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Title: Fine-scale genetic structure of the Y chromosome in a French-Canadian population and accurate estimation of Y-haplotype frequencies in forensic cases

Abstract:

In forensic DNA mixtures analysis, minor contributors to a stain can be hard to detect and quantify. In cases of mixtures involving a female major and a male minor contributor (e.g. on intimate swabs taken after sexual assaults), the analysis of Y chromosome STR typing can help to separate these contributors. According to the SWGDAM guidelines, the biggest challenge in forensic DNA interpretation is to use an appropriate reference database to estimate frequencies in the population of interest. The interpretation of DNA evidence is driven by two major premises: 1) reference samples used to calculate match probabilities are representative of the populations of interest and 2) the genetic diversity in the population is stable over the spatiotemporal scale at which a reference sample is applied. This may be particularly problematic with Y-STR haplotypes, as these markers are very sensitive to founder effects and to random genetic drift. Our objectives are 1) to test above assumptions using the French-Canadian population from Québec. A model combining genealogical and molecular data was used to extend the Y-STR population coverage achieved with typically small reference samples (hundreds of individuals). Thus, we were able to impute up to 300,593 Y-STR profiles to men who lived between 1608 (i.e foundation of the population) and 1960 from only 429 profiles of living men, accounting for mutation rate and genealogical errors. We focused on four regions best covered by this extended data set. In these regions a Y-STR profile could be imputed to >15% of the male population who lived between 1936 and 1960. Overall, our results show that using frequencies for the global population can strongly bias the calculation of a match probability at a local scale. Moreover, comparisons of samples with large international databases could make things even worse. This study illustrates the necessity to develop alternative approaches to evaluate Y-STR match probabilities.

Keywords: Forensic interpretation, genealogy, match probability, population genetics, short tandem repeat, Y chromosome