(TTTA)n polymorphism of CYP19 (aromatase gene) in Euro- and Afro-Brazilians

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Abstract

We investigated the polymorphic tetranucleotide repeat (TTTA)n located in the fourth intron of the CYP19 gene in two Brazilian populations. The frequencies of the five common alleles (A) in Euro- and Afro-Brazilians were, respectively: seven repeats (A5), 0.586 and 0.80; eight repeats (A4), 0.092 and 0.06; nine repeats (A3), 0.014 and 0.01; eleven repeats (A2), 0.284 and 0.09; twelve repeats (A1), 0.021 and 0.04. In addition, one Euro-Brazilian individual had a rare allele with 13 repeats. The allelic frequencies in Euro- and Afro-Brazilians differed statistically (p < 10^{-3}). The two samples were found to be in Hardy-Weinberg equilibrium (p = 0.828 and p = 0.995).

Key words: CYP19, gene polymorphism.

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Previous studies had shown that \textit{CYP19} might be involved in breast cancer susceptibility (Kristensen \textit{et al.}, 1998; 2000; Siegelmann-Danieli and Buetow, 1999; Miyoshi \textit{et al.}, 2000), probably due to its role in the conversion of C19 steroids into estrogens. Population differences in the frequency of the \textit{CYP19} polymorphism as we disclosed here between Afro- and Euro-derived Brazilian populations are crucial in the interpretation of these association studies.

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