

Jacques Rochette

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/10736743/publications.pdf>

Version: 2024-02-01

21
papers

1,914
citations

687363

13
h-index

642732

23
g-index

25
all docs

25
docs citations

25
times ranked

1806
citing authors

#	ARTICLE	IF	CITATIONS
1	Stormorken syndrome or York platelet syndrome: A clinician's dilemma. <i>Molecular Genetics and Metabolism Reports</i> , 2015, 2, 80.	1.1	7
2	Gain of Function Mutation in STIM1 (P.R304W) Is Associated with Stormorken Syndrome. <i>Human Mutation</i> , 2014, 35, 1221-1232.	2.5	101
3	Phenotypic expression of a novel C282Y/R226G compound heterozygous state in HFE hemochromatosis: Molecular dynamics and biochemical studies. <i>Blood Cells, Molecules, and Diseases</i> , 2014, 52, 27-34.	1.4	10
4	<i>PRRT2</i> links infantile convulsions and paroxysmal dyskinesia with migraine. <i>Neurology</i> , 2012, 79, 2097-2103.	1.1	90
5	Mutations in the Gene <i>PRRT2</i> Cause Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions. <i>Cell Reports</i> , 2012, 1, 2-12.	6.4	250
6	Iron overload in HFE C282Y heterozygotes at first genetic testing: a strategy for identifying rare HFE variants. <i>Haematologica</i> , 2011, 96, 507-514.	3.5	41
7	Novel familial cases of ICCA (infantile convulsions with paroxysmal choreoathetosis) syndrome. <i>Epileptic Disorders</i> , 2010, 12, 199-204.	1.3	12
8	Infantile Convulsions with Paroxysmal Dyskinesia (ICCA Syndrome) and Copy Number Variation at Human Chromosome 16p11. <i>PLoS ONE</i> , 2010, 5, e13750.	2.5	16
9	A novel HFE mutation (c.del478) results in nonsense-mediated decay of the mutant transcript in a hemochromatosis patient. <i>Blood Cells, Molecules, and Diseases</i> , 2009, 43, 194-198.	1.4	9
10	First Identification of a Point Mutation at Position -83 (G>A) of the β^2 -Globin Gene Promoter. <i>Hemoglobin</i> , 2009, 33, 274-278.	0.8	7
11	A Novel β^2 -Thalassemic Allele Due to a Two Nucleotide Deletion: β^276 (\sim GC). <i>Hemoglobin</i> , 2007, 31, 31-37.	0.8	6
12	Novel mutation in ferroportin 1 gene is associated with autosomal dominant iron overload. <i>Journal of Hepatology</i> , 2003, 39, 286-289.	3.7	79
13	Digenic inheritance of mutations in HAMP and HFE results in different types of haemochromatosis. <i>Human Molecular Genetics</i> , 2003, 12, 2241-2247.	2.9	227
14	Linkage of Benign Familial Infantile Convulsions to Chromosome 16p12-q12 Suggests Allelism to the Infantile Convulsions and Choreoathetosis Syndrome. <i>American Journal of Human Genetics</i> , 2001, 68, 788-794.	6.2	125
15	An Integrated Map of Human 6q22.3-q24 Including a 3-Mb High-Resolution BAC/PAC Contig Encompassing a QTL for Fetal Hemoglobin. <i>Genomics</i> , 2000, 64, 264-276.	2.9	12
16	Geography of HFE C282Y and H63D Mutations. <i>Genetic Testing and Molecular Biomarkers</i> , 2000, 4, 183-198.	1.7	265
17	Polymorphism in intron 4 of HFE does not compromise haemochromatosis mutation results. <i>Nature Genetics</i> , 1999, 23, 271-271.	21.4	38
18	Familial Infantile Convulsions and Paroxysmal Choreoathetosis: A New Neurological Syndrome Linked to the Pericentromeric Region of Human Chromosome 16. <i>American Journal of Human Genetics</i> , 1997, 61, 889-898.	6.2	294

#	ARTICLE	IF	CITATIONS
19	Dissecting the loci controlling fetal haemoglobin production on chromosomes 11p and 6q by the regressive approach. <i>Nature Genetics</i> , 1996, 12, 58-64.	21.4	181
20	Rapid analysis of α^+ ^{3.7} thalassaemia and β^+ ^{anti 3.7} triplication by enzymatic amplification analysis. <i>British Journal of Haematology</i> , 1993, 83, 105-111.	2.5	120
21	Level and composition of fetal hemoglobin expression in normal newborn babies are not dependent on β^2 cluster DNA haplotype. <i>American Journal of Hematology</i> , 1990, 34, 223-224.	4.1	5