

CURRICULUM VITAE – ELAINE RENE MARDIS

Date: June 7, 2018

1. Personal Information:

- a. Sex: Female
- b. Date of birth: 9/28/62
- c. Place of birth: North Platte, NE

2. Citizenship: US

3. Address and Telephone Numbers:

- a. **Office:** Nationwide Children's Hospital
575 Children's Crossroad
WB2153
Columbus, OH 43205
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(tel): 614-722-6521

4. Present position: Co-Executive Director, The Institute for Genomic Medicine at Nationwide Children's Hospital
Professor of Pediatrics, The Ohio State University College of Medicine

5. Education:

a. Undergraduate:

- 1984 B.S. University of Oklahoma, Norman OK
Department of Zoology
Highest Honors, Phi Beta Kappa
Undergraduate Research Project in *Drosophila* Genetics
Dr. Gerald Braver, advisor

b. Graduate:

- 1989 Ph.D. University of Oklahoma, Norman OK
Department of Chemistry and Biochemistry
Dr. Bruce A. Roe, advisor

c. Postgraduate:

- 1989-1993 Bio-Rad Laboratories, Hercules CA
Senior Staff Scientist
Life Sciences Division/Genetics Systems Research & Development
- 2000 Cold Spring Harbor Laboratories, Making and Using DNA Microarrays Course (Joseph DeRisi and Vishy Iyer, Instructors)

6. Academic Positions/Employment:

- 2017 Nationwide Foundation Endowed Chair in Genomic Medicine,

Nationwide Children's Hospital, Columbus OH

- 2016-present Co-Executive Director, The Institute for Genomic Medicine at Nationwide Children's Hospital, Professor of Pediatrics at the Ohio State University College of Medicine, Columbus OH
- 2014-2016 Robert E. and Louise F. Dunn Distinguished Professor of Medicine, Washington University School of Medicine, St. Louis, MO
- 2009-present Adjunct Professor, Division of Pharmacotherapy and Experimental Therapeutics, Eshelman School of Pharmacy, The University of North Carolina at Chapel Hill
- 2011-2016 Professor, Departments of Genetics and Molecular Microbiology Washington University School of Medicine, St. Louis, MO
- 2006-2011 Associate Professor, Departments of Genetics and Molecular Microbiology Washington University School of Medicine, St. Louis, MO
- 2000-2005 Assistant Professor, Department of Genetics Washington University School of Medicine, St. Louis, MO
- 2002- 2016 Co-Director, The Genome Institute) Washington University School of Medicine, St. Louis, MO
- 2002-2016 Assistant Professor, Department of Molecular Microbiology Washington University School of Medicine (adjunct appointment)
- 2000-2002 Assistant Director, Genome Sequencing Center Washington University School of Medicine, St. Louis, MO
- 1997-2000 Research Assistant Professor, Department of Genetics Washington University School of Medicine, St. Louis, MO
- 1993-2010 Technology Development Core group leader Washington University Genome Sequencing Center, St. Louis, MO
- 1993-1996 Research Instructor, Department of Genetics Washington University School of Medicine, St. Louis, MO

7. University and Hospital Appointments and Committees:

- 2007 Washington University School of Medicine Medical Genomics Curriculum Working Group member
- 2007 Washington University School of Medicine Research Planning Cardiovascular and Metabolic Diseases Subcommittee member
- 2006-2008 Children's Discovery Institute Group member, Brain Tumor and

	Congenital Heart Defect groups
2001-2009	Advisory Committee member, WUMS Microarray Core facility
2015-2016	Co-leader, Siteman Cancer Center Breast Cancer Research Program
2016-present	Member, Solid Tumor Therapeutics Program, OSU Comprehensive Cancer Center

8. Medical Licensure and Board Certification:

9. Military Service:

10. Honors and Awards:

- 1984 Phi Beta Kappa
- 1988 Graduate Teaching Excellence Award; Graduate College, University of Oklahoma
- 2009 US News and World Report, "Medical Pioneers"
- 2010 Scripps Research Institute Translational Medicine Award (for work in Cancer Genomics)
- 2010 Distinguished Alumni Award, North Platte High School, North Platte NE
- 2011 Distinguished Alumna of the College of Arts and Sciences, Oklahoma University
- 2012 George Engelmann Interdisciplinary Award, St. Louis Academy of Science
- 2013 "Q&A" article, Discover Magazine, The Year in Science, 100 top stories of 2013
- 2014 Thompson-Reuters Top 10 Most Cited Researcher (#4)
- 2016 American Association for Clinical Chemistry Morton K. Schwartz Award for Significant Contributions in Cancer Research Diagnostics
- 2017 Precision Medicine World Congress 2017 Luminary Award

11. Editorial Responsibilities:

- 1995-2000 Editorial Board Member, *BioTechniques* (Eaton Publishing)
- 1999-present Reviewer, *Genome Research* (Cold Spring Harbor Press)
- 2005-present Reviewer, *Nature*, *Science*, *New England Journal of Medicine*, *Cell*, *Genome Research* and *Nature Methods*
- 2007-2014 Editorial Board Member, *Genome Research* (Cold Spring Harbor Press)
- 2010-present Editorial Board Member, *Genome Biology* and *Genome Medicine*
- 2012-present Senior Editor, *Molecular Cancer Research* (AACR Press)
- 2013-present Associate Editor for biotechnology, *Annals of Oncology* (ESMO)
- 2013-present Associate Editor, *Disease Models and Mechanisms* (Company of Biologists)
- 2015-present Editor-in-Chief, *Molecular Case Studies* (Cold Spring Harbor Press)

12. Professional Societies and Organizations:

- 2017 AACR Annual Meeting (2018) Program Committee Chair
- 2017 AACR-Pezcoller Foundation Award committee member
- 2015-2018 AACR Board of Directors member
- 2015-present AACR Project GENIE Advisory Board member
- 2013-present AACR Special Conferences committee member
- 2013 Organizing Committee member, AACR Precision Medicine Series, Drug Sensitivity and Response: Improving Cancer Therapy (Orlando FL)
- 2014 Organizing Committee member, AACR Special Conference on the

Hematologic Malignancies: Translating Discoveries to Novel Therapies (Philadelphia PA)

2015 Conference Co-chair, AACR Precision Medicine Series: Integrating Clinical Genomics and Cancer Therapy (Salt Lake City UT)

2013-2014 AACR Cancer Progress Report Steering Committee member

2015-2016, 2011-2013 Program Committee member, American Association for Cancer Research Annual meeting

2009-present Member, American Association for Cancer Research

2013-2015 Co-organizer, Cold Spring Harbor Biology of Genomes meeting

2009-2011 Co-Organizer, Cold Spring Harbor Personal Genomes meeting

2009 Co-Organizer, Human Genome Variation 2009 meeting

2004-present Member, American Society of Human Genetics

2006- 2008 Ad hoc member, NHGRI \$1,000/\$100,000 Genome RFA Study Section

2006-2007 Ad hoc member, NSF Plant Genome Study Section

2004-2006 Scientific Advisory Board Member, Columbia University CEGS

2003-2009 Bioengineering Sciences and Technologies IRG study section

1999-present Co-Organizer and Advisory Board member, “Advances in Genome Biology and Technology” meeting

1999 Co-Organizer, “Automation in DNA Mapping & Sequencing”

1997-2002 Member, NIH SBIR and SIG study sections

1998-2002 Women’s Leadership Training Institute (WLTi), St. Louis, MO
Mentor

1998-1999 DOE Joint Genome Institute Advisory Committee

13. Major Invited Professorships and Lectureships:

2017 Invited Speaker, Keystone Meeting on Hematopoiesis, Banff, Alberta, Canada

Invited Speaker, Nobel Forum “Frontiers in Cancer Research and Therapy”, Stockholm, Sweden

Keynote Speaker, “Biology and Medicine of the Future” Symposium, Genoscope, Paris France

Invited Speaker, 110th Annual AACR Meeting, Joint AACR-ASCO Major Symposium on Tumor Heterogeneity, Washington DC

Invited Speaker and Session Chair, Biology of Genomes, Cold Spring Harbor Laboratories, Cold Spring Harbor, NY

Invited Speaker, 50th Annual European Society of Human Genetics meeting, Copenhagen Denmark

Invited panel member, Aspen Institute Ideas Festival, Aspen CO

Keynote speaker, Gordon Research Conference on Hormone-Driven Cancers, Newry ME

Keynote speaker, 2nd Annual European Congress on Immunotherapies in Cancer, Barcelona Spain

Keynote speaker, Next-Generation Clinical Trials meeting, Cold Spring Harbor Laboratories, NY

Keynote speaker, SINAIInnovations, Mt. Sinai School of Medicine, NYC, NY

University Lecture, University of Texas Southwestern Medical School, Dallas TX

Session Chair, AACR-EORTC-NCI Molecular Targets Meeting, Philadelphia PA

Plenary Speaker, B Debate Meeting on Women’s Cancers, Barcelona Spain

Session moderator, San Antonio Breast Cancer Symposium, San Antonio TX

- 2016 Invited Speaker, The Ohio State University Comprehensive Cancer Center, Grand Rounds series
Invited Speaker, Oregon Health and Sciences University, Basic and Translational Sciences Seminar Series
Organizer and Invited Speaker, Keystone Symposium on Cancer Genomics, Banff Alberta Canada
Keynote Speaker, ABRF Annual Meeting, Ft. Lauderdale Florida
Invited Speaker, Future of Genome Medicine IX Conference, La Jolla CA
Invited Speaker, B-Debate, Cancer Therapy Resistance, Barcelona Spain
Plenary Speaker (Opening Session), 109th Annual American Association for Cancer Research Meeting, New Orleans LA
Invited Speaker, Hinterzartener Circle on Cancer Research meeting, Cadenabbia Italy
Invited Speaker, NIH Director's Lecture, NIH Campus, Rockville MD
Invited Speaker, New Directions in Leukemia Research, Noosa, Queensland Australia
Abstract-selected speaker, Biology of Genomes meeting, Cold Spring Harbor Laboratories, New York
Invited Speaker, Oslo Personalized Medicine meeting, Oslo Norway
Invited Speaker, Current Topics in Genome Analysis lecture series, NHGRI, Bethesda MD
Keynote Speaker, 81st Cold Spring Harbor Symposium on Quantitative Biology, Cold Spring Harbor Laboratory, NY
Guest Speaker, 2016 University of Chicago Biological Sciences Divisional Academic Ceremony, Chicago IL
Keynote Speaker, Oncology Research and Education Day, University of Western Ontario, London Ontario Canada
Invited Speaker, WIN 2016 Symposium, Paris France
Invited Speaker, 57th Annual McKusick Course on Human Genetics, Bar Harbor ME
Invited Speaker, 25th Annual Short Course on Mouse Models in Cancer, Bar Harbor, ME
Keynote Speaker, Genome Informatics Meeting, Wellcome Trust Sanger Institute, Hinxton UK
Keynote Speaker, Regenstrief Institute Annual Retreat, Turkey Run State Park, IN
Invited Speaker, 9th Annual Symposium of the Beth Israel Deaconess Medical Center, Boston MA
Invited Speaker, Jensen Symposium on Breast Cancer, University of Cincinnati, Cincinnati OH
Invited Speaker, Cornell University Graduate Program Symposium, Ithaca, NY
Invited Speaker, International Colloquium "PERSPECTIVES IN GENOMICS (Bringing together two generations: Scientific Leaders and Young Scientists)", Cancun, Mexico
Keynote Speaker, Regenstrief Institute Retreat, Indiana University, Marshall IN
Invited Speaker, NGS in Molecular Pathology Symposium, Netherlands Cancer Institute, Amsterdam, Netherlands
Invited Speaker, Cell Press Symposium, "Hallmarks of Cancer", Ghent, Belgium
Invited Speaker, Institute Lecture, IFCC, Torino, Italy

- 2015 Invited Speaker, Canada Gairdner Symposium: Genomics & Cancer, Vancouver BC
Invited Speaker, Society for Immunotherapy in Cancer (SITC) Annual Meeting, Washington DC
Invited Speaker, AACR Special Conference: Basic Science of Sarcomas, Salt Lake City UT
Invited Speaker, AACR Special Conference on Advances in Breast Cancer Research, Bellevue, WA
Invited Speaker, American Society for Human Genetics Annual Meeting, Baltimore MD
Keynote Speaker, EACR-AACR-SIC Meeting, Florence Italy
Keynote Speaker, Gordon Research Conference, “Cancer Genetics and Epigenetics”, Lucca Italy
Keynote Speaker, Gordon Research Conference, “Cell Growth and Proliferation, Mt. Snow VT
Invited Speaker, Cold Spring Harbor Laboratories, “Biology of Cancer: Microenvironment, Metastasis, and Therapeutics”, Cold Spring Harbor NY
Invited Speaker, Oncology Nursing Society Annual Meeting, Orlando FL
Invited Speaker, UNC Lineberger Cancer Center Symposium, Chapel Hill NC
Invited Speaker, Herbert Irving Comprehensive Cancer Center Annual Symposium in Immunology, Columbia University NYC NY
Invited Speaker, Nature Genetics-Mount Sinai School of Medicine Lecture Series, “Genomics, Cancer and Big Data”, NYC NY
Invited Speaker, British Columbia Cancer Agency, Vancouver BC
- 2014 Keynote Speaker, 21st Annual Plant and Animal Genome Conference, San Diego CA
Keynote Speaker, Princess Margaret Cancer Centre Conference, Toronto Ontario
Keynote Speaker, Royal College of Pathology Australasia Annual Meeting, Melbourne Australia
Keynote Speaker, 33rd Annual Lorne Genome Meeting, Lorne Australia
Invited Lecture, Lowy Cancer Research Centre and Centenary Institute at the University of Sydney, Sydney Australia
Invited Speaker, Scripps Translational Medicine conference, La Jolla CA
Invited Speaker, Society for Surgical Oncology annual meeting, Phoenix AZ
Invited Speaker, Educational and Major Symposium Session, AACR Annual meeting, San Diego CA
Invited Speaker, Fondation IPSEN Cancer Genomics meeting, Chantilly France
Keynote Speaker: Vanderbilt University Cancer Center Retreat, Nashville TN
Invited Lecture and Session Discussant, American Society for Clinical Oncology annual meeting, Chicago IL
Invited Lecture, Berlin Summer Meeting
Organizer and Invited Lecture, AACR Precision Medicine Series: Mechanisms of Drug Resistance
Keynote Speaker, European Assoc. for Cancer Research Bi-annual meeting, Munich Germany
American College of Surgeons Lectureship, Alliance Cooperative Group meeting, Chicago IL
Organizing Committee and Invited Lecture, AACR Hematologic Malignancies: Translating Discoveries to Novel Therapies meeting, Philadelphia PA

- Invited Lecture, Japanese Cancer Association/American Association for Cancer Research Plenary session, Yokohama JAPAN
Plenary Speaker, Samsung Genomics Institute, Samsung Medical Center, Seoul, S. KOREA
Plenary Speaker, National Cancer Research Institute Annual Meeting, Liverpool, UK
Inaugural Lecture, The American College of Surgeons Lecture, Alliance Cooperative Group Annual Meeting, Chicago IL
Plenary Speaker, EORTC-NCI-AACR Molecular Targets and Cancer Therapeutics Meeting, Barcelona SPAIN
Invited Speaker, Meet the Oncology Expert, Institut Jules Bordet, Brussels BELGIUM
- 2013 Invited Lecture, Moores Cancer Center, UCSD, California
Invited Lecture, Future of Genomic Medicine, La Jolla CA
Keynote Speaker, Cancer Forum of the Canceropole CLARA, Lyon France
Invited Lectures (3), Annual Meeting of the American Association for Cancer Research, Washington D.C.
Grand Rounds speaker, Cancer Medicine Division, MD Anderson Cancer Center
Invited Speaker, Cancer Evolution Symposium, Brown University, Providence RI
Invited Speaker, Massachusetts General Hospital Cancer Center, Boston MA
Cartwright Lecture, University of Utah School of Medicine, Salt Lake City UT
Annual American College of Surgeons Lecture for Alliance Group Meetings, inaugural lectureship, Chicago IL
Keynote Lecture, International Conference on Systems Biology, Copenhagen Denmark
Invited Speaker, EACR Summer Conference on Cancer Genomics, Cambridge UK
Keynote speaker, International Conference on Systems Biology, Copenhagen Denmark
Invited speaker, Ninth International Conference on Pharmacodynamics of Anti-Cancer Agents, Northumberland UK
Keynote Speaker, Mayo Clinic Individualizing Medicine Conference, Rochester MN
Invited Speaker, AACR Special Conference on Advances in Breast Cancer Research, San Diego CA
Invited Speaker, AACR-NCI EORTC Conference on Molecular Targets and Cancer Therapeutics, Boston MA
Invited Speaker, Nature-CNIO Cancer Symposium, Madrid Spain
Invited Speaker, 2nd EMBL Conference on Cancer Genomics
Invited Lecture, Vanderbilt-Ingram Cancer Center Seminar Series, Nashville TN
- 2012 Keynote Speaker, Pacific Symposium on Biocomputing, Hawaii
Invited Lecture, Penn Genomic Frontiers Institute, University of Pennsylvania
Invited Speaker, AACR Special Conference on Advances in Prostate Cancer Research, Orlando FL
Invited Speaker, Pediatric Cancer Translational Genomics Conference, Phoenix AZ
Invited Speaker and Session Chairperson, 5th Annual Scripps Translational Medicine meeting, San Diego CA
Invited Speaker, Nobel Forum on “Frontiers in Cancer Research and Therapy”, Stockholm Sweden
Invited Speaker, NCI Symposium on Translational Genomics, Washington D.C.
Invited Lecture, Seminars in Oncology Lecture Series at the Dana Farber Cancer Institute, Boston MA

Plenary Speaker (Opening Session), AACR 103rd Annual Meeting, Chicago IL
 Invited Lecture, Weill Cornell Medical College, NYC NY
 Invited Speaker, NCI Cancer Center Directors Retreat, NIH Campus Washington D.C.
 Keynote Speaker, Medical University of South Carolina 2012 Spring Pathology
 Symposia, Kiawah Island S.C.
 Invited Speaker, Pezcoller Foundation Symposium, Trento Italy
 Invited Speaker and Session Chair, European Association for Cancer Research Annual
 Meeting, Barcelona Spain
 Plenary Speaker, American Association for Clinical Chemistry Annual Meeting, Los
 Angeles CA
 Invited Speaker, University of Virginia, Charlottesville, VA
 Invited Speaker, University of Southern California, Keck School of Medicine Cancer
 Center Grand Rounds (with Rick Wilson), Los Angeles CA
 Invited Speaker, Blaffer Lecture Series, MD Anderson Cancer Center, Houston TX
 Plenary Speaker, CRI Annual Symposium, Cancer Research UK, Cambridge, UK
 Plenary Speaker, Princess Takamatsu Cancer Research Foundation Symposium, Tokyo
 Japan
 Invited Speaker, Annual Meeting of the American Society of Hematology, Atlanta GA

2011 Plenary Speaker, Keystone Symposia A1 2011, Functional Consequences of Structural
 Variation in the Genome, Steamboat Springs, CA
 Plenary Speaker, ASCO GI Cancer Symposium, San Francisco, CA
 Invited Speaker, Hematologic Malignancies Symposium, Medical University of South
 Carolina (MUSC), Charleston, SC
 Invited Speaker, 4th Annual Scripps Translational Medicine meeting, San Diego CA
 Invited Speaker, 3 sessions, American Association for Cancer Research 102nd Annual
 Meeting, Orlando FL
 Keynote Speaker, International Animal Functional Genomics meeting, Dublin IR
 Invited Lecture, Institute for Pure and Applied Mathematics, “Next-generation
 sequencing technology and algorithms for primary data analysis.” Los Angeles CA
 Invited lecture, American Society for Clinical Oncology (ASCO) meeting, Education
 Session, Chicago IL
 Invited lecture, CALGB Correlative Science Symposium, Boston MA
 Invited lecture, Institute of Genetic Medicine Seminar Series at Johns Hopkins School of
 Medicine, Baltimore MD
 Plenary speaker, European Molecular Biology Organization (EMBO) annual meeting,
 Vienna Austria
 Invited lecture, AACR Frontiers in Basic Cancer Research meeting, San Francisco CA
 Invited lecture, Beyond the Genome meeting, Rockville MD
 Invited lecture, Center for Cancer Systems Biology, Stanford University, Palo Alto CA
 Invited lecture, AACR Translation of the Cancer Genome, Educational Session, San
 Francisco CA
 Invited lecture, American Society for Clinical Pathology (ASCP) annual meeting, Las
 Vegas NV
 Invited lecture, Annual Meeting of the German Association for Diagnostics, Potsdam
 Germany
 Invited lecture, San Antonio Breast Cancer Symposium mini-symposium “Genome Data
 for the Masses: Presentation of TCGA and ICGC Breast Tumor Data”. San Antonio TX

- 2010 Invited Lecture, Pioneers in Genomic Biology Lecture, University of Illinois at Champaign-Urbana
Invited Speaker, 9th Annual Department of Genome Sciences Symposium, University of Washington, Seattle WA
Invited Speaker, Major Symposium Session, American Association for Cancer Research 101st Annual Meeting, Washington DC
Invited speaker, Bioinformatics Workshop, American Society of Investigational Pathology, Anaheim CA
Keynote Speaker, Association of Biomolecular Resource Facilities Annual Meeting, Sacramento CA
Invited Speaker, American College of Surgeons Clinical Congress, Washington DC
Invited Speaker, Berlin Summer Meeting on Quantitative Genomics, Berlin Germany
Invited Speaker, Prostate Cancer Foundation, Washington DC
Keynote Speaker, European Conference on Computational Biology, Ghent, Belgium
Invited Speaker, Personalized Cancer Therapeutics, University of Chicago, Chicago, IL
Invited Speaker, Controversies: Adjuvant and Neoadjuvant Therapy of Breast Cancer Meeting, New York, NY
Invited Speaker, University of California, San Francisco's 5th Anniversary Symposium in Human Genetics, San Francisco, CA
Plenary Speaker, Annual Meeting, Association of Molecular Pathologists, San Jose, CA
Invited Speaker, San Antonio Breast Cancer Symposium 10th Educational Session, San Antonio, TX
- 2009 Invited speaker, American Association of Cancer Research (AACR) Annual Meeting, Major Scientific Session, "The Cancer Genome: Recent Advances"
Invited Lecture, Institute for Advanced Studies, Princeton University
Invited speaker, Human Genome Variation and Complex Structural Variation meeting, Tallinn, Estonia EU
Invited speaker, American Clinical and Medical Genetics (ACMG) annual meeting, Tampa FL
Invited speaker, INSERM meeting on Human Genome Variation, St. Raphael FR
- 2008 Invited Lecture, Royal Swedish Academy of Sciences, Stockholm
Invited Speaker, INSERM workshop on Next-Generation Sequencing, Paris FR
Invited Speaker, Association of Biomolecular Resource Facilities Annual Meeting, Tampa FL
- 2007 Invited Lecture, President's Council of Advisors on Science and Technology (PCAST), Personalized Medicine Panel, Washington D.C.
Plenary Speaker, Association of Biomedical Research Facilities (ABRF) Annual Meeting, Tampa FL
Invited Speaker/Session Chair, Cold Spring Harbor Laboratories' Biology of Genomes Meeting, Cold Spring Harbor NY
- 2006 Invited Speaker, NCCR-sponsored workshop "Genomic Resources for the Rhesus Macaque", Bethesda MD
Invited Speaker, Marmoset Research Group of the Americas Workshop, San Antonio TX

- Plenary Speaker, The 8th International Meeting on Human Genome Variation and Complex Genome Analysis, Hong Kong PRC
- 2005 Invited Speaker/Session Chair, The Biology of Genomes, Cold Spring Harbor, NY
- 2004 Invited Speaker, NCI/NHGRI workshop on “Exploring Cancer through Genomic Sequence Comparison”, Bethesda, MD
Invited Speaker, FASEB meeting, Washington D.C.
- 2003 Keynote Speaker, Laboratory Robotics Interest Group meeting, Indianapolis, IN
Invited Speaker, Association of Laboratory Automation meeting, Palm Springs, CA
Invited Speaker, Applied Biosystems’ iScience Japan meetings, Osaka and Tokyo
- 2002 Invited Speaker, GENSIPS meeting, Chapel Hill, NC
- 2000 Invited Speaker, G2K: Back to Science. Advances in DNA Sequencing and Technology, Marco Island, FL
- 1999 Invited Speaker, Automation in DNA Mapping and Sequencing, Hinxton, UK
Invited Speaker, Genome Sequencing and Biology, Cold Spring Harbor, NY
- 1998 Plenary Lecture, Automation in DNA Mapping & Sequencing, St. Louis, MO
- 1997 Plenary Lecture, Automation in DNA Mapping & Sequencing, Heidelberg, Germany
Invited Speaker, Human Genome Meeting 1997 (HUGO), Toronto, Ontario, Canada
- 1994 Plenary Lecture, Automation in DNA Mapping and Sequencing, Hinxton, UK

14. Consulting Relationships and Board Memberships:

2017	<u>PACT Pharma, Scientific Advisory Board, Hayward CA</u>
2016-present	<u>Interpreta LLC, Scientific/Business Advisory Board, La Jolla CA</u>
2015-present	<u>Caperna LLC, Scientific Advisory Board, Boston MA</u>
2014-present	<u>Lineberger Comprehensive Cancer Center, External Advisory Board, University of North Carolina at Chapel Hill</u>
2014-present	<u>Carl Woese Institute of Genomics, Scientific Advisory Board University of Illinois at Urbana-Champaign</u>
2014-present	<u>Regeneron Genomics Center, Scientific Advisory Board, Tarrytown NY</u>
2012-present	<u>United States Veteran’s Administration Million Veteran’s Program, Scientific Advisory Board</u>
2012-present	<u>MD Anderson Cancer Center, External Advisory Board, Houston TX</u>
2014-present	<u>Qiagen NV, Supervisory Board member, Science and Technology committee member</u>
2013-2017	<u>ZS Genetics, Scientific Advisory Board, member and chair</u>
2013-2016	<u>Qiagen/Ingenuity, Scientific Advisory Board</u>
2012-present	<u>DNANexus, Scientific Advisory Board</u>

2009-2013	<u>Pacific Biosciences, Scientific Advisory Board</u>
2007-2008	<u>Applera Corporation, Board of Directors</u> member, Audit and Finance Committee member
2005-present	<u>Leerink-Swann/MEDACorp, consultant</u>
1998-2007	<u>Orion Genomics LLC, St. Louis, MO, member of SAB</u>
2001-2003	<u>Sigma-Genosys, Inc., St. Louis, MO, member of SAB</u>
1998-2002	<u>MJ Bioworks, South San Francisco, CA</u>
1998-2000	<u>Paradigm Genetics, Inc., Research Triangle Park, NC</u>
1997-1998	<u>Millennium Pharmaceuticals, Inc., Cambridge, MA</u>
1996-1998	<u>Monsanto Co., St. Louis, MO</u>

15. Research Support:

COLLABORATIVE GRANTS:

1R01CA204115 (Fields/Flavell) 01/09/2017-12/31/2019
NIH \$3,405 (direct costs, Mardis)
Towards True Precision Oncology: Validation of a Comprehensively Humanized, Autologous Mouse Model

The broad objective of this proposal is to validate an *in vivo* model to evaluate human tumors in the context of a complete and intact human immune system in a completely personalized and autologous fashion.

SU2C-AACR-DT14-14 Supplement (Schreiber) 03/01/2017 – 02/28/2019
American Association for Cancer Research \$8,251 (direct costs, Mardis)
(Stand Up To Cancer)

Transforming Pancreatic Cancer From Death Sentence To Treatable Disease

The goal of this project is to perform a clinical trial of a personalized DNA vaccine plus immune modulation of the tumor microenvironment in patients with resected pancreas cancer.

Breast Cancer Research Foundation (Wolff, PI) 12/01/2015 – 11/30/2017
\$41,289 (direct costs, Mardis)

Aurora US Effort in Metastatic Breast Cancer

This project will design and implement a retrospective and subsequent prospective specimen collection, repository, processing, tracking and distribution system of paired breast cancer tumor specimens. Subsequently, approximately 100 retrospective tumor sets will be collected and undergo molecular profiling. Final efforts will establish bioinformatics and data storage systems.

1U01CA209936 (Griffith, PI) 10/01/2016 - 06/30/2019
NIH/NCI \$1,691 (direct costs, Mardis)

Development of Informatics Resources for Interpretation of Clinically Actionable Variants in Cancer

Clinical tumor sequencing platforms will increasingly identify large numbers of genomic alterations that are relevant to understanding cancer progression and improving clinical decision making for individual patients. The most critical bottleneck in the precision medicine workflow is at the interpretation step, stemming from a lack of resources to help with the prioritization and interpretation of these alterations in a clinical context. To

address this, we propose to develop a curated knowledgebase and supporting software tools for clinical interpretation of variants in cancer (CIViC) that allows rapid intersection of genomic alterations with evidence for their clinical actionability.

COMPLETED GRANTS:

P50DK64540 (Hultgren-PI, Mardis-CoPI) 09/30/2002-08/31/2007
NIH-NIDDK \$103,853 direct costs (Mardis)

ORWH: SCOR on Sex and Gender Factors Affecting Women's Health

This project seeks to elucidate the molecular and epidemiologic basis of acute and recurrent urinary tract infections (UTIs). My role in this grant was first to oversee the genome sequencing of the UTI89 strain of *E. coli* and secondly to oversee and design the microarray resources designed to enhance our understanding of gene expression as it relates to colonization of the bladder epithelium.

R01AI40882 (Goldman-PI, Mardis-CoPI) 08/01/2002-05/31/2008
NIH-NIAID \$133,194 direct costs (Mardis)

Comparative/Functional Genomics of *Histoplasma* and *Blastomyces*

The goal of this project is to produce a draft sequence for the fungal pathogen, *Blastomyces dermatitidis*, and compare its genome to those of the two *Histoplasma* strains funded by a previous NIAID grant. We then will add putative coding sequences as oligomer elements to the existing *Histoplasma* microarrays, use these microarrays to identify similarly regulated genes of functional significance in the pathogenic yeast phase, and then explore their function using knock-out studies in the respective fungal species.

MCLI200601 (Stewart-PI, Mardis-CoPI) 07/01/2007-06/31/2009
CDI

Enabling Applied Genomic Technologies in Pediatric Diseases

The goal of this project is to support the development of the needed infrastructure to produce copies of the library plates, DNA stocks, and viral production for shRNA libraries of mouse and human. Once produced, this reagent will allow individual investigators to ask innovative biological questions and rapidly identify putative therapeutic targets for a wide range of human diseases.

U01HL065962 (Roden) 06/04/2010-08/31/2010
NIH-NHLBI \$24,000 direct costs (Mardis)

Pharmacogenomics of Arrhythmia Therapy

This U01 supports the Vanderbilt Site of the Pharmacogenomics Research Network and Knowledge Base. Studies are proposed to 1) identify polymorphisms in ion channel and other genes controlling cardiac rhythm; to determine polymorphism frequency in ethnic groups; to screen for baseline gating changes conferred by non-synonymous coding region polymorphisms; 2) to determine the effects of defined polymorphisms on patients with atrial fibrillation and ventricular fibrillation; 3) to determine prospectively the impact of CYP2C9 polymorphisms on warfarin dose requirement and of ion channel polymorphisms on QT prolongation during antiarrhythmic drug therapy; 4) to test the hypothesis that the extent of drug-induced QT change is familial, by drug challenge in healthy sib-pairs.

U01CA114722 (Ellis-PI, Mardis-CoPI) 06/01/2006-05/31/2011

NIH-NCI \$2,500,000 direct costs

Biological Breast Cancer Classification by qRT-PCR

This is a translational research proposal that seeks to identify gene expression patterns for a subset of human genes using FFPE breast cancer tissues of various grades in order to classify their tissue of origin and predict best choices for treatment based on that classification.

RC2HL102927 (Graubert-PI, Mardis-CoPI) 09/29/2009-08/31/2011
NIH-NHLBI \$136,226 direct costs (Mardis)

Whole Genome Sequencing of Myelodysplastic Syndromes

The major goal of this project is to identify mutations important for initiation and progression of myelodysplastic syndromes. Whole genome sequencing will be performed using paired tumor/normal DNA samples from at least 10 patients with myelodysplastic syndrome.

RC1CA145073 (Link-PI, Mardis-CoPI) 09/01/2009-08/31/2011
NIH-NCI \$77,345 direct costs (Mardis)

Mutational Profiling of miRNAs in AML

This project aims to understand the genetic mutations that contribute to acute myelogenous leukemia (AML) by characterizing miRNA expression using massively parallel sequencing in the leukemic blasts of at least 40 patients with *de novo* AML, and by identifying genetic variants of miRNA genes and defining the frequencies of somatic mutations in 400 cases of *de novo* AML.

CDILI201094 (Stewart-PI, Mardis-CoPI) 07/01/2010-06/30/2013
CDI \$135,392 direct costs current year (Mardis)

Enabling Applied Genomic Technologies in Pediatric Diseases

This project will develop a vector-based RNAi mouse genome library and the enabling technologies to utilize the library in high-throughput cell-based assays.

U10CA0760 (Ota) 12/01/2008-11/30/2011
NIH-NCI

American College of Surgeons Oncology Group

Dr. Mardis serves as the chair of the Basic and Translational Science Committee for the American College of Surgeons Oncology Group, an NCI-sponsored surgical oncology effort that coordinates clinical oncology studies and trials with tumor banking across the United States. Dr. Mardis is responsible for forming the committee and coordinating its activities.

RC4NS072916 (Gutmann-PI, Mardis-CoPI) 09/01/2010-08/31/2013
NIH-NINDS \$416,487 direct costs current year (Mardis)

Integrative Approaches to Stroma-Directed Glioma Therapy

Brain tumors (gliomas) are the leading cause of cancer-related death in children with few successful treatments available. This proposal applies genomics and novel high throughput technologies to translate basic science discoveries into new and better treatments for children with low-grade glioma.

U54HG004968 (Weinstock) 05/22/2009-04/30/2014

NIH-NHGRI \$3,404,045 direct costs current year
Sequencing the Human Microbiome

The study will add 1,000 genomes to the catalog of bacterial reference sequences of the human microbiome by using automated methods to isolate organisms and high throughput DNA sequencing to construct genome sequences. The study will also further characterize the metagenomes of the 250 individuals by directly comparing DNA sequences from organisms in the microbiome, thus determining what is common or variant between subjects.

U01GM097119 (Mardis, PI) 07/15/2010-06/30/2015
NIH-NIGMS \$3,000,000 direct costs

Next Generation Sequencing Add-On - Pharmacogenomics Research Network (PGRN)

The Genome Center will participate as one of the sequencing centers in the Pharmacogenetics Research Network (PGRN), providing sequencing and analysis in collaboration with other network participants.

Siteman Cancer Center 753855 (Schreiber) 01/01/2012-12/31/2013
Cancer immunotherapies targeting tumor-specific mutational antigens identified by exome sequencing

The goals of this project are to determine how we can best apply our new method(s) of unique tumor antigen identification and functional prioritization to cancer immunotherapy and determine whether we can (a) predict which tumor bearing host would most likely respond to checkpoint blockade therapy and (b) which tumor-specific antigens are best incorporated into individualized cancer vaccines.

Breast Cancer Research Foundation (Rohan) 10/01/2013-04/30/2015
Prediction of Breast Cancer Using Next Generation Sequencing of DNA and RNA and Analysis of Risk Factors from Breast Cancer

This grant provides funds to dissect the genomic differences in DCIS lesions and invasive breast cancer lesions isolated from the same patient.

NIH-NCI 5P01CA101937 (Ley) 09/19/2003-03/31/2018
Genomics of Acute Myelogenous Leukemia

The primary goal in this project is to utilize high throughput genomics technologies to define the commonly mutated target genes in AML that are relevant to clinical outcome.

Susan G. Komen KG111025 (Gillanders) 10/05/2011-10/04/2016
Personalized Breast Cancer Vaccines Based on Genome Sequencing

The vision of the Breast Cancer Research Program at WUSM is that breast cancer genome sequencing will be used in the future to tailor therapies to individual breast cancer patients, and The Genome Center is one of only a few centers worldwide with the sequencing capacity and analytical prowess to explore this vision. The flexibility of the DNA vaccine platform, and the experience of investigators at WUSM provides the opportunity to rapidly translate a personalized breast cancer vaccine strategy into a phase I clinical trial.

Susan G. Komen PG12220321 (Ellis) 10/09/2012-10/08/2015

Cell Death Activation to Prevent Late Relapse in Breast Cancer

The aims of this project are: 1) To improve genomic models for the prediction of late relapse by incorporating gene copy and gene mutation status of significantly mutated genes in ER+ breast cancer into the PAM50 ROR prediction algorithm. 2) To develop genome-driven therapeutic hypotheses that target patients at risk for late relapse by activating cell death pathways that are suppressed through somatic mutation. 3) To conduct proof-of-principle Phase 2 clinical trials designed to demonstrate synthetic lethal effects in the neoadjuvant setting.

The V Foundation for Cancer Research (Lockhart) 10/01/2010-09/30/2014

Whole Genome Sequencing to Identify Predictive Markers & Novel Therapeutic Targets in Gastric Cancer

To identify and interrogate novel therapeutic targets discovered through whole genome sequencing of a cohort of patients with gastric and gastroesophageal junction cancers.

Barnes Jewish Hospital Foundation 774266 (Fields) 05/01/2013-09/30/2015

Identifying Mechanisms of Metastasis to Improve Outcomes in Metastatic Colorectal Cancer

This grant provides funds to perform genomic studies comparing colorectal cancer genomes and transcriptomes across primary and metastatic disease.

NIH-NCI 1R01CA16629301A1 (Natarajan/Ellis) 03/05/2013-10/31/2015

Genomic, Clinical, and Behavioral Signatures of Long-Term Breast Cancer Survival

This proposal is to assay the PAM50 gene signature on archived Women's Healthy Eating and Living (WHEL) primary tumor, and to independently validate and compare the PAM50 clinico-genomic signature, and the Adjuvant, PREDICT, and Oxford Prognostic Index (OPI) clinico-pathologic tools using the rich WHEL study dataset, we will then integrate clinical and genomic tumor features into existing models, include potentially modifiable behavioral and psychosocial risk factors; and incorporate statistical improvements such as time-varying or nonlinear prognostic effects, and competing risk of death from non-cancer mortality.

NHMRC 1032443 (Pamphlett) 01/01/2012-09/30/2015

Using Trios to Find the Cause of Motor Neuron Disease

Genetic mutations underlie sporadic ALS. The disease is not seen in other family members either because (1) The mutation arises *de novo*, or (2) The penetrance of the mutation is affected by other genetic variants or environmental factors. We aim to determine, using case-parent trios, whether either *de novo* mutations, or germline mutations with variable penetrance, underlie SALS.

U54HG003079 (Wilson-PI, Mardis-CoPI) 11/01/2011-10/31/2015

NIH-NHGRI \$12,629,327 direct costs current year

A Platform for Large-Scale Genomic Discovery

This grant provides the majority of funding for our genome sequencing activities, including production, analysis and annotation of genomes as well as technology development to support these efforts. As Director of Technology Development, my group has provided the automation, technology and procedures for high-throughput DNA

sequencing that enables genome sequences to be produced at ever-decreasing costs at constantly increasing rates.

NIH-NHGRI

02/01/2012-12/31/2015

1U01HG006517 (Ding)

A Turn-Key System for High-Throughput Variant Discovery and Interpretation

The goal of this project is to make the analysis tools and next-generation pipelines currently in place in large genome centers available to the wider community, both individually and as part of a complete informatics solution.

16. Clinical Title and Responsibilities:

17. Teaching Title and Responsibilities:

Lecturer, Medical Genetics course (first year medical students). (2008-2011)
“Genomic Medicine”

I participated with Dr. Allison Whelan and other faculty members to revise the curriculum for first year medical students in 2007-2008. Since that revision, I have provided 1-2 lectures to address the scope and breadth of personalized medicine based on genomics.

Co-Coursemaster, Bio 4342 “Research Explorations in Genomics” (2003 to present)

This undergraduate course was conceptualized and developed in collaboration with Dr. Sarah Elgin (Biology), and for which Howard Hughes Medical Foundation funding was provided to support the first three years. We have since had our funding renewed by HHMI for another three-year period. The course is limited to ~14 junior/senior undergraduates who receive hands-on training in genomic sequencing from library construction to analysis and annotation of their own projects. As such, it represents a unique learning opportunity that combines laboratory work with intensive computational experiences.

Invited lecturer, Bio 181 “Biology for non-majors”. (2004- 2016)

Invited lecturer, BME 140, “Introduction to Biomedical Engineering”. (2004-2010)

Invited lecturer, Genetics and Genomics of Disease track, WUMS DBBS curriculum (2014-2016)

Coursemaster and Instructor, Next Generation Sequencing Technologies and Applications Course, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY (2006-present)

Instructor, Advanced Genome Sequencing & Analysis course, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY (1997-2004)

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b. Submitted Manuscripts:

1. Gastier-Foster JM, Mardis ER. Clinical Assays of Nucleic Acids: Past, Present and Future. *JAMA*, submitted as an invited JAMA Insights on Genomics and Precision Medicine
2. Miller CA, Dahiya S, Li T, Fulton RS, Smyth MD, Dunn GP, Rubin JB, Mardis ER. Resistance promoting effects of ependymoma treatment revealed through genomic analysis of multiple recurrences in a single patient. *PLoS Medicine*

c. Unpublished resources

1999-2001: Supported a web-based troubleshooting guide for the ABI 3700 DNA Sequencer. This resource was produced at a time when the 3700 was being widely implemented in genome centers to support the scale-up of human genome sequencing, and provided error messages with proposed solutions, protocols and other supporting information to groups attempting to utilize this relatively new and often unreliable platform.

2004-2010: Providing web-based resources in support of our whole genome microarrays for *C. elegans*. These resources include protocols for RNA labeling, hybridization, washing, and scanning as well as troubleshooting help. We also provide the files needed to grid arrays that have been scanned by the two main scanning instruments, an online ordering system that enables end-users to purchase the arrays, and QC results from our array print runs that establish quality for end-users. This microarray resource has now achieved worldwide distribution.

2013-present: Drug-gene interaction database (DGIdb). This online resource serves as a clearinghouse for gene-based queries of the pharmacopeia for purposes of identifying therapies that may be pertinent to the treatment of genomic variants.

2015: Clinical Interpretations of Variants in Cancer Database (CIViC). A freely available online resource and API that provides highly curated information about sequence variants in known cancer genes including literature that supports their use in diagnosis, prognosis and/or therapeutic indications. CIViC acts as a centralized forum for curation, interpretation and debate about variants and their functional/targetable/prognostic or diagnostic impact.

d. Patents Granted/Applied For

“Method of and Apparatus for Transferring Micro Quantities of Liquid Samples to Discrete Locations.” R.K. Wilson, E.R. Mardis, D.A. Panussis, Inventors. Filed by Washington University: March 15, 1996. Patent number 5,849,598. Issued December 4, 1998.

“Single-step magnetic bead-based preparation of nucleic acids.” M.T. Hickenbotham and E.R. Mardis, Inventors. Filed by Washington University: December 31, 2002.

“Gene expression profiles to predict breast cancer outcomes.” M.J. Ellis, C. Perou, P. Bernard, E.R. Mardis, and T. O. Nielsen, Inventors. Filed by Washington University: November 30, 2010 and assigned Application No. 12/995,450.

“DNMT3A mutations for the classification of acute myeloid leukemia (AML) prognosis.” T.J. Ley, R.K. Wilson, E.R. Mardis, L. Ding and J. F. DiPersio, Inventors. Filed by Washington University: October 20, 2010.

“Patient-specific mutation-directed immunotherapy for cancer.” G.P. Linette, B. Carreno, V. Magrini, E.R. Mardis, Inventors. Filed by Washington University: September 4, 2014.