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Y-STR typing in the identification of genetic profile of the semen

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Abstract. Since relatively few genital injuries are discovered at the medical examination, the detection of other elements such as the presence of semen is important to confirm the presumption of sexual assaults. For many years, cytology and the detection of acid phosphatase were considered to provide forensic evidence. In the past 2 years commercial Y-STR kits were developed to enable forensic practitioners to use these multiplex systems in male specific identification. DNA male profile was obtained from evidence samples, using Y-STR, when only autosomal STR profile from victims was detected. © 2006 Published by Elsevier B.V.

Keywords: Y-chromosome; Short tandem repeats; DNA typing; Y-STR; Forensic casework

1. Introduction

The Y-chromosome is specific to male and therefore useful in many sexual assault case situations where the perpetrator's DNA needs to be identified in the presence of the female victim's DNA. A number of short tandem repeat (STR) markers have been identified on the Y-chromosome and developed into multiplexed polymerase chain reaction (PCR) assays that can help differentiate between unrelated males [1].

The Genetics and Biology Forensic Laboratory of the National Institute of Legal Medicine (Oporto Delegation) has been asked to solve criminal cases, among other analysis, being the majority of them as sexual female assaults. For a variety of reasons, some forensic evidences provide only autosomal female profile.

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2. Material and methods

DNA was extracted from samples collected in sexual female cases using the organic phenol–chloroform–isoamylalcohol method, without a differential extraction [2]. The loci were co-amplified using the PowerPlex® 16 System (Promega) or the AmpFlSTR® Identifiler™ PCR Amplification Kit (Applied Biosystems) for the autosomal STR systems and, for the Y-STR systems, the PowerPlex® Y System (Promega) and, in some situations to increase the degree of informativeness, the AmpFℓSTR® Yfiler™ (Applied Biosystems). The amplified products were detected and separated by capillary electrophoresis on an ABI PRISM® 310 Genetic Analyzer (Applied Biosystems). Fragment sizes were determined automatically using the Genescan® Analysis Software v 3.7 and allele designations using Genotyper® Software v. 3.7 (Applied Biosystems) typed by comparison with an allelic ladder.

3. Results

In our laboratory we have used a strategy in sexual assaults, since the beginning of 2004, that always includes the Y-STR analysis provided with the multiplex kits previously mentioned, even when other results were negative, including the lack of the male autosomal STR profile.

A total of 128 cases, including different evidence samples (stains, swabs), were analyzed using this methodology. In 22 of these cases, a Y-STR profile was obtained. In the majority of them, that profile was complete, spermatozoa were detected by cytology and the acid phosphatases were negative (Table 1).

4. Discussion

We have used a non-differential extraction protocol, because many times a complete separation of male and female samples is difficult to achieve and to prevent the loss of sperm due to the multiple manipulations required of this process used to separate the sperm from the non-sperm DNA fractions.

The ability to obtain the Y-STR haplotype of the male donor in the presence of a vast excess of female DNA confirmed the potential utility of these systems in admixed male/female DNA cases. This occurs when a minor male component is present, even when the detection of acid phosphatase and cytological were negative.

Table 1
Results obtained for the 22 cases with Y-STRs and without male autosomal profile

No. of cases	Evidence samples	Cytological	Acid phosphatases	Complete Y-STRs profile
3	Vaginal swab	Positive	Positive	Yes
10	Stain	–	Negative	Yes
2	Vaginal swab	Positive	Negative	Yes
1	Vaginal swab	Positive	Negative	No
1	Oral swab	–	Negative	No
3	Vaginal swab	Negative	Negative	Yes
2	Stain	–	Positive	Yes

Y-STR have also demonstrated advantages over the use of autosomal STR estimating the number of perpetrators, and simplifying interpretation due to single allele profile per individual for most loci. However, it has limitations, because Y-STR loci do not assort independently, so the frequencies of alleles found at each locus cannot be combined using the product rule. Therefore, examination of a considerable number of these loci is necessary to provide sufficient discrimination of haplotypes among male lineages. Furthermore, the profiles among paternal relatives cannot be distinguished.

References

[1] J. Butler, et al., Chromosomal duplications along the Y-chromosome and their potential impact on Y-STR Interpretation, J. Forensic Sci. 50 (4) (2005).

[2] A. Hall, J. Balantyne, Novel Y-STR typing strategies reveal the genetic profile of the semen donor in extended interval post-coital cervicovaginal samples, Forensic Sci. Int. 136 (2003) 58–72.