Jean-Charles Deybach

List of Publications by Year
in descending order

70 papers	4,747 citations	101543 36 h-index	102487 66 g-index
71 all docs	71 docs citations	71 times ranked	2945 citing authors

#	Article	IF	Citations
1	Phlebotomy as an efficient long-term treatment of congenital erythropoietic porphyria. Haematologica, 2021, 106, 913-917.	3.5	13
2	EXPLORE: A Prospective, Multinational, Natural History Study of Patients with Acute Hepatic Porphyria with Recurrent Attacks. Hepatology, 2020, 71, 1546-1558.	7.3	103
3	Erythroid-Progenitor-Targeted Gene Therapy Using Bifunctional TFR1 Ligand-Peptides in Human Erythropoietic Protoporphyria. American Journal of Human Genetics, 2019, 104, 341-347.	6.2	22
4	International Porphyria Molecular Diagnostic Collaborative: an evidence-based database of verified pathogenic and benign variants for the porphyrias. Genetics in Medicine, 2019, 21, 2605-2613.	2.4	16
5	Current and innovative emerging therapies for porphyrias with hepatic involvement. Journal of Hepatology, 2019, 71, 422-433.	3.7	24
6	Hepatocellular carcinoma in acute hepatic porphyrias: A Damocles Sword. Molecular Genetics and Metabolism, 2019, 128, 236-241.	1.1	32
7	Systemic Administered mRNA as Therapy for Metabolic Diseases. Trends in Molecular Medicine, 2019, 25, 3-5.	6.7	4
8	From a dominant to an oligogenic model of inheritance with environmental modifiers in acute intermittent porphyria. Human Molecular Genetics, 2018, 27, 1164-1173.	2.9	73
9	Red cells from ferrochelatase-deficient erythropoietic protoporphyria patients are resistant to growth of malarial parasites. Blood, 2015, 125, 534-541.	1.4	37
10	Afamelanotide for Erythropoietic Protoporphyria. New England Journal of Medicine, 2015, 373, 48-59.	27.0	206
11	Porphyrias: A 2015 update. Clinics and Research in Hepatology and Gastroenterology, 2015, 39, 412-425.	1.5	132
12	High prevalence of and potential mechanisms for chronic kidney disease in patients with acute intermittent porphyria. Kidney International, 2015, 88, 386-395.	5.2	84
13	Mitochondrial energetic defects in muscle and brain of aHmbs^{â˜'/â˜'}mouse model of acute intermittent porphyria. Human Molecular Genetics, 2015, 24, 5015-5023.	2.9	34
14	Human Erythroid 5-Aminolevulinate Synthase Mutations Associated with X-Linked Protoporphyria Disrupt the Conformational Equilibrium and Enhance Product Release. Biochemistry, 2015, 54, 5617-5631.	2.5	18
15	Antisense Oligonucleotide-Based Therapy in Human Erythropoietic Protoporphyria. American Journal of Human Genetics, 2014, 94, 611-617.	6.2	34
16	Acute intermittent porphyria causes hepatic mitochondrial energetic failure in a mouse model. International Journal of Biochemistry and Cell Biology, 2014, 51, 93-101.	2.8	51
17	Les porphyries hÃ©rÃ©ditaires : anomalies du mÃ©tabolisme de lâ€™hÃ¨me. Bulletin De L'Academie Nationale De Medecine, 2014, 198, 1069-1093.	0.0	0
18	Epistasis in iron metabolism: complex interactions between Cp, Mon1a, and Slc40a1 loci and tissue iron in mice. Mammalian Genome, 2013, 24, 427-438.	2.2	0

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19	The incidence of inherited porphyrias in Europe. Journal of Inherited Metabolic Disease, 2013, 36, 849-857.	3.6	220
20	Heme Biosynthesis and Pathophysiology of Porphyrias. Handbook of Porphyrin Science, 2013, , 89-118.	0.8	2
21	Molecular and functional analysis of the C-terminal region of human erythroid-specific 5-aminolevulinic synthase associated with X-linked dominant protoporphyria (XLDPP). Human Molecular Genetics, 2013, 22, 1280-1288.	2.9	39
22	Vemurafenib: an unusual <scp>UVA</scp>â€induced photosensitivity. Experimental Dermatology, 2013, 22, 297-298.	2.9	76
23	ABCB6 is dispensable for erythropoiesis and specifies the new blood group system Langereis. Nature Genetics, 2012, 44, 170-173.	21.4	127
24	An Uncommon Option for Surviving Bariatric Surgery: Regaining Weight!. American Journal of Medicine, 2012, 125, e1-e2.	1.5	7
25	Establishing a network of specialist Porphyria centres - effects on diagnostic activities and services. Orphanet Journal of Rare Diseases, 2012, 7, 93.	2.7	12
26	Protoporphyrin Retention in Hepatocytes and Kupffer Cells Prevents Sclerosing Cholangitis in Erythropoietic Protoporphyria Mouse Model. Gastroenterology, 2011, 141, 1509-1519.e3.	1.3	39
27	ALAS2 acts as a modifier gene in patients with congenital erythropoietic porphyria. Blood, 2011, 118, 1443-1451.	1.4	80
28	Sideroblastic anemia: molecular analysis of the ALAS2 gene in a series of 29 probands and functional studies of 10 missense mutations. Human Mutation, 2011, 32, 590-597.	2.5	56
29	European Specialist Porphyria Laboratories: Diagnostic Strategies, Analytical Quality, Clinical Interpretation, and Reporting As Assessed by an External Quality Assurance Program. Clinical Chemistry, 2011, 57, 1514-1523.	3.2	38
30	A genetic schizophrenia-susceptibility region located between the ANKK1 and DRD2 genes. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2010, 34, 492-499.	4.8	39
31	Porphyrias. Lancet, The, 2010, 375, 924-937.	13.7	644
32	Erythropoietic protoporphyria. Orphanet Journal of Rare Diseases, 2009, 4, 19.	2.7	178
33	Chediak-Steinbrinck-Higashi Syndrome. , 2009, , 314-314.		0
34	C-Terminal Deletions in the ALAS2 Gene Lead to Gain of Function and Cause X-linked Dominant Protoporphyria without Anemia or Iron Overload. American Journal of Human Genetics, 2008, 83, 408-414.	6.2	246
35	Increased plasma transferrin, altered body iron distribution, and microcytic hypochromic anemia in ferrochelatase-deficient mice. Blood, 2007, 109, 811-818.	1.4	58
36	Genetic Study of Variation in Normal Mouse Iron Homeostasis Reveals Ceruloplasmin as an HFE-Hemochromatosis Modifier Gene. Gastroenterology, 2007, 132, 679-686.	1.3	26

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37	Contribution of a Common Single-Nucleotide Polymorphism to the Genetic Predisposition for Erythropoietic Protoporphyria. American Journal of Human Genetics, 2006, 78, 2-14.	6.2	164
38	A mouse model provides evidence that genetic background modulates anemia and liver injury in erythropoietic protoporphyria. American Journal of Physiology - Renal Physiology, 2005, 288, G1208-G1216.	3.4	32
39	Mutations in human CPO gene predict clinical expression of either hepatic hereditary coproporphyria or erythropoietic harderoporphyria. Human Molecular Genetics, 2005, 14, 3089-3098.	2.9	44
40	Modulation of penetrance by the wild-type allele in dominantly inherited erythropoietic protoporphyria and acute hepatic porphyrias. Human Genetics, 2004, 114, 256-262.	3.8	40
41	Recovery from a variegate porphyria by a liver transplantation. Liver Transplantation, 2004, 10, 935-938.	2.4	39
42	The 3â€² region of the DRD2 gene is involved in genetic susceptibility to schizophrenia. Schizophrenia Research, 2004, 67, 75-85.	2.0	115
43	Acute Intermittent Porphyria: From Clinical to Molecular Aspects. , 2003, , 23-41.		7
44	Ancestral Founder of Mutation W283X in the Porphobilinogen Deaminase Gene among Acute Intermittent Porphyria Patients. Human Heredity, 2002, 54, 69-81.	0.8	17
45	The penetrance of dominant erythropoietic protoporphyria is modulated by expression of wildtype FECH. Nature Genetics, 2002, 30, 27-28.	21.4	237
46	Characterization of Mutations in the CPO Gene in British Patients Demonstrates Absence of Genotype-Phenotype Correlation and Identifies Relationship between Hereditary Coproporphyria and Harderoporphyria. American Journal of Human Genetics, 2001, 68, 1130-1138.	6.2	62
47	Influence of Age and Gender on the Clinical Expression of Acute Intermittent Porphyria Based on Molecular Study of Porphobilinogen Deaminase Gene Among Swiss Patients. Molecular Medicine, 2001, 7, 535-542.	4.4	39
48	New Missense Mutation in the Human Ferrochelatase Gene in a Family with Erythropoietic Protoporphyria: Functional Studies and Correlation of Genotype and Phenotype. Clinical Chemistry, 2001, 47, 1112-1113.	3.2	7
49	Haplotype Analysis in Determination of the Heredity of Erythropoietic Protoporphyria among Swiss Families. Journal of Investigative Dermatology, 2001, 117, 1521-1525.	0.7	10
50	Identification of a Prevalent Nonsense Mutation (W283X) and Two Novel Mutations in the Porphobilinogen Deaminase Gene of Swiss Patients with Acute Intermittent Porphyria. Human Heredity, 2000, 50, 247-250.	0.8	23
51	New insights into the pathogenesis of erythropoietic protoporphyria and their impact on patient care. European Journal of Pediatrics, 2000, 159, 719-725.	2.7	61
52	Mutations in the iron-sulfur cluster ligands of the human ferrochelatase lead to erythropoietic protoporphyria. Blood, 2000, 96, 1545-1549.	1.4	34
53	Hepatocellular carcinoma in patients with acute hepatic porphyria: frequency of occurrence and related factors. Journal of Hepatology, 2000, 32, 933-939.	3.7	124
54	Mutations in the iron-sulfur cluster ligands of the human ferrochelatase lead to erythropoietic protoporphyria. Blood, 2000, 96, 1545-1549.	1.4	1

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55	Inheritance in Erythropoietic Protoporphyria: A Common Wild-Type Ferrochelatase Allelic Variant With Low Expression Accounts for Clinical Manifestation. Blood, 1999, 93, 2105-2110.	1.4	140
56	Variegate Porphyria in Western Europe: Identification of PPOX Gene Mutations in 104 Families, Extent of Allelic Heterogeneity, and Absence of Correlation between Phenotype and Type of Mutation. American Journal of Human Genetics, 1999, 65, 984-994.	6.2	100
57	Epidemiology of hepatitis C and G in sporadic and familial porphyria cutanea tarda. Hepatology, 1998, 27, 848-852.	7.3	49
58	Mutations in the Ferrochelatase Gene of Four Spanish Patients with Erythropoietic Protoporphyria. Journal of Investigative Dermatology, 1998, 111, 406-409.	0.7	11
59	Molecular Characterization of Homozygous Variegate Porphyria. Human Molecular Genetics, 1998, 7, 1921-1925.	2.9	37
60	Acute Hepatic Porphyrias and Primary Liver Cancer. New England Journal of Medicine, 1998, 338, 1853-1854.	27.0	33
61	5-Aminolevulinic acid dehydratase deficiency porphyria: a twenty-year clinical and biochemical follow-up. Clinical Chemistry, 1998, 44, 1892-1896.	3.2	35
62	Three novel mutations in the coproporphyrinogen oxidase gene. Human Mutation, 1997, 9, 78-80.	2.5	20
63	Protoporphyrinogen Oxidase: Complete Genomic Sequence and Polymorphisms in the Human Gene. Biochemical and Biophysical Research Communications, 1996, 226, 226-230.	2.1	36
64	A molecular defect in coproporphyrinogen oxidase gene causing harderoporphyria, a variant form of hereditary coproporphyria. Human Molecular Genetics, 1995, 4, 275-278.	2.9	58
65	Detection of eleven mutations causing acute intermittent porphyria using denaturing gradient gel electrophoresis. Human Genetics, 1994, 93, 47-52.	3.8	50
66	Ferrochelatase Structural Mutant (Fechm1Pas) in the House Mouse. Genomics, 1993, 16, 645-648.	2.9	68
67	Decreased nocturnal plasma melatonin levels in patients with recurrent acute intermittent porphyria attacks. Life Sciences, 1993, 53, 621-627.	4.3	52
68	Heterogeneity of mutations in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria. Human Genetics, 1992, 88, 320-324.	3.8	50
69	Human Erythropoietic Protoporphyria: Two point mutations in the ferrochelatase gene. Biochemical and Biophysical Research Communications, 1991, 181, 594-599.	2.1	118
70	The mitochondrial location of protoporphyrinogen oxidase. FEBS Journal, 1985, 149, 431-435.	0.2	64