Abstract 28

CONFLICTING INTERPRETATION OF A Y-STR LOCUS FAMILY, DYF371; THE RESULT OF PCR CONDITIONS?

Allison Buchanan, Gabrielle Lehmicke and Tara Luckau
Human Origins Genotyping Laboratory, Arizona Research Laboratories, The University of Arizona, Tucson. AZ 85721

When genotyping short tandem repeats (STRs) of the non-recombinant Y-chromosome, consistent nomenclature and interpretation is essential, whether used for forensic, genealogical, or phylogenetic purposes. At the University of Arizona, the Human Origins Genotyping Laboratory (HOGL) offers Y-chromosome STR testing to the public for genealogical reconstruction through Family Tree DNA (www.familytreeDNA.com). In 2006, we expanded our testing resolution by adding 30 additional Y-chromosome loci. We included the locus DYS425 because it had already been established for genealogical reconstruction. This locus is complicated to analyze because it is one of at least four duplicated regions that comprise the DYF371 locus complex. We were prompted to scrutinize DYS425 when numerous null and low intensity duplication signatures were observed. To investigate this anomaly, a series of optimization reactions were run for the locus, varying both the annealing temperature and $MgCl_2$ concentration. When these different conditions were run on the same set of samples, multiple conflicting genotypes were observed. Our findings suggest that the low intensity duplication signature initially observed is likely due to mispriming of other DYF371 copies on the Y-chromosome. This is of particular concern because these genotypes have been reported in the academic literature, and our results demonstrate that a single sample can produce multiple genotypes depending on the PCR conditions. In an effort to assign consistent genotypes for DYS425, we have incorporated measures to increase the confidence in our calls. The marker DYS425 illustrates the importance of careful and consistent interpretation of results when working with STRs.