# NHLBI Trans Omics Precision MEDicine (NHLBI TOPMed) Program

ASHG Ancillary Workshop, "NHLBI TOPMed program: How to use WGS and other multi-omics data on over 140K genetically diverse and deeply phenotyped participants to advance your science"

Weiniu Gan, NHLBI



October 26, 2020



## NHLBI's mission

## Leading causes of death World, 2016



BBC

Source: IHME, Global Burden of Disease, Our World in Data

## **Roads toward Precision Medicine**



Gary H. Gibbons, M.D., Director of the National Heart, Lung, and Blood Institute (NHLBI), joined NHLBI in 2012 with his eyes on precision medicine.

NHLBI Workshop, April 9-10, 2014: Genomic Research in Preventing and Treating Heart, Lung, and Blood Diseases <a href="https://www.nhlbi.nih.gov/events/2014/genomic-research-preventing-and-treating-heart-lung-and-blood-diseases">https://www.nhlbi.nih.gov/events/2014/genomic-research-preventing-and-treating-heart-lung-and-blood-diseases</a>

NHLBI Trans Omics Precision MEDicine (TOPMed) Program, 2014—To create a large WGS/Omics data resource supporting discovery research toward precision medicine.  Dr. Gibbons' talk at 5:30 PM EDT, October 29, 2020, The Presidential Plenary Symposium



# Impact of Genetic Research

Source: Vital Statistics of the United States, NCHS.

## Age-Adjusted Death Rates for Coronary Heart Disease, U.S., 1950–2010 Actual Rate and Expected Rates if Rise Had Continued or Reached a Plateau t



## Programmatic Design

- Not to comprehensively identify genetic basis of a couple of common diseases
- But to build genomic databases for many heart, lung, blood diseases and sleep disorders (more than 20 diseases or conditions).
- Uses existing biospecimens from existing studies.
- 1. Minority Populations
- 2. Longitudinal epidemiology studies (e.g., Framingham Heart Study (FHS, 1948-)...)
- 3. Natural history of disease (e.g., Genetic Epidemiology of COPD (COPDGene, 2008-)...)
- 4. Cross sectional disease studies (e.g., My Life, Our Future: Genotyping for Progress in Hemophilia (MLOF)...)
- 5. Family based studies, for example (e.g., Cleveland Family Study...)

# TOPMed Progress – Total WGS/Omics funded in FY14-19



# Type of Omics Products (FY14-19)



# Next, TOPMed2.0

- Release more data to the community; 75% of WGS are released by dbGaP and NHLBI's cloud-based platform: BioData Catalyst.
- 2. Build collaborations with other data sources to future empower research communities
- Generate more Omics data to establish molecular/cellular phenotypes between TOPMed's clinical measurements and WGS data
- 4. Support discovery studies



# Acknowledgments

#### **TOPMed Team**

- Felecia Campbell
- Kristi Cooper
- Shu Hui Chen
- Yi-Ping Fu
- Sweta Ladwa
- Huiqing Li
- James Luo
- Julie Mikulla
- Mollie Minear
- Kyung Moon
- Demetria Pace
- Melanie Peel
- Pankaj Qasba
- Asif Rizwan
- Charlene Schramm
- Phyliss Sholinsky

#### **NHLBI leaders**

- Gina Wei
- David Goff
- Dina Paltoo
- Gary Gibbons

### **TOPMed DCC**

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## **TOPMed IRC**

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TOPMed Investigators

#### **TOPMed Data Generation** Centers

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- Stacey Gabriel, Robert Gerszten, the Broad Institute
- Richard Gibbs, Eric Boerwinkle Baylor College of Med
- Deborah Nickerson, U of Washington
- Nicolas Robine, Catherine Reeves, New York Genome Center
- David J. Van Den Berg, U. Southern Cal

### **BioData Catalyst**



