

# Jean-Charles Deybach

## List of Publications by Year in descending order

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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	Porphyrias. Lancet, The, 2010, 375, 924-937.	13.7	644
2	C-Terminal Deletions in the ALAS2 Gene Lead to Gain of Function and Cause X-linked Dominant Protoporphyrinemia without Anemia or Iron Overload. American Journal of Human Genetics, 2008, 83, 408-414.	6.2	246
3	The penetrance of dominant erythropoietic protoporphyria is modulated by expression of wildtype FECH. Nature Genetics, 2002, 30, 27-28.	21.4	237
4	The incidence of inherited porphyrias in Europe. Journal of Inherited Metabolic Disease, 2013, 36, 849-857.	3.6	220
5	Afamelanotide for Erythropoietic Protoporphyrinemia. New England Journal of Medicine, 2015, 373, 48-59.	27.0	206
6	Erythropoietic protoporphyria. Orphanet Journal of Rare Diseases, 2009, 4, 19.	2.7	178
7	Contribution of a Common Single-Nucleotide Polymorphism to the Genetic Predisposition for Erythropoietic Protoporphyrinemia. American Journal of Human Genetics, 2006, 78, 2-14.	6.2	164
8	Inheritance in Erythropoietic Protoporphyrinemia: A Common Wild-Type Ferrochelatase Allelic Variant With Low Expression Accounts for Clinical Manifestation. Blood, 1999, 93, 2105-2110.	1.4	140
9	Porphyrias: A 2015 update. Clinics and Research in Hepatology and Gastroenterology, 2015, 39, 412-425.	1.5	132
10	ABCB6 is dispensable for erythropoiesis and specifies the new blood group system Langereis. Nature Genetics, 2012, 44, 170-173.	21.4	127
11	Hepatocellular carcinoma in patients with acute hepatic porphyria: frequency of occurrence and related factors. Journal of Hepatology, 2000, 32, 933-939.	3.7	124
12	Human Erythropoietic Protoporphyrinemia: Two point mutations in the ferrochelatase gene. Biochemical and Biophysical Research Communications, 1991, 181, 594-599.	2.1	118
13	The 3' region of the DRD2 gene is involved in genetic susceptibility to schizophrenia. Schizophrenia Research, 2004, 67, 75-85.	2.0	115
14	EXPLORE: A Prospective, Multinational, Natural History Study of Patients with Acute Hepatic Porphyria with Recurrent Attacks. Hepatology, 2020, 71, 1546-1558.	7.3	103
15	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
16	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
17	ALAS2 acts as a modifier gene in patients with congenital erythropoietic porphyria. Blood, 2011, 118, 1443-1451.	1.4	80
18	Vemurafenib: an unusual UVA-induced photosensitivity. Experimental Dermatology, 2013, 22, 297-298.	2.9	76

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19	From a dominant to an oligogenic model of inheritance with environmental modifiers in acute intermittent porphyria. <i>Human Molecular Genetics</i> , 2018, 27, 1164-1173.	2.9	73
20	Ferrochelatase Structural Mutant (Fechm1Pas) in the House Mouse. <i>Genomics</i> , 1993, 16, 645-648.	2.9	68
21	The mitochondrial location of protoporphyrinogen oxidase. <i>FEBS Journal</i> , 1985, 149, 431-435.	0.2	64
22	Characterization of Mutations in the CPO Gene in British Patients Demonstrates Absence of Genotype-Phenotype Correlation and Identifies Relationship between Hereditary Coproporphyrinemia and Harderoporphyria. <i>American Journal of Human Genetics</i> , 2001, 68, 1130-1138.	6.2	62
23	New insights into the pathogenesis of erythropoietic protoporphyria and their impact on patient care. <i>European Journal of Pediatrics</i> , 2000, 159, 719-725.	2.7	61
24	A molecular defect in coproporphyrinogen oxidase gene causing harderoporphyria, a variant form of hereditary coproporphyrinemia. <i>Human Molecular Genetics</i> , 1995, 4, 275-278.	2.9	58
25	Increased plasma transferrin, altered body iron distribution, and microcytic hypochromic anemia in ferrochelatase-deficient mice. <i>Blood</i> , 2007, 109, 811-818.	1.4	58
26	Sideroblastic anemia: molecular analysis of the ALAS2 gene in a series of 29 probands and functional studies of 10 missense mutations. <i>Human Mutation</i> , 2011, 32, 590-597.	2.5	56
27	Decreased nocturnal plasma melatonin levels in patients with recurrent acute intermittent porphyria attacks. <i>Life Sciences</i> , 1993, 53, 621-627.	4.3	52
28	Acute intermittent porphyria causes hepatic mitochondrial energetic failure in a mouse model. <i>International Journal of Biochemistry and Cell Biology</i> , 2014, 51, 93-101.	2.8	51
29	Heterogeneity of mutations in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria. <i>Human Genetics</i> , 1992, 88, 320-324.	3.8	50
30	Detection of eleven mutations causing acute intermittent porphyria using denaturing gradient gel electrophoresis. <i>Human Genetics</i> , 1994, 93, 47-52.	3.8	50
31	Epidemiology of hepatitis C and G in sporadic and familial porphyria cutanea tarda. <i>Hepatology</i> , 1998, 27, 848-852.	7.3	49
32	Mutations in human CPO gene predict clinical expression of either hepatic hereditary coproporphyrinemia or erythropoietic harderoporphyria. <i>Human Molecular Genetics</i> , 2005, 14, 3089-3098.	2.9	44
33	Modulation of penetrance by the wild-type allele in dominantly inherited erythropoietic protoporphyria and acute hepatic porphyrias. <i>Human Genetics</i> , 2004, 114, 256-262.	3.8	40
34	Influence of Age and Gender on the Clinical Expression of Acute Intermittent Porphyria Based on Molecular Study of Porphobilinogen Deaminase Gene Among Swiss Patients. <i>Molecular Medicine</i> , 2001, 7, 535-542.	4.4	39
35	Recovery from a variegate porphyria by a liver transplantation. <i>Liver Transplantation</i> , 2004, 10, 935-938.	2.4	39
36	A genetic schizophrenia-susceptibility region located between the ANKK1 and DRD2 genes. <i>Progress in Neuro-Psychopharmacology and Biological Psychiatry</i> , 2010, 34, 492-499.	4.8	39

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37	Protoporphyrin Retention in Hepatocytes and Kupffer Cells Prevents Sclerosing Cholangitis in Erythropoietic Protoporphyrin Mouse Model. <i>Gastroenterology</i> , 2011, 141, 1509-1519.e3.	1.3	39
38	Molecular and functional analysis of the C-terminal region of human erythroid-specific 5-aminolevulinic synthase associated with X-linked dominant protoporphyria (XLDPP). <i>Human Molecular Genetics</i> , 2013, 22, 1280-1288.	2.9	39
39	European Specialist Porphyria Laboratories: Diagnostic Strategies, Analytical Quality, Clinical Interpretation, and Reporting As Assessed by an External Quality Assurance Program. <i>Clinical Chemistry</i> , 2011, 57, 1514-1523.	3.2	38
40	Molecular Characterization of Homozygous Variegate Porphyria. <i>Human Molecular Genetics</i> , 1998, 7, 1921-1925.	2.9	37
41	Red cells from ferrochelatase-deficient erythropoietic protoporphyria patients are resistant to growth of malarial parasites. <i>Blood</i> , 2015, 125, 534-541.	1.4	37
42	Protoporphyrinogen Oxidase: Complete Genomic Sequence and Polymorphisms in the Human Gene. <i>Biochemical and Biophysical Research Communications</i> , 1996, 226, 226-230.	2.1	36
43	5-Aminolevulinic acid dehydratase deficiency porphyria: a twenty-year clinical and biochemical follow-up. <i>Clinical Chemistry</i> , 1998, 44, 1892-1896.	3.2	35
44	Mutations in the iron-sulfur cluster ligands of the human ferrochelatase lead to erythropoietic protoporphyria. <i>Blood</i> , 2000, 96, 1545-1549.	1.4	34
45	Antisense Oligonucleotide-Based Therapy in Human Erythropoietic Protoporphyrin. <i>American Journal of Human Genetics</i> , 2014, 94, 611-617.	6.2	34
46	Mitochondrial energetic defects in muscle and brain of a <i>Hmbs</i> <sup>Δ</sup> mouse model of acute intermittent porphyria. <i>Human Molecular Genetics</i> , 2015, 24, 5015-5023.	2.9	34
47	Acute Hepatic Porphyrias and Primary Liver Cancer. <i>New England Journal of Medicine</i> , 1998, 338, 1853-1854.	27.0	33
48	A mouse model provides evidence that genetic background modulates anemia and liver injury in erythropoietic protoporphyria. <i>American Journal of Physiology - Renal Physiology</i> , 2005, 288, G1208-G1216.	3.4	32
49	Hepatocellular carcinoma in acute hepatic porphyrias: A Damocles Sword. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 236-241.	1.1	32
50	Genetic Study of Variation in Normal Mouse Iron Homeostasis Reveals Ceruloplasmin as an HFE-Hemochromatosis Modifier Gene. <i>Gastroenterology</i> , 2007, 132, 679-686.	1.3	26
51	Current and innovative emerging therapies for porphyrias with hepatic involvement. <i>Journal of Hepatology</i> , 2019, 71, 422-433.	3.7	24
52	Identification of a Prevalent Nonsense Mutation (W283X) and Two Novel Mutations in the Porphobilinogen Deaminase Gene of Swiss Patients with Acute Intermittent Porphyria. <i>Human Heredity</i> , 2000, 50, 247-250.	0.8	23
53	Erythroid-Progenitor-Targeted Gene Therapy Using Bifunctional TFR1 Ligand-Peptides in Human Erythropoietic Protoporphyrin. <i>American Journal of Human Genetics</i> , 2019, 104, 341-347.	6.2	22
54	Three novel mutations in the coproporphyrinogen oxidase gene. <i>Human Mutation</i> , 1997, 9, 78-80.	2.5	20

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55	Human Erythroid 5-Aminolevulinate Synthase Mutations Associated with X-Linked Protoporphyrin Disrupt the Conformational Equilibrium and Enhance Product Release. <i>Biochemistry</i> , 2015, 54, 5617-5631.	2.5	18
56	Ancestral Founder of Mutation W283X in the Porphobilinogen Deaminase Gene among Acute Intermittent Porphyria Patients. <i>Human Heredity</i> , 2002, 54, 69-81.	0.8	17
57	International Porphyria Molecular Diagnostic Collaborative: an evidence-based database of verified pathogenic and benign variants for the porphyrias. <i>Genetics in Medicine</i> , 2019, 21, 2605-2613.	2.4	16
58	Phlebotomy as an efficient long-term treatment of congenital erythropoietic porphyria. <i>Haematologica</i> , 2021, 106, 913-917.	3.5	13
59	Establishing a network of specialist Porphyria centres - effects on diagnostic activities and services. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 93.	2.7	12
60	Mutations in the Ferrochelatase Gene of Four Spanish Patients with Erythropoietic Protoporphyrin. <i>Journal of Investigative Dermatology</i> , 1998, 111, 406-409.	0.7	11
61	Haplotype Analysis in Determination of the Heredity of Erythropoietic Protoporphyrin among Swiss Families. <i>Journal of Investigative Dermatology</i> , 2001, 117, 1521-1525.	0.7	10
62	New Missense Mutation in the Human Ferrochelatase Gene in a Family with Erythropoietic Protoporphyrin: Functional Studies and Correlation of Genotype and Phenotype. <i>Clinical Chemistry</i> , 2001, 47, 1112-1113.	3.2	7
63	Acute Intermittent Porphyria: From Clinical to Molecular Aspects. , 2003, , 23-41.		7
64	An Uncommon Option for Surviving Bariatric Surgery: Regaining Weight!. <i>American Journal of Medicine</i> , 2012, 125, e1-e2.	1.5	7
65	Systemic Administered mRNA as Therapy for Metabolic Diseases. <i>Trends in Molecular Medicine</i> , 2019, 25, 3-5.	6.7	4
66	Heme Biosynthesis and Pathophysiology of Porphyrias. <i>Handbook of Porphyrin Science</i> , 2013, , 89-118.	0.8	2
67	Mutations in the iron-sulfur cluster ligands of the human ferrochelatase lead to erythropoietic protoporphyria. <i>Blood</i> , 2000, 96, 1545-1549.	1.4	1
68	Chediak-Steinbrinck-Higashi Syndrome. , 2009, , 314-314.		0
69	Epistasis in iron metabolism: complex interactions between Cp, Mon1a, and Slc40a1 loci and tissue iron in mice. <i>Mammalian Genome</i> , 2013, 24, 427-438.	2.2	0
70	Les porphyries héréditaires : anomalies du métabolisme de l'hème. <i>Bulletin De L'Academie Nationale De Medecine</i> , 2014, 198, 1069-1093.	0.0	0