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DISSERTATION SUMMARY

Analysis of Y-chromosomal microsatellites on archaeological and modern samples

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The special properties of the Y chromosome, which include haploidy, paternal inheritance and absence of recombination through most of its length (95%), make this chromosome a powerful tool for tracing and comparing paternal lineages of human populations.

Among its markers microsatellites or short tandem repeat loci (STR) are marked by tandemly repeated sequences of 2-5 bp. The specific number of repeats (or allele) is the same in men who belong to the same paternal lineage and usually remains unchanged from generation to generation but changes sometimes occur and the number of repeats may increase or decrease by one unit (Kimura and Ohta 1978).

Y-chromosomal STRs show sufficient variability among individuals in a population and high degree of geographical differentiation (Jobling and Tyler-Smith 1995). Thus, their polymorphic character makes them especially suited for forensic, genealogical and population genetic studies (Schultes et al. 1999).

According to these features 4 microsatellite markers were selected for reconstruction of paternal lineages of ancient Hungarians, originated from 10th century. These markers have simple structures, they are highly variable and short in length which qualifies them as an investigate tool in the study of ancient DNA (aDNA). Due to degradation and low amounts of aDNA the first attempts were unsuccessful. Therefore several methods have been introduced to enhance the efficiency of our analyses. We tried to repair the highly fragmented and heavily damaged aDNA with different enzymes (Bernardo et al. 2002) and to increase the amount of aDNA with random primers (Zhang et al. 1992). These trials supported the notion that the bones did not preserve any amplifiable Y-chromosomal DNA or our detection system does not have enough sensitivity.

Then the examinations were continued on a fully automated, highly accurate and very sensitive system (Automated Laser Fluorescent DNA Sequencer). Its function relies on the detection of laser-induced fluorescence of the carbocyanine dye (Cy5) with which the DNA fragments are labelled. Using appropriate internal molecular size markers it can detect as small as one basepair difference. So it appeared to be a good system to investigate Y-chromosomal length polymorphisms.

This method proved to be successful. In the case of 3 ancient bone samples Y-chromosomal STR markers have been revealed. Among the 4 selected markers we could examine 2 STRs, which yield amplification products in shorter ranges (108-140 bp and 147-179 bp). The markers, which characterize the human remains, occur mainly in the Asian continent in our Y chromosome STR database, but they can be found in European populations also.

The Y-chromosomal DNA analysis was extended to modern Hungarians and Szeklers too. We have data from 137 Hungarian and 115 Szekler men.

Comparing the allele frequency distributions of these two modern populations for 4 STR markers it was established that there is no significant genetic difference between the modern Hungarian and Szekler men and they contain Asian genetic elements beside the European ones. The statistical evaluation of the results confirmed the small genetic distances between Hungarians, Szeklers and Albanian, Greek, Romanian, Bulgarian, Bulgarian-Turkish, Turkish, Estonian, Iranian, Syrian, German, Austrian and Italian populations, while both modern ethnic groups differ significantly from Portuguese, Spanish, British, Basque, Finnish, Lithuanian and Japanese populations.

References

Di Bernardo G, Del Gaudio S, Cammarota M, Galderisi U, Cascino A, Cipollaro M (2002) Enzymatic repair of selected cross-linked homoduplex molecules enhances nuclear genes rescue from Pompeii and Herculaneum remains. Nucleic Acids Res 30:e16.

Jobling MA, Tyler-Smith C (1995) Fathers and sons: the Y chromosome and human evolution. Trends Genet 11:449-456.

Kimura M, Ohta T (1978) Stepwise mutation model and distribution of allelic frequencies in a finite population. Proc Natl Acad Sci USA 75:2868-2872.

Schultes T, Hummel S, Herrmann B (1999) Amplification of Y-chromosomal STRs from ancient skeletal material. Hum Genet 104:164-166.

Zhang L, Cui X, Schmitt K, Hubert R, Navidi W, Arnheim N (1992) Whole genome amplification from a single cell: Implications for genetic analysis. Proc Natl Acad Sci USA 89:5847-5851.

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