Stanford

Matthew Wheeler

Associate Professor of Medicine (Cardiovascular Medicine)

Medicine - Cardiovascular Medicine

CLINICAL OFFICE (PRIMARY)

- **Cardiovascular Medicine**

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Bio

BIO

I am a physician scientist with interests in cardiomyopathies, rare and undiagnosed diseases, therapeutics and genomics. I am a physician with interest and experience treating patients with hypertrophic cardiomyopathy, neuromuscular disease associated cardiomyopathies including adults with Duchenne and Becker muscular dystrophies, myotonic dystrophy, limb girdle muscular dystrophies, inherited dilated cardiomyopathies, advanced heart failure, mechanical circulatory support/ LVADs, heart and multiorgan transplant, and competitive athletes with cardiac disease. I am Director of the Stanford Center for Undiagnosed Diseases, a clinical site of the Undiagnosed Diseases Network.. I have extensive translational science efforts, as site PI for ongoing clinical trials for hypertrophic cardiomyopathy and dilated cardiomyopathy and for cardiomyopathy consortia including NONCOMPACT, PPCM and the Precision Medicine Study/DCM Consortium. I am Co-PI of the GREGoR Stanford Site, a research center of the GREGoR Consortium, and Co-PI of the NIH-funded Bioinformatics Center of the Molecular Transducers of Physical Activity Consortium. Our laboratory efforts focus on advancing diagnostic and therapeutic approaches in ultrarare diseases, with a focus on RNA-based diagnostics and therapeutics. I pursue projects and collaborations at the intersection of striated muscle genetics, genomics, therapeutics and clinical investigation.

CLINICAL FOCUS

- Cardiology
- Cardiomyopathy, Hypertrophic, Familial
- Heart Failure
- Mechanical Circulatory Support
- Undiagnosed Diseases
- Myotonic Dystrophy associated Cardiomyopathy
- Cardiovascular Genetics
- Cardiac Amyloid
- Heart Transplantation
- Arrhythmogenic Right Ventricular Cardiomyopathy
- Dilated Cardiomyopathies

- Muscular Dystrophy associated Cardiomyopathy
- Sports Cardiology
- Duchenne and Becker Muscular Dystrophy
- BAG3 associated cardiomyopathy
- Advanced Heart Failure and Transplant Cardiology

ACADEMIC APPOINTMENTS

- Associate Professor - University Medical Line, Medicine - Cardiovascular Medicine
- Member, Bio-X
- Member, Cardiovascular Institute
- Member, Wu Tsai Human Performance Alliance
- Member, Maternal & Child Health Research Institute (MCHRI)
- Member, Wu Tsai Neurosciences Institute

ADMINISTRATIVE APPOINTMENTS

- Director, Center for Undiagnosed Diseases at Stanford, (2014- present)
- Adult Medical Director, Center for Undiagnosed Diseases at Stanford, (2015- present)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Member, American Heart Association (2012 - present)
- Member, American College of Cardiology (2017 - present)
- Member, American Society of Human Genetics (2016 - present)

PROFESSIONAL EDUCATION

- Fellowship: Stanford University Advanced Heart Failure and Transplant Fellowship (2013) CA
- Fellowship: Stanford University Cardiovascular Medicine Fellowship (2012) CA
- Residency: Stanford University Internal Medicine Residency (2007) CA
- Medical Education: Pritzker School of Medicine University of Chicago Registrar (2005) IL
- Board Certification: Advanced Heart Failure and Transplant Cardiology, American Board of Internal Medicine (2014)
- Board Certification: Cardiovascular Disease, American Board of Internal Medicine (2012)
- Bachelor of Arts, Williams College , History and Biology (1998)

COMMUNITY AND INTERNATIONAL WORK

- Undiagnosed Diseases - Latin America

LINKS

- Wheeler lab: https://med.stanford.edu/mattlab.html
- Center for Undiagnosed Diseases at Stanford: http://undiagnosed.stanford.edu
- GREGoR Stanford Site: https://gregor.stanford.edu/
- MoTrPAC Data Hub: http://motrpac-data.org/
- Get a Second Opinion: https://stanfordhealthcare.org/second-opinion/overview.html

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

Translational research in rare and undiagnosed diseases. Basic and clinical research in cardiomyopathy genetics, mechanisms, screening, and treatment. Investigating novel agents for treatment of hypertrophic cardiomyopathy and new mechanisms in heart failure. Cardiovascular screening and genetics in competitive athletes, disease gene discovery in cardiomyopathy and rare disease. Informatics approaches to rare disease and multiomics. Molecular transducers of physical activity bioinformatics.

CLINICAL TRIALS

- A Prospective Registry Study to Assess Real-world Patient Characteristics, Treatment Patterns, and Longitudinal Outcomes in Patients Receiving Mavacamten and Other Treatments for Symptomatic Obstructive Hypertrophic Cardiomyopathy (Obstructive-HCM), Recruiting
- Clinical and Genetic Evaluation of Individuals With Undiagnosed Disorders Through the Undiagnosed Diseases Network, Recruiting
- International Consortium for Multimodality Phenotyping in Adults With Non-compaction, Recruiting
- A Study of ARRY-371797 (PF-07265803) in Patients With Symptomatic Dilated Cardiomyopathy Due to a Lamin A/C Gene Mutation, Not Recruiting
- A Study to Evaluate Mavacamten in Adults With Symptomatic Obstructive HCM Who Are Eligible for Septal Reduction Therapy, Not Recruiting
- Phase 3 Trial to Evaluate the Efficacy and Safety of Aficamten Compared to Placebo in Adults With Symptomatic oHCM (SEQUOIA-HCM), Not Recruiting
- Single-Ascending Dose Study of JK07 in Subjects With HFpEF, Not Recruiting
- Study of JK07 in Subjects With Heart Failure With Reduced Ejection Fraction (HFrEF), Not Recruiting

Teaching

STANFORD ADVISEES

Postdoctoral Faculty Sponsor

Pauline Brochet, Hector Mendez, Samuel Montalvo, Laurens van de Wiel

Postdoctoral Research Mentor

Laurens van de Wiel

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Cardiovascular Medicine (Fellowship Program)
- Human Genetics and Genetic Counseling (Masters Program)

Publications

PUBLICATIONS

- **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

- **Temporal dynamics of the multi-omic response to endurance exercise training.** *Nature*

  2024; 629 (8010): 174-183

- **The mitochondrial multi-omic response to exercise training across rat tissues.** *Cell metabolism*

Amar, D., Gay, N. R., Jimenez-Morales, D., Jean Beltran, P. M., Ramaker, M. E., Raja, A. N., Zhao, B., Sun, Y., Marwaha, S., Gaul, D. A., Hershman, S. G., Ferrasse, A., Xia, et al

2024

- **Effects of Mavacamten on Measures of Cardiopulmonary Exercise Testing Beyond Peak Oxygen Consumption: A Secondary Analysis of the EXPLORER-HCM Randomized Trial.** *JAMA cardiology*

  Wheeler, M. T., Olivotto, I., Elliott, P. M., Saberi, S., Owens, A. T., Maurer, M. S., Masri, A., Sehnert, A. J., Edelberg, J. M., Chen, Y., Florea, V., Malhotra, R., Wang, et al

  2023

- **Effect of beta-blocker therapy on the response to mavacamten in patients with symptomatic obstructive hypertrophic cardiomyopathy.** *European journal of heart failure*

  Wheeler, M. T., Jacoby, D., Elliott, P. M., Saberi, S., Hegde, S. M., Lakdawala, N. K., Myers, J., Sehnert, A. J., Edelberg, J. M., Li, W., Olivotto, I.

  2022

- **Guide to the female student athlete ECG: A comprehensive study of 3466 young, racially diverse athletes.** *The American journal of medicine*

  Harris, C. S., Froelicher, V. F., Wheeler, M. T.

  2022

- **2022 HRS expert consensus statement on evaluation and management of arrhythmic risk in neuromuscular disorders.** *Heart rhythm*

  Groh, W. J., Bhakta, D., Tomaselli, G. F., Aleong, R. G., Teixeira, R. A., Amato, A., Asirvatham, S. J., Cha, Y., Corrado, D., Duboc, D., Goldberger, Z. D., Horie, M., Hornyak, et al

  2022

- **Prevalence and Cumulative Risk of Familial Idiopathic Dilated Cardiomyopathy.** *JAMA*

  Huggins, G. S., Kinnamon, D. D., Haas, G. J., Jordan, E., Hofmeyer, M., Kransdorf, E., Ewald, G. A., Morris, A. A., Owens, A., Lowes, B., Stoller, D., Tang, W. H., Garg, et al

  1800; 327 (5): 454-463

- **TOWARDS TRANSCRIPTOMICS AS A PRIMARY TOOL FOR RARE DISEASE INVESTIGATION.** *Cold Spring Harbor molecular case studies*

  Montgomery, S. B., Bernstein, J. A., Wheeler, M. T.

  2022

- **Mono- and Biallelic Protein-Truncating Variants in Alpha-Actinin 2 Cause Cardiomyopathy Through Distinct Mechanisms.** *Circulation. Genomic and precision medicine*

  Lindholm, M. E., Jimenez-Morales, D., Zhu, H., Seo, K., Amar, D., Zhao, C., Raja, A., Madhvani, R., Abramowitz, S., Espenel, C., Sutton, S., Caleshu, C., Berry, et al

  2021: CIRCGEN121003419

- **Variable clinical severity in TANGO2 deficiency: Case series and literature review.** *American journal of medical genetics. Part A*

  Schymick, J., Leahy, P., Cowan, T., Ruzhnikov, M. R., Gates, R., Fernandez, L., Pramanik, G., Undiagnosed Diseases Network, Yarlagadda, V., Wheeler, M., Bernstein, J. A., Enns, G. M., Lee, C.

  2021

- **The genetics of human performance.** *Nature reviews. Genetics*

  Kim, D. S., Wheeler, M. T., Ashley, E. A.

  2021

- **Mapping the human genetic architecture of COVID-19.** *Nature*

  COVID-19 Host Genetics Initiative

  2021

- **Findings From Cardiovascular Evaluation of National Collegiate Athletic Association Division I Collegiate Student-Athletes After Asymptomatic or Mildly Symptomatic SARS-CoV-2 Infection.** *Clinical journal of sport medicine : official journal of the Canadian Academy of Sport Medicine*

  Hwang, C. E., Kussman, A., Christle, J. W., Froelicher, V., Wheeler, M. T., Moneghetti, K. J.

  2021

- **Mavacamten for treatment of symptomatic obstructive hypertrophic cardiomyopathy (EXPLORER-HCM): a randomised, double-blind, placebo-controlled, phase 3 trial.** *Lancet (London, England)*

  Olivotto, I., Oreziak, A., Barriales-Villa, R., Abraham, T. P., Masri, A., Garcia-Pavia, P., Saberi, S., Lakdawala, N. K., Wheeler, M. T., Owens, A., Kubanek, M., Wojakowski, W., Jensen, et al

2020

- **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

- **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

- **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

- **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

- **Effect of Moderate-Intensity Exercise Training on Peak Oxygen Consumption in Patients With Hypertrophic Cardiomyopathy A Randomized Clinical Trial** *JAMA-JOURNAL OF THE AMERICAN MEDICAL ASSOCIATION*
  Saberi, S., Wheeler, M., Bragg-Gresham, J., Hornsby, W., Agarwal, P. P., Attili, A., Concannon, M., Dries, A. M., Shmargad, Y., Salisbury, H., Kumar, S., Herrera, J., Myers, et al
  2017; 317 (13): 1349-1357

- **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)

- **The Undiagnosed Diseases Network: Characteristics of solvable applicants and diagnostic suggestions for non-accepted ones.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Mulvihill, J. J., Findley, L., Ni, W., Sinsheimer, J. S., Cole, F. S., Esteves, C., Bernstein, J. A., Newman, J. H., Wheeler, M. T., Mokry, J. R.
  2024: 101203

- **Impact of genome build on RNA-seq interpretation and diagnostics.** *American journal of human genetics*
  Ungar, R. A., Goddard, P. C., Jensen, T. D., Degalez, F., Smith, K. S., Jin, C. A., Bonner, D. E., Bernstein, J. A., Wheeler, M. T., Montgomery, S. B.
  2024

- **The impact of exercise on gene regulation in association with complex trait genetics.** *Nature communications*
  Vetr, N. G., Gay, N. R., MoTrPAC Study Group, Montgomery, S. B., Adkins, J. N., Albertson, B. G., Amar, D., Amper, M. A., Armenteros, J. J., Ashley, E., Avila-Pacheco, J., Bae, D., Balci, A. T., et al
  2024; 15 (1): 3346

- **Sexual dimorphism and the multi-omic response to exercise training in rat subcutaneous white adipose tissue.** *Nature metabolism*
  Many, G. M., Sanford, J. A., Sagendorf, T. J., Hou, Z., Nigro, P., Whytock, K. L., Amar, D., Caputo, T., Gay, N. R., Gaul, D. A., Hirshman, M. F., Jimenez-Morales, D., Lindholm, et al
  2024

- **Molecular Transducers of Physical Activity Consortium (MoTrPAC): Human Studies Design and Protocol.** *Journal of applied physiology (Bethesda, Md. : 1985)*
  Group, M. R., Jakicic, J. M., Kohrt, W. M., Houmard, J. A., Miller, M. E., Radom-Aizik, S., Rasmussen, B. B., Ravussin, E., Serra, M., Stowe, C. L., Trappe, S., AbouAssi, H., Adkins, et al
  2024

- **Immunological and hematological findings as major features in a patient with a new germline pathogenic CBL variant.** *American journal of medical genetics. Part A*
  Stellacci, E., Carter, J. N., Pannone, L., Stevenson, D., Moslehi, D., Venanzi, S., Bernstein, J. A., Tartaglia, M., Martinelli, S.

2024: e63627

- **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

- **Quantifying assumptions underlying peak oxygen consumption equations across the body mass spectrum.** *Clinical obesity*
  Busque, V., Christle, J. W., Moneghetti, K. J., Cauwenberghs, N., Kouznetsova, T., Blumberg, Y., Wheeler, M. T., Ashley, E., Haddad, F., Myers, J.
  2024: e12653

- **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

- **Prevalence of frequent premature ventricular contractions and nonsustained ventricular tachycardia in older women screened for atrial fibrillation in the Women's Health Initiative.** *Heart rhythm*
  Gomez, S. E., Larson, J., Hlatky, M. A., Rodriguez, F., Wheeler, M., Greenland, P., LaMonte, M., Froelicher, V., Stefanick, M. L., Wallace, R., Kooperberg, C., Tinker, L. F., Schoenberg, et al
  2024

- **Digitized Electrocardiography Measurements Support the Biological Plausibility of the Pathological Significance of ST Segments in Athletes.** *Clinical journal of sport medicine : official journal of the Canadian Academy of Sport Medicine*
  Montalvo, S., Froelicher, V. F., Hadley, D., Wheeler, M. T.
  2024

- **Recurring homozygous ACTN2 variant (p.Arg506Gly) causes a recessive myopathy.** *Annals of clinical and translational neurology*
  Donkervoort, S., Mohassel, P., O'Leary, M., Bonner, D. E., Hartley, T., Acquaye, N., Brull, A., Mozaffar, T., Saporta, M. A., Dyment, D. A., Sampson, J. B., Pajusalu, S., Austin-Tse, et al
  2024

- **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

- **Improving Reporting of Exercise Capacity Across Age Ranges Using Novel Workload Reference Equations.** *The American journal of cardiology*
  Santana, E. J., Christle, J. W., Cauwenberghs, N., Peterman, J. E., Busque, V., Gomes, B., Bagherzadeh, S. P., Moneghetti, K., Kuznetsova, T., Wheeler, M., Ashley, E., Harber, M. P., Arena, et al
  2024

- **Impact of genome build on RNA-seq interpretation and diagnostics.** *medRxiv : the preprint server for health sciences*
  Ungar, R. A., Goddard, P. C., Jensen, T. D., Degalez, F., Smith, K. S., Jin, C. A., Bonner, D. E., Bernstein, J. A., Wheeler, M. T., Montgomery, S. B.
  2024

- **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

- **Mavacamten Treatment for Symptomatic Obstructive Hypertrophic Cardiomyopathy: Interim Results From the MAVA-LTE Study, EXPLORER-LTE Cohort.** *JACC. Heart failure*

  Rader, F., Or#ziak, A., Choudhury, L., Saberi, S., Fermin, D., Wheeler, M. T., Abraham, T. P., Garcia-Pavia, P., Zwas, D. R., Masri, A., Owens, A., Hegde, S. M., Seidler, et al

  2024; 12 (1): 164-177

- **Biallelic variants in POLR3A encoding catalytic subunit of human RNA polymerase III cause primary microcephaly through perturbation of the mTOR signaling pathway**

  Makhdoom, E., Asif, M., Abu Bakar, M., Sheraz, K., Alawbethani, S., Hoehne, W., Anjum, I., Fatima, A., Baig, J., Georgomanolis, T., Budde, B., Irshad, S., Zastrow, et al

  SPRINGERNATURE.2024: 504-505

- **Quantitative metrics of the LV trabeculated layer by cardiac CT and cardiac MRI in patients with suspected noncompaction cardiomyopathy.** *European radiology*

  Manohar, A., Vigneault, D. M., Kwon, D. H., Caliskan, K., Budde, R. P., Hirsch, A., Lee, S. P., Lee, W., Owens, A., Litt, H., Haddad, F., Mistelbauer, G., Wheeler, et al

  2023

- **Assessing the Assisted Six-Minute Cycling Test as a Measure of Endurance in Non-Ambulatory Patients with Spinal Muscular Atrophy (SMA).** *Journal of clinical medicine*

  Tang, W. J., Gu, B., Montalvo, S., Dunaway Young, S., Parker, D. M., de Monts, C., Ataide, P., Ni Ghiollagain, N., Wheeler, M. T., Tesi Rocha, C., Christle, J. W., He, Z., Day, et al

  2023; 12 (24)

- **Long-term effects of mavacamten treatment in obstructive hypertrophic cardiomyopathy (HCM): updated cumulative analysis of the EXPLORER cohort of MAVA-long-term extension (LTE) study up to 120 weeks**

  Garcia-Pavia, P., Oreziak, A., Masri, A., Barriales-Villa, R., Abraham, T. P., Owens, A. T., Lakdawala, N. K., Saberi, S., Wang, A., Wheeler, M. T., Choudhury, L., Balaratnam, G., Fox, et al

  OXFORD UNIV PRESS.2023

- **Changes in standard of care (SOC) medication during long-term mavacamten treatment for obstructive hypertrophic cardiomyopathy (HCM): results from the EXPLORER cohort of MAVA-Long-Term Extension (LTE)**

  Lakdawala, N., Afshar, K., Barriales-Villa, R., Gimeno-Blanes, J., Michels, M., Saberi, S., Wheeler, M. T., Balaratnam, G., Shah, A., Chen, Y. M., Sehnert, A. J., Abraham, T. P.

  OXFORD UNIV PRESS.2023

- **Improved Cardiac Performance and Decreased Arrhythmia in Hypertrophic Cardiomyopathy With Non-#-Blocking R-Enantiomer Carvedilol.** *Circulation*

  Seo, K., Yamamoto, Y., Kirillova, A., Kawana, M., Yadav, S., Huang, Y., Wang, Q., Lane, K. V., Pruitt, B. L., Perez, M. V., Bernstein, D., Wu, J. C., Wheeler, et al

  2023

- **The functional impact of rare variation across the regulatory cascade.** *Cell genomics*

  Li, T., Ferraro, N., Strober, B. J., Aguet, F., Kasela, S., Arvanitis, M., Ni, B., Wiel, L., Hershberg, E., Ardlie, K., Arking, D. E., Beer, R. L., Brody, et al

  2023; 3 (10): 100401

- **Assessment of maximal effort for weaker individuals with NMD during the assisted six-minute cycling test**

  Blumberg, Y., De Monts, C., Tang, W., Montalvo, S., Ataide, P., Young, S., Wheeler, M., Ashley, E., Myers, J., Day, J., Christle, J., Duong, T.

  PERGAMON-ELSEVIER SCIENCE LTD.2023: S187

- **Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care** *JOURNAL OF PEDIATRICS*

  Halley, M. C., Young, J. L., Tang, C., Mintz, K. T., Lucas-Griffin, S., Maghiro, A., Ashley, E. A., Tabor, H. K., Undiagnosed Diseases Network

  2023; 261

- **Personalized digital behaviour interventions increase short-term physical activity: a randomized control crossover trial substudy of the MyHeart Counts Cardiovascular Health Study.** *European heart journal. Digital health*

  Javed, A., Kim, D. S., Hershman, S. G., Shcherbina, A., Johnson, A., Tolas, A., O'Sullivan, J. W., McConnell, M. V., Lazzeroni, L., King, A. C., Christle, J. W., Oppezzo, M., Mattsson, et al

  2023; 4 (5): 411-419

- **Generation of two induced pluripotent stem cell lines from Duchenne muscular dystrophy patients.** *Stem cell research*

  Liu, W., Zeng, W., Kong, X., Htet, M., Yu, R., Wheeler, M., Day, J. W., Wu, J. C.

2023; 72: 103207

- **Cardiac Remodeling in Subclinical Hypertrophic Cardiomyopathy: The VANISH Randomized Clinical Trial.** *JAMA cardiology*
  Vissing, C. R., Axelsson Raja, A., Day, S. M., Russell, M. W., Zahka, K., Lever, H. M., Pereira, A. C., Colan, S. D., Margossian, R., Murphy, A. M., Canter, C., Bach, R. G., Wheeler, et al
  2023
- **A defect in mitochondrial fatty acid synthesis impairs iron metabolism and causes elevated ceramide levels.** *Nature metabolism*
  Dutta, D., Kanca, O., Byeon, S. K., Marcogliese, P. C., Zuo, Z., Shridharan, R. V., Park, J. H., Lin, G., Ge, M., Heimer, G., Kohler, J. N., Wheeler, M. T., Kaipparettu, et al
  2023
- **Rare Variant Genetics and Dilated Cardiomyopathy Severity: The DCM Precision Medicine Study.** *Circulation*
  Hofmeyer, M., Haas, G. J., Jordan, E., Cao, J., Kransdorf, E., Ewald, G. A., Morris, A. A., Owens, A., Lowes, B., Stoller, D., Wilson Tang, W. H., Garg, S., Trachtenberg, 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
- **Genetic Architecture of Dilated Cardiomyopathy in Individuals of African and European Ancestry.** *JAMA*
  Jordan, E., Kinnamon, D. D., Haas, G. J., Hofmeyer, M., Kransdorf, E., Ewald, G. A., Morris, A. A., Owens, A., Lowes, B., Stoller, D., Tang, W. H., Garg, S., Trachtenberg, et al
  2023; 330 (5): 432-441
- **Assessment of Oxygen Pulse in Hypertrophic Cardiomyopathy-Reply.** *JAMA cardiology*
  Wheeler, M. T., Chen, Y. M., Myers, J.
  2023
- **Respiratory gas kinetics in patients with congestive heart failure during recovery from peak exercise.** *Clinics (Sao Paulo, Brazil)*
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