MITOCHONDRIAL DNA TESTING IN SWITZERLAND
30 MONTHS OF EXPERIENCE WITH FORENSIC CASEWORK

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Since 1998 forensic mitochondrial DNA analysis has been performed in our laboratory in 58 cases including homicide, hold-up and burglary, rape, aggression, and identification of missing person. These cases have represented about 23% of our casework.

338 evidence samples (171 hair shafts, 80 bloodstains, 31 saliva stains, 24 sweat stain, 9 bones, 8 nails, 8 muscles, 2 stools, 1 tooth, 1 urine, 1 semen stain, and 2 fixed tissues) and 112 reference samples were extracted and sequenced for HVI and HVII mtDNA control regions. Despite the fact that about 80% of the evidence samples yielded a negative result for nuclear DNA with the human dot quantitation system, the success rate of the HVI and HVII mtDNA typisation was 79% using a nested PCR amplification followed by a fluorescent based capillary nested sequencing method.

The results of the reference samples were exclusion in 57%, inclusion in 40%, and inconclusive in 3% of the cases. Considering that in some cases, different mtDNA haplotypes corresponding to different samples were observed, a total of 123 exclusions was obtained. 91% of the excluded sequences displayed 4 to 15 nucleotide differences in respect to the evidence sample sequences.

Concerning the included reference samples, we reported the frequency using the counting method (that is the number of times the sequence was observed in a Caucasian database (N=715): 73% of the sequences were never observed while the other sequences had frequencies between 1/715 and 33/715. Point mutation heteroplasmy were observed in 15% of the casework and in 4% of the reference samples. Furthermore, length heteroplasmy in HVI and HVII was found in 9% and 17% of the casework and in 8% and 9% of the reference samples, respectively.

In addition, the relevant problem of heteroplasmy is illustrated with some casework in which different biological materials were analyzed.