

WICSEC 2015

FORT WORTH
CITY OF COWBOYS AND CULTURE

Shaping the Future for Children

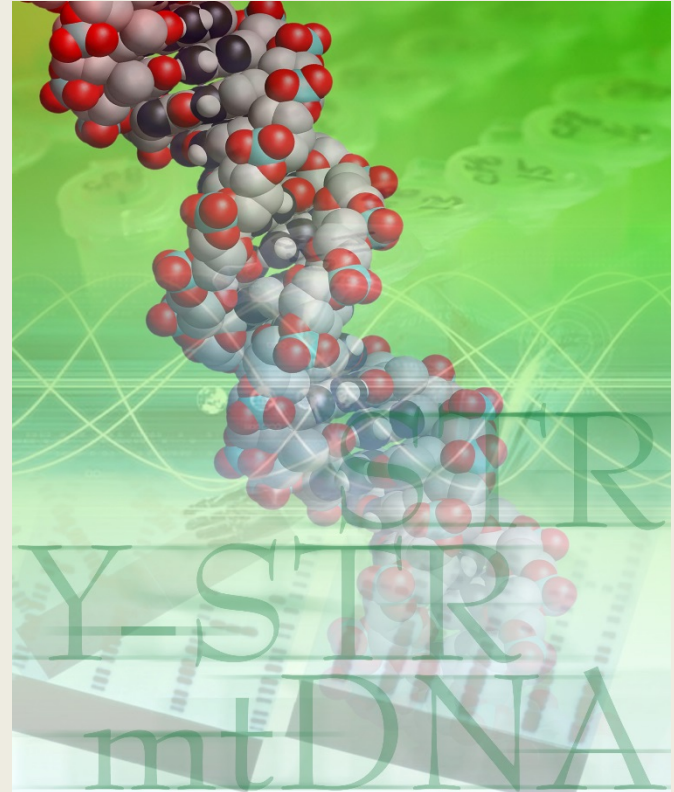
**Advances in DNA
Testing: The Art and
Science of Parentage**

Dr Michael L Baird



Overview

- u **DNA Basics**
- u **Biological Relationship Analysis**
- u **Databases and Calculations**
- u **Mutation**
- u **Chimera**
- u **Non-Invasive Prenatal Paternity**

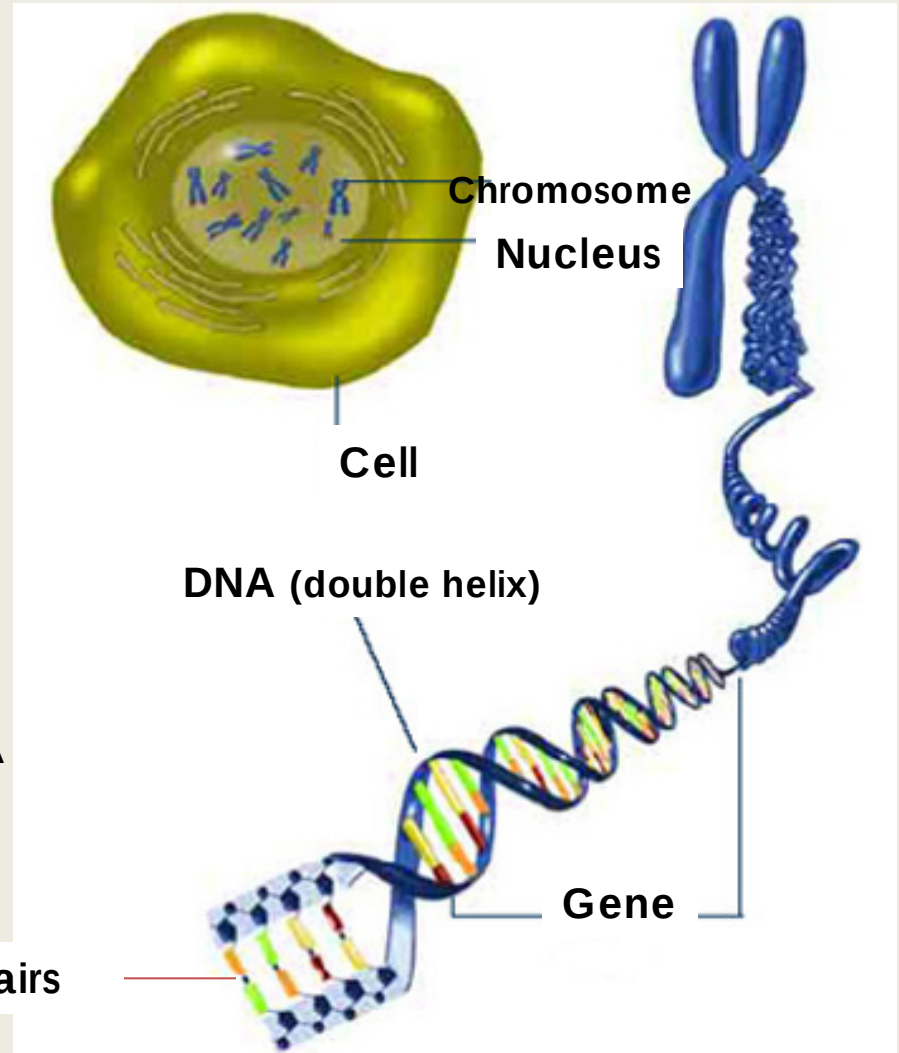


DNA Basics

About DNA

DNA=Deoxyribonucleic Acid

- DNA is found in most of the cells in the body
- DNA is the same in all of the cells in the body
- We inherit DNA from our parents
- No two people have the same DNA except identical twins



DNA Basics

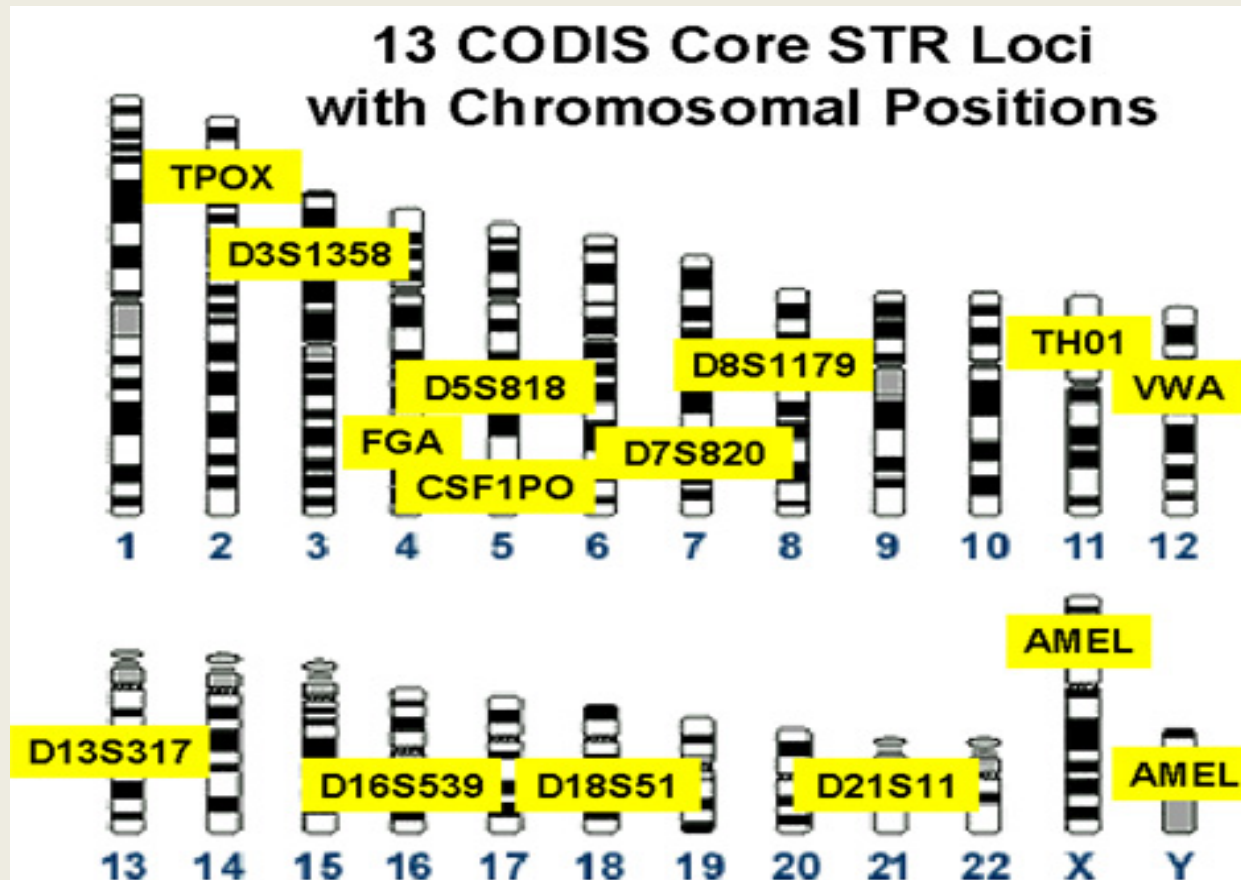
How DNA can be Used for Human Identification?

- 99.5% of everyone's DNA sequence is the same
- The unique markers (also called “loci”) found among the 0.5% of DNA that make humans different are used for human identification
- When we look at a collection of DNA loci from one person, we can generate a DNA profile
- A person's DNA profile is unique, just like a fingerprint



DNA Basics

Markers Examined for Identification



DNA Basics

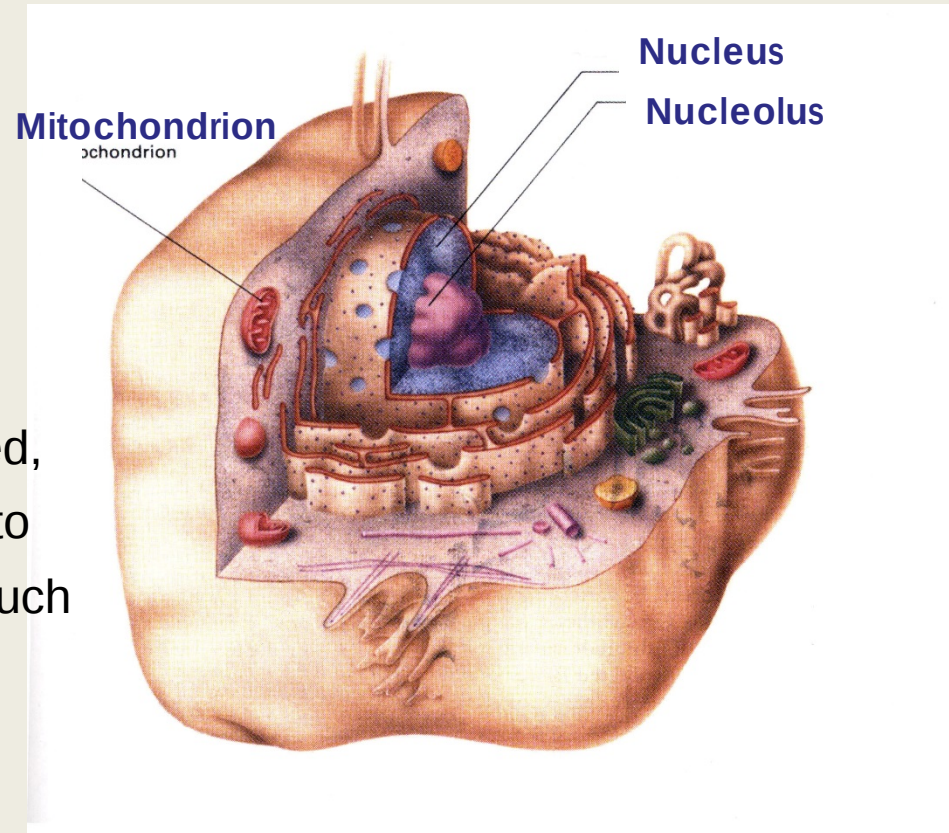
Testing Systems Available

STR: “Short Tandem Repeat”

unique to an individual, most powerful, regularly used for all types of samples

Mitochondrial DNA: maternally inherited, *not unique* to an individual, less prone to degradation, used for difficult samples such as bone, teeth, and hair without roots

Y-STR: male specific, paternally inherited, *not unique* to an individual, used for sexual assault cases



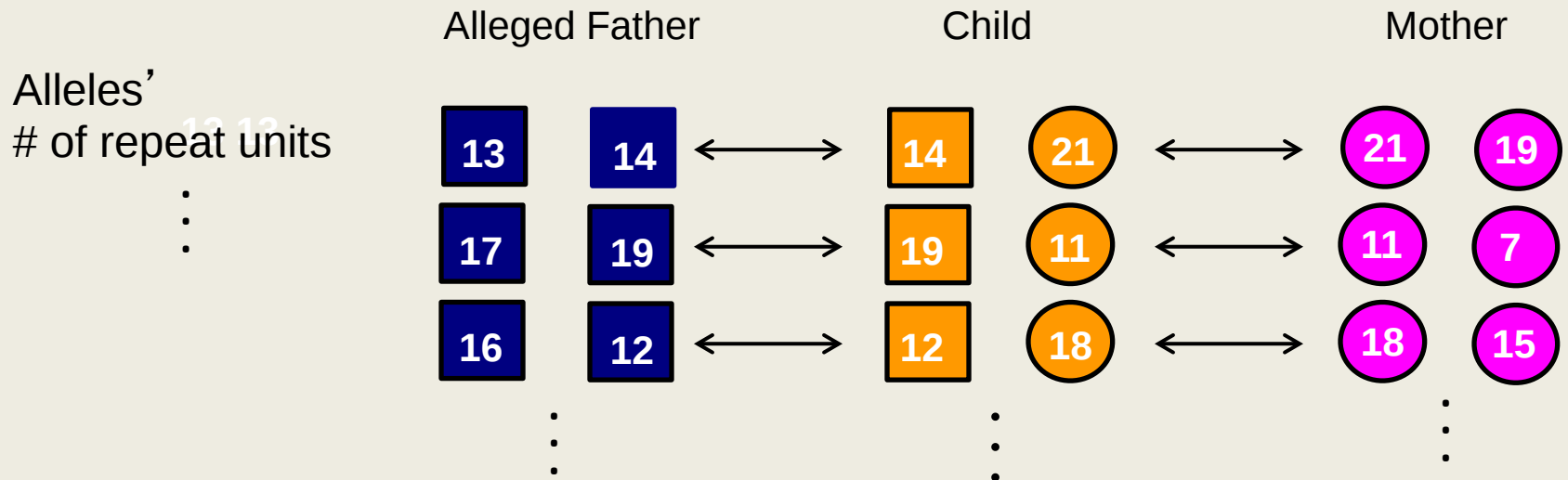
Family Relationship Premise

History of DNA Testing



Family Relationship Premise

Parentage DNA Analysis

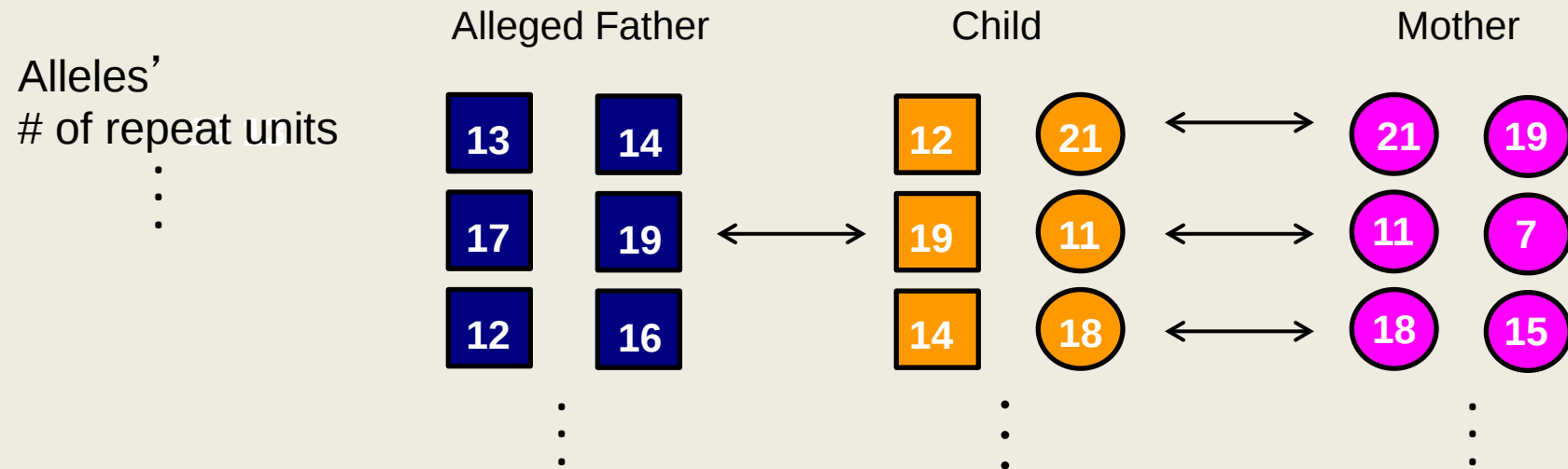


Inclusion-

he is the biological father

Family Relationship Premise

Parentage DNA Analysis



Exclusion-

he is not the biological father



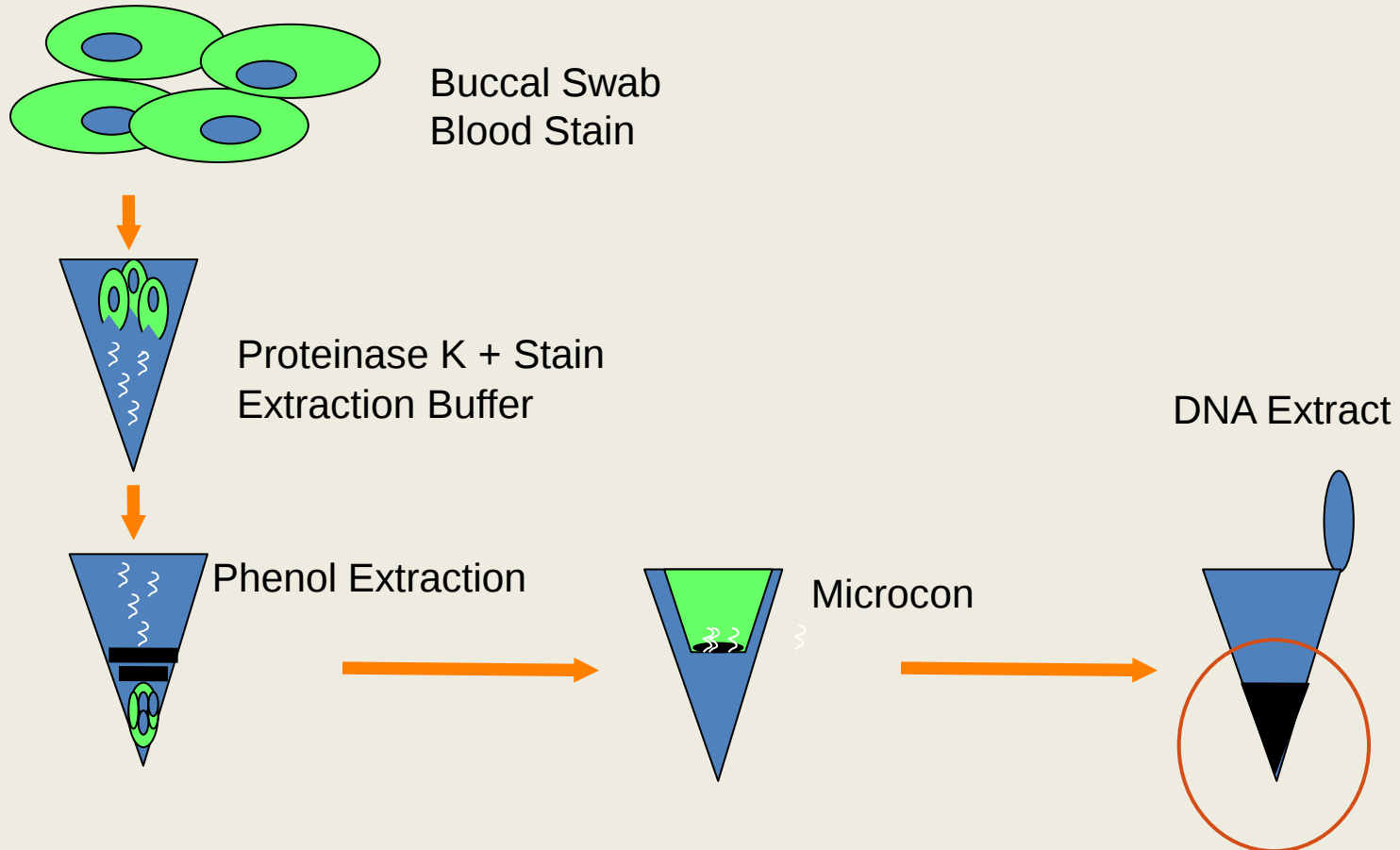






DNA Testing Process

Extract DNA



DNA Testing Process

Amplify STR Loci

After the DNA has been extracted and quantified, the STR loci are amplified in thermal cycler using the Polymerase Chain Reaction (PCR).

Thermal Cycler



Amplification



DNA Testing Process

Generate a DNA Profile

A DNA profile is obtained using the 310, or 3100 gene fragment analyzers.

ABI Prism 310 CE



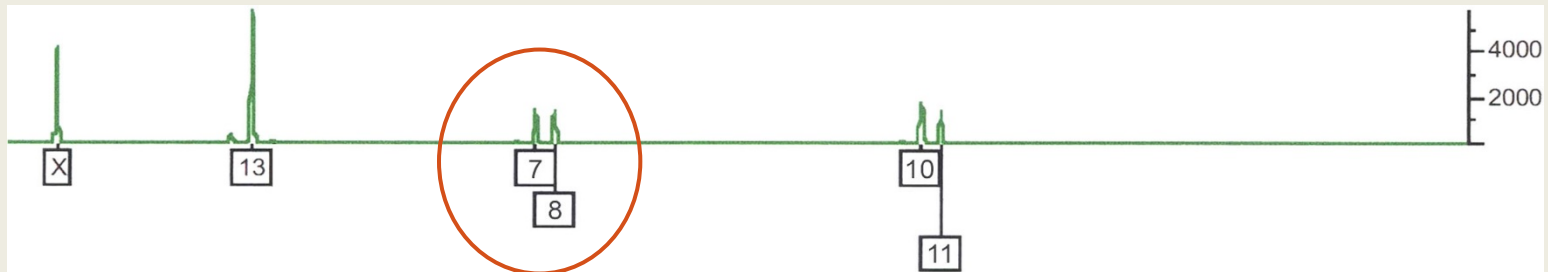
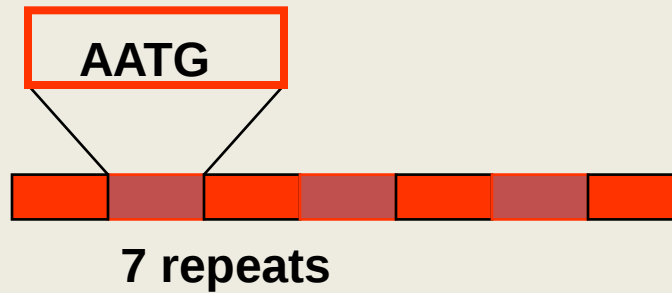
ABI Prism 3100 CE



STR DNA Analysis

Reading a DNA Profile

*Short
Tandem
Repeats
(STR)*



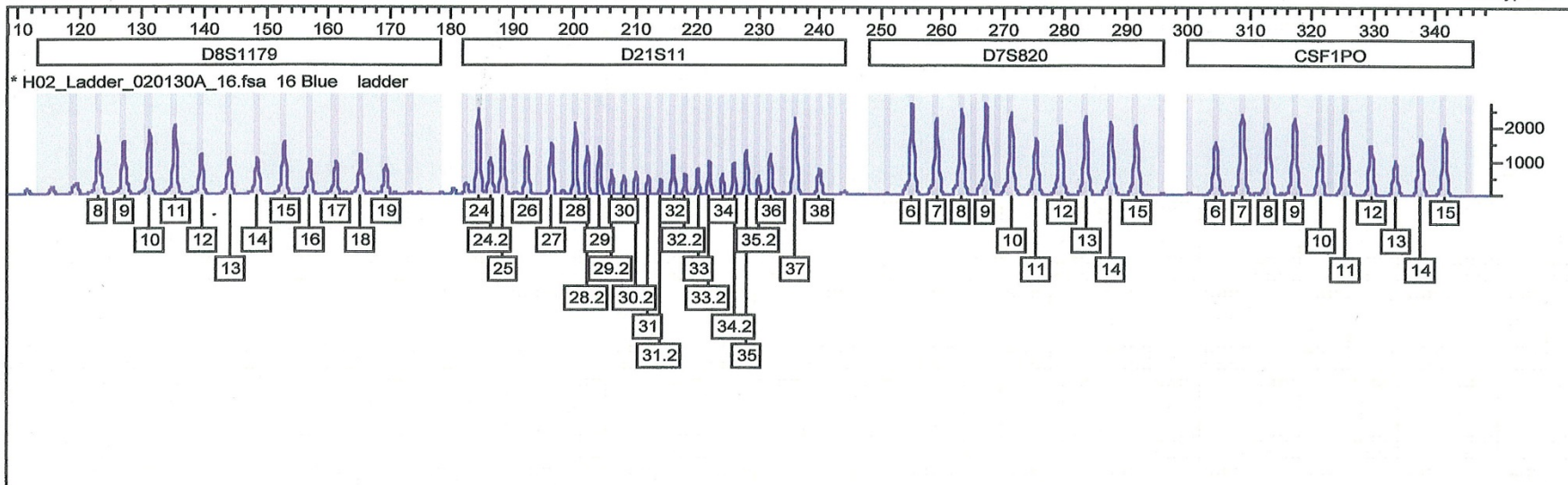
STR DNA Analysis

Common Alleles Detected



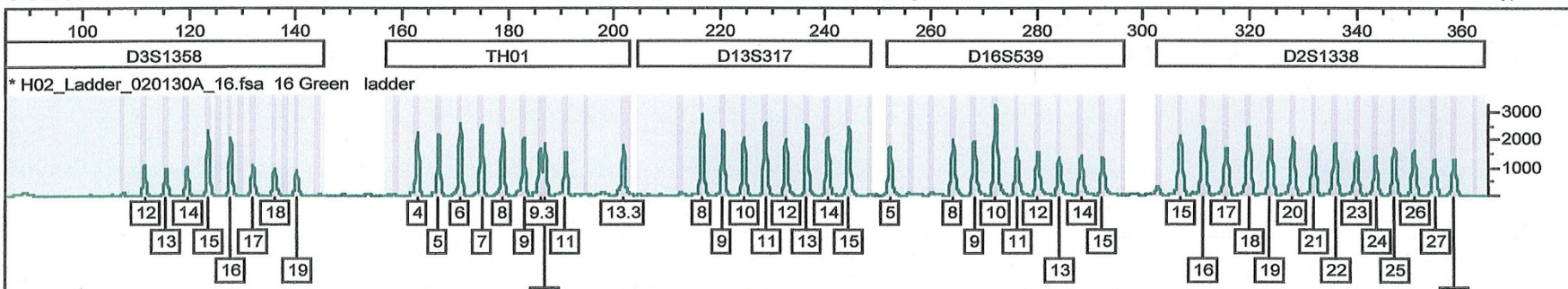
BLUE Ladder
Licensed to Dr. Richard Lee, DNA Diagnostics

8:22:37 AM Mon, Feb 18, 2002
Genotyper® 3.7



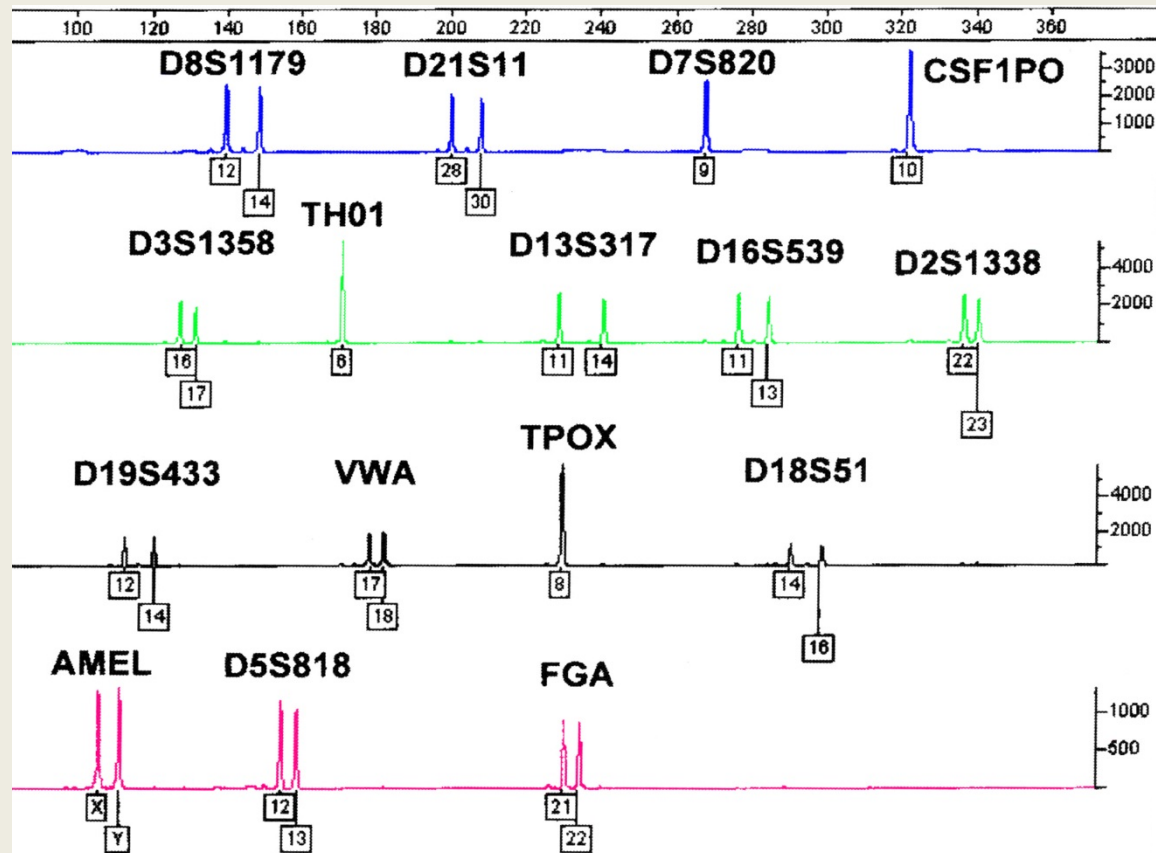
GREEN Ladder
Licensed to Dr. Richard Lee, DNA Diagnostics

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Genotyper® 3.7



STR DNA Analysis

STR Profile



Databases

Databases are composed of random, unrelated individuals that self identify their ethnicity

Most databases contain at least 200 data points (100 individuals)

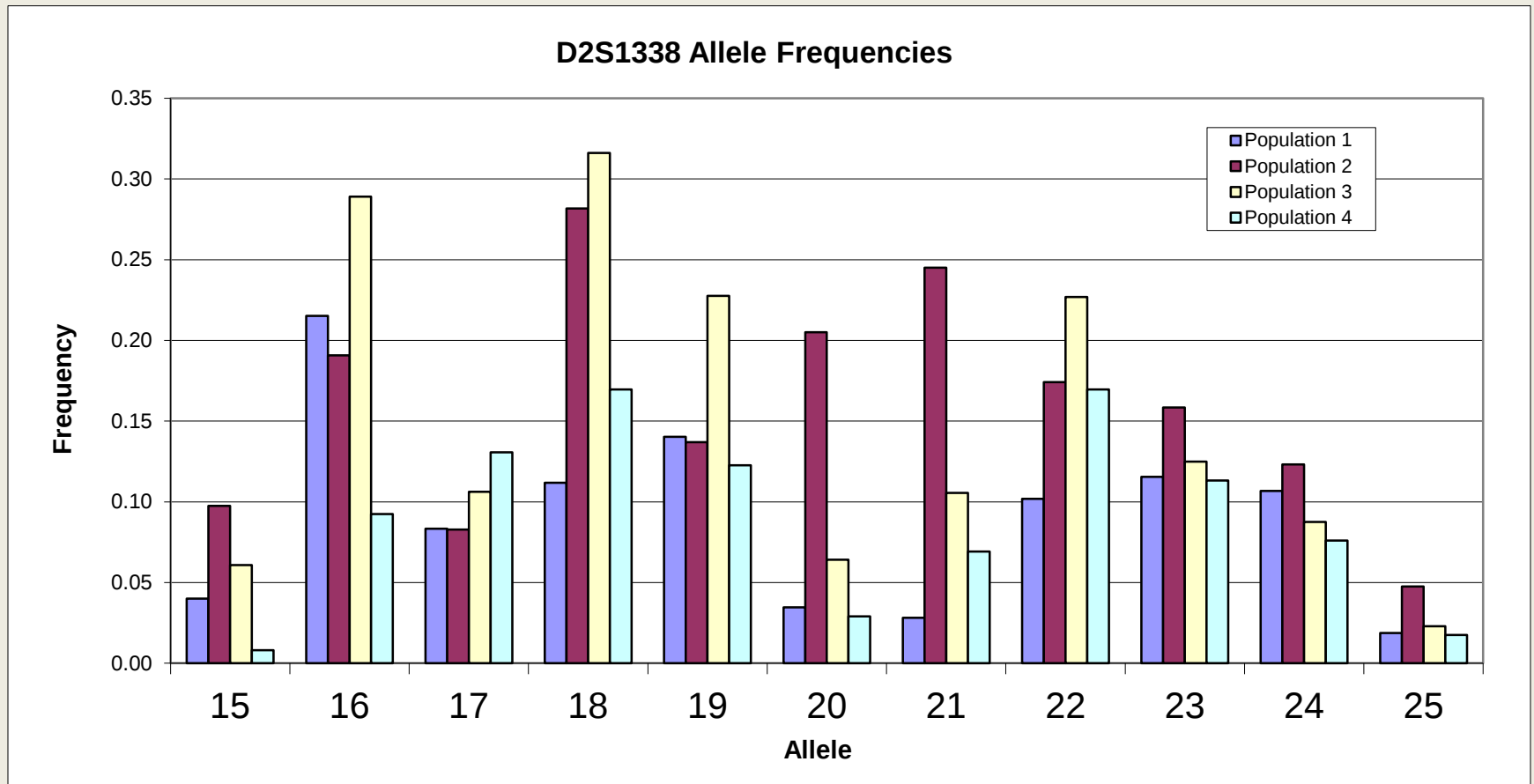
Databases are checked to determine whether they meet the assumptions of the Hardy-Weinberg equation

Databases for the same ethnicity may vary slightly, but usually produce results that are not significantly different

Allele Frequency at D2S1338

- | Allele | Population 1 | Population 2 | Population 3 | Population 4 |
|--------|--------------|--------------|--------------|--------------|
| 15 | 0.0400 | 0.0975 | 0.0607 | 0.0081 |
| 16 | 0.2152 | 0.1908 | 0.2890 | 0.0923 |
| 17 | 0.0834 | 0.0828 | 0.1062 | 0.1307 |
| 18 | 0.1118 | 0.2816 | 0.3161 | 0.1696 |
| 19 | 0.1403 | 0.1370 | 0.2276 | 0.1225 |
| 20 | 0.0346 | 0.2050 | 0.0642 | 0.0290 |
| 21 | 0.0282 | 0.2450 | 0.1055 | 0.0691 |
| 22 | 0.1018 | 0.1741 | 0.2269 | 0.1696 |
| 23 | 0.1154 | 0.1584 | 0.1249 | 0.1132 |
| 24 | 0.1067 | 0.1232 | 0.0874 | 0.0761 |
| 25 | 0.0187 | 0.0476 | 0.0229 | 0.0174 |

D3S1338



Paternity Index, P_I

- Likelihood Ratio
- Ratio between X and Y , $P_I = X / Y$
- X = chance the alleged father contributed the paternal allele to the child (0, 0.5, or 1)
- Y = chance a random, unrelated individual contributed the paternal allele to the child; the frequency of the paternal allele in the ethnic group of interest

Mutations

Observed when child contains alleles not present in either biological parent.

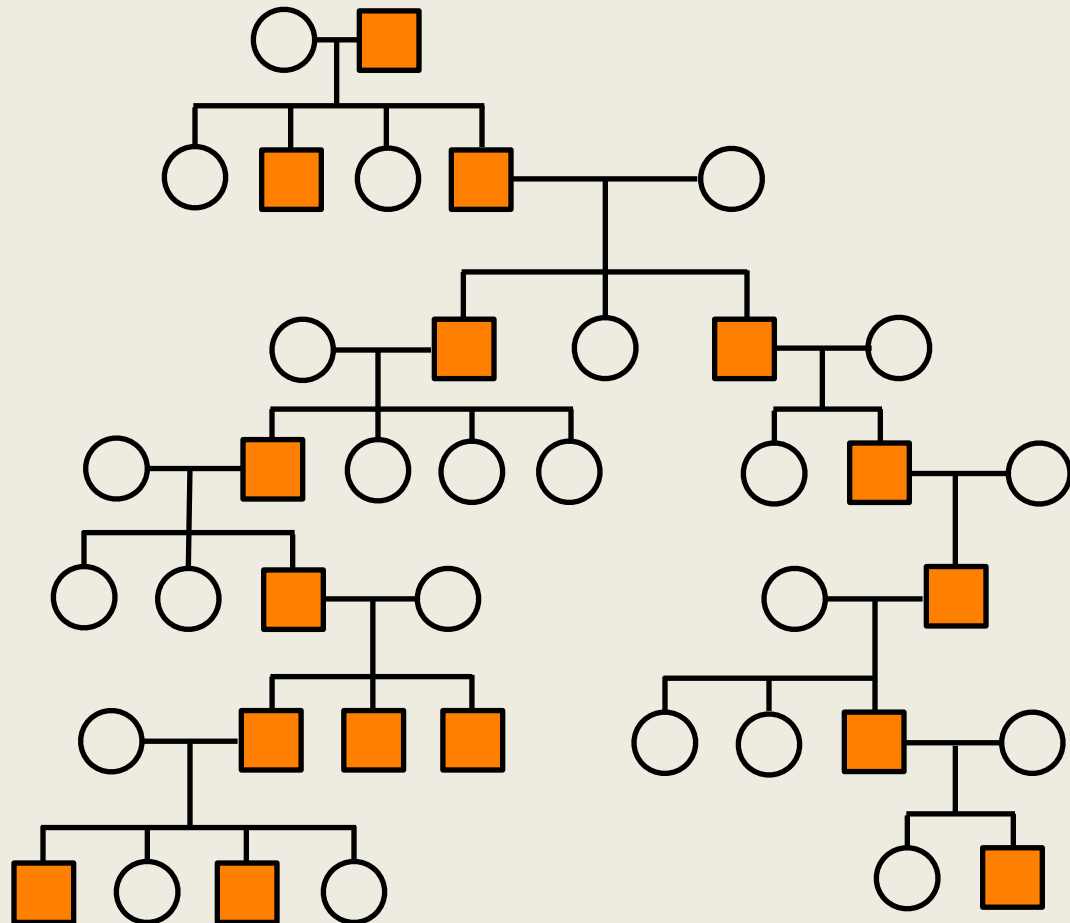
Often result of recombinational event in meiosis in the production of sperm or eggs.

With STR analysis, often leads to the presence of an allele one (1) repeat unit larger or smaller than the parent.

Non-Autosomal DNA Markers

Y-STR DNA

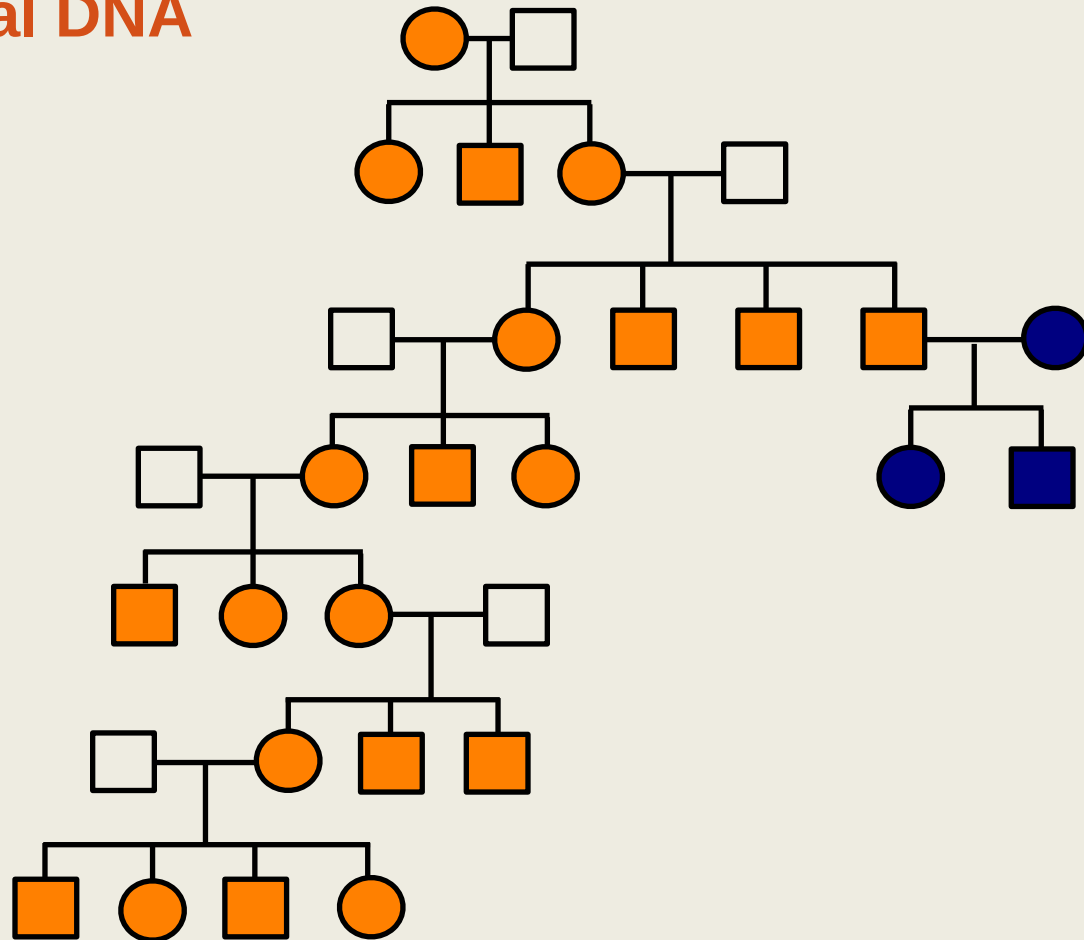
Pedigree
(7 generations)



Non-Autosomal DNA Markers

Mitochondrial DNA

Pedigree (7 generations)



Chimera Case

- AF excluded using a buccal sample
- Same AF included with semen sample
- DNA profile a mixture
- Origin of all alleles identified

Chimera Case

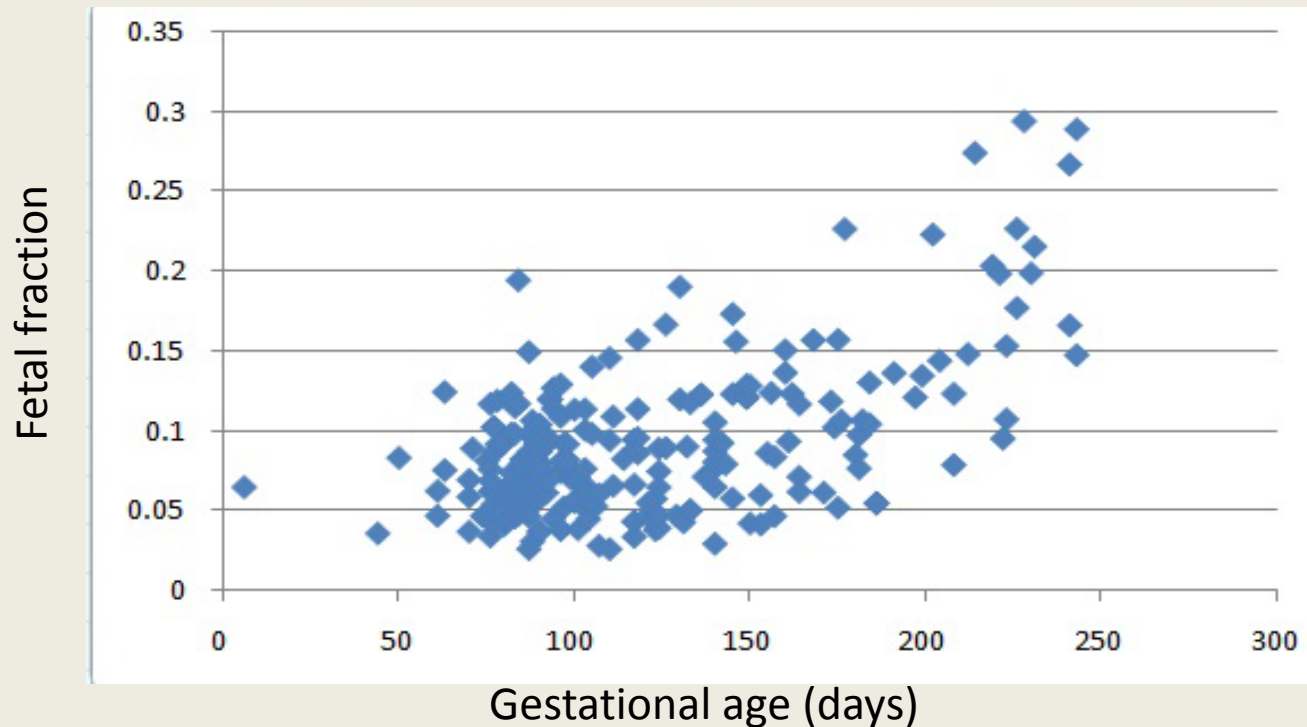
- Embryonic cells of dizygotic twins fused early in development
- Sperm developed in chimeric AF was of four different types
- AF falsely excluded when the buccal swab sample was used

Non-Invasive Prenatal

- Determine paternity as early as 9 weeks gestation from a blood sample of the pregnant woman
- Next generation sequencing of 2688 SNP loci
- Bioinformatics used to sort data and determine paternity

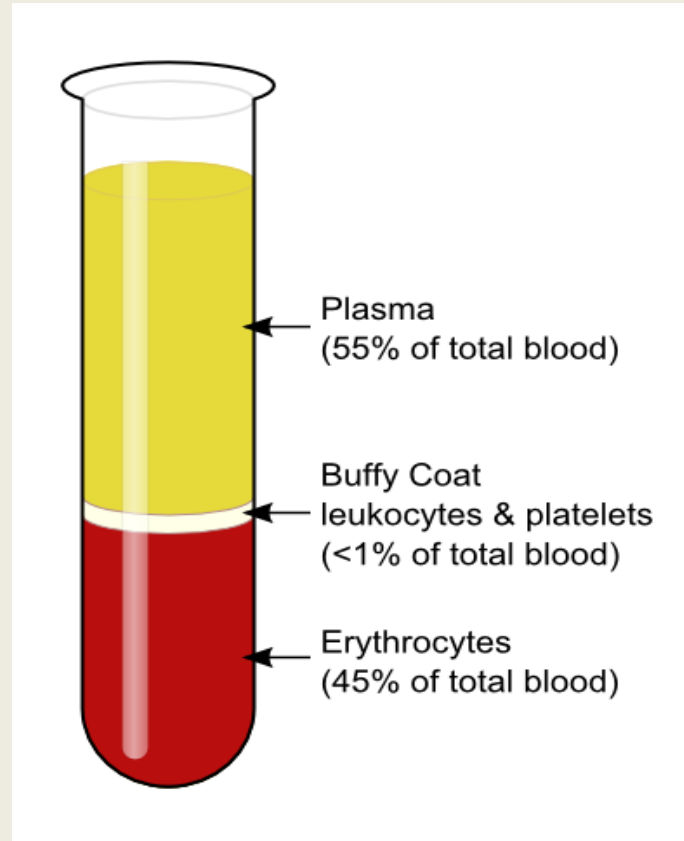
Fetal Fraction Varies Widely Across Individuals

Observed fetal fraction in processed maternal plasma



Non-Invasive Prenatal Testing

– Blood Sample



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Shaping the Future for Children

Advances in DNA Testing: The Art and Science of Parentage

Dr. George C. Maha

wicsec
western interstate CHILD SUPPORT enforcement council
Dalworthington

Legal Issues & Technical Issues

- Chain of Custody
- Twins
 - Twin Children
 - Twin Alleged Fathers
- Fraud

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

[...], a young woman gave birth to a daughter. Brueggeman was the father of this child. Initially, Brueggeman acknowledged the child as his, but he later developed doubts about his paternity and stopped supporting his child. [...], the mother of the child applied for public assistance. She identified Brueggeman as her child's father, and she assigned her child support rights to the State. [...], the Child Support Enforcement Division (CSED) filed a paternity action against Brueggeman.

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

Brueggeman privately arranged for genetic testing at the Valley Phlebotomy Service in Wasilla. [...], he directed the mother and child to go there so that the service could obtain genetic samples from them. The next day, Brueggeman had his friend (and later co-defendant) David Hood go to Valley Phlebotomy and pose as Brueggeman. Hood identified himself as Brueggeman, presented the service with Brueggeman's Alaska State identification card, signed his name as Brueggeman, and provided a sample for paternity testing.

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

Not surprisingly, genetic testing of this sample excluded "Brueggeman" (i.e., Hood) as the father of Brueggeman's child. Brueggeman filed the test results with the court and asked the court to dismiss the paternity case. But the State was not satisfied that Brueggeman was in fact the person who provided the sample, so the State asked the court to order another paternity test. Brueggeman opposed the motion and claimed that he was being harassed because he had already taken a paternity test.

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

Over Brueggeman's objection, the superior court granted the State's motion for another test. Brueggeman missed two scheduled appointments for this second test, the second on advice of counsel. Finally, the court granted the State's motion to compel Brueggeman to appear for testing.

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

While this litigation was proceeding, Brueggeman condoned and participated in a letter-writing campaign conducted by a woman who was a friend of his family. The aim of this campaign apparently was to convince various state legislators that the CSED was harassing an innocent citizen, so that these legislators would pressure the CSED and the Attorney General's Office to drop the paternity suit against Brueggeman. These letters contained language and assertions personally approved by Brueggeman. The letters accused the CSED of harassing Brueggeman by requesting a second paternity test. The letters also asserted that the child's mother was a promiscuous woman who had falsely accused Brueggeman of being the father of her child because she was poor and needed money.

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

Ultimately, Brueggeman (now represented by counsel) notified the court that he would submit to the additional paternity test under protest. However, Brueggeman once more conspired with Hood to defraud the court, the State, and the child's mother. [...], Hood appeared for the court-ordered test and once again identified himself as Brueggeman. He presented Brueggeman's identification and signed his name as Brueggeman. Hood then provided a sample for genetic testing.

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

And again, the genetic testing of Hood's body sample showed that "Brueggeman" (i.e., Hood) was not the father of Brueggeman's daughter. This time, however, the people who administered the test took Hood's photograph and thumb print. When the State showed this photograph to the mother of the child, she identified the person in the photograph as Hood, not Brueggeman.

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

Meanwhile, Brueggeman moved for summary judgment in the paternity case based on the result of the second genetic test. The State opposed Brueggeman's motion, offering an affidavit from the mother in which she identified Hood as the person in the photograph taken at the court-ordered test. Rather than admitting his fraud, Brueggeman answered with his own affidavit in which he falsely claimed that he had twice appeared and been tested, both at the original genetic test [...] and at the second, court-ordered test [...]. To further bolster his position, Brueggeman induced Hood to sign a false supporting affidavit. In his supporting affidavit, Hood falsely claimed that he had never taken a paternity test and, more specifically, that he had never done so for Brueggeman. .

Chain of Custody

State v. Brueggeman, 24 P.3d 583 (Alaska Ct. App. 2001)

In the face of these competing affidavits, the court ordered both Brueggeman and Hood to provide genetic samples and fingerprints. This third round of genetic testing confirmed that the genetic samples from the two prior tests could not have come from Brueggeman (and had probably come from Hood). The testing also showed that Brueggeman was the father of the child. Finally, the court-ordered fingerprinting showed that the thumb print taken at the time of the second test [...] was Hood's. Based on these facts, a grand jury indicted Brueggeman on one count of scheme to defraud, two counts of tampering with physical evidence, and four counts of perjury. The State also filed an information charging Brueggeman with attempted criminal non-support.

Imposters

- Laboratory is photographing test participants for your use
- If you retest make sure all the test participants are positively identified by someone who knows the participant.

Chain of Custody

J Forensic Sci, March 2007, Vol. 52, No. 2
doi:10.1111/j.1556-4029.2006.00373.x
Available online at: www.blackwell-synergy.com

CASE REPORT

Luis J. Martinez-Gonzalez,¹ M.S.; Jose A. Lorente,¹ M.D., Ph.D.; Esther Martinez-Espin,¹ M.S.; J. Carlos Alvarez,¹ Ph.D.; Miguel Lorente,¹ M.D., Ph.D.; Enrique Villanueva,¹ M.D., Ph.D.; and Bruce Budowle,² Ph.D.

Intentional Mixed Buccal Cell Reference Sample in a Paternity Case*

ScienceDaily

Your source for the latest research news

Web address:

[http://www.sciencedaily.com/releases/2008/04/
080410131603.htm](http://www.sciencedaily.com/releases/2008/04/080410131603.htm)

DNA Paternity Test Almost Fooled: Man Put Someone Else's Saliva In His Mouth

Twin Children

Fraternal twins were tested. The alleged father was excluded as the biological father of one twin. For the other twin he had a paternity index of 11,734,738 to 1 (Probability of paternity of 99.99+%). The trial court dismissed the petition to establish paternity because, in part, of a fear “that there might be disparity created between developing twin siblings, one of whom had [an] identifiable father and the other of whom did not.” Reversed. *Commissioner of Social Serv. v. Hector S.*, 628 N.Y.S.2d 270 (A.D. 1 Dept. 1995).

Twin Children

Man Fathered Only One Twin, Judge Says

Charles Toutant, New Jersey Law Journal

May 7, 2015

Twin Children

The twins' mother, identified as T.M., was in a relationship with A.S., and she initially identified him to [...] Social Services as the father of the twins when she applied for benefits, according to [the judge]. The board filed an application seeking to establish paternity of A.S. and to make him pay child support. But the mother testified in the case that she had sexual intercourse with A.S. and with another, unidentified man within the span of a week.

Twin Children

[The judge] ruled the man, identified in court documents as A.S., must pay child support only for the twin who was determined through a DNA test to be his offspring, but not for the other. [...] the DNA paternity test used in the case, conducted by Laboratory Corp. of America, accurately and reliably established the father's paternity for only one of the twins.

The ruling is one of first impression in New Jersey and is among only a handful of similar reported cases nationwide, according to the opinion.

Twin Children

- The ruling reinforces the well-settled law that the right to child support belongs to the child and that the obligation to pay support belongs to the parent, said Jeralyn Lawrence, chair of the family law section of the New Jersey State Bar Association. The judge correctly found that the father is obligated to pay child support for his own child, but not for the other twin, Lawrence said.
- Lawrence said the case illustrates the need for practitioners to understand the little-known phenomenon of twins with separate fathers.

Identical Twin Alleged Fathers

Identical twins will be identical (See:
Illinois Department of Public Aid
Masinelli v. Whitworth, 652 N.E.2d
458 (Ill.App. 4 Dist. 1995))

Identical Twin Alleged Fathers

**Finding the needle in the haystack:
Differentiating “identical” twins in paternity
testing and forensics by ultra-deep next
generation sequencing.**

Weber-Lehmann, J., et. al. Forensic Science International:
Genetics 9 (2014) 42–46

Fraud

- Alterations / Forgeries
 - Laboratory Reports
 - Letters
 - Child Support Orders
- New AABB Standard (January 2016) - *The facility shall have policies, processes, and procedures to evaluate and respond to possible altered or fabricated documents.*

Standard Conclusion - Inclusion

The alleged father cannot be excluded as the biological father of the child since they share genetic markers. Using the above systems, the probability of paternity is 99.99%, as compared to an untested, unrelated man of the Caucasian population.

Standard Conclusion - Exclusion

The alleged father is excluded from paternity in the following systems: D3S1358, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, TH, TPOX. These results indicate that the alleged father is not the biological father of the child.

Fraudulent Conclusion

The alleged father should be excluded as the biological father of the child. While there are some shared genetic markers in only part of the independent loci tested, the chances of probability of paternity from this Caucasian male tested are 79.25%. After carefully reviewing, a “recalculation” was performed, and since an error was found, the rightful total of paternity is 79.25%

Fraudulent Conclusion

The alleged father has established paternity in all of the systems above. These results indicate that the alleged father is the biological father of the child.

Fraudulent Conclusion

The alleged father is not excluded from paternity in the following systems:
D1S80, FES, VWF, and CYP19. Therefore, he is the biological father of the child.

Fraudulent Conclusion

The alleged father is included in above paternity. Therefore, test shows him to be the biological father of the child.

Fraudulent Conclusion

It have been concede that the alleged father is the Biological Father of the child. On a ratio scale; 1 to 4, the alleged father is 3.5%

Fraudulent Conclusion

Conclusions: The alleged father is excluded from paternity in the following system(s): None

It has been determined that he is the biological father of the child.

Fraudulent Conclusion

I further certify that the annex report was made in the regular course of business of this Laboratory. Due to Laboratory Test Interactions there has been an error in the conclusion of paternity. The chemical Depo-Provera Sterile Medroxyprogesterone acetate suspension given to the mother caused a chemical imbalance reaction in the following: D3S1358, VWA, and TPOX. Therefore I concluded that there was an error in the blood test result. This error has been rectified and therefore all other findings are considered normal.

Fraud

- Watch for reversals of report outcomes.
- Is the document's language typical of what you have seen in the past? If not, check its authenticity.
- Look at grammar and spelling
- Talk to the laboratory