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**Titre**

Y-chromosome and mtDNA diversity in a French-Canadian population and lessons for forensic genetics

**Abstract**

Knowledge of the dynamic of sex-linked markers (on the Y chromosome and mitochondrial DNA [mtDNA]) is necessary for multiple areas of genetics including forensics. However, fine-scale estimation of frequencies, i.e. relevant to a specific population of interest, remains a challenge, as these markers are sensitive to genetic drift and founder effects. This limitation impacts on the interpretation of DNA evidence. Typically, one assumes that 1) reference samples used to calculate match probabilities are representative of the populations of interest and 2) the genetic diversity is stable over time and space, but these assumptions may not be true. Our objective is to test this empirically using the French-Canadian population from Québec.

Models combining genealogical and molecular data can overcome limitations associated to limited samples in order to obtain a reliable estimation of frequencies, making it possible to test above assumptions underlying DNA interpretation. Those models allow to obtain a much larger coverage of the population from a small genetic sample. By linking long-term genealogical data from the French-Canadian population of Québec to Y chromosome data from 429 men connected to this genealogy, we were able to impute a haplotype for up to 400,000 men who lived between 1608 (i.e. the foundation of the population) and 1960. Likewise, we imputed a mtDNA haplotype to 2 million individuals from 875 genotyped individuals. We then determined how haplotype frequencies have changed through time and space in the population. Analysis of mtDNA showed that haplotype frequencies were relatively stable through time, but that they weren't distributed homogeneously on the territory. Differences between sub-populations were sometime more important when looking at a much finer scale. Results from ongoing spatio-temporal analysis of Y haplotype frequencies will also be presented. Those results are determining for forensic genetics because they challenge the application of haplotype frequencies from national or international databases to specific caseworks.

**Key Words:**

Population genetics, forensic DNA, Y-STR, mitochondrial DNA, genealogy