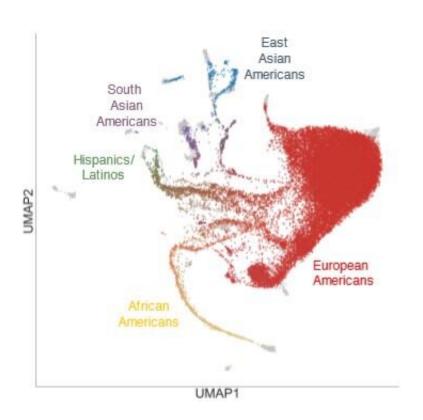


Genetic study offers comprehensive and diverse view of recent US population history

March 5 2020



A dimensionality reduction method, UMAP, applied to genetic data from the National Geographic Genographic Project, where each dot represents one individual, with colors determined from a linear combination of each individual's admixture proportions (colors correspond to each continental ancestry, with European = red, African = yellow, Native American = green, East Asian = blue, South Asian = purple). Credit: Dai et al./AJHG

Researchers have assembled one of the most comprehensive studies of



population genetics ever conducted in the United States, bringing together large-scale genetics data from more than 32,000 participants in the National Geographic Genographic Project. This new view on the US, appearing March 5 in the *American Journal of Human Genetics*, reveals a remarkable degree of complexity. Beyond offering an intriguing view of the nation's recent history, the findings also have important implications for health and medicine, the researchers say.

"We have inferred US <u>demographic history</u> at fine scale by combining genetic, geospatial, and multigenerational birth record data," says Alicia Martin (@genetisaur) of the Broad Institute of MIT and Harvard. "This provides important lessons for the future: if we want <u>genetic</u> <u>technologies</u> to benefit everyone, we need to rethink our current approach for <u>genetic studies</u> because they typically miss a huge swath of American—and more broadly human—diversity."

Although much effort has been made to understand the genetic diversity in the US, fine-scale patterns were less clear for minority populations, particularly for Latin American and African descendants with complex ancestral histories. There also had been little research on the population structure of individuals in the US with East Asian, South Asian, and Middle Eastern ancestry.

Martin and colleagues report that the new effort had several distinct advantages compared to other large-scale population genetics datasets. First, genetic data for each consenting participant from the Genographic Project are accessible to researchers around the world to answer anthropological questions. Most of the study's participants also reported their postal code along with birthplace and ethnicity data for themselves, their parents, and their grandparents, enabling fine-scale insights into recent history.

Their analysis revealed the following:



- Hispanic/Latino populations show a remarkably complex population structure, including geographically varying proportions of Native American, European, and African ancestry.
- The evidence uncovered a north-south barrier to migration among African Americans, consistent with the transatlantic slave trade and historical patterns of segregation between states.
- Some Asian Americans, a fast-growing group in the US that had been often overlooked in past genetic studies, show elevated patterns of relatedness consistent with intermarrying between close kin among their ancestors.
- People of European descent show a subtle, unevenly distributed structure. For example, Irish, Finnish, and Acadian (French Canadian) descendants are common in New England, the Northwest, and the South, respectively.

Martin also noted that Hispanic/Latino populations have native origins spanning North, Central, and South America over several generations. These roots vary in how much and how long ago they appear in Californians, New Mexicans, Texans, and Floridians on average.

The researchers say that these findings are "the tip of the iceberg." As the number of participants in this and other ongoing studies continues to grow, the view of US population history will become even clearer.

Martin says they will continue this work as the available data grow to include about 250,000 Americans. In future studies, they also plan to expand to people who may or may not live in the US as well, which, Martin says, will help to tease out relationships among underrepresented populations, such as Hispanic/Latino populations in Florida who have ancestral roots from Cuba, Puerto Rico, Mexico, and elsewhere.

More information: American Journal of Human Genetics, Dai et al.:



"Population histories of the United States revealed through fine-scale migration and haplotype analysis" www.cell.com/ajhg/fulltext/S0002-9297(20)30044-6, DOI: 10.1016/j.ajhg.2020.02.002

Provided by Cell Press

Citation: Genetic study offers comprehensive and diverse view of recent US population history (2020, March 5) retrieved 6 May 2024 from https://medicalxpress.com/news/2020-03-genetic-comprehensive-diverse-view-population.html

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