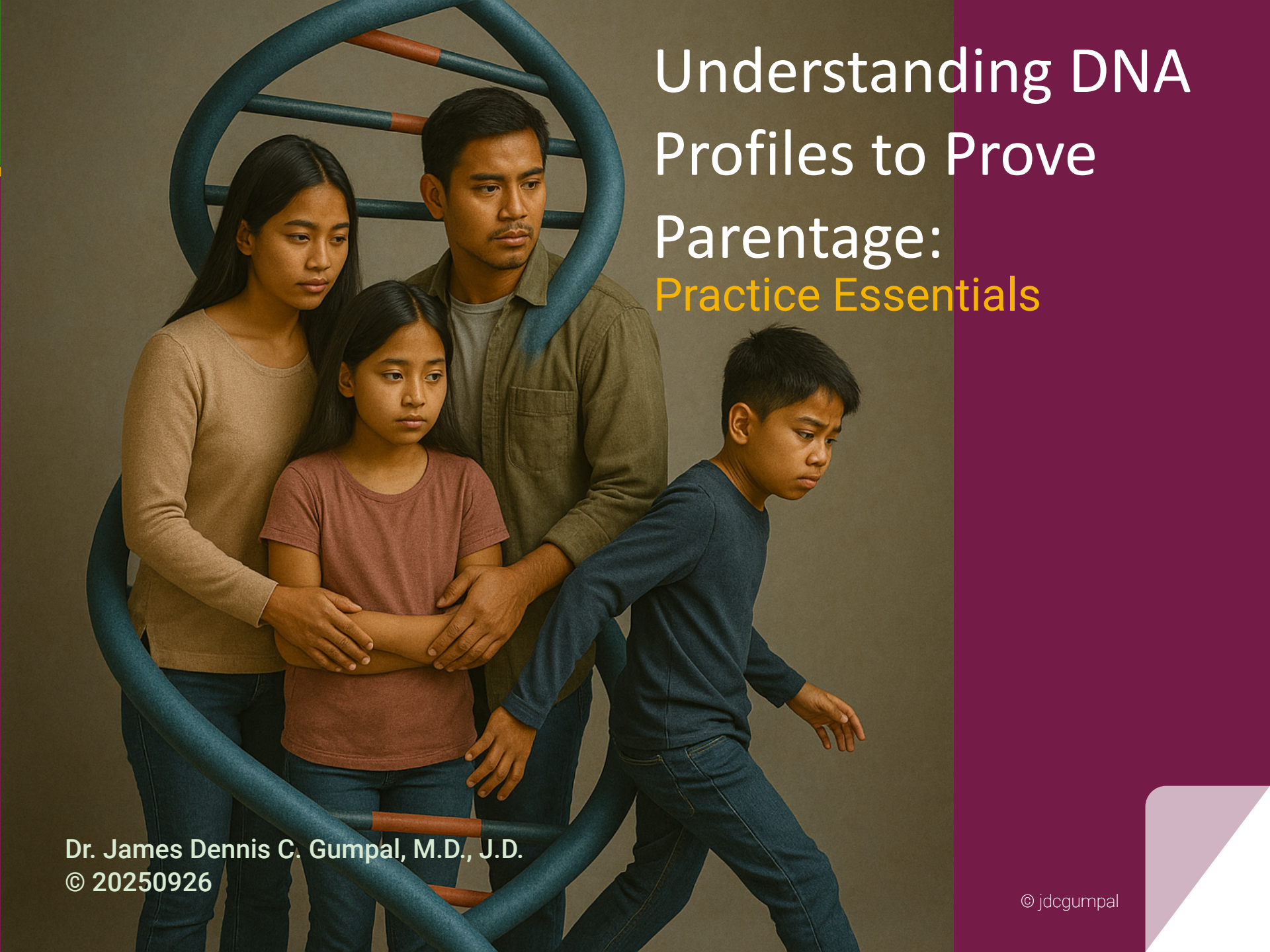


Understanding DNA Profiles to Prove Parentage: Practice Essentials



Dr. James Dennis C. Gumpal, M.D., J.D.
© 20250926

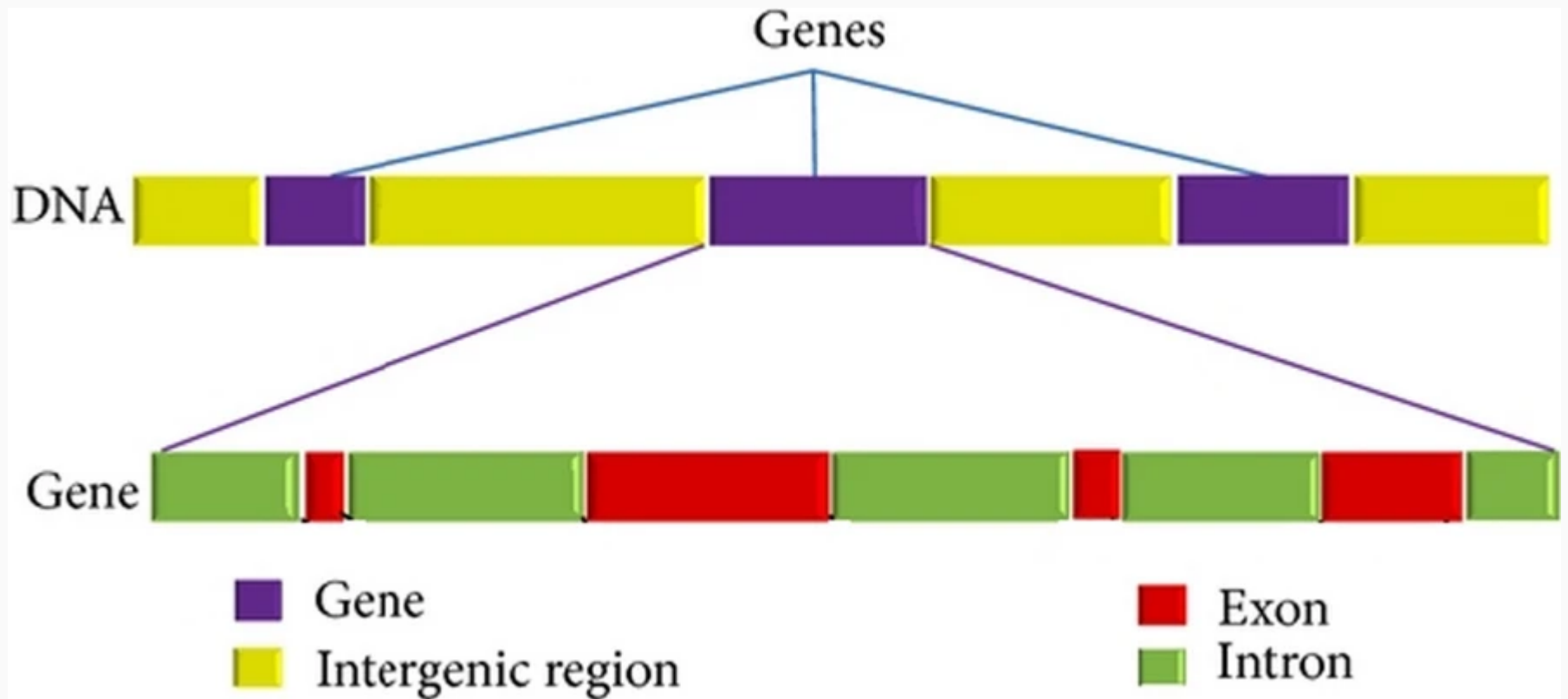
© jdcgumpal

DNA Profile Basics

- Genes
- Introns
- STRs
- Alleles
- Allele frequencies
- Paternity & Maternity Indices
- Probability of Paternity or Maternity



Introns - location of most STRs



Mother

Father



Short Tandem Repeats (STRs)

STRs = Short sequences of DNA (2–6 base pairs) repeated in tandem found in introns, which are highly variable among persons.

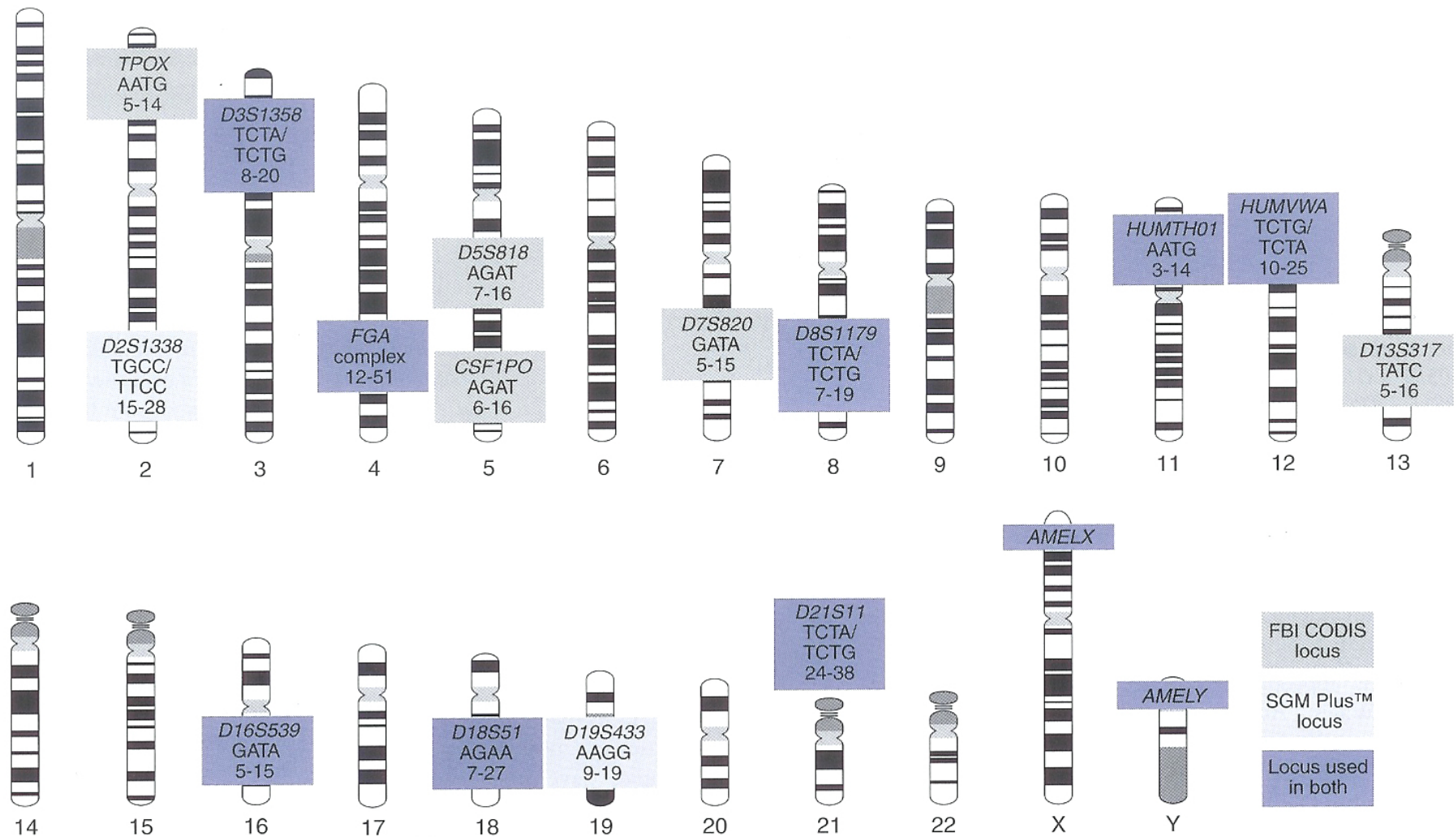
2 bp repeat = **dinucleotide** STR (e.g., **ATATATATATATATATAT...**)

3 bp repeat = **trinucleotide** STR (e.g., **CATCATCATCATCATCAT...**)

4 bp repeat = **tetranucleotide** STR (e.g., **GATAGATAGATAGATA...**)

most common in forensic DNA profiling

20 Loci CODIS + AMEL X and AMEL Y



CODIS STR Repeat Motifs, and Chromosome locations

Locus	Repeat Motif (STR)	Chromosome
CSF1PO	[AGAT]	5
D1S1656	[TAGA]/[TCTG])*	1
D2S441	[TAAA]/[TCTG]	2
D2S1338	[TACA]/[TCTG]	2
D3S318	[AGAT]	5
D5S817	[GATA	5
D10S248	[TCTA]/[TCTG]	7
D12S1349	[AGAT]	10
D13S391	[AGAT]/[TCTG]	12
D13S317	[TATC	13
D16S539	[GATA	16
D18S51	[AAGA]/[AAAG)	18
D21S11	[TCTA]/[TCTG]/ICA	21
FGA	[TTTC]/[CTTT]	4
TH01	[AATG	11
TPOX	[AATG	2
vWA	[TCTA]/[TCTG]	12
Amelogenin*	Sex-typing (X-Y insertion/deletion	X/Y

DNA Paternity Test



DNA Profile - Paternity

STR loci = specific places in the genome (complete set of DNA) where STRs (short tandem repeats) are found.

Alleles: numeric repeat counts at STR loci; the 2 numbers represent the STR inherited from a person's biological parents

Obligate paternal allele: the allele in a child's DNA profile that is not inherited from the mother and therefore must have come from the biological father.

Allele frequencies: the proportion of a specific allele in a given population showing how common that allele is compared to all others at the same genetic locus.

Locus	Child's Alleles	Mother's Alleles	Obligate paternal allele	Putative Father #1	Putative Father #2	Allele Frequencies
D3S1358	15, 18	15, 17	18	18, 19	12, 14	15 = 0.20; 17 = 0.25; 18 = 0.10; 19 = 0.08; 12 = 0.12; 14 = 0.15
vWA	16, 18	16, 16	18	17, 18	14, 15	16 = 0.18; 17 = 0.22; 18 = 0.12; 14 = 0.14; 15 = 0.09
FGA	22, 25	21, 22	25	24, 25	19, 20	21 = 0.11; 22 = 0.16; 24 = 0.10; 25 = 0.07; 19 = 0.14; 20 = 0.13
D7S820	8, 10	8, 9	10	10, 11	12, 13	8 = 0.19; 9 = 0.15; 10 = 0.12; 11 = 0.08; 12 = 0.13; 13 = 0.10
D8S1179	12, 14	12, 13	14	14, 15	9, 10	12 = 0.20; 13 = 0.14; 14 = 0.11; 15 = 0.09; 9 = 0.13; 10 = 0.10

Paternity Index (PI)

The **Paternity Index**, or **PI**, is a measure of how strongly the DNA evidence supports that a tested man is the biological father.

- It is computed using the formula $PI = 1/p$, where **p** represents the **allele frequency of the obligate paternal allele** — the allele in the child that came from the biological father and is not shared with the mother. (See column: PI of PF#1 = $1/p$)
- The PI for PF#2 in the table below cannot be mathematically computed because he is already genetically excluded as a father.

Locus	Child	Mother	Oblig. Paternal Allele	Putative Father #1 (PF#1)	Putative Father #2 (PF#2)	Frequency (p) PF#1	Frequency (p) PF#2	PI= 1/p (PF#1)	"no match no PI" (PF#2)
D3S1358	15, 18	15, 17	18	18, 19	12, 14	0.10	0	$1/0.10=10$	0 (excluded)
vWA	16, 18	16 16	18	17, 18	14, 15	0.12	0	$1/0.12=8.33$	0 (excluded)
FGA	22, 25	21, 22	25	24, 25	19, 20	0.07	0	$1/0.07=14.29$	0 (excluded)
D7S820	8, 10	8, 9	10	10, 11	12, 13	0.12	0	$1/0.12=8.33$	0 (excluded)
D8S1179	12, 14	12, 13	14	14, 15	9, 10	0.11	0	$1/0.11=9.09$	0 (excluded)

Combined Paternity Index

Combined Paternity Index (CPI) - multiply all PIs

PF#1: $10.00 \times 8.33 \times 14.29 \times 8.33 \times 9.09 \approx 90,187.59$

PF#2: product includes zeros $\rightarrow 0$ (excluded)

This means the putative father #1 is 90,187.59 times more likely to be the biological father of the child compared to any random person.

Probability of Paternity

Probability of Paternity (POP) (prior = 0.5):

$$\begin{aligned}\text{PF\#1: POP} &= \text{CPI}/(\text{CPI}+1) \\ &= 90,187.59/90,187.59+1 \\ &= 0.9999823703 \\ &= 99.9989\%\end{aligned}$$

PF#2: 0% (excluded)

This means that the putative father #1 is 99.9989% is most likely the biological father of the child, which under the Rule on DNA Evidence, creates a disputable presumption of paternity.



Maternity Test

© jdcgumpal

Maternity Index

Maternity Index (MI) —measures how likely a woman is to be the biological mother of a child compared to an unrelated woman.

Obligate maternal allele: the allele in a child's DNA profile that is not inherited from the father and therefore must have come from the biological mother.

- A higher MI value means stronger genetic evidence that she is the true mother.
- It is conceptually similar to the **Paternity Index (PI)** but instead evaluates maternity.

STR Locus	Child's alleles	Putative Mother #1	Obligate maternal allele	Putative Mother #2	Allele frequencies used (p)	p(obligate)	MI (PM#2 = 1/p)	MI (PM#1)
D3S1358	15, 18	12,14	18	16, 18	12:0.12; 14:0.15; 15:0.20; 18:0.10	0.10	10.000	0 (excluded)
vWA	16 ,18	14,15	16	16 ,16	14:0.14; 15:0.09; 16:0.18; 18:0.12	0.18	5.556	0 (excluded)
FGA	22, 25	19,20	25	24, 25	19:0.14; 20:0.13; 24:0.10; 22:0.16; 25:0.07	0.07	14.286	0 (excluded)
D7S820	8,10	12,13	8	8,9	8:0.19; 9:0.15; 10:0.12; 12:0.13; 13:0.10	0.19	5.263	0 (excluded)
D8S1179	12,14	9,10	12	12,13	9:0.13; 10:0.10; 12:0.20; 14:0.11; 13:0.14	0.20	5.000	0 (excluded)

Combined Maternity Index (CMI)

Combined Maternity Index (CMI) - multiply all MIs

PM#1 CMI = product of zeros = 0 (excluded)

PM#2 CMI = $10.000 \times 5.556 \times 14.286 \times 5.263 \times 5.000$
 $\approx 20,887$

This means that the putative mother #2 is 20,887 more likely to be the biological mother of the child.

Probability of Maternity (PoM)

Probability of Maternity (PoM)

(assuming the standard neutral prior = 0.5)

$$\text{PoM} = \text{CMI} / (\text{CMI} + 1)$$

$$\text{PM\#2 PoM} = 20,887 / (20,887 + 1) \approx 0.999952 \rightarrow \mathbf{99.995\%}$$

$$\text{PM\#1 PoM} = 0 / (0 + 1) = 0 \rightarrow 0\% \text{ (genetic exclusion)}$$

PoM Value	Interpretation	Meaning in Words
> 99.9%	Practically proven maternity	The DNA evidence overwhelmingly supports that the tested woman is the biological mother.
90–99%	Strong support for maternity	Highly likely that she is the biological mother.
50–89%	Moderate support	Some genetic similarity, but not conclusive — more testing or additional relatives needed.
≈ 50% (0.5)	Inconclusive	DNA evidence does not favor either side (equal likelihood).
< 50%	Evidence against maternity	DNA is more consistent with the tested woman being unrelated

This means that the DNA evidence overwhelmingly supports that the tested woman is the biological mother of the child.

References

- Butler, J. M. (2015). Advanced topics in forensic DNA typing: Interpretation. Academic Press.
→ Comprehensive discussion of STR loci, allele frequencies, and kinship index calculations.
- Butler, J. M. (2012). Forensic DNA typing: Biology, technology, and genetics of STR markers (2nd ed.). Elsevier Academic Press.
→ Explains STR markers, allele sharing, and computation of paternity and siblingship indices.
- Evett, I. W., & Weir, B. S. (1998). Interpreting DNA evidence: Statistical genetics for forensic scientists. Sinauer Associates.
→ Foundational work on likelihood ratios, kinship probabilities, and population genetics principles.
- Jobling, M. A., & Gill, P. (2004). Encoded evidence: DNA in forensic analysis. *Nature Reviews Genetics*, 5(10), 739–751. <https://doi.org/10.1038/nrg1455>
→ Overview of DNA profiling and its use in relationship testing and criminal forensics.
- National Research Council (U.S.) Committee on DNA Forensic Science. (1996). The evaluation of forensic DNA evidence. National Academies Press.
→ Establishes statistical and legal standards for interpreting DNA evidence.
- Tagliaro, F., & Bortolotti, F. (2021). DNA analysis in kinship testing. In A. J. Schütz (Ed.), *Forensic science and human identification* (pp. 101–124). Springer. https://doi.org/10.1007/978-3-030-60171-1_5
→ Discusses statistical interpretation of kinship tests, including full and half-siblingship indices.
- Supreme Court of the Philippines. (2007). A.M. No. 06-11-5-SC: Rule on DNA Evidence. Retrieved from <https://sc.judiciary.gov.ph>
→ Provides the legal framework for admitting and interpreting DNA evidence in Philippine courts, including the concept of “disputable presumption of paternity.”
- Gill, P., Fereday, L., Morling, N., & Schneider, P. M. (2006). The evolution of DNA databases—recommendations for new European STR loci. *Forensic Science International*, 156(2–3), 242–244. <https://doi.org/10.1016/j.forsciint.2005.11.016>
→ Explains STR locus selection and validation for kinship and identification testing.
- Pereira, R., et al. (2012). Evaluating forensic DNA kinship analysis methods. *Forensic Science International: Genetics*, 6(4), 469–475. <https://doi.org/10.1016/j.fsigen.2011.10.009>
→ Reviews statistical methods for computing and interpreting kinship likelihood ratios.