Parentage Statistics
Strength of Genetic Evidence In Parentage Testing

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PATERNITY TESTING

MOTHER

ALLEGED FATHER

CHILD

Two alleles for each autosomal genetic marker

Typical Paternity Test

Two possible outcomes of test:

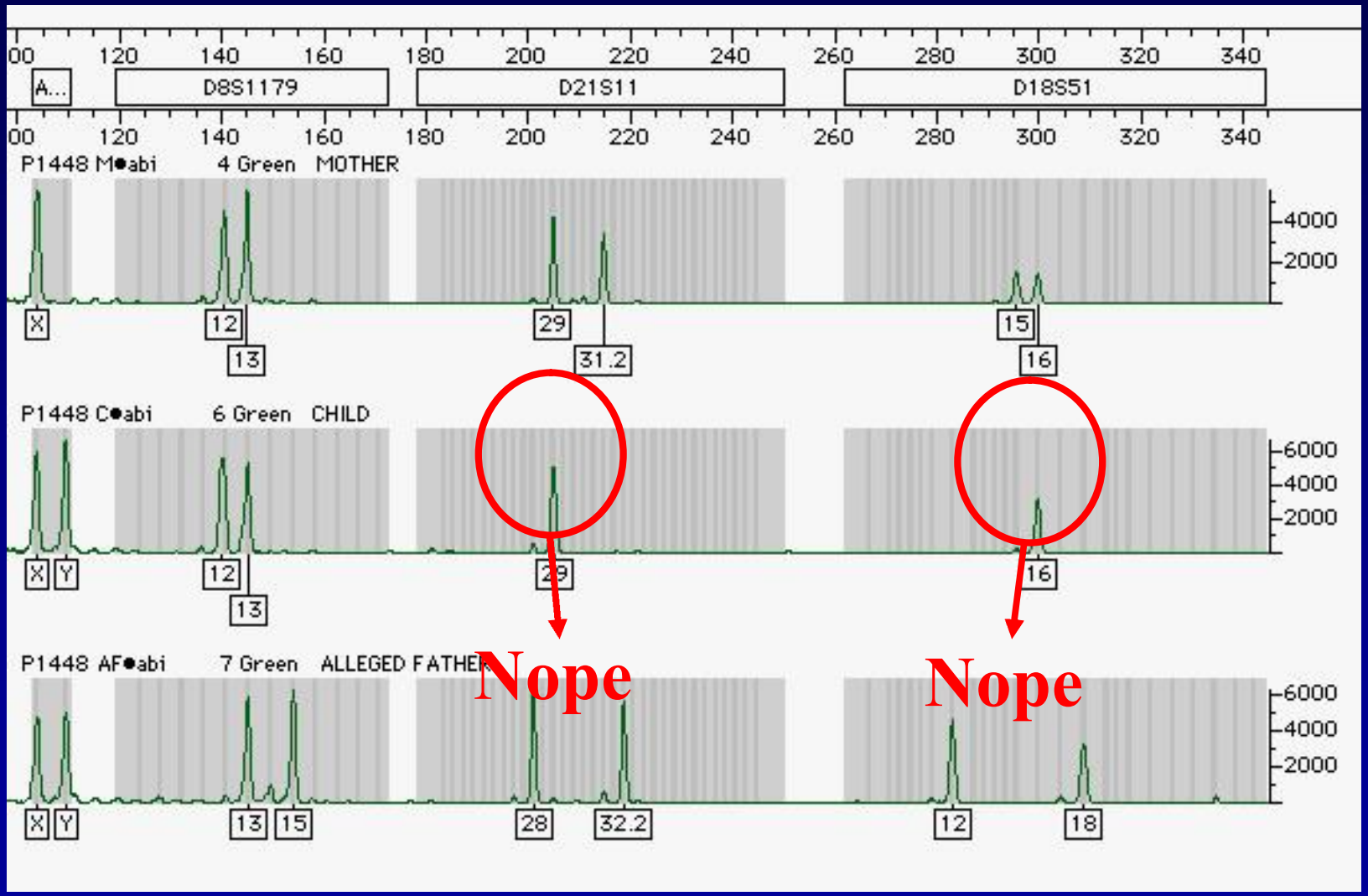
Inclusion

The obligate paternal alleles in the child all have corresponding alleles in the *Alleged Father*

Exclusion

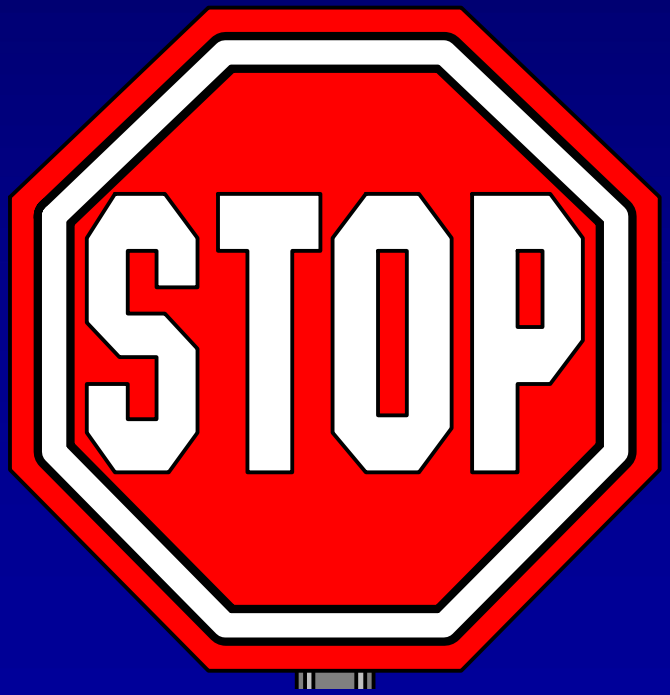
The obligate paternal alleles in the child DO NOT have corresponding alleles in the *Alleged Father*

Exclusion

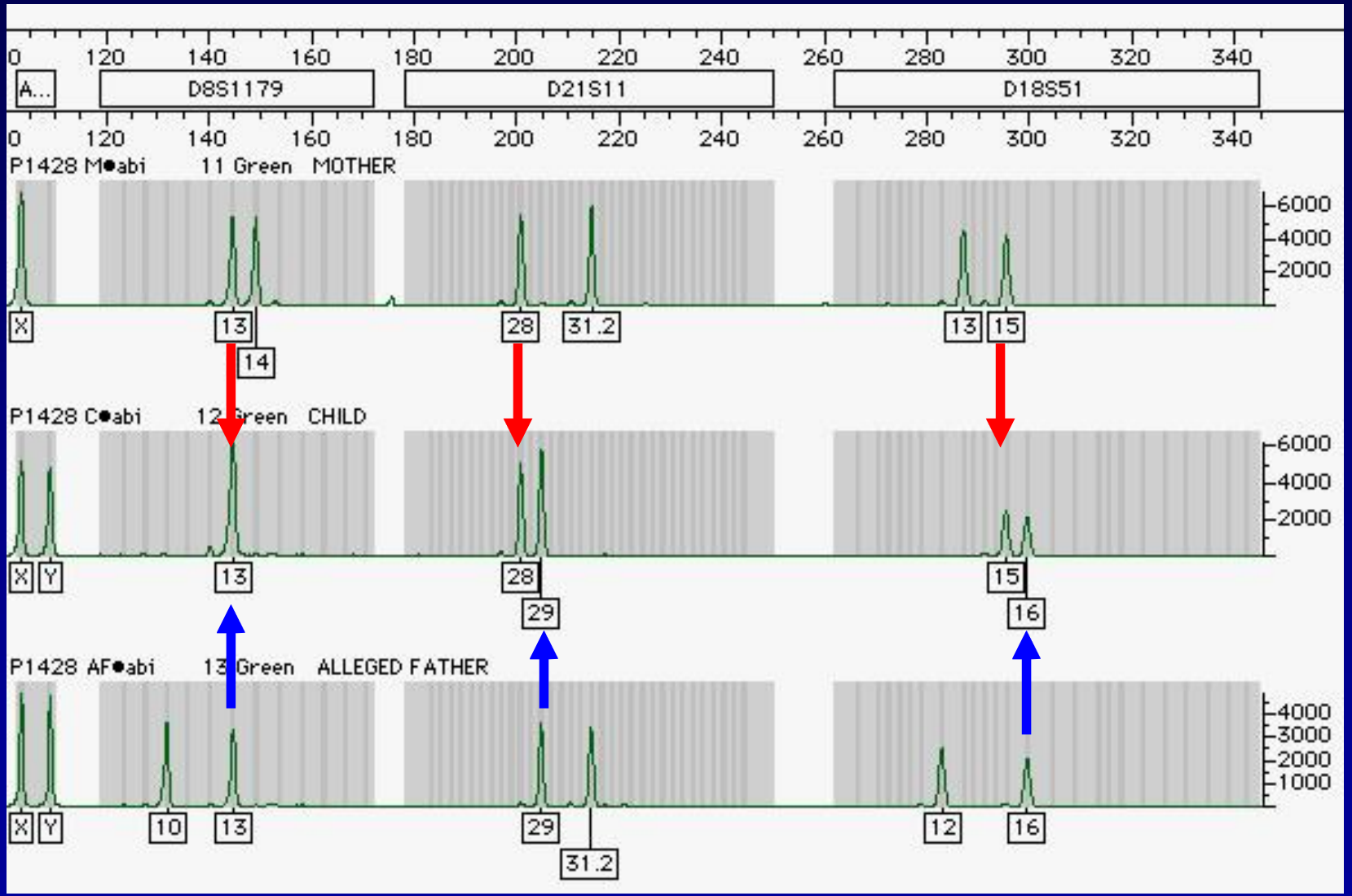

A...
D8S1179
D21S11
D18S51
P1448 M●abi 4 Green MOTHER
X
12
13
29
31.2
15
16
4000
2000
P1448 C●abi 6 Green CHILD
X
Y
12
13
29
16
6000
4000
2000
Nope
Nope
P1448 AF●abi 7 Green ALLEGED FATHER
X
Y
13
15
28
32.2
12
18
6000
4000
2000

Results

The Tested Man is Excluded as the Biological Father of the Child in Question



Inclusion


0 120 140 160 180 200 220 240 260 280 300 320 340
A...
D8S1179
D21S11
D18S51
P1428 M●abi 11 Green MOTHER
X
13
14
28
31.2
13
15
6000
4000
2000
P1428 C●abi 12 Green CHILD
X
Y
13
28
29
15
16
6000
4000
2000
P1428 AF●abi 13 Green ALLEGED FATHER
X
Y
10
13
29
31.2
12
16
4000
3000
2000
1000

Results

The Tested Man *Cannot be Excluded* as the Biological Father of the Child in Question

Several Statistical Values are Calculated to Assess the Strength of the Genetic Evidence

Language of Paternity Testing

PI	**Paternity Index**
CPI	**Combined Paternity Index**
W	**Probability of Paternity**
PE	**Probability of Exclusion**

Paternity Index
summarizes information provided by genetic testing

- Likelihood Ratio
- Probability that some event will occur under a set of conditions or assumptions
- Divided by the probability that the same event will occur under a set of different mutually exclusive conditions or assumptions

Paternity Index

- Observe three types – from a man, a woman, and a child
- Assume true trio – the man and woman are the true biologic parents of child
- Assume false trio – woman is the mother, man is not the father
- In the false trio, the child's father is a man of unknown type, selected at random from population (unrelated to mother and tested man)

Standard Paternity Index

- In paternity testing, the event is observing three phenotypes, those of a woman, man and child.
- The assumptions made for calculating the numerator (X) is that these three persons are a "true trio".
- For the denominator (Y) the assumptions is that the three persons are a "false trio".

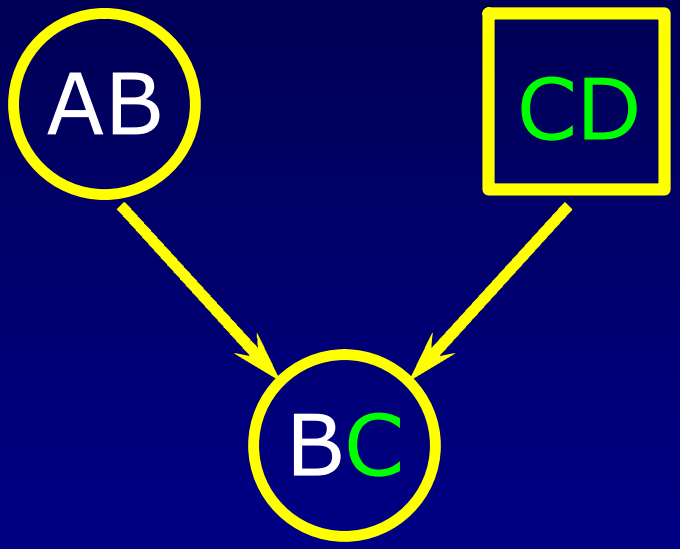
Paternity Analysis

Hypothetical case

DNA Analysis Results in Three Genotypes

Mother	**(AB)**
Child	**(BC)**
Alleged Father	**(CD)**

Paternity Analysis



An AB mother and a CD father can have four possible offspring:

AC, AD, BC, BD

Standard Paternity Index

PI determination in hypothetical DNA System

PI = X / Y

Numerator

X = **is the probability that (1) a woman randomly selected from a population is type AB, and (2) a man randomly selected from a population is type CD, and (3) their child is type BC.**

Standard Paternity Index

PI determination in hypothetical DNA System

PI = X / Y

Denominator

Y = is the probability that (1) a woman randomly selected from a population is type AB, (2) a man randomly selected and unrelated to either mother or child is type CD, and (3) the woman's child, unrelated to the randomly selected man is BC.

Standard Paternity Index

When mating is random, the probability that the untested alternative father will transmit a specific allele to his child is equal to the allele frequency in his race.

We can now look into how to actually calculate a Paternity Index

Hypothetical DNA Example

First Hypothesis

Numerator

Person	Type
Mother	AB
Child	BC
Alleged Father	CD

In order to explain this evidence Calculate Probability that

a) Woman randomly selected from population is type AB

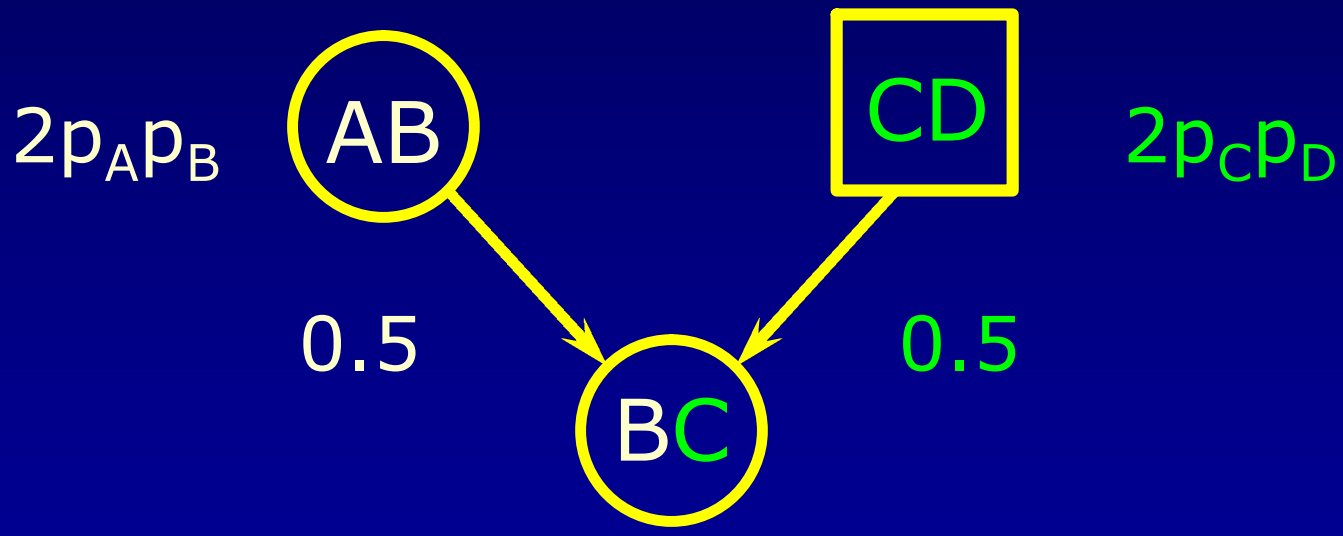
b) Man randomly selected from population is type CD, and

c) Their child is type BC

Paternity Analysis

Paternity Index

Numerator



Probability = $2p_Ap_B$ x $2p_Cp_D$ x 0.5 x 0.5

Hypothetical DNA Example

Second Hypothesis
Denominator

Person	Type
Mother	AB
Child	BC
Alleged Father	CD

In order to explain this evidence Calculate Probability that

a) Woman randomly selected from population is type AB

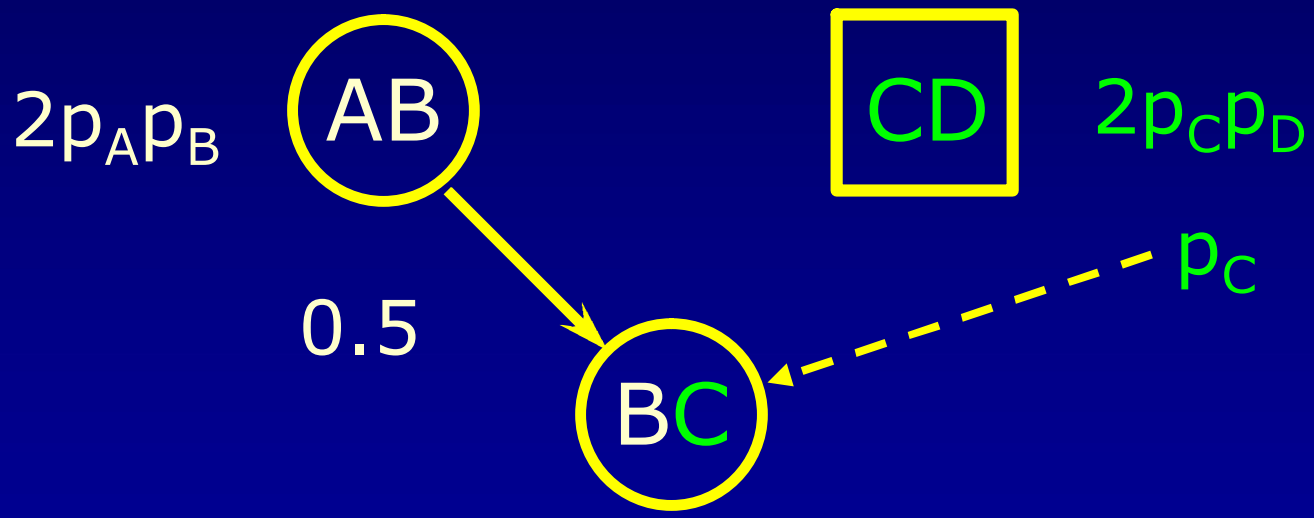
b) An alternative man randomly selected from population is type CD, and

c) The woman's child, fathered by random man, is type BC

Paternity Analysis

Paternity Index

Denominator



Probability = $2p_Ap_B$ x $2p_Cp_D$ x 0.5 x p_C

Paternity Analysis
Paternity Index

$$PI = \frac{2p_A p_B \times 2p_C p_D \times 0.5 \times 0.5}{2p_A p_B \times 2p_C p_D \times 0.5 \times p_C}$$

$$PI = \frac{0.5}{p_C}$$

Hypothetical DNA Example

Probability Statements

Person	Type
Mother	AB
Child	BC
Alleged Father	CD

One might say (Incorrectly)

a) Numerator is probability that tested man is the father, and

b) Denominator is probability that he is not the father

Hypothetical DNA Example

Probability Statement

Person	Type
Mother	AB
Child	BC
Alleged Father	CD

A **Correct** statement is

a) Numerator is probability of observed genotypes, given the tested man is the father, and

b) Denominator is probability of observed genotypes, given a random man is the father.

Incorrect Verbal Expression of the Paternity Index?

It is (X/Y) times more likely the tested man was the true biological father than an untested random man was the father

Correct Verbal Expression of the Paternity Index?

It is (X/Y) times more likely to see the genetic results if the tested man was the true biological father than if an untested random man was the father

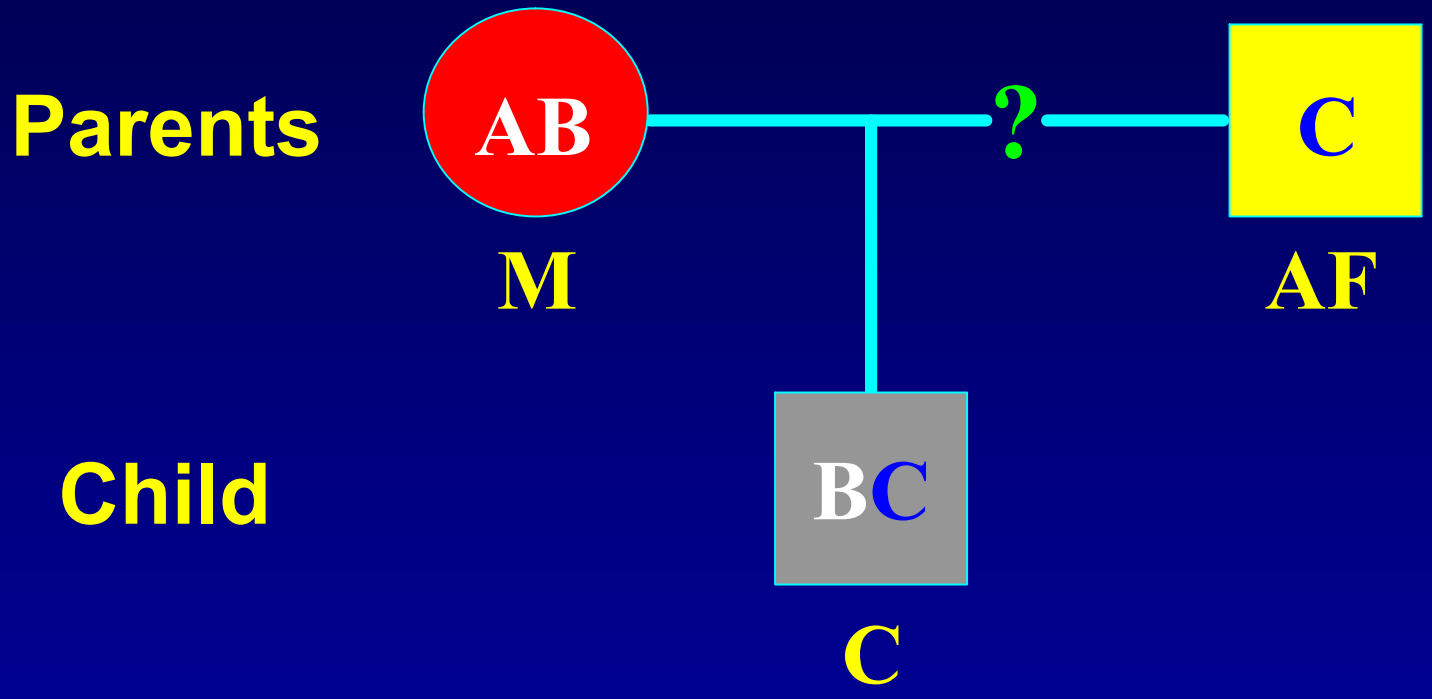
or

There is (X/Y) times more support for the genetic results if the tested man was the true biological father than if an untested random man was the father

There are 15 possible combinations of genotypes for a paternity trio

Paternity Index

M and C share one allele
and AF is homozygous for the obligatory allele



AF can only pass C allele

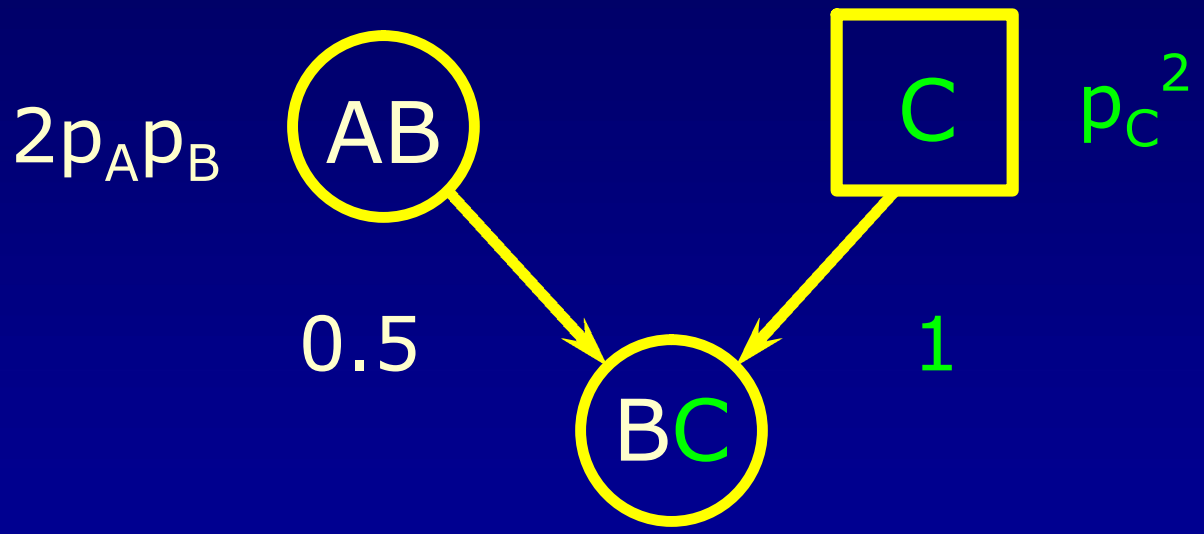
Random Man has p chance of passing the C allele

PI = 1/p

Paternity Analysis

Paternity Index

Numerator

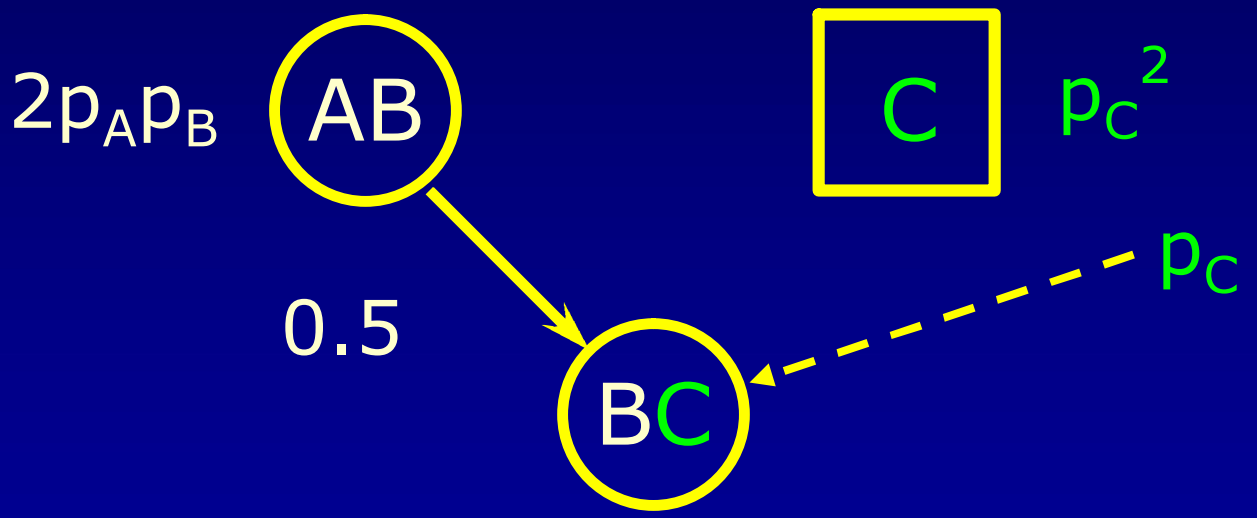


Probability = $2p_Ap_B \times p_C^2 \times 0.5 \times 1$

Paternity Analysis

Paternity Index

Denominator



Probability = $2p_Ap_B \times p_C^2 \times 0.5 \times p_C$

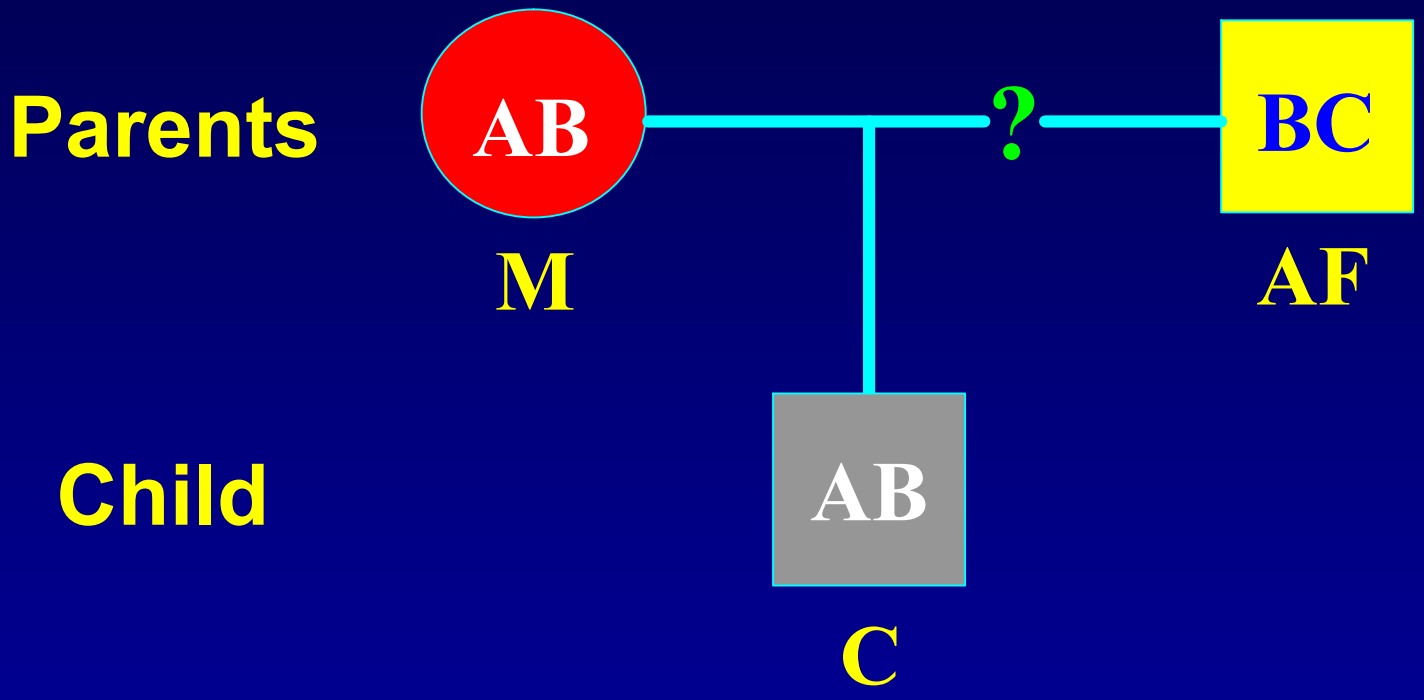
Paternity Analysis
Paternity Index

$$PI = \frac{2p_A p_B \times p_C^2 \times 0.5 \times 1}{2p_A p_B \times p_C^2 \times 0.5 \times p_C}$$

$$PI = \frac{1}{p_C}$$

Paternity Index

M and C share both alleles and
AF is heterozygous with one of the obligatory alleles



M has a 1 in 2 chance of passing A or B allele

AF has a 1 in 2 chance of passing B allele

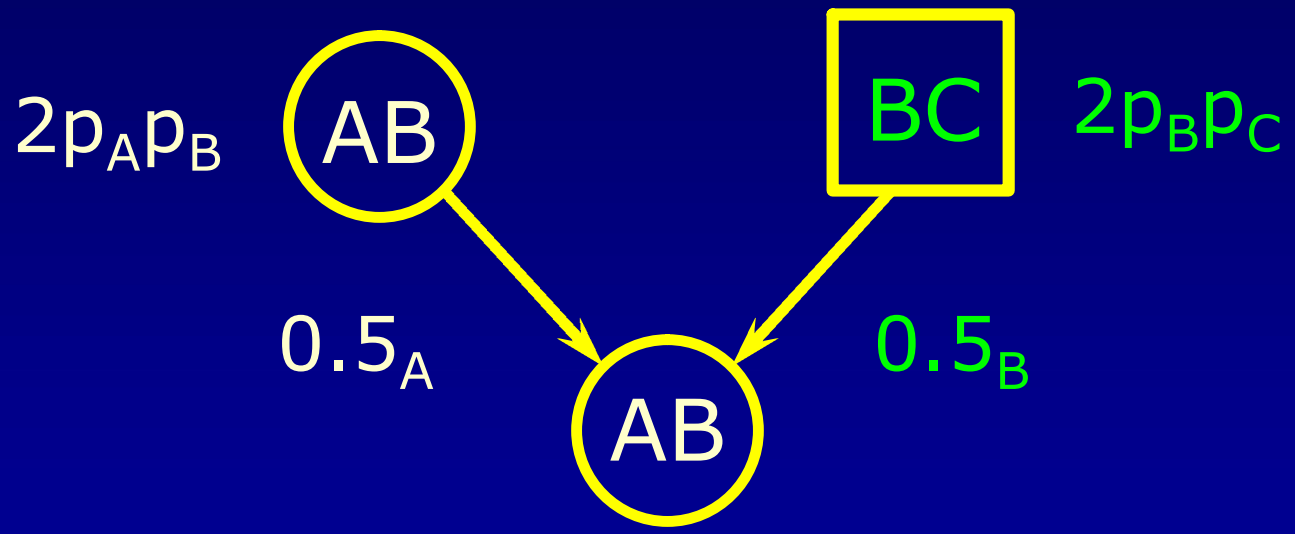
RM has (p + q) chance of passing the A or B alleles

PI = 0.5/(p+q)

Paternity Analysis

Paternity Index

Numerator

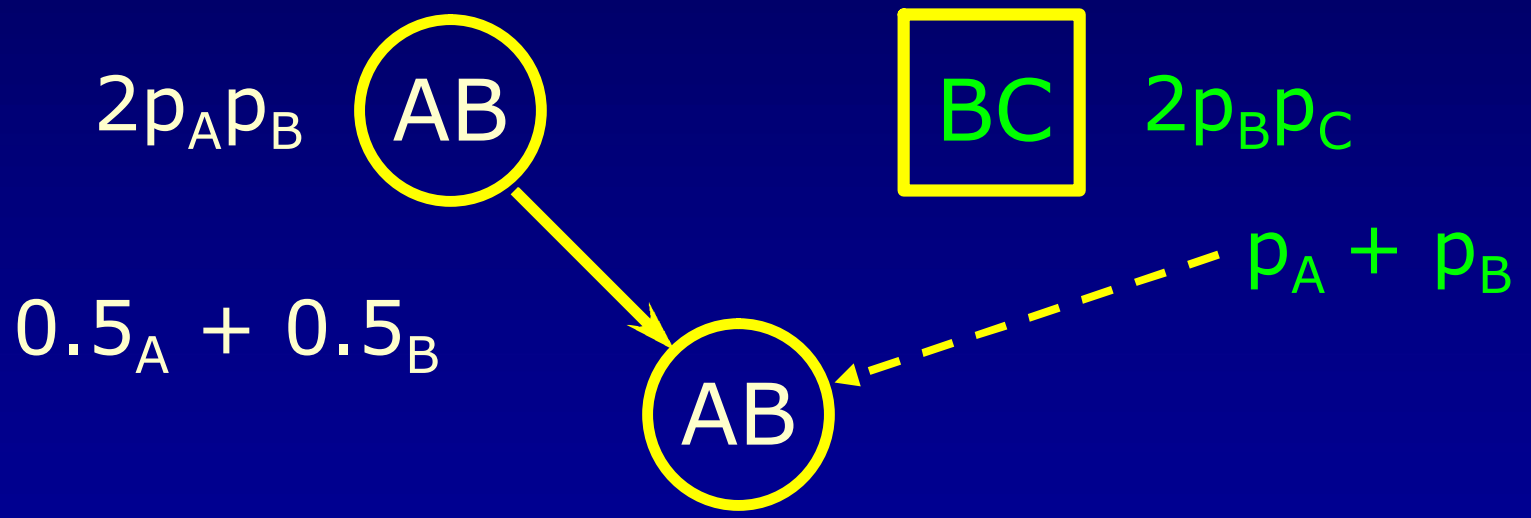


Probability = $2p_Ap_B$ x $2p_Bp_C$ x $0.5_{(mA)}$ x $0.5_{(fB)}$

Paternity Analysis

Paternity Index

Denominator



probability =

$$2p_Ap_B \times 2p_Bp_C \times (0.5_{(mA)} \times p_B + 0.5_{(mB)} \times p_A)$$

Paternity Analysis

Paternity Index

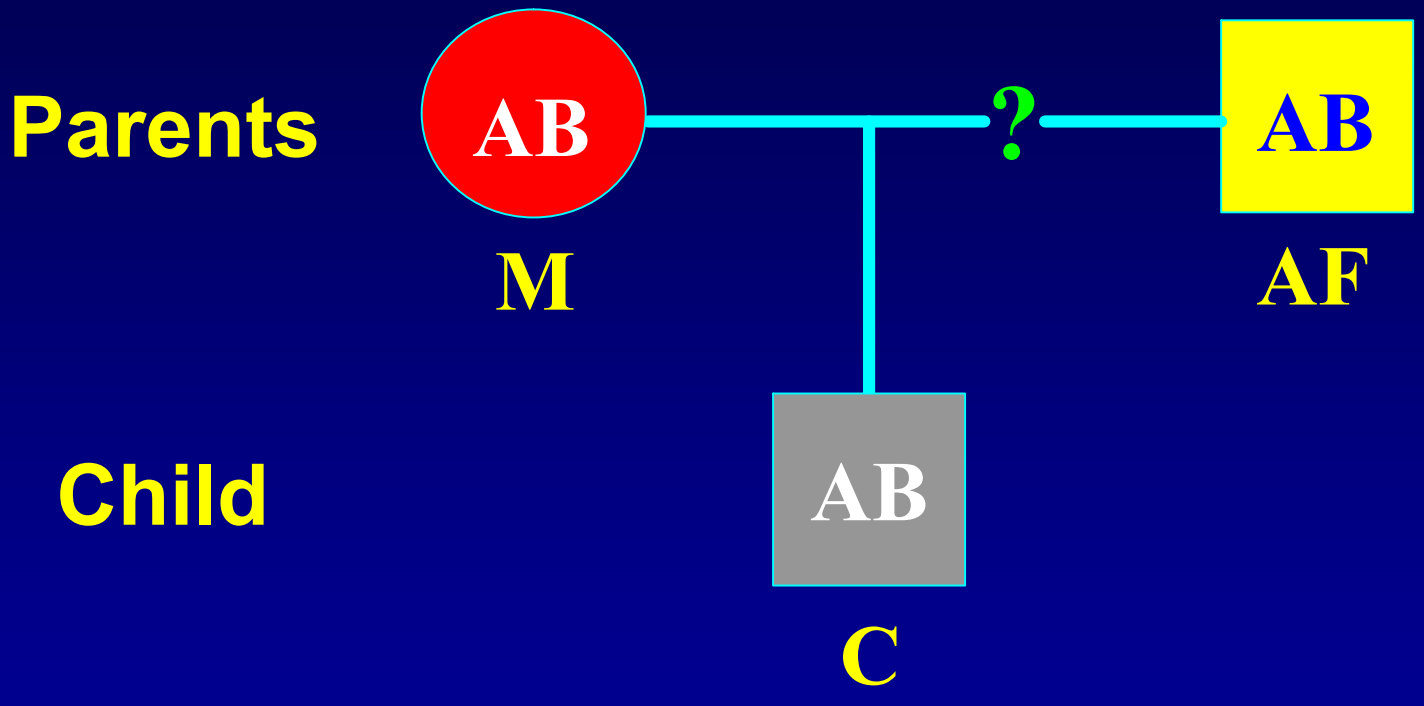
$$PI = \frac{2p_Ap_B \times 2p_Bp_C \times 0.5_{(mA)} \times 0.5_{(fB)}}{2p_Ap_B \times 2p_Bp_C \times (0.5_{(mB)} \times p_A + 0.5_{(mA)} \times p_B)}$$

$$PI = \frac{0.25}{0.5p_A + 0.5p_B}$$

$$PI = \frac{0.5}{p_A + p_B}$$

Paternity Index

M and C share both alleles and AF is heterozygous with both of the obligatory alleles



M has a 1 in 2 chance of passing A or B allele

AF has a 1 in 2 chance of passing A or B allele

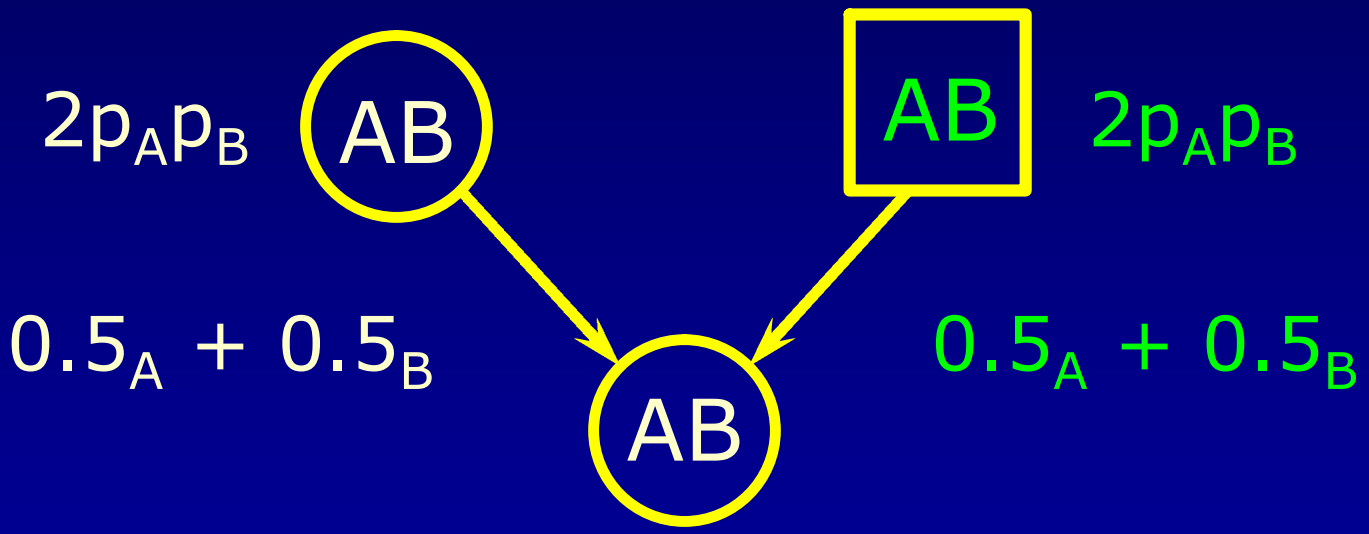
RM has (p + q) chance of passing the A or B alleles

PI = 1/(p+q)

Paternity Analysis

Paternity Index

Numerator



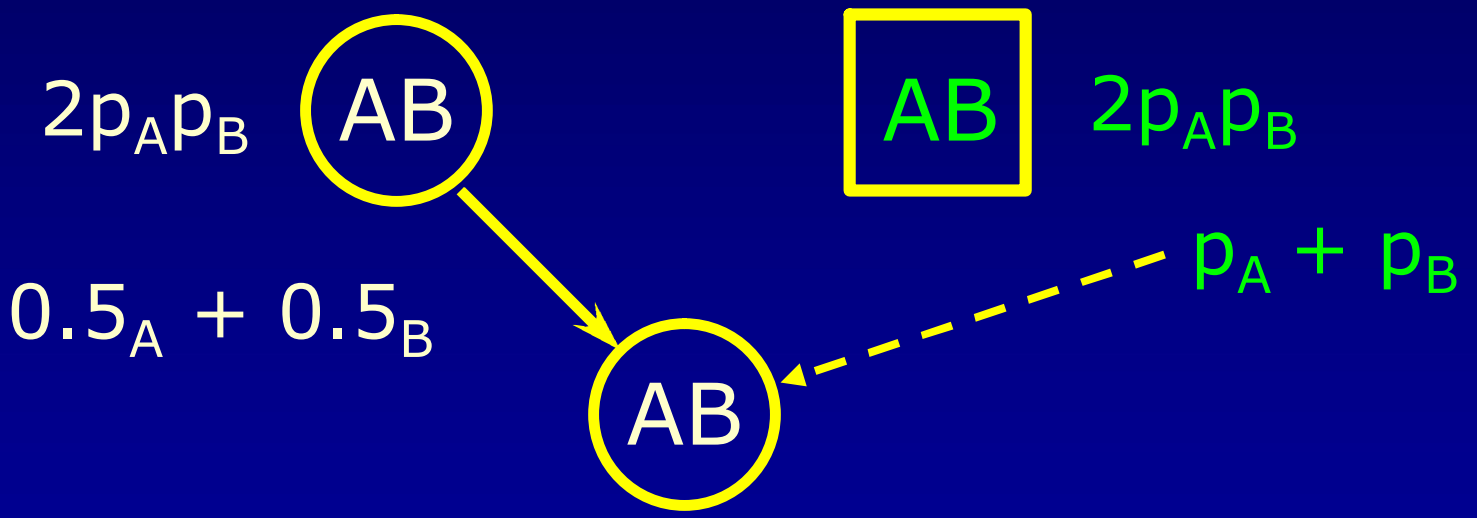
Probability =

$2p_Ap_B \times 2p_Ap_B \times (0.5_{(mA)} \times 0.5_{(fB)} + 0.5_{(mB)} \times 0.5_{(fA)})$

Paternity Analysis

Paternity Index

Denominator



probability =

$2p_Ap_B \times 2p_Ap_B \times (0.5_{(mA)} \times p_B + 0.5_{(mB)} \times p_A)$

Paternity Analysis

Paternity Index

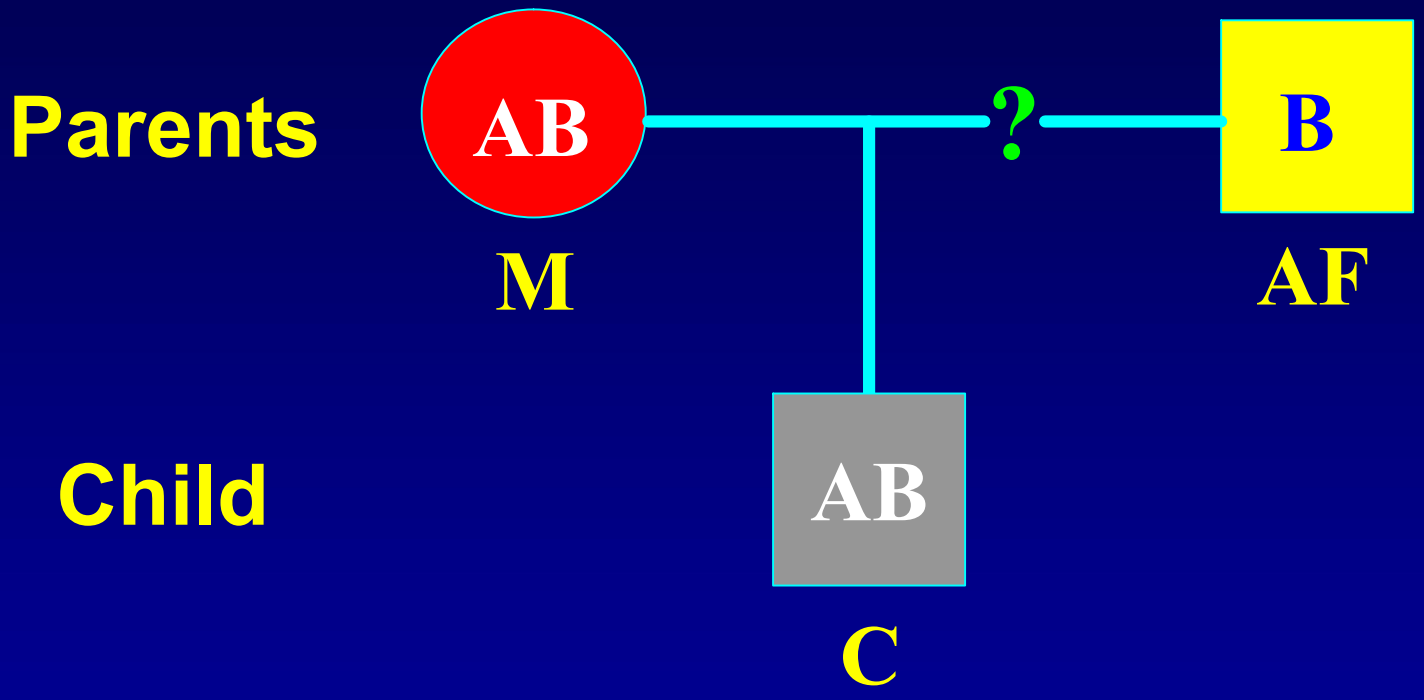
$$PI = \frac{2p_Ap_B \times 2p_Ap_B \times (0.5_{(mA)} \times 0.5_{(fB)} + 0.5_{(mB)} \times 0.5_{(fA)})}{2p_Ap_B \times 2p_Ap_B \times (0.5_{(mB)} \times p_A + 0.5_{(mA)} \times p_B)}$$

$$PI = \frac{0.5}{0.5p_A + 0.5p_B}$$

$$PI = \frac{1}{p_A + p_B}$$

Paternity Index

M and C share both alleles and
AF is homozygous with one of the obligatory alleles



M has a 1 in 2 chance of passing A or B allele

AF can only pass the B allele

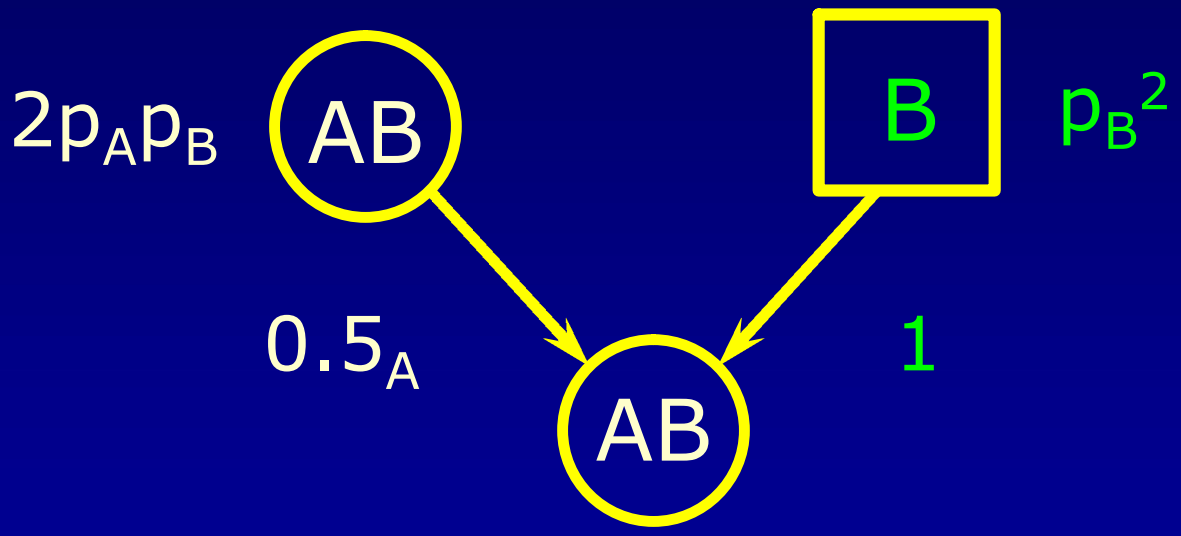
RM has (p + q) chance of passing the A or B alleles

PI = 1/(p+q)

Paternity Analysis

Paternity Index

Numerator

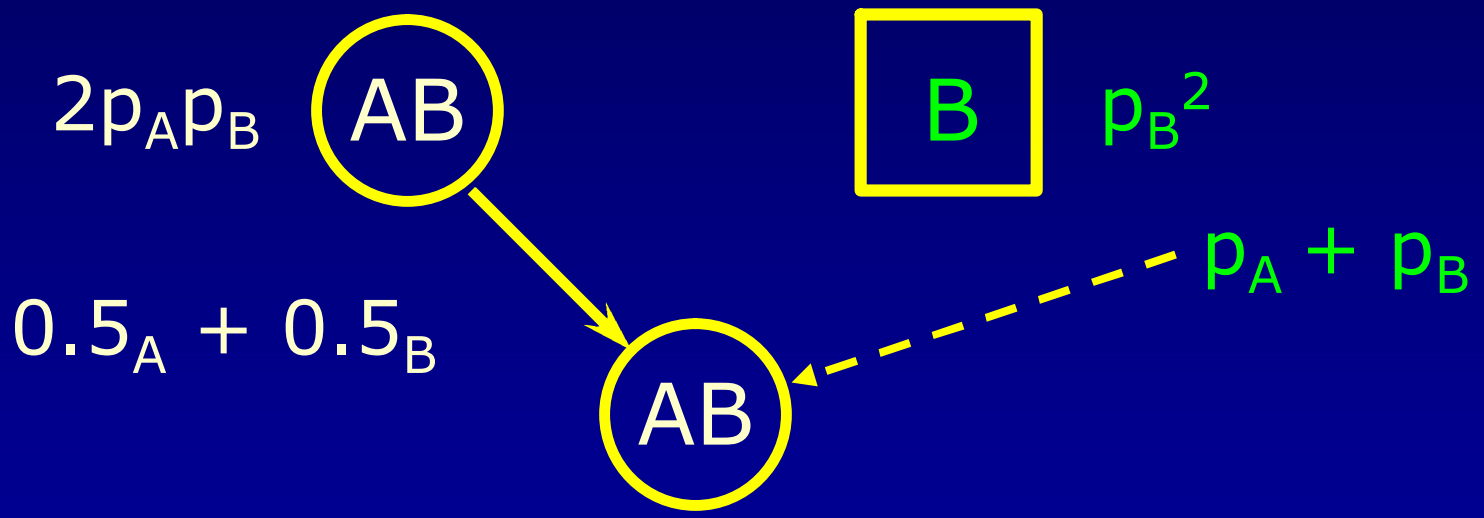


Probability = $2p_Ap_B$ x p_B^2 x $0.5_{(mA)}$ x $1_{(fB)}$

Paternity Analysis

Paternity Index

Denominator



probability =

$$2p_Ap_B \times p_B^2 \times (0.5_{(mA)} \times p_B + 0.5_{(mB)} \times p_A)$$

Paternity Analysis

Paternity Index

$$PI = \frac{2p_A p_B \times p_B^2 \times 0.5_{(mA)} \times 1_{(fB)}}{2p_A p_B \times p_B^2 \times (0.5_{(mB)} \times p_A + 0.5_{(mA)} \times p_B)}$$

$$PI = \frac{0.5}{0.5p_A + 0.5p_B}$$

$$PI = \frac{1}{p_A + p_B}$$

PI Formulas

Single locus, no null alleles, low mutation rate, codominance

M	C	AF	Numerator	Denominator	PI
A	A	AB	0.5	a	**0.5/a**
A	AB	AB	0.5	a	**0.5/a**
A	AB	BC	0.5	a	**0.5/a**
AB	A	AB	0.25	0.5a	**0.5/a**
AB	A	AC	0.25	0.5a	**0.5/a**
BC	AB	AB	0.25	0.5a	**0.5/a**
BC	AB	AC	0.25	0.5a	**0.5/a**
BD	AB	AC	0.25	0.5a	**0.5/a**

PI Formulas

Single locus, no null alleles, low mutation rate, codominance

M	C	AF	Numerator	Denominator	PI
A	A	A	1	a	**1/a**
AB	A	A	0.5	0.5a	**1/a**
B	AB	A	1	a	**1/a**
BC	AB	A	0.5	0.5a	**1/a**

PI Formulas

Single locus, no null alleles, low mutation rate, codominance

M	C	AF	Numerator	Denominator	PI
AB	AB	AC	0.25	0.5(a+b)	**0.5/(a+b)**

PI Formulas

Single locus, no null alleles, low mutation rate, codominance

M	C	AF	Numerator	Denominator	PI
AB	AB	A	0.5	0.5(a+b)	**1/(a+b)**
AB	AB	AB	0.5	0.5(a+b)	**1/(a+b)**

Combined Paternity Index

- When multiple genetic systems are tested, a PI is calculated for each system.
- This value is referred to as a System PI.
- If the genetic systems are inherited independently, the Combined Paternity Index (CPI) is the product of the System PI's

Combined Paternity Index

What "is" the CPI?

- **The CPI is a measure of the strength of the genetic evidence.**
- **It indicates whether the evidence fits better with the hypothesis that the man is the father or with the hypothesis that someone else is the father.**

Combined Paternity Index

- **The theoretical range for the CPI is from 0 to infinity**
- **A CPI of 1 means the genetic tests provides no information**
- **A CPI less than 1; the genetic evidence is more consistent with non-paternity than paternity.**
- **A CPI greater than 1; the genetic evidence supports the assertion that the tested man is the father.**

Probability of Paternity

- **The probability of paternity is a measure of the strengths of one's belief in the hypothesis that the tested man is the father.**
- **The correct probability must be based on all of the evidence in the case.**
- **The non-genetic evidence comes from the testimony of the mother, tested man, and other witnesses.**
- **The genetic evidence comes from the DNA paternity test.**

Probability of Paternity

- **The probability of paternity (W) is based upon Baye's Theorem, which provides a method for determining a posterior probability based upon the genetic results of testing the mother, child, and alleged father. In order to determine the probability of paternity, an assumption must be made (before testing) as to the prior probability that the tested man is the true biological father.**

Probability of Paternity

- **The prior probability of paternity is the strength of one's belief that the tested man is the father based only on the non-genetic evidence.**

Probability of Paternity

$$\text{Probability of Paternity (W)} = \frac{\text{CPI x P}}{[\text{CPI x P} + (1 - \text{P})]}$$

P = Prior Probability; it is a number greater than 0 and less than or equal to 1. In many criminal proceedings the Probability of Paternity is not admissible. In criminal cases, the accused is presumed innocent until proven guilty. Therefore, the defense would argue that the Prior Probability should be 0. You cannot calculate a posterior Probability of Paternity with a Prior Probability of 0.

Probability of Paternity

- **In the United States, the court system has made the assumption that the prior probability is equal to 0.5. The argument that is presented is that the tested man is either the true father or he is not. In the absence of any knowledge about which was the case, it is reasonable to give these two possibilities equal prior probabilities.**

Probability of Paternity

With a prior probability of 0.5, the Probability of Paternity (W) =

$$\frac{\text{CPI} \times 0.5}{[\text{CPI} \times 0.5 + (1 - 0.5)]}$$

$$= \frac{\text{CPI}}{\text{CPI} + 1}$$

Posterior Odds in Favor of Paternity

Posterior Odds = CPI x Prior Odds

Prior Odds = P / (1 - P)

Posterior Odds in Favor of Paternity =

CPI x [P / (1 - P)]

If the prior probability of paternity is 0.7, then the prior odds favoring paternity is 7 to 3. If a paternity test is done and the CPI is 10,000, then the Posterior Odds in Favor of Paternity =

10,000 x (0.7 / 0.3) = 23,333

Posterior Odds in Favor of Paternity = 23,333 to 1

Probability of Exclusion

- **The probability of exclusion (PE) is defined as the probability of excluding a random individual from the population given the alleles of the child and the mother.**
- **The genetic information of the tested man is not considered in the determination of the probability of exclusion**

Probability of Exclusion

- **The probability of exclusion (PE) is equal to the frequency of all men in the population who do not contain an allele that matches the obligate paternal allele of the child.**

Probability of Exclusion

$$PE = 1 - (a^2 + 2ab)$$

a = frequency of the allele the child inherited from the biological father (obligate paternal allele). The frequency of the obligate allele is determined for each of the major racial groups, and the most common frequency is used in the calculation.

Probability of Exclusion

$(a^2 + 2ab)$ = Probability of Inclusion

Probability of Inclusion is equal to the frequency of all men in the population who contain an allele that matches the obligate paternal allele of the child.

PE = 1 – Probability of Inclusion

Probability of Exclusion

$$PE = 1 - (a^2 + 2ab)$$

b = sum of the frequency of all alleles other than the obligate paternal allele.

$$b = (1 - a)$$

$$PE = 1 - [a^2 + 2a(1 - a)]$$

$$PE = 1 - [a^2 + 2a - 2a^2]$$

$$PE = 1 - [2a - a^2]$$

$$PE = 1 - 2a + a^2$$

$$PE = (1 - a)^2$$

Probability of Exclusion

If the Mother and Child are both phenotype AB, men who cannot be excluded are those who could transmit either an A or B allele (or both). In this case the:

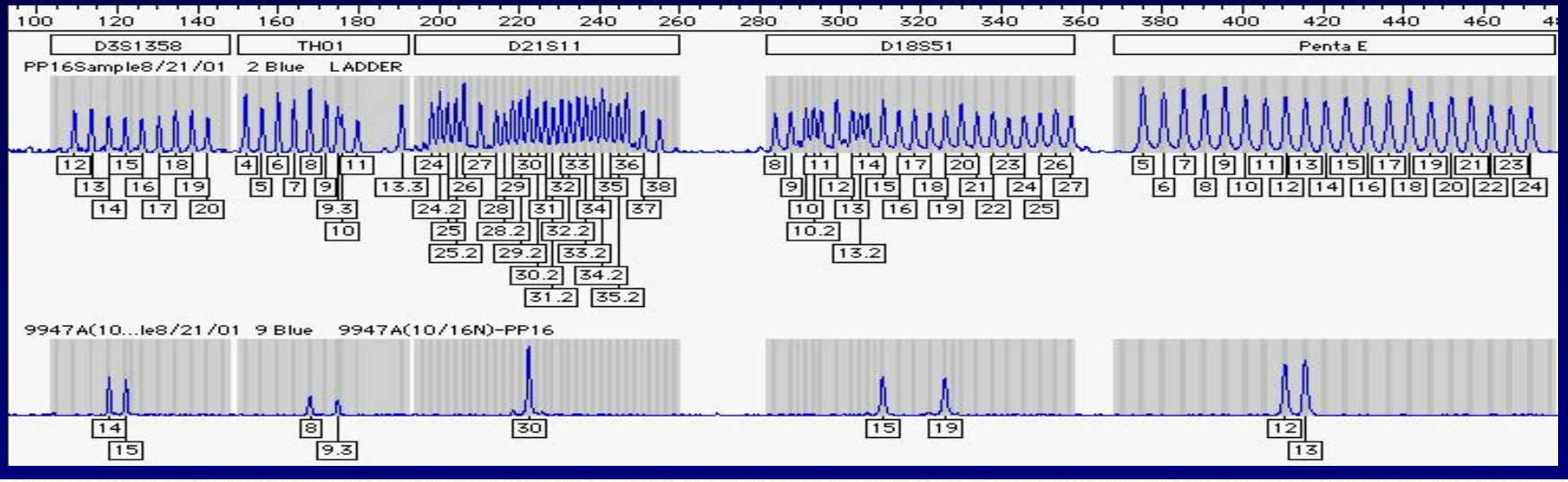
$$PE = [1 - (a + b)]^2$$

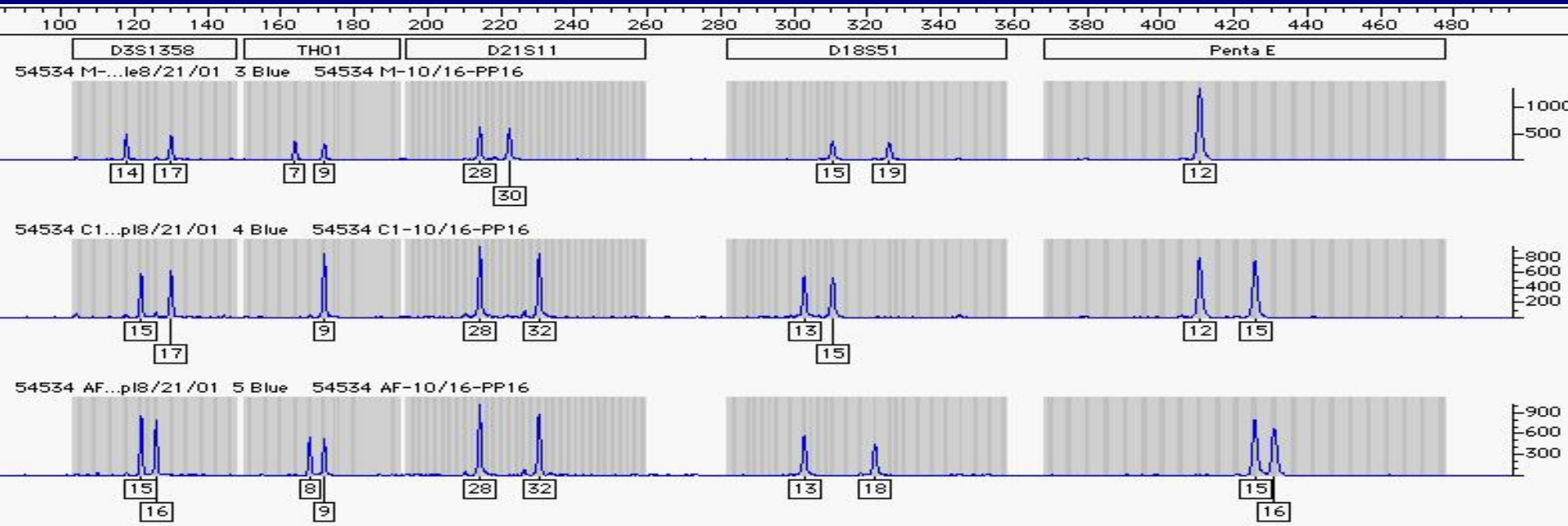
Combined Probability of Exclusion

The individual Probability of Exclusion is calculated for each of the genetic systems (loci) analyzed. The overall Probability of Excluding (CPE) a falsely accused man in a given case equals:

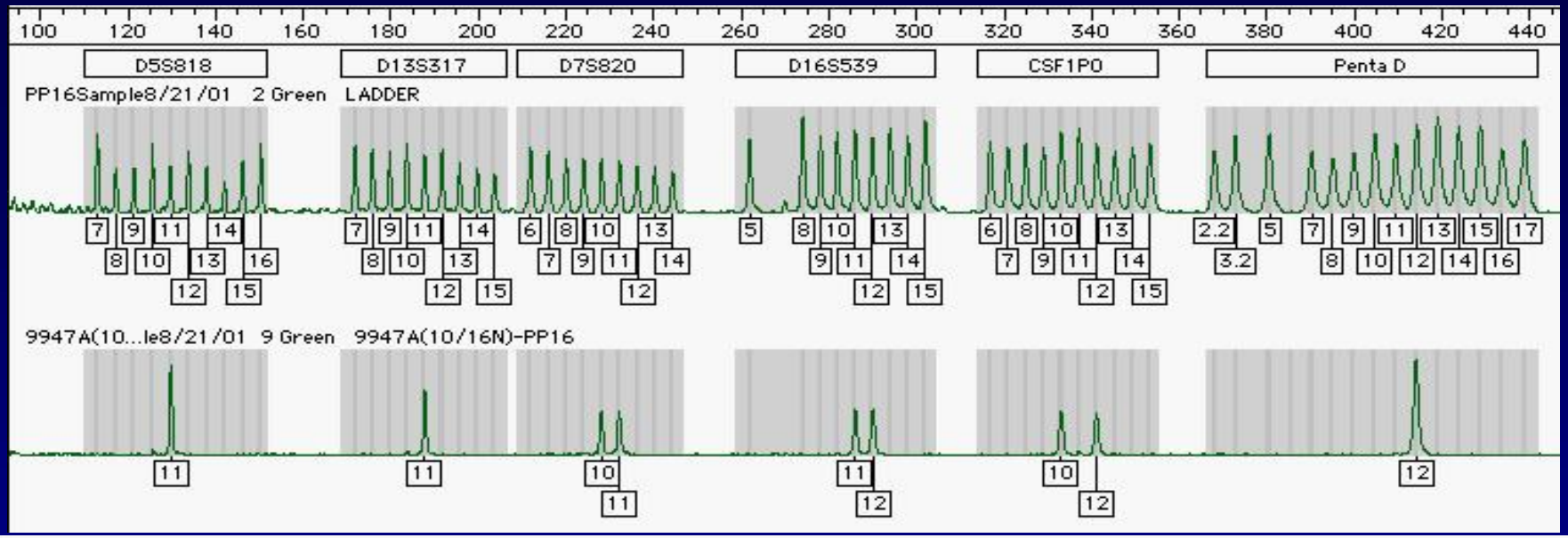
$$1 - [(1 - PE_1) \times (1 - PE_2) \times (1 - PE_3) \ldots \times (1 - PE_N)]$$

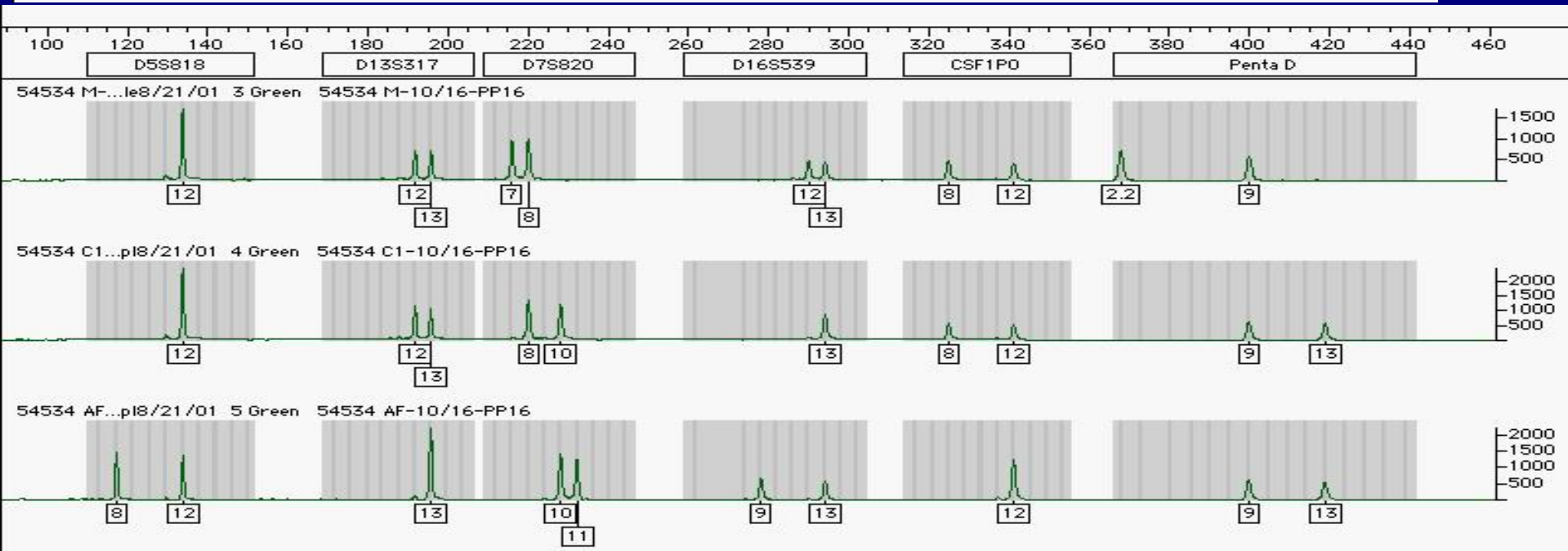
Paternity Trio P-54534



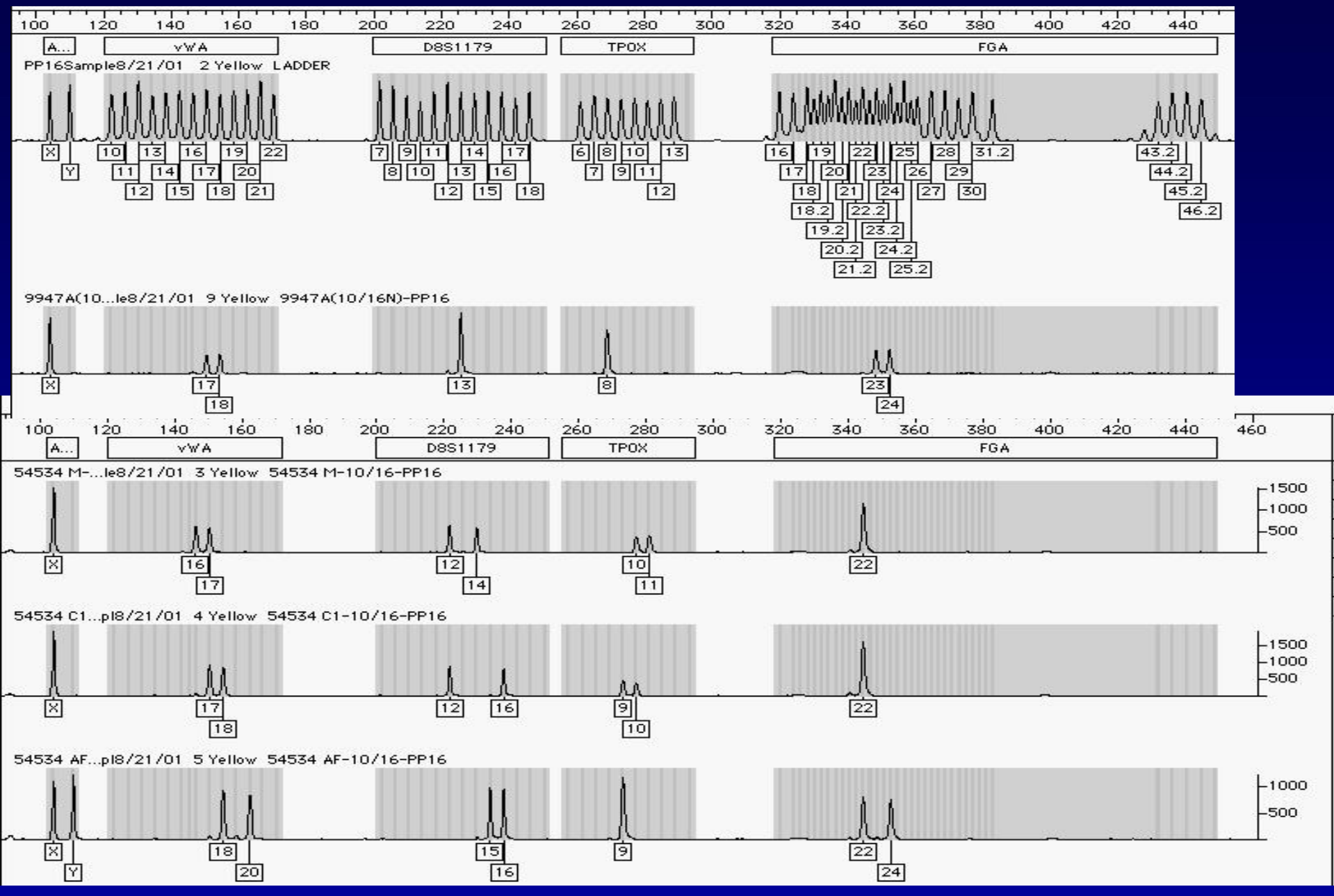


Paternity Trio P-54534





Paternity Trio P-54534



Paternity Trio P-54534

	M	C	AF	Allele Frequency
D3S1358	14	15p	15	15 = 0.2463
(3p)	17	17m	16	
HUMvWA31	16	17m	18	18 = 0.2219
(12p13.3 - p13.2)	17	18p	20	
FGA	22	22	22	22 = 0.1888
(4q28)			24	

Paternity Trio P-54534

	M	C	AF	PI Formula
D3S1358	14	15p	15	0.5/a
(3p)	17	17m	16	
HUMvWA31	16	17m	18	0.5/a
(12p13.3 - p13.2)	17	18p	20	
FGA	22	22	22	0.5/a
(4q28)			24	

Paternity Trio P-54534

	M	C	AF	Paternity Index
D3S1358	14	15p	15	2.03
(3p)	17	17m	16	
HUMvWA31	16	17m	18	2.25
(12p13.3 - p13.2)	17	18p	20	
FGA	22	22	22	2.65
(4q28)			24	

Paternity Trio P-54534

	M	C	AF	PE Formula
D3S1358	14	15p	15	$(1 - a)^2$
(3p)	17	17m	16	
HUMvWA31	16	17m	18	$(1 - a)^2$
(12p13.3 - p13.2)	17	18p	20	
FGA	22	22	22	$(1 - a)^2$
(4q28)			24	

Paternity Trio P-54534

	M	C	AF	PE
D3S1358	14	15p	15	**0.5680**
(3p)	17	17m	16	
HUMvWA31	16	17m	18	**0.6054**
(12p13.3 - p13.2)	17	18p	20	
FGA	22	22	22	**0.6580**
(4q28)			24	

Paternity Trio P-54534

	M	C	AF	Allele Frequency
D8S1179	12	12m	15	
(8)	14	16p	16	**16 = 0.0128**
D21S11	28	28m	28	
(21q11.2 - q21)	30	32p	32	**32 = 0.0153**
D18S51	15	13p	13	
(18q21.3)	19	15m	18	**13 = 0.1224**

Paternity Trio P-54534

	M	C	AF	PI Formula
D8S1179	12	12m	15	0.5/a
(8)	14	16p	16	
D21S11	28	28m	28	0.5/a
(21q11.2 - q21)	30	32p	32	
D18S51	15	13p	13	0.5/a
(18q21.3)	19	15m	18	

Paternity Trio P-54534

	M	C	AF	Paternity Index
D8S1179	12	12m	15	39.06
(8)	14	16p	16	
D21S11	28	28m	28	32.68
(21q11.2 - q21)	30	32p	32	
D18S51	15	13p	13	4.08
(18q21.3)	19	15m	18	

Paternity Trio P-54534

	M	C	AF	PE Formula
D8S1179	12	12m	15	$(1 - a)^2$
(8)	14	16p	16	
D21S11	28	28m	28	$(1 - a)^2$
(21q11.2 - q21)	30	32p	32	
D18S51	15	13p	13	$(1 - a)^2$
(18q21.3)	19	15m	18	

Paternity Trio P-54534

	M	C	AF	PE
D8S1179	12	12m	15	**0.9745**
(8)	14	16p	16	
D21S11	28	28m	28	**0.9696**
(21q11.2 - q21)	30	32p	32	
D18S51	15	13p	13	**0.7701**
(18q21.3)	19	15m	18	

Paternity Trio P-54534

	M	C	AF	Allele Frequency
D5S818	12	12	8	**12 = 0.3538**
(5q21 - q31)			12	
D13S317	12	12	13	**12 = 0.3087**
(13q22 - q31)	13	13		**13 = 0.1097**
D7S820	7	8m	10	**10 = 0.2906**
(7q)	8	10p	11	

Paternity Trio P-54534

	M	C	AF	PI Formula
D5S818	12	12	8	0.5/a
(5q21 - q31)			12	
D13S317	12	12	13	1/(a+b)
(13q22 - q31)	13	13		
D7S820	7	8m	10	0.5/a
(7q)	8	10p	11	

Paternity Trio P-54534

	M	C	AF	Paternity Index
D5S818	12	12	8	1.41
(5q21 - q31)			12	
D13S317	12	12	13	2.39
(13q22 - q31)	13	13		
D7S820	7	8m	10	1.72
(7q)	8	10p	11	

Paternity Trio P-54534

	M	C	AF	PE Formula
D5S818	12	12	8	$(1 - a)^2$
(5q21 - q31)			12	
D13S317	12	12	13	$[1 -(a+b)]^2$
(13q22 - q31)	13	13		
D7S820	7	8m	10	$(1 - a)^2$
(7q)	8	10p	11	

Paternity Trio P-54534

	M	C	AF	PE
D5S818 (5q21 - q31)	12	12	8 12	**0.4175**
D13S317 (13q22 - q31)	12 13	12 13	13	**0.3382**
D7S820 (7q)	7 8	8m 10p	10 11	**0.5032**

Paternity Trio P-54534

	M	C	AF	Allele Frequency
HUMCSF1PO	8	8	12	8 = 0.0123
(5q33.3 - q34)	12	12		12 = 0.3251
HUMTPOX	10	9p	9	9 = 0.1232
(2p23 - 2pter)	11	10m		
HUMTH01	7	9	8	9 = 0.1650
(11p15.5)	9		9	
D16S539	12	13	9	13 = 0.1634
(16p24 - p25)	13		13	

Paternity Trio P-54534

	M	C	AF	PI Formula
HUMCSF1PO	8	8	12	1/(a+b)
(5q33.3 - q34)	12	12		
HUMTPOX	10	9p	9	1/a
(2p23 - 2pter)	11	10m		
HUMTH01	7	9	8	0.5/a
(11p15.5)	9		9	
D16S539	12	13	9	0.5/a
(16p24 - p25)	13		13	

Paternity Trio P-54534

	M	C	AF	Paternity Index
HUMCSF1PO	8	8	12	2.96
(5q33.3 - q34)	12	12		
HUMTPOX	10	9p	9	8.12
(2p23 - 2pter)	11	10m		
HUMTH01	7	9	8	3.03
(11p15.5)	9		9	
D16S539	12	13	9	3.06
(16p24 - p25)	13		13	

Paternity Trio P-54534

	M	C	AF	PE Formula
HUMCSF1PO	8	8	12	
(5q33.3 - q34)	12	12		$[1 - (a+b)]^2$
HUMTPOX	10	9p	9	
(2p23 - 2pter)	11	10m		$(1 - a)^2$
HUMTH01	7	9	8	
(11p15.5)	9		9	$(1 - a)^2$
D16S539	12	13	9	
(16p24 - p25)	13		13	$(1 - a)^2$

Paternity Trio P-54534

	M	C	AF	PE
HUMCSF1PO (5q33.3 - q34)	8 12	8 12	12	**0.4390**
HUMTPOX (2p23 - 2pter)	10 11	9p 10m	9	**0.7687**
HUMTH01 (11p15.5)	7 9	9	8 9	**0.6972**
D16S539 (16p24 - p25)	12 13	13	9 13	**0.6999**

Paternity Trio P-54534
13 Core CODIS Loci

Combined Paternity Index	81,424,694
Probability of Paternity	99.99999%
Probability of Exclusion	99.99999%

Parentage Statistics in Non-Typical Cases

- **Mutation/Recombination – Tested man does not match at a single genetic locus**
- **Tested Man is not the biological father but is related to the biological father (brother, son, or father)**

Case Scenario

A mother, child, and alleged father have been analyzed with the 13 core CODIS STR loci, the alleged father cannot be excluded at 12 loci, however, there is a single non-matching system (single inconsistency), the alleged father does not contain the obligate paternal allele found in the child at one locus.

Three possible explanations can be considered:

1. The alleged father is excluded as the biological father of the child and is unrelated to the true biological father.
2. A mutation or recombination event has occurred altering the allele inherited from the AF by the child.
3. The tested man is not the biological father, but is a 1st order relative of the true biological father, and shares the majority of alleles contributed to the child with the biological father.

Single Inconsistencies in Paternity Testing

- The American Association of Blood Banks, in their standards for parentage testing laboratories, has recognized that mutations are naturally occurring genetic events, and the mutation frequency at a given locus shall be documented (5.4.2).
- Standard 6.4.1 – An opinion of non-paternity shall not be rendered on the basis of an exclusion at a single DNA locus (single inconsistency).

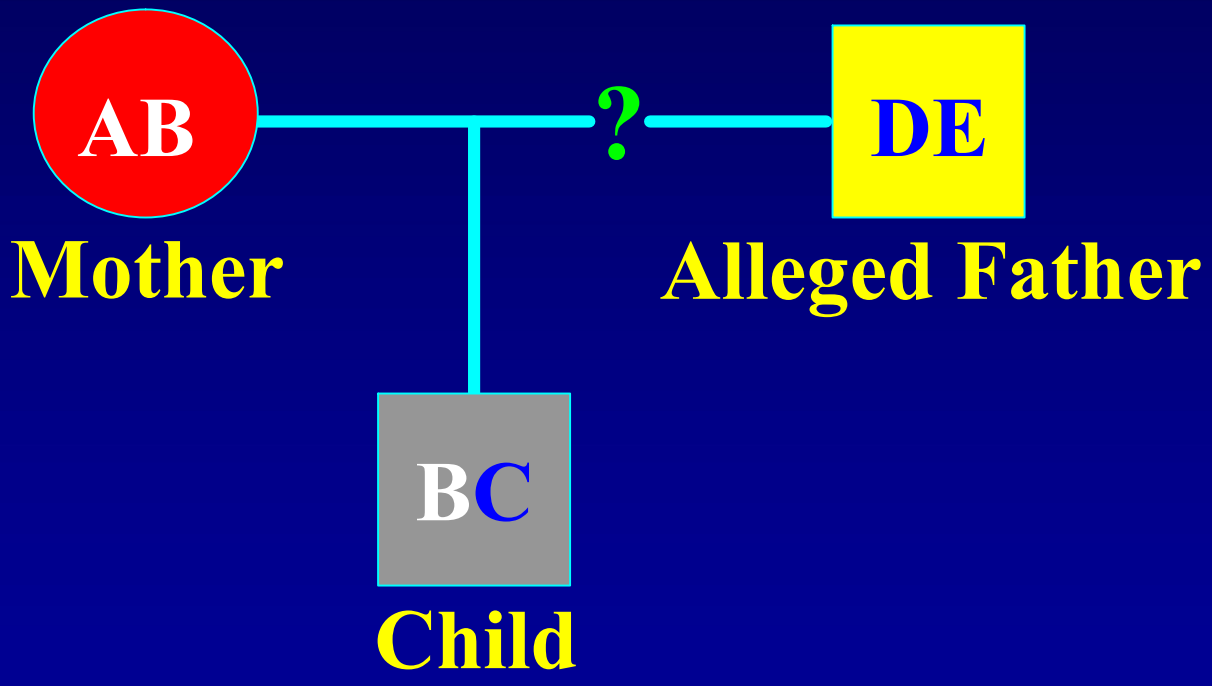
Mutations in Paternity Testing The "Two Exclusion Rule"

- A single inconsistency is not sufficient to render an opinion of non-paternity, therefore, two inconsistencies have been traditionally considered genetic evidence to exclude a tested man and to issue a finding of non-paternity. This rule has been commonly applied in both serological systems and RFLP testing. However, since STR analysis often examines a battery of a dozen or more systems it is not unexpected to occasionally see two inconsistencies in cases were the tested man is the true biological father.

Mutations in Paternity Testing Calculating a Paternity Index

- In cases with a single non-matching system, the laboratory cannot simply ignore the inconsistent locus. A paternity index must be calculated for the inconsistent locus, which takes into account the possibility of a mutation.
- The paternity index for a single inconsistency seen in the 13 Core CODIS STR loci is a relatively small number. The system PI is greater than zero but substantially less than one.

Single Inconsistency
Calculating a Paternity Index



Single Inconsistency

Numerator

Person	Type
Mother	AB
Child	BC
Alleged Father	DE

In order to explain this evidence Calculate Probability that

a) Woman randomly selected from population is type AB

b) Man randomly selected from population is type DE, and

c) Their child is type BC

Single Inconsistency

Numerator

Person	Type
Mother	AB
Child	BC
Alleged Father	DE

In order to explain this evidence the numerator must calculate the probability that a man without a C allele will contribute a C allele

X = P(man without C allele will contribute C allele)

= P(contributed gene will mutate) x P(mutated gene will be a C)

Single Inconsistency
Numerator

X = P(man without C will contribute C)

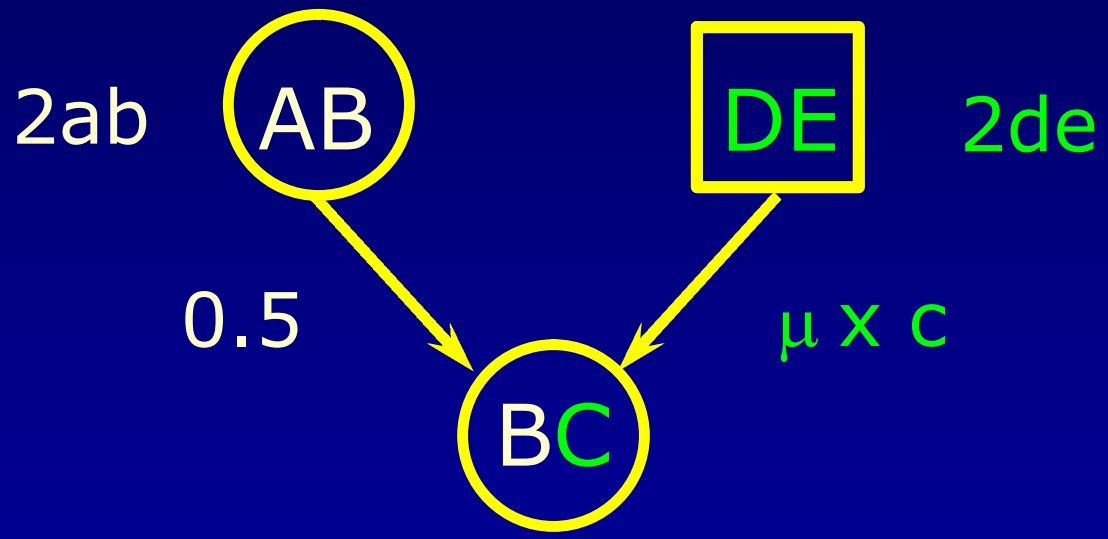
X = P(contributed gene will mutate)
x P(mutated gene will be a C)

μ = observed rate of mutations/meiosis for the locus

P(mutated gene will be a C) ie. Frequency of C allele = **c**

$$X = \mu \times c$$

Single Inconsistency
Calculating a Paternity Index
Numerator



Probability = 2ab x 2de x 0.5 x μ x c

Single Inconsistency
Denominator

Person	Type
Mother	AB
Child	BC
Alleged Father	DE

In order to explain this evidence Calculate Probability that

a) Woman randomly selected from population is type AB

b) An alternative man randomly selected from population is type DE, and

c) The woman's child, fathered by random man, is type BC

Single Inconsistency
Denominator

Person	Type
Mother	AB
Child	BC
Alleged Father	DE

In order to explain this evidence the denominator must calculate the probability that the paternal allele is C and a random man would have a genotype inconsistent with paternity at this locus

Y = P(paternal allele is C and random man has no C allele)

= P(paternal gene is C) x P(random man has no C allele)

Single Inconsistency
Denominator

Y = P(paternal allele is C and random man has no C allele)

= P(paternal gene is C) x P(random man has no C allele)

P(paternal allele will be a C) ie. Frequency of C allele = **c**

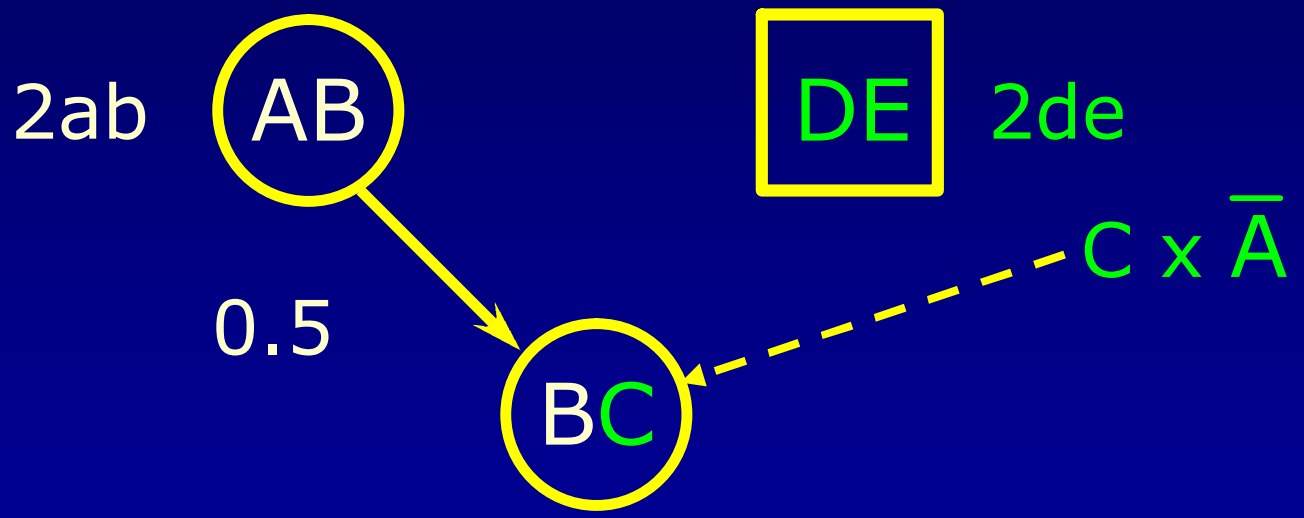
P(random man has no C allele) = probability of exclusion

The AABB does not use the case specific power of exclusion, but the mean power of exclusion ($\overline{A}$)

$$Y = c \cdot \overline{A}$$

Single Inconsistency
Calculating a Paternity Index

Denominator



Probability = 2ab x 2de x 0.5 x c x $\bar{A}$

Single Inconsistency Paternity Index

$$PI = \frac{2ab \times 2de \times 0.5 \times \mu \times c}{2ab \times 2de \times 0.5 \times c \times \bar{A}}$$

$$PI = \frac{\mu}{\bar{A}}$$

Mutation Rates and Mean Power of Exclusion for CODIS Core STR Loci

Locus	Mutation Rate	Mean PE
CSF1PO	0.0013	0.455
TPOX	0.0005	0.537
TH01	0.0003	0.503
vWA	0.0034	0.667
D16S539	0.0013	0.590
D7S820	0.0019	0.648
D13S317	0.0017	0.582
D5S818	0.0017	0.566

Mutation Rates and Mean Power of Exclusion for CODIS Core STR Loci

Locus	Mutation Rate	Mean PE
FGA	0.0030	0.750
D8S1179	0.0019	0.554
D18S51	0.0032	0.740
D21S11	0.0010	0.791
D3S1358	0.0010	0.596

Mutation Rates and Mean Power of Exclusion for Additional STR Loci

Locus	Mutation Rate	Mean PE
F13AO1	0.0009	0.577
FESFPS	0.0007	0.620
F13B	0.0005	0.507
LIPOL	0.0012	0.451
PENTA E	0.0012	0.797

Single Inconsistency
P-41411

	M	C	AF	PI Formula
HUMCSF1PO	12 8	12 8	12 10	0.5/(a+b)]
HUMTPOX	11 8	11 8	11 8	1/(a+b)
HUMTH01	7	9p 7m	9 6	0.5/a
HUMvWA31	19 18	19m 16p	17 15	$\mu/\bar{A}$ (0.0034/0.667)

Single Inconsistency
P-41411

	M	C	AF	Paternity Index
HUMCSF1PO	12 8	12 8	12 10	1.52
HUMTPOX	11 8	11 8	11 8	1.25
HUMTH01	7	9p 7m	9 6	3.03
HUMvWA31	19 18	19m 16p	17 15	0.005

Single Inconsistency
P-41411

	M	C	AF	PI Formula
D16S539	12	12m	12	0.5/a
		11p	11	
D7S820	10	11p	11	0.5/a
	9	9m	10	
D13S317	12	12m	11	0.5/a
	10	8p	8	
D5S818	13	11	11	1/a
	11			

Single Inconsistency
P-41411

	M	C	AF	Paternity Index
D16S539	12	12m	12	1.84
		11p	11	
D7S820	10	11p	11	2.48
	9	9m	10	
D13S317	12	12m	11	5.03
	10	8p	8	
D5S818	13	11	11	2.44
	11			

Single Inconsistency

P-41411

	M	C	AF	PI Formula
FGA	24 23	24m 21p	23 21	0.5/a
D18S51	17 14	17 14	14	1/(a+b)
D21S11	30 28	30m 29p	29 28	0.5/a
D3S1358	15 14	14	15 14	0.5/a
D8S1179	14 10	15p 14m	15 13	0.5/a

Single Inconsistency
P-41411

	M	C	AF	Paternity Index
FGA	24 23	24m 21p	23 21	2.88
D18S51	17 14	17 14	14	3.04
D21S11	30 28	30m 29p	29 28	2.76
D3S1358	15 14	14	15 14	3.56
D8S1179	14 10	15p 14m	15 13	4.56

Paternity Trio with a Single Inconsistency

12 STR without vWA

Combined Paternity Index	126,476
Probability of Paternity	99.9992%

Single Inconsistency at vWA

Combined Paternity Index	632
Probability of Paternity	99.84%

Single Inconsistencies in Paternity Testing

A mutation may be one of the possible explanations, the genetic results could suggest that a close relative (such as a brother, child or father) may be the biological father.

Single Inconsistencies in Paternity Testing

When considering brothers, on average a tested man and his brother will share 50% of their alleles… each can contribute these alleles in a random manner. This is also true between a father and son of a tested man.

Avuncular Index
AI

We can use the development of a likelihood ratio to test two competing hypotheses:

H_1: The tested man's brother is the biological father of the child

H_2: A random man is the biological father of the child

Avuncular Index

Numerator

H_1: The tested man's brother is the biological father of the child

$$H_1 = \frac{X + Y}{2}$$

$$H_1 = 0.5\,X + 0.5\,Y$$

Avuncular Index

Denominator

H_2: A random man is the biological father of the child

$$H_2 = Y$$

Avuncular Index
AI

The Avuncular Index for any system can be written as:

$$AI = \frac{0.5\,X + 0.5\,Y}{Y}$$

$$AI = \frac{PI + 1}{2}$$

Single Inconsistency
P-41411

	M	C	AF	Paternity Index	Avuncular Index
HUMCSF1PO	12 8	12 8	12 10	1.52	1.26
HUMTPOX	11 8	11 8	11 8	1.25	1.13
HUMTH01	7	9p 7m	9 6	3.03	2.02
HUMvWA31	19 18	19m 16p	17 15	0.005	0.50

Single Inconsistency
P-41411

	M	C	AF	Paternity Index	Avuncular Index
D16S539	12	12m	12	1.84	1.42
		11p	11		
D7S820	10	11p	11	2.48	1.74
	9	9m	10		
D13S317	12	12m	11	5.03	3.02
	10	8p	8		
D5S818	13	11	11	2.44	1.72
	11				

Single Inconsistency
P-41411

	M	C	AF	Paternity Index	Avuncular Index
FGA	24 23	24m 21p	23 21	2.88	1.94
D18S51	17 14	17 14	14	3.04	2.02
D21S11	30 28	30m 29p	29 28	2.76	1.88
D3S1358	15 14	14	15 14	3.56	2.28
D8S1179	14 10	15p 14m	15 13	4.56	2.78

Paternity Trio with a Single Inconsistency

13 Core CODIS STR Loci

Combined Paternity Index	632
Combined Avuncular Index	862

Single Inconsistency
P-41411

	M	C	AF	Paternity Index	Avuncular Index
F13AO1	7 12	7 12	12	4.83	2.92
FESFPS	11 12	11	11 12	1.41	1.21
F13B	9	9	8 9	2.06	1.53
LIPOL	10 11	10m 13p	13	16.95	8.98
PENTA E	14 15	13p 14m	13 15	3.85	2.43

Paternity Trio with a Single Inconsistency

18 STR Loci

Combined Paternity Index	578,603
Combined Avuncular Index	101,683

We can use a likelihood ratio to test two competing hypotheses:

H_1: The tested man (alleged father) is the biological father of the child

H_2: The tested man's brother is the biological father of the child

We can use a likelihood ratio to test two competing hypotheses:

$$\frac{\text{Combined Paternity Index}}{\text{Combined Avuncular Index}}$$

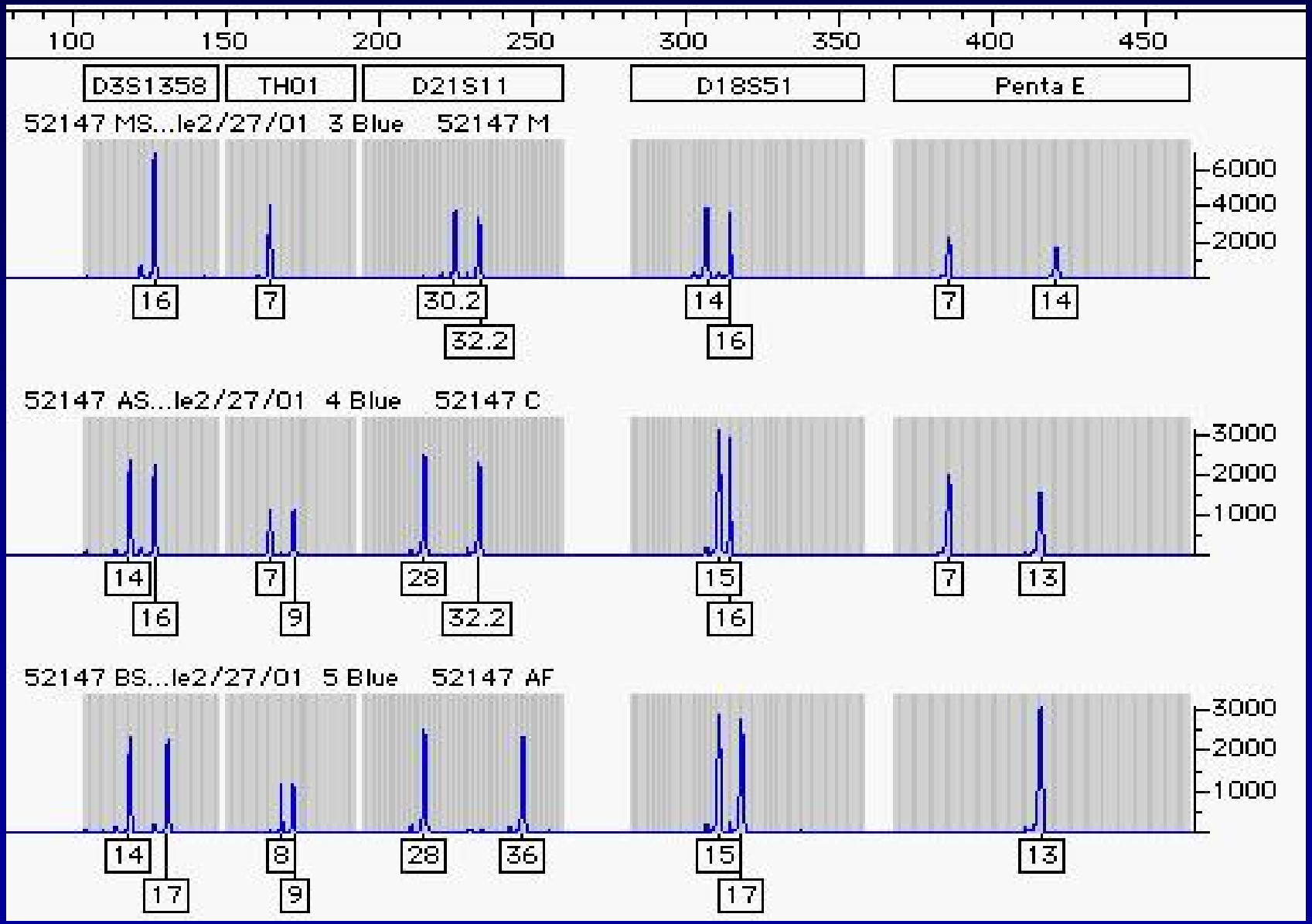
$$\frac{578{,}603}{101{,}683} = 5.69$$

The observed genetic results are 5.7-times more likely to occur under the scenario that the tested man is the father of the child, as opposed to the scenario that the tested man was the uncle of the child.

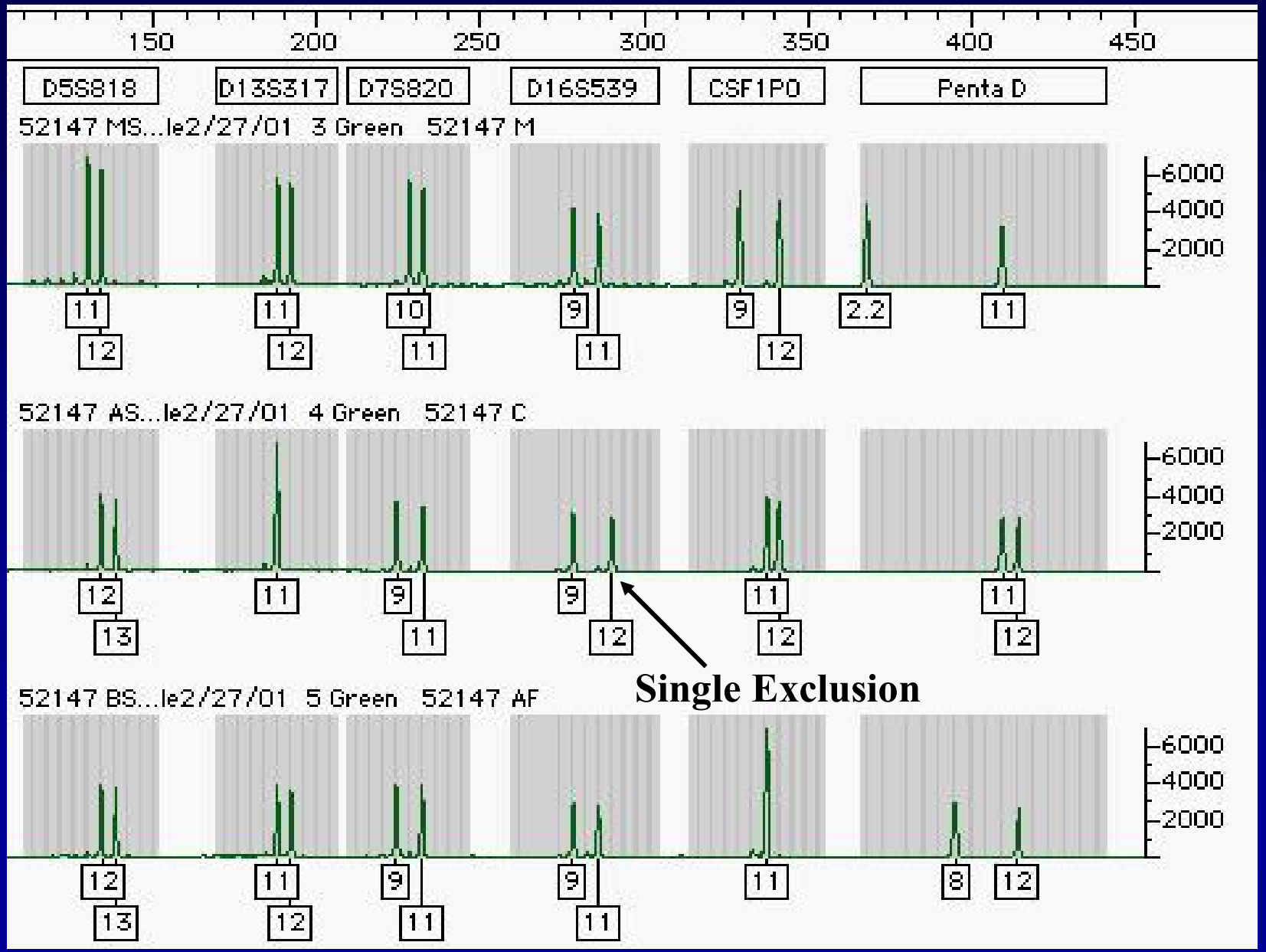
PowerPlex™ 16 System

Extremely Useful in Cases with a Single Non-Matching Locus

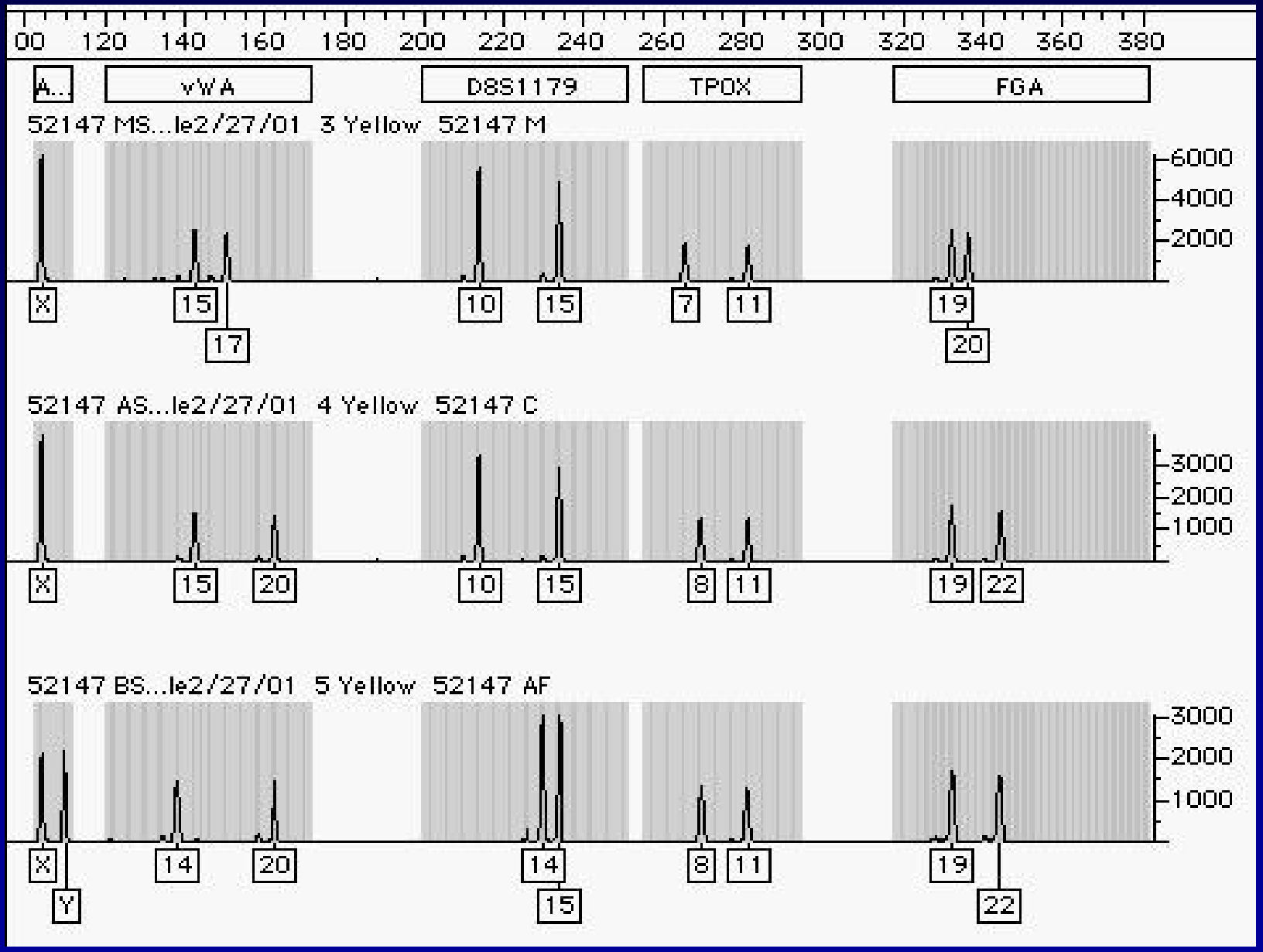
P-52147 Case of Single Exclusion



P-52147 Case of Single Exclusion



P-52147 Case of Single Exclusion



P-52147 Case of Single Exclusion
PowerPlex™ 16 System

13 STR loci minus Penta D & Penta E

Residual Combined Paternity Index	1,914
Probability of Exclusion	99.99997%
Probability of Paternity(prior=0.5)	99.95%

15 STR loci with Penta D & Penta E

Residual Combined Paternity Index	37,699
Probability of Exclusion	99.999998%
Probability of Paternity(prior=0.5)	99.997%

Popstats Cannot Correctly Calculate Parentage Statistics in Non-Typical Cases

Paternity Index
Only Man and Child Tested

- Observe two types – from a man and a child
- Assume true duo– the man is the father of the child
- Assume false duo – the man is not the father of the child (simply two individuals selected at random)
- In the false duo the child's father is a man of unknown type, selected at random from population (unrelated to tested man)

Paternity Index

Only Man and Child Tested

Hypothetical case

DNA Analysis Results in Two Genotypes

Mother	**Not Tested**
Child	**(AB)**
Alleged Father	**(AC)**

Motherless Paternity Index

PI determination in hypothetical DNA System

PI = X / Y

Numerator

X = **is the probability that (1) a man randomly selected from a population is type AC, and (2) his child is type AB.**

X = **Pr{AF passes *A*} x Pr {M passes *B*} + Pr{AF passes *B*} x Pr{M passes *A*}**

Motherless Paternity Index

PI determination in hypothetical DNA System

$$PI = X / Y$$

Denominator

Y = **is the probability that (1) a man randomly selected and unrelated to tested man is type AC, and (2) a child unrelated to the randomly selected man is AB.**

Y = **Pr{RM passes *A*} x Pr {M passes *B*} +**

Pr{RM passes *B*} x Pr{M passes *A*}

Motherless Paternity Index

- **When the mother's genetic data is present, Pr{M passes *A*} is 0, 0.5, or 1, and Pr{M passes *B*} is 0, 0.5, or 1**
- **Without the mother's data, Pr {M passes *A*} becomes the frequency of the gametic allele, *p and* Pr {M passes *B*} becomes the frequency of the gametic allele, *q* .**

Motherless Paternity Index

So, if we have a heterozygous child *AB*, and a heterozygous Alleged Father AC then

X = Pr{AF passes *A*} x Pr {M passes *B*} +

Pr{AF passes *B*} x Pr{M passes *A*}

X = Pr{AF passes *A*} x *q* + Pr{AF passes *B*} x *p*

Pr{AF passes *A*} = 0.5

Pr{AF passes *B*} = 0

X = 0.5 x *q* + 0 x *p*

X = 0.5*q*

Motherless Paternity Index

So, if we have a heterozygous child *AB*, and a heterozygous Alleged Father AC then

$$Y = \Pr\{RM \text{ passes } A\} \times \Pr\{M \text{ passes } B\} + \Pr\{RM \text{ passes } B\} \times \Pr\{M \text{ passes } A\}$$

$$Y = p \times q + q \times p$$

$$Y = 2pq$$

Motherless Paternity Index

So, if we have a heterozygous child *AB*, and a heterozygous Alleged Father AC then

PI = X / Y

X = 0.5*q*

Y = 2pq

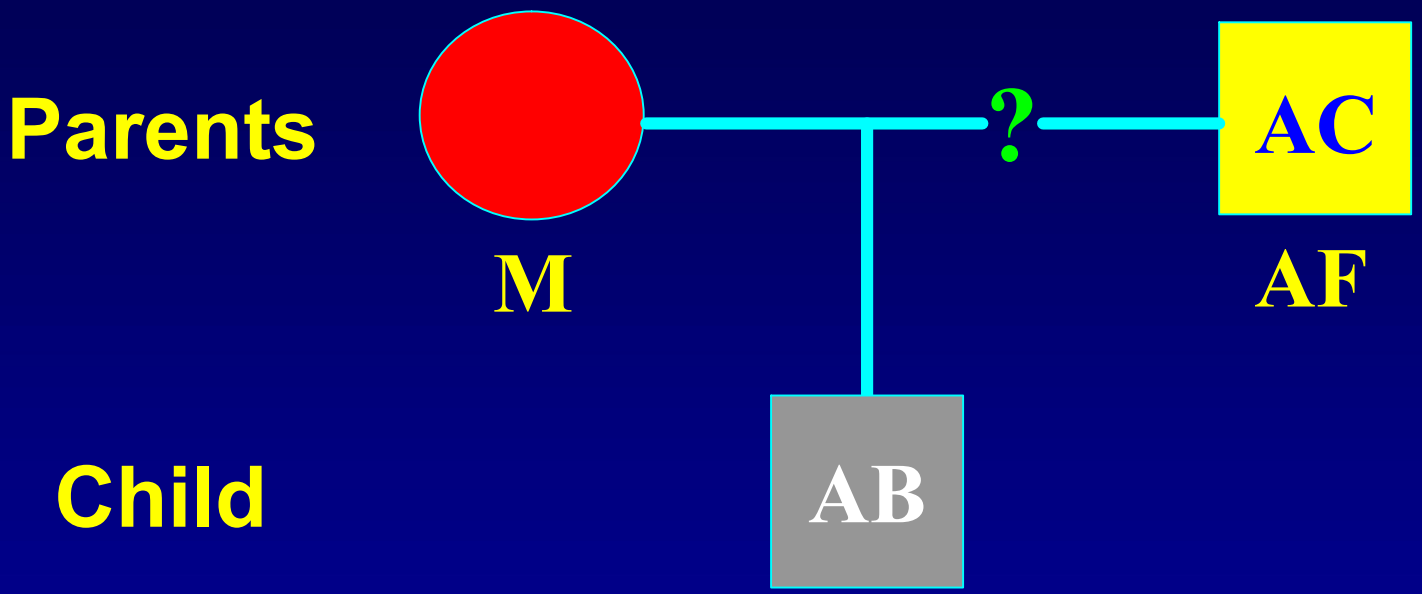
PI = 0.5*q* / 2pq

PI = 0.25/p

PI = 1/4p

Paternity Index

Only Man and Child Tested

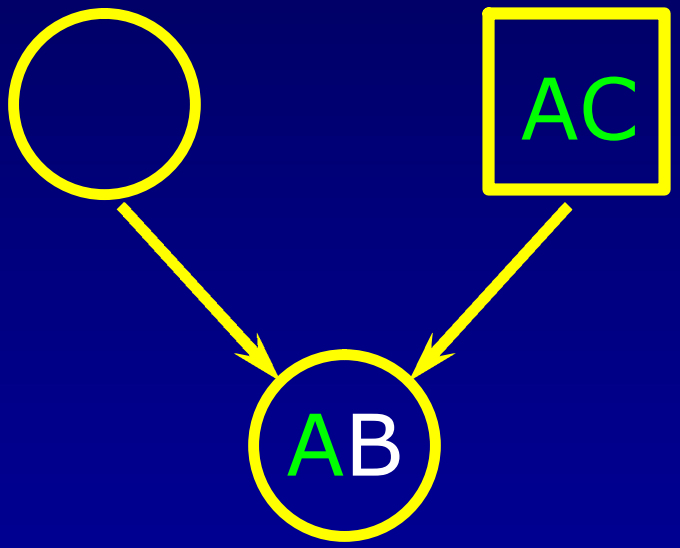


The untested Mother could have passed either the A or B allele

AF has a 1 in 2 chance of passing A allele

RM has (p + q) chance of passing the A or B allele

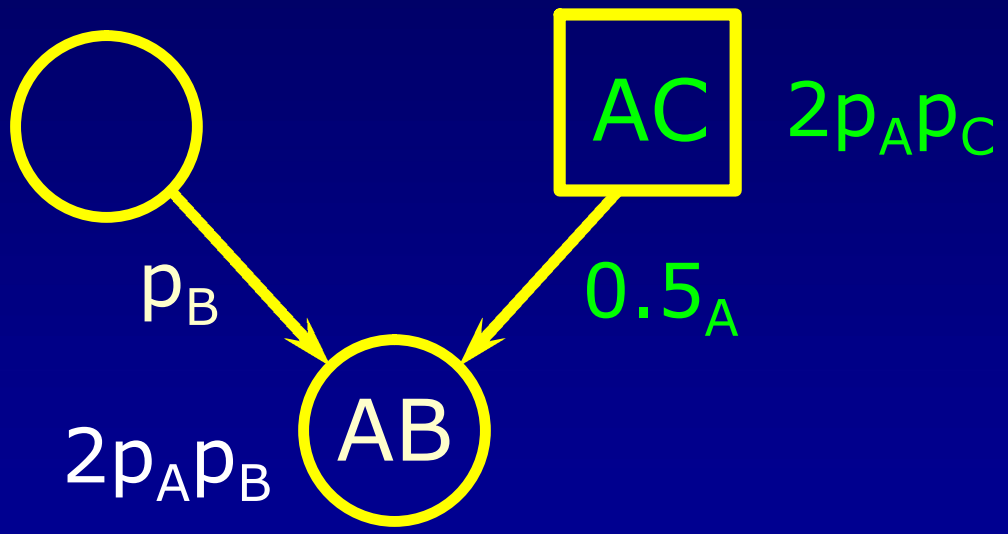
Paternity Index
Only Man and Child Tested



Paternity Index

Only Man and Child Tested

Numerator

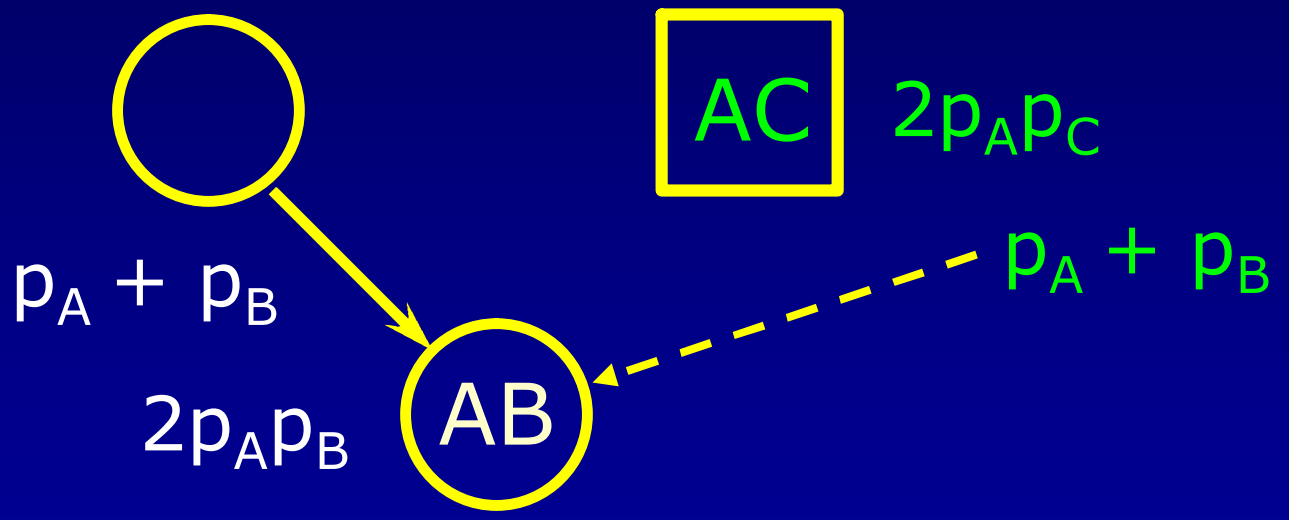


Probability = $2p_Ap_C$ x $2p_Ap_B$ x $0.5_{(fA)}$ x p_B

Paternity Index

Only Man and Child Tested

Denominator



probability =

$$2p_Ap_C \times 2p_Ap_B \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})$$

Paternity Index
Only Man and Child Tested

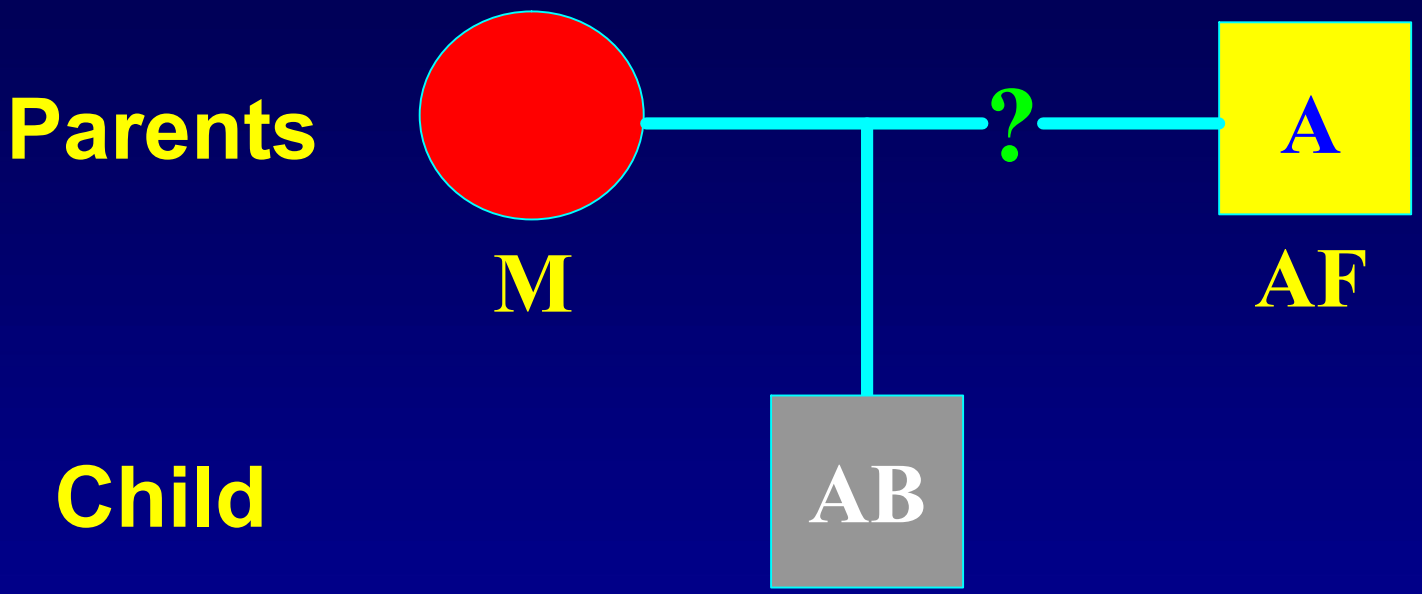
$$PI = \frac{2p_A p_B \times 2p_A p_C \times 0.5_{(mA)} \times p_B}{2p_A p_B \times 2p_A p_C \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})}$$

$$PI = \frac{0.5 p_B}{2p_A p_B}$$

$$PI = \frac{0.25}{p_A}$$

Paternity Index

Only Man and Child Tested

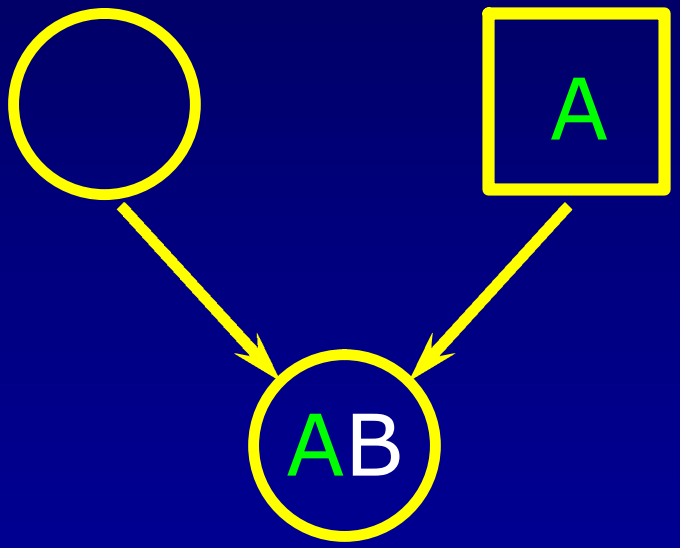


The untested Mother could have passed either the A or B allele

AF can only pass A allele

RM has (p + q) chance of passing the A or B allele

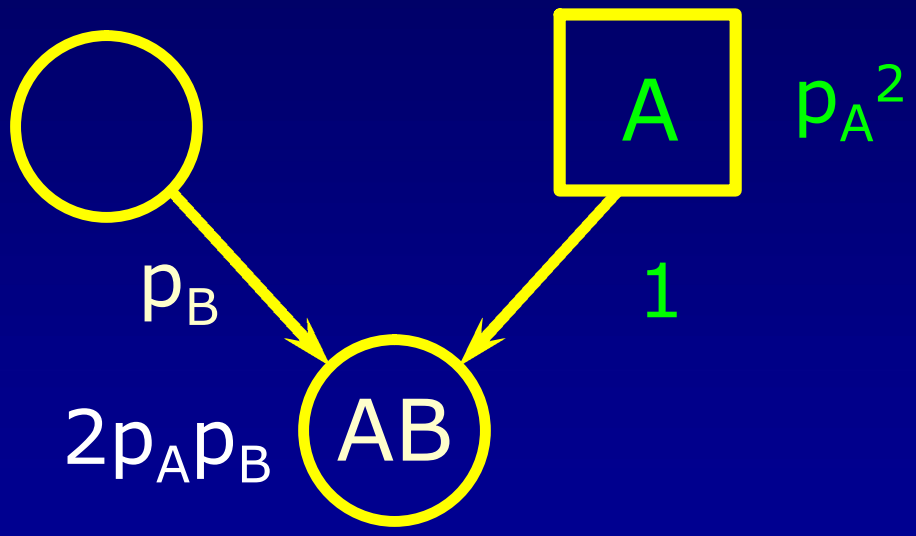
Paternity Index
Only Man and Child Tested



Paternity Index

Only Man and Child Tested

Numerator

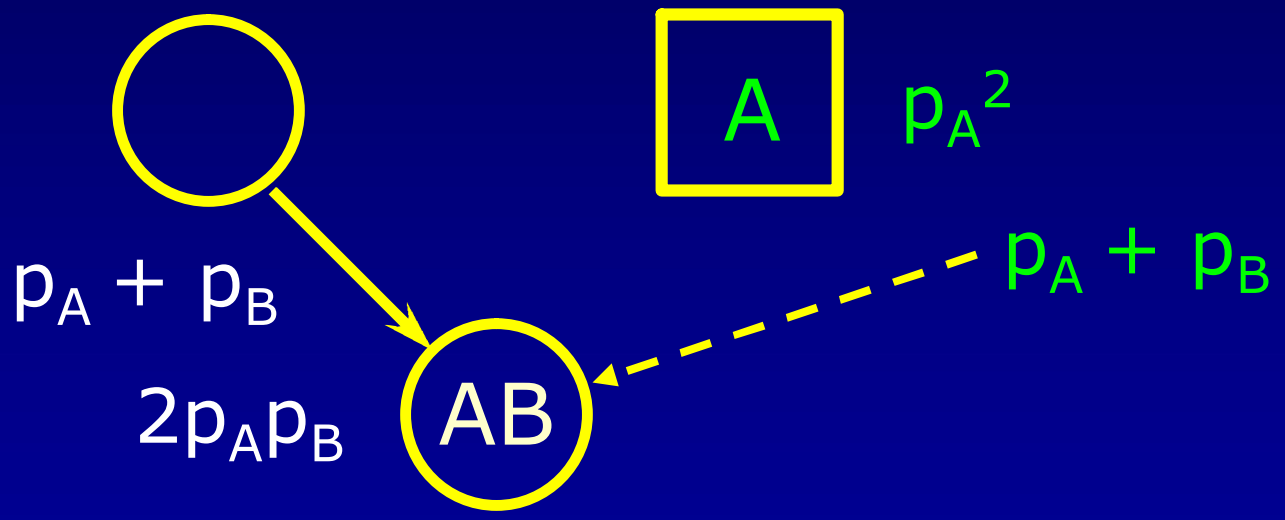


Probability = $p_A^2 \times 2p_Ap_B \times 1_{(fA)} \times p_B$

Paternity Index

Only Man and Child Tested

Denominator



probability =

$$p_A^2 \times 2p_Ap_B \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})$$

Paternity Index
Only Man and Child Tested

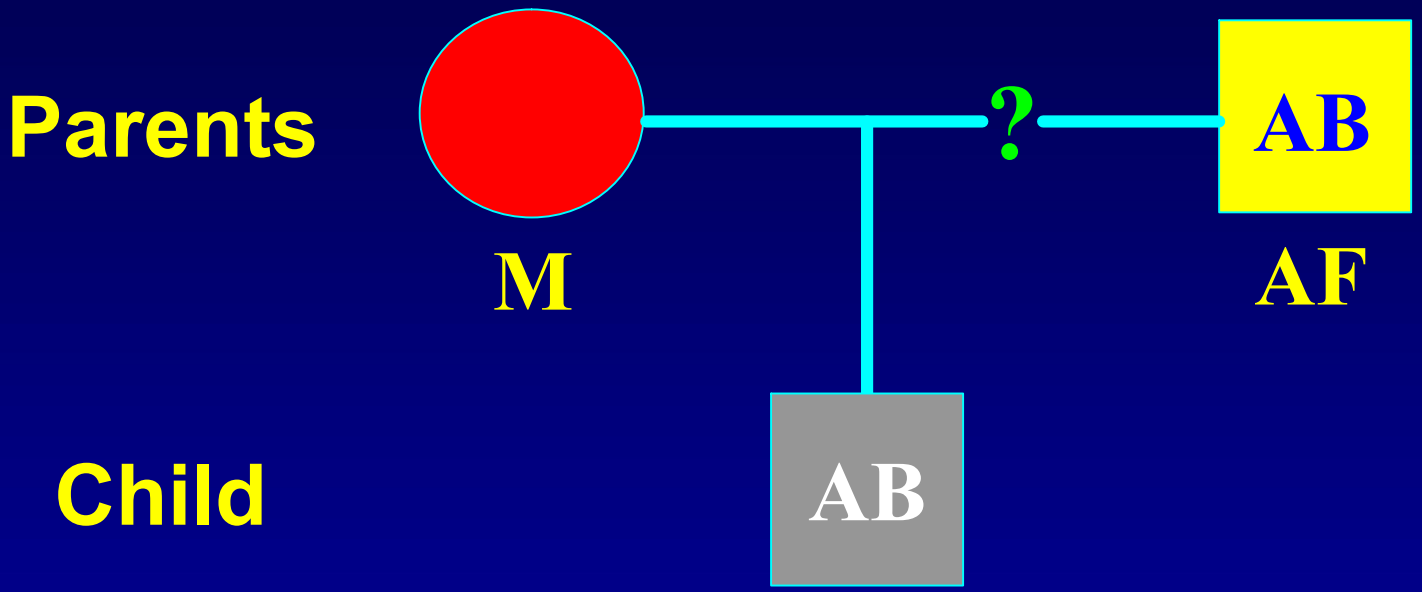
$$PI = \frac{\cancel{p_A^2 \times 2p_Ap_C} \times 1_{(mA)} \times p_B}{\cancel{p_A^2 \times 2p_Ap_C} \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})}$$

$$PI = \frac{\cancel{p_B}}{2p_A\cancel{p_B}}$$

$$PI = \frac{0.5}{p_A}$$

Paternity Index

Only Man and Child Tested

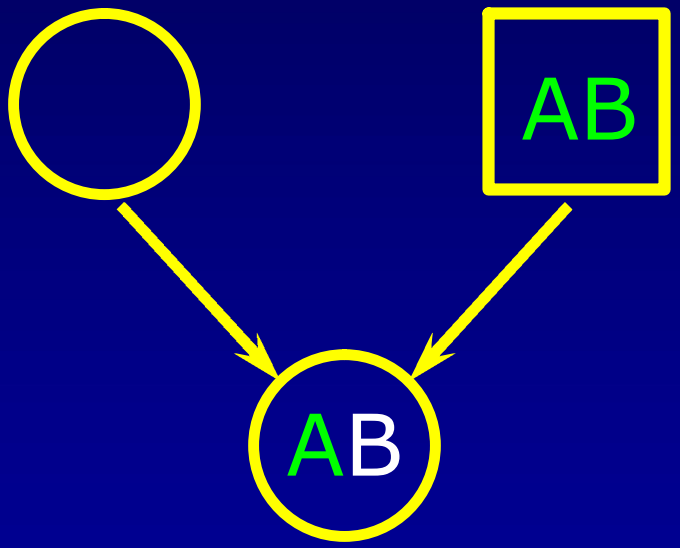


The untested Mother could have passed either the A or B allele

AF can pass either A or B allele

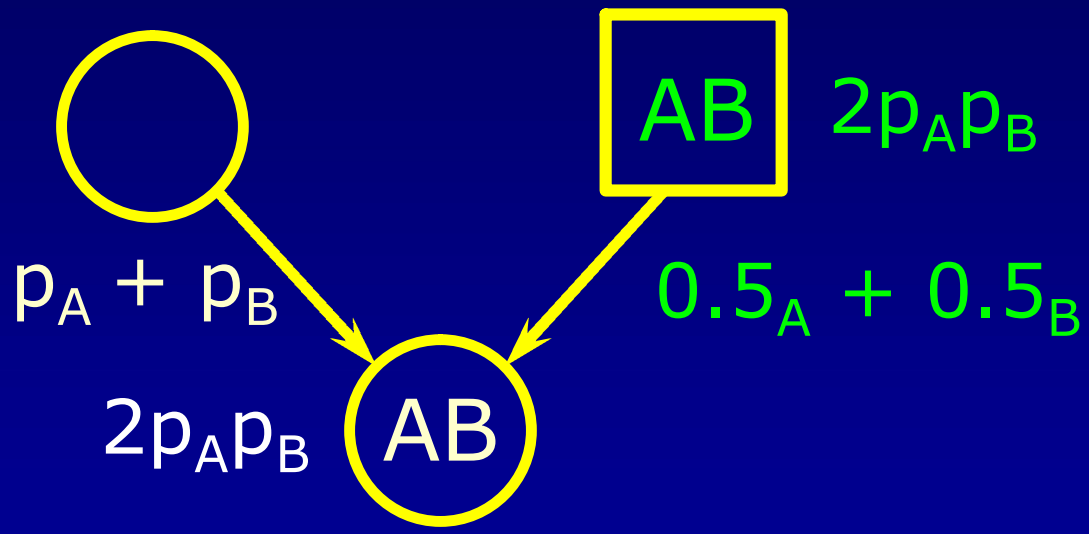
RM has (p + q) chance of passing the A or B allele

Paternity Index
Only Man and Child Tested



Paternity Index
Only Man and Child Tested

Numerator



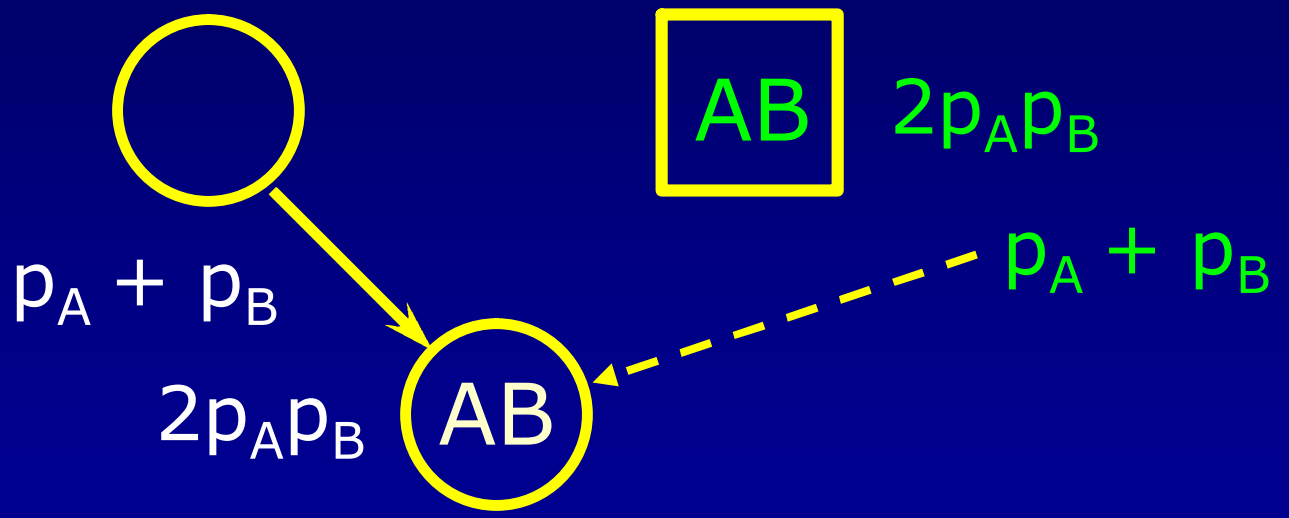
Probability =

$$2p_Ap_B \times 2p_Ap_B \times (0.5_{(fA)} \times p_B + 0.5_{(fB)} \times p_A)$$

Paternity Index

Only Man and Child Tested

Denominator



probability =

$$2p_Ap_B \times 2p_Ap_B \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})$$

Paternity Index
Only Man and Child Tested

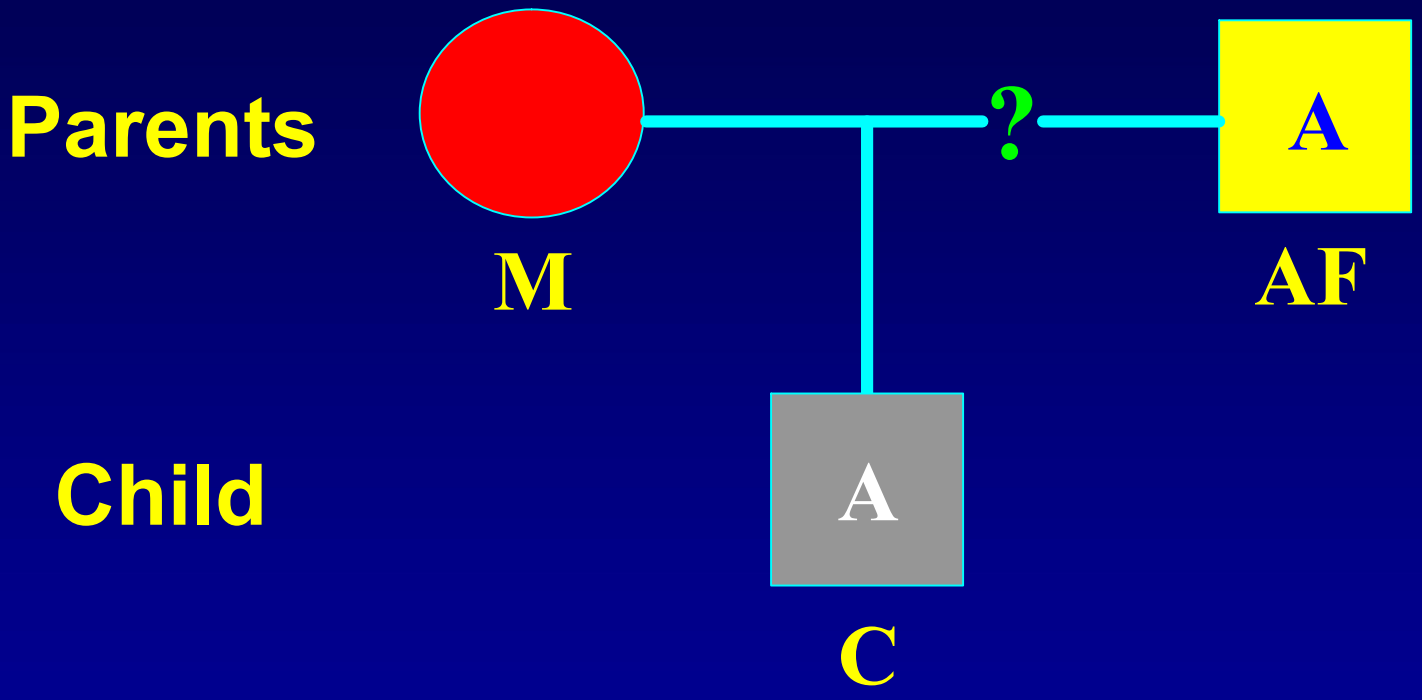
$$PI = \frac{\cancel{2p_A p_B} \times \cancel{2p_A p_B} \times (0.5_{(fA)} \times p_B + 0.5_{(fB)} \times p_A)}{\cancel{2p_A p_B} \times \cancel{2p_A p_B} \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})}$$

$$PI = \frac{0.5p_B + 0.5p_A}{2p_A p_B}$$

$$PI = \frac{p_A + p_B}{4p_A p_B}$$

Paternity Index

Only Man and Child Tested

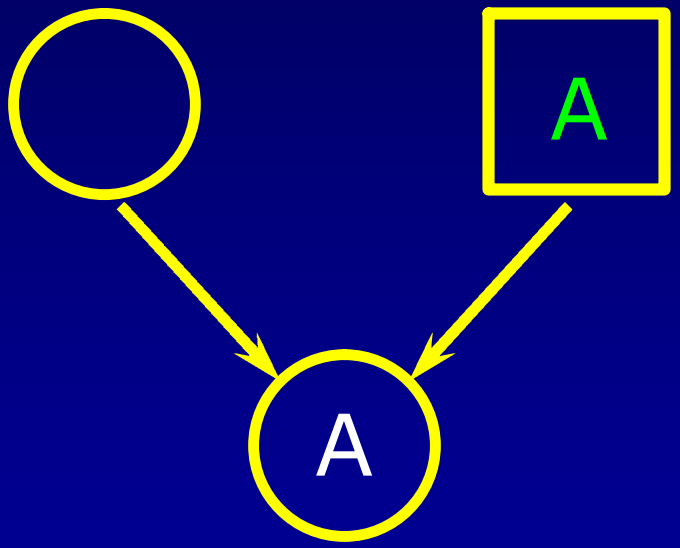


The untested Mother would have to pass an A allele

AF can pass only the A allele

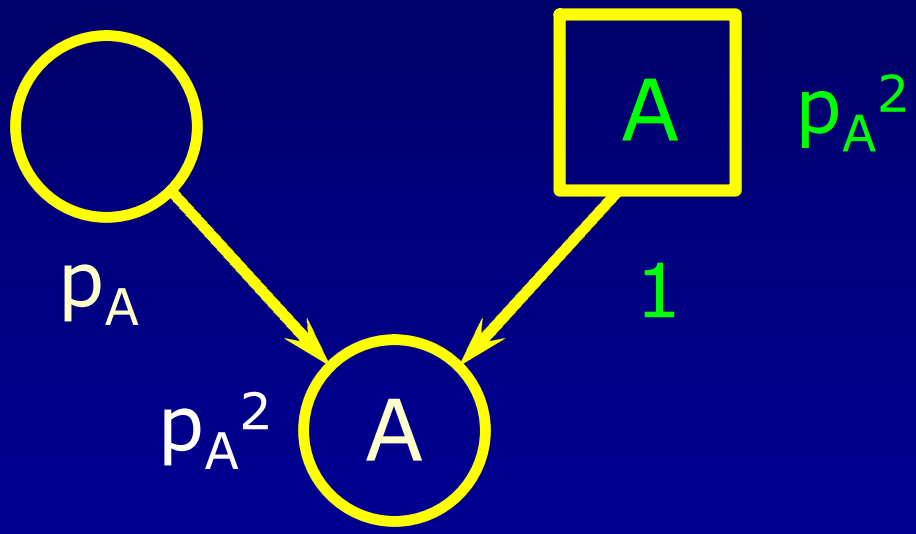
RM has p chance of passing the A allele

Paternity Index
Only Man and Child Tested



Paternity Index
Only Man and Child Tested

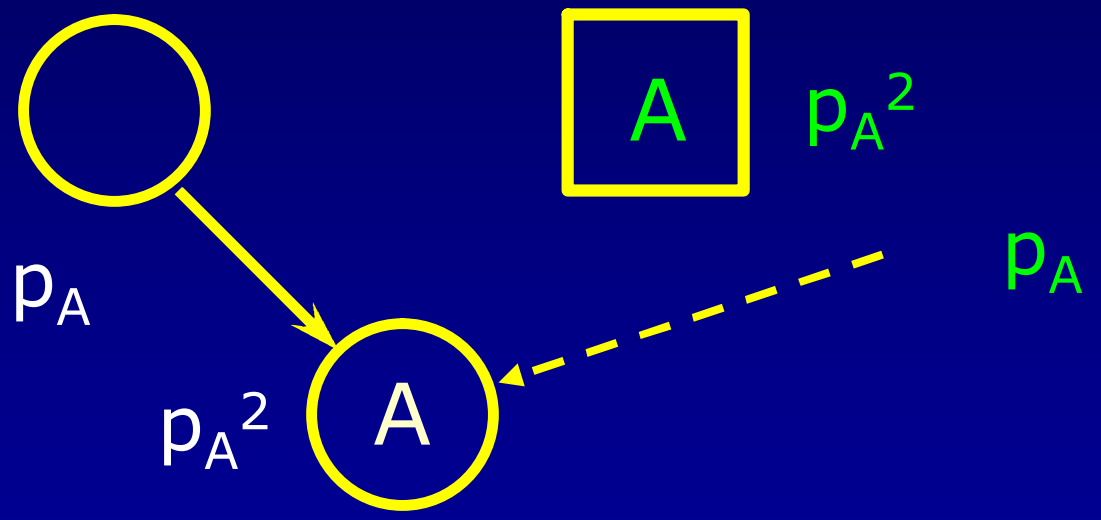
Numerator



Probability = $p_A^2 \times p_A^2 \times 1_{(fA)} \times p_A$

Paternity Index
Only Man and Child Tested

Denominator



probability = $p_A^2 \times p_A^2 \times p_{(mA)} \times p_{(fA)}$

Paternity Index
Only Man and Child Tested

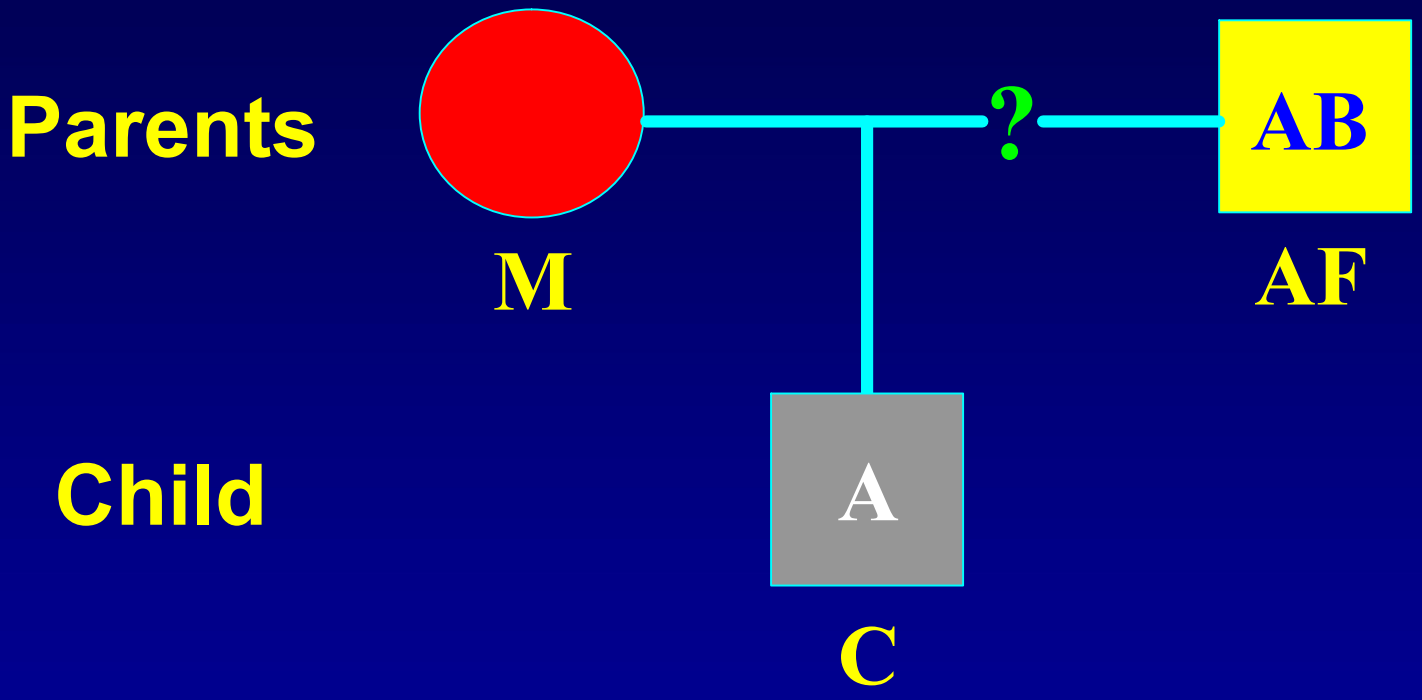
$$PI = \frac{\cancel{p_A^2} \times \cancel{p_A^2} \times 1_{(fA)} \times p_A}{\cancel{p_A^2} \times \cancel{p_A^2} \times p_{(mA)} \times p_{(fA)}}$$

$$PI = \frac{\cancel{p_A}}{\cancel{p_A} \times p_A}$$

$$PI = \frac{1}{p_A}$$

Paternity Index

Only Man and Child Tested

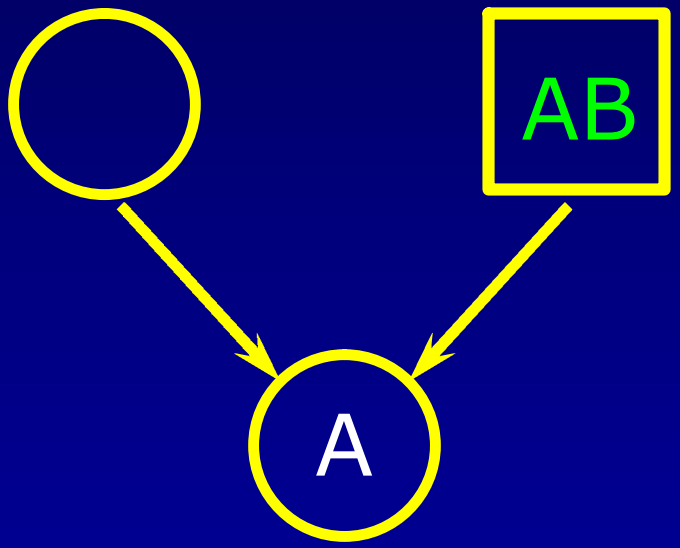


The untested Mother would have to pass an A allele

AF would have to pass the A allele

RM has p chance of passing the A allele

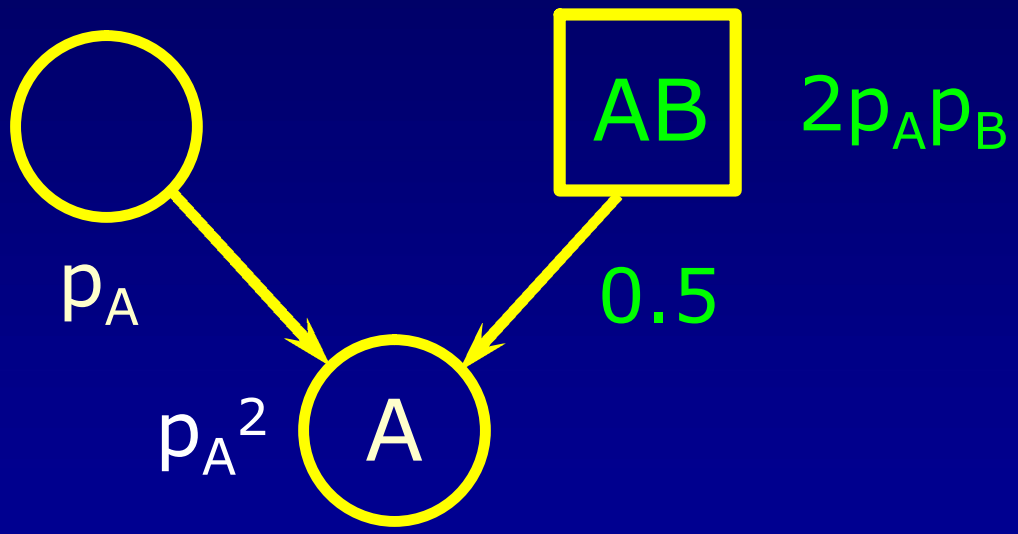
Paternity Index
Only Man and Child Tested



Paternity Index

Only Man and Child Tested

Numerator

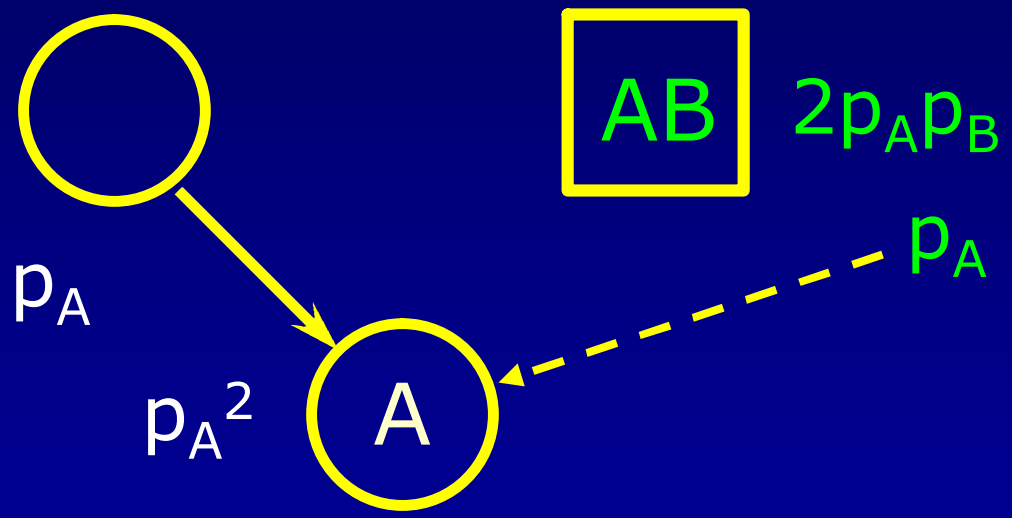


Probability = $2p_Ap_B \times p_A^2 \times 0.5_{(fA)} \times p_A$

Paternity Index

Only Man and Child Tested

Denominator



probability = $2p_Ap_B \times p_A^2 \times p_{(mA)} \times p_{(fA)}$

Paternity Index
Only Man and Child Tested

$$PI = \frac{\cancel{2p_A p_B} \times \cancel{p_A}^2 \times 0.5_{(fA)} \times p_A}{\cancel{2p_A p_B} \times \cancel{p_A}^2 \times p_{(mA)} \times p_{(fA)}}$$

$$PI = \frac{0.5\cancel{p_A}}{\cancel{p_A} \times p_A}$$

$$PI = \frac{0.5}{p_A}$$

Paternity Index
Only Man and Child Tested
Formulas

Single locus, no null alleles, low mutation rate, codominance

C	AF	Numerator	Denominator	PI	PE
AB	AC	0.5b	2ab	0.25/a	$[1-(a + b)]^2$
AB	AB	0.5(a+b)	2ab	(a+b)/4ab	$[1-(a + b)]^2$
AB	A	b	2ab	0.5/a	$[1-(a + b)]^2$
A	AC	0.5a	a^2	0.5/a	$(1-a)^2$
A	A	a	a^2	1/a	$(1-a)^2$

MOTHERLESS PATERNITY CASE P-41376

	C	AF	Allele Frequencies
HUMCSF1PO	10	11	10 = 0.25269
(5q33.3 - q34)	11	12	11 = 0.30049
HUMTPOX	8	8	8 = 0.54433
(2p23 - 2pter)	11	11	11 = 0.25369
HUMTH01	6	6	6 = 0.22660
(11p15.5)	9.3	7	9.3 = 0.30542
HUMvWA31	15	16	15 = 0.11224
(12p13.3 - p13.2)	16		16 = 0.20153

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PI Formula
HUMCSF1PO	10	11	**0.25/a**
(5q33.3 - q34)	11	12	
HUMTPOX	8	8	**(a+b)/4ab**
(2p23 - 2pter)	11	11	
HUMTH01	6	6	**0.25/a**
(11p15.5)	9.3	7	
HUMvWA31	15	16	**0.5/a**
(12p13.3 - p13.2)	16		

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PI
HUMCSF1PO	10	11	**0.83**
(5q33.3 - q34)	11	12	
HUMTPOX	8	8	**1.44**
(2p23 - 2pter)	11	11	
HUMTH01	6	6	**1.10**
(11p15.5)	9.3	7	
HUMvWA31	15	16	**2.48**
(12p13.3 - p13.2)	16		

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PE Formulas
HUMCSF1PO	10	11	$[1-(a+b)]^2$
(5q33.3 - q34)	11	12	
HUMTPOX	8	8	$[1-(a+b)]^2$
(2p23 - 2pter)	11	11	
HUMTH01	6	6	$[1-(a+b)]^2$
(11p15.5)	9.3	7	
HUMvWA31	15	16	$[1-(a+b)]^2$
(12p13.3 - p13.2)	16		

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PE
HUMCSF1PO	10	11	**0.1988**
(5q33.3 - q34)	11	12	
HUMTPOX	8	8	**0.0408**
(2p23 - 2pter)	11	11	
HUMTH01	6	6	**0.2190**
(11p15.5)	9.3	7	
HUMvWA31	15	16	**0.4709**
(12p13.3 - p13.2)	16		

MOTHERLESS PATERNITY CASE P-41376

	C	AF	Allele Frequencies
D16S539	12	11	12 = 0.33911
(16p24 - p25)	13	12	13 = 0.16337
D7S820	11	11	11 = 0.20197
(7q)	12	14	12 = 0.14030
D13S317	11	11	11 = 0.31888
(13q22 - q31)			
D5S818	11	11	11 = 0.41026
(5q21 - q31)	13	12	13 = 0.14615

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PI Formulas
D16S539 (16p24 - p25)	12 13	11 12	**0.25/a**
D7S820 (7q)	11 12	11 14	**0.25/a**
D13S317 (13q22 - q31)	11	11	**1/a**
D5S818 (5q21 - q31)	11 13	11 12	**0.25/a**

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PI
D16S539 (16p24 - p25)	12 13	11 12	**0.74**
D7S820 (7q)	11 12	11 14	**1.24**
D13S317 (13q22 - q31)	11	11	**3.14**
D5S818 (5q21 - q31)	11 13	11 12	**0.61**

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PE Formulas
D16S539 (16p24 - p25)	12 13	11 12	$[1-(a+b)]^2$
D7S820 (7q)	11 12	11 14	$[1-(a+b)]^2$
D13S317 (13q22 - q31)	11	11	$(1-a)^2$
D5S818 (5q21 - q31)	11 13	11 12	$[1-(a+b)]^2$

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PE
D16S539 (16p24 - p25)	12 13	11 12	**0.2475**
D7S820 (7q)	11 12	11 14	**0.4325**
D13S317 (13q22 - q31)	11	11	**0.4639**
D5S818 (5q21 - q31)	11 13	11 12	**0.1968**

MOTHERLESS PATERNITY CASE P-41376

	C	AF	Allele Frequencies
FGA (4q28)	19 21	19 25	19 = 0.05612 21 = 0.17347
D18S51 (18q21.3)	16	16 20	16 = 0.10714
D21S11 (21q11.2 - q21)	29	28 29	29 = 0.18112
D3S1358 (3p)	15 18	15 17	15 = 0.24631 18 = 0.16256
D8S1179 (8)	11 13	11 13	11 = 0.05867 13 = 0.33929

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PI Formulas
FGA (4q28)	19 21	19 25	**0.25/a**
D18S51 (18q21.3)	16	16 20	**0.5/a**
D21S11 (21q11.2 - q21)	29	28 29	**0.5/a**
D3S1358 (3p)	15 18	15 17	**0.25/a**
D8S1179 (8)	11 13	11 13	**(a+b)/4ab**

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PI
FGA (4q28)	19 21	19 25	**4.45**
D18S51 (18q21.3)	16	16 20	**4.67**
D21S11 (21q11.2 - q21)	29	28 29	**2.76**
D3S1358 (3p)	15 18	15 17	**1.02**
D8S1179 (8)	11 13	11 13	**5.00**

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PE Formulas
FGA (4q28)	19 21	19 25	$[1-(a+b)]^2$
D18S51 (18q21.3)	16	16 20	$(1-a)^2$
D21S11 (21q11.2 - q21)	29	28 29	$(1-a)^2$
D3S1358 (3p)	15 18	15 17	$[1-(a+b)]^2$
D8S1179 (8)	11 13	11 13	$[1-(a+b)]^2$

MOTHERLESS PATERNITY CASE P-41376

	C	AF	PE
FGA (4q28)	19 21	19 25	**0.5935**
D18S51 (18q21.3)	16	16 20	**0.7972**
D21S11 (21q11.2 - q21)	29	28 29	**0.6706**
D3S1358 (3p)	15 18	15 17	**0.3944**
D8S1179 (8)	11 13	11 13	**0.3625**

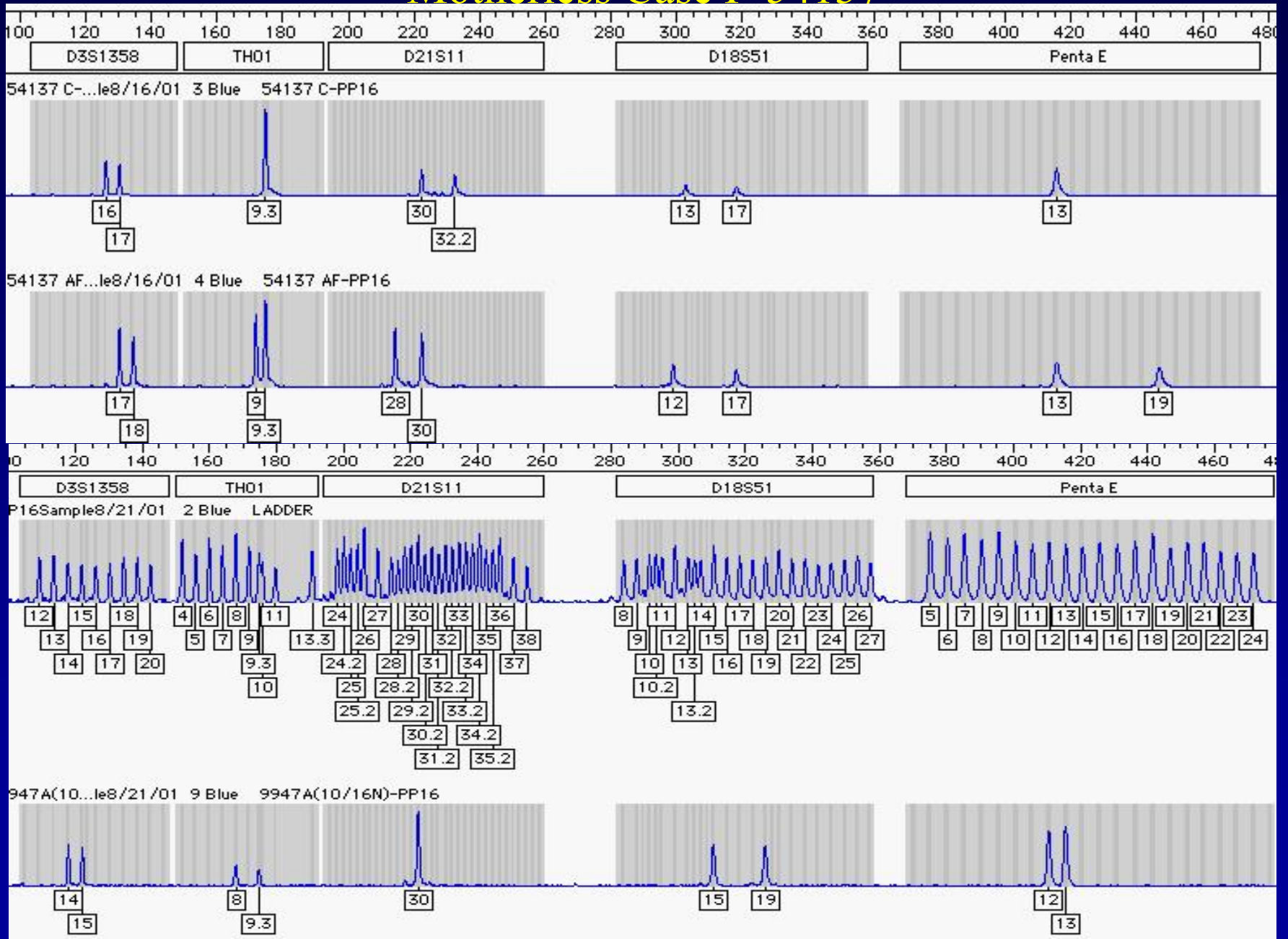
Motherless Paternity
13 Core CODIS Loci

Combined Paternity Index	1,676
Probability of Paternity	99.94%
Probability of Exclusion	99.94%

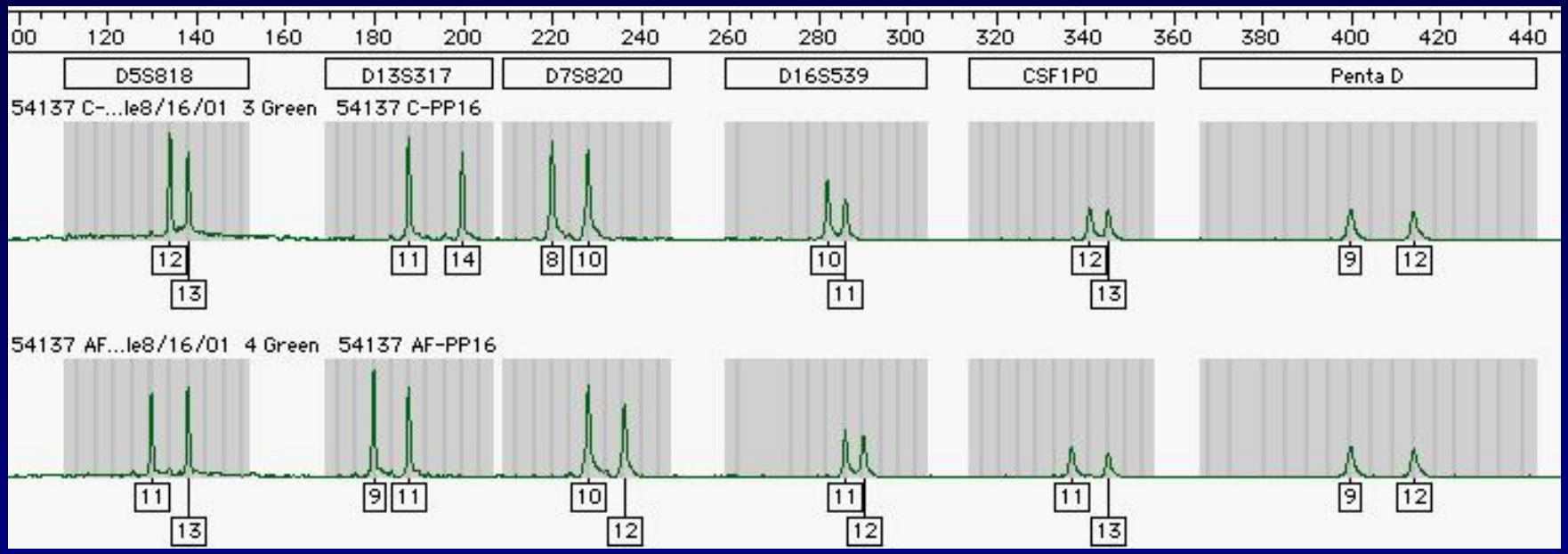
PowerPlex™ 16 System

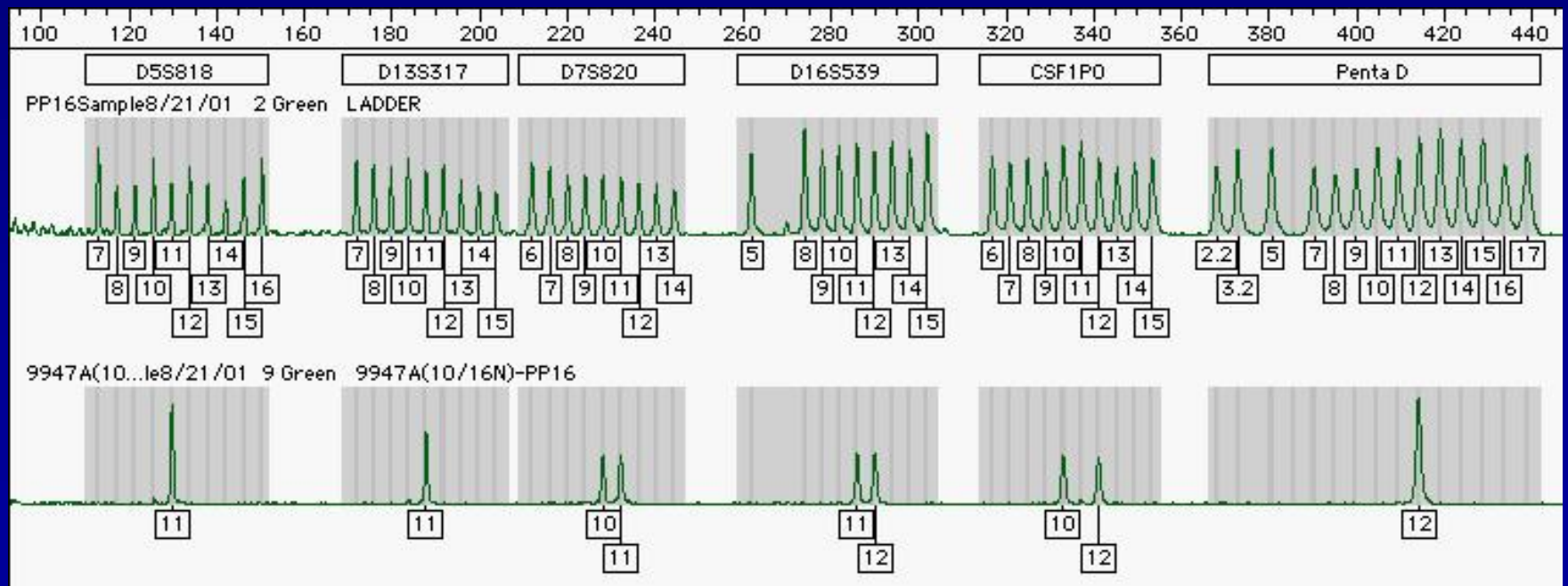
Extremely Useful in Cases
Where the Mother is Not Tested
(Motherless Cases)

PowerPlex™ 16
Motherless Case P-54137

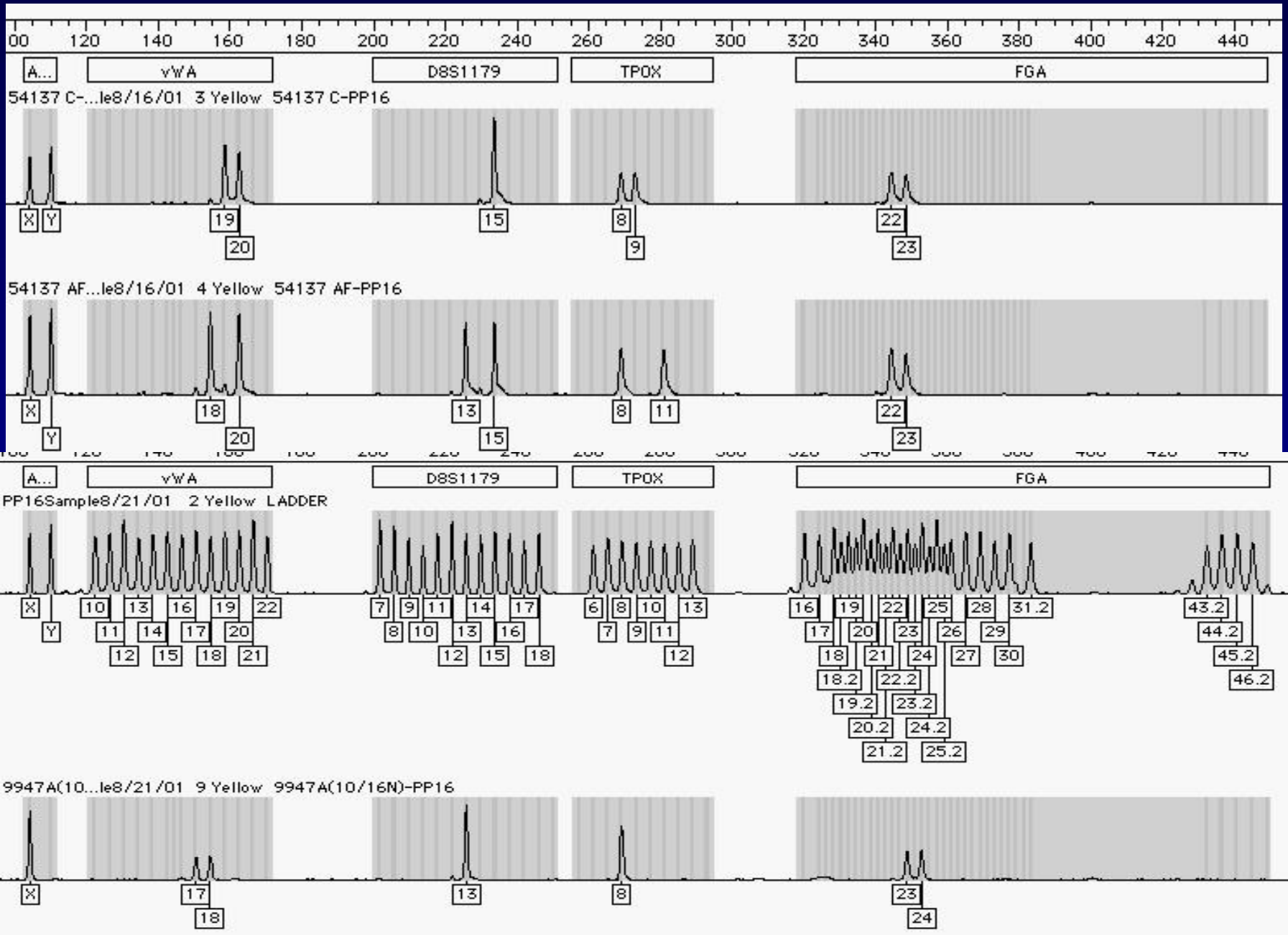


PowerPlex™ 16
Motherless Case P-54137





PowerPlex™ 16
Motherless Case P-54137



Motherless Case P-54137
PowerPlex™ 16 System

13 STR loci minus Penta D & Penta E

Combined Paternity Index	1,050
Probability of Exclusion	99.98%
Probability of Paternity$_{(prior=0.5)}$	99.90%

15 STR loci with Penta D & Penta E

Combined Paternity Index	12,340
Probability of Exclusion	99.997%
Probability of Paternity(prior=0.5)	99.992%

Parentage Statistics For The Identification Of Human Remains

Reverse Parentage Testing

Reverse Parentage Testing

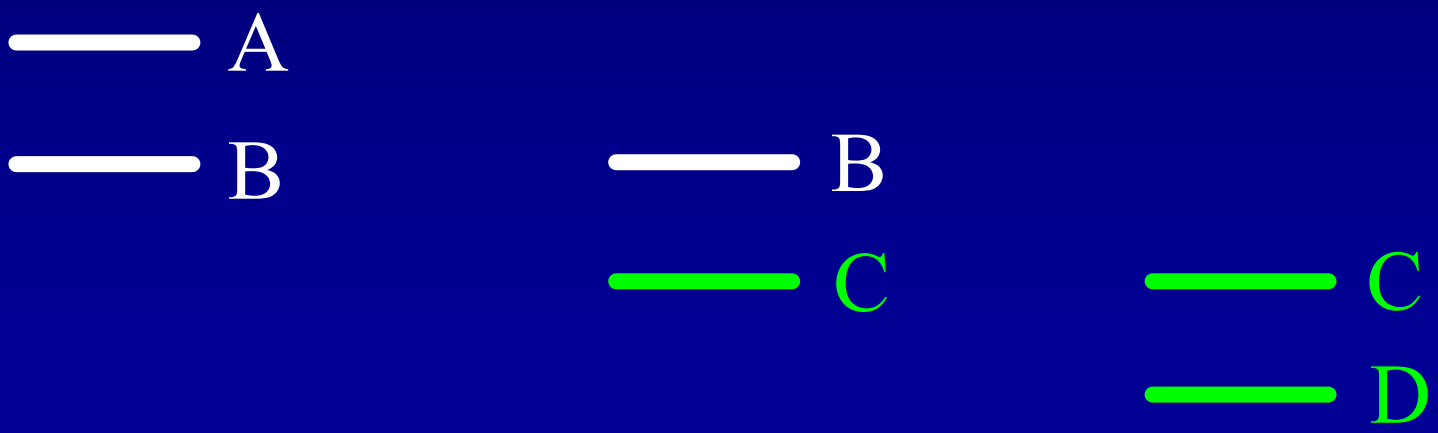
Applications

- Unidentified remains
- Victims of Mass Disasters
- Crime Scene Evidence
- Kidnapped or Abandoned Babies

REVERSE PARENTAGE INDEX

BODY IDENTIFICATION

ALLEGED MOTHER	EVIDENCE	ALLEGED FATHER



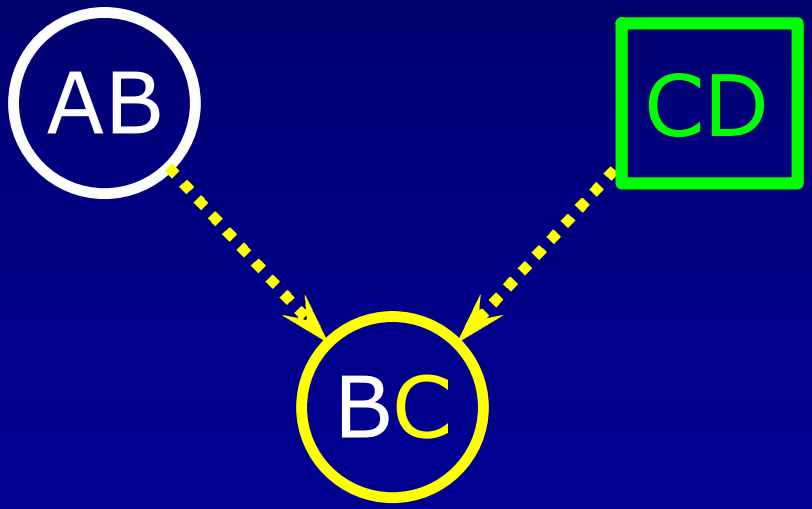
Reverse Parentage Testing

Three genotypes:

- Alleged Mother
- Child (missing)
- Alleged Father

Reverse Parentage Analysis

Missing child scenario



Reverse Parentage Index

RPI = X / Y

Numerator

X = **is the probability that (1) a woman randomly selected from a population is type AB, and (2) a man randomly selected from a population is type CD, and (3) their child is type BC.**

Reverse Parentage Index

RPI = X / Y

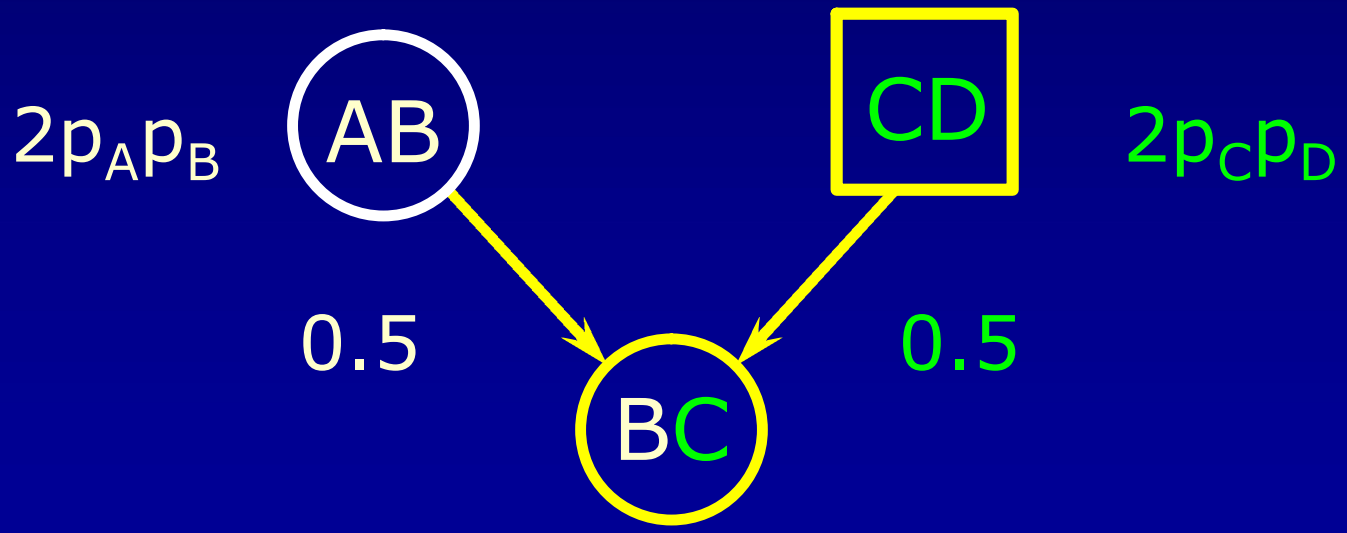
Denominator

Y = **is the probability that (1) a woman randomly selected from a population and unrelated to missing child is type AB, (2) a man randomly selected from a population and unrelated to missing child is type CD, and (3) a child, randomly selected from a population is BC.**

Reverse Parentage Analysis

Missing child scenario

Numerator

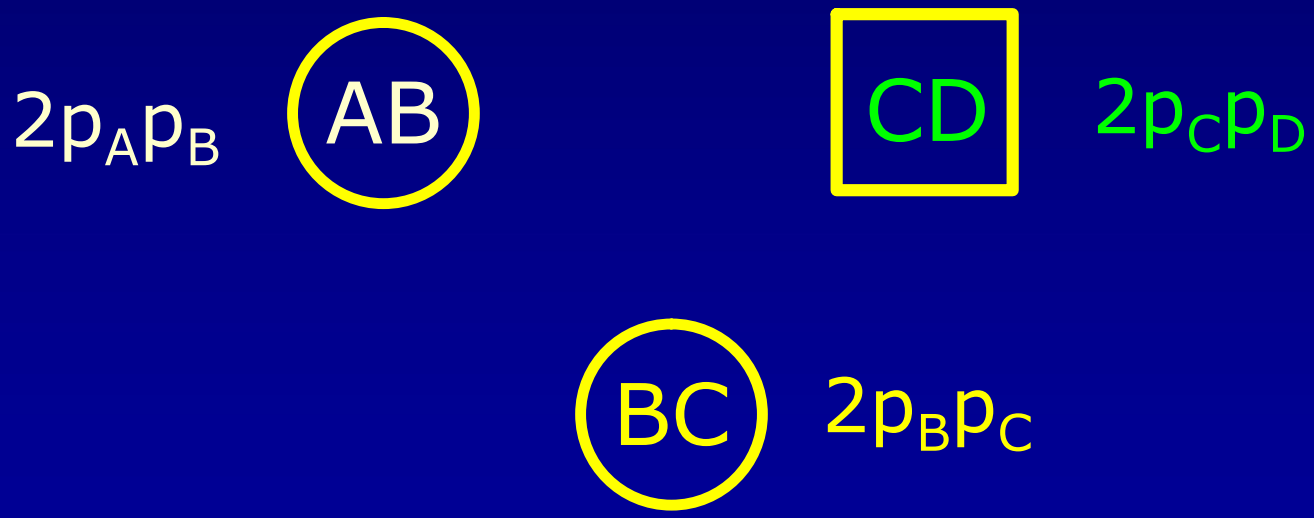


Probability = $2p_Ap_B$ x $2p_Cp_D$ x 0.5 x 0.5

Reverse Parentage Analysis

Missing child scenario

Denominator



Probability = $2p_Ap_B \times 2p_Cp_D \times 2p_Bp_C$

Reverse Parentage Analysis

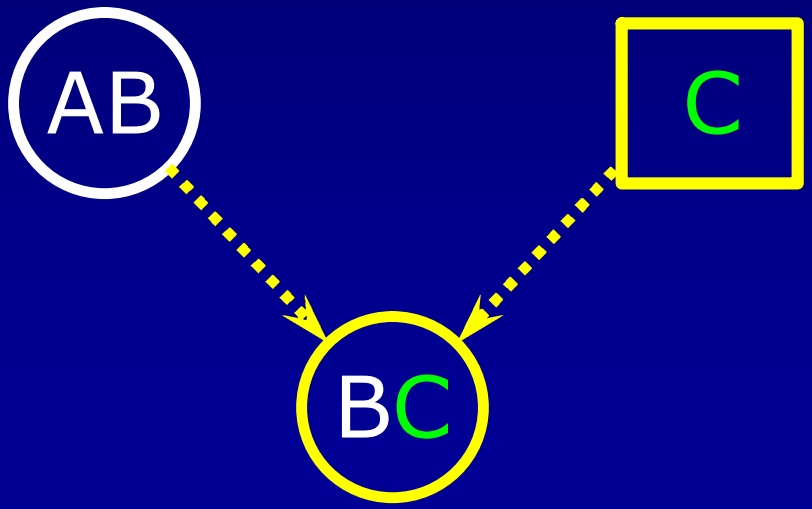
Missing child scenario

$$LR = \frac{2p_A p_B \times 2p_C p_D \times 0.5 \times 0.5}{2p_A p_B \times 2p_C p_D \times 2p_B p_C}$$

$$LR = \frac{0.25}{2p_B p_C}$$

Reverse Parentage Analysis

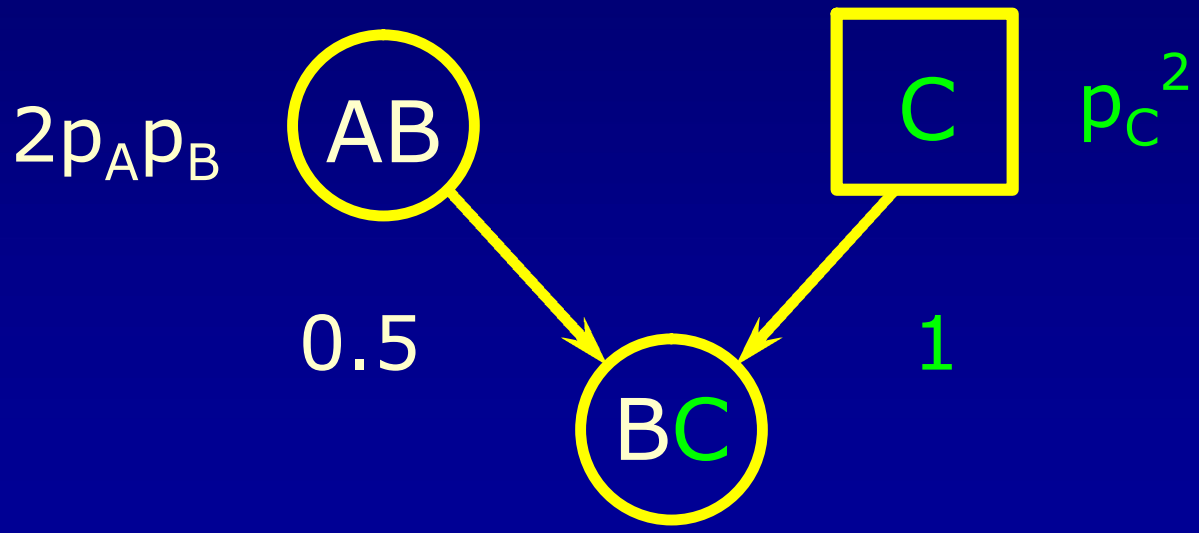
Missing child scenario



Reverse Parentage Analysis

Missing child scenario

Numerator

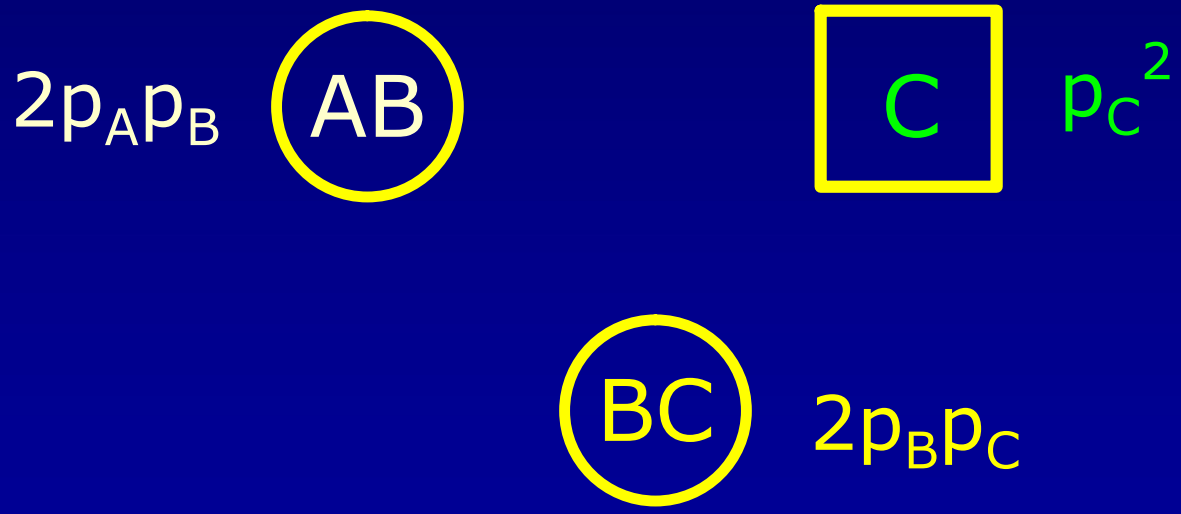


Probability = $2p_Ap_B \times p_C^2 \times 0.5 \times 1$

Reverse Parentage Analysis

Missing child scenario

Denominator



Probability = $2p_Ap_B \times p_C^2 \times 2p_Bp_C$

Reverse Parentage Analysis

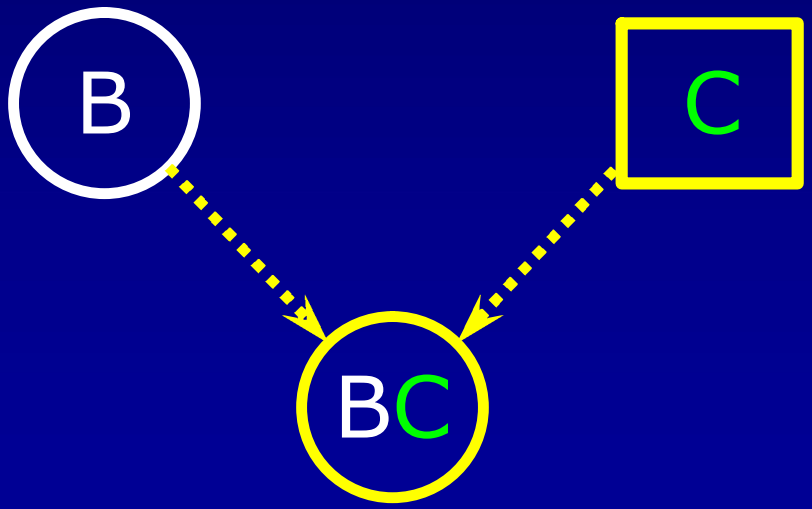
Missing child scenario

$$LR = \frac{p_A p_B \times p_C^2 \times 0.5 \times 1}{p_A p_B \times p_C^2 \times 2 p_B p_C}$$

$$\mathbf{LR = \frac{0.5}{2 p_B p_C}}$$

Reverse Parentage Analysis

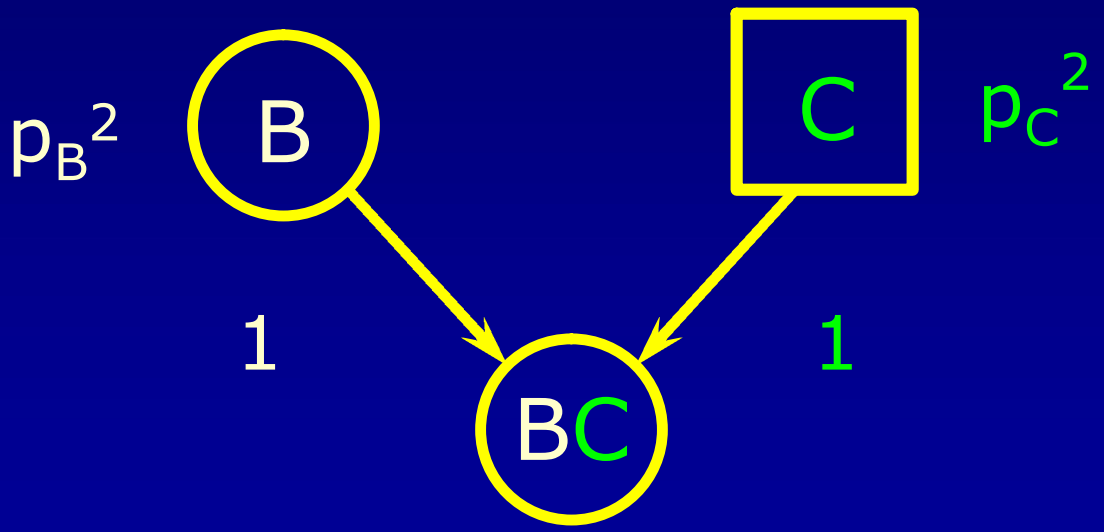
Missing child scenario



Reverse Parentage Analysis

Missing child scenario

Numerator

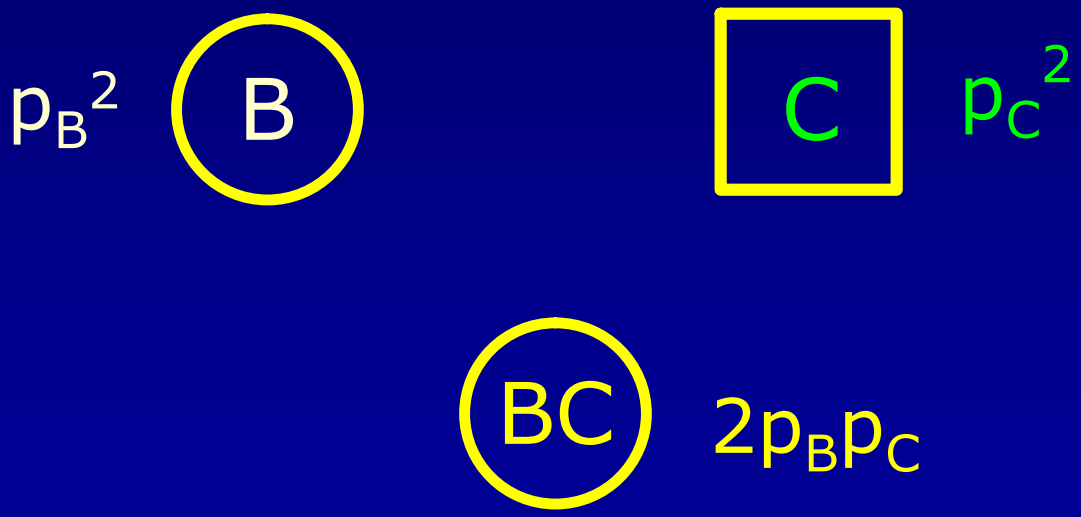


Probability = $p_B^2 \times p_C^2 \times 1 \times 1$

Reverse Parentage Analysis

Missing child scenario

Denominator



Probability = $p_B^2 \times p_C^2 \times 2p_Bp_C$

Reverse Parentage Analysis

Missing child scenario

$$LR = \frac{p_B^2 \times p_C^2 \times 1 \times 1}{p_B^2 \times p_C^2 \times 2p_Bp_C}$$

$$LR = \frac{1}{2p_Bp_C}$$

Having both parents to test in a reverse parentage test is indeed a luxury

Often, we are limited to one parent or possibly even siblings to attempt an identification

Single parent cases revert statistically to the "non-maternal" format we discussed earlier

Thank you!