmacology</i> , <b>2015</b> , 13, 202-8	3.3	15
39	Testing the Short-Term Efficacy of a Lipid-Lowering Nutraceutical in the Setting of Clinical Practice: A Multicenter Study. <i>Journal of Medicinal Food</i> , <b>2015</b> , 18, 1270-3	2.8	13
38	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. <i>Atherosclerosis</i> , <b>2017</b> , 265, 124-132	3.1	13
37	Pharmacological treatment of a Sardinian patient affected by Autosomal Recessive Hypercholesterolemia (ARH). <i>Journal of Clinical Lipidology</i> , <b>2015</b> , 9, 103-6	4.9	13
36	Multiple abnormally spliced ABCA1 mRNAs caused by a novel splice site mutation of ABCA1 gene in a patient with Tangier disease. <i>Clinica Chimica Acta</i> , <b>2010</b> , 411, 524-30	6.2	13
35	The study of familial hypercholesterolemia in Italy: A narrative review. <i>Atherosclerosis Supplements</i> , <b>2017</b> , 29, 1-10	1.7	12
34	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). <i>Journal of Clinical Lipidology</i> , <b>2020</b> , 14, 487-497.e1	4.9	12
33	Denaturing high-performance liquid chromatography in the detection of ABCA1 gene mutations in familial HDL deficiency. <i>Journal of Lipid Research</i> , <b>2005</b> , 46, 817-22	6.3	12
32	Timely diagnosis of sitosterolemia by next generation sequencing in two children with severe hypercholesterolemia. <i>Atherosclerosis</i> , <b>2017</b> , 262, 71-77	3.1	11
31	Long term substrate reduction therapy with ezetimibe alone or associated with statins in three adult patients with lysosomal acid lipase deficiency. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 24	4.2	10
30	A silent mutation of Niemann-Pick C1-like 1 and apolipoprotein E4 modulate cholesterol absorption in primary hyperlipidemias. <i>Journal of Clinical Lipidology</i> , <b>2013</b> , 7, 147-52	4.9	10
29	Baseline hs-CRP predicts hypertension remission in metabolic syndrome. <i>European Journal of Clinical Investigation</i> , <b>2019</b> , 49, e13128	4.6	9
28	Pseudoxanthoma elasticum and familial hypercholesterolemia: a deleterious combination of cardiovascular risk factors. <i>Atherosclerosis</i> , <b>2010</b> , 210, 173-6	3.1	9
27	A novel mutation of the apolipoprotein A-I gene in a family with familial combined hyperlipidemia. <i>Atherosclerosis</i> , <b>2008</b> , 198, 145-51	3.1	9
26	Molecular characterization of two patients with severe LCAT deficiency. <i>Nephrology Dialysis Transplantation</i> , <b>2007</b> , 22, 2379-82	4.3	9
25	A 33-year-old man with nephrotic syndrome and lecithin-cholesterol acyltransferase (LCAT) deficiency. Description of two new mutations in the LCAT gene. <i>Nephrology Dialysis Transplantation</i> , <b>2004</b> , 19, 1622-4	4.3	9
24	Efficacy of Nutraceutical Combination of Monacolin K, Berberine, and Silymarin on Lipid Profile and PCSK9 Plasma Level in a Cohort of Hypercholesterolemic Patients. <i>Journal of Medicinal Food</i> , <b>2020</b> , 23, 658-666	2.8	9
23	Plasma PCSK9 levels and lipoprotein distribution are preserved in carriers of genetic HDL disorders. <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , <b>2018</b> , 1863, 991-997	5	9

22	Clinical efficacy of eucaloric ketogenic nutrition in the COVID-19 cytokine storm: A retrospective analysis of mortality and intensive care unit admission. <i>Nutrition</i> , <b>2021</b> , 89, 111236	4.8	9
21	Serum lipoprotein (a) predicts acute coronary syndromes in patients with severe carotid stenosis. <i>European Journal of Clinical Investigation</i> , <b>2018</b> , 48, e12888	4.6	7
20	Lipoprotein glomerulopathy associated with a mutation in apolipoprotein e. <i>Clinical Medicine Insights: Case Reports</i> , <b>2013</b> , 6, 189-96	0.8	7
19	An apparent inconsistency in parent to offspring transmission of point mutations of LDLR gene in familial hypercholesterolemia. <i>Clinica Chimica Acta</i> , <b>2009</b> , 406, 75-80	6.2	7
18	A "de novo" mutation of the LDL-receptor gene as the cause of familial hypercholesterolemia. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2002</b> , 1587, 7-11	6.9	7
17	Homozygous familial hypercholesterolemia in Italy: Clinical and molecular features. <i>Atherosclerosis</i> , <b>2020</b> , 312, 72-78	3.1	7
16	A complex phenotype in a child with familial HDL deficiency due to a novel frameshift mutation in APOA1 gene (apoA-IGuastalla). <i>Journal of Clinical Lipidology</i> , <b>2015</b> , 9, 837-846	4.9	6
15	Short-term effect of rosuvastatin treatment on arterial stiffness in individuals with newly-diagnosed heterozygous familial hypercholesterolemia. <i>International Journal of Cardiology</i> , <b>2018</b> , 255, 215-220	3.2	5
14	LIPA gene mutations affect the composition of lipoproteins: Enrichment in ACAT-derived cholesteryl esters. <i>Atherosclerosis</i> , <b>2020</b> , 297, 8-15	3.1	4
13	Cholesterol Lowering Therapy: Treat to Target or Reduce the Global Risk? The Unresolved Problem of Residual Risk. <i>Current Pharmaceutical Design</i> , <b>2016</b> , 22, 5676-5686	3.3	4
12	Effects of a Mediterranean Diet, Dairy, and Meat Products on Different Phenotypes of Dyslipidemia: A Preliminary Retrospective Analysis. <i>Nutrients</i> , <b>2021</b> , 13,	6.7	3
11	Quantitative polymerase chain reaction and microchip electrophoresis to detect major rearrangements of the low-density lipoprotein receptor gene causing familial hypercholesterolemia. <i>Electrophoresis</i> , <b>2004</b> , 25, 3882-9	3.6	2
10	Diet and Nutraceutical Supplementation in Dyslipidemic Patients: First Results of an Italian Single Center Real-World Retrospective Analysis. <i>Nutrients</i> , <b>2020</b> , 12,	6.7	2
9	Long term follow-up of genetically confirmed patients with familial hypercholesterolemia treated with first and second-generation statins and then with PCSK9 monoclonal antibodies. <i>Atherosclerosis</i> , <b>2020</b> , 308, 6-14	3.1	2
8	A successful term pregnancy with severe hypertriglyceridaemia and acute pancreatitis. Clinical management and review of the literature. <i>Atherosclerosis Supplements</i> , <b>2019</b> , 40, 117-121	1.7	1
7	In vitro functional characterization of splicing variants of the APOB gene found in familial hypobetalipoproteinemia. <i>Journal of Clinical Lipidology</i> , <b>2019</b> , 13, 960-969	4.9	1
6	Serum osteopontin predicts glycaemic profile improvement in metabolic syndrome: A pilot study. <i>European Journal of Clinical Investigation</i> , <b>2021</b> , 51, e13403	4.6	1
5	Mediterranean Diet Adherence in a Sample of Italian Adolescents Attending Secondary School-The "#facciamoComunicazione" Project. <i>Nutrients</i> , <b>2021</b> , 13,	6.7	1

4	Long-term efficacy of lipoprotein apheresis and lomitapide in the treatment of homozygous familial hypercholesterolemia (HoFH): a cross-national retrospective survey. <i>Orphanet Journal of Rare Diseases</i> , <b>2021</b> , 16, 381	4.2	1
3	Homozygous familial hypercholesterolaemia in childhood - The first case report in Southeast Europe. <i>Atherosclerosis Supplements</i> , <b>2019</b> , 40, 122-124	1.7	0
2	Hepatic Elastometry and Glissonian Line in the Assessment of Liver Fibrosis. <i>Ultrasound in Medicine and Biology</i> , <b>2021</b> , 47, 947-959	3.5	0
1	Effect of a common missense variant in LIPA gene on fatty liver disease and lipid phenotype: New perspectives from a single-center observational study. <i>Pharmacology Research and Perspectives</i> , <b>2021</b> , 9, e00820	3.1	0