Isabel De Castro-OrÃ³s

List of Publications by Year
in descending order

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26 papers	508 citations	687363 13 h-index	677142 22 g-index
30 all docs	30 docs citations	30 times ranked	924 citing authors

#	Article	IF	Citations
1	The p.Leu167del Mutation in APOE Gene Causes Autosomal Dominant Hypercholesterolemia by Down-regulation of LDL Receptor Expression in Hepatocytes. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2113-2121.	3.6	71
2	Apolipoprotein E gene mutations in subjects with mixed hyperlipidemia and a clinical diagnosis of familial combined hyperlipidemia. Atherosclerosis, 2012, 222, 449-455.	0.8	61
3	Promoter variant â^'204AÂ>ÂC of the cholesterol 7Î±-hydroxylase gene: Association with response to plant sterols in humans and increased transcriptional activity in transfected HepG2 cells. Clinical Nutrition, 2011, 30, 239-246.	5.0	50
4	The genetic basis of familial hypercholesterolemia: inheritance, linkage, and mutations. The Application of Clinical Genetics, 2010, 3, 53.	3.0	49
5	Common Genetic Variants Contribute to Primary Hypertriglyceridemia Without Differences Between Familial Combined Hyperlipidemia and Isolated Hypertriglyceridemia. Circulation: Cardiovascular Genetics, 2014, 7, 814-821.	5.1	36
6	APOA5 variants predispose hyperlipidemic patients to atherogenic dyslipidemia and subclinical atherosclerosis. Atherosclerosis, 2015, 240, 98-104.	0.8	28
7	Functional analysis of LDLR promoter and 5â€² UTR mutations in subjects with clinical diagnosis of familial hypercholesterolemia. Human Mutation, 2011, 32, 868-872.	2.5	26
8	Genetic Variants of LDLR and PCSK9 Associated with Variations in Response to Antihypercholesterolemic Effects of Armolipid Plus with Berberine. PLoS ONE, 2016, 11, e0150785.	2.5	21
9	Frequency of rare mutations and common genetic variations in severe hypertriglyceridemia in the general population of Spain. Lipids in Health and Disease, 2016, 15, 82.	3.0	20
10	Assessment of plasma chitotriosidase activity, CCL18/PARC concentration and NP-C suspicion index in the diagnosis of Niemann-Pick disease type C: a prospective observational study. Journal of Translational Medicine, 2017, 15, 43.	4.4	19
11	Cosegregation of serum cholesterol with cholesterol intestinal absorption markers in families with primary hypercholesterolemia without mutations in LDLR, APOB, PCSK9 and APOE genes. Atherosclerosis, 2016, 246, 202-207.	0.8	15
12	A genetic variant in the LDLRpromoter is responsible for part of the LDL-cholesterol variability in primary hypercholesterolemia. BMC Medical Genomics, 2014, 7, 17.	1.5	14
13	Rare genetic variants with large effect on triglycerides in subjects with a clinical diagnosis of familial vs nonfamilial hypertriglyceridemia. Journal of Clinical Lipidology, 2016, 10, 790-797.	1.5	13
14	Bile acid synthesis precursors in familial combined hyperlipidemia: The oxysterols 24S-hydroxycholesterol and 27-hydroxycholesterol. Biochemical and Biophysical Research Communications, 2014, 446, 731-735.	2.1	12
15	LC-MS/MS analysis of plasma glucosylsphingosine as a biomarker for diagnosis and follow-up monitoring in Gaucher disease in the Spanish population. Clinical Chemistry and Laboratory Medicine, 2020, 58, 798-809.	2.3	12
16	Variantes de un solo nucleÃ³tido asociadas con la hipercolesterolemia poligÃ©nica en familias diagnosticadas de hipercolesterolemia familiar. Revista Espanola De Cardiologia, 2018, 71, 351-356.	1.2	10
17	Functional analysis of new 3â€² untranslated regions genetic variants in genes associated with genetic hypercholesterolemias. Journal of Clinical Lipidology, 2017, 11, 532-542.	1.5	9
18	Haplotype analyses, mechanism and evolution of common double mutants in the human LDL receptor gene. Molecular Genetics and Genomics, 2010, 283, 565-574.	2.1	7

#	Article	IF	Citations
19	Effect of different fat-enriched meats on non-cholesterol sterols and oxysterols as markers of cholesterol metabolism: Results of a randomized and cross-over clinical trial. Nutrition, Metabolism and Cardiovascular Diseases, 2015, 25, 853-859.	2.6	7
20	The fine line between familial and polygenic hypercholesterolemia. Clinical Lipidology, 2013, 8, 303-306.	0.4	6
21	New contributions to the study of common double mutants in the human LDL receptor gene. Die Naturwissenschaften, 2011, 98, 943-949.	1.6	5
22	Single Nucleotide Variants Associated With Polygenic Hypercholesterolemia in Families Diagnosed Clinically With Familial Hypercholesterolemia. Revista Espanola De Cardiologia (English Ed), 2018, 71, 351-356.	0.6	3
23	AnÃ¡lisis funcional de mutaciones en el promotor del LDLR y su relaciÃ³n con la hipercolesterolemia familiar. ClÃnica E InvestigaciÃ³n En Arteriosclerosis, 2011, 23, 119-124.	0.8	1
24	Serum plant sterols as surrogate markers of dietary compliance inÂfamilial dyslipidemias. Clinical Nutrition, 2015, 34, 490-495.	5.0	1
25	Searching new loci associated to autosomal dominant hypercholesterolemia. Atherosclerosis, 2014, 235, e54-e55.	0.8	0
26	Identifying variants in candidate genes for primary hypertriglyceridemia. Atherosclerosis, 2014, 235, e56.	0.8	0