



## Matthew T. Wheeler

Associate Professor of Medicine (Cardiovascular Medicine)  
Medicine - Cardiovascular Medicine

### CLINICAL OFFICE (PRIMARY)

- **Cardiovascular Medicine**

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### Bio

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#### BIO

Dr Wheeler is a physician scientist with interests in cardiomyopathies, rare and undiagnosed diseases, therapeutics and genomics. He has 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. He is Director of the Stanford Center for Undiagnosed Diseases, a clinical site of the Undiagnosed Diseases Network. He has extensive translational science efforts, as site PI for clinical trials for hypertrophic cardiomyopathy and dilated cardiomyopathy and for cardiomyopathy consortia including NONCOMPACT, PPCM and the Precision Medicine Study/DCM Consortium. He is 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. His laboratory efforts focus on advancing diagnostic and therapeutic approaches in ultrarare diseases, with a focus on iPSC models, RNA-based diagnostics and therapeutics. He pursues projects and collaborations at the intersection of rare disease genetics, genomics, therapeutics and clinical investigation.

#### CLINICAL FOCUS

- Cardiology
- Cardiomyopathy, Hypertrophic, Familial
- Cardiovascular Genetics
- Myotonic Dystrophy associated Cardiomyopathy
- Heart Failure
- Undiagnosed Diseases
- Mechanical Circulatory Support
- Cardiac Amyloid
- Heart Transplantation
- Muscular Dystrophy associated Cardiomyopathy

- Arrhythmogenic Right Ventricular Cardiomyopathy
- Rare Diseases
- Dilated Cardiomyopathies
- 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

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### 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

- Clinical and Genetic Evaluation of Individuals With Undiagnosed Disorders Through the Undiagnosed Diseases Network, Recruiting
- NONCOMPACT, Recruiting
- Study of JK07 in Patients With Chronic Heart Failure, Recruiting
- ARRY Lamin Gene Mutation, Not Recruiting
- CLCZ69612201, Not Recruiting
- DCM Precision Medicine Study sIRB, Not Recruiting
- Discover-HCM, Not Recruiting
- EXPLORER, Not Recruiting
- JK07.1.02 HFpEF, Not Recruiting
- Manage HF+, Not Recruiting
- MAVERICK, Not Recruiting
- Salubris JK07.1.01, Not Recruiting
- Valor- MYK017, Not Recruiting

## Teaching

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### STANFORD ADVISEES

#### Postdoctoral Faculty Sponsor

Allison Cheney, Eric Leslie, Hector Mendez, Samuel Montalvo, Laurens van de Wiel

#### Postdoctoral Research Mentor

Laurens van de Wiel

### GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Biomedical Data Science (Phd Program)
- Cardiovascular Medicine (Fellowship Program)
- Genetics (Phd Program)
- Human Genetics and Genetic Counseling (Masters Program)
- Medical Genetics (Fellowship Program)
- Molecular and Genetic Medicine (Fellowship Program)
- Neurosciences (Phd Program)
- Stem Cell Biology and Regenerative Medicine (Phd Program)

## Publications

### PUBLICATIONS

- **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
- **Distinct Mechanistic Features of Atrial Fibrillation in Hypertrophic Cardiomyopathy.** *Heart rhythm*  
Liu, M., Huang, S., Kawana, M., Wheeler, M. T., Parikh, V., Perez, M. V.  
2026
- **Biallelic variants in RNU2-2 cause the most prevalent known recessive neurodevelopmental disorder.** *Nature genetics*  
Greene, D., Mendez, R., Lees, J., Barbosa, M., Bruselles, A., Chiriatti, L., Ferraro, F., Mancini, C., Schot, R., Sleutels, F., Bertini, E., Bonner, D. E., Bouman, et al  
2026
- **Biallelic Variants in RNU6ATAC Result in a Minor Spliceopathy Characterized by Transcriptome-Wide Minor Intron Retention Events and Short Stature with Variable Multisystem Manifestations.** *HGG advances*  
Mendez, R., Arriaga, T. M., Ma, J., Bonner, D. E., Emami, S., Levy, R. J., Alsagheir, A., Alhaddad, B., Bakur, K., Ungar, R. A., Matalon, D. R., Miller, A. M., Nguyen, et al  
2026: 100588
- **Biallelic LAMP3 Variants in Five Families with Interstitial Lung Disease: Evidence of a Disease-Gene Association.** *Genetics in medicine : official journal of the American College of Medical Genetics*  
Keehan, L. A., Ono-Minagi, H., Hadhud, M., Rips, J., Hinds, D. M., Fischer, A. J., Bartlett, J. A., McCray, P. B., Qawasmi, N., Nathan, N., Louvrier, C., Desroziers, T., Damme, et al  
2026: 102531
- **GREGoR: accelerating genomics for rare diseases.** *Nature*  
Dawood, M., Heavner, B., Wheeler, M. M., Ungar, R. A., LoTempio, J., Wiel, L., Berger, S., Bernstein, J. A., Chong, J. X., Délot, E. C., Eichler, E. E., Lupski, J. R., Shojaie, et al  
2025; 647 (8089): 331-342
- **An optimized variant prioritization process for rare disease diagnostics: recommendations for Exomiser and Genomiser.** *Genome medicine*  
Cooperstein, I. B., Marwaha, S., Ward, A., Kobren, S. N., Carter, J. N., Wheeler, M. T., Marth, G. T.  
2025; 17 (1): 127
- **Transcriptome-wide outlier approach identifies individuals with minor spliceopathies.** *American journal of human genetics*  
Arriaga, T. M., Mendez, R., Ungar, R. A., Bonner, D. E., Matalon, D. R., Lemire, G., Goddard, P. C., Padhi, E. M., Miller, A. M., Nguyen, J. V., Ma, J., Smith, K. S., Scott, et al  
2025
- **Endurance Training Enhances Sex-Specific Cardioprotective Metabolism.** *Circulation research*  
Brochet, P., Montalvo, S., Lindholm, M. E., Jimenez-Morales, D., Jin, C. A., Rasmussen, B. B., Kraus, W. E., Yan, Z., Wheeler, M. T., Katz, D. H.  
2025
- **Cardiac CT fractal analysis of LV noncompaction and common cardiomyopathies.** *Journal of cardiovascular computed tomography*  
Manohar, A., Wong, A., Castillo, E., Gunderson, A., Mistelbauer, G., Bagherzadeh, S. P., Haddad, F., Caliskan, K., Budde, R. P., Hirsch, A., Lee, S. P., Lee, W., Owens, et al  
2025
- **Single Ascending-Dose Study of Selective ErbB4 Agonist JK07 in Heart Failure With Reduced Ejection Fraction.** *JACC. Basic to translational science*  
Tang, W. H., Steiner, J., Kassi, M., Wheeler, M. T., Spahillari, A., Sweitzer, N. K., Grodin, J. L., Solomon, N., Singhal, S., McEwen, A. M., Murphy, S. L.  
2025; 10 (9): 101352

- **Identification of candidate cardiomyopathy modifier genes through genome sequencing and RNA profiling.** *Frontiers in cardiovascular medicine*  
Lindholm, M. E., Abramowitz, S., Waggott, D. M., Grove, M. E., Dewey, F. E., Pan, C., Pavlovic, A., Shang, C., Huang, Y., Bensabath, L., Goldfeder, R. L., Cordero, P., Erbilgin, et al  
2025; 12: 1546493
- **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
- **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
- **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
- **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., Baldrige, 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., Homiyak, 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
- **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
  - **The genetics of human performance.** *Nature reviews. Genetics*  
Kim, D. S., Wheeler, M. T., Ashley, E. A.  
2021
  - **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*  
Saber, 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
  - **Sex and Age Specific Genetic Risk Across the Dilated and Arrhythmogenic Cardiomyopathy Spectrum: Insights From the SHARe Registry.** *Journal of the American College of Cardiology*  
Stroeks, S. L., Bart, N. K., Rossano, J., Claggett, B., Beelen, N. J., Buchan, R. J., Day, S., Fornaro, A., Halliday, B. P., Wheeler, M. T., Hammersley, D. J., Helms, A., Heymans, et al  
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
  - **Women's Health Initiative Strong and Healthy Silent Atrial Fibrillation Recording Study: Rationale, Study Design and Baseline Data.** *American heart journal*  
Gomez, S. E., Larson, J., Hlatky, M. A., Kooperberg, C., LaMonte, M., Tinker, L., Greenland, P., Albert, C., Froelicher, V., Wheeler, M., Stefanick, M. L., Perez, M. V.  
2026: 107478
  - **Defining baseline high-sensitivity cardiac troponin levels in healthy collegiate athletes.** *American journal of clinical pathology*  
Jamal, Y. A., Montiel, A., Jones, S., Nair, P., Parikh, V., Haddad, F., Wheeler, M. T., Hwang, C., Tso, J. V., Bowen, R. A.  
2026; 165 (5)
  - **Publisher Correction: The impact of exercise on gene regulation in association with complex trait genetics.** *Nature communications*  
Vetr, N. G., Gay, N. R., Montgomery, S. B.  
2026; 17 (1)

- **The Long Non-coding RNA Landscape of Endurance Exercise Training.** *Molecular metabolism*  
Bonilauri, B., Smith, G. R., Raja, A. N., Jimenez-Morales, D., Ahmed, A., Jin, C., Sparks, L. M., Walsh, M. J., Montgomery, S. B., Bodine, S. C., Ashley, E. A., Lindholm, M. E.  
2026: 102358
- **De Novo Variants Associated With Autosomal Recessive Conditions: Case Series and Implications for Genetic Testing and Counseling.** *American journal of medical genetics. Part A*  
Niehaus, A. D., Bonner, D. E., Carter, J., Avello, K., Jacob, N., Neu, M. B., Mendez, R., Qiao, W., Scott, S. A., Levy, R. J., Mattas, L., Schymick, J., Van Anandel, et al  
2026
- **The Common Fund Data Ecosystem (CFDE).** *bioRxiv : the preprint server for biology*  
Jurgens, J. A., Bueckle, A., Vora, J., Maurya, M. R., Ahooyi, T. M., Zheng, E., Stear, B., Wang, D., Ree, C., Ramachandran, S., Nekrutenko, A., Brandes, M., Thaker, et al  
2026
- **EVALUATION OF VENTILATORY THRESHOLDS DURING EXERCISE THROUGH THE PERCUTANEOUS OXYGEN SATURATION METHOD**  
Parsa, S., Abe, M., Christle, J., Myers, J. N., Wheeler, M., Kawana, M.  
ELSEVIER SCIENCE INC.2026: A1242
- **Generation and characterization of two human induced pluripotent stem cell lines from myotonic dystrophy type 1 patients.** *Stem cell research*  
Darji, P., Liu, W., Zeng, W., Chao, J. T., Pang, P. D., Wheeler, M. T., Perez, M., Wu, J. C.  
2026; 94: 103977
- **Generation of two induced pluripotent stem cell lines from hypertrophic cardiomyopathy patients carrying MYBPC3 mutations.** *Stem cell research*  
Wu, C. A., Wu, M. A., Zhao, S. R., Sun, J., Flores-Banuelos, A. G., Walther, P., Wheeler, M., Sayed, N.  
2026; 94: 103982
- **Multi-omic identification of key transcriptional regulatory programs during endurance exercise training in rats.** *Nature communications*  
Smith, G. R., Zhao, B., Lindholm, M. E., Raja, A., Viggars, M., Pincas, H., Gay, N. R., Sun, Y., Vangeti, S., Ge, Y., Nair, V. D., Sanford, J. A., Amper, et al  
2026
- **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)
- **Genetic Diagnosis and Discovery Enabled by Large Language Models.** *Advanced science (Weinheim, Baden-Wurtemberg, Germany)*  
Tu, T., Saab, K., Liu, W., Fang, Z., Cheng, Z., Spasic, S., Djuricic, M., Mohri, H., Ren, W., Palepu, A., Gottweis, J., Karthikesalingam, A., Kulkarni, et al  
2026: e18656
- **Generation of induced pluripotent stem cell lines from patients with Emery-Dreifuss muscular dystrophy.** *Stem cell research*  
Chorsi, M. S., Mui, B. J., Tan, R., Fernandez, H. M., Xu, L., Chao, J. T., Sampson, J. B., Wheeler, M. T., Wu, J. C.  
2026; 91: 103926
- **Assessment Of Ventilatory Thresholds During Exercise Using Percutaneous Oxygen Saturation Method**  
Parsa, S., Abe, M., Christle, J., Myers, J., Wheeler, M., Kawana, M.  
CHURCHILL LIVINGSTONE INC MEDICAL PUBLISHERS.2026
- **Spatial T1-Mapping of Cardiac Fibrosis Identifies Causal Protein Drivers through Deep Learning and Mendelian Randomization**  
Reddy, S., Loong, S., Xia, R., Chen, E., Cao, F., Steffner, K., Wheeler, M., Ashley, E., Gomes Botelho Quintas, B.  
LIPPINCOTT WILLIAMS & WILKINS.2025
- **"Nobody listened to us for years": Parents' experiences of provider communication in the diagnostic odyssey.** *Molecular genetics and metabolism*  
Nguyen, M. M., Mahfoozi, S., Bonner, D., Martschenko, D. O., Giri, A., Tang, C., Bernstein, J. A., Wheeler, M. T., Halley, M. C.

2025; 146 (4): 109283

- **RAPSN-Associated Congenital Myasthenic Syndrome due to Biallelic Single Nucleotide Variants at the Same Position.** *Case reports in genetics*  
Keehan, L., Carter, J. N., Kravets, E., Wheeler, M. T., Bernstein, J. A., Maselli, R. A., Sampson, J. B., Bachir, S.  
2025; 2025: 1882021
- **Sequentially constrained randomization in preclinical animal studies.** *Function (Oxford, England)*  
Rigdon, J., Walkup, M., Amar, D., Wheeler, M. T., Goodyear, L. J., Bodine, S., Esser, K., Esserman, D., Miller, M. E.  
2025
- **Assessing function and metabolic cost: development of the neuromuscular disease specific activity questionnaire (NAQ)**  
Duong, T., Wilcox, R., de Monts, C., Christle, J., Tang, W., Khonde, S., Wheeler, M., Ashley, E., Birkmeier, M., Meyer, J.  
PERGAMON-ELSEVIER SCIENCE LTD.2025
- **Prediction of VO2max and cardiopulmonary responses from ventilatory threshold VO2 in neuromuscular disease patients using machine learning**  
Duong, T., Montalvo, S., de Monts, C., Blumberg, Y., Vogt-Domke, S., Salvatore, S., Tang, W., Meyer, J., Wheeler, M., Day, J. W., Hageman, N., Christle, J.  
PERGAMON-ELSEVIER SCIENCE LTD.2025
- **Delayed exercise recovery ventilatory responses between individuals with neuromuscular disease**  
Duong, T., Montalvo, S., de Monts, C., Blumberg, Y., Vogt-Domke, S., Salvatore, S., Tang, W., Young, S., Wheeler, M., Day, J., Hageman, N., Taivassalo, T., Christle, et al  
PERGAMON-ELSEVIER SCIENCE LTD.2025
- **Biallelic variants in RNU2-2 cause the most prevalent known recessive neurodevelopmental disorder.** *medRxiv : the preprint server for health sciences*  
Greene, D., Mendez, R., Lees, J., Barbosa, M., Bruselles, A., Chiriatti, L., Mancini, C., Bertini, E. S., Bonner, D. E., Cassini, T. A., Ezell, K. M., Gomez-Ospina, N., Rives, et al  
2025
- **Implementing Precision Medicine for Dilated Cardiomyopathy: Insights From the DCM Consortium.** *Circulation. Genomic and precision medicine*  
Jordan, E., Ni, H., Parker, P., Kinnamon, D. D., Owens, A., Lowes, B., Shenoy, C., Martin, C. M., Judge, D. P., Fishbein, D. P., Stoller, D., Minami, E., Kransdorf, et al  
2025: e005078
- **Cardiopulmonary Recovery After Maximal Exercise in Individuals with Neuromuscular Disease and Limited Mobility.** *Journal of clinical medicine*  
Blumberg, Y., de Monts, C., Montalvo, S., Tang, W. J., Dunaway Young, S., Hageman, N., Sanchis-Gomar, F., Ashley, E. A., Amar, D., Myers, J., Wheeler, M. T., Day, J. W., Duong, et al  
2025; 14 (12)
- **Biomni: A General-Purpose Biomedical AI Agent.** *bioRxiv : the preprint server for biology*  
Huang, K., Zhang, S., Wang, H., Qu, Y., Lu, Y., Roohani, Y., Li, R., Qiu, L., Li, G., Zhang, J., Yin, D., Marwaha, S., Carter, et al  
2025
- **Scaled multidimensional assays of variant effect identify sequence-function relationships in hypertrophic cardiomyopathy.** *bioRxiv : the preprint server for biology*  
Yamamoto, Y., Chua, K., Ferrasse, A., Kirilova, A., De Jong, H. N., Floyd, B. J., Cadisch, C., Wiel, L., Wang, Q., O'Neill, M. J., Tabet, D., Staudt, D., Goryznski, et al  
2025
- **Proteomic Analysis of Valsartan for Attenuating Disease Evolution in Early Sarcomeric Hypertrophic Cardiomyopathy (VANISH) Clinical Trial.** *Circulation. Heart failure*  
Topriceanu, C. C., Vissing, C. R., Axelsson Raja, A., Day, S. M., Russell, M. W., Zahka, K., Pereira, A. C., Colan, S. D., Murphy, A. M., Canter, C., Bach, R. G., Wheeler, M. T., Rossano, et al  
2025: e012393
- **Prognostic Value of Compact Myocardial Thinning in Patients with Left Ventricular Non-compaction.** *The American journal of cardiology*

- Gegenava, T., Tukker, M., Caliskan, K., Hirsch, A., Manohar, A., Lee, S. P., Owens, A., Kwon, D. H., Ramchand, J., Wheeler, M. T., Tang, W. H., Nieman, K.  
2025
- **Sex-specific electrocardiographic criteria for left ventricular hypertrophy in young athletes.** *Heart rhythm*  
Montalvo, S., Tso, J. V., Christle, J. W., Hadley, D., Wheeler, M., Perez, M., Myers, J., Ashley, E., Froelicher, V.  
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  - **Proposed enhanced recommendations for interpretation of electrocardiographic screening of athletes.** *Progress in cardiovascular diseases*  
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