

Porphyrias Consortium

Book Chapters

1. Anderson KE, Singal AK. Variegated Porphyria. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP, eds. *GeneReviews*. Seattle WA: University of Washington, Seattle; 1993.
2. Balwani M, Bloomer J, Desnick R. X-Linked Protoporphyrinemia. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP, eds. *GeneReviews*. Seattle WA: University of Washington, Seattle; 1993. PMID: 23409301
3. Balwani M, Bloomer J, Desnick R. Erythropoietic Protoporphyrinemia, Autosomal Recessive. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP, eds. *GeneReviews*. Seattle WA: University of Washington, Seattle; 1993. PMID: 23016163
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6. Liu LU, Phillips J, Bonkovsky H. Hepatoerythropoietic Porphyria. In: Pagon RA, Adam MP, Bird TD, et al., eds. *GeneReviews(R)*. Seattle WA: University of Washington, Seattle; 1993. PMID: 24175354
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9. Anderson K, Sood G. Acute intermittent porphyria. *Best Practice*. London, UK: BMJ Publishing Group; 2008.
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13. Sood G, Anderson K. Clinical manifestations and diagnosis of acute intermittent porphyria. In: Rose B, ed. *UpToDate*. Waltham, MA: UpToDate; 2010.

14. Sood G, Anderson K. Management of acute intermittent porphyria. In: Rose B, ed. UpToDate. Waltham, MA: UpToDate; 2010.
15. Anderson K, Lee C, Balwani M, Desnick R. The porphyrias. In: Kliegman R, Stanton B, St. Geme J, Schor N, Behrman R, eds. *Nelson Textbook of Pediatrics*. 19 ed. Philadelphia, PA: Elsevier; 2011.
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17. Anderson KE. The porphyrias. In: Goldman L, Schafer AI, eds. *Goldman's Cecil Medicine*. 24th ed. Philadelphia: Elsevier Saunders; 2012:1363-1371.
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19. Phillips JD. Side chain modification during heme biosynthesis. In: Kadish KM, Smith KM, Guillard R, eds. *Handbook of Porphyrin Science*. Vol 16: World Scientific Publishing Company; 2012:283-337.
20. Bonkovsky HL, Guo J-T, Hou W, Li T, Narang T, Thapar M. Porphyrin and Heme Metabolism and the Porphyrias. *Comprehensive Physiology*: John Wiley & Sons, Inc.; 2013:365-401.
21. Gou E, Anderson K. The Porphyrias. In: Hamblin M, Huang Y, eds. *Handbook of Photomedicine*. Boca Raton, FL: Taylor and Francis; 2013.

Abstracts Presented at Conferences

1. Anderson K. CYP1A2*1F and GSTM1 Alleles Are Associated with Susceptibility to Porphyria Cutanea Tarda. Poster presented at: International Porphyrins & Porphyrias Meeting; April 2011; Cardiff, Wales.
2. Bishop D, Tchaikovskii V, Nazarenko I, Balwani M, Doheny D, Desnick RJ. Expression and characterization of the ALAS2 carboxy-terminal gain-of-function mutations causing X-linked protoporphyria (Abstract #1157F). Paper presented at: 12th International Congress of Human Genetics/61st Annual Meeting of The American Society of Human Genetics; October 14, 2011; Montreal, Canada.
3. Doheny D, Nazarenko I, Balwani M, Liu L, Naik H, Anderson K, Bissell DM, Bloomer JR, Bonkovsky HL, Kushner JP, Phillips J, Bishop D, Desnick RJ. Erythropoietic Protoporphyrins: Frequency of Mutations in the Ferrochelatase Gene Causing Autosomal Recessive Erythropoietic Protoporphyrinemia and Mutations in the 5'-Aminolevulinic Synthase 2 Gene Causing X-Linked Protoporphyrinemia (Abstract #1338T). Paper presented at: 12th International Congress of Human Genetics/61st Annual Meeting of The American Society of Human Genetics; October 14, 2011; Montreal, Canada.
4. Desnick RJ. Overview of RDCRN and Porphyrias Consortium Activities. Paper presented at: AASLD Annual Meeting; November 6, 2011; San Francisco, CA.
5. Wang B. PTF Experience from the Trainee Perspective. Paper presented at: AASLD Annual Meeting; November 6, 2011; San Francisco, CA.
6. Anderson K. The cutaneous porphyrias. Paper presented at: AASLD Annual Meeting; November 6, 2011; San Francisco, CA.

7. Desnick RJ. The Porphyrins: Cardinal Signs and Diagnosis/Treatment. Paper presented at: American College of Medical Genetics Annual Clinical Genetics Meeting; March 31, 2012; Charlotte, NC.
8. Singh A, Pierson K, Wilkinson G, Anderson K. Porphyrins: prevalence and frequency of testing in a national health care database. Paper presented at: Annual Meeting of the American Association for the Study of Liver Disease (AASLD); November 9-13, 2012; Boston, MA.
9. Balwani M, Desnick RJ. The Porphyrins: Advances in Diagnosis and Treatment. Paper presented at: American Society of Hematology, Educational Program; December 8-10, 2012; New Orleans, LA. PMID: 23233556
10. Ludtke A, Yasuda M, Balwani M, et al. First US Orthotopic Liver Transplant for Intractable Acute Intermittent Porphyria. *The American Society of Human Genetics 63rd Annual Meeting*. Boston, MA2013.
11. Ludtke A, Yasuda M, Lin G, et al. Acute Intermittent Porphyria: Identification of 23 Novel Hydroxymethylbilane Synthase Mutations and Functional Characterization of Six Novel Missense Mutations. *ACMG Annual Clinical Genetics Meeting*. Phoenix, AZ2013.
12. Balwani M, Bishop D, Nazarenko I, et al. Mutation analysis of 155 North American Patients with Erythropoietic Protoporphyrinemia reveals novel Ferrochelatase Mutations and a high prevalence of X-Linked Protoporphyrinemia due to previous and novel 5-Aminolevulinic Acid Synthase 2 mutations. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
13. Balwani M, Naik H, Peter I, et al. Erythropoietic Protoporphyrinemia and X-Linked Protoporphyrinemia in the United States: Results from the Longitudinal Study of the NIH/RDCRN Porphyrin Consortium. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
14. Bishop D, Tchaikovskii V, Nazarenko I, Desnick R. Synthase Gain-of-Function Mutations Causing X-Linked Protoporphyrinemia. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
15. Gou E, Singh A, Pierson K, Wilkinson G, Anderson K. Frequency of Porphyria Testing in a National Health Care Database. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
16. Larion S, Caballes F, Hwang S, et al. Circadian rhythms in acute intermittent porphyria—a pilot study. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
17. Ludtke A, Yasuda M, Gan L, et al. Acute Intermittent Porphyria: Identification of 19 Novel Hydroxymethylbilane Synthase Mutations and Functional Characterization of Four Novel

Missense Mutations. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.

18. Maddukuri V, Yazici C, Anderson K, et al. Acute intermittent porphyria [AIP] in the United States: features of the first 82 cases enrolled in the longitudinal study of the porphyria consortium [PC]. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
19. Mittal S, Yasuda M, Desnick R, Anderson K. Metabolic Analysis in Transgenic Mouse Models of Acute Intermittent Porphyria (AIP). Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
20. Naik H, Balwani M, Doheny D, Liu L, Desnick R. Experience with a Pilot Skype Internet Support Group for Symptomatic Patients with Acute Intermittent Porphyria. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
21. Phillips J, Warby C, Bergonia H, Marcero J, Parker C, Franklin M. Porphyria studies in Cyp1A2^{-/-} and wild type mice suggest that heme regulation of ALA-synthase transcription and mitochondrial membrane translocation can be separated based on heme supply-and-demand. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
22. Singal A, Jampana S, Kormos-Hallberg C, Anderson K. Low-dose hydroxychloroquine to treat or prevent relapse of porphyria cutanea tarda during hepatitis C treatment. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
23. Singal A, Gou E, Albuerne M, Kormos-Hallberg C, Anderson K. Relapse of porphyria cutanea tarda after achieving remission with phlebotomy or low dose hydroxychloroquine. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
24. Yazici C, Maddukuri V, Anderson K, et al. Hereditary coproporphyria [HCP] and variegated porphyria [VP] in the United States: Initial results from the longitudinal study of the porphyria consortium [PC]. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.
25. Wang B, Bissell D, Lai J, Cimino T, PorphyriasConsortium. A Combined Clinical Index for the Diagnosis of Acute Porphyria. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrins; May 16-18, 2013; Lucerne, Switzerland.

Conference Proceedings

1. Hou W, Tian Q, Lu QL, Schrum LW, Bonkovsky HL. Zinc protoporphyrin, a novel endogenous HCV NS3-4A protease inhibitor, displays anti-viral activity. [Abstract #926 Presidential Poster of Distinction, presented at Annual Meeting of the American Association for the Study of Liver Diseases]. *Hepatology*. 2011;54(S1):128A-359A.
2. Hwang SI, Lee YY, Park JO, Norton HJ, Clemens E, Schrum LW, Bonkovsky HL. The measurement of hepcidin from human urine and serum to study effects of a single dose of oral iron by an optimized LC-MS/MS method. [Abstract #1204 presented at Annual Meeting of the American Association for the Study of Liver Diseases]. *Hepatology*. 2011;54(S1):931A.
3. Tian Q, Hou W, Steuerwald NM, Schrum LW, Bonkovsky HL. Heme markedly up-regulates RNA-binding motif protein 24 gene expression in human hepatocytes. [Abstract #895 presented at the Annual Meeting of the American Association for the Study of Liver Diseases]. *Hepatology*. 2011;54(S1):780A.
4. Tian Q, Hou W, Zheng J, Schrum LW, Bonkovsky HL. LONP1-dependent breakdown of mitochondrial 5-aminolevulinic acid synthase protein by heme in human liver cells. [Abstract #904 presented at the Annual Meeting of the American Association for the Study of Liver Diseases]. *Hepatology*. 2011;54(S1):785A.

Journal Articles

1. Jalil S, Grady JJ, Lee C, Anderson KE. Associations among behavior-related susceptibility factors in porphyria cutanea tarda. *Clin. Gastroenterol. Hepatol.* Mar 2010;8(3):297-302, 302 e291. PMID: PMC2834813
2. Hou W, Tian Q, Zheng J, Bonkovsky HL. Zinc mesoporphyrin induces rapid proteasomal degradation of hepatitis C nonstructural 5A protein in human hepatoma cells. *Gastroenterology*. May 2010;138(5):1909-1919. PMID: PMC2860067
3. Gunn GB, Anderson KE, Patel AJ, et al. Severe radiation therapy-related soft tissue toxicity in a patient with porphyria cutanea tarda: a literature review. *Head Neck*. Aug 2010;32(8):1112-1117. PMID: PMC2891307
4. Dailey HA, Septer AN, Daugherty L, Thames D, Gerdes S, Stabb EV, Dunn AK, Dailey TA, Phillips JD. The *Escherichia coli* protein YfeX functions as a porphyrinogen oxidase, not a heme dechelataase. *MBio*. 2011;2(6):e00248-00211. PMID: PMC3215433
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7. Machaczka M, Klimkowska M, Regenthal S, Hagglund H. Gaucher disease with foamy transformed macrophages and erythrophagocytic activity. *J. Inherit. Metab. Dis.* Feb 2011;34(1):233-235. PMID: 21113739

8. Hasanoglu A, Balwani M, Kasapkara CS, Ezgu FS, Okur I, Tumer L, Cakmak A, Nazarenko I, Yu C, Clavero S, Bishop DF, Desnick RJ. Harderoporphyria due to homozygosity for coproporphyrinogen oxidase missense mutation H327R. *J. Inherit. Metab. Dis.* Feb 2011;34(1):225-231. PMID: PMC3091031
9. Wickliffe JK, Abdel-Rahman SZ, Lee C, Kormos-Hallberg C, Sood G, Rondelli CM, Grady JJ, Desnick RJ, Anderson KE. CYP1A2*1F and GSTM1 alleles are associated with susceptibility to porphyria cutanea tarda. *Mol. Med.* Mar-Apr 2011;17(3-4):241-247. PMID: PMC3060985
10. Lorenzo FRt, Phillips JD, Nussenzveig R, Lingam B, Koul PA, Schrier SL, Prchal JT. Molecular basis of two novel mutations found in type I methemoglobinemia. *Blood Cells. Mol. Dis.* Apr 15 2011;46(4):277-281. PMID: PMC3075332
11. Troadec MB, Warner D, Wallace J, Thomas K, Spangrude GJ, Phillips J, Khalimonchuk O, Paw BH, Ward DM, Kaplan J. Targeted deletion of the mouse Mitoferrin1 gene: from anemia to protoporphyria. *Blood.* May 19 2011;117(20):5494-5502. PMID: PMC3109720
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15. Tian Q, Li T, Hou W, Zheng J, Schrum LW, Bonkovsky HL. Lon peptidase 1 (LONP1)-dependent breakdown of mitochondrial 5-aminolevulinic acid synthase protein by heme in human liver cells. *J. Biol. Chem.* Jul 29 2011;286(30):26424-26430. PMID: PMC3143606
16. Zhang J, Yasuda M, Desnick RJ, Balwani M, Bishop D, Yu C. A LC-MS/MS method for the specific, sensitive, and simultaneous quantification of 5-aminolevulinic acid and porphobilinogen. *J. Chromatogr. B Analyt. Technol. Biomed. Life. Sci.* Aug 15 2011;879(24):2389-2396. PMID: PMC3269068
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18. Huang Z, Chen K, Xu T, Zhang J, Li Y, Li W, Agarwal AK, Clark AM, Phillips JD, Pan X. Sampangine inhibits heme biosynthesis in both yeast and human. *Eukaryot. Cell.* Nov 2011;10(11):1536-1544. PMID: PMC3209050
19. Hwang SI, Lee YY, Park JO, Norton HJ, Clemens E, Schrum LW, Bonkovsky HL. Effects of a single dose of oral iron on hepcidin concentrations in human urine and serum analyzed by a robust LC-MS/MS method. *Clin. Chim. Acta.* Nov 20 2011;412(23-24):2241-2247. PMID: PMC3207492
20. Phillips JD, Kushner JP, Bergonia HA, Franklin MR. Uroporphyrin in the Cyp1a2^{-/-} mouse. *Blood Cells. Mol. Dis.* Dec 15 2011;47(4):249-254. PMID: PMC3223295

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22. Ryan Caballes F, Sendi H, Bonkovsky HL. Hepatitis C, porphyria cutanea tarda and liver iron: an update. *Liver Int.* Jul 2012;32(6):880-893. PMCID: PMC3418709
23. Balwani M, Desnick RJ. The porphyrias: advances in diagnosis and treatment. *Blood.* Nov 29 2012;120(23):4496-4504. PMCID: PMC3512229
24. Singal AK, Kormos-Hallberg C, Lee C, Sadagoparamanujam VM, Grady JJ, Freeman DH, Jr., Anderson KE. Low-dose hydroxychloroquine is as effective as phlebotomy in treatment of patients with porphyria cutanea tarda. *Clin. Gastroenterol. Hepatol.* Dec 2012;10(12):1402-1409. PMCID: PMC3501544
25. Bishop DF, Tchaikovskii V, Nazarenko I, Desnick RJ. Molecular expression and characterization of erythroid-specific 5-aminolevulinic synthase gain-of-function mutations causing X-linked protoporphyria. *Mol. Med.* 2013;19:18-25. PMCID: PMC3592931
26. Balwani M, Doheny D, Bishop DF, et al. Loss-of-function ferrochelatase and gain-of-function erythroid-specific 5-aminolevulinic synthase mutations causing erythropoietic protoporphyria and x-linked protoporphyria in North American patients reveal novel mutations and a high prevalence of X-linked protoporphyria. *Mol. Med.* 2013;19:26-35. PMCID: PMC3646094
27. Bonkovsky HL. Risk factors for porphyria cutanea tarda -the iron/HFE connection. *Liver Int.* Jan 2013;33(1):162. PMID: 23121614
28. Bonkovsky HL, Hou W, Steuerwald N, et al. Heme status affects human hepatic messenger RNA and microRNA expression. *World J. Gastroenterol.* Mar 14 2013;19(10):1593-1601. PMID: 23538684, PMCID: PMC3602476
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31. Yasuda M, Gan L, Chen B, et al. RNAi-mediated silencing of hepatic *Alas1* effectively prevents and treats the induced acute attacks in acute intermittent porphyria mice. *Proc. Natl. Acad. Sci. U. S. A.* May 27 2014;111(21):7777-7782. PMID: 24821812, PMCID: PMC4040563
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34. Bonkovsky HL, Maddukuri VC, Yazici C, et al. Acute porphyrias in the USA: features of 108 subjects from porphyrias consortium. *Am. J. Med.* Dec 2014;127(12):1233-1241. PMID: 25016127
35. Bossi K, Lee J, Schmeltzer P, et al. Homeostasis of iron and hepcidin in erythropoietic protoporphyria. *Eur. J. Clin. Invest.* 2015;45(10):1032-1041. PMID: 26199063

36. Medlock AE, Shiferaw MT, Marcero JR, et al. Identification of the Mitochondrial Heme Metabolism Complex. *PLoS ONE*. 2015;10(8):e0135896. PMID: 26287972, PMCID: PMC4545792
37. Ramanujam VM, Anderson KE. Porphyrin Diagnostics-Part 1: A Brief Overview of the Porphyrins. *Current protocols in human genetics / editorial board, Jonathan L Haines [et al]*. 2015;86:17.20.11-26. PMID: 26132003, PMCID: PMC4640448
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39. Yasuda M, Erwin AL, Liu LU, et al. Liver Transplantation for Acute Intermittent Porphyrin: Biochemical and Pathologic Studies of the Explanted Liver. *Mol. Med*. 2015;21:487-495. PMID: 26062020, PMCID: PMC4607616
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Special Projects

1. Bloomer JR. Managing acute porphyrias: practice considerations in inpatient and outpatient settings. *Medscape Education Gastroenterology*. 2010. <http://www.medscape.org/viewarticle/730948>. Accessed February 28, 2013.
2. Anderson KE. Porphyrin-an overview. In: Rose BD, ed. *UpToDate*. Waltham, MA: UpToDate; 2010.
3. Sood G, Anderson KE. Acute intermittent porphyria. In: Rose BD, ed. *UpToDate*. Waltham, MA: UpToDate; 2010.

4. Singal A, Anderson KE. Porphyria cutanea tarda and hepatoerythropoietic porphyria. In: Rose BD, ed. *UpToDate*. Waltham, MA: UpToDate; 2010.
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6. Mittal S, Anderson KE. Erythropoietic porphyria. In: Rose BD, ed. *UpToDate*. Waltham, MA: UpToDate; 2011.
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