

Herve Puy

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

142
papers

5,727
citations

40
h-index

71
g-index

156
ext. papers

6,723
ext. citations

7.2
avg. IF

5.24
L-index

#	Paper	IF	Citations
142	Iron, Heme Synthesis and Erythropoietic Porphyrins: A Complex Interplay.. <i>Metabolites</i> , 2021 , 11,	5.6	1
141	Identification of novel mutations in a patient with congenital erythropoietic porphyria and efficient treatment by phlebotomy. <i>Molecular Genetics and Metabolism Reports</i> , 2021 , 27, 100722	1.8	3
140	Hepcidin and Iron Deficiency in Women One Year after Sleeve Gastrectomy: A Prospective Cohort Study. <i>Nutrients</i> , 2021 , 13,	6.7	1
139	Phlebotomy as an efficient long-term treatment of congenital erythropoietic porphyria. <i>Haematologica</i> , 2021 , 106, 913-917	6.6	7
138	A mutation in the iron-responsive element of is a modifier of disease severity in a patient suffering from associated erythropoietic protoporphyria. <i>Haematologica</i> , 2021 , 106, 2030-2033	6.6	2
137	Analytical comparison of ELISA and mass spectrometry for quantification of serum hepcidin in critically ill patients. <i>Bioanalysis</i> , 2021 , 13, 1029-1035	2.1	2
136	Renal Function Decline With Small Interfering RNA Silencing Aminolevulinic Acid Synthase 1 (ALAS1). <i>Kidney International Reports</i> , 2021 , 6, 1904-1911	4.1	6
135	TSPO2 translocates 5-aminolevulinic acid into human erythroleukemia cells. <i>Biology of the Cell</i> , 2020 , 112, 113-126	3.5	2
134	Iron chelation rescues hemolytic anemia and skin photosensitivity in congenital erythropoietic porphyria. <i>Blood</i> , 2020 , 136, 2457-2468	2.2	9
133	Kidney transplantation improves the clinical outcomes of Acute Intermittent Porphyrin. <i>Molecular Genetics and Metabolism</i> , 2020 , 131, 259-266	3.7	3
132	GLRX5 mutations impair heme biosynthetic enzymes ALA synthase 2 and ferrochelatase in Human congenital sideroblastic anemia. <i>Molecular Genetics and Metabolism</i> , 2019 , 128, 342-351	3.7	9
131	Erythroid-Progenitor-Targeted Gene Therapy Using Bifunctional TFR1 Ligand-Peptides in Human Erythropoietic Protoporphyria. <i>American Journal of Human Genetics</i> , 2019 , 104, 341-347	11	17
130	Regulation of globin-heme balance in Diamond-Blackfan anemia by HSP70/GATA1. <i>Blood</i> , 2019 , 133, 1358-1370	2.2	21
129	International Porphyrin 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	8.1	11
128	Regulation and tissue-specific expression of Aminolevulinic acid synthases in non-syndromic sideroblastic anemias and porphyrias. <i>Molecular Genetics and Metabolism</i> , 2019 , 128, 190-197	3.7	13
127	Results of a pilot study of isoniazid in patients with erythropoietic protoporphyria. <i>Molecular Genetics and Metabolism</i> , 2019 , 128, 309-313	3.7	7
126	A variant erythroferrone disrupts iron homeostasis in -mutated myelodysplastic syndrome. <i>Science Translational Medicine</i> , 2019 , 11,	17.5	27

125	Genetic background influences hepcidin response to iron imbalance in a mouse model of hemolytic anemia (Congenital erythropoietic porphyria). <i>Biochemical and Biophysical Research Communications</i> , 2019 , 520, 297-303	3.4	3
124	Extrahepatic hepcidin production: The intriguing outcomes of recent years. <i>World Journal of Clinical Cases</i> , 2019 , 7, 1926-1936	1.6	6
123	Functional erythropoietin-hepcidin axis in recombinant human erythropoietin independent haemodialysis patients. <i>Nephrology</i> , 2019 , 24, 751-757	2.2	3
122	Hepatocellular carcinoma in acute hepatic porphyrias: A Damocles Sword. <i>Molecular Genetics and Metabolism</i> , 2019 , 128, 236-241	3.7	16
121	Systemic Administered mRNA as Therapy for Metabolic Diseases. <i>Trends in Molecular Medicine</i> , 2019 , 25, 3-5	11.5	2
120	Dyserythropoiesis evaluated by the RED score and hepcidin:ferritin ratio predicts response to erythropoietin in lower-risk myelodysplastic syndromes. <i>Haematologica</i> , 2019 , 104, 497-504	6.6	7
119	Porphyria and kidney diseases. <i>CKJ: Clinical Kidney Journal</i> , 2018 , 11, 191-197	4.5	24
118	Gene Therapy in Patients with Transfusion-Dependent β Thalassemia. <i>New England Journal of Medicine</i> , 2018 , 378, 1479-1493	59.2	347
117	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	5.6	42
116	Urinary metabolic profiling of asymptomatic acute intermittent porphyria using a rule-mining-based algorithm. <i>Metabolomics</i> , 2018 , 14, 10	4.7	4
115	Involvement of hepcidin in iron metabolism dysregulation in Gaucher disease. <i>Haematologica</i> , 2018 , 103, 587-596	6.6	12
114	Iron deficiency diagnosed using hepcidin on critical care discharge is an independent risk factor for death and poor quality of life at one year: an observational prospective study on 1161 patients. <i>Critical Care</i> , 2018 , 22, 314	10.8	27
113	Characterization and origin of heme precursors in amniotic fluid: lessons from normal and pathological pregnancies. <i>Pediatric Research</i> , 2018 , 84, 80-84	3.2	1
112	High urinary ferritin reflects myoglobin iron evacuation in DMD patients. <i>Neuromuscular Disorders</i> , 2018 , 28, 564-571	2.9	6
111	Gene Therapy in a Patient with Sickle Cell Disease. <i>New England Journal of Medicine</i> , 2017 , 376, 848-855	59.2	418
110	Hemolytic anemia repressed hepcidin level without hepatocyte iron overload: lesson from G β her disease model. <i>Haematologica</i> , 2017 , 102, 260-270	6.6	9
109	Acute hepatic and erythropoietic porphyrias: from ALA synthases 1 and 2 to new molecular bases and treatments. <i>Current Opinion in Hematology</i> , 2017 , 24, 198-207	3.3	19
108	Iron status and inflammatory biomarkers in patients with acutely decompensated heart failure: early in-hospital phase and 30-day follow-up. <i>European Journal of Heart Failure</i> , 2017 , 19, 1075-1076	12.3	20

107	Iloprost Use in Patients with Persistent Intestinal Ischemia Unsuitable for Revascularization. <i>Annals of Vascular Surgery</i> , 2017 , 42, 128-135	1.7	4
106	Isoniazid inhibits human erythroid 5-aminolevulinate synthase: Molecular mechanism and tolerance study with four X-linked protoporphyria patients. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2017 , 1863, 428-439	6.9	11
105	A Variant of Peptide Transporter 2 Predicts the Severity of Porphyrin-Associated Kidney Disease. <i>Journal of the American Society of Nephrology: JASN</i> , 2017 , 28, 1924-1932	12.7	35
104	Impact of iron deficiency diagnosis using hepcidin mass spectrometry dosage methods on hospital stay and costs after a prolonged ICU stay: Study protocol for a multicentre, randomised, single-blinded medico-economic trial. <i>Anaesthesia, Critical Care & Pain Medicine</i> , 2017 , 36, 391-396	3	8
103	Mutation in human elevates levels of aminolevulinate synthase and protoporphyrin IX to promote erythropoietic protoporphyria. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017 , 114, E8045-E8052	11.5	50
102	GNPAT polymorphism rs11558492 is not associated with increased severity in a large cohort of HFE p.Cys282Tyr homozygous patients. <i>Hepatology</i> , 2017 , 65, 1069-1071	11.2	3
101	Fecal calprotectin in inflammatory bowel diseases: update and perspectives. <i>Clinical Chemistry and Laboratory Medicine</i> , 2017 , 55, 474-483	5.9	50
100	Cardiac iron overload in chronically transfused patients with thalassemia, sickle cell anemia, or myelodysplastic syndrome. <i>PLoS ONE</i> , 2017 , 12, e0172147	3.7	29
99	Hepcidin as a Major Component of Renal Antibacterial Defenses against Uropathogenic Escherichia coli. <i>Journal of the American Society of Nephrology: JASN</i> , 2016 , 27, 835-46	12.7	33
98	Reply. <i>Gastroenterology</i> , 2016 , 151, 771-2	13.3	2
97	Heterozygous Mutations in BMP6 Pro-peptide Lead to Inappropriate Hepcidin Synthesis and Moderate Iron Overload in Humans. <i>Gastroenterology</i> , 2016 , 150, 672-683.e4	13.3	61
96	Update from the Hgb-205 Phase 1/2 Clinical Study of Lentiglobin Gene Therapy: Sustained Clinical Benefit in Severe Hemoglobinopathies. <i>Blood</i> , 2016 , 128, 2311-2311	2.2	3
95	Anémies microcytaires rares. <i>Bulletin De L'Académie Nationale De Médecine</i> , 2016 , 200, 335-347	0.1	
94	A Dominant Mutation in Mitochondrial Unfoldase CLPX Results in Erythropoietic Protoporphyrin. <i>Blood</i> , 2016 , 128, 77-77	2.2	
93	Does IV Iron Induce Plasma Oxidative Stress in Critically Ill Patients? A Comparison With Healthy Volunteers. <i>Critical Care Medicine</i> , 2016 , 44, 521-30	1.4	8
92	Porphyrias: A 2015 update. <i>Clinics and Research in Hepatology and Gastroenterology</i> , 2015 , 39, 412-25	2.4	96
91	High prevalence of and potential mechanisms for chronic kidney disease in patients with acute intermittent porphyria. <i>Kidney International</i> , 2015 , 88, 386-95	9.9	55
90	Mitochondrial energetic defects in muscle and brain of a Hmbs ^{-/-} mouse model of acute intermittent porphyria. <i>Human Molecular Genetics</i> , 2015 , 24, 5015-23	5.6	25

89	Human Erythroid 5-Aminolevulinate Synthase Mutations Associated with X-Linked Protoporphyrin Disrupt the Conformational Equilibrium and Enhance Product Release. <i>Biochemistry</i> , 2015 , 54, 5617-31	3.2	16
88	LC-MS/MS method for hepcidin-25 measurement in human and mouse serum: clinical and research implications in iron disorders. <i>Clinical Chemistry and Laboratory Medicine</i> , 2015 , 53, 1557-67	5.9	39
87	Red cells from ferrochelatase-deficient erythropoietic protoporphyria patients are resistant to growth of malarial parasites. <i>Blood</i> , 2015 , 125, 534-41	2.2	25
86	Iron regulatory protein 1 sustains mitochondrial iron loading and function in frataxin deficiency. <i>Cell Metabolism</i> , 2015 , 21, 311-323	24.6	46
85	Performance of PIVKA-II for early hepatocellular carcinoma diagnosis and prediction of microvascular invasion. <i>Journal of Hepatology</i> , 2015 , 62, 848-54	13.4	157
84	Assessment of Cardiac Iron Overload in Chronically Transfused Patients with Thalassemia, Sickle Cell Anemia, and Myelodysplastic Syndromes. <i>Blood</i> , 2015 , 126, 2151-2151	2.2	
83	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	5.6	36
82	Urinary metabolic fingerprint of acute intermittent porphyria analyzed by (1)H NMR spectroscopy. <i>Analytical Chemistry</i> , 2014 , 86, 2166-74	7.8	16
81	Pro-oxidant effect of ALA is implicated in mitochondrial dysfunction of HepG2 cells. <i>Biochimie</i> , 2014 , 106, 157-66	4.6	18
80	Clinical measurement of Hepcidin-25 in human serum: Is quantitative mass spectrometry up to the job?. <i>EuPA Open Proteomics</i> , 2014 , 3, 60-67	0.1	18
79	Antisense oligonucleotide-based therapy in human erythropoietic protoporphyria. <i>American Journal of Human Genetics</i> , 2014 , 94, 611-7	11	23
78	Les porphyries héréditaires : anomalies du métabolisme de l'hème. <i>Bulletin De L'Académie Nationale De Médecine</i> , 2014 , 198, 1069-1093	0.1	
77	Erythropoietic Protoporphyrin Red Blood Cells Are Resistant to the Growth of Malarial Parasites. <i>Blood</i> , 2014 , 124, 2670-2670	2.2	
76	Epistasis in iron metabolism: complex interactions between Cp, Mon1a, and Slc40a1 loci and tissue iron in mice. <i>Mammalian Genome</i> , 2013 , 24, 427-38	3.2	
75	Hepcidin regulates intrarenal iron handling at the distal nephron. <i>Kidney International</i> , 2013 , 84, 756-66	9.9	47
74	Heme Biosynthesis and Pathophysiology of Porphyrins. <i>Handbook of Porphyrin Science</i> , 2013 , 89-118	0.3	1
73	Late-onset X-linked dominant protoporphyria: an etiology of photosensitivity in the elderly. <i>Journal of Investigative Dermatology</i> , 2013 , 133, 1688-90	4.3	10
72	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, 1280-8	5.6	35

71	ABCB6 is dispensable for erythropoiesis and specifies the new blood group system Langereis. <i>Nature Genetics</i> , 2012 , 44, 170-3	36.3	108
70	A management algorithm for congenital erythropoietic porphyria derived from a study of 29 cases. <i>British Journal of Dermatology</i> , 2012 , 167, 888-900	4	46
69	Congenital erythropoietic porphyria: a single-observer clinical study of 29 cases. <i>British Journal of Dermatology</i> , 2012 , 167, 901-13	4	53
68	Null alleles of ABCG2 encoding the breast cancer resistance protein define the new blood group system Junior. <i>Nature Genetics</i> , 2012 , 44, 174-7	36.3	93
67	An uncommon option for surviving bariatric surgery: regaining weight!. <i>American Journal of Medicine</i> , 2012 , 125, e1-2	2.4	4
66	Comprehensive cytochrome P450 CYP1A2 gene analysis in French caucasian patients with familial and sporadic porphyria cutanea tarda. <i>British Journal of Dermatology</i> , 2012 , 166, 425-9	4	6
65	Iron metabolism in patients with anorexia nervosa: elevated serum hepcidin concentrations in the absence of inflammation. <i>American Journal of Clinical Nutrition</i> , 2012 , 95, 548-54	7	24
64	Protoporphyrin retention in hepatocytes and Kupffer cells prevents sclerosing cholangitis in erythropoietic protoporphyria mouse model. <i>Gastroenterology</i> , 2011 , 141, 1509-19, 1519.e1-3	13.3	32
63	Acute porphyric attack mimicking HIV-associated progressive polyradiculoneuropathy. <i>Medicine Et Maladies Infectieuses</i> , 2011 , 41, 441-3	4	1
62	ALAS2 acts as a modifier gene in patients with congenital erythropoietic porphyria. <i>Blood</i> , 2011 , 118, 1443-51	2.2	72
61	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, 590-7	4.7	45
60	A homoallelic FECH mutation in a patient with both erythropoietic protoporphyria and palmar keratoderma. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2010 , 24, 1349-53	4.6	10
59	Iron regulatory proteins secure mitochondrial iron sufficiency and function. <i>Cell Metabolism</i> , 2010 , 12, 194-201	24.6	89
58	Porphyrias. <i>Lancet, The</i> , 2010 , 375, 924-37	40	506
57	Diagnostic accuracy of serum hepcidin for iron deficiency in critically ill patients with anemia. <i>Intensive Care Medicine</i> , 2010 , 36, 1044-8	14.5	70
56	Role of two nutritional hepatic markers (insulin-like growth factor 1 and transthyretin) in the clinical assessment and follow-up of acute intermittent porphyria patients. <i>Journal of Internal Medicine</i> , 2009 , 266, 277-85	10.8	22
55	Erythropoietic protoporphyria. <i>Orphanet Journal of Rare Diseases</i> , 2009 , 4, 19	4.2	142
54	Melatonin and environmental lighting regulate ALA-S gene expression and So porphyrin biosynthesis in the rat harderian gland. <i>Chronobiology International</i> , 2008 , 25, 851-67	3.6	10

53	Sequential regulation of ferroportin expression after erythrophagocytosis in murine macrophages: early mRNA induction by haem, followed by iron-dependent protein expression. <i>Biochemical Journal</i> , 2008 , 411, 123-31	3.8	103
52	C-terminal deletions in the ALAS2 gene lead to gain of function and cause X-linked dominant protoporphyria without anemia or iron overload. <i>American Journal of Human Genetics</i> , 2008 , 83, 408-14	11	208
51	Genetic study of variation in normal mouse iron homeostasis reveals ceruloplasmin as an HFE-hemochromatosis modifier gene. <i>Gastroenterology</i> , 2007 , 132, 679-86	13.3	22
50	Increased plasma transferrin, altered body iron distribution, and microcytic hypochromic anemia in ferrochelatase-deficient mice. <i>Blood</i> , 2007 , 109, 811-8	2.2	49
49	Contribution of a common single-nucleotide polymorphism to the genetic predisposition for erythropoietic protoporphyria. <i>American Journal of Human Genetics</i> , 2006 , 78, 2-14	11	140
48	Biochemical compared to molecular diagnosis in acute intermittent porphyria. <i>Journal of Inherited Metabolic Disease</i> , 2006 , 29, 157-61	5.4	4
47	Plasma N-terminal pro-brain natriuretic peptide and brain natriuretic peptide in assessment of acute dyspnea. <i>Biomedicine and Pharmacotherapy</i> , 2005 , 59, 20-4	7.5	43
46	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, G1208-16	5.1	25
45	Mutations in human CPO gene predict clinical expression of either hepatic hereditary coproporphyria or erythropoietic harderoporphyria. <i>Human Molecular Genetics</i> , 2005 , 14, 3089-98	5.6	38
44	Loss of heterozygosity on 10q and mutational status of PTEN and BMPR1A in colorectal primary tumours and metastases. <i>British Journal of Cancer</i> , 2004 , 90, 1230-4	8.7	23
43	Modulation of penetrance by the wild-type allele in dominantly inherited erythropoietic protoporphyria and acute hepatic porphyrias. <i>Human Genetics</i> , 2004 , 114, 256-62	6.3	32
42	Analytical correlation between plasma N-terminal pro-brain natriuretic peptide and brain natriuretic peptide in patients presenting with dyspnea. <i>Clinical Biochemistry</i> , 2004 , 37, 933-6	3.5	14
41	Acute Intermittent Porphyria: From Clinical to Molecular Aspects 2003 , 23-41		2
40	The penetrance of dominant erythropoietic protoporphyria is modulated by expression of wildtype FECH. <i>Nature Genetics</i> , 2002 , 30, 27-8	36.3	203
39	A molecular, enzymatic and clinical study in a family with hereditary coproporphyria. <i>Journal of Inherited Metabolic Disease</i> , 2002 , 25, 279-86	5.4	12
38	Ancestral founder of mutation W283X in the porphobilinogen deaminase gene among acute intermittent porphyria patients. <i>Human Heredity</i> , 2002 , 54, 69-81	1.1	13
37	Human hereditary hepatic porphyrias. <i>Clinica Chimica Acta</i> , 2002 , 325, 17-37	6.2	55
36	Hemochromatosis (HFE) and transferrin receptor-1 (TFRC1) genes in sporadic porphyria cutanea tarda (SPCT). <i>Cellular and Molecular Biology</i> , 2002 , 48, 33-41	1.1	7

35	Characterization of mutations in the CPO gene in British patients demonstrates absence of genotype-phenotype correlation and identifies relationship between hereditary coproporphyrria and harderoporphyria. <i>American Journal of Human Genetics</i> , 2001 , 68, 1130-8	11	48
34	Influence of Age and Gender on the Clinical Expression of Acute Intermittent Porphyrria Based on Molecular Study of Porphobilinogen Deaminase Gene Among Swiss Patients. <i>Molecular Medicine</i> , 2001 , 7, 535-542	6.2	27
33	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, 247-50	1.1	21
32	Porphobilinogen deaminase gene in African and Afro-Caribbean ethnic groups: mutations causing acute intermittent porphyria and specific intragenic polymorphisms. <i>Human Genetics</i> , 2000 , 107, 150-9	6.3	15
31	Hepatocellular carcinoma in patients with acute hepatic porphyria: frequency of occurrence and related factors. <i>Journal of Hepatology</i> , 2000 , 32, 933-9	13.4	98
30	Inheritance in Erythropoietic Protoporphyrria: A Common Wild-Type Ferrochelatase Allelic Variant With Low Expression Accounts for Clinical Manifestation. <i>Blood</i> , 1999 , 93, 2105-2110	2.2	114
29	Heme and acute inflammation role in vivo of heme in the hepatic expression of positive acute-phase reactants in rats. <i>FEBS Journal</i> , 1999 , 261, 190-6		18
28	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. <i>American Journal of Human Genetics</i> , 1999 , 65, 984-94	11	90
27	Evaluation of mutation screening by heteroduplex analysis in acute intermittent porphyria: comparison with denaturing gradient gel electrophoresis. <i>Clinica Chimica Acta</i> , 1999 , 279, 133-43	6.2	13
26	New mutations of the hydroxymethylbilane synthase gene in German patients with acute intermittent porphyria. <i>Molecular and Cellular Probes</i> , 1999 , 13, 443-7	3.3	9
25	Epidemiology of hepatitis C and G in sporadic and familial porphyria cutanea tarda. <i>Hepatology</i> , 1998 , 27, 848-52	11.2	43
24	Exon 1 donor splice site mutations in the porphobilinogen deaminase gene in the non-erythroid variant form of acute intermittent porphyria. <i>Human Genetics</i> , 1998 , 103, 570-5	6.3	26
23	Mutations in the ferrochelatase gene of four Spanish patients with erythropoietic protoporphyria. <i>Journal of Investigative Dermatology</i> , 1998 , 111, 406-9	4.3	8
22	Systematic analysis of molecular defects in the ferrochelatase gene from patients with erythropoietic protoporphyria. <i>American Journal of Human Genetics</i> , 1998 , 62, 1341-52	11	117
21	Molecular characterization of homozygous variegate porphyria. <i>Human Molecular Genetics</i> , 1998 , 7, 1921-5	5.5	45
20	Molecular Characterization of Homozygous Variegate Porphyria. <i>Human Molecular Genetics</i> , 1998 , 7, 1921-1925	5.6	36
19	Acute hepatic porphyrias and primary liver cancer. <i>New England Journal of Medicine</i> , 1998 , 338, 1853-4	59.2	28
18	Nitric oxide synthase inhibition and the induction of cytochrome P-450 affect heme oxygenase-1 messenger RNA expression after partial hepatectomy and acute inflammation in rats. <i>Critical Care Medicine</i> , 1998 , 26, 1683-9	1.4	13

17	Molecular analysis of porphobilinogen (PBG) deaminase gene mutations in acute intermittent porphyria: first study in patients of Slavic origin. <i>Scandinavian Journal of Clinical and Laboratory Investigation</i> , 1997 , 57, 217-24	2	15
16	Molecular epidemiology and diagnosis of PBG deaminase gene defects in acute intermittent porphyria. <i>American Journal of Human Genetics</i> , 1997 , 60, 1373-83	11	122
15	Acute intermittent porphyria: prevalence of mutations in the porphobilinogen deaminase gene in blood donors in France. <i>Journal of Internal Medicine</i> , 1997 , 242, 213-7	10.8	75
14	Three novel mutations in the coproporphyrinogen oxidase gene. <i>Human Mutation</i> , 1997 , 9, 78-80	4.7	16
13	Protoporphyrinogen oxidase: complete genomic sequence and polymorphisms in the human gene. <i>Biochemical and Biophysical Research Communications</i> , 1996 , 226, 226-30	3.4	32
12	Detection of four novel mutations in the porphobilinogen deaminase gene in French Caucasian patients with acute intermittent porphyria. <i>Human Heredity</i> , 1996 , 46, 177-80	1.1	12
11	Review: molecular pathogenesis of hepatic acute porphyrias. <i>Journal of Gastroenterology and Hepatology (Australia)</i> , 1996 , 11, 1046-52	4	22
10	Mutations in the protoporphyrinogen oxidase gene in patients with variegate porphyria. <i>Human Molecular Genetics</i> , 1996 , 5, 407-10	5.6	42
9	Increased delta aminolevulinic acid and decreased pineal melatonin production. A common event in acute porphyria studies in the rat. <i>Journal of Clinical Investigation</i> , 1996 , 97, 104-10	15.9	35
8	Porphobilinogen deaminase gene structure and molecular defects. <i>Journal of Bioenergetics and Biomembranes</i> , 1995 , 27, 197-205	3.7	29
7	Molecular abnormalities of coproporphyrinogen oxidase in patients with hereditary coproporphyria. <i>Journal of Bioenergetics and Biomembranes</i> , 1995 , 27, 215-9	3.7	16
6	Variegate porphyria: diagnostic value of fluorometric scanning of plasma porphyrins. <i>Clinica Chimica Acta</i> , 1995 , 238, 163-8	6.2	45
5	Decreased nocturnal plasma melatonin levels in patients with recurrent acute intermittent porphyria attacks. <i>Life Sciences</i> , 1993 , 53, 621-7	6.8	45
4	Thyroid hormone extraction by plasma exchange: a study of extraction rate. <i>Biomedicine and Pharmacotherapy</i> , 1992 , 46, 413-7	7.5	7
3	Accès palustre et anémie hémolytique potentiellement auto-immune chez un enfant drépanocytaire. <i>Médecine Et Maladies Infectieuses</i> , 1992 , 22, 746-748	4	
2	Immunological specificity of monoclonal antibodies to Chlamydia psittaci ovine abortion strain. <i>Immunology Letters</i> , 1990 , 23, 217-21	4.1	2
1	Porphyria898-907		