CURRICULUM VITAE

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H-index: 94 (Google Scholar; as of 5/30/2023)

EDUCATION

1999, BS University of Pittsburgh at Johnstown (Biology)
2002, MS Vanderbilt University (Applied Statistics)
2004, PhD Vanderbilt University (Statistical Genetics)
Genetic Programming Optimized Neural Networks for Detecting Gene-Gene Interactions

PROFESSIONAL EMPLOYMENT

Academic Appointments

2004-2009 Assistant Professor
Department of Molecular Physiology & Biophysics
Vanderbilt University

2004-2011 Investigator
Center for Human Genetics Research
Vanderbilt University

2009-2011 Associate Professor with tenure
Department of Molecular Physiology & Biophysics
Vanderbilt University

2009-2011 Associate Professor, Secondary Appointment
Department of Biomedical Informatics
Vanderbilt University

2011- 2014 Associate Professor with tenure
Department of Biochemistry and Molecular Biology
Pennsylvania State University

2011-2016	Director, Center for Systems Genomics The Huck Institutes of the Life Sciences Pennsylvania State University
2014-2016	Professor with tenure Department of Biochemistry and Molecular Biology Pennsylvania State University
2015-2016	Paul Berg Professor Department of Biochemistry and Molecular Biology Pennsylvania State University
2015-2017	Director, Biomedical and Translational Informatics Institute Professor of Biomedical and Translational Informatics Geisinger Health System
2016-2017	Chief Research Informatics Officer Geisinger Health System
2016-2018	Fixed-term, Part-time Professor Department of Biochemistry and Molecular Biology Pennsylvania State University
2017-2018	Faculty Pending, Department of Genetics University of Pennsylvania, Perelman School of Medicine
2017-2022	Associate Director for Bioinformatics Institute for Biomedical Informatics University of Pennsylvania, Perelman School of Medicine
2017-2023	Associate Director, Center for Precision Medicine University of Pennsylvania, Perelman School of Medicine
2017-present	Director, Center for Translational Bioinformatics Institute for Biomedical Informatics University of Pennsylvania, Perelman School of Medicine
2018-present	Professor with tenure, Department of Genetics University of Pennsylvania, Perelman School of Medicine
2022-present	Director, Institute for Biomedical Informatics University of Pennsylvania, Perelman School of Medicine
2022-present	Vice President for Research Informatics University of Pennsylvania Health System
2022-present	Edward Rose, M.D. and Elizabeth Kirk Rose, M.D. Professor University of Pennsylvania, Perelman School of Medicine

2023- present	Director, Division of Informatics, Department of Biostatistics, Epidemiology, and Informatics. University of Pennsylvania, Perelman School of Medicine

Other Professional Positions

2005-2010	Consultant, Boehringer-Ingelheim
2018-2022	Scientific Advisory Board, Cipherome
2019-2020	Scientific Advisory Board, Goldfinch Bio

Honors and Awards

1999	Graduated Summa Cum Laude, University of Pittsburgh, Johnstown
2000- 2002	NIH Breast Cancer Research Training Grant
2001	Vanderbilt University, Charles R. Park Student Travel Award
2002- 2003	NLM Bioinformatics Training Grant
2002	Vanderbilt University Graduate School Travel Grant
2003	Vanderbilt University Dissertation Enhancement Grant
2003	Vanderbilt University Graduate School Travel Grant
2003	Vanderbilt University, Charles R. Park Student Travel Award
2004	Best Paper Award, Genetic and Evolutionary Computation Conference
2006	*Genome Technology*, Rising Young Investigator Award
2010	Sloan Research Fellow
2011-2014	KAVLI Frontiers in Science fellow, National Academy of Science Jointly sponsored by the US National Academy of Sciences and The Kavli Foundation, the Kavli Frontiers of Science bring together some of the very best young scientists to discuss exciting advances and opportunities in their fields in a format that encourages informal collective, as well as one-on-one discussions among participants. It is an honor to be selected as a Kavli fellow, and Dr. Ritchie was selected four years in a row.
2014	Member, Thomas Reuters, Most Highly Cited Researchers
2015	Paul Berg Professor of Biochemistry and Molecular Biology, Penn State University
2016	E. Allen Deaver, High Potential Leadership Fellow, Geisinger Health System
2017	First Place, AMIA "Why Informatics" Video Contest
2020	Group on Information Resources (GIR) Excellence Award, American Association of Medical Colleges (AAMC) – for our team's PennChart Genomics Project
2020	Elected as a fellow in the American College of Medical Informatics (ACMI)
2021	Executive Leadership in Academic Medicine (ELAM) fellow
2021	Elected into the National Academy of Medicine (NAM)
2022	Edward Rose, M.D. and Elizabeth Kirk Rose, M.D. Professor, University of Pennsylvania, Perelman School of Medicine

Membership in Professional Societies

American Association for the Advancement of Science (AAAS)
American Society of Human Genetics (ASHG)

International Society for Computational Biology (ISCB)
American Medical Informatics Association (AMIA)
American College of Medical Informatics (ACMI)
National Academy of Medicine (NAM)

Administrative and Service Contributions

Institutional Service

2003-2004	Center for Human Genetics Research Core Oversight Board Vanderbilt University Medical Center, Nashville, TN
2005	Organizing Committee—CHGR Annual Genetics Symposium Vanderbilt University Medical Center, Nashville, TN
2006	ACCRE Study Group, co-chair Vanderbilt University, Nashville, TN
2006	Panel Member—Women in Academe Series Vanderbilt University Medical Center, Nashville, TN
2008	Faculty Recruitment Committee Center for Human Genetics Research
2009	Faculty Recruitment Committee, chair Center for Human Genetics Research
2009	Personalized Medicine Task Force – Vision 2020 Vanderbilt University
2009	IGP Internal Review Administrative Subcommittee Vanderbilt University
2004-2001	Faculty Director, CHGR Computational Genomics Core Vanderbilt University Medical Center, Nashville, TN
2007-2011	ACCRE Faculty Advisory Board Vanderbilt University, Nashville, TN
2008-2011	Director, Program in Computational Genomics Vanderbilt University, Center for Human Genetics Research
2011	Biobehavioral Health Genetics Faculty Search Committee The Pennsylvania State University, University Park, PA
2011-2012	PSU Presidential Task Force The Pennsylvania State University, University Park, PA
2011-2013	BMB Graduate Student Recruitment Committee The Pennsylvania State University, University Park, PA

2012-2013	Institute of Cyberscience, Search Committee The Pennsylvania State University, University Park, PA
2012-2013	iBIOS Bioinformatics & Genomics Graduate Program Admissions Committee The Pennsylvania State University, University Park, PA
2012-2013	Research Data and Computing Committee The Pennsylvania State University, University Park, PA
2012-2014	IT Strategic Planning Committee The Pennsylvania State University, University Park, PA
2014-2015	Chair, BMB Promotion & Tenure Committee The Pennsylvania State University, University Park, PA
2011-2014	BMB Junior Faculty Mentoring Committee The Pennsylvania State University, University Park, PA
2011-2016	Director, Center for System Genomics The Pennsylvania State University, University Park, PA
2012-2014	BMB Promotion & Tenure Committee (2014 – Chair) The Pennsylvania State University, University Park, PA
2014	Research CI Governance Task Force The Pennsylvania State University, University Park, PA
2014	ICS Coordinating Committee for Advanced CI The Pennsylvania State University, University Park, PA
2014	Academic Integrity Committee – RA10 The Pennsylvania State University, University Park, PA
2015-2017	Enterprise Data Strategy Steering Committee Geisinger Health System, Danville, PA
2016-2017	Research Space Committee, Chair Geisinger Health System Danville, PA
2016-2017	Chief Research Informatics Officer Geisinger Health System, Danville, PA
2018-2019	Faculty recruitment selection committee, Department of Genetics University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA
2018-2022	Senior IT Council (member) University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2018-2019 Clinical and Medical Bioinformatics Working Group
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019-2021 Clinical Research Informatics Oversight Committee
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019-2022 Research Data Domain Committee, co-chair
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019-present Clinical IS Genomics Oversight Committee
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019-2021 Biomedical Informatics Graduate Group Planning Committee
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019-2021 Epic Cosmos User Group member
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2020 DBEI Department Chair search Committee
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2020-present Committee on Appointments and Promotions
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2020-present Genomics and Computational Biology Graduate Group Committee on Equality, Diversity and Inclusion, co-chair
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2020-present Genomics and Computational Biology Graduate Group Executive Committee member
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022-present Senior IT Council (co-chair)
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022-2023 Pathology and Laboratory Medicine Department Chair search committee
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022-2023 Genetics/IBI Faculty search committee
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022-2023 Cardiology/Cardiovascular Institute Faculty search committee
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022-2023 Computational and Data-Dependent Research committee
University of Pennsylvania, Philadelphia, PA

National / International Service

2002	Grant Reviewer, "Small Grants Program for Cancer Epidemiology." PA-01-021, National Cancer Institute, National Institutes of Health
2004	Workshop Organizer, Biological Applications of Genetic and Evolutionary Computation
2005	Session Organizer, Computational Approaches for Pharmacogenomics, Pacific Symposium on Biocomputing
2005	Program Committee, Biological Applications of Genetic and Evolutionary Computation
2005	Workshop Organizer, Biological Applications of Genetic and Evolutionary Computation
2006	Grant Reviewer, "Toward Maximizing the Scientific Value of the Biologic Specimens from the Women's Health Initiative." RFP-NHLBI-WH-06-09, National Heart, Lung, and Blood Institute, National Institutes of Health
2006	Session Organizer, Computational Approaches for Pharmacogenomics, Pacific Symposium on Biocomputing
2006	Program Committee, Biological Applications of Genetic and Evolutionary Computation
2006	Workshop Organizer, Biological Applications of Genetic and Evolutionary Computation
2006	Program Committee, European Conference on Evolutionary Computation, Machine Learning and Data Mining in Bioinformatics
2006	Workshop Organizer, PGRN Data Analysis Workshop
2007	Program Committee, Genetic and Evolutionary Computation Conference
2007	Program Committee, European Conference on Evolutionary Computation, Machine Learning and Data Mining in Bioinformatics
2007	Grant Reviewer, "Facilitating Interdisciplinary Research via Methodological and Technological Innovation in the Behavioral and Social Sciences (R21)." RFA-RM-07-004, National Institutes of Health
2007	Grant Reviewer, "Small Grants Program in Cancer Epidemiology." PAR-06-294, National Cancer Institute (June and November), National Institutes of Health
2008	Program Committee, Genetic and Evolutionary Computation Conference
2008	Program Committee, International Conference on Intelligent Systems in Molecular Biology
2008	Grant Reviewer, "PA Department of Health Final Performance Reviews." Oak Ridge Associated Universities
2008	Grant Reviewer, "GWA Studies for the Genes, Environment, and Health Initiatives." RFA-HG-07-012, National Human Genome Research Institute, National Institutes of Health

2008	Grant Reviewer, "Toward Maximizing the Scientific Value of the Biologic Specimens from the Women's Health Initiative II." BAA HLBI-WH-09-01, National Heart, Lung, and Blood Institute, National Institutes of Health
2008	Grant Reviewer, "Genomic Parsing of Bipolar Disorder and Schizophrenia: Collaborative R01's (Teleconference)". Special Emphasis Panel/Scientific Review Group 2008/08 ZMH1 ERB-S (08). National Institute of Mental Health, National Institutes of Health
2008	Grant Reviewer, Israel Science Foundation's FIRST (Focal Initiatives in Research in Science and Technology)
2008	Grant Reviewer, "Small Grants Program in Cancer Epidemiology." PAR-06-294, National Cancer Institute (June), National Institutes of Health
2009	Grant Reviewer, "GWAS of Treatment Response in Randomized Clinical Trials – Study Investigators." RFA-HG-08-004, National Human Genome Research Institute, National Institutes of Health
2009	Meeting Participant, "The Challenge of Mapping GWAS Signals." Hosted by the Genes, Environment and Health Initiative (GEI), and National Cancer Institute. National Institutes of Health.
2009	Conference Chair, European Conference on Evolutionary Computation, Machine Learning and Data Mining in Bioinformatics
2010	Conference Chair, European Conference on Evolutionary Computation, Machine Learning and Data Mining in Bioinformatics
2010	Program Committee, Genetic and Evolutionary Computation Conference
2010	Program Committee, The 2010 International Conference on Intelligent Computing (ICIC 2010)
2010	Session Chair, Genetic and Evolutionary Computation Conference
2010	Program Committee, European Conference on Computational Biology
2010	Program Committee, Pattern Recognition in Bioinformatics
2010	Host, P-STAR Analysis Workshop, October 2010, Nashville, TN
2005-2010	Grant Reviewer, Alzheimer's Association
2006-2010	Pharmacogenetics Research Network (PGRN) Analysis Working Group Co-Chair
2011	Workshop Chair, "Systems Pharmacogenomics: Birds of a Feather", Pacific Symposium on Biocomputing 2011
2011	Conference Chair, European Conference on Evolutionary Computation, Machine Learning and Data Mining in Bioinformatics
2011	Program Committee, Asia-Pacific Translational Bioinformatics Conference
2011	External Examiner for Viva, University of Nottingham, Nottingham UK
2011	Program Committee, The 2011 International Conference on Intelligent Computing (ICIC 2011)

2011	Grant Reviewer, "Computational Tool Development and Integrative Data Analysis for LINCS (U01)", RFA- RM-10-005, Special Emphasis Panel, National Institutes of Health
2011	Grant Reviewer, Ad Hoc, Genomics, Computational Biology and Technology (GCAT), National Institutes of Health
2011	Bioinformatics Track Chair, Genetic and Evolutionary Computation Conference
2011	Program Committee, European Conference on Artificial Life 2011
2011	Program Committee, Pattern Recognition in Bioinformatics 2011
2011	Host, P-STAR Analysis Workshop, December 2011, Chicago, IL
2011	Session Chair, "Beyond GWAS: Integrating Transcriptome, Proteome, and Pathway Data in the Genetic Dissection of Complex Traits", International Congress on Human Genetics, October 2011, Montreal, Canada
2012	Workshop Chair, "Systems Pharmacogenomics", Pacific Symposium on Biocomputing
2012	Local Host, Genetic and Evolutionary Computation Conference, Philadelphia, PA
2012	Grant Reviewer, Genome Canada
2012	Program Committee, Asia-Pacific Translational Bioinformatics Conference
2012	External Examiner for PhD, University of Liege
2012	Host, P-STAR Analysis Workshop, December 2012, Austin TX
2012	Grant Reviewer, Special Emphasis Panel 2012/10 ZEY1 VSN, National Eye Institute
2012	Grant Reviewer, Special Emphasis Panel 2012/05 ZAI1 QV-I, National Institute of Allergy and Infectious Disease
2012	Grant Reviewer, Science Foundation Ireland
2012	Organizing Committee, KAVLI Frontiers in Science, American-Indonesian conference
2013	Organizing Committee, KAVLI Frontiers in Science, American-Indonesian conference
2013	Program Committee, Asia-Pacific Translational Bioinformatics Conference
2013	Guest Editor, PLOS Genetics
2013	Guest Editor, Frontiers in Genetics
2013	Program Committee, Translational Bioinformatics Conference
2013	Program Committee, The 2013 International Conference on Intelligent Computing
2013	Program Committee, Pattern Recognition in Bioinformatics 2013
2013	Host, P-STAR Analysis Workshop, December 2013, Nashville TN
2014	Organizing Committee, Pacific Symposium of Biocomputing
2014	Track Chair, Bioinformatics, ACM-BCB
2014	Program Committee, Translational Bioinformatics Conference
2014	Program Committee, ICCABS
2014	Program Committee, World Congress Psychiatric Genetics

2014	Program Committee, Asia-Pacific Translational Bioinformatics Conference
2014	Grant Reviewer, Medical Research Council (MRC), United Kingdom
2014	Organizing Committee, KAVLI Frontiers in Science, American-Indonesian conference
2015	Organizing Committee, Pacific Symposium on Biocomputing
2015	Program Committee, AMIA
2015	Program Committee, Translational Bioinformatics Conference
2016	Organizing Committee, Pacific Symposium on Biocomputing
2016	Program Committee, Translational Bioinformatics Conference
2017	Organizing Committee, Pacific Symposium on Biocomputing
2017	Vice Chair, AMIA Translational Bioinformatics Conference
2017	Co-Host, Translational Bioinformatics Conference
2017	Grant Reviewer, Canada Excellence Research Chair
2017	Grant Reviewer, Wellcome Trust, Sir Henry Dale Fellowship
2018	Organizing Committee, Pacific Symposium on Biocomputing
2007-2012	Grant Reviewer, Pennsylvania Performance Review, PA Department of Health
2007-2017	Member, Faculty of 1000 Biology
2008-2015	Member, Genetic Epidemiology Scientific Review Committee for the Department of Veterans Affairs
2009-2010	Executive Editor, Pharmacogenomics
2012-2018	Member, Genomics, Computational Biology and Technology (GCAT), National Institutes of Health
2015-2018	GWAS Catalog Scientific Advisory Board
2016-2018	Member, Parasite Award Committee
2007-2015	Managing Editor, BioData Mining
2008-present	Member Editorial Board, Pharmacogenomics
2011-present	Member Editorial Board, Pharmacogenetics & Genomics
2015-2022	Member, Standing Committee on Biological and Physical Sciences in Space (CBPSS), National Academy of Sciences
2015-present	Editor-in-chief, BioData Mining
2017-2021	Grant Reviewer, Genome Canada
2017-present	Institute for Computational Biology External Advisory Board, Case Western Reserve University
2017-2021	UCSF PREMIER (Precision Medicine in Rheumatology) External Advisory Committee
2017-present	NIH Advisory panel, All of Us Cohort Program
2018-present	NHGRI PhenX Steering Committee (Member)
2019-2020	Vice Chair, Translational Bioinformatics Track, AMIA Joint Summits
2019-present	NHGRI ANVIL External Consultant Committee
2019-2022	NIA Dog Aging Project, Scientific Advisory Board
2019	Fusion Personalized Medicine Conference co-chair

2019-present	Genome Canada Research Oversight Committee, Turvey LSARP Project
2019-present	Genome Canada Research Oversight Committee, Go PGx project
2019-present	Vanderbilt DBMI T32, Scientific Advisory Board
2019-2022	NHGRI LIGHT Workshop Planning Committee
2019-2022	ANVIL Data Ingestion Working Group
2019-present	iCOPE Advisory Board (Childhood Cancer project in Copenhagen, Denmark)
2020-present	UK Biobank International Advisory Board (chair starting in 2022)
2020-present	Annual Review of Biomedical Data Science Editorial Committee
2020-2022	H3ABioNet Scientific Advisory Board
2020	Grant Reviewer, COVID Special Emphasis Panel
2020	Grant Reviewer, Advancing Genomic Medicine, Special Emphasis Panel
2021	Grant Reviewer, NIH Director's Transformative Research Award (TRA)
2021-present	Member, NHGRI Genomic Data Science Working Group (GDSWG) - working group of the National Advisory Council for Human Genome Research (NACHGR)
2021-present	CalPenn Joint Steering Committee (chair starting in 2023)
2022-present	ClinGen PGx Workgroup
2023-present	ACMI 2024 symposium, program chair
2023-present	NHLBI TopMed External Scientific Panel

REVIEWER FOR JOURNALS

2005-Present	Ad Hoc Reviewer, Pacific Symposium on Biocomputing
2005-Present	Hoc Reviewer, Annals of Human Genetics
2005-Present	Ad Hoc Reviewer, Current Bioinformatics
2005-Present	Ad Hoc Reviewer, British Journal of Cancer
2006-Present	Ad Hoc Reviewer, BMC Cancer
2006-Present	Ad Hoc Reviewer, Epidemiology
2006-Present	Ad Hoc Reviewer, Human Genomics
2006-Present	Ad Hoc Reviewer, Clinical Pharmacology & Therapeutics
2006-Present	Ad Hoc Reviewer, International Journal Artificial Intelligence in Medicine
2006-Present	Ad Hoc Reviewer, Genetic Epidemiology
2006-Present	Ad Hoc Reviewer, Psychiatry Research
2007-Present	Ad Hoc Reviewer, BMC Medical Genomics
2007-Present	Managing Editor, BMC BioData Mining
2007-Present	Ad Hoc Reviewer, IBM Systems Journal
2007-Present	Ad Hoc Reviewer, Clinical Cancer
2007-Present	Ad Hoc Reviewer, Clinical Genetics
2007-Present	Ad Hoc Reviewer, Cancer Epidemiology Biomarkers & Prevention
2007-Present	Ad Hoc Reviewer, Cancer Informatics
2007-Present	Ad Hoc Reviewer, American Journal of Human Genetics
2008-Present	Ad Hoc Reviewer, Molecular Psychiatry
2008-Present	Ad Hoc Reviewer, Neuropsychiatric Genetics
2008-Present	Ad Hoc Reviewer, Neoplasia
2008-Present	Ad Hoc Reviewer, Statistics in Medicine
2008-Present	Ad Hoc Reviewer, Human Heredity

2009-Present	Ad Hoc Reviewer, Biometrical Journal
2009-Present	Ad Hoc Reviewer, PLOS Genetics
2009-Present	Ad Hoc Reviewer, Nature Reviews Drug Discovery
2009-Present	Ad Hoc Reviewer, BMC Systems Biology
2009-Present	Ad Hoc Reviewer, Human Immunology
2009-Present	Ad Hoc Reviewer, Bioinformatics
2009-Present	Executive Editor, Pharmacogenomics
2010-Present	Ad Hoc Reviewer, European Journal of Operational Research
2011-Present	Ad Hoc Reviewer, Frontiers in Genetics
2011-Present	Ad Hoc Reviewer, Intelligent Systems in Molecular Biology
2011-Present	Ad Hoc Reviewer, In Silico Biology
2011-Present	Ad Hoc Reviewer, Source Code for Biology and Medicine
2011-Present	Ad Hoc Reviewer, OMICS Integrative Biology
2011-Present	Ad Hoc Reviewer, Pharmacogenomics
2011-Present	Ad Hoc Reviewer, PLOS One
2011-Present	Ad Hoc Reviewer, BioData Mining
2012-Present	Ad Hoc Reviewer, Journal of the American Medical Association (JAMA)
2012-Present	Ad Hoc Reviewer, Cellular and Molecular Life Sciences
2012-Present	Ad Hoc Reviewer, Annals of Neurology
2012-Present	Ad Hoc Reviewer, Pharmacogenetics and Genomics
2012-Present	Ad Hoc Reviewer, Human Molecular Genetics
2013-Present	Ad Hoc Reviewer, BMC Bioinformatics
2013-Present	Ad Hoc Reviewer, Nucleic Acids Research
2013	Guest Editor, PLOS Genetics
2013	Guest Editor, Frontiers in Genetics
2013-Present	Ad Hoc Reviewer, Biostatistics
2013-Present	Ad Hoc Reviewer, European Journal of Human Genetics
2013-Present	Ad Hoc Reviewer, IEEE Transactions in Evolutionary Computation
2013-Present	Ad Hoc Reviewer, Nature Reviews Genetics
2013-Present	Ad Hoc Reviewer, BMC Genetics
2013-Present	Ad Hoc Reviewer, Expert Reviews of Molecular Diagnostics
2013-Present	Ad Hoc Reviewer, Science Translational Medicine
2014-Present	Ad Hoc Reviewer, PLOS Biology
2014-Present	Ad Hoc Reviewer, PLOS Computational Biology
2014-Present	Ad Hoc Reviewer, Nature
2014-Present	Ad Hoc Reviewer, The Pharmacogenomics Journal
2014-Present	Ad Hoc Reviewer, Briefings in Bioinformatics
2014-Present	Ad Hoc Reviewer, Circulation Cardiovascular Genetics
2014-Present	Ad Hoc Reviewer, Proceedings of the National Academy of Science
2014-Present	Ad Hoc Reviewer, Human Genetics
2015-Present	Ad Hoc Reviewer, Science
2015-Present	Ad Hoc Reviewer, EvoBio
2016-Present	Ad Hoc Reviewer, Human Mutation
2017-Present	Ad Hoc Reviewer, Expert Reviews of Precision Medicine
2018-Present	Ad Hoc Reviewer, Nature Communications

2018-Present	Ad Hoc Reviewer, Nature Genetics
2019-present	Ad Hoc Reviewer, JAMA
2019-present	Ad Hoc Reviewer, Molecular Genetics and Genomics
2020-present	Ad Hoc Reviewer, Genome Medicine
2020-present	Ad Hoc Reviewer, Nature Metabolism
2020-present	Ad Hoc Reviewer, Sleep
2020-present	Ad Hoc Reviewer, Nature Medicine
2021-present	Ad Hoc Reviewer, iScience
2022-present	Ad Hoc Reviewer, Science Translational Medicine
2022-present	Ad Hoc Reviewer, Cell Genomics
2023-present	Ad Hoc Reviewer, Nature Protocols

Teaching activities

Vanderbilt Contributions

2002	IGP Biostatistics Module, Lecturer Vanderbilt University
2004	Human Genetics—MPB 340, Lecturer Vanderbilt University
2004	IGP Biostatistics Module, Lecturer Vanderbilt University
2004	Health Systems Management Organization Course, Lecturer Vanderbilt University
2005	Human Genetics, HGEN 341, Lecturer Vanderbilt University
2005	Tutorials in Statistical and Population Genetics, HGEN 371, Co-Organizer Vanderbilt University
2006	Human Genetics, HGEN 341, Lecturer Vanderbilt University
2006	Tutorials in Statistical and Population Genetics, HGEN 371, Co-Organizer Vanderbilt University
2006	Cancer Biology Methodology Course, Lecturer Vanderbilt University
2007	Human Genetics, HGEN 341, Lecturer Vanderbilt University
2007	Tutorials in Statistical and Population Genetics, HGEN 371, Co-Organizer Vanderbilt University
2007	Cancer Biology Methodology Course, Lecturer Vanderbilt University
2008	Human Genetics, HGEN 341, Lecturer Vanderbilt University
2008	Organizer, Program in Computational Genomics Journal Club Vanderbilt University
2009	Human Genetics, HGEN 341, Lecturer Vanderbilt University
2009	Organizer, Program in Computational Genomics Journal Club Vanderbilt University
2009	Biomedical Informatics 310, Lecturer Vanderbilt University

2009 IGP, Human Genetics, Lecturer
Vanderbilt University
2010 Human Genetics, HGEN 341, Lecturer
Vanderbilt University
2010 Biomedical Informatics 310, Lecturer
Vanderbilt University
2011 Human Genetics, HGEN 341, Course Director
Vanderbilt University

The Pennsylvania State University Contributions

2012 Tutorials in Bioinformatics & Genomics, IBIOS 598B FA, Co-director
The Pennsylvania State University, University Park, PA

2012 Genomics, IBIOS/BMMB 551 FA, Lecturer
The Pennsylvania State University, University Park, PA

2013 Tutorials in Bioinformatics & Genomics, IBIOS 598B SP, Co-director
The Pennsylvania State University, University Park, PA

2013 Human Genomics and Biomedical Informatics, BMB498D, Director
The Pennsylvania State University, University Park, PA

2013 Tutorials in Bioinformatics & Genomics, IBIOS 598B FA, Co-director
The Pennsylvania State University, University Park, PA

2013 Genomics, IBIOS/BMMB 551 FA, Lecturer
The Pennsylvania State University, University Park, PA

2014 Critical Analysis in Bioinformatics and Genomics, IBIOS 541 SP, Co-director
The Pennsylvania State University, University Park, PA

2014 Human Genomics and Biomedical Informatics, BMB498D SP, Director
The Pennsylvania State University, University Park, PA

2014 Critical Analysis in Bioinformatics and Genomics, IBIOS 541 FA, Co-director
The Pennsylvania State University, University Park, PA

2014 Genomics, IBIOS/BMMB 551 FA, Lecturer
The Pennsylvania State University, University Park, PA

2015 Genomics, IBIOS/BMMB 551 FA, Lecturer
The Pennsylvania State University, University Park, PA

2016 Genomics, IBIOS/BMMB 551 FA, Lecturer
The Pennsylvania State University, University Park, PA

University of Pennsylvania Contributions

2018 MS1 Genetics Course, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019 BMIN504: Special Topics in Biomedical and Health Informatics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019 MS1 Genetics Course, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019 GCB533: Statistics for Genomics and Biomedical Informatics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2019 BMIN 505: Precision Medicine and Health Policy
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2020 BMIN504: Special Topics in Biomedical and Health Informatics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2020 BMIN 505: Precision Medicine and Health Policy, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2020 GCB533: Statistics for Genomics and Biomedical Informatics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2020 MS1 Genetics Course, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2021 BMIN504: Special Topics in Biomedical and Health Informatics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2021 BMIN505: Precision Medicine and Health Policy, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2021 BMIN502: Database and Data Integration in Biomedical Research, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2021 GCB/CAMB 752: Seminar in Genomics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2021 MS1 Genetics Course, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2021 GCB533: Statistics for Genomics and Biomedical Informatics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022 BMIN504: Special Topics in Biomedical and Health Informatics, Director
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022 BMIN505: Precision Medicine and Health Policy, Lecturer

University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022 BMIN502: Database and Data Integration in Biomedical Research, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022 GCB/CAMB 752: Seminar in Genomics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022 MS4, Frontiers 519 Medical Informatics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022 MS1 Genetics Course, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2022 GCB533: Statistics for Genomics and Biomedical Informatics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2023 BMIN504: Special Topics in Biomedical and Health Informatics, Director
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2023 BMIN502: Database and Data Integration in Biomedical Research, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2023 GCB/CAMB 752: Seminar in Genomics, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

2023 MS1 Genetics Course, Lecturer
University of Pennsylvania, Perelman School of Medicine, Philadelphia, PA

External Contributions

2009 Course Co-Director, Genetic Analysis of Complex Disease
Miami Institute of Human Genetics, University of Miami
2010 Course Co-Director, Genetic Analysis of Complex Disease
Miami Institute of Human Genetics, University of Miami
2010 Lecturer, Vanderbilt-NARI-NIE Informatics Training Program (VNNIT) Workshop, Pune, India
2010 "A comparison of analytical methods for genetic association studies," CDC Workshop, Atlanta, GA
2011 Course Co-Director, Genetic Analysis of Complex Disease
Miami Institute of Human Genetics, University of Miami
2011 "A comparison of analytical methods for genetic association studies II", CDC Workshop, Atlanta, GA
2011 "Next-generation Sequencing", CDC Workshop, Atlanta, GA
2012 Course Co-Director, Genetic Analysis of Complex Disease
Miami Institute of Human Genetics, University of Miami
2012 "Next-generation Sequencing", CDC Workshop, Atlanta, GA
2013 "Next-generation Sequencing", CDC Workshop, Atlanta, GA

2013	Course Co-Director, Genetic Analysis of Complex Disease Miami Institute of Human Genetics, University of Miami
2013	"Analysis Tools For Genetic Testing", CDC Workshop, Atlanta, GA
2014	"Public Health Genomics", CDC Workshop, Atlanta, GA
2014	"Gene-gene and Gene-environment interactions", UAB's 4th Annual NIGMS-funded Short Course on Statistical Genetics & Genomics, Birmingham, AL
2014	"Intro to Bioinformatics", CDC Workshop, Atlanta, GA
2014	"Precision Medicine: When Genomics Gets Personal", CDC Workshop, Atlanta, GA
2015	"Next Generation Sequencing: What is it How is it Used and What Can We Learn", CDC Workshop, Atlanta, GA
2021	"A tale of the 2020s, or a tale of the future?", Precision Medicine Roundtable, Genetics in Medicine class, University of Deusto in Bilbao, Spain. Host: Urko Marigorta

Advisory and Supervisory Responsibilities

High School Students

2007	Xun Miao Martin Luther King High School, Nashville TN

Undergraduate Students

2005	Gordon Lemmon Vanderbilt Summer Science Academy
2006	Theresa Fanelli Vanderbilt Summer Science Academy
2012-2013	Christian Suri The Pennsylvania State University
2013-2014	Tomek Dobranski The Pennsylvania State University
2013-2015	Anastasia Lucas The Pennsylvania State University
2020	David Cornejo APSA Virtual Summer Research Program
2020	Mitsy Dominguez APSA Virtual Summer Research Program
2020-2021	Ly Nguyen APSA Virtual Summer Research Program
2020-2021	Abigail Bossa APSA Virtual Summer Research Program

2020-2021 Chelsea Okeh
APSA Virtual Summer Research Program

2021-2022 Alexis Garofalo
Penn SUIP Summer Research Program

2022 Mariah Antopia
Penn SUIP Summer Research Program

Post-doctoral Fellows – Members of Lab

2009-2011 Sarah A. Pendergrass, PhD (now Rion K. Pendergrass)
Vanderbilt University School of Medicine
Currently, Scientific Manager in Human Genetics, Genentech

2009-2011 Rebecca Zuvich Essner, PhD
Vanderbilt University School of Medicine
Currently, Data Scientist, Advent Health

2013-2016 DoKyoon Kim, PhD
The Pennsylvania State University
Currently, Assistant Professor, University of Pennsylvania

2015-2017 Marta Byrska-Bishop, PhD
Geisinger Health System
Currently, Senior Bioinformatics Scientist, NY Genome Center

2016-2021 Jason Miller, PhD
University of Pennsylvania
Currently, Senior Scientist Statistical Genetics, Merck

2017-2022 Yogasudha Veturi, PhD
University of Pennsylvania

2018-2022 Theodore Drivas, MD, PhD
University of Pennsylvania

2020-2023 Milton Pividori, PhD
University of Pennsylvania, co-mentor with Dr. Casey Greene

2023-present Zinhle Cindi, PhD
University of Pennsylvania

Clinical Fellows – Members of Lab

2020-2022 Nosheen Reza, MD
University of Pennsylvania, MTR program, co-mentor with Dr. Tom Cappola

Graduate Students – Members of Lab

2004-2007 Alison A. Motsinger
Vanderbilt University, Ph.D. Program in Human Genetics
Currently Chief, Biostatistics & Computational Biology Branch and Principal Investigator, NIEHS

2005-2008 Todd L. Edwards
Vanderbilt University, Ph.D. Program in Human Genetics
Currently Associate Professor with tenure at Vanderbilt University (Department of Medicine, Center for Epidemiology)

2004-2009 William S. Bush
Vanderbilt University, Ph.D. Program in Human Genetics
Currently Tenure Track Associate Professor at Case-Western Reserve University (Department of Population and Quantitative Health Sciences)

2006-2010 Stephen D. Turner
Vanderbilt University, Ph.D. Program in Human Genetics
Currently senior informatics science & technology adviser at Signature Science, LLC.

2008-2012 Benjamin Grady
Vanderbilt University, Ph.D. Program in Human Genetics

2009-2013 Emily R. Holzinger
Vanderbilt University, Ph.D. Program in Human Genetics
Currently Senior Scientist, Bristol Myers Squibb

2009-2013 Carrie C Buchanan Moore
Vanderbilt University, Ph.D. Program in Human Genetics
Currently Surgical Resident, Duke University School of Medicine

2011-2013 Neerja Katiyar
Pennsylvania State University, Ph.D. Program in Bioinformatics & Genomics

2011-2015 Molly A Hall
Pennsylvania State University, Ph.D. Program in BMMB
Currently Tenure-track Asst. Prof at Penn State University

2012-2016 Ruowang Li
Pennsylvania State University, Ph.D. Program in Bioinformatics & Genomics
Currently Assistant Professor, Cedars-Sinai

2013-2018 Anna Okula Basile
Pennsylvania State University, Ph.D. Program in BMMB
Currently Bioinformatics Scientist, NY Genome Center

2014-2018 Anurag Verma

Pennsylvania State University, Ph.D. Program in Bioinformatics & Genomics
Currently Instructor, University of Pennsylvania, Perelman School of Medicine

2014-2018 Shefali Setia Verma
Pennsylvania State University, Ph.D. Program in Bioinformatics & Genomics
Currently Instructor, University of Pennsylvania, Perelman School of Medicine

2016-2020 Binglan Li
University of Pennsylvania, Genomics and Computational Biology Grad Group
Currently Postdoc, Stanford University

2016-2020 Xinyuan (Blair) Zhang
University of Pennsylvania, Genomics and Computational Biology Grad Group
Currently: Senior Statistical Geneticist at Regeneron Genetics Center

2018-2021 Joseph Park, MD-PhD student, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. Dan Rader
Currently: Resident, Weill Cornell School of Medicine

2018-2023 William Bone, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. Ben Voight
Currently: Scientist, Recursion

2018-2023 Pankhuri Singhal, Genetics Student in the Cellular and Molecular Biology program, University of Pennsylvania

2020-present Daniel Hui, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. Sarah Tishkoff

2020-present Brenda Zhao, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. Dokyoon Kim

2020-present John Crawford, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. Casey Greene

2021-present Karl Keat, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. Dokyoon Kim

2021-present Van Truong, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. John Wherry

2022-present Anni Moore, Genomics and Computational Biology Grad Group, University of Pennsylvania

2022-present Chris Jones, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. Sarah Tishkoff

2022-present David Zhang, MD-PhD student, Genomics and Computational Biology Grad Group, University of Pennsylvania, co-mentor with Dr. Dan Rader

2022-present Rachit Kumar, MD-PhD student, Genomics and Computational Biology Grad Group, University of Pennsylvania

Thesis Committees

2005-2006 Zheng (Roger) Liu
Microbiology & Immunology
Vanderbilt University School of Medicine

2005-2006 Tricia Thornton-Wells
Neuroscience Program
Vanderbilt University School of Medicine

2005-2007 Xueying (Sharon) Liang
Human Genetics Ph.D. Program
Vanderbilt University School of Medicine

2006-2008 Digna Velez
Human Genetics Ph.D. Program
Vanderbilt University School of Medicine

2007-2009 Daniel Kinnamon
Statistical Genetics
University of Miami Miller School of Medicine

2008-2009 Rebecca Zuvich
Human Genetics PhD Program
Vanderbilt University School of Medicine

2007-2010 Britney Grayson
Microbiology & Immunology
Vanderbilt University School of Medicine

2008-2010 Logan Dumitriscu
Human Genetics PhD Program
Vanderbilt University School of Medicine

2010-2012 Janina Jeff
Human Genetics PhD Program
Vanderbilt University School of Medicine

2013-2014 Matt Oetgens
Human Genetics PhD Program
Vanderbilt University School of Medicine

2013-2016 Jacob Hall
Human Genetics PhD Program
Vanderbilt University School of Medicine

2013-2017	Claire Reynolds Biochemistry, Microbiology and Molecular Biology PhD Program The Pennsylvania State University
2013-2015	Marta Byrska-Bishop Bioinformatics and Genomics PhD Program The Pennsylvania State University
2013-2016	Rohit Reja Bioinformatics and Genomics PhD Program The Pennsylvania State University
2018-2022	Laura Ritenour Cell and Molecular Biology (CAMB) PhD Program University of Pennsylvania, Perelman School of Medicine
2018-2021	David Lee Genomics and Computational Biology (GCB) PhD Program University of Pennsylvania, Perelman School of Medicine
2019-2021	Jennifer Shah Cell and Molecular Biology (CAMB) PhD Program University of Pennsylvania, Perelman School of Medicine
2019-2021	Victoria Arthur Epidemiology PhD Program University of Pennsylvania, Perelman School of Medicine
2020-2022	Benjamin Heil Genomics and Computational Biology (GCB) PhD Program University of Pennsylvania, Perelman School of Medicine
2020-2022	Alexa Woodward Epidemiology PhD program University of Pennsylvania, Perelman School of Medicine
2020-present	John Gregg Genomics and Computational Biology (GCB) PhD Program University of Pennsylvania, Perelman School of Medicine
2021-present	Jenea Adams Genomics and Computational Biology (GCB) PhD Program University of Pennsylvania, Perelman School of Medicine
2021-present	Kaylyn Clark Genomics and Computational Biology (GCB) PhD Program University of Pennsylvania, Perelman School of Medicine

2021-present Jacob Leiby
Genomics and Computational Biology (GCB) PhD Program
University of Pennsylvania, Perelman School of Medicine

2021-present Vivek Sriram
Genomics and Computational Biology (GCB) PhD Program
University of Pennsylvania, Perelman School of Medicine

2022-present Erica Suh
Genomics and Computational Biology (GCB) PhD Program
University of Pennsylvania, Perelman School of Medicine

2022-present Stacy Guzman
Biochemistry and Molecular Biology (BMB) PhD program
University of Pennsylvania, Perelman School of Medicine

MAJOR RESEARCH INTERESTS

Computational Genomics	Genetic Epidemiology
Translational Bioinformatics	Statistical Genetics
Epistasis	Systems Genomics
Pharmacogenomics	Computational Biology
Big Data	Biomedical Informatics
Evolutionary Computation	Cardiovascular Disease

SOFTWARE AND PATENTS

2001	Multifactor Dimensionality Reduction (MDR)
2003	MDR-PDT
2002	U.S. Provisional Patent: "Method and Apparatus for Multifactor Dimensionality Reduction"
2006	genomeSIMLA
2009	LD-Plus
2009	LD-Spline
2010	Synthesis-View
2012	ATHENA
2012	Biofilter 2.0
2012	BioBin
2012	PheWAS-view
2013	Phenogram
2016	iPhenogram

PODCASTS

The Biomedical Informatics Roundtable podcast, cohost
http://bmipodcast.org/

The CALM (Combining Academic and Life with Marylyn) podcast, host
https://marylynritchie.com/podcast

RESEARCH FUNDING

Active Funding

5R01 AI077505 (**MPI, Haas and Ritchie)** 04/01/2021-03/31/2026 1.2 calendar months
NIH/NIAID – Subcontract from Vanderbilt Total Award Amount (including Indirect Costs): $1,804,980
Pharmacogenomics of HIV Therapy
The goal of this project is to identify genetic variation associated with adverse reactions to HIV therapy, including PheWAS, epistasis, and gene-environment interaction analyses.

R01HL141232-01A1 (**MPI, Cappola and Musurunu**) 07/01/2019-06/30/2023 0.24 calendar months
NIH Total Award Amount (including Indirect Costs): $3,164,978
MTSS1 in Myocardial Disease
The goal of this project is to explore the relationship of MTSS1 to different types of heart failure, to establish that reducing MTSS1 activity is protective against heart failure in mouse models and human heart muscle cells, and to test a therapeutic strategy to reduce MTSS1 in the heart.

Team Science Grant (**PI, Nathanson**) 07/01/2019-6/30/2023 0.12 calendar months
Gray Foundation Total Award Amount (including Indirect Costs): $3,000,000
Determinants of immune activity and molecular features in BRCA 1 / 2 mutation carriers
We will build a biorepository of BRCA1/2 mutation-associated tumors, study the immune health of unaffected mutation carriers and correlate molecular features and immunogenicity in the tumors.

State of Pennsylvania Department of Health Nonformula Tobacco Settlement Act Grant:
Berrettini (PI) **Ritchie (CO-PI)** 6/1/2019 – 5/30/2023 0.12 calendar months
Total Award Amount (including Indirect Costs): $3,952,162
Pharmacogenetics of Opioid Use Disorder
The goal is to conduct genome-wide association studies of the efficacy of buprenorphine and methadone treatment for opioid use disorder.

1R01GM138597-01 (**PI, Kim**) 08/01/2020-07/31/2024 0.24 calendar months
NIH/NIGMS Total Award Amount (including Indirect Costs): $1,915,421
Unravelling genetic basis of comorbidity using EHR-linked biobank data
Inthis proposal,wewill construct a disease comorbidity map of 2.1 million patients using longitudinal EHRs in Penn Medicine (Aim 1), construct a disease-gene map derived from phenome-wide association study using Penn Medicine Biobank Participants (Aim 2), and developanovelscoringsystemusinggraph-basedmachine learningandpredictcomorbidityriskscores(CRS)foragivendisease(Aim3).

1U24HG010862-01A1 (**MPI, Klein and Ritchie**) 08/07/2020-05/31/2023 1.44 calendar months
NHGRI Total Award Amount (including Indirect Costs): $560,000
Pharmacogenomics Clinical Annotation Tool (PharmCAT)
The goal of this project is to build an annotation tool for all pharmacogenomic variants with clinical guidelines and to create a report with the clinical recommendations based on those variants.

R01 (**Subcontract with Mt. Sinai**) 09/01/20-08/31/23 0.12 calendar months
NIH Total Award Amount (including Indirect Costs): $292,380
Elucidating hereditary transthyretin-mediated heart failure risk using machine learning, polygenic risk and recall by genotype approaches in African ancestory individuals
The proposed work will allow us to determine whether an integrated, data-driven approach can improve identification of V122I carriers as well as lead to improved insights about the interplay between polygenic risk scores and V122I.

3R01RY023557-06 (**PI, O'Brien**) 09/01/2020-08/31/2025 1.2 calendar months
NIH/NEI Total Award Amount (including Indirect Costs): $6,645,134
Primary Open Angle African-American Glaucoma Genetics Study Renewal
This proposal will identify and investigate genetic variants of biologicimportance to thisdisease using genotypicand phenotypic data from the Primary Open-Angle African American Glaucoma Genetics study cohort. Our findingswill contribute to the development of more targeted screening and precise therapeutic interventions for this blinding disease.

1 R01 AG066833-01A1 (**MPI, Moore, Ritchie, Li**) 9/30/2021-08/31/2026 1.86 calendar months
NIH/NIA Total Award Amount (including Indirect Costs): $8,046,920
Artificial Intelligence Strategies for Alzheimer's Disease Research
The goal of this project is to develop artificial intelligence (AI) approaches for extracting unforeseen patterns from clinical, genetic, genomic, and imaging data that could lead to ideas for new drug development. Our proposed AI methods and software will beopen-source, user-friendly, and freely available for all to use.

1 P30 1P30AG073105-01 (**Ritchie**) 9/30/2021-08/31/2026 1.2 calendar months
NIH/NIA Total Award Amount (including Indirect Costs): $817,513
Networking and Mentoring Core of the Penn Artificial Intelligence and Technology (PennAITech) Collaboratory
Successful aging in the home can greatly improve quality of life, improve health outcomes, and reduce burden on the healthcare system. The goal of the overall project is to establish a national collaboratory for the development, evaluation, and implementation of artificial intelligence software and new technologies to facilitate health aging in the home. The Networking and Mentoring Core (Core E) will organize networking, mentoring, and dissemination opportunities for the awardees of the pilot programs.

1 R01HG010067 (**Ritchie, PI subcontract**) 06/12/22 – 04/30/23 0.6 calendar months
NIH/NHGRI Total Award Amount (including Indirect Costs): $1,396,502
Network-based algorithms for target identification and drug repositioning from genetic associations
The major goal of this application is to develop and evaluate algorithms that use tissue-specific functional relationship networks to identify new gene targets and to suggest opportunities for drug repositioning.

2-UL1-TR-001878-06 (**Ritchie, PI Informatics core**) 06/30/2021-05/31/2026 1.2 calendar months
NIH/NCATS Total Award Amount (including Indirect Costs): $12,720,728
Institutional Clinical and Translational Sciences Award
To provide translational informatics technologies and a knowledge base for more effectively conducting clinical and translational science.

U01-HL160277 **(Chirinos)** 9/10/2021 - 7/31/2026 0.18 calendar months
NIH/NHLBI Total Award Amount (including Indirect Costs): $1,354,700
HeartShare: Next-Generation Phenomics to Define Heart Failure Subtypes and Treatment Targets – Clinical Centers

The goal of the current proposal is to establish a large cohort of deeply phenotyped patients with HF to help with better understanding differential mechanisms of adverse outcomes in patients with HFpEF and identifying subgroups of patients that are likely to benefit from targeted therapy.

R01 HG012670-01 (**Ritchie, Nathanson, Schnoll, MPIs**) 07/01/2022- 06/30/2027 1.8 calendar months
NIH/NHGRI Total Award Amount (including Indirect Costs): $4,379,282
Using Behavioral Economics and Implementation Science to Advance the Use of Genomic Medicine Utilizing an EHR Infrastructure across a Diverse Health System
We propose a highly innovative study using implementation science to study nudges in the electronic health record (EHR) to influence both clinicians and patients to increase the use of genetic testing in clinical care, building upon the infrastructure that we have already built to deliver genomic medicine in the EHR

Completed Funding

2004-2005	Development award supported by CFAR - P30 AI54999 (PI) Genetic Basis of NNRTI Toxicity in HIV Patients
2004-2005	Development award supported by CFAR - P30 AI54999 (Co-PI) A Pilot Study of Mitochondrial Genetics and Peripheral Neuropathy during Antiretroviral Therapy
2004-2008	NIH R01 AG020135 (PI of subcontract) Revealing Epistasis in Alzheimer Disease
2005-2007	Development award supported by SPORE - P50 CA098131 (PI) Approaches to Genome-Wide Association in Sporadic Breast Cancer
2006-2009	AVCF- Vanderbilt (Co-investigator) DNA Capture Project – Dan Roden 9/28/2007-7/31/2011
2006-2009	NIH R21 HL0877226 (Co-investigator) Iron as a Nutritional Modifier of Toxic Neuropathy in HIV/AIDS – Asha Kallianpur 9/27/06-7/31/09
2007-2009	NIH R01 CA122756 (Co-investigator) Cell Cycle/Apoptosis Gene Variants and Breast Cancer – Jiyang Cai 12/1/2007-11/30/2011
2007-2009	NIH/NIA R01 AG027944 (Co-investigator) Genomic Convergence in Alzheimer Disease – Pericak-Vance, Univ. of Miami 12/01/2007-11/30/2012
2008-2009	NCCAM R01 AT004660 (Co-investigator) Dietary Calcium and Magnesium, Genetics and Colorectal Adenoma – Qi Dai 06/01/2008-05/31/2012
2006-2009	NIH R21 NS059330 (Co-investigator) Mitochondrial Genomics and Peripheral Neuropathy during HIV Therapy – Todd

	Hulgan
	3/15/2007-6/30/2010
2004-2010	NIH U01 HL065962 (PI of core)
	Pharmacogenomics of Arrhythmia Therapy – Dan Roden
	9/01/2005-8/31/2010
2007-2011	NIH U01 HG004603-01 (Co-investigator)
	Vanderbilt Genome-Electronic Records Project – Dan Roden
	9/28/2007-7/31/2011
2007-2011	NIH U01 HG004608 (PI of subcontract)
	Genome-Wide Study of Cataract and Low HDL in the Personalized Medicine Research Project – Catherine McCarty
	9/30/2007-9/29/2011
2009-2013	NIH/NLM 1R01LM009989-01A1 (Malin)
	Technologies to Enable Privacy in Biomedical Databanks
	07/01/2009-06/30/2013
2009-2011	NIH/NIGMS 1RC2GM092618-01 (Masys)
	VESPA: Vanderbilt Electronic Systems for Pharmacogenomic Assessment
	09/30/2009-09/29/2011
2010-2015	NIH U01 HL065962 (PI of core)
	Pharmacogenomics of Arrhythmia Therapy – Dan Roden
	9/01/2010-8/31/2015
2009-2013	NIH/NLM 1R01 LM010040-01(PI)
	Analysis Tool for Heritable and Environmental Network Associations
	07/01/2009-06/30/2013
2008-2013	NIH/NIAID 5R01 AI077505-03 (Haas)
	Pharmacogenomics of HIV Therapy
	07/08/2008-06/30/2013
2009-2013	NIH/NHGRI 5U01 HG004798-03
	Epidemiologic Architecture for Genes Linked to Environment (Crawford)
	07/17/2008-12/31/2013
2011-2015	NIH/NHGRI 5U01 HG006385 (Haines, Ritchie PI subcontract)
	eMERGE II Coordinating Center
	08/15/2011-07/31/2015
2013-2014	Geisinger contract (Ritchie PI)
	Geisinger Clinic Genetic Studies: Mining Electronic Health Records for Genetic Associations
	8/01/2013-7/31/2014
2010-2016	5U01 HL065962 (Roden, Ritchie PI U19-subcontract)
	Pharmacogenomics of Arrhythmia Therapy: P-STAR Network Resource
	09/01/2010-08/31/2016
2011-2016	5U01 HG006389 (McCarty, Ritchie PI subcontract)
	IRIS: Incorporating Research into Sight: eMERGE II
	08/15/2011-07/31/2016

2011-2016	3 UL1 RR033184-01 (Sinoway) CTSA (Clinical and Translational Science Award) 3/1/2011-2/29/2016
2015-2016	R01GM111913 (Mirshahi and Robishaw) An integrated approach to study GPCR variants associated with complex diseases. 06/01/2015-8/30/2016
2016-2017	1OT2OD024609-01 (Ledbetter and Ritchie) Precision Medicine at Geisinger 09/27/2016 – 05/31/2017
2015-2017	1 U01 HG008679-01 (Williams and Ritchie) EMR-Linked Biobank for Translational Genomics (part of eMERGE-III network) 09/01/2015-12/18/2017
2015-2017	SAP 4100070267 (Ritchie) (Pennsylvania Department of Health) Integrating Big Data for Biomedical Discovery: Methods, Tools, and Applications 6/1/2015-12/18/2017
2013-2017	2 P01 ES011269-11 (Van de Water, Ritchie sub) EPA: 83543201 (Van de Water, Ritchie sub) UC. Davis Center for Children's Environmental Health and Disease Prevention (CCEH) 06/01/2013 – 12/18/2017
2018-2019	P50GM115318-04S1 (Krauss, Ritchie PI Informatics Core) Pharmacogenomics of Statin Therapy (Informatics Core) – administrative supplement $245,954 09/01/2018-08/31/2019
2015-2021	P50GM115318-01 (Krauss, Ritchie PI Informatics Core) Pharmacogenomics of Statin Therapy (Informatics Core) $150,000 07/01/2015-08/30/2021
2015-2021	5R01 AI077505 (MPI, Haas and Ritchie) Pharmacogenomics of HIV Therapy $243,000 07/01/2015-12/31/2021
2016-2021	R01AI116794-01A1 (Moore) Biomedical Computing and Informatics Strategies for Infectious Disease Research $343,833 04/01/16 – 03/31/21

BIBLIOGRAPHY

Peer-Reviewed Research Papers

1. **Ritchie MD**, Hahn LW, Roodi N, Bailey LR, Dupont WD, Plummer WD, Parl FF, Moore JH. Multifactor dimensionality reduction reveals high-order interactions among estrogen metabolism genes in sporadic Breast Cancer. *American Journal of Human Genetics,* 69:138-147 (2001). PMID: 11404819, PMCID: PMC1226028
2. Moore JH, Hahn LW, **Ritchie MD**, Thornton TA, White BC. Application of genetic algorithms to the discovery of complex genetic models for simulation studies in human genetics. In: Langdon WB, Cantu-Paz E, Mathias K, Roy R, Davis D, Poli R, Balakrishnan K, Honavar V, Rudolph G, Wegener J, Bull L, Potter MA, Schultz AC, Miller JF, Burke E, and Jonoska N, eds.

Proceedings of the Genetic and Evolutionary Computation Conference, *Morgan Kaufmann Publishers*, San Francisco, 1150-55 (2002). PMID: 23413413, PMCID: PMC3569849

3. Donaldson JC, Dise RS, **Ritchie MD**, Hanks SK. Nephrocystin conserved domains involved in targeting to epithelial cell-cell junctions, interaction with filamins, and establishing cell polarity. *Journal of Biological Chemistry*, 277, 29028-29035 (2002). PMID: 12006559
4. Hahn LW, **Ritchie MD**, Moore JH. Multifactor dimensionality reduction software for detecting gene-gene and gene-environment interactions. *Bioinformatics*, 19, 376-382 (2003). PMID: 12584123
5. **Ritchie MD**, Hahn LW, Moore JH. Power of Multifactor Dimensionality Reduction for detecting gene-gene and gene-environment interactions in the presence of genotyping error, missing data, phenocopy, and genetic heterogeneity. *Genetic Epidemiology*, 24, 150-157 (2003). PMID: 12548676
6. **Ritchie MD**, White B, Parker JS, Hahn LW, and Moore JH. Optimization of neural network architecture improves the power to identify gene-gene interaction in common diseases. *BMC Bioinformatics*, 4:28 (2003). PMID: 12846935, PMCID: PMC183838
7. Moore JH, Thornton TA, **Ritchie MD**. Basic statistics. *Curr Protoc Hum Genet*. 2003 Aug; Appendix 3: Appendix 3M. doi: 10.1002/0471142905.hga03ms37. PMID: 18428338
8. Moore JH, Hahn LW, Ritchie MD, Thornton TA, White BC. Routine Discovery of Complex Genetic Models using Genetic Algorithms. *Appl Soft Comput.* 2004 Feb 1;4(1):79–86. PMID: 20948983, PMCID: PMC2952957
9. Cho YM, **Ritchie MD**, Moore JH, Moon MK, Lee YY, Yoon KH, Sung YA, Lang HC, Park JY, Lee KU, Shin HD, Kim SY, Lee HK, Park KS. Multifactor Dimensionality Reduction reveals a two-locus interaction associated with Type 2 Diabetes Mellitus. *Diabetologia*, 47:549-554 (2004). PMID: 14730379
10. Tsai CT, Lai LP, Chiang FT, Fallin D, Hwang JJ, **Ritchie MD**, Moore JH, Hsu KL, Tseng CD, Liau CS, Lin JL, Tseng YZ. Renin-angiotensin system gene polymorphisms and Atrial Fibrillation. *Circulation*, 109:1640-1646 (2004). PMID: 15023884
11. Williams SM, **Ritchie MD**, Phillips JA, Wong LJ, Felder RA, Jose PA, Moore JH. Multilocus analysis of hypertension: a hierarchical approach. *Human Heredity*, 57:28-38 (2004). PMID: 15133310
12. Coffey CS, Hebert PR, **Ritchie MD**, Krumholz HM, Morgan TM, Gaziano JM, Ridker PM, Moore JH. An application of conditional logistic regression and Multifactor Dimensionality Reduction for detecting gene-gene interactions on risk of Myocardial Infarction: the importance of model validation. *BMC Bioinformatics*, 5:49 (2004). PMID: 15119966, PMCID: PMC419697
13. **Ritchie MD**, Coffey CS, Moore JH. Genetic programming neural networks: a bioinformatics tool for human genetics. *Lecture Notes in Computer Science*, 3102: 438-448 (2004). PMID: 20948988, PMCID: PMC2952963
14. Shin N, Dise RS, Schneider-Mergener J, **Ritchie MD**, Kilkenny DM, Hanks SK. Subsets of the major tyrosine phosphorylation sites in Crk-associated substrate (CAS) are sufficient to promote cell migration. *Journal of Biological Chemistry*, 279(37): 38331-38337 (2004). PMID: 15247284
15. Vanhoose AM, **Ritchie MD**, Winder DG. Regulation of cAMP levels in area CA1 of hippocampus by Gi/o-coupled receptors is stimulus dependent in mice. *Neuroscience Letters*, 370(1):80-3 (2004). PMID: 15489022
16. Soares ML, Coelho T, Sousa A, **Ritchie MD**, Williams SM, Batalov S, Conceição I, de Lurdes M, Luís S, Saraiva MJ, Buxbaum JN. Susceptibility and modifier genes in familial amyloid polyneuropathy type I. *Human Molecular Genetics*, 14:543-53 (2005). PMid: 15649951
17. Bush WS, Motsinger AA, Dudek, SM, **Ritchie MD**. Can neural network constraints in GP provide power to detect genes associated with human disease? *Lecture Notes in Computer Science*, 3449:44-53 (2005). PMid: 20419044, PMCID: PMC2858417

18. Hulgan T, Haas DW, Haines JL, **Ritchie MD**, Robbins GK, Shafer RW, Clifford DB, Kallianpur A, Summar M, Canter JA. Mitochondrial haplogroups and peripheral neuropathy during antiretroviral therapy: an adult AIDS clinical trials group study. *AIDS*, 19(13):1341-1349 (2005). PMID: 16103764

19. Ma DQ, Whitehead PL, Menold MM, Martin ER, Ashley-Koch AE, Mei H, **Ritchie MD**, DeLong GR, Abramson RK, Wright HH, Cuccaro ML, Hussman JP, Gilbert JR, Pericak-Vance MA. Identification of significant association and gene-gene interaction on GABA receptor subunit genes in Autism. *American Journal of Human Genetics*, 77(3):377-88 (2005). PMID: 16080114, PMCID: PMC1226204

20. Martin ER, **Ritchie MD**, Kang S, Hahn L, Moore JH. A novel method to identify potential interactions in nuclear families: The MDR-PDT. *Genetic Epidemiology*, 30:111-23 (2006). PMID: 16374833

21. Takahashi H, Wilkinson GR, PhD, Nutescu EA, Morita T, **Ritchie MD,** Scordo MG, Pengo V, Barban M, Padrini R, Ieiri I, Otsubo K, Kashima T, Kimura S, Kijima S, Echizen H. Different contributions of polymorphisms in *VKORC1* and *CYP2C9* to intra- and inter-population differences in maintenance dose of warfarin in Japanese, Caucasians and African Americans. *Pharmacogenetics and Genomics*, 16:101-110 (2006). PMID: 16424822

22. Ashley-Koch A, Mei H, Jaworski J, Ma D, **Ritchie MD**, Menold M, Abramson R, Wright H, Cuccaro M, Gilbert J, Martin E, Pericak-Vance M. An analysis paradigm for investigating multi-locus effects in complex disease: examination of three GABA receptor subunit genes on 15q11-q13 as risk factors for Autistic disorder. *Annals of Human Genetics*, 70:281-92 (2006). PMID: 16674551

23. Motsinger AA, Lee SL, Mellick G, **Ritchie MD**. GPNN: power studies and applications of a neural network method for detecting gene-gene interactions in studies of human disease. *BMC Bioinformatics*, 7:39 (2006). PMID: 16436204, PMCID: PMC1388239

24. Motsinger AA, Donahue BS, Brown NJ, Roden DM, **Ritchie MD**. Risk factor interactions and genetic effects associated with post-operative Atrial Fibrillation. *Pacific Symposium on Biocomputing Proceedings*, 11:584-595 (2006). PMID: 17094271

25. Dudek SM, Motsinger AA, Velez DR, Williams SM, **Ritchie MD**. Data simulation software for whole-genome association studies and other studies in human genetics. *Pacific Symposium on Biocomputing Proceedings*, 11:499-510 (2006). PMID: 17094264

26. Sanda H, Yatabe J, Midorikawa S, Hashimoto S, Watanabe T, Moore JH, **Ritchie MD**, Williams SM, PExzullo JC, Eisner GM, Jose PA, Felder RA. Salt sensitivity and G-protein-coupled receptor kinase type 4 polymorphisms. Single-nucleotide polymorphisms for diagnosis of salt-sensitive hypertension. *Clinical Chemistry,* 52:352-360 (2006). PMID: 16439609

27. Motsinger AA, **Ritchie MD**. Multifactor Dimensionality Reduction: an analysis strategy for modeling and detecting gene-gene interactions in human genetics and pharmacogenomics studies. *Human Genomics*, 2:318-328. (2006). PMID: 16595076, PMCID: PMC3500181

28. Brassat D, Motsinger AA, Walker K, Steiner LL, Erlich HA, Barcellos L, Pericak-Vance MA, Hauser SL, Haines JL, Oksenberg JR, **Ritchie MD**. Multifactor dimensionality reduction reveals gene-gene interactions associated with Multiple Sclerosis in African Americans. *Genes and Immunity*, 7:310-315 (2006). PMID: 16625214, PMCID: PMC4339061

29. Motsinger AA, Dudek SM, Hahn LW, **Ritchie MD**. Comparison of neural network optimization approaches for studies of human genetics. *Lecture Notes in Computer Science*, 3907:103-114 (2006). PMID: 20191104, PMCID: PMC2828685

30. Motsinger AA, **Ritchie MD**, Dobrin SE. Clinical applications of whole-genome association studies (WGAS): future applications at the bedside. *Expert Reviews in Molecular Diagnostics*, 6:551-65 (2006). PMID: 16824029

31. Motsinger AA, Hahn LW, Dudek SM, Ryckman KK, **Ritchie MD**. Alternative cross-over strategies and selection techniques for grammatical evolution optimized neural networks.

Proceedings of Genetic and Evolutionary Computation Conference, Association for Machine Learning Press, New York, 947-949 (2006). PMID: 20634918, PMCID: PM2903763

32. Kallianpur AR, Hulgan T, Canter JA, **Ritchie MD**, Haines JL, Robbins GK, Shafer RW, Clifford DB, Haas DW. Hemochromatosis (HFE) gene mutations and peripheral neuropathy during antiretroviral therapy: an AIDS clinical trials group study. *AIDS*, 20:1503-1513 (2006). PMID: 16847405

33. Motsinger AA, **Ritchie MD**. The effect of reduction in cross-validation intervals on the performance of multifactor dimensionality reduction. *Genetic Epidemiology*, 30: 546-555 (2006). PMID: 16800004

34. Haas DW, Geraghty DE, Andersen J, Mar J, Motsinger AA, D'Aquila RT, Unutmaz D, Benson CA, **Ritchie MD**, Landay A. Immunogenetics of CD4 lymphocyte recovery during antiretroviral therapy: an AIDS clinical trials group study. *Journal of Infectious Disease*, 198:1098-1107 (2006). PMID: 16991084

35. Motsinger AA, **Ritchie MD,** Shafer RW, Robbins GK, Morse GD, Labbé L, Wilkinson GR, Clifford DB, D'Aquila RT, Johnson VA, De Gruttola V, Pollard RB, Merigan TC, Hirsch MS, Donahue JP, Kim RB, Haas DW. Multilocus gene-gene interactions and response to efavirenz-containing regimens: an adult AIDS clinical trials group study. *Pharmacogenetics and Genomics*, 16:837-845 (2006). PMID: 17047492

36. **Ritchie MD**, Haas DW, Motsinger AA, Donahue JP, Erdem H, Raffanti S, Rebiero P, George AL, Kim RB, Haines JL, Sterling TR. Drug transporter and metabolizing enzyme gene variants and non-nucleoside reverse transcriptase inhibitor hepatotoxicity. *Clinical Infectious Diseases*, 43:779-82 (2006). PMID: 16912956

37. Bush WS, Dudek SM, **Ritchie MD**. Parallel Multifactor Dimensionality Reduction: a tool for the large scale analysis of gene-gene interactions. *Bioinformatics*, 22:2173-4 (2006). PMID: 16809395, PMCID: PMC4939609

38. Crooke PS, **Ritchie MD,** Hachey DL, Dawling S, Roodi N, Parl FF. Estrogens, enzyme variants, and Breast Cancer. A risk model. *Cancer, Epidemiology, Biomarkers, and Prevention*, 15:1620-9 (2006). PMID: 16985022

39. Tsai CT, Hwang JJ, **Ritchie MD**, Moore JH, Fallin D, Chiang FT, Lai LP, Hwang JJ, Hsu KL, Tseng CD, Liau CS, Lin JL, Tseng YZ. Renin-angiotensin system gene polymorphisms and Coronary Artery disease in a large angiographic cohort: detection of high-order gene-gene interaction. *Atherosclerosis*, 195(1):172-80 (2007). PMID: 17118372

40. Motsinger AA, Reif DM, Dudek SM, **Ritchie MD.** Understanding the Evolutionary Process of Grammatical Evolution Neural Networks for Feature Selection in Genetic Epidemiology. *IEEE Symposium on Computational Intelligence in Bioinformatics and Computational Biology*, 1-8 (2006). PMID: 20634919, PMCID: PMC2903766

41. Motsinger AA ,Brassat D, Caillier SC, Erlich HA, Walker K, Steiner LL, Barcellos L, Pericak-Vance MA, Schmidt S, Gregory S, Hauser SL, Haines JL, Oksenberg JR, **Ritchie MD**. Complex gene-gene interactions in Multiple Sclerosis: a multi-factorial approach reveals associations with inflammatory genes. *Neurogenetics*, 8:11-20 (2007). PMID: 17024427

42. Nordgard SH, **Ritchie MD**, Motsinger AA, Lemmon G, Fjelstad S, Alnæs GIG, Jensrud SD, Berg M, Geisler S, Moore JH, Lønning PE, Børresen-Dale AL, Kristensen VN. ABCB1 and GST polymorphisms associated with TP53 status in breast cancer. *Pharmacogenetics and Genomics*, 17:127-36 (2007). PMID: 17301692, PMCID: PMC2954594

43. Canter JA, Summar ML, Smith HB, **Ritchie MD**, Motsinger AA, Christian K, Drinkwater D, Dyer KL, Kavanaugh A, Barr FE. Genetic variation in the mitochondrial enzyme carbamyl-phosphate synthetase I predisposes children to increased pulmonary artery pressure following surgical repair of congenital heart defects: a validated genetic association study. *Mitochondrion*, 7:204-210 (2007). PMID: 17188582, PMCID: PMC1929167

44. Velez DR, White BC, Motsinger AA, Bush WS, **Ritchie MD**, Williams SM, Moore JH. A balanced accuracy metric for epistasis modeling in imbalanced datasets using Multifactor Dimensionality Reduction. *Genetic Epidemiology*, 31:306-315 (2007). PMID: 17323372

45. Motsinger AA, Reif DM, Fanelli TJ, Davis AC, **Ritchie MD**. Linkage disequilibrium in genetic association studies improves the performance of grammatical evolution neural networks. *IEEE Symposium on Computational Intelligence in Bioinformatics and Computational Biology*, 1-8 (2007). PMID: 21572972, PMCID: PMC3092290, MIHMS 160137

46. Bush WS, Thornton-Wells TA, **Ritchie MD**. Association rule discovery has the ability to model complex genetic effects. *Proceedings of the 2007 IEEE Symposium on Computational Intelligence and Data Mining*, 624-629 (2007). PMID: 20953276, PMCID: PMC2954594

47. **Ritchie MD**, Motsinger AA, Bush WS, Coffey CS, Moore JH. Genetic programming neural networks: a powerful bioinformatics tool for human genetics. *Applied Soft Computing*, 7:471-479 (2007). PMID: 20948988, PMCID: PMC2952963

48. Ashley-Koch AE, Jaworski J, Ma D, Mei H, **Ritchie MD**, Skaar DA, Delong GR, Abramson RK, Wright HH, Cuccaro ML, Gilbert JR, Martin ER, Pericak-Vance MA. Investigation of potential gene-gene interactions between APOE and RELN contributing to Autism risk. *Psychiatric Genetics*, 17:221-226 (2007). PMID: 17621165, PMCID: PMC2749242

49. Darbar D, Motsinger AA, **Ritchie MD**, Gainer JV, Roden DM. Polymorphism modulates symptomatic response to antiarrhythmic drug therapy in patients with lone Atrial Fibrillation. *Heart Rhythm*, 4:743-749 (2007). PMID: 17556195, PMCID: PMC1948880

50. **Ritchie MD**, Bartlett J, Bush WS, Edwards TL, Motsinger AA, Torstenson ES. Exploring epistasis in candidate genes for Rheumatoid Arthritis. *BMC Proceedings*, 1 Suppl 1:S70 (2007). PMID: 18466572, PMCID: PMC2367541

51. Bartlett CW, Vieland VJ, Bartlett J, Bell JT, Bhattacharjee S, Clerget-Darpoux F, Bush WS, Edwards TL, Gao G, Halder I, Huang Y, Kotti S, Larkin EK, Li H, Motsinger AA, Mukhopadhyay N, Namkung J, Park T, **Ritchie MD**, Stein CM, Zhou JY. Discussing gene-gene interaction: warning--translating equations to English may result in jabberwocky. *Genetic Epidemiology*, 31 Suppl 1:S61-7 (2007). PMID: 18046759, PMCID: PMC2749242

52. Pretorius MM, Gainer JV, Van Guilder GP, Coelho EB, Luther JM, Fong P, Rosenbaum DD, Malave HA, Yu C, **Ritchie MD**, Vaughan DE, Brown NJ. The Bradykinin type 2 receptor BE1 polymorphism and ethnicity influence systolic blood pressure and vascular resistance. *Clinical Pharmacology and Therapeutics,* 83(1):122-9 (2008). PMID: 17522594, PMCID: PMC1948880

53. Canter JA, Haas DW, Kallianpur AR, **Ritchie MD**, Robbins GK, Shafer RW, Murdock DG, Hulgan T. The mitochondrial pharmacogenomics of haplogroup T: MTND2(*)LHON4917G and antiretroviral therapy-associated peripheral neuropathy. *Pharmacogenomics Journal*, 8(1):71-7 (2008). PMID: 17684475

54. Schwartz UI, **Ritchie MD**, Bradford Y, Dudek S, Frye-Anderson A, Kim RB, Roden DM, Stein CM. Genetic determinants of response to warfarin during initial anticoagulation. *NEJM*, 358(10):999-1008 (2008). doi: 10.1056/NEJMoa0708078. PMID: 18322281, PMCID: PMC3894627

55. Edwards TL, Bush WS, Turner SD, Dudek SM, Torstenson ES, Schmidt M, Martin E, **Ritchie MD**. Generating linkage disequilibrium patterns in data simulations using genomeSIMLA. *Lecture Notes in Computer Science,* 4973: 24-35 (2008). PMID: 19779634, PMCID: PMC749242

56. Darbar D, Kimbrough J, Jawaid A, McCray R, **Ritchie MD**, Roden DM. Persistent atrial fibrillation is associated with reduced risk of torsades de pointes in patients with drug-induced long QT syndrome. *J Am Coll Cardiol*, 51(8):836-42 (2008). PMID: 18294569, PMCID: PMC2271078

57. Motsinger-Reif AA, Dudek SM, Hahn LW, **Ritchie MD**. Comparison of approaches for machine learning optimization of neural networks for detecting gene-gene interactions in genetic

epidemiology. *Genetic Epidemiology*, 32(4):325-40 (2008). doi: 10.1002/gepi.20307. PMID: 18265411

58. Motsinger-Reif AA, Reif DM, Fanelli TJ, **Ritchie MD**. A comparison of analytical methods for genetic association studies. *Genetic Epidemiology*, 32(8):767-78 (2008). PMID: 18561203, PMCID: PMC2459220

59. Bush WS, Edwards TL, Dudek SM, McKinney BA, **Ritchie MD**. Alternative contingency table measures improves the power and detection of Multifactor Dimensionality Reduction. *BMC Bioinformatics*, 9:238 (2008). PMID: 18485205, PMCID: PMC2412877

60. Cooper GM, Johnson JA, Langaee TY, Feng H, Stanaway IB, Schwartz U, **Ritchie MD**, Stein CM, Roden DM, Smith JD, Veenstra DL, Rettie AE, Rieder MJ. A Genome-Wide Scan for Common Genetic Variants with a Large Influence on Warfarin Maintenance Dose. *Blood*, 112:1022-7 (2008). PMID: 18535201, PMCID: PMC2515139

61. Edwards TL, Lewis KG, Velez DR, Dudek SM, **Ritchie MD**. Exploring the Performance of Multifactor Dimensionality Reduction in Large Scale SNP Studies and in the Presence of Genetic Heterogeneity among Epistatic Disease Models. *Human Heredity*, 67(3):183-92 (2008). PMID: 19077437, PMCID: PMC3078287

62. Motsinger-Reif AA, Fanelli TJ, Davis AC, **Ritchie MD**. Power of grammatical evolution neural networks to detect gene-gene interactions in the presence of error. *BMC Research Notes*, 1:65 (2008). PMID: 18710518, PMCID: PMC2531119

63. Edwards TL, Wang X, Chen Q, Wormly B, Riley B, O'Neill FA, Walsh D, **Ritchie MD**, Kendler KS, Chen X. Interaction between interleukin 3 and dystrobrevin-binding protein 1 in schizophrenia. *Schizophrenia Research,* 106(2-3); 208-17 (2008). PMID: 18804346, PMCID: PMC2746913

64. Li C, Schwartz UI, **Ritchie MD**, Roden DM, Stein CM, Kurnik D. Relative contribution of *CYP2C9* and *VKORC1* genotypes and early INR response to the prediction of warfarin sensitivity during initiation of therapy. *Blood*, 113(17):3925-30 (2009). doi: 10.1182/blood-2008-09-176859. PMID: 19074728, PMCID: PMC2673121

65. Edwards TL, Pericak-Vance M, Gilbert JR, Haines JL, Martin ER, **Ritchie MD**. An association analysis of Alzheimer disease candidate genes detects an ancestral risk haplotype clade in ACE and putative multilocus association between ACE, A2M, and LRRTM3. *American Journal of Medical Genetics Part B, Neuropsychiatric Genetics*, 67(3):183-92 (2009). doi: 10.1002/ajmg.b.30899. PMID: 19105203, PMCID: PMC2821734

66. Hardison NE, Fanelli TJ, Dudek SM, Reif DM, **Ritchie MD**, Motsinger AA. A balanced accuracy fitness function leads to robust analysis using grammatical evolution neural networks in the case of class imbalance. *GECCO*, 353-4 (2008). PMID: 21197143, PMCID: PMC3011228

67. Bush WS, Dudek SM, **Ritchie MD**. Biofilter: a knowledge-integration system for the multi-locus analysis of genome-wide association studies. *Pacific Symposium on Biocomputing*, 368-79 (2009). PMID: 19209715, PMCID: PMC2859610

68. Body SC, Collard CD, Shernan SK, Fox AA, Liu KY, **Ritchie MD**, Perry TE, Muehlschlegel JD, Aranki S, Donahue BS, Pretorius M, Estrada J, Ellinor PT, Newton-Cheh C, Seidman CE, Seidman, Hermann DS, Lichtner P, Meitinger T, Pfeufer A, Kääb S, Brown NJ, Roden DM, Darbar D. Variation in the 4q25 Chromosomal Locus Predicts Atrial Fibrillation after Coronary Artery Bypass Graft Surgery. *Circulation Cardiovascular Genetics*, 2(5):499-506 (2009). doi: 10.1161/CIRCGENETICS.109.849075. PMID: 20031626, PMCID: PMC2801871

69. **International Warfarin Pharmacogenetics Consortium**, Klein TE, Altman RB, Eriksson N, Gage BF, Kimmel SE, Lee MT, Limdi NA, Page D, Roden DM, Wagner MJ, Caldwell MD, Johnson JA. Estimation of the warfarin dose with clinical and pharmacogenetic data. *NEJM*, 360(8):753-64 (2009). PMID: 19228618, PMCID: PMC2722908

70. Bush WS, Crawford DC, Alexander C, George A, Roden DM, **Ritchie MD**. Genetic variation in the rhythmonome: ethnic variation and haplotype structure in candidate genes for arrhythmias.

Pharmacogenomics, 10(6):1043-53 (2009). doi: 10.2217/pgs.09.67. PMID: 19530973, PMCID: PMC2746955

71. Turner SD, **Ritchie MD**, Bush WS. Conquering the Needle-in-a-Haystack: How Correlated Input Variables Beneficially Alter the Fitness Landscape for Neural Networks. *Lect Notes Comput Sci*, 5483:80-91 (2009). PMID: 19578549, PMCID: PMC2704607

72. Turner SD, Crawford DC, **Ritchie MD**. Methods for optimizing statistical analyses in pharmacogenomics research. *Expert Rev. Clin. Pharmacol.* 2(5), 559-570 (2009). PMID: 20221410, PMCID: PMC2835152

73. **Ritchie MD**. Using Prior Knowledge for Genome-Wide Association Studies in Multiple Sclerosis: a model for identifying pathways important for common, complex disease. *Genome Medicine*, 1(6):65 (2009). doi: 10.1186/gm65. PMID: 19566917, PMCID: PMC2703874

74. Edwards TL, Torstenson ES, Martin EM, **Ritchie MD**. A cross-validation procedure for general pedigrees and matched odds ratio fitness metric implemented for the multifactor dimensionality reduction pedigree disequilibrium test MDR-PDT and cross-validation: power studies. *Genetic Epidemiology*, 34(2):194-9 (2010). doi: 10.1002/gepi.20447. PMID: 19697353, PMCID: PMC2811750

75. Canter JA, Robbins GK, Selph D, Clifford DB, Kallianpur AR, Shafer R, Levy S, Murdock DG, **Ritchie MD**, Haas DW, Hulgan T, for the AIDS Clinical Trials Group Study 384 and New York Concept Sheet 273 Teams. African Mitochondrial DNA Subhaplogroups and Peripheral Neuropathy during Antiretroviral Therapy. *The Journal of Infectious Diseases*, 201(11):1703-7 (2010). doi: 10.1086/652419. PMID: 20402593, PMCID: PMC2862090

76. Bush WS, Chen G, Torstenson ES, **Ritchie MD**. LD-Spline: Mapping SNPs on genotyping platforms to genomic regions using patterns of linkage disequilibrium. *BMC BioData Mining,* 2(1):7 (2009). doi: 10.1186/1756-0381-2-7. PMID: 19954552, PMCID: PMC2795743

77. Grady BJ, Torstenson ES, Dudek SM, Giles J, Sexton D, **Ritchie MD**. Finding unique filter sets in PLATO: a precursor to efficient interaction analysis in GWAS data. *Pacific Symposium on Biocomputing*, 315-26 (2010). PMID: 19908384, PMCID: PMC2903053

78. Bush WS, Dudek SM, Torstenson ES, **Ritchie MD**. Visualizing SNP statistics in the context of linkage disequilibrium using LD-Plus. *Bioinformatics*, 26(4):578-9 (2010). doi: 10.1093/bioinformatics/btp678. PMID: 20130027, PMCID: PMC2820673

79. Edwards TL, Turner SD, Torstenson ES, Dudek SM, Martin EM, **Ritchie MD**. A general framework for formal tests of interaction after exhaustive search methods with applications to MDR and MDR-PDT. *PLOS ONE,* 5(2):e9363 (2010). doi: 10.1371/journal.pone.0009363. PMID: 20186329, PMCID: PMC2826406

80. Limdi NA, Wadelius M, Cavallari L, Eriksson N, Crawford DC, Lee MT, Chen CH, Motsinger-Reif A, Sagreiya H, Liu N, Wu AH, Gage BF, Jorgensen A, Pirmohamed M, Shin JG, Suarez-Kurtz G, Kimmel SE, Johnson JA, Klein TE, Wagner MJ; **International Warfarin Pharmacogenetics Consortium**. Warfarin pharmacogenetics: a single VKORC1 polymorphism is predictive of dose across 3 racial groups. *Blood*. 115(18):3827-34 (2010). doi: 10.1182/blood-2009-12-255992. PMID: 20203262, PMCID: PMC2865873

81. Turner SD, Dudek SM, **Ritchie MD**. Grammatical Evolution of Neural Networks for Discovering Epistasis among Quantitative Trait Loci. *Lect Notes Comput Sci*. 6023:86-97. (2010). PMID: 20526438, PMCID: PMC2880550

82. Denny JC, **Ritchie MD**, Basford M, Pulley J, Bastarache L, Brown-Gentry K, Wang D, Masys DR, Roden DM, Crawford DC. PheWAS: Demonstrating the feasibility of a phenome-wide scan to discover gene-disease associations. *Bioinformatics*, 26(9):1205-10 (2010). doi: 10.1093/bioinformatics/btq126. PMID: 20335276, PMCID: PMC2859132

83. Zabaleta J, Piazuelo MB, Camargo MC, Correa P, Ochoa A, Sierra R, **Ritchie MD**, Turner S, Fontham E, Delgado AG, Schneider B. Association of haplotypes of inflammation-related genes with gastric preneoplastic lesions in African Americans and Caucasians. *International Journal of*

Cancer, 128(3):668-75 (2011). doi: 10.1002/ijc.25385. PMID: 20473875, PMCID: PMC2964400

84. Cattaert T, Urrea V, Naj AC, De Lobel L, De Wit V, Fu M, John JM, Shen H, Calle LM, **Ritchie MD**, Edwards T, Van Steen K. FAM-MDR: a flexible family-based multifactor dimensionality reduction technique to detect epistasis using related individuals. *PlosONE*, 5(4):e10304 (2010). PMID: 20421984, PMCID: PMC2858665

85. Ribaudo HJ, Liu H, Schwab M, Schaeffeler E, Eichelbaum M, Motsinger-Reif AA, **Ritchie MD**, Zanger UM, Acosta EP, Morse GD, Gulick RM, Robbins GK, Clifford D, Haas DW. Impact of CYP2B6, ABCB1 and CYP3A5 Polymorphisms on Efavirenz Pharmacokinetics and Treatment Response: An AIDS Clinical Trials Group Study. *Journal of Infectious Disease*, 202(5):717-22 (2010). doi: 10.1086/655470. PMID: 20662624, PMCID: PMC2919241

86. Wilke RA, Berg RL, Linneman JG, Peissig P, Starren J, **Ritchie MD**, McCarty CA. Quantification of the clinical modifiers impacting high-density lipoprotein cholesterol in the community: personalized medicine research project. *Prev Cardiol*, Spring; 13(2):63-8 (2010). doi: 10.1111/j.1751-7141.2009.00055.x. PMID: 20377807, PMCID: PMC 2862090

87. Holzinger ER, Buchanan CC, Dudek SC, Torstenson EC, Turner SD, **Ritchie MD**. Initialization Parameter Sweep in ATHENA: Optimizing Neural Networks for Detecting Gene-Gene Interactions in the Presence of Small Main Effects. *GECCO 2010*, 12: 203-210 (2010). PMID: 21152364, PMCID: PMC 2997651

88. Dumitrescu L, **Ritchie MD**, Brown-Gentry K, Pulley JJ, Basford M, Denny J, Oksenberg JR, Roden DM, Haines JL, Crawford DC. Assessing the accuracy of observer-reported ancestry in a biorepository linked to electronic medical records. *Genetics in Medicine*, 12(10):648-50. (2010) doi: 10.1097/GIM.0b013e3181efe2df. PMID: 20733501, PMCID: PMC2952033

89. **Ritchie MD**, Denny JC, Crawford DC, Havens A, Weiner J, Pulley JM, Basford M, Balser JR, Masys DR, Haines JL, Roden DM. Robust replication of genotype-phenotype associations across multiple diseases in an electronic medical record. *American Journal of Human Genetics*, 86(4):560-72 (2010). doi: 10.1016/j.ajhg.2010.03.003. PMID: 20362271, PMCID: PMC2850440

90. Turner SD, Dudek SM, **Ritchie MD**. ATHENA: A Knowledge-Based Hybrid Backpropagation-Grammatical Evolution Neural Network Algorithm for Discovering Epistasis among Quantitative Trait Loci. *BMC BioData Mining*, 3(1):5 (2010). doi: 10.1186/1756-0381-3-5. PMID: 20875103, PMCID: PMC 2955681

91. Pendergrass SA, Dudek SM, Crawford DC, **Ritchie MD**. Synthesis-View: visualization and interpretation of SNP association results for multi-cohort, multi-phenotype data and meta-analysis. *BMC BioData Mining*, 3(1): 10 (2010). doi: 10.1186/1756-0381-3-10. PMID: 21162740 , PMCID: PMC 3012023

92. Hulgan T, Haubrich R, Riddler SA, Tebas P, **Ritchie MD**, McComsey GA, Haas DW, Canter JA. European Mitochondrial DNA Haplogroups and Metabolic Changes during Antiretroviral Therapy in AIDS Clinical Trials Group Study A5142. *AIDS*, (1):37-47. (2011) doi: 10.1097/QAD.0b013e32833f9d02. PMID: 20871389 PMCID: PMC2995830

93. McCarty CA, Chisholm RL, Chute C, Kullo I, Jarvik G, Larson EB, Li R, Masys DR, **Ritchie MD**, Roden DM, Struewing J, Wolf WA; the eMERGE Team. (2011) The eMERGE Network: A consortium of biorepositories linked to electronic medical records data for conducting genomic studies. *BMC Genomics*, 4(1):13. doi: 10.1186/1755-8794-4-13. PMID: 21269473, PMCID: PMC3038887

94. Denny JC, **Ritchie MD**, Crawford DC, Schildcrout JS, Ramirez AH, Pulley JM, Basford MA, Masys DR, Haines JL, Roden DM. (2011) Identification of genomic predictors of atrioventricular conduction: Using electronic medical records as a tool for genome science. *Circulation*, 122: 2016-2021. doi: 10.1161/CIRCULATIONAHA.110.948828. PMID: 21041692, PMCID: PMC2991609

95. Turner SD, Dudek SM, **Ritchie MD**. Incorporating Domain Knowledge into Evolutionary Computing for Discovering Gene-Gene Interaction. *11th Int'l Conference on Parallel Problem*

Solving From Nature (PPSN), Lecture Notes in Computer Science. 6238:394-403. (2011) PMID: 20938492, PMCID: PMC2951735

96. Grady BJ, Torstenson ES, McLaren PJ, de Bakker P, Haas DW, Robbins GK, Gulick RM, Haubrich R, Ribaudo H, **Ritchie MD**. Use of biological knowledge to inform the analysis of gene-gene interactions involved in modulating virologic failure with efavirenz-containing treatment regimens in art-naïve actg clinical trials participants. *Pacific Symposium on Biocomputing,* 253-64. (2011) PMID: 21121053, PMCID: PMC 3094912

97. Pendergrass S, Dudek SM, Roden DM, Crawford DC, **Ritchie MD**. Visual integration of results from a large DNA Biobank (BioVU) using Synthesis-View. *Pacific Symposium on Biocomputing,* 265-275 (2011). PMID: 21121054, PMCID: PMC 3065108

98. **Ritchie MD**. Using Biological Knowledge to Uncover the Mystery in the Search for Epistasis in Genome-Wide Association Studies. *Ann. of Human Genetics,* 75(1): 172-82. (2011). PMID: 21158748, PMCID: PMC3092784

99. Bush WS, McCauley JL, DeJager PL, Dudek SM, Hafler DA, Gibson RA, Matthews PM, Kappos L, Naegelin Y, Polman CH, Hauser SL, Oksenberg J, Haines JL, **Ritchie MD.** A knowledge-driven interaction analysis reveals novel mechanism of MS susceptibility. *Genes & Immunity*, 2011 Feb 24 (Epub ahead of print). PMID: 21346779, PMCID: PMC 3136581

100. Turner SD, Armstrong L, Bradford Y, Carlson C, Crawford DC, Crenshaw AT, de Andrede M, Doheny K, Haines JL, Hayes G, Jarvik G, Jiang L, Ling H, Kullo I, Li R, Manolio TA, Matsumoto M, McCarty CA, McDavid A, Mirel D, Paschall J, Pugh E, Rasmussen LV, Wilke RA, Zuvich RL, **Ritchie MD**. Quality Control procedures for Genome-Wide Association Studies. *Current Protocols in Human Genetics*, Chapter 1: Unit1.19. (2011) PMID: 21234875, PMCID: PMC 3066182

101. Cattaert T, Calle ML, Dudek SM, Mahachie John JM, Van Lishout F, Urrea V, **Ritchie MD**, Van Steen K. Model-Based Multifactor Dimensionality Reduction for detecting epistasis in case-control data in the presence of noise. *Ann Hum Genet*, 75(1): 78-89 (2011). PMID: 21158747, PMCID: PMC 3059142

102. Lehr T, Yuan J, Zeumer D, Jayadev S, **Ritchie MD**. Rule based classifier for the analysis of gene-gene and gene-environment interactions in genetic association studies. *BMC Biodata Mining*, 4(1): 4 (2011). PMID: 21362183, PMCID: PMC 3060133

103. Yaspan BL, Bush WS, Torstenson ES, Ma D, Pericak-Vance MA, **Ritchie MD**, Sutcliffe JS, Haines JL. Genetic analysis of biological pathway data through genomic randomization. *Human Genetics*, 2011 Jan 30 (Epub ahead of print). PMID: 21279722, PMCID: PMC3107984

104. Pendergrass SA, Brown-Gentry K, Dudek SM, Torstenson ES, Ambite JL, Avery CL, Buyske S, Cai C, Fesinmeyer MD, Haiman C, Heiss G, Hindorff LA, Hsu C-N, Jackson RD, Kooperberg C, Le Marchand L, Lin Y, Matise TC, Moreland L, Monroe K, Reiner AP, Wallace R, Wilkens LR, Crawford DC, **Ritchie MD**. The Use of Phenotype-Wide association study (PheWAS) for exploration of novel genotype-phenotype relationships and pleiotropy discovery. *Genetic Epidemiology*, 35: 410-22 (2011) PMID: 21594894, PMCID: PMC3116446

105. Holzinger ER, Dudek SM, Torstenson EC, **Ritchie MD**. ATHENA Optimization: The Effect of Initial Parameter Settings Across Different Genetic Models. *Lect Notes Comput Sci*. 2011 Jan 1;6623(2011):48-58. PMID: 21966082, PMCID: PMC 3182152

106. Matise T, Ambite JL, Buyske, Cole S, Crawford DC, Haiman C, Heiss G, Kooperberg C, Le Marchand L, Manolio T, North K, Peters R, **Ritchie MD**, Hindorff L, Haines JL, for PAGE. The next PAGE in understanding complex traits: study design for analysis of Population Architecture using Genetics and Epidemiology. *American Journal of Epidemiology*, 2011 Oct 1;174(7):849-59. PMID: 21836165, PMCID: PMC3176830

107. Grady BJ, Torstenson ES, **Ritchie MD**. The effects of linkage disequilibrium in large scale SNP datasets for MDR. *Biodata Mining*, 2011 May 5; 4(1):11. PMID: 21545716, PMCID: PMC 3108918

108. Hsu Ch-N, Kuo Ch-J, Cai C, Pendergrass SA, **Ritchie MD**, Ambite JL. Learning Phenotype Mapping for Integrating Large Genetic Data. *Proceedings of BioNLP 2011 Workshop*, June 2011, 19-27.

109. Turner SD, Berg RL, Linneman JG, Peissig PL, Crawford DC, Denny JC, Roden DM, McCarty CA, **Ritchie MD,** Wilke RA. Knowledge-Driven Multi-Locus Analysis Reveals Gene-Gene Interactions Influencing HDL Cholesterol Level in Two Independent EMR-Linked Biobanks. *PLoS One.* 2011 May 11;6(5):e19586. doi: 10.1371/journal.pone.0019586. PMID: 21589926, PMCID: PMC 3092760

110. Kullo IJ, Ding K, Shameer K, McCarty CA, Jarvik GP, Denny JC, **Ritchie MD**, Zi Y, Crosslin DR, Chisholm RL, Manolio TA, Chute CG. Complement Receptor 1 Gene Variants are Associated with Erythrocyte Sedimentation Rate. *AJHG*, 2011 Jul 15;89(1):131-8. PMID: 21700265, PMCID: PMC 3135803

111. Xu H,Jiang M,Oetjens M,Bowton EA,Ramirez AH,Jeff JM,Basford MA,Pulley JM,Cowan JD,Wang X,**Ritchie MD**,Masys DR,Roden DM,Crawford DC,Denny JC . Facilitating pharmacogenetic studies using electronic health records and natural-language processing: a case study of warfarin. *J Am Med Inform Assoc.* 2011 Jul 1;18(4):387-91. PMID: 21672908, PMCID: PMC 3128409

112. Avery CL, He Q, North KE, Ambite JL, Boerwinkle E, Fornage M, Hindorff LA, Kooperbert C, Meigs JB, Pankow JS, Pendergrass SA, Psaty BM, **Ritchie MD**, Rotter ji, Taylor KD, Wilkens LR, Heiss G, Lin DY. A Phenomics-Based Strategy Identifies Loci on *APOC1, BRAP,* and *PLCG1* Associated with Metabolic Syndrome Phenotype Domains. October 2011 *PLOS Genetics*. PMID: 2202282, PMCID: PMC 3192835

113. Delaney JT, Ramirez AH, Bowton E, Pulley JM, Basford MA, Schildcrout JS, Shi Y, Oetjens M, Cleator JH, Jahangir E, **Ritchie MD**, Masys DR, Crawford DC, Roden DM, Denny JC. Predicting clopidogrel response using DNA samples linked to an electronic health record. *CPT*, 2012 Feb;91(2):257-63. PMID: 22190063, PMCID: PMC 3621954

114. Mechanic LE, Cheng HS, Amos CI, Chatterjee N, Cox NJ, Divi RL, Fan R, Harris EL, Jacobs K, Kraft P, Leal SM, McAllister K, Moore JH, Paltoo DN, Province MA, Ramos EM, **Ritchie MD**, roeder K, Schaid DJ, Stephens M, Thomas DC, Weinberg CR, Witte JS, Zhang S, Zöllner S, Feuer EJ, Gillanders EM. Next Generation Analytic Tools for Large Scale Genetic Epidemiology Studeis of Complex Diseases. *Genetic Epidemiology*, 2011 Dec 6. doi: 10.1002/gepi.20652. [Epub ahead of print]. PMID: 22147673, PMCID: PMC 3368075

115. Crosslin DR, McDavid A, Weston N, Nelson SC, Zheng X, Hart E, deAndrade M, Kullo IJ, McCarty CA, Doheny KF, Pugh E, Kho A, Hays MG, Pretel S, Saip A, **Ritchie MD**, Crawford DC, Crane PK, Newton K, Li R, Mirel D, Crenshaw A, Larson EB, Carlson C, Jarvik GP, eMERGE . Genetic variants associated with the white blood cell count in 13,923 subjects in the eMERGE Network. *Human Genetics.* October 2011 PMID: 22037903 PMCID: PMC 22037903

116. Haas DW, Kuritzkes D, **Ritchie MD**, Smur S, Gage BF, Maartens G, Masys D, Fellay J, Phillips E, Ribaudo HJ, Freedberg KA, Petropoulos C, Manolio TA, Gulick RM, Haubrich R, Kim P, Dehlinger M, Abebe R, Telenti A. Pharmacogenomics of HIV Therapy: Summary of a Workshop Sponsored by the National Institute of Allergy and Infectious Diseases. *HIV Clinical Trials.* Sept/Oct 2011 12; 12(5): 277-85 PMID: 22180526, PMCID: PMC3322423

117. Denny JC, Crawford DC, **Ritchie MD**, Bielinski SJ, Basford MA, Bradford Y, Chai HS, Bastarache L, Zuvich R, Peissig P, Carrell D, Ramirez AH, Pathak J, Wilke RA, Rasmussen L, Wang X, Pacheco JA, Kho AN, Hayes MG, Weston N, Matsumoto M, Kopp PA, Newton KM, Jarvik GP, Li R, Manolio TA, Kullo IJ, Chute CG, Chisholm RL, Larson EB, McCarty CA, Masys DR, Roden DM, de Andre M. Variants Near FOXE1 Are Associated with Hypothyroidism and Other Thyroid Conditions: Using Electronic Medical Records for Genome- and Phenome-wide Studies. *The American Journal of Human Genetics* 89, 529-542, October 7, 2011. PMID: 21981779, PMCID: PMC3192835

118. Kääb S, Crawford DC, Sinner MF, Behr ER, Schulze-Bahr E, Guicheney P, Bishopric NH, Myerburg RJ, Schott JJ, Pfeufer A, Beckmann BM, Martens E, Zhang T, Stallmeyer B,

Sumhagen S, Denjoy I, Bardai A, Van Gelder IC, Jamshidi Y, Dalageorgou C, Marshal V, Jeffery S, Shakir S, Camm AJ, Steinbeck G, Perz S, Lichtner P, Meitinger T, Peters A, Wichmann HE, Ingram C, Bradford Y, Carter S, Norris K, **Ritchie MD**, George AL, Roden DM. A Large Candidate Gene Survey Identifies the *KCNE1* D85N Polymorphism as a Possible Modulator of Drug-induced Torsades de Pointes. *Circ Cardiovasc Genet*. 2011 11; (1) PMID: 22100668, PMCID: PMC3237759

119. Grady BJ, Samuels DC, Robbins GK, Selph D, Canter JA, Pollard RB, Haas DW, Shafer R, Kalams SA, Murdock DG, **Ritchie MD**, Hulgan T for the ACTG 384 and DACS 250 study teams. Mitochondrial Genomics and CD4T-cell Count Recovery after Antiretroviral Therapy Initiation in AIDS Clinical Trials Group Study 384. *Journal of Acquired Immune Deficiency Syndromes*, 2011 Dec 1;58(4):363-70. PMID: 21792066, PMCID: PMC320417

120. Birdwell KA, Grady B, Choi L, Xu H, Bian A, Denny JC, Jiang M, Vranic G, Basford M, Cowan JD, Richardson DM, Robinson MP, Ikizler TA, **Ritchie MD**, Stein CM, Hass DW. Use of a DNA Biobank Linked to Electronic Medical Records to Characterize Pharmacogenomic Predictors of Tacrolimus Dose Requirement in Kidney Transplant Recipients. *Pharmacogenetics and Genomics.* 2012 Jan;22(1):32-42 PMID: 22108237, PMCID: PMC3237759

121. Fullerton S, Wolf W, Brothers K, Clayton E, Crawford D, Denny J, Greenland P, Koenig B, Leppig K, Lindor L, MccARTHY C, McGuire A, Mc Peek Hinz G, Mirel D, Ramos E, **Ritchie MD**, Smith M, Waudby C, Burke W, Jarvic G. Return of Individual Research Results from Genome-wide Association Studies: Experience of the Electronic Medical Records & Genomics (eMERGE) Network. *Genetics in Medicine*. 2012 Apr;14(4):424-31 PMID: 22361898, PMCID: PMC3297180

122. **Ritchie MD**, Rowan S, Kucera G, Stubblefield T, Blair M, Carter S, Roden DM, Darbar D. J Chromosome 4q25 variants are genetic modifiers of rare ion channel mutations associated with familial atrial fibrillation. *Am Coll Cardiol.* 2012 Sep 25;60(13):1173-81 PMID: 22818067, PMCID: PMC3448817

123. Ramirez AH, Shi Y, Schildcrout JS, Delaney JT, Xu H, Oetjens MT, Zuvich RL, Basford MA, Bowton E, Jiang M, Speltz P, Zink R, Cowan J, Pulley JM, **Ritchie MD**, Masys DR, Roden DM, Crawford DC, Denny JC. Predicting warfarin dosage in European-Americans and African-Americans using DNA samples linked to an electronic health record. *A Pharmacogenomics.* 2012 Mar;13(4):407-18. Epub 2012 Feb 13. PMID: 22329724, PMCID: PMC3361510

124. Buchanan CC, Torstenson ES, Bush WS, **Ritchie MD**. A comparison of cataloged variation between International HapMap Consortium and 1000 Genomes Project data. *J Am Med Inform Assoc*. 2012 Mar-Apr;19(2):289-94. PMID: 22319179, PMCID: PMC3277631

125. Zuvich RL, Armstrong LL, Bielinski SJ, Bradford Y, Carlson CS, Crawford DC, Crenshaw AT, de Andrade M, Doheny KF, Haines JL, Hayes MG, Jarvik GP, Jiang L, Kullo IJ, Li R, Ling H, Manolio TA, Matsumoto ME, McCarty A, McDavid AN, Mirel DB, Olson LM, Paschall JE, Pugh EW, Rasmussen LV, Rasmussen-Torvik LJ, Turner SD, Wilke RA, **Ritchie MD**. Pitfalls of merging GWAS data: lessons learned in the eMERGE network and quality control procedures to maintain high data quality. *Genet Epidemiol*. 2011 Dec;35(8):887-98. doi: 10.1002/gepi.20639. PMID: 22125226, PMCID: PMC3592376

126. Ellinor PT, Lunetta KL, Albert CM, Gudbjartsson DF, Glazer NL, **Ritchie MD**, Smith AV, Arking DE, Müller M, Krijthe BP, Lubitz SA, Bis JC, Chung MK, Dörr M, Ozaki K, Roberts JD, Smith JG, Holm H, Pfeufer A, Sinner MF, Smith NL, Smith JD, Rienstra M, Rice KM, Van Wagoner DR, Magnani JW, Wakili R, Clauss S, Arnar DO, Rotter JI, Steinbeck G, Launer LJ, Davies RW, Borkovich M, Harris TB, Lin H, Völker U, Völzke H, Milan DJ, Hofman A, Boerwinkle E, Chen LY, Soliman E, Voight BF, Li G, Chakravarti A, Kubo M, Tedrow U, Rose LM, Ridker PM, Conen D, Tsunoda T, Furukawa T, Sotoodehnia N, Xu S, Kamatani N, Levy DL, Nakamura Y, Parvez B, Thorsteinsdottir U, Muhammad R, Psaty BM, Meitinger T, Perz S, Wichmann H-E, Witteman JCM, Kao WHL, Kathiresan S, Roden DM, Kong A, Uitterlinden AG, Rivadeneira F, McKnight B, Sjögren M, Gollob MH, Melander O, Tanaka T, Stricker BHCh, Felix SB, Alonso A, Gudnason V, Darbar D, Barnard J, Stefansson K, Heckbert SR, Chasman D, Benjamin EJ, Kääb S. Meta-Analysis in the AFGen Consortium Identifies Seven

Novel Loci for Atrial Fibrillation. *Nature Genetics*. 2012 Apr 29;44(6):670-5 PMID: 22544366, PMCID: PMC3366038

127. Ding K, Shameer K, Jouni H, Masys DR, Jarvik GP, Kho AN, **Ritchie MD**, McCarty CA, Chute CG, Manolio TA, Kullo IJ. Genetic Loci Implicated in Erythroid Differentiation and Cell Cycle Regulation are Associated with Red Blood Cell Traits. *Mayo Clin Proc. 2012 May;87(5):461-74.* PMID: 22560525, PMCID: PMC3538470

128. Stranger BE, Björkegren J, Dolan ME, **Ritchie MD**. Systems and genome-wide approaches unite to provide a route to personalized medicine. *Genome Med*. 2012 Mar 30;4(3):29. PMID: 22494390, PMCID: PMC3446279

129. Peters U, North KE, Sethupathy P, Buyske S, Haessler J, Fesinmeyer MD, Jackson RD, Kuller L, Rajkovic A, Lim U, Cheng I, Schumacher F, Wilkens L, Li R, Monda K, Ehret G, Nguyen KH, Cooper R, Lewis CE, Leppert M, Irvin MR, Gu CC, Houston D, Bůžková P, **Ritchie MD**, Matise TC, Le Marchand L, Hindorff LA, Crawford DC, Haiman CA, Kooperberg CL. A systematic mapping approach of 16q12.2/FTO and BMI in over 20,000 African Americans narrows in on the underlying functional variation: results from the Population Architecture using Genomics and Epidemiology (PAGE) Study. *PLoS Genetics*, 2013 January; 9(1): e1003171. PMID: 23341774, PMCID: PMC3547789

130. Holzinger ER, Grady B, **Ritchie MD**, Ribaudo HJ, Acosta EP, Morse GD, Gulick RM, Robbins GK, Clifford DB, Daar ES, McLaren P, Hass DW. Genome-Wide Association Study of Plasma Efavirenz Pharmacokinetics in AIDS clinical Trials Group Protocols implicates several CYP2B6 variants. *Pharmacogenetics and Genomics*. 2012 Dec;22(12):858-67. PMID: 23080225, PMCID: PMC3614365

131. Holzinger ER, Hulgan T, Ellis RJ, Samuels DC, **Ritchie MD**, Haas DW, Kallianpur AR, Bloss CS, Clifford DB, Collier AC, Gelman BB, Marra CM, McArthur JC, McCutchan JA, Morgello S, Simpson DM, Franklin DR, Rosario D, Selph D, Letendre S, Grant I, for the CHARTER group. Mitochondrial DNA variation and HIV-associated sensory neuropathy in CHARTER. *Journal of Neurovirology*, 2012 Dec;18(6):511-20. PMID: 23073667, PMCID: PMC3587171

132. S.A. Pendergrass, K. Brown-Gentry, S. Dudek, E.S. Torstenson, R.Goodloe, J.L. Ambite, C.L. Avery, S. Buyske, P. Bůžková, C. Cai, E. Deelman, M.D. Fesinmeyer,C. Haiman G. Heiss, L.A. Hindorff, C.-N. Hsu; R. D. Jackson; C. Kooperberg,L. LeMarchand, Y. Lin, T. C. Matise, K.R. Monroe, L. Moreland, A. Reiner, R. Wallace, L. R. Wilkens, D.C. Crawford, **M.D. Ritchie.** Phenome-Wide Association Study (PheWAS) for Detection of Pleiotropy within the Population Architecture Using Genomics and Epidemiology (PAGE) Network. *PLoS Genetics*, 2013 January; 9(1): e1003087. PMID: 23382687, PMCID: PMC3561060

133. Graff, Mariaelisa; Gordon-Larsen, Penny; Lim, Unhee; Fowke, Jay; Love, Shelly-Ann; Fesinmeyer, Megan; Wilkens, Lynne; Vertilus, Shawyntee; **Ritchie, Marylyn**; Prentice, Ross; Pankow, James; Monroe, Kristine; Manson, JoAnn; Marchand, Loic; Kuller, Lewis; Kolonel, Laurence; Hong, Ching; Henderson, Brian; Haessler, Jeff; Gross, Myron; Goodloe, Robert; Franceschini, Nora; Carlson, Christopher; Buyske, Steven; Buzkova, Petra; Hindorff, Lucia; Matise, Tara; Crawford, Dana; Haiman, Chris; Peters, Ulrike; North, Kari. The Influence of Obesity related SNPs on BMI across the Life Course: the PAGE Study, *Diabetes*. 2013 Jan 8. PMID: 23300277, PMC ID: PMC3636619

134. Crosslin DR, McDavid A, Weston N, Zheng X, Hart E, de Andrade M, Kullo IJ, McCarty CA, Doheny KF, Pugh E, Kho A, Hayes MG, **Ritchie MD**, Saip A, Crawford DC, Crane PK, Newton K, Carrell DS, Gallego CJ, Nalls MA, Li R, Mirel DB, Crenshaw A, Couper DJ, Tanaka T, van Rooij FJ, Chen MH, Smith AV, Zakai NA, Yango Q, Garcia M, Liu Y, Lumley T, Folsom AR, Reiner AP, Felix JF, Dehghan A, Wilson JG, Bis JC, Fox CS, Glazer NL, Cupples LA, Coresh J, Eiriksdottir G, Gudnason V, Bandinelli S, Frayling TM, Chakravarti A, van Duijn CM, Melzer D, Levy D, Boerwinkle E, Singleton AB, Hernandez DG, Longo DL, Witteman JC, Psaty BM, Ferrucci L, Harris TB, O'Donnell CJ, Ganesh SK; CHARGE Hematology Working Group, Larson EB, Carlson CS, Jarvik GP; The electronic Medical Records and Genomics (eMERGE) Network. Genetic variation associated with circulating monocyte count in the eMERGE

Network. *Hum Mol Genet*. 2013 May 15;22(10):2119-27. Epub 2013 Jan 12. PMID: 23314186, PMCID: PMC3633369

135. Fesinmeyer MD, North KE, Lim U, Buzková P, Crawford DC, Haessler J, Gross MD, Fowke JH, Goodloe R, Love SA, Graff M, Carlson CS, Kuller LH, Matise TC, Hong CP, Henderson BE, Allen M, Rohde RR, Mayo P, Schnetz-Boutaud N, Monroe KR, **Ritchie MD**, Prentice RL, Kolonel LN, Manson JE, Pankow J, Hindorff LA, Franceschini N, Wilkens LR, Haiman CA, Marchand L, Peters U. Effects of smoking on the genetic risk of obesity: the population architecture using genomics and epidemiology study. *BMC Med Genet*. 2013 Jan 11;14(1):6. PMID: 23311614, PMCID: PMC3564691

136. Kim D, Kim S, Risacher SL, Shen L, **Ritchie MD**, Weiner MW, Saykin AJ, Nho K; the Alzheimer's Disease Neuroimaging Initiative (ADNI). A Graph-Based Integration of Multimodal Brain Imaging Data for the Detection of Early Mild Cognitive Impairment (E-MCI). *Multimodal Brain Image Anal* (2013). 2013; 8159:159-169. PMID: 25383392, PMCID: PMC4224282

137. Spencer KL, Malinowski J, Carty CL, Franceschini N, Fernández-Rhodes L, Young A, Cheng I, **Ritchie MD**, Haiman CA, Wilkens L, Chunyuanwu, Matise TC, Carlson CS, Brennan K, Park A, Rajkovic A, Hindorff LA, Buyske S, Crawford DC. Genetic variation and reproductive timing: African American women from the Population Architecture using Genomics and Epidemiology (PAGE) Study. *PLoS One*. 2013;8(2):e55258. doi: 10.1371/journal.pone.0055258. Epub 2013 Feb 12. PMID: 23424626, PMCID: PMC3570525

138. Rasmussen-Torvik LJ, Pacheco JA, Wilke RA, Thompson WK, **Ritchie MD**, Kho AN, Muthalagu A, Hayes MG, Armstrong LL, Scheftner DA, Wilkins JT, Zuvich RL, Crosslin D, Roden DM, Denny JC, Jarvik GP, Carlson CS, Kullo IJ, Bielinski SJ, McCarty CA, Li R, Manolio TA, Crawford DC, Chisholm RL. High density GWAS for LDL cholesterol in African Americans using electronic medical records reveals a strong protective variant in APOE. *Clin Transl Sci*. 2012 Oct;5(5):394-9. doi: 10.1111/j.1752-8062.2012.00446.x. Epub 2012 Aug 23. PMID: 23067351, PMCID: PMC3521536

139. Fesinmeyer MD, North KE, **Ritchie MD**, Lim U, Franceschini N, Wilkens LR, Gross MD, Bůžková P, Glenn K, Quibrera PM, Fernández-Rhodes L, Li Q, Fowke JH, Li R, Carlson CS, Prentice RL, Kuller LH, Manson JE, Matise TC, Cole SA, Chen CT, Howard BV, Kolonel LN, Henderson BE, Monroe KR, Crawford DC, Hindorff LA, Buyske S, Haiman CA, Le Marchand L, Peters U. Genetic Risk Factors for BMI and Obesity in an Ethnically Diverse Population: Results from the Population Architecture Using Genomics and Epidemiology (PAGE) Study. *Obesity* (Silver Spring). 2013 Apr;21(4);835-46. doi: 10.1002/oby.20268. PM 23712987, PMC 3482415

140. Schildcrout JS*, Denny JC*, Bowton E, Gregg W, Pulley JM, Basford MA, Cowan J, Ramirez AH, Crawford DC, **Ritchie MD**, Peterson JF, Masys DR, Wilke R, Roden DM. Optimizing drug outcomes with pharmacogenomics: A case for preemptive genotyping. *Clin Pharmacol Ther*. 2012 Aug;92(2):235-42. doi: 10.1038/clpt.2012.66. Epub 2012 Jun 27. PMID: 22739144, PMCID: PMC3785311

141. Pendergrass SA, Dudek SM, Crawford DC, **Ritchie MD**. Visually integrating and exploring high throughput Phenome-Wide Association Study (PheWAS) results using PheWAS-View. *BioData Min.* 2012 Jun 8;5(1):5. PMID: 22682510, PMC ID: PMC3476448

142. Pendergrass SA, Verma SS, Holzinger ER, Moore CB, Wallace JR, Dudek SM, Huggins W, Kitchner T, Waudby C, Berg R, McCarty CA, **Ritchie MD**. Next-Generation Analysis of Cataracts: Determining Knowledge Driven Gene-Gene Interactions Using Biofilter, and Gene-Environment Interactions Using the PhenX Toolkit. *Pacific Symposium on Biocomputing* 18:147-158(2013) PMID 23424120, PMCID: PMC3615413

143. Crawford DC, Goodloe R, Brown-Gentry K, Wilson S, Roberson J, Gillani NB, **Ritchie MD**, Dilks HH, Bush WS. Characterization of the Metabochip in Diverse Populations from the International HapMap Project in the Epidemiologic Architecture for Genes Linked to Environment (EAGLE) Project. *Pacific Symposium on Biocomputing* 18:188-199(2013) PMID: 23424124, PMCID: PMC3584704

144. Moore CB, Wallace JR, Frase AT, Pendergrass SA, **Ritchie MD**. Using BioBin to Explore Rare Variant Population Stratification. *Pacific Symposium on Biocomputing* 18:332-343(2013) PMID: 23424138, PMCID: PMC3638724

145. Bush WS, Boston J, Pendergrass SA, Dumitrescu L, Goodloe R, Brown-Gentry K, Wilson S, McClellan R Jr, Torstenson E, Basford MA, Spencer KL, **Ritchie MD**, Crawford DC. Enabling High-Throughput Genotype-Phenotype Associations in the Epidemiologic Architecture for Genes Linked to Environment (EAGLE) Project as Part of the Population Architecture Using Genomics and Epidemiology (PAGE) Study. *Pacific Symposium on Biocomputing* 18:373-384(2013) PMID: 23424142, PMCID: PMC3579641

146. Holzinger ER, Dudek SM, Frase AT, Krauss RM, Medina MW, **Ritchie MD**. ATHENA: A Tool for Meta-Dimensional Analysis Applied to Genotypes and Gene Expression Data to Predict HDL Cholesterol Levels. *Pacific Symposium on Biocomputing* 18:385-396(2013) PMID: 23424143, PMCID: PMC3587764

147. Moore CB, Wallace JR, Frase AT, Pendergrass SA, **Ritchie MD**. BioBin: A bioinformatics tool for automating the binning of rare variants using publicly available biological knowledge. *BMC Medical Genomics*. 2013; 6(Suppl 2): S6. PMID: 23819467, PMCID: PMC3654874

148. **Ritchie MD**, Deny JC, Zuvich RL, Crawford DC, Schildcrout JS, Bastarache L, Ramirez AH, Mosley JD, Pulley JM, Basford MA, Bradford Y, Rasmussen LV, Pathak J, Chute CG, Kullo IJ, McCarty C, Chisholm RL, Kho AN, Carlson CS, Larson EB, Jarvik GP, Sotoodehnia N, Manolio TA, Li R, Masys DR, Haines JL, Roden DM. Genome- and phenome-wide analysis of cardiac conduction identifies markers of arrhythmia risk. *Circulation*, 2013. PMID: 23463857, PMCID: PMC3713791

149. McGeachie MJ, Stahl EA, Himes BE, Pendergrass SA, Lima JJ, Irvin CG, Peters SP, **Ritchie MD**, Plenge RM, Tantisira KG. Polygenic Heritability Estimates in Pharmacogenetics: Focus on Asthma and Related Phenotypes. *Pharmacogenet Genomics.* 2013 Jun;23(6):324-328. PMID: 23532052, PMCID: PMC3767309

150. Gottesman O, Kuivaniemi H, Tromp G, Faucett WA, Li R, Manolio TA, Sanderson SC, Kannry J, Zinberg R, Basford MA, Brilliant M, Carey DJ, Chisholm RL, Chute CG, Connolly JJ, Crosslin D, Denny JC, Gallego CJ, Haines JL, Hakonarson H, Harley J, Jarvik GP, Kohane I, Kullo IJ, Larson EB, McCarty C, Ritchie MD, Roden DM, Smith ME, Böttinger EP, Williams MS; eMERGE Network. The Electronic Medical Records and Genomics (eMERGE) Network: past, present, and future. *Genet Med*. 2013 Oct;15(10):761-71. doi: 10.1038/gim.2013.72. Epub 2013 Jun 6. Review. PMID: 23743551, PMCID: PMC3795928

151. Park SL, Cheng I, Pendergrass SA, Kucharska-Newton AM, Lim U, Ambite JL, Caberto CP, Monroe KR, Schumacher F, Hindorff LA, Oetjens MT, Wilson¬ S, Goodloe RJ, Love SA,. Henderson BE, Kolonel LN, Haiman CA, Crawford DC, North KE, Heiss G, **Ritchie MD**, Wilkens LR, Le Marchand L. Association of the FTO Obesity Risk Variant rs8050136 With Percent Energy Intake from Fat in Multiple Ethnic/Racial Populations: The PAGE Study. *American Journal of Epidemiology*, in press (2013). PMID: 23820787, PMCID: PMC3755639

152. Jeff JM, **Ritchie MD**, Denny JC, Kho AN, Ramirez AH, Crosslin D, Armstrong L, Basford MA, Wolf WA, Pacheco JA, Chisholm RL, Roden DM, Hayes G, Crawford DC. Generalization of Variants Identified by Genome-wide Association Studies for ECG Traits in African Americans. *Ann Hum Genet.* 2013 Mar 28. doi: 10.1111/ahg.12023. PMID: 23534349, PMCID: PMC3743946

153. Zhang L, Franceschini N, Buzkova P, Wassel CL, Roman MJ, North KE, Crawford DC, Boston J, Brown-Gentry KD, Cole SA, Deelman E, Goodloe R, Heiss G, Jenny NS, Jorgensen NW, Matise TC, McClellan Jr BE, **Ritchie MD**, Wilson S, Kao WHL. Lack of Associations of Coronary Heart Disease Risk Genetic Variants and Subclinical Atherosclerosis in Four U.S. Populations: the Population Architecture using Genomics and Epidemiology (PAGE) Study. *Atherosclerosis*. 2013 Jun;228(2):390-9. doi: 10.1016/j.atherosclerosis.2013.02.038. Epub 2013 Mar 13. PMID: 23587283, PMCID: PMC 3717342

154. Ding K, de Andrade M, Manolio T, Crawford D, Rasmussen-Torvik L, **Ritchie M**, Denny J, Masys D, Jouni H, Pacheco J, Kho A, Roden D, Chisholm R, Kullo I, Genetic Variants That Confer Resistance to Malaria are Associated with Red Blood Cell Traits in African Americans: An Electronic Medical Record-Based Genome Wide Association Study. *G3* (Bethesda). 2013 5; PMID: 23696099, PMCID: PMC3704235

155. Oetjens M, Denny J, **Ritchie M**, Gillani N, Richardson D, Restrepo N, Pulley J, Dilks H, Basford M, Bowton E, Masys D, Wilke R, Roden D, Crawford D, Assessment of a pharmacogenomic marker panel in a polypharmacy population identified from electronic medical records. *Pharmacogenomics.* 2013 05; 14(7): 735-44. PMID: 23651022, PMCID: PMC3725600

156. Girirajan S, Johnson R, Tassone F, Balciuniene J, Katiyar N, Fox K, Baker C, Srikanth A, Yeoh K, Khoo S, Nauth T, Hansen R, **Ritchie M**, Hertz-Picciotto I, Eichler E, Pessah I, Selleck S, Global increases in both common and rare copy number load associated with autism. *Hum. Mol. Genet.* 2013 4; (2): PMID: 23535821, PMCID: PMC3690969

157. McDavid A, Crane P, Newton KM, Crosslin D, McCormik W, Weston N, Ehrlich K, Hart E, Harrison R, Kukull WA, Rottscheit C, Peissing P, Stefanski E, McCartha CA, Suvich RL, **Ritchie MD**, Schellenberg G, deAndrade M, Kullo I, Li R, Mirel D, Crenshaw A, Bowed JD, Li G, Tsuang D, McCurry S, Teri L, Larson E, Jarvik GP, Carlson CS. Enhancing the Power of Genetic Association Studies through the Use of Silver Standard Cases Derived from Electronic Medical Records. *PLoS One.* 2013 Jun 10;8(6):e63481. PMID: 23762230, PMCID: PMC3677889

158. Hulgan T, Stein JH, Cotter BR, Murdock DG, **Ritchie MD**, Dube MP, Gerschenson M, Haas DW, and Torriani FJ. Mitochondrial DNA Variation and Changes in Adiponectin and Endothelial Function in HIV-infected Adults after Antiretroviral Therapy Initiation. *AIDS Res Hum Retroviruses* 2013 Aug 14. [Epub ahead of print]. PMID: 23944767 PMCID: PMC3785797

159. **Ritchie MD**. Erratum to: The success of pharmacogenomics in moving genetic association studies from bench to bedside: study design and implementation of precision medicine in the post-GWAS era. *Hum Genet*. 132:1437. PMID: 22923055, PMCID: PMC3432217

160. Shameer K, Denny JC, Ding K, Jouni H, Crosslin DR, de Andrade M, Chute CG, Peissig P, Pacheco JA, Li R, Bastarache L, Kho AN, **Ritchie MD,** Masys DR, Chisholm RL, Larson EB, McCarty CA, Roden DM, Jarvik GP, Kullo IJ. A genome- and phenome-wide association study to identify genetic variants influencing platelet count and volume and their pleiotropic effects. *Hum Genet*. 2013 Sep 12. PMID: 24026423, PMCID:PMC3880605

161. Pare G, Kubo M, Byrd JB, McCarty CA, Woodard-Grice A, Teo KK, Anand SS, Zuvich RL, Bradford Y, Ross S, Nakamura Y, **Ritchie M**, Brown NJ. Genetic variants associated with angiotensin-converting enzyme inhibitor-associated angioedema. *Pharmacogenet Genomics.* 2013 Sep;23(9):470-8. doi: 10.1097/FPC.0b013e328363c137. PMID: 23838604, PMCID: PMC3904664

162. 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. doi: 10.1371/journal.pgen.1003959. Epub 2013 Dec 26. PMID: 24385916, PMCID: PMC3873241

163. Holzinger ER, Dudek SM, Frase AT, Pendergrass SA, **Ritchie MD**. ATHENA: the analysis tool for heritable and environmental network associations. *Bioinformatics*. 2014 Mar 1;30(5):698–705 PMID: 24149050 PMCID: PMC3933870

164. 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.* 19:200-211. PMID: 24297547, PMCID: PMC4037237

165. Behr ER, **Ritchie MD**, Tanaka T, Kääb S, Crawford DC, Nicoletti P, Floratos A, Sinner MF, Kannankeril PJ, Wilde AA, Bezzina CR, Schulze-Bahr E, Zumhagen S, Guicheney P, Bishopric

NH, Marshall V, Shakir S, Dalageorgou C, Bevan S, Jamshidi Y, Bastiaenen R, Myerburg RJ, Schott JJ, Camm AJ, Steinbeck G, Norris K, Altman RB, Tatonetti NP, Jeffery S, Kubo M, Nakamura Y, Shen Y, George AL Jr, Roden DM. Genome wide analysis of drug-induced torsades de pointes: lack of common variants with large effect sizes. *PLoS One.* 2013 Nov 6;8(11):e78511. doi: 0.1371/journal.pone.0078511. PMID: 24223155, PMCID: PMC3819377

166. Denny JC, Bastarache L, **Ritchie MD**, Carroll RJ, Zink R, Mosley JD, Field JR, Pulley JM, Ramirez AH, Bowton E, Basford MA, Carrell DS, Peissig PL, Kho AN, Pacheco JA, Rasmussen LV, Crosslin DR, Crane PK, Pathak J, Bielinski SJ, Pendergrass SA, Xu H, Hindorff LA, Li R, Manolio TA, Chute CG, Chisholm RL, Larson EB, Jarvik GP, Brilliant MH, McCarty CA, Kullo IJ, Haines JL, Crawford DC, Masys DR, Roden DM. Systematic comparison of phenome-wide association study of electronic medical record data and genome-wide association study data. *Nat Biotechnol.* 31:1102-1111. PMID: 24270849, PMCID: PMC3969265

167. Wolfe D, Dudek S, **Ritchie MD,** Pendergrass SA. Visualizing genomic information across chromosomes with PhenoGram. *BioData Min.* 2013 Oct 16;6(1):18. PMID: 24131735, PMCID: PMC4015356

168. Gong J, Schumacher F, Lim U, Hindorff LA, Haessler J, Buyske S, Carlson CS, Rosse S, Bůžková P, Fornage M, Gross M, Pankratz N, Pankow JS, Schreiner PJ, Cooper R, Ehret G, Gu CC, Houston D, Irvin MR, Jackson R, Kuller L, Henderson B, Cheng I, Wilkens L, Leppert M, Lewis CE, Li R, Nguyen KD, Goodloe R, Farber-Eger E, Boston J, Dilks HH, **Ritchie MD,** Fowke J, Pooler L, Graff M, Fernandez-Rhodes L, Cochrane B, Boerwinkle E, Kooperberg C, Matise TC, Le Marchand L, Crawford DC, Haiman CA, North KE, Peters U. Fine Mapping and Identification of BMI Loci in African Americans. *Am J Hum Genet*. 2013 Oct 3;93(4):661-71. PMID: 24094743, PMCID: PMC3791273

169. Carlson CS, Matise TC, North KE, Haiman CA, Fesinmeyer MD, Buyske S, Schumacher FR, Peters U, Franceschini N, **Ritchie MD**, Duggan DJ, Spencer KL, Dumitrescu L, Eaton CB, Thomas F, Young A, Carty C, Heiss G, Le Marchand L, Crawford DC, Hindorff LA, Kooperberg CL; PAGE Consortium. Generalization and dilution of association results from European GWAS in populations of non-European ancestry: the PAGE study. *PLoS Biol.* 2013 Sep;11(9):e1001661. Epub 2013 Sep 17. PMID: 24068893, PMCID: PMC3775722

170. Pendergrass SA, Frase A, Wallace J, Wolfe D, Katiyar N, Moore C, **Ritchie MD.** Genomic analyses with biofilter 2.0: knowledge driven filtering, annotation, and model development. *BioData Min.* 2013 Dec 30;6(1):25. PMID: 24378202, PMCID: PMC3917600

171. Kim D, Shin H, Sohn KA, Verma A, **Ritchie MD**, Kim JH. Incorporating inter-relationships between different levels of genomic data into cancer clinical outcome prediction. *Methods*. Epub 2014 Feb 18. pii: S1046-2023(14)00033-4. doi: 10.1016/j.ymeth.2014.02.003. PMID: 24561168, PMC ID:PMC4664202

172. 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 Aug;14(4):336-42. doi: 10.1038/tpj.2014.2. Epub 2014 Feb 11. PMID: 24513692, PMCID: PMC4111770

173. Kim D, Li R, Dudek SM, **Ritchie MD**. ATHENA: Identifying interactions between different levels of genomic data associated with cancer clinical outcomes using grammatical evolution neural network. *BioData Min.* 2013 Dec 20;6(1):23. doi: 10.1186/1756-0381-6-23. PMID: 24359638, PMCID: PMC3912499

174. Mosley JD, Van Driest SL, Larkin EK, Weeke PE, Witte JS, Wells QS, Karnes JH, Guo Y, Bastarache L, Olson LM, McCarty CA, Pacheco JA, Jarvik GP, Carrell DS, Larson EB, Crosslin DR, Kullo IJ, Tromp G, Kuivaniemi H, Carey DJ, **Ritchie MD**, Denny JC, Roden DM. Mechanistic phenotypes: an aggregative phenotyping strategy to identify disease mechanisms using GWAS data. *PLoS ONE*. 2013 Dec 12;8(12):e81503. PMID: 24349080, PMCID: PMC3861317

175. McCarty CA, Berg R, Rottscheit CM, Waudby CJ, Kitchner T, Brilliant M, **Ritchie MD.** Validation of PhenX measures in the personalized medicine research project for use in gene/environment studies. *BMC Med Genomics.* 2014 Jan 14;7:3. doi: 10.1186/1755-8794-7-3. PMID: 24423110, PMCID: PMC3896802

176. Jeff JM, Armstrong LL, **Ritchie MD,** Denny JC, Kho AN, Basford MA, Wolf WA, Pacheco JA, Li R, Chisholm RL, Roden DM, Hayes MG, Crawford DC. Admixture mapping and subsequent fine-mapping suggests a biologically relevant and novel association on chromosome 11 for type 2 diabetes in African Americans. *PLoS ONE.* 2014 Mar 3;9(3):e86931. doi: 10.1371/journal.pone.0086931. eCollection 2014. PMID: 24595071, PMCID: PMC3940426

177. McCarty CA, Huggins W, Aiello AE, Bilder RM, Hariri A, Jernigan TL, Newman E, Sanghera DK, Strauman TJ, Zeng Y, Ramos EM, Junkins HA, PhenX RISING network. PhenX RISING: real world implementation and sharing of PhenX measures. *BMC Med Genomics.* 2014 Mar 20;7:16. doi: 10.1186/1755-8794-7-16. PMID:24650325, PMCID: 3994539

178. Johnson DH, Venuto C, **Ritchie MD**, Morse GD, Daar ES, McLaren PJ, Haas DW. Genomewide association study of atazanavir pharmacokinetics and hyperbilirubinemia in AIDS Clinical Trials Group protocol A5202. *Pharmacogenet Genomics.* 2014 Apr;24(4):195–203. doi: 10.1097/FPC.0000000000000034. PMID: 24557078, PMCID: PMC4059003

179. Sun X, Lu Q, Mukheerjee S, Crane PK, Elston R, **Ritchie MD.** Analysis pipeline for the epistasis search - statistical versus biological filtering. *Front Genet.* 2014 Apr 30;5:106. doi: 10.3389/fgene.2014.00106. eCollection 2014. PMID: 24817878, PMCID: PMC4012196

180. Arking DE, Pulit SL, Crotti L, van der Harst P, Munroe PB, Koopmann TT, Sotoodehnia N, Rossin EJ, Morley M, Wang X, Johnson AD, Lundby A, Gudbjartsson DF, Noseworthy PA, Eijgelsheim M, Bradford Y, Tarasov KV, Dörr M, Müller-Nurasyid M, Lahtinen AM, Nolte IM, Smith AV, Bis JC, Isaacs A, Newhouse SJ, Evans DS, Post WS, Waggott D, Lyytikäinen LP, Hicks AA, Eisele L, Ellinghaus D, Hayward C, Navarro P, Ulivi S, Tanaka T, Tester DJ, Chatel S, Gustafsson S, Kumari M, Morris RW, Naluai AT, Padmanabhan S, Kluttig A, Strohmer B, Panayiotou AG, Torres M, Knoflach M, Hubacek JA, Slowikowski K, Raychaudhuri S, Kumar RD, Harris TB, Launer LJ, Shuldiner AR, Alonso A, Bader JS, Ehret G, Huang H, Kao WH, Strait JB, Macfarlane PW, Brown M, Caulfield MJ, Samani NJ, Kronenberg F, Willeit J; CARe Consortium; COGENT Consortium, Smith JG, Greiser KH, Meyer Zu Schwabedissen H, Werdan K, Carella M, Zelante L, Heckbert SR, Psaty BM, Rotter JI, Kolcic I, Polašek O, Wright AF, Griffin M, Daly MJ; DCCT/EDIC, Arnar DO, Hólm H, Thorsteinsdottir U; eMERGE Consortium, Denny JC, Roden DM, Zuvich RL, Emilsson V, Plump AS, Larson MG, O'Donnell CJ, Yin X, Bobbo M, D'Adamo AP, Iorio A, Sinagra G, Carracedo A, Cummings SR, Nalls MA, Jula A, Kontula KK, Marjamaa A, Oikarinen L, Perola M, Porthan K, Erbel R, Hoffmann P, Jöckel KH, Kälsch H, Nöthen MM; HRGEN Consortium, den Hoed M, Loos RJ, Thelle DS, Gieger C, Meitinger T, Perz S, Peters A, Prucha H, Sinner MF, Waldenberger M, de Boer RA, Franke L, van der Vleuten PA, Beckmann BM, Martens E, Bardai A, Hofman N, Wilde AA, Behr ER, Dalageorgou C, Giudicessi JR, Medeiros-Domingo A, Barc J, Kyndt F, Probst V, Ghidoni A, Insolia R, Hamilton RM, Scherer SW, Brandimarto J, Margulies K, Moravec CE, Greco M FD, Fuchsberger C, O'Connell JR, Lee WK, Watt GC, Campbell H, Wild SH, El Mokhtari NE, Frey N, Asselbergs FW, Mateo Leach I, Navis G, van den Berg MP, van Veldhuisen DJ, Kellis M, Krijthe BP, Franco OH, Hofman A, Kors JA, Uitterlinden AG, Witteman JC, Kedenko L, Lamina C, Oostra BA, Abecasis GR, Lakatta EG, Mulas A, Orrú M, Schlessinger D, Uda M, Markus MR, Völker U, Snieder H, Spector TD, Arnlöv J, Lind L, Sundström J, Syvänen AC, Kivimaki M, Kähönen M, Mononen N, Raitakari OT, Viikari JS, Adamkova V, Kiechl S, Brion M, Nicolaides AN, Paulweber B, Haerting J, Dominiczak AF, Nyberg F, Whincup PH, Hingorani AD, Schott JJ, Bezzina CR, Ingelsson E, Ferrucci L, Gasparini P, Wilson JF, Rudan I, Franke A, Mühleisen TW, Pramstaller PP, Lehtimäki TJ, Paterson AD, Parsa A, Liu Y, van Duijn CM, Siscovick DS, Gudnason V, Jamshidi Y, Salomaa V, Felix SB, Sanna S, **Ritchie MD**, Stricker BH, Stefansson K, Boyer LA, Cappola TP, Olsen JV, Lage K, Schwartz PJ, Kääb S, Chakravarti A, Ackerman MJ, Pfeufer A, de Bakker PI, Newton-Cheh C. Genetic association study of QT interval highlights role for calcium signaling

pathways in myocardial repolarization. *Nat Genet.* 2014 Aug;46(8):826-36. doi: 10.1038/ng.3014. Epub 2014 Jun 22. PMID: 24952745, PMCID: PMC4124521

181. Rasmussen-Torvik LJ, Stallings SC, Gordon AS, Almoguera B, Basford MA, Bielinski SJ, Brautbar A, Brilliant MH, Carrell DS, Connolly JJ, Crosslin DR, Doheny KF, Gallego CJ, Gottesman O, Kim DS, Leppig KA, Li R, Lin S, Manzi S, Mejia AR, Pacheco JA, Pan V, Pathak J, Perry CL, Peterson JF, Prows CA, Ralston J, Rasmussen LV, **Ritchie MD**, Sadhasivam S, Scott SA, Smith M, Vega A, Vinks AA, Volpi S, Wolf WA, Bottinger E, Chisholm RL, Chute CG, Haines JL, Harley JB, Keating B, Holm IA, Kullo IJ, Jarvik GP, Larson EB, Manolio T, McCarty CA, Nickerson DA, Scherer SE, Williams MS, Roden DM, Denny JC. Design and Anticipated Outcomes of the eMERGE-PGx Project: A Multicenter Pilot for Preemptive Pharmacogenomics in Electronic Health Record Systems. *Clin Pharmacol Ther.* 2014 Jun 24. doi: 10.1038/clpt.2014.137. [Epub ahead of print]. PMID: 24960519, PMCID: PMC4169732

182. 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; Cross Consortia Pleiotropy (XC-Pleiotropy) Group; Cohorts for Heart and; Aging Research in Genetic Epidemiology (CHARGE); Genetic Investigation of Anthropometric Traits (GIANT) Consortium; Global Lipids Genetics Consortium (GLGC); Meta-Analyses of Glucose; Insulin-related traits Consortium (MAGIC); Global BPgen (GBPG) Consortium; ADIPOGen Consortium; Women's Genome Health Study (WGHS); Howard University Family Study (HUFS), 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 SL, Loos RJ, Larson MG, Hsu YH, 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):317-38. doi: 10.1016/j.ymgme.2014.04.007. Epub 2014 May 9. PMID: 24981077, PMCID: PMC4122618

183. Crawford DC, Crosslin DR, Tromp G, Kullo IJ, Kuivaniemi H, Hayes MG, Denny JC, Bush WS, Haines JL, Roden DM, McCarty CA, Jarvik GP, **Ritchie MD**. eMERGEing progress in genomics-the first seven years. *Front Genet.* 2014 Jun 17;5:184. doi: 10.3389/fgene.2014.00184. eCollection 2014. PMID: 24987407, PMCID: PMC4060012

184. Kim D, Joung JG, Sohn KA, Shin H, Park YR, **Ritchie MD**, Kim JH. Knowledge boosting: a graph-based integration approach with multi-omics data and genomic knowledge for cancer clinical outcome prediction. *J Am Med Inform Assoc.* 2014 Jul 7. doi: 10.1136/amiajnl-2013-002481. PMID: 25002459, PMCID: PMC4433357

185. 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 Jun 30;7:10. doi: 10.1186/1756-0381-7-10. PMID: 25071867, PMCID: PMC4112852

186. 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 Aug 5;5:250. doi: 10.3389/fgene.2014.00250. PMID: 25177340, PMCID: PMCID: PMC4134007.

187. Kim D, Li R, Dudek SM, Frase AT, Pendergrass SA, **Ritchie MD**. Knowledge-driven genomic interactions: an application in ovarian cancer. *BioData Min.* 2014 Sep 9;7:20. doi: 10.1186/1756-0381-7-20. *eCollection* 2014. PMID: 25214892, PMCID: PMC4161273.

188. Namjou B, Marsolo K, Caroll RJ, Denny JC, **Ritchie MD**, Verma SS, Lingren T, Porollo A, Cobb BL, Perry C, Kottyan LC, Rothenberg ME, Thompson SD, Holm IA, Kohane IS, Harley JB. Phenome-wide association study (PheWAS) in EMR-linked pediatric cohorts, genetically links PLCL1 to speech language development and IL5-IL13 to Eosinophilic Esophagitis. *Front Genet.* 2014 Nov 18;5:401. doi: 10.3389/fgene.2014.00401. *eCollection* 2014. PMID: 25477900, PMCID: PMC4235428.

189. 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 4;10(12):e1004678. doi: 10.1371/journal.pgen.1004678. eCollection 2014 Dec. PMID: 25474351, PMCID: PMC4256091

190. Malinowski JR, Denny JC, Bielinski SJ, Basford MA, Bradford Y, Peissig PL, Carrell D, Crosslin DR, Pathak J, Rasmussen L, Pacheco J, Kho A, Newton KM, Li R, Kullo IJ, Chute CG, Chisholm RL, Jarvik GP, Larson EB, McCarty CA, Masys DR, Roden DM, de Andrade M, **Ritchie MD**, Crawford DC. Genetic Variants Associated with Serum Thyroid Stimulating Hormone (TSH) Levels in European Americans and African Americans from the eMERGE Network. *PLoS One.* 2014 Dec 1;9(12):e111301. doi: 10.1371/journal.pone.0111301. *eCollection* 2014. PMID: 25436638, PMCID: PMC4249871.

191. Crosslin DR, Tromp G, Burt A, Kim DS, Verma SS, Lucas AM, Bradford Y, Crawford DC, Armasu SM, Heit JA, Hayes MG, Kuivaniemi H, **Ritchie MD**, Jarvik GP, de Andrade M; electronic Medical Records and Genomics (eMERGE) Network. Controlling for population structure and genotyping platform bias in the eMERGE multi-institutional biobank linked to electronic health records. *Front Genet.* 2014 Nov 4;5:352. doi: 10.3389/fgene.2014.00352. *eCollection* 2014. PMID: 25414722, PMCID: PMC4220165.

192. Chen HS, Hutter CM, Mechanic LE, Amos CI, Bafna V, Hauser ER, Hernandez RD, Li C, Liberles DA, McAllister K, Moore JH, Paltoo DN, Papanicolaou GJ, Peng B, **Ritchie MD**, Rosenfeld G, Witte JS, Gillanders EM, Feuer EJ. Genetic simulation tools for post-genome wide association studies of complex diseases. *Genet Epidemiol.* 2015 Jan;39(1):11-9. doi: 10.1002/gepi.21870. Epub 2014 Nov 4. PMID: 25371374, PMCID: PMC4270837.

193. **Ritchie MD**, Verma SS, Hall MA, Goodloe RJ, Berg RL, Carrell DS, Carlson CS, Chen L, Crosslin DR, Denny JC, Jarvik G, Li R, Linneman JG, Pathak J, Peissig P, Rasmussen LV, Ramirez AH, Wang X, Wilke RA, Wolf WA, Torstenson ES, Turner SD, McCarty CA. Electronic medical records and genomics (eMERGE) network exploration in cataract: several new potential susceptibility *loci. Mol Vis.* 2014 Sep 19;20:1281-95. eCollection 2014. PMID: 25352737, PMCID: PMC4168835.

194. 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):1017-25. doi: 10.1161/CIRCRESAHA.116.304398. Epub 2014 Oct 17. PMID: 25326128, PMIDC: PMC58174.

195. Crosslin DR, Carrell DS, Burt A, Kim DS, Underwood JG, Hanna DS, Comstock BA, Baldwin E, de Andrade M, Kullo IJ, Tromp G, Kuivaniemi H, Borthwick KM, McCarty CA, Peissig PL, Doheny KF, Pugh E, Kho A, Pacheco J, Hayes MG, **Ritchie MD**, Verma SS, Armstrong G, Stallings S, Denny JC, Carroll RJ, Crawford DC, Crane PK, Mukherjee S, Bottinger E, Li R, Keating B, Mirel DB, Carlson CS, Harley JB, Larson EB, Jarvik GP. Genetic variation in the HLA region is associated with susceptibility to herpes zoster. *Genes Immun.* 2014 Oct 9;0. doi: 10.1038/gene.2014.51. PMID: 25297839, PMCID: PMC4308645.

196. Wood AR, Esko T, Yang J, Vedantam S, Pers TH, Gustafsson S, Chu AY, Estrada K, Luan J, Kutalik Z, Amin N, Buchkovich ML, Croteau-Chonka DC, Day FR, Duan Y, Fall T, Fehrmann R, Ferreira T, Jackson AU, Karjalainen J, Lo KS, Locke AE, Mägi R, Mihailov E, Porcu E,

Randall JC, Scherag A, Vinkhuyzen AA, Westra HJ, Winkler TW, Workalemahu T, Zhao JH, Absher D, Albrecht E, Anderson D, Baron J, Beekman M, Demirkan A, Ehret GB, Feenstra B, Feitosa MF, Fischer K, Fraser RM, Goel A, Gong J, Justice AE, Kanoni S, Kleber ME, Kristiansson K, Lim U, Lotay V, Lui JC, Mangino M, Mateo Leach I, Medina-Gomez C, Nalls MA, Nyholt DR, Palmer CD, Pasko D, Pechlivanis S, Prokopenko I, Ried JS, Ripke S, Shungin D, Stancáková A, Strawbridge RJ, Sung YJ, Tanaka T, Teumer A, Trompet S, van der Laan SW, van Setten J, Van Vliet-Ostaptchouk JV, Wang Z, Yengo L, Zhang W, Afzal U, Arnlöv J, Arscott GM, Bandinelli S, Barrett A, Bellis C, Bennett AJ, Berne C, Blüher M, Bolton JL, Böttcher Y, Boyd HA, Bruinenberg M, Buckley BM, Buyske S, Caspersen IH, Chines PS, Clarke R, Claudi-Boehm S, Cooper M, Daw EW, De Jong PA, Deelen J, Delgado G, Denny JC, Dhonukshe-Rutten R, Dimitriou M, Doney AS, Dörr M, Eklund N, Eury E, Folkersen L, Garcia ME, Geller F, Giedraitis V, Go AS, Grallert H, Grammer TB, Gräßler J, Grönberg H, de Groot LC, Groves CJ, Haessler J, Hall P, Haller T, Hallmans G, Hannemann A, Hartman CA, Hassinen M, Hayward C, Heard-Costa NL, Helmer Q, Hemani G, Henders AK, Hillege HL, Hlatky MA, Hoffmann W, Hoffmann P, Holmen O, Houwing-Duistermaat JJ, Illig T, Isaacs A, James AL, Jeff J, Johansen B, Johansson Å, Jolley J, Juliusdottir T, Junttila J, Kho AN, Kinnunen L, Klopp N, Kocher T, Kratzer W, Lichtner P, Lind L, Lindström J, Lobbens S, Lorentzon M, Lu Y, Lyssenko V, Magnusson PK, Mahajan A, Maillard M, McArdle WL, McKenzie CA, McLachlan S, McLaren PJ, Menni C, Merger S, Milani L, Moayyeri A, Monda KL, Morken MA, Müller G, Müller-Nurasyid M, Musk AW, Narisu N, Nauck M, Nolte IM, Nöthen MM, Oozageer L, Pilz S, Rayner NW, Renstrom F, Robertson NR, Rose LM, Roussel R, Sanna S, Scharnagl H, Scholtens S, Schumacher FR, Schunkert H, Scott RA, Sehmi J, Seufferlein T, Shi J, Silventoinen K, Smit JH, Smith AV, Smolonska J, Stanton AV, Stirrups K, Stott DJ, Stringham HM, Sundström J, Swertz MA, Syvänen AC, Tayo BO, Thorleifsson G, Tyrer JP, van Dijk S, van Schoor NM, van der Velde N, van Heemst D, van Oort FV, Vermeulen SH, Verweij N, Vonk JM, Waite LL, Waldenberger M, Wennauer R, Wilkens LR, Willenborg C, Wilsgaard T, Wojczynski MK, Wong A, Wright AF, Zhang Q, Arveiler D, Bakker SJ, Beilby J, Bergman RN, Bergmann S, Biffar R, Blangero J, Boomsma DI, Bornstein SR, Bovet P, Brambilla P, Brown MJ, Campbell H, Caulfield MJ, Chakravarti A, Collins R, Collins FS, Crawford DC, Cupples LA, Danesh J, de Faire U, den Ruijter HM, Erbel R, Erdmann J, Eriksson JG, Farrall M, Ferrannini E, Ferrières J, Ford I, Forouhi NG, Forrester T, Gansevoort RT, Gejman PV, Gieger C, Golay A, Gottesman O, Gudnason V, Gyllensten U, Haas DW, Hall AS, Harris TB, Hattersley AT, Heath AC, Hengstenberg C, Hicks AA, Hindorff LA, Hingorani AD, Hofman A, Hovingh GK, Humphries SE, Hunt SC, Hypponen E, Jacobs KB, Jarvelin MR, Jousilahti P, Jula AM, Kaprio J, Kastelein JJ, Kayser M, Kee F, Keinanen-Kiukaanniemi SM, Kiemeney LA, Kooner JS, Kooperberg C, Koskinen S, Kovacs P, Kraja AT, Kumari M, Kuusisto J, Lakka TA, Langenberg C, Le Marchand L, Lehtimäki T, Lupoli S, Madden PA, Männistö S, Manunta P, Marette A, Matise TC, McKnight B, Meitinger T, Moll FL, Montgomery GW, Morris AD, Morris AP, Murray JC, Nelis M, Ohlsson C, Oldehinkel AJ, Ong KK, Ouwehand WH, Pasterkamp G, Peters A, Pramstaller PP, Price JF, Qi L, Raitakari OT, Rankinen T, Rao DC, Rice TK, **Ritchie M**, Rudan I, Salomaa V, Samani NJ, Saramies J, Sarzynski MA, Schwarz PE, Sebert S, Sever P, Shuldiner AR, Sinisalo J, Steinthorsdottir V, Stolk RP, Tardif JC, Tönjes A, Tremblay A, Tremoli E, Virtamo J, Vohl MC; Electronic Medical Records and Genomics (eMEMERGEGE) Consortium; MIGen Consortium; PAGEGE Consortium; LifeLines Cohort Study, Amouyel P, Asselbergs FW, Assimes TL, Bochud M, Boehm BO, Boerwinkle E, Bottinger EP, Bouchard C, Cauchi S, Chambers JC, Chanock SJ, Cooper RS, de Bakker PI, Dedoussis G, Ferrucci L, Franks PW, Froguel P, Groop LC, Haiman CA, Hamsten A, Hayes MG, Hui J, Hunter DJ, Hveem K, Jukema JW, Kaplan RC, Kivimaki M, Kuh D, Laakso M, Liu Y, Martin NG, März W, Melbye M, Moebus S, Munroe PB, Njølstad I, Oostra BA, Palmer CN, Pedersen NL, Perola M, Pérusse L, Peters U, Powell JE, Power C, Quertermous T, Rauramaa R, Reinmaa E, Ridker PM, Rivadeneira F, Rotter JI, Saaristo TE, Saleheen D, Schlessinger D, Slagboom PE, Snieder H, Spector TD, Strauch K, Stumvoll M, Tuomilehto J, Uusitupa M, van der Harst P, Völzke H, Walker M, Wareham NJ, Watkins H, Wichmann HE, Wilson JF, Zanen P, Deloukas P, Heid IM, Lindgren CM, Mohlke KL, Speliotes EK, Thorsteinsdottir U, Barroso I, Fox CS, North KE, Strachan DP, Beckmann JS, Berndt SI, Boehnke M, Borecki IB, McCarthy MI, Metspalu A, Stefansson K, Uitterlinden AG, van Duijn CM, Franke L, Willer CJ, Price AL, Lettre G, Loos RJ, Weedon MN, Ingelsson E, O'Connell JR, Abecasis GR, Chasman DI, Goddard ME, Visscher

PM, Hirschhorn JN, Frayling TM. Defining the role of common variation in the genomic and biological architecture of adult human height. *Nat Genet.* 2014 Nov;46(11):1173-86. doi: 10.1038/ng.3097. Epub 2014 Oct 5. PMID: 25282103, PMCID: PMC250049.

197. Restrepo NA, Spencer KL, Goodloe R, Garrett TA, Heiss G, Bůžková P, Jorgensen N, Jensen RA, Matise TC, Hindorff LA, Klein BE, Klein R, Wong TY, Cheng CY, Cornes BK, Tai ES, **Ritchie MD**, Haines JL, Crawford DC. Genetic determinants of age-related macular degeneration in diverse populations from the PAGE study. *Invest Ophthalmol Vis Sci.* 2014 Sep 9;55(10):6839-50. doi: 10.1167/iovs.14-14246. PMID: 25205864, PMCID: PMC4214207.

198. Ng MC, Shriner D, Chen BH, Li J, Chen WM, Guo X, Liu J, Bielinski SJ, Yanek LR, Nalls MA, Comeau ME, Rasmussen-Torvik LJ, Jensen RA, Evans DS, Sun YV, An P, Patel SR, Lu Y, Long J, Armstrong LL, Wagenknecht L, Yang L, Snively BM, Palmer ND, Mudgal P, Langefeld CD, Keene KL, Freedman BI, Mychaleckyj JC, Nayak U, Raffel LJ, Goodarzi MO, Chen YD, Taylor HA Jr, Correa A, Sims M, Couper D, Pankow JS, Boerwinkle E, Adeyemo A, Doumatey A, Chen G, Mathias RA, Vaidya D, Singleton AB, Zonderman AB, Igo RP Jr, Sedor JR; FIND Consortium, Kabagambe EK, Siscovick DS, McKnight B, Rice K, Liu Y, Hsueh WC, Zhao W, Bielak LF, Kraja A, Province MA, Bottinger EP, Gottesman O, Cai Q, Zheng W, Blot WJ, Lowe WL, Pacheco JA, Crawford DC; **eMERGE Consortium**; DIAGRAM Consortium, Grundberg E; MuTHER Consortium, Rich SS, Hayes MG, Shu XO, Loos RJ, Borecki IB, Peyser PA, Cummings SR, Psaty BM, Fornage M, Iyengar SK, Evans MK, Becker DM, Kao WH, Wilson JG, Rotter JI, Sale MM, Liu S, Rotimi CN, Bowden DW; MEta-analysis of type 2 DIabetes in African Americans Consortium. Meta-analysis of genome-wide association studies in African Americans provides insights into the genetic architecture of type 2 diabetes. *PLoS Genet.* 2014 Aug 7;10(8):e1004517. doi: 10.1371/journal.pgen.1004517. eCollection 2014 Aug. PMID: 25102180, PMCID: PMC4125087.

199. Jeff JM, Brown-Gentry K, Goodloe R, **Ritchie MD**, Denny JC, Kho AN, Armstrong LL, McClellan B Jr, Mayo P, Allen M, Jin H, Gillani NB, Schnetz-Boutaud N, Dilks HH, Basford MA, Pacheco JA, Jarvik GP, Chisholm RL, Roden DM, Hayes MG, Crawford DC. Replication of SCN5A Associations with Electrocardio-graphic Traits in African Americans from Clinical and Epidemiologic Studies. *Evol Comput Mach Learn Data Min Bioinform.* 2014;2014:939-951. doi: 10.1007/978-3-662-45523-4_76. PMID: 25590050; PMCID: PMC4290789.

200. Verma SS, de Andrade M, Tromp G, Kuivaniemi H, Pugh E, Namjou-Khales B, Mukherjee S, Jarvik GP, Kottyan LC, Burt A, Bradford Y, Armstrong GD, Derr K, Crawford DC, Haines JL, Li R, Crosslin D, **Ritchie MD**. Imputation and quality control steps for combining multiple genome-wide datasets. Front Genet. 2014 Dec 11;5:370. doi: 10.3389/fgene.2014.00370. *eCollection* 2014. PMID: 25566314, PMCID: PMC4263197.

201. Pendergrass SA, Verma SS, Hall MA, Holzinger ER, Moore CB, Wallace JR, Dudek SM, Huggins W, Kitchner T, Waudby C, Berg R, Mccarty CA, **Ritchie MD**. Next-generation analysis of cataracts: determining knowledge driven gene-gene interactions using biofilter, and gene-environment interactions using the Phenx Toolkit*. *Pac Symp Biocomput.* 2015:495-505. doi: PMID: 25741542

202. Locke AE, Kahali B, Berndt SI, Justice AE, Pers TH, Day FR, Powell C, Vedantam S, Buchkovich ML, Yang J, Croteau-Chonka DC, Esko T, Fall T, Ferreira T, Gustafsson S, Kutalik Z, Luan J, Mägi R, Randall JC, Winkler TW, Wood AR, Workalemahu T, Faul JD, Smith JA, Hua Zhao J, Zhao W, Chen J, Fehrmann R, Hedman ÅK, Karjalainen J, Schmidt EM, Absher D, Amin N, Anderson D, Beekman M, Bolton JL, Bragg-Gresham JL, Buyske S, Demirkan A, Deng G, Ehret GB, Feenstra B, Feitosa MF, Fischer K, Goel A, Gong J, Jackson AU, Kanoni S, Kleber ME, Kristiansson K, Lim U, Lotay V, Mangino M, Mateo Leach I, Medina-Gomez C, Medland SE, Nalls MA, Palmer CD, Pasko D, Pechlivanis S, Peters MJ, Prokopenko I, Shungin D, Stančáková A, Strawbridge RJ, Ju Sung Y, Tanaka T, Teumer A, Trompet S, van der Laan SW, van Setten J, Van Vliet-Ostaptchouk JV, Wang Z, Yengo L, Zhang W, Isaacs A, Albrecht E, Ärnlöv J, Arscott GM, Attwood AP, Bandinelli S, Barrett A, Bas IN, Bellis C, Bennett AJ, Berne C, Blagieva R, Blüher M, Böhringer S, Bonnycastle LL, Böttcher Y, Boyd HA, Bruinenberg M, Caspersen IH, Ida Chen YD, Clarke R, Daw EW, de Craen AJ, Delgado G, Dimitriou M, Doney AS, Eklund N, Estrada K, Eury E, Folkersen L, Fraser RM, Garcia ME,

Geller F, Giedraitis V, Gigante B, Go AS, Golay A, Goodall AH, Gordon SD, Gorski M, Grabe HJ, Grallert H, Grammer TB, Gräßler J, Grönberg H, Groves CJ, Gusto G, Haessler J, Hall P, Haller T, Hallmans G, Hartman CA, Hassinen M, Hayward C, Heard-Costa NL, Helmer Q, Hengstenberg C, Holmen O, Hottenga JJ, James AL, Jeff JM, Johansson Å, Jolley J, Juliusdottir T, Kinnunen L, Koenig W, Koskenvuo M, Kratzer W, Laitinen J, Lamina C, Leander K, Lee NR, Lichtner P, Lind L, Lindström J, Sin Lo K, Lobbens S, Lorbeer R, Lu Y, Mach F, Magnusson PK, Mahajan A, McArdle WL, McLachlan S, Menni C, Merger S, Mihailov E, Milani L, Moayyeri A, Monda KL, Morken MA, Mulas A, Müller G, Müller-Nurasyid M, Musk AW, Nagaraja R, Nöthen MM, Nolte IM, Pilz S, Rayner NW, Renstrom F, Rettig R, Ried JS, Ripke S, Robertson NR, Rose LM, Sanna S, Scharnagl H, Scholtens S, Schumacher FR, Scott WR, Seufferlein T, Shi J, Vernon Smith A, Smolonska J, Stanton AV, Steinthorsdottir V, Stirrups K, Stringham HM, Sundström J, Swertz MA, Swift AJ, Syvänen AC, Tan ST, Tayo BO, Thorand B, Thorleifsson G, Tyrer JP, Uh HW, Vandenput L, Verhulst FC, Vermeulen SH, Verweij N, Vonk JM, Waite LL, Warren HR, Waterworth D, Weedon MN, Wilkens LR, Willenborg C, Wilsgaard T, Wojczynski MK, Wong A, Wright AF, Zhang Q; LifeLines Cohort Study, Brennan EP, Choi M, Dastani Z, Drong AW, Eriksson P, Franco-Cereceda A, Gådin JR, Gharavi AG, Goddard ME, Handsaker RE, Huang J, Karpe F, Kathiresan S, Keildson S, Kiryluk K, Kubo M, Lee JY, Liang L, Lifton RP, Ma B, McCarroll SA, McKnight AJ, Min JL, Moffatt MF, Montgomery GW, Murabito JM, Nicholson G, Nyholt DR, Okada Y, Perry JR, Dorajoo R, Reinmaa E, Salem RM, Sandholm N, Scott RA, Stolk L, Takahashi A, Tanaka T, Van't Hooft FM, Vinkhuyzen AA, Westra HJ, Zheng W, Zondervan KT; ADIPOGen Consortium; AGEN-BMI Working Group; CARDIOGRAMplusC4D Consortium; CKDGen Consortium; GLGC; ICBP; MAGIC Investigators; MuTHER Consortium; MIGen Consortium; PAGE Consortium; ReproGen Consortium; GENIE Consortium; International Endogene Consortium, Heath AC, Arveiler D, Bakker SJ, Beilby J, Bergman RN, Blangero J, Bovet P, Campbell H, Caulfield MJ, Cesana G, Chakravarti A, Chasman DI, Chines PS, Collins FS, Crawford DC, Cupples LA, Cusi D, Danesh J, de Faire U, den Ruijter HM, Dominiczak AF, Erbel R, Erdmann J, Eriksson JG, Farrall M, Felix SB, Ferrannini E, Ferrières J, Ford I, Forouhi NG, Forrester T, Franco OH, Gansevoort RT, Gejman PV, Gieger C, Gottesman O, Gudnason V, Gyllensten U, Hall AS, Harris TB, Hattersley AT, Hicks AA, Hindorff LA, Hingorani AD, Hofman A, Homuth G, Hovingh GK, Humphries SE, Hunt SC, Hyppönen E, Illig T, Jacobs KB, Jarvelin MR, Jöckel KH, Johansen B, Jousilahti P, Jukema JW, Jula AM, Kaprio J, Kastelein JJ, Keinanen-Kiukaanniemi SM, Kiemeney LA, Knekt P, Kooner JS, Kooperberg C, Kovacs P, Kraja AT, Kumari M, Kuusisto J, Lakka TA, Langenberg C, Le Marchand L, Lehtimäki T, Lyssenko V, Männistö S, Marette A, Matise TC, McKenzie CA, McKnight B, Moll FL, Morris AD, Morris AP, Murray JC, Nelis M, Ohlsson C, Oldehinkel AJ, Ong KK, Madden PA, Pasterkamp G, Peden JF, Peters A, Postma DS, Pramstaller PP, Price JF, Qi L, Raitakari OT, Rankinen T, Rao DC, Rice TK, Ridker PM, Rioux JD, **Ritchie MD**, Rudan I, Salomaa V, Samani NJ, Saramies J, Sarzynski MA, Schunkert H, Schwarz PE, Sever P, Shuldiner AR, Sinisalo J, Stolk RP, Strauch K, Tönjes A, Trégouët DA, Tremblay A, Tremoli E, Virtamo J, Vohl MC, Völker U, Waeber G, Willemsen G, Witteman JC, Zillikens MC, Adair LS, Amouyel P, Asselbergs FW, Assimes TL, Bochud M, Boehm BO, Boerwinkle E, Bornstein SR, Bottinger EP, Bouchard C, Cauchi S, Chambers JC, Chanock SJ, Cooper RS, de Bakker PI, Dedoussis G, Ferrucci L, Franks PW, Froguel P, Groop LC, Haiman CA, Hamsten A, Hui J, Hunter DJ, Hveem K, Kaplan RC, Kivimaki M, Kuh D, Laakso M, Liu Y, Martin NG, März W, Melbye M, Metspalu A, Moebus S, Munroe PB, Njølstad I, Oostra BA, Palmer CN, Pedersen NL, Perola M, Pérusse L, Peters U, Power C, Quertermous T, Rauramaa R, Rivadeneira F, Saaristo TE, Saleheen D, Sattar N, Schadt EE, Schlessinger D, Slagboom PE, Snieder H, Spector TD, Thorsteinsdottir U, Stumvoll M, Tuomilehto J, Uitterlinden AG, Uusitupa M, van der Harst P, Walker M, Wallaschofski H, Wareham NJ, Watkins H, Weir DR, Wichmann HE, Wilson JF, Zanen P, Borecki IB, Deloukas P, Fox CS, Heid IM, O'Connell JR, Strachan DP, Stefansson K, van Duijn CM, Abecasis GR, Franke L, Frayling TM, McCarthy MI, Visscher PM, Scherag A, Willer CJ, Boehnke M, Mohlke KL, Lindgren CM, Beckmann JS, Barroso I, North KE, Ingelsson E, Hirschhorn JN, Loos RJ, Speliotes EK. Genetic studies of body mass index yield new insights for obesity biology. *Nature.* 2015 Feb 12;518(7538):197-206. doi: 10.1038/nature14177. PMID: 25673413, PMCID: PMC4382211.

203. Shungin D, Winkler TW, Croteau-Chonka DC, Ferreira T, Locke AE, Mägi R, Strawbridge RJ, Pers TH, Fischer K, Justice AE, Workalemahu T, Wu JM, Buchkovich ML, Heard-Costa NL, Roman TS, Drong AW, Song C, Gustafsson S, Day FR, Esko T, Fall T, Kutalik Z, Luan J, Randall JC, Scherag A, Vedantam S, Wood AR, Chen J, Fehrmann R, Karjalainen J, Kahali B, Liu CT, Schmidt EM, Absher D, Amin N, Anderson D, Beekman M, Bragg-Gresham JL, Buyske S, Demirkan A, Ehret GB, Feitosa MF, Goel A, Jackson AU, Johnson T, Kleber ME, Kristiansson K, Mangino M, Mateo Leach I, Medina-Gomez C, Palmer CD, Pasko D, Pechlivanis S, Peters MJ, Prokopenko I, Stančáková A, Ju Sung Y, Tanaka T, Teumer A, Van Vliet-Ostaptchouk JV, Yengo L, Zhang W, Albrecht E, Ärnlöv J, Arscott GM, Bandinelli S, Barrett A, Bellis C, Bennett AJ, Berne C, Blüher M, Böhringer S, Bonnet F, Böttcher Y, Bruinenberg M, Carba DB, Caspersen IH, Clarke R, Daw EW, Deelen J, Deelman E, Delgado G, Doney AS, Eklund N, Erdos MR, Estrada K, Eury E, Friedrich N, Garcia ME, Giedraitis V, Gigante B, Go AS, Golay A, Grallert H, Grammer TB, Gräßler J, Grewal J, Groves CJ, Haller T, Hallmans G, Hartman CA, Hassinen M, Hayward C, Heikkilä K, Herzig KH, Helmer Q, Hillege HL, Holmen O, Hunt SC, Isaacs A, Ittermann T, James AL, Johansson I, Juliusdottir T, Kalafati IP, Kinnunen L, Koenig W, Kooner IK, Kratzer W, Lamina C, Leander K, Lee NR, Lichtner P, Lind L, Lindström J, Lobbens S, Lorentzon M, Mach F, Magnusson PK, Mahajan A, McArdle WL, Menni C, Merger S, Mihailov E, Milani L, Mills R, Moayyeri A, Monda KL, Mooijaart SP, Mühleisen TW, Mulas A, Müller G, Müller-Nurasyid M, Nagaraja R, Nalls MA, Narisu N, Glorioso N, Nolte IM, Olden M, Rayner NW, Renstrom F, Ried JS, Robertson NR, Rose LM, Sanna S, Scharnagl H, Scholtens S, Sennblad B, Seufferlein T, Sitlani CM, Vernon Smith A, Stirrups K, Stringham HM, Sundström J, Swertz MA, Swift AJ, Syvänen AC, Tayo BO, Thorand B, Thorleifsson G, Tomaschitz A, Troffa C, van Oort FV, Verweij N, Vonk JM, Waite LL, Wennauer R, Wilsgaard T, Wojczynski MK, Wong A, Zhang Q, Hua Zhao J, Brennan EP, Choi M, Eriksson P, Folkersen L, Franco-Cereceda A, Gharavi AG, Hedman ÅK, Hivert MF, Huang J, Kanoni S, Karpe F, Keildson S, Kiryluk K, Liang L, Lifton RP, Ma B, McKnight AJ, McPherson R, Metspalu A, Min JL, Moffatt MF, Montgomery GW, Murabito JM, Nicholson G, Nyholt DR, Olsson C, Perry JR, Reinmaa E, Salem RM, Sandholm N, Schadt EE, Scott RA, Stolk L, Vallejo EE, Westra HJ, Zondervan KT; ADIPOGen Consortium; CARDIOGRAMplusC4D Consortium; CKDGen Consortium; GEFOS Consortium; GENIE Consortium; GLGC; ICBP; International Endogene Consortium; LifeLines Cohort Study; MAGIC Investigators; MuTHER Consortium; PAGE Consortium; ReproGen Consortium, Amouyel P, Arveiler D, Bakker SJ, Beilby J, Bergman RN, Blangero J, Brown MJ, Burnier M, Campbell H, Chakravarti A, Chines PS, Claudi-Boehm S, Collins FS, Crawford DC, Danesh J, de Faire U, de Geus EJ, Dörr M, Erbel R, Eriksson JG, Farrall M, Ferrannini E, Ferrières J, Forouhi NG, Forrester T, Franco OH, Gansevoort RT, Gieger C, Gudnason V, Haiman CA, Harris TB, Hattersley AT, Heliövaara M, Hicks AA, Hingorani AD, Hoffmann W, Hofman A, Homuth G, Humphries SE, Hyppönen E, Illig T, Jarvelin MR, Johansen B, Jousilahti P, Jula AM, Kaprio J, Kee F, Keinanen-Kiukaanniemi SM, Kooner JS, Kooperberg C, Kovacs P, Kraja AT, Kumari M, Kuulasmaa K, Kuusisto J, Lakka TA, Langenberg C, Le Marchand L, Lehtimäki T, Lyssenko V, Männistö S, Marette A, Matise TC, McKenzie CA, McKnight B, Musk AW, Möhlenkamp S, Morris AD, Nelis M, Ohlsson C, Oldehinkel AJ, Ong KK, Palmer LJ, Penninx BW, Peters A, Pramstaller PP, Raitakari OT, Rankinen T, Rao DC, Rice TK, Ridker PM, **Ritchie MD**, Rudan I, Salomaa V, Samani NJ, Saramies J, Sarzynski MA, Schwarz PE, Shuldiner AR, Staessen JA, Steinthorsdottir V, Stolk RP, Strauch K, Tönjes A, Tremblay A, Tremoli E, Vohl MC, Völker U, Vollenweider P, Wilson JF, Witteman JC, Adair LS, Bochud M, Boehm BO, Bornstein SR, Bouchard C, Cauchi S, Caulfield MJ, Chambers JC, Chasman DI, Cooper RS, Dedoussis G, Ferrucci L, Froguel P, Grabe HJ, Hamsten A, Hui J, Hveem K, Jöckel KH, Kivimaki M, Kuh D, Laakso M, Liu Y, März W, Munroe PB, Njølstad I, Oostra BA, Palmer CN, Pedersen NL, Perola M, Pérusse L, Peters U, Power C, Quertermous T, Rauramaa R, Rivadeneira F, Saaristo TE, Saleheen D, Sinisalo J, Slagboom PE, Snieder H, Spector TD, Thorsteinsdottir U, Stumvoll M, Tuomilehto J, Uitterlinden AG, Uusitupa M, van der Harst P, Veronesi G, Walker M, Wareham NJ, Watkins H, Wichmann HE, Abecasis GR, Assimes TL, Berndt SI, Boehnke M, Borecki IB, Deloukas P, Franke L, Frayling TM, Groop LC, Hunter DJ, Kaplan RC, O'Connell JR, Qi L, Schlessinger D, Strachan DP, Stefansson K, van Duijn CM, Willer CJ, Visscher PM, Yang J, Hirschhorn JN, Zillikens MC, McCarthy MI, Speliotes EK, North KE, Fox CS, Barroso I, Franks PW, Ingelsson E, Heid IM, Loos RJ, Cupples LA, Morris

AP, Lindgren CM, Mohlke KL. New genetic loci link adipose and insulin biology to body fat distribution. *Nature.* 2015 Feb 12;518(7538):187-96. doi: 10.1038/nature14132. PMID: 25673412, PMCID: PMC4338562.

204. Kim D, Li R, Dudek SM, Wallace JR, **Ritchie MD**. Binning somatic mutations based on biological knowledge for predicting survival: an application in renal cell carcinoma. *Pac Symp Biocomput*. 2015:96-107. PMID: 25592572, PMCID: PMC4299944.

205. **Ritchie MD**, de Andrade M, Kuivaniemi H. The foundation of precision medicine: integration of electronic health records with genomics through basic, clinical, and translational research. Front Genet. 2015 Mar 17;6:104. doi: 10.3389/fgene.2015.00104. *eCollection* 2015. PMID: 25852745, PMCID: PMC4362332.

206. 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 Anaylsis of Age-Related Cataract in the eMERGE Network. *Genet Epidemiol*. 2015 Jul;39(5):387-84. doi: 10.1002/gepi.21902. Epub 2015 May 17. PMID: 25982363, PMCID: PMC4550090

207. 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 9;2(1):ofu113. doi: 10.1093/ofid/ofu113. eCollection 2015 Jan. PMID: 25884002, PMCID: PMC4396430.

208. Crosslin DR, Robertson PD, Carrell DS, Gordon AS, Hanna DS, Burt A, Fullerton SM, Scrol A, Ralston J, Leppig K, Hartzler A, Baldwin E, Andrade Md, Kullo IJ, Tromp G, Doheny KF, **Ritchie MD**, Crane PK, Nickerson DA, Larson EB, Jarvik GP. Prospective participant selection and ranking to maximize actionable pharmacogenetic variants and discovery in the eMERGE Network. *Genome Med.* 2015 Jul 3;7(1):67. doi: 10.1186/s 13073-015-0181-z. eCollection 2015. PMID: 26221186, PMCID: PMC4517371

209. 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):137-46. Doi: 10.1159/000381805. Epub 2015 Jul 28. PMID: 26201699, PMCID: PMC4528966

210. 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):111-23. Doi: 10.1159/000381851. Epub 2015 Jul 28. PMID: 26201697

211. Mosley JD, Shaffer CM, Van Driest SL, Weeke PE, Wells QS, Karnes JH, Velez Edwards DR, Wei WQ, Teixeira PL, Bastarache L, Crawford DC, Li R, Manolio TA, Bottinger EP, McCarty CA, Linneman JG, Brilliant MH, Pacheco JA, Thompson W, Chisholm RL, Jarvik GP, Crosslin DR, Carrell DS, Baldwin E, Ralston J, Larson EB, Grafton J, Scrol A, Jouni H, Kullo IJ, Tromp G, Borthwick KM, Kuivaniemi H, Carey DJ, **Ritchie MD**, Bradford Y, Verma SS, Chute CG, Veluchamy A, Siddiqui MK, Palmer CN, Doney A, MahmoudPour SH, Maitland-van der Zee AH, Morris AD, Denny JC, Roden DM. A genome-wide association study identifies variants in KCNIP4 associated with ACE inhibitor-induced cough. *Pharmacogenomics j*. 2015 Jul 14. doi: 10.1038/tpj.2015.51. [Epub ahead of print] PMID: 26169577, PMCID: PMC4713364

212. Freitag DF, Butterworth AS, Willeit P, Howson JM, Burgess S, Kaptoge S, Young R, Ho WK, Wood AM, Sweeting M, Spackman S, Staley JR, Ramond A, Harshfield E, Nielsen SF, Grande P, Lange LA, Bown MJ, Jones GT, Scott RA, Bevan S, Porcu E, Thorleifsson G, Zeng L, Kessler T, Do R, Nikpay M, Zhang W, Hopewell JC, Kleber M, Delgado GE, Nelson CP, Goel A, Bis JC, Dehghan A, Ligthart S, Smith AV, Qu L, van 't Hof FN, de Bakker PI, Baas AF, van Rij A, Tromp G, Kuivaniemi H, Ritchie MD, Verma SS, Crawford DC, Malinowski J, de Andrade M, Kullo IJ, Peissig PL, McCarty CA, Böttinger EP, Gottesman O, Crosslin DR, Carrell DS, Rasmussen-Torvik LJ, Pacheco JA, Huang J, Timpson NJ, Kettunen J, Ala-Korpela

M, Mitchell GF, Parsa A, Wilkinson IB, Gorski M, Li Y, Franceschini N, Keller MF, Ganesh SK, Langefeld CD, Bruijn L, Brown MA, Evans DM, Baltic S, Ferreira MA, Baurecht H, Weidinger S, Franke A, Lubitz SA, Müller-Nurasyid M, Felix JF, Smith NL, Sudman M, Thompson SD, Zeggini E, Panoutsopoulou K, Nalls MA, Singleton A, Polychronakos C, Bradfield JP, Hakonarson H, Easton DF, Thompson D, Tomlinson IP, Dunlop M, Hemminki K, Morgan G, Eisen T, Goldschmidt H, Allan JM, Henrion M, Whiffin N, Wang Y, Chubb D, Houlston RS, Iles MM, Bishop DT, Law MH, Hayward NK, Luo Y, Nejentsev S, Barbalic M, Crossman D, Sanna S, Soranzo N, Markus HS, Wareham NJ, Rader DJ, Reilly M, Assimes T, Harris TB, Hofman A, Franco OH, Gudnason V, Tracy R, Psaty BM, Farrall M, Watkins H, Hall AS, Samani NJ, März W, Clarke R, Collins R, Kooner JS, Chambers JC, Kathiresan S, McPherson R, Erdmann J, Kastrati A, Schunkert H, Stefánsson K, Walston JD, Tybjærg-Hansen A, Alam DS, Majumder AA, Di Angelantonio E, Chowdhury R, Nordestgaard BG, Saleheen D, Thompson SG, Danesh J. Cardiometabolic effects of genetic upregulation of the interleukin 1 receptor antagonist: a Mendelian randomisation analysis. *Lancet Diabetes Endocrinol*. 2015 Apr;3(4):243-53. doi: 10.1016/S2213-8587(15)00034-0. Epub 2015 Feb 26. PMID: 25726324 PMCID: PMC4648058

213. Kim D, Li R, Dudek SM, **Ritchie MD**. Predicting censored survival data based on the interactions between meta-dimensional omics data in breast cancer. *J Biomed Inform*. 2015 Aug;56:220-8. doi: 10.1016/j.jbi.2015.05.019. PMID: 26048077, PMCID: PMC4550096

214. Pendergrass SA, **Ritchie MD**. Phenome-Wide Association Studies: Leveraging Comprehensive Phenotypic and Genotypic Data for Discovery. *Curr Genet Med Rep*. 2015 Jun 1;3(2):92-100. PMID: 26146598, PMCID: PMC4489156

215. Namjou B, Marsolo K, Lingren T, **Ritchie MD**, Verma SS, Cobb BL, Perry C, Kitchner TE, Brilliant MH, Peissig PL, Borthwick KM, Williams MS, Grafton J, Jarvik GP, Holm IA, Harley JB. A GWAS Study on Liver Function Test Using eMERGE Network Participants. *PLoS One.* 2015 Sep 28;10(9):e0138677. doi: 10.1371/journal.pone.0138677. PMID: 2641371, PMCID: PMC4586138

216. Li YR, van Setten J, Verma SS, Lu Y, Holmes MV, Gao H, Lek M, Nair N, Chandrupatla H, Chang B, Karczewski KJ, Wong C, Mohebnasab M, Mukhtar E, Phillips R, Tragante V, Hou C, Steel L, Lee T, Garifallou J, Guettouche T, Cao H, Guan W, Himes A, van Houten J, Pasquier A, Yu R, Carrigan E, Miller MB, Schladt D, Akdere A, Gonzalez A, Llyod KM, McGinn D, Gangasani A, Michaud Z, Colasacco A, Snyder J, Thomas K, Wang T, Wu B, Alzahrani AJ, Al-Ali AK, Al-Muhanna FA, Al-Rubaish AM, Al-Mueilo S, Monos DS, Murphy B, Olthoff KM, Wijmenga C, Webster T, Kamoun M, Balasubramanian S, Lanktree MB, Oetting WS, Garcia-Pavia P, MacArthur DG, de Bakker PI, Hakonarson H, Birdwell KA, Jacobson PA, **Ritchie MD**, Asselbergs FW, Israni AK, Shaked A, Keating BJ. Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. *Genome Med*. 2015 Oct 1;7:90. doi: 10.1186/s13073-015-0211-x. PMID: 26423053, PMCID: PMC4589899

217. Keating BJ, van Setten J, Jacobson PA, Holmes MV, Verma SS, Chandrupatla HR, Nair N, Gao H, Li YR, Chang BL, Wong C, Phillips R, Cole BS, Mukhtar E, Zhang W, Cao H, Mohebnasab M, Hou C, Lee T, Steel L, Shaked O, Garifallou J, Miller MB, Karczewski KJ, Akdere A, Gonzalez A, Lloyd KM, McGinn D, Michaud Z, Colasacco A, Lek M, Fu Y, Pawashe M, Guettouche T, Himes A, Perez L, Guan W, Wu B, Schladt D, Menon M, Zhang Z, Tragante V, de Jonge N, Otten HG, de Weger RA, van de Graaf EA, Baan CC, Manintveld OC, De Vlaminck I, Piening BD, Strehl C, Shaw M, Snieder H, Klintmalm GB, O'Leary JG, Amaral S, Goldfarb S, Rand E, Rossano JW, Kohli U, Heeger P, Stahl E, Christie JD, Fuentes MH, Levine JE, Aplenc R, Schadt EE, Stranger BE, Kluin J, Potena L, Zuckermann A, Khush K, Alzahrani AJ, Al-Muhanna FA, Al-Ali AK, Al-Ali R, Al-Rubaish AM, Al-Mueilo S, Byrne EM, Miller D, Alexander SI, Onengut-Gumuscu S, Rich SS, Suthanthiran M, Tedesco H, Saw CL, Ragoussis J, Kfoury AG, Horne B, Carlquist J, Gerstein MB, Reindl-Schwaighofer R, Oberbauer R, Wijmenga C, Palmer S, Pereira AC, Segovia J, Alonso-Pulpon LA, Comez-Bueno M, Vilches C, Jaramillo N, de Borst MH, Naesens M, Hao K, MacArthur DG, Balasubramanian S, Conlon PJ, Lord GM, **Ritchie MD**, Snyder M, Olthoff KM, Moore JH, Petersdorf EW, Kamoun M, Wang J, Monos DS, de Bakker PI, Hakonarson H, Murphy B, Lankree MB, Garcia-Pavia P, Oetting WS,

Birdwell KA, Bakker SJ, Israni AK, Shaked A, Asselbergs FW. Design and Implementation of the International Genetics and Translational Research in Transplantation Network. *Transplantation.* 2015 Nov;99(11):2401-12. doi: 10.1097/TP.0000000000000913. PMID: 26479416, PMCID: PMC4623847

218. De R, Verma SS, Drenos F, Holzinger ER, Holmes MV, Hall MA, Crosslin DR, Carrell DS, Hakonarson H, Jarvik G, Larson E, Pacheco JA, Rasmussen-Torvik LJ, Moore CB, Asselbergs FW, Moore JH, Ritchie MD, Keating BJ, Gilbert-Diamond D. Identifying gene-gene interactions that are highly associated with Body Mass Index using Quantitative Multifactor Dimensionality Reduction (QMDR). *BioData Min.* 2015 Dec 14;8:41. doi: 10.1186/s13040-015-0074-0. PMID: 26674805, PMCID: PMC4678717

219. Van Driest SL, Wells QS, Stallings S, Bush WS, Gordon A, Nickerson DA, Kim JH, Crosslin DR, Jarvik GP, Carrell DS, Ralston JD, Larson EB, Bielinski SJ, Olson JE, Ye Z, Kullo IJ, Abul-Husn NS, Scott SA, Bottinger E, Almoguera B, Connolly J, Chiavacci R, Hakonarson H, Rasmussen-Torvik LJ, Pan V, Persell SD, Smith M, Chisholm RL, Kitchner TE, He MM, Brilliant MH, Wallace JR, Doheny KF, Shoemaker MB, Li R, Manolio TA, Callis TE, Macaya D, Williams MS, Carey D, Kapplinger JD, Ackerman MJ, **Ritchie MD**, Denny JC, Roden DM. Association of Arrhythmia-Related Genetic Variants With Phenotypes Documented in Electronic Medical Records. *JAMA*. 2016 Jan 5;315(1):47-57. doi: 10.1001/jama.2015.17701. PMID: 26746457, PMCID: PMC4758131

220. 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.* 2016;21:57-68. PMID: 26776173, PMCID: PMC4722952

221. Verma A, Leader JB, Verma SS, Frase A, Wallace J, Dudek S, Lavage DR, Van Hout CV, Dewey FE, Penn J, Lopez A, Overton JD, Carey DJ, Ledbetter DH, Kirchner HL, **Ritchie MD**, Pendergrass SA. Integrating Clinical Laboratory Measures and ICD-9 Code Diagnoses in Phenome-Wide Association Studies. *Pac Symp Biocomput.* 2016;21:168-79. PMID: 26776183, PMCID: PMCID4718547

222. Basile AO, Wallace JR, Peissig P, McCarty CA, Brilliant M, **Ritchie MD**. Knowledge Driven Binning and PheWAS Analysis in Marshfield Personalized Medicine Research Project Using BioBin. *Pac Symp Biocomput.* 2016;21:249-60. PMID: 26776191, PMCID: PMC4824557

223. Kim D, Lucas A, Glessner J, Verma SS, Bradford Y, Li R, Frase AT, Hakonarson H, Peissig P, Brilliant M, **Ritchie MD**. Biofilter as a Functional Annotation Pipeline for Common and Rare Copy Number Burden. *Pac Symp Biocomput*. 2016;21:357-68. PMID: 26776200, PMCID: PMC4722964

224. Hohman TJ, Bush WS, Jiang L, Brown-Gentry KD, Torstenson ES, Dudek SM, Mukherjee S, Naj A, Kunkle BW, **Ritchie MD**, Martin ER, Schellenberg GD, Mayeux R, Farrer LA, Pericak-Vance MA, Haines JL, Thornton-Wells TA; Alzheimer's Disease Genetics Consortium. Discovery of gene-gene interactions across multiple independent data sets of late onset Alzheimer disease from the Alzheimer Disease Genetics Consortium. *Neurobiol Aging*. 2016 Feb;38:141-50. doi: 10.1016/j.neurobiolaging.2015.10.031. PMID: 26827652, PMCID: PMC4735733

225. Bush WS, Crosslin DR, Owusu-Obeng A, Wallace J, Almoguera B, Basford MA, Bielinski SJ, Carrell DS, Connolly JJ, Crawford D, Doheny KF, Gallego CJ, Gordon AS, Keating B, Kirby J, Kitchner T, Manzi S, Mejia AR, Pan V, Perry CL, Peterson JF, Prows CA, Ralston J, Scott SA, Scrol A, Smith M, Stallings SC, Veldhuizen T, Wolf W, Volpi S, Wiley K, Li R, Manolio T, Bottinger E, Brilliant MH, Carey D, Chisholm RL, Chute CG, Haines JL, Hakonarson H, Harley JB, Holm IA, Kullo IJ, Jarvik GP, Larson EB, McCarty CA, Williams MS, Denny JC, Rasmussen-Torvik LJ, Roden DM, **Ritchie MD**. Genetic variation among 82 pharmacogenes: The PGRNseq data from the eMERGE network. *Clin Pharmacol Ther*. 2016 Aug;100(2):160-9. doi: 10.1002/cpt.350. PMID: 26857349, PMCID: PMC5010878

226. Simonti CN, Vernot B, Bastarache L, Bottinger E, Carrell DS, Chisholm RL, Crosslin DR, Hebbring SJ, Jarvik GP, Kullo IJ, Li R, Pathak J, **Ritchie MD**, Roden DM, Verma SS, Tromp G,

Prato JD, Bush WS, Akey JM, Denny JC, Capra JA. The phenotypic legacy of admixture between modern humans and Neandertals. *Science*. 2016 Feb 12;351(6274):737-41. doi: 10.1126/science.aad2149. PMID: 26912863, PMCID: PMC4849557

227. Dewey FE, Gusarova V, O'Dushlaine C, Gottesman O, Trejos J, Hunt C, Van Hout CV, Habegger L, Buckler D, Lai KM, Leader JB, Murray MF, **Ritchie MD**, Kirchner HL, Ledbetter DH, Penn J, Lopez A, Borecki IB, Overton JD, Reid JG, Carey DJ, Murphy AJ, Yancopoulos GD, Baras A, Gromada J, Shuldiner AR. Inactivating Variants in ANGPTL4 and Risk of Coronary Artery Disease. *N Engl J Med*. 2016 Mar 24;374(12):1123-33. doi: 10.1056/NEJMoa1510926. PMID: 26933753, PMCID: PMC4900689

228. Williams MS, **Ritchie MD**, Payne PR. Interdisciplinary training to build an informatics workforce for precision medicine. *Appl Transl Genom*. 2015 Aug 4;6:28-30. doi: 10.1016/j.atg.2015.07.003. PMID: 27054076, PMCID: PMC4803783

229. 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. *Bioinformatics*. 2016 Aug 1;32(15):2361-3. doi: 10.1093/bioinformatics/btw130. PMID: 27153576, PMCID: PMC4965631

230. Li R, Dudek SM, Kim D, Hall MA, Bradford Y, Peissig PL, Brilliant MH, Linneman JG, McCarty CA, Bao L, **Ritchie MD**. Identification of genetic interaction networks via an evolutionary algorithm evolved Bayesian network. *BioData Min*. 2016 May 10;9:18. doi: 10.1186/s13040-016-0094-4. PMID: 27168765, PMCID: PMC4862166

231. 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;17(8):853-66. doi: 10.2217/pgs-2015-0007. PMID: 27249515, PMCID: PMC5352965

232. Hall MA, Moore JH, **Ritchie MD**. Embracing Complex Associations in Common Traits: Critical Considerations for Precision Medicine. *Trends Genet*. 2016 Aug;32(8):470-84. doi: 10.1016/j.tig.2016.06.001. Review. PMID: 27392675

233. van 't Hof FN, Ruigrok YM, Lee CH, Ripke S, Anderson G, de Andrade M, Baas AF, Blankensteijn JD, Böttinger EP, Bown MJ, Broderick J, Bijlenga P, Carrell DS, Crawford DC, Crosslin DR, Ebeling C, Eriksson JG, Fornage M, Foroud T, von Und Zu Fraunberg M, Friedrich CM, Gaál EI, Gottesman O, Guo DC, Harrison SC, Hernesniemi J, Hofman A, Inoue I, Jääskeläinen JE, Jones GT, Kiemeney LA, Kivisaari R, Ko N, Koskinen S, Kubo M, Kullo IJ, Kuivaniemi H, Kurki MI, Laakso A, Lai D, Leal SM, Lehto H, LeMaire SA, Low SK, Malinowski J, McCarty CA, Milewicz DM, Mosley TH, Nakamura Y, Nakaoka H, Niemelä M, Pacheco J, Peissig PL, Pera J, Rasmussen-Torvik L, **Ritchie MD**, Rivadeneira F, van Rij AM, Santos-Cortez RL, Saratzis A, Slowik A, Takahashi A, Tromp G, Uitterlinden AG, Verma SS, Vermeulen SH, Wang GT; Aneurysm Consortium; Vascular Research Consortium of New Zealand., Han B, Rinkel GJ, de Bakker PI. Shared Genetic Risk Factors of Intracranial, Abdominal, and Thoracic Aneurysms. *J Am Heart Assoc*. 2016 Jul 14;5(7). pii: e002603. doi: 10.1161/JAHA.115.002603. PMID: 2741816, PMCID: PMC5015357

234. Verma A, Basile AO, Bradford Y, Kuivaniemi H, Tromp G, Carey D, Gerhard GS, Crowe JE Jr, **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 Aug 10;11(8):e0160573. doi: 10.1371/journal.pone.0160573. PMID: 27508393, PMCID: PMC4980020

235. 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 Aug 12;9 Suppl 1:32. doi: 10.1186/s12920-016-0191-8. PMID: 27535653, PMCID: PMC4989894

236. Moore CC, Basile AO, Wallace JR, Frase AT, **Ritchie MD**. A biologically informed method for detecting rare variant associations. *BioData Min*. 2016 Aug 30;9(1):27. doi: 10.1186/s13040-016-0107-3. PMID: 27582876, PMCID: PMC5006419

237. Verma SS, Cooke Bailey JN, Lucas A, Bradford Y, Linneman JG, Hauser MA, Pasquale LR, Peissig PL, Brilliant MH, McCarty CA, Haines JL, Wiggs JL, Vrabec TR, Tromp G, **Ritchie MD**; eMERGE Network.; NEIGHBOR Consortium. Epistatic Gene-Based Interaction Analyses for Glaucoma in eMERGE and NEIGHBOR Consortium. *PLoS Genet*. 2016 Sep 13;12(9):e1006186. doi: 10.1371/journal.pgen.1006186. PMID: 27623284, PMCID: PMC5021356

238. De R, Verma SS, Holzinger E, Hall M, Burt A, Carrell DS, Crosslin DR, Jarvik GP, Kuivaniemi H, Kullo IJ, Lange LA, Lanktree MB, Larson EB, North KE, Reiner AP, Tragante V, Tromp G, Wilson JG, Asselbergs FW, Drenos F, Moore JH, **Ritchie MD**, Keating B, Gilbert-Diamond D. Identifying gene-gene interactions that are highly associated with four quantitative lipid traits across multiple cohorts. *Hum Genet*. 2017 Feb;136(2):165-178. doi: 10.1007/s00439-016-1738-7. PMID: 27848076

239. Yoneyama S, Yao J, Guo X, Fernandez-Rhodes L, Lim U, Boston J, Buzková P, Carlson CS, Cheng I, Cochran B, Cooper R, Ehret G, Fornage M, Gong J, Gross M, Gu CC, Haessler J, Haiman CA, Henderson B, Hindorff LA, Houston D, Irvin MR, Jackson R, Kuller L, Leppert M, Lewis CE, Li R, Le Marchand L, Matise TC, Nguyen KD, Chakravarti A, Pankow JS, Pankratz N, Pooler L, **Ritchie MD**, Bien SA, Wassel CL, Chen YD, Taylor KD, Allison M, Rotter JI, Schreiner PJ, Schumacher F, Wilkens L, Boerwinkle E, Kooperberg C, Peters U, Buyske S, Graff M, North KE. Generalization and fine mapping of European ancestry-based central adiposity variants in African ancestry populations. *Int J Obes (Lond)*. 2017 Feb;41(2):324-331. doi: 10.1038/ijo.2016.207. PMID: 27867202, PMCID: PMC5296276

240. 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*. 2016;22:533-544. PMID: 27897004

241. Jones GT, Tromp G, Kuivaniemi H, Gretarsdottir S, Baas AF, Giusti B, Strauss E, Van't Hof FN, Webb TR, 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 KM, Smelser DT, Crosslin DR, de Andrade M, Ryer EJ, McCarty CA, Böttinger 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 AR, Bumpstead S, Badger SA, Clough RE, Cockerill G, Hafez H, Scott DJ, Futers TS, Romaine SP, Bridge K, Griffin KJ, Bailey MA, Smith A, Thompson MM, van Bockxmeer FM, Matthiasson SE, Thorleifsson G, Thorsteinsdottir U, Blankensteijn JD, Teijink JA, Wijmenga C, de Graaf J, Kiemeney LA, Lindholt JS, Hughes A, 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 EE, Björkegren JL, Lipovich L, Drolet AM, Verhoeven EL, Zeebregts CJ, Geelkerken RH, van Sambeek MR, van Sterkenburg SM, de Vries JP, Stefansson K, Thompson JR, de Bakker PI, Deloukas P, Sayers RD, Harrison SC, 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*. 2017 Jan 20;120(2):341-353. doi: 10.1161/CIRCRESAHA.116.308765. PMID: 27899403, PMCID: PMC5253231

242. Schmidt AF, Swerdlow DI, Holmes MV, Patel RS, Fairhurst-Hunter Z, Lyall DM, Hartwig FP, Horta BL, Hyppönen E, Power C, Moldovan M, van Iperen E, Hovingh GK, Demuth I, Norman K, Steinhagen-Thiessen E, Demuth J, Bertram L, Liu T, Coassin S, Willeit J, Kiechl S, Willeit K, Mason D, Wright J, Morris R, Wanamethee G, Whincup P, Ben-Shlomo Y, McLachlan S, Price JF, Kivimaki M, Welch C, Sanchez-Galvez A, Marques-Vidal P, Nicolaides A, Panayiotou

AG, Onland-Moret NC, van der Schouw YT, Matullo G, Fiorito G, Guarrera S, Sacerdote C, Wareham NJ, Langenberg C, Scott R, Luan J, Bobak M, Malyutina S, Pająk A, Kubinova R, Tamosiunas A, Pikhart H, Husemoen LL, Grarup N, Pedersen O, Hansen T, Linneberg A, Simonsen KS, Cooper J, Humphries SE, Brilliant M, Kitchner T, Hakonarson H, Carrell DS, McCarty CA, Kirchner HL, Larson EB, Crosslin DR, de Andrade M, Roden DM, Denny JC, Carty C, Hancock S, Attia J, Holliday E, O'Donnell M, Yusuf S, Chong M, Pare G, van der Harst P, Said MA, Eppinga RN, Verweij N, Snieder H; LifeLines Cohort study group., Christen T, Mook-Kanamori DO, Gustafsson S, Lind L, Ingelsson E, Pazoki R, Franco O, Hofman A, Uitterlinden A, Dehghan A, Teumer A, Baumeister S, Dörr M, Lerch MM, Völker U, Völzke H, Ward J, Pell JP, Smith DJ, Meade T, Maitland-van der Zee AH, Baranova EV, Young R, Ford I, Campbell A, Padmanabhan S, Bots ML, Grobbee DE, Froguel P, Thuillier D, Balkau B, Bonnefond A, Cariou B, Smart M, Bao Y, Kumari M, Mahajan A, Ridker PM, Chasman DI, Reiner AP, Lange LA, **Ritchie MD**, Asselbergs FW, Casas JP, Keating BJ, Preiss D, Hingorani AD; UCLEB consortium., Sattar N. PCSK9 genetic variants and risk of type 2 diabetes: a mendelian randomisation study. *Lancet Diabetes Endocrinol*. 2017 Feb;5(2):97-105. doi: 10.1016/S2213-8587(16)30396-5. PMID: 27908689, PMCID: PMC5266795

243. Dewey FE, Murray MF, Overton JD, Habegger L, Leader JB, Fetterolf SN, O'Dushlaine C, Van Hout CV, 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). pii: aaf6814. doi: 10.1126/science.aaf6814. PMID: 28008009

244. Abul-Husn NS, Manickam K, Jones LK, Wright EA, Hartzel DN, Gonzaga-Jauregui C, O'Dushlaine C, Leader JB, Lester Kirchner H, Lindbuchler DM, Barr ML, Giovanni MA, **Ritchie MD**, Overton JD, Reid JG, Metpally RP, Wardeh AH, Borecki IB, Yancopoulos GD, Baras A, Shuldiner AR, Gottesman O, Ledbetter DH, Carey DJ, Dewey FE, Murray MF. Genetic identification of familial hypercholesterolemia within a single U.S. health care system. *Science*. 2016 Dec 23;354(6319). pii: aaf7000. doi: 10.1126/science.aaf7000. PMID: 28008010

245. Kim D, Li R, Lucas A, Verma SS, Dudek SM, **Ritchie MD**. Using knowledge-driven genomic interactions for multi-omics data analysis: metadimensional models for predicting clinical outcomes in ovarian carcinoma. *J Am Med Inform Assoc*. 2016 Dec 31. pii: ocw165. doi: 10.1093/jamia/ocw165. PMID: 28040685, PMCID: PMC5391734

246. 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. doi: 10.1097/FPC.0000000000000263. PMID: 28099408, PMCID: PMC5285297

247. Shan Y, Tromp G, Kuivaniemi H, Smelser DT, Verma SS, **Ritchie MD**, Elmore JR, Carey DJ, Conley YP, Gorin MB, Weeks DE. Genetic risk models: Influence of model size on risk estimates and precision. *Genet Epidemiol*. 2017 Feb 15. doi: 10.1002/gepi.22035. PMID: 28198095

248. Heit JA, Armasu SM, McCauley BM, Kullo IJ, Sicotte H, Pathak J, Chute CG, Gottesman O, Bottinger EP, Denny JC, Roden DM, Li R, **Ritchie MD**, de Andrade M. Identification of unique venous thromboembolism-susceptibility variants in African-Americans. *Thromb Haemost*. 2017 Feb 16. doi: 10.1160/TH16-08-0652. PMID: 28203683

249. Dumitrescu L, **Ritchie MD**, Denny JC, El Rouby NM, McDonough CW, Bradford Y, Ramirez AH, Bielinski SJ, Basford MA, Chai HS, Peissig P, Carrell D, Pathak J, Rasmussen LV, Wang X, Pacheco JA, Kho AN, Hayes MG, Matsumoto M, Smith ME, Li R, Cooper-DeHoff RM,

Kullo IJ, Chute CG, Chisholm RL, Jarvik GP, Larson EB, Carey D, McCarty CA, Williams MS, Roden DM, Bottinger E, Johnson JA, de Andrade M, Crawford DC. Genome-wide study of resistant hypertension identified from electronic health records. *PLoS One*. 2017 Feb 21;12(2):e0171745. doi: 10.1371/journal.pone.0171745. PMID: 28222112, PMCID: PMC5319785

250. 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 of 30,716 subjects with exome sequencing. *Genet Med.* 2017 May 4. doi: 10.1038/gim.2017.40. PMID: 28471438

251. Kim D, Volk H, Girirajan S, Pendergrass S, Hall MA, Verma SS, Schmidt RJ, Hansen RL, Ghosh D, Ludena-Rodriguez Y, Kim K, **Ritchie MD**, Hertz-Picciotto I, Selleck SB. The joint effect of air pollution exposure and copy number variation on risk for autism. *Autism Res.* 2017 Apr 27. doi: 10.1002/aur.1799. PMID: 28448694

252. Fernández-Rhodes L, Gong J, Haessler J, Franceschini N, Graff M, Nishimura KK, Wang Y, Highland HM, Yoneyama S, Bush WS, Goodloe R, **Ritchie MD,** Crawford D, Gross M, Fornage M, Buzkova P, Tao R, Isasi C, Avilés-Santa L, Daviglus M, Mackey RH, Houston D, Gu CC, Ehret G, Nguyen KH, Lewis CE, Leppert M, Irvin MR, Lim U, Haiman CA, Le Marchand L, Schumacher F, Wilkens L, Lu Y, Bottinger EP, Loos RJ, Sheu WH, Guo X, Lee WJ, Hai Y, Hung YJ, Absher D, Wu IC, Taylor KD, Lee IT, Liu Y, Wang TD, Quertermous T, Juang JJ, Rotter JI, Assimes T, Hsiung CA, Chen YI, Prentice R, Kuller LH, Manson JE, Kooperberg C, Smokowski P, Robinson WR, Gordon-Larsen P, Li R, Hindorff L, Buyske S, Matise TC, Peters U, North KE. Trans-ethnic fine-mapping of genetic loci for body mass index in the diverse ancestral populations of the Population Architecture using Genomics and Epidemiology (PAGE) Study reveals evidence for multiple signals at established loci. *Hum Genet.* 2017 Apr 8. doi: 10.1007/s00439-017-1787-6. PMID: 28391526

253. Greene CS, Garmire LX, Gilbert JA, **Ritchie MD**, Hunter LE. Celebrating parasites. *Nat Genet.* 2017 Mar 30;49(4):483-484. doi: 10.1038/ng.3830. No abstract available. PMID: 28358134

254. Holzinger ER, Verma SS, Moore CB, Hall M, De R, Gilbert-Diamond D, Lanktree MB, Pankratz N, Amuzu A, Burt A, Dale C, Dudek S, Furlong CE, Gaunt TR, Kim DS, Riess H, Sivapalaratnam S, Tragante V, van Iperen EPA, Brautbar A, Carrell DS, Crosslin DR, Jarvik GP, Kuivaniemi H, Kullo IJ, Larson EB, Rasmussen-Torvik LJ, Tromp G, Baumert J, Cruickshanks KJ, Farrall M, Hingorani AD, Hovingh GK, Kleber ME, Klein BE, Klein R, Koenig W, Lange LA, März W, North KE, Charlotte Onland-Moret N, Reiner AP, Talmud PJ, van der Schouw YT, Wilson JG, Kivimaki M, Kumari M, Moore JH, Drenos F, Asselbergs FW, Keating BJ, **Ritchie MD**. Discovery and replication of SNP-SNP interactions for quantitative lipid traits in over 60,000 individuals. *BioData Min.* 2017 Jul 24;10:25. doi: 10.1186/s13040-017-0145-5. eCollection 2017. PMID: 28770004

255. Li R, Kim D, **Ritchie MD.** Methods to analyze big data in pharmacogenomics research. *Pharmacogenomics.* 2017 Jun;18(8):807-820. doi: 10.2217/pgs-2016-0152. Epub 2017 Jun 14. PMID: 28612644

256. Kim D, Basile AO, Bang L, Horgusluoglu E, Lee S, **Ritchie MD**, Saykin AJ, Nho K. Knowledge-driven binning approach for rare variant association analysis: application to neuroimaging biomarkers in Alzheimer's disease. *BMC Med Inform Decis Mak.* 2017 May 18;17(Suppl 1):61. doi: 10.1186/s12911-017-0454-0. PMID: 28539126

257. Dewey FE, Gusarova V, Dunbar RL, O'Dushlaine C, Schurmann C, Gottesman O, McCarthy S, Van Hout CV, Bruse S, Dansky HM, Leader JB, Murray MF, **Ritchie MD**, Kirchner HL, Habegger L, Lopez A, Penn J, Zhao A, Shao W, Stahl N, Murphy AJ, Hamon S, Bouzelmat A, Zhang R, Shumel B, Pordy R, Gipe D, Herman GA, Sheu WHH, Lee IT, Liang KW, Guo X, Rotter JI, Chen YI, Kraus WE, Shah SH, Damrauer S, Small A, Rader DJ, Wulff AB, Nordestgaard BG, Tybjærg-Hansen A, van den Hoek AM, Princen HMG, Ledbetter DH, Carey

DJ, Overton JD, Reid JG, Sasiela WJ, Banerjee P, Shuldiner AR, Borecki IB, Teslovich TM, Yancopoulos GD, Mellis SJ, Gromada J, Baras A. Genetic and Pharmacologic Inactivation of ANGPTL3 and Cardiovascular Disease. *N Engl J Med.* 2017 Jul 20;377(3):211-221. doi: 10.1056/NEJMoa1612790. Epub 2017 May 24. PMID: 28538136

258. Rasmussen-Torvik LJ, Almoguera B, Doheny KF, Freimuth RR, Gordon AS, Hakonarson H, Hawkins JB, Husami A, Ivacic LC, Kullo IJ, Linderman MD, Manolio TA, Obeng AO, Pellegrino R, Prows CA, Ritchie MD, Smith ME, Stallings SC, Wolf WA, Zhang K, Scott SA. Concordance between Research Sequencing and Clinical Pharmacogenetic Genotyping in the eMERGE-PGx Study. *J Mol Diagn.* 2017 Jul;19(4):561-566. doi: 10.1016/j.jmoldx.2017.04.002. Epub 2017 May 11. PMID: 28502727

259. 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 S, Moore JH, **Ritchie MD**. PLATO software provides analytic framework for investigating complexity beyond genome-wide association studies. *Nature Communications,* 2017 Oct 27;8(1):1167 PMID: 29079728, PMCID: PMC5660079

260. McAllister K, Mechanic LE, Amos C, Aschard H, Blair IA, Chatterjee N, Conti D, Gauderman WJ, Hsu L, Hutter CM, Jankowska MM, Kerr J, Kraft P, Montgomery SB, Mukherjee B, Papanicolaou GJ, Patel CJ, Ritchie MD, Ritz BR, Thomas DC, Wei P, Witte JS. Current Challenges and New Opportunities for Gene-Environment Interaction Studies of Complex Diseases. *Am J Epidemiol.* 2017 Oct 1;186(7):753-761. PMID: 28978193

261. **Ritchie MD,** Davis JR, Aschard H, Battle A, Conti D, Du M, Eskin E, Fallin MD, Hsu L, Kraft P, Moore JH, Pierce BL, Bien SA, Thomas DC, Wei P, Montgomery SB. Incorporation of Biological Knowledge Into the Study of Gene-Environment Interactions. *Am J Epidemiol.* 2017 Oct 1;186(7):771-777. PMID: 28978191

262. Basile AO, Byrska-Bishop M, Wallace J, Frase A, and **Ritchie MD.** Novel features and enhancements in BioBin, a tool for the biologically inspired binning and association analysis of rare variants. *Bioinformatics.* 2017 Feb 1;34(3):527-529. PMID: 28968757. PMCID: PMC5860358

263. Mechanic LE, Lindström S, Daily KM, Sieberts SK, Amos CI, Chen HS, Cox NJ, Dathe M, Feuer EJ, Guertin MJ, Hoffman J, Liu Y, Moore JH, Myers CL, **Ritchie MD**, Schildkraut J, Schumacher F, Witte JS, Wang W, Williams SM; U4C Challenge Participants; U4C Challenge Data Contributors, Gillanders EM. Up For A Challenge (U4C): Stimulating innovation in breast cancer genetic epidemiology. *PLoS Genet.* 2017 Sep 28;13(9):e1006945. PMID: 28957327. PMCID: PMC5619686

264. Veturi Y, **Ritchie MD.** How powerful are summary-based methods for identifying expression-trait associations under different genetic architectures? *Pac Symp Biocomput.* 2018; 23:228-239. PMID: 29218884. PMCID: PMC5785784

265. Li B, Verma SS, Veturi YC, Verma A, Bradford Y, Haas DW, **Ritchie MD**. Evaluation of PrediXcan for prioritizing GWAS associations and predicting gene expression. *Pac Symp Biocomput.* 2018;23:448-459. PMID: 29218904. PMCID: PMC5749400

266. Buchanan AH, Manickam K, Meyer MN, Wagner JK, Hallquist MLG, Williams JL, Rahm AK, Williams MS, Chen ZE, Shah CK, Garg TK, Lazzeri AL, Schwartz MLB, Lindbuchler DM, Fan AL, Leeming R, Servano PO 3rd, Smith AL, Vogel VG, Abul-Husn NS, Dewey FE, Lebo MS, Mason-Suares HM, **Ritchie MD**, Davis FD, Carey DJ, Feinberg DT, Faucett WA, Ledbetter DH, Murray MF. Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. *Genet Med.* 2017 Oct 26. PMID: 29261187

267. Nielsen JB, Fritsche LG, Zhou W, Teslovich TM, Holmen OL, Gustafsson S, Gabrielsen ME, Schmidt EM, Beaumont R, Wolford BN, Lin M, Brummett CM, Preuss MH, Refsgaard L, Bottinger EP, Graham SE, Surakka I, Chu Y, Skogholt AH, Dalen H, Boyle AP, Oral H, Herron TJ, Kitzman J, Jalife J, Svendsen JH, Olesen MS, Njølstad I, Løchen ML, Baras A, Gottesman O, Marcketta A, O'Dushlaine C, **Ritchie MD**, Wilsgaard T, Loos RJF, Frayling TM, Boehnke M, Ingelsson E, Carey DJ, Dewey FE, Kang HM, Abecasis GR, Hveem K, Willer CJ. Genome-

wide Study of Atrial Fibrillation Identifies Seven Risk Loci and Highlights Biological Pathways and Regulatory Elements Involved in Cardiac Development. *Am J Hum Genet.* 2018 Jan 4;102(1):103-115. PMID: 29290336

268. Volpi S, Bult CJ, Chisholm RL, Deverka PA, Ginsburg GS, Jacob HJ, Kasapi M, McLeod HL, Roden DM, Williams MS, Green ED, Rodriguez LL, Aronson S, Cavallari LH, Denny JC, Dressler LG, Johnson JA, Klein TE, Leeder JS, Piquette-Miller M, Perera M, Rasmussen-Torvik LJ, Rehm HL, **Ritchie MD,** Skaar TC, Wagle N, Weinshilboum R, Weitzel KW, Wildin R, Wilson J, Manolio TA, Relling MV. Research Directions in the Clinical Implementation of Pharmacogenomics: An Overview of US Programs and Projects. *Clin Pharmacol Ther*. 2018 Feb 20. doi: 10.1002/cpt.1048. PMID:29460415.

269. 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. doi: 10.1038/s41598-018-22834-4. PMID:29545597; PMCID:PMC5854600

270. 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 Mar 19. pii: S0002-9297(18)30062-4. doi: 10.1016/j.ajhg.2018.02.017. PMID:29606303

271. Verma A, Bradford Y, Dudek SM, Verma SS, Pendergrass SA, **Ritchie MD**. A simulation study investigating power of Phenome-Wide Association Studies. *BMC Bioinformatics,* 2018 Apr 4;19(1):120. doi: 10.1186/s12859-018-2135-0. PMID:29618318

272. van Setten J, Brody JA, Jamshidi Y, Swenson BR, Butler AM, Campbell H, Del Greco FM, Evans DS, Gibson Q, Gudbjartsson DF, Kerr KF, Krijthe BP, Lyytikäinen LP, Müller C, Müller-Nurasyid M, Nolte IM, Padmanabhan S, **Ritchie MD**, Robino A, Smith AV, Steri M, Tanaka T, Teumer A, Trompet S, Ulivi S, Verweij N, Yin X, Arnar DO, Asselbergs FW, Bader JS, Barnard J, Bis J, Blankenberg S, Boerwinkle E, Bradford Y, Buckley BM, Chung MK, Crawford D, den Hoed M, Denny JC, Dominiczak AF, Ehret GB, Eijgelsheim M, Ellinor PT, Felix SB, Franco OH, Franke L, Harris TB, Holm H, Ilaria G, Iorio A, Kähönen M, Kolcic I, Kors JA, Lakatta EG, Launer LJ, Lin H, Lin HJ, Loos RJF, Lubitz SA, Macfarlane PW, Magnani JW, Leach IM, Meitinger T, Mitchell BD, Munzel T, Papanicolaou GJ, Peters A, Pfeufer A, Pramstaller PP, Raitakari OT, Rotter JI, Rudan I, Samani NJ, Schlessinger D, Silva Aldana CT, Sinner MF, Smith JD, Snieder H, Soliman EZ, Spector TD, Stott DJ, Strauch K, Tarasov KV, Thorsteinsdottir U, Uitterlinden AG, Van Wagoner DR, Völker U, Völzke H, Waldenberger M, Jan Westra H, Wild PS, Zeller T, Alonso A, Avery CL, Bandinelli S, Benjamin EJ, Cucca F, Dörr M, Ferrucci L, Gasparini P, Gudnason V, Hayward C, Heckbert SR, Hicks AA, Jukema JW, Kääb S, Lehtimäki T, Liu Y, Munroe PB, Parsa A, Polasek O, Psaty BM, Roden DM, Schnabel RB, Sinagra G, Stefansson K, Stricker BH, van der Harst P, van Duijn CM, Wilson JF, Gharib SA, de Bakker PIW, Isaacs A, Arking DE, Sotoodehnia N. PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. *Nat Commun.* 2018 Jul 25;9(1):2904. doi: 10.1038/s41467-018-04766-9. PMID: 30046033

273. Lee JJ, Wedow R, Okbay A, Kong E, Maghzian O, Zacher M, Nguyen-Viet TA, Bowers P, Sidorenko J, Karlsson Linnér R, Fontana MA, Kundu T, Lee C, Li H, Li R, Royer R, Timshel PN, Walters RK, Willoughby EA, Yengo L; 23andMe Research Team; COGENT (Cognitive Genomics Consortium); Social Science Genetic Association Consortium, Alver M, Bao Y, Clark DW, Day FR, Furlotte NA, Joshi PK, Kemper KE, Kleinman A, Langenberg C, Mägi R, Trampush JW, Verma SS, Wu Y, Lam M, Zhao JH, Zheng Z, Boardman JD, Campbell H, Freese J, Harris KM, Hayward C, Herd P, Kumari M, Lencz T, Luan J, Malhotra AK, Metspalu A, Milani L, Ong KK, Perry JRB, Porteous DJ, Ritchie MD, Smart MC, Smith BH, Tung JY, Wareham NJ, Wilson JF, Beauchamp JP, Conley DC, Esko T, Lehrer SF, Magnusson PKE, Oskarsson S, Pers TH, Robinson MR, Thom K, Watson C, Chabris CF, Meyer MN, Laibson DI, Yang J, Johannesson M, Koellinger PD, Turley P, Visscher PM, Benjamin DJ, Cesarini D. Gene discovery and polygenic prediction from a genome-wide association study of educational

attainment in 1.1 million individuals. *Nat Genet.* 2018 Jul 23. doi: 10.1038/s41588-018-0147-3. [Epub ahead of print. PMID: 30038396

274. Gusarova V, O'Dushlaine C, Teslovich TM, Benotti PN, Mirshahi T, Gottesman O, Van Hout CV, Murray MF, Mahajan A, Nielsen JB, Fritsche L, Wulff AB, Gudbjartsson DF, Sjögren M, Emdin CA, Scott RA, Lee WJ, Small A, Kwee LC, Dwivedi OP, Prasad RB, Bruse S, Lopez AE, Penn J, Marcketta A, Leader JB, Still CD, Kirchner HL, Mirshahi UL, Wardeh AH, Hartle CM, Habegger L, Fetterolf SN, Tusie-Luna T, Morris AP, Holm H, Steinthorsdottir V, Sulem P, Thorsteinsdottir U, Rotter JI, Chuang LM, Damrauer S, Birtwell D, Brummett CM, Khera AV, Natarajan P, Orho-Melander M, Flannick J, Lotta LA, Willer CJ, Holmen OL, **Ritchie MD**, Ledbetter DH, Murphy AJ, Borecki IB, Reid JG, Overton JD, Hansson O, Groop L, Shah SH, Kraus WE, Rader DJ, Chen YI, Hveem K, Wareham NJ, Kathiresan S, Melander O, Stefansson K, Nordestgaard BG, Tybjærg-Hansen A, Abecasis GR, Altshuler D, Florez JC, Boehnke M, McCarthy MI, Yancopoulos GD, Carey DJ, Shuldiner AR, Baras A, Dewey FE, Gromada J. Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes. *Nat Commun.* 2018 Jun 13;9(1):2252. doi: 10.1038/s41467-018-04611-z. PMID: 29899519. PMCID: PMC5997992

275. Haas DW, Bradford Y, Verma A, Verma SS, Eron JJ, Gulick RM, Riddler SA, Sax PE, Daar ES, Morse GD, Acosta EP, **Ritchie MD**. Brain neurotransmitter transporter/receptor genomics and efavirenz central nervous system adverse events. *Pharmacogenet Genomics*. 2018 Jul;28(7):179-187. doi: 10.1097/FPC.0000000000000341. PMID:29847509

276. Li R, Kim D, Wheeler HE, Dudek SM, Dolan ME, **Ritchie MD**. Integration of genetic and functional genomics data to uncover chemotherapeutic induced cytotoxicity. *Pharmacogenomics J*. 2018 May 25. doi: 10.1038/s41397-018-0024-6. [Epub ahead of print]PMID:29795408

277. Verma SS, Lucas A, Zhang X, Veturi Y, Dudek S, Li B, Li R, Urbanowicz R, Moore JH, Kim D, **Ritchie MD**. Collective feature selection to identify crucial epistatic variants. *BioData Min.* 2018 Apr 19;11:5. doi: 10.1186/s13040-018-0168-6. eCollection 2018. PMID:29713383. PMCID:PMC5907720

278. Bergmeijer TO, Reny JL, Pakyz RE, Gong L, Lewis JP, Kim EY, Aradi D, Fernandez-Cadenas I, Horenstein RB, Lee MTM, Whaley RM, Montaner J, Gensini GF, Cleator JH, Chang K, Holmvang L, Hochholzer W, Roden DM, Winter S, Altman RB, Alexopoulos D, Kim HS, Déry JP, Gawaz M, Bliden K, Valgimigli M, Marcucci R, Campo G, Schaeffeler E, Dridi NP, Wen MS, Shin JG, Simon T, Fontana P, Giusti B, Geisler T, Kubo M, Trenk D, Siller-Matula JM, Ten Berg JM, Gurbel PA, Hulot JS, Mitchell BD, Schwab M, **Ritchie MD**, Klein TE, Shuldiner AR; ICPC Investigators. Genome-wide and candidate gene approaches of clopidogrel efficacy using pharmacodynamic and clinical end points-Rationale and design of the International Clopidogrel Pharmacogenomics Consortium (ICPC). *Am Heart J.* 2018 Apr;198:152-159. doi: 10.1016/j.ahj.2017.12.010. Epub 2017 Dec 17.PMID:29653637

279. Mosley JD, Feng Q, Wells QS, Van Driest SL, Shaffer CM, Edwards TL, Bastarache L, Wei WQ, Davis LK, McCarty CA, Thompson W, Chute CG, Jarvik GP, Gordon AS, Palmer MR, Crosslin DR, Larson EB, Carrell DS, Kullo IJ, Pacheco JA, Peissig PL, Brilliant MH, Linneman JG, Namjou B, Williams MS, **Ritchie MD**, Borthwick KM, Verma SS, Karnes JH, Weiss ST, Wang TJ, Stein CM, Denny JC, Roden DM. A study paradigm integrating prospective epidemiologic cohorts and electronic health records to identify disease biomarkers. *Nat Commun.* 2018 Aug 30;9(1):3522. PMID:30166544 . PMCID:PMC6117367

280. 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

281. 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. doi: 10.1016/j.ajhg.2018.11.006. Epub 2018 Dec 29. PMID:30598166

282. 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. doi: 10.1186/s12859-018-2591-6. PMID:30669967

283. Li B, Veturi Y, Bradford Y, Verma SS, Verma A, Lucas AM, Haas DW, **Ritchie MD**. Influence of tissue context on gene prioritization for predicted transcriptome-wide association studies. *Pac Symp Biocomput.* 2019;24:296-307. PMID:30864331

284. Zhang X, Veturi Y, Verma S, Bone W, Verma A, Lucas A, Hebbring S, Denny JC, Stanaway IB, Jarvik GP, Crosslin D, Larson EB, Rasmussen-Torvik L, Pendergrass SA, Smoller JW, Hakonarson H, Sleiman P, Weng C, Fasel D, Wei WQ, Kullo I, Schaid D, Chung WK, **Ritchie MD.** Detecting potential pleiotropy across cardiovascular and neurological diseases using univariate, bivariate, and multivariate methods on 43,870 individuals from the eMERGE network. *Pac Symp Biocomput.* 2019;24:272-283. PMID:30864329

285. Safarova MS, Satterfield BA, Fan X, Austin EE, Ye Z, Bastarache L, Zheng N, Ritchie MD, Borthwick KM, Williams MS, Larson EB, Scrol A, Jarvik GP, Crosslin DR, Leppig K, Rasmussen-Torvik LJ, Pendergrass SA, Sturm AC, Namjou B, Shah AS, Carroll RJ, Chung WK, Wei WQ, Feng Q, Stein CM, Roden DM, Manolio TA, Schaid DJ, Denny JC, Hebbring SJ, de Andrade M, Kullo IJ. A phenome-wide association study to discover pleiotropic effects of PCSK9, APOB, and LDLR. *NPJ Genom Med.* 2019 Feb 11;4:3. doi: 10.1038/s41525-019-0078-7. eCollection 2019. PMID:30774981

286. Moore JH, Boland MR, Camara PG, Chervitz H, Gonzalez G, Himes BE, Kim D, Mowery DL, **Ritchie MD,** Shen L, Urbanowicz RJ, Holmes JH. Preparing next-generation scientists for biomedical big data: artificial intelligence approaches. *Per Med.* 2019 May 1;16(3):247-257. doi: 10.2217/pme-2018-0145. Epub 2019 Feb 14. PMID:30760118

287. Hellwege JN, Stallings S, Torstenson ES, Carroll R, Borthwick KM, Brilliant MH, Crosslin D, Gordon A, Hripcsak G, Jarvik GP, Linneman JG, Devi P, Peissig PL, Sleiman PAM, Hakonarson H, **Ritchie MD**, Verma SS, Shang N, Denny JC, Roden DM, Velez Edwards DR, Edwards TL. Heritability and genome-wide association study of benign prostatic hyperplasia (BPH) in the eMERGE network. *Sci Rep.* 2019 Apr 15;9(1):6077. doi: 10.1038/s41598-019-42427-z. PMID:30988330

288. Miller JE, Metpally RP, Person TN, Krishnamurthy S, Dasari VR, Shivakumar M, Lavage DR, Cook AM, Carey DJ, **Ritchie MD,** Kim D, Gogoi R; DiscovEHR collaboration. Correction to: Systematic characterization of germline variants from the DiscovEHR study endometrial carcinoma population. *BMC Med Genomics.* 2019 May 22;12(1):65. doi: 10.1186/s12920-019-0523-6. PMID:31118041

289. Miller JE, Veturi Y, **Ritchie MD.** Innovative strategies for annotating the "relationSNP" between variants and molecular phenotypes. *BioData Min.* 2019 May 14;12:10. doi: 10.1186/s13040-019-0197-9. eCollection 2019. Review. PMID:31114635

290. Miller JE, Metpally RP, Person TN, Krishnamurthy S, Dasari VR, Shivakumar M, Lavage DR, Cook AM, Carey DJ, **Ritchie MD**, Kim D, Gogoi R; DiscovEHR collaboration. Systematic characterization of germline variants from the DiscovEHR study endometrial carcinoma population. *BMC Med Genomics.* 2019 May 3;12(1):59. doi: 10.1186/s12920-019-0504-9. Erratum in: *BMC Med Genomics.* 2019 May 22;12(1):65. PMID:31053132

291. eMERGE Consortium. Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *Am J Hum Genet.* 2019 Sep 5;105(3):588-605. doi: 10.1016/j.ajhg.2019.07.018. Epub 2019 Aug 22. PMID:31447099

292. Park J, Levin MG, Haggerty CM, Hartzel DN, Judy R, Kember RL, Reza N; Regeneron Genetics Center, **Ritchie MD**, Owens AT, Damrauer SM, Rader DJ. A genome-first approach to

aggregating rare genetic variants in LMNA for association with electronic health record phenotypes. *Genet Med.* 2019 Aug 6. doi: 10.1038/s41436-019-0625-8. [Epub ahead of print] PMID:31383942

293. Manduchi E, Orzechowski PR, **Ritchie MD**, Moore JH. Exploration of a diversity of computational and statistical measures of association for genome-wide genetic studies. *BioData Min.* 2019 Jul 9;12:14. doi: 10.1186/s13040-019-0201-4. eCollection 2019. PMID:31320928

294. Namjou B, Lingren T, Huang Y, Parameswaran S, Cobb BL, Stanaway IB, Connolly JJ, Mentch FD, Benoit B, Niu X, Wei WQ, Carroll RJ, Pacheco JA, Harley ITW, Divanovic S, Carrell DS, Larson EB, Carey DJ, Verma S, **Ritchie MD**, Gharavi AG, Murphy S, Williams MS, Crosslin DR, Jarvik GP, Kullo IJ, Hakonarson H, Li R; eMERGE Network, Xanthakos SA, Harley JB. GWAS and enrichment analyses of non-alcoholic fatty liver disease identify new trait-associated genes and pathways across eMERGE Network. *BMC Med.* 2019 Jul 17;17(1):135. doi: 10.1186/s12916-019-1364-z. PMID:31311600

295. Sangkuhl K, Whirl-Carrillo M, Whaley RM, Woon M, Lavertu A, Altman RB, Carter L, Verma A, **Ritchie MD**, Klein TE. Pharmacogenomics Clinical Annotation Tool (PharmCAT). *Clin Pharmacol Ther.* 2019 Jul 15. doi: 10.1002/cpt.1568. [Epub ahead of print]. PMID:31306493

296. Haggerty CM, Damrauer SM, Levin MG, Birtwell D, Carey DJ, Golden AM, Hartzel DN, Hu Y, Judy R, Kelly MA, Kember RL, Lester Kirchner H, Leader JB, Liang L, McDermott-Roe C, Babu A, Morley M, Nealy Z, Person TN, Pulenthiran A, Small A, Smelser DT, Stahl RC, Sturm AC, Williams H, Baras A, Margulies KB, Cappola TP, Dewey FE, Verma A, Zhang X, Correa A, Hall ME, Wilson JG, **Ritchie MD,** Rader DJ, Murray MF, Fornwalt BK, Arany Z. Genomics-First Evaluation of Heart Disease Associated With Titin-Truncating Variants. *Circulation.* 2019 Jul 2;140(1):42-54. doi: 10.1161/CIRCULATIONAHA.119.039573. Epub 2019 Jun 20. Erratum in: Circulation. 2019 Oct 15;140(16):e727. PMID:31216868

297. Glessner JT, Li J, Desai A, Palmer M, Kim D, Lucas AM, Chang X, Connolly JJ, Almoguera B, Harley JB, Jarvik GP, **Ritchie MD**, Sleiman PMA, Roden DM, Crosslin D, Hakonarson H. CNV Association of Diverse Clinical Phenotypes from eMERGE reveals novel disease biology underlying cardiovascular disease. *Int J Cardiol.* 2019 Jul 19. pii: S0167-5273(19)30191-3. doi: 10.1016/j.ijcard.2019.07.058. [Epub ahead of print] PMID:31447229

298. Lewis JP, Backman JD, Reny JL, Bergmeijer TO, Mitchell BD, **Ritchie MD,** Déry JP, Pakyz RE, Gong L, Ryan K, Kim EY, Aradi D, Fernandez-Cadenas I, Lee MTM, Whaley RM, Montaner J, Gensini GF, Cleator JH, Chang K, Holmvang L, Hochholzer W, Roden DM, Winter S, Altman R, Alexopoulos D, Kim HS, Gawaz M, Bliden K, Valgimigli M, Marcucci R, Campo G, Schaeffeler E, Dridi NP, Wen MS, Shin JG, Fontana P, Giusti B, Geisler T, Kubo M, Trenk D, Siller-Matula JM, Ten Berg JM, Gurbel PA, Schwab M, Klein TE, Shuldiner AR. Pharmacogenomic Polygenic Response Score Predicts Ischemic Events and Cardiovascular Mortality in Clopidogrel-Treated Patients. *Eur Heart J Cardiovasc Pharmacother.* 2019 Sep 3. pii: pvz045. doi: 10.1093/ehjcvp/pvz045. PMID:31504375

299. Schmidt AF, Holmes MV, Preiss D, Swerdlow DI, Denaxas S, Fatemifar G, Faraway R, Finan C, Valentine D, Fairhurst-Hunter Z, Hartwig FP, Horta BL, Hypponen E, Power C, Moldovan M, van Iperen E, Hovingh K, Demuth I, Norman K, Steinhagen-Thiessen E, Demuth J, Bertram L, Lill CM, Coassin S, Willeit J, Kiechl S, Willeit K, Mason D, Wright J, Morris R, Wanamethee G, Whincup P, Ben-Shlomo Y, McLachlan S, Price JF, Kivimaki M, Welch C, Sanchez-Galvez A, Marques-Vidal P, Nicolaides A, Panayiotou AG, Onland-Moret NC, van der Schouw YT, Matullo G, Fiorito G, Guarrera S, Sacerdote C, Wareham NJ, Langenberg C, Scott RA, Luan J, Bobak M, Malyutina S, Pająk A, Kubinova R, Tamosiunas A, Pikhart H, Grarup N, Pedersen O, Hansen T, Linneberg A, Jess T, Cooper J, Humphries SE, Brilliant M, Kitchner T, Hakonarson H, Carrell DS, McCarty CA, Lester KH, Larson EB, Crosslin DR, de Andrade M, Roden DM, Denny JC, Carty C, Hancock S, Attia J, Holliday E, Scott R, Schofield P, O'Donnell M, Yusuf S, Chong M, Pare G, van der Harst P, Said MA, Eppinga RN, Verweij N, Snieder H; Lifelines Cohort authors, Christen T, Mook-Kanamori DO; ICBP Consortium, Gustafsson S, Lind L, Ingelsson E, Pazoki R, Franco O, Hofman A, Uitterlinden A, Dehghan A, Teumer A, Baumeister S, Dörr M, Lerch MM, Völker U, Völzke H, Ward J, Pell JP, Meade T, Christophersen IE,

Maitland-van der Zee AH, Baranova EV, Young R, Ford I, Campbell A, Padmanabhan S, Bots ML, Grobbee DE, Froguel P, Thuillier D, Roussel R, Bonnefond A, Cariou B, Smart M, Bao Y, Kumari M, Mahajan A, Hopewell JC, Seshadri S; METASTROKE Consortium of the ISGC, Dale C, Costa RPE, Ridker PM, Chasman DI, Reiner AP, **Ritchie MD**, Lange LA, Cornish AJ, Dobbins SE, Hemminki K, Kinnersley B, Sanson M, Labreche K, Simon M, Bondy M, Law P, Speedy H, Allan J, Li N, Went M, Weinhold N, Morgan G, Sonneveld P, Nilsson B, Goldschmidt H, Sud A, Engert A, Hansson M, Hemingway H, Asselbergs FW, Patel RS, Keating BJ, Sattar N, Houlston R, Casas JP, Hingorani AD. Phenome-wide association analysis of LDL-cholesterol lowering genetic variants in PCSK9. *BMC Cardiovasc Disord*. 2019 Oct 29;19(1):240. doi: 10.1186/s12872-019-1187-z. PMID:31664920

300. Mosley JD, Benson MD, Smith JG, Melander O, Ngo D, Shaffer CM, Ferguson JF, Herzig MS, McCarty CA, Chute CG, Jarvik GP, Gordon AS, Palmer MR, Crosslin DR, Larson EB, Carrell DS, Kullo IJ, Pacheco JA, Peissig PL, Brilliant MH, Kitchner TE, Linneman JG, Namjou B, Williams MS, **Ritchie MD**, Borthwick KM, Kiryluk K, Mentch FD, Sleiman PM, Karlson EW, Verma SS, Zhu Y, Vasan RS, Yang Q, Denny JC, Roden DM, Gerszten RE, Wang TJ. Probing the Virtual Proteome to Identify Novel Disease Biomarkers. *Circulation.* 2018 Nov 27;138(22):2469-2481. doi: 10.1161/CIRCULATIONAHA.118.036063. PMID:30571344

301. Lucas AM, Palmiero NE, McGuigan J, Passero K, Zhou J, Orie D, **Ritchie MD**, Hall MA. CLARITE Facilitates the Quality Control and Analysis Process for EWAS of Metabolic-Related Traits. *Front Genet.* 2019 Dec 18;10:1240. PMID: 31921293

302. Joo YY, Actkins K, Pacheco JA, Basile AO, Carroll R, Crosslin DR, Day F, Denny JC, Velez Edwards DR, Hakonarson H, Harley JB, Hebbring SJ, Ho K, Jarvik GP, Jones M, Karderi T, Mentch FD, Meun C, Namjou B, Pendergrass S, **Ritchie MD**, Stanaway IB, Urbanek M, Walunas TL, Smith M, Chisholm RL, Kho AN, Davis L, Hayes MG; International PCOS Consortium. A polygenic and phenotypic risk prediction for Polycystic Ovary Syndrome evaluated by Phenome-wide association studies. *Clin Endocrinol Metab.* 2020 Jan 9. pii: dgz326. PMID:31917831

303. Pendergrass SA, Buyske S, Jeff JM, Frase A, Dudek S, Bradford Y, Ambite JL, Avery CL, Buzkova P, Deelman E, Fesinmeyer MD, Haiman C, Heiss G, Hindorff LA, Hsu CN, Jackson RD, Lin Y, Le Marchand L, Matise TC, Monroe KR, Moreland L, North KE, Park SL, Reiner A, Wallace R, Wilkens LR, Kooperberg C, **Ritchie MD**, Crawford DC. A phenome-wide association study (PheWAS) in the Population Architecture using Genomics and Epidemiology (PAGE) study reveals potential pleiotropy in African Americans. *PLoS One.* 2019 Dec 31;14(12):e0226771. PMID: 31891604

304. Erlandson KM, Bradford Y, Samuels DC, Brown T, Sun J, Wu K, Tassiopoulos K, **Ritchie MD**, Haas D, Hulgan T. Mitochondrial DNA Haplogroups and Frailty in Adults Living with HIV. *AIDS Res Hum Retroviruses.* 2019 Dec 10. PMID: 31822125

305. Murray MF, Kenny EE, **Ritchie MD**, Rader DJ, Bale AE, Giovanni MA, Abul-Husn NS. COVID-19 outcomes and the human genome. *Genet Med.* 2020 May 12. doi: 10.1038/s41436-020-0832-3. PMID: 32393819

306. Verma SS, Bergmeijer TO, Gong L, Reny JL, Lewis JP, Mitchell BD, Alexopoulos D, Aradi D, Altman RB, Bliden K, Bradford Y, Campo G, Chang K, Cleator JH, Déry JP, Dridi NP, Fernandez-Cadenas I, Fontana P, Gawaz M, Geisler T, Gensini GF, Giusti B, Gurbel PA, Hochholzer W, Holmvang L, Kim EY, Kim HS, Marcucci R, Montaner J, Backman JD, Pakyz RE, Roden DM, Schaeffeler E, Schwab M, Shin JG, Siller-Matula JM, Ten Berg JM, Trenk D, Valgimigli M, Wallace J, Wen MS, Kubo M, Lee MTM, Whaley R, Winter S, Klein TE, Shuldiner AR, **Ritchie MD**; ICPC Investigators. Genome-wide association study of platelet reactivity and cardiovascular response in patients treated with clopidogrel: a study by the International Clopidogrel Pharmacogenomics Consortium (ICPC). *Clin Pharmacol Ther.* 2020 May 30. doi: 10.1002/cpt.1911. PMID: 32472697

307. Lau-Min KS, Asher SB, Chen J, Domchek SM, Feldman M, Joffe S, Landgraf J, Speare V, Varughese LA, Tuteja S, VanZandbergen C, **Ritchie MD**, Nathanson KL. Real-world integration

of genomic data into the electronic health record: the PennChart Genomics Initiative. Genet Med. 2020 Dec 10. doi: 10.1038/s41436-020-01056-y. Online ahead of print. PMID: 33299147

308. Levin MG, Judy R, Gill D, Vujkovic M, Verma SS, Bradford Y; Regeneron Genetics Center, **Ritchie MD**, Hyman MC, Nazarian S, Rader DJ, Voight BF, Damrauer SM. Genetics of height and risk of atrial fibrillation: A Mendelian randomization study. PLoS Med. 2020 Oct 8;17(10):e1003288. doi:10.1371/journal.pmed.1003288. PMID: 33031386. PMCID: PMC7544133

309. Bergmeijer TO, Yasmina A, Vos GJA, Janssen PWA, Hackeng CM, Kelder JC, Verma SS, **Ritchie MD**, Gong L, Klein TE, Icpc Investigators, de Boer A, Klungel OH, Ten Berg JM, Deneer VHM. Effect of CYP3A4*22 and PPAR-α Genetic Variants on Platelet Reactivity in Patients Treated with Clopidogrel and Lipid-Lowering Drugs Undergoing Elective Percutaneous Coronary Intervention. Genes (Basel). 2020 Sep 11;11(9):1068. doi: 10.3390/genes11091068. PMID: 32932966. PMCID: PMC7564055

310. Kember RL, Merikangas AK, Verma SS, Verma A, Judy R; Regeneron Genetics Center, Damrauer SM, **Ritchie MD**, Rader DJ, Bućan M. Polygenic Risk of Psychiatric Disorders Exhibits Cross-trait Associations in Electronic Health Record Data From European Ancestry Individuals. Biol Psychiatry. 2020 Jul 6:S0006-3223(20)31734-0. doi: 10.1016/j.biopsych.2020.06.026. Online ahead of print. PMID: 32919613.

311. Leonard MA, Cindi Z, Bradford Y, Bourgi K, Koethe J, Turner M, Norwood J, Woodward B, Erdem H, Basham R, Baker P, Rebeiro PF, Sterling TR, Hulgan T, Daar ES, Gulick R, Riddler SA, Sinxadi P, **Ritchie MD**, Haas DW. Efavirenz Pharmacogenetics and Weight Gain following Switch to Integrase Inhibitor-containing Regimens. Clin Infect Dis. 2020 Aug 23:ciaa1219. doi: 10.1093/cid/ciaa1219. Online ahead of print. PMID: 32829410

312. Li R, Duan R, Zhang X, Lumley T, Pendergrass S, Bauer C, Hakonarson H, Carrell DS, Smoller JW, Wei WQ, Carroll R, Velez Edwards DR, Wiesner G, Sleiman P, Denny JC, Mosley JD, **Ritchie MD**, Chen Y, Moore JH. Lossless integration of multiple electronic health records for identifying pleiotropy using summary statistics. Nat Commun. 2021 Jan 8;12(1):168. PMID: 33420026. PMCID: PMC7794298.

313. Rosenthal EA, Crosslin DR, Gordon AS, Carrell DS, Stanaway IB, Larson EB, Grafton J, Wei WQ, Denny JC, Feng QP, Shah AS, Sturm AC, **Ritchie MD,** Pacheco JA, Hakonarson H, Rasmussen-Torvik LJ, Connolly JJ, Fan X, Safarova M, Kullo IJ, Jarvik GP. Association between triglycerides, known risk SNVs and conserved rare variation in SLC25A40 in a multi-ancestry cohort. BMC Med Genomics. 2021 Jan 6;14(1):11. PMID: 33407432. PMCID: PMC7789246

314. Park J, Lucas AM, Zhang X, Chaudhary K, Cho JH, Nadkarni G, Dobbyn A, Chittoor G, Josyula NS, Katz N, Breeyear JH, Ahmadmehrabi S, Drivas TG, Chavali VRM, Fasolino M, Sawada H, Daugherty A, Li Y, Zhang C, Bradford Y, Weaver J, Verma A, Judy RL, Kember RL, Overton JD, Reid JG, Ferreira MAR, Li AH, Baras A, LeMaire SA, Shen YH, Naji A, Kaestner KH, Vahedi G, Edwards TL, Chen J, Damrauer SM, Justice AE, Do R, **Ritchie MD**, Rader DJ. Exome-wide evaluation of rare coding variants using electronic health records identifies new gene-phenotype associations. Nat Med. 2021 Jan;27(1):66-72. PMID: 33432171.

315. Bone WP, Siewert KM, Jha A, Klarin D, Damrauer SM; VA Million Veteran Program, Chang KM, Tsao PS, Assimes TL, **Ritchie MD**, Voight BF. Multi-trait association studies discover pleiotropic loci between Alzheimer's disease and cardiometabolic traits. Alzheimers Res Ther. 2021 Feb 4;13(1):34. doi: 10.1186/s13195-021-00773-z. PMID: 33541420

316. Sun L, Surya S, Le AN, Desai H, Doucette A, Gabriel P, **Ritchie MD**, Rader D, Maillard I, Bange E, Huang AC, Vonderheide RH, DeMichele A, Verma A, Mamtani R, Maxwell KN. Rates of COVID-19-Related Outcomes in Cancer Compared With Noncancer Patients. JNCI Cancer Spectr. 2021 Jan 21;5(1):pkaa120. doi: 10.1093/jncics/pkaa120. eCollection 2021 Feb. PMID: 33554040

317. Horowitz JE, Kosmicki JA, Damask A, Sharma D, Roberts GHL, Justice AE, Banerjee N, Coignet MV, Yadav A, Leader JB, Marcketta A, Park DS, Lanche R, Maxwell E, Knight SC, Bai

X, Guturu H, Sun D, Baltzell A, Kury FSP, Backman JD, Girshick AR, O'Dushlaine C, McCurdy SR, Partha R, Mansfield AJ, Turissini DA, Li AH, Zhang M, Mbatchou J, Watanabe K, Gurski L, McCarthy SE, Kang HM, Dobbyn L, Stahl E, Verma A, Sirugo G, **Ritchie MD**, Jones M, Balasubramanian S, Siminovitch K, Salerno WJ, Shuldiner AR, Rader DJ, Mirshahi T, Locke AE, Marchini J, Overton JD, Carey DJ, Habegger L, Cantor MN, Rand KA, Hong EL, Reid JG, Ball CA, Baras A, Abecasis GR, Ferreira MA. Common genetic variants identify targets for COVID-19 and individuals at high risk of severe disease. medRxiv. 2021 Feb 19:2020.12.14.20248176. doi: 10.1101/2020.12.14.20248176. Preprint. PMID: 33619501

318. Drivas TG, Lucas A, Zhang X, Ritchie MD. Mendelian pathway analysis of laboratory traits reveals distinct roles for ciliary subcompartments in common disease pathogenesis. Am J Hum Genet. 2021 Mar 4;108(3):482-501. doi: 10.1016/j.ajhg.2021.02.008. Epub 2021 Feb 25. PMID: 33636100

319. Liu X, Verma A, Ramage H, Garcia G, Myers RL, Lucas A, Michaelson JJ, Coryell W, Kumar A, Charney A, Kazanietz MG, Rader DJ, **Ritchie MD**, Berrettini WH, Damoiseaux R, Arumugaswami V, Schultz D, Cherry S, Klein PS. Targeting the Coronavirus Nucleocapsid Protein through GSK-3 Inhibition. medRxiv. 2021 Feb 22:2021.02.17.21251933. doi: 10.1101/2021.02.17.21251933. Preprint. PMID: 33655282

320. Kosmicki JA, Horowitz JE, Banerjee N, Lanche R, Marcketta A, Maxwell E, Bai X, Sun D, Backman JD, Sharma D, Kang HM, O'Dushlaine C, Yadav A, Mansfield AJ, Li AH, Watanabe K, Gurski L, McCarthy SE, Locke AE, Khalid S, O'Keeffe S, Mbatchou J, Chazara O, Huang Y, Kvikstad E, O'Neill A, Nioi P, Parker MM, Petrovski S, Runz H, Szustakowski JD, Wang Q, Wong E, Cordova-Palomera A, Smith EN, Szalma S, Zheng X, Esmaeeli S, Davis JW, Lai YP, Chen X, Justice AE, Leader JB, Mirshahi T, Carey DJ, Verma A, Sirugo G, **Ritchie MD**, Rader DJ, Povysil G, Goldstein DB, Kiryluk K, Pairo-Castineira E, Rawlik K, Pasko D, Walker S, Meynert A, Kousathanas A, Moutsianas L, Tenesa A, Caulfield M, Scott R, Wilson JF, Baillie JK, Butler-Laporte G, Nakanishi T, Lathrop M, Richards JB, Jones M, Balasubramanian S, Salerno W, Shuldiner AR, Marchini J, Overton JD, Habegger L, Cantor MN, Reid JG, Baras A, Abecasis GR, Ferreira MA. A catalog of associations between rare coding variants and COVID-19 outcomes. medRxiv. 2021 Feb 27:2020.10.28.20221804. doi: 10.1101/2020.10.28.20221804. Preprint. PMID: 33655273

321. Lee DSM, Park J, Kromer A, Baras A, Rader DJ, **Ritchie MD,** Ghanem LR, Barash Y. Disrupting upstream translation in mRNAs is associated with human disease. Nat Commun. 2021 Mar 9;12(1):1515. doi: 10.1038/s41467-021-21812-1. PMID: 33750777

322. Shang N, Khan A, Polubriaginof F, Zanoni F, Mehl K, Fasel D, Drawz PE, Carrol RJ, Denny JC, Hathcock MA, Arruda-Olson AM, Peissig PL, Dart RA, Brilliant MH, Larson EB, Carrell DS, Pendergrass S, Verma SS, **Ritchie MD**, Benoit B, Gainer VS, Karlson EW, Gordon AS, Jarvik GP, Stanaway IB, Crosslin DR, Mohan S, Ionita-Laza I, Tatonetti NP, Gharavi AG, Hripcsak G, Weng C, Kiryluk K. Medical records-based chronic kidney disease phenotype for clinical care and "big data" observational and genetic studies. NPJ Digit Med. 2021 Apr 13;4(1):70. doi: 10.1038/s41746-021-00428-1. PMID: 33850243

323. Li B, Veturi Y, Verma A, Bradford Y, Daar ES, Gulick RM, Riddler SA, Robbins GK, Lennox JL, Haas DW, **Ritchie MD**. Tissue specificity-aware TWAS (TSA-TWAS) framework identifies novel associations with metabolic, immunologic, and virologic traits in HIV-positive adults. PLoS Genet. 2021 Apr 26;17(4):e1009464. doi: 10.1371/journal.pgen.1009464. Online ahead of print. PMID: 33901188

324. Chung WK, Brothers K, Bradbury A, Chanprasert S, Orlando L, Torkamani A, Zierhut H, **Ritchie MD**, Phillips M, Schoden J, Maiese D, Hendershot T, Hamilton CM, Ramos EM. Genomic medicine implementation protocols in the PhenX Toolkit: tools for standardized data collection. Genet Med. 2021 Sep;23(9):1783-1788. doi: 10.1038/s41436-021-01183-0. Epub 2021 May 10. PMID: 33972721; PMCID: PMC8108409

325. Hall MA, Wallace J, Lucas AM, Bradford Y, Verma SS, Müller-Myhsok B, Passero K, Zhou J, McGuigan J, Jiang B, Pendergrass SA, Zhang Y, Peissig P, Brilliant M, Sleiman P, Hakonarson H, Harley JB, Kiryluk K, Van Steen K, Moore JH, **Ritchie MD**. Novel EDGE encoding method

enhances ability to identify genetic interactions. PLoS Genet. 2021 Jun 4;17(6):e1009534. doi: 10.1371/journal.pgen.1009534. eCollection 2021 Jun. PMID: 34086673; PMCID: PMC8208534

326. Kosmicki JA, Horowitz JE, Banerjee N, Lanche R, Marcketta A, Maxwell E, Bai X, Sun D, Backman JD, Sharma D, Kury FSP, Kang HM, O'Dushlaine C, Yadav A, Mansfield AJ, Li AH, Watanabe K, Gurski L, McCarthy SE, Locke AE, Khalid S, O'Keeffe S, Mbatchou J, Chazara O, Huang Y, Kvikstad E, O'Neill A, Nioi P, Parker MM, Petrovski S, Runz H, Szustakowski JD, Wang Q, Wong E, Cordova-Palomera A, Smith EN, Szalma S, Zheng X, Esmaeeli S, Davis JW, Lai YP, Chen X, Justice AE, Leader JB, Mirshahi T, Carey DJ, Verma A, Sirugo G, **Ritchie MD**, Rader DJ, Povysil G, Goldstein DB, Kiryluk K, Pairo-Castineira E, Rawlik K, Pasko D, Walker S, Meynert A, Kousathanas A, Moutsianas L, Tenesa A, Caulfield M, Scott R, Wilson JF, Baillie JK, Butler-Laporte G, Nakanishi T, Lathrop M, Richards JB; Regeneron Genetics Center; UKB Exome Sequencing Consortium, Jones M, Balasubramanian S, Salerno W, Shuldiner AR, Marchini J, Overton JD, Habegger L, Cantor MN, Reid JG, Baras A, Abecasis GR, Ferreira MAR. Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals. Am J Hum Genet. 2021 Jul 1;108(7):1350-1355. doi: 10.1016/j.ajhg.2021.05.017. Epub 2021 Jun 3. PMID: 34115965; PMCID: PMC8173480

327. Veturi Y, Lucas A, Bradford Y, Hui D, Dudek S, Theusch E, Verma A, Miller JE, Kullo I, Hakonarson H, Sleiman P, Schaid D, Stein CM, Edwards DRV, Feng Q, Wei WQ, Medina MW, Krauss RM, Hoffmann TJ, Risch N, Voight BF, Rader DJ, **Ritchie MD**. A unified framework identifies new links between plasma lipids and diseases from electronic medical records across large-scale cohorts. Nat Genet. 2021 Jul;53(7):972-981. doi: 10.1038/s41588-021-00879-y. Epub 2021 Jun 17. PMID: 34140684

328. Zhang C, Verma A, Feng Y, Melo MCR, McQuillan M, Hansen M, Lucas A, Park J, Ranciaro A, Thompson S, Rubel MA, Campbell MC, Beggs W, Hirbo J, Mpoloka SW, Mokone GG; Regeneron Genetic Center, Nyambo T, Meskel DW, Belay G, Fokunang C, Njamnshi AK, Omar SA, Williams SM, Rader D, **Ritchie MD**, de la Fuente Nunez C, Sirugo G, Tishkoff S. Impact of natural selection on global patterns of genetic variation, and association with clinical phenotypes, at genes involved in SARS-CoV-2 infection. medRxiv. 2021 Aug 7:2021.06.28.21259529. doi: 10.1101/2021.06.28.21259529. Preprint. PMID: 34230933; PMCID: PMC8259910

329. Eric V, Yi V, Murdock D, Kalla SE, Wu TJ, Sabo A, Li S, Meng Q, Tian X, Murugan M, Cohen M, Kovar C, Wei WQ, Chung WK, Weng C, Wiesner GL, Jarvik GP, Muzny D, Gibbs RA; eMERGE Consortium. Neptune: an environment for the delivery of genomic medicine.Genet Med. 2021 Jul 13. doi: 10.1038/s41436-021-01230-w. Online ahead of print. PMID: 34257418

330. Drivas TG, Lucas A, **Ritchie MD**. eQTpLot: a user-friendly R package for the visualization of colocalization between eQTL and GWAS signals. BioData Min. 2021 Jul 17;14(1):32. doi: 10.1186/s13040-021-00267-6. PMID: 34273980; PMCID: PMC8285863

331. Banday AR, Stanifer ML, Florez-Vargas O, Onabajo OO, Zahoor MA, Papenberg BW, Ring TJ, Lee CH, Andreakos E, Arons E, Barsh G, Biesecker LG, Boyle DL, Burnett-Hartman A, Carrington M, Chang E, Choe PG, Chrisholm RL, Dalgard C, Edberg J, Erdmann N, Feigelson HS, Firestein GS, Gehring AJ, Ho M, Holland S, Hutchinson AA, Im H, Ison MG, Kim HB, Kreitman RJ, Korf BR, Mirabello L, Pacheco JA, Peluso MJ, Rader DJ, Redden DT, **Ritchie MD,** Rosenbloom B, Sant Anna HP, Savage S, Siouti E, Triantafyllia V, Vargas JM, Verma A, Vij V, Wesemann DR, Yeager M, Yu X, Zhang Y, Boulant S, Chanock SJ, Feld JJ, Prokunina-Olsson L. Genetic regulation of OAS1 nonsense-mediated decay underlies association with risk of severe COVID-19. medRxiv. 2021 Jul 13:2021.07.09.21260221. doi: 10.1101/2021.07.09.21260221. Preprint. PMID: 34282422; PMCID: PMC8288155

332. Holmes JH, Beinlich J, Boland MR, Bowles KH, Chen Y, Cook TS, Demiris G, Draugelis M, Fluharty L, Gabriel PE, Grundmeier R, Hanson CW, Herman DS, Himes BE, Hubbard RA, Kahn CE Jr, Kim D, Koppel R, Long Q, Mirkovic N, Morris JS, Mowery DL, **Ritchie MD**, Urbanowicz R, Moore JH. Why Is the Electronic Health Record So Challenging for Research and Clinical Care? Methods Inf Med. 2021 May;60(1-02):32-48. doi: 10.1055/s-0041-1731784. Epub 2021 Jul 19. PMID: 34282602

333. Park J, Packard EA, Levin MG, Judy RL; Regeneron Genetics Center, Damrauer SM, Day SM, **Ritchie MD**, Rader DJ. A genome-first approach to rare variants in hypertrophic cardiomyopathy genes MYBPC3 and MYH7 in a medical biobank. Hum Mol Genet. 2021 Sep 20:ddab249. doi: 10.1093/hmg/ddab249. Online ahead of print. PMID: 34542152

334. Liu X, Verma A, Garcia G Jr, Ramage H, Lucas A, Myers RL, Michaelson JJ, Coryell W, Kumar A, Charney AW, Kazanietz MG, Rader DJ, **Ritchie MD**, Berrettini WH, Schultz DC, Cherry S, Damoiseaux R, Arumugaswami V, Klein PS. Targeting the coronavirus nucleocapsid protein through GSK-3 inhibition. Proc Natl Acad Sci U S A. 2021 Oct 19;118(42):e2113401118. PMID: 34593624

335. Hartwell EE, Merikangas AK, Verma SS, **Ritchie MD**; Regeneron Genetics Center, Kranzler HR, Kember RL. Genetic liability for substance use associated with medical comorbidities in electronic health records of African- and European-ancestry individuals. Addict Biol. 2022 Jan;27(1):e13099. PMID: 34611967

336. Verma A, Tsao N, Thomann L, Ho YL, Iyengar S, Luoh SW, Carr R, Crawford D, Efird JT, Huffman J, Hung A, Ivey K, Levin M, Lynch J, Natarajan P, Pyarajan S, Bick A, Costa L, Genovese G, Hauger R, Madduri R, Pathak G, Polimanti R, Voight B, Vujkovic M, Zekavat M, Zhao H, **Ritchie MD**; VA Million Veteran Program COVID-19 Science Initiative, Chang KM, Cho K, Casas JP, Tsao PS, Gaziano JM, O'Donnell C, Damrauer S, Liao K. A Phenome-Wide Association Study of genes associated with COVID-19 severity reveals shared genetics with complex diseases in the Million Veteran Program. medRxiv. 2021 Oct 15:2021.05.18.21257396. doi: 10.1101/2021.05.18.21257396. Preprint. PMID: 34642702

337. Buchanan AH, Manickam K, Meyer MN, Wagner JK, Hallquist MLG, Williams JL, Rahm AK, Williams MS, Chen ZE, Shah CK, Garg TK, Lazzeri AL, Schwartz MLB, Lindbuchler AM, Fan AL, Leeming R, Servano PO 3rd, Smith AL, Vogel VG, Abul-Husn NS, Dewey FE, Lebo MS, Mason-Suares HM, **Ritchie MD,** Davis FD, Carey DJ, Feinberg DT, Faucett WA, Ledbetter DH, Murray MF. Correction to: Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. Genet Med. 2021 Dec;23(12):2470. PMID: 34646007

338. Abramoff BA, Dillingham TR, Caldera FE, **Ritchie MD,** Pezzin LE. Inpatient Rehabilitation Outcomes After Severe COVID-19 Infections: A Retrospective Cohort Study. Am J Phys Med Rehabil. 2021 Dec 1;100(12):1109-1114. PMID: 34657085

339. Li B, **Ritchie MD**. From GWAS to Gene: Transcriptome-Wide Association Studies and Other Methods to Functionally Understand GWAS Discoveries. Front Genet. 2021 Sep 30;12:713230. PMID: 34659337

340. Arthur VL, Li Z, Cao R, Oetting WS, Israni AK, Jacobson PA, **Ritchie MD**, Guan W, Chen J. A Multi-Marker Test for Analyzing Paired Genetic Data in Transplantation. Front Genet. 2021 Oct 13;12:745773. PMID: 34721531

341. Graham SE, Clarke SL, Wu KH, Kanoni S, Zajac GJM, Ramdas S, Surakka I, Ntalla I, … **Ritchie MD**, … Mohlke KL, Frayling TM, Hirschhorn JN, Kathiresan S; VA Million Veteran Program; Global Lipids Genetics Consortium*, Boehnke M, Natarajan P, Peloso GM, Brown CD, Morris AP, Assimes TL, Deloukas P, Sun YV, Willer CJ. The power of genetic diversity in genome-wide association studies of lipids. Nature. 2021 Dec;600(7890):675-679. PMID: 34887591

342. Wang L, Desai H, Verma SS, Le A, Hausler R, Verma A, Judy R, Doucette A, Gabriel PE; Regeneron Genetics Center, Nathanson KL, Damrauer SM, Mowery DL, **Ritchie MD,** Kember RL, Maxwell KN. Performance of polygenic risk scores for cancer prediction in a racially diverse academic biobank. Genet Med. 2021 Nov 30:S1098-3600(21)05367-3. PMID: 34906489

343. Haas DW, Abdelwahab MT, van Beek SW, Baker P, Maartens G, Bradford Y, **Ritchie MD**, Wasserman S, Meintjes G, Beeri K, Gandhi NR, Svensson EM, Denti P, Brust JCM. Pharmacogenetics of Between-Individual Variability in Plasma Clearance of Bedaquiline and Clofazimine in South Africa. J Infect Dis. 2022 Jan 29:jiac024. PMID: 35091749

344. Bellomo TR, Bone WP, Chen BY, Gawronski KAB, Zhang D, Park J, Levin M, Tsao N, Klarin D, Lynch J, Assimes TL, Gaziano JM, Wilson PW, Cho K, Vujkovic M, O'Donnell CJ, Chang KM, Tsao PS, Rader DJ, **Ritchie MD,** Damrauer SM, Voight BF. Multi-Trait Genome-Wide Association Study of Atherosclerosis Detects Novel Pleiotropic Loci. Front Genet. 2022 Feb 2;12:787545. PMID: 35186008

345. Hartwell EE, Merikangas AK, Verma SS, **Ritchie MD**; Regeneron Genetics Center, Kranzler HR, Kember RL. Genetic liability for substance use associated with medical comorbidities in electronic health records of African- and European-ancestry individuals. Addict Biol. 2022 Jan;27(1):e13099. doi: 10.1111/adb.13099. Epub 2021 Oct 5. PMID: 34611967, PMCID: PMC9254745

346. Wang L, Desai H, Verma SS, Le A, Hausler R, Verma A, Judy R, Doucette A, Gabriel PE; Regeneron Genetics Center, Nathanson KL, Damrauer SM, Mowery DL, **Ritchie MD**, Kember RL, Maxwell KN. Performance of polygenic risk scores for cancer prediction in a racially diverse academic biobank. Genet Med. 2022 Mar;24(3):601-609. doi: 10.1016/j.gim.2021.10.015. Epub 2021 Nov 30. PMID: 34906489

347. Park J, Packard EA, Levin MG, Judy RL; Regeneron Genetics Center, Damrauer SM, Day SM, **Ritchie MD**, Rader DJ. A genome-first approach to rare variants in hypertrophic cardiomyopathy genes MYBPC3 and MYH7 in a medical biobank. Hum Mol Genet. 2022 Mar 3;31(5):827-837. doi: 10.1093/hmg/ddab249. PMID: 34542152, PMCID: PMC8895730

348. Horowitz JE, Kosmicki JA, Damask A, Sharma D, Roberts GHL, Justice AE, Banerjee N, Coignet MV, Yadav A, Leader JB, Marcketta A, Park DS, Lanche R, Maxwell E, Knight SC, Bai X, Guturu H, Sun D, Baltzell A, Kury FSP, Backman JD, Girshick AR, O'Dushlaine C, McCurdy SR, Partha R, Mansfield AJ, Turissini DA, Li AH, Zhang M, Mbatchou J, Watanabe K, Gurski L, McCarthy SE, Kang HM, Dobbyn L, Stahl E, Verma A, Sirugo G; Regeneron Genetics Center, **Ritchie MD**, Jones M, Balasubramanian S, Siminovitch K, Salerno WJ, Shuldiner AR, Rader DJ, Mirshahi T, Locke AE, Marchini J, Overton JD, Carey DJ, Habegger L, Cantor MN, Rand KA, Hong EL, Reid JG, Ball CA, Baras A, Abecasis GR, Ferreira MAR. Genome-wide analysis provides genetic evidence that ACE2 influences COVID-19 risk and yields risk scores associated with severe disease. Nat Genet. 2022 Apr;54(4):382-392. doi: 10.1038/s41588-021-01006-7. Epub 2022 Mar 3. PMID: 35241825, PMCID: PMC9005345

349. Verma A, Tsao NL, Thomann LO, Ho YL, Iyengar SK, Luoh SW, Carr R, Crawford DC, Efird JT, Huffman JE, Hung A, Ivey KL, Levin MG, Lynch J, Natarajan P, Pyarajan S, Bick AG, Costa L, Genovese G, Hauger R, Madduri R, Pathak GA, Polimanti R, Voight B, Vujkovic M, Zekavat SM, Zhao H, **Ritchie MD**; VA Million Veteran Program COVID-19 Science Initiative, Chang KM, Cho K, Casas JP, Tsao PS, Gaziano JM, O'Donnell C, Damrauer SM, Liao KP. A Phenome-Wide Association Study of genes associated with COVID-19 severity reveals shared genetics with complex diseases in the Million Veteran Program. PLoS Genet. 2022 Apr 28;18(4):e1010113. doi: 10.1371/journal.pgen.1010113. eCollection 2022 Apr. PMID: 35482673, PMCID: PMC9049369

350. Wen J, Fu CHY, Tosun D, Veturi Y, Yang Z, Abdulkadir A, Mamourian E, Srinivasan D, Skampardoni I, Singh A, Nawani H, Bao J, Erus G, Shou H, Habes M, Doshi J, Varol E, Mackin RS, Sotiras A, Fan Y, Saykin AJ, Sheline YI, Shen L, **Ritchie MD**, Wolk DA, Albert M, Resnick SM, Davatzikos C; iSTAGING consortium, ADNI, BIOCARD, and BLSA. Characterizing Heterogeneity in Neuroimaging, Cognition, Clinical Symptoms, and Genetics Among Patients With Late-Life Depression. JAMA Psychiatry. 2022 May 1;79(5):464-474. doi: 10.1001/jamapsychiatry.2022.0020. PMID: 35262657, PMCID: PMC8908227

351. Cindi Z, Kawuma AN, Maartens G, Bradford Y, Venter F, Sokhela S, Chandiwana N, Wasmann RE, Denti P, Wiesner L, **Ritchie MD**, Haas DW, Sinxadi P. Pharmacogenetics of dolutegravir plasma exposure among Southern Africans living with HIV. J Infect Dis. 2022 May 4:jiac174. doi: 10.1093/infdis/jiac174. Online ahead of print. PMID: 35512135

352. Zhang C, Verma A, Feng Y, Melo MCR, McQuillan M, Hansen M, Lucas A, Park J, Ranciaro A, Thompson S, Rubel MA, Campbell MC, Beggs W, Hirbo J, Wata Mpoloka S, George Mokone G; Regeneron Genetic Center, Nyambo T, Wolde Meskel D, Belay G, Fokunang C, Njamnshi

AK, Omar SA, Williams SM, Rader DJ, **Ritchie MD**, de la Fuente-Nunez C, Sirugo G, Tishkoff SA. Impact of natural selection on global patterns of genetic variation and association with clinical phenotypes at genes involved in SARS-CoV-2 infection. Proc Natl Acad Sci U S A. 2022 May 24;119(21):e2123000119. doi: 10.1073/pnas.2123000119. Epub 2022 May 17. PMID: 35580180, PMCID: PMC9173769

353. Zhang X, Lucas AM, Veturi Y, Drivas TG, Bone WP, Verma A, Chung WK, Crosslin D, Denny JC, Hebbring S, Jarvik GP, Kullo I, Larson EB, Rasmussen-Torvik LJ, Schaid DJ, Smoller JW, Stanaway IB, Wei WQ, Weng C, Ritchie MD. Large-scale genomic analyses reveal insights into pleiotropy across circulatory system diseases and nervous system disorders. Nat Commun. 2022 Jun 14;13(1):3428. doi: 10.1038/s41467-022-30678-w. PMID: 35701404, PMCID: PMC9198016

354. Kousathanas A, Pairo-Castineira E, Rawlik K, Stuckey A, Odhams CA, Walker S, Russell CD, Malinauskas T, Wu Y, Millar J, Shen X, Elliott KS, Griffiths F, Oosthuyzen W, Morrice K, Keating S, Wang B, Rhodes D, Klaric L, Zechner M, Parkinson N, Siddiq A, Goddard P, Donovan S, Maslove D, Nichol A, Semple MG, Zainy T, Maleady-Crowe F, Todd L, Salehi S, Knight J, Elgar G, Chan G, Arumugam P, Patch C, Rendon A, Bentley D, Kingsley C, Kosmicki JA, Horowitz JE, Baras A, Abecasis GR, Ferreira MAR, Justice A, Mirshahi T, Oetjens M, Rader DJ, **Ritchie MD**, Verma A, Fowler TA, Shankar-Hari M, Summers C, Hinds C, Horby P, Ling L, McAuley D, Montgomery H, Openshaw PJM, Elliott P, Walsh T, Tenesa A; GenOMICC investigators; 23andMe investigators; COVID-19 Human Genetics Initiative, Fawkes A, Murphy L, Rowan K, Ponting CP, Vitart V, Wilson JF, Yang J, Bretherick AD, Scott RH, Hendry SC, Moutsianas L, Law A, Caulfield MJ, Baillie JK. Whole-genome sequencing reveals host factors underlying critical COVID-19. Nature. 2022 Jul;607(7917):97-103. doi: 10.1038/s41586-022-04576-6. Epub 2022 Mar 7. PMID: 35255492, PMCID: PMC9259496

355. Liu H, Doke T, Guo D, Sheng X, Ma Z, Park J, Vy HMT, Nadkarni GN, Abedini A, Miao Z, Palmer M, Voight BF, Li H, Brown CD, **Ritchie MD**, Shu Y, Susztak K. Epigenomic and transcriptomic analyses define core cell types, genes and targetable mechanisms for kidney disease. Nat Genet. 2022 Jul;54(7):950-962. doi: 10.1038/s41588-022-01097-w. Epub 2022 Jun 16. PMID: 35710981

356. Wiley K, Findley L, Goldrich M, Rakhra-Burris TK, Stevens A, Williams P, Bult CJ, Chisholm R, Deverka P, Ginsburg GS, Green ED, Jarvik G, Mensah GA, Ramos E, Relling MV, Roden DM, Rowley R, Alterovitz G, Aronson S, Bastarache L, Cimino JJ, Crowgey EL, Del Fiol G, Freimuth RR, Hoffman MA, Jeff J, Johnson K, Kawamoto K, Madhavan S, Mendonca EA, Ohno-Machado L, Pratap S, Taylor CO, **Ritchie MD**, Walton N, Weng C, Zayas-Cabán T, Manolio TA, Williams MS. A research agenda to support the development and implementation of genomics-based clinical informatics tools and resources. J Am Med Inform Assoc. 2022 Jul 12;29(8):1342-1349. doi: 10.1093/jamia/ocac057. PMID: 35485600, PMCID: PMC9277642

357. Chen BY, Bone WP, Lorenz K, Levin M, Ritchie MD, Voight BF. ColocQuiaL: A QTL-GWAS colocalization pipeline. Bioinformatics. 2022 Jul 27;38(18):4409-11. doi: 10.1093/bioinformatics/btac512. Online ahead of print. PMID: 35894642, PMCID: PMC9477517

358. Banday AR, Stanifer ML, Florez-Vargas O, Onabajo OO, Papenberg BW, Zahoor MA, Mirabello L, Ring TJ, Lee CH, Albert PS, Andreakos E, Arons E, Barsh G, Biesecker LG, Boyle DL, Brahier MS, Burnett-Hartman A, Carrington M, Chang E, Choe PG, Chisholm RL, Colli LM, Dalgard CL, Dude CM, Edberg J, Erdmann N, Feigelson HS, Fonseca BA, Firestein GS, Gehring AJ, Guo C, Ho M, Holland S, Hutchinson AA, Im H, Irby L, Ison MG, Joseph NT, Kim HB, Kreitman RJ, Korf BR, Lipkin SM, Mahgoub SM, Mohammed I, Paschoalini GL, Pacheco JA, Peluso MJ, Rader DJ, Redden DT, **Ritchie MD**, Rosenblum B, Ross ME, Anna HPS, Savage SA, Sharma S, Siouti E, Smith AK, Triantafyllia V, Vargas JM, Vargas JD, Verma A, Vij V, Wesemann DR, Yeager M, Yu X, Zhang Y, Boulant S, Chanock SJ, Feld JJ, Prokunina-Olsson L. Genetic regulation of OAS1 nonsense-mediated decay underlies association with COVID-19 hospitalization in patients of European and African ancestries. Nat Genet. 2022 Aug;54(8):1103-1116. doi: 10.1038/s41588-022-01113-z. Epub 2022 Jul 14. PMID: 35835913, PMCID: PMC9355882

359. Tcheandjieu C, Zhu X, Hilliard AT, Clarke SL, Napolioni V, Ma S, Lee KM, Fang H, Chen F, Lu Y, Tsao NL, Raghavan S, Koyama S, Gorman BR, Vujkovic M, Klarin D, Levin MG, Sinnott-Armstrong N, Wojcik GL, Plomondon ME, Maddox TM, Waldo SW, Bick AG, Pyarajan S, Huang J, Song R, Ho YL, Buyske S, Kooperberg C, Haessler J, Loos RJF, Do R, Verbanck M, Chaudhary K, North KE, Avery CL, Graff M, Haiman CA, Le Marchand L, Wilkens LR, Bis JC, Leonard H, Shen B, Lange LA, Giri A, Dikilitas O, Kullo IJ, Stanaway IB, Jarvik GP, Gordon AS, Hebbring S, Namjou B, Kaufman KM, Ito K, Ishigaki K, Kamatani Y, Verma SS, Ritchie MD, Kember RL, Baras A, Lotta LA; Regeneron Genetics Center; CARDIoGRAMplusC4D Consortium; Biobank Japan; Million Veteran Program, Kathiresan S, Hauser ER, Miller DR, Lee JS, Saleheen D, Reaven PD, Cho K, Gaziano JM, Natarajan P, Huffman JE, Voight BF, Rader DJ, Chang KM, Lynch JA, Damrauer SM, Wilson PWF, Tang H, Sun YV, Tsao PS, O'Donnell CJ, Assimes TL. Large-scale genome-wide association study of coronary artery disease in genetically diverse populations. Nat Med. 2022 Aug;28(8):1679-1692. doi: 10.1038/s41591-022-01891-3. Epub 2022 Aug 1.PMID: 35915156, PMCID: PMC9419655

360. COVID-19 Host Genetics Initiative. A first update on mapping the human genetic architecture of COVID-19. Nature. 2022 Aug;608(7921):E1-E10. doi: 10.1038/s41586-022-04826-7. Epub 2022 Aug 3. PMID: 35922517, PMCID: PMC9352569

361. Ramdas S, Judd J, Graham SE, … **Ritchie MD**, Jousilahti P, Salomaa V, Hveem K, Åsvold BO, Kubo M, Kamatani Y, Okada Y, Murakami Y, Kim BJ, Thorsteinsdottir U, Stefansson K, Zhang J, Chen YE, Ho YL, Lynch JA, Tsao PS, Chang KM, Cho K, O'Donnell CJ, Gaziano JM, Wilson P, Mohlke KL, Frayling TM, Hirschhorn JN, Kathiresan S, Boehnke M; Million Veterans Program; Global Lipids Genetics Consortium, Struan Grant, Natarajan P, Sun YV, Morris AP, Deloukas P, Peloso G, Assimes TL, Willer CJ, Zhu X, Brown CD. A multi-layer functional genomic analysis to understand noncoding genetic variation in lipids. Am J Hum Genet. 2022 Aug 4;109(8):1366-1387. doi: 10.1016/j.ajhg.2022.06.012.PMID: 35931049, PMCID: PMC9388392

362. Haas DW, Abdelwahab MT, van Beek SW, Baker P, Maartens G, Bradford Y, **Ritchie MD**, Wasserman S, Meintjes G, Beeri K, Gandhi NR, Svensson EM, Denti P, Brust JCM. Pharmacogenetics of Between-Individual Variability in Plasma Clearance of Bedaquiline and Clofazimine in South Africa. J Infect Dis. 2022 Aug 12;226(1):147-156. doi: 10.1093/infdis/jiac024. PMID: 35091749, PMCID: PMC9373148

363. Chand GB, Singhal P, Dwyer DB, Wen J, Erus G, Doshi J, Srinivasan D, Mamourian E, Varol E, Sotiras A, Hwang G, Dazzan P, Kahn RS, Schnack HG, Zanetti MV, Meisenzahl E, Busatto GF, Crespo-Facorro B, Pantelis C, Wood SJ, Zhuo C, Shinohara RT, Shou H, Fan Y, Koutsouleris N, Kaczkurkin AN, Moore TM, Verma A, Calkins ME, Gur RE, Gur RC, Ritchie MD, Satterthwaite TD, Wolf DH, Davatzikos C. Schizophrenia Imaging Signatures and Their Associations With Cognition, Psychopathology, and Genetics in the General Population. Am J Psychiatry. 2022 Sep;179(9):650-660. doi: 10.1176/appi.ajp.21070686. Epub 2022 Apr 12. PMID: 35410495, PMCID: PMC9444886

364. Lau-Min KS, McKenna D, Asher SB, Bardakjian T, Wollack C, Bleznuck J, Biros D, Anantharajah A, Clark DF, Condit C, Ebrahimzadeh JE, Long JM, Powers J, Raper A, Schoenbaum A, Feldman M, Steinfeld L, Tuteja S, VanZandbergen C, Domchek SM, **Ritchie MD**, Landgraf J, Chen J, Nathanson KL. Impact of integrating genomic data into the electronic health record on genetics care delivery. Genet Med. 2022 Sep 15:S1098-3600(22)00900-5. doi: 10.1016/j.gim.2022.08.009. Online ahead of print. PMID: 36107166

365. Yengo L, Vedantam S, Marouli E, Sidorenko J, Bartell E, Sakaue S, Graff M, Eliasen AU, Jiang Y, Raghavan S, Miao J, Arias JD, Graham SE, Mukamel RE, Spracklen CN, Yin X, Chen SH, Ferreira T, Highland HH, Ji Y, Karaderi T, Lin K, Lüll K, Malden DE, Medina-Gomez C, Machado M, Moore A, Rüeger S, Sim X, Vrieze S, Ahluwalia TS, Akiyama M, Allison MA, Alvarez M, Andersen MK, Ani A, Appadurai V, Arbeeva L, Bhaskar S, Bielak LF, Bollepalli S, Bonnycastle LL, Bork-Jensen J, Bradfield JP, Bradford Y, Braund PS, Brody JA, Burgdorf KS, Cade BE, Cai H, Cai Q, Campbell A, Cañadas-Garre M, Catamo E, Chai JF, Chai X, Chang LC, Chang YC, Chen CH, Chesi A, Choi SH, Chung RH, Cocca M, Concas MP, Couture C, Cuellar-Partida G, Danning R, Daw EW, Degenhard F, Delgado GE, Delitala A, Demirkan A, Deng X,

Devineni P, Dietl A, Dimitriou M, Dimitrov L, Dorajoo R, Ekici AB, Engmann JE, Fairhurst-Hunter Z, Farmaki AE, Faul JD, Fernandez-Lopez JC, Forer L, Francescatto M, Freitag-Wolf S, Fuchsberger C, Galesloot TE, Gao Y, Gao Z, Geller F, Giannakopoulou O, Giulianini F, Gjesing AP, Goel A, Gordon SD, Gorski M, Grove J, Guo X, Gustafsson S, Haessler J, Hansen TF, Havulinna AS, Haworth SJ, He J, Heard-Costa N, … A saturated map of common genetic variants associated with human height. Nature. 2022 Oct;610(7933):704-712. doi: 10.1038/s41586-022-05275-y. Epub 2022 Oct 12. PMID: 36224396. PMCID: PMC9605867.

366. Ding Z, Ritchie MD, Voight BF, Hwang WT. Estimating the effect size of a hidden causal factor between SNPs and a continuous trait: a mediation model approach. BMC Bioinformatics. 2022 Oct 13;23(1):420. doi: 10.1186/s12859-022-04977-4. PMID: 36229773. PMCID: PMC9559019
367. Li B, Sangkuhl K, Keat K, Whaley RM, Woon M, Verma S, Dudek S, Tuteja S, Verma A, Whirl-Carrillo M, Ritchie MD, Klein TE. How to Run the Pharmacogenomics Clinical Annotation Tool (PharmCAT). Clin Pharmacol Ther. 2022 Nov 9. doi: 10.1002/cpt.2790. Online ahead of print. PMID: 36350094
368. Levin MG, Tsao NL, Singhal P, Liu C, Vy HMT, Paranjpe I, Backman JD, Bellomo TR, Bone WP, Biddinger KJ, Hui Q, Dikilitas O, Satterfield BA, Yang Y, Morley MP, Bradford Y, Burke M, Reza N, Charest B; Regeneron Genetics Center; Judy RL, Puckelwartz MJ, Hakonarson H, Khan A, Kottyan LC, Kullo I, Luo Y, McNally EM, Rasmussen-Torvik LJ, Day SM, Do R, Phillips LS, Ellinor PT, Nadkarni GN, Ritchie MD, Arany Z, Cappola TP, Margulies KB, Aragam KG, Haggerty CM, Joseph J, Sun YV, Voight BF, Damrauer SM. Genome-wide association and multi-trait analyses characterize the common genetic architecture of heart failure. Nat Commun. 2022 Nov 14;13(1):6914. doi: 10.1038/s41467-022-34216-6. PMID: 36376295. PMCID: PMC9663424
369. Truong VQ, Woerner JA, Cherlin TA, Bradford Y, Lucas AM, Okeh CC, Shivakumar MK, Hui DH, Kumar R, Pividori M, Jones SC, Bossa AC, Turner SD, Ritchie MD, Verma SS. Quality Control Procedures for Genome-Wide Association Studies. Curr Protoc. 2022 Nov;2(11):e603. doi: 10.1002/cpz1.603. PMID: 36441943
370. Verma SS, Keat K, Li B, Hoffecker G, Risman M; Regeneron Genetics Center; Sangkuhl K, Whirl-Carrillo M, Dudek S, Verma A, Klein TE, Ritchie MD, Tuteja S. Evaluating the frequency and the impact of pharmacogenetic alleles in an ancestrally diverse Biobank population. J Transl Med. 2022 Nov 28;20(1):550. doi: 10.1186/s12967-022-03745-5. PMID: 36443877. PMCID: PMC9703665
371. Park J, MacLean MT, Lucas AM, Torigian DA, Schneider CV, Cherlin T, Xiao B, Miller JE, Bradford Y, Judy RL; Regeneron Genetics Center; Verma A, Damrauer SM, Ritchie MD, Witschey WR, Rader DJ. Exome-wide association analysis of CT imaging-derived hepatic fat in a medical biobank. Cell Rep Med. 2022 Dec 20;3(12):100855. doi: 10.1016/j.xcrm.2022.100855. Epub 2022 Dec 12. PMID: 36513072. PMCID: PMC9798024.
372. Keat K, Hui D, Xiao B, Bradford Y, Cindi Z, Daar ES, Gulick R, Riddler SA, Sinxadi P, Haas DW, Ritchie MD. Leveraging Multi-Ancestry Polygenic Risk Scores for Body Mass Index to Predict Antiretroviral Therapy-Induced Weight Gain. Pac Symp Biocomput. 2023;28:233-244. PMID: 36540980
373. Hui D, Xiao B, Dikilitas O, Freimuth RR, Irvin MR, Jarvik GP, Kottyan L, Kullo I, Limdi NA, Liu C, Luo Y, Namjou B, Puckelwartz MJ, Schaid D, Tiwari H, Wei WQ, Verma S, Kim D, Ritchie MD. Quantifying factors that affect polygenic risk score performance across diverse ancestries and age groups for body mass index. Pac Symp Biocomput. 2023;28:437-448. PMID: 36540998. PMCID: PMC10018532
374. Verma A, Damrauer SM, Naseer N, Weaver J, Kripke CM, Guare L, Sirugo G, Kember RL, Drivas TG, Dudek SM, Bradford Y, Lucas A, Judy R, Verma SS, Meagher E, Nathanson KL, Feldman M, Ritchie MD, Rader DJ, For The Penn Medicine BioBank. The Penn Medicine BioBank: Towards a Genomics-Enabled Learning Healthcare System to Accelerate Precision Medicine in a Diverse Population. J Pers Med. 2022 Nov 29;12(12):1974. doi: 10.3390/jpm12121974. PMID: 36556195. PMCID: PMC9785650
375. Kanoni S, Graham SE, Wang Y, Surakka I, Ramdas S, Zhu X, Clarke SL, Bhatti KF, Vedantam S, Winkler TW, Locke AE, Marouli E, Zajac GJM, Wu KH, Ntalla I, Hui Q, Klarin D, Hilliard AT, Wang Z, Xue C, Thorleifsson G, Helgadottir A, Gudbjartsson DF, Holm H, Olafsson I, Hwang MY, Han S, Akiyama M, Sakaue S, Terao C, Kanai M, Zhou W, Brumpton BM, Rasheed H, Havulinna AS, Veturi Y, Pacheco JA, Rosenthal EA, Lingren T, Feng Q, Kullo IJ,

Narita A, Takayama J, Martin HC, Hunt KA, Trivedi B, Haessler J, Giulianini F, Bradford Y, Miller JE, Campbell A, Lin K, Millwood IY, Rasheed A, Hindy G, Faul JD, Zhao W, Weir DR, Turman C, Huang H, Graff M, Choudhury A, Sengupta D, Mahajan A, Brown MR, Zhang W, Yu K, Schmidt EM, Pandit A, Gustafsson S, Yin X, Luan J, Zhao JH, Matsuda F, Jang HM, Yoon K, Medina-Gomez C, Pitsillides A, Hottenga JJ, Wood AR, Ji Y, Gao Z, Haworth S, Yousri NA, Mitchell RE, Chai JF, Aadahl M, Bjerregaard AA, Yao J, Manichaikul A, Hwu CM, Hung YJ, Warren HR, Ramirez J, Bork-Jensen J, Kårhus LL, Goel A, Sabater-Lleal M, Noordam R, Mauro P, Matteo F, McDaid AF, Marques-Vidal P, Wielscher M, Trompet S, Sattar N, Møllehave LT, Munz M, Zeng L, Huang J, Yang B, Poveda A, Kurbasic A, L… Implicating genes, pleiotropy, and sexual dimorphism at blood lipid loci through multi-ancestry meta-analysis. Genome Biol. 2022 Dec 27;23(1):268. doi: 10.1186/s13059-022-02837-1. PMID: 36575460. PMCID: PMC9793579

376. Hui D, Mehrabi S, Quimby AE, Chen T, Chen S, Park J, Li B; Regeneron Genetics Center; Penn Medicine Biobank; Ruckenstein MJ, Rader DJ, Ritchie MD, Brant JA, Epstein DJ, Mathieson I. Gene burden analysis identifies genes associated with increased risk and severity of adult-onset hearing loss in a diverse hospital-based cohort. PLoS Genet. 2023 Jan 19;19(1):e1010584. doi: 10.1371/journal.pgen.1010584. eCollection 2023 Jan. PMID: 36656851. PMCID: PMC9888707

377. Zhou W, Kanai M, Wu KH, Rasheed H, Tsuo K, Hirbo JB, Wang Y, Bhattacharya A, Zhao H, Namba S, Surakka I, Wolford BN, Lo Faro V, Lopera-Maya EA, Läll K, Favé MJ, Partanen JJ, Chapman SB, Karjalainen J, Kurki M, Maasha M, Brumpton BM, Chavan S, Chen TT, Daya M, Ding Y, Feng YA, Guare LA, Gignoux CR, Graham SE, Hornsby WE, Ingold N, Ismail SI, Johnson R, Laisk T, Lin K, Lv J, Millwood IY, Moreno-Grau S, Nam K, Palta P, Pandit A, Preuss MH, Saad C, Setia-Verma S, Thorsteinsdottir U, Uzunovic J, Verma A, Zawistowski M, Zhong X, Afifi N, Al-Dabhani KM, Al Thani A, Bradford Y, Campbell A, Crooks K, de Bock GH, Damrauer SM, Douville NJ, Finer S, Fritsche LG, Fthenou E, Gonzalez-Arroyo G, Griffiths CJ, Guo Y, Hunt KA, Ioannidis A, Jansonius NM, Konuma T, Lee MTM, Lopez-Pineda A, Matsuda Y, Marioni RE, Moatamed B, Nava-Aguilar MA, Numakura K, Patil S, Rafaels N, Richmond A, Rojas-Muñoz A, Shortt JA, Straub P, Tao R, Vanderwerff B, Vernekar M, Veturi Y, Barnes KC, Boezen M, Chen Z, Chen CY, Cho J, Smith GD, Finucane HK, Franke L, Gamazon ER, Ganna A, Gaunt TR, Ge T, Huang H, Huffman J, Katsanis N, Koskela JT, Lajonchere C, Law MH, Li L, Lindgren CM, Loos RJF, MacGregor S, Matsuda K, Olsen CM… Global Biobank Meta-analysis Initiative: Powering genetic discovery across human disease. Cell Genom. 2022 Oct 12;2(10):100192. doi: 10.1016/j.xgen.2022.100192. eCollection 2022 Oct 12. PMID: 36777996. PMCID: PMC9903716

378. Small AM, Peloso GM, Linefsky J, Aragam J, Galloway A, Tanukonda V, Wang LC, Yu Z, Sunitha Selvaraj M, Farber-Eger EH, Baker MT, Setia-Verma S, Lee SSK, Preuss M, Ritchie MD, Damrauer SM, Rader DJ, Wells QS, Loos R, Lubitz SA, Thanassoulis G, Cho K, Wilson PWF; VA Million Veteran Program; Natarajan P, O'Donnell CJ. Multiancestry Genome-Wide Association Study of Aortic Stenosis Identifies Multiple Novel Loci in the Million Veteran Program. Circulation. 2023 Mar 21;147(12):942-955. doi: 10.1161/CIRCULATIONAHA.122.061451. Epub 2023 Feb 20. PMID: 36802703

379. Singhal P, Veturi Y, Dudek SM, Lucas A, Frase A, van Steen K, Schrodi SJ, Fasel D, Weng C, Pendergrass R, Schaid DJ, Kullo IJ, Dikilitas O, Sleiman PMA, Hakonarson H, Moore JH, Williams SM, Ritchie MD, Verma SS. Evidence of epistasis in regions of long-range linkage disequilibrium across five complex diseases in the UK Biobank and eMERGE datasets. Am J Hum Genet. 2023 Apr 6;110(4):575-591. doi: 10.1016/j.ajhg.2023.03.007. PMID: 37028392

380. Suzuki K, Hatzikotoulas K, Southam L, Taylor HJ, Yin X, Lorenz KM, Mandla R, Huerta-Chagoya A, Rayner NW, Bocher O, Ana Luiza de SVA, Sonehara K, Namba S, Lee SSK, Preuss MH, Petty LE, Schroeder P, Vanderwerff B, Kals M, Bragg F, Lin K, Guo X, Zhang W, Yao J, Kim YJ, Graff M, Takeuchi F, Nano J, Lamri A, Nakatochi M, Moon S, Scott RA, Cook JP, Lee JJ, Pan I, Taliun D, Parra EJ, Chai JF, Bielak LF, Tabara Y, Hai Y, Thorleifsson G, Grarup N, Sofer T, Wuttke M, Sarnowski C, Gieger C, Nousome D, Trompet S, Kwak SH, Long J, Sun M, Tong L, Chen WM, Nongmaithem SS, Noordam R, Lim VJY, Tam CHT, Joo YY, Chen CH, Raffield LM, Prins BP, Nicolas A, Yanek LR, Chen G, Brody JA, Kabagambe E, An P, Xiang AH, Choi HS, Cade BE, Tan J, Alaine Broadaway K, Williamson A, Kamali Z, Cui J, Adair LS, Adeyemo A, Aguilar-Salinas CA, Ahluwalia TS, Anand SS, Bertoni A, Bork-Jensen J, Brandslund I, Buchanan TA, Burant CF, Butterworth AS, Canouil M, Chan JCN, Chang LC,

Chee ML, Chen J, Chen SH, Chen YT, Chen Z, Chuang LM, Cushman M, Danesh J, Das SK, Janaka de Silva H, Dedoussis G, Dimitrov L, Doumatey AP, Du S, Duan Q, Eckardt KU, Emery LS, Evans DS, Evans MK, Fischer K, Floyd JS, Ford I, Franco OH, Frayling … Multi-ancestry genome-wide study in >2.5 million individuals reveals heterogeneity in mechanistic pathways of type 2 diabetes and complications. medRxiv. 2023 Mar 31:2023.03.31.23287839. doi: 10.1101/2023.03.31.23287839. Preprint. PMID: 37034649. PMCID: PMC10081410

381. Cindi Z, Kawuma AN, Maartens G, Bradford Y, Sokhela S, Chandiwana N, Venter WDF, Wasmann RE, Denti P, Wiesner L, Ritchie MD, Haas DW, Sinxadi P. Pharmacogenetics of tenofovir clearance among Southern Africans living with HIV. Pharmacogenet Genomics. 2023 Jun 1;33(4):79-87. doi: 10.1097/FPC.0000000000000495. Epub 2023 Mar 6. PMID: 37098852
382. Mateza S, Bradford Y, Maartens G, Sokhela S, Chandiwana NC, Venter WDF, Post FA, Ritchie MD, Haas DW, Sinxadi P. Pharmacogenetics of tenofovir renal toxicity in HIV-positive Southern Africans. Pharmacogenet Genomics. 2023 Apr 24. doi: 10.1097/FPC.0000000000000491. Online ahead of print. PMID: 37099271
383. Dwyer DB, Chand GB, Pigoni A, Khuntia A, Wen J, Antoniades M, Hwang G, Erus G, Doshi J, Srinivasan D, Varol E, Kahn RS, Schnack HG, Meisenzahl E, Wood SJ, Zhuo C, Sotiras A, Shinohara RT, Shou H, Fan Y, Schaulfelberger M, Rosa P, Lalousis PA, Upthegrove R, Kaczkurkin AN, Moore TM, Nelson B, Gur RE, Gur RC, Ritchie MD, Satterthwaite TD, Murray RM, Di Forti M, Ciufolini S, Zanetti MV, Wolf DH, Pantelis C, Crespo-Facorro B, Busatto GF, Davatzikos C, Koutsouleris N, Dazzan P. Psychosis brain subtypes validated in first-episode cohorts and related to illness remission: results from the PHENOM consortium. Mol Psychiatry. 2023 May 5. doi: 10.1038/s41380-023-02069-0. Online ahead of print. PMID: 37147389
384. Reza N, Levin MG, Vidula MK, Bravo PE, Damrauer SM, Ritchie MD; Regeneron Genetics Center; Chahal CAA, Owens AT. Prevalence of Pathogenic Variants in Dilated Cardiomyopathy-Associated Genes in Patients Evaluated for Cardiac Sarcoidosis. Circ Genom Precis Med. 2023 May 17:e003850. doi: 10.1161/CIRCGEN.122.003850. Online ahead of print. PMID: 37194596.
385. Singhal P, Verma SS, Ritchie MD. Gene Interactions in Human Disease Studies-Evidence Is Mounting. Annu Rev Biomed Data Sci. 2023 May 17. doi: 10.1146/annurev-biodatasci-102022-120818. Online ahead of print. PMID: 37196359.
386. Raizen DM, Mullington J, Anaclet C, Clarke G, Critchley H, Dantzer R, Davis R, Drew KL, Fessel J, Fuller PM, Gibson EM, Harrington M, Lipkin WI, Klerman EB, Klimas N, Komaroff AL, Koroshetz W, Krupp L, Kuppuswamy A, Lasselin J, Lewis LD, Magistretti PJ, Matos HY, Miaskowski C, Miller AH, Nath A, Nedergaard M, Opp MR, Ritchie MD, Rogulja D, Rolls A, Salamone JD, Saper C, Whittemore V, Wylie G, Younger J, Zee PC, Heller HC. Beyond the Symptom: The Biology of Fatigue. Sleep. 2023 May 24:zsad069. doi: 10.1093/sleep/zsad069. Online ahead of print. PMID: 37224457.

Peer-Reviewed Review Papers

1. Moore JH, **Ritchie MD**. STUDENTJAMA. The challenges of whole-genome approaches to common diseases**.** *JAMA*, 291:1642-1643 (2004). PMID: 15069055
2. **Ritchie MD**. Trends in genomic variation: a view of some of the latest technologies. *Drug Discovery Today*, 10(21):1417-1418 (2005). PMID: 16243260
3. **Ritchie MD**. Bioinformatics approaches for detecting gene-gene and gene-environment interactions in studies of human disease: a neurological focus. *Neurosurgical Focus*, 19(4):E2 (2005). PMID: 16241104
4. **Ritchie MD**, Motsinger AA. Multifactor Dimensionality Reduction for detecting gene-gene and gene-environment interactions in pharmacogenomics studies. *Pharmacogenomics*, 6(8):823-34 (2005). PMID: 16296945
5. McKinney BA, **Ritchie MD**, Moore JH. Machine learning for detecting gene-gene interactions. *Applied Bioinformatics*, 5:77-88 (2006). PMID: 16722772, PMCID: PMC3244050

6. Motsinger A, Haas D, Hulgan T, **Ritchie M**. Human genomic association studies: a primer for infectious disease specialists. *Journal of Infectious Disease,* 195:1737-44 (2007). PMID: 17492588

7. Crawford DC, **Ritchie MD**, Rieder MJ. Identifying the genotype behind the phenotype: a role model found in VKORC1 and its association with warfarin dosing. *Pharmacogenomics*, 8:487-96 (2007). PMID: 17465713, PMCID: PMC3112050

8. **Ritchie MD**, Edwards TL, Fanelli TJ, Motsinger AA. Genetic heterogeneity is not as threatening as you might think. *Genetic Epidemiology*, 31:797-800 (2007). PMID: 17654613

9. Motsinger AA, **Ritchie MD**, Reif DM. Novel methods for detecting epistasis in pharmacogenomics studies. *Pharmacogenomics*, 8:1229-1241 (2007). PMID: 17924838

10. Motsinger AA, **Ritchie MD**. Neural networks for genetic epidemiology: past, present, and future. *BMC BioData Mining*, 1:3 (2008). doi: 10.1186/1756-0381-1-3. PMID: 18822147, PMCID: PMC2553772

11. Vineis P, Brennan P, Canzian F, Ioannidis JPA, Matullo G, **Ritchie M**, Stromberg U, Taioli E, Thompson J. Expectations and challenges stemming from genome-wide association studies. *Mutagenesis*, 23(6):439-44 (2008). doi: 10.1093/mutage/gen042. Epub 2008 Sep 2. PMID: 18765424

12. Srinivasan BS, Chen J, Cheng C, Conti D, Duan S, Fridley BL, Gu X, Haines JL, Jorgenson E, Kraja A, Lasky-Su J, Li L, Rodin A, Wang D, Province M, and **Ritchie MD**. Methods for analysis in pharmacogenomics: Lessons from the Pharmacogenetics Research Network (PGRN) Analysis Group. *Pharmacogenomics*, 10:243-51 (2009). doi: 10.2217/14622416.10.2.243. PMID: 19207025, PMCID: PMC2737060

13. **Ritchie MD**, Bush WS. Genome simulation approaches for synthesizing in silico datasets for human genomics. *Adv Genet*, 72: 1-24 (2010). doi: 10.1016/B978-0-12-380862-2.00001-1. PMID: 21029846

14. Grady BJ and **Ritchie MD**. Statistical optimization of pharmacogenomics association studies: Key Considerations from Study Design to Analysis. *CPPM*, 9:1; 41-66. 2011 PMID: 21887206, PMCID: PMC3163263

15. Holzinger ER, **Ritchie MD**. Integrating heterogeneous high-throughput data for meta-dimensional pharmacogenomics and disease-related studies. *Pharmacogenomics.* 2012 Jan;13(2):213-22. doi: 10.2217/pgs.11.145. PMID: 22256870, PMCID: PMC3350322

16. **Ritchie MD**. The success of pharmacogenomics in moving genetic association studies from bench to bedside: study design and implementation of precision medicine in the post-GWAS era. *Hum Genet.* 2012 Oct;131(10):1615-26. doi: 10.1007/s00439-012-1221-z. Epub 2012 Aug 25. PMID: 22923055, PMCID: PMC3432217

17. 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):2025-48. doi: 10.2217/pgs.14.144. PMID: 25521360, PMCID: PMC4308414.

18. **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. doi: 10.1038/nrg3868. Epub 2015 Jan 13. Review. PMID: 25582081

19. Klein TE, **Ritchie MD**. PharmCAT: A Pharmacogenomics Clinical Annotation Tool. *Clin Pharmacol Ther*. 2017 Dec 1. PMID: 29194583

20. Verma A, **Ritchie MD**. Current Scope and Challenges in Phenome-Wide Association Studies. *Current Epidemiology Reports*. December 2017, Volume 4, Issue 4, pp 321-329. PMID:29545989; PMCID:PMC5846687

21. Verma SS, **Ritchie MD.** Another Round of "Clue" to Uncover the Mystery of Complex Traits. *Genes* (Basel). 2018 Jan 25;9(2). pii: E61. PMID: 29370075

22. Basile AO, **Ritchie MD**. Informatics and machine learning to define the phenotype. *Expert Rev Mol Diagn.* 2018 Mar;18(3):219-226. doi: 10.1080/14737159.2018.1439380. Epub 2018 Feb 16. PMID:29431517
23. **Ritchie MD**, Van Steen K. The search for gene-gene interactions in genome-wide association studies: challenges in abundance of methods, practical considerations, and biological interpretation. *Ann Transl Med*. 2018 Apr;6(8):157. doi: 10.21037/atm.2018.04.05. PMID:29862246. PMCID: PMC5952010
24. **Ritchie MD,** Large-Scale Analysis of Genetic and Clinical Patient Data. *Annual Rev Biomedical Data Science*. 2018. Vol. 1:263-274.
25. Li R, Chen Y, **Ritchie MD**, Moore JH. Electronic health records and polygenic risk scores for predicting disease risk. *Nat Rev Genet.* 2020 Mar 31. doi: 10.1038/s41576-020-0224-1. PMID: 32235907.

Non-Peer Reviewed Papers/Chapters/Books

1. Moore JH, Thornton TA, **Ritchie MD**. Basic statistics. In: Dracopoli NC, Haines JL, Korf BR, Moir DT, Morton CC, Seidman CE, Seidman JG, Smith DR. (eds). *Current Protocols in Human Genetics*. Wiley-Liss, Inc., New York, A.3M.1-A.3M.10 (2003).
2. **Ritchie MD**, White BC, Parker JS, Hahn LW, Moore JH. Optimization of neural networks using genetic programming improves detection and modeling of gene-gene interactions in studies of human diseases. *Genetic and Evolutionary Computation Conference Proceedings* (2003). PMID: 12846935, PMCID: PMC183838
3. **Ritchie MD**, Moore JH. Biological applications of genetic and evolutionary computation. *Genetic and Evolutionary Computation Conference Workshop Proceedings* (2004).
4. **Ritchie MD**. Model Validation in Biological Applications of Genetic and Evolutionary Computation. *Genetic and Evolutionary Computation Conference Workshop Proceedings* (2004).
5. **Ritchie MD**. A review of computational approaches for detecting interactions. Educational Program. *American Association of Cancer Research* (2005).
6. Wilke RA, Carrillo MW, **Ritchie MD**. Pacific symposium on biocomputing - computational approaches for pharmacogenomics. *Pharmacogenomics*, 6(2):111-3 (2005). PMID: 15882130
7. **Ritchie MD**, Carillo MW, Wilke RA. Computational approaches for pharmacogenomics. *Pacific Symposium on Biocomputing Proceedings.* 2005:245-7. PMID: 15759630
8. Carillo MW, Wilke RA, **Ritchie MD**. Computational approaches for pharmacogenomics. *Pacific Symposium on Biocomputing Proceedings*, 11:544-546 (2006). PMID: 15759630
9. Williams SM, Canter JA, Crawford DC, Moore JH, **Ritchie MD**, Haines JL. Problems with genome-wide association studies. *Science.* 2007 Jun 29;316(5833):1840-2. PMID: 17605173
10. Aguilar-Ruiz J, Moore JH, **Ritchie MD**. Filling the gap between Biology and Computer Science. *BioData Mining*, 1:1 (2008). doi: 10.1186/1756-0381-1-1. PMID: 18822148, PMCID: PMC2547862
11. Haines JL, Crawford DC, **Ritchie MD**. A Primer in Statistical Methods in Genetics. In: Roden DM (ed), *Cardiovascular Genetics and Genomic*s, Blackwell publishing (2009).
12. Pizzuti C, **Ritchie MD**, Giacobini M (eds). *Evolutionary Computation, Machine Learning and Data Mining in Bioinformatics: 7th European Conference, Proceedings EvoBIO 2009*. Springer publishing (2009).
13. Moore JH, Clegg J, Marchiori E, **Ritchie MD**, and Smith S. Artificial Evolution Methods in the Biological and Biomedical Sciences. *JAEA*. 2009.

14. Pizzuti C, **Ritchie MD**, Giacobini M (eds). *Evolutionary Computation, Machine Learning and Data Mining in Bioinformatics: 8th European Conference, Proceedings EvoBIO 2010*. Springer publishing (2010).
15. **Ritchie MD**, Bush WS. Genome Simulation: Approaches for Synthesizing *In Silico* Datasets for Human Genomics. In: *Advances in Genetics, Vol. 72*, Elsevier Inc (2010). doi: 10.1016/B978-0-12-380862-2.00001-1. PMID: 21029846
16. Moore JH, **Ritchie MD**. The central role of biological data mining in connecting diverse disciplines. *BioData Mining* 2013, 6:14. doi: 10.1186/1756-0381-6-14. PMID: 23937773, PMCID: PMC3765075
17. **Ritchie MD,** Reducing Dimensionality in the Search for Gene-Gene Interactions. In: Between the Lines of Genetic Code. *Elsevier Inc* (2014).
18. **Ritchie MD.** Finding the Epistasis Needles in the Genome-Wide Haystack. In: Epistasis: Methods and Protocols, *Methods Mol Biol.* 2015;1253:19-33. doi: 10.1007/978-1-4939-2155-3_2. PMID: 25403525
19. **Ritchie MD,** Study design and analysis approaches in pharmacogenomics research. In: *Pharmacogenomics: Applications to Patient Care*, Third Edition. ACCP (2015).
20. **Ritchie MD**, Moore JH, Kim JH. Translational Bioinformatics: Biobanks in the Precision Medicine Era. *Pac Symp Biocomput*. 2020;25:743-747. PMID: 31797645
21. Moore JH, Barnett I, Boland MR, Chen Y, Demiris G, Gonzalez-Hernandez G, Herman DS, Himes BE, Hubbard RA, Kim D, Morris JS, Mowery DL, **Ritchie MD**, Shen L, Urbanowicz R, Holmes JH. Ideas for how informaticians can get involved with COVID-19 research. Version 2. *BioData Min*. 2020 May 12;13:3. doi: 10.1186/s13040-020-00213-y. eCollection 2020. PMID: 32419848; PMCID: PMC7216865
22. Scott WS, **Ritchie MD** (eds.) Genetic Analysis of Complex Disease, 3rd edition. Wiley, in press 2021. https://www.wiley.com/en-us/Genetic+Analysis+of+Complex+Disease%2C+3rd+Edition-p-9781118123911 ISBN: 978-1-118-12391-1

ABSTRACT CONTRIBUTED PRESENTATIONS

2003 "Optimization of Neural Networks using Genetic Programming to Improve Detection and Modeling of Gene-Gene Interactions in Studies of Human Diseases," Biological Applications in Genetic and Evolutionary Computation, Chicago, IL (Host: Wolfgang Banzhaf, peer-reviewed)

2003 "Optimization of Neural Networks using Genetic Programming to Improve Detection and Modeling of Gene-Gene Interactions in Studies of Human Diseases," Genetic and Evolutionary Computation Conference, Chicago, IL (Host: James Foster)

2004 "How Can We Detect Gene-Gene Interactions in Pharmacogenomic Studies?" Pharmacogenetics Research Network - Statistics Workshop, Los Angeles, CA (Host: Mike Province)
2004 "Model Validation Strategies in Biological Applications of Genetic and Evolutionary Computation," Biological Applications of Genetic and Evolutionary Computation Workshop, Seattle, WA (Host: Jason Moore, peer-reviewed)

2004 "GPNN as a Bioinformatics Tool for Human Genetics," Genetic and Evolutionary Computation Conference, Seattle, WA (Host: James Foster, peer reviewed)

2004 "MDR Reveals Gene-Gene Interactions Associated with Multiple Sclerosis," American Society of Human Genetics, Toronto, Canada, peer-reviewed

2004 "Gene-Gene Interaction Associated with Response to Treatment of Unipolar Major Depression Disorder," Cold Spring Harbor Laboratory: Pharmacogenomics, Cold Spring Harbor, NY, Peer-reviewed

2005 "Computational Approaches for Pharmacogenomics," Session Introduction at the Pacific Symposium on Biocomputing, The Big Island, HI (Host: Russ Altman, peer-reviewed)

2005 "Computational Approaches for Pharmacogenomics," Leader of Discussion Session at the Pacific Symposium on Biocomputing, The Big Island, HI (Host: Russ Altman)

2005 "Can Neural Network Constraints in GP Provide Power to Detect Genes Associated with Human Disease?" European Workshop on Evolutionary Computation and Bioinformatics, Lausanne, Switzerland (Host: Elena Marchiori, peer-reviewed)

2005 "Multifactor Dimensionality Reduction and the Power to Detect Pharmacogenomic Effects in Treatment of Tenureitis," Pharmacogenetics Research Network Statistics Workshop, Chicago, IL (Host: Nancy Cox and Mike Province, peer-reviewed)

2005 "Challenges and Strategies for Whole Genome Association Studies," American Society of Human Genetics, Salt Lake City, UT (Host: David Cox, peer-reviewed) *Given by Dr. Jonathan Haines due to maternity leave*

2006 "Tutorial on Computational Approaches for Pharmacogenomics," Pacific Symposium on Biocomputing, Maui, Hawaii (Host: Larry Hunter, peer-reviewed) *Given by Alison A. Motsinger due to maternity leave*

2006 "GenomeSim: Data Simulation Software for Whole-Genome Association Studies," Pacific Symposium on Biocomputing, Maui, HI (Host: Russ Altman, peer-reviewed) *Given by Alison A. Motsinger due to maternity leave*

2006 "Grammatical Evolution Neural Networks: A Powerful Gene-Gene Interaction Detection Method," Genetic Analysis Workshop Special Session, St. Petersburg, FL (Host: Jean MacCleur, peer-reviewed)

2006 "The Potential of a Sequential Replication Approach for Whole Genome Association Studies," International Genetic Epidemiology Society, Tampa, FL (Host: Jeanne Cashman, peer-reviewed)

2006 "PLATO: A Platform for the Analysis, Translation, and Organization of Large Scale Data," Pharmacogenetics Research Network Analysis Workshop, Nashville, TN (Host: Mike Province, peer-reviewed)

2008 "Thinking about Epistasis in Whole Genome Association Studies," Pharmacogenetics Research Network Analysis Workshop, Memphis, TN (Host: Mary Relling, peer-reviewed)

2009 "Genome-wide association study identifies novel genomic regions associated with drug-induced Long QT Syndrome," CSHL-Wellcome Trust Pharmacogenomics Meeting, Hinxton, UK. (Host: Steve Leeder, peer-reviewed)

2009 "Genome-wide association study identifies novel genomic regions associated with drug-induced Long QT Syndrome," American Society for Human Genetics, Honolulu, HI. (peer-reviewed)

2010 "Using Biological Knowledge to Guide the Search for Epistasis in Genome-Wide Association Studies", American Society of Human Genetics, Washington DC. peer-reviewed

2010 "Chromosome 4q25 variants are genetic modifiers of rare ion channel mutations associated with familial atrial fibrillation", American Society of Human Genetics, Washington DC. peer-reviewed

2010 "Rare and Common Variants for Atrial Fibrillation", PGRN Statistical Analysis Workshop, Nashville, TN. peer-reviewed

2011 "Meta-dimensional analysis of phenotypes", International Congress on Human Genetics, Montreal, Canada, peer-reviewed

2012 "Meta-dimensional Analysis Of Complex Biological Traits Using ATHENA", Genetic Programming Theory in Practice, Ann Arbor, MI. peer-reviewed

2013 "Using Biological Knowledge in Modern Genomic Analysis of Complex Traits", Epistasis Discovery in Genetic Epidemiology (EDGE) 2013 Workshop, Key West, FL

2013 "Next generation analysis of human genome sequence", Pharmacogenomics Research Network Scientific Meeting, Chicago IL. peer-reviewed

2014 "Rare Variants in the Pharmacogenomics Studies ", American Society of Clinical Pharmacology and Therapeutics (ASCPT), Atlanta, GA

2015 "Binning Somatic Mutations based on Biological Knowledge for Predicting Survival: an Application in Renal Cell Carcinoma", Pacific Symposium on Biocomputing (PSB), Big Island, Hawaii

2016 "The Public Sharing of Genomic Data from the DiscovEHR Collaboration", American Society of Human Genetics, Vancouver, Canada

INVITED PRESENTATIONS

2001 "Approaches to Detecting Epistasis in Sporadic Breast Cancer," The Second Annual Genetics Symposium, Meharry Medical College, Nashville, TN (Host: Ellen Wright Clayton)

2001 "An Introduction to Neural Network Models for Statistical Data Analysis," Quantitative Brown Bag Seminar Series, Vanderbilt University, Nashville, TN (Host: Niels Waller)

2001 "Quantitative Resources on the WWW," Quantitative Brown Bag Seminar Series, Vanderbilt University, Nashville, TN (Host: Niels Waller)

2001 "Gene-Gene Interactions in Common, Complex Diseases," Molecular Physiology and Biophysics Seminar Series, Vanderbilt University, Nashville, TN (Host: Alan Cherrington)

2002 "Genetic Programming Optimization of Neural Network Architecture for the Detection of Gene-Gene Interactions," Biomathematics Seminar Series, Vanderbilt University, Nashville, TN (Host: Peter Hinow)

2002 "Neural Networks for Detecting Gene-Gene Interactions," Molecular Physiology and Biophysics Seminar Series, Vanderbilt University, Nashville, TN (Host: Alan Cherrington)

2003 "Data Reduction and Pattern Recognition Approaches for Complex Genetic Analysis," Statistical Genetics Seminar Series, Rockefeller University, New York, NY (Host: Jurg Ott, invited)

2003 "Data Reduction and Pattern Recognition Approaches for Complex Genetic Analysis," Clinical Epidemiology and Biostatistics Faculty Candidate Seminar, University of Pennsylvania, Philadelphia, PA (Host: Tim Rebbeck, invited)

2003 "Data Reduction and Pattern Recognition Approaches for Complex Genetic Analysis," Bioinformatics Faculty Candidate Seminar, Medical University of South Carolina, Charleston, SC (Host: Eberhard Voit, invited)

2003 "Data Reduction and Pattern Recognition Approaches for Complex Genetic Analysis," Human Genetics Faculty Candidate Seminar Series, Wake Forest University, Winston-Salem, NC (Host: Stephen Rich, invited)

2003 "Computational Approaches for Detecting Gene-Gene Interactions," Intelligent Systems for Molecular Biology Conference, Brisbane, Australia (Host: Mark Ragan, invited)

2003 "Optimization of Neural Networks using Genetic Programming to Improve Detection and Modeling of Gene-Gene Interactions in Studies of Human Diseases," National Library of Medicine Training Directors' Conference, Bethesda, MD (Host: Carol Bean, invited)

2003 "Neural Networks for Detecting Gene-Gene Interactions," Epidemiology Faculty Candidate Seminar Series, Wake Forest University, Winston-Salem, NC (Host: Lynne Wagenknecht, invited)

2003 "Data Mining and Pattern Recognition Approaches for Detecting Gene-Gene Interactions," Statistical Genetics Interview Seminar, Bristol-Myers Squibb, Hopewell, NJ (Host: Kim Zerba, invited)

2003 "Computational Approaches for Detecting Gene-Gene Interactions," Molecular Physiology & Biophysics Faculty Candidate Seminar Series, Vanderbilt University, Nashville, TN (Host: Alan Cherrington, Jonathan Haines)

2003 "Neural Networks for Detecting Gene-Gene Interactions," Department of Human Genetics, University of Pittsburgh, Pittsburgh, PA (Host: Bob Ferrell, invited)

2004 "Analysis of Genome-Wide Epistasis via Dimensionality Reduction Procedures," International Biometric Society, Eastern North American Region 2004, Pittsburgh, PA (Host: David Allison, invited)

2004 "How to Look for Gene-Gene Interactions using Multifactor Dimensionality Reduction," Vanderbilt University GCRC Skills Workshop, Nashville, TN (Host: Dan Byrne)

2004 "Genome-Wide Association Studies: Challenges, Strategies, and MDR," Vanderbilt University Genetic Interest Group Seminar Series, Nashville, TN (Host: Kelly McDermott)

2004 "Neural Networks Approaches for Genetic Studies," Marshfield Clinic Research Foundation Scientific Seminar Series, Marshfield, WI (Host: Russell Wilke, invited)

2004 "Dissecting the Genetics of Complex Disease," National Society of Genetic Counselors, Washington, DC, invited

2005 "Parallel Multifactor Dimensionality Reduction: Applications in Rheumatoid Arthritis," Invited Presentation, Universitat Politechnica, Barcelona, Spain (Host: Sara Marsal, invited)

2005 "Computational Approaches to Detecting Interactions," Educational Session on Gene-Gene Interactions in Cancer Etiology at the AACR, Anaheim, CA (Host: Jason Moore, invited)

2005 "Multifactor Dimensionality Reduction for Detecting Epistasis," Joint Conference of the Classification Society of North America and the Interface Foundation of North America, St. Louis, MO (Host: Rob Culverhouse, invited)

2005 "Multifactor Dimensionality Reduction for the Analysis of Pharmacogenomics Data," Beyond Genome 2005: Genomic Variation, San Francisco, CA (Host: Catherine Lisciandra, peer-reviewed)

2005 "Detecting Gene-Gene Interactions Using Multifactor Dimensionality Reduction," Joint Statistical Meetings, Minneapolis, MN (Host: Cheng Cheng, invited)

2005 "Multifactor Dimensionality Reduction for the Analysis of Epistasis in Studies of Human Disease and Pharmacogenomics," Boehringer-Ingelheim Invited Special Seminar, Danbury, CT (Host: Supriya Jayadev, invited)

2005 "Data Mining Approaches in Biological Sciences," Fields Institute on Data Mining, Toronto, Canada (Host: Helmut Kroger, invited)
Given by Will Bush due to maternity leave

2006 "The Complexities of Data Analysis in Human Genetics," Complex Data Conference, Vanderbilt University Kennedy Center, Nashville, TN (Host: Jennifer Blackford, invited)

2006 "Multifactor Dimensionality Reduction for Detecting Gene-Gene and Gene-Environment Interactions," Vanderbilt University Center for AIDS Research, Nashville, TN (Host: Todd Hulgan, invited)

2006 "Challenges and Strategies for Whole-Genome Association Studies," Boehringer Ingelheim Invited Speaker, Danbury, CT (Host: Jing Yuan, invited)

2007 "Computational Genetics Approaches in Cancer Epidemiology," University of Miami Bio-behavioral Oncology and Cancer Control Program Meeting, Miami, FL (Host: Jennifer Hu, invited)

2007 "The MDR Approach," ECNIS WP 7-8 Workshop, "AFTER GWAS: an Exercise in Problem Solving," Venice, Italy (Host: Paolo Vineis, invited)

2007 "Computational Genomics in the Whole Genome Era," University of Virginia, Charlottesville, NC (Host: Stephen Rich, invited)

2008 "Computational Genomics in the Whole Genome Era," Washington University in St. Louis, St. Louis, MO (Host: Mike Province, invited)

2008 "A Bio-filter for Systems Biology," NEWGENERIS Workshop, Athens, Greece (Host: S.A. Kyrtopoulos, invited)

2008 "Epistasis Modeling," Keystone Symposium on Towards Identifying the Pathophysiology of Autistic Syndromes, Santa Fe, NM (Host: Joseph Piven and Pat Levitt, invited)

2008 "Epistasis, Stratification, and Confounding: Lessons from Genomic Analyses," ASCPT, Orlando, FL (Host: Issam Zineh, invited)

2008 "Computational Genomics in the GWAS Era," NIEHS, Raleigh-Durham, NC (Host: Gregg Dinse, invited)

2009 Invited Seminar, University of Alabama Department of Genetics, Birmingham, AL (Host: Hemant Tiwari, invited)

2009 "Computational Strategies for Genome-Wide Association Studies," Korea Pharmacogenetics Research Network Annual Symposium, Seoul, Korea (Host: Ju Han Kim, invited)

2009 "Individual Opt-Out: Efficacy, Challenges, and Concerns," American Society for Human Genetics, Honolulu, HI. (Host: Laura Rodriguez, invited)

2009 "Genome Wide Association Studies," Annual Education Conference of the National Society of Genetic Counselors, Atlanta, GA.

2010 "Summary of the state-of-the-art for gene-gene interaction analysis," CDGCA, Belgium, Host: Kristel Van Steen, invited

2010 "Beyond GWAS: Integrating Transcriptome, Proteome, and Pathway Data in the Genetic Dissection of Complex Traits", Huck Institutes of Life Sciences, Pennsylvania State University, State College, PA

2011 "Pharmacogenetics using the electronic medical record and a large institutional biobank", Dartmouth Medical School, Lebanon, NH

2011 "Bringing Biology Back to Human Genomics", University of Nottingham, Nottingham, UK

2011 "Surfing the Tsunami of Human Genetic Data", University of Idaho, Moscow, ID

2011 "Phenotype-Genotype Associations in DNA Biobanks Linked to Electronic Medical Records", Asia-Pacific Translational Bioinformatics Conference, Seoul, Korea

2011 "Using Biological Knowledge in Modern Genomic Analysis of Complex Traits", University of California at Davis (Host: Isaac Pessah)

2011 "Using biological knowledge to inform complex genetics analysis", NIEHS meeting Bioinformatics and Computational Approaches to Integrate Genes and Environment in Autism Research, Durham, NC

2011 "Beyond GWAS: Integrating Transcriptome, Proteome, and Pathway Data in the Genetic Dissection of Complex Traits", Flash Poster Talk, Kavli Frontiers in Science, National Academy of Science, Irvine, CA

2011 "Genomics and Pharmacogenomics using the electronic medical record and a large institutional biobank", Geisinger Clinic, Weis Center for Research, Danville, PA (Host: David Carey)

2012 "Meta-Dimensional Analysis of Phenotypes to Dissect the Architecture of Complex Traits", Keystone Symposium, Complex Traits: Genomics and Computational Approaches, Breckenridge, CO

2012 "Mining electronic health records for genomic research – experiences of the eMERGE network". Inaugural Symposium of Penn State Hershey Institute for Personalized Medicine, Hershey, PA

2012 "Searching for the missing heritability in gene-gene and gene-environment interactions", University of Georgia Symposium, Methodological and Statistical Issues in Gene-Environment and Gene-Gene Interaction Research, Atlanta, GA

2012 "The phenome and pleiotropy: Dissecting the architecture of complex traits", AACR-MEG Special Conference on Post-GWAS Horizons in Molecular Epidemiology: Digging Deeper into the Environment, Hollywood, FL

2012 "Meta-Dimensional Analysis for Dissecting the Architecture of Complex Traits", University of Liege, Liege, Belgium

2013 "Overview of pharmacogenomics, gene-gene interactions, system genomics", Eastern North America Region International Biometric Society 2013, Orlando, FL

2013 "Analytic approaches for complex trait analysis", American Association of Cancer Research Educational Session, Washington DC

2013 "The Next Frontier in Genomics: Data Analysis Strategies", Ohio State University Comprehensive Cancer Center Annual Scientific Meeting, Columbus OH

2013 "Complex genetic analysis: Searching for the missing heritability", Ohio State University, Columbus OH

2013 "Analysis technologies to go beyond a single gene: the quest for understanding genetic architecture", University of Maryland, Baltimore MD

2013 "Bioinformatics tools for incorporating biological knowledge into genetic analysis", Advances in Statistical Methods for Cancer Genetic Epidemiology. Memorial Sloan-Kettering, New York, NY. Host: Sanjay Shete.

2013 "Bringing Biology back to Genomics". University of Pittsburgh, Pittsburgh, PA. Host: Robert Ferrell.

2013 "The quest for understanding genetic architecture", University of Chicago, Chicago IL. Host: Barbara Stranger.

2014 "Challenges in Modeling Interactions", EDGE: Epistasis Discovery in Genetic Epidemiology, Key West, Florida, Host: Jason Moore.

2014 "Top Ten Challenges in Data Simulations", Genetic Simulation Tools Post-GWAS, National Cancer Institute Workshop, Host: Elizabeth Gillanders and Leah Mechanic

2014 "Making sense of genome-phenome relationships", The Biology of Genomes, Cold Spring Harbor, NY, Host: Carlos Bustamante

2014 "Mining the EMR for genome-phenome associations", Big Data in Biomedicine, Stanford University, Host: Russ Altman

2014 "Data fusion in the omic universe", Mid-Atlantic Genetic Epidemiology & Statistics (MAGES), Children's Hospital of Philadelphia-U Penn, Host: Marcella Devoto

2014 "Big Data", KAVLI Frontiers in Science, Medan Indonesia, Host: Edward Patte

2014 "Exploring the Genetic Architecture of Complex Traits ", UC Davis, Host: Irva Hertz-Piccoto

2014 "Exploring Genome-Phenome Relationships in an Electronic Health Record", CTSI Lecture, Penn State Medical Center, Hershey, PA, Host: Larry Sinoway

2014 "Mining electronic health records and genomics", KOSMI, Pusan S. Korea, Host: Ju Han Kim

2015 "Significance of Gene-Gene Interactions/Epistasis", Pacific Symposium on Biocomputing (PSB), Big Island, Hawaii, Host: Sarah Pendergrass

2015 "Exploration of the EHR Universe: Big Data for Predictive Genomics", Indo-US Bilateral Conferences-cum-Workshop: Big Data Analysis and Translation in Disease Biology (Big Data and Disease)-2015, New Delhi, India, Host: Indira Ghosh

2015 "Genomics and the Electronic Health Record - a dynamic duo made for Precision Medicine", Biomedical Informatics Annual Retreat keynote, Ohio State University, Columbus, Ohio, Host: Phillip Payne

2015 "Genomics and the Electronic Health Record - a dynamic duo made for Precision Medicine", Institute for Computational Biology Symposium, Case-Western Reserve University, Host: Dana Crawford

2015 "A game of Clue – which 'omics are guilty?" IGES-ASHG Joint Symposium, Baltimore, Maryland

2015 "Electronic health records and genomics: A dynamic duo for precision medicine", Presidential Symposium, American Society of Human Genetics, Baltimore, Maryland, Host: Neil Risch

2015 "Using genomics and electronic health records for precision medicine - thinking about the future", Pharmacogenomics Research Network: Research in Progress seminar series, Webinar, Host: Kathy Giacomini

2015 "Exploring the Use of Electronic Health Records and Genomics for Precision Medicine", Biotech Symposia, Simons Foundation and the New York Bioinformatics Group, New York, New York, Host: Doron Betel

2015 "Electronic health records and genomics – a dynamic duo for precision medicine", Integrative Biostatistics Research for Imaging, Genomics, & High-throughput Technologies in Precision Medicine (iBRIGHT), Houston, Texas, Host: Pei Weng

2015 "Electronic health records and genomics – a dynamic duo for precision medicine", Translational Bioinformatics Conference (TBC), Tokyo, Japan, Host: Tomohiro Sawa

2016 "Integrating Genomics into the Electronic Health Record for Precision Medicine", HCSRN Precision Oncology Symposium, Atlanta, Georgia, Host: Larry Kushi

2016 "Precision Medicine – The Beginning of a Long Voyage", Keynote at Annual Retreat, Institute for Biomedical Informatics (IBI) and the Graduate Group in Genomics and Computational Biology (GCB), University of Pennsylvania, Philadelphia, Pennsylvania, Hosts: Li-San Wang and Ben Voight

2016 "Methods for Integrating Data to Uncover Genotype-phenotype Interactions", American Society for Bone and Mineral Research (ASBMR), Atlanta, Georgia, Host: Fernando Rivadeneira

2016 "DNAnexus: The Geisinger Experience", DNAnexus Connect, San Francisco, California, Host: Angela Anderson

2016 "Genomics and Precision Medicine: approaches and challenges", University of Pennsylvania, Philadelphia, Pennsylvania, Host: Dan Rader

2017 "Applications in Multi-omics data integration", No Boundary Thinking Workshop, Pacific Symposium on Biocomputing, Big Island, Hawaii, Host: Jason Moore

2017 "Phenotypes: The next frontier for precision medicine" Precison Medicine Fusion Conference, Cancun, Mexico (my talk by video broadcast), Host: Michael Snyder

2017 "PharmCAT: Pharmacogenomics Clinical Annotation Tool", Precison Medicine Fusion Conference, Cancun, Mexico (my talk by video broadcast), Host: Michael Snyder

2017 "PharmCAT: A Tool for Pharmacogenomics Implementation", Clinical Pharmacogenetics Implementation Consortium Meeting, Washington DC, Host: Mary Relling

2017 "PharmCAT: A Tool for Pharmacogenomics Implementation", NHGRI Genomic Medicine X: Pharmacogenomics, Silver Spring, Maryland, Host: Teri Manolio

2017 "Machine Learning Strategies in the Genome and the Phenome – Toward a Better Understanding of Complex Traits", Department of Biomedical Informatics, Harvard Medical School, Boston, Massachusetts, Host: Isaac Kohane

2017 "Multi-omics data integration", European Society of Human Genetics annual meeting, Copenhagen, Denmark

2017 "Meta-dimensional methods for 'omics data integration", Retreat talk, Department of Genetics, University of Pennsylvania

2017 "Precision Medicine: Study Designs, Tools, and Applications", Tutorial, Translational Bioinformatics Conference, Long Beach, CA, Host: Dokyoon Kim

2017 "What does the future hold in precision medicine?", Illumina workshop, American Society of Human Genetics annual meeting, Orlando, FL. Host: Jay Kaufman

2017 "Machine learning strategies in the Genome and the Phenome", University of Virginia seminar, Host: Charles Farber

2017 "It's All in the Genes: The Science Behind Customizing Patient Care", ASHP midyear clinical meeting and exposition, Ornaldo, FL. Host: Cynthia Von Heerigan

2018 "Machine Learning in the Genome and Phenome to Understand Complex Traits: Discovery Science for Precision Medicine", University of California, San Francisco, PREMIER Symposium. Host: Lindsey Criswell

2018 "PheWAS in Populations: Exploring the relationship between the genome and the phenome", Lorne Genome Conference 2018, Lorne Australia. Host: Traude Beilharz

2018 "Exploring the relationship between the genome and the phenome", Melbourne Integrative Genomics (MIG) at The University of Melbourne. Host: Andrew Siebel

2018 "PheWAS in Populations: Exploring the relationship between the genome and the phenome", Institute for Computational Biomedicine (ICB) seminar, Weill Cornell. Hosts: Drs. Doron Betel and Olivier Elemento

2018 "Experiences from Electronic Health Records (EHRs) and what we can expect in the future", Informing Environmental Health Decisions Through Data Integration Workshop, National Academy of Science, Washington DC, Host: Ben Wender

2018 "Exploring the Genome and the Phenome – Toward a Better Understanding of Complex Traits", Academic Health Center, University of Minnesota, Duluth campus. Host: Catherine McCarty

2018 "Exploring the Genome and the Phenome – Toward a Better Understanding of Complex Traits", 3rd Midwest Bioinformatics Conference, University of Missouri, Columbia, MO. Host: Mark Hoffman.

2018 "Exploring the Genome and the Phenome with Machine Learning", Copenhagen Novo Nordisk Foundation (NNF) symposium. Copenhagen, Denmark. Host: Ramneek Gupta

2018 "Using your spidey sense to guide research using machine learning in electronic health records", Denmark Technical University, Big Data Workshop, Copenhagen, Denmark, Host: Ramneek Gupta

2018 "Pharmacogenomics: Personalizing Medicine since 2005" Keynote at Penn 2nd annual precision medicine symposium, Philadelphia PA, Host: David Roth

2018 "Gene-Environment interaction = the missing piece of precision medicine?", Total Exposure Health 2018 Conference, Bethesda, MD, Host Justin Teeguarden

2018 "Machine Learning Strategies in the Genome and the Phenome – Toward a Better Understanding of Complex Traits", NY Genome Center, New York, NY, Host: Nicolas Robine

2018 "Exploring the World of Phenomics", IGES Educational Symposium, San Diego, CA, Host: Stephanie A. Santorico

2018 "Meta-dimensional analysis of multi-omics data", CHOP/UPENN Mid Atlantics Bioinformatics conference, Philadelphia, PA, Host: Kai Wang

2018 "Exploring the world of phenomics in electronic health records", CIFAR Genetic Networks symposium, Toronto, Canada, Host: Charlie Boone

2019 "Training the next generation of biomedical scientists for success in today's world" Pacific Symposium on Biocomputing, Big Island, HI, Host: Teri Klein

2019 "Machine Learning Strategies in the Genome and the Phenome – Toward a Better Understanding of Complex Traits", Vanderbilt University, Nashville, TN, Host: Nancy Cox

2019 "Exploring meta-dimensional models of multi-omics data using machine learning" NMBIST, Sante Fe, NM, Host: Lisana Chavez

2019 "Phenomics in Electronic Health Records", CHOP Scientific Symposium, Philadelphia, PA, Host: Yi Xing.

2019 "When Precision Medicine Get Personal", Fusion Personalized Medicine Conference, Puerto Vallerta, Mexico, Host: David Roth

2019 "Machine Learning to Identify Phenotypes from Electronic Health Records", Fusion Personalized Medicine Conference, Puerto Vallerta, Mexico, Host: David Roth

2019 "Embracing the Complexity of Complex Traits", Workshop on Statistical Challenges in Medical Data Science, Ascona, Switzerland, Host: Niko Beerenwinkel

2019 "Phenotype opportunities from electronic health records", International Society of Pharmacoepidemiology (ISPE), Philadelphia, PA, Host: Leah Sansbury

2019 "EHR phenomics for precision medicine", University of Pennsylvania Department of Genetics retreat, Bear Creek, PA, Host: Dan Rader

2019 "Leveraging Translational Research for the Advancement of Drug Discovery and Understanding of Complex Disease", DNAnexus Connect, Boston, MA, Host: Brady Davis

2019 "The power of the EHR for genome-phenome exploration", VA Million Veterans Program Research meeting, Philadelphia, PA, Host: Kyong-Mi Kim

2019 "The power of the EHR for genome-phenome exploration", First Penn Conference on Big Data in Population Health Sciences, Philadelphia, PA, Host: Jason Moore

2019 "Exploring genomics and phenomics with machine learning toward an improved understanding of complex traits", EMBO|EMBL Symposium "Systems Genetics: From genotypes to complex traits", Heidelberg, Germany, Host: Tuuli Lappalainen, PhD

2019 "Translational bioinformatics using the electronic health record: harnessing the power of clinical data", Institute for Translational Medicine and Therapeutics Symposium, Philadelphia, PA, Host: Garret Fitzgerald

2019 "Exploring the phenome and the relationship with the genome using EHR-linked biobanks", American Society of Human Genetics, Houston, TX, Host: ASHG Program Committee

2019 "Implementing genomics into clinical care – a vision for the future of medicine", Thomas Jefferson Departmental seminar, Clinical Pharmacology, Philadelphia, PA, Host: Scott Waldman

2019 "Pharmacogenomics for precision medicine", American Society of Heath System Pharmacists (ASHP), Las Vegas, NV, Host: ASHP program committee

2020 "Why use biobanks for precision medicine", Pacific Symposium on Biocomputing, Big Island, Hawaii, Host: Ju Han Kim

2020 "Making Precision Medicine Mainstream with Health Data: Challenges to Acceleration: Informatics and Analytics", Precision Medicine World Conference, San Jose, CA, Host: Tal Behar

2020 "The power of electronic health records for precision medicine research", Calico Life Sciences invited seminar, San Francisco, CA, Host: David Botstein

2020 "Machine learning for identifying phenotypes in electronic health records", International Molecular Medicine Tri-Conference, San Francisco, CA, Host: Emily Le.

2020 "Translational bioinformatics using the electronic health record: harnessing the power of clinical data", Northwestern University, Feinberg School of Medicine, Distinguished Lectures in Life Sciences, Chicago, IL, Host: Justin Starren

2020 "The power of medical biobanks for precision medicine", Columbia University Precision Medicine Initiative Conference, Virtual Conference, Host: Tom Maniatis

2020 "COMPACTS: COVID Observational and Multiomic Penn and Community Together Study", Cancer Center COVID-19 Retreat, University of Pennsylvania, Abramson Cancer Center, Virtual Conference, Host: Angela DiMichele

2020 "The Future is Now: how technology and AI have advanced genomics and medicine", UCSF AI4All Camp ,Closing Keynote, Virtual Program, Host: Mirina Sirota and Tomiko Oskotsky

2020 "Electronic Health Records – Fueling Precision Medicine", 9th Annual Big Data in Biomedicine Symposium, Georgetown University, Virtual Conference, Host: Subha Madhavan

2020 "Electronic Health Records – Fueling Precision Medicine", Margaret Prine Joy Lecture in Reproductive Sciences, Magee Women's Research Institute Retreat, Virtual Conference, Host: Judith Yanowitz

2020 "Medical Biobanks for Precision Medicine Research", Cancer Outcomes Seminar at the University of Alabama at Birmingham, Virtual seminar, Host: Smita Bhatia

2020 "Predictive Analytics: An Introduction to Machine Learning", Precision Medicine Leaders' Summit, Virtual Conference, Host: The Journal of Precision Medicine

2021 “Electronic Health Records – Fueling Precision Medicine Research and Learning Healthcare Systems”, The Adler Lecture at Scheie Eye Institute’s Grand Rounds, Department of Opthalmology, University of Pennsylvania, Virtual lecture, Host: Dr. Joan O’Brien.

2021 “Making use of our genome for life: strategic opportunities and challenges”, NHGRI Genomic Medicine XIII: Developing a Clinical Genomic Informatics Research Agenda, Virtual meeting, Host: Ken Wiley and Marc Williams

2021 “Polygenic Risk Scores: For some or for all?”, Importance and Challenges of Increasing Ethnic Diversity in Human Genomics Research Symposium, Virtual symposium by the Center for Global Genomics and Health Equity and the Department of Genetics at the University of Pennsylvania, Host: Sarah Tishkoff and Dan Rader

2021 “Deep respresenation learning of electronic health records to unlock patient stratification at scale”, DBMI Journal Club at Harvard University, Virtual journal club, Host: Isaac Kohane

2021 “Leveraging Electronic Health Records for Precision Health”, Department of Medicine Grand Rounds, University of Pennsylvania Perelman School of Medicine, Host: Michael Parmacek

2021 “Leveraging Electronic Health Records for Precision Health”, IEEE EMBS conference on Biomedical and Health Informatics (BHI2021) keynote speaker, Virtual conference, Host: May Wang

2021 “PharmCAT: Current Status and Strategies for the Future”, Pharmacogenomics Research Network, Research in Progress Series, co-presenter with Dr. Michelle Whirl-Carrillo, Host: Jun Yang

2021 “Use of EHR for Understanding disease”, BEYOND THE SYMPTOM: THE BIOLOGY OF FATIGUE A Joint NIH Blueprint/Sleep Research Society Workshop, Virtual Workshop, Host: Vicky H Whittemore

2021 “The power of medical biobanks for precision medicine”, Genetics and Pharmacogenomics (GpGx) seminar series at Merck & Co., Inc., Virtual seminar, Host: Myung K. Shin
2021 “The power of medical biobanks for precision medicine”, Precision Medicine Resource at the Irving Institute for Clinical and Translational Research, Columbia University, virtual seminar, Host: Dr. Krzysztof Kiryluk, Dr. Ronald Wapner, and Dr. Wendy Chung

2021 “AI for Precision Medicine”, Workshop on Trustworthy AI in Medicine, University of Pennsylvania, Perelman School of Medicine and School of Engineering, Host: Qi Long

2022 “Leveraging Electronic Health Records for Precision Health”, Grand Rounds, Division of Cardiology, University of Pennsylvania, Perelman School of Medicine, Host: Thomas Cappola

2022 “Surviving Season 8 of the Pandemic”, Wellness Seminar Series, Department of Radiology, University of Pennsylvania, Perelman School of Medicine, Host: Despina Kontos

2022 “Future Directions of the NHGRI Analysis, Visualization, and Informatics Lab-space (AnVIL)”, NHGRI Council Meeting, Virtual, Host: Eric Green

2022 “PharmCAT: Pharmacogenomics Clinical Annotation Tool”, CDC NGS Workgroup, Virtual

2022 “Next generation work life balance amid the COVID-19 pandemic”, American Society of Human Genetics (ASHG) webinar, Virtual, Host: Career Development Committee of ASHG – Lucia Hindorff

2022 “Precision Medicine Research in Electronic Health Records”, Clinical and Translational Research Certificate of Added Qualification (CTR-CAQ) graduate program seminar, Baylor College of Medicine, Virtual, Host: Sundararajah ‘Thev’ Thevananther and Carolyn Smith

2022 “The Power of the Electronic Health Record for Precision Medicine”, Penn Cardiovascular Institute (CVI) seminar series, University of Pennsylvania, Philadelphia, PA, Host: Rajan Jain

2022 “Taking a Big Data View of Aging”, NIAWorkshop on Genetic Variation and Aging, Seattle, WA, Host: Daniel Promislow and Pankaj Kapahi

2022 “From Genomics to Genomic Medicine: Embracing the complexity”, NHGRI Center for Precision Health Research seminar, virtual, Host: Josh Denny

2022 “Exploring genetics and electronic health record data to identify sex differences in disease”, Penn FOCUS Fall Leadership Mentoring Conference, Successful Strategies for Women in Academic Medicine, virtual, Host: Mira Mamtani

2022 “Medical Biobanks: A Powerful Resource for Precision Medicine”, Penn-Stanford CVI Symposium, Philadelphia, PA, Host: Dr. Daniel Kelly

2022 “Using the electronic health record to advance translational research”, P30 symposium on High Dimensional Analytics in Digestive and Liver Disease: From Single Cell to the Whole Human, Philadelphia, PA, Host: Dr. Gary Wu

2022 “Beyond GWAS: innovative ways to use genomics for complex disease research”, MPaCT T2D Symposium: Genome Sequencing and Precision Medicine for Type 2 Diabetes, Centre of Genomic Regulation, Barcelona, Spain, Host: Carme Sanahuja

2022 “Precision medicine research dimensions made accessible by electronic health records”, IEEE BIBM, Las Vegas, NV, Host: Qi Long

2023 “ Mind the Gaps: How to Fill the Holes in EHR Data”. American College of Medical Informatics (ACMI) Annual Symposium, San Juan, Puerto Rico, Host: Kevin Johnson

2023 “Statistical Approaches for Integrating Environmental Data and Omics Data in Cancer Epidemiology Studies”, panelist, virtual workshop sponsored by NIEHS, Host: Kimberly McCallister

2023 “From Genomics to Genomic Medicine: Embracing the complexity.” Seminar, Department of Biostatistics, Epidemiology, and Informatics, University of Pennsylvania, Perelman School of Medicine. Host: Jeff Morris.

2023 “Using Large Biobanks, Medical Records, and Genetic Data for Disease Understanding”. Keynote at the 14th Biennial International Podocyte Conference, Philadelphia, PA. Host: Katalin Suzstak

2023 “Pharmacogenomics: Improving Quality and Safety of Healthcare”. ASHP Summer Meeting, Baltimore, MD. Host: Cynthia Von Heeringen

2023 “ Integrated risk scores for precision medicine”, Pharmacogenomics Research Network (PGRN) Annual Meeting, Memphis, TN. Host: Marisa Medina