Dr. Sarah A. Pendergrass

Assistant Professor, Biomedical and Translational Informatics Institute Geisinger Health System

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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, 2015-present Geisinger Health System

Research Interests: Characterization of the relationship between genetic variation, phenotypic outcome, and common complex disease. Analyses have included electronic health record data from the Geisinger Health System Biorepository the *MyCode Community Health Initiative*, the Electronic Medical Records and Genomics (eMERGE) network, the Pharmacogenomics Research Network (PGRN), the Aids Clinical Trials Group (ACTG), as well as epidemiological studies.

Research Faculty, Center for System Genomics, Department of Biochemistry and
Molecular Biology
Lab of Dr. Marylyn Ritchie

Postdoctoral Research Fellow, Center for Human Genetics Research, Vanderbilt University, Nashville TN Advisors: Dr. Marylyn Ritchie, Dr. Dana Crawford

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"

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"

Awards and Grants:

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

Current Funding:

No assignment # (PENDERGRASS, SARAH) 9/1/2016-6/30/2017 0.72 cal. mos. GHS GT Collaboration

IDENTIFICATION OF GENES AND DISEASES ASSOCIATED WITH DEVELOPMENTAL ROBUSTNESS Phenotypic robustness is a predicted consequence of stabilizing selection and it has been proposed to confer resistance to disease. To date, this hypothesis has proved exceedingly difficult to demonstrate in humans. We will take advantage of a unique combination of datasets to perform a thorough analysis of a promising marker of developmental robustness and identify the genetic causes and clinical consequences of phenotypic heterogeneity.

No assignment # (KIRCHNER, H. LESTER) 8/15/2016-4/30/2020 2.4 cal. mos. University of Pennsylvania

APPROACHES TO GENETIC HETEROGENEITY OF OBSTRUCTIVE SLEEP APNEA

This study proposes to use both genome- and phenome-based approaches to identify OSA-related genes and variants using large patient cohorts from multiple institutions in the United States.

U01GH008679 (WILLIAMS, MARC; RITCHIE, MARYLYN) 9/1/2015 -5/31/2019 1.8 cal. mos. NIH

EMR-LINKED BIOBANK FOR TRANSLATIONAL GENOMICS (EMERGE III)

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

No assignment # (RITCHIE, MARYLYN) 6/1/2015-5/31/2019 1.2 cal. mos. Pennsylvania Department of Health

INTEGRATING BIG DATA FOR BIOMEDICAL DISCOVERY: METHODS, TOOLS, AND APPLICATIONS. We propose 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.

Teaching/Mentoring Experience

Postdoctoral Fellows Mentored

Dr. Chris Bauer Dr. Yanfei Zhang

Graduate Students Mentored

Brett Beaulieu-Jones — University of Pennsylvania Anurag Verma — The Pennsylvania State University Thang Li — The Pennsylvania State University

Courses Taught

"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:

Mentorship Committee Member for Dr. Kevin Ho 2017

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 accepted, Nicole Restrepo, Mariusz Butkiewicz have offer letters)

Contact with The Commonwealth Medical College to recruit gap year students

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:

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 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:

"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

2010 Annual Meeting of the International Genetic Epidennology Society, Boston, MA, October 10, 2010

Peer Reviewed Publications

- 1. Verma A, Bradford Y, Dudek S, Verma SS, Pendergrass SA, Ritchie MD. A simulation study investigating power estimates in Phenome-Wide Association Studies. bioRxiv. 2017;115550.
- 2. 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
- 3. 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
- 4. 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

- 5. 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
- 6. 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
- 7. 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
- 8. Verma SS, Lucas AM, Lavage DR, Leader JB, Metpally R, Krishnamurthy S, Dewey F, Borecki I, Lopez A, Overton J, Penn J, Reid J, Pendergrass SA, Breitwieser G, Ritchie MD. IDENTIFYING GENETIC ASSOCIATIONS WITH VARIABILITY IN METABOLIC HEALTH AND BLOOD COUNT LABORATORY VALUES: DIVING INTO THE QUANTITATIVE TRAITS BY LEVERAGING LONGITUDINAL DATA FROM AN EHR. Pac Symp Biocomput Pac Symp Biocomput. 2016;22:533–544. PMID: 27897004
- 9. Verma A, Verma SS, Pendergrass SA, Crawford DC, Crosslin DR, Kuivaniemi H, Bush WS, Bradford Y, Kullo I, Bielinski SJ, Li R, Denny JC, Peissig P, Hebbring S, De Andrade M, Ritchie MD, Tromp G. eMERGE Phenome-Wide Association Study (PheWAS) identifies clinical associations and pleiotropy for stop-gain variants. BMC Med Genomics. 2016;9 Suppl 1:32. PMID: 27535653
- Verma A, Basile AO, Bradford Y, Kuivaniemi H, Tromp G, Carey D, Gerhard GS, Crowe JE, Ritchie MD, Pendergrass SA. Phenome-Wide Association Study to Explore Relationships between Immune System Related Genetic Loci and Complex Traits and Diseases. PloS One. 2016;11(8):e0160573. PMID: 27508393
- 11. Oetjens MT, Bush WS, Denny JC, Birdwell K, Kodaman N, Verma A, Dilks HH, Pendergrass SA, Ritchie MD, Crawford DC. Evidence for extensive pleiotropy among pharmacogenes. Pharmacogenomics. 2016 Jun 1;0. PMID: 27249515
- 12. Butkiewicz M, Cooke Bailey JN, Frase A, Dudek S, Yaspan BL, Ritchie MD, Pendergrass SA, Haines JL. Pathway analysis by randomization incorporating structure-PARIS: an update. Bioinforma Oxf Engl. 2016 Mar 7; PMID: 27153576
- 13. Verma A, Leader JB, Verma SS, Frase AT, Wallace J, Dudek S, Lavage D, Van Hout C, Dewey FE, Penn J, Lopez A, Overton JD, Carey DJ, Ledbetter DH, Kirchner LH, Ritchie MD, Pendergrass SA. Integrating clinical laboratory measures and ICD-9 code diagnoses in phenome-wide association studies. Pac Symp Biocomput. 2016;
- 14. Leader JB, Pendergrass SA, Verma A, Carey DJ, Hartzel D, Ritchie MD, Kirchner LH. Contrasting Association Results between Existing PheWAS Phenotype Definition Methods and Five Validated Electronic Phenotypes. AMIA Annu Symp Proc AMIA Symp AMIA Symp. 2015;
- 15. Dumitrescu L, Restrepo NA, Goodloe R, Boston J, Farber-Eger E, Pendergrass SA, Bush WS, Crawford DC. Towards a phenome-wide catalog of human clinical traits impacted by genetic ancestry. BioData Min. 2015;8:35. PMCID: PMC4642611
- 16. Tyler AL, Crawford DC, Pendergrass SA. The detection and characterization of pleiotropy: discovery, progress, and promise. Brief Bioinform. 2015 Jul 28; PMID: 26223525

- 17. Crawford DC, Goodloe R, Farber-Eger E, Boston J, Pendergrass SA, Haines JL, Ritchie MD, Bush WS. Leveraging Epidemiologic and Clinical Collections for Genomic Studies of Complex Traits. Hum Hered. 2015;79(3–4):137–146. PMCID: PMC4528966
- 18. Pendergrass SA, Verma A, Okula A, Hall MA, Crawford DC, Ritchie MD. Phenome-Wide Association Studies: Embracing Complexity for Discovery. Hum Hered. 2015;79(3–4):111–123. PMID: 26201697
- 19. Pendergrass SA, Ritchie MD. Phenome-Wide Association Studies: Leveraging Comprehensive Phenotypic and Genotypic Data for Discovery. Curr Genet Med Rep. 2015 Apr 4;3(2):92–100.
- 20. Hall MA, Verma SS, Wallace J, Lucas A, Berg RL, Connolly J, Crawford DC, Crosslin DR, de Andrade M, Doheny KF, Haines JL, Harley JB, Jarvik GP, Kitchner T, Kuivaniemi H, Larson EB, Carrell DS, Tromp G, Vrabec TR, Pendergrass SA, McCarty CA, Ritchie MD. Biology-Driven Gene-Gene Interaction Analysis of Age-Related Cataract in the eMERGE Network. Genet Epidemiol. 2015 May 17; PMID: 25982363
- 21. Moore CB, Verma A, Pendergrass S, Verma SS, Johnson DH, Daar ES, Gulick RM, Haubrich R, Robbins GK, Ritchie MD, Haas DW. Phenome-wide Association Study Relating Pretreatment Laboratory Parameters With Human Genetic Variants in AIDS Clinical Trials Group Protocols. Open Forum Infect Dis. 2015 Jan 1;2(1):ofu113.
- 22. Hall MA, Verma SS, Wall DP, Moore JH, Keating B, Campbell DB, Gibson G, Asselbergs FW, Pendergrass S. Session introduction: 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. Pac Symp Biocomput Pac Symp Biocomput. 2015;20:156–160. PMID: 25592577
- 23. Ritchie MD, Holzinger ER, Li R, Pendergrass SA, Kim D. Methods of integrating data to uncover genotype-phenotype interactions. Nat Rev Genet. 2015 Feb;16(2):85–97. PMID: 25582081
- 24. Chhibber A, Kroetz DL, Tantisira KG, McGeachie M, Cheng C, Plenge R, Stahl E, Sadee W, Ritchie MD, Pendergrass SA. Genomic architecture of pharmacological efficacy and adverse events. Pharmacogenomics. 2014 Dec;15(16):2025–2048. PMID: 25521360
- 25. Barrie ES, Weinshenker D, Verma A, Pendergrass SA, Lange LA, Ritchie MD, Wilson JG, Kuivaniemi H, Tromp G, Carey DJ, Gerhard GS, Brilliant MH, Hebbring SJ, Cubells JF, Pinsonneault JK, Norman GJ, Sadee W. Regulatory polymorphisms in human DBH affect peripheral gene expression and sympathetic activity. Circ Res. 2014 Dec 5;115(12):1017–1025. PMCID: PMC4258174
- 26. Kim D, Li R, Dudek SM, Frase AT, Pendergrass SA, Ritchie MD. Knowledge-driven genomic interactions: an application in ovarian cancer. BioData Min. 2014;7:20. PMCID: PMC4161273
- 27. Cronin RM, Field JR, Bradford Y, Shaffer CM, Carroll RJ, Mosley JD, Bastarache L, Edwards TL, Hebbring SJ, Lin S, Hindorff LA, Crane PK, Pendergrass SA, Ritchie MD, Crawford DC, Pathak J, Bielinski SJ, Carrell DS, Crosslin DR, Ledbetter DH, Carey DJ, Tromp G, Williams MS, Larson EB, Jarvik GP, Peissig PL, Brilliant MH, McCarty CA, Chute CG, Kullo IJ, Bottinger E, Chisholm R, Smith ME, Roden DM, Denny JC. Phenome-wide association studies demonstrating pleiotropy of genetic variants within FTO with and without adjustment for body mass index. Front Genet. 2014;5:250. PMCID: PMC4134007
- 28. Kraja AT, Chasman DI, North KE, Reiner AP, Yanek LR, Kilpeläinen TO, Smith JA, Dehghan A, Dupuis J, Johnson AD, Feitosa MF, Tekola-Ayele F, Chu AY, Nolte IM, Dastani Z, Morris A, Pendergrass SA, Sun YV, Ritchie MD, Vaez A, Lin H, Ligthart S, Marullo L, Rohde R, Shao Y, Ziegler MA, Im HK, Schnabel RB, Jørgensen T, Jørgensen ME, Hansen T, Pedersen O, Stolk RP, Snieder H, Hofman A, Uitterlinden AG, Franco OH, Ikram MA, Richards JB, Rotimi C, Wilson JG, Lange L, Ganesh SK, Nalls M, Rasmussen-Torvik LJ, Pankow JS, Coresh J, Tang W, Linda Kao WH, Boerwinkle E, Morrison AC, Ridker PM, Becker DM, Rotter JI, Kardia SLR, Loos RJF, Larson MG, Hsu Y-H, Province MA, Tracy R, Voight BF, Vaidya D, O'Donnell CJ, Benjamin EJ, Alizadeh BZ, Prokopenko I, Meigs JB, Borecki IB. Pleiotropic genes for metabolic syndrome and inflammation. Mol Genet Metab. 2014 Aug;112(4):317–338.

- 29. Hall MA, Verma A, Brown-Gentry KD, Goodloe R, Boston J, Wilson S, McClellan B, Sutcliffe C, Dilks HH, Gillani NB, Jin H, Mayo P, Allen M, Schnetz-Boutaud N, Crawford DC, Ritchie MD, Pendergrass SA. Detection of pleiotropy through a Phenome-wide association study (PheWAS) of epidemiologic data as part of the Environmental Architecture for Genes Linked to Environment (EAGLE) study. PLoS Genet. 2014 Dec;10(12):e1004678. PMCID: PMC4256091
- 30. Ciesielski TH, Pendergrass SA, White MJ, Kodaman N, Sobota RS, Huang M, Bartlett J, Li J, Pan Q, Gui J, Selleck SB, Amos CI, Ritchie MD, Moore JH, Williams SM. Diverse convergent evidence in the genetic analysis of complex disease: coordinating omic, informatic, and experimental evidence to better identify and validate risk factors. BioData Min. 2014;7:10. PMCID: PMC4112852
- 31. Mitchell SL, Hall JB, Goodloe RJ, Boston J, Farber-Eger E, Pendergrass SA, Bush WS, Crawford DC. Investigating the relationship between mitochondrial genetic variation and cardiovascular-related traits to develop a framework for mitochondrial phenome-wide association studies. BioData Min. 2014;7:6. PMCID: PMC4021623
- 32. Kocarnik JM, Pendergrass SA, Carty CL, Pankow JS, Schumacher FR, Cheng I, Durda P, Ambite JL, Deelman E, Cook NR, Liu S, Wactawski-Wende J, Hutter C, Brown-Gentry K, Wilson S, Best LG, Pankratz N, Hong C-P, Cole SA, Voruganti VS, Bůžkova P, Jorgensen NW, Jenny NS, Wilkens LR, Haiman CA, Kolonel LN, Lacroix A, North K, Jackson R, Le Marchand L, Hindorff LA, Crawford DC, Gross M, Peters U. Multiancestral analysis of inflammation-related genetic variants and C-reactive protein in the population architecture using genomics and epidemiology study. Circ Cardiovasc Genet. 2014 Apr 1;7(2):178–188. PMCID: PMC4104750
- 33. Chhibber A, Mefford J, Stahl EA, Pendergrass SA, Baldwin RM, Owzar K, Li M, Winer EP, Hudis CA, Zembutsu H, Kubo M, Nakamura Y, McLeod HL, Ratain MJ, Shulman LN, Ritchie MD, Plenge RM, Witte JS, Kroetz DL. Polygenic inheritance of paclitaxel-induced sensory peripheral neuropathy driven by axon outgrowth gene sets in CALGB 40101 (Alliance). Pharmacogenomics J. 2014 Feb 11; PMID: 24513692
- 34. Mitchell SL, Goodloe R, Brown-Gentry K, Pendergrass SA, Murdock DG, Crawford DC. Characterization of mitochondrial haplogroups in a large population-based sample from the United States. Hum Genet. 2014 Feb 1; PMID: 24488180
- 35. Pendergrass S, Girirajan S, Selleck S. Uncovering the etiology of autism spectrum disorders: genomics, bioinformatics, environment, data collection and exploration, and future possibilities. Pac Symp Biocomput Pac Symp Biocomput. 2014;19:422–426. PMID: 24297568
- 36. Hall MA, Dudek SM, Goodloe R, Crawford DC, Pendergrass SA, Peissig P, Brilliant M, McCarty CA, Ritchie MD. Environment-wide association study (EWAS) for type 2 diabetes in the Marshfield Personalized Medicine Research Project Biobank. Pac Symp Biocomput Pac Symp Biocomput. 2014;200–211. PMCID: PMC4037237
- 37. Tyler AL, Crawford DC, Pendergrass SA. Detecting and characterizing pleiotropy: new methods for uncovering the connection between the complexity of genomic architecture and multiple phenotypes- session introduction. Pac Symp Biocomput Pac Symp Biocomput. 2014;19:183–187. PMID: 24297545
- 38. Moore CB, Wallace JR, Wolfe DJ, Frase AT, Pendergrass SA, Weiss KM, Ritchie MD. Low frequency variants, collapsed based on biological knowledge, uncover complexity of population stratification in 1000 genomes project data. PLoS Genet. 2013 Dec;9(12):e1003959. PMCID: PMC3873241
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