THE ANALYSIS OF MC1R POLYMORPHISMS CAN BE USED AS A TOOL TO PREDICT COMPLEX PHENOTYPIES, SUCH AS SKIN AND HAIR COLOR IN BRAZILIAN POPULATION?

Gonçalves FT[1]; Fridman C[1]., Department of Legal Medicine, Ethics and Occupational Health, Medical School, University of São Paulo

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Human pigmentation traits, including color variation in skin, eye, and hair, belong to the most visible and differentiating human traits. The genetic basis underlying variation in human pigmentation has been the subject of intensive research by investigators in a variety of life science communities, including forensic purposes, in which the use of these trials can help identifying missing person or guide some police investigations. The two types of melanin synthesized in well defined chemical reactions are the protective dark colored eumelanin and the sulphur containing light red-yellow pheomelanin. Eumelanin together with pheomelanin constitute the two main pigments of the skin and hair. The events leading to the melanogenesis are controlled by different genes, that we can highlight the melanocortin 1 receptor gene (MC1R), which encodes a protein in melanocytes responsible for melanin synthesis regulation. Polymorphisms in *MC1R*, which result in a loss of function of the receptor, are associated with increased pheomelanin production, which leads to lighter skin and hair color in European population. The aim of this study was evaluated the association between the polymorphisms in the only one exon of *MC1R* gene and hair and skin color in a sample of 401 individuals of admixed population from Brazil , intending to use the data in forensic genetics casework in several situations. No deviation in Hardy–Weinberg equilibrium was observed for all the polymorphisms analyzed. We found a strong association between the SNP *rs885479* (G>A) and yellow skin color (OR:148.14; CI: 6.95-315.2 to polymorphic homozygous genotype and OR: 14.0; IC: 2.68-72.99 to heterozygous genotype). In the analyses of hair color, we found associations between the red hair color and the heterozygous genotype to the SNP *rs1805007* (C>T - OR: 73.89; CI: 3.27-166.4) and *rs1110400* (T>C OR: 43.18; CI: 1.83-1020.8). Our data corroborate the findings of other studies in homogeneous populations, suggesting that the analysis of *MC1R* polymorphisms can be used as a tool to access some phenotypic traits and use the obtained in attempt to help forensic investigations. We are increasing our sample and additional analyzes are ongoing to confirm these results. These results are part of a major project which the aim is to study the correlation of several pigmentation genes and skin, hair and eye color.
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