Stefano Bertolini

List of Publications by Year in descending order

70 papers	3,469 citations	201674 27 h-index	138484 58 g-index
71 all docs	71 docs citations	71 times ranked	4083 citing authors

#	Article	IF	Citations
1	Search for familial hypercholesterolemia patients in an Italian community: A real-life retrospective study. Nutrition, Metabolism and Cardiovascular Diseases, 2022, , .	2.6	3
2	Worldwide experience of homozygous familial hypercholesterolaemia: retrospective cohort study. Lancet, The, 2022, 399, 719-728.	13.7	69
3	Serum osteopontin predicts glycaemic profile improvement in metabolic syndrome: A pilot study. European Journal of Clinical Investigation, 2021, 51, e13403.	3.4	8
4	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.	2.7	12
5	Homozygous familial hypercholesterolemia in Italy: Clinical and molecular features. Atherosclerosis, 2020, 312, 72-78.	0.8	25
6	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.	0.8	5
7	Angiopoietin-like protein 3 (ANGPTL3) deficiency and familial combined hypolipidemia. Journal of Biomedical Research, 2019, 33, 73.	1.6	25
8	Novel mutations of SAR1B gene in four children with chylomicron retention disease. Journal of Clinical Lipidology, 2019, 13, 554-562.	1.5	6
9	InÂvitro functional characterization of splicing variants of the APOB gene found in familial hypobetalipoproteinemia. Journal of Clinical Lipidology, 2019, 13, 960-969.	1.5	3
10	Baseline hsâ€CRP predicts hypertension remission in metabolic syndrome. European Journal of Clinical Investigation, 2019, 49, e13128.	3.4	24
11	Autosomal Recessive Hypercholesterolemia. Journal of the American College of Cardiology, 2018, 71, 279-288.	2.8	38
12	Serum lipoprotein (a) predicts acute coronary syndromes in patients with severe carotid stenosis. European Journal of Clinical Investigation, 2018, 48, e12888.	3.4	13
13	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.	2.7	12
14	Impact of rare variants in autosomal dominant hypercholesterolemia causing genes. Current Opinion in Lipidology, 2017, 28, 267-272.	2.7	6
15	Timely diagnosis of sitosterolemia by next generation sequencing in two children with severe hypercholesterolemia. Atherosclerosis, 2017, 262, 71-77.	0.8	16
16	Familial hypercholesterolemia: The Italian Atherosclerosis Society Network (LIPIGEN). Atherosclerosis Supplements, 2017, 29, 11-16.	1.2	53
17	Spectrum of mutations in Italian patients with familial hypercholesterolemia: New results from the LIPIGEN study. Atherosclerosis Supplements, 2017, 29, 17-24.	1.2	65
18	The study of familial hypercholesterolemia in Italy: A narrative review. Atherosclerosis Supplements, 2017, 29, 1-10.	1.2	16

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19	Incidental finding of severe hypertriglyceridemia in children. Role of multiple rare variants in genes affecting plasma triglyceride. Journal of Clinical Lipidology, 2017, 11, 1329-1337.e3.	1.5	9
20	LDL receptor phenotype and response to treatment in an italian cohort of FH patients: A retrospective analysis. Atherosclerosis, 2017, 263, e238.	0.8	0
21	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.	0.8	19
22	Characterization of Italian patients with familial hypercholesterolemia: The lipigen study. Atherosclerosis, 2017, 263, e235.	0.8	0
23	Phenotypic variability in 4 homozygous familial hypercholesterolemia siblings compound heterozygous for LDLR mutations. Journal of Clinical Lipidology, 2016, 10, 944-952.e1.	1.5	8
24	Clinical and genetic features of 3 patients with familial chylomicronemia due to mutations in GPIHBP1 gene. Journal of Clinical Lipidology, 2016, 10, 915-921.e4.	1.5	22
25	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.	1.5	6
26	Spectrum of mutations of the LPL gene identified in Italy in patients with severe hypertriglyceridemia. Atherosclerosis, 2015, 241, 79-86.	0.8	55
27	Pharmacological treatment of a Sardinian patient affected by Autosomal Recessive Hypercholesterolemia (ARH). Journal of Clinical Lipidology, 2015, 9, 103-106.	1.5	15
28	A 3-day-old neonate with severe hypertriglyceridemia from novel mutations of the GPIHBP1 gene. Journal of Clinical Lipidology, 2015, 9, 265-270.	1.5	22
29	The history of Autosomal Recessive Hypercholesterolemia (ARH). From clinical observations to gene identification. Gene, 2015, 555, 23-32.	2.2	67
30	Identifying genetic risk variants for coronary heart disease in familial hypercholesterolemia: an extreme genetics approach. European Journal of Human Genetics, 2015, 23, 381-387.	2.8	15
31	Lipoproteins, Stroke and Statins. Current Vascular Pharmacology, 2015, 13, 202-208.	1.7	17
32	Microsomal transfer protein (MTP) inhibitionâ€"a novel approach to the treatment of homozygous hypercholesterolemia. Annals of Medicine, 2014, 46, 464-474.	3.8	24
33	Whole exome sequencing of familial hypercholesterolaemia patients negative forLDLR/APOB/PCSK9mutations. Journal of Medical Genetics, 2014, 51, 537-544.	3.2	104
34	Lysosomal acid lipase deficiency â€" An under-recognized cause of dyslipidaemia and liver dysfunction. Atherosclerosis, 2014, 235, 21-30.	0.8	232
35	Spectrum of mutations and phenotypic expression in patients with autosomal dominant hypercholesterolemia identified in Italy. Atherosclerosis, 2013, 227, 342-348.	0.8	128
36	A three month-old infant with severe hyperchylomicronemia: Molecular diagnosis and extracorporeal treatment. Atherosclerosis Supplements, 2013, 14, 73-76.	1.2	25

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37	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.	1.5	11
38	Severe Hypertriglyceridemia in a Newborn with Monogenic Lipoprotein Lipase Deficiency: An Unconventional Therapeutic Approach with Exchange Transfusion. JIMD Reports, 2013, 13, 59-64.	1.5	18
39	Clinical characteristics and plasma lipids in subjects with familial combined hypolipidemia: a pooled analysis. Journal of Lipid Research, 2013, 54, 3481-3490.	4.2	76
40	Familial combined hypolipidemia due to mutations in theANGPTL3gene. Clinical Lipidology, 2013, 8, 81-95.	0.4	5
41	Lipoprotein Glomerulopathy Associated with a Mutation in Apolipoprotein E. Clinical Medicine Insights: Case Reports, 2013, 6, CCRep.S12209.	0.7	12
42	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
43	Novel mutations of ABCA1 transporter in patients with Tangier disease and familial HDL deficiency. Molecular Genetics and Metabolism, 2012, 107, 534-541.	1.1	28
44	Lysosomal lipase deficiency: Molecular characterization of eleven patients with Wolman or cholesteryl ester storage disease. Molecular Genetics and Metabolism, 2012, 105, 450-456.	1.1	71
45	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.	3.0	68
46	Two novel rare variants of APOA5 gene found in subjects with severe hypertriglyceridemia. Clinica Chimica Acta, 2011, 412, 2194-2198.	1.1	17
47	Altered mRNA splicing in lipoprotein disorders. Current Opinion in Lipidology, 2011, 22, 93-99.	2.7	13
48	Mechanisms and genetic determinants regulating sterol absorption, circulating LDL levels, and sterol elimination: implications for classification and disease risk. Journal of Lipid Research, 2011, 52, 1885-1926.	4.2	76
49	Pseudoxanthoma elasticum and familial hypercholesterolemia: A deleterious combination of cardiovascular risk factors. Atherosclerosis, 2010, 210, 173-176.	0.8	12
50	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.	1.1	15
51	The Type of LDLR Gene Mutation Predicts Cardiovascular Risk in Children with Familial Hypercholesterolemia. Journal of Pediatrics, 2009, 155, 199-204.e2.	1.8	62
52	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.	5.8	54
53	An apparent inconsistency in parent to offspring transmission of point mutations of LDLR gene in Familial Hypercholesterolemia. Clinica Chimica Acta, 2009, 406, 75-80.	1.1	7
54	Functional analysis of two novel splice site mutations of APOB gene in familial hypobetalipoproteinemia. Molecular Genetics and Metabolism, 2009, 96, 66-72.	1.1	14

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55	Cholesteryl Ester Storage Disease (CESD) due to novel mutations in the LIPA gene. Molecular Genetics and Metabolism, 2009, 97, 143-148.	1.1	59
56	Prevalence and clinical features of heterozygous carriers of autosomal recessive hypercholesterolemia in Sardinia. Atherosclerosis, 2009, 207, 162-167.	0.8	27
57	Correlation between Progetto Cuore risk score and early cardiovascular damage in never treated subjects. Cardiovascular Ultrasound, 2008, 6, 47.	1.6	4
58	A novel mutation of the apolipoprotein A-I gene in a family with familial combined hyperlipidemia. Atherosclerosis, 2008, 198, 145-151.	0.8	9
59	Molecular diagnosis of hypobetalipoproteinemia: An ENID review. Atherosclerosis, 2007, 195, e19-e27.	0.8	152
60	Autosomal recessive hypercholesterolemia (ARH) and homozygous familial hypercholesterolemia (FH): A phenotypic comparison. Atherosclerosis, 2006, 188, 398-405.	0.8	84
61	Adaptor Protein ARH Is Recruited to the Plasma Membrane by Low Density Lipoprotein (LDL) Binding and Modulates Endocytosis of the LDL/LDL Receptor Complex in Hepatocytes. Journal of Biological Chemistry, 2005, 280, 38416-38423.	3.4	31
62	Genetic polymorphisms affecting the phenotypic expression of familial hypercholesterolemia. Atherosclerosis, 2004, 174, 57-65.	0.8	77
63	Autosomal recessive hypercholesterolaemia in Sardinia, Italy, and mutations in ARH: a clinical and molecular genetic analysis. Lancet, The, 2002, 359, 841-847.	13.7	150
64	Autosomal Recessive Hypercholesterolemia Caused by Mutations in a Putative LDL Receptor Adaptor Protein. Science, 2001, 292, 1394-1398.	12.6	539
65	Paradoxes in longevity: sequence analysis of mtDNA haplogroup J in centenarians. European Journal of Human Genetics, 2001, 9, 701-707.	2.8	116
66	Three novel missense mutations in the glucokinase gene (G80S; E221K; G227C) in Italian subjects with maturity-onset diabetes of the young (MODY). Human Mutation, 1998, 12, 136-136.	2.5	19
67	LDL Apheresis with Dextran Sulfate and Angiotensin Receptor Antagonist (Losartan). Artificial Organs, 1997, 21, 334-335.	1.9	6
68	Presence of soluble amyloid Î²â€“peptide precedes amyloid plaque formation in Down's syndrome. Nature Medicine, 1996, 2, 93-95.	30.7	342
69	Lipoprotein (a) is increased in acute coronary syndromes (unstable angina pectoris and myocardial) Tj ETQq1 1 0.784314 rgBT /Overlo 526-529.	1.8	8
70	The Role of Registers in Increasing Knowledge and Improving Management of Children and Adolescents Affected by Familial Hypercholesterolemia: the LIPIGEN Pediatric Group. Frontiers in Genetics, 0, 13, .	2.3	4