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Mapping gene variation in sub-Saharan African populations

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ABSTRACT

The present study examined the distribution of six genetic variants (*CYP17A1*, *CYP3A4, SRD5A2, KLK3, AR*) in the androgen biosynthesis and metabolism pathway, in 14 sub-Saharan African populations. These polymorphisms have been implicated in several complex diseases, most notably prostate cancer. In order to elucidate the frequencies of these genetic variants, PCR-RFLP and STR based methodologies were employed. Consistent with previously reported results, the frequency distribution of the gene variants in the examined populations greatly coincided with prostate cancer incidence and geographic origin. Populations of African descent had the highest frequencies of the alleles that are postulated to increase risk to prostate cancer, whilst Asian populations had the lowest. Also, there were evident differences in the frequencies of these variants between populations of different continental origin particularly between African and Eurasian populations. The distribution of these genetic variants was further used to assess the spectrum of variation within Africa. The results were greatly aligned with those previously reported, providing further support to the origin and evolution of modern humans from Africa as well as other historic events.