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Y-chromosomal microsatellites in the Finns

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We have evaluated 16 Y-chromosomal microsatellites for forensic purposes. The loci were amplified in two multiplex PCRs, a 9-plex (http://www.ystr.charite.de./index_gr.html) and a 10-plex (Ch.M. Rutberg and J.M. Butler, pers. comm.), with three common Y microsatellite loci. Of these 16 loci 9 belong to the widely used Y-chromosomal minimal haplotype (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a and DYS385b), and the additional 7 loci were DYS435, DYS436, DYS437, DYS438, DYS439, Y GATA A7.1 and Y GATA H4. The PCR products were subsequently analysed using capillary electrophoresis.

For population studies and PCR validation purposes, DNA samples from 200 Finnish males, 20 female samples and various male animal samples were analysed with both multiplex PCRs. To compare the sensitivity of the 9-plex and the 10-plex reactions, we performed the multiplex PCRs with samples from a dilution series of a known amount of DNA.

Our results show that the 9-plex PCR is more sensitive than the 10-plex PCR. In addition, the 10-plex PCR showed constant, unspecific peaks which were detected in male and in female samples.

When using the set of minimal haplotype markers, our 200 Finnish male DNA samples fell into 85 different haplotypes, 64 (75%) of them occurring only once in our data. When we were using data from 10-plex, we found 72 different haplotypes, 52 (72%) occurring only once. Combining the data, together 120 different haplotypes were found, and from these 99 (82.5%) occurred once. In the 9-plex PCR, 26.5% of the Finns had the most common haplotype. Similarly in the 10-plex PCR also 26.5% of Finns shared the most common haplotype. However, when the two marker sets were combined, the most common haplotype was shared only in 13% of the samples.

The power of discrimination values of separate microsatellite loci varied in the 9-plex PCR between 0.448 (DYS392) and 0.706 (DYS385a and b together) and in the additional seven loci between 0.020 (DYS436) and 0.523 (Y GATA H4).

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We further divided our samples according to the geographical origin from where the sample donors had their family roots. This resulted in six geographically defined sub-populations (Häme, Karjala, Pohjanmaa, Satakunta, Savo and Varsinais-Suomi), each containing 30 individuals. The most common haplotype in Finns (0.130) was found in four of these sub-populations (10/30 in Savo, 7/30 in Pohjanmaa, 3/30 in Karjala and 2/30 in Häme), but in the sub-populations located in south-western Finland (Satakunta and Varsinais-Suomi) this haplotype was not observed, and no other haplotype was found to be dominating. Altogether, the power of discrimination values of individual loci were the lowest among individuals from Savo (DYS435 being the only exception).

In conclusion, the 9-plex PCR appears more sensitive and produces no unspecific or female-derived peaks, whereas in the 10-plex constant, unspecific peaks were observed in male and in female DNA samples. For forensic purposes the 9-plex PCR is more discriminating among the Finns than the 10-plex PCR. This is mostly due to the fact that in the 10-plex, two loci (DYS435, DYS436) had very little variation, with only two or three alleles and 98–99% of the samples shared the one most common allele. However, by combining these two multiplex PCRs a high resolution Y-chromosomal DNA profile can be achieved for criminal investigations and for establishing family relationships. From a population genetics point of view, our observations are in accordance with earlier studies that show decreased variation among individuals from Eastern Finland. Our findings also support the theory that only a small number of individuals have inhabited the Savo area in historical times.