

Popstats 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:

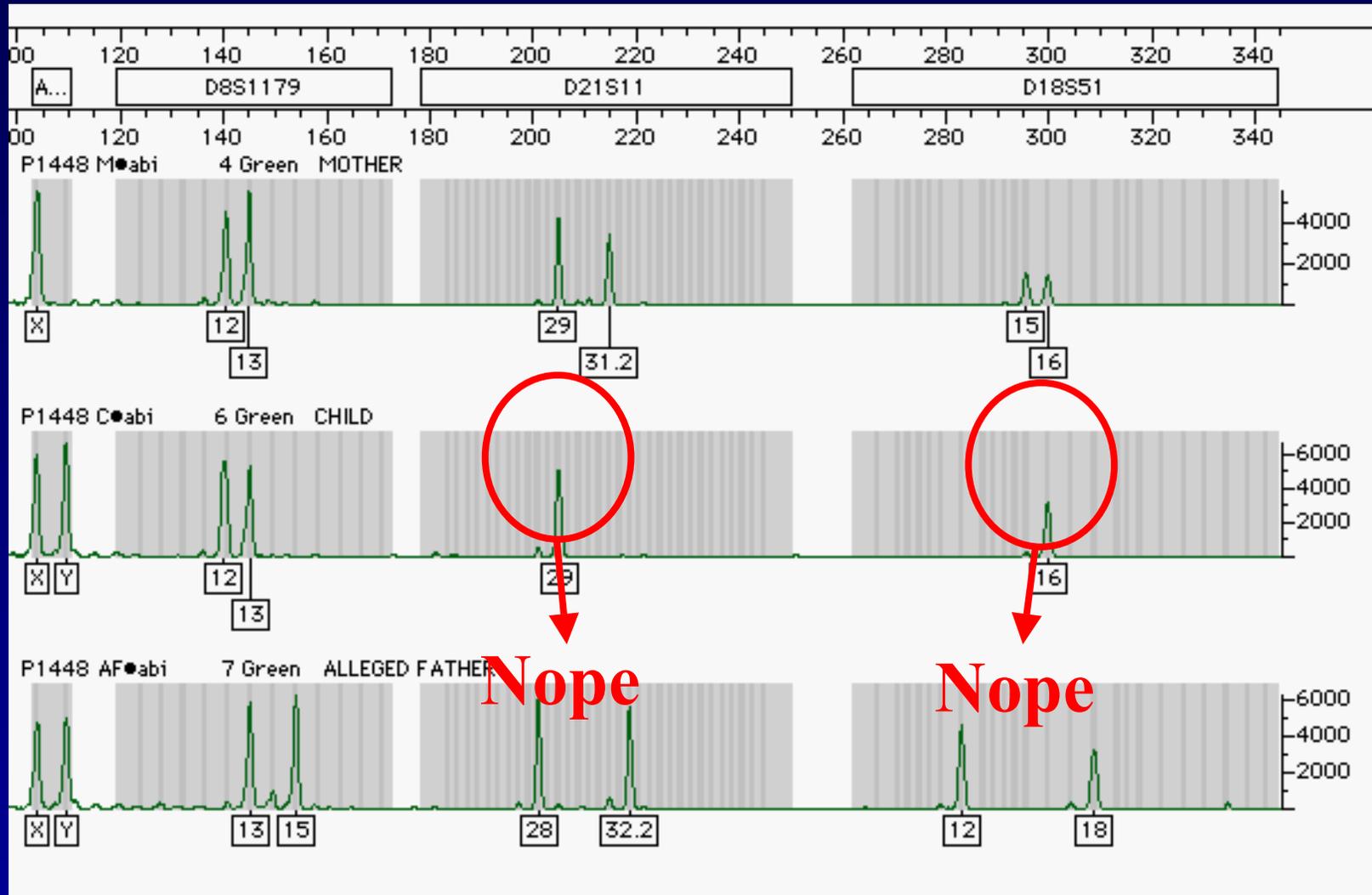
Inclusion

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

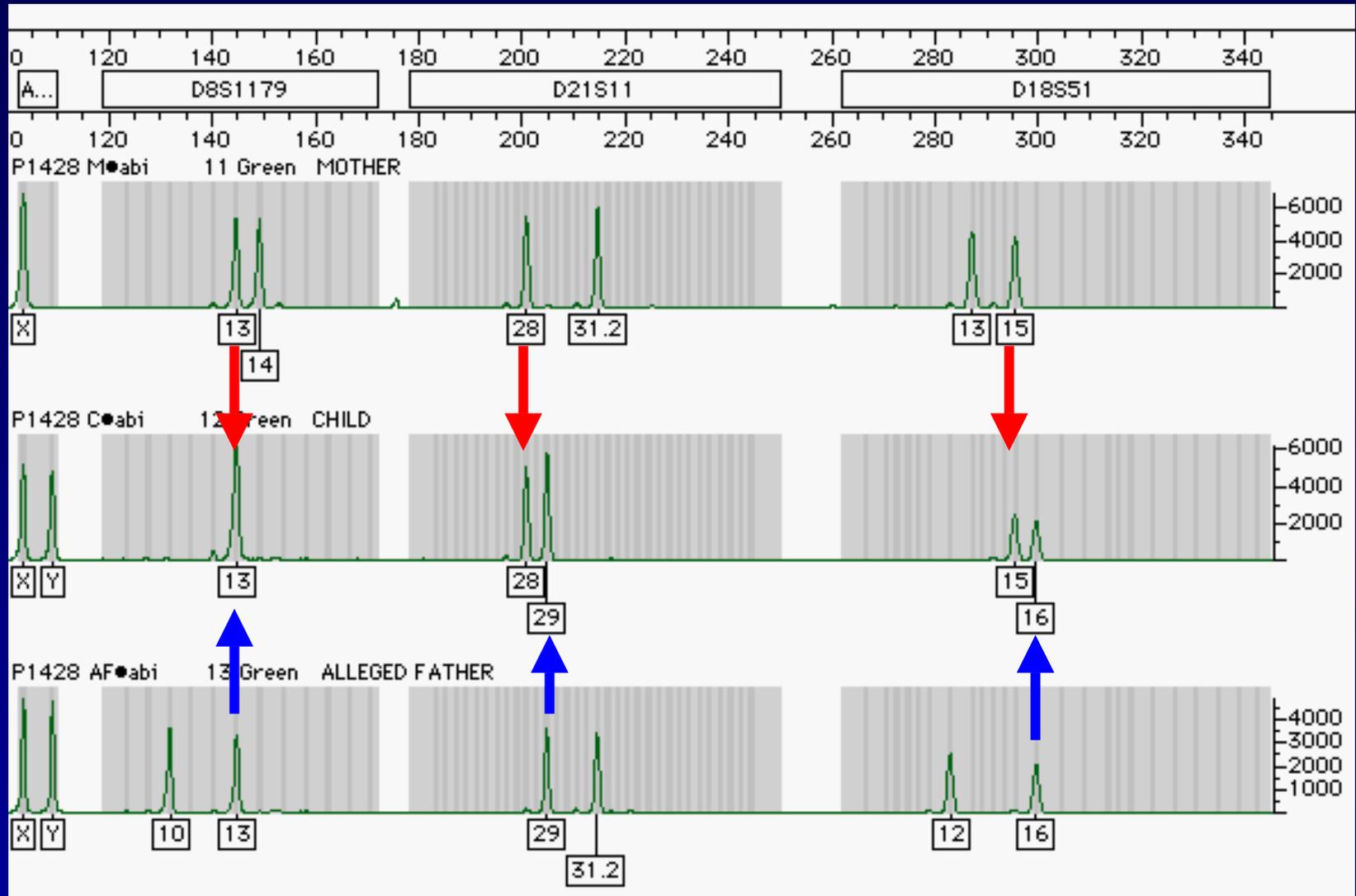


Results

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



Inclusion



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



Paternity Analysis

Hypothetical case

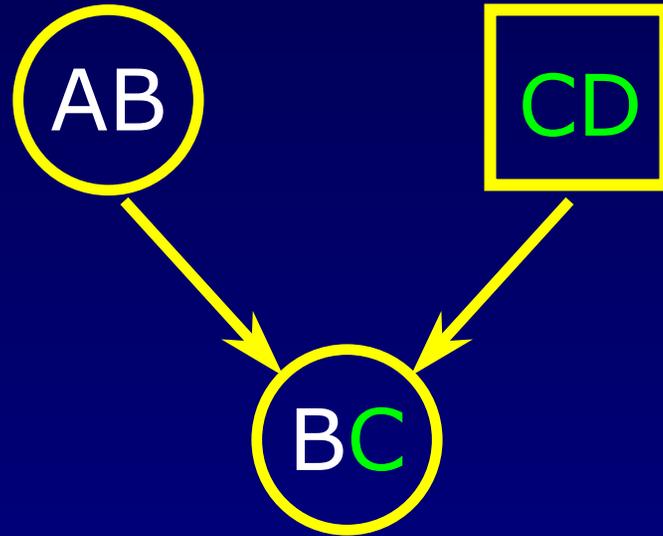
DNA Analysis Results in Three Genotypes

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



Numerator

<u>Person</u>	<u>Type</u>
Mother	AB
Child	BC
Alleged Father	CD

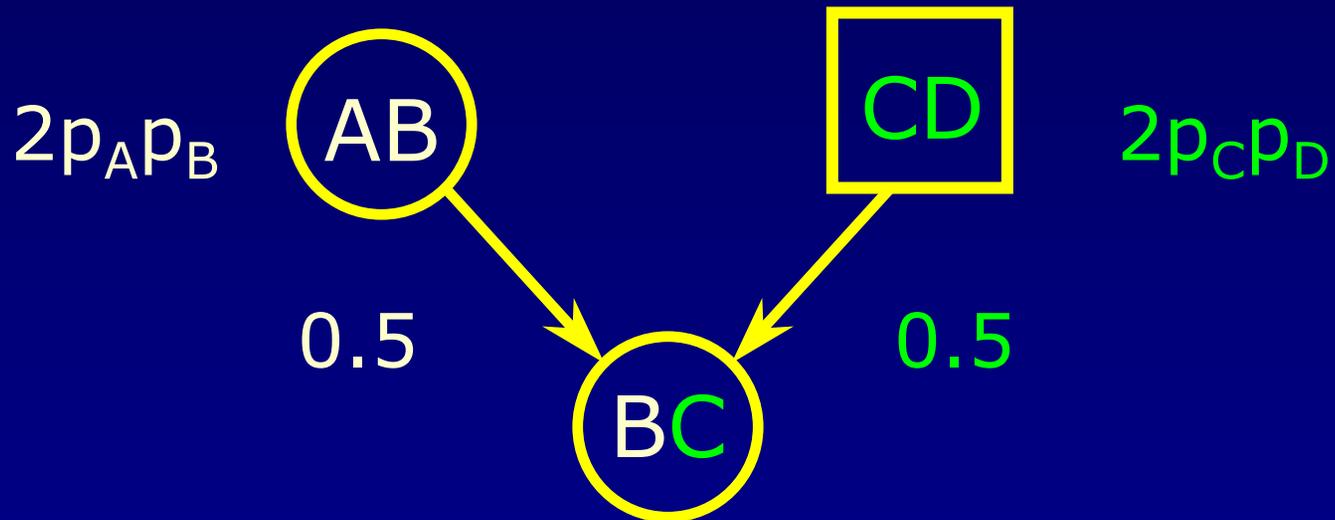
In order to explain this evidence Calculate **Probability** that

- Woman randomly selected from population is type AB
- Man randomly selected from population is type **CD**, and
- Their child is type **BC**

Paternity Analysis

Paternity Index

Numerator



$$\text{Probability} = 2p_A p_B \times 2p_C p_D \times 0.5 \times 0.5$$

Hypothetical DNA Example

Second Hypothesis

Denominator

<u>Person</u>	<u>Type</u>
Mother	AB
Child	BC
Alleged Father	CD

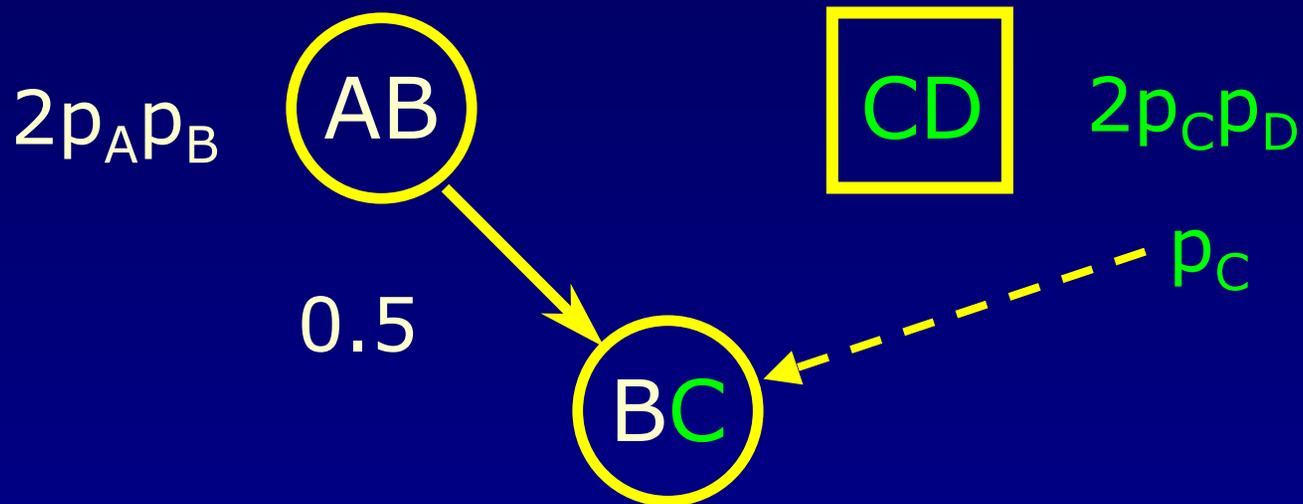
In order to explain this evidence Calculate **Probability** that

- Woman randomly selected from population is type AB
- An alternative man randomly selected from population is type **CD**, and
- The woman's child, fathered by random man, is type **BC**

Paternity Analysis

Paternity Index

Denominator



$$\text{Probability} = 2p_A p_B \times 2p_C p_D \times 0.5 \times p_C$$

Paternity Analysis

Paternity Index

$$PI = \frac{2p_{A|B} \times 2p_{C|D} \times 0.5 \times 0.5}{2p_{A|B} \times 2p_{C|D} \times 0.5 \times p_C}$$
$$PI = \frac{0.5}{p_C}$$

Hypothetical DNA Example

Probability Statements

<u>Person</u>	<u>Type</u>
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

<u>Person</u>	<u>Type</u>
Mother	AB
Child	BC
Alleged Father	CD

A **Correct** statement is

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

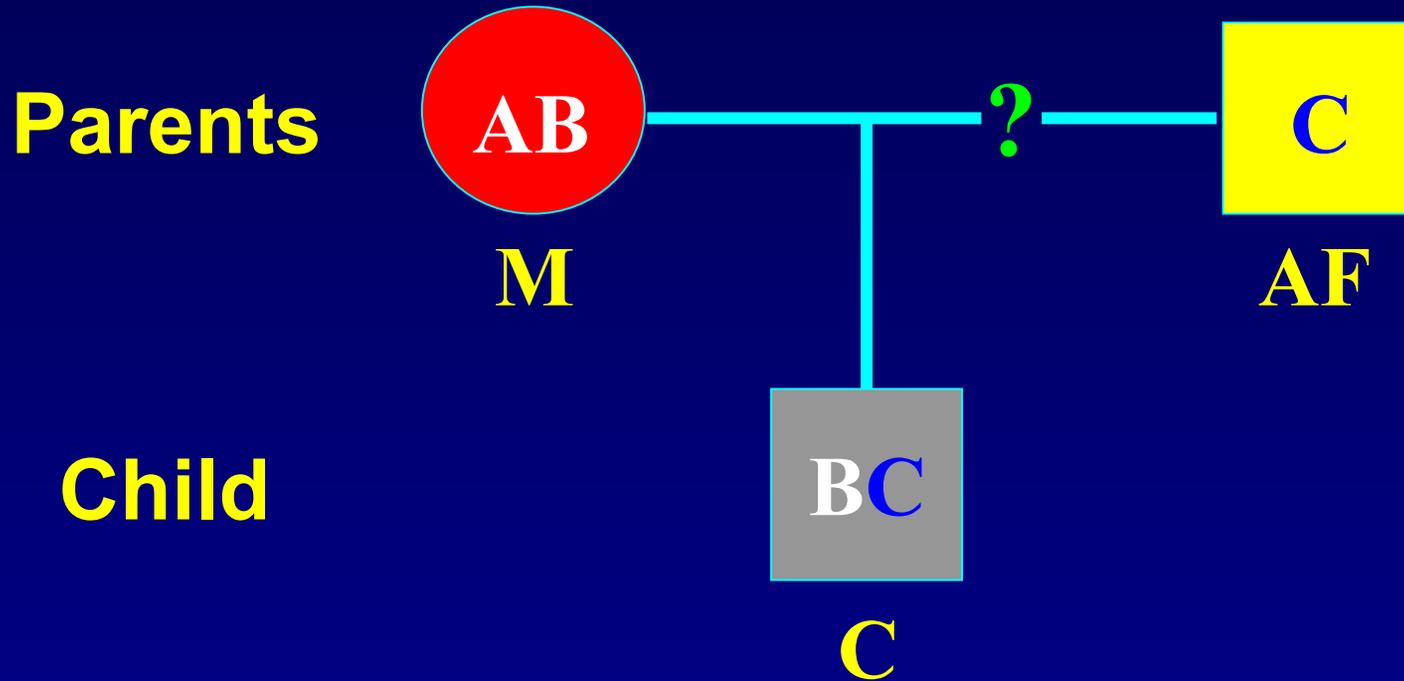
There is (X/Y) times more support for the genetic results if the tested man was the true biological father than if an untested random man was the father

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

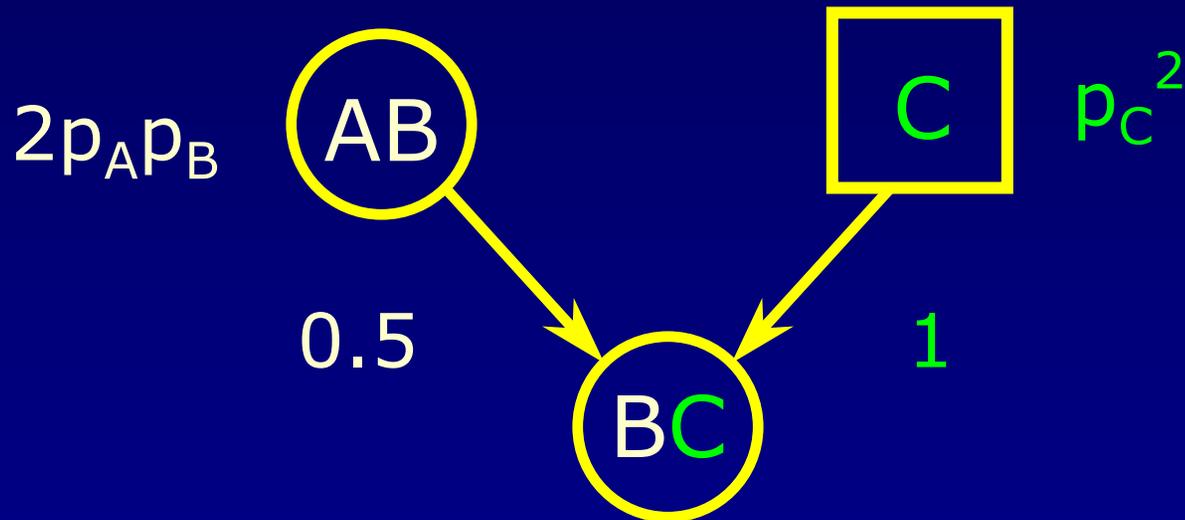
Random Man has p chance of passing the C allele

$$PI = 1/p$$

Paternity Analysis

Paternity Index

Numerator

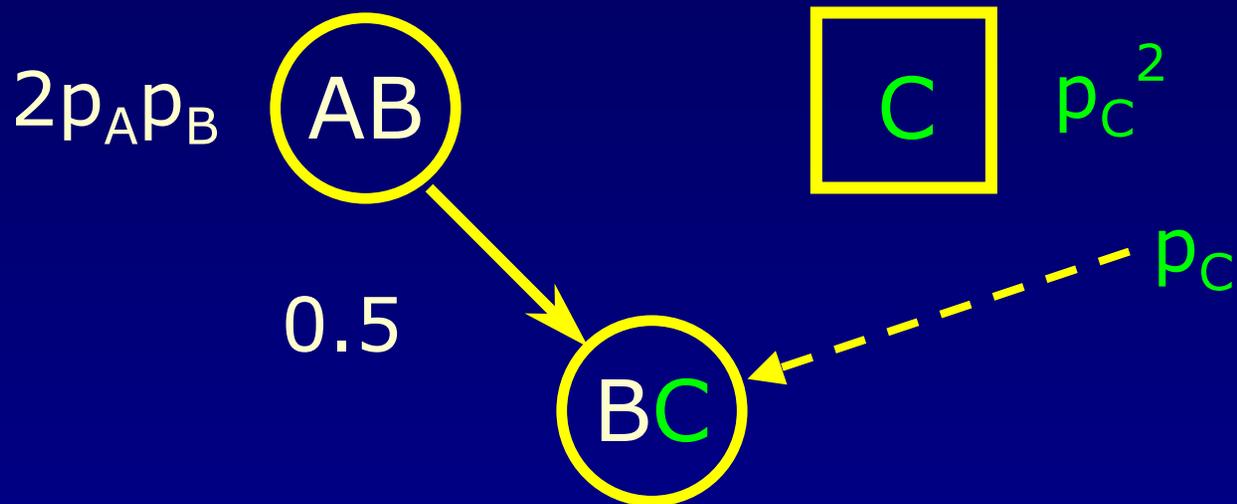


$$\text{Probability} = 2p_A p_B \times p_C^2 \times 0.5 \times 1$$

Paternity Analysis

Paternity Index

Denominator



$$\text{Probability} = 2p_A p_B \times p_C^2 \times 0.5 \times p_C$$

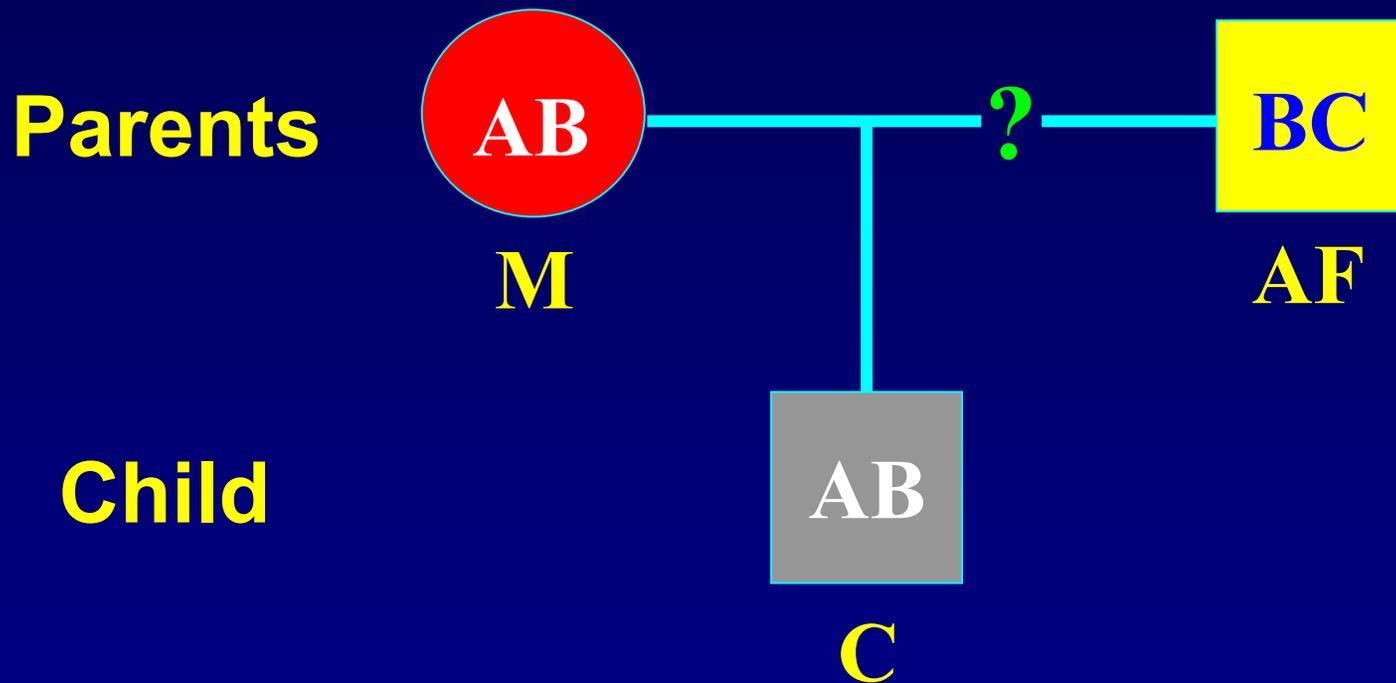
Paternity Analysis

Paternity Index

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

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

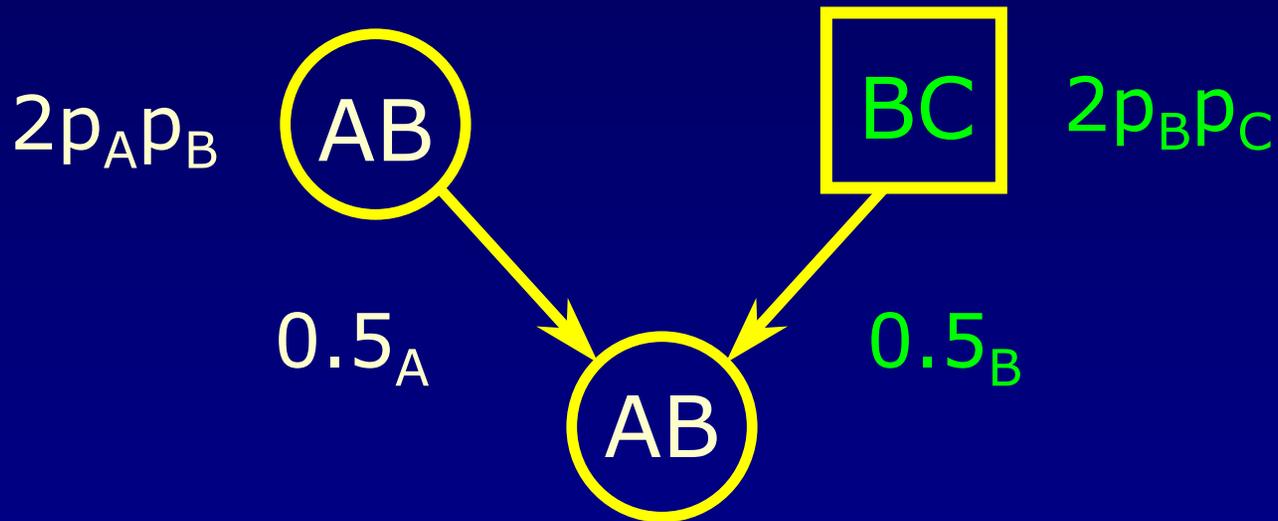
RM has $(p + q)$ chance of passing the A or B alleles

$$PI = 0.5/(p+q)$$

Paternity Analysis

Paternity Index

Numerator

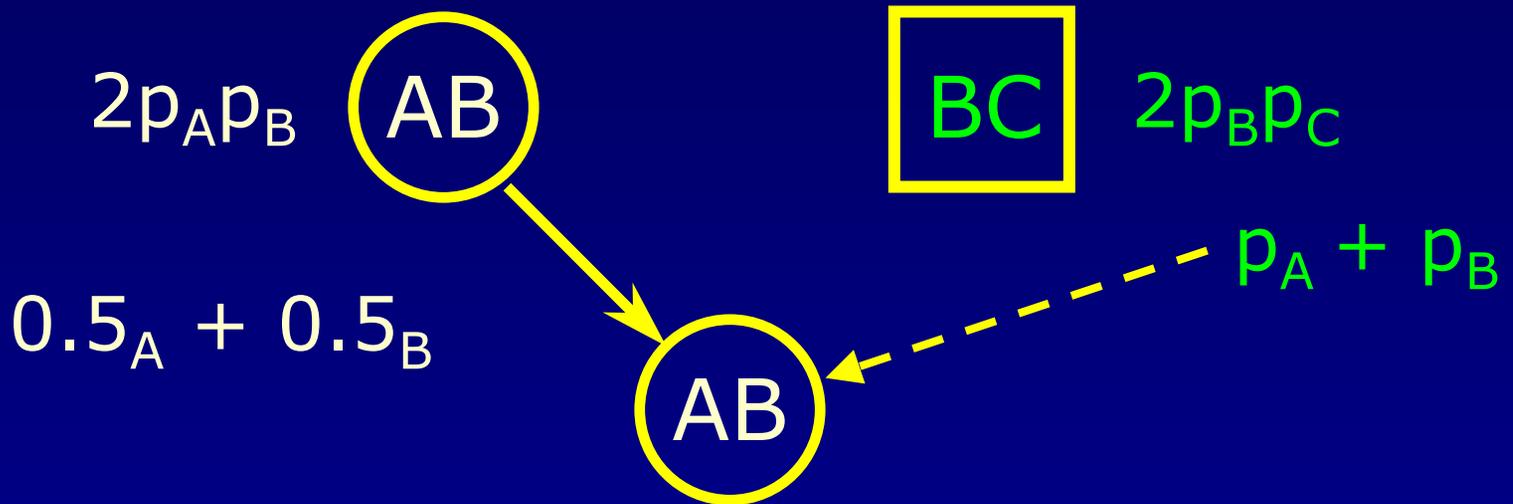


$$\text{Probability} = 2p_A p_B \times 2p_B p_C \times 0.5_{(mA)} \times 0.5_{(fB)}$$

Paternity Analysis

Paternity Index

Denominator



probability =

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

Paternity Analysis

Paternity Index

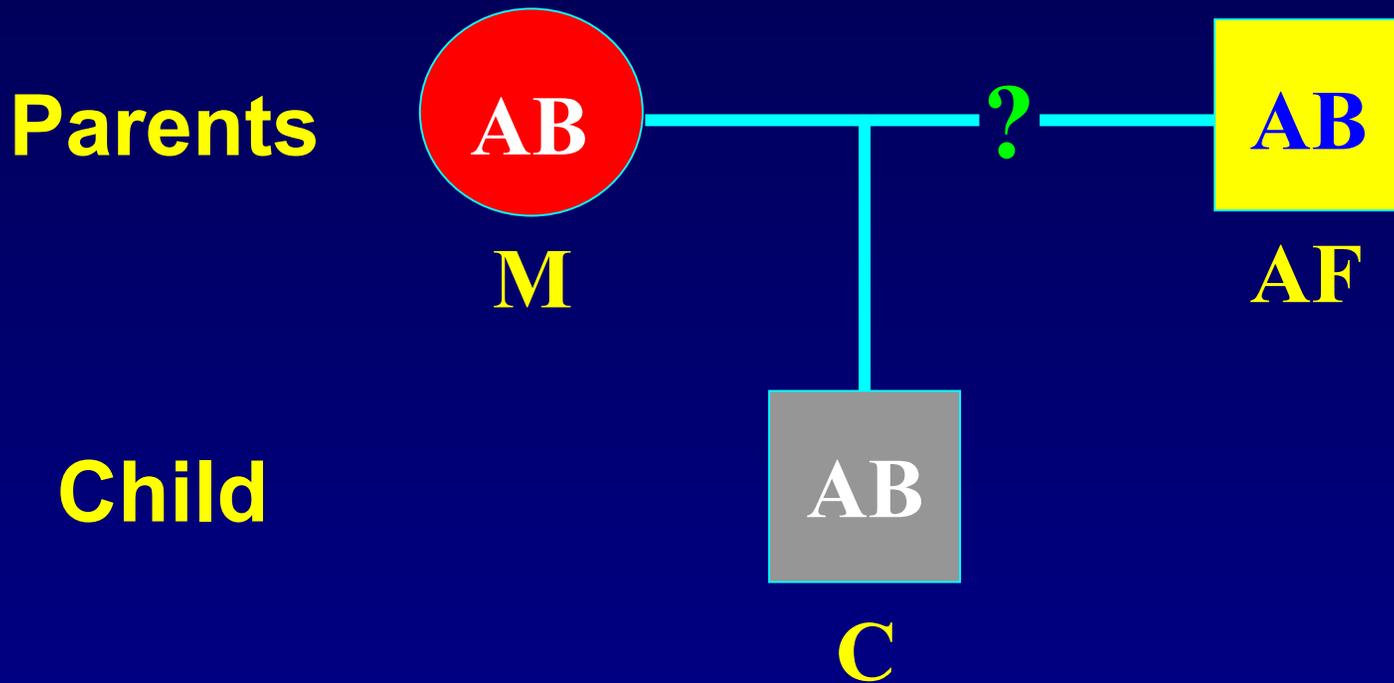
$$PI = \frac{2p_A p_B \times 2p_B p_C \times 0.5_{(mA)} \times 0.5_{(fB)}}{2p_A p_B \times 2p_B p_C \times (0.5_{(mB)} \times p_A + 0.5_{(mA)} \times p_B)}$$

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

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

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

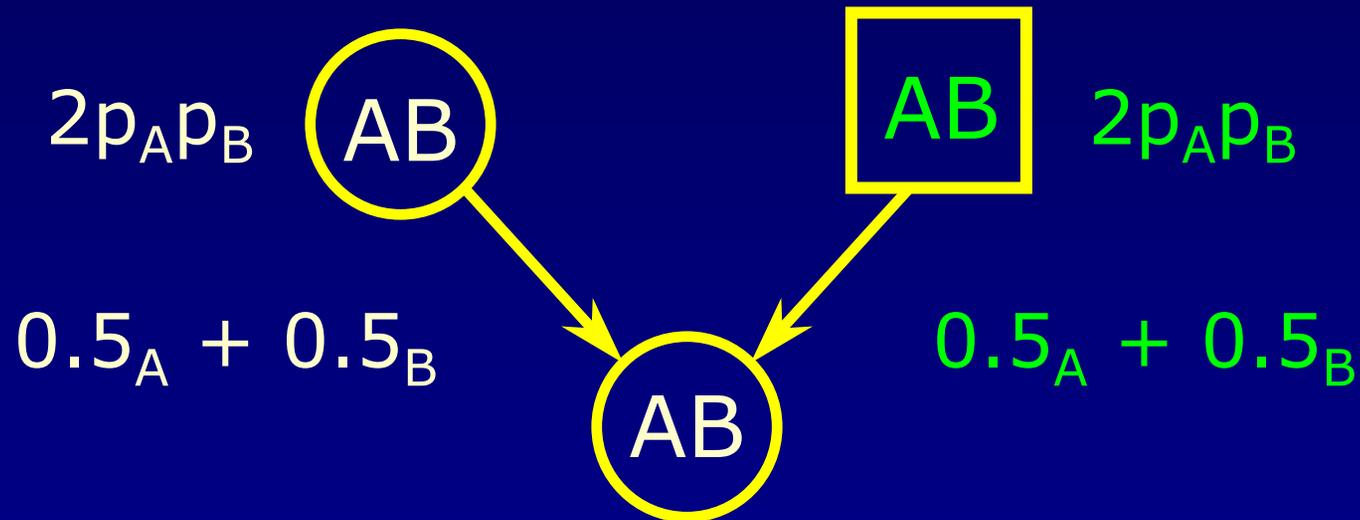
RM has $(p + q)$ chance of passing the A or B alleles

$$PI = 1/(p+q)$$

Paternity Analysis

Paternity Index

Numerator



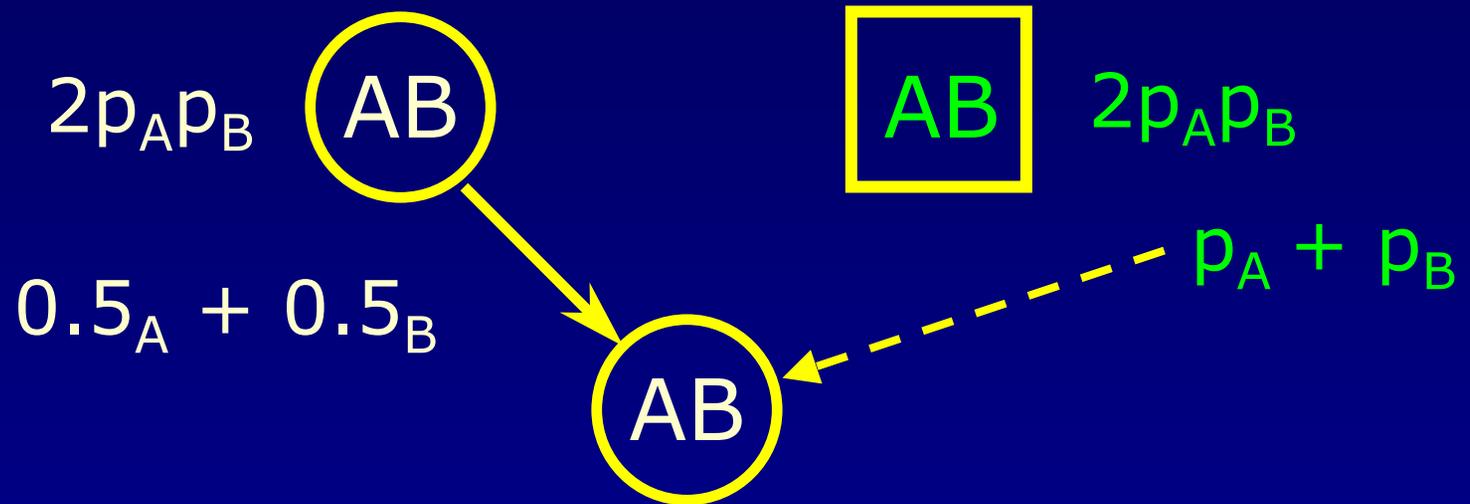
Probability =

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

Paternity Analysis

Paternity Index

Denominator



probability =

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

Paternity Analysis

Paternity Index

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

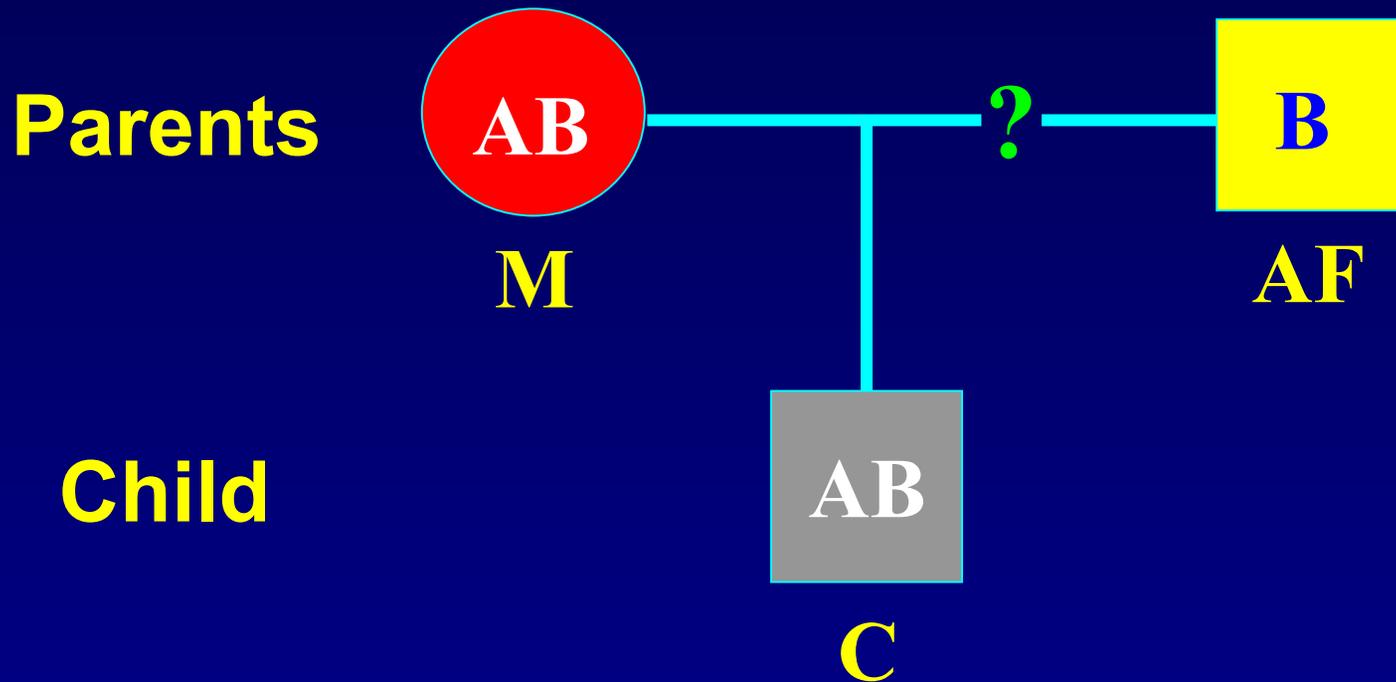
$$PI = \frac{0.5}{0.5p_A + 0.5p_B}$$

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

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

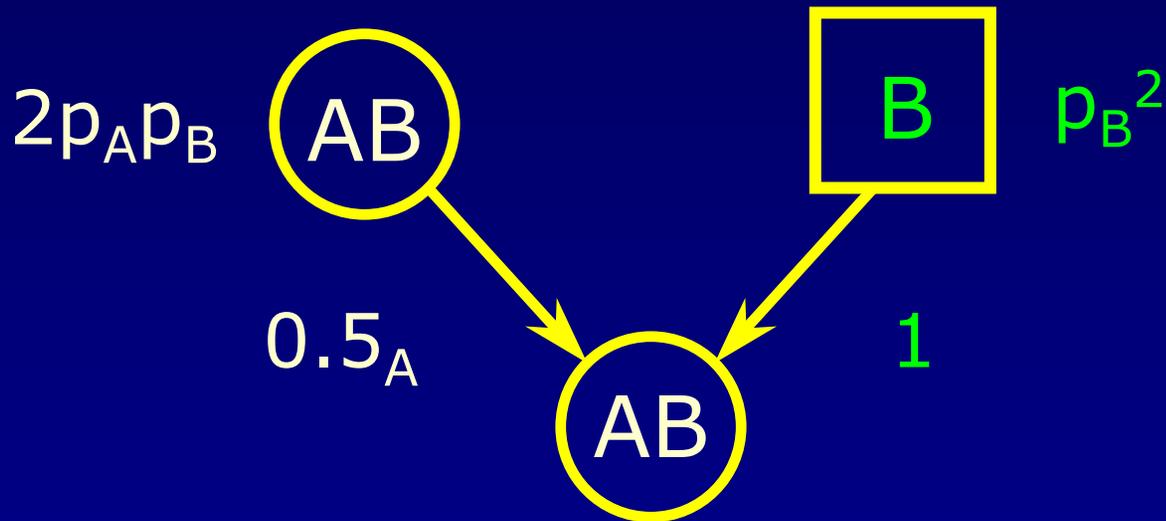
RM has $(p + q)$ chance of passing the A or B alleles

$$PI = 1/(p+q)$$

Paternity Analysis

Paternity Index

Numerator

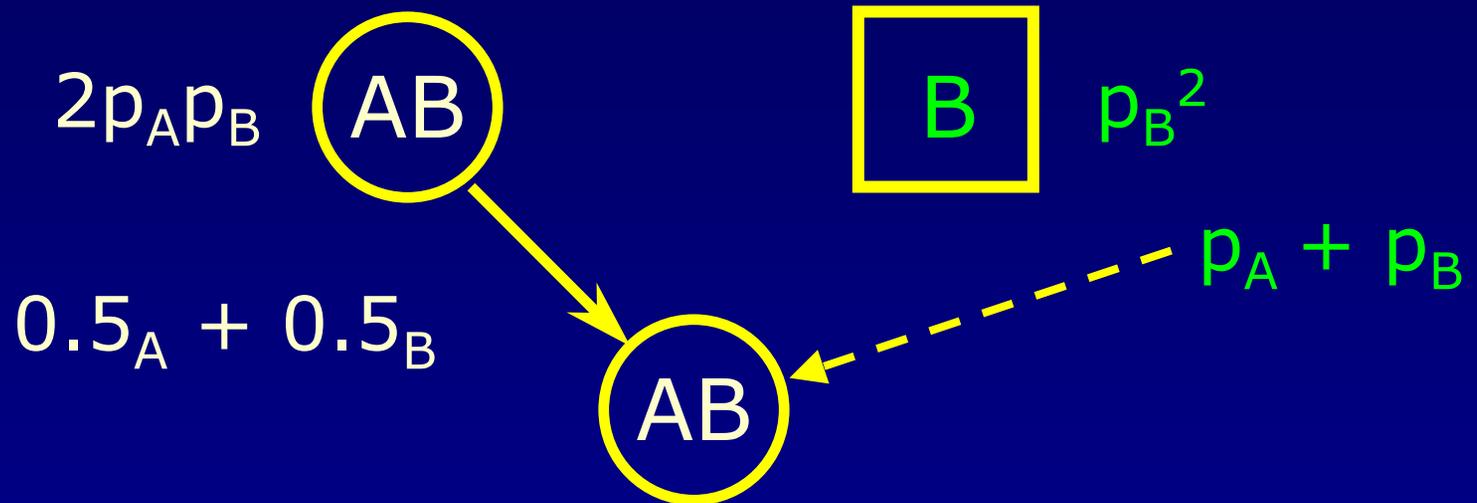


$$\text{Probability} = 2p_A p_B \times p_B^2 \times 0.5_{(mA)} \times 1_{(fB)}$$

Paternity Analysis

Paternity Index

Denominator



probability =

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

Paternity Analysis

Paternity Index

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

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

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

PI Formulas

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

<u>M</u>	<u>C</u>	<u>AF</u>	<u>Numerator</u>	<u>Denominator</u>	<u>PI</u>
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

<u>M</u>	<u>C</u>	<u>AF</u>	<u>Numerator</u>	<u>Denominator</u>	<u>PI</u>
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

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

PI Formulas

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

<u>M</u>	<u>C</u>	<u>AF</u>	<u>Numerator</u>	<u>Denominator</u>	<u>PI</u>
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

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

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

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) =

$$\begin{aligned} & \frac{\text{CPI} \times 0.5}{[\text{CPI} \times 0.5 + (1 - 0.5)]} \\ &= \frac{\text{CPI}}{\text{CPI} + 1} \end{aligned}$$

Posterior Odds in Favor of Paternity

$$\text{Posterior Odds} = \text{CPI} \times \text{Prior Odds}$$

$$\text{Prior Odds} = P / (1 - P)$$

$$\text{Posterior Odds in Favor of Paternity} =$$

$$\text{CPI} \times [P / (1 - P)]$$

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 =

$$10,000 \times (0.7 / 0.3) = 23,333$$

$$\text{Posterior Odds in Favor of Paternity} = 23,333 \text{ to } 1$$

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:

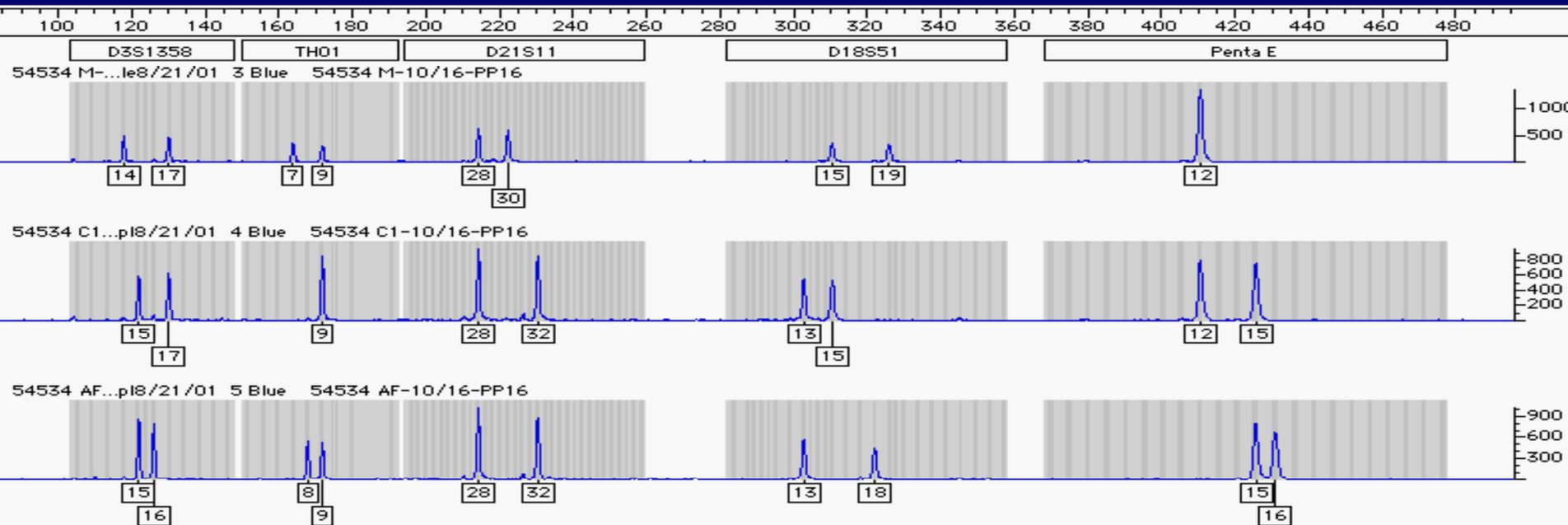
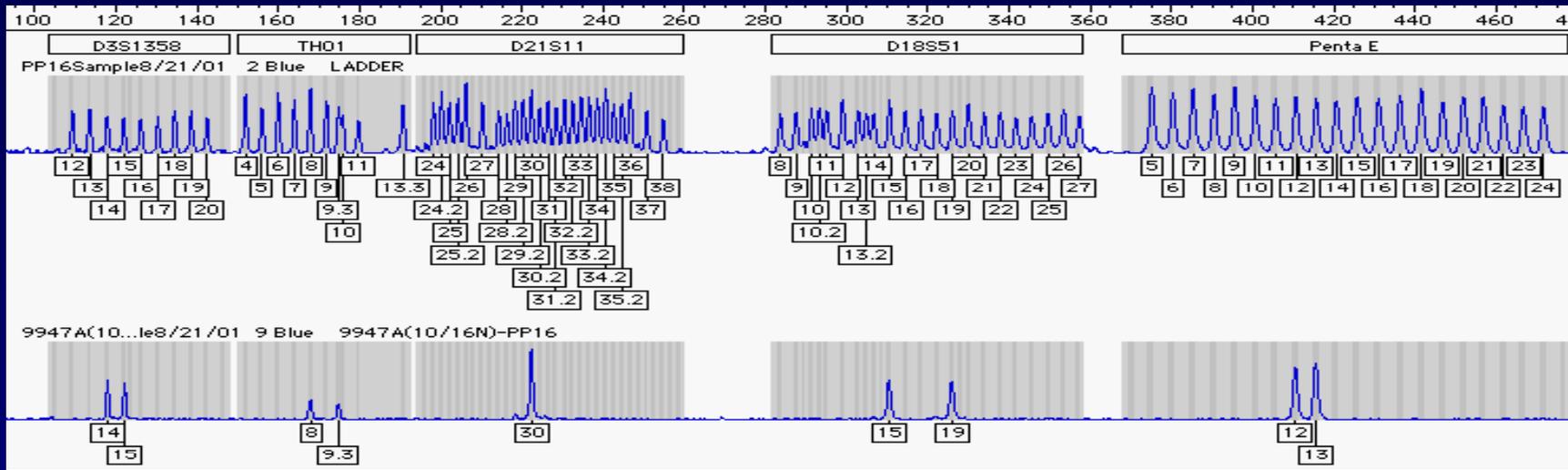
$$PE = [1 - (a + b)]^2$$

Combined Probability of Exclusion

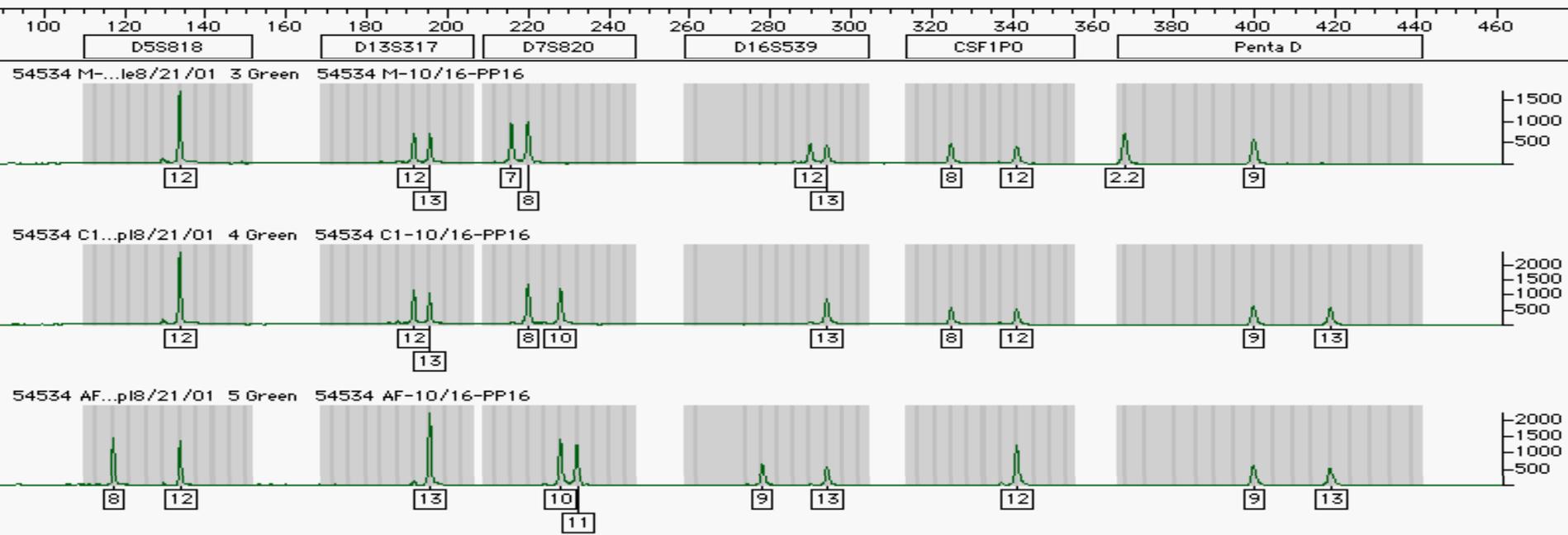
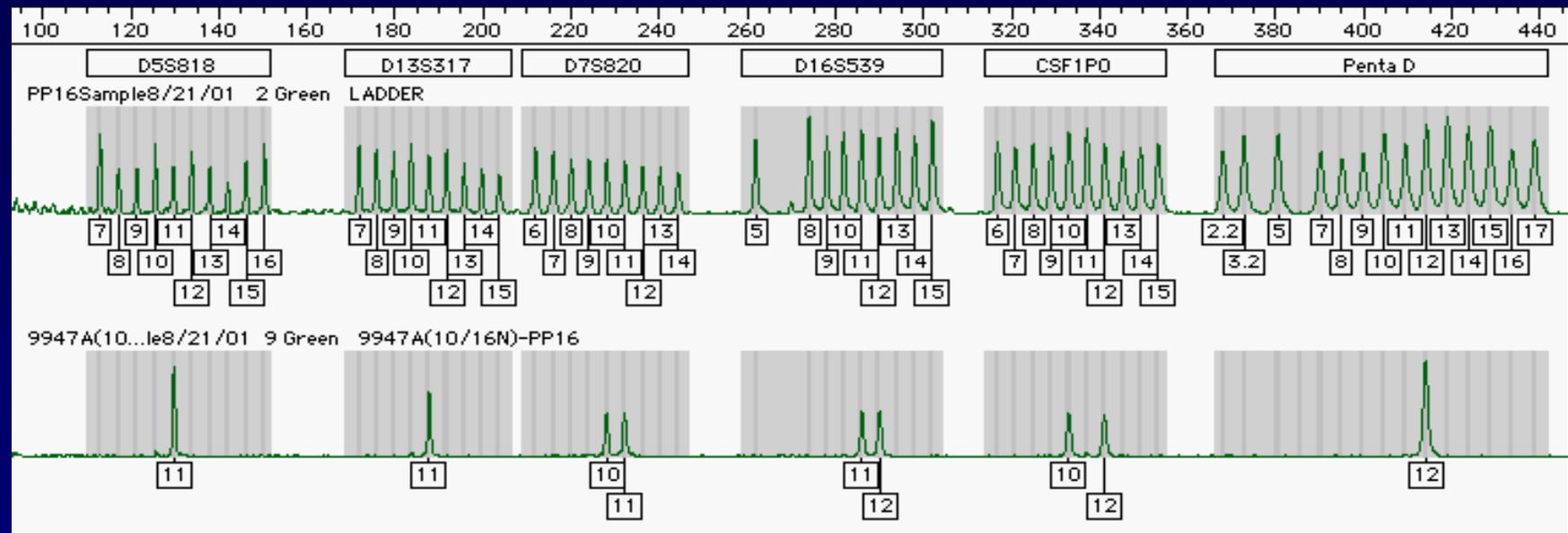
The individual Probability of Exclusion is calculated for each of the genetic systems (loci) analyzed. The overall Probability of Excluding (CPE) a falsely accused man in a given case equals:

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

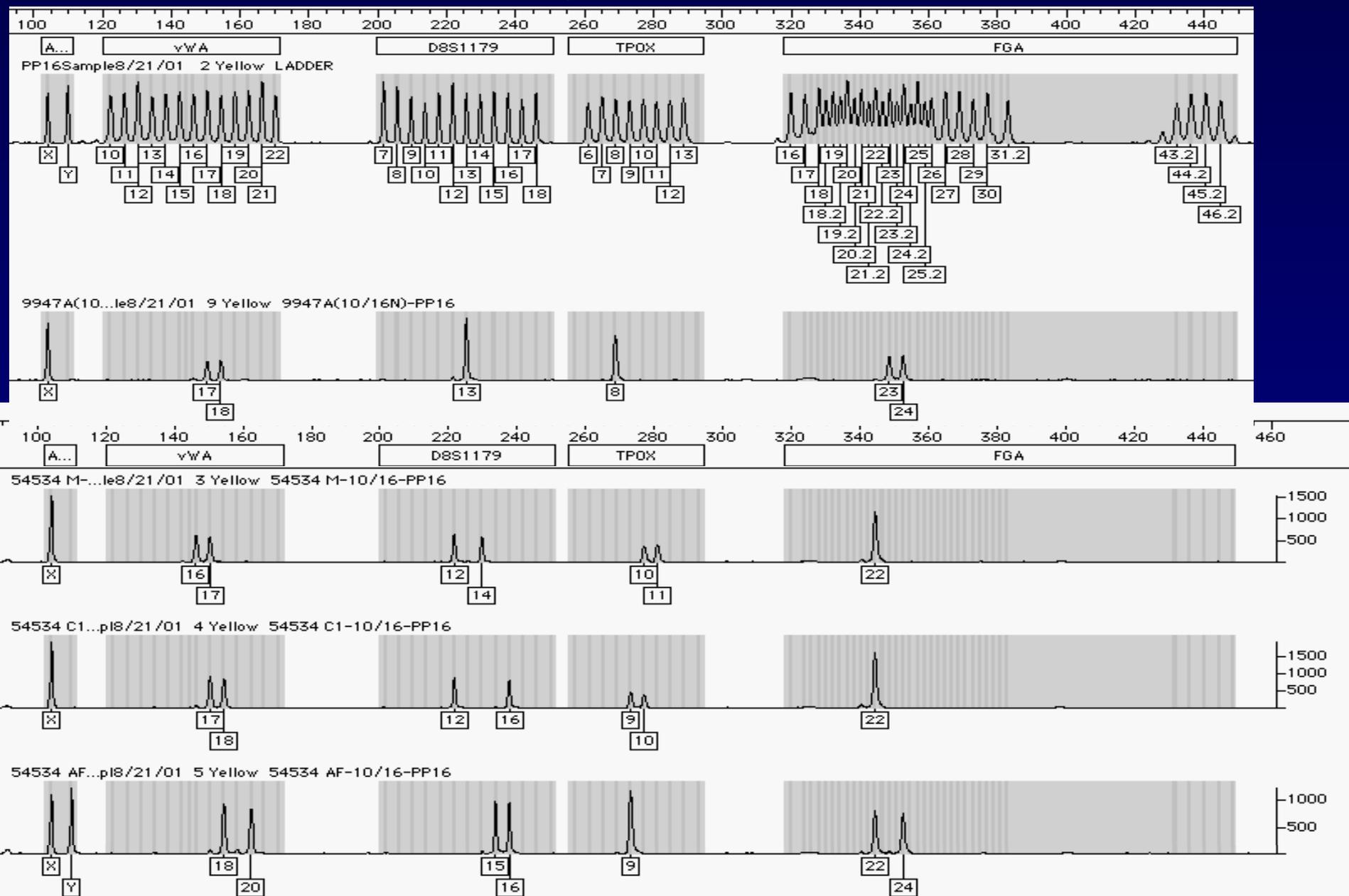
Paternity Trio P-54534



Paternity Trio P-54534



Paternity Trio P-54534



Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

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

Paternity Trio P-54534

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

Paternity Trio P-54534

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

Paternity Trio P-54534

	M	C	AF	PE Formula
HUMCSF1PO (5q33.3 - q34)	8 12	8 12	12	$[1 - (a+b)]^2$
HUMTPOX (2p23 - 2pter)	10 11	9p 10m	9	$(1 - a)^2$
HUMTH01 (11p15.5)	7 9	9	8 9	$(1 - a)^2$
D16S539 (16p24 - p25)	12 13	13	9 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	99.999999%
Probability of Exclusion	99.999999%

Popstats

Parentage Calculations

PopStats can only do basic
parentage statistics!



Popstats Calculations

Band/Allele Occurrence Frequency

VNTR/RFLP Loci

- Fixed-bin Method
- Floating-bin method
- Bin-determination windows
- Default bin method

PCR Loci

Minimum Allele Frequency

Rounding and Significant Figures Applied to Band/Allele Frequency

Genotype Probability

Probability of A Locus Genotype

Default Locus Genotype Probability Formulae and Settings

Combined Genotype Probability of a DNA Profile

Inverse Genotype Probabilities

Rounding and Significant Figures in Genotype Probabilities

Forensic-Single Sample Case

Forensic-multiple samples Case

Confidence Limits

Relatedness Statistics

Interim Ceiling Principle

Forensic Mixture Case

Default Mixture Formula and Settings

Rounding and Significant Figures

Parentage Case

- Case 1
- Case 2
- Case 3
- Case 4
- Case 5
- Case 6
- Case 7

RFLP Band Values Exceeds the Maximum Band Values Allowed

Parentage Case

In the Popstats Parentage Case type, only the pure parentage calculation is performed. Currently there is no reverse parentage or single-parent statistics calculation implemented in Popstats. Three DNA profiles are looked at in the Parentage case type: that of the known parent (KP), the child (C), and the alleged parent (AP).

Because the known parent and child relationship is assumed certain, at least one band/allele of the known parent and child must match at every locus.

Popstats compares the child and alleged parent profiles at each locus, and computes three statistical values: Parentage Index (PI), Probability of Exclusion (PE), and Probability of Parentage (W).

The Parentage Index is a likelihood ratio based on two conditional probabilities: the probability of parentage given that the alleged parent is the biological parent, and the probability that the alleged parent is not the biological parent. The general formula is as follows:

$$PI = \frac{\text{Probability of the genetic observations conditional on parentage}}{\text{Probability of the genetic observations conditional on non -parentage}}$$

The exact formula of PI in terms of band/allele occurrence frequencies depends on the obligate parental band/allele and the homozygosity of the alleged parent.

Parentage Case

The Probability of Parentage (W) is based upon Bayes' Theorem. It provides a method for determining a posterior probability for parentage based upon the DNA profiles of the known parent, child and alleged parent. To perform the Probability of Parentage calculation, the user must assign a value for the prior probability that the alleged parent is the true biological parent. The equation used to calculate W is as follows:

$$W = \frac{PI \cdot (\text{prior probability})}{PI \cdot (\text{prior probability}) + [1 - (\text{prior probability})]}$$

Therefore, the parentage probability of a parentage test for a locus in a population group is

$$W_{\text{locus}} = \frac{(PI_{\text{locus}}) \cdot P_{\text{prior}}}{(PI_{\text{locus}}) \cdot P_{\text{prior}} + (1 - P_{\text{prior}})}$$

where P_{prior} is the prior probability. The prior probability is user configurable and its default value is set to the neutral value of 0.5. That is, we assume there is a 50% probability that the alleged parent is the biological parent of the child and a 50% probability that the alleged parent is not the biological parent of the child.

Parentage Case

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 known parent. The DNA profile of the alleged parent is not considered in calculating the PE. The Probability of Exclusion is equal to the frequency of all the people in the population who do not contain an allele that matches the obligate paternal allele of the child.

If the phenotypes of the known parent, child, and alleged parent do not match at one locus, then "inconclusive" is declared for both the locus and the entire parentage test. If the phenotypes of the known parent, child, and alleged parent do not match at two or more loci, then "no match" is declared for the entire parentage test. The following table lists all the "matched" cases and their corresponding formulae for PI and PE.

Where

$$\hat{p}_i \text{ is defined to be } \hat{p}_i = \max\{p_{\min}, p_i\},$$

p and q is the band/allele occurrence frequency of the child

p_{\min} is the user-configured minimum frequency

E is the factor to be used as a conservative correction in PI for the RFLP homozygotes in the alleged parent's profile.

The default value for E is 0.5.

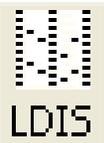
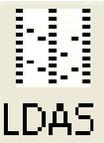
Parentage Case

The parentage index (PI), probability of exclusion (PE), and probability of parentage, W , are

$$PI = \prod_{loci} (PI_{locus})$$

$$PE = 1 - \prod_{loci} [1 - (PE_{locus})]$$

$$W = \frac{(PI) \cdot P_{prior}}{(PI) \cdot P_{prior} + (1 - P_{prior})}$$



Known Parent:

P-54534-M

Locus	Allele 1	Allele 2	
<input checked="" type="checkbox"/> D3S1358	14	17	
<input checked="" type="checkbox"/> VWA	16	17	
<input checked="" type="checkbox"/> FGA	22		
<input checked="" type="checkbox"/> D8S1179	12	14	
<input checked="" type="checkbox"/> D21S11	28	30	
<input checked="" type="checkbox"/> D18S51	15	19	
<input checked="" type="checkbox"/> D5S818	12		
<input checked="" type="checkbox"/> D13S317	12	13	
<input checked="" type="checkbox"/> D7S820	7	8	
<input checked="" type="checkbox"/> CSF1PO	8	12	
<input checked="" type="checkbox"/> TPOX	10	11	
<input checked="" type="checkbox"/> TH01	7	9	
<input checked="" type="checkbox"/> D16S539	12	13	

Child:

P-54534-C

Allele 1	Allele 2	
15	17	
17	18	
22		
12	16	
28	32	
13	15	
12		
12	13	
8	10	
8	12	
9	10	
9		
13		

Alleged Parent:

P-54534-AF

Allele 1	Allele 2	
15	16	
18	20	
22	24	
15	16	
28	32	
13	18	
8	12	
13		
10	11	
12		
9		
8	9	
9	13	

Popstats 5.3 - [Parentage Statistics]

File Edit Profile Case Type Configuration Window Help



CAU

BLK

Locus	PE(%)	PI	W(%)	Match?
D3S1358	5.6806E+01	2	6.6997E+01	Yes
VWA	6.0544E+01	2	6.9262E+01	Yes
FGA	6.5805E+01	3	7.2590E+01	Yes
D8S1179	9.7456E+01	39	9.7504E+01	Yes
D21S11	9.6963E+01	33	9.7031E+01	Yes
D18S51	7.7001E+01	4	8.0321E+01	Yes
D5S818	4.1745E+01	1	5.8555E+01	Yes
D13S317	3.3826E+01	2	7.0502E+01	Yes
D7S820	5.0325E+01	2	6.3243E+01	Yes
CSF1PO	4.3904E+01	3	7.4772E+01	Yes
TPOX	7.6878E+01	8	8.9031E+01	Yes
TH01	6.9722E+01	3	7.5188E+01	Yes
D16S539	6.9990E+01	3	7.5369E+01	Yes
Total	1.000E+02	81,800,000	1.000E+02	Yes

PE = 99.99999%

W = 99.99999%

Popstats 5.3 - [Parentage Statistics]

File Edit Profile Case Type Configuration Window Help



CAU

BLK

Locus	PE(%)	PI	W(%)	Match?
D3S1358	5.0339E+01	2	6.3251E+01	Yes
VWA	7.4632E+01	4	7.8604E+01	Yes
FGA	6.0062E+01	2	6.8966E+01	Yes
D8S1179	9.1317E+01	11	9.1844E+01	Yes
D21S11	9.7220E+01	36	9.7276E+01	Yes
D18S51	8.9189E+01	9	8.9993E+01	Yes
D5S818	4.1525E+01	1	5.8439E+01	Yes
D13S317	1.5296E+01	2	6.2154E+01	Yes
D7S820	4.5725E+01	2	6.0694E+01	Yes
CSF1PO	3.7736E+01	3	7.2166E+01	Yes
TPOX	6.6945E+01	6	8.4617E+01	Yes
TH01	7.3068E+01	3	7.7495E+01	Yes
D16S539	6.9706E+01	3	7.5177E+01	Yes

Total	1.000E+02	26,950,000	1.000E+02	Yes
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Popstats 5.3 - [Parentage Statistics]

File Edit Profile Case Type Configuration Window Help



CAU

BLK

SEH

Locus	PE(%)	PI	W(%)	Match?
D3S1358	4.1809E+01	1	5.8589E+01	Yes
VWA	6.6016E+01	3	7.2727E+01	Yes
FGA	7.2386E+01	3	7.7018E+01	Yes
D8S1179	9.4829E+01	19	9.5021E+01	Yes
D21S11	9.7397E+01	38	9.7447E+01	Yes
D18S51	7.7828E+01	4	8.0932E+01	Yes
D5S818	4.6690E+01	2	6.1222E+01	Yes
D13S317	4.3891E+01	3	7.4766E+01	Yes
D7S820	5.3773E+01	2	6.5215E+01	Yes
CSF1PO	4.0107E+01	3	7.3169E+01	Yes
TPOX	8.4034E+01	12	9.2311E+01	Yes
TH01	6.6357E+01	3	7.2950E+01	Yes
D16S539	7.0141E+01	3	7.5472E+01	Yes
Total	1.000E+02	93,130,000	1.000E+02	Yes

Popstats 5.3 - [Parentage Statistics]

File Edit Profile Case Type Configuration Window Help



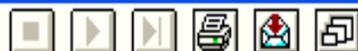
CAU

BLK

SEH

SWH

Locus	PE(%)	PI	W(%)	Match?
D3S1358	3.2971E+01	1	5.4007E+01	Yes
VWA	6.4867E+01	3	7.1984E+01	Yes
FGA	6.7684E+01	3	7.3823E+01	Yes
D8S1179	9.5141E+01	20	9.5311E+01	Yes
D21S11	9.7555E+01	41	9.7599E+01	Yes
D18S51	6.8890E+01	3	7.4627E+01	Yes
D5S818	5.0325E+01	2	6.3243E+01	Yes
D13S317	4.1641E+01	3	7.3817E+01	Yes
D7S820	4.8136E+01	2	6.2019E+01	Yes
CSF1PO	3.5486E+01	2	7.1210E+01	Yes
TPOX	9.3412E+01	30	9.6759E+01	Yes
TH01	8.0479E+01	5	8.2932E+01	Yes
D16S539	8.0389E+01	5	8.2864E+01	Yes
Total	1.000E+02	284,100,000	1.000E+02	Yes



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Popstats 5.3

Page 1

Parentage Calculations Part 1

Fixed Bin

Database: C:\CODISII\CODIS\POPDATA\FBI\STR
 User Name: eisenber
 Boundaries: 0 2000 10000
 Windows: > 0.025 <> 0.025 <> 0.080

Locus	Known Parent P-54534-M		Child P-54534-C		Alleged Parent P-54534-AF		Equation Number	Match?
D3S1358	14	17	15 op	17	15	16	2	Yes
VWA	16	17	17	18 op	18	20	4	Yes
FGA	22		22 op		22	24	2	Yes
D8S1179	12	14	12	16 op	15	16	4	Yes
D21S11	28	30	28	32 op	28	32	4	Yes
D18S51	15	19	13 op	15	13	18	2	Yes
D5S818	12		12 op		8	12	2	Yes
D13S317	12	13	12 op	13 op	13		6	Yes
D7S820	7	8	8	10 op	10	11	4	Yes
CSF1PO	8	12	8 op	12 op	12		6	Yes
TPOX	10	11	9 op	10	9		1	Yes
TH01	7	9	9 op		8	9	2	Yes
D16S539	12	13	13 op		9	13	2	Yes

op = obligate parentage allele

Popstats Help

Equation Numbers

Parentage Case

Case 1

Case 2

Case 3

Case 4

Case 5

Case 6

Case 7

Equation Number

1

Parentage - Case 1

For RFLP/VNTR loci:

$$PI_{\text{locus}} = \frac{E}{\hat{p}}, \quad PE_{\text{locus}} = (1 - \hat{p})^2$$

For PCR loci:

$$PI_{\text{locus}} = \frac{1}{\hat{p}}, \quad PE_{\text{locus}} = (1 - \hat{p})^2$$

TPOX

M	C	AF
10	9	9
11	10	op

Equation Number

2

Parentage - Case 2

$$PI_{\text{locus}} = \frac{0.5}{\hat{p}}, \quad PE_{\text{locus}} = (1 - \hat{p})^2$$

D3S1358

M	C	AF
14	15 _{op}	9
17	17	

FGA

M	C	AF
22	22 _{op}	22
		24

D18S51

M	C	AF
15	13 _{op}	13
19	15	18

TH01

M	C	AF
7	9 _{op}	8
9		9

Equation Number

3

Parentage - Case 3

For VNTR/RFLP loci:

$$PI_{\text{locus}} = \frac{E}{q}, \quad PE_{\text{locus}} = (1 - q)^2$$

For PCR loci:

$$PI_{\text{locus}} = \frac{1}{q}, \quad PE_{\text{locus}} = (1 - q)^2$$

M C AF

P P Q

R Q_{op}

M C AF

P Q_{op} Q

Q

M C AF

Q Q_{op} Q

Equation Number

4

Parentage - Case 4

$$PI_{\text{locus}} = \frac{0.5}{q}, PE_{\text{locus}} = (1 - q)^2$$

VWA

D8S1179

D21S11

D7S820

M	C	AF
16	17	18
17	18 _{op}	20

M	C	AF
12	12	15
14	16 _{op}	16

M	C	AF
28	28	28
30	32 _{op}	32

M	C	AF
7	8	10
8	10 _{op}	11

Equation Number

5

Parentage - Case 5

$$PI_{\text{locus}} = \frac{1}{p + q}, \quad PE_{\text{locus}} = [1 - (p + q)]^2$$

M	C	AF	M	C	AF	M	C	AF
P	P	P or	P	P	Q or	P	P	P
Q	Q		Q	Q		Q	Q	Q

Equation Number

6

Parentage - Case 6

For VNTR/RFLP loci:

$$PI_{bcus} = \frac{E}{p + q}, PE_{bcus} = [1 - (p + q)]^2$$

For PCR loci:

$$PI_{bcus} = \frac{1}{p + q}, PE_{bcus} = [1 - (p + q)]^2$$

CSF1PO

M	C	AF
8	8 _{op}	12
12	12 _{op}	

Equation Number

7

Parentage - Case 7

$$PI_{\text{locus}} = \frac{0.5}{p + q}, PE_{\text{locus}} = [1 - (p + q)]^2$$

M

C

AF

P

P

P

or

M

C

AF

Q

Q

S

Q

Q

S

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Page 1

Parentage Calculations Part 2

Fixed Bin

Database: C:\CODISII\CODIS\POPDATA\FBI\STR

User Name: eisenber

Boundaries: 0 2000 10000

Windows: > 0.025 <> 0.025 <> 0.080

Population Group: CAU

Locus	Probability of Exclusion	Parentage Index	Probability of Parentage	Match?
D3S1358	5.6806E+01 %	2.0300E+00	6.6997E+01 %	Yes
VWA	6.0544E+01 %	2.2533E+00	6.9262E+01 %	Yes
FCA	6.5805E+01 %	2.6483E+00	7.2590E+01 %	Yes
D8S1179	9.7456E+01 %	3.9062E+01	9.7504E+01 %	Yes
D21S11	9.6963E+01 %	3.2680E+01	9.7031E+01 %	Yes
D18S51	7.7001E+01 %	4.0816E+00	8.0321E+01 %	Yes
D5S818	4.1745E+01 %	1.4128E+00	5.8555E+01 %	Yes
D13S317	3.3826E+01 %	2.3901E+00	7.0502E+01 %	Yes
D7S820	5.0325E+01 %	1.7206E+00	6.3243E+01 %	Yes
CSF1P0	4.3904E+01 %	2.9638E+00	7.4772E+01 %	Yes
TPOX	7.6878E+01 %	8.1169E+00	8.9031E+01 %	Yes
TH01	6.9722E+01 %	3.0303E+00	7.5188E+01 %	Yes
D16S539	6.9990E+01 %	3.0600E+00	7.5369E+01 %	Yes
Total	1.000E+02 %	8.180E+07	1.000E+02 %	Yes

Population Group: BLK

Locus	Probability of Exclusion	Parentage Index	Probability of Parentage	Match?
D3S1358	5.0339E+01 %	1.7212E+00	6.3251E+01 %	Yes
VWA	7.4632E+01 %	3.6738E+00	7.8604E+01 %	Yes
FGA	6.0062E+01 %	2.2222E+00	6.8966E+01 %	Yes
D8S1179	9.1317E+01 %	1.1261E+01	9.1844E+01 %	Yes
D21S11	9.7220E+01 %	3.5714E+01	9.7276E+01 %	Yes
D18S51	8.9189E+01 %	8.9928E+00	8.9993E+01 %	Yes
D5S818	4.1525E+01 %	1.4061E+00	5.8439E+01 %	Yes
D13S317	1.5296E+01 %	1.6423E+00	6.2154E+01 %	Yes
D7S820	4.5725E+01 %	1.5442E+00	6.0694E+01 %	Yes
CSF1PO	3.7736E+01 %	2.5927E+00	7.2166E+01 %	Yes
TPOX	6.6945E+01 %	5.5006E+00	8.4617E+01 %	Yes
TH01	7.3068E+01 %	3.4435E+00	7.7495E+01 %	Yes
D16S539	6.9706E+01 %	3.0285E+00	7.5177E+01 %	Yes
Total	1.000E+02 %	2.695E+07	1.000E+02 %	Yes

Population Group: SEN

Locus	Probability of Exclusion	Parentage Index	Probability of Parentage	Match?
D3S1358	4.1809E+01 %	1.4148E+00	5.8589E+01 %	Yes
VWA	6.6016E+01 %	2.6667E+00	7.2727E+01 %	Yes
FGA	7.2386E+01 %	3.3512E+00	7.7018E+01 %	Yes
D8S1179	9.4829E+01 %	1.9084E+01	9.5021E+01 %	Yes
D21S11	9.7397E+01 %	3.8168E+01	9.7447E+01 %	Yes
D18S51	7.7828E+01 %	4.2445E+00	8.0932E+01 %	Yes
D5S818	4.6690E+01 %	1.5788E+00	6.1222E+01 %	Yes
D13S317	4.3891E+01 %	2.9630E+00	7.4766E+01 %	Yes
D7S820	5.3773E+01 %	1.8748E+00	6.5215E+01 %	Yes
CSF1PO	4.0107E+01 %	2.7270E+00	7.3169E+01 %	Yes
TPOX	8.4034E+01 %	1.2005E+01	9.2311E+01 %	Yes
TH01	6.6357E+01 %	2.6969E+00	7.2950E+01 %	Yes
D16S539	7.0141E+01 %	3.0769E+00	7.5472E+01 %	Yes

Total	1.000E+02 %	9.313E+07	1.000E+02 %	Yes
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Popstats 5.3

Page 2

Parentage Calculations Part 2

Fixed Bin

Database: C:\CODISII\CODIS\POPDATA\FBI\STR

User Name: eisenber

Boundaries: 0 2000 10000

Windows: > 0.025 <> 0.025 <> 0.080

Population Group: SWH

Locus	Probability of Exclusion	Parentage Index	Probability of Parentage	Match?
D3S1358	3.2971E+01 %	1.1743E+00	5.4007E+01 %	Yes
VWA	6.4867E+01 %	2.5694E+00	7.1984E+01 %	Yes
FGA	6.7684E+01 %	2.8201E+00	7.3823E+01 %	Yes
D8S1179	9.5141E+01 %	2.0325E+01	9.5311E+01 %	Yes
D21S11	9.7555E+01 %	4.0650E+01	9.7599E+01 %	Yes
D18S51	6.8890E+01 %	2.9412E+00	7.4627E+01 %	Yes
D5S818	5.0325E+01 %	1.7206E+00	6.3243E+01 %	Yes
D13S317	4.1641E+01 %	2.8193E+00	7.3817E+01 %	Yes
D7S820	4.8136E+01 %	1.6329E+00	6.2019E+01 %	Yes
CSF1PO	3.5486E+01 %	2.4734E+00	7.1210E+01 %	Yes
TPOX	9.3412E+01 %	2.9851E+01	9.6759E+01 %	Yes
TH01	8.0479E+01 %	4.8591E+00	8.2932E+01 %	Yes
D16S539	8.0389E+01 %	4.8356E+00	8.2864E+01 %	Yes
Total	1.000E+02 %	2.841E+08	1.000E+02 %	Yes

Specifications

Conservative Practice for Handling Single Paternal Bands = 0.5

Prior probability = 0.5

**Popstats Cannot Correctly
Calculate Parentage Statistics
in
Non-Typical Cases**

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)**

Popstats 5.3 - [Parentage Target Profile]

File Edit Profile Case Type Configuration Window Help



- LDAS
- LDIS
-
-

Known Parent:

P-41411-M

Child:

P-41411-C

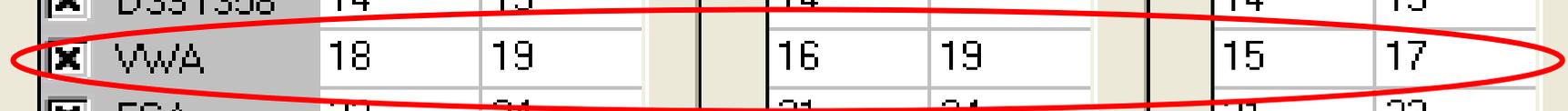
Alleged Parent:

P-41411-AF

	Locus	Allele 1	Allele 2	
<input checked="" type="checkbox"/>	D3S1358	14	15	
<input checked="" type="checkbox"/>	VWA	18	19	
<input checked="" type="checkbox"/>	FGA	23	24	
<input checked="" type="checkbox"/>	D8S1179	10	14	
<input checked="" type="checkbox"/>	D21S11	28	30	
<input checked="" type="checkbox"/>	D18S51	14	17	
<input checked="" type="checkbox"/>	D5S818	11	13	
<input checked="" type="checkbox"/>	D13S317	10	12	
<input checked="" type="checkbox"/>	D7S820	9	10	
<input checked="" type="checkbox"/>	CSF1PO	8	12	
<input checked="" type="checkbox"/>	TPOX	8	11	
<input checked="" type="checkbox"/>	TH01	7		
<input checked="" type="checkbox"/>	D16S539	12		

Allele 1	Allele 2	
14		
16	19	
21	24	
14	15	
29	30	
14	17	
11		
8	12	
9	11	
8	12	
8	11	
7	9	
11	12	

Allele 1	Allele 2	
14	15	
15	17	
21	23	
13	15	
28	29	
14		
11		
8	11	
8	11	
10	12	
8	11	
6	9	
11	12	



Popstats 5.3 - [Parentage Statistics]

File Edit Profile Case Type Configuration Window Help



CAU

BLK

Locus	PE(%)	PI	W(%)	Match?
D3S1358	7.2881E+01	4	7.8076E+01	Yes
VWA				No
FGA	6.8310E+01	3	7.4239E+01	Yes
D8S1179	7.9263E+01	5	8.2008E+01	Yes
D21S11	6.7060E+01	3	7.3411E+01	Yes
D18S51	4.5011E+01	3	7.5239E+01	Yes
D5S818	3.4775E+01	2	7.0907E+01	Yes
D13S317	8.1090E+01	5	8.3403E+01	Yes
D7S820	6.3680E+01	2	7.1225E+01	Yes
CSF1PO	4.3904E+01	1	5.9709E+01	Yes
TPOX	4.0804E+00	1	5.5617E+01	Yes
TH01	6.9722E+01	3	7.5188E+01	Yes
D16S539	5.2955E+01	2	6.4742E+01	Yes
Total	1.000E+02	122,900	1.000E+02	Inconclusive

Parentage Calculations Part 1

Fixed Bin

Database: C:\CODISII\CODIS\POPDATA\FBI\STR

User Name: eisenber

Boundaries: 0 2000 10000

Windows: > 0.025 <> 0.025 <> 0.080

Locus	Known Parent P-41411-M		Child P-41411-C		Alleged Parent P-41411-AF		Equation Number	Match?
D3S1358	14	15	14 op		14	15	2	Yes
VWA	18	19	16	19	15	17		No
FGA	23	24	21 op	24	21	23	2	Yes
D8S1179	10	14	14	15 op	13	15	4	Yes
D21S11	28	30	29 op	30	28	29	2	Yes
D18S51	14	17	14 op	17 op	14		6	Yes
D5S818	11	13	11 op		11		1	Yes
D13S317	10	12	8 op	12	8	11	2	Yes
D7S820	9	10	9	11 op	8	11	4	Yes
CSF1P0	8	12	8 op	12 op	10	12	7	Yes
TPOX	8	11	8 op	11 op	8	11	5	Yes
TH01	7		7	9 op	6	9	4	Yes
D16S539	12		11 op	12	11	12	2	Yes

op = obligate parentage allele

Parentage Calculations Part 2

Fixed Bin

Database: C:\CODISII\CODIS\POPDATA\FBI\STR

User Name: eisenber

Boundaries: 0 2000 10000

Windows: > 0.025 <> 0.025 <> 0.080

Population Group: CAU

Locus	Probability of Exclusion	Parentage Index	Probability of Parentage	Match?
D3S1358	7.3891E+01 %	3.5613E+00	7.8076E+01 %	Yes
VWA				No
FGA	6.8310E+01 %	2.8818E+00	7.4239E+01 %	Yes
D8S1179	7.9263E+01 %	4.5579E+00	8.2008E+01 %	Yes
D21S11	6.7060E+01 %	2.7609E+00	7.3411E+01 %	Yes
D18S51	4.5011E+01 %	3.0386E+00	7.5239E+01 %	Yes
D5S818	3.4775E+01 %	2.4372E+00	7.0907E+01 %	Yes
D13S317	8.1090E+01 %	5.0251E+00	8.3403E+01 %	Yes
D7S820	6.3680E+01 %	2.4752E+00	7.1225E+01 %	Yes
CSF1P0	4.3904E+01 %	1.4819E+00	5.9709E+01 %	Yes
TPOX	4.0804E+00 %	1.2531E+00	5.5617E+01 %	Yes
TH01	6.9722E+01 %	3.0303E+00	7.5188E+01 %	Yes
D16S539	5.2955E+01 %	1.8362E+00	6.4742E+01 %	Yes
Total	1.000E+02 %	1.229E+05	1.000E+02 %	Inconclusive

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 where the tested man is the true biological father.

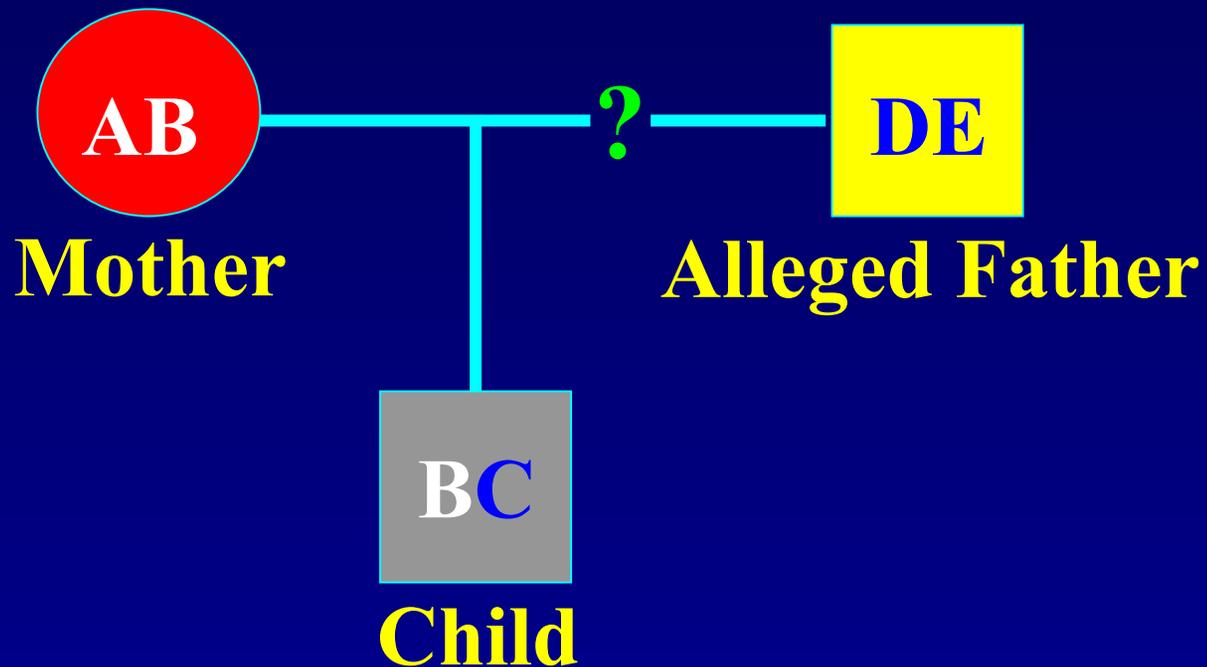
Mutations in Paternity Testing

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

<u>Person</u>	<u>Type</u>
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

<u>Person</u>	<u>Type</u>
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

$$X = P(\text{man without } \mathbf{C} \text{ allele will contribute } \mathbf{C} \text{ allele}) \\ = P(\text{contributed gene will mutate}) \times P(\text{mutated gene will be a } \mathbf{C})$$

Single Inconsistency

Numerator

$$X = P(\text{man without } C \text{ will contribute } C)$$

$$X = P(\text{contributed gene will mutate}) \\ \times P(\text{mutated gene will be a } C)$$

μ = observed rate of mutations/meiosis for the locus

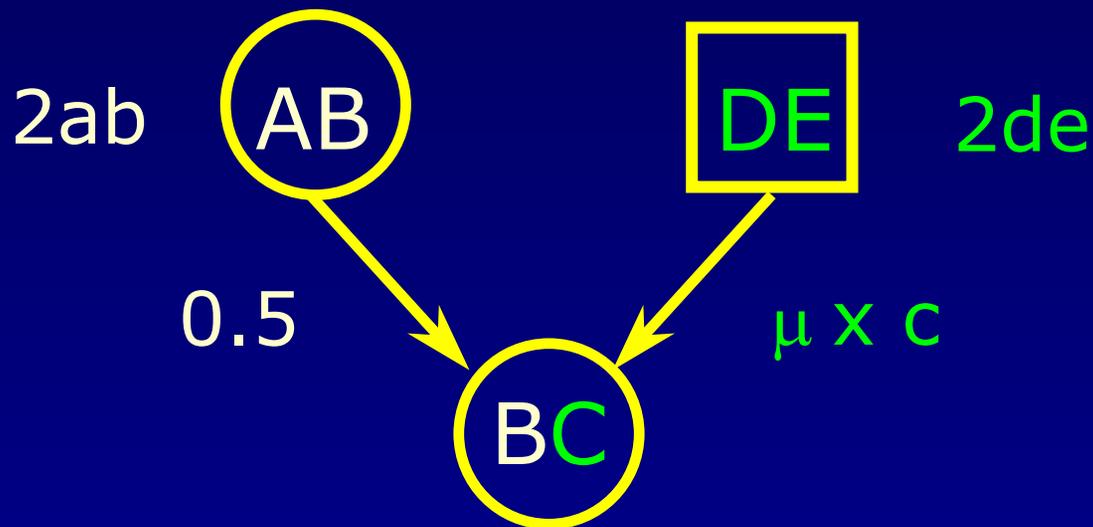
$P(\text{mutated gene will be a } C)$ ie. Frequency of C allele = c

$$X = \mu \times c$$

Single Inconsistency

Calculating a Paternity Index

Numerator



$$\text{Probability} = 2ab \times 2de \times 0.5 \times \mu \times c$$

Single Inconsistency

Denominator

<u>Person</u>	<u>Type</u>
Mother	AB
Child	BC
Alleged Father	DE

In order to explain this evidence Calculate **Probability** that

- Woman randomly selected from population is type AB
- An alternative man randomly selected from population is type **DE**, and
- The woman's child, fathered by random man, is type **BC**

Single Inconsistency

Denominator

<u>Person</u>	<u>Type</u>
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

$$Y = P(\text{paternal allele is } \mathbf{C} \text{ and random man has no } \mathbf{C} \text{ allele}) \\ = P(\text{paternal gene is } \mathbf{C}) \times P(\text{random man has no } \mathbf{C} \text{ allele})$$

Single Inconsistency Denominator

$$Y = P(\text{paternal allele is } C \text{ and random man has no } C \text{ allele})$$
$$= P(\text{paternal gene is } C) \times P(\text{random man has no } C \text{ allele})$$

$P(\text{paternal allele will be a } C) \text{ ie. Frequency of } C \text{ allele} = c$

$P(\text{random man has no } C \text{ allele}) = \text{probability of exclusion}$

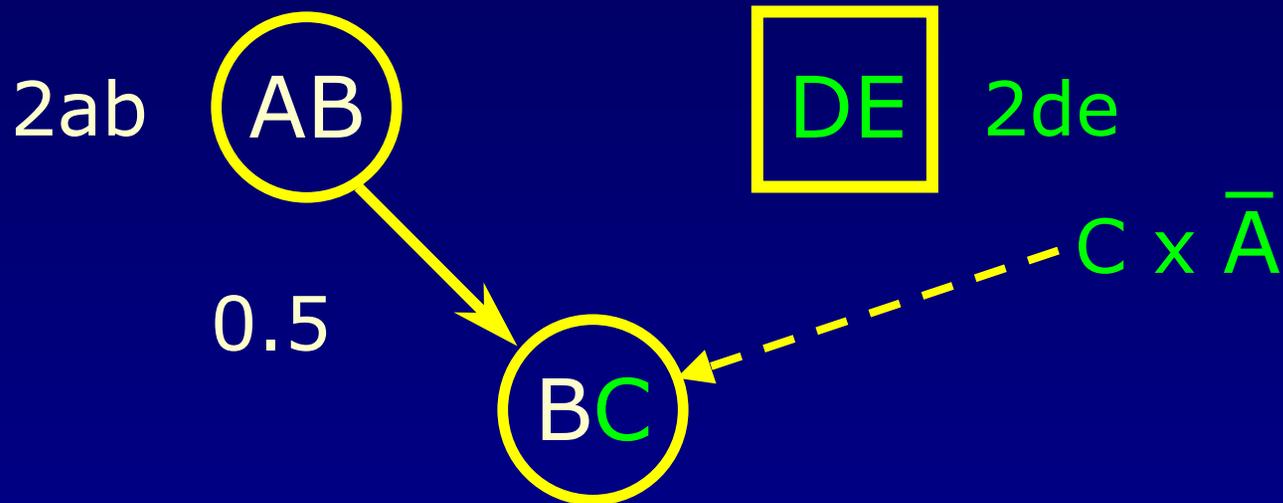
The AABB does not use the case specific power of exclusion,
but the mean power of exclusion (\bar{A})

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

Single Inconsistency

Calculating a Paternity Index

Denominator



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

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

<u>Locus</u>	<u>Mutation Rate</u>	<u>Mean PE</u>
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

<u>Locus</u>	<u>Mutation Rate</u>	<u>Mean PE</u>
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

<u>Locus</u>	<u>Mutation Rate</u>	<u>Mean PE</u>
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	$\frac{\mu}{\bar{A}}$ $(0.0034/0.667)$

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 11p	12 11	$0.5/a$
D7S820	10 9	11p 9m	11 10	$0.5/a$
D13S317	12 10	12m 8p	11 8	$0.5/a$
D5S818	13 11	11	11	$1/a$

Single Inconsistency

P-41411

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

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

H₂: 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 11p	12 11	1.84	1.42
D7S820	10 9	11p 9m	11 10	2.48	1.74
D13S317	12 10	12m 8p	11 8	5.03	3.02
D5S818	13 11	11	11	2.44	1.72

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:

Combined Paternity Index

Combined Avuncular Index

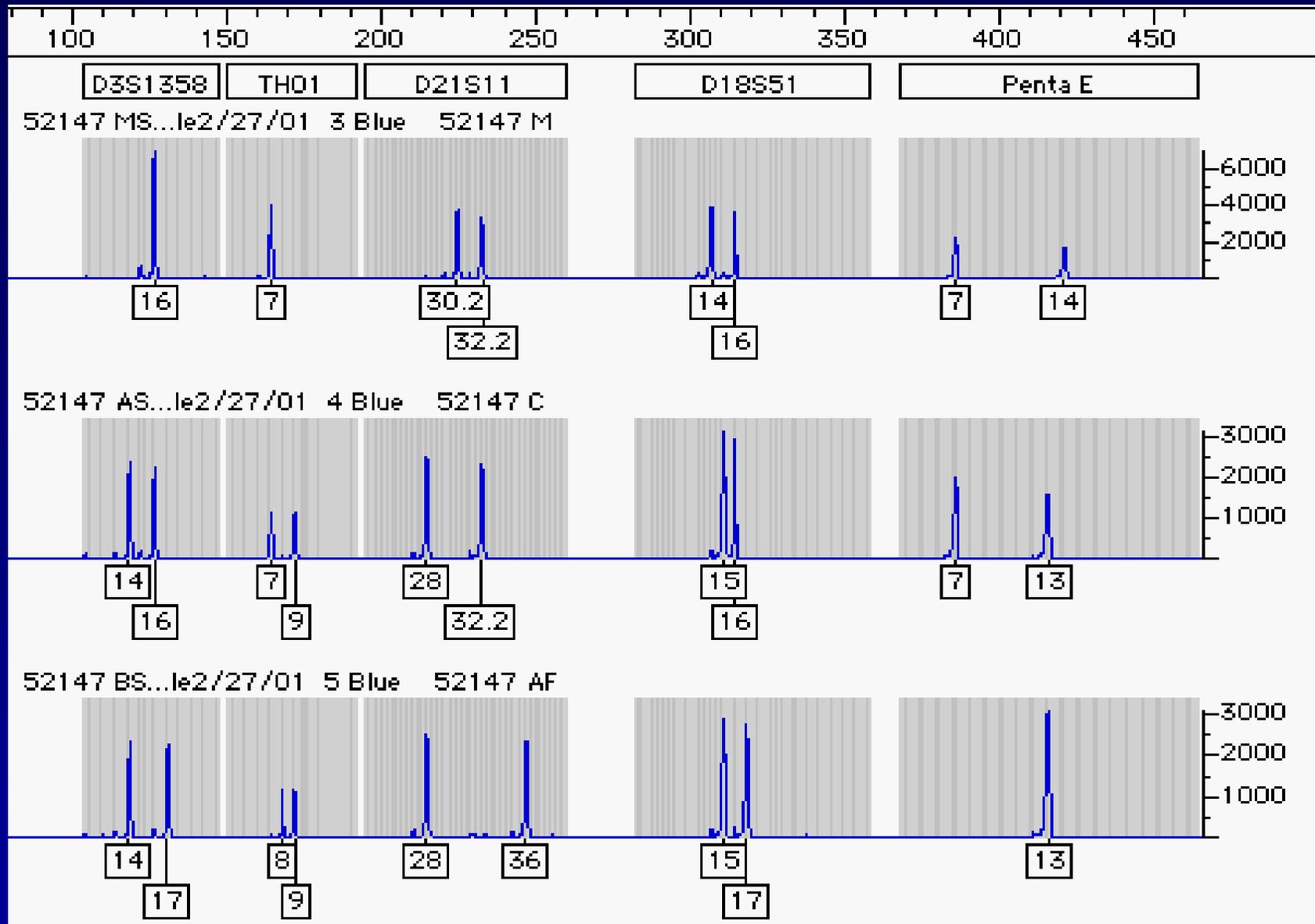
$$\frac{578,603}{101,683} = 5.69$$

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.

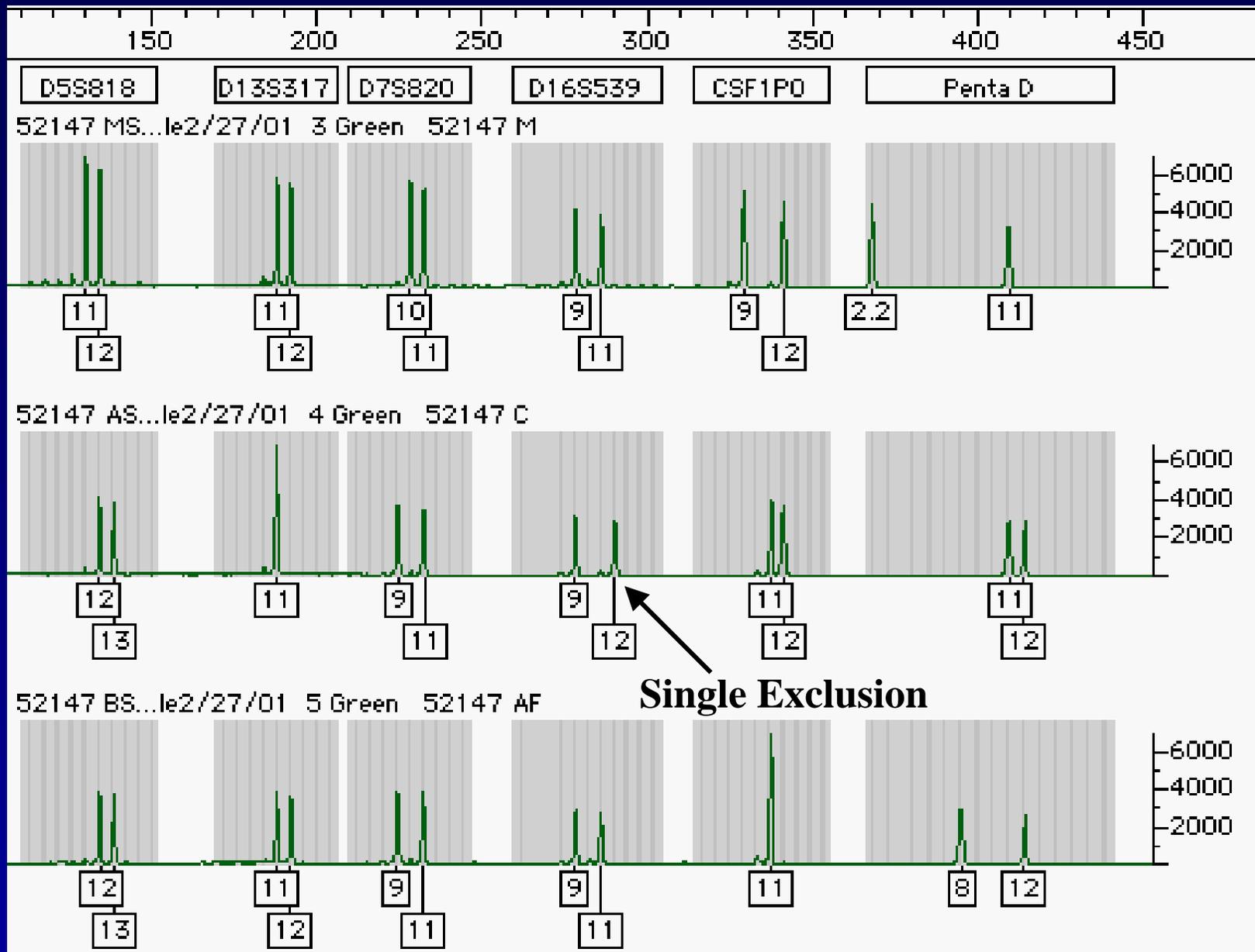
PowerPlex™ 16 System

Extremely Useful in Cases with
a Single Non-Matching Locus

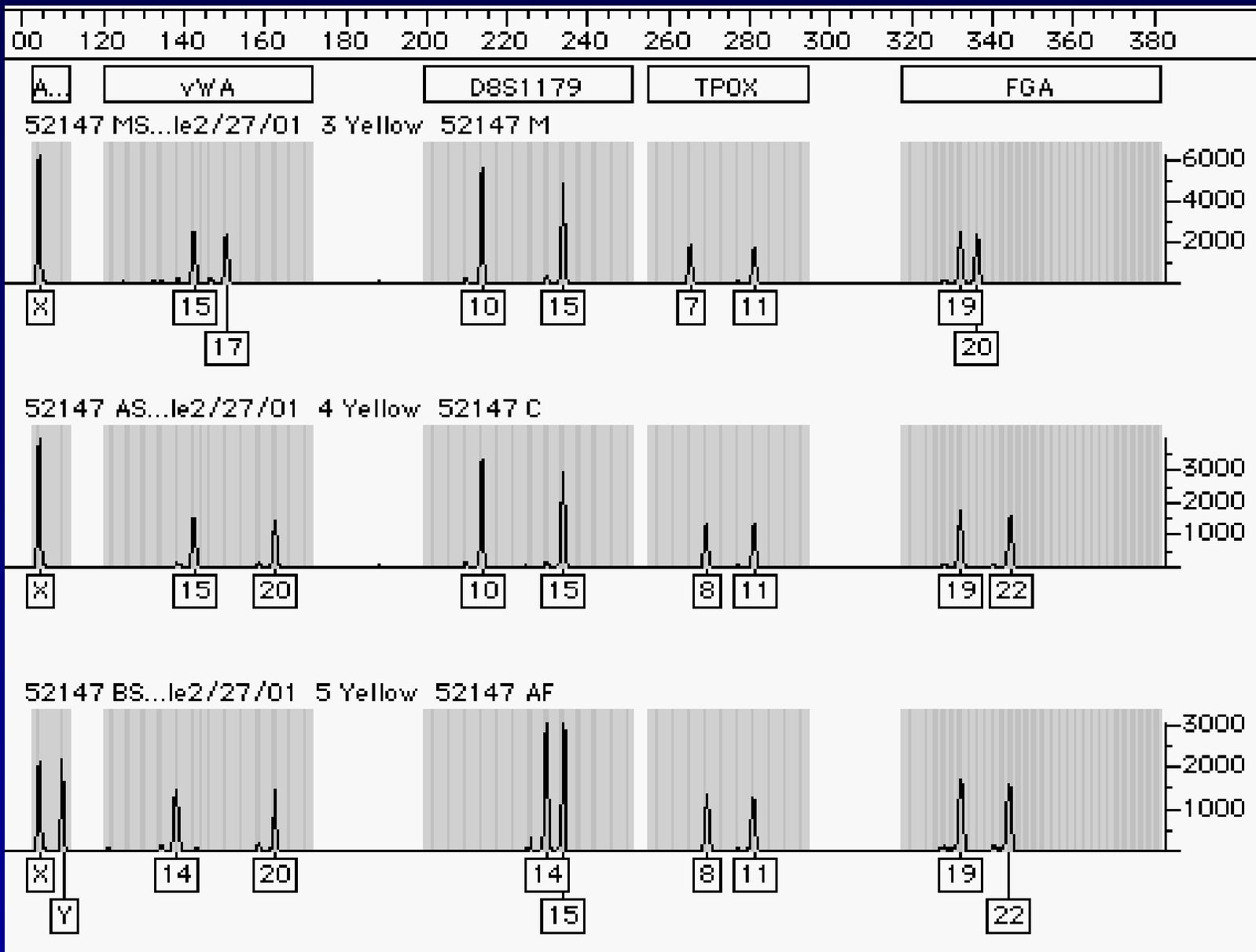
P-52147 Case of Single Exclusion



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**

What if We Don't Have the Mother's Genetic Data?

Popstats Cannot Calculate the Paternity Statistics
Without the Known Parent (Mother)

We can still develop a likelihood
estimation for parentage.

Lets examine the following logic:

Popstats 5.3 - [Parentage Target Profile]

File Edit Profile Case Type Configuration Window Help



LDAS
LDIS

Known Parent:
Not Tested

	Locus	Allele 1	Allele 2
<input type="checkbox"/>	D3S1358		
<input type="checkbox"/>	VWA		
<input type="checkbox"/>	FGA		
<input type="checkbox"/>	D8S1179		
<input type="checkbox"/>	D21S11		
<input type="checkbox"/>	D18S51		
<input type="checkbox"/>	D5S818		
<input type="checkbox"/>	D13S317		
<input type="checkbox"/>	D7S820		
<input type="checkbox"/>	CSF1PO		
<input type="checkbox"/>	TPOX		
<input type="checkbox"/>	TH01		
<input type="checkbox"/>	D16S539		

Child:
P-54137-C

Allele 1	Allele 2
16	17
19	20
22	23
15	
30	32.2
13	17
12	13
11	14
8	10
12	13
8	9
9.3	
10	11

Alleged Parent:
P-54137-AF

Allele 1	Allele 2
17	18
18	20
22	23
13	15
28	30
12	17
11	13
9	11
10	12
11	13
8	11
9	9.3
11	12

Popstats Can only Calculate with a Complete Trio
(Mother, Child, Alleged Father)

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.

$$X = \Pr\{AF \text{ passes } A\} \times \Pr\{M \text{ passes } B\} + \Pr\{AF \text{ passes } B\} \times \Pr\{M \text{ passes } A\}$$

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.

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

Motherless Paternity Index

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

$$X = \Pr\{\text{AF passes } A\} \times \Pr\{\text{M passes } B\} + \Pr\{\text{AF passes } B\} \times \Pr\{\text{M passes } A\}$$

$$X = \Pr\{\text{AF passes } A\} \times q + \Pr\{\text{AF passes } B\} \times p$$

$$\Pr\{\text{AF passes } A\} = 0.5$$

$$\Pr\{\text{AF passes } B\} = 0$$

$$X = 0.5 \times q + 0 \times p$$

$$X = 0.5q$$

Motherless Paternity Index

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

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

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

$$Y = 2pq$$

Motherless Paternity Index

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

$$PI = X / Y$$

$$X = 0.5q$$

$$Y = 2pq$$

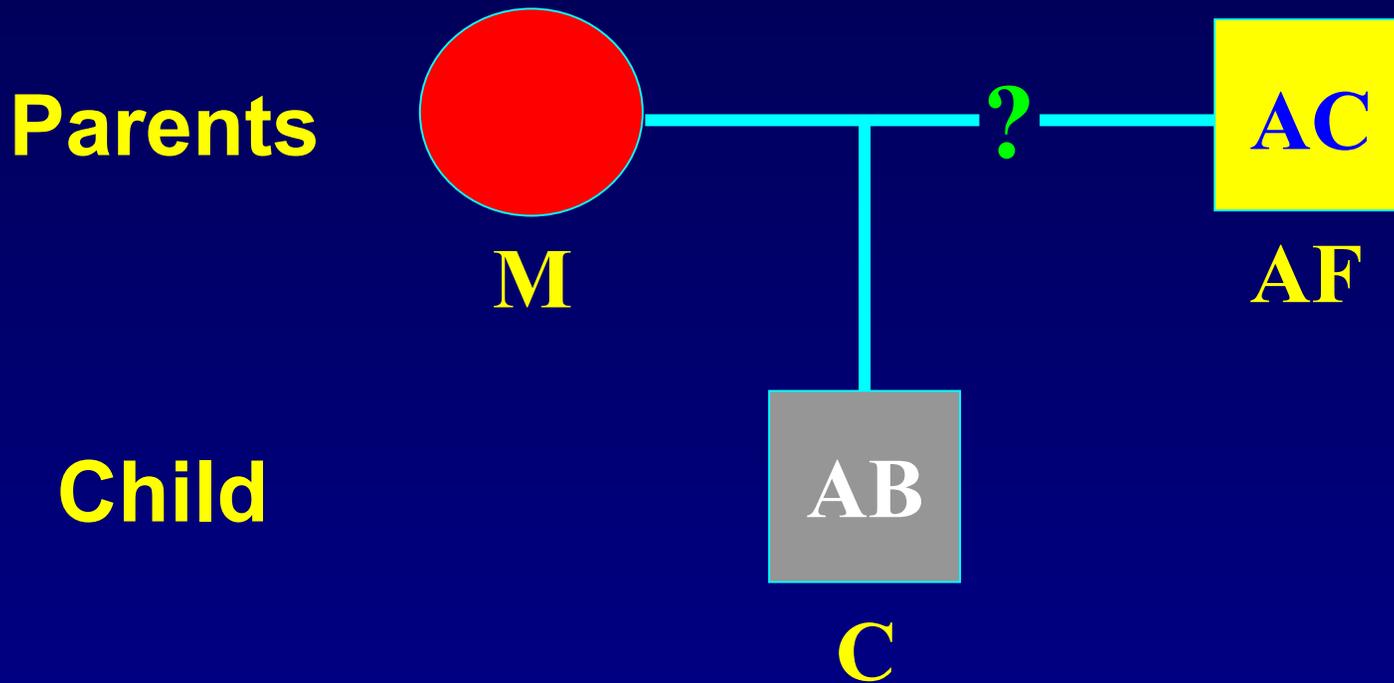
$$PI = 0.5q / 2pq$$

$$PI = 0.25/p$$

$$PI = 1/4p$$

Paternity Index

Only Man and Child Tested



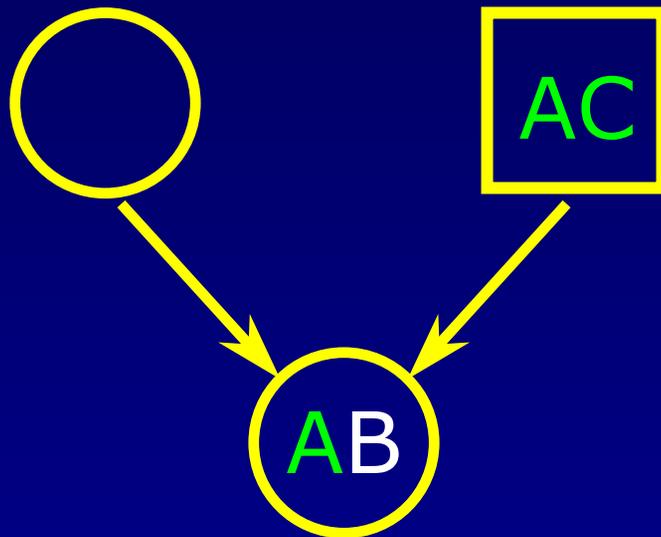
The untested Mother could have passed either the A or B allele

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

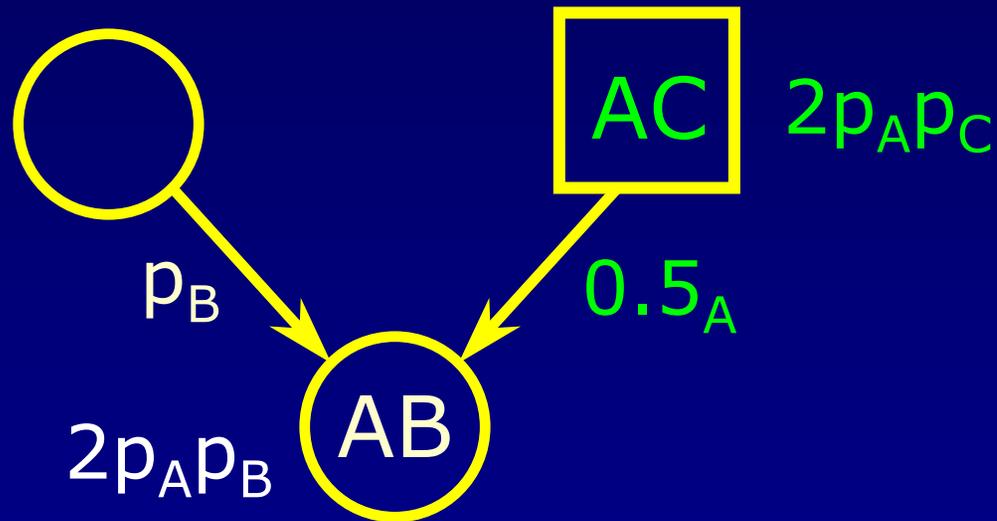
Only Man and Child Tested



Paternity Index

Only Man and Child Tested

Numerator

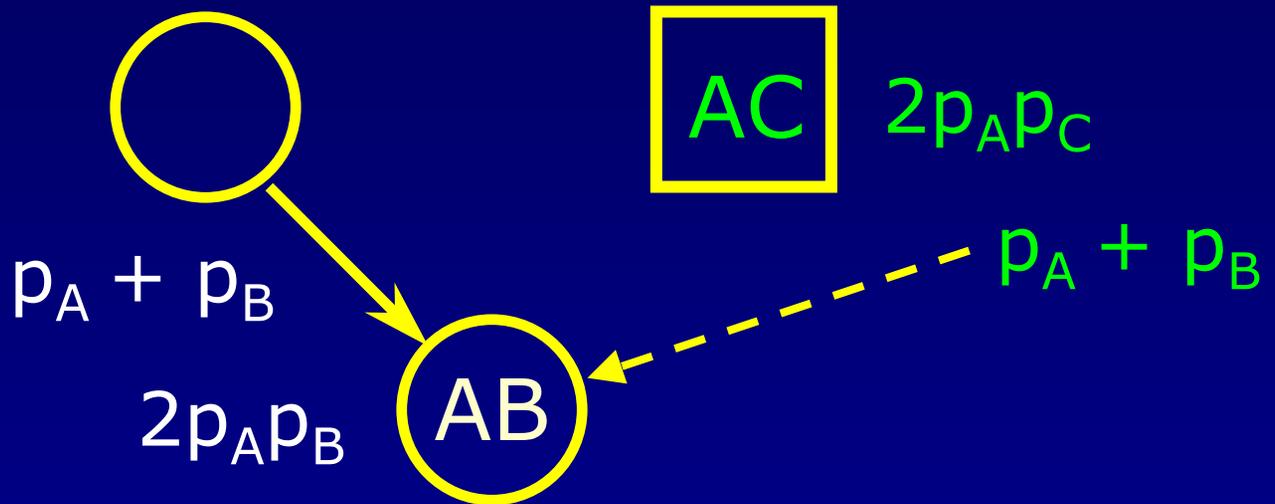


$$\text{Probability} = 2p_A p_C \times 2p_A p_B \times 0.5_{(fA)} \times p_B$$

Paternity Index

Only Man and Child Tested

Denominator



probability =

$$2p_A p_C \times 2p_A p_B \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})$$

Paternity Index

Only Man and Child Tested

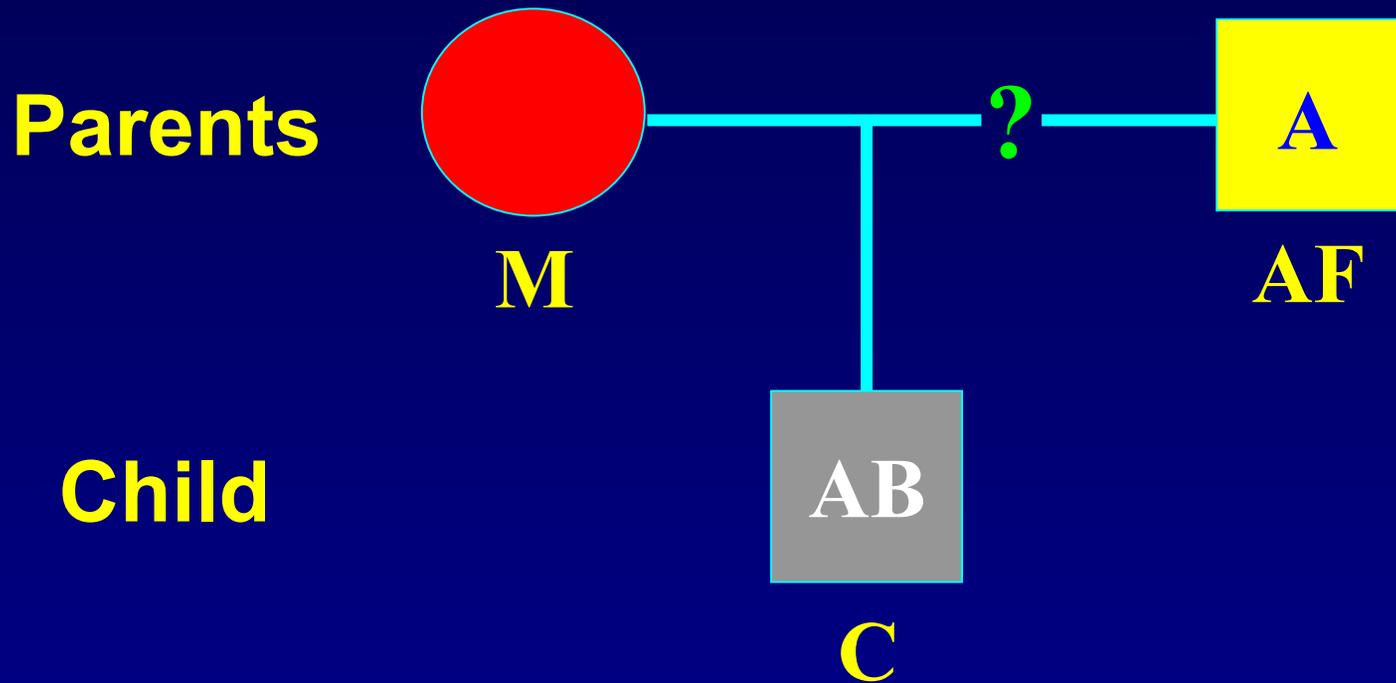
$$PI = \frac{\cancel{2p_A p_B} \times \cancel{2p_A p_C} \times 0.5_{(mA)} \times p_B}{\cancel{2p_A p_B} \times \cancel{2p_A p_C} \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})}$$

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

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

Paternity Index

Only Man and Child Tested



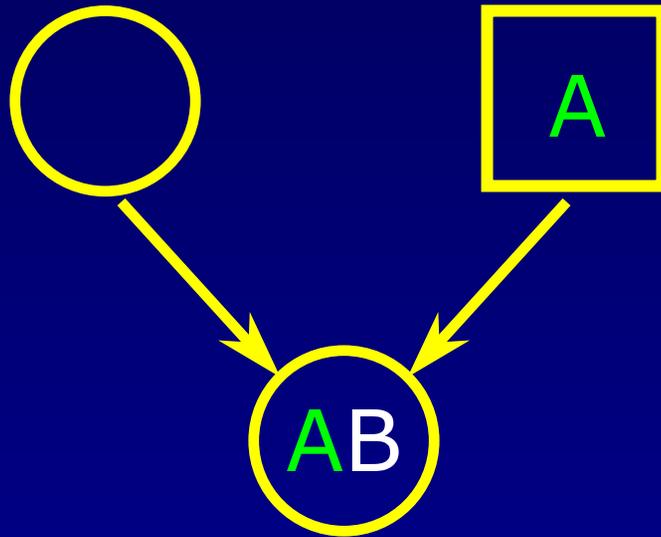
The untested Mother could have passed either
the A or B allele

AF can only pass A allele

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

Paternity Index

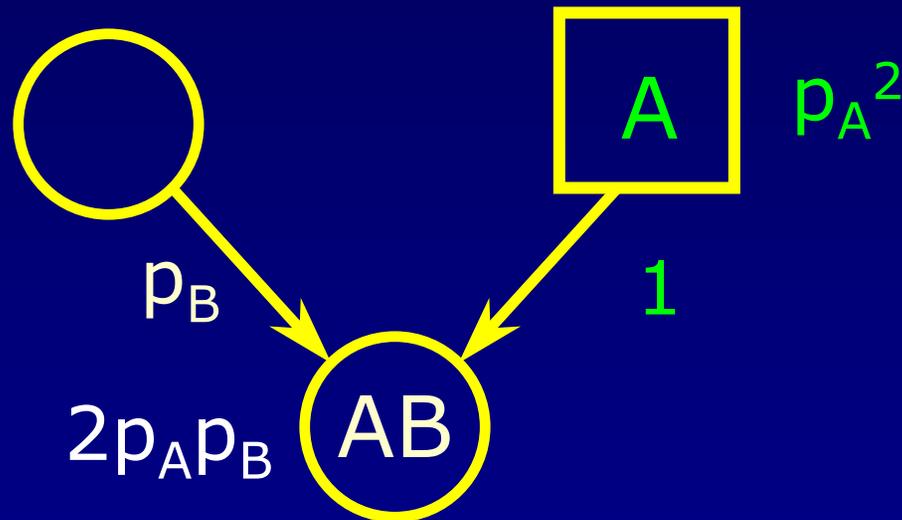
Only Man and Child Tested



Paternity Index

Only Man and Child Tested

Numerator

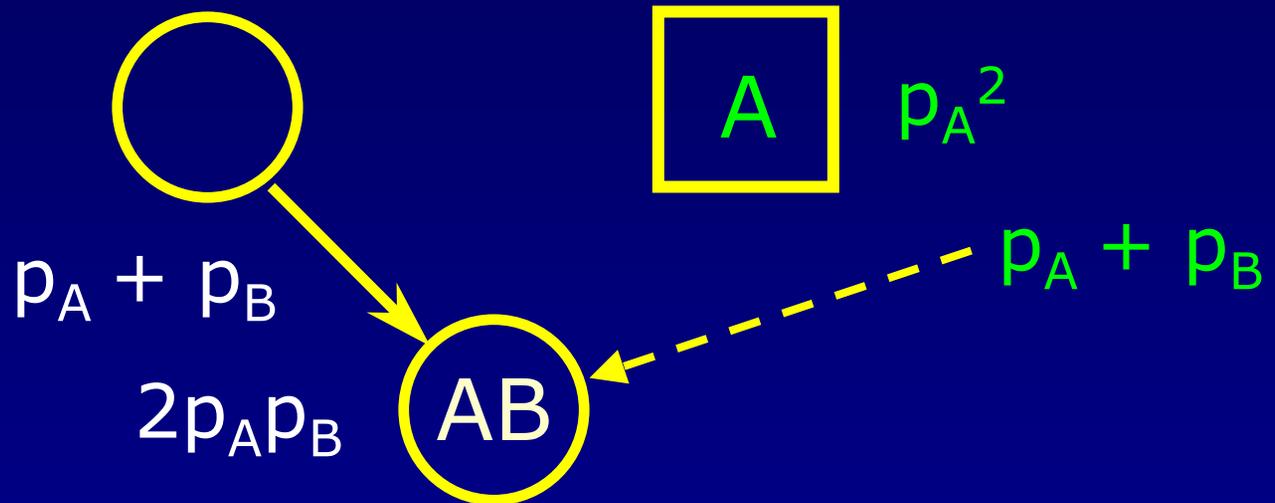


$$\text{Probability} = p_A^2 \times 2p_A p_B \times 1_{(fA)} \times p_B$$

Paternity Index

Only Man and Child Tested

Denominator



probability =

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

Paternity Index

Only Man and Child Tested

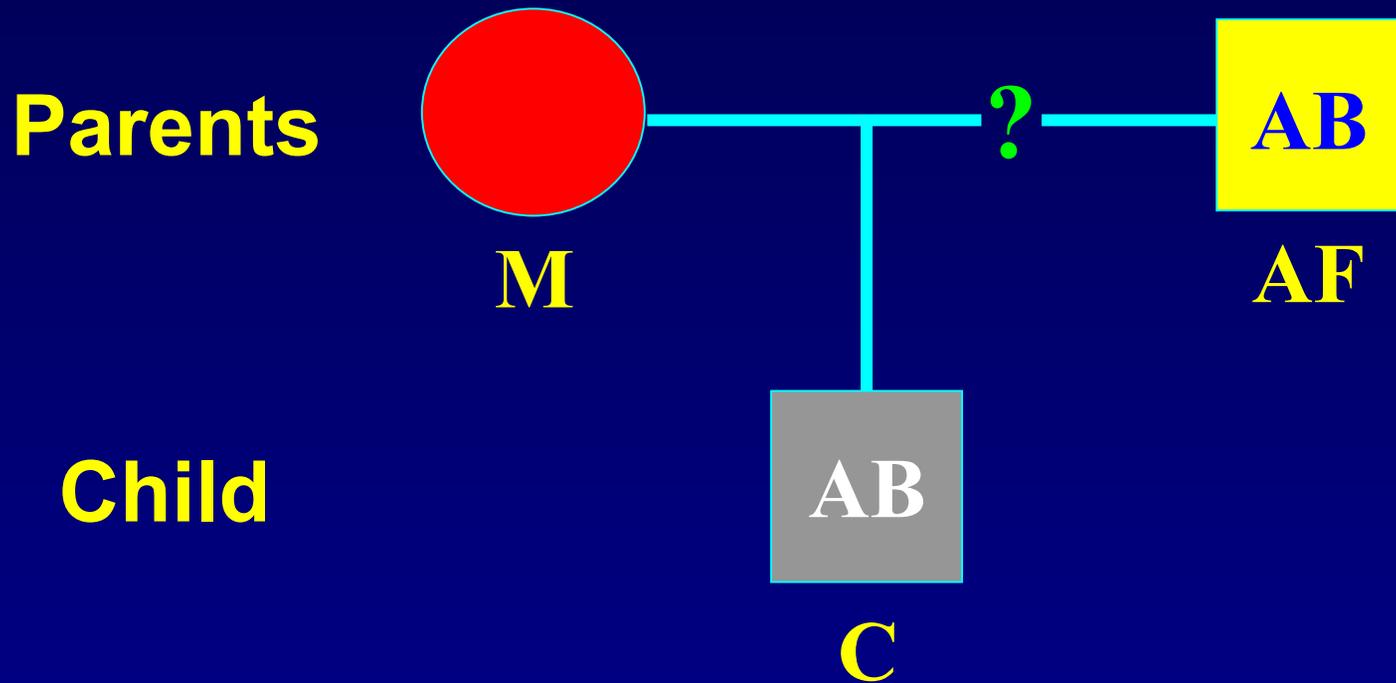
$$PI = \frac{p_A^2 \times 2p_A p_C \times 1_{(mA)} \times p_B}{p_A^2 \times 2p_A p_C \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})}$$

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

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

Paternity Index

Only Man and Child Tested



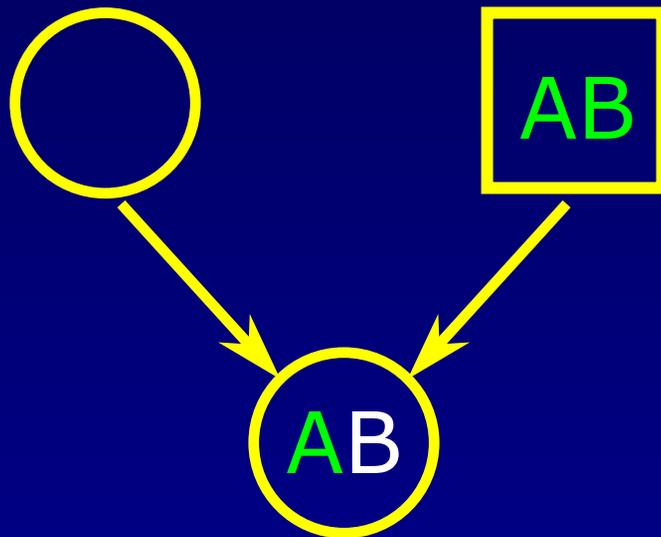
The untested Mother could have passed either the A or B allele

AF can pass either A or B allele

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

Paternity Index

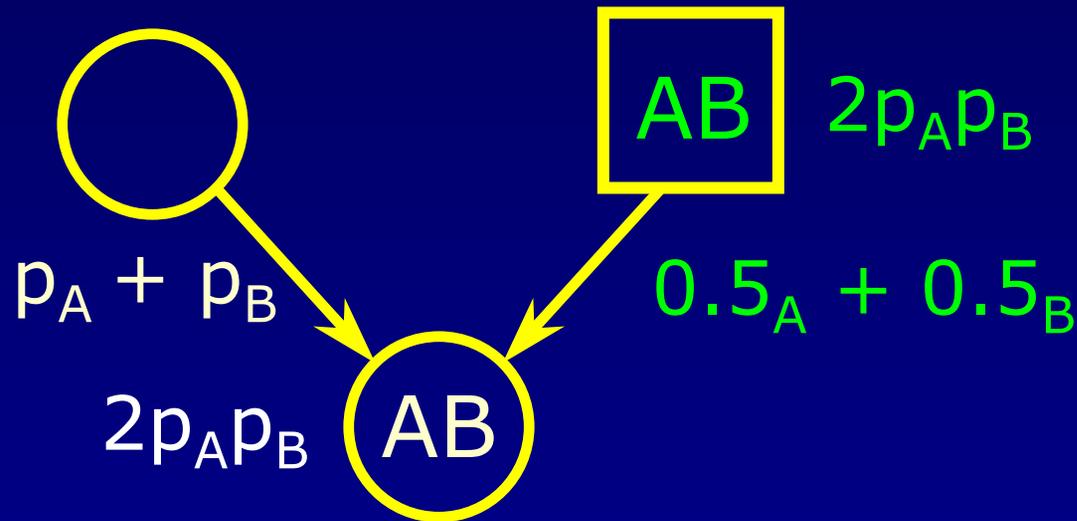
Only Man and Child Tested



Paternity Index

Only Man and Child Tested

Numerator



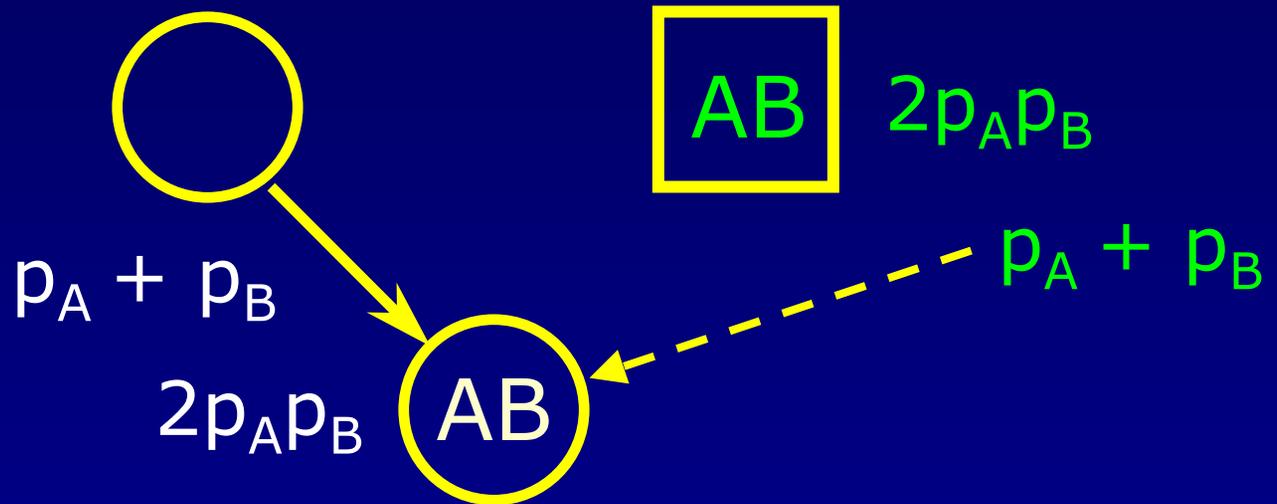
Probability =

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

Paternity Index

Only Man and Child Tested

Denominator



probability =

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

Paternity Index

Only Man and Child Tested

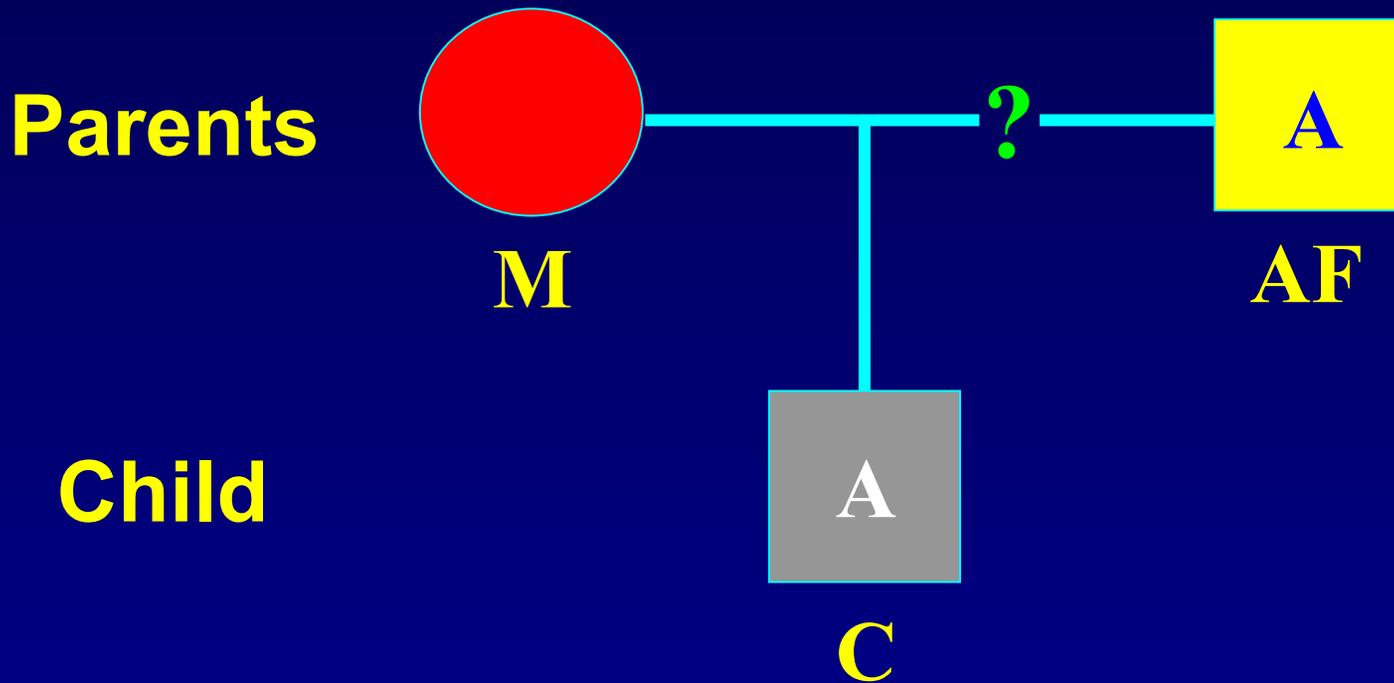
$$PI = \frac{\cancel{2p_A p_B} \times \cancel{2p_A p_B} \times (0.5_{(fA)} \times p_B + 0.5_{(fB)} \times p_A)}{\cancel{2p_A p_B} \times \cancel{2p_A p_B} \times (p_{(mA)} \times p_{(fB)} + p_{(mB)} \times p_{(fA)})}$$

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

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

Paternity Index

Only Man and Child Tested



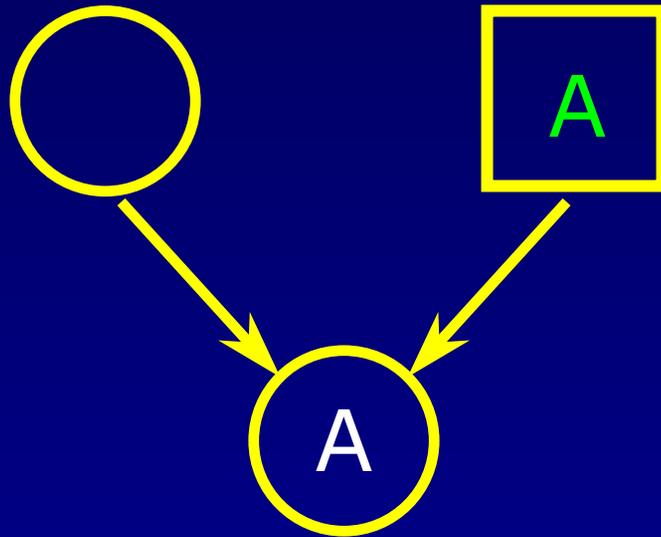
The untested Mother would have to pass an A allele

AF can pass only the A allele

RM has p chance of passing the A allele

Paternity Index

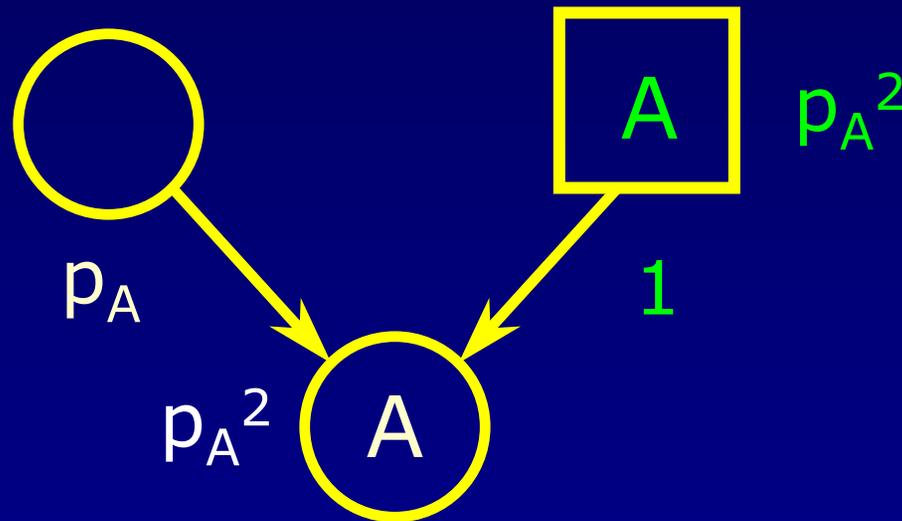
Only Man and Child Tested



Paternity Index

Only Man and Child Tested

Numerator

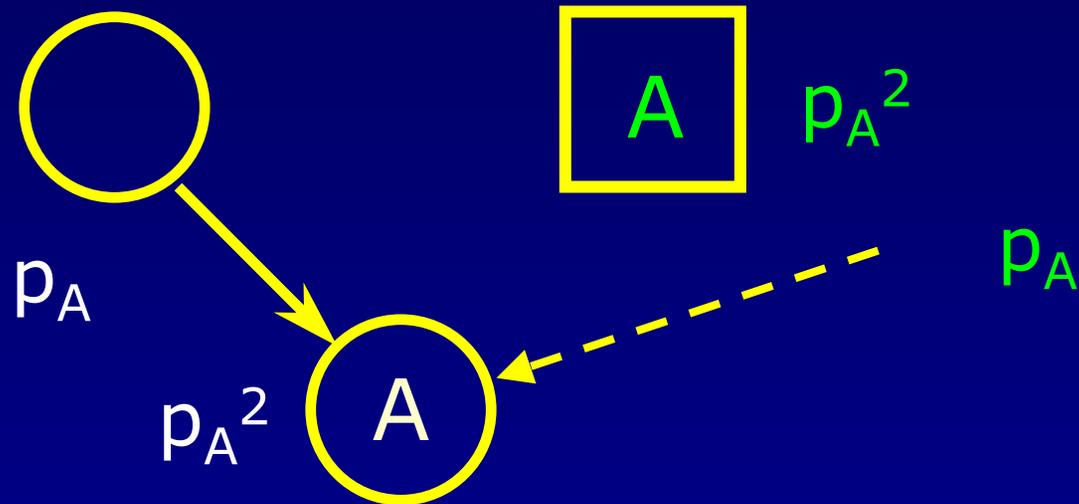


$$\text{Probability} = p_A^2 \times p_A^2 \times 1_{(fA)} \times p_A$$

Paternity Index

Only Man and Child Tested

Denominator



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

Paternity Index

Only Man and Child Tested

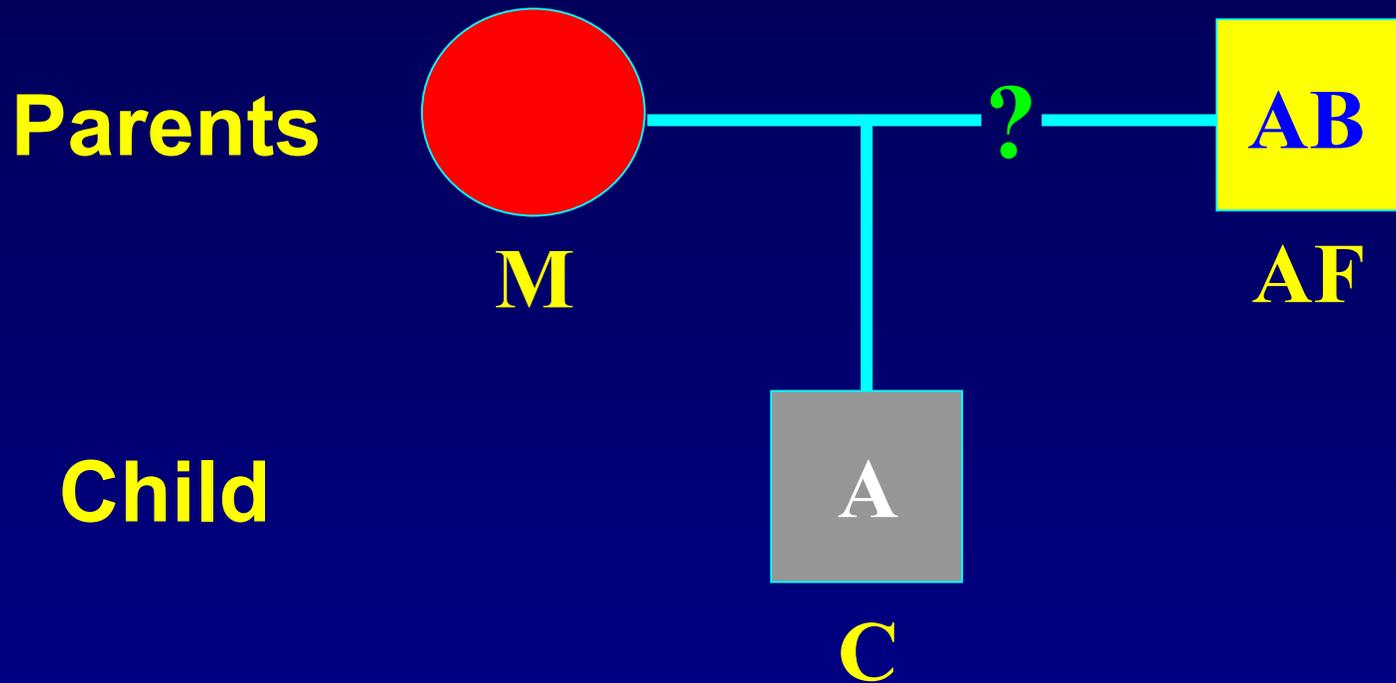
$$PI = \frac{\cancel{p_A^2} \times \cancel{p_A^2} \times 1_{(fA)} \times p_A}{\cancel{p_A^2} \times \cancel{p_A^2} \times p_{(mA)} \times p_{(fA)}}$$

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

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

Paternity Index

Only Man and Child Tested



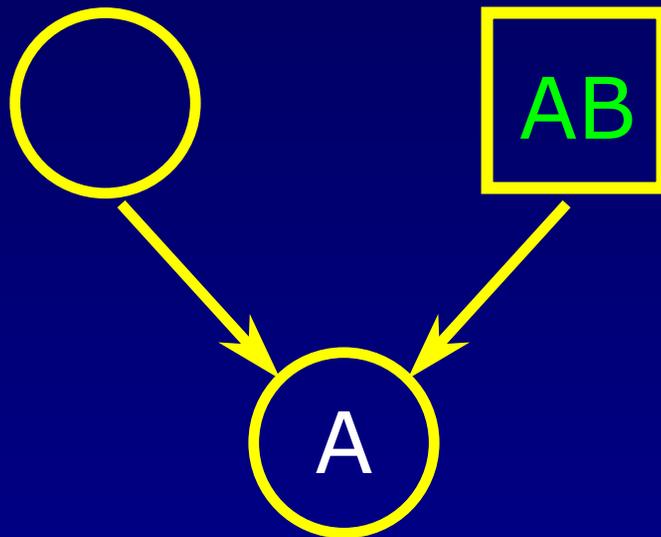
The untested Mother would have to pass an A allele

AF would have to pass the A allele

RM has p chance of passing the A allele

Paternity Index

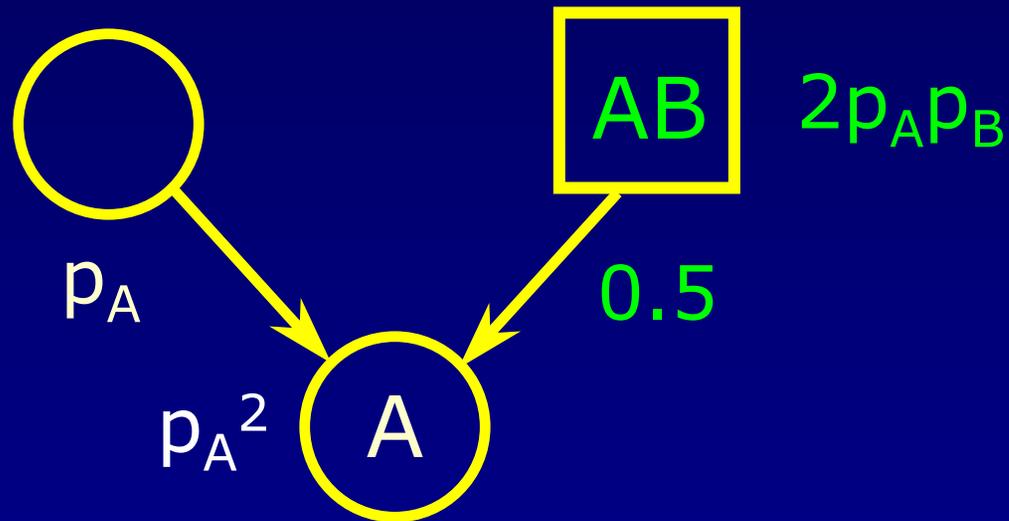
Only Man and Child Tested



Paternity Index

Only Man and Child Tested

Numerator

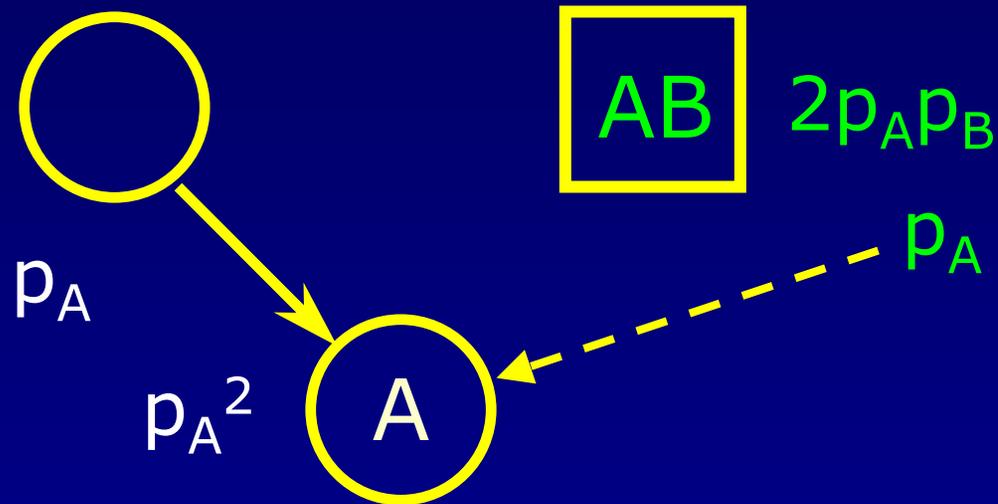


$$\text{Probability} = 2p_A p_B \times p_A^2 \times 0.5_{(fA)} \times p_A$$

Paternity Index

Only Man and Child Tested

Denominator



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

Paternity Index

Only Man and Child Tested

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

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

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

Paternity Index

Only Man and Child Tested

Formulas

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

<u>C</u>	<u>AF</u>	<u>Numerator</u>	<u>Denominator</u>	<u>PI</u>	<u>PE</u>
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 (5q33.3 - q34)	10 11	11 12	10 = 0.25269 11 = 0.30049
HUMTPOX (2p23 - 2pter)	8 11	8 11	8 = 0.54433 11 = 0.25369
HUMTH01 (11p15.5)	6 9.3	6 7	6 = 0.22660 9.3 = 0.30542
HUMvWA31 (12p13.3 - p13.2)	15 16	16	15 = 0.11224 16 = 0.20153

MOTHERLESS PATERNITY

CASE P-41376

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

MOTHERLESS PATERNITY

CASE P-41376

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

MOTHERLESS PATERNITY

CASE P-41376

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

MOTHERLESS PATERNITY

CASE P-41376

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

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

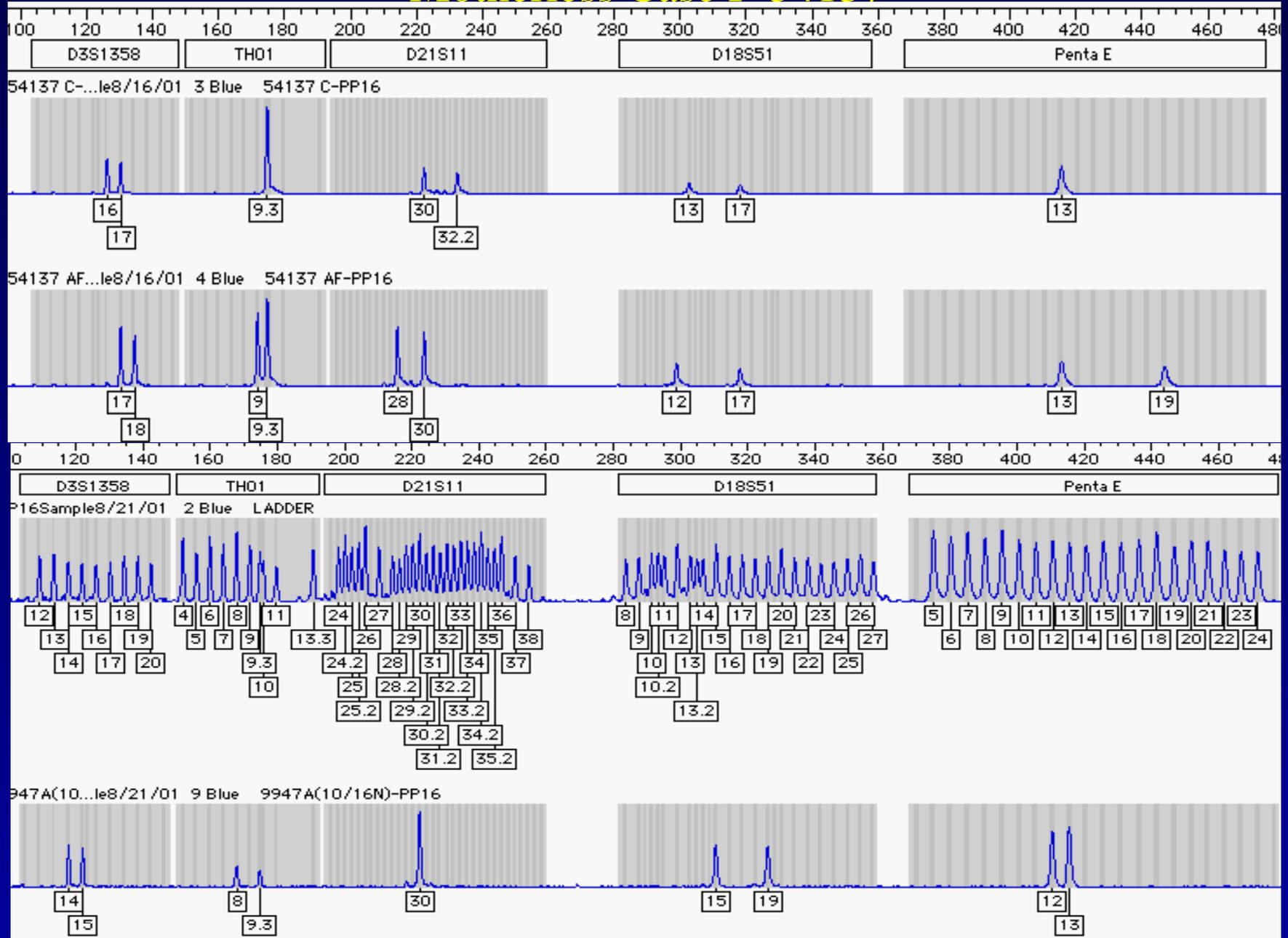
Combined Paternity Index	1,676
Probability of Paternity	99.94%
Probability of Exclusion	99.94%

PowerPlex™ 16 System

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

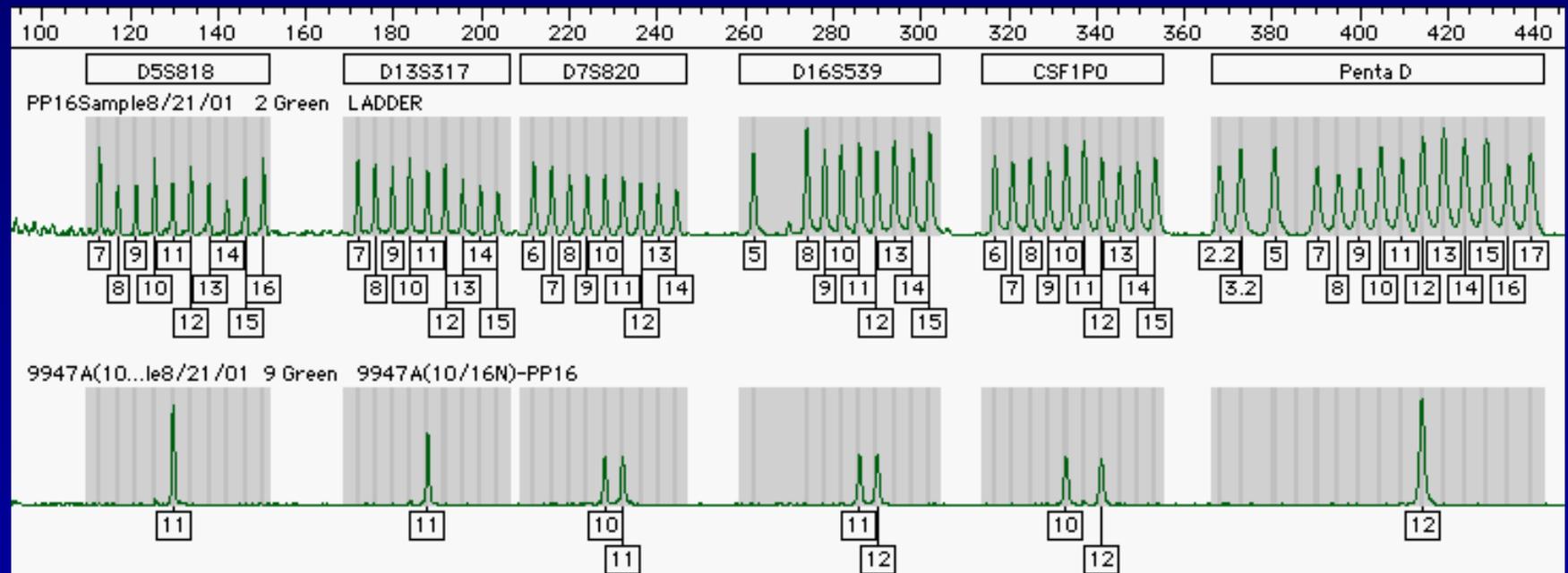
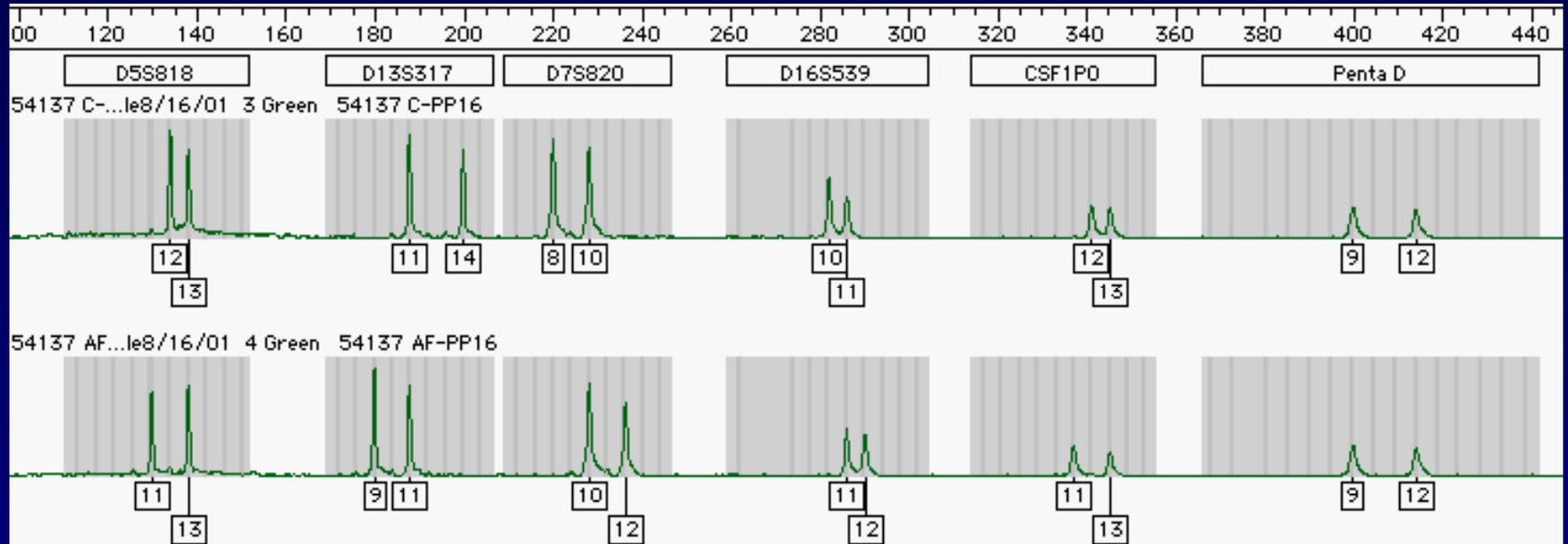
PowerPlex™ 16

Motherless Case P-54137

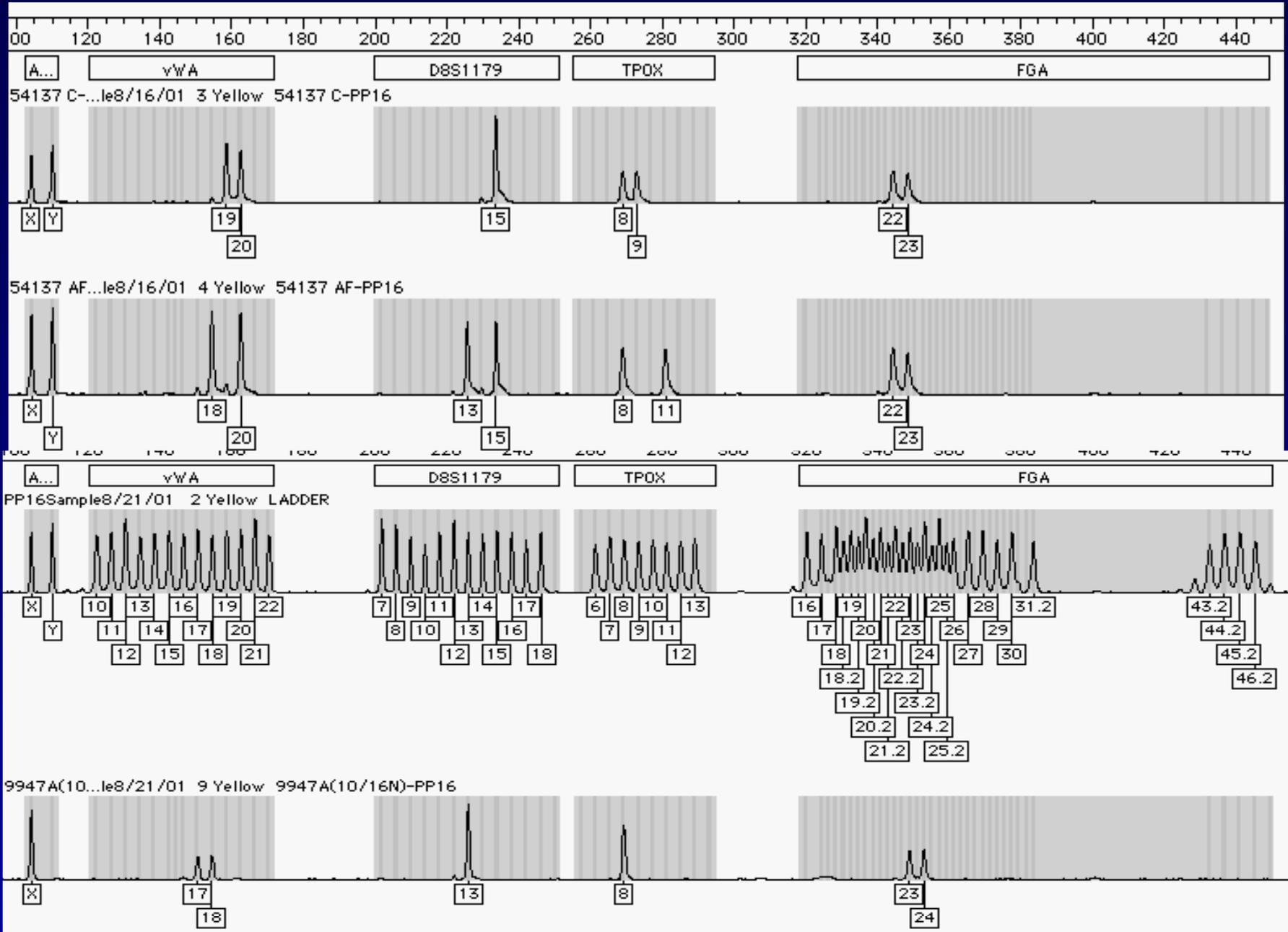


PowerPlex™ 16

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%

**Popstats Cannot Correctly
Calculate Parentage Statistics
in
Non-Typical Cases**

**Popstats Cannot Currently
Calculate 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
-------------------	----------	-------------------

— A

— B

— B

— C

— C

— D

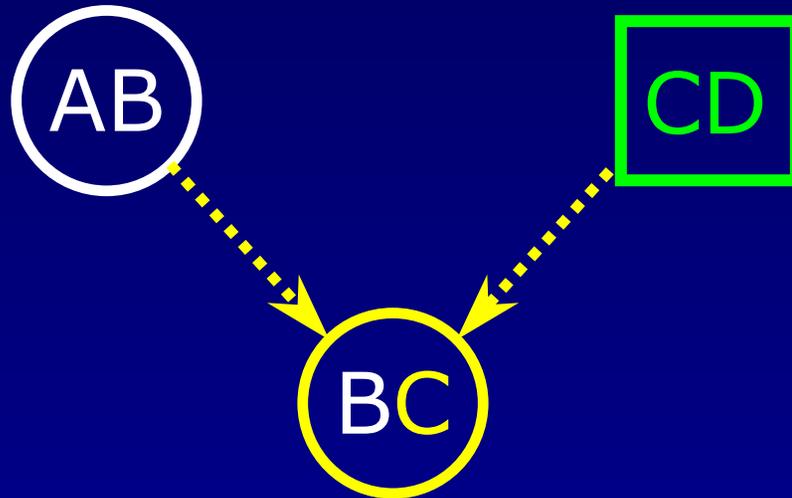
Reverse Parentage Testing

Three genotypes:

- Alleged Mother
- Child (missing)
- Alleged Father

Reverse Parentage Analysis

Missing child scenario



Reverse Parentage Index

$$\text{RPI} = \mathbf{X} / \mathbf{Y}$$

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

$$\text{RPI} = X / Y$$

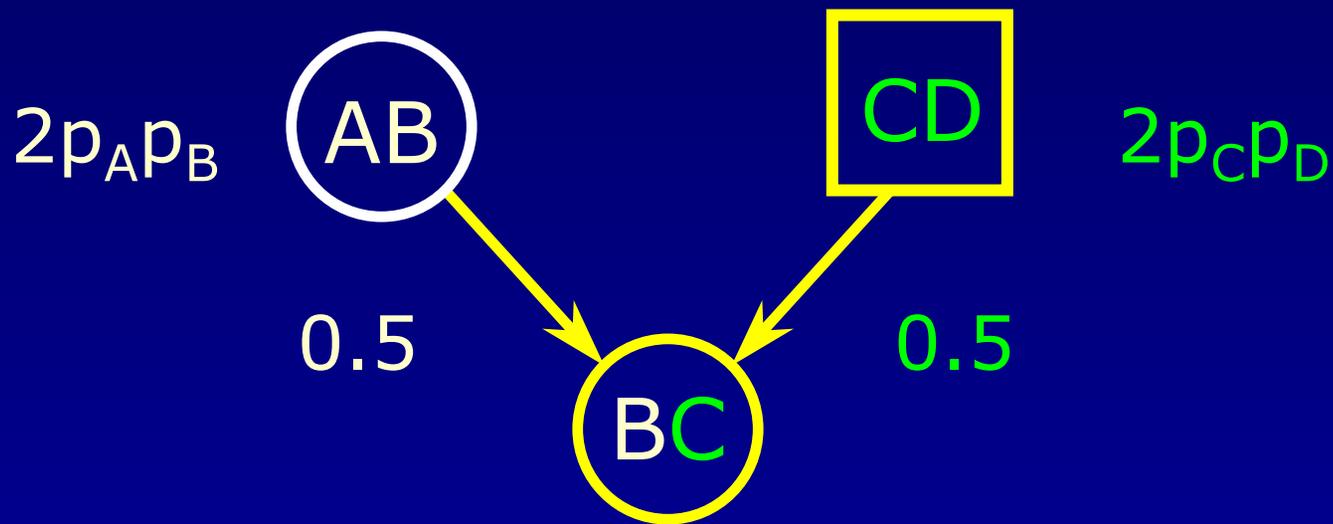
Denominator

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

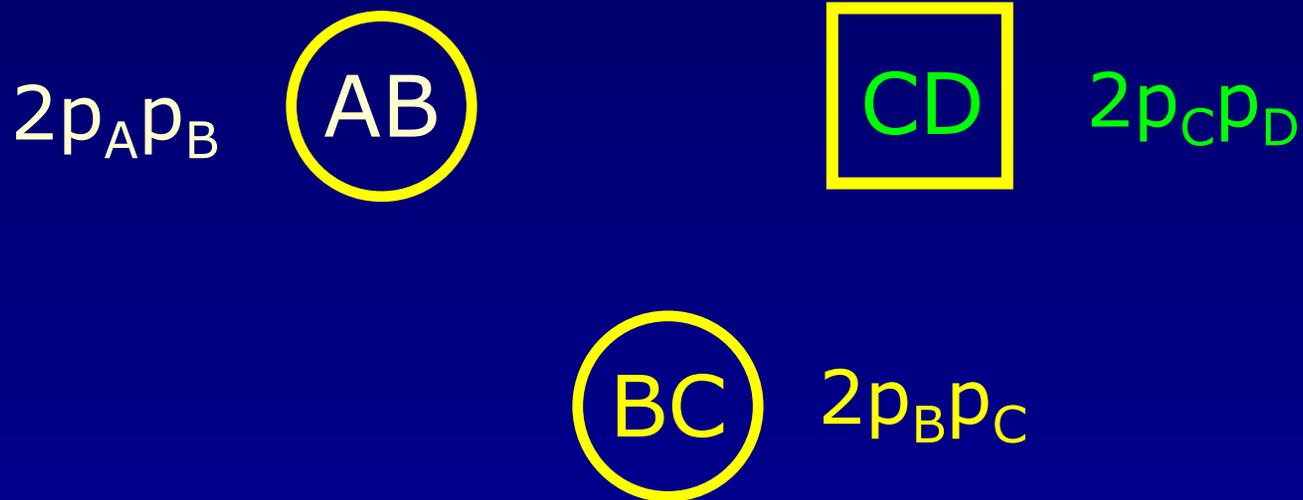


$$\text{Probability} = 2p_A p_B \times 2p_C p_D \times 0.5 \times 0.5$$

Reverse Parentage Analysis

Missing child scenario

Denominator



$$\text{Probability} = 2p_A p_B \times 2p_C p_D \times 2p_B p_C$$

Reverse Parentage Analysis

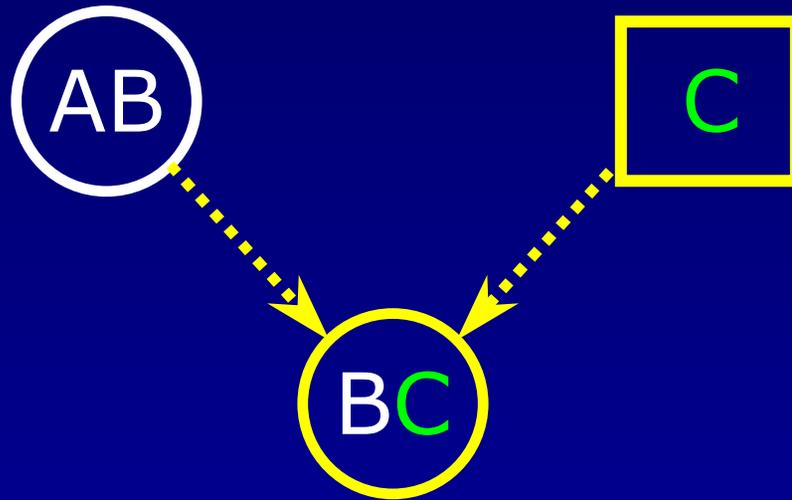
Missing child scenario

$$\text{LR} = \frac{\cancel{2p_{APB}} \times \cancel{2p_{CPD}} \times 0.5 \times 0.5}{\cancel{2p_{APB}} \times \cancel{2p_{CPD}} \times 2p_B p_C}$$

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

Reverse Parentage Analysis

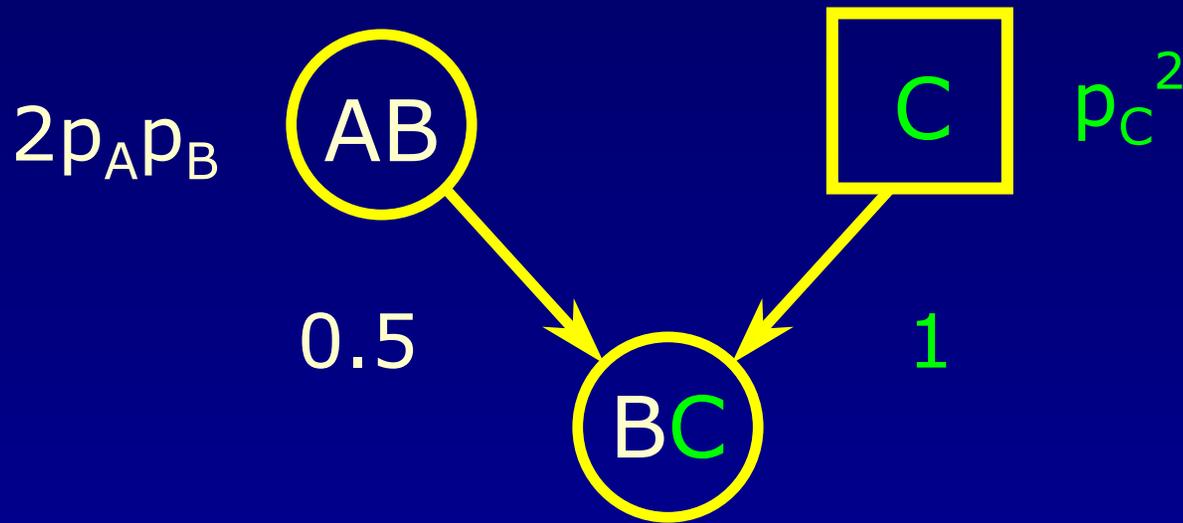
Missing child scenario



Reverse Parentage Analysis

Missing child scenario

Numerator



$$\text{Probability} = 2p_A p_B \times p_C^2 \times 0.5 \times 1$$

Reverse Parentage Analysis

Missing child scenario

Denominator

$$2p_A p_B \quad \textcircled{AB} \quad \square_C \quad p_C^2$$

$$\textcircled{BC} \quad 2p_B p_C$$

$$\text{Probability} = 2p_A p_B \times p_C^2 \times 2p_B p_C$$

Reverse Parentage Analysis

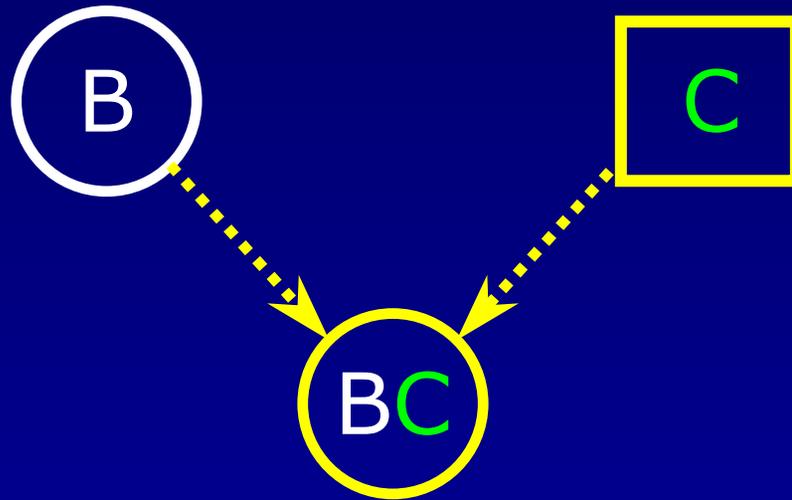
Missing child scenario

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

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

Reverse Parentage Analysis

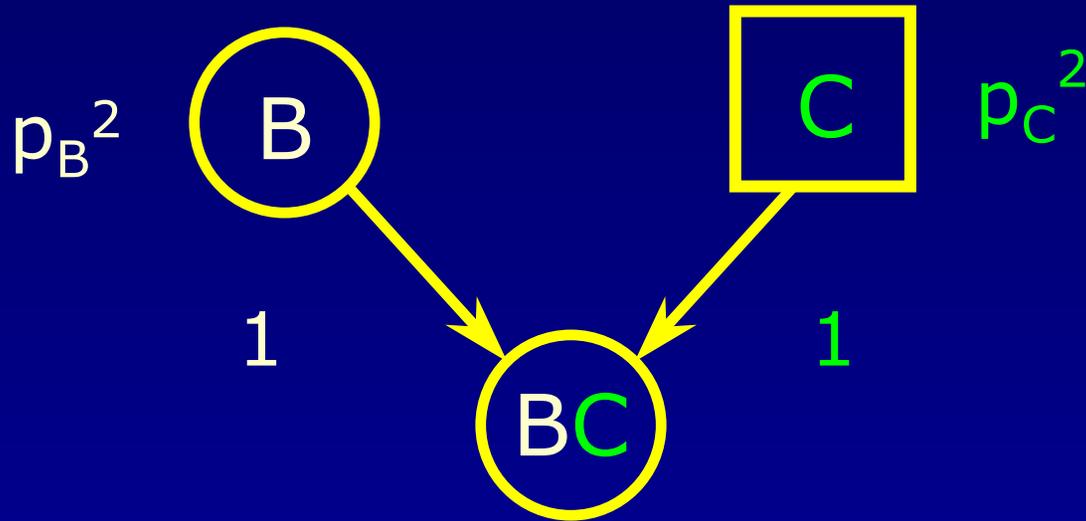
Missing child scenario



Reverse Parentage Analysis

Missing child scenario

Numerator



$$\text{Probability} = p_B^2 \times p_C^2 \times 1 \times 1$$

Reverse Parentage Analysis

Missing child scenario

Denominator

$$p_B^2 \quad \textcircled{B} \quad \square C \quad p_C^2$$

$$\textcircled{BC} \quad 2p_B p_C$$

$$\text{Probability} = p_B^2 \times p_C^2 \times 2p_B p_C$$

Reverse Parentage Analysis

Missing child scenario

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

$$\text{LR} = \frac{1}{2p_B p_C}$$

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!

