Precision Medicine for All: Ensuring Diversity in Participants and in Practice
Case Western Reserve University
Thursday September 29, 2016

Biorepositories, data collection, and data analysis in US territories

Jacob L. McCauley, PhD
Associate Professor of Human Genetics and Pathology
Dr. John T. Macdonald Foundation Department of Human Genetics
John P. Hussman Institute for Human Genomics
University of Miami, Miller School of Medicine
"I want the country that eliminated polio and mapped the human genome to lead a new era of medicine ... Tonight, I’m launching a new Precision Medicine Initiative to bring us closer to curing diseases like cancer and diabetes -- and to give all of us access to the personalized information we need to keep ourselves and our families healthier."

President Obama on personalized medicine
State of the Union Address
Jan. 20, 2015

President’s 2016 Budget will provide a $215 million investment

• $130 million to NIH for development of a voluntary national research cohort of a million or more volunteers (through engaged participation and open, responsible data sharing).

• $70 million to the National Cancer Institute (NCI), part of NIH, to scale up efforts to identify genomic drivers in cancer and apply that knowledge in the development of more effective approaches to cancer treatment.

“The proposed initiative has two main components: a near-term focus on cancers and a longer-term aim to generate knowledge applicable to the whole range of health and disease.”

NIH Precision Medicine Initiative

THE PRECISION MEDICINE INITIATIVE

WHAT IS IT?

Precision medicine is an emerging approach for disease prevention and treatment that takes into account people's individual variations in genes, environment, and lifestyle.

The Precision Medicine Initiative will generate the scientific evidence needed to move the concept of precision medicine into clinical practice.

WHY NOW?

The time is right because of:

- Sequencing of the human genome
- Improved technologies for biomedical analysis
- New tools for using large datasets

LONGER TERM GOALS

Create a research cohort of > 1 million American volunteers who will share genetic data, biological samples, and diet/lifestyle information, all linked to their electronic health records if they choose.

- Pioneer a new model for doing science that emphasizes engaged participants, responsible data sharing, and privacy protection.

Research based upon the cohort data will:

- Advance pharmacogenomics, the right drug for the right patient at the right dose
- Identify new targets for treatment and prevention
- Test whether mobile devices can encourage healthy behaviors
- Lay scientific foundation for precision medicine for many diseases

A Complex but Robust Infrastructure is needed….
Guiding Principles to PM Efforts

• Creating a dynamic and inclusive governance structure
• Building trust and accountability through transparency
• Respecting participant preferences
• Empowering participants through access to information
• Ensuring responsible data sharing, access, and use
• Maintaining data security, quality and integrity

Bottom line: Samples and data will last for a long time, participants want assurances that these things are protected and utilized appropriately!
<table>
<thead>
<tr>
<th>Title</th>
<th>ID Number</th>
<th>Earliest Submission Date</th>
<th>Application Due Date</th>
</tr>
</thead>
<tbody>
<tr>
<td>Precision Medicine Initiative® Cohort Program Direct Volunteers Pilot Studies (OTA)*</td>
<td>OT-PM-16-001</td>
<td>November 16, 2015</td>
<td>December 22, 2015</td>
</tr>
<tr>
<td>Communication Support for the Precision Medicine Initiative® Research Programs at NIH (OTA)*</td>
<td>OT-PM-16-002</td>
<td>November 16, 2015</td>
<td>December 22, 2015</td>
</tr>
<tr>
<td>Precision Medicine Initiative® Cohort Program Biobank (U24)</td>
<td>RFA-PM-16-004</td>
<td>January 4, 2016</td>
<td>February 4, 2016</td>
</tr>
<tr>
<td>Precision Medicine Initiative® Cohort Program Coordinating Center (U2C)</td>
<td>RFA-PM-16-001</td>
<td>January 17, 2016</td>
<td>February 17, 2016</td>
</tr>
<tr>
<td>Precision Medicine Initiative® Cohort Program Healthcare Provider Organization Enrollment Centers (UG3/UH3)</td>
<td>RFA-PM-16-002</td>
<td>January 17, 2016</td>
<td>February 17, 2016</td>
</tr>
<tr>
<td>Precision Medicine Initiative® Cohort Program Participant Technologies Center (U24)</td>
<td>RFA-PM-16-003</td>
<td>January 17, 2016</td>
<td>February 17, 2016</td>
</tr>
</tbody>
</table>
Precision Medicine: Clearly a Hot Topic

- Several PM related Conferences/Forums
- Focus on Participant Engagement
  (must implement into actual practice)
- Precision
  (genetic information holds promise, but is it ready to deliver in the clinical setting?)
- Phenotype
  (deep and structured, will the existing EHR data work?)
Large existing US-based Cohorts

**eMERGE**

MyCode® Community Health Initiative

Geisinger’s MyCode project: the link to personalized medicine

**CHARGE**

*Its founding member cohorts include:*
- Age, Gene, Environment, Susceptibility Study -- Reykjavik
- Atherosclerosis Risk in Communities Study
- Cardiovascular Health Study
- Framingham Heart Study
- Rotterdam Study

*Additional core cohorts include:*
- Coronary Artery Risk Development in Young Adults
- Family Heart Study
- Health, Aging, and Body Composition Study
- Jackson Heart Study
- Multi-Ethnic Study of Atherosclerosis
About UK Biobank
UK Biobank is a major national health resource, and a registered charity in its own right, with the aim of improving the prevention, diagnosis and treatment of a wide range of serious and life-threatening illnesses – including cancer, heart diseases, stroke, diabetes, arthritis, osteoporosis, eye disorders, depression and forms of dementia. **UK Biobank recruited 500,000 people** aged between 40-69 years in 2006-2010 from across the country to take part in this project. They have undergone measures, provided blood, urine and saliva samples for future analysis, detailed information about themselves and agreed to have their health followed.
Technical capabilities already exist: 
HIHG Biorepository Overview

One of the largest academic Biorepositories in the United States.

- ~4,000 square foot facility in BRB
- Contains >157,000 unique individual participants
- Tracking 1,190,148 total aliquots
- Supports 75+ individual studies
- Supports numerous national and international disease consortia
Biorepository

Sample Storage Equipment

- 5 Double-door refrigerators (4°C)
- 2 Freezers (-20°C)
- 23 Ultra-low Freezers (-80°C)
- 2 MVE 1520 Liquid nitrogen dewars (-196°C)

<table>
<thead>
<tr>
<th>Aliquot Type</th>
<th>Item Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood for DNA</td>
<td>41630</td>
</tr>
<tr>
<td>Blood for RNA</td>
<td>19544</td>
</tr>
<tr>
<td>Frozen Brain</td>
<td>237</td>
</tr>
<tr>
<td>Fixed Brain</td>
<td>130</td>
</tr>
<tr>
<td>Tissue Blocks</td>
<td>3555</td>
</tr>
<tr>
<td>Tissue Slides</td>
<td>6581</td>
</tr>
<tr>
<td>Buffy Coat</td>
<td>3045</td>
</tr>
<tr>
<td>Cell Line</td>
<td>25662</td>
</tr>
<tr>
<td>DNA</td>
<td>193364</td>
</tr>
<tr>
<td>Filter Card</td>
<td>21672</td>
</tr>
<tr>
<td>Hair</td>
<td>1</td>
</tr>
<tr>
<td>Plasma</td>
<td>77901</td>
</tr>
<tr>
<td>RNA</td>
<td>174</td>
</tr>
<tr>
<td>Saliva</td>
<td>455</td>
</tr>
<tr>
<td>Serum</td>
<td>38884</td>
</tr>
<tr>
<td>Tissue</td>
<td>3325</td>
</tr>
<tr>
<td>Urine</td>
<td>3401</td>
</tr>
<tr>
<td>White Blood Cells</td>
<td>328</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>439889</strong></td>
</tr>
</tbody>
</table>
Biorepository: Security/Back-ups

A Secure and Comprehensive Facility

- Yokogawa DAQMaster Monitoring System
- Sensaphone (Secondary Monitoring)
- Back-up Air Conditioning
- 10 Ton spot Chiller (tertiary backup)
- Emergency Power
Biorepository: LIMS Tracking/Retrieval

- LIMS tracks all locations for samples, racks, shelves, and freezers
- All samples are ‘coded’ (no PHI data is used on labels or stored within the LIMS)
- Samples are retrieved and their locations updated to “checked-out” to a technician and a process
- Samples remain in “check-out” until technician acknowledges their return to storage
We have the genomic building blocks
So if we have the tools... why?

- Racial and ethnic minorities are underrepresented in genomic research.
- “Those most in need must not be the last to benefit from genetic research.”
- What affects participation?

(Bustamante 2011)
Factors that affect Participation

• TRUST

• Past experiences (either personal or historical examples)
  (e.g. trust between aboriginal communities in Australia and geneticists)

• Personal motivations
  • You or your family/friends are affected by a disease
  • Desire to better understand your own health
  • Etc.

“Geneticists attempt to heal rifts with Aboriginals”
Are the majority of complex diseases ready for PM?

What samples and data exist for diverse participants groups?
Examples of ongoing biobanking efforts and complex genetic disease studies of individuals with diverse ancestral and cultural backgrounds
ERICH Study (Stroke)

This study is supported by funds from the NINDS U01-069763 (Dr. Daniel Woo)

- Ethnic/Racial Variations of Intracerebral Hemorrhage (ERICH)
- Large prospective multi-center case-control study focused on identification of genetic and epidemiological risk factors for ICH
- 19 Clinical Recruitment Centers encompassing 42 sites
- Goal 6,000 participants
  - 3,000 cases of ICH among white, blacks, and hispanics
  - 3,000 demographically matched controls
- HIHG serves as central BioBank
- >6,000 samples (as of 9/1/16)
Inflammatory Bowel Disease (IBD)

IBD Pathogenesis

- Establish sample collection (blood, tissue, DNA)
- Explore genetics, environment, and microbiome
- Collect data electronically within the clinic setting

~1890 total study participants
696 Hispanic/ 1,188 non-Hispanic

86% of patients approached agreed to participate and provide a blood sample
Multiple Sclerosis Genetics

- MS is an unpredictable inflammatory demyelinating disease of the central nervous system
- Affected individuals have a spectrum of chronic symptoms and disabilities
- Genetic association studies in individuals of European descent have identified:
  - 110 autosomal MS risk variants
    - 103 discrete loci
    - Outside Major Histocompatibility Complex (MHC)
    - Explain 18% of the genetic risk
  - 27% of the genetic risk is explained after including the MHC
- International Multiple Sclerosis Genetics Consortium (IMSGC) expanded non-MHC variation to include:
  - 200 autosomal non-MHC risk variants
  - 1 X chromosome risk variant

Incidence (per 100,000 inhabitants)

More than 45

Less than 15

https://www.imsgc.org/
Replication analysis identifies TYK2 as a multiple sclerosis susceptibility factor

Maria Ban1, An Goris1, Ádálaug R Lorenzén4, Ámie Baker1, Tania Mihalova5, Gillian Inganna1, David R Booth1, Robert N Heard1, Graeme J Stewart5, Elke Bogaert1, Bénédicte Dubois4, Hanne F Harbo3, Elisabeth G Celius2, Anne Spurkland1, Richard Strange1, Olve Hawkins1, Neil P Robertson1, Frank Dudbridge1, James Watson1, Philip L de Jager4, 10, 11, David Hafler1, John D Rioux1, 2, Adrian J Irvin1, Jacob L McCauley4, Margaret Pericak-Vance11, Jorge R Olsenberg15, Stephen L Hauser15, David Sexton16, Jonathan Haines16, and Stephen Sawcer1, The Wellcome Trust Case–Control Consortium (WTCCC) and Alastair Compston1

The International Multiple Sclerosis Genetics Consortium

Genetic variation in the IL7RA/IL7 pathway increases multiple sclerosis susceptibility

Rebecca L. Zuvich1, Jacob L. McCauley1, Jorge R. Olsenberg1, Stephen J. Sawcer1, Philip L. De Jager1, International Multiple Sclerosis Genetics Consortium1, Cristin Aubin1, Anne H. Cross1, Laura Piccio1, Neelum T. Aggarwal1, Denis Evans1, David A. Hafler1, Alastair Compston1, Stephen L. Hauser1, Margaret A. Pericak-Vance1, and Jonathan L. Haines1

Hum Genet
DOI 10.1007/s00439-004-0789-4

ORIGINAL INVESTIGATION

Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis

International Multiple Sclerosis Genetics Consortium (IMSGC)

Interleukin 7 receptor α chain (IL7R) shows allelic and functional association with multiple sclerosis

Simon G Gregory1, 2, Silke Schmidt1, 2, Puneet Sethi1, Jorge R Olsenberg1, John Hart1, Angela Prokop3, Stacy J Gaillier1, Maria Ban1, An Goris1, Lisa F Barcelos1, Robin Lincock1, Jacob L McCauley1, Stephen J Sawcer1, DAS Compston1, Bénédicte Dubois4, Stephane L Hauser1, Mariano A Garcia-Blanco2, Margaret A Pericak-Vance1, and Jonathan L Haines1, for the Multiple Sclerosis Genetics Group

The role of the CD58 locus in multiple sclerosis

Philip L De Jager1, 4, 12, Clara Boedeker-Allart1, Udo M. Meurer1, Ariel T. Arber1, Linda Ottoboni3, Lisa Barcelos1, Jacob L McCauley1, Stephen Sawcer1, An Goris1, Joanna Szaflarska, Roman Yelensky5, Alison Price6, Virgin Lepage5, Nick Patterson5, Paul W de Bakker5, Dong Tran5, Cristin Aubin1, Susan Polychronakos7, Elizabeth Rossin8, Xinli Han9, Charles W. Ashley10, Edwin Cho10, John D. Rioux11, Margaret A. Pericak-Vance1, Adrian Johnson1, David R. Booth1, Gema M. Stewart1, Asano Poltorak12, Laura Peltomaa12, Bénédicte Dubois4, Jonathan L. Hauser1, Howard L. Weiner5, Alastair Compston1, Stephen L. Hauser1, and David M. Hafler1, 2, 5

Krieger-Miller Institute for Molecular Genetics, Center for Neurogenetics, Nemours Children's Hospital, Starship Children's Hospital, Children's Hospital of Philadelphia, World Class Hospital, and Children's National Health System, Philadelphia, PA 19142, USA; and Department of Neurology, School of Medicine, University of California, San Francisco, California, CA 94143, USA; and the Wellcome Trust Case-Control Consortium (WTCCC)

Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis

The International Multiple Sclerosis Genetics Consortium1 & the Wellcome Trust Case Control Consortium2

COMPREHENSIVE FOLLOW-UP OF THE FIRST GENOME-WIDE ASSOCIATION STUDY OF MULTIPLE SCLEROSIS

KIF21B and TMEM39A as additional susceptibility loci

The International Multiple Sclerosis Genetics Consortium
Individual disease results: post-ImmunoChip

**MS** (45 new loci/104 total)

**Celiac** (13 new loci/40 total)

**Psoriasis** (15 new loci/36 total)

**IBD** (71 new loci/163 total)

**RA** (14 new loci/48 total)

**Ankylosing Spondylitis** (13 new loci/31 total)
Precision Medicine for All:

This will require us to understand whether our findings in populations of primarily European ancestry extend to other populations affected by disease.
Why study racial and ethnically diverse populations in MS?

Are the risk loci identified in European populations relevant to disease risk in African-Americans or Hispanic Americans?

- Differences in LD structure can help to localize signal (i.e. fine-mapping)
- Understanding etiology of disease
- Ethics – what is the relevance of current scientific knowledge to other populations?
- Potential identification of new loci
- Teasing apart gene-gene and gene-environment interaction in admixed populations
- Better understanding the observed clinical differences across race/ethnic groups
MS: Hispanics and African Americans

Largest minority populations in the US (2010 Census Data)

~16% Hispanic/Latino and ~12% African American

Classically lower MS prevalence than individuals of primarily European descent, but complicated:

- Ethnic heterogeneity
- Lack of consistent diagnostic criteria
- Changing disease awareness

Disease heterogeneity seen:

- African Americans often exhibit greater disease severity (Buchanan R. *Ethn Dis.* 2010)
- Hispanics present more often with Optic Neuritis (Amezcua L. *Mult. Scler.* 2011)

Data from the Multiple Sclerosis International Federation (MSIF)

PREVALENCE BY COUNTRY (2013)

Our MS Ascertainment for Genetic Research in Florida

>1,200 participants recruited thus far
Alliance for Research in Hispanic MS (ARHMS): www.arhms.org

ARHMS Recruitment Centers

Jacob McCauley, PhD
UM Miller School of Medicine
Miami, FL 33136

Lilyana Amezcua, MD, MS
USC Keck School of Medicine
Los Angeles, CA 90089

Jorge Oksenberg, PhD
UCSF School of Medicine
San Francisco, CA 94158

Angel Chinea, MD
San Juan MS Center
Guaynabo, PR 00968

Figure: Adopted from the Rural Health Information Hub (https://www.ruralhealthinfo.org/rural-maps/mapfiles/hispanic.jpg)
Illustrates 2010 Census Summary Data on U.S. Hispanic/Latino population
## Initial Hispanic Dataset

<table>
<thead>
<tr>
<th>Location</th>
<th>MS Case</th>
<th>Control</th>
</tr>
</thead>
<tbody>
<tr>
<td>University of Miami</td>
<td>561 (48)</td>
<td>1102 (83)</td>
</tr>
<tr>
<td>University of California, San Francisco</td>
<td>227 (19)</td>
<td>63 (5)</td>
</tr>
<tr>
<td>University of Southern California</td>
<td>196 (17)</td>
<td></td>
</tr>
<tr>
<td>Caribbean Neurological Center</td>
<td>99 (8)</td>
<td>96 (7)</td>
</tr>
<tr>
<td>Brigham and Women’s Hospital</td>
<td>95 (8)</td>
<td>69 (5)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Genotype data available:</th>
</tr>
</thead>
<tbody>
<tr>
<td>– Illumina ExomeChip+ Beadchip</td>
</tr>
</tbody>
</table>
Global Ancestry: Hispanics

- Miami
- Hispanics
- USC
- UCSF
- Puerto Rico

- European ancestry
- African ancestry
- Native American ancestry
MS Ascertainment in Puerto Rico

January 2015

January 2016
Our Experiences in Puerto Rico

• Building trust: We spent 4 years in discussions and engagement before we collected the first sample
• MS patients and their families are very eager and motivated to participate
• We have a great clinician champion (Dr. Chinea)
• The logistics of sample collection at various sites across the island can be a challenge (especially finding FedEx)
• With each ascertainment trip we have become more efficient in our processes
• Clinical data collection is a little more challenging
• Next Trip November 2016
MS Collaborative Discussions in Cuba

April 2016
Some Challenges in Cuba

- Governmental restrictions/limitations on sample and data sharing
- More open to collaboration, but every university is flying in to ‘build’ collaborations
- Clinicians/Researchers want to grow their expertise and collaborate….. not simply provide samples and data for export to the US
- There are distinct unmet needs of patients, research may be a secondary pursuit in the eyes of many MS patients and their families
- Again, building trust will take time
Pilot Assessment of Motivations for Participation

To investigate the reasons for participation among Hispanic and non-Hispanic enrollees in a genetic risk for disease study.

Study involves a blood draw, family history questionnaire, and medical records review.
Participants enrolled via ascertainment for a study of genetic risk for multiple sclerosis.

N=101 individuals (95 agreed to participate)
80% Hispanic
20% M: 80% F

Education ranges (%)
- < high school: 5%
- High school: 24%
- Some college/AA: 35%
- College grad: 29%
- Post-graduate: 8%

Age ranges (%)
- 18-25: 7%
- 26-35: 17%
- 36-45: 21%
- 46-55: 27%
- 56-65: 20%
- >66: 8%
Methods

Surveyed reasons for participation
Provided 11 options

- Cure
- Knowledge
- Relative with disease
- Doctor recommendation
- Encouraged by others
- Treatment
- Have MS
- Future generations
- Reimbursement
- Other
- Not sure

“I want to help find a cure for MS.”
“I suffer from MS.”
“The doctor asked/recommended that I participate.”
“To help future generations.”
Results

% of endorsements per reason*  
(N=95)

- cure: 56%
- disease-self: 46%
- future: 37%
- treatment: 22%
- science: 21%
- doctor rec: 10%
- encouraged: 10%
- disease-relative: 1%
- other: 1%

*not sure and compensation were not endorsed
# Results

Comparison of endorsement rates per reason in Hispanics vs. non-Hispanics*

<table>
<thead>
<tr>
<th>Reason</th>
<th>Hispanic</th>
<th>non-Hispanic</th>
<th>**p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cure for MS</td>
<td>56%</td>
<td>56%</td>
<td>1.00</td>
</tr>
<tr>
<td>Have MS</td>
<td>52%</td>
<td>19%</td>
<td>0.03</td>
</tr>
<tr>
<td>Help future generations</td>
<td>34%</td>
<td>50%</td>
<td>0.26</td>
</tr>
<tr>
<td><strong>Find new treatments</strong></td>
<td>17%</td>
<td>50%</td>
<td><strong>0.007</strong></td>
</tr>
<tr>
<td>Improve science</td>
<td>19%</td>
<td>31%</td>
<td>0.32</td>
</tr>
<tr>
<td>Doctor recommendation</td>
<td>11%</td>
<td>0%</td>
<td>0.35</td>
</tr>
<tr>
<td>Encouraged by others</td>
<td>8%</td>
<td>19%</td>
<td>0.17</td>
</tr>
</tbody>
</table>

*relative with disease and other were not analyzed due to low numbers of endorsements

**Fisher’s Exact Test
## Results

Comparison of endorsements in younger vs. older participants (N=52)

<table>
<thead>
<tr>
<th>Reason</th>
<th>Younger (18-35)</th>
<th>Older (&gt;56)</th>
<th><strong>p-value</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Cure for MS</td>
<td>74%</td>
<td>39%</td>
<td>0.02</td>
</tr>
<tr>
<td>Have MS</td>
<td>44%</td>
<td>42%</td>
<td>1.00</td>
</tr>
<tr>
<td>Help future generations</td>
<td>30%</td>
<td>39%</td>
<td>0.76</td>
</tr>
<tr>
<td>Find new treatments</td>
<td>30%</td>
<td>12%</td>
<td>0.16</td>
</tr>
<tr>
<td>Improve science</td>
<td>26%</td>
<td>12%</td>
<td>0.27</td>
</tr>
<tr>
<td>Doctor recommendation</td>
<td>4%</td>
<td>15%</td>
<td>0.35</td>
</tr>
<tr>
<td>Encouraged by others</td>
<td>13%</td>
<td>12%</td>
<td>1.00</td>
</tr>
</tbody>
</table>

*relative with disease and other were not analyzed due to low numbers of endorsements

**Fisher’s Exact Test
Preliminary Motivation Results

• **Among Hispanics with MS**, participation in genetic research for MS is strongly associated with having MS.

• **Among non-Hispanics with MS**, participation in genetic research for MS is strongly associated with the desire to find new treatments.

• **Among younger individuals with MS**, participation in genetic research for MS is strongly associated with the desire to find a cure for MS.
‘We SEEC Precision in Medicine’
South-East Enrollment Center (SEEC)

Adopted from the Rural Health Information Hub
(https://www.ruralhealthinfo.org/rural-maps/demographics)
Illustrates 2010 Census Summary Data
Morehouse School of Medicine
- Historically Black College
- 350,000 patients per year
- 1,500 employees

University of Florida Gainesville
- Largest State University in FL
- 1.2M patients with EHR records
- 12,000 employees

University of Miami
- Largest private university in FL
- 1.1M patients per year
- 20,000 employees

Emory University/Emory Healthcare
- Largest private university in GA
- 2.8M patients in 2015
- 24,000 employees
Florida and Georgia
- 30 million individuals in GA and FL
- FL is the 3rd most populous state
- GA is the 9th most populous
- Unique climate and environment

Atlanta metro area (6.2M people)
- 30% African-American

North Florida (4.1M people)
- 32% AA and 18% Hispanics

South Florida metro area (5.5M people)
- 70% Hispanic, highly diverse
- Caribbean blacks

South-East Enrollment Center (SEEC)
Vanderbilt-Miami-Meharry Center for Precision Medicine and Population Health

• Foster research using precision medicine approaches to address racial/ethnic health disparities

• Develop novel methods to integrate individual, contextual and environmental data

• Propel novel health disparities research leveraging genomic and phenotypic data

• Develop ethical, deliberate, socially and culturally acceptable methods for engaging racial and ethnic minorities and vulnerable populations in precision medicine research
Practical Challenges to Consider

- Patient and Research Community Education
- Resources to more actively engage diverse participants
- General long-term sustainability of these resources (paying for storage of both samples and data over the long haul)
- How will large well-publicized national US efforts affect participation in smaller focused biobank and data collections? (i.e. will it help or hurt, how much is dependent on the success of the national effort)
- Diversity isn’t only racial/ethnic, how do we engage/include rural participants and those across the SES spectrum?
- Resources to more actively engage diverse participants
Some Keys to Success

• Build Trust within your participant community
• Be mindful of how results are reported and respect the participant community
• Well-trained personnel (preferably from the community you are engaging)
• Stay engaged with participants (if they want to be engaged) (i.e. newsletters updating your research progress)
• PATIENCE (building trust and biobanks take time)
Translating Genetic Findings into Precision Medicine for ALL

What can we achieve?

• Better understanding of the pathophysiology of disease
• Identify targets for more ‘precise’ treatments
• Provide better predictions of who might be diagnosed with disease
• Give us improved predictions of the disease course
• Lead to knowledge that can help find a cure or prevent disease

Strong need for large well-characterized (genetics, environmental exposures, treatment history, imaging) cohorts with a diverse set of research biospecimens (DNA, RNA, PBMCs, etc.) and ideally longitudinal engagement of diverse participant populations
Acknowledgements

**MS Studies (Hispanics & African Americans)**

Alliance for Research in Hispanic MS (ARHMS):
www.arhms.org

University of Miami
– Silvia Delgado
– Patricia Manrique
– Ashley Beecham
– Gary Beecham
– Michael Cucarro
– Margaret Pericak-Vance

University of Southern California
– Lilyana Amezcua

University of California, San Francisco
– Jorge Oksenberg
– Noriko Isobe

Universidad Central Del Caribe School of Medicine
– Angel Chinea

**IBD Studies (Hispanics)**

University of Miami
– Maria Abreu
– Oriana Damas
– Alejandra Quintero

**ERICH (stroke) Studies (Hispanics & African Americans)**

University of Cincinnati
– Daniel Woo (PI)

**Center for Excellence in Precision Medicine and Population Health (Hispanics & African Americans)**

Vanderbilt University
– Consuelo Wilkins (PI)
– Nancy Cox (PI)

Meharry Medical College
– Maria de Fatima Lima (PI)

University of Miami
– Roy Weiss (PI)

**South-East Enrollment Center**

University of Miami
– Stephan Zuchner (PI)
– Margaret Pericak-Vance (PI)
– Olveen Carraquillo (PI)

University of Florida
– Elizabeth Shenkman (PI)
– William Hogan (PI)

Emory University
– Michael Zwick (PI)

Morehouse School of Medicine
– Priscilla Igho-Pemu (PI)

**Funding/Support**

**MS Studies**

National Multiple Sclerosis Society (RG 4680A1/1)
NIH/NINDS (R01NS096212)

**IBD Studies**

NIH/NIDDK (R01DK104844)

**Stroke Studies**

NIH/NINDS (U01NS069763)

Center for Excellence in Precision Medicine and Population Health
NIH/NIMHD (U54MD010722)

Most importantly our Participants and their Families!