

500 YEARS LATER: UNDERSTANDING THE GENETIC ANCESTRY OF THE SOUTHEAST REGION, BRAZIL

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Southeast region of Brazil is the main economic region, the most populous and with the major cultural and genetic miscegenation of the country. Due to its historical context, it mainly includes descendants of Portuguese and Italians, but also has strong influence of Amerindians, Africans, Spaniards and Germans. The influence of Arabs and Asians is very small. In this work, the ancestral proportions of the population of the Southeast region were evaluated through a genetic data compilation with diverse markers (Y-SNPs, AIM-INDELS, mtDNA and X-INDELS) in individuals from the states of São Paulo, Rio de Janeiro, Minas Gerais and Espírito Santo, which form this region. The proportion of paternal lineages (Y-SNPs) with European origin is much higher (86.57%), being 10.86% of African origin and 1.98% native American. Instead, the proportion of African origin (44.1%) predominates in the maternal lineages (mtDNA), followed by Native American with 29.2%, European with 26.2% and only 0.5% for Asian. With the autosomal AIM-INDELS the ancestral proportion obtained was 59.12% European, 28.12% African and 12.76% Native American. The autosomal chromosomes tend to reflect an average between the ancestry proportions of the Y chromosome and mtDNA. No Asian ancestry was identified with these markers. In preliminary analyzes with X-INDELS in samples from São Paulo, we found the following ancestral proportions: 29.4% African, 49.7% European and 20.9% Native American. This genetic miscegenation occurred since the formation of this region, when Portuguese colonizers mated with native women and later with slaves brought from Africa.

1. Figueiredo RF, Ambrosio IB, Braganholi DF, Chemale G, Martins JA, Gomes V, Gusmão L and Cicarelli RMB. Male-specific contributions to the Brazilian population of Espírito Santo. *International Journal of Legal Medicine*. 2015; doi:10.1007/s00414-015-1214-2.
2. Ambrosio IB, Braganholi DF, Polverari FS and Cicarelli RMB. Mitochondrial DNA 30-SNP data confirm high prevalence of African lineages in the population of Espírito Santo, Brazil. *Forensic Science International: Genetics Supplement Series*. 2015; Volume: 5, Pages e346–e347.
3. Manta FSN, Pereira R, Vianna R, Araújo ARB, Gitaí DLG, Da Silva DA, Wolfgramm EV, Pontes IM, Aguiar JI, Moraes MO, Carvalho EF and Gusmão L. Revisiting the genetic ancestry of Brazilians using autosomal AIM-Indels. *PLoS One*. 2012; Volume: 8, 2012. doi: 10.1371/journal.pone.0075145