SEQUENCE POLYMORPHISM IN THE HUMAN MELANOCORTIN 1 RECEPTOR GENE AS AN INDICATOR OF THE RED HAIR PHENOTYPE

Penny J Noake*, Eileen A Grimes, John Lee-Edghill, Andrew Urquhart
Forensic Science Service, Solihull, West Midlands, UK

In circumstances where there is no eyewitness evidence to a crime and a biological sample has been left at the scene, it is of benefit to establish some indication of the person's physical characteristics, such as the colour of their hair.

Although human pigmentation displays a wide variety of phenotypes and is thought to be under the control of several genetic loci, recent work has shown that in many cases the red hair phenotype can be associated with variants of a single gene, the melanocortin I receptor (*MC1r*). The action of alpha-melanocyte stimulating hormone on this receptor controls the switch between the syntheses of a red/yellow form of melanin (phaeomelanin) to the black/brown (eumelanin) photoprotective form. Red hair contains higher levels of phaeomelanin than hair of other colours.

A solid-phase fluorescent minisequencing protocol for screening DNA samples for the presence of 12 point mutations in the human melanocortin 1 receptor gene (*MC1r*), eight of which are associated with the red hair phenotype, has been validated for application in forensic investigations. Red hair displays a recessive mode of inheritance. A minisequencing profile, which shows homozygosity for one of these mutations or the presence of two different mutations would strongly indicate that the sample donor is red haired. The absence of any red hair causing mutations would indicate that the sample donor is not red haired.

A database of *MC1r* genotypes has been compiled. These data show that in addition to the test being used to give an indication of the likelihood of the profile if the DNA originated from an individual with red hair, *MC1r* results are indicative of the donor's ethnicity. The results may therefore be used to complement the ethnic inference service currently offered by the FSS, which is based on the results from 10 short tandem repeat (STR) loci.

*Corresponding author.