Stanford

Chloe Reuter

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

Bio

BIO

Chloe Reuter is a Genetic Counselor and Clinical Associate Professor in the Division of Cardiovascular Medicine at Stanford University School of Medicine. She is Principal Investigator of an NIH-funded research program focused on developing frameworks for the clinical translation of multi-omic technologies in rare diseases.

Ms. Reuter provides genetic counseling care at the Stanford Center for Inherited Cardiovascular Diseases, specializing in inherited cardiovascular conditions including cardiomyopathies, arrhythmia syndromes, and sudden cardiac death. She has expertise in interpreting complex genomic results from multi-gene panels, exome sequencing, and whole genome sequencing for patients with rare and undiagnosed diseases.

Ms. Reuter's research centers on translating cutting-edge genomic technologies into clinical practice. She develops frameworks for integrating multi-omic data (genomic, transcriptomic, and other omics) into diagnostic workflows and contributes to evidence-based guidelines for variant interpretation and clinical curation. Her work addresses the ethical and practical challenges of implementing these technologies, with particular focus on patient engagement, equitable access to precision medicine, and protocols for returning complex research results. She also conducts genetic counseling practice research, examining patient experiences, psychosocial assessment tools, and healthcare delivery models including telehealth services.

She has contributed to multiple national genomics initiatives including the Undiagnosed Diseases Network and the GREGoR Consortium, applying comprehensive genomic approaches to solve medical mysteries and improve diagnostic outcomes for patients with rare diseases.

ACADEMIC APPOINTMENTS

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

HONORS AND AWARDS

- Loan Repayment Program Award, National Institutes of Health (2022 - 2027)

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

- **Cardiovascular Genetic Counseling Is Associated With Improved Patient-Reported Outcomes Across Clinical Indications and Settings.** *Circulation. Genomic and precision medicine*
  Murray, B., Gordon, C., Christian, S., Dzwiniel, T., Tichnell, C., Brown, E., MacCarrick, G., McClellan, R., Reuter, C., Ison, H., Carter, J., Moscarello, T., Murtha, et al
  2026; 19 (5): e005514

- **Characterizing psychosocial assessments across genetic counseling sessions through qualitative content analysis.** *Journal of genetic counseling*
  Ruiz, K., Narain, A., Nguyen, T. P., Pollard, E., Joseph, G., Moscarello, T., Campion, M., Reuter, C.
  2026; 35 (1): e70161

- **Assessing and attending to psychosocial concerns in genetic counseling: Proposing the BATHE method.** *Journal of genetic counseling*
  Moscarello, T., Higgs, E., Pollard, E., Monroe, M., Nguyen, T. P., Campion, M., Reuter, C. M.
  2024

- **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

- **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

- **MetaDome 2027: a comprehensively updated resource for aggregating missense variant evidence across homologous human protein domains.** *Nucleic acids research*
  Wiel, L., Ferraro, F., Yu, J., Zhen, J., Nachun, D., Mendez, R., Reuter, C. M., Cui, J. L., Bonner, D. E., Carter, J. N., Marwaha, S., van de Vorst, M., Emami, et al
  2026

- **MetaDome 2027: a comprehensively updated resource for aggregating missense variant evidence across homologous human protein domains.** *bioRxiv : the preprint server for biology*
  Wiel, L., Ferraro, F., Yu, J., Zhen, J., Nachun, D., Mendez, R., Reuter, C. M., Cui, J. L., Bonner, D. E., Carter, J. N., Marwaha, S., van de Vorst, M., Emami, et al
  2026

- **"It didn't feel like anything unusual because we had already been through so much": Disability-Related Research Experiences of Families with Children Enrolled in the Undiagnosed Diseases Network.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Mintz, K. T., Altamirano, E. N., Halley, M. C., Barton, K. S., Cho, M. K., Bernstein, J. A., Carter, J. N., Undiagnosed Diseases Network, Tabor, H. K., Quinlan, A., Elkadri, A., Beggs, A. H., La Spada, A. R., et al
  2026: 102663

- **Ensilication preserves high-molecular weight native DNA for clinical long-read sequencing.** *Genome biology*
  Ferrasse, A., Mendez, R., Gorzynski, J. E., Reuter, C., Carter, J. N., Blas, M., Bernstein, J. A., Wheeler, M. T., Banal, J. L., Ashley, E. A.
  2026; 27 (1)

- **Population-scale detection of methylation outliers from long-read genome sequencing.** *medRxiv : the preprint server for health sciences*
  Jensen, T. D., Kaur, R., Bonner, D. E., Nguyen, J., Reuter, C. M., Ashley, E. A., Bernstein, J. A., Wheeler, M. T., Montgomery, S. B.
  2026

- **Author Correction: Biallelic variants in the noncoding RNA gene RNU4-2 cause a recessive neurodevelopmental syndrome with distinct white matter changes.** *Nature genetics*
  Rius, R., Blakes, A. J., Chen, Y., De Jonghe, J., Lecoquierre, F., Dawes, R., Cogne, B., Kim, H. C., Alvi, J. R., Amblard, F., Ansari, M., Arlt, A., Austin-Tse, et al
  2026

- **Biallelic variants in the noncoding RNA gene RNU4-2 cause a recessive neurodevelopmental syndrome with distinct white matter changes.** *Nature genetics*
  Rius, R., Blakes, A. J., Chen, Y., De Jonghe, J., Lecoquierre, F., Dawes, R., Cogne, B., Kim, H. C., Alvi, J. R., Amblard, F., Ansari, M., Arlt, A., Austin-Tse, et al
  2026

- **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

- **DNASE1L3 Deficiency With Novel Missense Variant: Enzymatic and Plasma Fragmentomic Evidence of Pathogenicity and Partial Response to JAK Blockade** *ACR OPEN RHEUMATOLOGY*
  Tenorio, A., Sugio, T., Cheng, J., Bonner, D. E., Esfahani, M., Kasinathan, S., Hsu, J. J., Moyer, A., Vera, L., Carter, J., Reuter, C. M., Marwaha, S., Balboni, et al
  2026; 8 (2)

- **DNASE1L3 Deficiency With Novel Missense Variant: Enzymatic and Plasma Fragmentomic Evidence of Pathogenicity and Partial Response to JAK Blockade.** *ACR open rheumatology*
  Leon Tenorio, A. A., Sugio, T., Cheng, J., Bonner, D. E., Esfahani, M. S., Kasinathan, S., Hsu, J. J., Moyer, A., Pimentel Vera, L., Carter, J., Reuter, C. M., Marwaha, S., Balboni, et al
  2026; 8 (2): e70184

- **Pathogenic variants in myoferlin (MYOF) cause arrhythmogenic cardiomyopathy**
  Kirillova, A., Aytekin, M., Chan, I., Mina, A., Shao, Y., Kirdar, A., Romito, O., Kalepalli, N., Zhang, D., Al Aaraj, Y., Reuter, C., Annis, J., Yue, et al
  LIPPINCOTT WILLIAMS & WILKINS.2025

- **Stanford Center for Undiagnosed Diseases: a decade of advancing genetic diagnosis and discovery**
  Bonner, D., Carter, J., Reuter, C., Marwaha, S., Kravets, E., Hom, J., Sampson, J., Fisher, P., Ashley, E., Tabor, H., Halley, M., Wheeler, M., Bernstein, et al
  SPRINGERNATURE.2025: 877

- **Clinical Validity of Autosomal Dominant ALPK3 Loss-of-Function Variants as a Cause of Hypertrophic Cardiomyopathy.** *Circulation. Genomic and precision medicine*
  Hespe, S., Singer, E. S., Reuter, C., Murray, B., Jordan, E., Chowns, J., Peters, S., Mayers, M., Gray, B., Hershberger, R. E., Owens, A., Semsarian, C., Waddell, et al
  2025: e004976

- **Integration of transcriptomics and long-read genomics prioritizes structural variants in rare disease.** *Genome research*
  Jensen, T. D., Ni, B., Reuter, C. M., Gorzynski, J. E., Fazal, S., Bonner, D., Ungar, R. A., Goddard, P. C., Raja, A., Ashley, E. A., Bernstein, J. A., Zuchner, S., Greicius, et al
  2025

- **Genes Associated With Hypertrophic Cardiomyopathy: A Reappraisal by the ClinGen Hereditary Cardiovascular Disease Gene Curation Expert Panel.** *Journal of the American College of Cardiology*
  Hespe, S., Waddell, A., Asatryan, B., Owens, E., Thaxton, C., Adduru, M. L., Anderson, K., Brown, E. E., Hoffman-Andrews, L., Jordan, E., Josephs, K., Mayers, M., Peters, et al
  2025; 85 (7): 727-740

- **Arrhythmic Risk Stratification of Carriers of Filamin C Truncating Variants.** *JAMA cardiology*
  Filamin C Registry Consortium, Gigli, M., Stolfo, D., Barbati, G., Graw, S., Chen, S. N., Merlo, M., Medo, K., Gregorio, C., Dal Ferro, M., Paldino, A., Perotto, M., Peter van Tintelen, J., et al
  2025

- **Researcher views on returning results from multi-omics data to research participants: insights from The Molecular Transducers of Physical Activity Consortium (MoTrPAC) Study.** *BMC medical ethics*
  Ormond, K. E., Stanclift, C., Reuter, C. M., Carter, J. N., Murphy, K. E., Lindholm, M. E., Wheeler, M. T.

2025; 26 (1): 22

- **Detailed tandem repeat allele profiling in 1,027 long-read genomes reveals genome-wide patterns of pathogenicity.** *bioRxiv : the preprint server for biology*

  Danzi, M. C., Xu, I. R., Fazal, S., Dolzhenko, E., Pellerin, D., Weisburd, B., Reuter, C., Sampson, J., Folland, C., Wheeler, M., O'Donnell-Luria, A., Wuchty, S., Ravenscroft, et al

  2025

- **Re-evaluating the clinical validity of hypertrophic cardiomyopathy genes**

  Hespe, S., Waddell, A., Asatryan, B., Owens, E., Thaxton, C., Adduru, M., Anderson, K., Brown, E., Hoffman-Andrews, L., Jordan, E., Mayers, M., Peters, S., Stafford, et al

  SPRINGERNATURE.2024: 1393-1394

- **Re-evaluating the clinical validity of hypertrophic cardiomyopathy genes**

  Hespe, S., Waddell, A., Asatryan, B., Owens, E., Thaxton, C., Adduru, M., Anderson, K., Brown, E., Hoffman-Andrews, L., Jordan, E., Mayers, M., Peters, S., Stafford, et al

  SPRINGERNATURE.2024: 1393-1394

- **The current landscape of clinical exome and genome reanalysis in the U.S.** *Journal of genetic counseling*

  Frees, M., Carter, J. N., Wheeler, M. T., Reuter, C.

  2024

- **Large-scale mutational analysis identifies UNC93B1 variants that drive TLR-mediated autoimmunity in mice and humans.** *The Journal of experimental medicine*

  Rael, V. E., Yano, J. A., Huizar, J. P., Slayden, L. C., Weiss, M. A., Turcotte, E. A., Terry, J. M., Zuo, W., Thiffault, I., Pastinen, T., Farrow, E. G., Jenkins, J. L., Becker, et al

  2024; 221 (8)

- **ClinGen Hereditary Cardiovascular Disease Gene Curation Expert Panel: Reappraisal of Genes associated with Hypertrophic Cardiomyopathy.** *medRxiv : the preprint server for health sciences*

  Hespe, S., Waddell, A., Asatryan, B., Owens, E., Thaxton, C., Adduru, M. L., Anderson, K., Brown, E. E., Hoffman-Andrews, L., Jordan, E., Josephs, K., Mayers, M., Peters, et al

  2024

- **De novo variants in the RNU4-2 snRNA cause a frequent neurodevelopmental syndrome.** *Nature*

  Chen, Y., Dawes, R., Kim, H. C., Ljungdahl, A., Stenton, S. L., Walker, S., Lord, J., Lemire, G., Martin-Geary, A. C., Ganesh, V. S., Ma, J., Ellingford, J. M., Delage, et al

  2024

- **Loss of function of FAM177A1, a Golgi complex localized protein, causes a novel neurodevelopmental disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Kohler, J. N., Legro, N. R., Baldridge, D., Shin, J., Bowman, A., Ugur, B., Jackstadt, M. M., Shriver, L. P., Patti, G. J., Zhang, B., Feng, W., McAdow, A. R., Goddard, et al

  2024: 101166

- **De novo variants in the non-coding spliceosomal snRNA gene RNU4-2 are a frequent cause of syndromic neurodevelopmental disorders.** *medRxiv : the preprint server for health sciences*

  Chen, Y., Dawes, R., Kim, H. C., Stenton, S. L., Walker, S., Ljungdahl, A., Lord, J., Ganesh, V. S., Ma, J., Martin-Geary, A. C., Lemire, G., D'Souza, E. N., Dong, et al

  2024

- **Integration of transcriptomics and long-read genomics prioritizes structural variants in rare disease.** *medRxiv : the preprint server for health sciences*

  Jensen, T. D., Ni, B., Reuter, C. M., Gorzynski, J. E., Fazal, S., Bonner, D., Ungar, R. A., Goddard, P. C., Raja, A., Ashley, E. A., Bernstein, J. A., Zuchner, S., Greicius, et al

  2024

- **Regional Variation in Cardiovascular Genes Enables a Tractable Genome Editing Strategy.** *Circulation. Genomic and precision medicine*

  Krysov, V. A., Wilson, R. H., Ten, N. S., Youlton, N., De Jong, H. N., Sutton, S., Huang, Y., Reuter, C. M., Grove, M. E., Wheeler, M. T., Ashley, E. A., Parikh, V. N.

  2024: e004370

- **Exome and genome sequencing in a heterogeneous population of patients with rare disease: Identifying predictors of a diagnosis.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Pucel, J., Briere, L. C., Reuter, C., Gochyyev, P., LeBlanc, K.
  2024: 101115

- **RExPRT: a machine learning tool to predict pathogenicity of tandem repeat loci.** *Genome biology*
  Fazal, S., Danzi, M. C., Xu, I., Kobren, S. N., Sunyaev, S., Reuter, C., Marwaha, S., Wheeler, M., Dolzhenko, E., Lucas, F., Wuchty, S., Tekin, M., Züchner, et al
  2024; 25 (1): 39

- **Arrhythmias including Atrial Fibrillation and Congenital Heart Disease in Kleefstra Syndrome: a possible epigenetic link.** *Europace : European pacing, arrhythmias, and cardiac electrophysiology : journal of the working groups on cardiac pacing, arrhythmias, and cardiac cellular electrophysiology of the European Society of Cardiology*
  Vasireddi, S. K., Draksler, T. Z., Bouman, A., Kummeling, J., Wheeler, M., Reuter, C., Srivastava, S., Harris, J., Fisher, P. G., Narayan, S. M., Wang, P. J., Badhwar, N., Kleefstra, et al
  2024

- **Emery-Dreifuss Muscular Dystrophy 1 is associated with high risk of malignant ventricular arrhythmias and end-stage heart failure.** *European heart journal*
  Cannie, D. E., Syrris, P., Protonotarios, A., Bakalakos, A., Pruny, J. F., Ditaranto, R., Martinez-Veira, C., Larrañaga-Moreira, J. M., Medo, K., Bermúdez-Jiménez, F. J., Ben Yaou, R., Leturq, F., Mezcua, et al
  2023

- **Beyond the exome: What's next in diagnostic testing for Mendelian conditions.** *American journal of human genetics*
  Wojcik, M. H., Reuter, C. M., Marwaha, S., Mahmoud, M., Duyzend, M. H., Barseghyan, H., Yuan, B., Boone, P. M., Groopman, E. E., Délot, E. C., Jain, D., Sanchis-Juan, A., Starita, et al
  2023; 110 (8): 1229-1248

- **Proactive Variant Effect Mapping Aids Diagnosis in Pediatric Cardiac Arrest.** *Circulation. Genomic and precision medicine*
  Floyd, B. J., Weile, J., Kannankeril, P. J., Glazer, A. M., Reuter, C. M., MacRae, C. A., Ashley, E. A., Roden, D. M., Roth, F. P., Parikh, V. N.
  2023

- **Beyond the exome: what's next in diagnostic testing for Mendelian conditions.** *ArXiv*
  Wojcik, M. H., Reuter, C. M., Marwaha, S., Mahmoud, M., Duyzend, M. H., Barseghyan, H., Yuan, B., Boone, P. M., Groopman, E. E., Délot, E. C., Jain, D., Sanchis-Juan, A., Starita, et al
  2023

- **Variant Location Is a Novel Risk Factor for Individuals With Arrhythmogenic Cardiomyopathy Due to a Desmoplakin (DSP) Truncating Variant.** *Circulation. Genomic and precision medicine*
  Hoorntje, E. T., Burns, C., Marsili, L., Corden, B., Parikh, V. N., Te Meerman, G. J., Gray, B., Adiyaman, A., Bagnall, R. D., Barge-Schaapveld, D. Q., van den Berg, M. P., Bootsma, M., Bosman, et al
  2022: e003672

- **A concurrent dual analysis of genomic data augments diagnoses: experiences of two clinical sites in the Undiagnosed Diseases Network.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Spillmann, R. C., Tan, Q. K., Reuter, C., Schoch, K., Kohler, J., Bonner, D., Zastrow, D., Alkelai, A., Baugh, E., Cope, H., Marwaha, S., Wheeler, M. T., Bernstein, et al
  2022

- **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*
  Deuitch, 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., Baldridge, 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., Beleford, 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)

- **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

- **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

- **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

- **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., Cheatle-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 HypertrophicCardiomyopathy.** *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)