Gerald Le Gac

List of Publications by Year
in descending order

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23	561	758635 12	642321 23
papers	citations	h-index	g-index

26	26	26	859
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prevalence of HFEâ€related haemochromatosis and secondary causes of hyperferritinaemia and their association with iron overload in 1059 French patients treated by venesection. Alimentary Pharmacology and Therapeutics, 2022, 55, 1016-1027.	1.9	3
2	Splicing analysis of SLC40A1 missense variations and contribution to hemochromatosis type 4 phenotypes. Blood Cells, Molecules, and Diseases, 2021, 87, 102527.	0.6	5
3	Missense RHD single nucleotide variants induce weakened D antigen expression by altering splicing and/or protein expression. Transfusion, 2021, 61, 2468-2476.	0.8	1
4	A novel hypomorphic splice variant in EIF2B5 gene is associated with mild ovarioleukodystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 1574-1579.	1.7	3
5	Assessment of branch point prediction tools to predict physiological branch points and their alteration by variants. BMC Genomics, 2020, 21, 86.	1.2	33
6	Molecular model of the ferroportin intracellular gate and implications for the human iron transport cycle and hemochromatosis type 4A. FASEB Journal, 2019, 33, 14625-14635.	0.2	11
7	Functional analysis of novel RHD variants: splicing disruption is likely to be a common mechanism of variant D phenotype. Transfusion, 2019, 59, 1367-1375.	0.8	12
8	The SLC40A1 R178Q mutation is a recurrent cause of hemochromatosis and is associated with a novel pathogenic mechanism. Haematologica, 2018, 103, 1796-1805.	1.7	19
9	Novel diagnostic tool for prediction of variant spliceogenicity derived from a set of 395 combined in silico/in vitro studies: an international collaborative effort. Nucleic Acids Research, 2018, 46, 7913-7923.	6.5	71
10	Diagnostic value of targeted nextâ€generation sequencing in suspected hemochromatosis patients with a single copy of the HFE p.Cys282Tyr causative allele. American Journal of Hematology, 2017, 92, E664-E666.	2.0	4
11	The p.Leu96Pro Missense Mutation in the BMP6 Gene Is Repeatedly Associated With Hyperferritinemia in Patients ofÂFrench Origin. Gastroenterology, 2016, 151, 769-770.	0.6	11
12	Mutational status of synchronous and metachronous tumor samples in patients with metastatic non-small-cell lung cancer. BMC Cancer, 2016, 16, 210.	1.1	26
13	Heterozygous Mutations in BMP6 Pro-peptide Lead to Inappropriate Hepcidin Synthesis and Moderate Iron Overload in Humans. Gastroenterology, 2016, 150, 672-683.e4.	0.6	73
14	Extensive functional analyses of RHD splice site variants: Insights into the potential role of splicing in the physiology of Rh. Transfusion, 2015, 55, 1432-1443.	0.8	25
15	Characterization of the second <scp>HFE</scp> gross deletion highlights the potential importance of Aluâ€mediated recombination in haemochromatosis. British Journal of Haematology, 2015, 168, 759-762.	1.2	0
16	Comprehensive functional annotation of 18 missense mutations found in suspected hemochromatosis type 4 patients. Human Molecular Genetics, 2014, 23, 4479-4490.	1.4	46
17	Establishment of a mediumâ€throughput approach for the genotyping of <scp>RHD</scp> variants and report of nine novel rare alleles. Transfusion, 2013, 53, 1821-1828.	0.8	39
18	Homozygous deletion of HFE produces a phenotype similar to the HFE p.C282Y/p.C282Y genotype. Blood, 2008, 112, 5238-5240.	0.6	16

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19	The molecular genetics of haemochromatosis. European Journal of Human Genetics, 2005, 13, 1172-1185.	1.4	91
20	The Q283P amino-acid change in HFE leads to structural and functional consequences similar to those described for the mutated 282Y HFE protein. Human Genetics, 2005, 117, 467-475.	1.8	14
21	Impact of HFEgenetic testing on clinical presentation of hereditary hemochromatosis: new epidemiological data. BMC Medical Genetics, 2005, 6, 24.	2.1	15
22	Complete Scanning of the Hereditary Hemochromatosis Gene (HFE) by Use of Denaturing HPLC. Clinical Chemistry, 2001, 47, 1633-1640.	1.5	31
23	Nramp2 Analysis in Hemochromatosis Probands. Blood Cells, Molecules, and Diseases, 2000, 26, 312-319.	0.6	1