SNP ANALYSIS OF THE Y CHROMOSOME

<u>Kerri Dugan</u>¹, Elizabeth Olivastro¹, Peter Underhill² and Bruce Budowle³ ¹Counterterrorism and Forensic Science Research Unit, FBI Academy, Quantico, VA ²Stanford University, Department of Genetics, Stanford, CA ³Forensic Analysis Branch, Federal Bureau of Investigation, Washington D.C.

Analysis of single nucleotide polymorphism (SNP) markers on the Y chromosome could provide valuable information in some cases of paternity, missing persons, mass disasters, and violent crimes. Since more than 250 SNP sites on the Y chromosome have been discovered, it is important to identify those markers that will be useful in forensic analyses. Because of the population sub-structuring associated with genetic markers on the non-recombinant portion of the Y chromosome, the Y SNP markers can be evaluated using phylogenetic analyses.

We have examined the phylogenetic tree for SNP sites that potentially could be useful in forensic analyses. The approach that has been used in this study is to 1) identify the Y haplogroups that predominantly comprise relevant, major U.S. population groups and 2) select markers that facilitate differentiation between individuals within haplogroups. The markers lie within haplogroup defining branches of the tree rather than at the base branch points. A preliminary set of 20 Y SNP markers has been selected to provide discrimination of individuals within each forensically relevant haplogroup.

Multiplex primer extension assays for evaluation of these markers are being developed. Once a working assay is available, population studies will be carried out to resolve which candidate Y SNP markers are informative in a forensic context. This approach will allow a critical study of Y SNP markers and should provide a foundation on which to build a forensic Y SNP detection assay.