1000 Genomes admixture tutorial

Alicia Martin
Massachusetts General Hospital, Broad Institute
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armartin@broadinstitute.org
How has human evolutionary history shaped genetic -> phenotypic diversity?

• What insights into human history can be made by assessing genetic divergence among global populations?

• How do selective pressures shape complex phenotypic variation?

• How does haplotype structure inform recent demographic history?
Insights into human demography

• Admixture among divergent groups
• Population divergence
• Population expansions/contractions
  • Recent (up to ~200 generations) haplotype-based inference
  • Older (100s - 1000s of generations) site frequency-based inference
Substantial global genetic diversity in 1000 Genomes
Varying admixture proportions across groups in the Americas

Native American ancestry in the ASW

Hispanic/Latino ancestry in the ASW

African descent ancestry in the ASW

Reference panel
Admixture as a lens into recent demography
Admixture as a lens into recent demography
Admixture as a lens into recent demography
Hispanic/Latino European admixture originated from Spain

Moreno-Estrada, et al. (2013). PLoS Genetics
Sex-biased demographic events

MXL

NAT chrX vs NAT autosome

EUR chrX vs EUR autosome

AFR chrX vs AFR autosome

PUR chrX vs NAT autosome

PUR chrX vs EUR autosome

PUR chrX vs AFR autosome
Migration events in southern Africans

- Khoesan are among most genetically diverged modern human populations, reside in southern Africa.
- Bantu expansion reached southern Africa ~1500 years ago.
- Dutch settlers beginning ~1700.
- Pigmentation allele frequencies

Gravel, S. (2012). Genetics
https://github.com/sgravel/tracts
Recent haplotype-based demographic inference in Finland

![Graph showing mean R² between SNPs and Ne (x 10³) over generations before present (g) for different populations.](image)
Summary

• Much more population-level information from genotype, sequence data than PCA coordinates

• Admixed samples are easiest case to look into recent history, can look at detailed populations history

• 1000 Genomes samples provide reference panel for demographic inference into new samples

• Haplotype sharing analogously enables demographic history inference in the absence of admixture
Resources

- ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20140818_ancestry_deconvolution/
- https://github.com/armartin/ancestry_pipeline
Thank you!

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