

Y-STRS

Son, Give Up Your Gun: Presenting Y-STR Results in Court

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With the eagerly awaited PowerPlex® Y System^(a) now released, it is probably the right moment to carefully think about the forensic use of Y-STRs. Interpretation and presentation of matching Y-STR profiles in court can be prone to serious misunderstandings. Here I would like to concentrate on the most obvious issue: sharing of identical Y-STR profiles among male relatives.

INTRODUCTION

In the previous issue of *Profiles in DNA* (1), Gusmão and Carracedo provided an excellent introduction to important technical aspects of Y-STRs. Here I will concentrate where they ended: the interpretation and presentation of Y-STRs in court.

Because of its patrilineal inheritance pattern, Y-STR profiles (or haplotypes) co-segregate with surnames in many western societies. A detailed discussion on this topic was given by Jobling (2), and for an illustrative example see Sykes and Irven (3). Irrespective of this co-segregation, it is a simple fact of life (and genetics) that all patrilineal male relatives of a single multigeneration pedigree share an identical Y-STR profile. In addition, in many European countries, male relatives tend to live close together in a rather small geographic area. As a consequence of this, presenting a Y-STR match between a crime sample and a suspect is complex. The example below merely illustrates some aspects of this complexity.

THE "D" PEDIGREE

Figure 1 illustrates a simple 5-generation pedigree, connecting all Dutch males with the surname "D". There are 4 main branches with 26 males currently still alive. Most members (21 males) live within a radius of 25km of each other in the south of the Netherlands (Figure 2). Members of a fourth branch live in two other areas, each separated from each other and the core area by at least 100km. The last member recently "migrated" out of the core area to the west coast of the Netherlands. Recent census data indicated that the core area is inhabited by 95,000 males (all ages). Thus among males in this area, the frequency of the D surname and Y-STR haplotype is 0.022% (or 1 in 4,545). If we assume (i) that we have sampled all males with the D surname and (ii) that the D haplotype is unique for this pedigree, the frequency of the haplotype and surname across Holland is about 0.0003% (1 in 307,700).

All males of this pedigree share a 16-Y-STR haplotype. Of these, 8 loci represent the Y-STR database minimal haplotype 14-12-28-23-10-11-13-13,14 (DYS19-389)/II-390-391-392-393-385a/b). A recent search in this database at:

ystr.charite.de resulted in 81 matches among 12,802 minimal haplotypes, representing a mean frequency of 0.63% (1 in 159) across all haplotypes sampled among Europeans. The geographical distribution of these 81 matches is shown in Figure 3. To our surprise, and despite its frequent occurrence among many German population samples, the D haplotype was not found among the 275 Dutch and 125 Belgian haplotypes present in this database.

A Y-STR database search provides essential information about the geographic distribution and variation of the frequency among populations of a Y-STR haplotype.

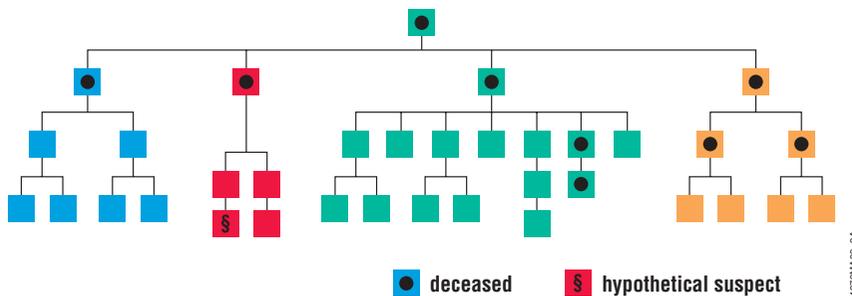


Figure 1. The 5-generation pedigree connecting all males in the Netherlands with the surname "D". Each distinct branch is indicated with a different color.

THE CRIME, THE MATCH, THE REPORT, AND THE EXPLANATION

Let us assume that a serious crime was committed and a suspect was apprehended. From a single crime sample, only a Y-STR profile could be obtained. Autosomal STRs only produced a profile matching the victim. Most likely due to preferential amplification, the autosomal STR profile of the suspect was completely masked by the victim's profile. Based on this scenario, it is safe to conclude that this Y-STR match could be important, if not crucial, especially if this is the only incriminating evidence linking this suspect to the victim and the crime. In such situations my written conclusion is always as follows:

"The Y-STR profile of the crime sample matches the Y-STR profile of the suspect. Therefore we cannot exclude the suspect as being the donor of the crime sample. In addition, we cannot exclude all patrilineal related male relatives and an unknown number of unrelated males as being the donor of the crime sample."

This extremely conservative conclusion is an open invitation for questions from the prosecution. Often these relate to the lack of an estimate of the frequency of this Y-STR profile. I answer these questions using the following example:

1. Consider a pedigree, such as the D pedigree, and assume that our suspect is one of the males in this pedigree (indicated by § in Figure 1).

In addition to our suspect, there are 25 close relatives, 20 of which are from the same geographic (and social) environment, and are equally likely candidates without any other prior incriminating evidence. Even if there is such incriminating evidence, I am usually not informed about this at the time of writing my DNA report.

2. Consider the information obtained from searching the Y-STR Database with the D-haplotype. Search results indicate a frequency of approximately 0.63% (or 1 in 159) but note the absence of the D haplotype in Holland.

Reporting an estimated Y-STR profile frequency is very misleading in such a situation. In the case of an autosomal STR profile, both the defense and the prosecution are accustomed to the use of match likelihood as a statistical estimate of the number of unrelated individuals with the same DNA profile as that of the suspect and the crime sample. For Y-STR profiles, it is safe to assume that there are other related males with identical profiles. When I report the database frequency of this profile (0.63% or 1 in 159), I thereby severely

underestimate this probability. If I have all the pedigree D information at hand and report the region-specific profile frequency of 0.022% (1 in 4,545), my frequency estimate seems more accurate, but the probability of another related male with the same profile would still be 100%.

3. Consider the given fact that two identical Y-STR haplotypes can be identical due to random mutation.

This is more difficult to explain to those not familiar with Y-profiles, but nevertheless I use examples that were published previously (4,5). It follows that even the probability of another unrelated male displaying a similar profile is not negligible.

DISCUSSION

Superficially, one could ask "why bother using these new markers if one cannot use them in court?". This reply has been given to me many times. I do not believe that Y-STRs are without use and should not be used in court. On the contrary, I think they are very useful. I also do not state that



Figure 2. Map of the The Netherlands showing the place of current residence of all Dutch males with the surname "D". Colors correspond with those from Figure 1. Unnumbered circles represent a single male. Numbers in a circle indicate the number of males of each distinct branch.

Y-STRS

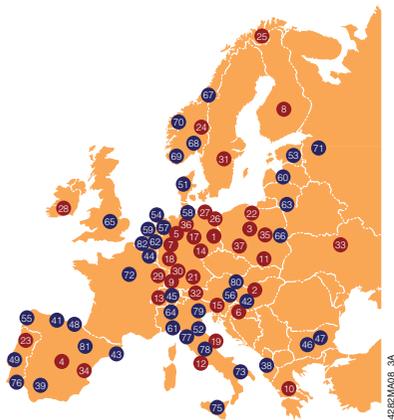


Figure 3. Geographical distribution of the Y-STR Database hits of minimal haplotype D. Blue dots represent populations not containing the D haplotype; red dots indicate centers containing one or more hits with the D haplotype.

the Y-STR Database is useless (as some people think), only that one should realize when and how to use the information therein. Information obtained from the Y-STR Database should be seen as qualitative, not quantitative. A database search provides essential information about the geographic distribution and variation of the frequency among populations of a Y-STR haplotype. This knowledge can be very important since it could suggest that, even if the Y-haplotype has not been observed in your relevant population, it is probably there. Criticism of my careful approach ignores the many cases where suspects are excluded on the basis of nonmatching Y-STR profiles. Even in the case of matching profiles, one presents the prosecution with one valuable piece of technical information, namely that we do not exclude the suspect from being the perpetrator. It is their job to use this piece of information. This sometimes leads to the apprehension of a male relative of the original suspect as the true perpetrator of the crime (6).

Gusmão and Carracedo (1) are correct in warning against multiplying Y-STR allele frequency estimates. Here I also warn against the use of Y-STR haplotype frequencies, which should not be confused with autosomal STR match likelihoods. However, some DNA experts have no problem multiplying the Y-STR haplotype frequency with the autosomal STR-derived match likelihood obtained from the same crime sample. This approach ignores the issue of male relatives and confuses two unrelated statistical estimates.

CONCLUSION

The use of Y-STRs for forensic purposes will increase several orders of magnitude now that the PowerPlex® Y System has been released. This product facilitates the sensitive and reliable typing of a 12-locus Y-STR haplotype. Based on first hand experience, this system is helpful in resolving previously hopeless cases. Now the many difficult technical aspects of obtaining a reliable Y-STR profile are history; we should concentrate on the use of this information in court. The issues discussed in this article merely scratch the surface. There are other points (some of which easily extend to the use of mitochondrial DNA profiles), that should also be studied. For the time being, I hope that anyone who uses this kind of evidence does so carefully. One can rarely be certain whether it is the father or the son who holds the smoking gun.

ACKNOWLEDGMENTS

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Editor's Note: Approaches to statistical interpretation of Y-STR data vary. The opinions expressed in this article do not necessarily reflect those of Promega Corporation.

REFERENCES

- Gusmão, L. and Carracedo, A. (2003) Y chromosome-specific STRs. *Profiles in DNA* **6(1)**, 3–6.
- Jobling, M.A. (2001) In the name of the father: Surnames and genetics. *Trends Genet.* **17**, 353–7.
- Sykes, B. and Irven, C. (2000) Surnames and the Y-chromosome. *Am. J. Hum. Genet.* **66**, 1417–9.
- de Knijff, P. (2000) Y-chromosomes shared by descent or by state. In: *Archaeogenetics: DNA and the population prehistory of Europe*. Renfrew, C. and Boyle, K. (eds.) The McDonald Institute, Cambridge, 301–4.
- de Knijff, P. (2000) Messages through bottlenecks: On the combined used of slow and fast evolving polymorphic markers on the human Y chromosome. *Am. J. Hum. Genet.* **67**, 1055–61.
- Detlaff-Kakol, A. and Pawlowski R. (2002) First Polish DNA "manhunt"—an application of Y-chromosome STRs. *Int. J. Leg. Med.* **116**, 289–91.