CURRICULUM VITAE (updated 1/13/2022)

PERSONAL INFORMATION

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EDUCATIONAL PREPARATION AND DEGREES RECEIVED

University of Wisconsin-Madison B.S. Chemistry 1977
University of Wisconsin-Madison M.D. 1981

AWARDS

ALWIN SCHULTZ AWARD Presented by the University of Wisconsin to the medical student who showed extraordinary concern and caring in dealing with terminally ill patients and their families. 1980

HOUGHTON AWARD Presented by the State Medical Society of Wisconsin to the medical student, "who through scholastic excellence, extracurricular achievement, and interest in medical organization, shows high promise of becoming a complete physician." 1981

Best paper by a postgraduate professional--Northwest Pediatric Society meeting Fall 1992.

CLARITY Challenge (Children's Leadership Award for the Reliable Interpretation and appropriate Transmission of Your genomic information)—Honorable Mention for the Simulconsult/Geisinger team. The Challenge goal is to identify best methods and practices for the analysis, interpretation and reporting of individuals' DNA sequence data, to provide the most meaningful results to clinicians, patients and families.

Bio-IT World Best Practice Award in Genomics Grand Prize Overcoming the clinical interpretation bottleneck using integrated genome-phenome analysis. 2013

POSTGRADUATE EDUCATION

University of Utah Department of Pediatrics. Categorical internship and residency in pediatrics. June 1981-July 1984

Fellowship Medical Genetics University of Wisconsin-Madison 1992-1995.

Diplomate Intermountain Healthcare Institute for Healthcare Delivery Research Advanced Training Program in Quality Improvement 2007.

CERTIFICATION

Diplomate National Boards Parts I, II, and III.
Certified American Board of Pediatrics September 1986 Not participating in maintenance of certification (not required for certification of that date)
Certified American Board of Medical Genetics-Clinical Genetics September 1996, Recertified 2006. In compliance with ABMG Expanded Maintenance of Certification Current cycle until December 31, 2019.
Certified American Board of Preventive Medicine—Clinical Informatics January 1, 2015 expires January 31, 2025.

LICENSURE

State of Pennsylvania MD444648
State of Utah 5760101-1205 (inactive)
State of Wisconsin 31830-020 (inactive)
State of California (inactive)
State of Michigan (inactive)

PROFESSIONAL AFFILIATIONS AND FELLOWSHIPS

Fellow-American Academy of Pediatrics
Fellow-American College of Medical Genetics
Elected Fellow of the American College of Medical Informatics 2016
American Medical Association
Member-Section on Genetics and Birth Defects American Academy of Pediatrics
American Society of Human Genetics
American Medical Informatics Association

PROFESSIONAL ACTIVITIES

August 1984-July 1986 Private pediatric practice 170 S. Howell St. Hillsdale, MI 49242. (National Health Service Corps private practice option)

August 1986-December 1990 General pediatrician Riverside Medical Clinic 3660 Arlington Ave. Riverside, CA 92506.

January 1991-December 2004 Pediatrician/Medical Geneticist Gundersen Lutheran Medical Center

1999-2004 Associate Medical Director Gundersen Lutheran Health Plan.
Primary Responsibilities:

- Technology Assessment and coverage determinations.
- Oversee Quality program including HEDIS, CAHPS, and QAPI (for Medicare + Choice product). Rates are, with very few exceptions, above state and national averages. Gundersen Lutheran Health Plan was ranked the #1 Medicare + Choice health plan in the country based on customer service. Quality scores on Medicare HEDIS exempted the plan from CMS audit in the quality area.
- Direct the Health Plan's disease management programs in diabetes and congestive heart failure. This is being done in conjunction with a disease management initiative of our major provider system.
- Responsible for credentialing the provider network and quality peer review.

January 2005-December 2011 Director, Intermountain Healthcare Clinical Genetics Institute

January 2012-July 1, 2020 Professor of research and Director Genomic Medicine Institute, Geisinger

July 1, 2020-present Professor and Director Emeritus Genomic Medicine Institute Geisinger

June 2017 Professor Geisinger Commonwealth School of Medicine

HOSPITAL AFFILIATIONS

Gundersen Lutheran Medical Center LaCrosse, WI Jan. 1991-2004 Full privileges.

Franciscan Skemp Healthcare LaCrosse, WI Nov. 1992-2004 Consulting privileges.

LDS Hospital Salt Lake City, UT 2005-2011 Consulting privileges

Primary Children's Hospital Salt Lake City, UT 2005-2011 Consulting privileges

Geisinger Medical Center 2012-present full genetics privileges

Geisinger Wyoming Valley 2012-present full genetics privileges

LOCAL AND STATE COMMITTEES

Chair Recruitment Committee Riverside Medical Clinic 7/87- 1990.

Director of Pediatric Educational Programs Riverside Community Hospital 7/89-12/90.

Chair Perinatal Advisory Council of Riverside-West. Moderator and organizer of annual Perinatal Conference. 1989-1990

Medical Director LaCrosse Regional Genetic Services Project Nov. 1992- 2004.

Chair Personnel Committee Gundersen Clinic. 1993-2004.

Member Credentials Committee Lutheran Hospital 1992-2004. Acting Chair 2000.

Member Integrative Medicine Committee Gundersen Lutheran Medical Center 1998-2004.

Chair Credentials Sub-Committee Gundersen Lutheran Health Plan 1996-2004.

Chair Quality Improvement Sub-Committee Gundersen Lutheran Health Plan 2000-2004.

Secretary-Treasurer Northwestern Pediatric Society 1994-1996.

Medical Director's Committee Wisconsin Association of Health Plans 1999-2004.

Member transitional residency review committee, Gundersen Lutheran Medical Center, 2004.

Chair LDS Hospital Credentials Committee 2006-2007.

Chair Intermountain Healthcare Urban Central Region Credentials Committee 2007-present.

Member Intermountain Healthcare Primary Care Clinical Programs Guidance Council

Member Intermountain Healthcare Research Operations Council

Member Intermountain Healthcare Research Guidance Council

Member Intermountain Healthcare Intellectual Property Committee

Intermountain representative to Resource for Genetic Epidemiology Committee (Oversight of Utah Population Database)

Member Utah Department of Health Genetic Advisory Committee 2005-2011

Member Utah Department of Health Newborn Screening Advisory Committee 2005-2011

NATIONAL COMMITTEES

Chair Health Systems Subcommittee Section on Genetics and Birth Defects American Academy of Pediatrics 2000-2008.

RUC advisor Section on Genetics and Birth Defects American Academy of Pediatrics 2003-2008.

Participant Strategic Planning workshop American College of Medical Genetics 1/2000.

Director American College of Medical Genetics and Genomics Board (elected 2006. Term began March, 2007) to March 2013.

American College of Medical Genetics and Genomics Vice President for Clinical Genetics 2009-2013.

American College of Medical Genetics and Genomics President-Elect 2019-present

Member Committee on the Economics of Genetic Services American College of Medical Genetics 3/2000-12/2007.

Chair Committee on the Economics of Genetic Services American College of Medical Genetics 11/2000-2006
Major Accomplishments:

- Publication of Reimbursement Manual for members of the ACMG.
- Sponsor educational workshops at annual ACMG meeting on economic issues
- Participate in CPT and ICD processes to improve coding and reimbursement for genetic services.
    - Obtained CPT code for Genetic Counselors
- Assisted in preparation of a grant proposal to HRSA addressing delivery of genetic services. The purpose of the grant is two-fold: Phase 1 involves collecting data from a variety of practice settings on genetic care delivery, effectiveness of care and cost/benefit. Phase 2 will convene working groups to analyze the data and develop recommendations to the Maternal Child Health Bureau (MCHB) regarding demonstration projects to improve access to, and enhance quality and cost effectiveness of genetic services.
- Develop official ACMG statement on Genetics and Managed Care. (http://www.acmg.net/resources/policy-019.pdf)
- Supported Genetic Counseling licensure efforts in several states
- Consultant to the National Society of Genetic Counselor's Billing and Reimbursement Workgroup

Ex-officio member Intellectual Property Committee American College of Medical Genetics 2000-2006.

Focus of this committee is to address the impact of patents on the availability and cost of genetics tests.

Co-chair American College of Medical Genetics Workgroup to develop Professional Practice Guidelines for the use of Pharmacogenomic testing for Warfarin dosing. 2006

Organizer American College of Medical Genetics Special Interest Group on Quality Improvement in Clinical Genetics (Officially recognized as a SIG in March 2007). Chair 2007-2009.

Chair ACMG Ad Hoc Committee on the Value of a Genetic Diagnosis 2011-2013

Member American College of Medical Genetics and Genomics Foundation Development Committee 2013-present.

Chair Sub-Committee on Professional Recognition American College of Medical Genetics 3/2000-2002

Member Maternal and Child Health Task Force American Association of Health Plans 1999-2003.

Member Emerging Technology Committee American Association of Health Plans 2001-2004.

Member Access Workgroup Secretary's Advisory Committee on Genetic Testing 2001-2002

Member Access and Reimbursement Workgroup Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS) 2003-2010.

Member Secretary's Advisory Committee for Genetics, Health and Society (2007-2010).
Chapter lead and participant Workgroup preparing Oversight of Genetic Testing Report.
Lead Comparative Effectiveness Research and Clinical Utility Workgroup

Reviewer for CDC Collaboration, Education and Test Translation (CETT) program. 2006-2010.

Member Personalized Health Care workgroup of the American Health Information Community (2006-2008) Participating in Family History, Genetic/Genomic Testing and Clinical Decision Support workgroups.

Member National Center for Biomedical Informatics Medical Genetics Working Group (2010-present)

Member eMERGE External Scientific Panel (2011)

Member EGAPP Working Group (2012-2018)

Member Clinical Pharmacogenetic Implementation Consortium (2012-2015)
Member CPIC Informatics Working Group (2013-present)

Member American Medical Informatics Association Genomics and Translational Bioinformatics Working Group (2013-present)

Institute of Medicine Electronic Health Record Action Collaborative (2014-present)

Board of Directors representing Geisinger Health Care Systems Research Network (2013-2016)

NIH activities

Study Section—Member Biomedical Computing and Health Informatics Study Section 7/1/2015-6/30/2019. Co-Chair 2017-2019

Served as member of several NHGRI special study sections. Chair NHGRI Special Study section Mendelian Disease program 11/2020.

Member of National Human Genome Research Institute (NHGRI) Genomic Medicine Working Group Planning committee (2011-present)

Member NHGRI Intersociety Coordinating Committee (2013-2014)
Chair of Use Case Workgroup (2013-2015)

Chair NHGRI Clinician Education Workgroup (2012-2014)

Invited participant Precision Medicine Stakeholder meeting 2015

Chair External Review Committee Uniformed Services University's TAG-C/CHIRP program. 2018.

External Advisory Committee T32 Genomic Medicine Training Program University of Florida (2018-present).

Member Technical Evaluation Panel Office of the National Coordinator of Health IT Sync for Genes Phase 4 (2020-present)

EXTERNAL SCIENTIFIC ADVISORY BOARDS

Pharmacogenomics Clinical Annotation Tool Scientific Advisory Board (2021-present)

Washington University Informatics Institute 2019-present

University of Michigan Precision Health 2019-present

Online Mendelian Inheritance in Man (OMIM) External Scientific Advisory Board (2018-present)

Clinical Pharmacogenetics Implementation Consortium Scientific Advisory Board (2015-present)

NIH Undiagnosed Diseases Program External Scientific Panel (2015-present)

CHARGE Syndrome Foundation (1995)

National Marden-Walker Organization (1999).

ACADEMIC APPOINTMENTS/EDUCATIONAL RESPONSIBILITIES

Current

Professor of Genomics Department of Clinical Science, Geisinger Commonwealth School of Medicine 5/19/2017 to present

Adjunct Professor of Biomedical Informatics Ohio State University 4/1/2015 to present

Adjunct Professor of Biomedical Informatics University of Utah 2019-present

Past

Clinical Assistant Professor of Pediatrics University of Wisconsin-Madison 7/1/94-6/30/98.

Clinical Associate Professor of Pediatrics University of Wisconsin-Madison 7/1/98-2004.

Small group facilitator Genetics 721 (Med-1 course at UW-Madison)

Lecturer and attending in Pediatrics Med-3 UW-Madison Pediatrics rotation at Gundersen Lutheran Medical Center (Western Clinical Campus of the University of Wisconsin Medical School.

Clinical Associate Professor, Department of Clinical Science, University of Wisconsin-LaCrosse 12/19/01-2004.

Co-director and lecturer of advanced biology seminar class “Drugs and Disease”.

Lecturer on genetics for Pathophysiology course for Physician’s Assistant program

Full Professor LDS Hospital Clinical Academic Faculty. Joint appointment Departments of Internal Medicine and Pediatrics. (2005-2011)

Genetics lectures to Internal Medicine Residents at Intermountain Healthcare, University of Utah and Veteran’s Administration Hospital.

Clinical Professor of Pediatrics-Division of Medical Genetics University of Utah School of Medicine (2005-2011)

Lead seminars for 1st and 2nd year genetic counseling students at University of Utah
Clinical site for 2nd year genetic counseling students at University of Utah

Adjunct Associate Professor of Biomedical Informatics University of Utah School of Medicine (February 2008-2011)

RESEARCH FUNDING

ACTIVE

External
Federal Prime

1R01HG011799-01 (WILLIAMS, MARC) 08/10/2021 – 05/31/2026 2.4 cal. mos.
NIH (NHGRI) **Role PI** $975,703
REAL-TIME GENOMIC DIAGNOSIS AT THE POINT OF CARE

Complex diseases frustrate patients and create a burden on healthcare systems with multiple inpatient hospitalizations and frequent testing. Creating a pathway that would allow physicians to utilize genetic information for real time genetic diagnosis could prevent much of this burden. Utilizing a High Impact Phenotype Identification System (HIPIS) we would like to develop dynamic virtual genetic panels that should impact clinicians' decisions at point of care.

1U01HL146188-01A1 (WILLIAMS, MARC) 03/20/2020 – 02/28/2025 1.2 cal. mos.
NIH (NHLBI) **Role MPI** $538,094
COLLABORATIVE RESEARCH TO EXPLORE GENETIC VARIATION AND PHENOTYPIC SPECTRUM OF ELASTIN AND RELATED GENES

In the past, the medical community identified human diseases by combining patients with similar phenotypes and looking for shared genetic changes. This approach is limited by our clinical ability to see appropriate connections between phenotypic features in patients. This project uses a gene-first approach in a large research cohort with associated exome sequences to identify the full range of medically relevant human phenotypes (common and rare) associated with variants in genes coding for elastic fiber proteins followed by deep phenotyping in individual patients to learn more about the mechanism of disease and its clinical impact. This is a unique grant mechanism as the NHLBI intramural program is a subcontract to Geisinger. One of the goals of the project is to identify Geisinger patients who could be offered the opportunity to have an evaluation at the NIH clinical center for the purposes of better understand the clinical phenotype of these disorders. **Overlap** None

R01HG009671-01A1 (BUCHANAN, ADAM) 9/19/2018 – 06/30/2023 0.6 cal. mos.
NIH (NHGRI) **Role CoI** $514,799

REPORTING ADULT-ONSET GENOMIC RESULTS TO PEDIATRIC BIOBANK PARTICIPANTS AND PARENTS

This project will evaluate the psychosocial outcomes and the health behaviors from the return of genomic results including adult-onset conditions for children and their parents.
Overlap None.

R01CA211723 (RAHM, ALANNA) 8/1/2017 - 7/31/2022 0.6 cal. mos.
NIH (NCI) **Role CoI** $112,171
IMPLEMENTING UNIVERSAL LYNCH SYNDROME SCREENING ACROSS MULTIPLE HEALTHCARE SYSTEMS: IDENTIFYING STRATEGIES TO FACILITATE AND MAINTAIN PROGRAMS IN DIFFERENT ORGANIZATIONAL CONTEXTS

The goal of this proposal is to utilize the CFIR and other tools from implementation science to describe, analyze, and compare variations in LS screening program implementation across multiple healthcare systems with the overarching goal to create a comprehensive model and customizable approach to implementation for LS screening programs.
Overlap None.

1R01HL148246-01 (STURM, AMY) 7/1/2019-6/302024 0.6 cal. Mos.
NIH (NHLBI) **Role CoI** $513,080

IDENTIFICATION METHODS, PATIENT ACTIVATION, AND CASCADE TESTING FOR FH: IMPACT-FH

Identifying the >1 million individuals with familial hypercholesterolemia (FH) in the U.S. will lead to reduced morbidity and mortality due to atherosclerotic cardiovascular disease. To achieve population health impact of identifying individuals with this genetic condition, systematic cascade testing to identify all affected at-risk relatives must also commence. However, both under-identification of index patients, and low uptake of cascade testing, limits the potential population health impact. Therefore, this proposal will address these critical gaps in translational cardiovascular medicine by studying 1) innovative index patient identification methods via both phenotypic algorithms that utilize electronic health record (EHR) data, and genomic analysis of next-generation sequencing data; 2) the effectiveness of patient-centered approaches for family communication assistance to promote patient activation toward cascade testing uptake; and 3) feasibility, acceptability, and cost of these methods using an implementation science framework towards the goal of defining best practices for FH identification and cascade testing. **Overlap** None

Federal Subaward

5R01HG009694-02 (HAO, JING) 7/1/2017 - 6/30/2021

0.6 cal. mos.
Subcontract to Vanderbilt University (NIH-NHGRI) **Role CoI** $42,867
RATIONAL INTERGRATION OF CLINICAL SEQUENCING (RISE)

The aims are to understand the impact of drivers of economic value in genomic screening, assess the impact
or real-world use data on genomic screening policy implications, and identify key evidence gaps and research priorities in genomic screening. **Overlap** None. **In no cost extension.**

3U01HG011166-01S1 (PETERSON, JOSH) 6/1/2020 - 5/31/2021
1.2 cal. mos.
Subcontract to Vanderbilt **Role Site PI** $100,000

THE EMERGE RISK ASSESSMENT NETWORK - COORDINATING CENTER COVID-19

Supplement

This is a 1-year supplement to eMERGE Phase IV. The purpose of this supplement is to work with eMERGE sites to develop, test, and disseminate a set of phenotypes relevant to the study of health conditions associated with infection with COVID-19. **NB This award started much later than anticipated, so while technically complete, it will continue into early 2022**

HHSN275201800005C (BROWER, AMY, WILLIAMS, MARC) 9/26/2018 - 9/25/2023
1.8 cal. mos.
Subcontract to American College of Medical Genetics and GenomICs (NIH-NICHD) **CoI**

$104,891

THE NEWBORN SCREENING TRANSLATIONAL RESEARCH NETWORK (NBSTRN)

The NBSTRN is a project that creates a centralized repository of information generated by newborn screening programs across the United States. This is a key component of the Hunter Kelly Newborn Screening Program at the Eunice Kennedy Shriver National Institutes of Child Health and Human Development through a contract with the American College of Medical Genetics and Genomics.

PENDING

1 R01 HG012262-01 (Peterson; Hao; Veenstra) 12/01/2021 – 11/30/2026 0.6 ca. mos.
NIH/NHGRI $ 192,330
RATIONAL INTEGRATION OF POLYGENIC RISK SCORES (RIPS)
There has been an extraordinary growth in recent years of new techniques to predict common diseases using a new genetic intervention, polygenic risk scores (PRS). The Rational Integration of Polygenic Risk Scores (RIPS) project will use economic modeling to project the lifetime clinical impact and cost-effectiveness of screening a diverse population with PRS to predict the

onset of heart disease, breast cancer, and colorectal cancer. The models produced by this study will account for the cost of genetic testing, the cost and effectiveness of the preventative care needed to reduce risk, and the potential pitfalls associated with using these novel interventions in underrepresented and underserved populations. **Overlap** None **NB This was reviewed in July 2021 and scored a 24. Awaiting NHGRI Council review.**

R61HL161775 (Jones, Gidding) 12/01/2021 – 11/30/2026 1.2 ca. mos.
NIH/NHLBI **Role Co-I** $259,727
COLLABORATIVE APPROACH TO REACH EVERYONE WITH FAMILIAL HYPERCHOLESTEROLEMIA: CARE-FH
Diagnosis rates of familial hypercholesterolemia (FH) are low in the United States, despite multiple guidelines and recommendations for screening and treatment of high cholesterol, to prevent heart attacks in those affected. Using a stepped-wedge design, we plan to utilize tools from implementation science to improve uptake, acceptability, and sustainability of FH diagnostic programs in primary care settings. If successful, this study will provide tools generalizable to other health care systems to improve FH diagnosis rates. **Overlap** None

HHSN275201800005C (Amy Brower) 9/26/2018 - 9/25/2023 1.8 cal. mos.
Subcontract to American College of Medical Genetics and GenomICs (NIH-NICHD) **Role MPI**
$104,891
THE NEWBORN SCREENING TRANSLATIONAL RESEARCH NETWORK (NBSTRN)
The NBSTRN is a project that creates a centralized repository of information generated by newborn screening programs across the United States. This is a key component of the Hunter Kelly Newborn Screening Program at the Eunice Kennedy Shriver National Institutes of Child Health and Human Development through a contract with the American College of Medical Genetics and Genomics.

PENDING

R01 No assignment # (WILLIAMS, MARC) 4/1/2021 - 3/31/2026 3.0 cal. mos.
NIH (NHGRI) **Role PI** $599,430
REAL-TIME GENETIC DIAGNOSIS AT THE POINT OF CARE
The goal of the project is to test the proposition that we can identify symptoms or diseases where having an individual's genetic sequence information can have a significant impact on diagnosis or treatment. We can then build algorithms to capture these symptoms from an individual's electronic medical record as they are being seen by a physician and offer a genetic diagnosis while the patient is still in the office. This rapid genetic diagnosis has the potential to

significantly improve care through more targeted treatment and the avoidance of unnecessary visits and tests. As this is a new capability for the electronic medical record we must determine the best way to integrate such capability in a way that provides the best benefit for the patient and the physician. We will also study methods of delivering genetic diagnoses and information surrounding those diagnoses to physicians so they can provide optimal informed treatment to the patient. **Overlap** None.

R01HL157211 (JONES, LANEY) 04/01/2021 – 03/31/2026 0.6 cal. mos.
NIH (NHLBI) **Role CoI** $3,103,915

TESTING REAL-WORLD EVIDENCE-BASED APPROACHES TO TREAT FH: TREAT-FH

The goal of this project is to develop, tailor, and test implementation strategies to improve treatment of familial hypercholesterolemia.
Overlap: None (coordinated with work of IMPACT-FH)

Completed Research Support

DHHS Health Resources and Services Administration Congressionally-Mandated Health Information Technology Grants. PI for Intermountain Healthcare Award date 9/1/2010 10% effort. 18 month grant. (My role terminated on Dec. 22, 2011 upon leaving Intermountain Healthcare)

National Library of Medicine SimulConsult An Evidence-Based, Open Database Approach to Diagnostic Decision Support. Role Senior Expert

Washington State Department of Health Grant # N14531 Intermountain Healthcare Genetic Use Case. Total award $5000 Due date 5/31/2007 Completed 2008
Goal: Develop a case study for clinical genetic services in a private not for profit integrated health care delivery system
Role: Coordinate data acquisition and serve a key informant for site visit team.

University of Washington Subcontract No. 343192 (from CDC contract) entitled "Should Genetic Testing be used to Guide Warfarin Therapy? An Evidence Based Cost-Utility Analysis." Total Award $17,492. Ended 2007
Goal: Evaluate cost of implementing genetic testing to assist Warfarin dosing in clinical practice. Done in parallel with the CoumaGen prospective randomize controlled-trial, but funded independently.
Role: Institutional PI. Clinical consultant to economic modeling team.

Genetic Alliance Project ID No. 1002481 (HRSA funded) entitled Genetic Alliance Grant: "Utilizing Family Traditions and Oral History for Health Promotion." Total award $183,148.42. 3 year grant ending May 9, 2009.
Goal: Development and implementation of community based family history tools.

Role: Institutional PI. Lead qualitative research effort on how primary care physicians use family history information they collect. Clinical consultant for development of family history collection tools. Evaluation of provider response to information.

Microsoft HealthVault award to develop Family History collection tool in the electronic patient portal. $150,000 1 year award for 2009
Goal: Develop and implement an electronic family history collection tool in the Intermountain Healthcare MyHealth Patient Portal. Develop ability to upload information from this tool to a Microsoft HealthVault Personal Health Record.
Role: Clinical consultant for development of family history collection tools.

INTEROPERABLE DECISION SUPPORT TO IMPROVE DIAGNOSTIC WORKFLOW ACROSS MULTIPLE EHRS R44LM011585 Co-I 5% effort 4/1/2013 3/31/2015

Geisinger E-Genomic Medicine Program (GEM) NHGRI 1U01HG006382-01 Co-PI Geisinger Health System 20% effort. 7/1/2011 - 6/30/2015

Cardiovascular Research Network HMO Research Network - Anticoagulation Treatment and Long-Term Outcomes after Venous Thromboembolism 0.6 Calendar months Assumed site PI role January 2014. Site Principal Investigator. 1.2 Calendar months The goal of this project is to use the electronic health record to improve the identification and management of patients who have experience venous thromboembolism.

EMR-Linked Biobank for Translational Genomics U01HG8679 9/1/2015-3/31/2020 NCE 8/31/2020 Role: PI Goal: eMERGE is a national network organized and funded by the National Human Genome Research Institute (NHGRI) that combines DNA biorepositories with electronic medical record (EMR) systems for large scale, high-throughput genetic research in support of implementing genomic medicine.

Washington University K12 Program in T4 Implementation Research K12HL137842-02 07/01/2018 – 06/30/2020 Role Site Mentor I mentor a junior faculty member on a project focused on implementation of a program to improve the care of patients with Familial Hypercholesterolemia.

A Genotype- Phenotype Archiving and Communication System (G-PACS) 1R43HG010322 09/19/2018 – 08/31/2019 Role: MPI The project proposes to develop a prototype platform for a Genotype-Phenotype Archiving and Communication System, analogous to the radiology PACS, with archiving and communication of 2 types of information – patient findings and annotated genomic variants, enabling genome-phenome analysis using these components.

Clinically Relevant Genetic Variants Resource: A Unified Approach For Identifying Genetic Variants For Clinical Use NHGRI Site PI 08/01/2017 – 07/31/2018
Goal: The goal of this project is to create an annotated genomic variant repository that is available for use by laboratorians and clinicians. My role is to lead the team that will explore interfaces between the resource and the electronic health record.

ENHANCING GENOMIC LABORATORY REPORTS TO ENHANCE COMMUNICATION AND EMPOWER PATIENTS. PCORI PI 9/1/2013 12/31/2016 Goal: The goal of this project is to create a patient facing genomic test report and do a prospective comparative effectiveness trial using this report compared to standard of care.

GENOMIC MEDICINE IMPLEMENTATION: THE PERSONALIZED MEDICINE PROGRAM U01 HG007269 Subaward FDSP00010620 (Economic modeling project for pharmacogenomics prevention of Stevens-Johnson syndrome). 5/1/2014 to 4/30/2015 The goal of this project is to create a generic decision model that will allow rapid determination of cost-effectiveness through the input of a set of key variables.

U01GH008679 (WILLIAMS, MARC) 9/1/2015 - 3/31/2020
2.4 cal. mos.
NIH (NHGRI) Role PI $577,739
EMR-LINKED BIOBANK FOR TRANSLATIONAL GENOMICS (EMERGE III)
The goals of this study are to use existing biospecimens, genotype and sequence data and EMR generated phenotypes for discovery in the proposed disorders: familial hypercholesterolemia (FH) and chronic rhinosinusitis (CRS), develop and test approaches for implementation of genomic information in clinical practice and to explore, develop and implement novel approaches for family-centered communication around clinically relevant genomic results. The original grant period was to end 5/31/2019, but a one-year extension (through a supplement) was approved so the project will now end 3/31/2020.
Overlap None.
No cost extension completed 8/31/2020.

1U41HG009650-01 (WILLIAMS, MARC) 9/12/2017 - 7/31/20211.56 cal. mos.
Subcontract to University of North Carolina (NIH-NHGRI) $35,454
THE CLINICAL GENOME RESOURCE – EXPERT CURATION AND EHR INTEGRATION
ClinGen is a National Institutes of Health (NIH)-funded resource dedicated to building an authoritative central resource that defines the clinical relevance of genes and variants for use in precision medicine and research.
Overlap None.

RESEARCH COORDINATION

Supervisor Genetics Summer Fellowship Gundersen Lutheran Medical Foundation 1996-2004.

EDITORIAL POSITIONS

Editorial Board Wisconsin Medical Journal 1998-2004.

Ad Hoc Peer Reviewer for: New England Journal of Medicine, Journal of the American Medical Association (JAMA), Nature Medicine, Annals of Internal Medicine, Journal of the American Heart Association, American Journal of Medical Genetics, Genetics in Medicine, Mayo Clinic Proceedings, European Journal of Pediatrics, Clinical Dysmorphology, Clinical Genetics, Medical Decision Making, Personalized Medicine, Journal of Medical Genetics, Effective Health

Care, Journal of Pediatrics, American Journal of Medical Quality, Science Translational Medicine, Postgraduate Medical Journal, Journal of Personalized Medicine, Journal of Healthcare Engineering, Molecular Diagnosis and Therapy, Applied Clinical Informatics, Human Mutation, Learning Health Systems, BMC Medical Genetics, Health Affairs, Neuroscience, Molecular Genetics and Metabolism Reports, Children.

Guest Editor: American Journal of Medical Genetics Part C (3 issues of Seminars in Medical Genetics); Genetics in Medicine (1 special issue); BMC Medicine Big Data Article Collection (2019)

Standing reviewer for American Medical Informatics Association Informatics Summit, and Annual Meeting.

PUBLICATIONS

Available at NCBI:
http://www.ncbi.nlm.nih.gov/sites/myncbi/marc.williams.1/bibliography/46499458/public/?sort=date&direction=ascending

Kivlin JD, Fineman RM, **Williams MS**. Phenotypic Variation in the del(12p) Syndrome. Am J Med Genet 22:769-779, 1985.

Williams MS, Josephson KD, Wargowski DS. Marden-Walker Syndrome: a case report and a critical review of the literature. Clin Dysmorphol 2:211-219(1993).

Williams MS, Rooney BL, Williams JL, Josephson KD, Pauli RM. Investigation of Thermoregulatory Characteristics in Patients with Prader-Willi Syndrome. Am J Med Genet 49:302- 307(1994).

Williams MS, Josephson KD, Pauli RM. Patterson-Lowry Rhizomelic Dysplasia: a Possible Second Example. Clin Dysmorphol 4:216-221, (1995).

Richards ML, Gundersen AE, **Williams MS**. Cystic Neuroblastoma of Infancy. J Pediatr Surg 30:1354-1357, 1995.

Pauli RM, **Williams MS**, Josephson KD, Tint GS. Smith-Lemli-Opitz Syndrome: Thirty Year Follow-up of "S" of the "RSH" Syndrome. Am J Med Genet, 68:260-262, 1997.

Aswegan AL, Josephson KD, Mowbray R, Pauli RM Spritz RA, **Williams MS**. Autosomal Dominant Hypohidrotic Ectodermal Dysplasia in a large family. Am J Med Genet 72:462-467, 1997.

Williams MS, Josephson KD. Unusual autosomal recessive lymphatic anomalies in two unrelated Amish families. Am J Med Genet 73:286-289, 1997.

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