A R59W mutation in human protoporphyrinogen oxidas activity and is prevalent in South Africans with variegat

Nature Genetics

13, 95-97

DOI: 10.1038/ng0596-95

Citation Report

#	Article	IF	Citations
1	Protoporphyrinogen Oxidase: Complete Genomic Sequence and Polymorphisms in the Human Gene. Biochemical and Biophysical Research Communications, 1996, 226, 226-230.	2.1	36
2	Molecular basis of variegate porphyria: a de novo insertion mutation in the protoporphyrinogen oxidase gene. Human Genetics, 1996, 99, 126-129.	3.8	26
3	The South African malady. Nature Genetics, 1996, 13, 7-8.	21.4	25
4	The Leptin Receptor Activates Janus Kinase 2 and Signals for Proliferation in a Factor-Dependent Cell Line. Molecular Endocrinology, 1997, 11, 393-399.	3.7	282
5	Expression, purification, and characteristics of mammalian protoporphyrinogen oxidase. Methods in Enzymology, 1997, 281, 340-349.	1.0	14
6	Physiological response to long-term peripheral and central leptin infusion in lean and obeseâ€‰mice. Proceedings of the National Academy of Sciences of the United States of America, 1997, 94, 8878-8883.	7.1	937
7	The little imitator--porphyria: a neuropsychiatric disorder.. Journal of Neurology, Neurosurgery and Psychiatry, 1997, 62, 319-328.	1.9	131
8	The acute porphyrias. Lancet, The, 1997, 349, 1613-1617.	13.7	171
9	A Molecular View of the Neurologic Porphyrias. Clinics in Laboratory Medicine, 1997, 17, 73-83.	1.4	5
10	Lack of the R59W South African founder effect mutation in protoporphyrinogen oxidase in a Britsh patient with homozxygous variegate porphyria. British Journal of Dermatology, 1997, 136, 292-292.	1.5	0
11	Erythropoietic Protoporphyria: Four Novel Frameshift Mutations in the Ferrochelatase Gene. Journal of Investigative Dermatology, 1997, 109, 688-691.	0.7	13
12	Enzymes of heme biosynthesis. Journal of Biological Inorganic Chemistry, 1997, 2, 411-417.	2.6	35
13	Hepatic porphyrias in children. Journal of Inherited Metabolic Disease, 1997, 20, 237-246.	3.6	69
14	Detection of a R173W Mutation in the Porphobilinogen Deaminase Gene in the Nova Scotian â€œForeign Protestantâ€•Population with Acute Intermittent Porphyria: a Founder Effect. Clinical Biochemistry, 1997, 30, 607-612.	1.9	37
15	Update on enzyme and molecular defects in porphyria. Photodermatology Photoimmunology and Photomedicine, 1998, 14, 66-69.	1.5	25
16	Recurrent missense mutation in the protoporphyrinogen oxidase gene underlies variegate porphyria. , 1998, 79, 22-26.		13
17	Identification and characterisation of a deletion (537delAT) in the protoporphyrinogen oxidase gene in a South African variegate porphyria family. Human Mutation, 1998, 12, 403-407.	2.5	15
18	The genetic basis of "Scarsdale Gourmet Diet" variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene. Archives of Dermatological Research, 1998, 290, 441-445.	1.9	14

#	Article	IF	Citations
19	Variegate Porphyria: Identification of a Nonsense Mutation in the Protoporphyrinogen Oxidase Gene. Journal of Investigative Dermatology, 1998, 110, 449-451.	0.7	20
20	Homozygous Variegate Porphyria: Identification of Mutations on Both Alleles of the Protoporphyrinogen Oxidase Gene in a Severely Affected Proband. Journal of Investigative Dermatology, 1998, 110, 452-455.	0.7	39
21	The Biochemistry of Heme Synthesis in Porphyria and in the Porphyrinurias. Clinics in Dermatology, 1998, 16, 203-223.	1.6	22
22	Genetic Defects in the Porphyrias: Types and Significance. Clinics in Dermatology, 1998, 16, 225-233.	1.6	56
23	Linkage Disequilibrium Analysis in a Recently Founded Population: Evaluation of the Variegate Porphyria Founder in South African Afrikaners. American Journal of Human Genetics, 1998, 62, 1254-1258.	6.2	46
24	Identification of Genetic Markers Associated with Gilles de la Tourette Syndrome in an Afrikaner Population. American Journal of Human Genetics, 1998, 63, 839-846.	6.2	69
25	HEPATIC PORPHYRIAS. Clinics in Liver Disease, 1998, 2, 77-102.	2.1	6
26	Ionizing radiation and genetic risks. Mutation Research - Reviews in Mutation Research, 1998, 411, 129-178.	5.5	47
27	Molecular analysis reveals a high mutation frequency in the first untranslated exon of the PPOX gene and largely excludes variegate porphyria in a subset of clinically affected Afrikaner families. Molecular and Cellular Probes, 1998, 12, 293-300.	2.1	10
28	Molecular characterization of homozygous variegate porphyria. Human Molecular Genetics, 1998, 7, 1921-1925.	2.9	49
29	Molecular Characterization of Homozygous Variegate Porphyria. Human Molecular Genetics, 1998, 7, 1921-1925.	2.9	37
30	The Genetic Bases of the Porphyrias. Skin Pharmacology and Physiology, 1998, 11, 297-309.	2.5	25
31	Diagnosis of Porphyric Syndromes: A Practical Approach in the Era of Molecular Biology. Seminars in Liver Disease, 1998, 18, 57-65.	3.6	57
32	Variegate Porphyria. Seminars in Liver Disease, 1998, 18, 33-41.	3.6	68
33	Identification of an FAD Superfamily Containing Protoporphyrinogen Oxidases, Monoamine Oxidases, and Phytoene Desaturase. Journal of Biological Chemistry, 1998, 273, 13658-13662.	3.4	71
34	Molecular basis of variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene.. Journal of Medical Genetics, 1998, 35, 244-247.	3.2	11
35	Variegate Porphyria: Past, Present and Future. Skin Pharmacology and Physiology, 1998, 11, 310-320.	2.5	28
36	Spectrum of Mutations in the HFE Gene Implicated in Haemochromatosis and Porphyria. Human Molecular Genetics, 1999, 8, 1517-1522.	2.9	85

#	Article	IF	Citations
37	Characteristics of Protoporphyrinogen Oxidase. , 1999, , 245-277.		6
38	The cutaneous porphyrias: a review. British Journal of Dermatology, 1999, 140, 573-581.	1.5	98
39	Mutations in the translation initiation codon of the protoporphyrinogen oxidase gene underlie variegate porphyria. Clinical and Experimental Dermatology, 1999, 24, 296-301.	1.3	17
40	Haplotype Analysis of Families with Erythropoietic Protoporphyria and Novel Mutations of the Ferrochelatase Gene. Journal of Investigative Dermatology, 1999, 113, 87-92.	0.7	20
41	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
42	A novel mutation (1320InsT) identified in two Argentine families with variegate porphyria. Human Mutation, 2000, 16, 96-96.	2.5	9
43	Three novel mutations in the protoporphyrinogen oxidase gene in Japanese patients with variegate porphyria. Clinical Biochemistry, 2000, 33, 495-500.	1.9	7
44	Ionizing radiation and genetic risks. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2000, 453, 129-181.	1.0	14
45	Two deletion mutations in the hydroxymethylbilane synthase gene in two unrelated Japanese patients with acute intermittent porphyria. Journal of Human Genetics, 2000, 45, 263-268.	2.3	9
46	Porphyrias. British Journal of Anaesthesia, 2000, 85, 143-153.	3.4	98
47	Significant Evidence for Linkage Disequilibrium over a 5-cM Region among Afrikaners. Genomics, 2000, 66, 87-92.	2.9	61
48	Homozygous Variegate Porphyria in South Africa: Genotypic Analysis in Two Cases. Molecular Genetics and Metabolism, 2000, 69, 323-330.	1.1	29
49	Diagnosis of variegate porphyria - hard to get?. Scandinavian Journal of Clinical and Laboratory Investigation, 2000, 60, 605-610.	1.2	14
50	Uncommon Mutations and Polymorphisms in the Hemochromatosis Gene. Genetic Testing and Molecular Biomarkers, 2000, 4, 151-161.	1.7	75
51	Identification of the First Variegate Porphyria Mutation in an Indigenous Black South African and Further Evidence for Heterogeneity in Variegate Porphyria. Molecular Genetics and Metabolism, 2001, 73, 91-96.	1.1	10
52	Expression and Characterization of Six Mutations in the Protoporphyrinogen oxidase gene among Finnish Variegate Porphyria Patients. Molecular Medicine, 2001, 7, 320-328.	4.4	14
53	Mutation Screening of the Entire Coding Region of the Protoporphyrinogen Oxidase Gene Using Denaturing Gradient Gel Electrophoresis and Denaturing HPLC. Clinical Chemistry, 2001, 47, 1115-1117.	3.2	5
54	Identification of a Founder Mutation in the Protoporphyrinogen Oxidase Gene in Variegate Porphyria Patients from Chile. Human Heredity, 2001, 51, 160-168.	0.8	25

#	Article	IF	Citations
55	Homozygous variegate porphyria: a compound heterozygote with novel mutations in the protoporphyrinogen oxidase gene. British Journal of Dermatology, 2001, 144, 866-869.	1.5	28
56	Homozygous Variegate Porphyria: 20â€‰y Follow-Up and Characterization of Molecular Defect. Journal of Investigative Dermatology, 2001, 116, 610-613.	0.7	28
57	A Spectrum of Novel Mutations in the Protoporphyrinogen Oxidase Gene in 13 Families with Variegate Porphyria. Journal of Investigative Dermatology, 2001, 116, 821-823.	0.7	13
58	Diversity of Autosomal Dominant Diseases in Populations of Russia. Russian Journal of Genetics, 2001, 37, 290-301.	0.6	10
59	Analysis of Diversity of Autosomal Recessive Diseases in Populations of Russia. Russian Journal of Genetics, 2001, 37, 1312-1322.	0.6	9
60	Molecular and genealogical evidence for a founder effect in Fanconi anemia families of the Afrikaner population of South Africa. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 5734-5739.	7.1	100
61	Hereditary neuropathies. , 2002, , 1129-1142.		0
62	Human hereditary hepatic porphyrias. Clinica Chimica Acta, 2002, 325, 17-37.	1.1	66
63	Porfirias agudas: aspectos laboratoriais. Revista Brasileira De Ciencia Do Solo, 2002, 38, 249.	1.3	1
64	Isonymy and Isolation by Distance in the Netherlands. Human Biology, 2002, 74, 263-283.	0.2	26
65	Variegate porphyria in Western Australian Aboriginal patients. Internal Medicine Journal, 2002, 32, 445-450.	0.8	2
66	Clinical and biochemical characteristics and genotypeâ€“phenotype correlation in Finnishvariegate porphyria patients. European Journal of Human Genetics, 2002, 10, 649-657.	2.8	49
67	Evidence for a founder effect for pseudoxanthoma elasticum in the Afrikaner population of South Africa. Human Genetics, 2002, 111, 331-338.	3.8	35
68	Genetic analysis of Variegate Porphyria (VP) in Italy: Identification of six novel mutations in the protoporphyrinogen oxidase (PPOX) gene. Human Mutation, 2003, 21, 448-448.	2.5	6
69	Famous people and genetic disorders: From monarchs to geniuses?A portrait of their genetic illnesses. American Journal of Medical Genetics Part A, 2003, 118A, 187-196.	2.4	8
70	Nine novel mutations in the protoporphyrinogen oxidase gene in Swedish families with variegate porphyria. Clinical Genetics, 2003, 64, 122-130.	2.0	12
71	Kinetic and physical characterisation of recombinant wild-type and mutant human protoporphyrinogen oxidases. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2003, 1650, 10-21.	2.3	28
72	Physicianâ€™s Guide to the Laboratory Diagnosis of Metabolic Diseases. , 2003, , .		87

#	Article	IF	Citations
73	Acute Porphyrias: A Case Report and Review. American Journal of Psychiatry, 2003, 160, 450-459.	7.2	52
74	Coproporphyrinogen III and Protoporphyrinogen IX Oxidases. , 2003, , 75-92.		2
75	Variegate Porphyria. , 2003, , 93-120.		7
76	Porphyria: A Diagnostic Approach. , 2003, , 211-245.		13
77	Approaches to Treatment and Prevention of Human Porphyrias. , 2003, , 247-284.		5
78	Plasma Fluorescence Scanning and Fecal Porphyrin Analysis for the Diagnosis of Variegate Porphyria: Precise Determination of Sensitivity and Specificity with Detection of Protoporphyrinogen Oxidase Mutations as a Reference Standard. Clinical Chemistry, 2004, 50, 915-923.	3.2	64
79	Clinical and molecular characterization of lipoid proteinosis in Namaqualand, South Africa. British Journal of Dermatology, 2004, 151, 413-423.	1.5	98
80	A systematic study of the clinical and biochemical expression of variegate porphyria in a large South African family. British Journal of Dermatology, 2004, 151, 465-471.	1.5	33
81	Founder mutations among the Dutch. European Journal of Human Genetics, 2004, 12, 591-600.	2.8	63
82	Porphyric neuropathy. Muscle and Nerve, 2004, 30, 410-422.	2.2	128
83	Plasma fluorescence scanning did not detect latent variegate porphyria in nine patients with non-p.R59W mutations. Pathology, 2005, 37, 324-326.	0.6	1
84	Overrepresentation of the founder PPOX gene mutation R59W in a South African patient with severe clinical manifestation of porphyria. Experimental Dermatology, 2005, 14, 50-55.	2.9	3
85	Molecular mechanisms of dominant expression in porphyria. Journal of Inherited Metabolic Disease, 2005, 28, 277-286.	3.6	71
86	Circulating leptin levels in juvenile idiopathic arthritis: a marker of nutritional status?. Annals of the Rheumatic Diseases, 2005, 64, 149-152.	0.9	18
87	Life-threatening dermatoses due to metabolic and endocrine disorders. Clinics in Dermatology, 2005, 23, 258-266.	1.6	6
88	Recommendations for the Diagnosis and Treatment of the Acute Porphyrias. Annals of Internal Medicine, 2005, 142, 439.	3.9	485
90	Porphyries hÃ©patiques. EMC - HÃ©patologie, 2006, 1, 1-8.	0.0	0
91	Genetic studies in variegate porphyria in Spain. Identification of gene mutations and family study for carrier detection. Journal of the European Academy of Dermatology and Venereology, 2006, 20, 060804053334005-???.	2.4	9

#	Article	IF	Citations
92	A Chilean boy with severe photosensitivity and finger shortening: the first case of homozygous variegate porphyria in South America. British Journal of Dermatology, 2006, 154, 368-371.	1.5	18
93	Modern diagnosis and management of the porphyrias. British Journal of Haematology, 2006, 135, 281-292.	2.5	164
94	Lack of clinical manifestation of hereditary haemochromatosis in South African patients with multiple sclerosis. Metabolic Brain Disease, 2006, 21, 105-116.	2.9	13
95	Mitochondrial targeting of human protoporphyrinogen oxidase. Cell Biology International, 2006, 30, 416-426.	3.0	12
96	Biosynthesis of heme in mammals. Biochimica Et Biophysica Acta - Molecular Cell Research, 2006, 1763, 723-736.	4.1	389
97	Dual porphyria with mutations in both the UROD and HMBS genes. Annals of Clinical Biochemistry, 2006, 43, 80-82.	1.6	18
98	Crystal Structure of Protoporphyrinogen Oxidase from Myxococcus xanthus and Its Complex with the Inhibitor Acifluorfen. Journal of Biological Chemistry, 2006, 281, 38625-38633.	3.4	61
100	Functional definition of the tobacco protoporphyrinogen IX oxidase substrate-binding site. Biochemical Journal, 2007, 402, 575-580.	3.7	44
101	Heme Biosynthesis and the Porphyrias. , 0, , 677-693.		1
102	Cloning and expression of zebrafish genes encoding the heme synthesis enzymes uroporphyrinogen III synthase (UROS) and protoporphyrinogen oxidase (PPO). DNA Sequence, 2007, 18, 54-60.	0.7	3
103	Genetic and biochemical studies in Argentinean patients with variegate porphyria. BMC Medical Genetics, 2008, 9, 54.	2.1	12
104	Molecular characterization of erythropoietic protoporphyria in South Africa. British Journal of Dermatology, 2008, 159, 182-191.	1.5	33
105	montalcino, A zebrafish model for variegate porphyria. Experimental Hematology, 2008, 36, 1132-1142.	0.4	36
106	The biochemistry of heme biosynthesis. Archives of Biochemistry and Biophysics, 2008, 474, 238-251.	3.0	269
107	Familial and Sporadic Porphyria Cutanea Tarda: Characterization and Diagnostic Strategies. Clinical Chemistry, 2009, 55, 795-803.	3.2	39
108	Multiplex ligation-dependent probe amplification: a novel approach for genetic diagnosis of Porphyria. Journal of Human Genetics, 2009, 54, 479-487.	2.3	13
109	Diagnostic Strategies for Autosomal Dominant Acute Porphyrias: Retrospective Analysis of 467 Unrelated Patients Referred for Mutational Analysis of the HMBS, CPOX, or PPOX Gene. Clinical Chemistry, 2009, 55, 1406-1414.	3.2	74
110	Chapter 6 Disruption of Heme Synthesis by Polyhalogenated Aromatics. Advances in Molecular Toxicology, 2009, 3, 161-210.	0.4	4

#	Article	IF	Citations
111	Site-directed mutagenesis and computational study of the Y366 active site in Bacillus subtilis protoporphyrinogen oxidase. Amino Acids, 2009, 37, 523-530.	2.7	11
112	Understanding the Mechanism of Drug Resistance Due to a Codon Deletion in Protoporphyrinogen Oxidase through Computational Modeling. Journal of Physical Chemistry B, 2009, 113, 4865-4875.	2.6	47
113	Spectrum of genetic variation at the ABCC6 locus in South Africans: Pseudoxanthoma elasticum patients and healthy individuals. Journal of Dermatological Science, 2009, 54, 198-204.	1.9	10
114	Clinic and genetic evaluation of variegate porphyria (VP) in a large family from the Balearic Islands. Journal of Inherited Metabolic Disease, 2009, 32, 59-66.	3.6	4
115	The acute hepatic porphyrias: Current status and future challenges. Bailliere's Best Practice and Research in Clinical Gastroenterology, 2010, 24, 593-605.	2.4	52
116	Porphyrias at a glance: diagnosis and treatment. Internal and Emergency Medicine, 2010, 5, 73-80.	2.0	18
117	The porphyrias: pathophysiology. Internal and Emergency Medicine, 2010, 5, 65-71.	2.0	13
118	Structural insight into unique properties of protoporphyrinogen oxidase from Bacillus subtilis. Journal of Structural Biology, 2010, 170, 76-82.	2.8	55
119	Porphyrias. Lancet, The, 2010, 375, 924-937.	13.7	644
121	Porphyria. , 0, , 231-236.		0
122	Structural insight into human variegate porphyria disease. FASEB Journal, 2011, 25, 653-664.	0.5	54
123	A review of the clinical presentation, natural history and inheritance of variegate porphyria: its implausibility as the source of the â€˜Royal Maladyâ€™. Journal of Clinical Pathology, 2012, 65, 200-205.	2.0	28
124	Review of hepatocellular cancer, hypertension and renal impairment as late complications of acute porphyria and recommendations for patient follow-up. Journal of Clinical Pathology, 2012, 65, 976-980.	2.0	57
126	Integrative role of neuropeptides and cytokines in cancer anorexiaâ€“cachexia syndrome. Clinica Chimica Acta, 2012, 413, 1025-1034.	1.1	46
127	Establishing a network of specialist Porphyria centres - effects on diagnostic activities and services. Orphanet Journal of Rare Diseases, 2012, 7, 93.	2.7	12
128	Fifty years of porphyria at the University of Cape Town. South African Medical Journal, 2012, 102, 422.	0.6	11
129	Extended haplotype studies in South African and Dutch variegate porphyria families carrying the recurrent p.R59W mutation confirm a common ancestry. British Journal of Dermatology, 2012, 166, 261-265.	1.5	12
130	Retracing an old journey in variegate porphyria. British Journal of Dermatology, 2012, 166, 237-237.	1.5	0

#	Article	IF	Citations
131	Quantitative structureâ€“activity relationships of 1,3,4-thiadiazol-2(3H)-ones and 1,3,4-oxadiazol-2(3H)-ones as human protoporphyrinogen oxidase inhibitors. Bioorganic and Medicinal Chemistry, 2012, 20, 296-304.	3.0	35
132	Inherited Porphyrias. , 2013, , 1-32.		3
133	Mutation versus polymorphism in evolution. Genomics, 2013, 101, 211-212.	2.9	5
134	Clinical and Molecular Epidemiology of the Porphyrias. Handbook of Porphyrin Science, 2013, , 119-150.	0.8	2
135	Clinical and Laboratory Diagnosis of the Porphyrias. Handbook of Porphyrin Science, 2013, , 369-414.	0.8	1
136	Variegate Porphyria. Handbook of Porphyrin Science, 2013, , 263-298.	0.8	0
137	Role of genetic testing in the management of patients with inherited porphyria and their families. Annals of Clinical Biochemistry, 2013, 50, 204-216.	1.6	48
138	Quantitative Structural Insight into Human Variegate Porphyria Disease. Journal of Biological Chemistry, 2013, 288, 11731-11740.	3.4	37
139	The neurological manifestations of the acute porphyrias. South African Medical Journal, 2014, 104, 285.	0.6	2
140	Heme biosynthesis and the porphyrias. , 0, , 509-525.		0
141	Seven Novel Mutations in Bulgarian Patients with Acute Hepatic Porphyrias (AHP). JIMD Reports, 2014, 16, 57-64.	1.5	2
142	Mechanistic insights into the substrate recognition of PPO: toward the rational design of effective inhibitors. Future Medicinal Chemistry, 2014, 6, 597-599.	2.3	11
144	A challenging diagnosis for potential fatal diseases: Recommendations for diagnosing acute porphyrias. European Journal of Internal Medicine, 2014, 25, 497-505.	2.2	34
145	A Practical Guide to Human Cancer Genetics. , 2014, , .		8
146	Hexahydrophthalimideâ€“benzothiazole hybrids as a new class of protoporphyrinogen oxidase inhibitors: synthesis, structureâ€“activity relationship, and DFT calculations. New Journal of Chemistry, 2014, 38, 4510.	2.8	15
147	Molecular Phylogeny and Intricate Evolutionary History of the Three Isofunctional Enzymes Involved in the Oxidation of Protoporphyrinogen IX. Genome Biology and Evolution, 2014, 6, 2141-2155.	2.5	49
148	Haplotype Study in Argentinean Variegate Porphyria Patients. Human Heredity, 2015, 80, 139-143.	0.8	3
149	Porphyria Diagnosticsâ€”Part 1: A Brief Overview of the Porphyrias. Current Protocols in Human Genetics, 2015, 86, 17.20.1-17.20.26.	3.5	83

#	Article	IF	Citations
150	Understanding the genetic diversity of South Africaâ€™s peoples. South African Medical Journal, 2015, 105, 544.	0.6	1
151	Acute Hepatic Porphyria. Journal of Clinical and Translational Hepatology, 2015, 3, 17-26.	1.4	63
152	Severe porphyric neuropathy - importance of screening for porphyria in Guillain-BarrÃ© syndrome. South African Medical Journal, 2015, 106, 44.	0.6	19
153	Hepatocellular carcinoma in variegate porphyria: a case report and literature review. Annals of Clinical Biochemistry, 2015, 52, 407-412.	1.6	11
154	Complicity of haem in some adverse drug-reactions. Toxicology Research, 2015, 4, 1128-1142.	2.1	2
155	A novel FKRP-related muscular dystrophy founder mutation in South African Afrikaner patients with a phenotype suggestive of a dystrophinopathy. South African Medical Journal, 2016, 107, 80.	0.6	6
158	Acute hepatic porphyrias: Recommendations for evaluation and longâ€term management. Hepatology, 2017, 66, 1314-1322.	7.3	122
159	Update review of the acute porphyrias. British Journal of Haematology, 2017, 176, 527-538.	2.5	133
160	Porfirias agudas: enfoque diagnÃ³stico y terapÃ©utico. Acta Colombiana De Cuidado Intensivo, 2017, 17, 276-294.	0.2	0
161	Porphyrias and photosensitivity: pathophysiology for the clinician. Postgraduate Medicine, 2018, 130, 673-686.	2.0	8
162	Common and Founder Mutations for Monogenic Traits in Sub-Saharan African Populations. Annual Review of Genomics and Human Genetics, 2018, 19, 149-175.	6.2	9
163	Heme Biosynthesis and Its Disorders. , 2018, , 497-513.e6.		1
164	Recent advances on porphyria genetics: Inheritance, penetrance & molecular heterogeneity, including new modifying/causative genes. Molecular Genetics and Metabolism, 2019, 128, 320-331.	1.1	59
165	Pitfalls of relying on genetic testing only to diagnose inherited metabolic disorders in non-western populations - 5 cases of pyruvate dehydrogenase deficiency from South Africa. Molecular Genetics and Metabolism Reports, 2020, 24, 100629.	1.1	5
166	Mitochondrial molecular genetic results in a South African cohort: divergent mitochondrial and nuclear DNA findings. Journal of Clinical Pathology, 2022, 75, 34-38.	2.0	4
167	When genetic and surname analyses meet historical sources: The C56R mutation associated with factor XI deficiency as a marker of human migration during the Spanish Reconquista. Medical Hypotheses, 2020, 141, 109709.	1.5	0
168	Givosiran, a novel treatment for acute hepatic porphyrias. Expert Review of Precision Medicine and Drug Development, 2021, 6, 9-18.	0.7	12
169	Inherited Porphyrias. , 2021, , 373-411.		1

#	Article	IF	Citations
170	Disorders of porphyrin metabolism. , 2021, , 503-528.		0
171	Heme Biosynthesis and the Porphyrias in Children. , 2021, , 530-547.		0
172	The hydrogen bonding network involved Arg59 in human protoporphyrinogen IX oxidase is essential for enzyme activity. Biochemical and Biophysical Research Communications, 2021, 557, 20-25.	2.1	4
173	Monokaryotic Pleurotus sapidus Strains with Intraspecific Variability of an Alkene Cleaving DyP-Type Peroxidase Activity as a Result of Gene Mutation and Differential Gene Expression. International Journal of Molecular Sciences, 2021, 22, 1363.	4.1	14
174	Inherited Disorders of Haem Synthesis:. , 2009, , 89-100.		3
175	The Porphyrias. , 2003, , 593-613.		2
176	The Porphyrias. , 2006, , 1391-1432.		3
177	The Biology of Inherited Disorders of the Gastrointestinal Tract Part II: Pancreatic and Hepatobiliary Disorders. Journal of Pediatric Gastroenterology and Nutrition, 1998, 26, 437-445.	1.8	2
178	Diseases of the Blood. , 2000, , 673-728.		0
179	Diseases of the Blood. , 2006, , 745-779.		0
180	Diseases of the Blood. , 2006, , 745-779.		0
181	Porphyrias. , 2010, , 1077-1092.		0
183	Founder mutations among the Dutch*. , 2013, , 3-12.		2
184	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
185	Formalgenetik. , 2015, , 459-523.		0
187	Formalgenetik. , 2020, , 569-642.		0
188	Porfirias agudas: manifestaciones inespecÃficas y manejo terapÃ©utico especÃfico. Repertorio De Medicina Y Cirugia, 2020, 29, .	0.1	0
189	Human Hereditary Porphyrias. , 0, , 1343-1351.		1

#	Article	IF	Citations
190	Disorders of Erythrocyte Metabolism Including Porphyria. , 0, , 171-212.		3
191	Heterologous expression and purification of recombinant human protoporphyrinogen oxidase IX: A comparative study. PLoS ONE, 2021, 16, e0259837.	2.5	2
192	RNAi therapy with givosiran significantly reduces attack rates in acute intermittent porphyria. Journal of Internal Medicine, 2022, 291, 593-610.	6.0	11
193	Molecular characterization of a novel His333Arg variant of human protoporphyrinogen oxidase IX. Biochemical and Biophysical Research Communications, 2022, 588, 182-186.	2.1	1
194	The Porphyrias. JDDG - Journal of the German Society of Dermatology, 2022, 20, 316-331.	0.8	2
195	Die Porphyrien. JDDG - Journal of the German Society of Dermatology, 2022, 20, 316-333.	0.8	0
196	Update on the diagnosis and management of the autosomal dominant acute hepatic porphyrias. Journal of Clinical Pathology, 2022, 75, 537-543.	2.0	3
197	Development of Protoporphyrinogen IX Oxidase Inhibitors for Sustainable Agriculture. ACS Symposium Series, 0, , 11-41.	0.5	3
198	Generation and characterization of human U-2 OS cell lines with the CRISPR/Cas9-edited protoporphyrinogen oxidase IX gene. Scientific Reports, 2022, 12, .	3.3	1
199	An interaction network in Bacillus subtilis coproporphyrinogen oxidase is essential for the oxidation of protoporphyrinogen <scp>IX</scp>. Proteins: Structure, Function and Bioinformatics, 2023, 91, 1163-1172.	2.6	2
200	MANAGEMENT OF ACUTE AND CUTANEOUS PORPHYRIAS. International Journal of Clinical Practice, 2002, 56, 272-278.	1.7	47
201	Porphyric neuropathy in black South Africans: a case series. Wits Journal of Clinical Medicine, 2023, 5, .	0.0	0
202	The Biology of Inherited Disorders of the Gastrointestinal Tract Part II: Pancreatic and Hepatobiliary Disorders. Journal of Pediatric Gastroenterology and Nutrition, 1998, 26, 437-445.	1.8	0