

Dynamics of genetic admixture in Brazilian populations

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Credit: Marcelo Calvet/Wikipedia

(Phys.org)—Human genomic diversity studies provide a window to population movements across regions and societies throughout history. Generally, South America has been underrepresented in such studies, but

recognizing that Brazil provides a classical model of population admixture, an international group of researchers recently conducted a population-based, genome-wide analysis of three Brazilian populations.

Their study, the EPIGEN Brazil Initiative, represents the most comprehensive genomic analysis of any South American population to date. They have published their results in the *Proceedings of the National Academy of Sciences*.

The population of Brazil consists of the post-Columbian admixture between Amerindians, European colonizers and immigrants, and African slaves. The authors note that Brazil was the destiny of 40 percent of the African diaspora that characterized the period of the slave trade. Brazil received seven times more slaves than the United States during that period.

From 6,487 admixed Brazilian individuals, the researchers genotyped nearly 2.2 million single nucleotide polymorphisms (SNPs, colloquially called "snips"), which are DNA sequence variations that occur commonly within populations. They studied three population-based cohorts from different regions with distinct socioeconomic backgrounds.

This population-based approach allowed the researchers to identify and quantify [ancestry](#) SNP components of three representative Brazilian populations for the first time; they developed an approximate Bayesian analysis to infer properties of population admixture; they identified how genetic structure was influenced by ancestry-related social history; and they were able to study the interactions of admixture, kinship, and inbreeding on patterns of deleterious genetic mutations.

The populations in question were from Salvador, a coastal city with 2.7 million inhabitants; Bambuí, a city with a population of around 15,000; and Pelotas, a city of 214,000 inhabitants. Across these three regions, the

researchers traced a historical pattern of sex-biased preferential mating between men with predominant European ancestry and women with predominant African or Amerindian ancestry.

Families from Salvador and Pelotas, the cities with the largest populations, had the lowest rates of consanguinity. By contrast, Bambuí, consisting of the smallest population in the study, had the highest family structure and the most inbreeding, which was correlated with European ancestry.

Genomic ancestry in Brazil correlates with a set of phenotypes such as self-reported ethnicity, skin color, and social aspects such as socioeconomic status. The authors write, "... after five centuries of admixture, Brazilians still preferentially mate with individuals with similar ancestry (and its correlated morphological phenotypes and socioeconomic characteristics), a trend also observed in Mexicans and Puerto Ricans."

This was particularly true in the cities of Pelotas and Bambuí, which have higher proportions of individuals with markedly predominant ancestries. In Salvador, however, the [population](#) is far more admixed, likely due to a combination of factors including a longer history of admixture and the relatively homogenous socioeconomic status of the inhabitants, according to the authors.

The authors investigated how European ancestry shapes the amount of deleterious genetic variants in admixed individuals across domains of heterozygosity and homozygosity. They report that in Latin-American populations, the history of continental admixture comprises the main determinant of the presence of deleterious variants, but in a much more complex way than they expected, and likely unrelated to local demographic history.

They suggest that future studies on Northern Brazilian populations or those in the Central-West might reveal larger dynamics of Amerindian ancestry. They also speculate that studies of large urban centers that historically serve as destinations for immigration might reveal the influence and impacts of other global ancestry components.

More information: "Origin and dynamics of admixture in Brazilians and its effect on the pattern of deleterious mutations." *PNAS* 2015 ; published ahead of print June 29, 2015, [DOI: 10.1073/pnas.1504447112](https://doi.org/10.1073/pnas.1504447112)

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