Different Distribution of Mitochondrial DNA Length Heteroplasmy across Human Blood, Tissues and Hairs

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Length heteroplasmy within the homopolymeric C-stretchs of the mitochondrial HV1 and HV2 regions is one of the most often observed neutral polymorphisms in normal populations, and accordingly it has been used to investigate the segregation of mtDNA not only in the germ cell lineage but also in somatic tissues. In this context, we analyzed the HV1 and HV2 length heteroplasmy using fluorescence-labeled PCR primers in postmortem samples collected from 25 individuals ranging in age from 5 to 73 years. A blood, a brain, a heart, a liver, a skeletal muscle and five hair shaft samples were collected during autopsies of the each of the 25 individuals. Most heteroplasmic peak patterns across blood and various tissue samples in an individual seemed somewhat different, but they showed no significant shift in the major length variant in a mixture. On the other hand, considerable variation in the mtDNA profiles was observed in 5 of 7 and 13 of 16 heteroplasmic individuals' hair samples for HV1 and HV2 regions, respectively. More interestingly, *de novo* generation of length heteroplasmy was observed in 1 and 3 homoplasmic individuals' hair samples in HV1 and HV2 regions, respectively. In addition, an individual who was heteroplasmic for all tissue and blood samples in HV2 region showed the extinction of heteroplasmy in 1 of 5 hair samples. Specialized forensic laboratories currently use mtDNA sequence analysis for identification of telogen hairs and human remains. Prior to the routine use of mtDNA typing for personal identification, any different mtDNA profiles across each tissue and hair from individuals in the normal population should be considered. The mtDNA segregation during development and extremely high mutation frequency in hairs will be discussed.