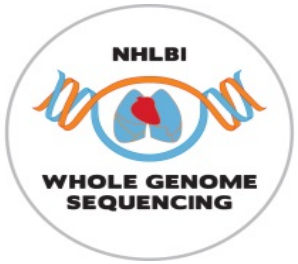


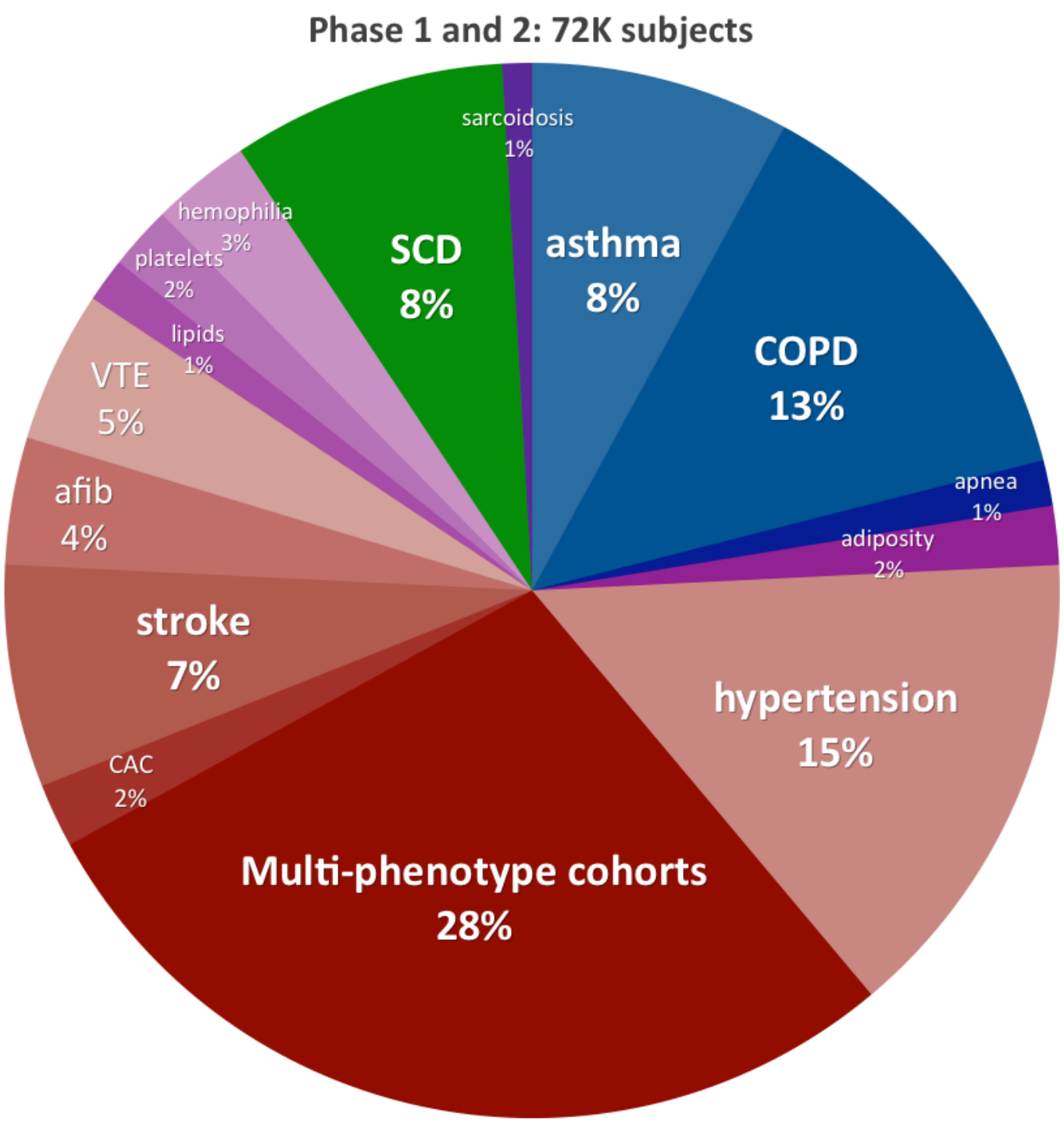
Cross-Cohort Collaboration Consortium
March 11, 2017
Portland, Oregon

Brief TOPMed Introduction

TOPMed Data Coordinating Center
Cathy Laurie – cclaurie@uw.edu

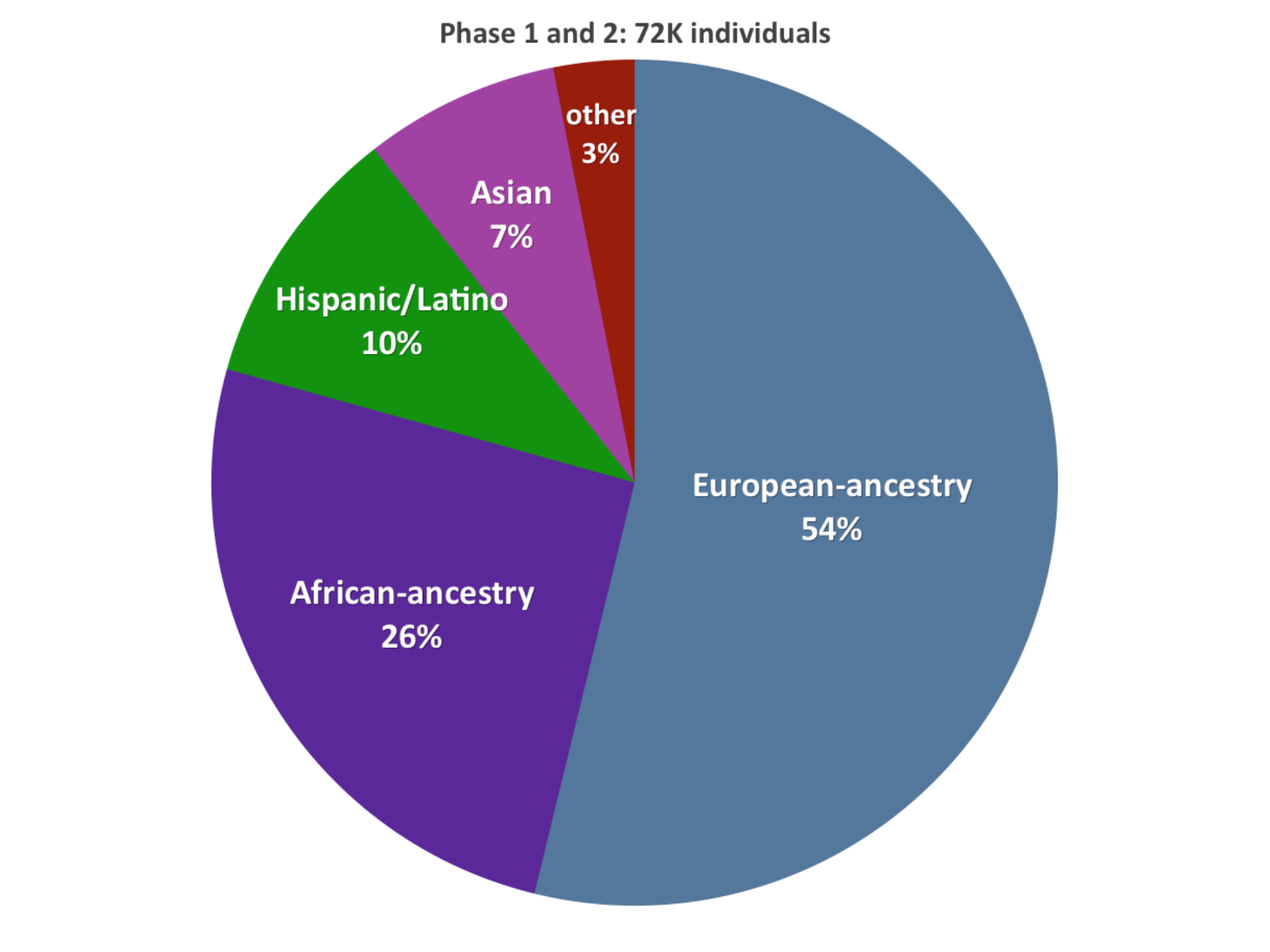
Trans-Omics for Precision Medicine (TOPMed)

- Part of the broader Precision Medicine Initiative
- Identify risk factors for heart, lung, blood and sleep disorders
  - Cross-study analyses to maximize power
  - Requires phenotype harmonization
- Develop targeted and personalized treatments
- Whole-genome sequencing – began Oct 2014, ~70k whole genomes at 30x completed, expect ~120k by end of year 3
- Additional ‘omics planned – RNAseq, metabolomics, Methylation, Proteomics



37 Different studies

Major Cohorts:
Framingham
Jackson
MESA
ARIC
HCHS/SOL
CARDIA
WHI


Phase 1 and 2: 72K individuals
other
3%
Asian
7%
Hispanic/Latino
10%
African-ancestry
26%
European-ancestry
54%

Current Working Groups

Anthropometry – Adiposity
Asthma
Atherosclerosis
Blood Pressure
Bone Mineralization
COPD
Diabetes
EKG – Arrhythmia
Epigenetics
Family Studies
Heart Failure – Cardiac Function & Morphology
Hematology & Hemostasis
Inflammation Biomarkers
Kidney Function
Lipids
Lung Function
Metabolomics
Mitochondrial DNA
PFT
Population Genetics
Reproductive Health
Sarcoidosis
Sickle Cell Disease
Sleep
Smoking
Stroke
Structural Variation
VTE

www.nhlbiwgs.org

Upcoming event of interest: Analysis Commons Hands-On Workshop in Houston, January 24-25, 2017. This is a hands-on workshop to help facilitate genotype-phenotype analysis and discovery using WGS data.
Agenda
Register here: https://www.eventbrite.com/e/analysis-commons-hands-on-workshop-tickets-28585102812?utm-medium=discovery&utm-campaign=social&utm-content=attendeeshare&aff=escb&utm-source=cp&utm-term=listing

Whole Genome Sequencing in the NHLBI Trans-Omics for Precision Medicine

Trans-Omics for Precision Medicine (TOPMed), sponsored by the National Institutes of Health's National Heart, Lung and Blood Institute (NHLBI), is a program to generate scientific resources to enhance our understanding of fundamental biological processes that underlie heart, lung, blood and sleep disorders (HLBS). It is part of a broader *Precision Medicine Initiative*, which aims to provide disease treatments that are tailored to an individual's unique genes and environment. TOPMed will contribute to this initiative through the integration of whole-genome sequencing (WGS) and other –omics (e.g., metabolic profiles, protein and RNA expression patterns) data with molecular, behavioral, imaging, environmental, and clinical data. In doing so, this program seeks to uncover factors that increase or decrease the risk of disease, identify subtypes of disease, and develop more targeted and personalized treatments.

The *Whole Genome Sequencing* (WGS) project is part of NHLBI's TOPMed program and serves as an initial step for the larger initiative. In recent years, genetic research of complex disease using Genome-Wide Association Study (GWAS) and Exome-sequencing approaches has resulted in an unprecedented explosion of genetic discovery. However, a large portion of heritability in complex diseases remains elusive. Whole Genome Sequencing (WGS) will provide a comprehensive view of the genome, an opportunity to further understand the genetic architecture relevant to HLBS disorders, and an unprecedented resource to the scientific community.

The WGS project started in 2014 to generate deep WGS data for studies with diverse ancestries and extensive characterization of HLBS-related traits. The current TOPMed project studies have a variety of study

« December »

S	M	T	W	T	F	S
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11	12	13	14	15	16	17
18	19	20	21	22	23	24
25	26	27	28	29	30	31

iCal

Note: All events are noted in Pacific time.

- No Sample and Data Processing call
  12/28/16
- Lipids Working Group weekly call
  12/28/16 -
  06:00 am to 07:00 am
  PST
- Orientation for TOPMed