# Genetic Analysis of Disease in Diverse Individuals

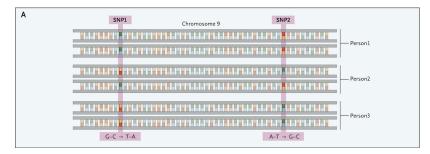
LA's Best 6/17/2024

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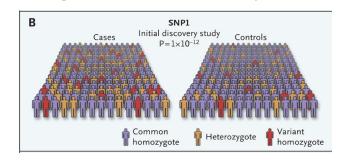


## Genome-wide association studies

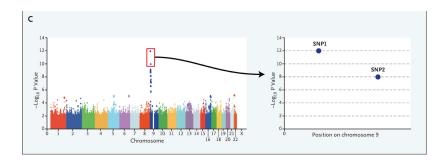
### Quantify common genetic variation (SNPs)



#### Large cohorts and simple models



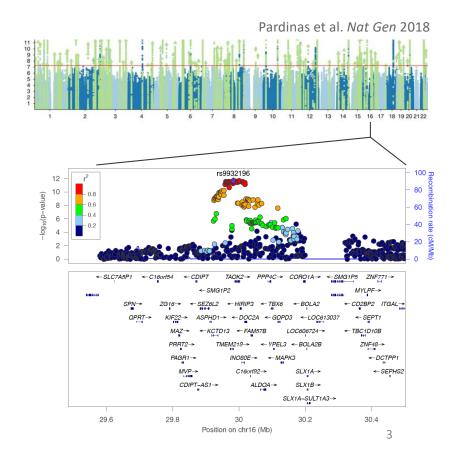
#### Test for association between SNP and disease



Manolio TA. N Engl J Med 2010

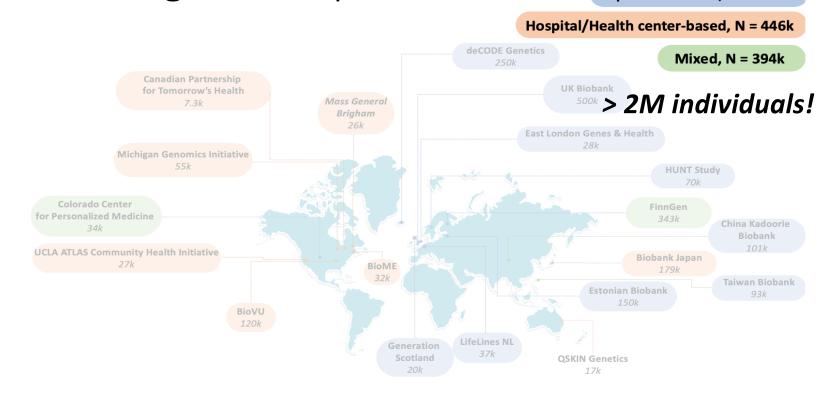
## GWAS does not provide *causal mechanisms*

- GWAS provides regions associated with disease risk
- What are the mechanisms driving disease risk?
- Most associations are non-coding
  - SNPs with regulatory function are strong candidates

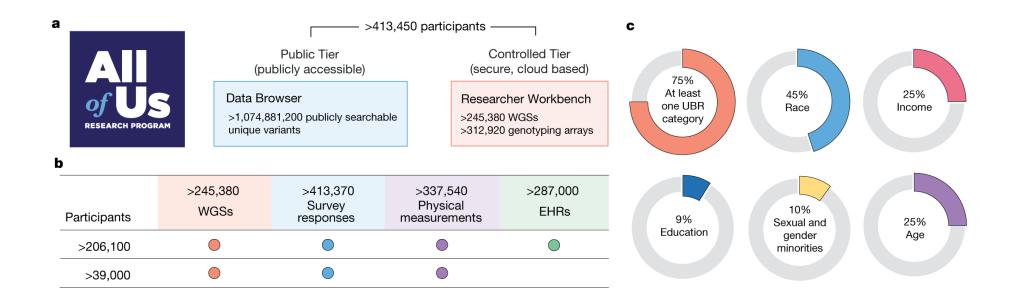


Maurano et al., *Science* 2013; Aguet et al., *Science* 2020; ENCODE et al., *Nature* 2020

Global biobanks reflect *massive* scale of available genome/phenome



## All of US Biobank



# LA's Best Project: All of Us Analysis

#### • Project Aims:

- Learn to navigate genetic + phenotypic data on AOU portal
- Select phenotype and construct analysis group (non EHR related!)
- Visualize diverse ancestry components using PCA
- Perform GWAS

#### Deliverables

• Small presentation/writeup on any potential genetic findings