wyndham forensic group inc.
analysis & support

WHY Y's?

Strengths and Limitations of Male-Specific Y-STR Testing

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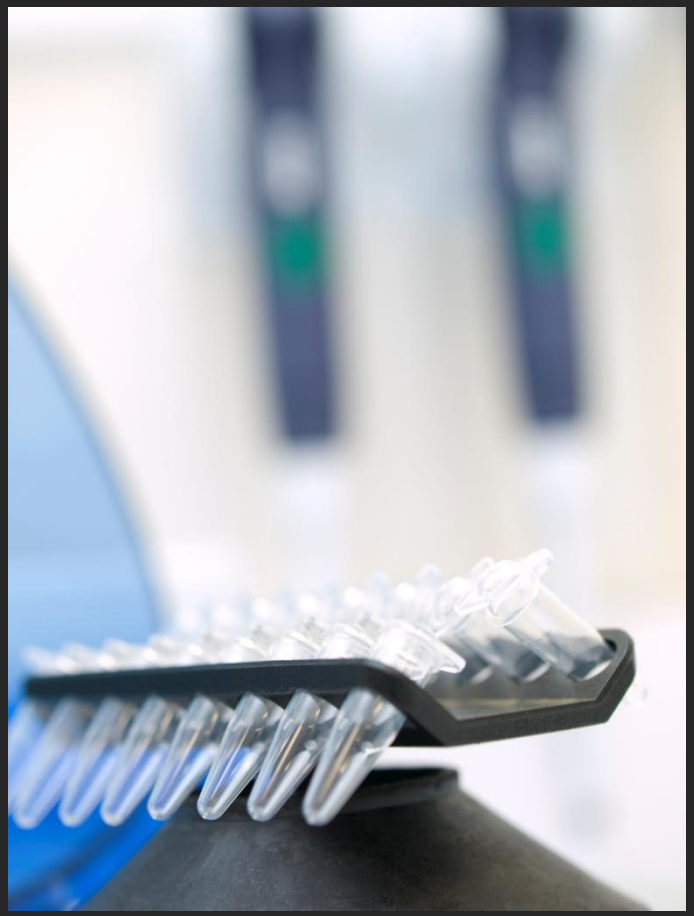
Senior Associates

General Information Presentation
Fall 2011

OUTLINE

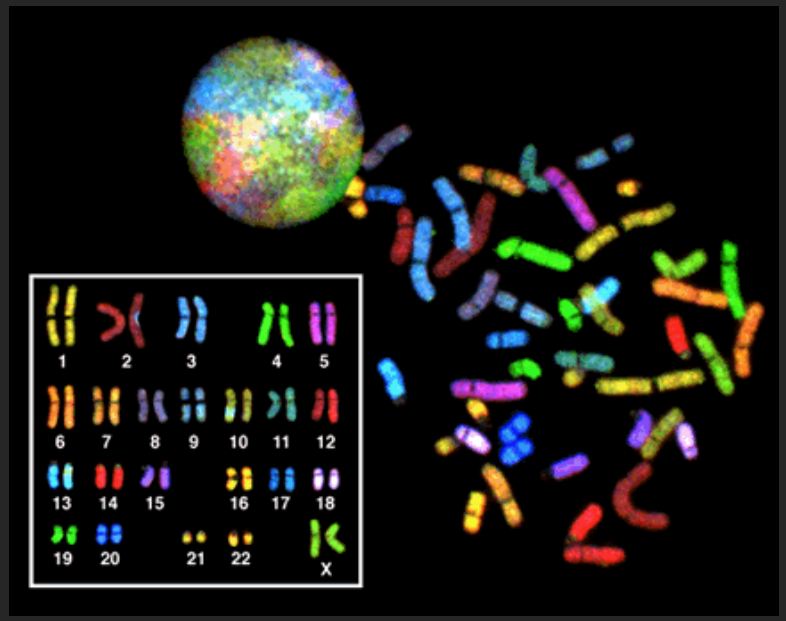


- Introduction
- Autosomal vs Y-STRs
- Implications for Forensic Science
- Conclusion

INTRODUCTION

- In the fall of 2011, Wyndham Forensic Group added Y-STR testing to its suite of forensic DNA capabilities.
- This presentation is designed to highlight a number of features of this technology and its application to forensic science.
- What is Y-STR testing? Why is it different than the traditional STR testing you may be accustomed to? When is it employed?
- Find the answers to these and other questions in the following slides.

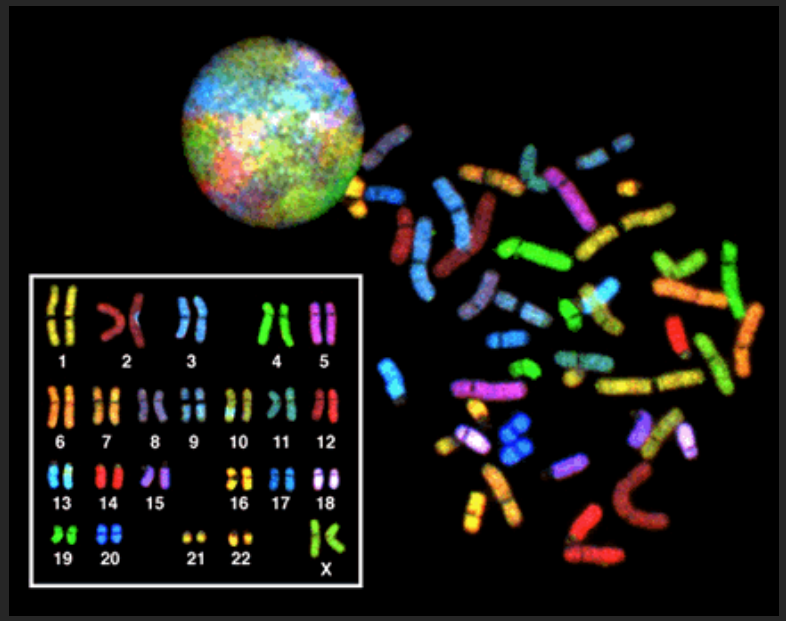
AUTOSOMAL vs Y STRs



Source: US National Human Genome Research Institute

- STR (Short Tandem Repeat) testing has been the gold standard for forensic DNA analysis since the mid 1990's.
- STRs are specific locations within one's DNA where there is a high degree of variability between individuals – this variability is what allows forensic scientists to readily distinguish individuals through their DNA.
- STRs are located throughout the human genome, on both the autosomal chromosomes (22 pairs) and on the sex chromosomes (1 pair).

AUTOSOMAL vs Y STRs



Source: US National Human Genome Research Institute

- Traditional STR testing, which sometimes leads to astronomical match probabilities and which is employed to generate profiles for inclusion in crime scene and offender databases, is performed on sites located throughout the autosomal chromosomes.
- Y-STR testing is performed on sites located on the Y chromosome, which is present only in males, and this has important implications for forensic science.

AUTOSOMAL vs Y STRs

D3S1358	16,17
vWA	15,19
FGA	21,24
THO1	6,8
TPOX	8,11
CSF1PO	9,12
D5S818	11,12
D13S317	12,12
D7S820	8,11

These are examples of DNA profiles.

DYS391	10
DYS389I	13
DYS439	10
DYS389II	30
DYS438	11
DYS437	14
DYS19	17
DYS392	11
DYS393	13
DYS390	25

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In each case, the leftmost column indicates the locations on the DNA where the tests were performed.

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These are examples of DNA profiles.

In each case, the leftmost column indicates the locations on the DNA where the tests were performed.

The rightmost column indicates the test results.

DYS391	10
DYS389I	13
DYS439	10
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DYS438	11
DYS437	14
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DYS392	11
DYS393	13
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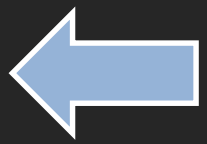

The DNA profile on the left is an example of an autosomal STR profile.

There are 9 sets of test results, and two results noted for each set.

AUTOSOMAL vs Y STRs

The DNA profile on the right is an example of a Y-STR profile.

There are 10 sets of test results, and one result noted for each set.

DYS391	10
DYS389I	13
DYS439	10
DYS389II	30
DYS438	11
DYS437	14
DYS19	17
DYS392	11
DYS393	13
DYS390	25

AUTOSOMAL vs Y STRs

D3S1358	16,17
vWA	15,19
FGA	21,24
THO1	6,8
TPOX	8,11
CSF1PO	9,12
D5S818	11,12
D13S317	12,12
D7S820	8,11

LOCUS

A specific test site on the DNA. (plural = loci)

DYS391	10
DYS389I	13
DYS439	10
DYS389II	30
DYS438	11
DYS437	14
DYS19	17
DYS392	11
DYS393	13
DYS390	25

AUTOSOMAL vs Y STRs

D3S1358	16,17
vWA	15,19
FGA	21,24
THO1	6,8
TPOX	8,11
CSF1PO	9,12
D5S818	11,12
D13S317	12,12
D7S820	8,11

ALLELE

A variant form of DNA

DYS391	10
DYS389I	13
DYS439	10
DYS389II	30
DYS438	11
DYS437	14
DYS19	17
DYS392	11
DYS393	13
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AUTOSOMAL vs Y STRs

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D5S818	11,12
D13S317	12,12
D7S820	8,11

ALLELE

A variant form of DNA.

At autosomal STR loci, each person has two alleles, one passed on from one's mother and one from one's father.

Although any given person can only possess two, the general population contains multiple alleles at each STR locus.

So, while the profile depicted here is '11,12' at D5S818, another unrelated person may be '10,13', for example, or any one of numerous other combinations.

DYS391	10
DYS389I	13
DYS439	10
DYS389II	30
DYS438	11
DYS437	14
DYS19	17
DYS392	11
DYS393	13
DYS390	25

AUTOSOMAL vs Y STRs

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THO1	6,8
TPOX	8,11
CSF1PO	9,12
D5S818	11,12
D13S317	12,12
D7S820	8,11

ALLELE

A variant form of DNA.

At Y-STR loci, each male has only one allele passed on from one's father.

Although any given male can only possess one, the general male population contains multiple alleles at each Y-STR locus.

So, while the profile depicted here is '10' DYS439, another unrelated male may be '12', for example, or one of a number of other alternatives.

DYS391	10
DYS389I	13
DYS439	10
DYS389II	30
DYS438	11
DYS437	14
DYS19	17
DYS392	11
DYS393	13
DYS390	25

AUTOSOMAL vs Y STRs

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CSF1PO	9,12
D5S818	11,12
D13S317	12,12
D7S820	8,11

PASSING ON ALLELES

Let's say the autosomal profile on the left is from a male and the autosomal profile on the right is from a female.

If these two people have a child, that child can inherit any combination of alleles represented at each locus.

Example – **D5S818**

Child could equally likely be:
10,11 or
10,12 or
11,13 or
12,13

D3S1358	14,16
vWA	17,18
FGA	21,24
THO1	7,7
TPOX	9,11
CSF1PO	9,10
D5S818	10,13
D13S317	12,12
D7S820	9,10

AUTOSOMAL vs Y STRs

D3S1358	16,17
vWA	15,19
FGA	21,24
THO1	6,8
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CSF1PO	9,12
D5S818	11,12
D13S317	12,12
D7S820	8,11

PASSING ON ALLELES

Let's say the autosomal profile on the left is from a male and the autosomal profile on the right is from a female.

If these two people have a child, that child can inherit any combination of alleles represented at each locus.

Example – **TPOX**

Child could equally likely be:
8,9 or
8,11 or
9,11 or
11,11

D3S1358	14,16
vWA	17,18
FGA	21,24
THO1	7,7
TPOX	9,11
CSF1PO	9,10
D5S818	10,13
D13S317	12,12
D7S820	9,10

AUTOSOMAL vs Y STRs

D3S1358	16,17
vWA	15,19
FGA	21,24
THO1	6,8
TPOX	8,11
CSF1PO	9,12
D5S818	11,12
D13S317	12,12
D7S820	8,11

PASSING ON ALLELES

Let's say the autosomal profile on the left is from a male and the autosomal profile on the right is from a female.

If these two people have a child, that child can inherit any combination of alleles represented at each locus.

The same chance pairings occur independently at each of the autosomal loci, which accounts for the tremendous diversity observed in people's DNA profiles.

D3S1358	14,16
vWA	17,18
FGA	21,24
THO1	7,7
TPOX	9,11
CSF1PO	9,10
D5S818	10,13
D13S317	12,12
D7S820	9,10

AUTOSOMAL vs Y STRs

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DYS19	17
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DYS393	13
DYS390	25

PASSING ON ALLELES

The Y chromosome is exclusive to males.

If the male represented by the Y-STR profile on the left has a female child, then none of the alleles represented in the Y-STR profile will be passed on.

If the male has a male child, then all of the alleles will be passed on, exactly as is, to the male child.

AUTOSOMAL vs Y STRs

DYS391	10
DYS389I	13
DYS439	10
DYS389II	30
DYS438	11
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DYS393	13
DYS390	25

PASSING ON ALLELES

Therefore, it can be said that if a certain male person possesses the Y-STR profile on the left, so too does:

- each of his full brothers
- his father
- his grandfather on his father's side
- his uncles on his father's side, as well as their sons
- etc…

IMPLICATIONS FOR FORENSIC SCIENCE

Two main features of Y-STR testing have important implications for forensic science:

1. Testing for Y-STRs specifically targets male DNA and ignores female DNA.

   This effectively means that when male DNA is present in small quantities in mixtures of female and male DNA, the use of Y-STR analysis greatly increases the chance of detecting the low level male contributions, generating results otherwise not possible using autosomal analysis.

IMPLICATIONS FOR FORENSIC SCIENCE

Example

As part of validating its Y-STR testing system, Wfg examined a vaginal swab from a volunteer collected 12hrs post-coitus, during which there was no ejaculation.

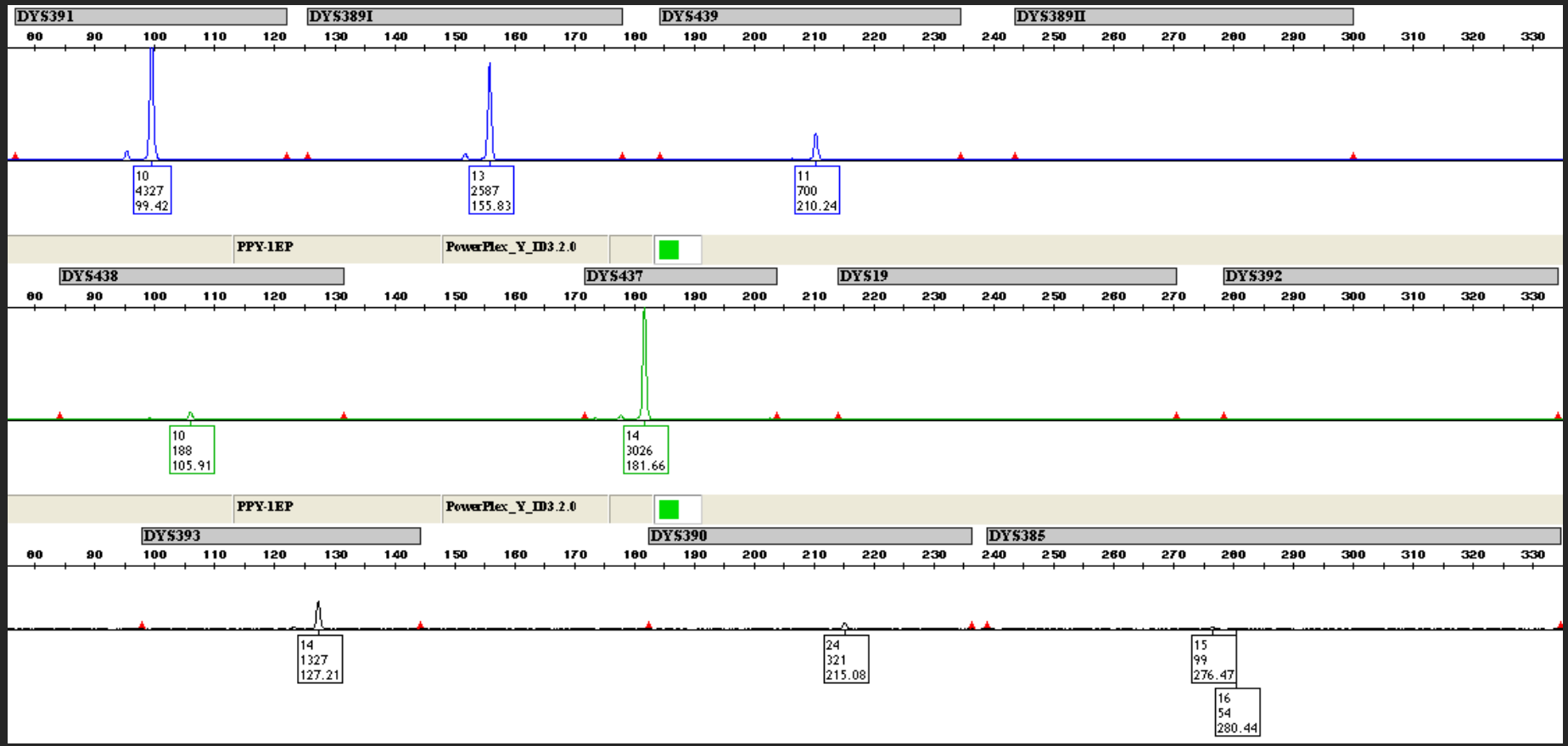
Analysis of the sample using autosomal STR technology detected a female profile only (the donor of the swab) and failed to detect any male DNA whatsoever.

Analysis of the same sample using Y-STR technology (depicted on next slide) detected a partial male Y-STR profile matching that of the female's consensual partner.

It was determined that there was approximately 11,000 times as much female DNA present in the sample as there was male DNA. Since the detection of minor sources of male DNA in female:male mixtures becomes unreliable with autosomal systems at ratios of as little as 30:1, this result exemplifies the value of Y-STR testing in select instances where autosomal analysis would be of no value.

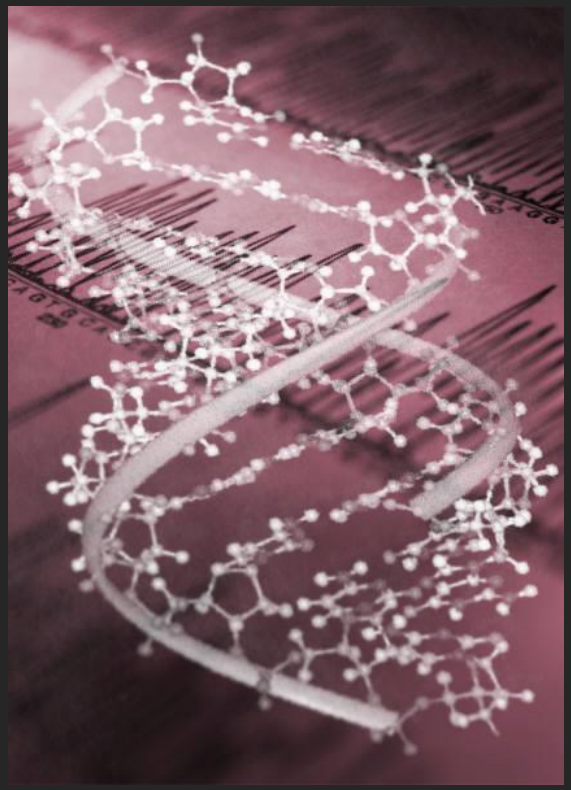
IMPLICATIONS FOR FORENSIC SCIENCE

Example: Partial Y-STR profile from post-coital vaginal swab (no ejaculation)



IMPLICATIONS FOR FORENSIC SCIENCE

Two main features of Y-STR testing have important implications for forensic science:



2. Y-STRs are passed on unaltered from fathers to their sons.

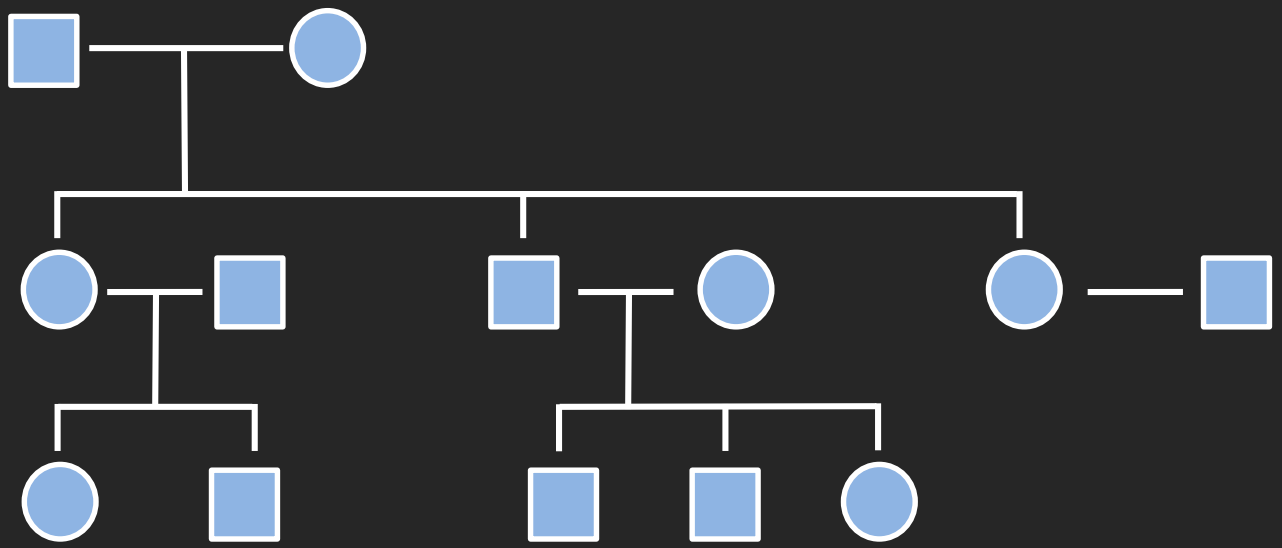
   In forensic science, this has an upside and a downside. On the downside, it effectively means that Y-STR profiles are not as discriminating as profiles generated using autosomal systems. All paternally-related males have the same Y-STR profile and thus cannot be distinguished using this test. Also, although unrelated males are generally expected to be distinguished using this test, random match probabilities are significantly higher (i.e. more common) than those generated with autosomal systems.

   On the upside, Y-STRs can be of great value in establishing familial relationships and can even be used to establish the DNA profile of a person of interest or a missing person from whom a sample cannot be obtained.

IMPLICATIONS FOR FORENSIC SCIENCE

Example

The following pedigree represents an average family. Square symbols represent males and round symbols represent females.



IMPLICATIONS FOR FORENSIC SCIENCE

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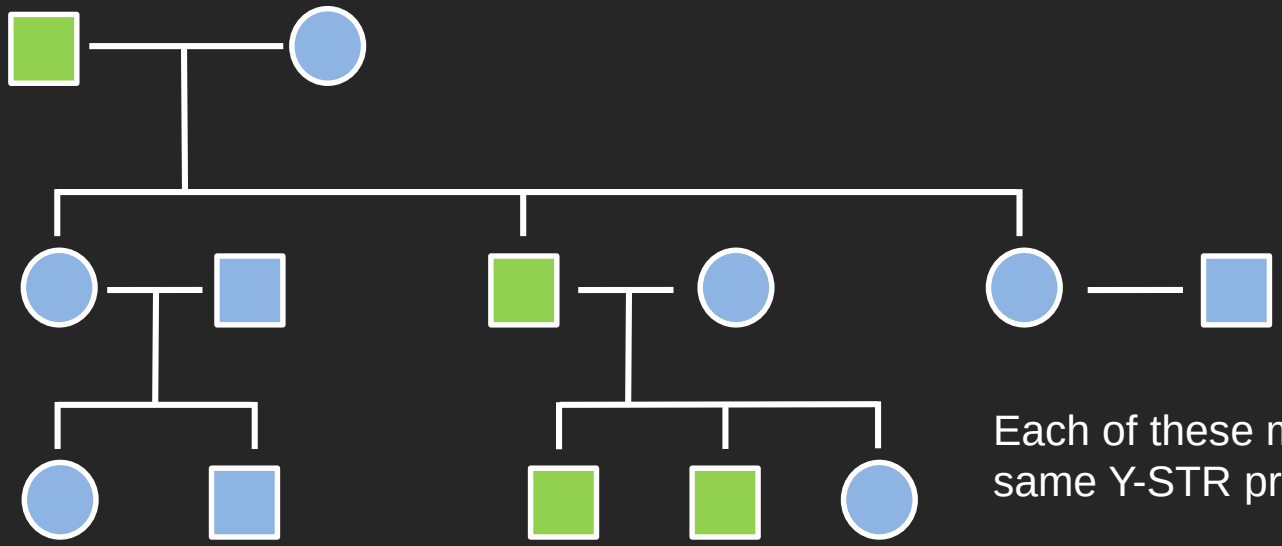
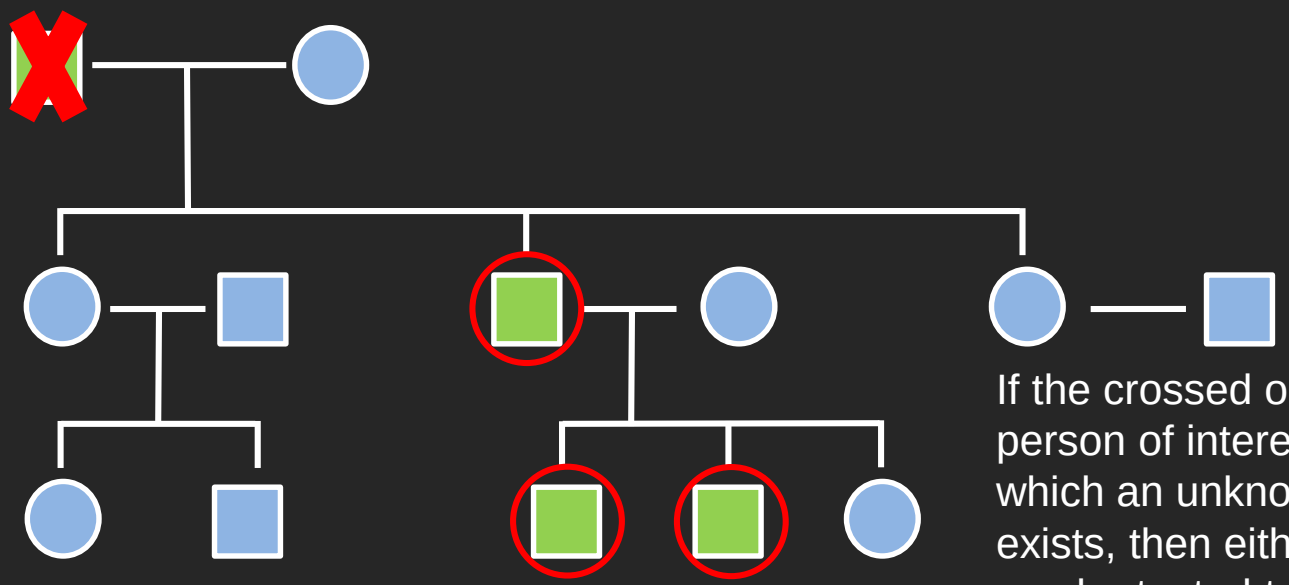


Each of these males will share the same Y-STR profile.

IMPLICATIONS FOR FORENSIC SCIENCE

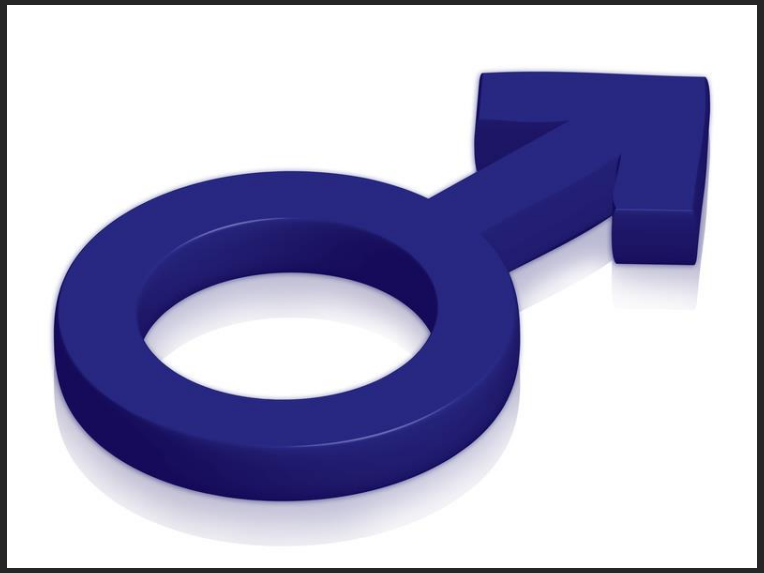
Example

The following pedigree represents an average family. Square symbols represent males and round symbols represent females.



If the crossed out male (deceased) is a person of interest in a cold case for which an unknown Y-STR profile exists, then either of the circled males can be tested to establish whether or not the person of interest can be excluded.

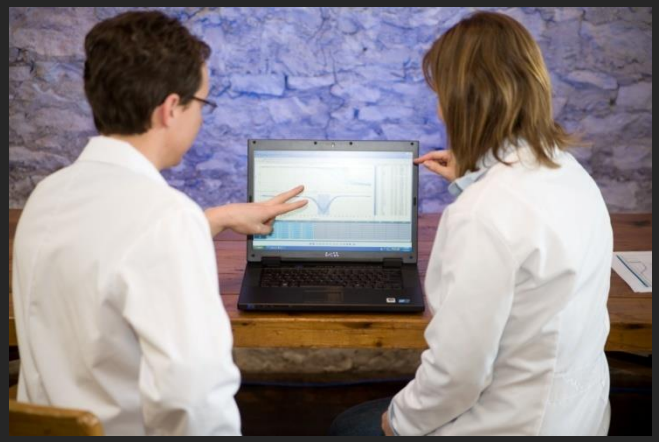
CONCLUSION



Y-STR testing is available at Wyndham Forensic Group as of November 2011. This technology adds an important tool to enhance forensic DNA testing in select cases:

- Sexual assaults and other cases with female:male mixtures of DNA
- Select relationship testing cases
- Select missing persons cases

ABOUT Wfg



- Privately-owned Canadian company committed to the timely delivery of forensic biology/DNA laboratory test results and opinions to all interested parties in the justice system.
- ISO17025 accredited by the Standards Council of Canada
- Services:
  - Forensic Biology/DNA Testing
  - Forensic Biology/DNA Casework Consultation
  - Training and Professional Development
  - Laboratory Management and QA Consultation
  - Laboratory Auditing

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