



Chloe Reuter

- Clinical Associate Professor, Medicine - Cardiovascular Medicine
- Clinical Associate Professor, Pediatrics - Medical Genetics

Bio

ACADEMIC APPOINTMENTS

- Clinical Associate Professor, Medicine - Cardiovascular Medicine
- Clinical Associate Professor, Pediatrics - Medical Genetics

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Member, National Society of Genetic Counselors (2015 - present)

PROFESSIONAL EDUCATION

- Board Certification, American Board of Genetic Counseling (2016)
- MS, Stanford School of Medicine , Human Genetics and Genetic Counseling (2016)
- BA, Cornell University , Biological Sciences (2014)

Publications

PUBLICATIONS

- **Arrhythmogenic Cardiomyopathy: Mechanisms, Genetics, and Their Clinical Implications** *CURRENT CARDIOVASCULAR RISK REPORTS*
Reuter, C. M., Dries, A. M., Parikh, V. N.
2021; 15 (5)
- **De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation.** *American journal of human genetics*
Mao, D. n., Reuter, C. M., Ruzhnikov, M. R., Beck, A. E., Farrow, E. G., Emrick, L. T., Rosenfeld, J. A., Mackenzie, K. M., Robak, L. n., Wheeler, M. T., Burrage, L. C., Jain, M. n., Liu, et al
2020
- **Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students.** *Journal of genetic counseling*
Grove, M. E., White, S. n., Fisk, D. G., Rego, S. n., Dagan-Rosenfeld, O. n., Kohler, J. N., Reuter, C. M., Bonner, D. n., Wheeler, M. T., Bernstein, J. A., Ormond, K. E., Hanson-Kahn, A. K.
2019
- **Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease.** *The New England journal of medicine*
Splinter, K. n., Adams, D. R., Bacino, C. A., Bellen, H. J., Bernstein, J. A., Cheatle-Jarvela, A. M., Eng, C. M., Esteves, C. n., Gahl, W. A., Hamid, R. n., Jacob, H. J., Kikani, B. n., Koeller, et al
2018
- **Patient experiences with clinical confirmatory genetic testing after using direct-to-consumer raw DNA and third-party genetic interpretation services.** *Translational behavioral medicine*
Nguyen Dolphyn, T. T., Ormond, K. E., Weissman, S. M., Kim, H. J., Reuter, C. M.

2022

- **Causative Variants for Inherited Cardiac Conditions in a Southeast Asian Population Cohort.** *Circulation. Genomic and precision medicine*
Tomar, S., Klinzing, D. C., Kit, C. C., Gan, L. H., Moscarello, T., Reuter, C., Ashley, E. A., Foo, R.
2022: CIRCGEN121003536
- **Phenotypic Expression, Natural History and Risk Stratification of Cardiomyopathy Caused by Filamin C Truncating Variants.** *Circulation*
Gigli, M., Stolfo, D., Graw, S., Merlo, M., Gregorio, C., Chen, S. N., Dal Ferro, M., Paldino, A., De Angelis, G., Brun, F., Jirikowic, J., Salcedo, E. E., Turja, et al
2021
- **Correction to: The genetic architecture of Plakophilin 2 cardiomyopathy.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Dries, A. M., Kirillova, A., Reuter, C. M., Garcia, J., Zouk, H., Hawley, M., Murray, B., Tichnell, C., Pilichou, K., Protonotarios, A., Medeiros-Domingo, A., Kelly, M. A., Baras, et al
2021
- **Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation.** *Journal of genetic counseling*
Kohler, J. N., Kelley, E. G., Boyd, B. M., Sillari, C. H., Marwaha, S., Undiagnosed Diseases Network, Wheeler, M. T., Acosta, M. T., Adam, M., Adams, D. R., Agrawal, P. B., Alejandro, M. E., Alvey, J., et al
2021
- **The genetic architecture of Plakophilin 2 cardiomyopathy.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Dries, A. M., Kirillova, A., Reuter, C. M., Garcia, J., Zouk, H., Hawley, M., Murray, B., Tichnell, C., Pilichou, K., Protonotarios, A., Medeiros-Domingo, A., Kelly, M. A., Baras, et al
2021
- **"Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey.** *Journal of genetic counseling*
Deutch, N. T., Beckman, E., Halley, M. C., Young, J. L., Reuter, C. M., Kohler, J., Bernstein, J. A., Wheeler, M. T., Undiagnosed Diseases Network, Ormond, K. E., Tabor, H. K.
2021
- **A resource of lipidomics and metabolomics data from individuals with undiagnosed diseases** *SCIENTIFIC DATA*
Kyle, J. E., Stratton, K. G., Zink, E. M., Kim, Y., Bloodsworth, K. J., Monroe, M. E., Bacino, C. A., Bacino, C. A., Hanchard, N. A., Lewis, R. A., Rosenfeld, J. A., Scott, D. A., Tran, et al
2021; 8 (1): 114
- **Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain** *GENETICS IN MEDICINE*
Marbach, F., Stoyanov, G., Erger, F., Stratakis, C. A., Settas, N., London, E., Rosenfeld, J. A., Torti, E., Haldeman-Englert, C., Sklirou, E., Kessler, E., Ceulemans, S., Nelson, et al
2021
- **Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Kobren, S. N., Baldrige, D., Velinder, M., Krier, J. B., LeBlanc, K., Esteves, C., Pusey, B. N., Zuchner, S., Blue, E., Lee, H., Huang, A., Bastarache, L., Bican, et al
2021
- **Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Schoch, K. n., Esteves, C. n., Bican, A. n., Spillmann, R. n., Cope, H. n., McConkie-Rosell, A. n., Walley, N. n., Fernandez, L. n., Kohler, J. N., Bonner, D. n., Reuter, C. n., Stong, N. n., Mulvihill, et al
2020
- **Broad Genetic Testing in a Clinical Setting Uncovers a High Prevalence of Titin Loss-of-Function Variants in Very Early-Onset Atrial Fibrillation.** *Circulation. Genomic and precision medicine*
Goodyer, W. R., Dunn, K., Caleshu, C., Jackson, M., Wylie, J., Moscarello, T., Platt, J., Reuter, C., Smith, A., Trela, A., Ceresnak, S. R., Motonaga, K. S., Ashley, et al
2019

- **Pathological overlap of Arrhythmogenic Right Ventricular Cardiomyopathy and Cardiac Sarcoidosis.** *Circulation. Genomic and precision medicine*
Kerkar, A., Hazard, F., Caleshu, C. A., Shah, R. L., Reuter, C., Ashley, E. A., Parikh, V. N.
2019
- **Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing.** *Journal of genetic counseling*
Reuter, C. M., Kohler, J. N., Bonner, D., Zastrow, D., Fernandez, L., Dries, A., Marwaha, S., Davidson, J., Brokamp, E., Herzog, M., Hong, J., Macnamara, E., Rosenfeld, et al
2019
- **Understanding variants of uncertain significance in the era of multigene panels: Through the eyes of the patient** *JOURNAL OF GENETIC COUNSELING*
Reuter, C., Chun, N., Pariani, M., Hanson-Kahn, A.
2019; 28 (4): 878–86
- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
Kumar, A., Zastrow, D. B., Kravets, E. J., Belefond, D., Ruzhnikov, M. R. Z., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Mackenzie, K. M., Eng, et al
2019; 179 (6): 966–77
- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing** *JOURNAL OF GENETIC COUNSELING*
Zastrow, D. B., Kohler, J. N., Bonner, D., Reuter, C. M., Fernandez, L., Grove, M. E., Fisk, D. G., Yang, Y., Eng, C. M., Ward, P. A., Bick, D., Worthey, E. A., Fisher, et al
2019; 28 (2): 213–28
- **Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students** *JOURNAL OF GENETIC COUNSELING*
Grove, M. E., White, S., Fisk, D. G., Rego, S., Dagan-Rosenfeld, O., Kohler, J. N., Reuter, C. M., Bonner, D., Wheeler, M. T., Bernstein, J. A., Ormond, K. E., Hanson-Kahn, A. K., Undiagnosed Dis Network
2019; 28 (2): 466–76
- **Regional Variation in RBM20 Causes a Highly Penetrant Arrhythmogenic Cardiomyopathy.** *Circulation. Heart failure*
Parikh, V. N., Caleshu, C., Reuter, C., Lazzeroni, L. C., Ingles, J., Garcia, J., McCaleb, K., Adesiyun, T., Sedaghat-Hamedani, F., Kumar, S., Graw, S., Gigli, M., Stolfo, et al
2019; 12 (3): e005371
- **Direct-to-consumer raw genetic data and third-party interpretation services: more burden than bargain?** *GENETICS IN MEDICINE*
Moscarello, T., Murray, B., Reuter, C. M., Demo, E.
2019; 21 (3): 539-541
- **Regional Variation in RBM20 Causes a Highly Penetrant Arrhythmogenic Cardiomyopathy** *CIRCULATION-HEART FAILURE*
Parikh, V. N., Caleshu, C., Reuter, C., Lazzeroni, L. C., Ingles, J., Garcia, J., McCaleb, K., Adesiyun, T., Sedaghat-Hamedani, F., Kumar, S., Graw, S., Gigli, M., Stolfo, et al
2019; 12 (3)
- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing.** *Journal of genetic counseling*
Zastrow, D. B., Kohler, J. N., Bonner, D. n., Reuter, C. M., Fernandez, L. n., Grove, M. E., Fisk, D. G., Yang, Y. n., Eng, C. M., Ward, P. A., Bick, D. n., Worthey, E. A., Fisher, et al
2019; 28 (2): 213–28
- **Genomics in medicine: a novel elective rotation for internal medicine residents.** *Postgraduate medical journal*
Geng, L. N., Kohler, J. N., Levonian, P. n., Bernstein, J. A., Ford, J. M., Ahuja, N. n., Witteles, R. n., Hom, J. n., Wheeler, M. n.
2019
- **Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts.** *Nature medicine*
Frésard, L. n., Smail, C. n., Ferraro, N. M., Teran, N. A., Li, X. n., Smith, K. S., Bonner, D. n., Kernohan, K. D., Marwaha, S. n., Zappala, Z. n., Balliu, B. n., Davis, J. R., Liu, et al
2019

- **Understanding variants of uncertain significance in the era of multigene panels: Through the eyes of the patient.** *Journal of genetic counseling*
Reuter, C. n., Chun, N. n., Pariani, M. n., Hanson-Kahn, A. n.
2019
- **Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease** *NEW ENGLAND JOURNAL OF MEDICINE*
Splinter, K., Adams, D. R., Bacino, C. A., Bellen, H. J., Bernstein, J. A., Cheattle-Jarvela, A. M., Eng, C. M., Esteves, C., Gahl, W. A., Hamid, R., Jacob, H. J., Kikani, B., Koeller, et al
2018; 379 (22): 2131–39
- **Regional Variation in RBM20 Causes a Highly Penetrant Arrhythmogenic Cardiomyopathy.**
Parikh, V. N., Caleshu, C., Reuter, C., Lazzeroni, L., Ingles, J., Kumar, S., Garcia, J., McCaleb, K., Adesiyun, T., Dedaghat-Hamedani, F., Graw, S., Gigli, M., Stolfo, et al
LIPPINCOTT WILLIAMS & WILKINS.2018
- **Clinical Cardiovascular Genetic Counselors Take a Leading Role in Team-based Variant Classification** *JOURNAL OF GENETIC COUNSELING*
Reuter, C., Grove, M. E., Orland, K., Spoonamore, K., Caleshu, C.
2018; 27 (4): 751–60
- **Genome Sequencing in Hypertrophic Cardiomyopathy.** *Journal of the American College of Cardiology*
Ashley, E. A., Reuter, C. M., Wheeler, M. T.
2018; 72 (4): 430–33
- **Genome Sequencing in Hypertrophic Cardiomyopathy** *JOURNAL OF THE AMERICAN COLLEGE OF CARDIOLOGY*
Ashley, E. A., Reuter, C. M., Wheeler, M. T.
2018; 72 (4): 430-433
- **Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder** *AMERICAN JOURNAL OF HUMAN GENETICS*
Olahova, M., Yoon, W., Thompson, K., Jangam, S., Fernandez, L., Davidson, J. M., Kyle, J. E., Grove, M. E., Fisk, D. G., Kohler, J. N., Holmes, M., Dries, A. M., Huang, et al
2018; 102 (3): 494–504
- **A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network.** *The Journal of pediatrics*
Reuter, C. M., Brimble, E. n., DeFilippo, C. n., Dries, A. M., Enns, G. M., Ashley, E. A., Bernstein, J. A., Fisher, P. G., Wheeler, M. T.
2018
- **Clinical Characteristics of the GLA N215S Variant and Implications for the Diagnosis and Management of Nonclassic Fabry Disease** *CIRCULATION-CARDIOVASCULAR GENETICS*
Reuter, C., Platt, J.
2017; 10 (5)