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Y chromosome in forensic casework and paternity testing

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Abstract. The aim of the study is to demonstrate the power obtained by combining autosomal and Y chromosome STRs in certain case scenarios. Although autosomal STRs are in use and highly informative, Y STRs are extremely useful and provide additional information that could not be provided by autosomal STRs alone.

Keywords: Y STRs; Forensic casework; Paternity testing

1. Introduction

The determination of the gender of the source of forensic biological materials can be of investigative and evidentiary value [5]. Recently, Y STR loci have gained an increased attention from both forensic and paternity testing groups [3]. Y STRs loci typing is particularly useful for typing the male DNA component in male/female mixture evidence samples [4]. Typing closely linked loci that do not undergo recombination is of particular interest in deficiency paternity testing and in determining paternal lineages [2,6]. This study reports four example cases where Y STRs combined with autosomal STRs helped to reach a conclusion of potential source with greater confidence.

2. Material and methods

DNA was extracted using the standard organic procedure. Autosomal STRs were typed using either the AmpFLSTR® Profiler Plus™ and AmpFLSTR® Cofiler™ Kits, or the AmpFLSTR® Identifiler™ Kit (Applied Biosystems). Profiling of Y STRs was conducted

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using Y-Plex™ 6 STR kit and Y-Plex™ 5 STR kit (Reliagene, USA) according to the manufacturers' protocols.

2.1. Case 1: burned human remains

A fire and explosion took place in a firework games warehouse. Nine bodies were recovered of which relatives could identify four of them. Of the five remaining severely charred bodies, two examples are presented. In the first example, a corpse and five body fragments were recovered from under the fire debris 40 days after the incident. Autosomal STRs profiling showed a match among the recovered fragments and corpse. A man who reported his brother missing could not be excluded as a relative. Since only the brother was available for testing, Y STRs were typed and found to match the profile of the remains. The second example consisted of a partially burned body. A man whose full brother used to work in the same place visually identified one of the corpses to resemble his brother. Comparison at 15 autosomal STRs showed no shared alleles at seven loci. While the evidentiary value for sibship was not strong, the man could not be excluded as a possible

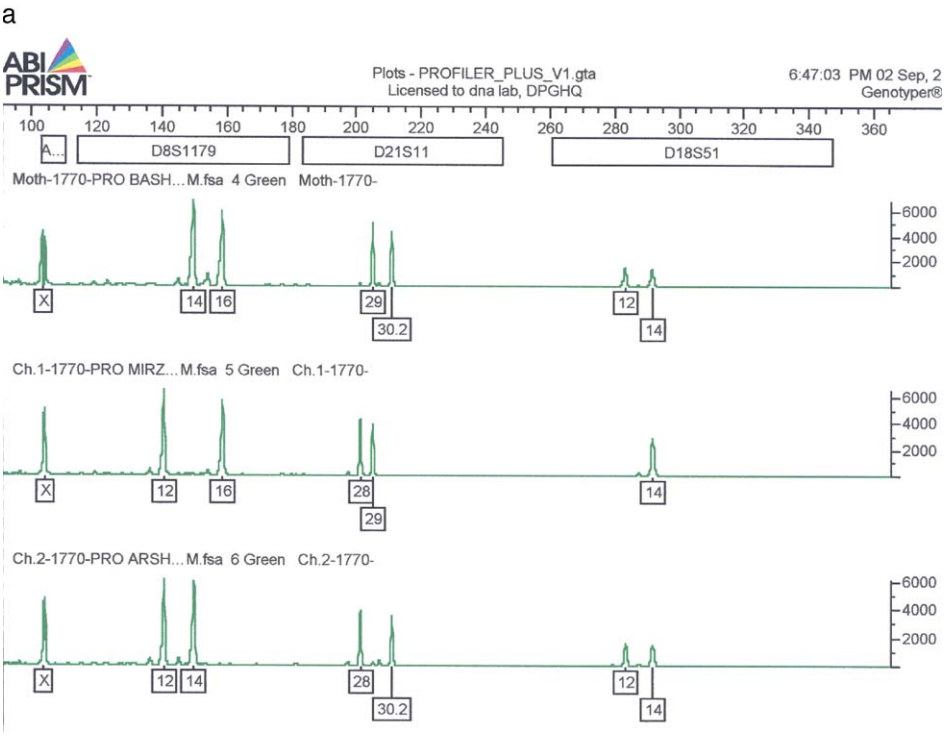

Fig. 1. (a) Autosomal STR profiles of the mother and two sons. (b) The YSTR profiles of the sons showing the same haplotype.

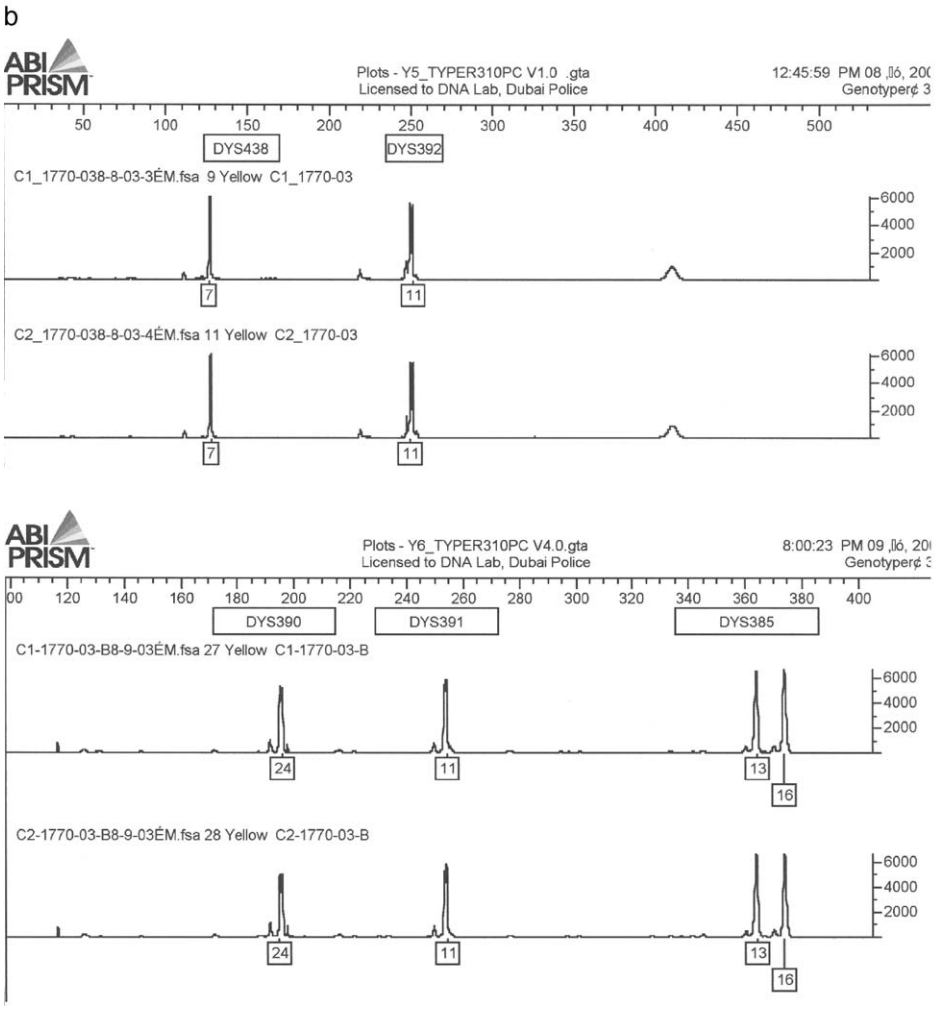

Fig. 1 (*continued*).

brother. The Y STR profile comparison clearly was different and provided a simple and a straightforward exclusion.

2.2. Case 2: sexual assault

A 14-year-old girl who had been missing from her home was found with an evidence of sexual abuse. The results obtained by autosomal STR analysis showed a mixture of more than one individual in the male fraction. The Y STR results were consistent with two males comprising the sample. One suspect was identified and could not be excluded as a contributor of the profile by both autosomal and Y STR typing. The second contributor has yet to be identified.

2.3. Case 3: forensic paternity

A woman accused a man of being the father of her child. The man denied the allegation. The trio was profiled using 15 autosomal STR markers. There were two differences found: a possible one step mutation at the CSF1PO locus on the mother's side and a null allele, i.e., a possible primer binding site mutation, at the D5S818 locus on the father side. The Y STR profiles were identical in the child and alleged father. Thus, the data could not exclude the alleged father as the biological father.

2.4. Case 4: gender confirmation

A putative mother and her two sons were referred to the lab to assess maternity. The father was not available for testing. The amelogenin results showed only an X in all three individuals. Testing with Y STR markers demonstrated that the putative sons did carry a Y chromosome and shared an identical Y haplotype at all 11 markers used (Fig. 1). The amelogenin anomaly is likely the result of a mutation or deletion in the region of the Y chromosome where the amelogenin primers reside [4].

3. Conclusion

Y STRs can supplement the highly robust autosomal STRs used in both forensic and paternity testing and in some instances provide information where autosomal markers fail to do so. Y STR typing could be used to assist in establishing kinship, to help resolve complicated paternity cases, to provide a simple straightforward means of exclusion in motherless paternities and in one of the most challenging applications in the field of forensics, i.e. mass disasters [1]. Although Y STRs are not as discriminating as autosomal STRs, they offer certain advantages in the analyses of different types of mixture samples. Autosomal STR profiles obtained in such cases at times provide inconclusive results. In conclusion, autosomal and Y STRs combined provide a powerful tool for certain forensic and paternity cases. Reliable Y STR typing has augmented the forensic arsenal for resolving complicated cases.

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