



B8 Heteroplasmy Pattern in D-Loop Region of Mitochondrial DNA

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After attending this presentation, attendees will learn about the heteroplasmy pattern in D-loop region of mitochondrial DNA.

This presentation will impact the forensic community by demonstrating how heteroplasmy pattern in the D-loop would help when interpreting the sequencing results for the mitochondrial DNA.

Mitochondrial DNA (mtDNA) is polymorphic in the D-loop region. This polymorphism is used in various areas, among which individual identification in forensics and confirmation of maternal lineage are the common usage. The mtDNA is small and circular, and exists as a lot of copy number even within a cell. With these characteristics mtDNA has become a valuable tool in individual identification, especially when the conventional typing for the autosomal STRs are unavailable such as in old degraded samples, bone and nail.

Several may be obstacles when using mtDNA. Heteroplasmy is one of those. There may exist different types of mtDNA within a cell or even within a mitochondrion. When someone performs mtDNA sequencing, heteroplasmy must be considered. It may be difficult to tell case of different origin from case of same origin showing heteroplasmy. Conversely the insight into the heteroplasmy may increase the discrimination power of mtDNA typing or may give some idea for how the polymorphism of mtDNA occurs.

Several have reported about cases of heteroplasmy, but these do not seem to be enough. We have screened 433 maternally unrelated Koreans using DGGE and have found 82 cases of heteroplasmy in HV1. We have sequenced all the samples and decided how heteroplasmy exists. The pattern of heteroplasmy and several related results including heteroplasmy pattern among different organs within an individual or age relationship will be presented.

Heteroplasmy, D-Loop, Mitochondrial DNA