Stanford

James Priest

Adjunct Clinical Assistant Professor, Pediatrics - Cardiology

CLINICAL OFFICE (PRIMARY)

- **LPCH Heart Center**

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Bio

BIO

Working as a physician-scientist across industry and academia my scientific worldview is centered on the use of human genetics for target discovery & validation, indication selection in early clinical development, and maximizing meaningful signal in clinical trial design. I have expanded our genetic understanding of human disease and translated discoveries from the laboratory into clinically actionable diagnoses and novel therapeutic approaches yielding recognition in the international press and multiple patents. My broad expertise combines epidemiology, statistical genetics, machine learning, and molecular biology with clinical practice in pediatrics, cardiology, and genetic medicine. I have a track-record of building high-performing teams in challenging environments, solving difficult scientific problems, and contributing key insight throughout the lifecycle of commercial therapeutic development.

CLINICAL FOCUS

- Pediatric Cardiology
- hyperlipidemia
- Familial Hypercholesterolemia

ACADEMIC APPOINTMENTS

- Member, Cardiovascular Institute
- Member, Maternal & Child Health Research Institute (MCHRI)
- Member, Wu Tsai Neurosciences Institute

PROFESSIONAL EDUCATION

- Board Certification: Pediatric Cardiology, American Board of Pediatrics (2022)

- Medical Education: Stanford University School of Medicine (2008) CA
- Fellowship: Stanford University Pediatric Cardiology Fellowship (2016) CA
- Residency: University of Washington Pediatric Residency (2011) WA
- Board Certification: Pediatrics, American Board of Pediatrics (2011)
- MA, University of California Berkeley , Molecular Biology (2004)

LINKS

- Priest Lab github repo: https://github.com/priestlab/

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

Over the last thirty years, our fundamental understanding of the genetics and pathogenesis of congenital heart disease has lagged the tremendous advances in the surgical and clinical care of infants with this group of disorders. With my combined research training in genomics and clinical training in pediatric cardiology I endeavor to close this gap with investigation into the genetic basis of congenital heart malformations and developing new models of disease. My goal is translate an improved molecular genetic and developmental understanding of congenital heart disease from the laboratory into clinically actionable models, diagnostics, and ultimately therapeutic interventions.

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Cardiovascular Medicine (Fellowship Program)
- Pediatric Cardiology (Fellowship Program)

Publications

PUBLICATIONS

- **Mendelian Randomization Suggests a Causal Link Between Glycemic Traits and Thoracic Aortic Structures and Diseases.** *JACC. Basic to translational science*
  Daria, T., Iyer, K., Alkhairo, H., Kho, P. F., Suzuki, K., Hatzikotoulas, K., Southam, L., Taylor, H. J., Yin, X., Mandla, R., Huerta-Chagoya, A., Rayner, W. N., Levin, et al
  2025: 101390

- **Reduced Expression of MTSS1 Increases Sarcomere Number and Improves Contractility in Select Forms of Monogenic DCM.** *JACC. Basic to translational science*
  Kleppe, H., Budan, A., Zhang, L., Majetic, M., Shenwai, R., Levinson, A. R., Cisne-Thompson, O., Zepeda, B., MacKay, C. E., Farshidfar, F., Tsui, J. H., Figarska, S. M., Hoey, et al
  2025; 10 (11): 101391

- **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

- **Epistasis regulates genetic control of cardiac hypertrophy.** *Nature cardiovascular research*
  Wang, Q., Tang, T. M., Youlton, M., Weldy, C. S., Kenney, A. M., Ronen, O., Hughes, J. W., Chin, E. T., Sutton, S. C., Agarwal, A., Li, X., Behr, M., Kumbier, et al
  2025

- **Combining genetic proxies of drug targets and time-to-event analyses from longitudinal observational data to identify target patient populations.** *BMC cardiovascular disorders*

Zhang, L., Kulkarni, P., Farshidfar, F., Tingley, W., Hoey, T., Wang, W., Priest, J. R., Figarska, S. M.
2025; 25 (1): 353

- **The ERBB2 c.1795C>T, p.Arg599Cys variant is associated with left ventricular outflow tract obstruction defects in humans.** *HGG advances*
  Ampuja, M., Ericsson, S., Paatero, I., Chowdhury, I., Villman, J., Broberg, M., Ramste, A., Balboa, D., Ojala, T., Chong, J. X., Bamshad, M. J., Priest, J. R., Varjosalo, et al
  2025: 100446

- **Molecular convergence of risk variants for congenital heart defects leveraging a regulatory map of the human fetal heart.** *medRxiv : the preprint server for health sciences*
  Ma, X. R., Conley, S. D., Kosicki, M., Bredikhin, D., Cui, R., Tran, S., Sheth, M. U., Qiu, W. L., Chen, S., Kundu, S., Kang, H. Y., Amgalan, D., Munger, et al
  2024

- **Genome-wide association studies highlight novel risk loci for septal defects and left-sided congenital heart defects.** *BMC genomics*
  Broberg, M., Ampuja, M., Jones, S., Ojala, T., Rahkonen, O., Kivelä, R., Priest, J., Palotie, A., Ollila, H. M., Helle, E.
  2024; 25 (1): 256

- **Genomic and transcriptomic data analyses highlight KPNB1 and MYL4 as novel risk genes for congenital heart disease**
  Broberg, M., Ampuja, M., Jones, S., Ojala, T., Kivela, R., Priest, J., Ollila, H., Helle, E.
  SPRINGERNATURE.2024: 775

- **Learning epistatic polygenic phenotypes with Boolean interactions.** *PloS one*
  Behr, M., Kumbier, K., Cordova-Palomera, A., Aguirre, M., Ronen, O., Ye, C., Ashley, E., Butte, A. J., Arnaout, R., Brown, B., Priest, J., Yu, B.
  2024; 19 (4): e0298906

- **Epistasis regulates genetic control of cardiac hypertrophy.** *Research square*
  Wang, Q., Tang, T. M., Youlton, N., Weldy, C. S., Kenney, A. M., Ronen, O., Hughes, J. W., Chin, E. T., Sutton, S. C., Agarwal, A., Li, X., Behr, M., Kumbier, et al
  2023

- **Epistasis regulates genetic control of cardiac hypertrophy.** *medRxiv : the preprint server for health sciences*
  Wang, Q., Tang, T. M., Youlton, N., Weldy, C. S., Kenney, A. M., Ronen, O., Hughes, J. W., Chin, E. T., Sutton, S. C., Agarwal, A., Li, X., Behr, M., Kumbier, et al
  2023

- **Rare variants in CAPN2 increase risk for isolated hypoplastic left heart syndrome.** *HGG advances*
  Blue, E. E., White, J. J., Dush, M. K., Gordon, W. W., Wyatt, B. H., White, P., Marvin, C. T., Helle, E., Ojala, T., Priest, J. R., Jenkins, M. M., Almli, L. M., Reefhuis, et al
  2023; 4 (4): 100232

- **A second update on mapping the human genetic architecture of COVID-19** *NATURE*
  Kanai, M., Andrews, S. J., Cordioli, M., Stevens, C., Neale, B. M., Daly, M., Ganna, A., Kanai, M., Andrews, S. J., Cordioli, M., Pathak, G. A., Ganna, A., Iwasaki, et al
  2023; 621 (7977): E7-+

- **Oligogenic Architecture of Rare Noncoding Variants Distinguishes 4 Congenital Heart Disease Phenotypes.** *Circulation. Genomic and precision medicine*
  Yu, M., Aguirre, M., Jia, M., Gjoni, K., Cordova-Palomera, A., Munger, C., Amgalan, D., Rosa Ma, X., Pereira, A., Tcheandjieu, C., Seidman, C., Seidman, J., Tristani-Firouzi, et al
  2023: e003968

- **Genetic Determinants of the Interventricular Septum Are Linked to Ventricular Septal Defects and Hypertrophic Cardiomyopathy.** *Circulation. Genomic and precision medicine*
  Yu, M., Harper, A. R., Aguirre, M., Pittman, M., Tcheandjieu, C., Amgalan, D., Grace, C., Goel, A., Farrall, M., Xiao, K., Engreitz, J., Pollard, K. S., Watkins, et al
  2023: e003708

- **Relationship Between Ascending Thoracic Aortic Diameter and Blood Pressure: A Mendelian Randomization Study.** *Arteriosclerosis, thrombosis, and vascular biology*
  DePaolo, J., Levin, M. G., Tcheandjieu, C., Priest, J., Gill, D., Burgess, S., Damrauer, S. M., Chirinos, J. A.

2023

- **Machine Learning for Automated Mitral Regurgitation Detection from Cardiac Imaging**
  Xiao, K., Learned-Miller, E., Kalogerakis, E., Priest, J., Fiterau, M.
  edited by Greenspan, H., Madabhushi, A., Mousavi, P., Salcudean, S., Duncan, J., Syeda-Mahmood, T., Taylor, R.
  SPRINGER INTERNATIONAL PUBLISHING AG.2023: 236-246

- **Maternal first trimester metabolic profile in pregnancies with transposition of the great arteries.** *Birth defects research*
  Huida, J., Ojala, T., Ilvesvuo, J., Surcel, H., Priest, J. R., Helle, E.
  2022

- **A first update on mapping the human genetic architecture of COVID-19** *NATURE*
  Pathak, G. A., Polimanti, R., Karjalainen, J., Daly, M., Ganna, A., Daly, M. J., Stevens, C., Kanai, M., Liao, R. G., Trankiem, A., Balaconis, M. K., Nguyen, H., Solomonson, et al
  2022: E1-E10

- **Leveraging Machine Learning for Translational Genetics of Cardiovascular Imaging.** *Journal of the American College of Cardiology*
  Priest, J. R.
  2022; 80 (5): 498-499

- **Maternal and perinatal obesity induce bronchial obstruction and pulmonary hypertension via IL-6-FoxO1-axis in later life.** *Nature communications*
  Selle, J., Dinger, K., Jentgen, V., Zanetti, D., Will, J., Georgomanolis, T., Vohlen, C., Wilke, R., Kojonazarov, B., Klymenko, O., Mohr, J., V Koningsbruggen-Rietschel, S., Rhodes, et al
  2022; 13 (1): 4352

- **High heritability of ascending aortic diameter and trans-ancestry prediction of thoracic aortic disease.** *Nature genetics*
  Tcheandjieu, C., Xiao, K., Tejeda, H., Lynch, J. A., Ruotsalainen, S., Bellomo, T., Palnati, M., Judy, R., Klarin, D., Kember, R. L., Verma, S., Palotie, A., Daly, et al
  2022

- **Computational estimates of annular diameter reveal genetic determinants of mitral valve function and disease.** *JCI insight*
  Yu, M., Tcheandjieu, C., Georges, A., Xiao, K., Tejeda, H., Dina, C., Le Tourneau, T., Fiterau, M., Judy, R., Tsao, N. L., Amgalan, D., Munger, C. J., Engreitz, et al
  2022; 7 (3)

- **Comprehensive Genetic Testing for Pediatric Hypertrophic Cardiomyopathy Reveals Clinical Management Opportunities and Syndromic Conditions.** *Pediatric cardiology*
  Gal, D. B., Morales, A., Rojahn, S., Callis, T., Garcia, J., Priest, J. R., Truty, R., Vatta, M., Nussbaum, R. L., Esplin, E. D., Hollander, S. A.
  2021

- **Disruption of Protein Quality Control of Human Ether-a-go-go Related Gene K+ Channel Results in Profound Long QT Syndrome.** *Heart rhythm*
  Ledford, H. A., Ren, L., Thai, P. N., Park, S., Timofeyev, V., Sirish, P., Xu, W., Emigh, A. M., Priest, J. R., Perez, M. V., Ashley, E. A., Yarov-Yarovoy, V., Yamoah, et al
  2021

- **Single-cell transcriptomic landscape of cardiac neural crest cell derivatives during development.** *EMBO reports*
  Chen, W., Liu, X., Li, W., Shen, H., Zeng, Z., Yin, K., Priest, J. R., Zhou, Z.
  2021: e52389

- **Mapping the human genetic architecture of COVID-19.** *Nature*
  COVID-19 Host Genetics Initiative
  2021

- **Congenital heart disease risk loci identified by genome-wide association study in European patients.** *The Journal of clinical investigation*
  Lahm, H., Jia, M., DreSSen, M., Wirth, F. F., Puluca, N., Gilsbach, R., Keavney, B., Cleuziou, J., Beck, N., Bondareva, O., Dzilic, E., Burri, M., Konig, et al
  2020

- **Inherited Extremes of Aortic Diameter Confer Risk for a Specific Class of Congenital Heart Disease.** *Circulation. Genomic and precision medicine*

Tcheandjieu, C., Zanetti, D., Yu, M., Priest, J. R.
2020

- **Exome-Based Case-Control Analysis Highlights the Pathogenic Role of Ciliary Genes in Transposition of the Great Arteries.** *Circulation research*
  Liu, X. n., Chen, W. n., Li, W. n., Priest, J. R., Fu, Y. n., Pang, K. J., Ma, B. n., Han, B. n., Liu, X. n., Hu, S. n., Zhou, Z. n.
  2020

- **A phenome-wide association study of 26 mendelian genes reveals phenotypic expressivity of common and rare variants within the general population.** *PLoS genetics*
  Tcheandjieu, C. n., Aguirre, M. n., Gustafsson, S. n., Saha, P. n., Potiny, P. n., Haendel, M. n., Ingelsson, E. n., Rivas, M. A., Priest, J. R.
  2020; 16 (11): e1008802

- **Cardiac Imaging of Aortic Valve Area from 34,287 UK Biobank Participants Reveal Novel Genetic Associations and Shared Genetic Comorbidity with Multiple Disease Phenotypes.** *Circulation. Genomic and precision medicine*
  Córdova-Palomera, A. n., Tcheandjieu, C. n., Fries, J. n., Varma, P. n., Chen, V. S., Fiterau, M. n., Xiao, K. n., Tejeda, H. n., Keavney, B. n., Cordell, H. J., Tanigawa, Y. n., Venkataraman, G. n., Rivas, et al
  2020

- **Ivy: Instrumental Variable Synthesis for Causal Inference**
  Kuangy, Z., Sala, F., Sohoni, N., Wu, S., Cordova-Palomera, A., Dunnmon, J., Priest, J., Re, C.
  edited by Chiappa, S., Calandra, R.
  ADDISON-WESLEY PUBL CO.2020: 398–409

- **Adults With Mild-to-Moderate Congenital Heart Disease Demonstrate Measurable Neurocognitive Deficits.** *Journal of the American Heart Association*
  Perrotta, M. L., Saha, P. n., Zawadzki, R. n., Beidelman, M. n., Ingelsson, E. n., Lui, G. K., Priest, J. R.
  2020: e015379

- **Clonally expanding smooth muscle cells promote atherosclerosis by escaping efferocytosis and activating the complement cascade.** *Proceedings of the National Academy of Sciences of the United States of America*
  Wang, Y. n., Nanda, V. n., Direnzo, D. n., Ye, J. n., Xiao, S. n., Kojima, Y. n., Howe, K. L., Jarr, K. U., Flores, A. M., Tsantilas, P. n., Tsao, N. n., Rao, A. n., Newman, et al
  2020

- **Association of congenital cardiovascular malformation and neuropsychiatric phenotypes with 15q11.2 (BP1-BP2) deletion in the UK Biobank.** *European journal of human genetics : EJHG*
  Williams, S. G., Nakev, A. n., Guo, H. n., Frain, S. n., Tenin, G. n., Liakhovitskaia, A. n., Saha, P. n., Priest, J. R., Hentges, K. E., Keavney, B. D.
  2020

- **Maternal Obesity and Diabetes Mellitus as Risk Factors for Congenital Heart Disease in the Offspring.** *Journal of the American Heart Association*
  Helle, E. n., Priest, J. R.
  2020: e011541

- **Association between the 4p16 genomic locus and different types of congenital heart disease: results from adult survivors in the UK Biobank.** *Scientific reports*
  Cordova-Palomera, A., Priest, J. R.
  2019; 9 (1): 16515

- **Phenome-wide Burden of Copy-Number Variation in the UK Biobank.** *American journal of human genetics*
  Aguirre, M., Rivas, M. A., Priest, J.
  2019

- **Risk factors associated with the development of double-inlet ventricle congenital heart disease** *BIRTH DEFECTS RESEARCH*
  Paige, S. L., Yang, W., Priest, J. R., Botto, L. D., Shaw, G. M., Collins, R., Natl Birth Defects Prevention
  2019; 111 (11): 640–48

- **Substantial Cardiovascular Morbidity in Adults With Lower-Complexity Congenital Heart Disease** *CIRCULATION*
  Saha, P., Potiny, P., Rigdon, J., Morello, M., Tcheandjieu, C., Romfh, A., Fernandes, S. M., McElhinney, D. B., Bernstein, D., Lui, G. K., Shaw, G. M., Ingelsson, E., Priest, et al

2019; 139 (16): 1889–99

- **Risk factors associated with the development of double-inlet ventricle congenital heart disease.** *Birth defects research*
  Paige, S. L., Yang, W., Priest, J. R., Botto, L. D., Shaw, G. M., Collins, R. T., National Birth Defects Prevention Study
  2019
- **NEUROCOGNITIVE DEFICITS IN ADULT CONGENITAL HEART DISEASE: DOES CORONARY ARTERY DISEASE ADD INSULT TO INJURY?**
  Morello, M. L., Beidelman, M., Saha, P., Ingelsson, E., Shaw, G., Lui, G., Priest, J.
  ELSEVIER SCIENCE INC.2019: 566
- **Loss of function, missense, and intronic variants in NOTCH1 confer different risks for left ventricular outflow tract obstructive heart defects in two European cohorts** *GENETIC EPIDEMIOLOGY*
  Helle, E., Cordova-Palomera, A., Ojala, T., Saha, P., Potiny, P., Gustafsson, S., Ingelsson, E., Bamshad, M., Nickerson, D., Chong, J. X., Ashley, E., Priest, J. R., Univ Washington Ctr Mendelia
  2019; 43 (2): 215–26
- **Substantial Cardiovascular Morbidity in Adults with Lower-Complexity Congenital Heart Disease.** *Circulation*
  Saha, P., Potiny, P., Rigdon, J., Morello, M., Tcheandjieu, C., Romfh, A., Fernandes, S. M., McElhinney, D. B., Bernstein, D., Lui, G. K., Shaw, G. M., Ingelsson, E., Priest, et al
  2019
- **Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources** *NUCLEIC ACIDS RESEARCH*
  Koehler, S., Carmody, L., Vasilevsky, N., Jacobsen, J. O. B., Danis, D., Gourdine, J., Gargano, M., Harris, N. L., Matentzoglu, N., McMurry, J. A., Osumi-Sutherland, D., Cipriani, V., Balhoff, et al
  2019; 47 (D1): D1018–D1027
- **IMPACT OF CARDIAC ALGORITHM ON CYTOGENETIC TESTING**
  Floyd, B. J., Hintz, S. R., Suarez, C. J., Cherry, A., Yu, L., Benitz, W., Priest, J. R., Wright, G. E., Bhombal, S., Davis, A., Chock, V. Y., Weigel, N., Kobayashi, et al
  BMJ PUBLISHING GROUP.2019: 207
- **Single-Cell RNA-Seq of the Developing Cardiac Outflow Tract Reveals Convergent Development of the Vascular Smooth Muscle Cells.** *Cell reports*
  Liu, X. n., Chen, W. n., Li, W. n., Li, Y. n., Priest, J. R., Zhou, B. n., Wang, J. n., Zhou, Z. n.
  2019; 28 (5): 1346–61.e4
- **Weakly supervised classification of rare aortic valve malformations using unlabeled cardiac MRI sequences** *Nature Communications*
  Fries, J. A., Varma, P., Chen, V. S., Xiao, K., Tejeda, H., Saha, P., Dunnmon, J., Chubb, H., Maskatia, S., Fiterau, M., Delp, S., Ashley, E., Ré, et al
  2019; 10
- **Loss of function, missense, and intronic variants in NOTCH1 confer different risks for left ventricular outflow tract obstructive heart defects in two European cohorts.** *Genetic epidemiology*
  Helle, E., Cordova-Palomera, A., Ojala, T., Saha, P., Potiny, P., Gustafsson, S., Ingelsson, E., Bamshad, M., Nickerson, D., Chong, J. X., University of Washington Center for Mendelian Genomics, Ashley, E., Priest, J. R.
  2018
- **Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources.** *Nucleic acids research*
  Kohler, S., Carmody, L., Vasilevsky, N., Jacobsen, J. O., Danis, D., Gourdine, J., Gargano, M., Harris, N. L., Matentzoglu, N., McMurry, J. A., Osumi-Sutherland, D., Cipriani, V., Balhoff, et al
  2018
- **CONGENITAL HEART DISEASE CONFERS SUBSTANTIAL RISK OF ACQUIRED CARDIOVASCULAR DISEASE AMONGST BRITISH ADULTS**
  Saha, P., Potiny, P., Tcheandjieu, C., Fernandes, S. M., Romfh, A., Bernstein, D., Lui, G. K., Ingelsson, E., Priest, J.
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- **Ring Finger Protein 207 Degrades T613M Kv11.1 Channel**
  Ledford, H. A., Park, S., Sirish, P., Xu, W., Emigh, A. M., Timofeyev, V., Priest, J. R., Perez, M. V., Ashley, E. A., Yarov-Yarovoy, V., Zhang, X., Chiamvimonvat, N.
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  Zanetti, D. n., Tikkanen, E. n., Gustafsson, S. n., Priest, J. R., Burgess, S. n., Ingelsson, E. n.
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- **A primer to clinical genome sequencing.** *Current opinion in pediatrics*
  Priest, J. R.
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  Li, G., Xu, A., Sim, S., Priest, J. R., Tian, X., Khan, T., Quertermous, T., Zhou, B., Tsao, P. S., Quake, S. R., Wu, S. M.
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  Priest, J. R., Gawad, C., Kahlig, K. M., Yu, J. K., O'Hara, T., Boyle, P. M., Rajamani, S., Clark, M. J., Garcia, S. T., Ceresnak, S., Harris, J., Boyle, S., Dewey, et al
  2016; 113 (41): 11555-11560
- **Standards of Evidence and Mechanistic Inference in Autosomal Recessive Hypercholesterolemia** *ARTERIOSCLEROSIS THROMBOSIS AND VASCULAR BIOLOGY*
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- **Prepregnancy Diabetes and Offspring Risk of Congenital Heart Disease A Nationwide Cohort Study** *CIRCULATION*
  Oyen, N., Diaz, L. J., Leirgul, E., Boyd, H. A., Priest, J., Mathiesen, E. R., Quertermous, T., Wohlfahrt, J., Melbye, M.
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  2016; 8 (1): 24-?
- **Maternal Midpregnancy Glucose Levels and Risk of Congenital Heart Disease in Offspring** *JAMA PEDIATRICS*
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  2015; 169 (12): 1112-1116
- **Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome** *HUMAN MUTATION*
  Cordeddu, V., Yin, J. C., Gunnarsson, C., Virtanen, C., Drunat, S., Lepri, F., De Luca, A., Rossi, C., Ciolfi, A., Pugh, T. J., Bruselles, A., Priest, J. R., Pennacchio, et al
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- **Sequence to Medical Phenotypes: A Framework for Interpretation of Human Whole Genome DNA Sequence Data.** *PLoS genetics*
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- **Molecular diagnosis of long QT syndrome at 10 days of life by rapid whole genome sequencing.** *Heart rhythm*
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