Stanford



Hetanshi Naik

Associate Professor (Teaching) of Genetics and, by courtesy, of Pediatrics

Bio

BIO

Hetanshi Naik is an Associate Professor in the Department of Genetics and the Research Director of the MS Program in Human Genetics and Genetic Counseling. She is a board certified genetic counselor and clinical researcher with clinical expertise in the inborn errors of heme biosynthesis, the Porphyrias, lysosomal storage disorders (LSDs), and pharmacogenomics, and research expertise in clinical trials, patient reported outcomes (PROs), qualitative methods, and study design.

Her research interests include developing and evaluating PROs for genetic disorders and genomics, in particular assessing PROs as outcomes for clinical trials, pharmacogenomics implementation, and genetic counseling education and processes, as well as utilizing digital health technologies to improve clinical care, genetic counseling, patient reporting, trial efficacy, and outcomes.

ACADEMIC APPOINTMENTS

- Associate Professor-Teaching, Genetics
- Associate Professor-Teaching (By courtesy), Pediatrics - Medical Genetics
- Member, Maternal & Child Health Research Institute (MCHRI)

ADMINISTRATIVE APPOINTMENTS

- Research Director, MS Program in Human Genetics and Genetic Counseling, Stanford University School of Medicine, (2022- present)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Research Faculty Working Group, Association of Genetic Counseling Program Directors (2022 - present)
- Board of Directors, United Porphyrias Association (2021 - present)
- Member, Rare Disease Diversity Coalition (2021 - present)
- Diversity Committee, Rare Diseases Clinical Research Network (2021 - 2025)
- Founding Member, American Porphyrias Expert Collaborative (APEX) (2020 - present)
- Data Standards Committee, Rare Diseases Clinical Research Network (2020 - 2024)

PROFESSIONAL EDUCATION

- PhD, Icahn School of Medicine at Mount Sinai , Clinical Research (2018)
- MS, Mount Sinai School of Medicine , Genetic Counseling (2010)
- BSc, McMaster University , Biology major, Genetics specialization (2008)

Teaching

COURSES

2026-27

- Genetic Counseling Research Seminar: GENE 282A (Win)
- Genetic Counseling Research Seminar: GENE 282B (Spr)
- Patient-Centered Care and Genetic Therapies for Rare Diseases: GENE 294 (Spr)

2025-26

- Genetic Counseling Research Seminar: GENE 282A (Win)
- Genetic Counseling Research Seminar: GENE 282B (Spr)
- Patient-Centered Care and Genetic Therapies for Rare Diseases: GENE 294 (Spr)

2024-25

- Genetic Counseling Research Seminar: GENE 282A (Win)
- Genetic Counseling Research Seminar: GENE 282B (Spr)
- Patient-Centered Care and Genetic Therapies for Rare Diseases: BIOS 426 (Spr)

2023-24

- Genetic Counseling Research Seminar: GENE 282A (Win)
- Genetic Counseling Research Seminar: GENE 282B (Spr)

STANFORD ADVISEES

Master's Program Advisor

Ria Datwani, Sammie Harker, Annie Heckler, Kira Kopec, Clarissa Otto, Elly Qunell, Carly Sauter, Elizabeth Troll, Samantha Tsai, Ines de Valle Diego

Publications

PUBLICATIONS

- **The psychosocial impact of erythropoietic protoporphyria in childhood: a guide for parents.** *Molecular genetics and metabolism*
  Gjestson, K., McGinty, M., Williams, L. R., Leppert, C., Wheeden, K., Naik, H.
  2026; 149 (3): 110293
- **The burden of an ultra-rare inherited skin disorder: Congenital erythropoietic porphyria.** *Molecular genetics and metabolism*
  Naik, H., Huang, S. D., Williams, L. R., Wheeden, K., Erwin, A. L.
  2026; 149 (3): 110290
- **Needs, Challenges, and Opportunities for Genetic Counselors in Vietnam: A Qualitative Interview Study With Healthcare Leaders and Genetics Professionals.** *Journal of genetic counseling*
  Burke, M. L., Abad, P. J., M'Barek, B., Raney, A. M., Bruce, J. S., Bereknyei Merrell, S., Naik, H., Ormond, K. E.
  2026; 35 (5): e70303
- **Clinical course and biomarkers in patients with type 1 Gaucher disease and Parkinson's disease: A retrospective cohort study** *MOLECULAR GENETICS AND METABOLISM REPORTS*
  Iverson, A., Sinha, D. B., Fierro, L., Stauffer, C., Naik, H., Alcalay, R. N., Balwani, M.
  2026; 49
- **Erythropoietic protoporphyria life impact and genetic health trajectory (LIGHT) study in Europe: a cross-sectional online questionnaire.** *Orphanet journal of rare diseases*
  Naik, H., Wheeden, K., Mathias, S. D., Norregaard, C., Amaefule, A. Q., Chin, M., Balwani, M.
  2026; 21 (1)

- **Introduction to the Special Issue on Research Methods in Genetic Counseling: Diverse Methods, Shared Purpose.** *Journal of genetic counseling*
  Myers, M. F., Naik, H., Wainstein, T.
  2026; 35 (4): e70261
- **Knowledge and Attitudes on Germline Pharmacogenomic Testing Among Clinical Oncologists.** *JCO precision oncology*
  Monroe, M., Naik, H., Ford, J. M., Whirl-Carrillo, M., Klein, T. E., Hertz, D. L., Scott, S. A.
  2026; 10 (8): e2600353
- **Trauma-Informed Care: An Investigation Into Knowledge, Attitudes, and Practices Among Genetic Counselors.** *Journal of genetic counseling*
  Chandler, L., Naik, H., Kletter, H., Farrelly, E., Smith, E. E.
  2026; 35 (4): e70273
- **Future-use consent and human virome research: ethical challenges.** *Journal of virology*
  de Melo-Martín, I., Naik, H., Yracheta, J., Li-Pook-Than, J.
  2026: e0042026
- **Complementary and alternative medicines and cannabis use among individuals with acute intermittent porphyria.** *Molecular genetics and metabolism*
  Tomasdottir, A., Hedstrom, K., Overbey, J. R., McDonough, C., Balwani, M., Naik, H.
  2026; 148 (4): 110203
- **Optimizing pharmacogenomics workflows: a qualitative evaluation of clinical service delivery models.** *Pharmacogenomics*
  Huang, E., Obeng, A. O., Stuart, S. A., Naik, H.
  2026: 1-7
- **Knowledge and perspectives on pharmacogenomic-guided antidepressant treatment among psychiatrists and primary care practitioners.** *Pharmacogenetics and genomics*
  Schmidt, K. T., Naik, H., Nguyen, T. P., Bishop, J. R., Bousman, C. A., Singh, A. B., Klein, T. E., Scott, S. A.
  2026
- **Accessibility and harmonization of biochemical tests for diagnosis and monitoring of Porphyrias in the United States: Recommendations by members of the American Porphyrias Expert Collaborative (APEX).** *Molecular genetics and metabolism*
  Anderson, K. E., Desnick, R. J., Bonkovsky, H. L., Balwani, M., Barnes, S., Beaven, S. W., Yeung, A. K., Erwin, A. L., Freeman, M., Huda, R., Kazamel, M., Leaf, R. K., Keel, et al
  2026; 148 (4): 110185
- **Inclusive Fertility: Investigating the Lack of Diversity in Sperm Donation.** *Journal of racial and ethnic health disparities*
  Anderson, O. G., Taylor, M., Naik, H., Isley, L., Hornberger, K.
  2026
- **Delivering pharmacogenomic results via electronic health record patient portals: Impacts of health literacy and genetics knowledge.** *Patient education and counseling*
  Kile, S. L., Owusu-Obeng, A., Scott, S. A., Naik, H.
  2026; 149: 109654
- **Guidelines for the management of acute porphyria: recommendations from the International Porphyria Network** *LANCET HAEMATOLOGY*
  Edel, Y., Stein, P. E., Kawtharany, H., Aarsand, A. K., Badminton, M., Balwani, M. C., Bonkovsky, H. L., Cappellini, M., Cassiman, D., Deybach, J., Gill, L., Harper, P., Hift, et al
  2026; 13 (5): e338-e350
- **Guidelines for the management of acute porphyria: recommendations from the International Porphyria Network.** *The Lancet. Haematology*
  Edel, Y., Stein, P. E., Kawtharany, H., Aarsand, A. K., Badminton, M., Balwani, M. C., Bonkovsky, H. L., Cappellini, M. D., Cassiman, D., Deybach, J. C., Gill, L., Harper, P., Hift, et al
  2026; 13 (5): e338-e350
- **A mixed methods approach to understanding patient outcomes in cardiovascular genetic counseling.** *Journal of genetic counseling*
  Lewis, E., Naik, H., Cragun, D., Zierhut, H., Reuter, C., Ison, H.
  2026; 35 (2): e70179

- **Complementary and alternative medicines and cannabis use among individuals with erythropoietic protoporphyria.** *Molecular genetics and metabolism*
  Draisin, E., Rome, C. P., Hedstrom, K., Overbey, J., Balwani, M., Naik, H.
  2025; 147 (1): 109703

- **Clinical trial design essentials for genetic counselors.** *Journal of genetic counseling*
  James, C. A., Overbey, J. R., Bier, L., Caleshu, C., Zierhut, H., Copeland, M., Naik, H.
  2025; 34 (6): e70150

- **Genetic exceptionalism and genomic contextualism among Asian Americans: a qualitative study.** *Journal of community genetics*
  Huang, S. D., Martschenko, D., Scherer, C. R., Chiang, K. F., Chang, K., Naik, H.
  2025; 17 (1): 2

- **Impact of delay in diagnosis in patients with erythropoietic protoporphyria: A cross-sectional survey study.** *JAAD international*
  Tran, B., Raef, H., Rebeiz, L., Wheeden, K., Elmariah, S. B., Anderson, K. E., Leaf, R. K., Naik, H., Dickey, A. K.
  2025; 22: 60-63

- **Validation of the sunlight exposure diary and the erythropoietic protoporphyria impact questionnaire (EPIQ).** *Orphanet journal of rare diseases*
  Naik, H., Mathias, S. D., Turner, M. P., O'Grady, M., Norregaard, C., Colwell, H. H., Savage, W., Chin, M.
  2025; 20 (1): 492

- **The Burden of Erythropoietic Protoporphyria (EPP): Results From the EPP Life Impact and Genetic Health Trajectory (LIGHT) Study.** *The Journal of dermatology*
  Naik, H., Balwani, M., Colwell, H. H., Mathias, S. D., Wheeden, K., Norregaard, C., Chin, M., Savage, W.
  2025

- **Towards an integrated resource for pharmacogenomics (PGx): Survey findings from the genomic medicine communities.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Klein, C. J., Gong, L., Caudle, K. E., Naik, H., Empey, P. E., Hoffman, J. M., Scherer, S., Iwuchukwu, O. F., Gregornik, D., Monte, A. A., Swen, J. J., Scott, S. A., Whirl-Carrillo, et al
  2025: 101529

- **PSYCHOMETRIC VALIDATION OF THE SUNLIGHT EXPOSURE DIARY AND ERYTHROPOIETIC PROTOPORPHYRIA IMPACT QUESTIONNAIRE (EPIQ) USING DATA FROM THE AURORA CLINICAL STUDY IN ERYTHROPOIETIC PROTOPORPHYRIA (EPP)**
  Mathias, S. D., Turner, M. P., O'Grady, M., Norregaard, C., Colwell, H. H., Savage, W., Naik, H., Chin, M.
  ELSEVIER SCIENCE INC.2025: S285

- **A Prospective, Blinded Study of Symptom Prevalence and Specificity of Porphyrin Precursors in Carriers of Acute Hepatic Porphyria.** *Liver international : official journal of the International Association for the Study of the Liver*
  Merati, M., Jayakumar, N., Kapoor, Y., Naik, H., Balwani, M., Anderson, K. E., Bonkovsky, H. L., Desnick, R. J., McGuire, B., Phillips, J., Bissell, D. M., Wang, B.
  2025; 45 (7): e70156

- **Self-Reported Liver Disease and the Burden of Erythropoietic Protoporphyria.** *JIMD reports*
  Naik, H., Wheeden, K., Colwell, H. H., Mathias, S. D., Norregaard, C., Chin, M., Savage, W., Balwani, M.
  2025; 66 (3): e70015

- **Liver involvement in a large cohort of patients with erythropoietic protoporphyria or X-linked protoporphyria.** *Hepatology communications*
  Levy, C., Naik, H., Overbey, J., Hedstrom, K., Wang, K., McDonough, C., Freeman, M., Keel, S. B., Erwin, A. L., Dickey, A. K., Leaf, R. K., Quigley, J., Mazepa, et al
  2025; 9 (3)

- **Patient experience with acute hepatic porphyria before and after long-term givosiran treatment in a qualitative interview study** *MOLECULAR GENETICS AND METABOLISM REPORTS*
  Naik, H., Brown, M., Meninger, S., Lombardelli, S.
  2025; 42

- **Integrating Pharmacogenomics into the Broader Construct of Genomic Medicine: Efforts by the ClinGen Pharmacogenomics Working Group (PGxWG).** *Clinical chemistry*
  Gong, L., Klein, C. J., Caudle, K. E., Moyer, A. M., Scott, S. A., Whirl-Carrillo, M., Klein, T. E.

2025; 71 (1): 36-44

- **The EPP LIGHT Study: A Survey Describing the Burden of Erythropoietic Protoporphyria and X-Linked Protoporphyria**
  Naik, H., Balwani, M., Colwell, H. H., Mathias, S. D., Wheeden, K., Norregaard, C., Chin, M., Savage, W.
  ELSEVIER.2024: 3631-3632
- **INVESTIGATING SOCIAL FACTORS CONTRIBUTING TO THE LACK OF DIVERSITY AMONG SPERM DONORS IN THE UNITED STATES**
  Anderson, O. G., Taylor, M. A., Naik, H., Isley, L., Hornberger, K.
  ELSEVIER SCIENCE INC.2024: E61
- **Health-related quality of life in patients with diverse rare diseases: An online survey.** *Genetics in medicine open*
  Rao, A., Yabumoto, M., Ward-Lev, E., Miller, E. G., Naik, H., Halley, M. C.
  2024; 2: 101889
- **Afamelanotide for Treatment of the Protoporphyrias: Impact on Quality of Life and Laboratory Parameters in a US Cohort.** *Life (Basel, Switzerland)*
  Leaf, R. K., Naik, H., Jiang, P. Y., Elmariah, S. B., Hodges, P., Mead, J., Trinidad, J., Saberi, B., Tran, B., Valiante, S., Mernick, F., Leaf, D. E., Anderson, et al
  2024; 14 (6)
- **Research biobank participants attitudes towards genetic exceptionalism and health record confidentiality.** *Journal of community genetics*
  Dortenzio, V., Rhodes, R., Merkelson, A., Naik, H.
  2024
- **Knowledge and attitudes on implementing cardiovascular pharmacogenomic testing.** *Clinical and translational science*
  Russell, C., Campion, M., Grove, M. E., Matsuda, K., Klein, T. E., Ashley, E., Naik, H., Wheeler, M. T., Scott, S. A.
  2024; 17 (3): e13737
- **Development and Content Validation of Novel Patient-Reported Outcome Measures to Assess Disease Severity and Change in Patients with Erythropoietic Protoporphyria: The EPP Impact Questionnaire (EPIQ).** *Patient related outcome measures*
  Mathias, S. D., Burke, L., Colwell, H. H., Mensing, G., Savage, W., Naik, H.
  2024; 15: 17-30
- **Pharmacogenomic knowledge and awareness among diverse patients treated with angiotensin converting enzyme inhibitors.** *Pharmacogenomics*
  Naik, H., O'Connor, M. Y., Sanderson, S. C., Pinnell, N., Dong, M., Wiegand, A., Obeng, A. O., Abul-Husn, N. S., Scott, S. A.
  2023
- **Development and content validation of a sunlight exposure diary in patients with erythropoietic protoporphyria.** *Journal of patient-reported outcomes*
  Mathias, S. D., Burke, L., Colwell, H. H., Mensing, G., Savage, W., Naik, H.
  2023; 7 (1): 119
- **Evidence based consensus guidelines for diagnosis and management of Protoporphyria-Related liver dysfunction in erythropoietic protoporphyria and X-Linked protoporphyria.** *Hepatology (Baltimore, Md.)*
  Levy, C., Dickey, A. K., Wang, B., Thapar, M., Naik, H., Keel, S. B., Saberi, B., Beaven, S. W., Rudnick, S. R., Elmariah, S. B., Erwin, A. L., Goddu, R. J., Hedstrom, et al
  2023
- **Patient and Provider Experiences and Views on the Use of Telehealth in Genetics Clinics in Response to the COVID-19 Pandemic.** *Telemedicine journal and e-health : the official journal of the American Telemedicine Association*
  Martinez, J. M., Zinberg, R. E., Diaz, G. A., Naik, H.
  2023
- **Ledipasvir/Sofosbuvir Is Effective as Sole Treatment of Porphyria Cutanea Tarda with Chronic Hepatitis C.** *Digestive diseases and sciences*
  Bonkovsky, H. L., Rudnick, S. P., Ma, C. D., Overbey, J. R., Wang, K., Faust, D., Hallberg, C., Hedstrom, K., Naik, H., Moghe, A., Anderson, K. E.
  2023; 68 (6): 2738-2746
- **Elective genetic testing: Genetics professionals' perspectives and practices.** *Journal of genetic counseling*
  Miura, M. S., Suckiel, S. A., Naik, H., Soper, E. R., Abul-Husn, N. S.
  2023; 32 (3): 607-617

- **Frequency of Epistaxis and Telangiectasia in patients with Hereditary Hemorrhagic Telangiectasia (HHT) in comparison with the General Population: Curaçao Diagnostic Criteria Revisited.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  McDonald, J., Kornish, J., Stevenson, D. A., Hanson-Kahn, A., Balch, H., James, J., Naik, H., Whitehead, K. J.
  2023: 100865

- **Key terms and definitions in acute porphyrias: Results of an international Delphi consensus led by the European porphyria network.** *Journal of inherited metabolic disease*
  Stein, P. E., Edel, Y., Mansour, R., Mustafa, R. A., Sandberg, S.
  2023

- **A pilot study of oral iron therapy in erythropoietic protoporphyria and X-linked protoporphyria.** *Molecular genetics and metabolism reports*
  Balwani, M., Naik, H., Overbey, J. R., Bonkovsky, H. L., Bissell, D. M., Wang, B., Phillips, J. D., Desnick, R. J., Anderson, K. E.
  2022; 33: 100939

- **Prospective observational pilot study of quantitative light dosimetry in erythropoietic protoporphyria.** *Journal of the American Academy of Dermatology*
  Dickey, A., Rebeiz, L., Raef, H., Leaf, R. K., Elmariah, S., Naik, H., Anderson, K., Conley, J., Iyasere, C., Zhao, S., Birkenfeld, J. S., Arroyo-Gallego, T., Wheeden, et al
  2022

- **Evidence-based consensus guidelines for the diagnosis and management of erythropoietic protoporphyria and X-linked protoporphyria.** *Journal of the American Academy of Dermatology*
  Dickey, A. K., Naik, H., Keel, S. B., Levy, C., Beaven, S. W., Elmariah, S. B., Erwin, A. L., Goddu, R. J., Hedstrom, K., Leaf, R. K., Kazamel, M., Mazepa, M., Philpotts, et al
  2022

- **Attitudes on pharmacogenomic results as secondary findings among medical geneticists.** *Pharmacogenetics and genomics*
  Bartos, M. N., Scott, S. A., Jabs, E. W., Naik, H.
  2022

- **Hepatocellular Carcinoma in Acute Hepatic Porphyrias: Results from the Longitudinal Study of the U.S. Porphyrias Consortium.** *Hepatology (Baltimore, Md.)*
  Saberi, B., Naik, H., Overbey, J. R., Erwin, A. L., Anderson, K. E., Bissell, D. M., Bonkovsky, H. L., Phillips, J. D., Wang, B., K Singal, A., M McGuire, B., Desnick, R. J., Balwani, et al
  2021; 73 (5): 1736-1746

- **Pharmacogenomic education among genetic counseling training programs in North America.** *Journal of genetic counseling*
  Loudon, E., Scott, S. A., Rigobello, R., Scott, E. R., Zinberg, R., Naik, H.
  2021

- **Experiences from the epicenter: Professional impact of the COVID-19 pandemic on genetic counselors in New York.** *American journal of medical genetics. Part C, Seminars in medical genetics*
  Bergstrom, K. L., Brander, T. E., Breen, K. E., Naik, H.
  2021; 187 (1): 28-36

- **Evidence in the UK Biobank for the underdiagnosis of erythropoietic protoporphyria.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Dickey, A. K., Quick, C., Ducamp, S., Zhu, Z., Feng, Y. A., Naik, H., Balwani, M., Anderson, K. E., Lin, X., Phillips, J. E., Rebeiz, L., Bonkovsky, H. L., McGuire, et al
  2021; 23 (1): 140-148

- **Gaucher disease and SARS-CoV-2 infection: Experience from 181 patients in New York.** *Molecular genetics and metabolism*
  Fierro, L., Nesheiwat, N., Naik, H., Narayanan, P., Mistry, P. K., Balwani, M.
  2021; 132 (1): 44-48

- **Digital Health Applications for Pharmacogenetic Clinical Trials.** *Genes*
  Naik, H., Palaniappan, L., Ashley, E. A., Scott, S. A.
  2020; 11 (11)

- **EXPLORE: A Prospective, Multinational, Natural History Study of Patients with Acute Hepatic Porphyria with Recurrent Attacks.** *Hepatology (Baltimore, Md.)*

Gouya, L., Ventura, P., Balwani, M., Bissell, D. M., Rees, D. C., Stölzel, U., Phillips, J. D., Kauppinen, R., Langendonk, J. G., Desnick, R. J., Deybach, J. C., Bonkovsky, H. L., Parker, et al
2020; 71 (5): 1546-1558

- **Evaluating the Patient-Reported Outcomes Measurement Information System scales in acute intermittent porphyria.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Naik, H., Overbey, J. R., Montgomery, G. H., Winkel, G., Balwani, M., Anderson, K. E., Bissell, D. M., Bonkovsky, H. L., Phillips, J. D., Wang, B., McGuire, B., Keel, S., Levy, et al
  2020; 22 (3): 590-597

- **The N370S/R496H genotype in type 1 Gaucher disease - Natural history and implications for pre symptomatic diagnosis and counseling.** *Molecular genetics and metabolism reports*
  Zeid, N., Stauffer, C., Yang, A., Naik, H., Fierro, L., Ganesh, J., Balwani, M.
  2020; 22: 100567

- **Knowledge and attitudes of Parkinson's disease risk in the Gaucher population.** *Journal of genetic counseling*
  Zaretsky, L., Zeid, N., Naik, H., Alcalay, R. N., Balwani, M.
  2020; 29 (1): 105-111

- **Knowledge and attitudes on pharmacogenetics among pediatricians.** *Journal of human genetics*
  Rahawi, S. n., Naik, H. n., Blake, K. V., Owusu Obeng, A. n., Wasserman, R. M., Seki, Y. n., Funanage, V. L., Oishi, K. n., Scott, S. A.
  2020; 65 (5): 437–44

- **Implementing a pharmacogenetic-driven algorithm to guide dual antiplatelet therapy (DAPT) in Caribbean Hispanics: protocol for a non-randomised clinical trial.** *BMJ open*
  Hernandez-Suarez, D. F., Melin, K. n., Marin-Maldonado, F. n., Nunez, H. J., Gonzalez, A. F., Gonzalez-Sepulveda, L. n., Rivas-Tumanyan, S. n., Naik, H. n., Ruaño, G. n., Scott, S. A., Duconge, J. n.
  2020; 10 (8): e038936

- **Clinical Pharmacogenetic Testing and the Posttest Counseling Conundrum.** *Clinical pharmacology and therapeutics*
  Rigobello, R. n., Rahawi, S. n., Wallsten, R. n., Cody, N. n., Nicoletti, P. n., Owusu Obeng, A. n., Naik, H. n., Dillon, M. W., Scott, S. A.
  2020; 108 (5): 924–28

- **Evaluating quality of life tools in North American patients with erythropoietic protoporphyria and X-linked protoporphyria.** *JIMD reports*
  Naik, H., Overbey, J. R., Desnick, R. J., Anderson, K. E., Bissell, D. M., Bloomer, J., Bonkovsky, H. L., Phillips, J. D., Wang, B., Singal, A., Balwani, M.
  2019; 50 (1): 9-19

- **Psychosocial issues in erythropoietic protoporphyria - the perspective of parents, children, and young adults: A qualitative study.** *Molecular genetics and metabolism*
  Naik, H., Shenbagam, S., Go, A. M., Balwani, M.
  2019; 128 (3): 314-319

- **Diagnostic Delay in Erythropoietic Protoporphyria.** *The Journal of pediatrics*
  Lala, S. M., Naik, H., Balwani, M.
  2018; 202: 320-323.e2

- **Parkinson's disease prevalence in Fabry disease: A survey study.** *Molecular genetics and metabolism reports*
  Wise, A. H., Yang, A., Naik, H., Stauffer, C., Zeid, N., Liong, C., Balwani, M., Desnick, R. J., Alcalay, R. N.
  2018; 14: 27-30

- **Clinical, Biochemical, and Genetic Characterization of North American Patients With Erythropoietic Protoporphyria and X-linked Protoporphyria.** *JAMA dermatology*
  Balwani, M., Naik, H., Anderson, K. E., Bissell, D. M., Bloomer, J., Bonkovsky, H. L., Phillips, J. D., Overbey, J. R., Wang, B., Singal, A. K., Liu, L. U., Desnick, R. J.
  2017; 153 (8): 789-796

- **Institutional profile: translational pharmacogenomics at the Icahn School of Medicine at Mount Sinai.** *Pharmacogenomics*
  Scott, S. A., Owusu Obeng, A. n., Botton, M. R., Yang, Y. n., Scott, E. R., Ellis, S. B., Wallsten, R. n., Kaszemacher, T. n., Zhou, X. n., Chen, R. n., Nicoletti, P. n., Naik, H. n., Kenny, et al
  2017; 18 (15): 1381–86

- **Acute Intermittent Porphyria in children: A case report and review of the literature.** *Molecular genetics and metabolism*
  Balwani, M., Singh, P., Seth, A., Debnath, E. M., Naik, H., Doheny, D., Chen, B., Yasuda, M., Desnick, R. J.
  2016; 119 (4): 295-299
- **Experiences and concerns of patients with recurrent attacks of acute hepatic porphyria: A qualitative study.** *Molecular genetics and metabolism*
  Naik, H., Stoecker, M., Sanderson, S. C., Balwani, M., Desnick, R. J.
  2016; 119 (3): 278-283
- **The D519G Polymorphism of Glyceronephosphate O-Acyltransferase Is a Risk Factor for Familial Porphyria Cutanea Tarda.** *PloS one*
  Farrell, C. P., Overbey, J. R., Naik, H., Nance, D., McLaren, G. D., McLaren, C. E., Zhou, L., Desnick, R. J., Parker, C. J., Phillips, J. D.
  2016; 11 (9): e0163322
- **Pitfalls in Erythrocyte Protoporphyrin Measurement for Diagnosis and Monitoring of Protoporphyrias.** *Clinical chemistry*
  Gou, E. W., Balwani, M., Bissell, D. M., Bloomer, J. R., Bonkovsky, H. L., Desnick, R. J., Naik, H., Phillips, J. D., Singal, A. K., Wang, B., Keel, S., Anderson, K. E.
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