International Congress Series 1239 (2003) 487–490

Y-STR typing in forensic analysis

N. von Wurmb-Schwark [a,*], S. Petermann [b], R. Wegener [b]

[a] *Institute of Legal Medicine, Christian-Albrechts-Universität Kiel, 24105 Kiel, Germany*
[b] *Institute of Legal Medicine, University of Rostock, 18055 Rostock, Germany*

Abstract

We reinvestigated 20 cases of rape/sexual maltreatment from the years 2000/2001 (i.e. 61 different samples) that had already been investigated by STR typing (using Profiler Plus and mentype kit). In 10 cases, male DNA has been detected in stains or vaginal swabs, samples from the other 10 cases did not show any traces of male DNA. Ten Y-specific loci were amplified in two multiplex PCRs, mpI: DYS393, DYS19, DYS392, DYS385; mpII: DYS390, DYS391, DYS389I and II. Even though amplification of Y-specific loci was not more sensitive and did not lead to any other conclusions than autosomal STR typing alone, in specific cases Y-STR determination gave important information, e.g. when the male suspect was unknown or when mixed stains from more than one male person had to be investigated.

Keywords: Y-STR; Forensic analysis; Multiplex; ABIPrism 310

1. Introduction

Y-chromosomal polymorphic markers have been extensively investigated in forensic medicine for male identification and paternity testing or in evolutionary studies [1–3]. These markers are inherited along a male lineage, are not subjected to recombination and are characterized by a moderate number of polymorphic loci. Y-STRs have a particular importance in cases where mixtures of male and female DNA have to be analyzed. These mixtures are frequently encountered in stains from cases of sexual assault and are often difficult to evaluate by autosomal STR analysis alone.

* Corresponding author.
E-mail address: nvonwurmb@rechtsmedinzin.uni-kiel.de (N. von Wurmb-Schwark).

0531-5131/03
PII: S0531-5131(02)00591-5

The aim of our study was to compare the results obtained by nuclear STR typing to those from Y-STR typing regarding:

- sensitivity of Y-STR typing,
- stability of DNA amplification (degraded DNA),
- power of discrimination.

2. Material and methods

Altogether, 61 samples were reinvestigated, including 12 buccal swabs, 30 vaginal swabs, 4 fingernail samples, and 15 possible semen or saliva stains from different surfaces (e.g. blankets, carpet, skin). The DNA from trace material and buccal swabs was extracted using the Invisorb Forensic kit (Invitek, Rödermark, Germany). Blood samples were extracted using the QuiampQuick Gene Kit (Qiagen) according to the manufacturer's instructions.

Amplification of Y-STRs was done in two multiplex PCRs according to Kayser et al. [4] and 10 ng total DNA or 10 μl of DNA solution were employed as template. A total of 10 pMol each of betaglobin primers were added to the multiplex PCRs to amplify a 164-bp fragment specific for autosomal DNA. A 3 μl of each amplified PCR product plus 21.5 μl formamide and 0.5 μl ROX500 standard was analyzed by capillary electrophoresis in denaturating polymer (POP4) on an AbiPrism310 DNA Sequencer (Applied Biosystem). Allele assignment was performed by comparison with self-established ladders and determination of fragment sizes.

3. Results and discussion

3.1. Proficiency of the multiplex PCR system

The loci investigated in this study showed different PCR efficiencies. Locus DYS392 was amplified with the lowest efficiency, followed by DYS19. Surprisingly, DYS385 as well as DYS389I and II with the longest fragments to be amplified often gave the best results. Sometimes they even were the only detectable loci indicating that the DNA was not degraded and that it should be possible to amplify all the other shorter loci. The proportion of the stutter peaks did not exceed 10%, which made it easy to recognize male DNA contents.

3.2. Detection of betaglobin specific DNA

Simultaneous amplification of a betaglobin specific 164 bp fragment in our multiplex PCRs did not interfere negatively with amplification of Y-specific loci concerning sensitivity or specificity. This provided a useful PCR control. A positive betaglobin signal and no Y-specific fragments showed that the PCR was working and that only female DNA was present in our sample (Fig. 1), ruling out a PCR failure due to inhibiting substances (even though male DNA is present).

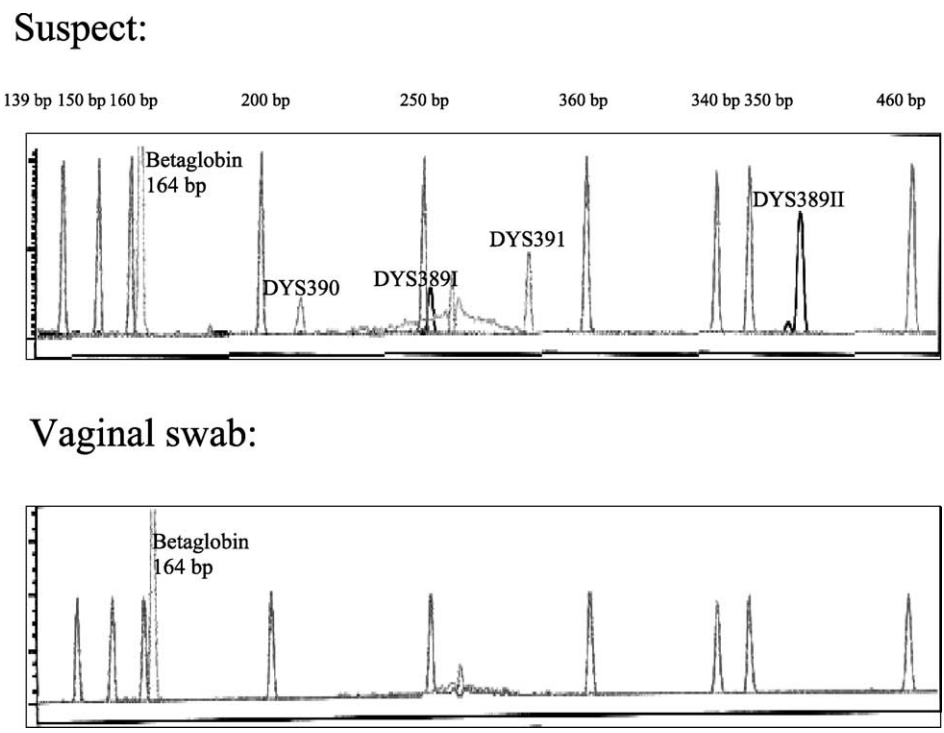

Fig. 1. Simultaneous amplification of autosomal betaglobin DNA and Y chromosomal loci. Electropherogram, AbiPrism 310. A 164-bp fragment of the betaglobin gene was co-amplified with Y-specific loci from mpII (DYS390, DYS391, DYS389I, II) without any modification to the PCR protocol. Shown is the genotype of a suspect using 10 ng template DNA (top) and the amplification pattern from a vaginal swab (bottom) which only shows a betaglobin fragment indicating that there is no male DNA in the sample.

3.3. Comparison to autosomal DNA analysis

Investigation of samples that were negative for male DNA by Profiler Plus and Mentype amplification did not yield any positive Y-specific signals. This indicates that in those cases, no false conclusions were drawn by autosomal STR typing. However, 9 out of 39 samples that were positive for male DNA using the commercial profiling kits did not show any Y-specific fragments. In three cases, a DNA mixture obtained from vaginal swabs was detected indicating at least two persons. Evaluating ProfilerPlus and Mentype PCRs, only two or three loci, respectively, could be determined as being loci from the suspect. Employing Y-STR analysis, we obtained three new haplotypes that were not found in the European Y-STR data collection, which currently contains around 7000 haplotypes from 51 population samples [5].

Thus, application of Y chromosomal STRs to forensic casework might be useful and more informative than autosomal STR typing (plus amelogenin) alone, especially if there are DNA mixtures from two or more male persons or if the male suspect is unknown and only mixed DNA (from victim and suspect) is available [6].

References

[1] R. Chakraborty, Paternity testing with genetic markers: are Y-linked genes more efficient than autosomal ones? Am. J. Med. Genet. 21 (1985) 297–305.
[2] M.A. Joblin, A. Pandya, C. Tyler-Smith, The Y chromosome in forensic analysis and paternity testing, Int. J. Legal Med. 110 (1997) 118–124.
[3] A.B. Spurdle, T. Jenkins, The Y chromosome as a tool for studying human evolution, Curr. Opin. Genet. Dev. 2 (1992) 487–491.
[4] M. Kayser, A. Cagli, D. Corach, et al., Evaluation of X chromosomal STRs: A multicenter study, Int. J. Legal Med. 110 (3) (1997) 125–133, 141–149.
[5] Q. Schiermeier, Letter to nature: Y chromosome analysis urged for sex crimes, Nature 413 (2001) 6.
[6] M. Prinz, K. Boll, H. Baum, B. Shaler, Multiplexing of Y-chromosome specific STRs and performance for mixed samples, For. Sci. Int. 85 (1997) 209–218.