Le impronte genetiche: un testimone silenzioso contro l'assassino

Mimmo Turano
- PLS -
Napoli 10 settembre 2021

IL PROFILO GENETICO INDIVIDUALE O GENOTIPO

Indagini di filiazione

Profilo genetico del figlio

Profilo genetico del padre

Profilo genetico della madre

Investigazioni criminali

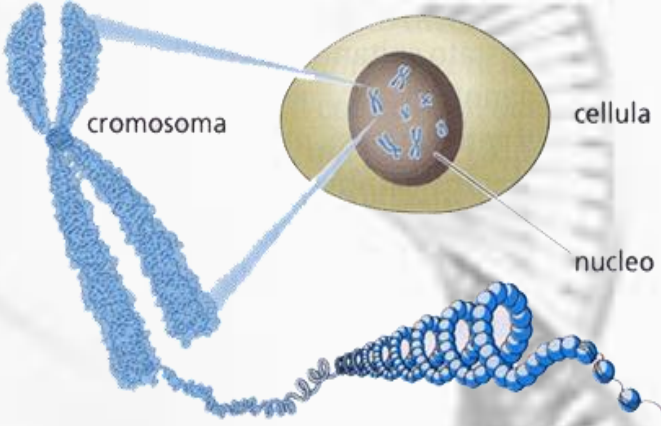
Profilo genetico da Tracce biologiche

Profilo genetico di banche dati

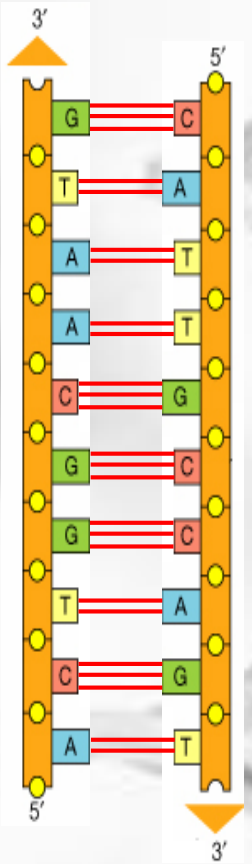
Profilo genetico di indagati

IL DNA

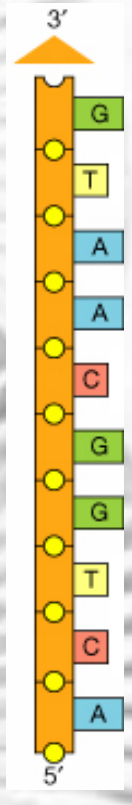
CCGGGCGACTCGGGGAACGGGGGTGGGGGGATGG
CGGCGTGTCGGCCTCTTCACCGCCCGGCCTTAAG
GCGCGAGAGTGGGAGCCCCTCGCGGTGACTTGGA
GGGTCGCCGTCGAAAGACATCTGCCGTGCTGCCC
TGCTGCCAGCCCGGACGCCGCCGTTACAGCTCCA
AGAAGAAAAGGAGCGGAGGGACCCCGGAGCAGGG



TACAGACAGAAGAAGCTT.....
ATGTCTGTCTTCTTCGAA.....

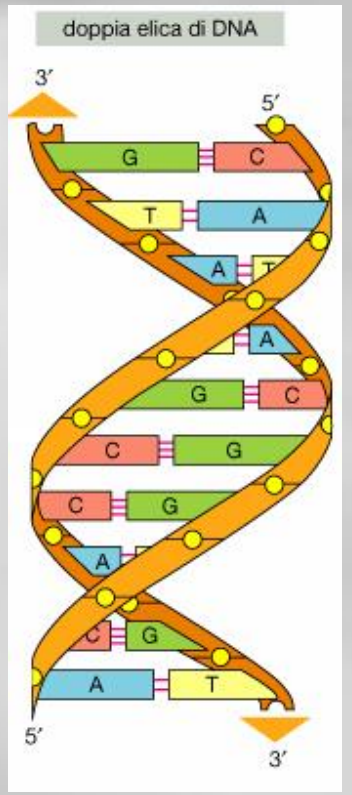


Doppio filamento di DNA

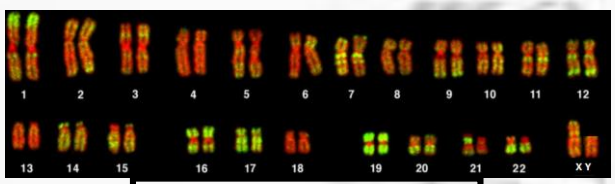


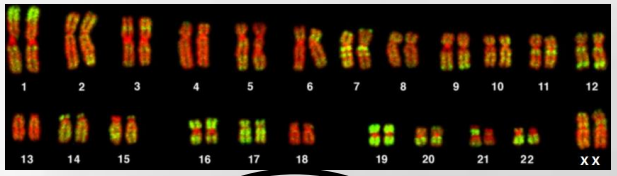
Singolo filamento di DNA

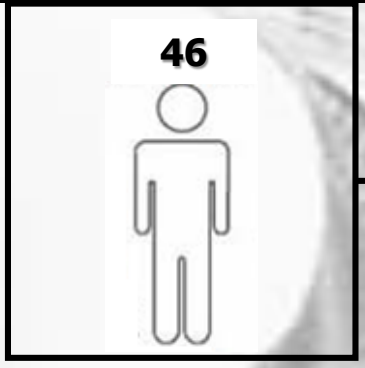
Nucleotidi



I CROMOSOMI


1 2 3 4 5 6 7 8 9 10 11 12
13 14 15 16 17 18 19 20 21 22 XY


1 2 3 4 5 6 7 8 9 10 11 12
13 14 15 16 17 18 19 20 21 22 XX


46


46


23
23


23 23

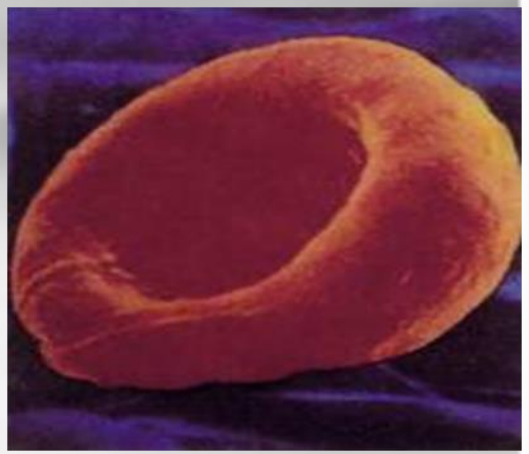

23 23

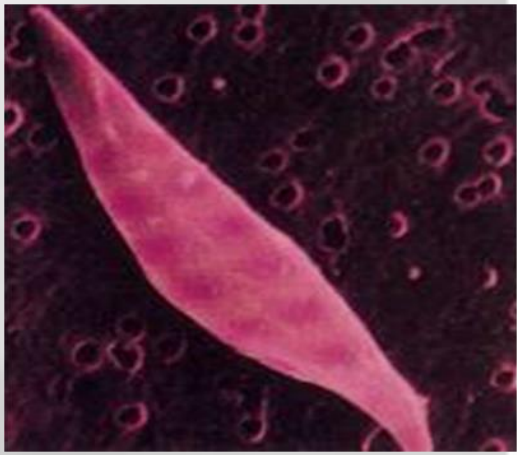
LA VARIABILITA' GENETICA INDIVIDUALE

MUTAZIONE

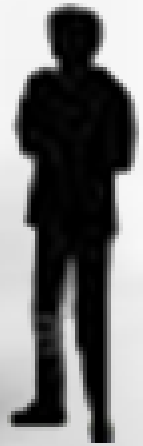
…CGAATGAGCGACG…

…CGAATGGGCGACG…





I MICROSATELLITI O SHORT TANDEM REPEAT (STR)

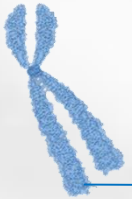


Cromosoma paterno

ATCGAAGTTCCCGAAATTAGC**GATGATGATGAT**CGAATGAGCGACGTTGGC…

ALLELE 4

Cromosoma materno



ATCGAAGTTCCCGAAATTAGC**GATGATGATGATGATGAT** CGAATGAGCGA…

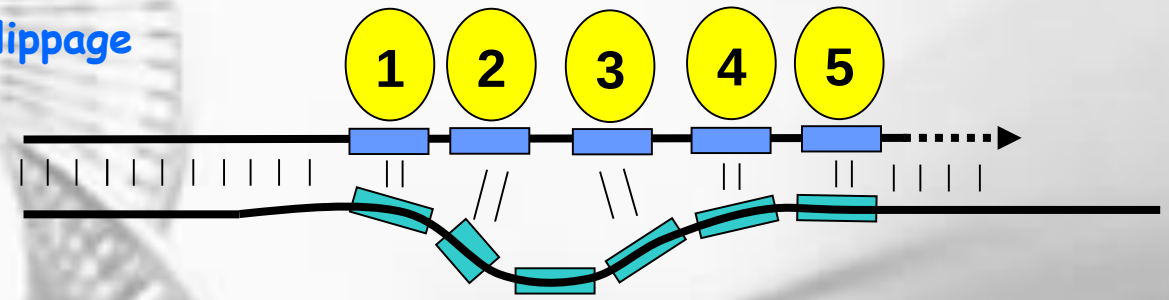
ALLELE 6

locus

GENOTIPO 4,6

STR

Slippage



Allele 3

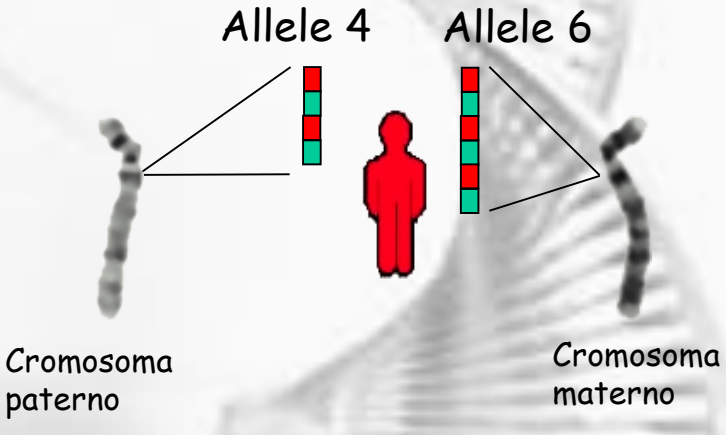
Allele 4

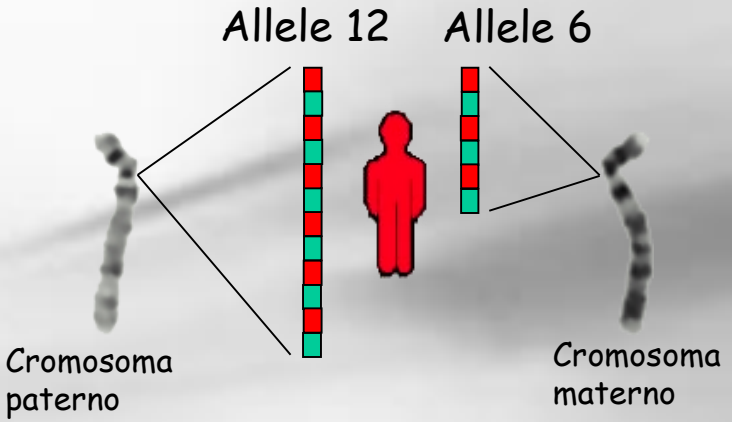
Allele 80

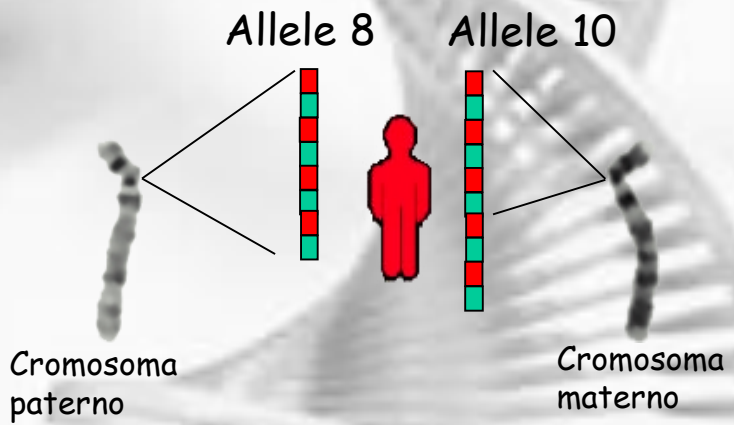
Genotipi del locus FGA:
80 alleli, 3240 genotipi

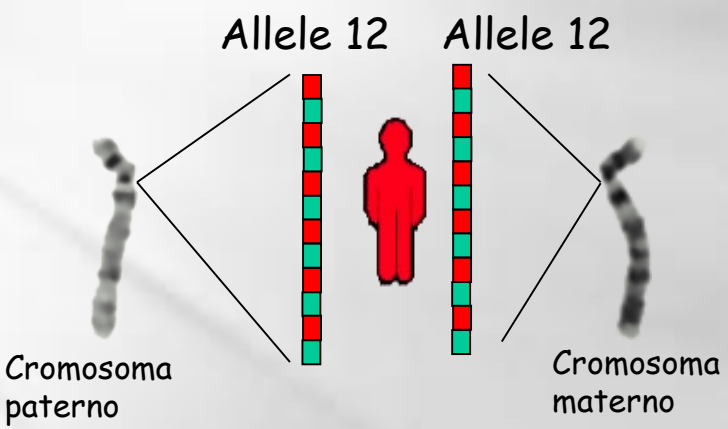
GENOTIPI:
n (n+1)/2

3240 GENOTIPI



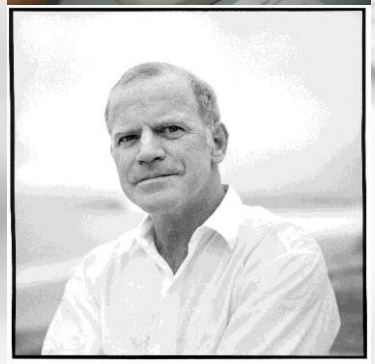




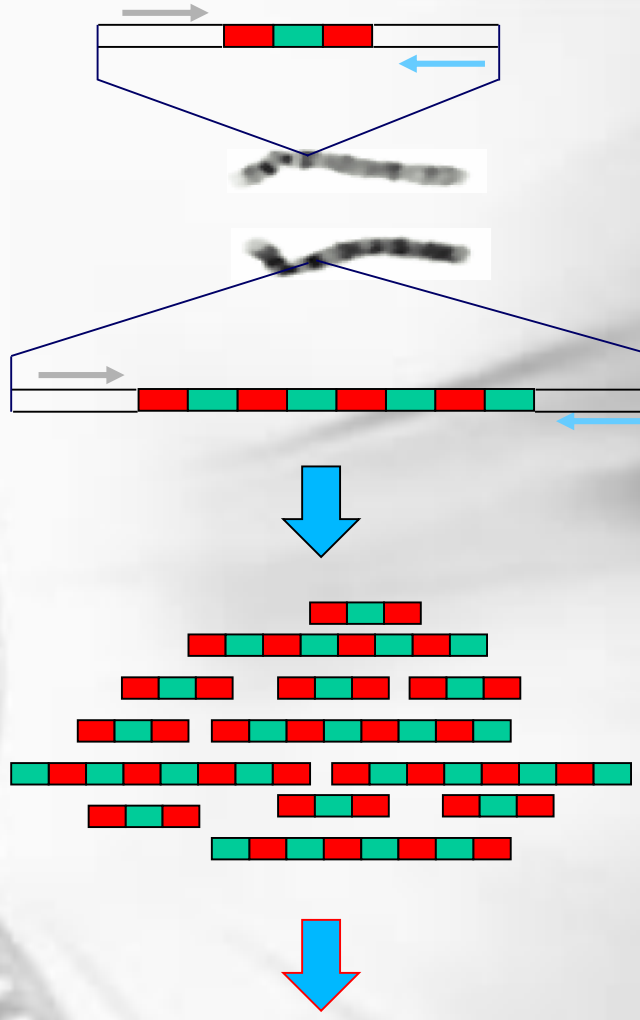


E COSI' VIA…….

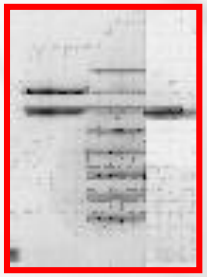
PCR DI UN SINGOLO LOCUS STR



Kary Mullis

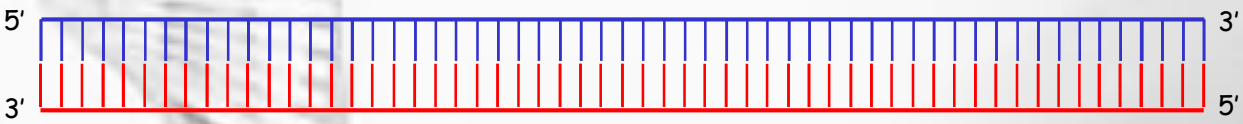


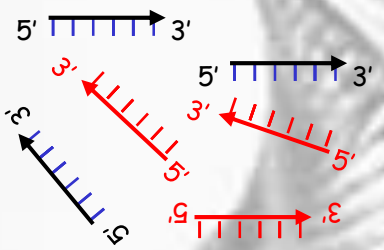
Eterozigoti :
I due alleli hanno lunghezza diversa



Omozigoti:
entrambi gli alleli hanno la stessa lunghezza

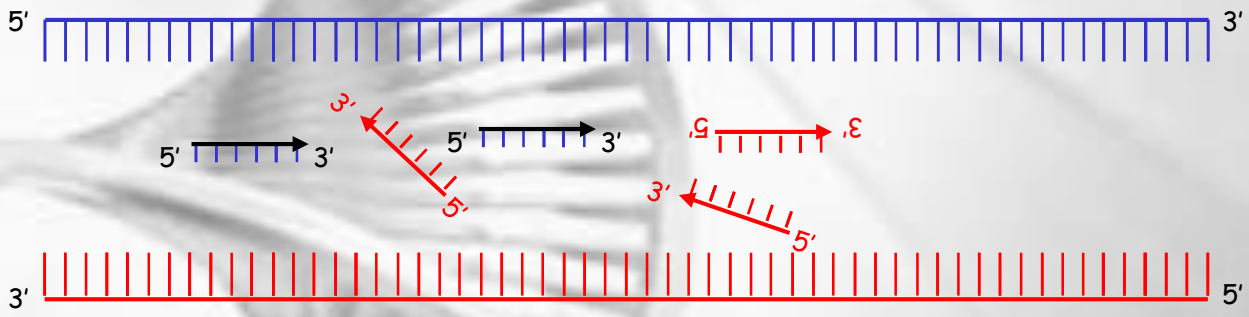
POLYMERASE CHAIN REACTION (PCR)





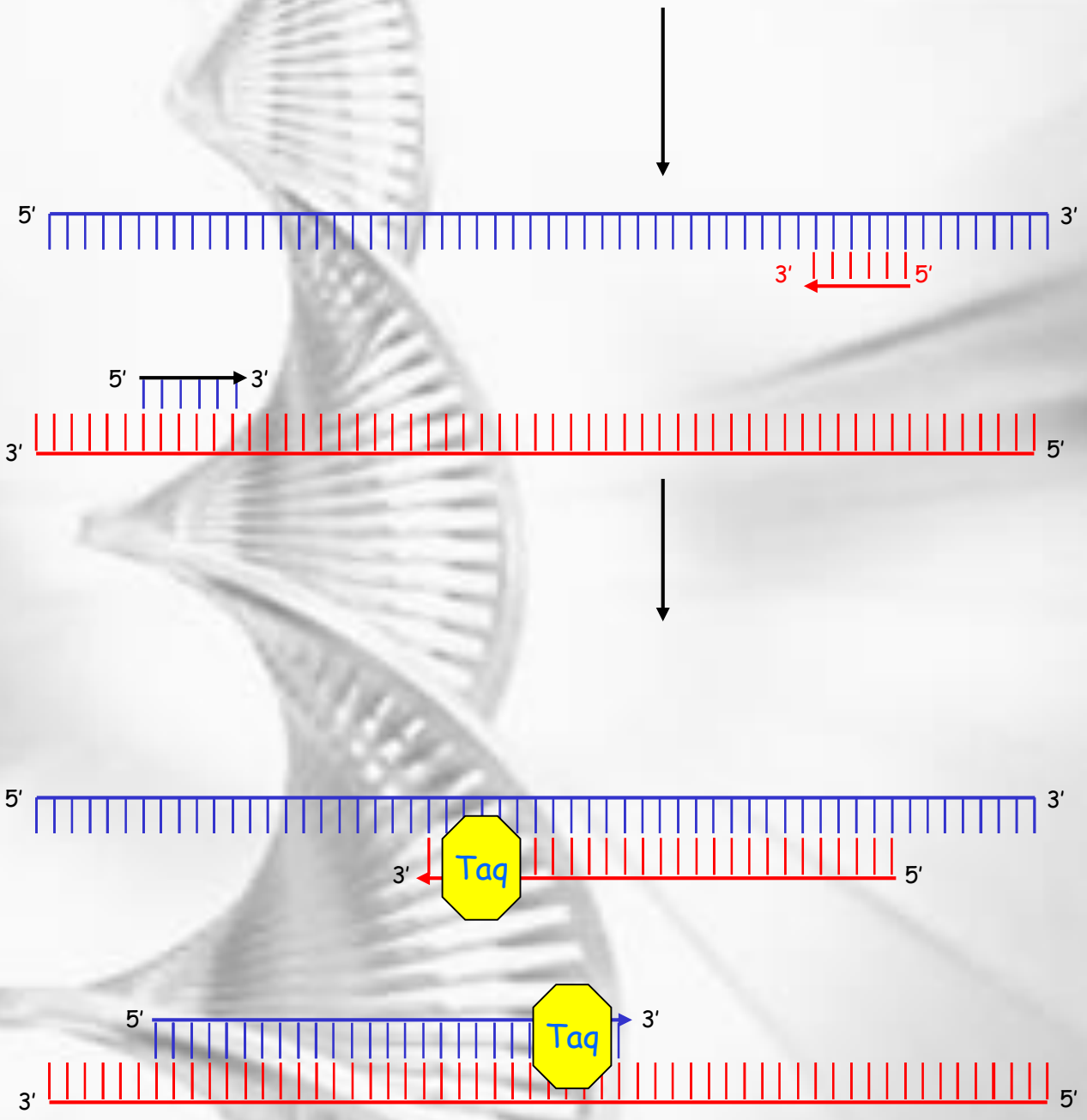
- Primers
- dNTPs (dATP, dGTP, dTTP, dCTP)
- Template (DNA o cDNA)
- Buffer
- Thermal Stable DNA Polymerase (Taq)

UNIRE I COMPONENTI NEL TUBINO DA PCR



DENATURATION

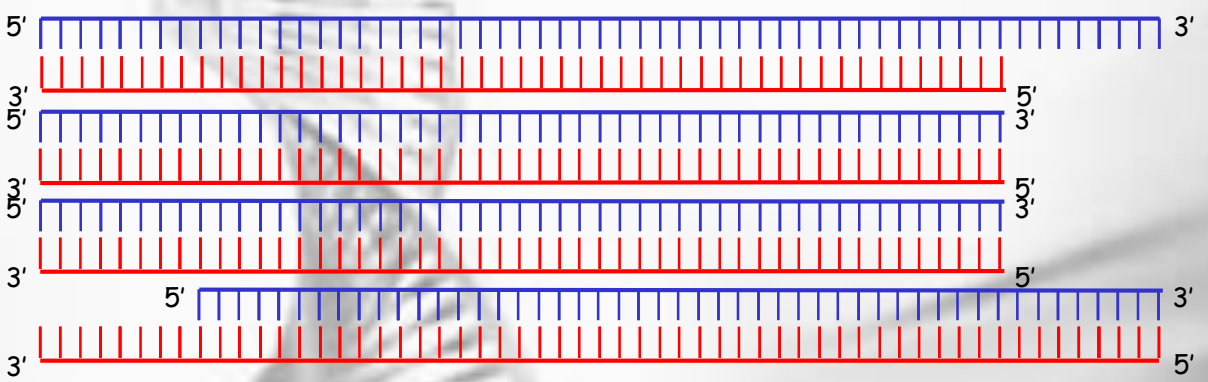
95 °C



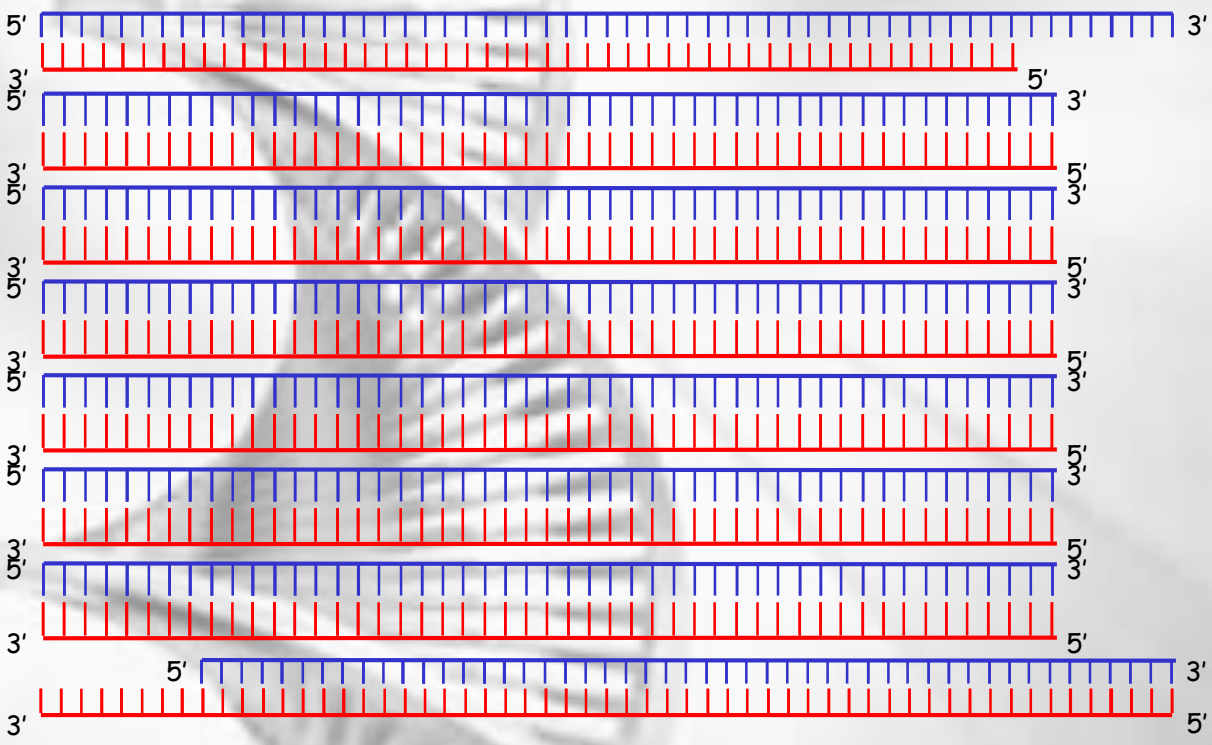
ANNEALING
VARIABLE TEMP.

EXTENSION
72 °C

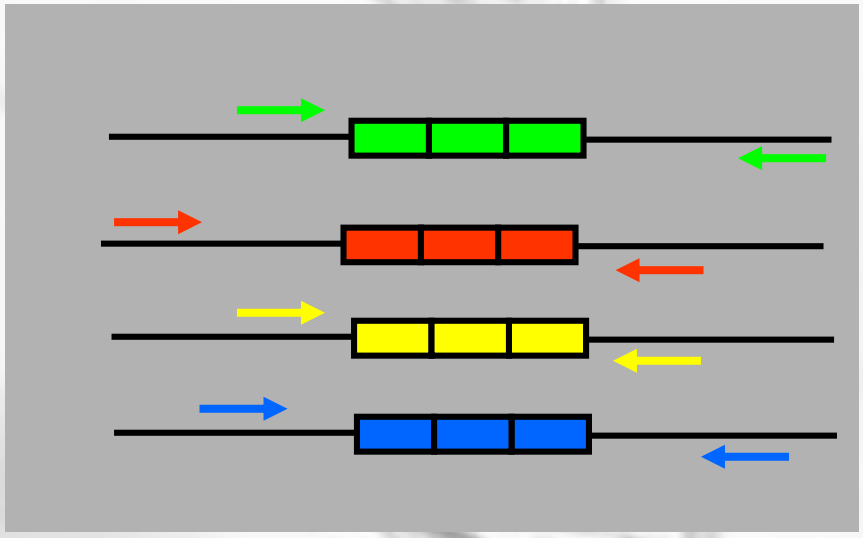
Ciclo 2 → 4 molecole

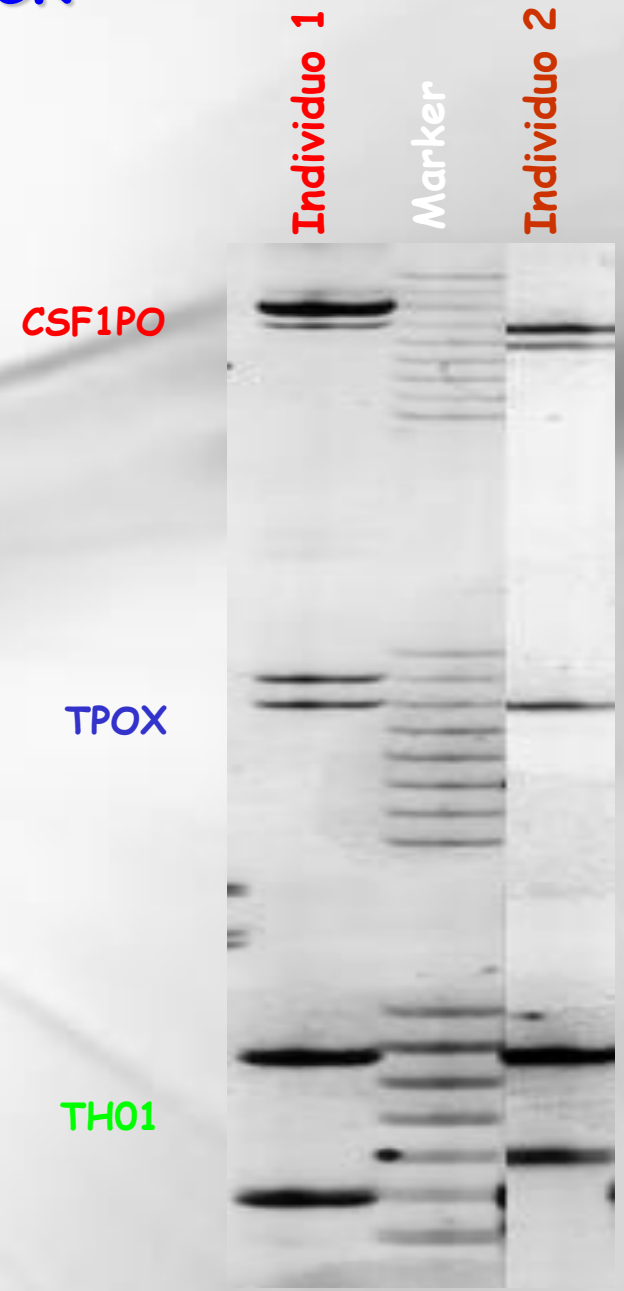


Ciclo 3 → 8 molecole



MULTIPLEX PCR



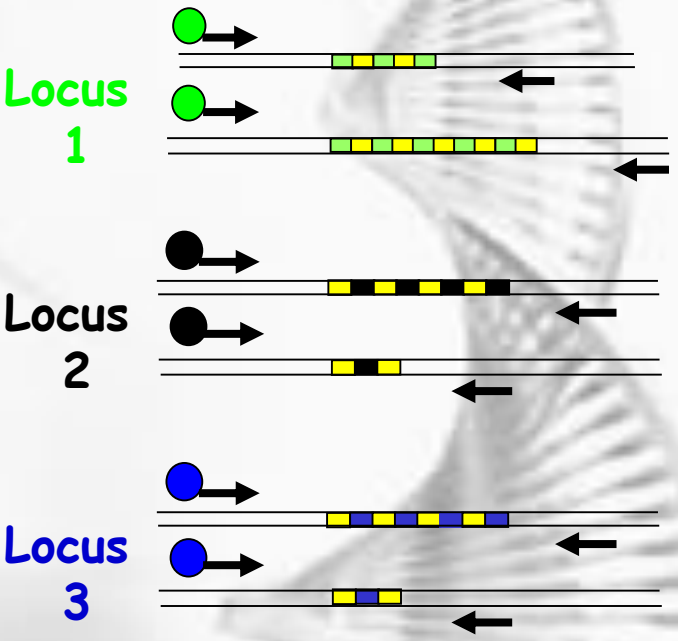


IL SISTEMA CODIS (1996-1997)
COmbined DNA Index System

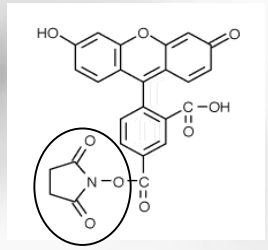
LOCUS	Sequence	chromosome position	GenBank accession	Alleles
TPOX	gaat	2p25.3	M68651	15
D3S1358	[tctg][tcta]	3p21.3	NT_00597	24
FGA	cttt	4q31.3	M64982	80
D5S818	agat	5q23.2	G08446	15
CSF1PO	taga	5q33.1	X14720	20
D7S820	gata	7q21.11	G08616	30
D8S1179	[tcta][tctg]	8q24.13	G08710	17
TH01	tcat	11p15.5	D00269	20
vWA	[tctg][tcta]	12.p13.31	M258S8	28
D13S317	tatc	13q31.1	G09017	17
D16S539	gata	16q24.1	G07925	19
D18S51	agaa	18q21.33	L18333	51
D21S11	[tcta][tctg]	21q21.1	AP000433	82

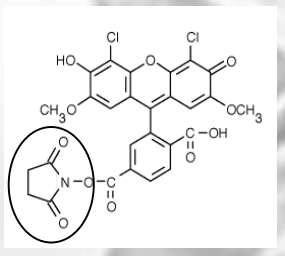
Random match probability = $1/10^{12}$

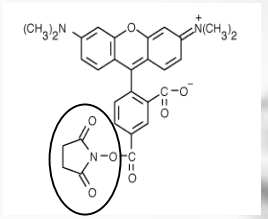
ELETTROFORESI CAPILLARE

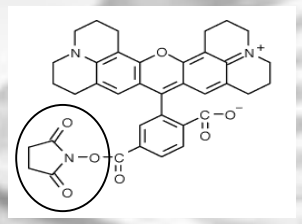


FAM (Blue) JOE (Green)

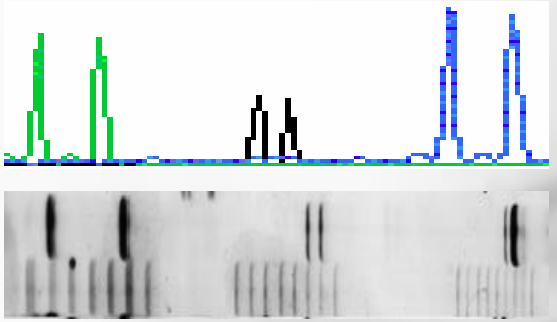


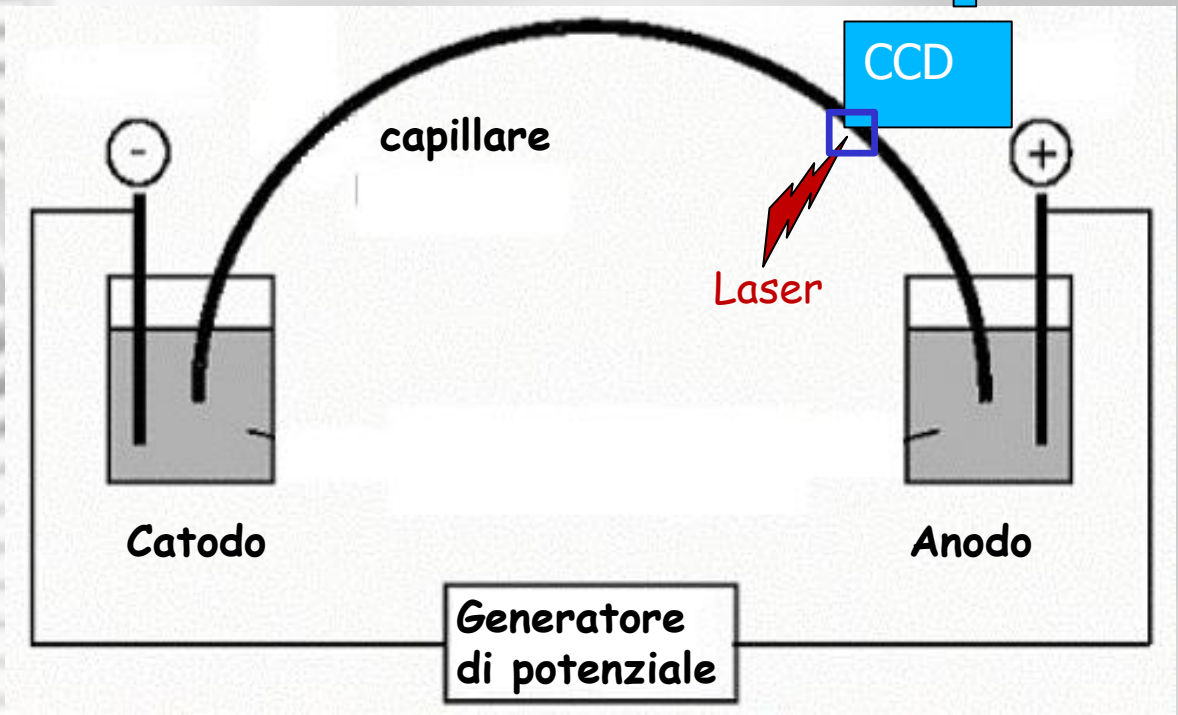




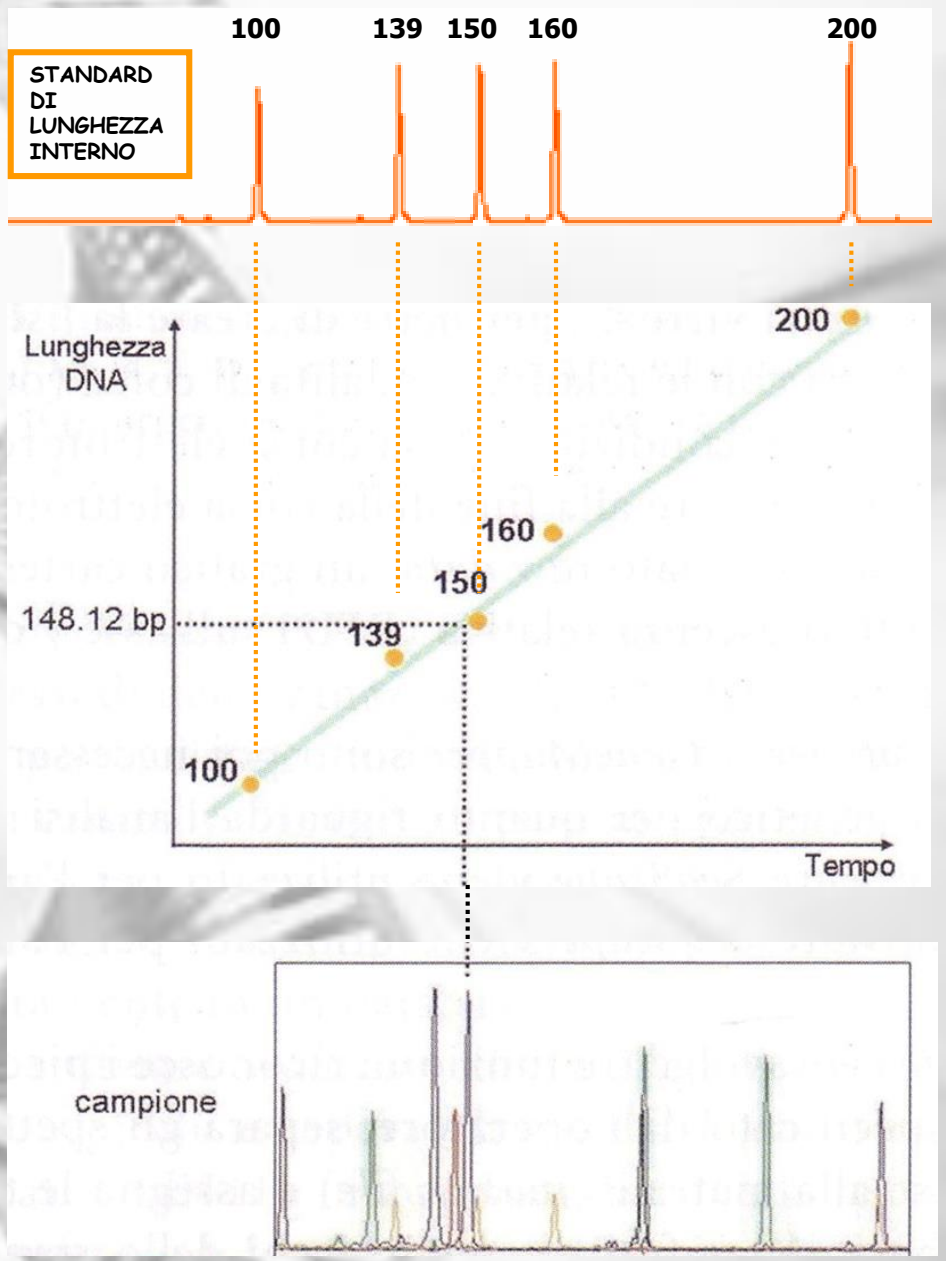


TAMRA (Yellow) ROX (Red)

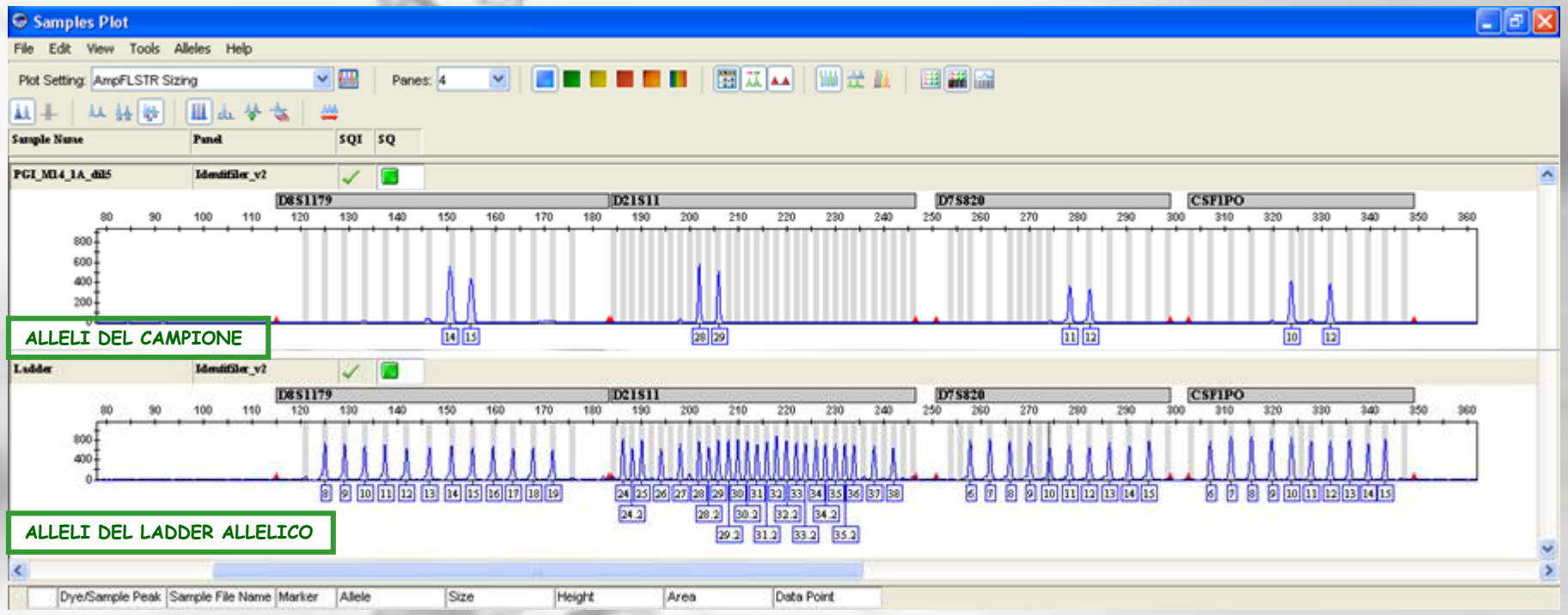




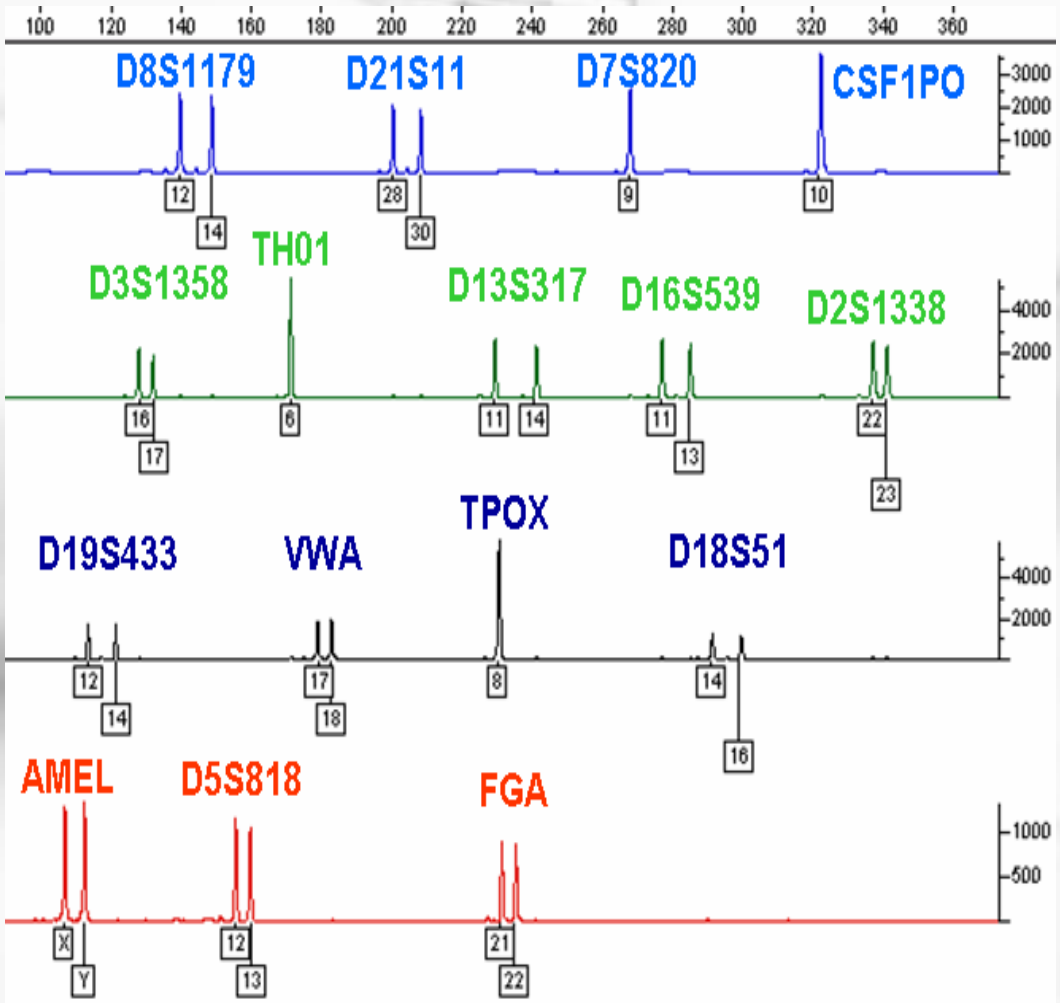
ASSEGNAZIONE DEL PESO MOLECOLARE DEI PICCHI DEL CAMPIONE



ASSEGNAZIONE ALLELICA



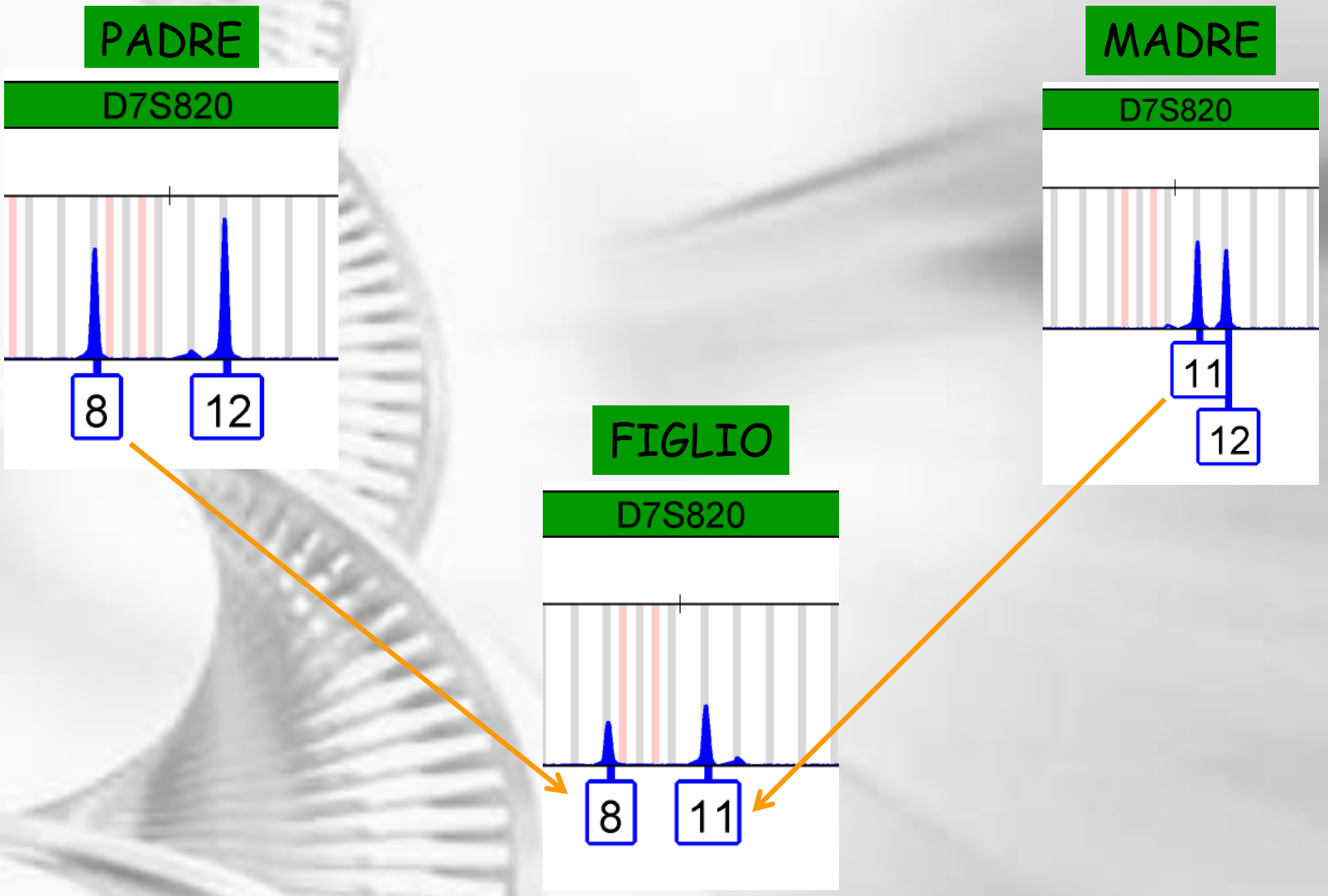
RISULTATO



AmpFlSTR®Identifiler™

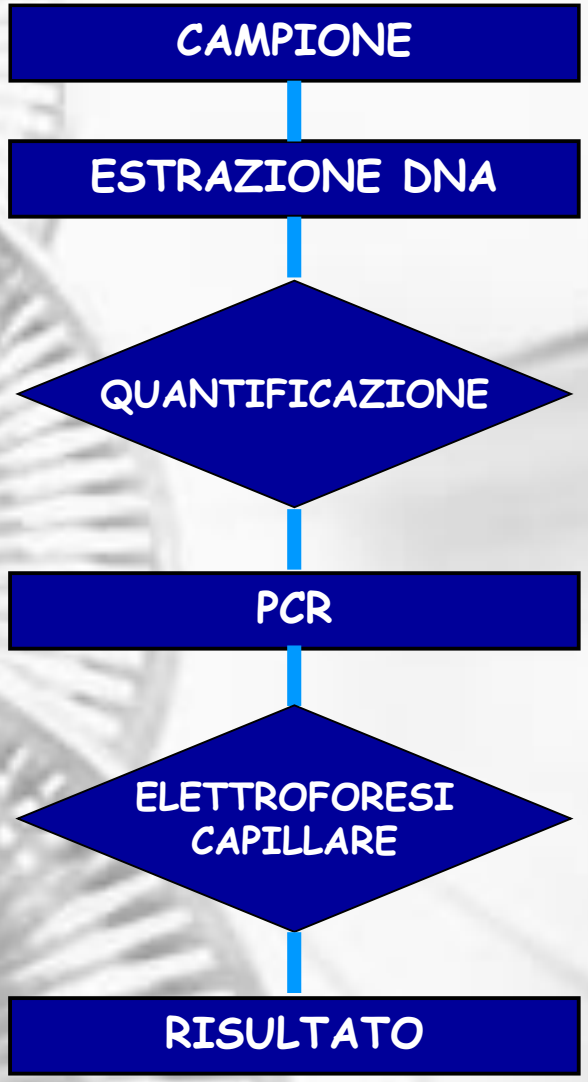
LOCUS	ALLELI	
D8S1179	12	14
D21S11	28	30
D7S820	9	9
CSF1PO	10	10
D3S1358	16	17
TH01	6	6
D13S317	11	14
D16S539	11	13
D2S1338	22	23
D19S433	12	14
vWA	17	18
TPOX	8	8
D18S51	14	16
AMEL	X	Y
D5S818	12	13
FGA	21	22

ATTRIBUZIONE DI PATERNITA'



RISULTATO: PROFILO GENETICO

LOCUS STR	PADRE	FIGLIA	MADRE
D8S1179	13 / 14	13 / 13	13 / 13
D21S11	30 / 32	30 / 34	29 / 34
D7S820	10 / 12	10 / 12	10 / 10
CSF1PO	10 / 12	10 / 10	10 / 11
D3S1358	15 / 18	15 / 18	15 / 15
TH01	8 / 9	8 / 11	11 / 11
D13S317	9 / 11	11 / 11	8 / 11
D16S539	9 / 9	9 / 11	11 / 11
D2S1338	19 / 20	20 / 20	17 / 20
D19S433	15 / 15	13 / 15	13 / 13
vWA	14 / 16	16 / 16	14 / 16
TPOX	8 / 11	8 / 11	8 / 11
D18S51	12 / 13	12 / 12	12 / 18
D5S818	11 / 12	11 / 12	12 / 12
FGA	22 / 25	22 / 24	21 / 24
AMELOGENIN	X / Y	X / X	X / X


CAMPIONE
ESTRAZIONE DNA
QUANTIFICAZIONE
PCR
ELETTROFORESI CAPILLARE
RISULTATO

Laboratorio di identificazione e tracciabilità molecolare

GRAZIE PER L'ATTENZIONE