

# Lineage Markers: Y Chromosome and mtDNA Testing

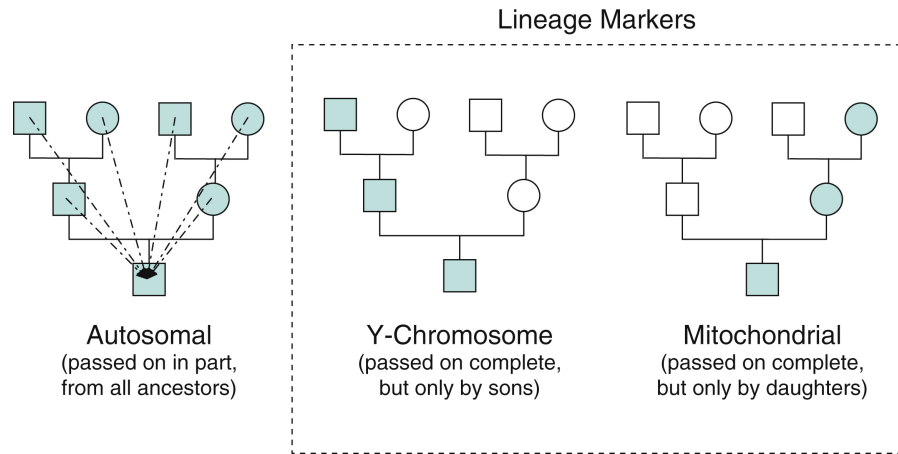
DNA is the messenger, which illuminates (our connection to the past), handed down from generation to generation, carried, literally, in the bodies of (our) ancestors. Each message traces a journey through time and space....

—Bryan Sykes, *The Seven Daughters of Eve*

## LINEAGE MARKERS

Autosomal DNA markers, such as the 13 core short tandem repeat (STR) loci, are shuffled with each generation because half of an individual's genetic information comes from his or her father and half from his or her mother. However, the Y chromosome (ChrY) and mitochondrial DNA (mtDNA) markers that will be discussed in this chapter represent 'lineage markers.' They are passed down from generation to generation without changing (except for mutational events). Maternal lineages can be traced with mitochondrial DNA sequence information, whereas paternal lineages can be followed with Y-chromosome markers (Figure 16.1).

With lineage markers, the genetic information from each marker is referred to as a haplotype rather than a genotype because there is only a single allele per individual. Because Y chromosome markers are linked on the same chromosome and are not shuffled with each generation, the statistical calculations for a random match probability cannot involve the product rule. Therefore, haplotypes obtained from lineage markers can never be as effective in differentiating between two individuals as genotypes from autosomal markers that are unlinked and segregate separately from generation to generation. However, as will be discussed in this chapter, Y chromosome, X chromosome, and mitochondrial DNA markers can play an important role in forensic investigations as well as other applications.

**FIGURE 16.1**

*Illustration of inheritance patterns from recombining autosomal genetic markers and the lineage markers from the Y chromosome and mitochondrial DNA.*

## Y-CHROMOSOME DNA ANALYSIS

The human Y chromosome (ChrY) is the second smallest human chromosome with a length of approximately 60 million nucleotides. The tips of the Y chromosome, which are called the *pseudoautosomal regions*, recombine with their sister sex X-chromosome homologous regions. The remainder of the Y chromosome (~95%) is known as the *nonrecombining portion of the Y chromosome*, or NRY. The NRY remains the same from father to son unless a mutation occurs. Genetic polymorphisms including STRs and SNPs that occur along the NRY can be used to track male genetic pedigrees as they are passed on intact from father to son barring any mutations that may occur.

### Applications of ChrY testing

Y-chromosome DNA testing is important for a number of different applications of human genetics including forensic evidence examination, paternity testing, historical investigations, studying human migration patterns throughout history, and genealogical research (Table 16.1).

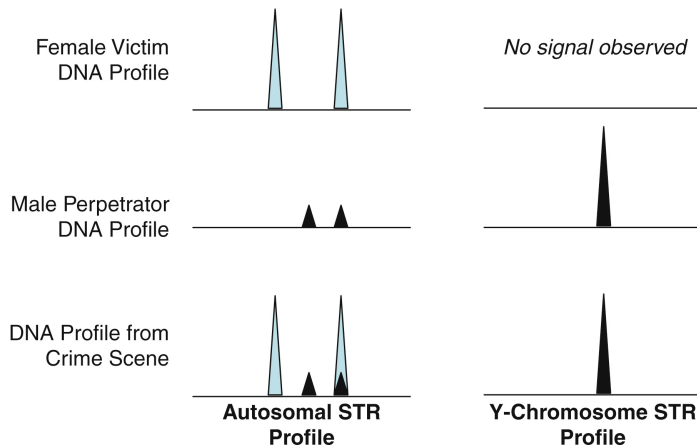
The value of the Y chromosome in forensic DNA testing is that it is found only in males. The SRY (sex-determining region of the Y) gene determines maleness. Since a vast majority of crimes where DNA evidence is helpful, particularly sexual assaults, involve males as the perpetrators, DNA tests designed to only examine the male portion can be valuable.

With Y-chromosome tests, interpretable results can be obtained in some cases where autosomal tests are limited by the evidence, such as high levels of

**Table 16.1** Areas of use in Y-chromosome testing.

Use	Advantage
Forensic casework on sexual assault evidence	Male-specific amplification (can avoid differential extraction to separate sperm and epithelial cells)
Verification of amelogenin Y-deficient males	Analysis of multiple regions along the Y chromosome that should not be affected by deletion of the amelogenin region
Paternity testing	Male children can be tied to fathers in motherless paternity cases or testing of male relatives if father is unavailable
Missing persons investigations	Patrilineal male relatives may be used for reference samples
Human migration and evolutionary studies	Lack of recombination enables comparison of male individuals separated by large periods of time
Historical and genealogical research	Surnames usually retained by males; can make links where paper trail is limited

Female-Male Mixture Performance with Autosomal vs. Y-Chromosome DNA Markers

**FIGURE 16.2**

*Schematic illustrating the types of autosomal or Y-STR profiles that might be observed with sexual assault evidence where mixtures of high amounts of female DNA may mask the STR profile of the perpetrator.*

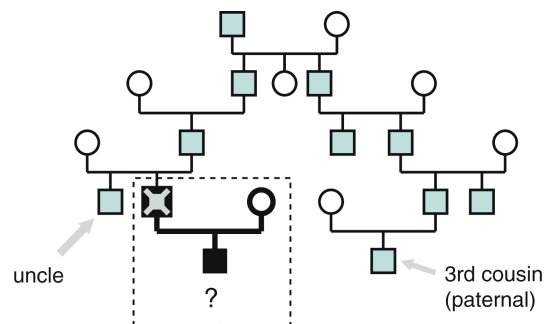
*Y-STR testing permits isolation of the male component without having to perform a differential lysis.*

female DNA in the presence of minor amounts of male DNA (Figure 16.2). These situations include sexual assault evidence from azoospermic or vasectomized males and blood–blood or saliva–blood mixtures where the absence of sperm prevents a successful differential extraction for isolation of male DNA. In addition, the number of individuals involved in a ‘gang rape’ may be

easier to decipher with Y-chromosome results than with highly complicated autosomal STR mixtures. The use of Y-chromosome-specific PCR primers can improve the chances of detecting low levels of the perpetrator's DNA in a high background of the female victim's DNA. Y-chromosome tests have also been used to verify amelogenin Y-deficient males.

The same feature of the Y chromosome that gives it an advantage in forensic testing, namely maleness, is also its biggest limitation. A majority of the Y chromosome is transferred directly from father to son (see Figure 16.1) without recombination to shuffle its genes and provide greater variety to future generations. Random mutations are the only mechanisms for variation over time between paternally related males. Thus, while exclusions in Y-chromosome DNA testing results can aid forensic investigations, a match between a suspect and evidence only means that the individual in question could have contributed the forensic stain—as could a brother, father, son, uncle, paternal cousin, or even a distant cousin from his paternal lineage! Needless to say, inclusions with Y-chromosome testing are not as meaningful as autosomal STR matches from a random match probability point of view.

On the other hand, the presence of relatives having the same Y chromosome (Figure 16.3) expands the number of possible reference samples in missing persons investigations and mass disaster victim identification efforts. Deficient paternity tests where the father is dead or otherwise unavailable for testing are benefited if Y-chromosome markers are used. However, an autosomal DNA test is always preferred if possible since it provides a higher power of discrimination.



**FIGURE 16.3**

*An example pedigree showing patrilineal inheritance where all shaded males have the same Y chromosome barring any mutations. To help identify the person in question, any of the other males with the same patrilineage could provide a reference sample to assist in a missing persons investigation, mass disaster victim identification, or deficient paternity test (boxed region) where the father is deceased or otherwise not available for testing.*

The Y chromosome has also become a popular tool for tracing historical human migration patterns through male lineages. Anthropological, historical, and genealogical questions can be answered through Y-chromosome results. For example, Y-chromosome results in 1998 linked modern-day descendants of Thomas Jefferson and Eston Hemings, leading to the controversial conclusion that Jefferson fathered the slave (D.N.A. Box 16.1).

### D.N.A. Box 16.1 The Thomas Jefferson–Sally Hemings Affair

In 1802, a year after becoming president of the United States, Thomas Jefferson was publicly accused by a Richmond, Virginia, newspaper of fathering a child by his slave, Sally Hemings. While it is uncertain how this accusation arose, the connection between Thomas Jefferson and his slave Sally Hemings has been a source of controversy for almost 200 years. In November 1998, the prestigious scientific journal *Nature* published a report that introduced DNA evidence into this historical controversy. The report, entitled 'Jefferson fathered slave's last child,' used Y-chromosome DNA markers to trace the Jefferson male line to a descendant of Sally Hemings's youngest son, Eston Hemings. The study involved 19 samples collected from living individuals who represented the Jefferson and Hemings line as well as other people who potentially could have been Jefferson's offspring or the father of Eston Hemings. These samples were tested at 19 different sites on the Y chromosome.

The study began in 1996 when Dr. Eugene Foster, a retired pathology professor, began tracking down living male-line relatives of President Thomas Jefferson. To show whether or not President Jefferson had fathered a child with Sally Hemings, direct male descendants were needed from both the Jefferson and the Hemings lines. Unfortunately, Jefferson's only legitimate son died in infancy. His two daughters who lived to adulthood obviously did not carry his Y chromosome and therefore their descendants were not useful in this study. There were two other possibilities for direct male-line descendants, Thomas Jefferson's brother Randolph and his father's brother Field. The last of the direct male descendants of Jefferson's brother Randolph died in the 1920s or 1930s so Dr. Foster turned to the relatives of President Jefferson's paternal uncle, Field Jefferson. Seven living descendants of Field Jefferson were located. Five of them agreed to cooperate in the study and had their blood drawn for Y-chromosome marker testing purposes.

On the Hemings side of the equation, it was even more difficult to come up with an abundance of living male relatives. Sally Hemings had at least six children: Harriet (1795–1797), Beverly (1798–post 1822), Harriet (1801–post 1822), an unnamed daughter (1799–1800), Madison (1805–1877), and Eston (1808–1856). According to the oral history of the descendants of Thomas Woodson (1790–1879), he was Sally Hemings's first child. Sally's son Beverly and daughter Harriet are listed as dying post 1822 because they disappeared into white society in the Washington, DC, area in the year 1822. Of the three known male sons from Sally Hemings, only descendants of Madison and Eston could possibly be located since Beverly's fate is unknown. Madison's Y-chromosome line ended in the mid-1800s when one of his three sons vanished into white society and the other two had no children. Thus, Eston Hemings's descendants remained the last chance to find a male-line descendant of the man who fathered Sally Hemings's children.

Eston Hemings was born on 21 May 1808, at Monticello where he lived until President Jefferson's death in 1826, at which time he was freed. Eventually he married and moved to Ohio and finally to Madison, Wisconsin, where he died and was buried in 1856. Eston assumed the surname of Jefferson when he left Virginia and gave everyone the impression that he was white because of his light skin color. Eston Hemings Jefferson had two sons and a daughter. His youngest son, Beverly Jefferson, lived from 1838–1908 and had one son. This son, Carl Smith Jefferson, lived from 1876–1941 and had two sons, William Magill Jefferson (1907–1956) and Carl Smith Jefferson, Jr. (1910–1948). Only William had a son. This son, John Weeks Jefferson, was born in 1946. As the *only* living male descendant of Eston Hemings, John Weeks Jefferson's blood was drawn to help answer the question of whether or not President Thomas Jefferson was Eston Hemings's father.

(Continued)

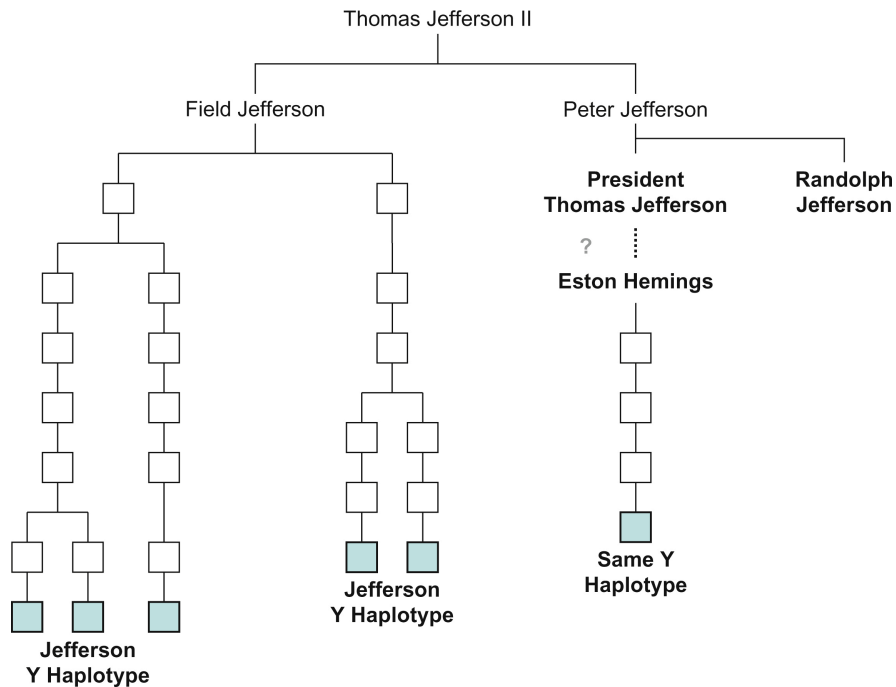
## D.N.A. Box 16.1 (Continued)

Another important set of samples for testing was gathered from direct male-line descendants of Samuel and Peter Carr, who were Thomas Jefferson's nephews, the sons of his sister. According to Thomas Jefferson's grandchildren Thomas Jefferson Randolph and Ellen Coolidge, Samuel and Peter Carr were the fathers of the children of Sally Hemings and her sister. Dr. Foster collected three blood samples from living descendants of John Carr, the grandfather of Samuel and Peter Carr. Finally, five male descendants from several old-line Virginia families around Charlottesville were sampled to serve as control samples. These controls were tested to provide a 'background' signal with the idea that potential similarities in the Y-chromosome tests due to geographic proximity needed to be eliminated.

In all, 19 Y-chromosome markers were examined in this study. These included 11 STRs, seven SNPs, and one minisatellite MSY1, which proved to be the most polymorphic marker. All 19 regions of the Y chromosome examined in this study matched between the Jefferson and Hemings descendants.

These DNA results were viewed by Dr. Foster and his coauthors as evidence for President Thomas Jefferson fathering the last child of Sally Hemings. The John Carr lines differed significantly from the Jefferson–Hemings results with at least 7 of the 19 tested DNA markers giving different results. Thus, neither Samuel Carr nor Peter Carr was the father of Eston Hemings. The results of the Virginia old-line families were not reported, presumably because these samples served their purpose as effective controls and revealed no unusual Y-chromosome patterns.

Shortly after the results of Dr. Foster's study were announced, an alternative scenario was proposed. Could some other male Jefferson have fathered Eston Hemings? All the results in this study conclusively show is that there is a genetic match between descendants of Eston Hemings and Thomas Jefferson's uncle, Field Jefferson. Was it historically possible for another male Jefferson to have fathered Sally Hemings's children? The Thomas Jefferson Memorial Foundation, a private,



*Ancestry of Thomas Jefferson and Eston Hemings male lines. The shaded boxes represent the samples tested by Foster et al. (1998) in their Jefferson Y-chromosome study. A male descendant of Eston Hemings, son of Thomas Jefferson's slave Sally Hemings, was found to have a Y-chromosome haplotype that matched male descendants of Field Jefferson, President Thomas Jefferson's uncle. Although he nor none of his descendants was tested, Randolph Jefferson, President Jefferson's brother, would also possess the same Y-chromosome haplotype.*

nonprofit organization established in 1923 that owns and operates Monticello with the goal of preservation and education, conducted a yearlong investigation into the historical record.

According to this careful historical investigation, 25 adult male descendants of Thomas Jefferson's father Peter and his uncle Field lived in Virginia during the 1794–1807 period of Sally Hemings's pregnancies (Monticello, 2000). Most of them lived over 100 miles from Monticello and make no appearance in Thomas Jefferson's correspondence documents. Several male Jeffersons including President Jefferson's brother Randolph and his sons did live in the area of Monticello and visited occasionally. However, the historical records fail to indicate that any of these individuals were present at Monticello 9 months before the births of Sally Hemings's children. This information combined with the fact that Thomas Jefferson was present at Monticello during the time of conception of each of Sally Hemings's six children led to the 26 January 2000 Thomas Jefferson Memorial Foundation report that he was the father of all of Sally Hemings's children.

A more recent study by a 13-member Scholars Commission of the Thomas Jefferson Heritage Society unanimously agreed that the allegations of a relationship are 'by no means proven.' The findings of this group are reported in a 565-page report available at the Heritage Society's Web site: <http://www.tjheritage.org>. This report notes that the original DNA study indicated only that a Jefferson male had fathered

one of Sally Hemings's children and that the available DNA evidence could not specify Thomas Jefferson as the father *to the exclusion of all other possibilities*. Thomas Jefferson's younger brother Randolph, who was known to fraternize with the Monticello slaves, is considered a likely possibility by many members of the Scholars Commission. Randolph and other family members would have visited Monticello when President Jefferson was home and therefore the circumstantial evidence of Thomas Jefferson being present on the plantation when Sally Hemings conceived might not be as strong as originally presented.

This study of Jefferson lineage DNA demonstrates one of the major disadvantages of Y-chromosome DNA testing, namely that results only indicate connection to a male lineage and are not specific to an individual like autosomal STR profiles can be. While a Jefferson Y-chromosome match exists between his descendants and those of Sally Hemings, the matter can probably never be definitely solved by Y-chromosome information alone.

*Sources:*

- Foster, E. A., et al. (1998). Jefferson fathered slave's last child. *Nature*, 396, 27–28.
- Monticello. (2000). Thomas Jefferson Memorial Foundation Research Committee Report on Thomas Jefferson and Sally Hemings, 26 January 2000. Available at <http://www.monticello.org>
- Murray, B., & Duffy, B. (1998). *US News and World Report*, 1998, November 9. pp. 59–63.

## ChrY markers

Y-chromosome DNA analysis can be performed with either Y-STRs or Y-SNPs. Since Y-STRs change more rapidly (mutation rate  $\sim 1$  in  $10^3$ ) compared to Y-SNPs (mutation rate  $\sim 1$  in  $10^9$ ), Y-STR results exhibit more variability and thus have greater use in forensic applications. Typically Y-STRs are described as defining haplotypes while Y-SNPs define haplogroups. Y-SNPs can be useful in DNA ancestry studies.

Although more than 400 Y-STR loci have been mapped along the human Y chromosome, only a small set of core loci are routinely analyzed. In 1997, the European forensic community settled on a core set of Y-STR markers or 'minimal haplotype' that includes DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393, and DYS385 a/b. Most Y-chromosome data to date has been generated with these loci. In 2003, the U.S. Scientific Working Group on DNA Analysis Methods (SWGDM) recommended a core set of Y-STRs that includes the nine markers in the minimal haplotype plus DYS438 and DYS439. These loci are available in commercial Y-STR kits (see next section).

Although new markers will be added to databases as their value is demonstrated and they become part of commercially available kits, these 11 established markers are likely to continue to be important in future Y-STR research.

### Y-STR kits

As discussed previously in Chapter 8, forensic scientists rely heavily on commercially available kits to perform DNA testing. Table 16.2 lists the loci present in the two most widely used Y-STR kits (Table 16.3): PowerPlex Y (Promega Corporation) and Yfiler (Applied Biosystems). Note that all of the European and U.S. core Y-STR loci are included in both kits with PowerPlex Y having one additional locus (DYS437) and Yfiler have six additional loci (DYS437, DYS448, DYS456, DYS458, DYS635, and GATA-H4). A Yfiler kit PCR amplification result is shown in Figure 16.4.

**Table 16.2** Characteristics of commonly used Y-chromosome STR loci.

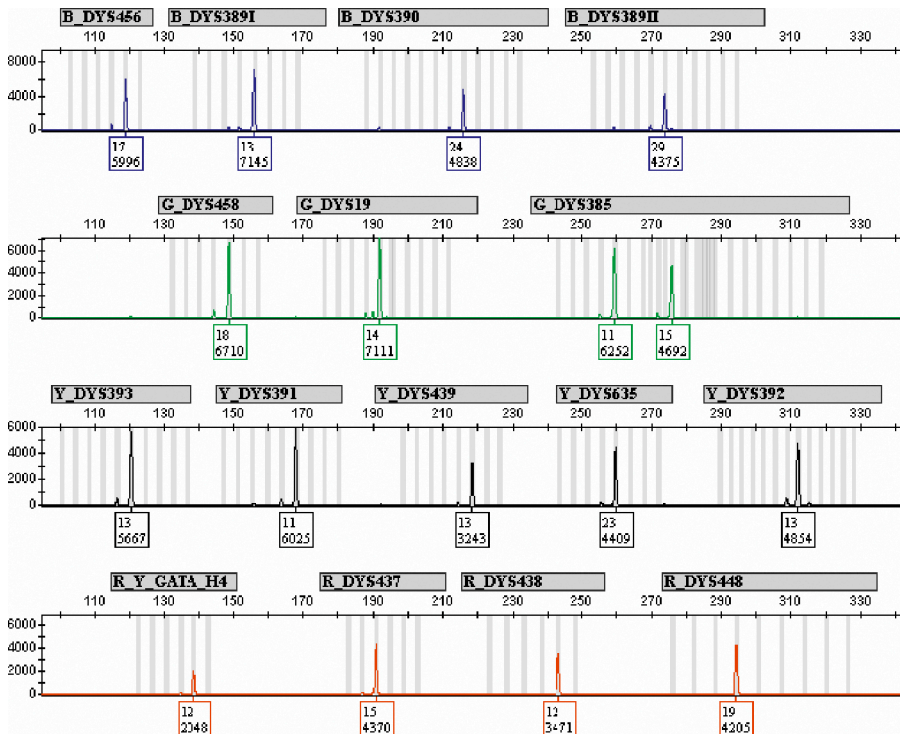
STR Marker	Position (Mb)	Repeat Motif	Allele Range	Mutation Rate
DYS393	3.19	AGAT	8–17	0.08%
DYS456	4.33	AGAT	13–18	0.72%
DYS458	7.93	GAAA	14–20	1.1%
DYS19	10.13	TAGA	10–19	0.24%
DYS391	12.61	TCTA	6–14	0.29%
DYS635	12.89	TSTA	17–27	0.42%
DYS437	12.98	TCTR	13–17	0.15%
DYS439	13.03	AGAT	8–15	0.54%
DYS389 I/II	13.12	TCTR	9–17/24–34	0.19%/0.30%
DYS438	13.38	TTTTTC	6–14	0.05%
DYS390	15.78	TCTR	17–28	0.24%
GATA-H4	17.25	TAGA	8–13	0.25%
DYS385 a/b	19.26	GAAA	7–28	0.22%
DYS392	21.04	TAT	6–20	0.06%
DYS448	22.78	AGAGAT	17–24	0.18%

*Adapted from Butler, J. M. (2006). Genetics and genomics of core short tandem repeat loci used in human identity testing. Journal of Forensic Sciences, 51, 253–265; Decker, A. E., et al. (2007). The impact of additional Y-STR loci on resolving common haplotypes and closely related individuals. Forensic Sciences International: Genetics, 1, 215–217; and Decker, A. E., et al. (2008). Analysis of mutations in father–son pairs with 17 Y-STR loci. Forensic Sciences International: Genetics, 2, e31–e35.*

**Table 16.3** Commercially available Y-STR kits. Characteristics of each locus may be found in Table 16.2.

Kit Name (Source)	Dye Color	Loci Amplified Arranged by Size
PowerPlex Y (Promega Corporation)	Blue	DYS391, <u>DYS389I</u> , <u>DYS439</u> , <u>DYS389II</u>
	Green	<u>DYS438</u> , <u>DYS437</u> , <u>DYS19</u> , <u>DYS392</u>
	Yellow	DYS393, <u>DYS390</u> , <u>DYS385a/b</u>
Yfiler (Applied Biosystems)	Blue	<u>DYS456</u> , <u>DYS389I</u> , <u>DYS390</u> , <u>DYS389II</u>
	Green	<u>DYS458</u> , <u>DYS19</u> , <u>DYS385a/b</u>
	Yellow	DYS393, <u>DYS391</u> , <u>DYS439</u> , <u>C4</u> , <u>DYS392</u>
	Red	<u>H4</u> , <u>DYS437</u> , <u>DYS438</u> , <u>DYS448</u>

Note: An internal size standard is typically run in the fourth or fifth dye position on multicolor fluorescence detection systems for allele sizing purposes. The underlined loci, such as *DYS437* present in the PowerPlex Y kit, are not part of the core Y-STR loci.



**FIGURE 16.4** Yfiler result from a single-source male of European ancestry. Allele calls and peak heights are shown under each peak. (Figure courtesy of Amy Decker, NIST.)

### Y-STR haplotype databases

A number of online Y-STR databases exist (Table 16.4). The forensic databases contain collections of anonymous individuals and can be used to estimate the frequency of specified Y-STR haplotypes. The genetic genealogy databases, such as Ysearch and Ybase, contain Y-STR haplotype information gathered by genetic genealogy companies with different sets of loci from males trying to make genealogical connections. Thus, the haplotypes in these genealogy databases are associated with specific individuals and family names.

The largest and most widely used forensic and general population genetics Y-STR database, known as the Y-STR Haplotype Reference Database (YHRD), was created by Lutz Roewer and colleagues at Humbolt University in Berlin, Germany, and has been available online since 2000. As of December 2008, YHRD contains results from more than 65,000 samples representing greater than 500 different groups of sample submissions from various populations around the world.

A U.S.-population-specific Y-STR Database (US Y-STR) was launched in December 2007 to enable haplotype frequency estimates on five different U.S. groups using the 11 SWGDAM recommended loci. The original version of US Y-STR contained 4796 African American profiles, 820 Asian, 5047 Caucasians, 2260 Hispanics, and 983 Native Americans. In some cases, further subdivision of these five primary groups can be examined if desired. Where possible, US Y-STR has attempted to ensure that no duplicates are present through examining autosomal STR typing results on any samples possessing the same Y-STR profile. Having both autosomal and Y-STR data can be helpful in cases of common Y-STR haplotypes.

**Table 16.4** Summary of available online Y-STR databases (as of December 2008).

Database	Number of Samples (Population Groups)	Number of Y-STR Markers Tested	Web Site
Y-STR Haplotype Reference Database (YHRD)	65,165 (517 groups)	7 to 17	<a href="http://www.YHRD.org">http://www.YHRD.org</a>
US Y-STR Database (US Y-STR)	13,906 (5 groups)	11 to 17	<a href="http://www.usystrdatabase.org">http://www.usystrdatabase.org</a>
Ysearch	65,869 (genealogists)	Up to 100	<a href="http://www.ysearch.org">http://www.ysearch.org</a>
Ybase	13,830 (genealogists)	Up to 49	<a href="http://www.ybase.org">http://www.ybase.org</a>
Sorenson Molecular Genealogy Foundation	>30,000 (genealogists)	Up to 43	<a href="http://www.smgf.org">http://www.smgf.org</a>

*Note:* For updated information, see [http://www.cstl.nist.gov/biotech/strbase/y\\_strs.htm](http://www.cstl.nist.gov/biotech/strbase/y_strs.htm)

Several genetic genealogy Y-STR haplotype databases are also available online. These databases are typically not used for Y-STR forensic haplotype frequency estimates, but could be helpful in trying to associate a family surname with a particular haplotype if this information was desired in an investigation. These genetic genealogy databases contain information from the minimal haplotype loci, a subset of the minimal haplotype loci, or additional Y-STRs, and therefore cannot always be searched across all loci of interest.

## Interpretation of Y-STR results

Since the Y chromosome is passed down unchanged (except for mutations) from father to son, the observation of a match with Y-STRs does not carry the same power of discrimination and weight in court as an autosomal STR match would. The lack of recombination between Y-chromosome markers means that Y-STR results have to be combined into a haplotype for searching available databases as well as estimating the rarity of a particular haplotype.

Generally speaking there are three possible interpretations resulting from comparing Y-STR haplotypes produced from question (Q) and known (K) samples: (1) *exclusion* because the Y-STR profiles are different and could not have originated from the same source, (2) *inconclusive* where data are insufficient to render an interpretation or ambiguous results were obtained, or (3) *failure to exclude* (or *inclusion*) because the Y-STR haplotype results from the Q-K comparison are the same and could have originated from the same source.

When the Q and K samples (e.g., evidence and suspect haplotypes) do not match, then Y-STR typing is helpful in demonstrating the exclusion. However, estimating the strength of a match when a suspect's Y-STR haplotype cannot be excluded is more problematic because barring any mutations, paternal relatives (e.g., all brothers, male children, father, uncles, paternal grandfather, and paternal cousins) would be expected to have the same Y-STR profile.

Since it is common practice to place some significance on the likelihood of a random match with unrelated individuals, statistics derived from population data can be applied. Estimates for a random match with Y-STR haplotypes (and mtDNA sequence information) are done by the *counting method*, where the number of times the haplotype of interest is observed is divided by the total number of haplotypes in the database used. The size of the database used for the counting method makes a difference when trying to estimate the rarity of a Y-STR profile. The larger the number of unrelated individuals in the database (i.e., the denominator in the counting method calculation), the better the statistics will be for a random match frequency estimate.

The Y-STR profile observed earlier in [Figure 16.4](#) is found zero times in 10, 243 17-locus haplotypes found in YHRD and zero times in 4163 17-locus

### D.N.A. Box 16.2 Calculation of Y-STR Haplotype Frequency Estimates Using the Counting Method

In cases where a Y-STR haplotype is observed a particular number of times ( $X$ ) in a database containing  $N$  profiles, its frequency ( $p$ ) can be calculated as follows:

$$p = X/N$$

A 95% upper bound confidence interval (CI) can be placed on the profile's frequency using:

$$p + 1.96 \sqrt{\frac{p(1-p)}{N}}$$

In cases where the profile has not been observed in a database, the upper bound CI is

$$1 - \alpha^{1/N} \quad \text{or} \quad 1 - (0.05)^{1/N}$$

where  $\alpha$  is the confidence coefficient (0.05 for a 95% confidence interval) and  $N$  is the number of individuals in the database.

Using the haplotype results from the 17-locus Yfiler profile seen in Figure 16.4, a search was performed using the YHRD and US Y-STR databases. Subsets of the same haplotype

were also searched using the Y-STR loci present in the 12-locus PowerPlex Y, 11-locus SWGDAM recommended, 9-locus European minimal haplotype, and 7-locus minimal haplotype minus DYS385 a/b.

Note that the size of the database ( $N$ )—and therefore the denominator of the counting method—changes depending on the number of Y-STR loci searched because these databases contain fewer complete 17-locus Yfiler haplotypes compared to the usual full set of minimal haplotype results. As with any investigation, the relevance of using an entire world survey of Y-STR haplotypes rather than a specific ethnic or geographic group for a population comparison must also be considered within the scope of the case. The results from several different population databases could be utilized in a case report for comparison purposes. In the end, comparisons would only be reported where results were available for both the Q and K samples.

Sources:

Y-STR Haplotype Reference Database (YHRD). <http://www.yhrd.org>  
US Y-STR Database. <http://www.usystrdatabase.org>

YHRD	Search Results	Upper Bound CI	~1 in Every...
17-locus	0/10243	0.029%	3420
12-locus	3/13751	0.047%	2150
11-locus	18/36174	0.073%	1375
9-locus	307/63369	0.539%	186
7-locus	2099/65165	3.36%	30

US Y-STR	Search Results	Upper Bound CI	~1 in Every...
17-locus	0/4163	0.072%	1390
12-locus	7/10865	0.112%	892
11-locus	19/13906	0.198%	505
9-locus	128/13906	1.08%	93
7-locus	846/13906	6.48%	15

haplotypes found in US Y-STR. D.N.A. Box 16.2 works through some example calculations with searches using subsets of the 17 Yfiler loci.

Frequency estimates calculated with the counting method, while not as powerful as those produced with unlinked autosomal STRs, may nevertheless be informative in many forensic casework scenarios and provide another piece of evidence in the overall framework of a case.