Mafalda Bourbon

List of Publications by Citations

69 papers	1,638 citations	24 h-index	39 g-index
74 ext. papers	2,124 ext. citations	4.6 avg, IF	4.41 L-index

#	Paper	IF	Citations
69	Clinical Genetic Testing for Familial Hypercholesterolemia: JACC Scientific Expert Panel. *Journal of the American College of Cardiology*, **2018**, 72, 662-680	15.1	215
68	Familial hypercholesterolaemia: A global call to arms. *Atherosclerosis*, **2015**, 243, 257-9	3.1	123
67	Overview of the current status of familial hypercholesterolaemia care in over 60 countries - The EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC). *Atherosclerosis*, **2018**, 277, 234-255	3.1	93
66	Characterization of Autosomal Dominant Hypercholesterolemia Caused by PCSK9 Gain of Function Mutations and Its Specific Treatment With Alirocumab, a PCSK9 Monoclonal Antibody. *Circulation: Cardiovascular Genetics*, **2015**, 8, 823-31		76
65	Familial hypercholesterolaemia in Portugal. *Atherosclerosis*, **2008**, 196, 633-42	3.1	71
64	Characterization of a novel cellular defect in patients with phenotypic homozygous familial hypercholesterolemia. *Journal of Clinical Investigation*, **1999**, 104, 619-28	15.9	67
63	Pooling and expanding registries of familial hypercholesterolaemia to assess gaps in care and improve disease management and outcomes: Rationale and design of the global EAS Familial Hypercholesterolaemia Studies Collaboration. *Atherosclerosis Supplements*, **2016**, 22, 1-32	1.7	60
62	Novel functional APOB mutations outside LDL-binding region causing familial hypercholesterolaemia. *Human Molecular Genetics*, **2014**, 23, 1817-28	5.6	58
61	ClinVar database of global familial hypercholesterolemia-associated DNA variants. *Human Mutation* , **2018**, 39, 1631-1640	4.7	55
60	Determinants of variable response to statin treatment in patients with refractory familial hypercholesterolemia. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2001**, 21, 832-7	9.4	54
59	How good is controlled attenuation parameter and fatty liver index for assessing liver steatosis in general population: correlation with ultrasound. *Liver International*, **2014**, 34, e111-7	7.9	52
58	Analysis of publicly available LDLR, APOB, and PCSK9 variants associated with familial hypercholesterolemia: application of ACMG guidelines and implications for familial hypercholesterolemia diagnosis. *Genetics in Medicine*, **2018**, 20, 591-598	8.1	51
57	Mutational analysis and genotype-phenotype relation in familial hypercholesterolemia: The SAFEHEART registry. *Atherosclerosis*, **2017**, 262, 8-13	3.1	42
56	Update of the Portuguese Familial Hypercholesterolaemia Study. *Atherosclerosis*, **2010**, 212, 553-8	3.1	42
55	Genetic diagnosis of familial hypercholesterolaemia: the importance of functional analysis of potential splice-site mutations. *Journal of Medical Genetics*, **2009**, 46, 352-7	5.8	35
54	Functional Analysis of LDLR (Low-Density Lipoprotein Receptor) Variants in Patient Lymphocytes to Assess the Effect of Evinacumab in Homozygous Familial Hypercholesterolemia Patients With a Spectrum of LDLR Activity. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2019**, 39, 2248-2260	9.4	33
53	Lysosomal acid lipase deficiency: A hidden disease among cohorts of familial hypercholesterolemia?. *Journal of Clinical Lipidology*, **2017**, 11, 477-484.e2	4.9	33

52	Advantages and versatility of fluorescence-based methodology to characterize the functionality of LDLR and class mutation assignment. *PLoS ONE*, **2014**, 9, e112677	3.7	29
51	Structural analysis of APOB variants, p.(Arg3527Gln), p.(Arg1164Thr) and p.(Gln4494del), causing Familial Hypercholesterolaemia provides novel insights into variant pathogenicity. *Scientific Reports* , **2015**, 5, 18184	4.9	28
50	Clinical utility of the polygenic LDL-C SNP score in familial hypercholesterolemia. *Atherosclerosis*, **2018**, 277, 457-463	3.1	28
49	The importance of an integrated analysis of clinical, molecular, and functional data for the genetic diagnosis of familial hypercholesterolemia. *Genetics in Medicine*, **2015**, 17, 980-8	8.1	27
48	Mutational analysis of a cohort with clinical diagnosis of familial hypercholesterolemia: considerations for genetic diagnosis improvement. *Genetics in Medicine*, **2016**, 18, 316-24	8.1	27
47	Characterization of the first PCSK9 gain of function homozygote. *Journal of the American College of Cardiology*, **2015**, 66, 2152-2154	15.1	27
46	Low-density lipoprotein receptor mutational analysis in diagnosis of familial hypercholesterolemia. *Current Opinion in Lipidology*, **2017**, 28, 120-129	4.4	26
45	Variable phenotypic expression of homozygous familial hypobetalipoproteinaemia due to novel APOB gene mutations. *Clinical Genetics*, **2008**, 74, 267-73	4	21
44	A rare polymorphism in the low density lipoprotein (LDL) gene that affects mRNA splicing. *Atherosclerosis*, **2007**, 195, e17-20	3.1	20
43	In´vitro functional characterization of missense mutations in the LDLR gene. *Atherosclerosis*, **2012**, 225, 128-34	3.1	18
42	Comparison of the characteristics at diagnosis and treatment of children with heterozygous familial hypercholesterolaemia (FH) from eight European countries. *Atherosclerosis*, **2020**, 292, 178-187	3.1	17
41	Cardiovascular risk assessment of dyslipidemic children: analysis of biomarkers to identify monogenic dyslipidemia. *Journal of Lipid Research*, **2014**, 55, 947-55	6.3	16
40	The familial hypercholesterolaemia phenotype: Monogenic familial hypercholesterolaemia, polygenic hypercholesterolaemia and other causes. *Clinical Genetics*, **2020**, 97, 457-466	4	15
39	Clinical and molecular aspects of familial hypercholesterolemia in Ibero-American countries. *Journal of Clinical Lipidology*, **2017**, 11, 160-166	4.9	14
38	Hepatitis B and C prevalence in Portugal: disparity between the general population and high-risk groups. *European Journal of Gastroenterology and Hepatology*, **2016**, 28, 640-4	2.2	14
37	Global perspective of familial hypercholesterolaemia: a cross-sectional study from the EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC). *Lancet, The*, **2021**, 398, 1713-1725	40	14
36	Genetic variation at the CYP2C19 gene associated with metabolic syndrome susceptibility in a South Portuguese population: results from the pilot study of the European Health Examination Survey in Portugal. *Diabetology and Metabolic Syndrome*, **2014**, 6, 23	5.6	12
35	Further evidence of novel APOB mutations as a cause of familial hypercholesterolaemia. *Atherosclerosis*, **2018**, 277, 448-456	3.1	12

34	Portuguese Familial Hypercholesterolemia Study: presentation of the study and preliminary results. *Revista Portuguesa De Cardiologia*, **2006**, 25, 999-1013	1	10
33	Phenotypical, Clinical, and Molecular Aspects of Adults and Children With Homozygous Familial Hypercholesterolemia in Iberoamerica. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2020**, 40, 2508-2515	9.4	9
32	Preliminary spectrum of genetic variants in familial hypercholesterolemia in Argentina. *Journal of Clinical Lipidology*, **2017**, 11, 524-531	4.9	7
31	Immune cell changes in response to a swimming training session during a 24-h recovery period. *Applied Physiology, Nutrition and Metabolism*, **2016**, 41, 476-83	3	7
30	Brown Algae Potential as a Functional Food against Hypercholesterolemia: Review. *Foods*, **2021**, 10,	4.9	7
29	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification.. *Genetics in Medicine*, **2021**,	8.1	6
28	Hypercholesterolemia--a disease with expression from childhood. *Revista Portuguesa De Cardiologia* , **2013**, 32, 379-86	1	5
27	Cardiovascular risk profile of high school students: a cross-sectional study. *Revista Portuguesa De Cardiologia*, **2014**, 33, 525-34	1	5
26	Inheritance of two different alleles of the low-density lipoprotein (LDL)-receptor gene carrying the recurrent Pro664Leu mutation in a patient with homozygous familial hypercholesterolaemia. *Clinical Genetics*, **1999**, 56, 225-31	4	5
25	Comparison of the mutation spectrum and association with pre and post treatment lipid measures of children with heterozygous familial hypercholesterolaemia (FH) from eight European countries. *Atherosclerosis*, **2021**, 319, 108-117	3.1	5
24	Pharmacogenetic profile of a South Portuguese population: results from the pilot study of the European Health Examination Survey in Portugal. *Public Health Genomics*, **2015**, 18, 139-50	1.9	4
23	Hypercysteinemia, A Potential Risk Factor for Central Obesity and Related Disorders in Azores, Portugal. *Journal of Nutrition and Metabolism*, **2019**, 2019, 1826780	2.7	4
22	Sortilin and the risk of cardiovascular disease. *Revista Portuguesa De Cardiologia*, **2013**, 32, 793-9	1	4
21	Worldwide experience of homozygous familial hypercholesterolaemia: retrospective cohort study.. *Lancet, The*, **2022**,	40	4
20	Prevalence and risk factors of fatty liver in Portuguese adults. *European Journal of Clinical Investigation*, **2020**, 50, e13235	4.6	4
19	Hypercholesterolemia A disease with expression since childhood. *Revista Portuguesa De Cardiologia (English Edition)*, **2013**, 32, 379-386	0	3
18	No Evidence for Lower Levels of Serum Vitamin D in the Presence of Hepatic Steatosis. A Study on the Portuguese General Population. *International Journal of Medical Sciences*, **2018**, 15, 1778-1786	3.7	3
17	Applicability of the low-density lipoprotein cholesterol gene score in a South European population. *Atherosclerosis*, **2017**, 263, e99-e100	3.1	2

16	«Papa Bem»: investir na literacia em saúde para a prevenção da obesidade infantil. *Revista Portuguesa De Saude Publica*, **2015**, 33, 12-23		2
15	Familial hypercholesterolemia: Molecular characterization of possible cases from the Azores Islands (Portugal). *Meta Gene*, **2014**, 2, 638-45	0.7	2
14	What Is the Role of the New Index Relative Fat Mass (RFM) in the Assessment of Nonalcoholic Fatty Liver Disease (NAFLD)?. *Obesity Surgery*, **2020**, 30, 560-568	3.7	2
13	LDLR variants functional characterization: Contribution to variant classification. *Atherosclerosis*, **2021**, 329, 14-21	3.1	2
12	Frequency and clinical and molecular aspects of familial hypercholesterolemia in an endocrinology unit in Ciudad Bolívar, Venezuela. *Endocrinologia, Diabetes Y Nutrición*, **2017**, 64, 432-439	1.3	1
11	The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification		1
10	Pharmacogenomics of statins and familial hypercholesterolemia. *Current Opinion in Lipidology*, **2021**, 32, 96-102	4.4	1
9	Machine learning modelling of blood lipid biomarkers in familial hypercholesterolaemia versus polygenic/environmental dyslipidaemia. *Scientific Reports*, **2021**, 11, 3801	4.9	1
8	Applicability of Martin-Hopkins formula and comparison with Friedewald formula for estimated low-density lipoprotein cholesterol in e_COR study population. *Revista Portuguesa De Cardiologia (English Edition)*, **2021**, 40, 715-724	0	1
7	Characterization of Two Variants at Met 1 of the Human Gene Encoding the Same Amino Acid but Causing Different Functional Phenotypes. *Biomedicines*, **2021**, 9,	4.8	1
6	Recommendations for genetic testing in cardiology: Review of major international guidelines. *Revista Portuguesa De Cardiologia*, **2020**, 39, 597-610	1	0
5	Performance comparison of different classification algorithms applied to the diagnosis of familial hypercholesterolemia in paediatric subjects.. *Scientific Reports*, **2022**, 12, 1164	4.9	0
4	Polygenic contribution for familial hypercholesterolemia (FH). *Current Opinion in Lipidology*, **2021**, 32, 392-395	4.4	0
3	Familial hypercholesterolemia **2021**, 323-348		0
2	Frequency and clinical and molecular aspects of familial hypercholesterolemia in an endocrinology unit in Ciudad Bolívar, Venezuela. *Endocrinología Diabetes Y Nutrición (English Ed.)*, **2017**, 64, 432-439	0.1	
1	Recommendations for genetic testing in cardiology: Review of major international guidelines. *Revista Portuguesa De Cardiologia (English Edition)*, **2020**, 39, 597-610	0	