

Isabel De Castro-OrÃ³s

List of Publications by Year in descending order

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26
papers

508
citations

686830

13
h-index

676716

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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 2113-2121.	1.8	71
2	Apolipoprotein E gene mutations in subjects with mixed hyperlipidemia and a clinical diagnosis of familial combined hyperlipidemia. <i>Atherosclerosis</i> , 2012, 222, 449-455.	0.4	61
3	Promoter variant $\hat{\sim}204A\hat{\>}\hat{A}C$ of the cholesterol $7\hat{\pm}$ -hydroxylase gene: Association with response to plant sterols in humans and increased transcriptional activity in transfected HepG2 cells. <i>Clinical Nutrition</i> , 2011, 30, 239-246.	2.3	50
4	The genetic basis of familial hypercholesterolemia: inheritance, linkage, and mutations. <i>The Application of Clinical Genetics</i> , 2010, 3, 53.	1.4	49
5	Common Genetic Variants Contribute to Primary Hypertriglyceridemia Without Differences Between Familial Combined Hyperlipidemia and Isolated Hypertriglyceridemia. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 814-821.	5.1	36
6	APOA5 variants predispose hyperlipidemic patients to atherogenic dyslipidemia and subclinical atherosclerosis. <i>Atherosclerosis</i> , 2015, 240, 98-104.	0.4	28
7	Functional analysis of LDLR promoter and 5 $\hat{\epsilon}^2$ UTR mutations in subjects with clinical diagnosis of familial hypercholesterolemia. <i>Human Mutation</i> , 2011, 32, 868-872.	1.1	26
8	Genetic Variants of LDLR and PCSK9 Associated with Variations in Response to Antihypercholesterolemic Effects of Armolipid Plus with Berberine. <i>PLoS ONE</i> , 2016, 11, e0150785.	1.1	21
9	Frequency of rare mutations and common genetic variations in severe hypertriglyceridemia in the general population of Spain. <i>Lipids in Health and Disease</i> , 2016, 15, 82.	1.2	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. <i>Journal of Translational Medicine</i> , 2017, 15, 43.	1.8	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. <i>Atherosclerosis</i> , 2016, 246, 202-207.	0.4	15
12	A genetic variant in the LDLR promoter is responsible for part of the LDL-cholesterol variability in primary hypercholesterolemia. <i>BMC Medical Genomics</i> , 2014, 7, 17.	0.7	14
13	Rare genetic variants with large effect on triglycerides in subjects with a clinical diagnosis of familial vs nonfamilial hypertriglyceridemia. <i>Journal of Clinical Lipidology</i> , 2016, 10, 790-797.	0.6	13
14	Bile acid synthesis precursors in familial combined hyperlipidemia: The oxysterols 24S-hydroxycholesterol and 27-hydroxycholesterol. <i>Biochemical and Biophysical Research Communications</i> , 2014, 446, 731-735.	1.0	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. <i>Clinical Chemistry and Laboratory Medicine</i> , 2020, 58, 798-809.	1.4	12
16	Variantes de un solo nucleÃ3tido asociadas con la hipercolesterolemia poligÃ©nica en familias diagnosticadas de hipercolesterolemia familiar. <i>Revista Espanola De Cardiologia</i> , 2018, 71, 351-356.	0.6	10
17	Functional analysis of new $3\hat{\epsilon}^2$ untranslated regions genetic variants in genes associated with genetic hypercholesterolemias. <i>Journal of Clinical Lipidology</i> , 2017, 11, 532-542.	0.6	9
18	Haplotype analyses, mechanism and evolution of common double mutants in the human LDL receptor gene. <i>Molecular Genetics and Genomics</i> , 2010, 283, 565-574.	1.0	7

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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. <i>Nutrition, Metabolism and Cardiovascular Diseases</i> , 2015, 25, 853-859.	1.1	7
20	The fine line between familial and polygenic hypercholesterolemia. <i>Clinical Lipidology</i> , 2013, 8, 303-306.	0.4	6
21	New contributions to the study of common double mutants in the human LDL receptor gene. <i>Die Naturwissenschaften</i> , 2011, 98, 943-949.	0.6	5
22	Single Nucleotide Variants Associated With Polygenic Hypercholesterolemia in Families Diagnosed Clinically With Familial Hypercholesterolemia. <i>Revista Espanola De Cardiologia (English Ed)</i> , 2018, 71, 351-356.	0.4	3
23	Análisis funcional de mutaciones en el promotor del LDLR y su relación con la hipercolesterolemia familiar. <i>Clínica E Investigación En Arteriosclerosis</i> , 2011, 23, 119-124.	0.4	1
24	Serum plant sterols as surrogate markers of dietary compliance in familial dyslipidemias. <i>Clinical Nutrition</i> , 2015, 34, 490-495.	2.3	1
25	Searching new loci associated to autosomal dominant hypercholesterolemia. <i>Atherosclerosis</i> , 2014, 235, e54-e55.	0.4	0
26	Identifying variants in candidate genes for primary hypertriglyceridemia. <i>Atherosclerosis</i> , 2014, 235, e56.	0.4	0