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	Autosomal Recessive Hypercholesterolemia Caused by Mutations in a Putative LDL Receptor Adaptor Protein. Science, 2001, 292, 1394-1398.	12.6	539
2	Presence of soluble amyloid Î²â€“peptide precedes amyloid plaque formation in Down's syndrome. Nature Medicine, 1996, 2, 93-95.	30.7	342
3	Lysosomal acid lipase deficiency â€“ An under-recognized cause of dyslipidaemia and liver dysfunction. Atherosclerosis, 2014, 235, 21-30.	0.8	232
4	Molecular diagnosis of hypobetalipoproteinemia: An ENID review. Atherosclerosis, 2007, 195, e19-e27.	0.8	152
5	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
6	Spectrum of mutations and phenotypic expression in patients with autosomal dominant hypercholesterolemia identified in Italy. Atherosclerosis, 2013, 227, 342-348.	0.8	128
7	Paradoxes in longevity: sequence analysis of mtDNA haplogroup J in centenarians. European Journal of Human Genetics, 2001, 9, 701-707.	2.8	116
8	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
9	Whole exome sequencing of familial hypercholesterolaemia patients negative forLDLR/APOB/PCSK9mutations. Journal of Medical Genetics, 2014, 51, 537-544.	3.2	104
10	Autosomal recessive hypercholesterolemia (ARH) and homozygous familial hypercholesterolemia (FH): A phenotypic comparison. Atherosclerosis, 2006, 188, 398-405.	0.8	84
11	Genetic polymorphisms affecting the phenotypic expression of familial hypercholesterolemia. Atherosclerosis, 2004, 174, 57-65.	0.8	77
12	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
13	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
14	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
15	Worldwide experience of homozygous familial hypercholesterolaemia: retrospective cohort study. Lancet, The, 2022, 399, 719-728.	13.7	69
16	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
17	The history of Autosomal Recessive Hypercholesterolemia (ARH). From clinical observations to gene identification. Gene, 2015, 555, 23-32.	2.2	67
18	Spectrum of mutations in Italian patients with familial hypercholesterolemia: New results from the LIPIGEN study. Atherosclerosis Supplements, 2017, 29, 17-24.	1.2	65

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19	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
20	Cholesteryl Ester Storage Disease (CESD) due to novel mutations in the LIPA gene. Molecular Genetics and Metabolism, 2009, 97, 143-148.	1.1	59
21	Spectrum of mutations of the LPL gene identified in Italy in patients with severe hypertriglyceridemia. Atherosclerosis, 2015, 241, 79-86.	0.8	55
22	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
23	Familial hypercholesterolemia: The Italian Atherosclerosis Society Network (LIPIGEN). Atherosclerosis Supplements, 2017, 29, 11-16.	1.2	53
24	Autosomal Recessive Hypercholesterolemia. Journal of the American College of Cardiology, 2018, 71, 279-288.	2.8	38
25	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
26	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
27	Prevalence and clinical features of heterozygous carriers of autosomal recessive hypercholesterolemia in Sardinia. Atherosclerosis, 2009, 207, 162-167.	0.8	27
28	A three month-old infant with severe hyperchylomicronemia: Molecular diagnosis and extracorporeal treatment. Atherosclerosis Supplements, 2013, 14, 73-76.	1.2	25
29	Angiopoietin-like protein 3 (ANGPTL3) deficiency and familial combined hypolipidemia. Journal of Biomedical Research, 2019, 33, 73.	1.6	25
30	Homozygous familial hypercholesterolemia in Italy: Clinical and molecular features. Atherosclerosis, 2020, 312, 72-78.	0.8	25
31	Microsomal transfer protein (MTP) inhibitionâ€"a novel approach to the treatment of homozygous hypercholesterolemia. Annals of Medicine, 2014, 46, 464-474.	3.8	24
32	Baseline hsâ€CRP predicts hypertension remission in metabolic syndrome. European Journal of Clinical Investigation, 2019, 49, e13128.	3.4	24
33	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
34	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
35	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
36	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

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37	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
38	Two novel rare variants of APOA5 gene found in subjects with severe hypertriglyceridemia. Clinica Chimica Acta, 2011, 412, 2194-2198.	1.1	17
39	Lipoproteins, Stroke and Statins. Current Vascular Pharmacology, 2015, 13, 202-208.	1.7	17
40	Timely diagnosis of sitosterolemia by next generation sequencing in two children with severe hypercholesterolemia. Atherosclerosis, 2017, 262, 71-77.	0.8	16
41	The study of familial hypercholesterolemia in Italy: A narrative review. Atherosclerosis Supplements, 2017, 29, 1-10.	1.2	16
42	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
43	Pharmacological treatment of a Sardinian patient affected by Autosomal Recessive Hypercholesterolemia (ARH). Journal of Clinical Lipidology, 2015, 9, 103-106.	1.5	15
44	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
45	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
46	Altered mRNA splicing in lipoprotein disorders. Current Opinion in Lipidology, 2011, 22, 93-99.	2.7	13
47	Serum lipoprotein (a) predicts acute coronary syndromes in patients with severe carotid stenosis. European Journal of Clinical Investigation, 2018, 48, e12888.	3.4	13
48	Pseudoxanthoma elasticum and familial hypercholesterolemia: A deleterious combination of cardiovascular risk factors. Atherosclerosis, 2010, 210, 173-176.	0.8	12
49	Lipoprotein Glomerulopathy Associated with a Mutation in Apolipoprotein E. Clinical Medicine Insights: Case Reports, 2013, 6, CCRep.S12209.	0.7	12
50	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
51	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
52	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
53	A novel mutation of the apolipoprotein A-I gene in a family with familial combined hyperlipidemia. Atherosclerosis, 2008, 198, 145-151.	0.8	9
54	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

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55	Lipoprotein (a) is increased in acute coronary syndromes (unstable angina pectoris and myocardial) Tj ETQq1 1 0.784314 rgBT /Overlock 526-529.	1.8	8
56	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
57	Serum osteopontin predicts glycaemic profile improvement in metabolic syndrome: A pilot study. European Journal of Clinical Investigation, 2021, 51, e13403.	3.4	8
58	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
59	LDL Apheresis with Dextran Sulfate and Angiotensin Receptor Antagonist (Losartan). Artificial Organs, 1997, 21, 334-335.	1.9	6
60	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
61	Impact of rare variants in autosomal dominant hypercholesterolemia causing genes. Current Opinion in Lipidology, 2017, 28, 267-272.	2.7	6
62	Novel mutations of SAR1B gene in four children with chylomicron retention disease. Journal of Clinical Lipidology, 2019, 13, 554-562.	1.5	6
63	Familial combined hypolipidemia due to mutations in theANGPTL3gene. Clinical Lipidology, 2013, 8, 81-95.	0.4	5
64	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
65	Correlation between Progetto Cuore risk score and early cardiovascular damage in never treated subjects. Cardiovascular Ultrasound, 2008, 6, 47.	1.6	4
66	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
67	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
68	Search for familial hypercholesterolemia patients in an Italian community: A real-life retrospective study. Nutrition, Metabolism and Cardiovascular Diseases, 2022, , .	2.6	3
69	LDL receptor phenotype and response to treatment in an italian cohort of FH patients: A retrospective analysis. Atherosclerosis, 2017, 263, e238.	0.8	0
70	Characterization of Italian patients with familial hypercholesterolemia: The lipigen study. Atherosclerosis, 2017, 263, e235.	0.8	0