Porphyrias Consortium

Book Chapters

1. Anderson KE, Singal AK. Variegate 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 Protoporphyria. 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 Protoporphyria, Autosomal Recessive. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP, eds. *GeneReviews*. Seattle WA: University of Washington, Seattle; 1993. PMID: 23016163
4. Bissell DM, Wang B, Cimino T, Lai J. Hereditary Coproporphyria. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP, eds. *GeneReviews*. Seattle WA: University of Washington, Seattle; 1993. PMID: 23236641
5. Erwin A, Balwani M, Desnick RJ. Congenital Erythropoietic Porphyria. In: Pagon RA, Adam MP, Bird TD, et al., eds. GeneReviews(R). Seattle WA: University of Washington, Seattle; 1993. PMID: 24027798
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
7. Liu LU, Phillips J, Bonkovsky H. Porphyria Cutanea Tarda, Type II. In: Pagon RA, Adam MP, Bird TD, et al., eds. GeneReviews(R). Seattle WA: University of Washington, Seattle; 1993. PMID: 23741761
8. Phillips JD, Kushner JP. The Porphyrias. In: Orkin SH, Nathan DG, Ginsburg D, Look AT, Fisher DE, Lux SE, eds. *Nathan and Oski's Hematology of Infancy and Childhood*. 7th ed. Philadelphia: Saunders 2008.
9. Anderson K, Sood G. Acute intermittent porphyria. *Best Practice*. London, UK: BMJ Publishing Group; 2008.
10. Sood G, Anderson K. Porphyria Cutanea Tarda. *Best Practice*. London, UK: BMJ Publishing Group; 2008.
11. Sood G, Anderson K. Porphyrias. In: Crowther M, Ginsberg J, Schunemann H, Meyer R, Lottenberg R, eds. *Evidence-Based Hematology*. United Kingdom: Blackwell Publishing Ltd; 2008:229-237.
12. Phillips JD, Anderson KE. The Porphyrias. In: Kaushansky K, Buetler E, Seligsohn U, Lichtman MA, Kipps TJ, Prchal JT, eds. *Williams Hematology*. 8th ed. New York: McGraw-Hill Medical; 2010:839-863.
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.

16. Phillips JD, Anderson K. The Porphyrias. In: Lichtman MA, Kaushansky K, Kipps TJ, Prchal JT, Levi MM, eds. *Williams Manual of Hematology*. 8th ed: McGraw-Hill Professional; 2011.

17. Anderson KE. The porphyrias. In: Goldman L, Schafer AI, eds. *Goldman's Cecil Medicine*. 24th ed. Philadelphia: Elsevier Saunders; 2012:1363-1371.

18. Lourenco CM, Lee C, Anderson KE. Disorders of heme biosynthesis In: J-M S, Van den Berghe G, Walter JH, eds. *Inborn Metabolic Diseases: Diagnosis and Treatment*. 5th ed. Berlin: Springer-Verlag; 2012:519-532.

19. Phillips JD. Side chain modification during heme biosynthesis. In: Kadish KM, Smith KM, Guilard 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 Protoporphyrias: Frequency of Mutations in the Ferrochelatase Gene Causing Autosomal Recessive Erythropoietic Protoporphyria and Mutations in the 5'-Aminolevulinate Synthase 2 Gene Causing X-Linked Protoporphyria (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 Porphyrias: 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. Porphyrias: 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 Porphyrias: 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 Protoporphyria reveals novel Ferrochelatase Mutations and a high prevalence of X-Linked Protoporphyria due to previous and novel 5-Aminolevulinate Synthase 2 mutations. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrias; May 16-18, 2013; Lucerne, Switzerland.
13. Balwani M, Naik H, Peter I, et al. Erythropoietic Protoporphyria and X-Linked Protoporphyria in the United States: Results from the Longitudinal Study of the NIH/RDCRN Porphyrias Consortium. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrias; May 16-18, 2013; Lucerne, Switzerland.
14. Bishop D, Tchaikovskii V, Nazarenko I, Desnick R. Synthase Gain-of-Function Mutations Causing X-Linked Protoporphyria. Paper presented at: Annual Assembly of the Swiss Society of Clinical Chemistry & International Congress of Porphyrins and Porphyrias; 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 Porphyrias; 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 Porphyrias; 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 Porphyrias; 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 Porphyrias; 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 Porphyrias; 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 Porphyrias; 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 Porphyrias; 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 Porphyrias; 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 Porphyrias; May 16-18, 2013; Lucerne, Switzerland.

24. Yazici C, Maddukuri V, Anderson K, et al. Hereditary coproporphyria [HCP] and variegate 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 Porphyrias; 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 Porphyrias; 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-aminolevulinicacid 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. PMCID: 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. PMCID: 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. PMCID: 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 dechelatase. *MBio.* 2011;2(6):e00248-00211. PMCID: PMC3215433
5. Li T, Bonkovsky HL, Guo JT. Structural analysis of heme proteins: implications for design and prediction. *BMC Struct. Biol.* 2011;11:13. PMCID: PMC3059290
6. Bishop DF, Clavero S, Mohandas N, Desnick RJ. Congenital erythropoietic porphyria: characterization of murine models of the severe common (C73R/C73R) and later-onset genotypes. *Mol. Med.* 2011;17(7-8):748-756. PMCID: PMC3146604
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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC3109720

12. Lakner AM, Bonkovsky HL, Schrum LW. microRNAs: fad or future of liver disease. *World J. Gastroenterol.* May 28 2011;17(20):2536-2542. PMCID: PMC3103811

13. Boynton TO, Gerdes S, Craven SH, Neidle EL, Phillips JD, Dailey HA. Discovery of a gene involved in a third bacterial protoporphyrinogen oxidase activity through comparative genomic analysis and functional complementation. *Appl. Environ. Microbiol.* Jul 2011;77(14):4795-4801. PMCID: PMC3147383

14. Wang Y, Langer NB, Shaw GC, Yang G, Li L, Kaplan J, Paw BH, Bloomer JR. Abnormal mitoferrin-1 expression in patients with erythropoietic protoporphyria. *Exp. Hematol.* Jul 2011;39(7):784-794. PMCID: PMC3143264

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. PMCID: 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. PMCID: PMC3269068

17. To-Figueras J, Phillips JD, Gonzalez-Lopez JM, Badenas C, Madrigal I, Gonzalez-Romaris EM, Ramos C, Aguirre JM, Herrero C. Hepatoerythropoietic porphyria due to a novel mutation in the uroporphyrinogen decarboxylase gene. *Br. J. Dermatol.* Sep 2011;165(3):499-505. PMCID: PMC3818800

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. PMCID: 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. PMCID: PMC3207492

20. Phillips JD, Kushner JP, Bergonia HA, Franklin MR. Uroporphyria in the Cyp1a2-/- mouse. *Blood Cells. Mol. Dis.* Dec 15 2011;47(4):249-254. PMCID: PMC3223295

21. Wang L, He F, Bu J, Liu X, Du W, Dong J, Cooney JD, Dubey SK, Shi Y, Gong B, Li J, McBride PF, Jia Y, Lu F, Soltis KA, Lin Y, Namburi P, Liang C, Sundaresan P, Paw BH, Li DY, Phillips JD, Yang Z. ABCB6 mutations cause ocular coloboma. *Am. J. Hum. Genet.* Jan 13 2012;90(1):40-48. PMCID: PMC3257322

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-aminolevulinate 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-aminolevulinate 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

29. Larion S, Caballes FR, Hwang SI, et al. Circadian rhythms in acute intermittent porphyria--a pilot study. *Eur. J. Clin. Invest.* Jul 2013;43(7):727-739. PMID: 23650938, PMC: PMC3687345

30. Clavero S, Ahuja Y, Bishop DF, et al. Diagnosis of feline acute intermittent porphyria presenting with erythrodontia requires molecular analyses. *Vet. J.* Dec 2013;198(3):720-722. PMID: 24239138, PMCID: PMC3963809

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

32. Singal AK, Parker C, Bowden C, Thapar M, Liu L, McGuire BM. Liver transplantation in the management of porphyria. Hepatology. Sep 2014;60(3):1082-1089. PMID: 24700519

33. Yien YY, Robledo RF, Schultz IJ, et al. TMEM14C is required for erythroid mitochondrial heme metabolism. *J. Clin. Invest.* Oct 2014;124(10):4294-4304. PMID: 25157825, PMCID: PMC4191016

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. O'Brien TR, Pfeiffer RM, Paquin A, et al. Comparison of functional variants in IFNL4 and IFNL3 for association with HCV clearance. *J Hepatol.* 2015;63(5):1103-1110. PMID: 26186989, PMCID: PMC4615534

38. Ramanujam VM, Anderson KE. Porphyria Diagnostics-Part 1: A Brief Overview of the Porphyrias. *Current protocols in human genetics / editorial board, Jonathan L Haines [et al].* 2015;86:17.20.11-26. PMID: 26132003, PMCID: PMC4640448

39. Wang G, Bonkovsky HL, de Lemos A, Burczynski FJ. Recent insights into the biological functions of liver fatty acid binding protein 1. *J Lipid Res.* 2015;56(12):2238-2247. PMID: 26443794, PMCID: PMC4655993

40. Yasuda M, Erwin AL, Liu LU, et al. Liver Transplantation for Acute Intermittent Porphyria: Biochemical and Pathologic Studies of the Explanted Liver. *Mol. Med.* 2015;21:487-495. PMID: 26062020, PMCID: PMC4607616

41. Dailey HA, Gerdes S, Dailey TA, Burch JS, Phillips JD. Noncanonical coproporphyrin-dependent bacterial heme biosynthesis pathway that does not use protoporphyrin. *Proc. Natl. Acad. Sci. U. S. A.* Feb 17 2015;112(7):2210-2215. PMID: 25646457, PMCID: PMC4343137

42. Bissell DM, Lai JC, Meister RK, Blanc PD. Role of Delta-aminolevulinic Acid in the Symptoms of Acute Porphyria. *Am. J. Med.* Mar 2015;128(3):313-317. PMID: 25446301, PMCID: PMC4339446

43. Langendonk JG, Balwani M, Anderson KE, et al. Afamelanotide for Erythropoietic Protoporphyria. *N. Engl. J. Med.* Jul 2 2015;373(1):48-59. PMID: 26132941

44. Bergonia HA, Franklin MR, Kushner JP, Phillips JD. A method for determining delta-aminolevulinic acid synthase activity in homogenized cells and tissues. *Clin. Biochem.* Aug 2015;48(12):788-795. PMID: 25959086, PMCID: PMC4522353

45. Farrell CP, Parker CJ, Phillips JD. Exome sequencing for molecular characterization of non-HFE hereditary hemochromatosis. *Blood Cells. Mol. Dis.* Aug 2015;55(2):101-103. PMID: 26142323, PMCID: PMC4491409

46. Gou EW, Balwani M, Bissell DM, et al. Pitfalls in Erythrocyte Protoporphyrin Measurement for Diagnosis and Monitoring of Protoporphyrias. *Clin. Chem.* Dec 2015;61(12):1453-1456. PMID: 26482161, PMCID: PMC4744648

47. Balwani M, Singh P, Seth A, et al. Acute Intermittent Porphyria in children: A case report and review of the literature. *Mol Genet Metab.* 2016;119(4):295-299. PMID: 27769855, PMCID: PMC5154763

48. Chen B, Solis-Villa C, Hakenberg J, et al. Acute Intermittent Porphyria: Predicted Pathogenicity of HMBS Variants Indicates Extremely Low Penetrance of the Autosomal Dominant Disease. *Hum Mutat.* 2016;37(11):1215-1222. PMID: 27539938, PMCID: PMC5063710

49. Merkel PA, Manion M, Gopal-Srivastava R, et al. The partnership of patient advocacy groups and clinical investigators in the rare diseases clinical research network. *Orphanet J. Rare Dis.* 2016;11(1):66. PMID: 27194034, PMCID: PMC4870759

50. Naik H, Stoecker M, Sanderson SC, Balwani M, Desnick RJ. Experiences and concerns of patients with recurrent attacks of acute hepatic porphyria: A qualitative study. *Mol Genet Metab.* 2016. PMID: 27595545

51. Piel RB, 3rd, Shiferaw MT, Vashisht AA, et al. A Novel Role for Progesterone Receptor Membrane Component 1 (PGRMC1): A Partner and Regulator of Ferrochelatase. *Biochemistry (Mosc).* 2016;55(37):5204-5217. PMID: 27599036, PMCID: PMC5278647

52. French JB, Bonacini M, Ghabril M, Foureau D, Bonkovsky HL. Hepatotoxicity Associated with the Use of Anti-TNF-alpha Agents. *Drug Saf.* Mar 2016;39(3):199-208. PMID: 26692395, PMCID: PMC4752395

53. Balwani M, Wang B, Anderson KE, et al. Acute Hepatic Porphyrias: Recommendations for Evaluation and Long Term Management. *Hepatology.* 2017. PMID: 28605040

54. Singal AK, Venkata KVR, Jampana S, Islam FU, Anderson KE. Hepatitis C Treatment in Patients With Porphyria Cutanea Tarda. *Am J Med Sci.* 2017;353(6):523-528. PMID: 28641714

55. Yien YY, Ducamp S, van der Vorm LN, et al. Mutation in human CLPX elevates levels of delta-aminolevulinate synthase and protoporphyrin IX to promote erythropoietic protoporphyria. *Proc Natl Acad Sci U S A.* 2017;114(38):E8045-e8052. PMID: 28874591, PMCID: PMC5617249

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. Porphyria-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.

5. Singal A, Anderson KE. Variegate porphyria. In: Rose BD, ed. *UpToDate*. Waltham, MA: UpToDate; 2010.

6. Mittal S, Anderson KE. Erythropoietic porphyria. In: Rose BD, ed. *UpToDate*. Waltham, MA: UpToDate; 2011.

7. Mittal S, Anderson KE. Congenital erythropoietic porphyria. In: Rose BD, ed. *UpToDate*. Waltham, MA: UpToDate; 2013.

8. Anderson KE, Singal A. Variegate porphyria In: Pagon RA, Bird TD, Dolan CR, et al., eds. *GeneReviews [Internet].* Vol 1993-. Seattle, Washington: University of Washington, Seattle; 2013: http://www.ncbi.nlm.nih.gov/books/NBK121283/. PMID: 23409300

9. Bloomer JR. Porphyria, ALA-D. 2010; http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/324/viewAbstract. Accessed February 29, 2013.

10. Anderson KE. Porphyria, variegate. 2010; http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/324/viewAbstract. Accessed April 4, 2014.