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Multilocus Genetic Risk Profiling of Atherosclerosis and Venous Thromboembolism in Azores and Mainland Portuguese Populations

Genome-wide association studies (GWAS) have identified multiple genetic variants associated with cardiovascular disease risk, yet population-specific differences remain insufficiently characterized, particularly in geographically isolated regions. This study investigated whether the frequencies of single nucleotide polymorphisms (SNPs) associated with atherosclerosis (AT) and venous thromboembolism (VTE) differs between populations from Azorean archipelago and mainland Portugal, and how these compare with other global populations. A total of 19 SNPs related to AT and VTE were analysed in samples from the Azores and mainland Portugal, and allele frequencies were compared with European, Asian, and African reference populations using chi-square tests. Multilocus genetic profile (MGP) analyses were performed to estimate cumulative genetic risk for both conditions. No significant differences were observed between Azorean and mainland Portuguese populations at the single-variant level; however, eight SNPs in ADAMTS7, PCSK9, APOE, and LDLR genes showed significant differences compared with European populations ($p < 0.05$). MGP analysis indicated that 11.2% of Azoreans and 7.4% of mainland individuals present a high genetic risk for atherosclerosis, whereas VTE risk was higher in the mainland population (6.5%) than in the Azores (4.1%). Significant intra-archipelago differences were observed, with the Eastern group showing the highest VTE risk and the Central group the highest AT risk. Overall, genetic risk patterns were broadly consistent with European populations, but regional differences suggest that local genetic background and socio-cultural factors, including diet and physical activity, may modulate disease risk. These findings highlight the importance of population-specific genetic assessments and support their integration into preventive and personalized cardiovascular healthcare strategies, particularly in small and isolated populations such as the Azores.

Keywords: Azores; Portugal; Atherosclerosis; Thromboembolism; Genetic predisposition to disease; Cardiovascular disease.



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Biography

Cláudia Margarida Aguiar Castelo Branco is a researcher in Human Genetics specializing in population genetics, cardiovascular disease risk, and molecular diagnosis. Her research is oriented toward translational and implementation studies of genetic testing in small hospital environments, with a focus on improving personalized medicine in the Azores archipelago, a ultraperipheral region. She has contributed to multidisciplinary projects combining genetic, clinical, and public health data.