Self-declared ethnicity and genomic ancestry in prostate cancer patients from Brazil

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ABSTRACT. Some studies of polymorphisms in prostate cancer (PCa) analyze individuals in a uniform manner, regardless of genetic ancestry. However, PCa aggressiveness differs between subjects of African descent and those of European extraction. Thus, genetic ancestry

analysis may be used to detect population stratification in case-control association studies. We genotyped 11 ancestry informative markers to estimate the contributions of African, European, and Amerindian ancestries in a case-control sample of 213 individuals from Bahia State, Northeast Brazil, including 104 PCa patients. We compared this data with self-reported ancestry and the stratification of cases by PCa aggressiveness according to Gleason score. A larger African genetic contribution (44%) was detected among cases, and a greater European contribution (61%) among controls. Self-declaration data revealed that 74% of PCa patients considered themselves non-white (black and brown), and 41.3% of controls viewed themselves as white. Our data showed a higher degree of European ancestry among fast-growing cancer cases than those of intermediate and slow development. This differs from many previous studies, in which the prevalence of African ancestry has been reported for all grades. Differences were observed between degrees of PCa aggressiveness in terms of genetic ancestry. In particular, the greater European contribution among patients with high-grade PCa indicates that a population's genetic structure can influence case-control studies. This investigation contributes to our understanding of the genetic basis of tumor aggressiveness among groups of different genetic ancestries, especially admixed populations, and has significant implications for the assessment of inter-population heterogeneity in drug treatment effects.

Key words: Admixed population; Ancestry informative markers; Single nucleotide polymorphism; African ancestry; European ancestry; Amerindian ancestry