CASE REPORT

Male DNA Fingerprints Say More

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In 2002, a woman was found with a smashed skull and covered in blood but still alive in her Berlin apartment. Her life was saved by intensive medical care. Later she told the police that she had let a man into her apartment, and he had immediately attacked her. The man was subletting the apartment next door. The evidence collected at the scene and in the neighboring apartment included a baseball cap, two towels and a glass. The evidence was sent to the state police laboratory in Berlin and was analyzed with conventional autosomal STR markers. Stains on the baseball cap and on one towel revealed a pattern consistent with that of the tenant, whereas two different male DNA profiles were found on a second bath towel and on the glass. The tenant was eliminated as a suspect because he was absent at the time of the offense, but two unknown men who shared the apartment were suspected. Unfortunately, the apartment had been used by many individuals of both European and African nationalities, so the initial search for the two men became very difficult.

The police obtained a court order for Y-chromosomal STR analysis (Y-STR haplotyping) to gain information about the unknown men's population affiliation. Y-STR haplotyping has been performed at the Institute of Legal Medicine, Humboldt University, Berlin, since 1991 to resolve male/female DNA admixtures in vaginal swabs and fingernails. In several high-profile cases, the Y-STR haplotyping method has been applied to gain a likelihood-based assessment of the population origin of a DNA sample. The specificity of Y-chromosome STR markers for specific populations is much higher than that of autosomal loci, and Y-STR haplotyping can often differentiate between closely related and admixed populations from the European continent. Prerequisites for such phylogeographic analyses are large reference databases containing Y-STR haplotypes from hundreds of different populations. The Y-STR Haplotype Reference Database (see www.yhrd.org), a collaborative project involving more than 100 forensic laboratories worldwide, proved useful to infer the population origin of the unknown male.

Y-STR analysis was performed on the four DNA samples collected from the apartment to establish the minimal Y-STR haplotype profiles (see Table 1). Since the police already knew the tenant's nationality, this search was ordered as a control to test the quality of the method. Searching for the tenant's Y-STR profile in the reference databases provided a clue about his ancestry—Southern Europe. The results pointed in the expected direction; the man was from Italy, which is right in the center of the radiant frequency distribution of the detected haplotype profile (see Figure 1). The analysis results from the two unknown DNA samples surprised the police. The autosomal profiles of these samples did not point to relatedness, but complete analysis of the Y-STR profiles for all 9 loci of the minimal haplotype profile strongly suggested a paternal relatedness of the two suspects. A father/son relationship could be excluded since the autosomal profiles did not share alleles in two out of eleven analyzed STR loci. They could be brothers, cousins or otherwise paternally related. A population query against the Y-STR Haplotype Reference Database (YHRD) was not as sensitive as in the case of the tenant because the two suspects' haplotypes were significantly less frequent (0.05% compared to 0.3% in about 15,500 Europeans). Nevertheless, the search result indicated a patrilineage of European ancestry and African descent was unlikely.

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Also, none of the African, African-American or African-Caribbean haplotypes (n = 1,587) matched the profile of the suspects; instead 7 matches were found in European populations (Figure 1).

The police were able to track down the tenant in Italy, and with his help, establish the identity of one of the unknown men, who was also Italian. When questioning this man, the police used the information that he had shared the apartment in Berlin with a paternal relative. This relative was identified as his nephew. Because of the close-knit relationship within the family, this information would probably not have been easily retrieved from the uncle without the prior knowledge. The nephew was suspected of the attempted murder in Berlin. He was later arrested in Italy, where he had committed another violent robbery.

Table 1. Autosome and Y-chromosome profiles of 2 paternally related persons who left traces at the crime scene. Exclusion constellations for a father/son relationship are marked in red.

STR	"Nephew"	"Uncle"
Amelogenin	X,Y	X,Y
D3S1358 I/II	15,17	14,16
vWA	16,17	15,16
FGA	20,21	21,22
D8S1179	10,12	12,15
D21S11	30,33.2	29,33.2
D18S51	15,16	15,19
D5S818	12,13	11,11
D13S317	11,13	11,11
D7S820	8,11	8,10
TH01	8,9	8,9.3
SE33	22,26.2	21,22
DYS19	14	14
DYS389I	12	12
DYS389II	28	28
DYS390	24	24
DYS391	10	10
DYS392	11	11
DYS393	12	12
DYS385	14,17	14,17

This actual case report illustrates the usefulness of the Y-STR haplotyping method, which was developed more than 10 years ago but rose to worldwide attention only recently with the development of highly sensitive multiplex PCR kits. Besides the analysis of mixed stains, especially in rape cases, the method has a wider range of applications as exemplified in this case report—determination of male relatedness and male population affiliation. The latter application has already been used in Germany, Switzerland and Sweden, mainly to assist in the identification of severely decomposed bodies or to retrieve intelligence information on an unknown culprit's identity.

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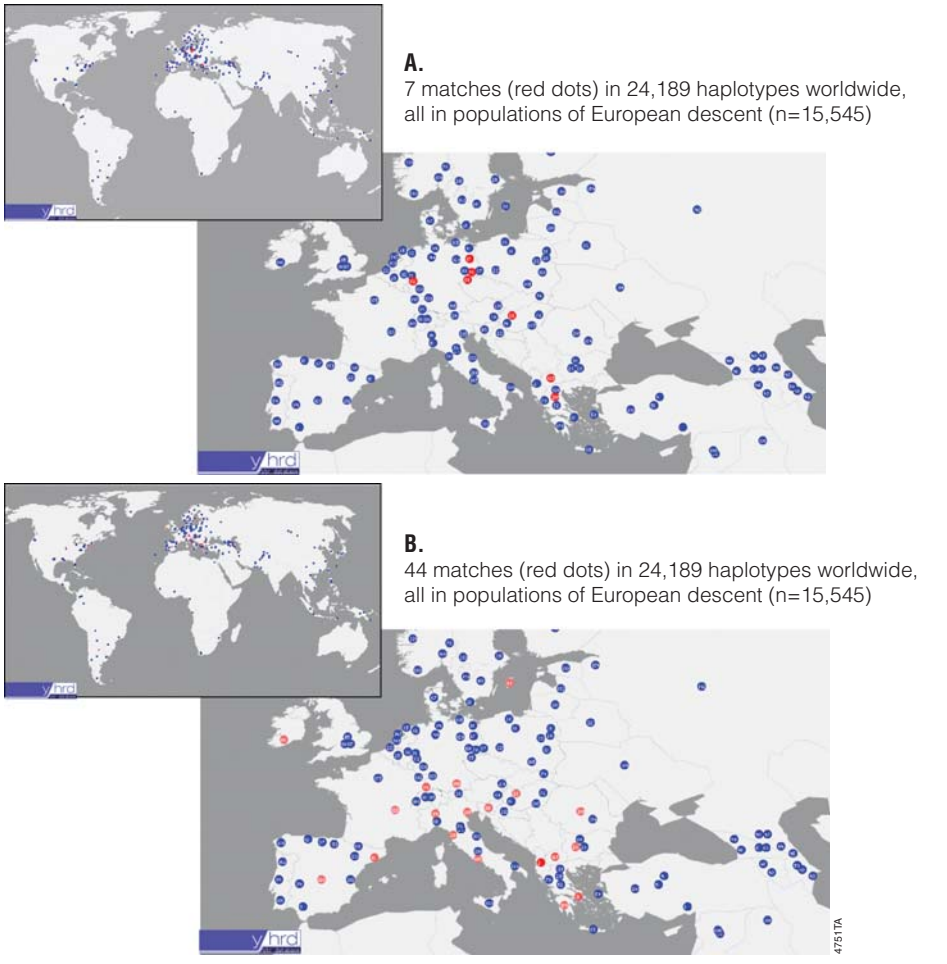

A.
7 matches (red dots) in 24,189 haplotypes worldwide, all in populations of European descent (n=15,545)

B.
44 matches (red dots) in 24,189 haplotypes worldwide, all in populations of European descent (n=15,545)

Figure 1. Y-STR Haplotype Reference Database query for the "uncle/nephew" and "tenant" haplotypes. Panel A. Search results for the "uncle/nephew" haplotype: 14,12,28,24,10,11,12,14–17 **Panel B.** Search results for the "tenant" haplotype: 14,13,29,24,11,13,13,11–11. From the YHRD-Release 13, July 1, 2004.