Significance of DNA Profiling in Paternity cases

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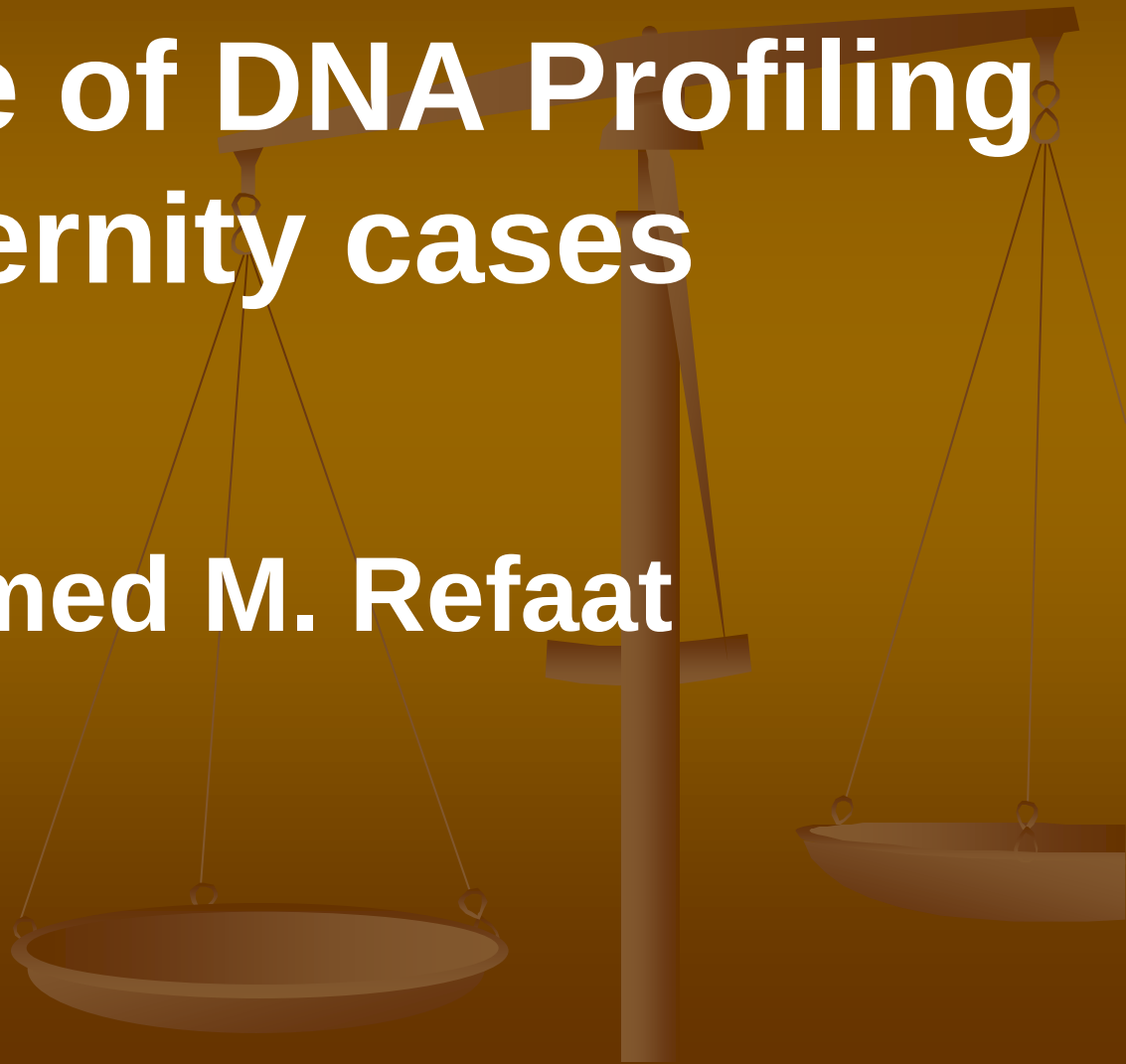
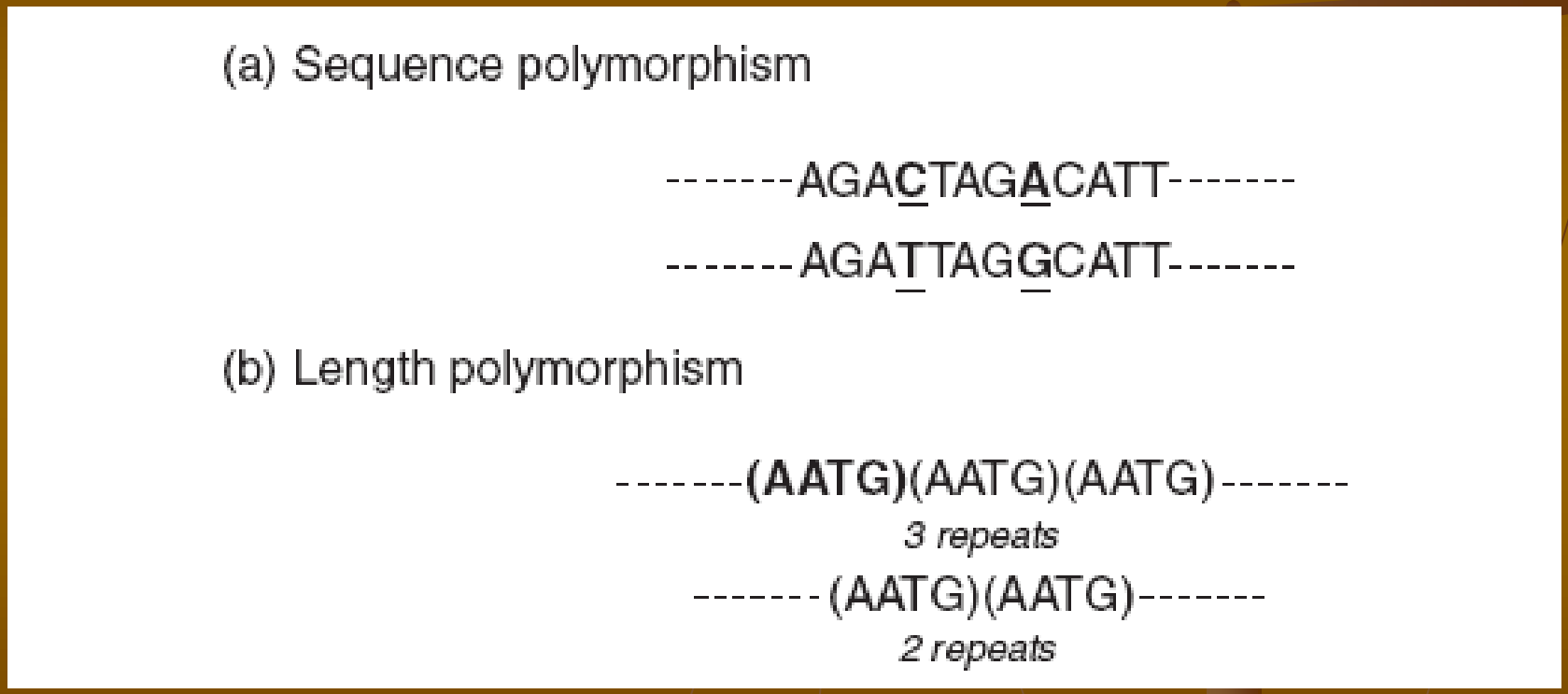
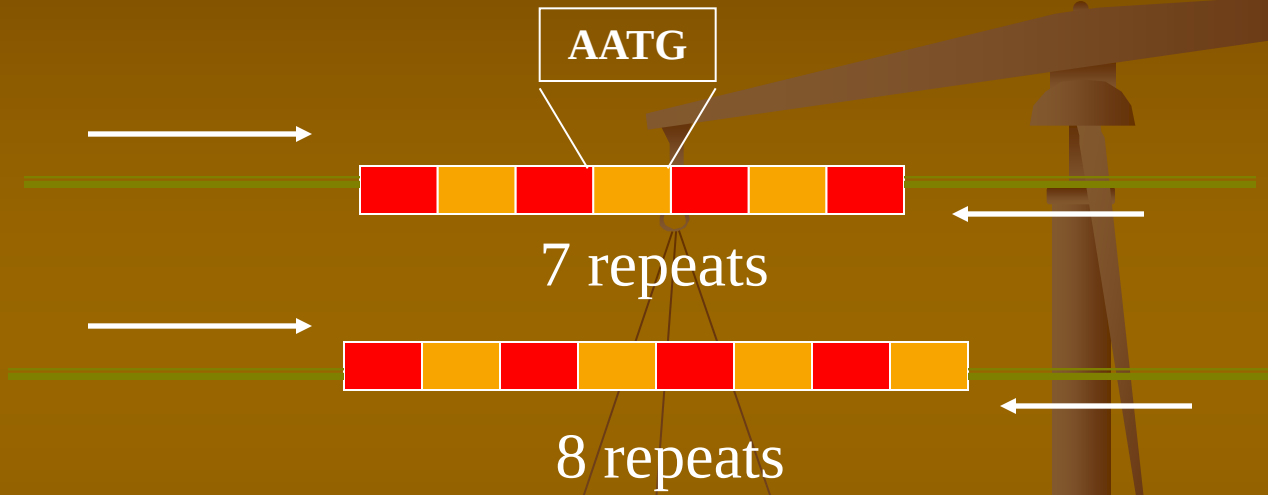


Types of DNA Polymorphism



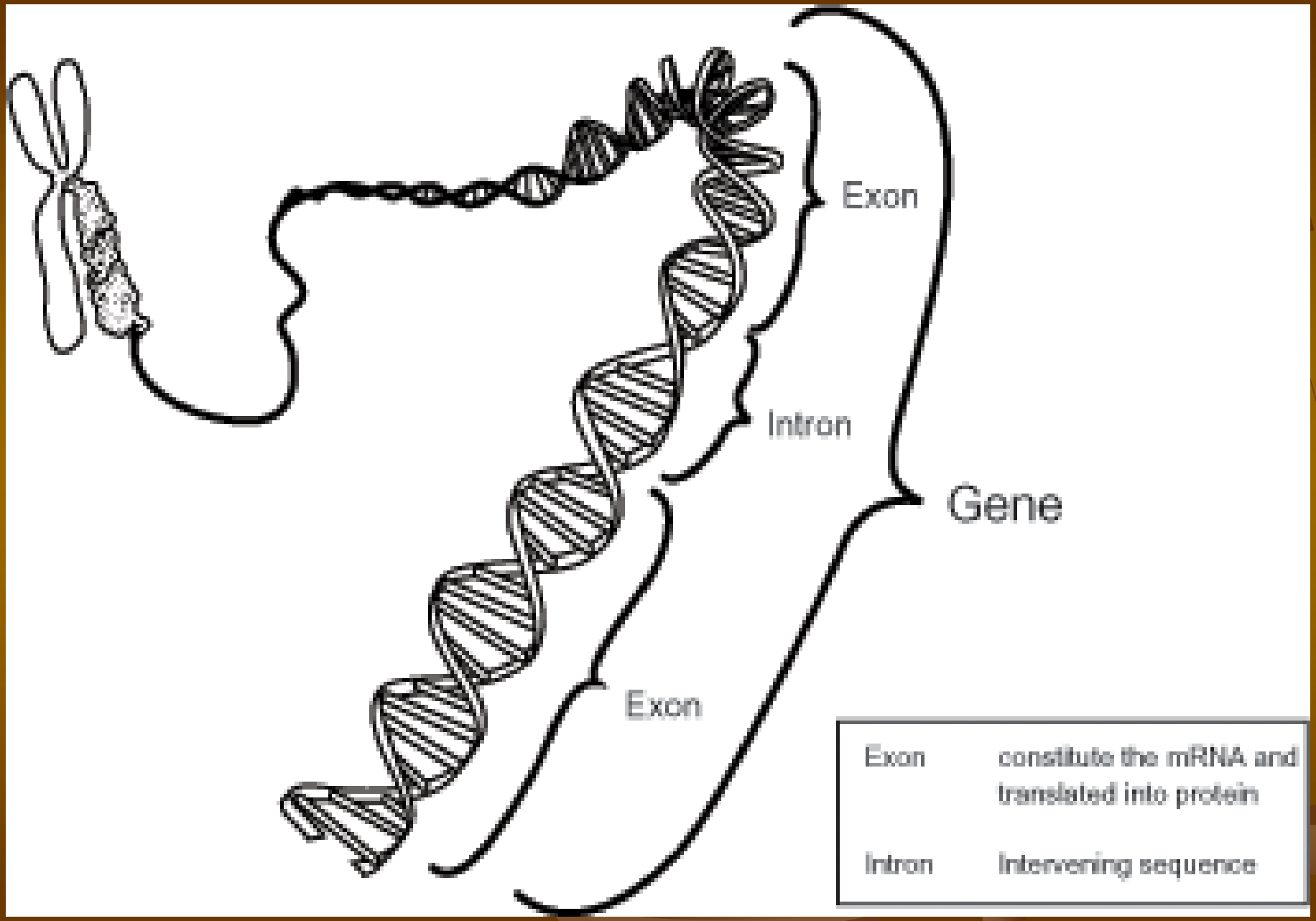
Short Tandem Repeats (STRs)

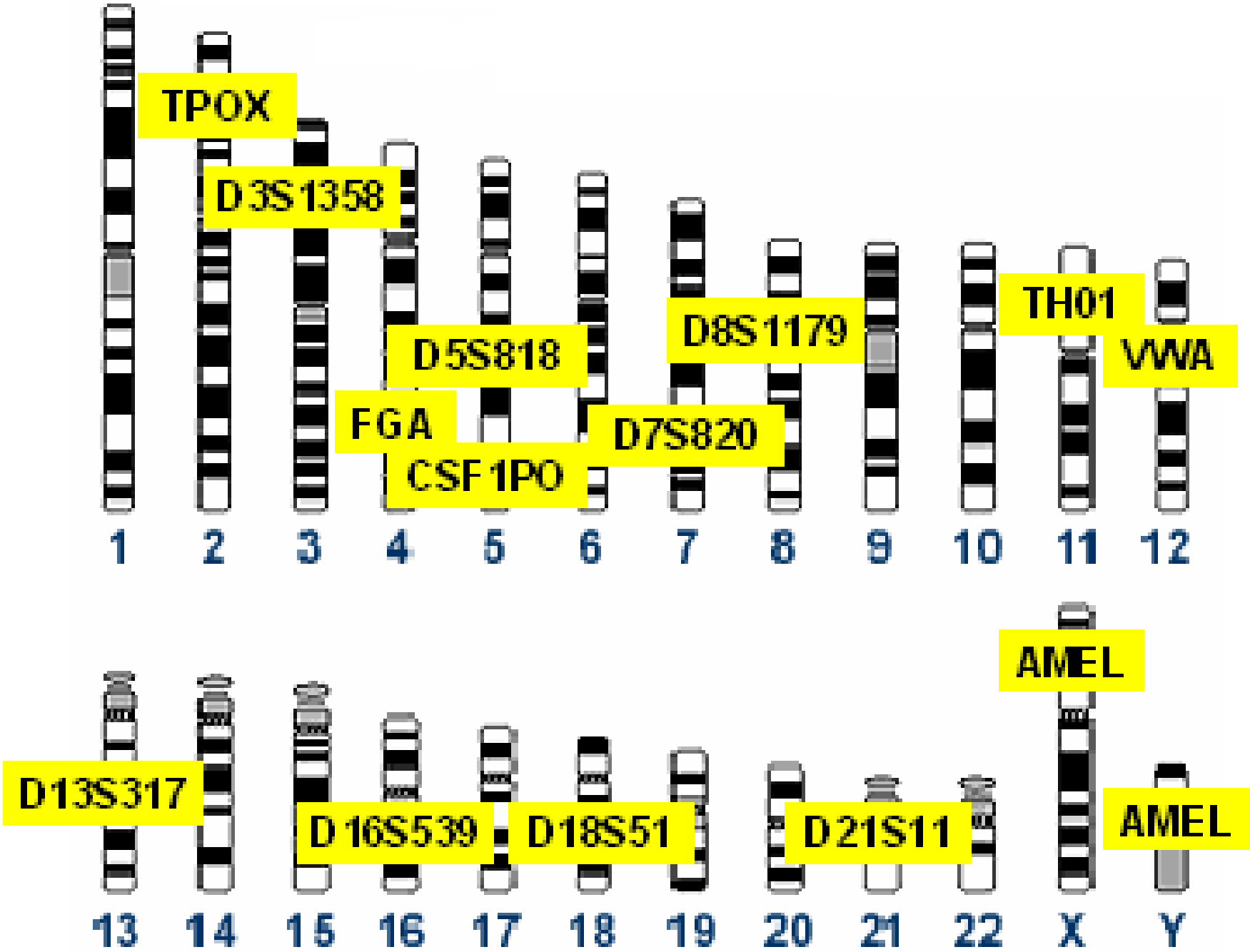


the repeat region is variable between samples while the flanking regions where PCR primers bind are constant

Homozygote = both alleles are the same length

Heterozygote = alleles differ and can be resolved from one another


Exon
Intron
Gene
Exon
Exon
constitute the mRNA and translated into protein
Intron
Intervening sequence


TPOX
D3S1358
D5S818
FGA
CSF1PO
D7S820
D8S1179
TH01
VWA
1
2
3
4
5
6
7
8
9
10
11
12
D13S317
D16S539
D18S51
D21S11
AMEL
AMEL
13
14
15
16
17
18
19
20
21
22
X
Y

Properties of STR Markers

- High discriminating power, usually > 0.9, with observed heterozygosity > 70%
- Separate chromosomal locations to ensure that closely linked loci are not chosen
- STRs are highly discriminating between unrelated and even closely related individuals.
- Low stutter characteristics
- Low mutation rate
- Predicted length of alleles that fall in the range of 90–500 bp with smaller sizes better suited for analysis of degraded DNA samples.

Advantages of STR Analysis Technique over traditional VNTR Analysis Technique:

1. The process can be used with degraded samples (since shorter fragments of DNA can be analyzed).
2. The PCR process permits analysis of extremely small amounts of DNA.
3. The potential number of loci is very large. This is particularly important if siblings or other relatives are involved.
4. The process is rapid; it may be completed in a day or two.
5. The system lends itself to multiplexing and automation.

Parentage Testing

The need to identify the parents of children is as old as history. A famos biblical example is that of two women coming to King Solomon and each arguing that a living infant was hers. Solomon's solution was to have a sword produced and threaten to cut the child in two, thus providing each mother with a half. One mother agreed to let the other have the child; the other mother agreed to let the child be killed. Solomon decided the 'true' mother to be the one who chose to give up the child rather than have it killed, and awarded the child to her.

Parentage identification has three forms:

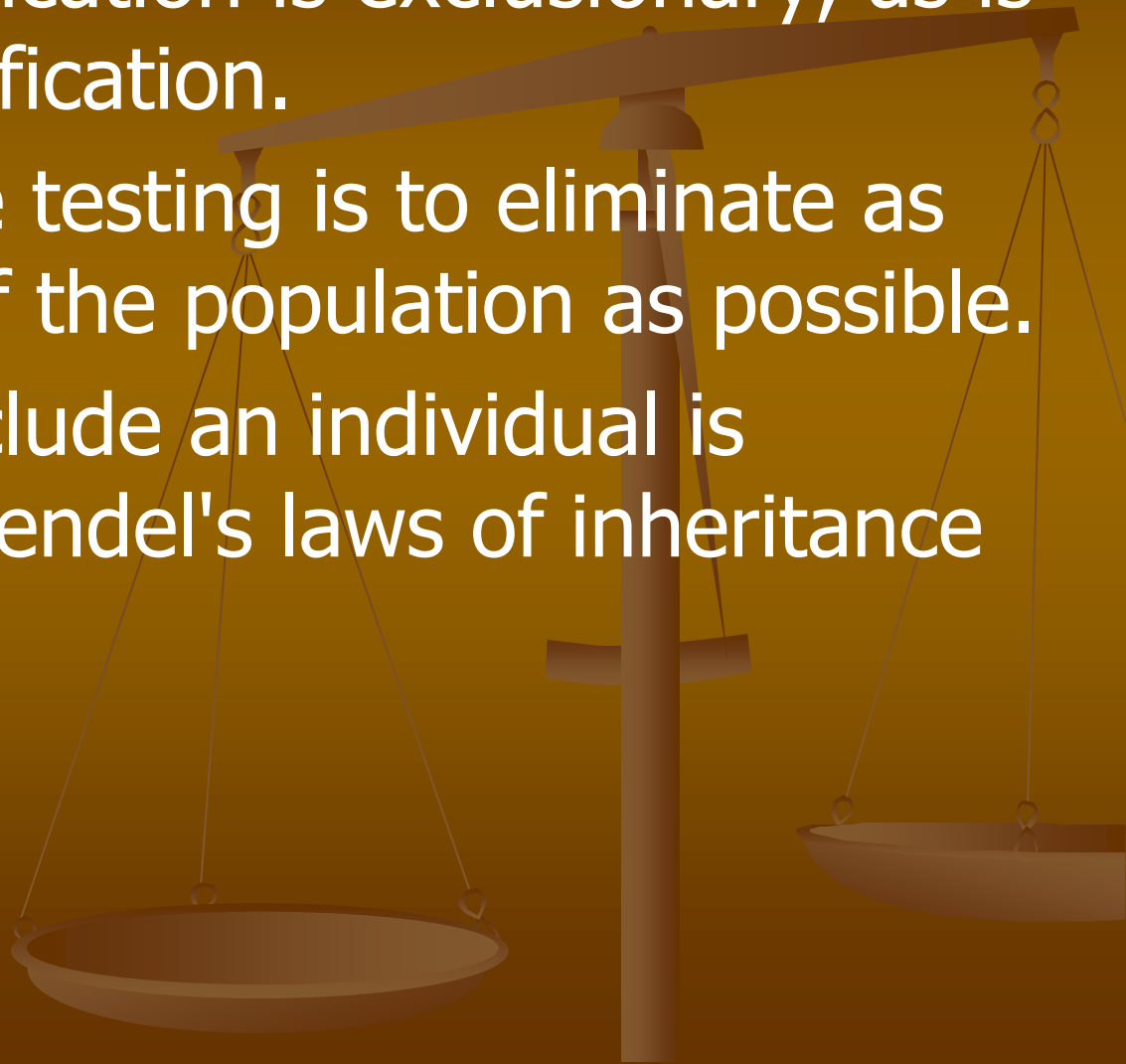
paternity identification, maternity identification and parentage identification.

Paternity identification is widely used for purposes of providing financial support to children. Paternity identification can also be important in cases of inheritance.

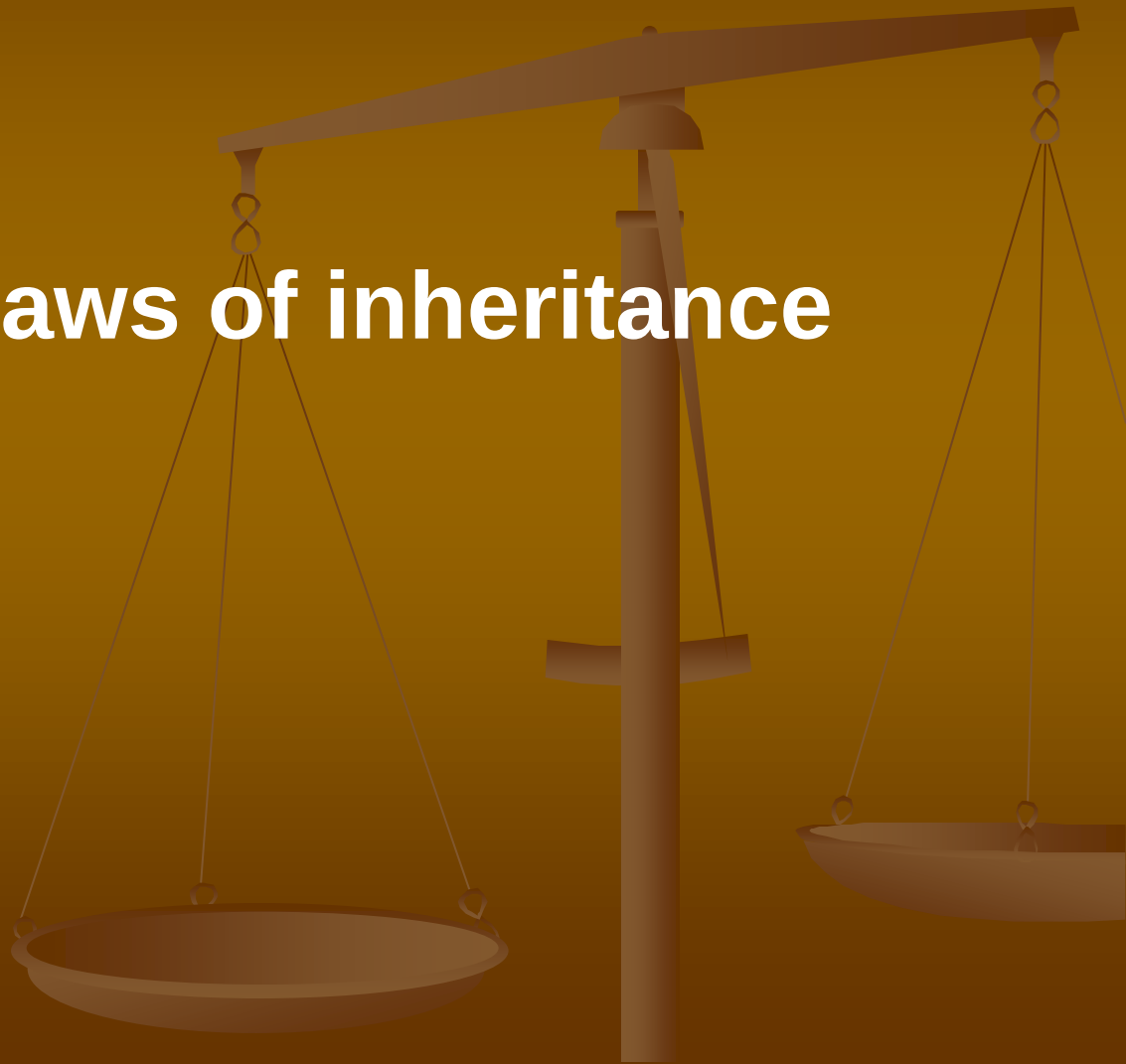
From the forensic standpoint, paternity identification can be important in cases of sexual assault in which a pregnancy occurs

Genetics of Parentage identification

- Parentage identification is exclusionary, as is all forensic identification.
- The object of the testing is to eliminate as large a portion of the population as possible.
- The ability to exclude an individual is determined by Mendel's laws of inheritance



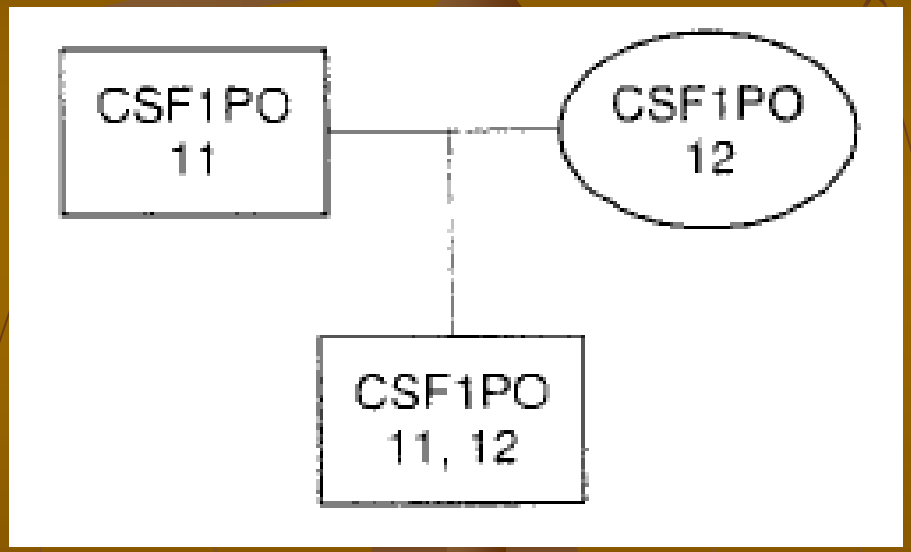
Mendel's laws of inheritance



Mendelian Laws of Parentage Testing

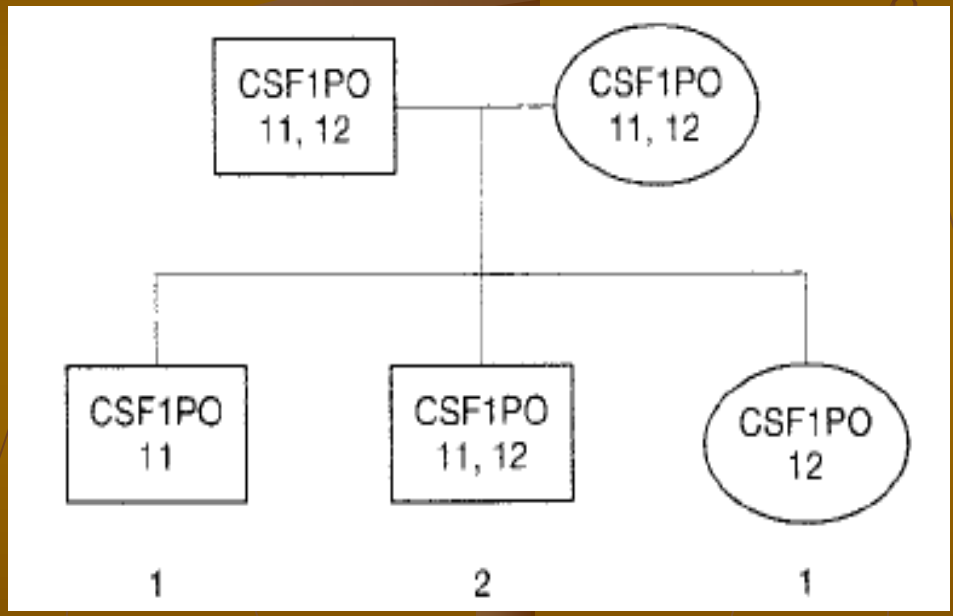
Mendel's First Law

- Mating of two parents homozygous (both alleles the same) for alleles 11 and 12 at locus CSF1PO.
- All children produced are heterozygous (both alleles different) for alleles 11 and 12.



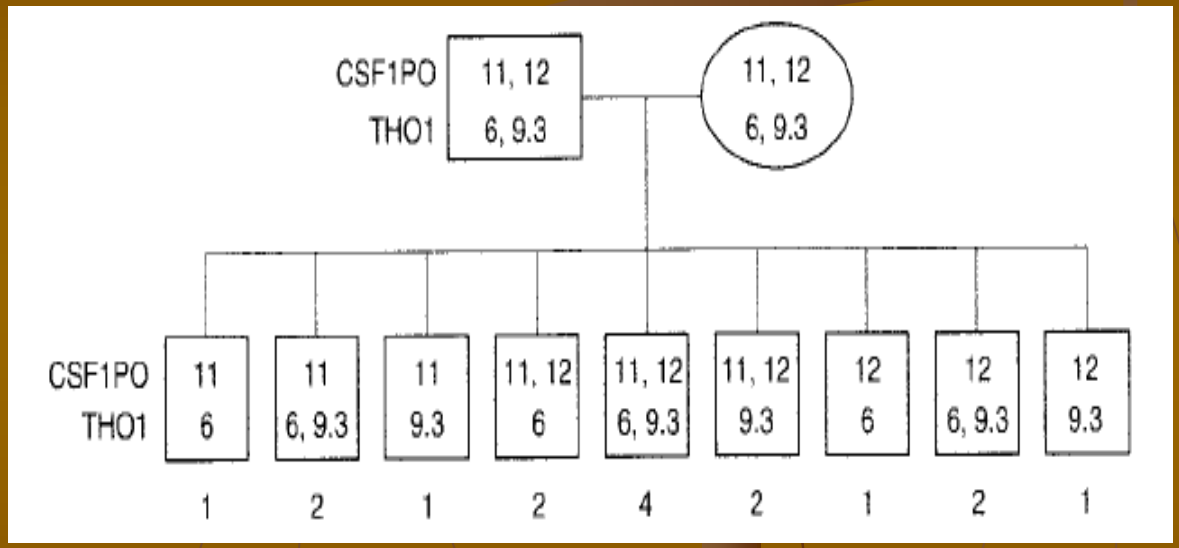
Mendel's Second Law

- Mating of two parents heterozygous for alleles 11 and 12 at locus CSF1PO produce offspring that are homozygous for CSF1PO 11, heterozygous CSF1PO 11,12 and homozygous CSF1PO 12 in the ratio of 1:2:1.



Mendel's Third Law (independent assortment).

- Mating of two parents heterozygous for two independent inherited traits: CSF1PO 11,12 and THO1 6,9.3.
- The expected ratio of the different types is represented under each child.



Paternity rules based on Mendel's laws

Paternity Rule 1

A child cannot have a genetic marker which is absent in both parents

	Inclusion	*Exclusion*
Mother	VWA 16,**17**	VWA 16,**17**
Child	VWA ***15***,**17**	VWA **17**,***18***
Alleged father	VWA ***15***,17	VWA 15,**16**

Paternity Rule 2

A child must inherit one of a pair of genetic markers from each parent

	Inclusion	*Exclusion*
Mother	VWA 16,**17**	VWA **16,17**
Child	VWA ***16,17***	VWA ***16,17***
Alleged father	VWA ***16***,18	VWA 15,18

Paternity Rule 3

A child cannot have a pair of identical genetic markers unless both parents have the marker

	Inclusion	*Exclusion*
Mother	VWA 16,**17**	VWA 16,**17**
Child	VWA ***17***	VWA ***17***
Alleged father	VWA ***17***,18	VWA 15,18

Paternity Rule 4

A child must have the genetic marker which is present as an identical pair in one parent

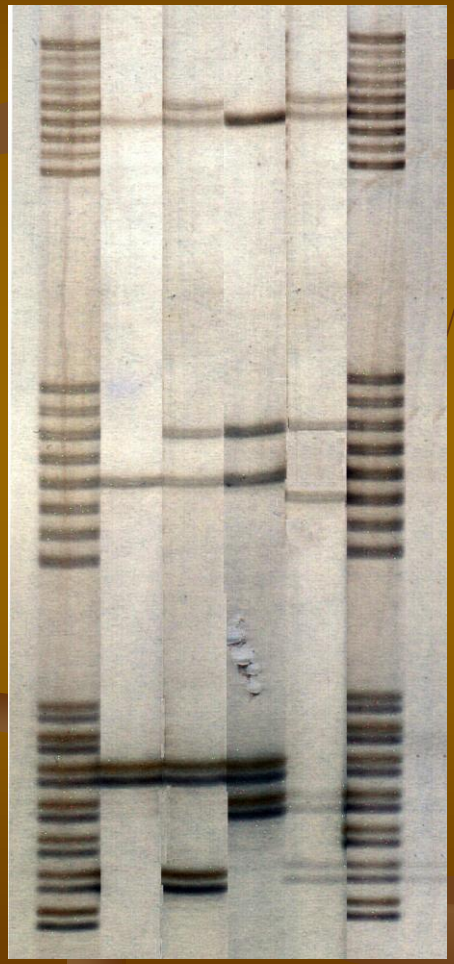
	Inclusion	*Exclusion*
Mother	VWA **18**	VWA **18**
Child	VWA ***17***,**18**	VWA ***17***,**18**
Alleged father	VWA ***17***	VWA 16

Paternity case

- The parents of a new born twin (two daughters) claiming at the court that the hospital in which the delivery of the twin has occurred had exchanged one of both infants with another.
- This is because a nurse in the hospital had said to both parents that their twin was a daughter and a son.
- The court decide to perform DNA analysis to prove whether the allegation of both parents is correct or not.

Paternity case …

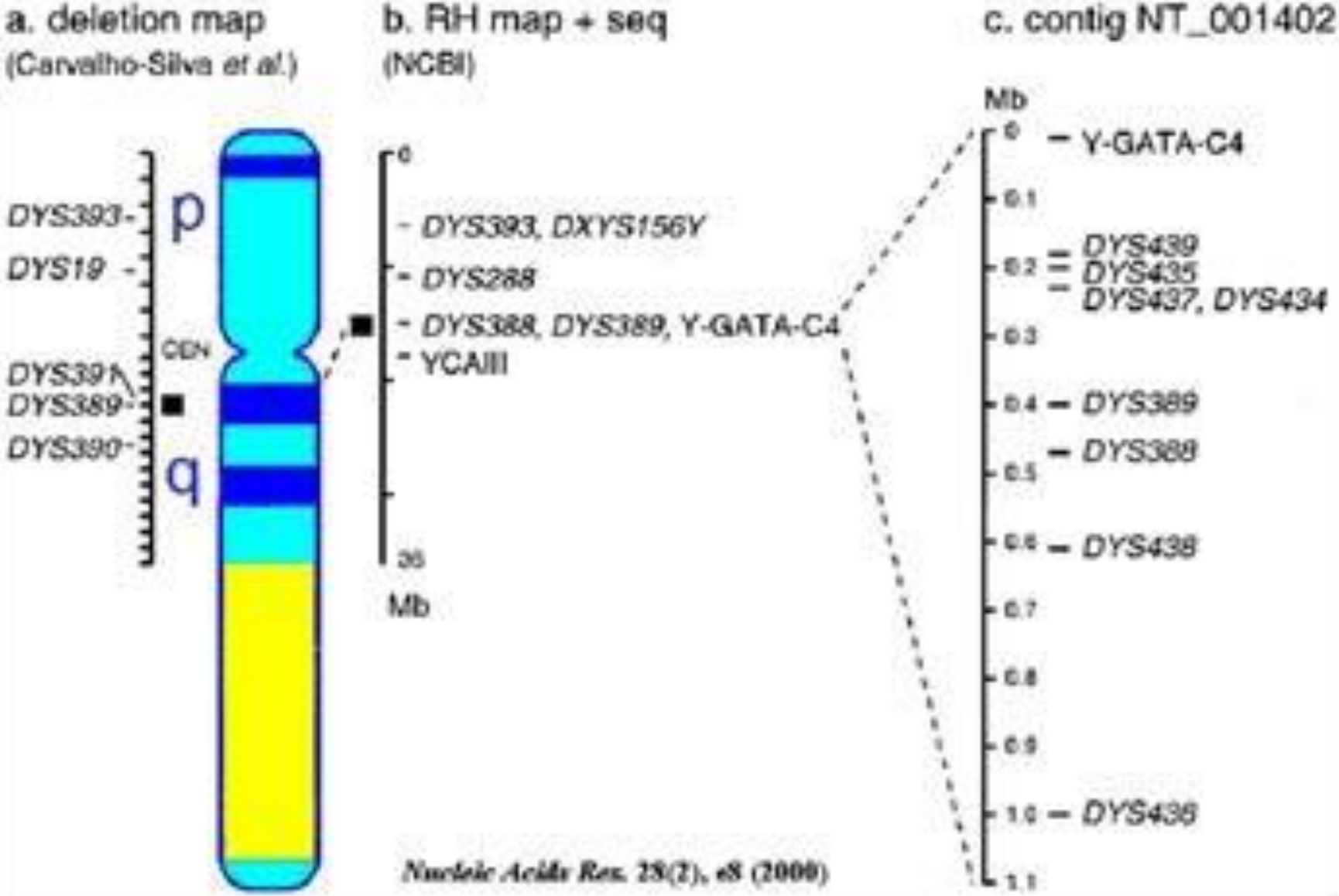
- DNA profiling using STR technology is done for the parents and the two infants.
- The conclusion of analysis had proved that the allegation of both parents was incorrect and both daughters are attributed to their biological parents submitting the complaint.



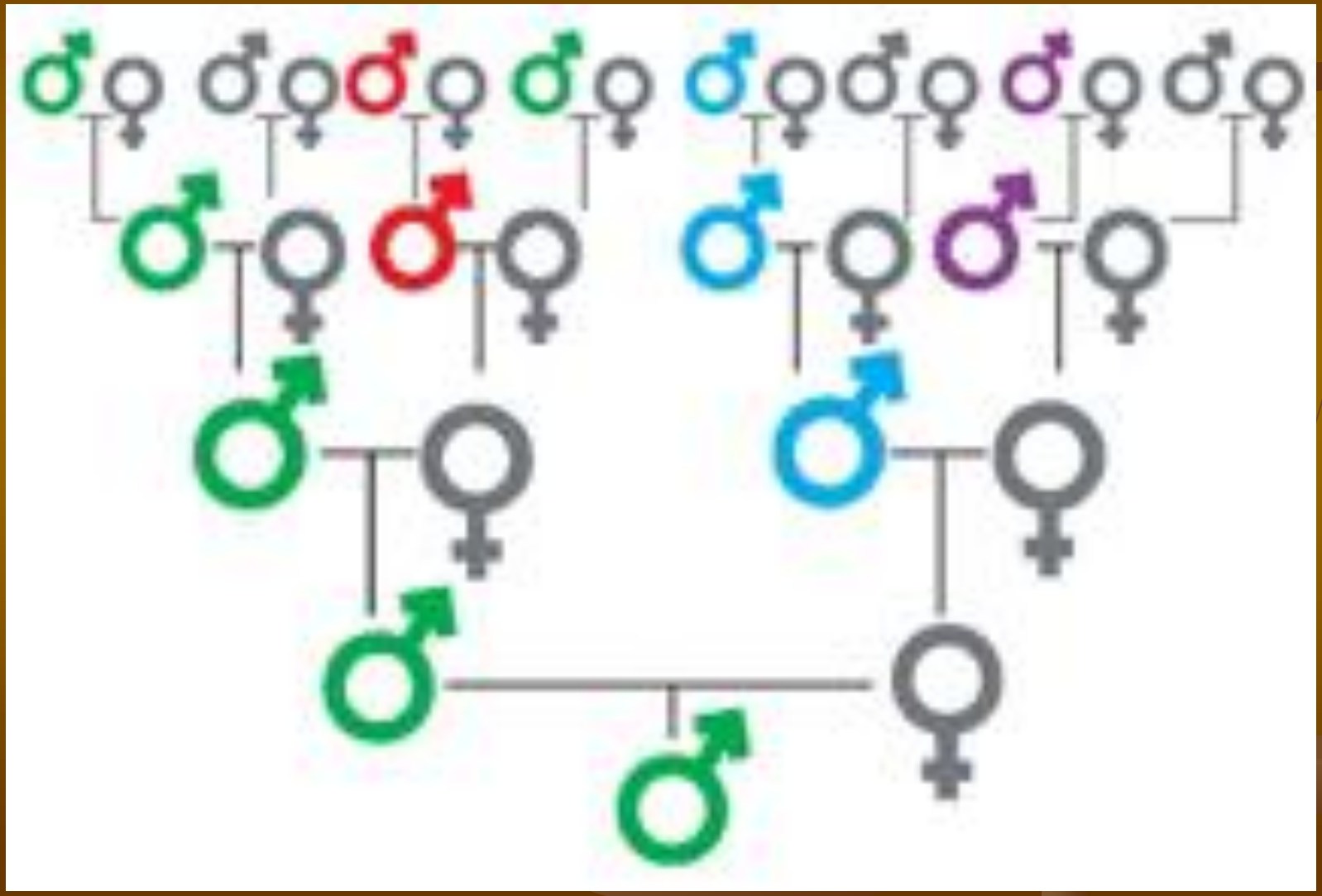
L F C1 C2 M L

Y-Chromosome Analysis

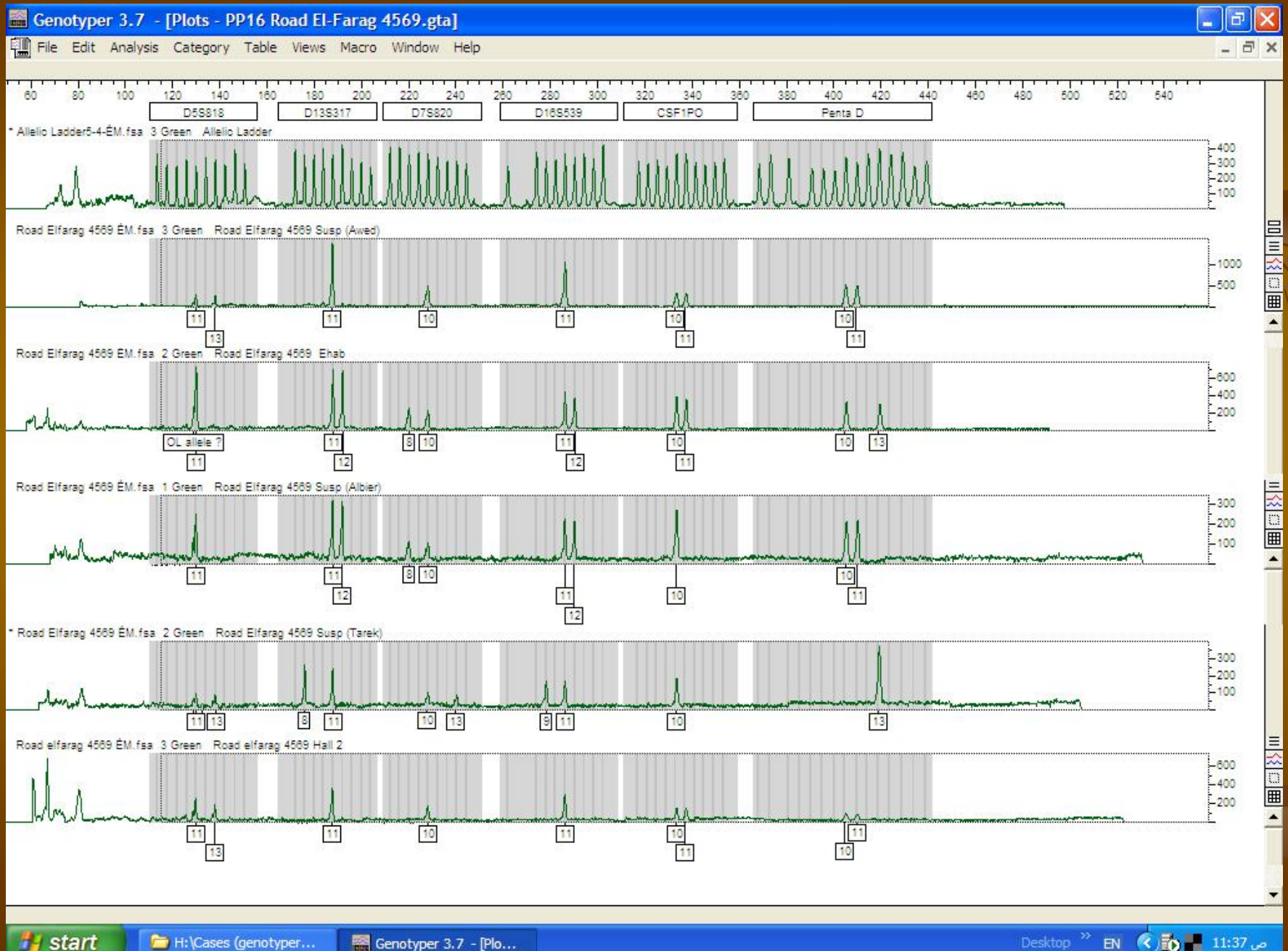
Y Chromosome STR Markers



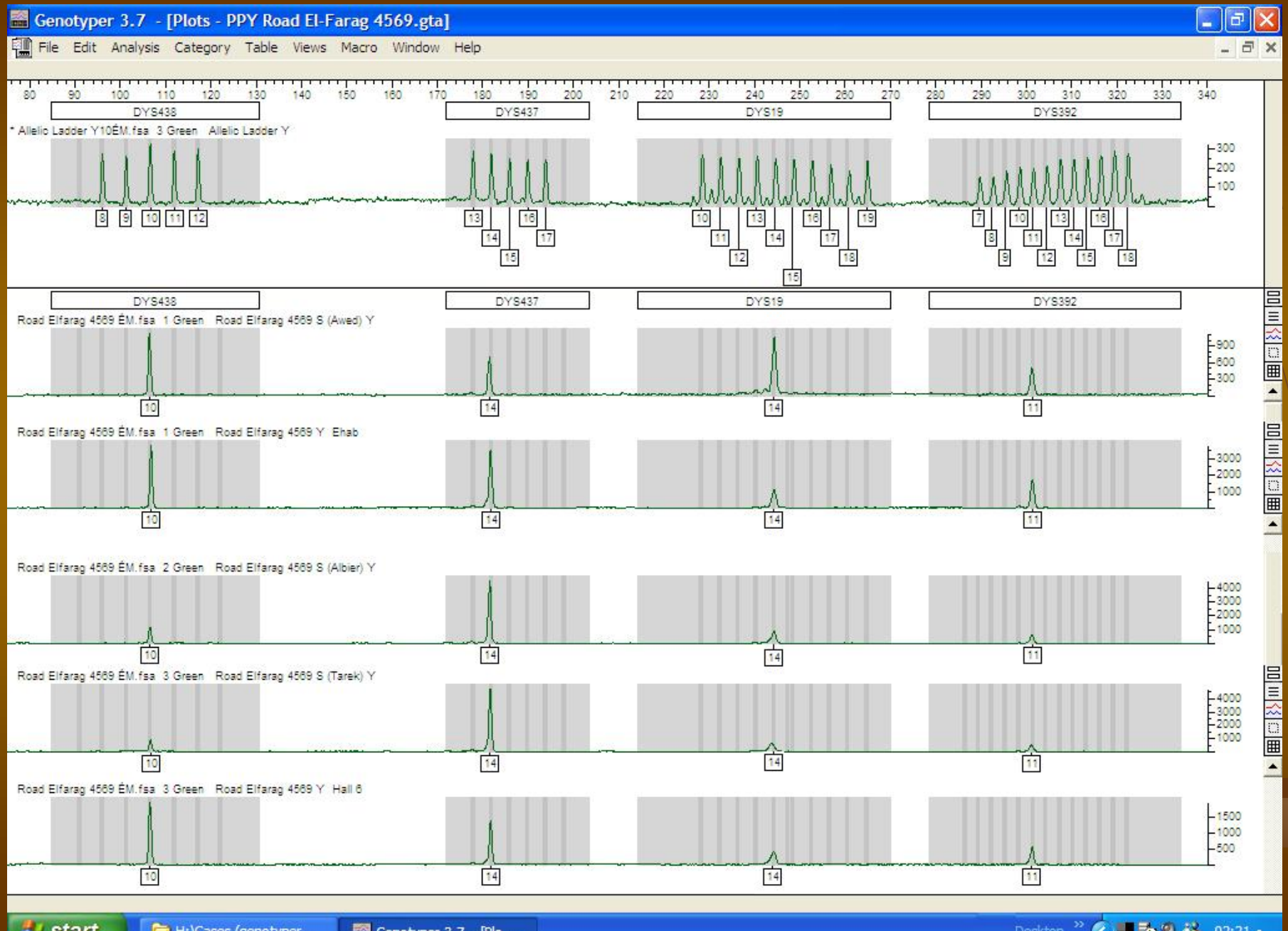
Y-chromosome Inheritance Pattern



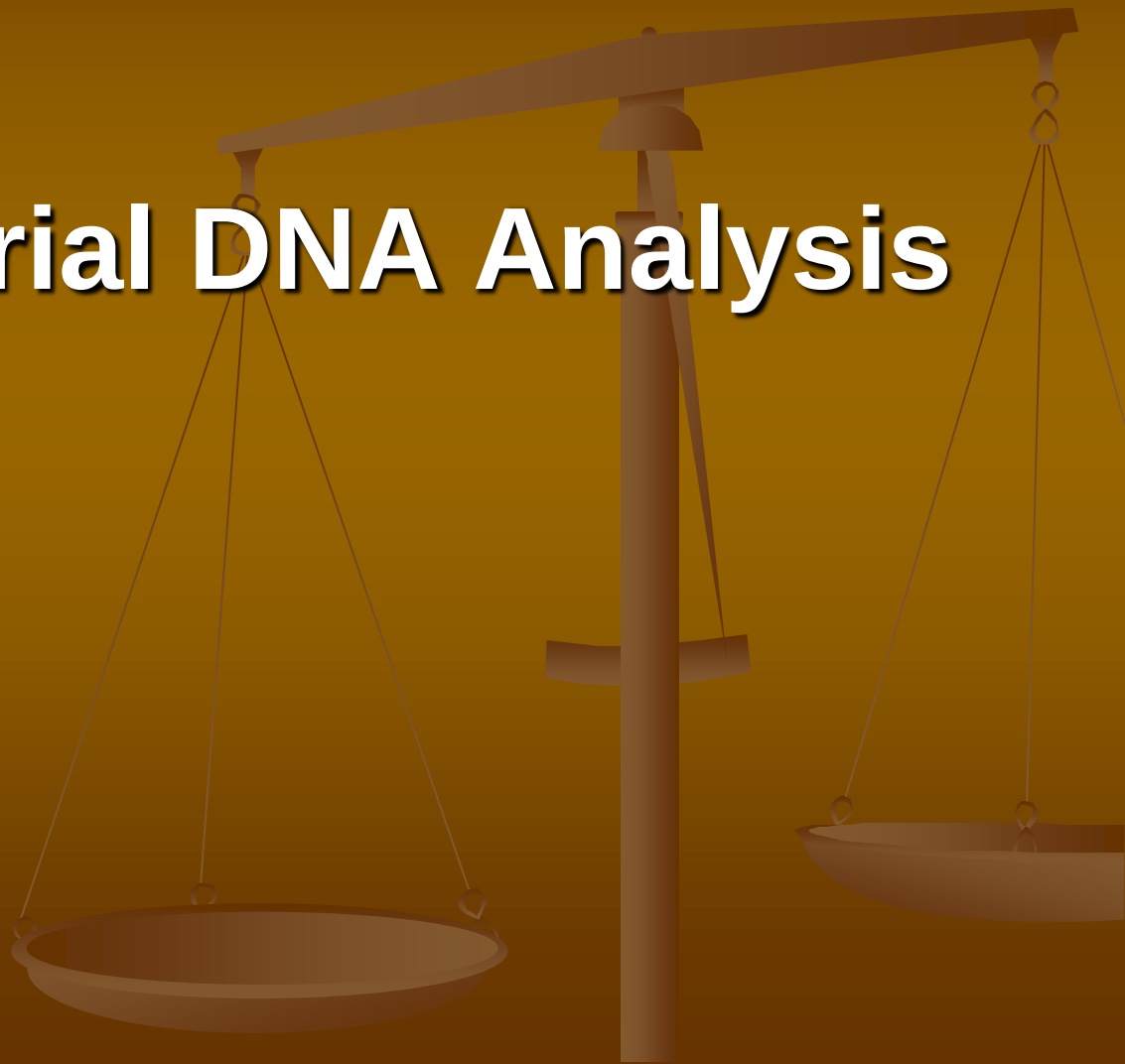
Identification Case



Identification Case …

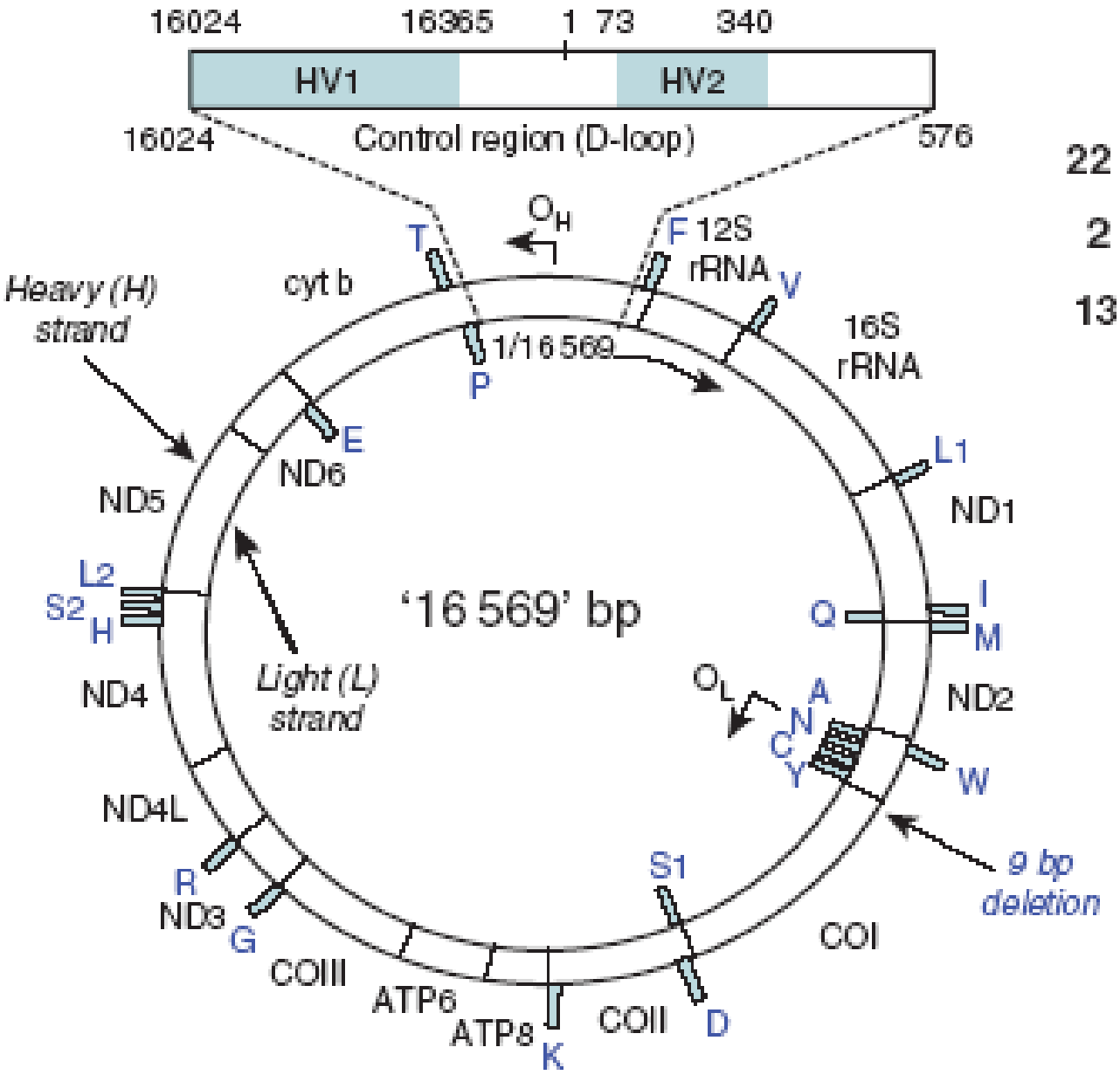


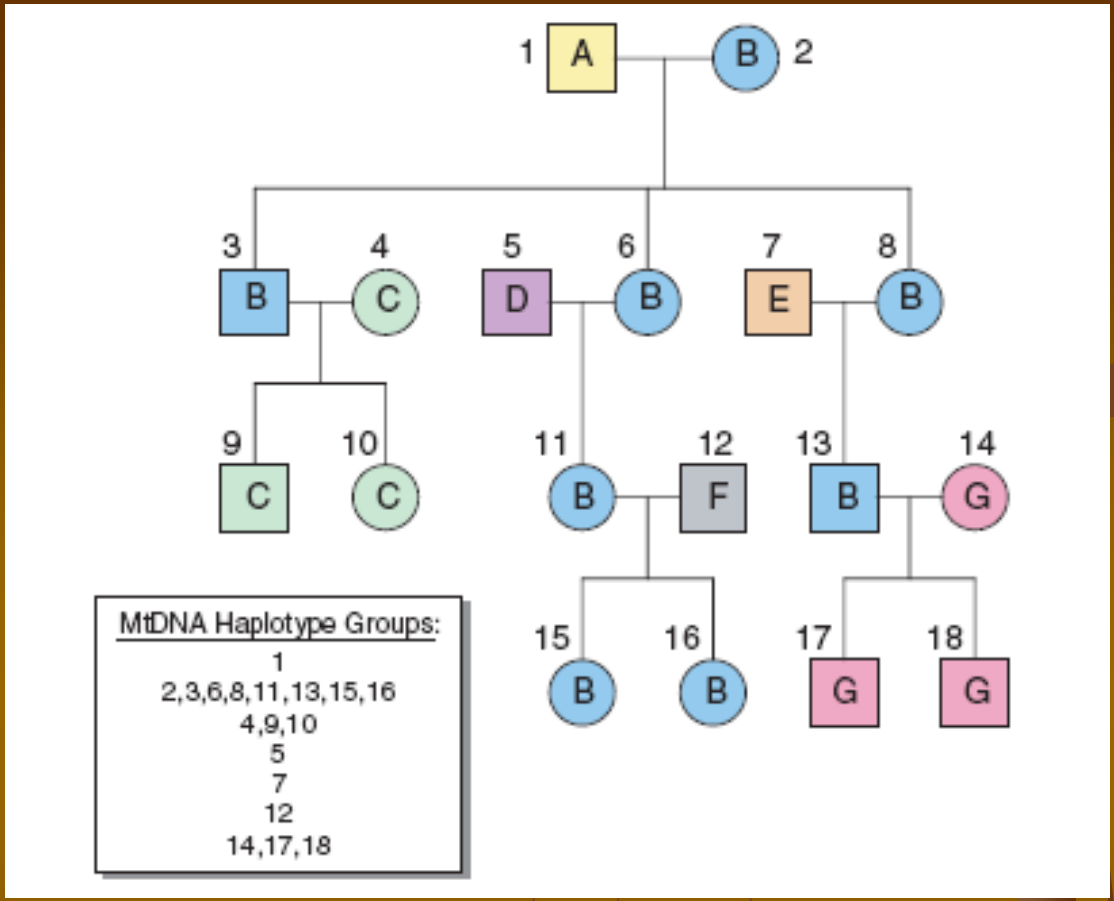
Mitochondrial DNA Analysis



MATERNAL INHERITANCE OF mtDNA

- At fertilization only the sperm's nucleus enters the egg and joins directly with the egg's nucleus.
- The sperm does not contribute other cellular components.
- When the zygote cell divides and a blastocyst develops, the cytoplasm and other cell parts save the nucleus are consistent with the mother's original egg cell.
- Thus, mitochondria with their mtDNA molecules are passed directly from the mother's egg cell to all offsprings independent of any male influence.


16024
16365
1
73
340
HV1
HV2
16024
Control region (D-loop)
576
22 tRNAs
2 rRNAs
13 genes
Heavy (H) strand
cyt b
T
O_H
F
12S rRNA
V
16S rRNA
P
1/16 569
E
ND6
ND5
L1
ND1
L2
S2
H
'16 569' bp
Q
I
M
Light (L) strand
O_L
A
N
C
Y
ND4
ND2
W
ND4L
R
ND3
G
COIII
ATP6
ATP8
K
COII
S1
D
COI
9 bp deletion



The figure shows the **mtDNA Inheritance Pattern**.

- Note that individual 16 will possess the same mtDNA type as seven of the other represented individuals (e.g., 2, 3, 6, 8, 11, 13, and 15).

SCIENCE OF MITOCHONDRIAL DNA

- In mtDNA analysis, the order of nitrogen bases provides the forensic scientist with a basis for distinguishing between unrelated individuals.
- A phone number analogy can illustrate the importance of the order of the bases in DNA. The phone number 555-1234 would reach one particular individual when dialed, whereas a phone number containing the same digits in another order, such as 555-4321, would contact an entirely different individual.
- In a similar manner, forensic scientists can use the order of bases in mtDNA to distinguish between unrelated individuals, thus mtDNA analysis is considered a type of Sequence polymorphism unlike nuclear (somatic and y-chromosome) analysis which is attributed to Length polymorphism.

SCIENCE OF MITOCHONDRIAL DNA

- In humans, individuals inherit mitochondrial DNA strictly from their mothers.
- Thus, the mtDNA sequences obtained from maternally related individuals, such as a brother and a sister or a mother and a daughter, will exactly match each other in the absence of a mutation.
- This characteristic of mtDNA is advantageous in missing person cases as any maternal relative of the missing individual can supply reference samples.

Comparison of Human Nuclear DNA & Mitochondrial DNA Markers

Characteristics	Nuclear DNA (nucDNA)	Mitochondrial DNA (mtDNA)
Size of genome	~3.2 billion bp	~16 569 bp
Copies per cell	2 (1 allele from each parent)	Can be > 1000
Percent of total DNA content per cell	99.75%	0.25%
Structure	Linear; packaged in chromosomes	Circular
Inherited from	Father and Mother	Mother
Chromosomal pairing	Diploid	Haploid
Generational recombination	Yes	No
Replication repair	Yes	No
Unique	Unique to individual (except identical twins)	Not unique to individual (same as maternal relatives)
Mutation rate	Low	At least 5–10 times nucDNA
Reference sequence	Described in 2001 by the Human Genome Project	Described in 1981 by Anderson and co-workers

STEPS IN mtDNA ANALYSIS

- Currently, the forensic analysis of mtDNA remains labor-intensive.
- Several biological techniques are combined to obtain an mtDNA sequence from a sample.
- Steps of mtDNA analysis process include primary visual analysis; sample preparation; DNA processing, including extraction, amplification, quantification, and sequencing; and data analysis.

Thank you