Dr. Sarah A. Pendergrass

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Education:

Ph.D. in Genetics, Dartmouth College, Hanover, NH	2009
M.S. in Engineering, Thayer School of Engineering, Hanover, NH	2004
B.A. in Physics, Smith College, Northampton, MA	2001

Research Experience:

Assistant Professor **Biomedical and Translational Informatics, Geisinger Health System** **2015-present**

Research Interests: My research focus is to develop and deploy new methodologies and strategies for the analysis of complex multi-dimensional datasets including genetic data, for biological discovery and ultimately for improvements in clinical care. I am a genetic Epidemiologist, bioinformatician, and data scientist. I am a nationally recognized expert in Phenome-Wide Association Studies (PheWAS), an approach that I have contributed to and have been developing and advancing starting in 2009. I have been expanding the use of electronic health record (EHR) data for research, including the use of medication and drug dosage information, quantitative lab measures, longitudinal data, and the use of imaging data. These strategies are moving the research field beyond genetic associations focused only on disease diagnoses, and incorporate more of the complexity and heterogeneity present in disease. I am also pursuing research in pharmacogenomics, one of the most actionable areas for bringing translational and precision medicine to the clinic. My projects have included data from the Geisinger Biorepository the *MyCode Community Health Initiative*, the Electronic Medical Records and Genomics (eMERGE) network and the Aids Clinical Trials Group (ACTG).

Research Faculty – Staff Scientist, Center for System Genomics, 2011-2014
Department of Biochemistry and Molecular Biology, Pennsylvania State University,
State College PA
Lab of Dr. Marylyn Ritchie
Highlighted Accomplishments: Participated in the first PheWAS using clinical trials data and wrote idea papers on future analyses for PheWAS. Participated in the first pharmacogenomic focused PhewAS. Pursued pharmacogenomic projects with collaborators from the Pharmacogenomic Resource Network (PGRN) subgroup the PGRN statistical analysis resource (P-STAR). Collaborated on the development of Biobin, a software tool for gene-based association testing where rare genetic data can be binned by genes as well as by pathways. Led the new and expanded build of the software Biofilter for annotating and filtering data via multiple publicly available sources of genetic annotation. Led the development of the data visualization software PhenoGram for viewing genetic association results across chromosomal ideograms and PheWAS-view for integrating multiple phewas results into a single plot. Data visualization from these software have been used in many publications and presentations by other researchers. Led a workshop and a session at the Pacific Symposium on Biocomputing.

Postdoctoral Research Fellow, Center for Human Genetics Research, Vanderbilt University, Nashville TN 2009-2011

Advisors: Dr. Marylyn Ritchie, Dr. Dana Crawford

Highlighted Accomplishments: Performed the first two published PheWAS using epidemiological study data, instead of EHR based data, using data from the Population Architecture Using Genomics and Epidemiology (PAGE) network, including data from the National Health and Nutrition Examination Survey (NHANES). I also developed a novel data visualization software "Synthesis View" for plotting multiple tracks of genetic association summary data.

Ph.D. research, Dartmouth College, Hanover, NH

Advisors: Dr. Michael Whitfield, Dr. Jason Moore

Thesis: "Gene expression subsets and biomarkers in the genome-wide expression profiles of systemic sclerosis"

Projects: Systemic sclerosis (SSc) is a rare and debilitating autoimmune disease, and patients present with highly heterogeneous symptoms and severity. I pursued gene expression analyses using skin biopsies and peripheral blood samples from individuals with SSc. The focus was to use these gene expression signatures to identify subsets of patients with the intention of eventually identifying the risk of different outcomes for SSc patients, and biomarkers for better and more specific treatments for subsets of patients with SSc. I confirmed gene expression signatures for subsets of SSc patients. I also identified different subsets of patients with limited SSc.

M.S. research, Thayer School of Engineering, Dartmouth College, Hanover, NH

Advisors: Dr. Paul Meany, Dr. Marvin Doyley

Thesis: "Microwave imaging device design and improvement related to dielectric property measurement and experimentation"

Projects: Participated in the development and testing of intravascular ultrasound modifications to detect thrombotic events beyond calcification levels. Participated in the development and testing of alternative imaging modalities for breast cancer detection.

Awards:

Healthcare Innovation and Transformation Initiative Grant 2016

The Pennsylvania State University CTSI KL2 Award 2015

Genome Technology 8th Annual Young Investigator, December 13, 2013

Young Investigator Award, 10th International Workshop on Scleroderma, 2008

Keystone Symposia Scholarship Recipient, 2008

NIAMS Autoimmunity and Connective Tissue Training Grant 2006 – 2008

Teaching/Mentoring Experience

Research Assistant Mentored

Elliot Cha, now a medical student at the Geisinger Commonwealth Medical College
Returned for a summer internship 2018

Postdoctoral Fellows Mentored

Dr. Chris Bauer, currently postdoctoral fellow in my lab
Dr. Yanfei Zhang, currently postdoctoral fellow in the lab of Mike (Ming Ta) Lee

Graduate Students Mentored

Brett Beaulieu-Jones – University of Pennsylvania, graduating December 2017, lab of Dr. Jason Moore

Anurag Verma – The Pennsylvania State University, graduated December 2017, lab or Dr. Marylyn Ritchie

Courses Taught

"Strategies for Enhancing the Value and Utility of Electronic Health Record Research", Case Western Reserve University, Institute for Computational Biology Symposium, September 28, 2017

"Precision Medicine: Prediction and Prevention", The Pennsylvania State University, April 5, 2017

"Precision Health Care at Geisinger", Nursing Research and Evidence Based Practice Conference, Danville PA, November 4, 2016

"From Learning Health Care to Genetic Association Research: Precision Medicine in Action" The Pennsylvania State University, October 10, 2016

"Precision Medicine, GWAS, Rare Variant Analyses, and Copy Number Variant analyses" Dr. Les Kirchner's Geisinger Course, June 29, 2016

"Data Visualization" Case Western Reserve, February 19, 2016

"Personalized Medicine: When Genomics Gets Personal" The Pennsylvania State University, March 16, 2015

"Workshop Introduction Putting the Pieces Together: Precision Medicine Discovery from Electronic Health Records" Case Western Reserve Symposium, September 22, 2015

Taught bioinformatics courses at the Centers for Disease Control and Prevention (CDC), February 23, 2015

Taught bioinformatics workshop at the Big Data Analysis and Translation in Disease Biology
(Big Data and Disease) JNU, New Delhi, January 18- 22, 2015

Taught bioinformatics courses at the Centers for Disease Control and Prevention (CDC), September 30-October 3, 2014

"Detecting And Characterizing Pleiotropy: New Methods For Uncovering The Connection Between The Complexity Of Genomic Architecture And Multiple Phenotypes" Pacific Symposium on Biocomputing, January 4, 2014

"Next Generation Sequencing: Applications for Infectious Disease" Centers for Disease Control and Prevention (CDC), February 28, 2012

Taught data visualization module for the "Comparison of Analytical Methods for Genetic Association Studies" course at the Centers for Disease Control and Prevention (CDC), February 25, 2011

Participant in Dartmouth Center for Learning seminars for teacher training, mentoring, and professional development 2006 - 2008

Teaching Assistant, Course: Molecular and Computational Genomics, 2006

Teaching Assistant, Course: Introduction to Genetics, 2006

Skills:

Bioinformatics, Biostatistics, Data Visualization Development, Genomic Analysis, Programming, Numerical Methods

Memberships:

American Society of Human Genetics

American Medical Informatics Association

Service:

Grant reviews Genome Canada 2017-2018

Mentorship Committee Member for Dr. Kevin Ho 2017-2018

BD2K Training Program: Committee for Evaluating and Choosing Students for BD2K Program 2016-2017

Director Data Science Core from May 2016 to January, 2017

Special Emphasis Panel Reviewer: Udall Centers of Excellence in Parkinson's Disease, National Institute of Neurological Disorders and Stroke 2016-2017

PLoS Genetics guest editor 2016-2017

Recruitment of 3 staff scientists (Matthew Oetjens, Nicole Restrepo, Mariusz Butkiewicz)

Recruitment of Geisinger Commonwealth Medical College gap year students (Elliot Cha)

Contacted Smith College and Mt. Holyoke College, and Spellman College to recruit undergraduate interns for the PA Cure grant – one intern accepted the summer of 2016

Event Organization:

Workshop on extracting lifestyle/behavior and exposures from electronic health records, Institute for Computational Biology Symposium, Case Western Reserve University, "Being Precise in Precision Medicine: Measuring Exposures in Diverse Populations", September 28, 2017

Pacific Symposium on Biocomputing Session "Patterns in Biomedical Data – How Do We Find Them?" January 2017

American Society of Human Genetics, Educational Session, Translating Genomic Knowledge into Clinical Practice: "Using electronic health record data and biorepositories, from experimental discovery to clinical decision support: progress and promise", October 10, 2015

Institute for Computational Biology Symposium, Case Western Reserve University, Workshop: "Putting the Pieces Together: Precision Medicine Discovery from Electronic Health Records" and Subsequent Talk: "PheWAS: Embracing Complexity for Discovery", September 22, 2015

Pacific Symposium on Biocomputing session "Characterizing the Importance of Environmental Exposures, Interactions between the Environment and Genetic Architecture, and Genetic Interactions: New Methods for Understanding the Etiology of Complex Traits and Disease", January, 2015

Organized and led a three hour Data Visualization Workshop for the Bioinformatics and Genomics Retreat for the Huck Institutes of the Life Sciences at the Pennsylvania State University, September 13, 2014

Bioinformatic Track Program Committee, ACM Conference on Bioinformatics, Computational Biology and Health, September 20-23, 2014

Co-led a peer-reviewed paper session at the Pacific Symposium on Biocomputing, titled "Detecting And Characterizing Pleiotropy: New Methods For Uncovering The Connection Between The Complexity Of Genomic Architecture And Multiple Phenotypes", January 6, 2014

Co-led a workshop at the Pacific Symposium on Biocomputing, titled "Uncovering the Etiology of Autism Spectrum Disorders: Genomics, Bioinformatics, Environment, Data Collection and Exploration, and Future Possibilities", January 4, 2014

Invited Presentations:

"Redefining our Understanding of Disease, Outcome, and the Impact of Genetic Architecture through Electronic Health Records" – John's Hopkins University, February 4, 2019

"Redefining our Understanding of Disease, Outcome, and the Impact of Genetic Architecture through Electronic Health Records" – University of Pennsylvania, January 23, 2019

"Redefining our Understanding of Disease, Outcome, and the Impact of Genetic Architecture through Electronic Health Records" – Washington University School of Medicine in St. Louis, October 9, 2018

"Using Electronic Health Records to Redefine our Understanding of Disease, Outcome, Phenotype, and the Impact of Genetic Architecture" – University of Colorado, June 20, 2018

"From Learning Health Care to Genetic Research: Precision Medicine In Action" – Dartmouth College, April 25, 2018

"Redefining our Understanding of Disease, Outcome, Phenotype and the Impact of Genetic Architecture through Electronic Health Records", Innovation Center for Biomedical Informatics (ICBI), Georgetown University, February 15, 2018

"From Learning Health Care to Genetic Research: Precision Medicine In Action at Geisinger Health System" Data Science Week, National Center for Biotechnology Information (NCBI), September 13, 2017

"Precision Health Care at Geisinger: Prediction and Prevention" Geisel School of Medicine at Dartmouth, Big Data in the Life Sciences Symposium, May 24, 2017

"Genomic Research in Action: Phenome-Wide Association Studies in Diverse Populations" Society for Advancement of Chicanos/Hispanics and Native Americans in Science (SACNAS) Conference, October 14, 2016

"Your Health Risks: Prediction to Prevention" Penn State Lectures on the Frontiers of Science, Pennsylvania State University, January 23, 2016

"Contrasting Association Results between Existing PheWAS Phenotype Definition Methods and Five Validated Electronic Phenotypes" American Medical Informatics Association, November 15, 2015

eMERGE Phenome-Wide Association Study (PheWAS) Identifies Clinical Associations and Pleiotropy for Stop-Gain Variants" Translational Bioinformatics Conference Tokyo Japan, November 8, 2015

"EHRs and genomic discovery: phenome-wide association studies and other adventures in pleiotropy" Society for Advancement of Chicanos/Hispanics and Native Americans in Science (SACNAS) Conference, October 30, 2015

"Electronic Health Records and Genomics: Embracing Complexity for Discovery" EMBL-EBI Industry Meeting, Waltham MA, October 29, 2015

"Mining Electronic Health Records for Discovery" The Pennsylvania State University Huck Graduate Students Advisory Committee, September 24, 2015

"Workshop Introduction Putting the Pieces Together: Precision Medicine Discovery from Electronic Health Records" Case Western Reserve Symposium, September 22, 2015

"Mining Electronic Health Records for Discovery" Case Western Reserve Symposium, September 22, 2015

"Biofilter 2.0 for Advanced Predictive Model Development, Testing, and Hypothesis Generation using Expert Domain Knowledge Resources"
American Medical Informatics Association – Translational Bioinformatics, San Francisco, CA, June 7, 2014

"Visualizing Multiple Types of Genomic Information Across Chromosomes With PhenoGram"
American Medical Informatics Association – Translational Bioinformatics, San Francisco, CA, June 7, 2014

"Adding Value to Large Genomic Epidemiology Studies: Phenome Wide Association Studies for Exploring the Relationship Between the Phenome and Genome"
American Association for Cancer Research, June 5, 2014

"Phenome Wide Association Study (PheWAS) for Detection of Pleiotropy within the Population Architecture using Genomics and Epidemiology (PAGE) Network "
Translational Bioinformatics Conference, Seoul, Korea, October 4, 2013

"Elucidating the Genetic Architecture of Complex Disorders: Challenges and New Approaches"
Drexel University, Philadelphia PA, May 22, 2013
"Visually integrating and exploring high throughput results using PheWAS-view, PhenoGram, and Synthesis-View"
PGRN Statistical Analysis Resource, December 4, 2012

"Visually Integrating and Exploring High Throughput Phenome-Wide Association (PheWAS) Results Using PheWAS-view and PhenoGram"
Annual Meeting of the American Society of Human Genetics, San Francisco, November 8, 2012

"Identification of Pleiotropy with a Phenome-Wide Association Study (PheWAS) using two National Health and Nutrition Examination Surveys (NHANES)"
Translational Bioinformatics Conference, JeJu Island, Korea, October 15, 2012

"Challenges and Approaches for Elucidating the Genetic Architecture of Complex Disorders"
Environmental Epidemiology Network Meeting, International Meeting for Autism Research, Toronto Canada, May 17, 2012

"A Phenome-wide Exploration of Genotype-Phenotype Associations and Pleiotropy using Metabochip in the PAGE Study"
Keystone Symposium Complex Traits: Genomics and Computational Approaches, Breckenridge, Colorado, February 21, 2012

"A Phenome-wide Exploration of Novel Genotype-Phenotype Associations and Pleiotropy using Metabochip in the PAGE Network"
Annual Meeting of the International Genetic Epidemiology Society, Heidelberg, Germany, September 20, 2011

"Visual Integration of Results Using Synthesis-View"
Gene Environment Association Studies (GENEVA) Steering Committee Meeting, Washington D.C., January 18, 2011

"Visual Integration of Results from a Large DNA Biobank (BioVU) using Synthesis-View" Pacific Symposium on Biocomputing, Big Island, Hawaii, January 5, 2011

"Phenotype-Wide Association Study (PheWAS) for Detection of Pleiotropy within the Multi-Ethnic Cohorts of the Population Architecture Using Genomics and Epidemiology (PAGE) network"
Annual Meeting of the American Society of Human Genetics, Washington D.C., November 2, 2010
"Phenotype-Wide Association Study (PheWAS) for Exploration of Novel SNP and Phenotype Relationships within PAGE"
2010 Annual Meeting of the International Genetic Epidemiology Society, Boston, MA, October 10, 2010

Grants/Funding

Current:

(no assignment yet), NIH R01, award in process

AN ATLAS OF CLINICAL ASSOCIATIONS MAPPING TO VARIANTS WITHIN PROTEIN STRUCTURES
Role: PI
The goal of this project is develop to tools and software for projecting low-frequency genetic variation (captured by genomic sequencing) into protein structural space, and to perform grouped-variant association tests to establish an online atlas, mapping functional protein regions to the thousands of clinical traits they potentially influence.

1R01HG010067, NIH. University of Pennsylvania 4/1/2018-3/1/2023
NETWORK-BASED ALGORITHMS FOR TARGET IDENTIFICATION AND DRUG REPOSITIONING FROM GENETIC ASSOCIATIONS
Role: Co-investigator
We will use the PheWAS algorithmic infrastructure within DNANexus, modifying as necessary, for performing PheWAS using ICD-9 codes, clinical lab variables, and high quality phenotypes derived from algorithms, and then using these results with the algorithmic developments of Dr. Casey Greene as detailed in the grant.

1R01HD093671, NIH. Vanderbilt University. 11/1/2017-8/31/2020
LARGE-SCALE STUDIES IN EMERGE TO DISCOVER THE GENETIC DETERMINANTS OF UTERINE FIBROIDS
Role: PI subcontract
Identifying common and rare variants associated with fibroids through genome-wide association studies (GWAS), whole genome, and exome resequencing of eMERGE participants, which will allow discoveries in coding, regulatory, and structural variants across a wide range of allele frequencies.

R01DA044015, NIH. Janet Robishaw, Vanessa Troiani and WH Berrettini (PIs) 09/1/17-5/31/22
CLINICAL AND GENETIC STUDY OF PRESCRIPTION OPIOID ADDICTION
Role: Co-Investigator
These studies will establish a foundation of critical knowledge of the clinical, genetic, and brain-related factors that influence the risk of developing POA. Such results will drive the development of more accurate predictive models for assessing abuse risk in individuals treated with opioids for chronic pain.

1R01HL134015, NIH. University of Pennsylvania. 08/15/16-04/30/20
APPROACHES TO GENETIC HETEROGENEITY OF OBSTRUCTIVE SLEEP APNEA
Role: Co-Investigator
Identification of genetic biomarkers for the risk of sleep apnea, as well as the identification of new risk factors for sleep apnea. The identification of new patient subsets within patients with sleep apnea.

SAP #4100070267 Pennsylvania Department of Health. 06/01/15-05/31/19
INTEGRATING BIG DATA FOR BIOMEDICAL DISCOVERY: METHODS, TOOLS, AND APPLICATIONS.
Role: Co-Investigator
A multidisciplinary project to develop a series of advanced algorithms, methodologies, and software for integrating and analyzing multiple types of biomedical big data and to apply these innovative approaches for better understanding and treatment of obesity and obesity-related comorbidities.
Role: Co-Investigator

U01GH008679, NIH. Williams, Marc (PI) 09/01/15-05/31/19
EMR-LINKED BIOBANK FOR TRANSLATIONAL GENOMICS (EMERGE III)
Role: Co-Investigator
The goals of this study are to use existing biospecimens, genotype and sequence data and EMR generated phenotypes for discovery in the proposed disorders: familial hypercholesterolemia (FH) and chronic rhinosinusitis (CRS), develop and test approaches for implementation of genomic information in clinical practice and to explore, develop and implement novel approaches for family-centered communication around clinically relevant genomic results

Completed Research Support:

FDA 12/15/2017-09/28/2018
GENERIC DRUG MONITORING WITHIN LARGE HEALTH PROVIDERS
Role: Co-Investigator

The goal of this study is to detect adverse events with generic drugs within the medication data of Geisinger and compare/contrast with Marshfield Clinic

No assignment # GHS GT Collaboration. Pendergrass, Sarah (PI) 9/1/2016-6/30/2017
IDENTIFICATION OF GENES AND DISEASES ASSOCIATED WITH DEVELOPMENTAL ROBUSTNESS
We will take advantage of a unique combination of datasets to identify markers of developmental robustness and identify the genetic causes and clinical consequences of phenotypic heterogeneity.

Publications Under Review/In Preparation

A Catalogue of Molecular Targets for Kidney Function from Genetic Analyses of a Million Individuals. CKDGEN Consortium including Dr. Pendergrass and analyst Navya Josyula. *Accepted Nature Genetics*

Phenotypic and Pharmacogenomics Identification of Intraoperative Phenylephrine Responses. Yanfei Zhang, Mark S. Poler, Vida Abedi, Sarah A. Pendergrass, Ming Ta M. Lee, *BMC Medicine*

Electronic Health Records Elucidate Relationships Between Genetics, the Anatomy of the Eye, and Ocular Diseases. Christopher R. Bauer, Nicole Restrepo, Elliott D.K. Cha, Annalise B. Paaby, Dustin Hartzel, Sarah A Pendergrass *Submitted for Review American Journal of Human Genetics*

Reviewed Publications

1. Zhang X, Basile AO, Pendergrass SA, Ritchie MD. Real world scenarios in rare variant association analysis: the impact of imbalance and sample size on the power in silico. BMC Bioinformatics. 2019 Jan 22;20(1):46.

2. Pendergrass SA, Crist RC, Jones LK, Hoch JR, Berrettini WH. The importance of buprenorphine research in the opioid crisis. Mol Psychiatry. 2019 Jan 7; PMID: 30617273

3. Pendergrass SA, Crawford DC. Using Electronic Health Records To Generate Phenotypes For Research. Curr Protoc Hum Genet. 2019 Jan;100(1):e80. PMCID: PMC6318047

4. Verma A, Bang L, Miller JE, Zhang Y, Lee MTM, Zhang Y, Byrska-Bishop M, Carey DJ, Ritchie MD, Pendergrass SA, Kim D, DiscovEHR Collaboration. Human-Disease Phenotype Map Derived from PheWAS across 38,682 Individuals. Am J Hum Genet. 2019 Jan 3;104(1):55–64. PMID: 30598166

5. Hall T, Stanaway I, Carrell D, Carroll R, Denny JC, Hakonarson HH, Larson EB, Mentch F, Peissig PL, Pendergrass S, Rosenthal E, Jarvik GP, Crosslin DR. Unfolding of hidden white blood cell count phenotypes for gene discovery using latent class mixed modelin. Genes Immun.

6. Cha EDK, Veturi Y, Agarwal C, Patel A, Arbabshirani MR, Pendergrass SA. Using Adipose Measures from Health Care Provider-Based Imaging Data for Discovery. J Obes. 2018;2018:3253096. PMCID: PMC6180992

7. Stanaway IB, Hall TO, Rosenthal EA, Palmer M, Naranbhai V, Knevel R, Namjou-Khales B, Carroll RJ, Kiryluk K, Gordon AS, Linder J, Howell KM, Mapes BM, Lin FTJ, Joo YY, Hayes MG, Gharavi AG, Pendergrass SA, Ritchie MD, de Andrade M, Croteau-Chonka DC, Raychaudhuri S, Weiss ST, Lebo M, Amr SS, Carrell D, Larson EB, Chute CG, Rasmussen-Torvik LJ, Roy-Puckelwartz MJ, Sleiman P, Hakonarson H, Li R, Karlson EW, Peterson JF, Kullo IJ, Chisholm R, Denny JC, Jarvik GP, eMERGE Network, Crosslin DR. The eMERGE genotype set of 83,717 subjects imputed to ~40 million variants genome wide and association with the herpes zoster medical record phenotype. Genet Epidemiol. 2018 Oct 8; PMID: 30298529

8. Verma A, Bradford Y, Dudek S, Lucas AM, Verma SS, Pendergrass SA, Ritchie MD. A simulation study investigating power estimates in phenome-wide association studies. BMC Bioinformatics. 2018 Apr 4;19(1):120.

9. Jones L, Rahm A, Manickam K, Butry L, Lazzeri A, Cocoran T, Komar D, Josyula N, Pendergrass S, Sturm A, Murray M. Healthcare Utilization and Patients' Perspectives After Receiving a Positive Genetic Test for Familial Hypercholesterolemia. Circ Genomic Precis Med. 2018 Aug 13;11(8):e002146.

10. Ji X, Niu X, Qian J, Martucci V, Pendergrass SA, Gorlov IP, Amos CI, Denny JC, Massion PP, Aldrich MC. A Phenome-Wide Association Study Uncovers a Role for Autoimmunity in the Development of Chronic Obstructive Pulmonary Disease. Am J Respir Cell Mol Biol. 2018 Jun 1;58(6):777–779.

11. Verma A, Lucas A, Verma SS, Zhang Y, Josyula N, Khan A, Hartzel DN, Lavage DR, Leader J, Ritchie MD, Pendergrass SA. PheWAS and Beyond: The Landscape of Associations with Medical Diagnoses and Clinical Measures across 38,662 Individuals from Geisinger. Am J Hum Genet. 2018 Apr 5;102(4):592–608. PMCID: PMC5985339

12. Verma SS, Josyula N, Verma A, Zhang X, Veturi Y, Dewey FE, Hartzel DN, Lavage DR, Leader J, Ritchie MD, Pendergrass SA. Rare variants in drug target genes contributing to complex diseases, phenome-wide. Sci Rep. 2018 Mar 15;8(1):4624.

13. Helle EIT, Biegley P, Knowles JW, Leader JB, Pendergrass S, Yang W, Reaven GR, Shaw GM, Ritchie M, Priest JR. First Trimester Plasma Glucose Values in Women without Diabetes are Associated with Risk for Congenital Heart Disease in Offspring. J Pediatr. 195:275–278.

14. Hall MA, Wallace J, Lucas A, Kim D, Basile AO, Verma SS, McCarty CA, Brilliant MH, Peissig PL, Kitchner TE, Verma A, Pendergrass SA, Dudek SM, Moore JH, Ritchie MD. PLATO software provides analytic framework for investigating complexity beyond genome-wide association studies. Nat Commun. 2017 Oct 27;8(1):1167. PMCID: PMC5660079

15. Verma A, Bradford Y, Verma SS, Pendergrass SA, Daar ES, Venuto C, Morse GD, Ritchie MD, Haas DW. Multiphenotype association study of patients randomized to initiate antiretroviral regimens in AIDS Clinical Trials Group protocol A5202. Pharmacogenet Genomics. 2017 Mar;27(3):101–111. PMCID: PMC5285297

16. Beaulieu-Jones BK, Lavage DR, Snyder JW, Moore JH, Pendergrass SA, Bauer CR. Characterizing and Managing Missing Structured Data in Electronic Health Records: Data Analysis. JMIR Med Inform. 2018;6(1):e11.

17. Haggerty CM, James CA, Calkins H, Tichnell C, Leader JB, Hartzel DN, Nevius CD, Pendergrass SA, Person TN, Schwartz M, Ritchie MD, Carey DJ, Ledbetter DH, Williams MS, Dewey FE, Lopez A, Penn J, Overton JD, Reid JG, Lebo M, Mason-Suares H, Austin-Tse C, Rehm HL, Delisle BP, Makowski DJ, Mehra VC, Murray MF, Fornwalt BK. Electronic Health Record Phenotype in Subjects with Genetic Variants Associated with Arrhythmogenic Right Ventricular Cardiomyopathy: A Study in 30,716 Subjects with Exome Sequencing. Genet Med Off J Am Coll Med Genet. 2017 Nov;19(11):1245–1252. PMCID: PMC5671380

18. Dewey FE, Murray MF, Overton JD, Habegger L, Leader JB, Fetterolf SN, O'Dushlaine C, Hout CVV, Staples J, Gonzaga-Jauregui C, Metpally R, Pendergrass SA, Giovanni MA, Kirchner HL, Balasubramanian S, Abul-Husn NS, Hartzel DN, Lavage DR, Kost KA, Packer JS, Lopez AE, Penn J, Mukherjee S, Gosalia N, Kanagaraj M, Li AH, Mitnaul LJ, Adams LJ, Person TN, Praveen K, Marcketta A, Lebo MS, Austin-Tse CA, Mason-Suares HM, Bruse S, Mellis S, Phillips R, Stahl N, Murphy A, Economides A, Skelding KA, Still CD, Elmore JR, Borecki IB, Yancopoulos GD, Davis FD, Faucett WA,

Gottesman O, Ritchie MD, Shuldiner AR, Reid JG, Ledbetter DH, Baras A, Carey DJ. Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study. Science. 2016 Dec 23;354(6319):aaf6814. PMID: 28008009

19. Jones GT, Tromp G, Kuivaniemi H, Gretarsdottir S, Baas AF, Giusti B, Strauss E, van 't Hof FN, Webb T, Erdman R, Ritchie MD, Elmore JR, Verma A, Pendergrass S, Kullo IJ, Ye Z, Peissig PL, Gottesman O, Verma SS, Malinowski J, Rasmussen-Torvik LJ, Borthwick K, Smelser DT, Crosslin DR, de Andrade M, Ryer EJ, McCarty CA, Bottinger EP, Pacheco JA, Crawford DC, Carrell DS, Gerhard GS, Franklin DP, Carey DJ, Phillips VL, Williams MJ, Wei W, Blair R, Hill AA, Vasudevan TM, Lewis DR, Thomson IA, Krysa J, Hill GB, Roake J, Merriman TR, Oszkinis G, Galora S, Saracini C, Abbate R, Pulli R, Pratesi C, Saratzis A, Verissimo A, Bumpstead SJ, Badger SA, Clough RE, Cockerill GW, Hafez H, Scott DJ, Futers TS, Romaine SP, Bridge K, Griffin KJ, Bailey MA, Smith A, Thompson MM, van Bockxmeer F, Matthiasson SE, Thorleifsson G, Thorsteinsdottir U, Blankensteijn JD, Teijink JA, Wijmenga C, de Graaf J, Kiemeney LA, Lindholt JS, Hughes AE, Bradley DT, Stirrups K, Golledge J, Norman PE, Powell JT, Humphries SE, Hamby SE, Goodall AH, Nelson CP, Sakalihasan N, Courtois A, Ferrell RE, Eriksson P, Folkersen L, Franco-Cereceda A, Eicher JD, Johnson AD, Betsholtz C, Ruusalepp A, Franzén O, Schadt E, Björkegren JL, Lipovich L, Drolet AM, Verhoeven E, Zeebregts CJ, Geelkerken RH, van Sambeek MR, van Sterkenburg SM, de Vries J-PP, Stefansson K, Thompson JR, de Bakker PI, Deloukas P, Sayers RD, Harrison S, van Rij AM, Samani NJ, Bown MJ. Meta-Analysis of Genome-Wide Association Studies for Abdominal Aortic Aneurysm Identifies Four New Disease-Specific Risk Loci. Circ Res. 2016 Nov 29; PMID: 27899403

20. Basile AO, Verma A, Byrska-Bishop M, Pendergrass SA, Darabos C, Lester Kirchner H. PATTERNS IN BIOMEDICAL DATA-HOW DO WE FIND THEM? Pac Symp Biocomput Pac Symp Biocomput. 2016;22:177–183. PMID: 27896973

21. Bauer CR, Lavage D, Snyder J, Leader J, Mahoney JM, Pendergrass SA. OPENING THE DOOR TO THE LARGE SCALE USE OF CLINICAL LAB MEASURES FOR ASSOCIATION TESTING: EXPLORING DIFFERENT METHODS FOR DEFINING PHENOTYPES. Pac Symp Biocomput Pac Symp Biocomput. 2016;22:356–367. PMID: 27896989

22. Verma SS, Frase AT, Verma A, Pendergrass SA, Mahony S, Haas DW, Ritchie MD. PHENOME-WIDE INTERACTION STUDY (PheWIS) IN AIDS CLINICAL TRIALS GROUP DATA (ACTG). Pac Symp Biocomput Pac Symp Biocomput. 2016;21:57–68. PMCID: PMC4722952

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