

HERC2 GENE AS A PHENOTYPIC PREDICTOR IN BRAZILIAN POPULATION FOR FORENSIC PURPOSES

Cintia Fridman, Lígia Viotto

Departamento de Medicina Legal, Ética Médica e Medicina Social e do Trabalho, Faculdade de Medicina da Universidade de São Paulo (FMUSP)

The forensic genetics, particularly the Forensic DNA Phenotyping (FDP), became an important tool which may be used to help criminal investigations, and some human identifications, due to its ability to suggest, with good accuracy, the possible phenotypes for externally visible characteristics (EVCs) in samples of unknown origin. Many studies in homogenous populations have reported genes associated with phenotypic diversity of skin, eyes and hair color. Herein, we evaluated the association between one SNP present in the *HERC2* gene with skin, hair and eyes color in Brazilian population in order to point out the possible use of this marker in admixed populations.

The rs6497292 SNP was analyzed in 596 volunteers who answered a questionnaire where they self-reported about their skin, eye and hair colors. This SNP has specific patterns in different populations worldwide; the ancestral allele G is associated with characteristics of dark phenotype. As comparison, it was found in a frequency of 9% in European populations and 63% in an African one.

Samples were amplified by simple PCR reaction and lastly we used PCR-based Taqman assay (Applied Biosystems Inc.) for the allelic discrimination of rs6497292 (*HERC2*). Our analysis revealed that the variant allele (A) appeared in higher frequency, along with the polymorphic homozygous genotype (AA), 77.4% and 61.4% respectively. When compared to the ancestral allele (G) and its homozygous genotype (GG) frequencies, 22.5% and 6.5% respectively, no deviation from the Hardy-Weinberg equilibrium for this polymorphism was observed. Further, the homozygous genotype AA was associated with light traits, as white skin color ($p < 0.001$; OR 9.3; CI 3.6-23.8), and no-dark eyes as green eyes ($p < 0.001$; OR 20.1; CI 2.7-150.4) and light brown eyes ($p < 0.009$; OR 4.5; CI 1.3-15.3). There was no significant associations observed within hair color.

Thus, our study corroborated the association of the polymorphic genotype (AA) of the rs6497292 SNP with characteristics of light pigmentation, showing the importance of the FDP technique in the forensic investigation field in admixture populations, with further studies being essential to better understanding the process. This polymorphism is part of a major project that is analyzing a group of 38 SNPs associated with FDP in Brazilian population and in the end the statistic calculations will be done with all as a set. Supported: FAPESP, HC-LIM40