Abstract

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Title: Forensic DNA Profiling of a Population Sample from Upper (South) Egypt

The work presented in this thesis was carried out to assess the genetic diversity and establish a forensic population database for a southern population sample inhabiting Upper Egypt. The Egyptian population encompasses several ethnic groups that need to be genetically characterised and evaluated for possible substructure before subsequent forensic use. Upper Egyptians are one of the major ethnic groups, exhibiting cultural and linguistic differences with the Northern population inhabiting Lower Egypt.

A sample of 265 unrelated individuals from five of Upper Egypt’s governorates was collected with informed consent, and profiled using fifteen autosomal STR loci contained in the AmpF®STR Identifiler™ PCR Amplification Kit. Statistical analyses indicated neither departure from expectations of Hardy-Weinberg equilibrium in most of the loci nor dependence of alleles between loci, allowing multiple locus profile frequency estimation using the product rule. All loci were polymorphic; the most discriminating is D18S51 while the least is TPOX. The combined power of exclusion was 0.9999986824 and the combined match probability was $1.93 \times 10^{-18}$.

Additionally, seventeen Y-STR loci included in the AmpF®STR Yfiler™ PCR Amplification Kit were typed in 208 males from the same population. Of 204 observed haplotypes, 200 were unique (96.6%) and four were found twice each. The 17-locus discriminating power was 0.9998. DYS385a/b and DYS458 showed the highest diversities with a high frequency of microvariant and new alleles (22% of the sample) in DYS458 locus. Other loci revealed duplicated, novel and null alleles such as DYS437, DYS635 and DYS448 respectively.

The generated data on Upper Egyptians from both STR systems were compared with data on other global, regional and local populations. Locally, some genetic differentiation was observed with other ethnic groups including the Northern population, suggesting likely population substructure among Egyptians. Global and regional comparisons demonstrated a regional genetic continuity among populations of the Nile valley, Middle East and Arabian Peninsula.