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Population study and validation of the Y–STR pentaplex for use in forensic case work

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Abstract

With the aim of using Y-chromosomal polymorphic markers in Danish crime cases and deficiency paternity cases, a validation study of a Y–STR pentaplex was performed. The Y–STR pentaplex included the systems DYS19, DYS389I/II, DYS390 and DYS393. In order to obtain frequency data, a population study of 200 male Danes and 91 male Eskimoes was performed. In order to validate the Y–STR pentaplex for use in crime cases, various ratios of mixtures of DNA from two individuals (male and female or male and male) were investigated. Mixed stains from Danish crime cases were used to compare the sensitivity of the Y–STR pentaplex with that of autosomal STR analysis.

Keywords: Y chromosome; STR; Population database; Forensics

1. Introduction

Analysis of short tandem repeats (STRs) located on the Y chromosome has been shown to be a powerful tool in rape cases as well as in deficiency paternity cases [1]. Here, we describe the validation of a Y–STR pentaplex [2] for use in forensic case work in Danish crime cases and deficiency paternity cases. The Y–STR pentaplex included the systems DYS19, DYS389I/II, DYS390 and DYS393.

2. Materials and methods

Amplification conditions of the Y–STR pentaplex were as described by Gusmao et al. [2]. For the population study, approximately 0.5 ng of DNA purified by the Chelex method

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was used. A total of 200 male Danes and 91 male Eskimoes randomly selected from Danish paternity cases were analysed.

For stain analysis, a total of 21 mixed stains (blood or semen), each containing DNA from a male and a female individual and six blood stains, each containing DNA from two male individuals were analysed. The results were compared to the results of the autosomal STR analyses. For autosomal analyses, either the AmpFISTR® SGM Plus™ (Applied Biosystems) or a quadruplex STR analysis consisting of the systems HumTH01, HumvWA, HumF13A01 and HumFES [3] were used.

Furthermore, 15 blood stains each containing DNA from a single male, four blood stains each containing DNA from a single female and six stains of semen were analysed. All results were compared to results from the reference samples.

3. Results

Analysis of 200 male Danes and 91 male Eskimoes revealed a total of 113 different haplotypes. Of these, only 12 haplotypes were present in both populations. The gene diversity of the haplotype distribution was 0.96 in Danes and 0.94 in Eskimoes.

Among 93 Danish father–son pairs, two inconsistencies were found in DYS389I/II, while a single inconsistency was found in DYS19. The father–son pairs had previously been investigated for at least five autosomal VNTR-systems, and in the VNTR-systems, the paternity indices exceeded 10,000. No inconsistency was found in the VNTR-systems.

Investigations on the sensitivity of the Y–STR pentaplex showed that the pentaplex produced reliable results using as little as 0.05–0.1 ng of template DNA. Stutter peaks were mainly observed in the systems DYS390, DYS393 and DYS389II. In all systems, the peak heights of the stutter peaks were below 15% of the heights of the alleles.

Analyses of mixtures each containing DNA from a female and a male showed that the Y–STR pentaplex worked satisfactorily in all five systems if the amount of female DNA was below 5 ng and if the ratio of female DNA to male DNA was less than 1000:1. If the amount of female DNA exceeded 5 ng, a product in DYS393 arising from female DNA was seen. However, it has been reported [4] that replacing one of the DYS393 primers solves this problem. If the ratio of female DNA to male DNA exceeded 1000:1, artifact peaks in DYS389II were observed in some cases.

Investigations of mixtures containing DNA from two men revealed that a full Y–STR profile from both men could be obtained if the ratio of male 1/male 2 did not exceed 10:1.

In total, 21 stains containing mixtures of male and female DNA and six stains containing mixtures of DNA from two males were investigated. The compositions of the stains were based on the autosomal STR analyses. A comparison of the Y–STR results with those of the autosomal STR analyses was performed. In three investigated stains, only a partial profile of the man was obtained in the autosomal STR analysis, while a full profile was obtained in the Y–STR pentaplex analysis. In two stains, the autosomal STR analysis showed a mixed profile of the woman and one of the men in the case, while the Y–STR analysis showed a full profile of one man and a partial profile of another man. The profiles of the two men were consistent with the profiles of the reference samples of the two men in the case. In a single stain, the Y–STR analysis gave

no profile even though the autosomal STR analysis had shown a partial profile of the man in the case.

4. Discussion

The population study of male Danes and male Eskimoes showed that only 12 out of 113 haplotypes were present in both populations. This result shows the importance of obtaining population data of Y–STRs from all ethnic groups that are to be investigated in routine forensic case work.

The detection limit of the Y–STR pentaplex was shown to be 0.05–0.1 ng DNA. Compared to autosomal STR analysis, the Y–STR pentaplex seemed to be more sensitive in the analysis of mixed stains containing small amounts of male DNA.

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