EXPERIENCES WITH USING Y CHROMOSOME SPECIFIC STRS IN FORENSIC CASEWORK

Mechthild Prinz, Asako Ishii, Maribel Sansone, Howard Baum, Robert Shaler

Department of Forensic Biology, Office of Chief Medical Examiner, 520 First Avenue, New York City, NY

INTRODUCTION

Especially in rape cases, Y chromosome specific STRs have the advantage of enabling the investigator to target only the male DNA in a mixture of body fluids. There have been a number of reports about the application of Y-STRs in casework (e.g. 1, 2, 3), but all of these reports are from outside of the United States. In order to be able to perform Y-STR testing in the US, our laboratory conducted a two year validation according to TWGDAM guidelines for a multiplex consisting of three fluorescently labeled Y-STR primers on the ABD/PE 377 platform. The three primer pairs amplify four polymorphic Y chromosome STR loci: DYS19, DYS390, DYS389I and DYS389II (4, 5). The validation was completed in September of 1998 (manuscript in preparation). Since then, the new test has been used in more than 500 cases, totaling over 1000 samples.

CASEWORK APPLICATION

In one year of casework, 915 evidence samples from 587 different cases were tested using the Y STR multiplex. Additionally to that number 250 known blood samples were tested for comparison purposes. Y-STR testing is routinely performed on all cases which fall under one of the following scenarios:

A: After differential lysis the semen evidence is consistent with being a mixture of DNA of two individuals with the victim as one contributor.
In these cases Y STR results are used to eliminate the possibility of multiple semen donors.

B: After differential lysis the semen evidence shows a mixture with an indication of the possible presence of multiple semen donors.
In these cases Y STR results are used to determine the number of semen donors.

C: After differential lysis the semen evidence only shows the victim's type.
In these cases the swab remains fraction is tested using Y-STR's in order to show the presence of a minor component of male DNA. In these cases it is also helpful to repeat the extraction using a non-differential approach which doesn't involve the risk of losing sperm cells during the differential lysis wash steps.

D: The case involves non-sperm cell evidence as aspermic semen, saliva or blood mixed with female DNA. In these cases autosomal test systems very often only reveal the victim's DNA type. Y-STR testing is used to specifically detect the male component.

E: The case currently doesn't have a suspect.
If enough semen containing evidence for three extractions is present, a quick non-differential lysis and Y-STR screen is performed so that only the cases where enough male DNA could be detected can go on to the more time consuming differential lysis.

Mixtures of more than one semen donor:
Many of these samples tested could be shown to be mixtures of more than one semen donor. This number might still be an underestimation, since with the limited discrimination rate of the current four-locus haplotype it is possible for two semen donors to have the same Y-STR profile. Semen on 12% of the vaginal and anal swabs and 21% of the underwear stains contained DNA from more than one male individual. See Table 1) for details. The determination of the minimum number of semen donors is helpful in multiple rape cases because it can corroborate the victim's account of the crime. It is also helpful for the interpretation of mixtures seen using autosomal typing systems. Figure 1) shows an example for a mixture result where a single semen type was found on a swab taken from the inside of a condom and a mixture of two DNA's was found on the outside of

the condom. Since the victim was female, the second male type on the outside of the condom is attributed to sperm cells that were already present in the vaginal tract before the rape.

Autosomal testing reveals only the victim's DNA type:
In several cases, also from other jurisdictions, autosomal STR typing on the evidence did not show any alleles foreign to the victim, while the Y-STR test yielded a male DNA type. Of course in many cases even though the stains or swabs were positive for the presence of semen the Y-STR test will still be negative, because the swabs really do not contain any or only insufficient amounts of cellular male material. Table 2 shows the percentage of samples that gave positive or negative results. No results were obtained on 19% of the samples tested. The equivalent number for one year of casework is not available for the autosomal systems but the 19% compare favorably to the 41% negative or victim's type only results for 56 samples validation samples (manuscript in preparation).

One type of evidence where the male and female component of a mixture of body fluids cannot be separated are amylase positive stains or swabs. The Y-STR success rate for saliva evidence is not very high. Table 3 shows the results for seventeen samples. Half of the samples did not yield any DNA type at all, but for some samples the Y-multiplex was the only system that could detect the male component in the mixed samples.

CONCLUSIONS

The Y multiplex has been shown to be a valuable DNA typing tool. It is mostly being used as an additional test system supporting and clarifying the autosomal test results. In cases where the Y STR type remains the only DNA result, the statistical power is very limited and all paternal relatives are also included. On the other hand an exclusion is unambiguous and in many cases it is sufficient to discriminate between two defendants.

So far there have been two motions for Frye hearings connected to Y-STR testing but in both cases the motion was withdrawn. In other cases, Y chromosome specific results have been accepted without any questions. We do not consider Y-STR testing a novel technique since PCR and fluorescent STR technology are well established and have been accepted in New York State Supreme Courts multiple times. The statistical approach of counting haplotypes differs from autosomal STR testing, but here a parallel to mitochondrial DNA testing can be drawn. Efforts to increase the Y-STR haplotype population database are underway worldwide.

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Table 1) Mixtures of more than one male individual on semen evidence

sample type	total number tested with positive results	number of mixtures	% of all positive items
vaginal/anal swabs	330	40	12%
panties/underwear	170	37	21%
other items	240	20	8%

Table 2) Results for Y-STR testing in one year of casework

number of samples tested	915
no result	175 (19%)
full or partial result	740 (81%)

Table 3) PCR typing results for 18 amylase positive swabs and stains

number of samples	Y STR result	autosomal result
7 vaginal swabs 1 dried secretions swab 2 underwear stains	no alleles detected	not tested
4 vaginal swabs	partial profile	victim's type only
1 vaginal swab	full profile	not tested
2 vaginal swabs	full profile	victim's type only
1 body swab	full profile	full clean profile

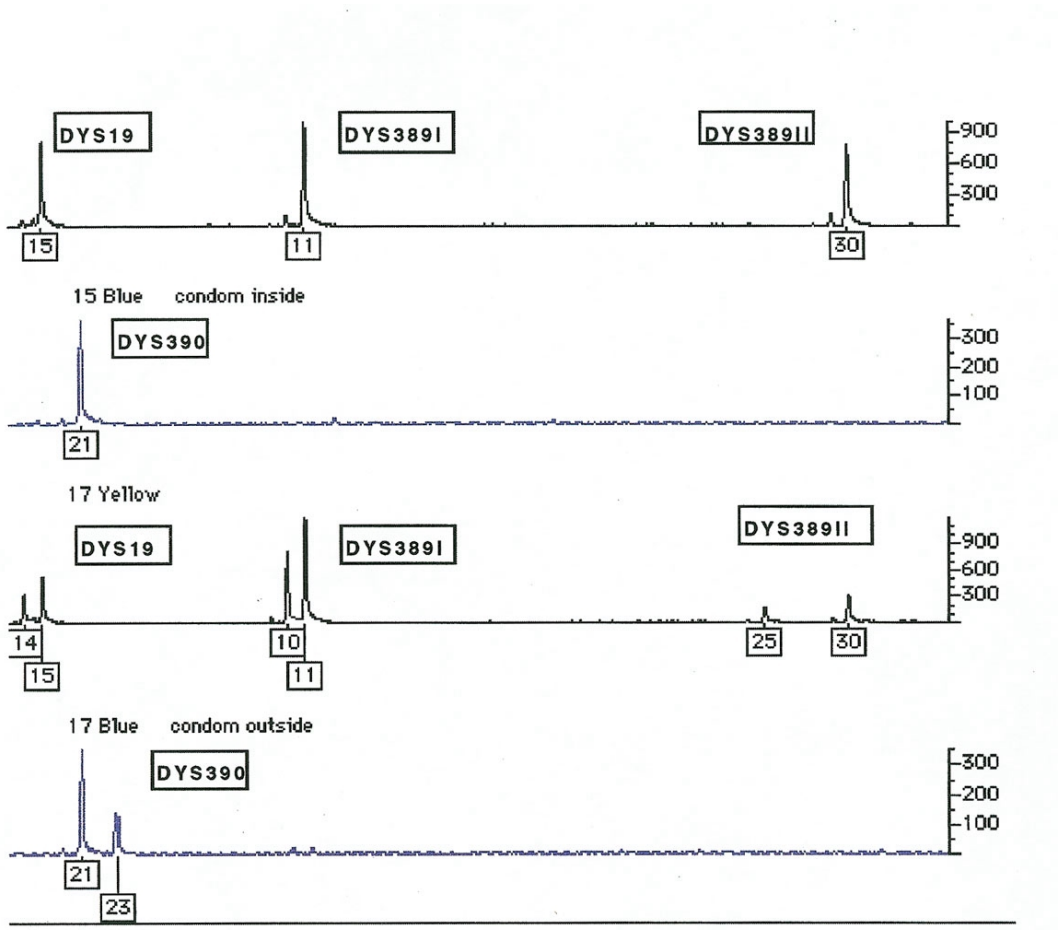

Figure 1 Two electropherograms with Y Multiplex typing results for two swabs taken from a condom are shown. DYS19, 389I and 389II are labeled in yellow, DYS390 is labeled in blue. The haplotype on the swab taken from the inside is consistent with coming from a single semen donor. The swab taken from the outside shows the same haplotype as the major component. One additional allele is seen at each locus, so this DNA must have come from at least two semen donors.