

Appendix C: Excerpt from “The Identification of Unknown Undocumented Border Crossers (UBCs): Recommendations for the Improvement of Current Methods” [41]

(PAGES 9 THROUGH 10)

Why MtDNA Typing is not enough

Mitochondrial DNA alone has very low discriminating power because many individuals within a population share the same mtDNA type. The probability that two samples would match purely by chance is proportional to the frequency of the type observed in the population. African populations are the most genetically diverse populations in the world. MtDNA HVSI sequence variation observed in 2,847 African individuals from 50 populations...indicates only ~70% discrimination capacity.... Thus, even for the most diverse populations in the world, on average there is a 30% probability (one minus the discrimination capacity) that two unrelated individuals will match their HVSI sequence by chance.

The sharing of mtDNA types is even higher in Native American populations where mtDNA diversity is low compared to African, Asian, or European populations. Prior to the Spanish conquest, the Americas were inhabited by populations whose ancestors crossed the Bering Land Bridge from Asia somewhere between 20,000-10,000 years ago. Because all Native American populations were established by relatively few individuals, many Native Americans share the same mtDNA type. Within the past 500 years, a significant number of immigrants from Europe and Africa have assimilated into populations living in the Americas, bringing new mtDNA into populations and increasing genetic diversity. The degree of European or African admixture varies greatly depending on the population. Populations living in urban environments (e.g., Mexico City, Guatemala City) generally have a larger degree of European admixture (22-86%) than indigenous populations living in the central or western regions. Notably, the degree of admixture is not the same for males and females. During the recent European and African colonizations of the Americas, more males than females have come to regions in Latin America. Thus, mtDNA is inherited only from the mother shows lower rates of admixture (and lower diversity) than nuclear DNA....

Low mtDNA diversity in a population means that there is a very high probability that two individuals from this population share the same mtDNA type even if they are not from the same family. For example, in three Hispanic populations in the southwestern US, Mexico, and Costa Rica, the probability that two samples will match by chance ranges from 21.4% (cosmopolitan Mexico City and Monterrey, Mexico) to 67.5% (Central Valley, Costa Rica).... Therefore, to avoid “false positives” when using mtDNA, some other piece(s) of information must also be considered. The low discriminating power inherent in the mtDNA region can be overcome by adding a STR profile. The combination of mtDNA and STRs has the potential to accurately identify nearly all unknown individuals.

Nuclear STRs (CODIS Markers)

The vast majority of forensic and missing persons cases that utilize DNA technology use the 13 STR CODIS markers because they provide extremely high discriminating power (i.e., low false positive rate). The 13 CODIS markers have a high power because there is a high variability at

each of the 13 unlinked (i.e., independent) STRs. The probability that one individual matches another individual in the population by chance is a function of the cumulative multiplied frequencies of alleles at each of the 13 CODIS markers. This typically results in exclusion probabilities that are less than one in a billion....

Because of the high copy number... extracting mtDNA from bone is relatively easy compared to nDNA. Nuclear DNA from bone is often highly degraded and many times fewer than 13 STRs work. To address this limitation, John Butler and colleagues at the National Institute of Science and Technology (NIST) developed mini-STRs that permit the successful amplification of all 13 CODIS STRs even when DNA samples are highly degraded.