

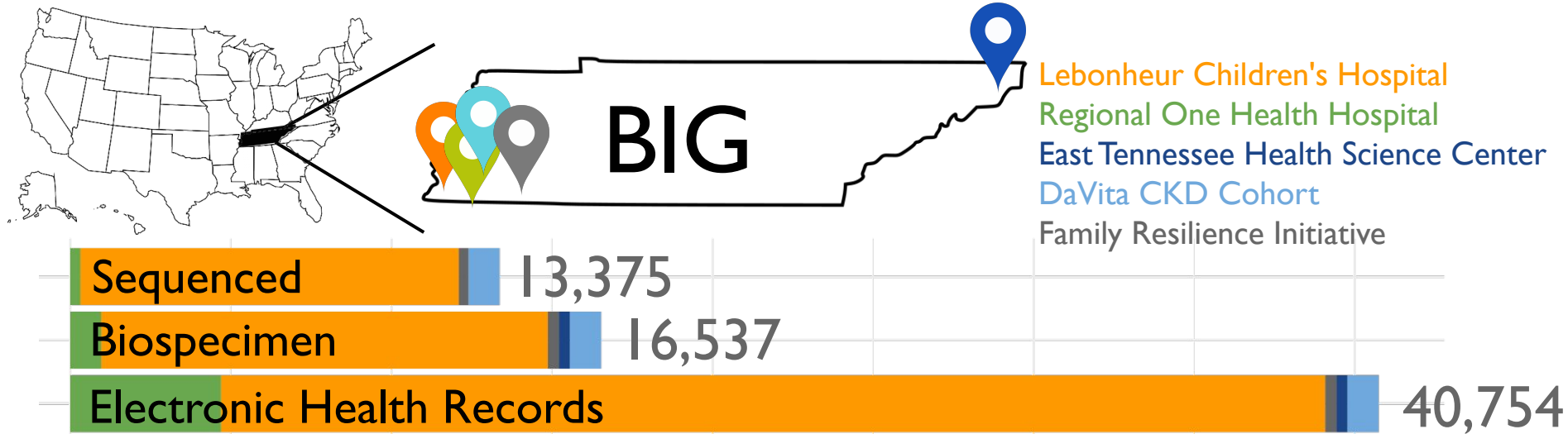
Identity-by-descent in HPRCv2

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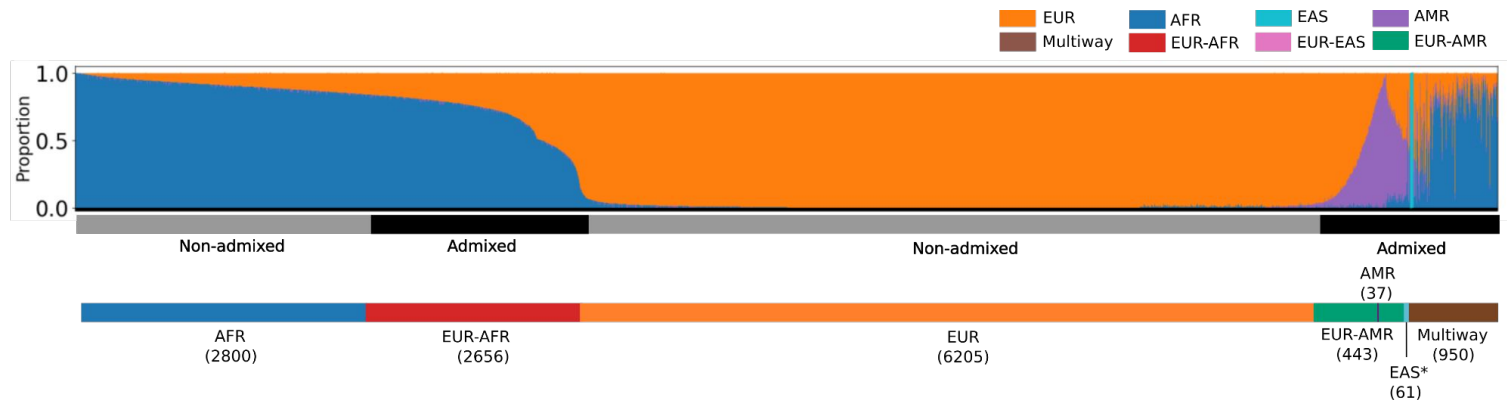
University of Tennessee Health Science Center

2025 June 3

The Biorepository for Integrative Genomics initiative



Ancestry inference in BIG



- Four continental level ancestries
- Relevant proportion of admixed individuals
- High genetic diversity geographically concentrated

nature communications

Article

<https://doi.org/10.1038/s41467-025-59375-0>

Insights from the Biorepository and Integrative Genomics pediatric resource

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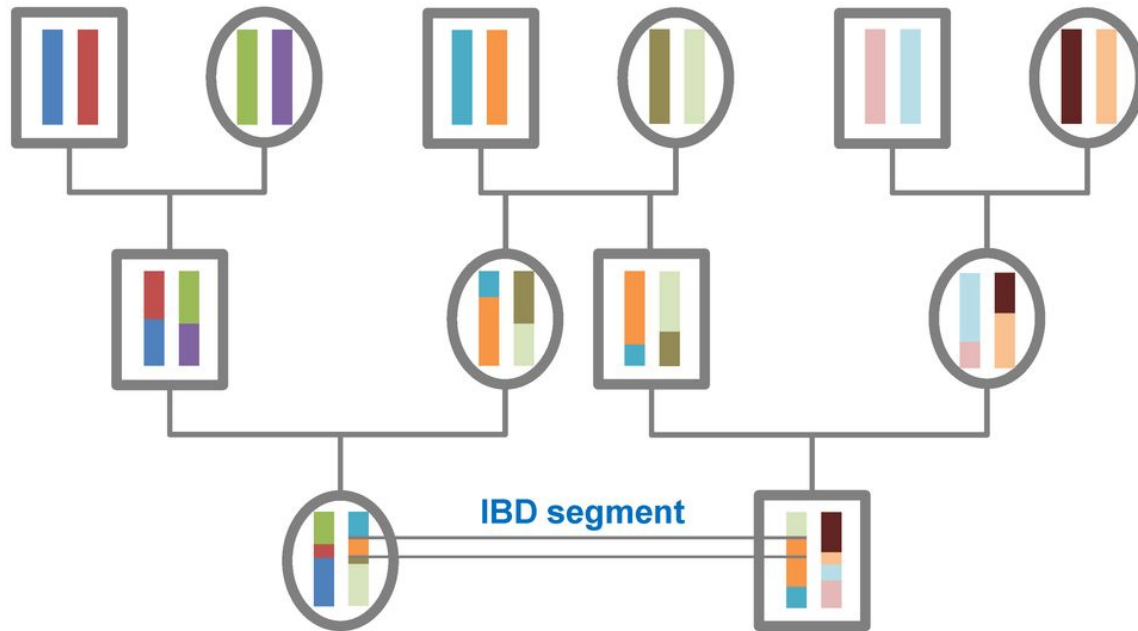
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Check for updates

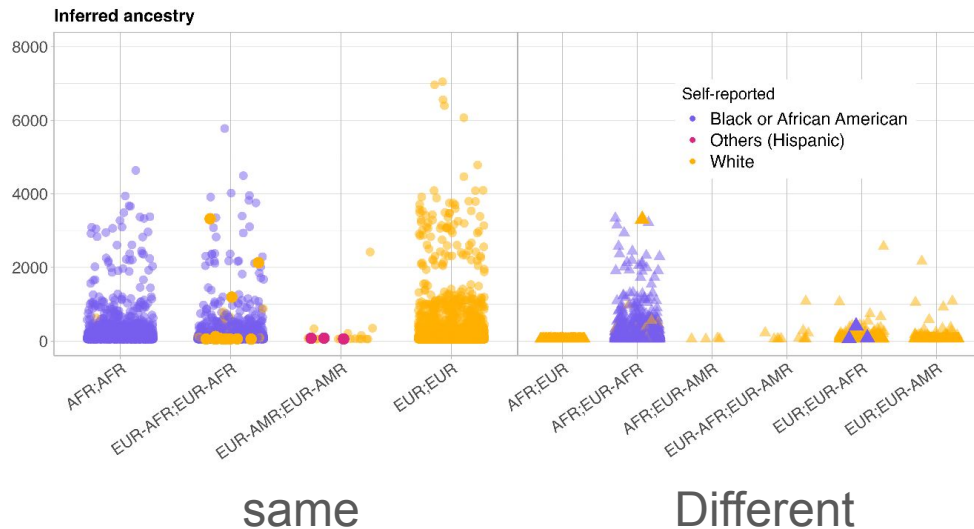
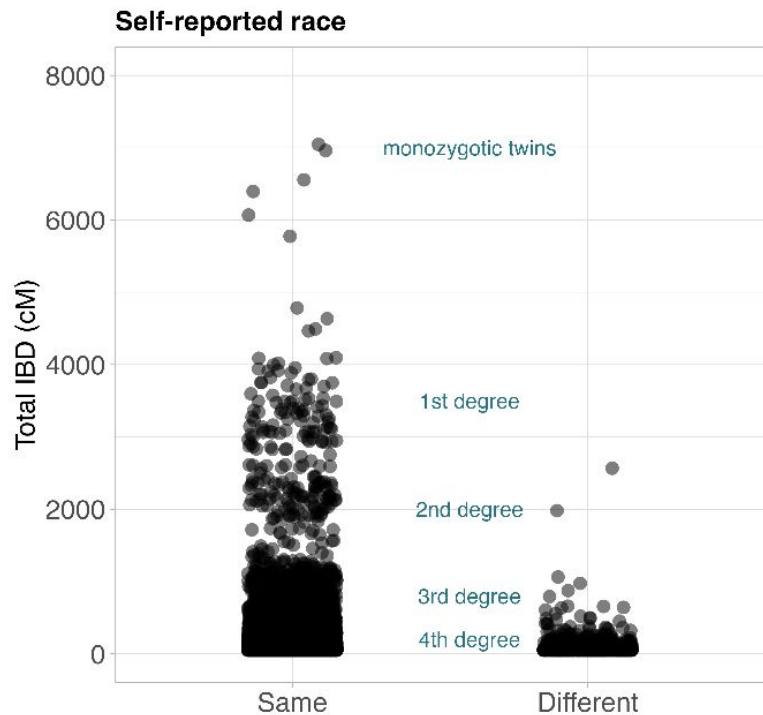
Silvia Buonaiuto^{1,10}, Franco Marsico^{1,2,10}, Akram Mohammed³, Lokesh K. Chinthala³, Ernestine K. Amos-Abanyie¹, Regeneron Genetics Center*, Piotr Prins¹, Khyobeni Mozhui^{1,4}, Robert J. Rooney^{1,5}, Robert W. Williams^{1,6}, Robert L. Davis², Terri H. Finkel⁷, Chester W. Brown^{1,8} & Vincenza Colonna^{1,2,5}

Buonaiuto* , Marsico* , Nat Comm, 2025

Identity by Descent (IBD)

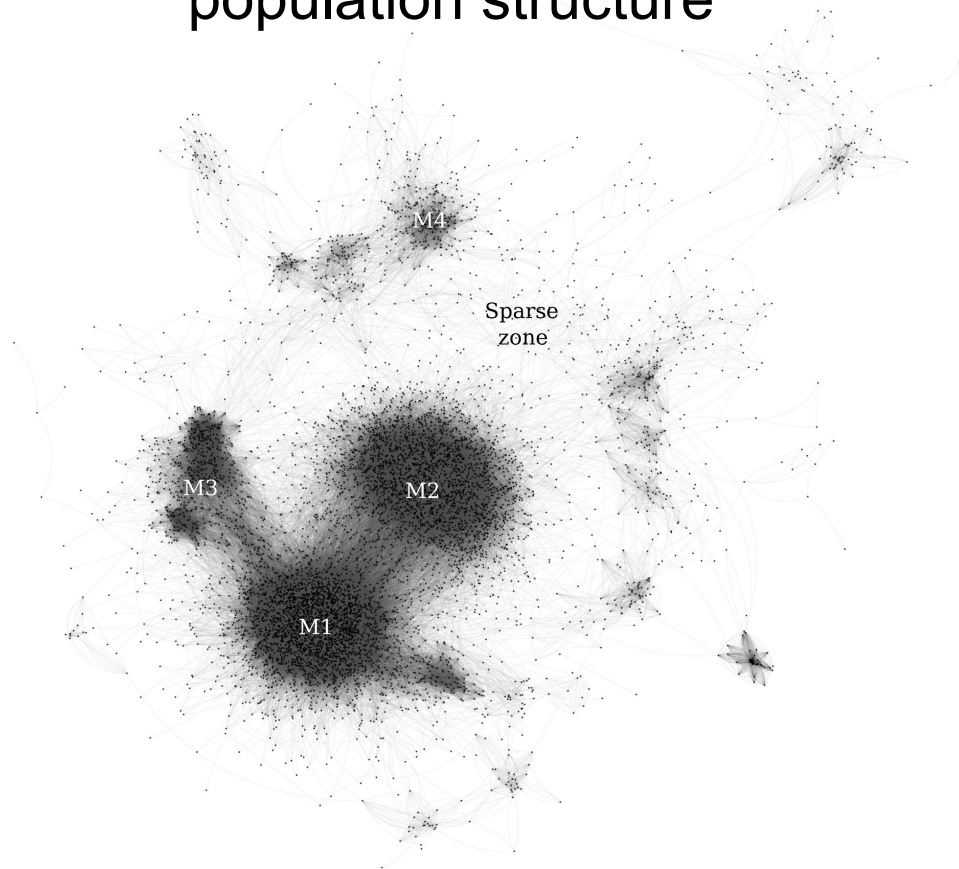


Self-reported race and genetic ancestry through the lens of relatedness

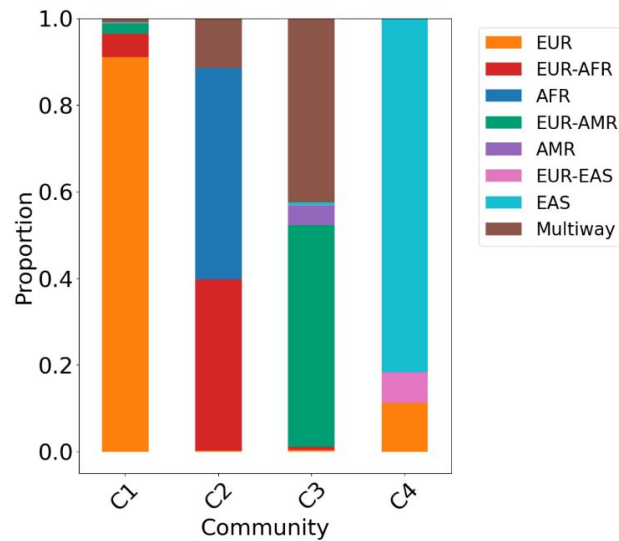


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From pairwise IBD to population structure



Leiden algorithm for community detection

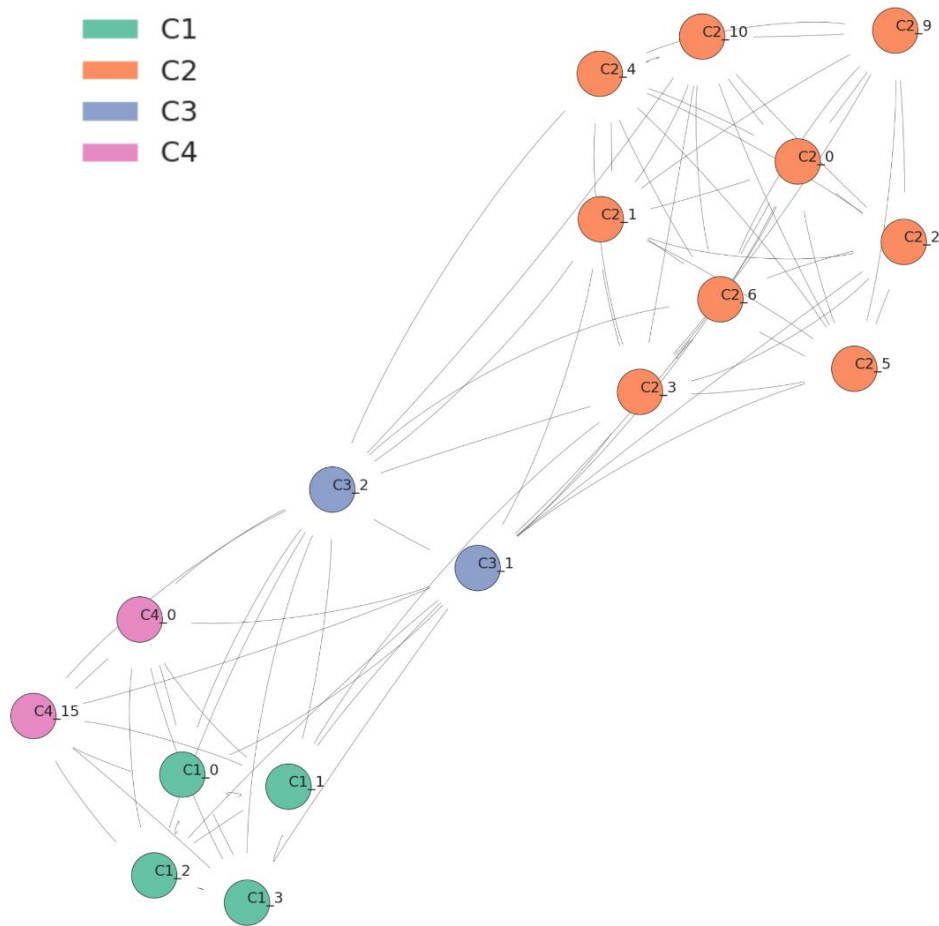


Beyond ancestry: grouping individuals based on how much genome they actually share

Identity-by-descent captures Shared Environmental
Factors at Biobank Scale

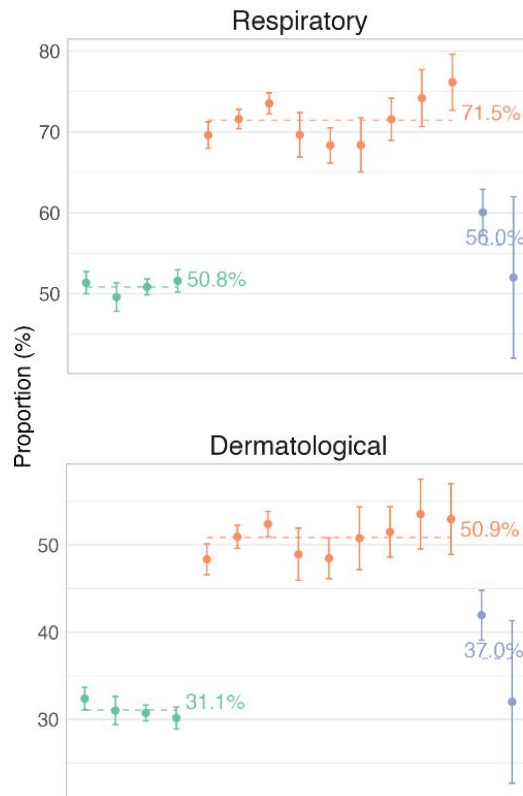
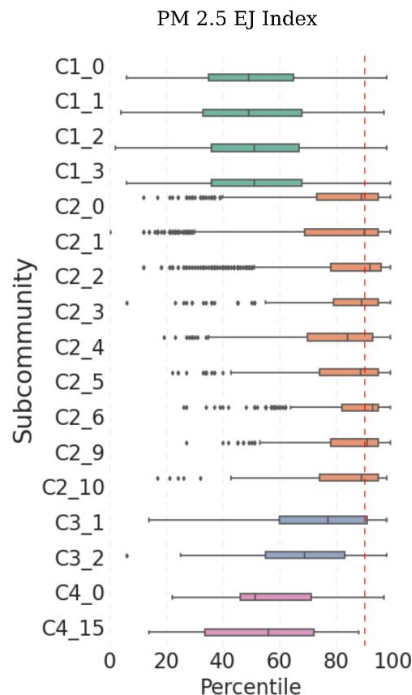
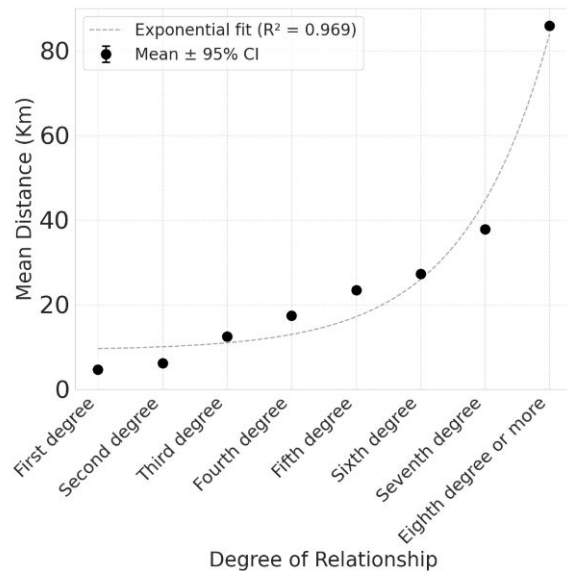
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<https://www.biorxiv.org/content/10.1101/2025.05.03.652048v1.full>



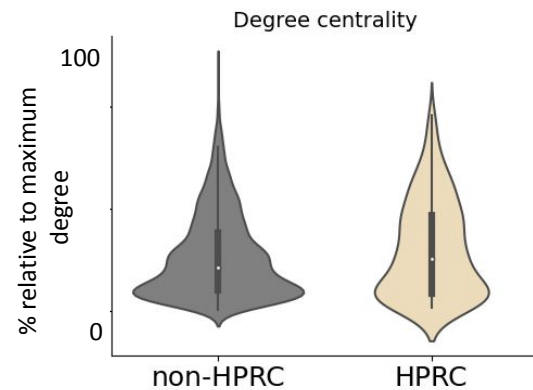
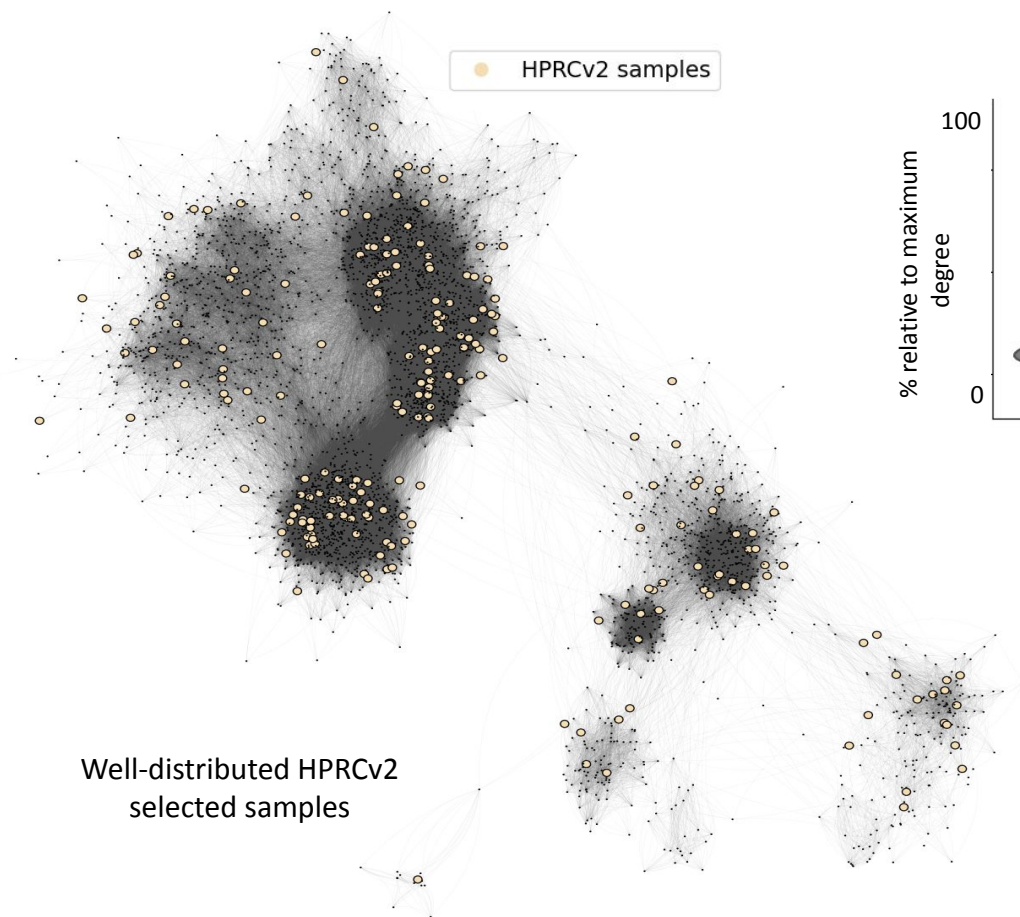
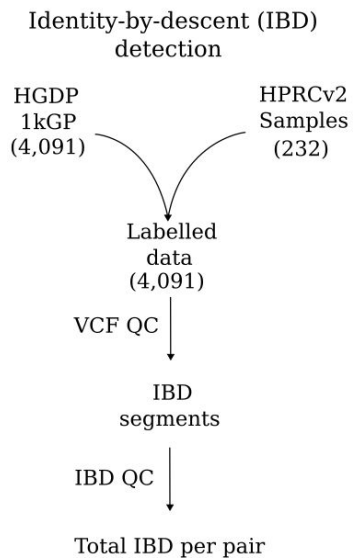
Identity-by-descent captures shared environment

Genealogical and geographic distance



Can a similar approach be
useful on HPRCv2?

Preliminary/pilot results

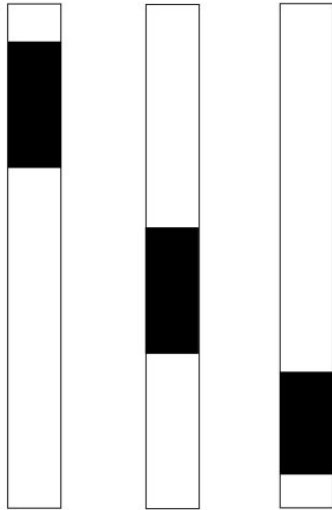


Higher centrality in HPRC samples

HPRCv2



HGDP-1kGP

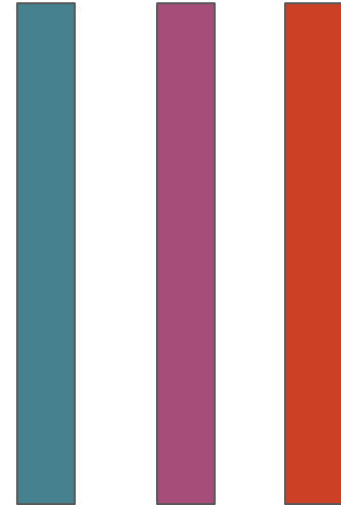


Haplotype informativeness: Compute the amount of cM explained by IBD sharing between the selected samples and public resources (HGDP-1kGP).

HGDP
1kGP



HPRCv2



Proportion already explained: Compute the percentage (amount of cM) already explored by IBD sharing of a potential HGDP-1kGP sample to be incorporated to HPRCv2.

Thank you

<https://github.com/MarsicoFL/HPRCv2-IBD>