Jacques Rochette

List of Publications by Year
in descending order

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	Familial Infantile Convulsions and Paroxysmal Choreoathetosis: A New Neurological Syndrome Linked to the Pericentromeric Region of Human Chromosome 16. American Journal of Human Genetics, 1997, 61, 889-898.	6.2	294
2	Geography ofHFEC282Y and H63D Mutations. Genetic Testing and Molecular Biomarkers, 2000, 4, 183-198.	1.7	265
3	Mutations in the Gene PRRT2 Cause Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions. Cell Reports, 2012, 1, 2-12.	6.4	250
4	Digenic inheritance of mutations in HAMP and HFE results in different types of haemochromatosis. Human Molecular Genetics, 2003, 12, 2241-2247.	2.9	227
5	Dissecting the loci controlling fetal haemoglobin production on chromosomes 11p and 6q by the regressive approach. Nature Genetics, 1996, 12, 58-64.	21.4	181
6	Linkage of Benign Familial Infantile Convulsions to Chromosome 16p12-q12 Suggests Allelism to the Infantile Convulsions and Choreoathetosis Syndrome. American Journal of Human Genetics, 2001, 68, 788-794.	6.2	125
7	Rapid analysis of â€Î±^{3.7} thalassaemia and Î±Î±Î±^{anti 3.7} triplication by enzymatic amplification analysis. British Journal of Haematology, 1993, 83, 105-111.	2.5	120
8	Gainâ€ofâ€Function Mutation in STIM1 (P.R304W) Is Associated with Stormorken Syndrome. Human Mutation, 2014, 35, 1221-1232.	2.5	101
9	PRRT2 links infantile convulsions and paroxysmal dyskinesia with migraine. Neurology, 2012, 79, 2097-2103.	1.1	90
10	Novel mutation in ferroportin 1 gene is associated with autosomal dominant iron overload. Journal of Hepatology, 2003, 39, 286-289.	3.7	79
11	Iron overload in HFE C282Y heterozygotes at first genetic testing: a strategy for identifying rare HFE variants. Haematologica, 2011, 96, 507-514.	3.5	41
12	Polymorphism in intron 4 of HFE does not compromise haemochromatosis mutation results. Nature Genetics, 1999, 23, 271-271.	21.4	38
13	Infantile Convulsions with Paroxysmal Dyskinesia (ICCA Syndrome) and Copy Number Variation at Human Chromosome 16p11. PLoS ONE, 2010, 5, e13750.	2.5	16
14	An Integrated Map of Human 6q22.3â€“q24 Including a 3-Mb High-Resolution BAC/PAC Contig Encompassing a QTL for Fetal Hemoglobin. Genomics, 2000, 64, 264-276.	2.9	12
15	Novel familial cases of ICCA (infantile convulsions with paroxysmal choreoathetosis) syndrome. Epileptic Disorders, 2010, 12, 199-204.	1.3	12
16	Phenotypic expression of a novel C282Y/R226G compound heterozygous state in HFE hemochromatosis: Molecular dynamics and biochemical studies. Blood Cells, Molecules, and Diseases, 2014, 52, 27-34.	1.4	10
17	A novel HFE mutation (c.del478) results in nonsense-mediated decay of the mutant transcript in a hemochromatosis patient. Blood Cells, Molecules, and Diseases, 2009, 43, 194-198.	1.4	9
18	First Identification of a Point Mutation at Position â^'83 (G>A) of the Î²-Globin Gene Promoter. Hemoglobin, 2009, 33, 274-278.	0.8	7

#	Article	IF	Citations
19	Stormorken syndrome or York platelet syndrome: A clinician's dilemma. Molecular Genetics and Metabolism Reports, 2015, 2, 80.	1.1	7
20	A Novel Î²-Thalassemic Allele Due to a Two Nucleotide Deletion: Î²76 (âˆ’GC). Hemoglobin, 2007, 31, 31-37.	0.8	6
21	Level and composition of fetal hemoglobin expression in normal newborn babies are not dependent on Î² cluster DNA haplotype. American Journal of Hematology, 1990, 34, 223-224.	4.1	5