

## THE ANCESTRAL ROOTS AND EVOLUTION OF BRAZILIANS

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For the past 15 years our group has been employing DNA-based genetic markers to study the origins and structure of Brazilian populations. We first demonstrated the existence of a founder effect on the Y-chromosome of Native Americans, which was used to show that the origin of Amerindian populations was Central Siberia. We then applied the molecular tools of Y-chromosome analysis and mitochondrial DNA analysis to the study of the Brazilian population. Among individuals self-classified as Whites, the vast majority of Y-chromosomes proved to be of European origin. On the other hand, mtDNA revealed that the majority of matrilineages were of Amerindian and African origin, a much higher level of genetic admixture than had previously been thought. These data configured a picture of strong directional mating between European males and Amerindian and African females, which agrees with the known history of the peopling of Brazil. In parallel studies we found that only 48% of the Y-chromosomes, but 85% of the mtDNA haplogroups of Afro-Brazilians were characteristic of sub-Saharan Africans. These data are also indicative of sexually biased mating in the evolution of Brazilians. Using these mtDNA data as reference, it was possible for the first time to obtain an estimate of the relative ancestral contribution of West-Central, West and Southeast Africa to Brazilian Blacks. We also studied the correlation between genetic ancestry and skin color in Brazilians. The data essentially showed that in Brazil color is a weak individual predictor of African ancestry. To assess more accurately the contribution of the African, European and Amerindian ancestral roots to the genomic ancestry of Brazilians, we developed a set of 40 insertion-deletion (indel) polymorphisms that has been validated on a study of worldwide human populations as useful ancestry informative markers. A variety of other genomic markers has also been applied to analyze the genetic structure of the Brazilian population, including X-chromosome haplotypes and indels on the long arm of chromosome 5. Altogether, our data show that, regardless of their skin color, the overwhelming majority of Brazilians simultaneously has a significant degree of African, European and Amerindian ancestry. The only paradigm to deal with genetic variation in Brazilians is on a person-to-person basis, as singular individuals with unique genomes and exclusive life histories. Our results establish a genetic backdrop for public policy.