

Distribution of local ancestry and evidence of positive selection in Brazilian individuals

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Introduction: In genome-wide association studies, admixed populations have the advantage of increase the probability to identify causal variants due to creation of local ancestry tracts across the genome. Local ancestry has been inferred in admixed populations, but poorly studied in Brazilian individuals. Therefore, our main goals were to describe the distribution of local ancestry and to identify deviations across the genome in a Brazilian sample.

Materials and Methods: We evaluated 264 individuals obtained within the scope of the Brazilian Initiative on Precision Medicine, and compared with data from the 1000 Genome Project and to additional 43 Native American samples from previous studies. Genotyping calling was performed by Genome-Wide Human SNP Array 6.0 platform. Individual and genotype filtering was performed by PLINK 1.9 software, which was also used to evaluate population structure by principal component analysis. Global and local ancestry was estimated by ADMIXTURE and RFMIX algorithm, respectively.

Results: As expected, Brazilian sample presented high proportion of South European ancestry, followed by West African and Native American ancestries. However, local ancestry revealed a decreased European ancestry proportion followed by an excess of Native American on chromosome (ch) 8p23.1.

Discussion: We showed that this deviation is due to haplotypes created by inversion events. In addition, Brazilian non-inverted haplotypes were found to be more similar to Native Americans than European haplotypes, different than what was found for other admixed populations (Puerto Rican and Colombian samples). We also found signals of recent positive selection on ch 8p23.1, and one gene (*PPP1R3B*) located within the target region is related with diet and have already been associated with type 2 diabetes and obesity, which strength the positive selection hypothesis.

Conclusions: Our results add an important local ancestry information for both Brazilian and other admixed populations. In addition, our finding is also relevant for precision medicine, since it shows that a result derived from a strict population study may also have implications for medicine.