Stanford



Matthew T. Wheeler

Associate Professor of Medicine (Cardiovascular Medicine)

Medicine - Cardiovascular Medicine

CLINICAL OFFICE (PRIMARY)

- **Cardiovascular Medicine**

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Bio

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 Prof-Univ Med 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

- 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
- Study of JK07 in Patients With Chronic Heart Failure, Recruiting
- A Mobile Application to Promote Self-management and Improve Outcomes in Heart Failure, Not Recruiting
- A Phase 2 Study of Mavacamten in Adults With Symptomatic Non-Obstructive Hypertrophic Cardiomyopathy (nHCM), Not Recruiting
- 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), Not 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
- Clinical Study to Evaluate Mavacamten (MYK-461) in Adults With Symptomatic Obstructive Hypertrophic Cardiomyopathy, Not Recruiting
- DCM Precision Medicine Study, Not Recruiting
- Single-Ascending Dose Study of JK07 in Subjects With HFpEF, Not Recruiting
- Study of Efficacy of Oral Sacubitril/Valsartan in Adult Patients With Non-obstructive Hypertrophic Cardiomyopathy, Not Recruiting
- Study of JK07 in Subjects With Heart Failure With Reduced Ejection Fraction (HFrEF), Not Recruiting

Teaching

STANFORD ADVISEES

Postdoctoral Faculty Sponsor

Allison Cheney, Samuel Montalvo

Doctoral Dissertation Reader (NonAC)

Esther Robb

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

- **Autonomous biomedical research with an artificial intelligence agent.** *Science (New York, N.Y.)*
  Huang, K., Zhang, S., Wang, H., Qu, Y., Lu, Y., Li, R., Roohani, Y., Qiu, L., Cao, S., Li, G., Zhang, J., Yin, D., Wierenga, et al
  2026: eadz4351

- **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 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

- **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., 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

- **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*
  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

- **Normative Values for QRS Duration in Athletes aged 12-35 years undergoing Pre-participation Exams.** *Heart rhythm*
  Montalvo, S., Tso, J. V., Ferrari, F., Ashley, E., Wheeler, M., Froelicher, V. F.
  2026

- **Interplay Between Body Mass Index and Patient-Reported Health Status in Early-Stage Hypertrophic Cardiomyopathy**
  Ostrominski, J., Claggett, B., Axelsson Raja, A., Day, S., Russell, M., Zahka, K. G., Pereira, A., Colan, S. D., Murphy, A. M., Canter, C., Bach, R. G., Wheeler, M., Rossano, et al
  LIPPINCOTT WILLIAMS & WILKINS.2026: A4980

- **Non-Invasive CVP Measurement Predicts Length of Stay and Readmission in Hospitalized Heart Failure Patients: Results of the ezCVP-HF2 Study**
  Torelli, S., Aung, P., Wu, E., Verma, A., Nguyen, V., Gjermeni, D., Cheng, P., Morimoto, H., Wheeler, M., Nguyen, P. K., Kawana, M.
  LIPPINCOTT WILLIAMS & WILKINS.2026: A16149

- **Acute Exercise Drives Transient Immune Dysregulation: Immune Score Analysis in the MoTrPAC Cohort**
  Brochet, P., Smith, G., Jin, C., Jimenez-Morales, D., Many, G., Lindholm, M. E., Smith, K., Khatri, P., Sealfon, S., Walsh, M., Montgomery, S. B., Snyder, M., Wheeler, et al
  LIPPINCOTT WILLIAMS & WILKINS.2026: A18787

- **Effectiveness and Safety of Mavacamten Across Sarcomere Gene Variant Subgroups: Evidence from MARVEL-HCM**
  Alsidawi, S., Owens, A. T., Wharton, R., Martinez, M. W., Mehra, V. C., Wheeler, M., de Feria, A., Reza, N., Emery, M., Belvin, T., Wang, A., Kim, M., Maksabedian Hernandez, et al
  LIPPINCOTT WILLIAMS & WILKINS.2026: A6743

- **Resting Diastolic Dysfunction Predicts Ventilatory Inefficiency and Clinical Outcomes in Hypertrophic Cardiomyopathy**
  Pepin, M., Osullivan, J. W., Gjermeni, D., Santana, E., Mosher, B., Malunjkar, S., Tso, J., Lewis, E. F., Ashley, E., Christle, J., Myers, J., Parikh, V. N., Kawana, et al
  LIPPINCOTT WILLIAMS & WILKINS.2026: A4638

- **Multi-Omic Integration Improves Prediction of Cardiorespiratory Fitness**
  Vetr, N., Montalvo, S., Jaeger, B., Rao, P., Many, G., Amar, D., Jin, C., Robbins, J. M., Burant, C., Gerszten, R. E., Moreau, K., Hastie, T., Tibshirani, et al
  LIPPINCOTT WILLIAMS & WILKINS.2026: A18057

- **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
- **Integrating sequence-to-omics predictions and personal multi-omics for rare variant prioritization.** *medRxiv : the preprint server for health sciences*
  Lake, J., Robb, E., Rosa, V., Arriaga, T. M., Barrick, R., Berger, S. I., Heavner, B. D., King, C. H., Ko, A., LoTempio, J., Nachun, D., Kundaje, A., Bonner, et al
  2026
- **Domain-associated phenotypic patterns with MACF1 variants.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Gogate, N., Jolly, A., Rosenfeld, J. A., Bahena-Carbajal, P., Carrasco, D., Bernstein, J. A., Bonner, D. E., Busa, T., Cristian, I., D'Souza, P., Friedman, J., Gorokhova, S., Haaf, et al
  2026: 102776
- **Genotype and arrhythmic risk in dilated cardiomyopathy.** *European heart journal*
  Padalia, K., Fornaro, A., Claggett, B., Jason, M., Moritz, S., Medo, K., Graw, S., Hespe, S., Ingles, J., Ho, C. Y., Helms, A. S., Shore, S., Sinagra, et al
  2026
- **Body Mass Index, Valsartan, and Myocardial Remodeling in Early-Stage Hypertrophic Cardiomyopathy: A Secondary Analysis of the Randomized VANISH Trial.** *Journal of the American Heart Association*
  Ostrominski, J. W., Claggett, B. L., Jerosch-Herold, M., Raja, A. A., Day, S. M., Russell, M. W., Zahka, K., Pereira, A. C., Colan, S. D., Murphy, A. M., Canter, C., Bach, R. G., Wheeler, et al
  2026: e050131
- **Accelerometry As An Independent Predictor For Cardiorespiratory Fitness In Sedentary Adults: A MoTrPac Study Analysis**
  Leslie, E., Montalvo, S., Chen, S., Fanning, J., Jakicic, J. M., Johannsen, N. M., Melanson, E. L., Jack Rejeski, W., Katz, D., Ashley, E., Wheeler, M. T.
  LIPPINCOTT WILLIAMS & WILKINS.2026
- **Mapping Sedentary Human Fitness And Clinical Features: Multi-omic Baseline Architecture And Acute Exercise Dynamics In The Molecular Transducers Of Physical Activity Consortium (MoTrPAC) Project**
  Montalvo, S., Rao, P., Vetr, N. G., Sutton, K., Leslie, E., Brochet, P., Jin, C. A., Many, G. M., Iyer, G., Ahn, C., Taylor, T., Amar, D., Brandt, et al
  LIPPINCOTT WILLIAMS & WILKINS.2026
- **Tandem repeat pathogenicity signature unlocks oculopharyngeal distal myopathy discoveries**
  Danzi, M., Xu, I., van de Vondel, L., Facchini, S., Curro, R., Ravenscroft, G., Wheeler, M., Cortese, A., Zuchner, S.
  PERGAMON-ELSEVIER SCIENCE LTD.2026
- **Atrial Fibrillation in Genotyped Dilated Cardiomyopathy: Epidemiology, Risk Factors, and Outcomes: Insights From the SHaRe Registry.** *Circulation. Heart failure*
  Balakrishnan, I. D., Heymans, A. B., Claggett, B., Stewart, G. C., Seidman, C. E., Stroeks, S. L., Beelen, N. J., Venner, M. F., Owens, A. T., Day, S. M., Helms, A. S., Shore, S., Wheeler, et al
  2026: e014322
- **Arriving at a diagnosis: Effective strategies used by the Undiagnosed Diseases Network.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Hwang, S., Brown, R. M., Baldridge, D., Baldwin, E. E., Beggs, A. H., Bernstein, J. A., Blue, E., Borja, N. A., Botto, L. D., Briere, L. C., Cassini, T., Esteves, C., Fieg, et al
  2026: 102718
- **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

- **The effect of sex and endurance exercise training on the incretin signaling pathway in 17 rat tissues.** *Physiological genomics*
  Jin, C., Many, G., Reynoso Spurrier, C. S., Borsellino, P., Wheeler, M. T., Adkins, J. N., Snyder, M. P., Newgard, C. B., Radom-Aizik, S., Cooper, D. M., Bodine, S. C., Schenk, S., Lindholm, et al
  2026

- **BHLHE22 monoallelic and biallelic variants cause a neurodevelopmental disorder with agenesis of the corpus callosum, intellectual disability, abnormal muscle tone and movement abnormalities.** *Journal of medical genetics*
  Le, C., Kalayci, T., Uyguner, Z., Karaman, B., Demirören, T., Beck, B., Winnerling, N., George, E., DiTroia, S., Héron, D., Sabatier, I., Rodan, L. H., Girisha, et al
  2026

- **Temporal multi-omic analysis uncovers sex-biased molecular programs underlying skeletal muscle adaptation to endurance training.** *Cell reports*
  Many, G. M., Jin, C., Day, N. J., Iyer, G., Smith, G., Voos, K., Sanford, J. A., Bareja, A., Jimenez-Morales, D., Leach, D. T., Sagendorf, T. J., Ahmed, A., Li, et al
  2026; 45 (8): 117823

- **Scaled Multidimensional Assays of Variant Effect Identify Sequence-Function Relationships in Hypertrophic Cardiomyopathy.** *Circulation*
  Yamamoto, Y., Chua, K., Staudt, D., Ferrasse, A., Kirillova, A., De Jong, H. N., Floyd, B. J., Cadisch, C., Wiel, L., Wang, Q., O'Neill, M. J., Tabet, D., Goryznski, et al
  2026

- **Variant Site-Specific Natural History of Titin-Induced Cardiomyopathy: An International Multicenter Registry.** *Circulation. Genomic and precision medicine*
  Perotto, M., Radesich, C., Paldino, A., Gandin, I., Verdonschot, J. A., Kim, Y., Balakrishnan, I. D., Stroekes, S., Medo, K., Gigli, M., Lenarduzzi, S., Girotto, G., Claggett, et al
  2026: e005422

- **Characterization of induced pluripotent stem cell lines from patients of African American ancestry.** *Stem cell research*
  Wu, M. A., Wu, C. A., Zhao, S. R., Sun, J., Flores-Banuelos, A. G., Walther, P., Wheeler, M., Sayed, N.
  2026; 95: 104053

- **RNU4ATAC-opathy: Clinical, molecular and transcriptomic insights from a large cohort.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Matalon, D. R., Duker, A. L., Arriaga, T. M., Russell, K., Mendez, H. R., Bonner, D. E., Harley, M. E., Singer-Berk, M., Wojcik, M. H., Pais, L., DiTroia, S., O'Leary, M., Cassini, et al
  2026: 102633

- **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.
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