Mathilde Di-Filippo

List of Publications by Year
in descending order

21	573	759233 12	713466 21
papers	citations	h-index	g-index

22	22	22	1026
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Phenotypic and genotypic characterization of familial hypercholesterolemia in French adult and pediatric populations. Journal of Clinical Lipidology, 2022, 16, 298-305.	1.5	0
2	Whole Exome/Genome Sequencing Joint Analysis of a Family with Oligogenic Familial Hypercholesterolemia. Metabolites, 2022, 12, 262.	2.9	1
3	APOB CRISPR-Cas9 Engineering in Hypobetalipoproteinemia: A Promising Tool for Functional Studies of Novel Variants. International Journal of Molecular Sciences, 2022, 23, 4281.	4.1	6
4	APOE Molecular Spectrum in a French Cohort with Primary Dyslipidemia. International Journal of Molecular Sciences, 2022, 23, 5792.	4.1	4
5	Phenotypic Differences Between Polygenic and Monogenic Hypobetalipoproteinemia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2021, 41, e63-e71.	2.4	12
6	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.	1.6	9
7	Bis(monoacylglycero)phosphate, a new lipid signature of endosome-derived extracellular vesicles. Biochimie, 2020, 178, 26-38.	2.6	24
8	PCSK9 post-transcriptional regulation: Role of a 3â€²UTR microRNA-binding site variant in linkage disequilibrium with c.1420G. Atherosclerosis, 2020, 314, 63-70.	0.8	7
9	Development of a new expanded nextâ€generation sequencing panel for genetic diseases involved in dyslipidemia. Clinical Genetics, 2020, 98, 589-594.	2.0	15
10	Normal serum ApoB48 and red cells vitamin E concentrations after supplementation in a novel compound heterozygous case of abetalipoproteinemia. Atherosclerosis, 2019, 284, 75-82.	0.8	10
11	Usefulness of the genetic risk score to identify phenocopies in families with familial hypercholesterolemia?. European Journal of Human Genetics, 2018, 26, 570-578.	2.8	22
12	Efficacy of two vitamin E formulations in patients with abetalipoproteinemia and chylomicron retention disease. Journal of Lipid Research, 2018, 59, 1640-1648.	4.2	16
13	A case of hypocholesterolemia and steatosis in a carrier of a PCSK9 loss-of-function mutation and polymorphisms predisposing to nonalcoholic fatty liver disease. Journal of Clinical Lipidology, 2017, 11, 1101-1105.	1.5	10
14	Lipid profile and cardiovascular risk factors in pediatric liver transplant recipients. Pediatric Transplantation, 2016, 20, 241-248.	1.0	9
15	Global molecular analysis and APOE mutations in a cohort of autosomal dominant hypercholesterolemia patients in France. Journal of Lipid Research, 2016, 57, 482-491.	4.2	29
16	Multiple microRNA regulation of lipoprotein lipase gene abolished by 3â€²UTR polymorphisms in a triglyceride-lowering haplotype harboring p.Ser474Ter. Atherosclerosis, 2016, 246, 280-286.	0.8	23
17	An APOA5 3â€² UTR Variant Associated with Plasma Triglycerides Triggers APOA5 Downregulation by Creating a Functional miR-485-5p Binding Site. American Journal of Human Genetics, 2014, 94, 129-134.	6.2	58
18	Homozygous MTTP and APOB mutations may lead to hepatic steatosis and fibrosis despite metabolic differences in congenital hypocholesterolemia. Journal of Hepatology, 2014, 61, 891-902.	3.7	116

#	Article	IF	Citations
19	Molecular and functional analysis of two new MTTP gene mutations in an atypical case of abetalipoproteinemia. Journal of Lipid Research, 2012, 53, 548-555.	4.2	42
20	GPIHBP1 C89F Neomutation and Hydrophobic C-Terminal Domain G175R Mutation in Two Pedigrees with Severe Hyperchylomicronemia. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1675-E1679.	3.6	56
21	Molecular Spectrum of Autosomal Dominant Hypercholesterolemia in France. Human Mutation, 2010, 31, E1811-E1824.	2.5	99