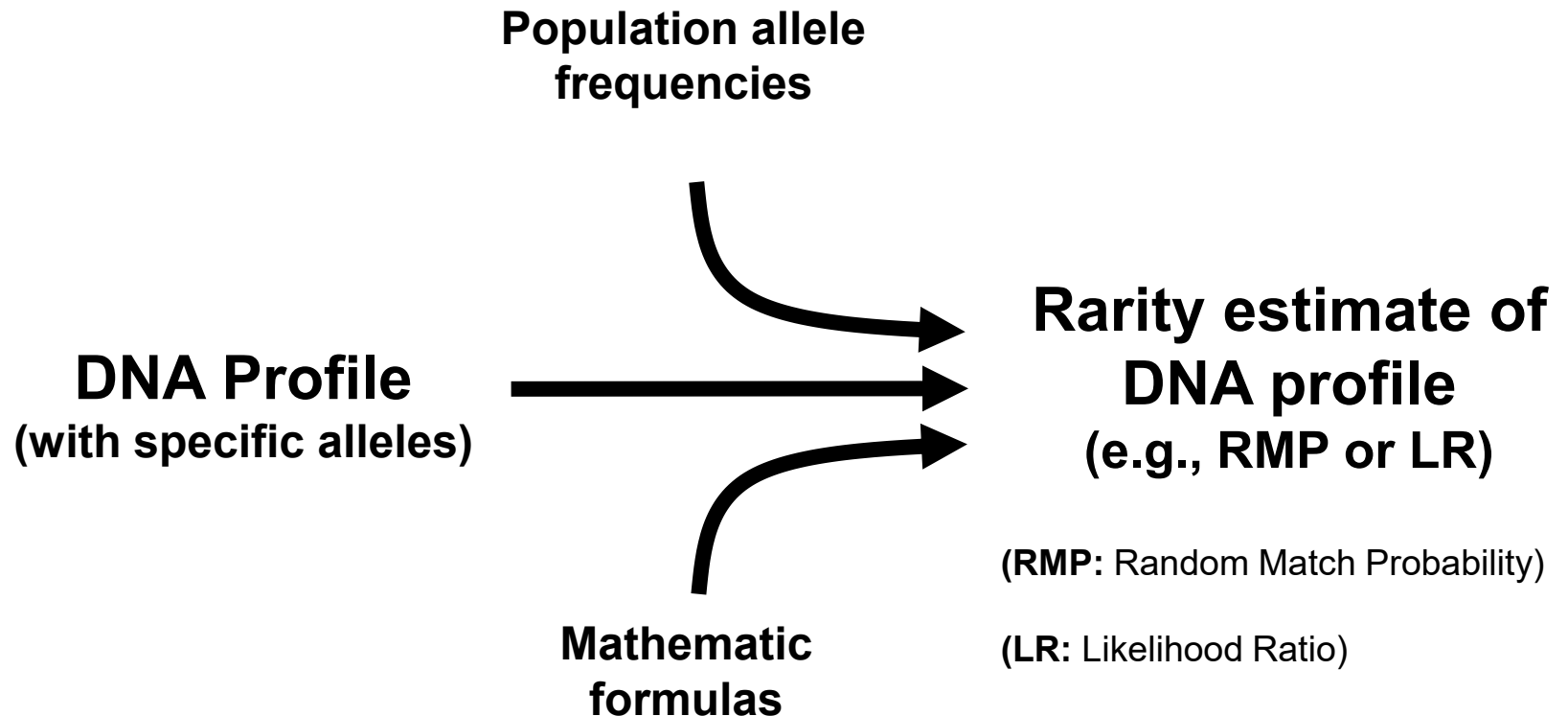


GENETICS AND POPULATION GENETICS

Forensic Genetics



Crime committed

↓ *Biological material transferred*

**Evidence sample
“Q” (Question)**

Steps Involved

Collection

Sample Storage

Characterization

Extraction

Quantitation

Amplification

STR Markers

Separation/
Detection

Data/Results
Evaluation

Statistical
Interpretation

Biology

Technology

Genetics

Profile put on database

*May match another
(K')*

Database Search

Exclusion (no match)

Q ≠ K

Q

DNA Profile
Comparison

K

Q = K

Inclusion (match)

Report

(with statistical weight)

**Q-K comparison during
forensic DNA testing**

Suspect developed

↓

**Reference sample
“K” (Known)**

Steps Involved

Collection

Sample Storage

Extraction

Quantitation

Amplification

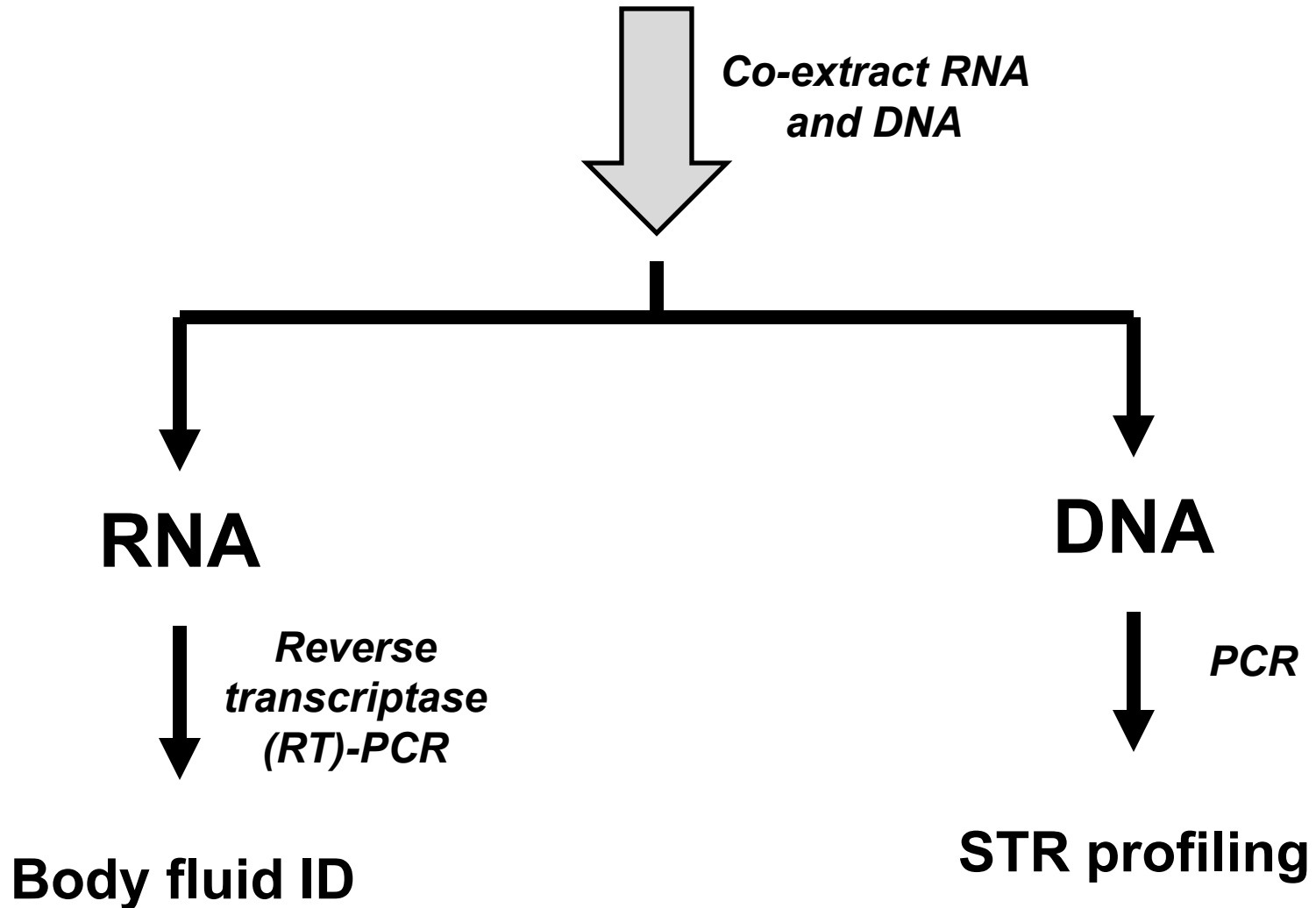
STR Markers

Separation/
Detection

Data/Results
Evaluation

Profile put on database

Evidentiary Sample (stain from crime scene)



Polymorphisms in DNA sequences

Single base substitutions / indels: Sequence polymorphisms (SNP)

-----AGACTAGACATT-----

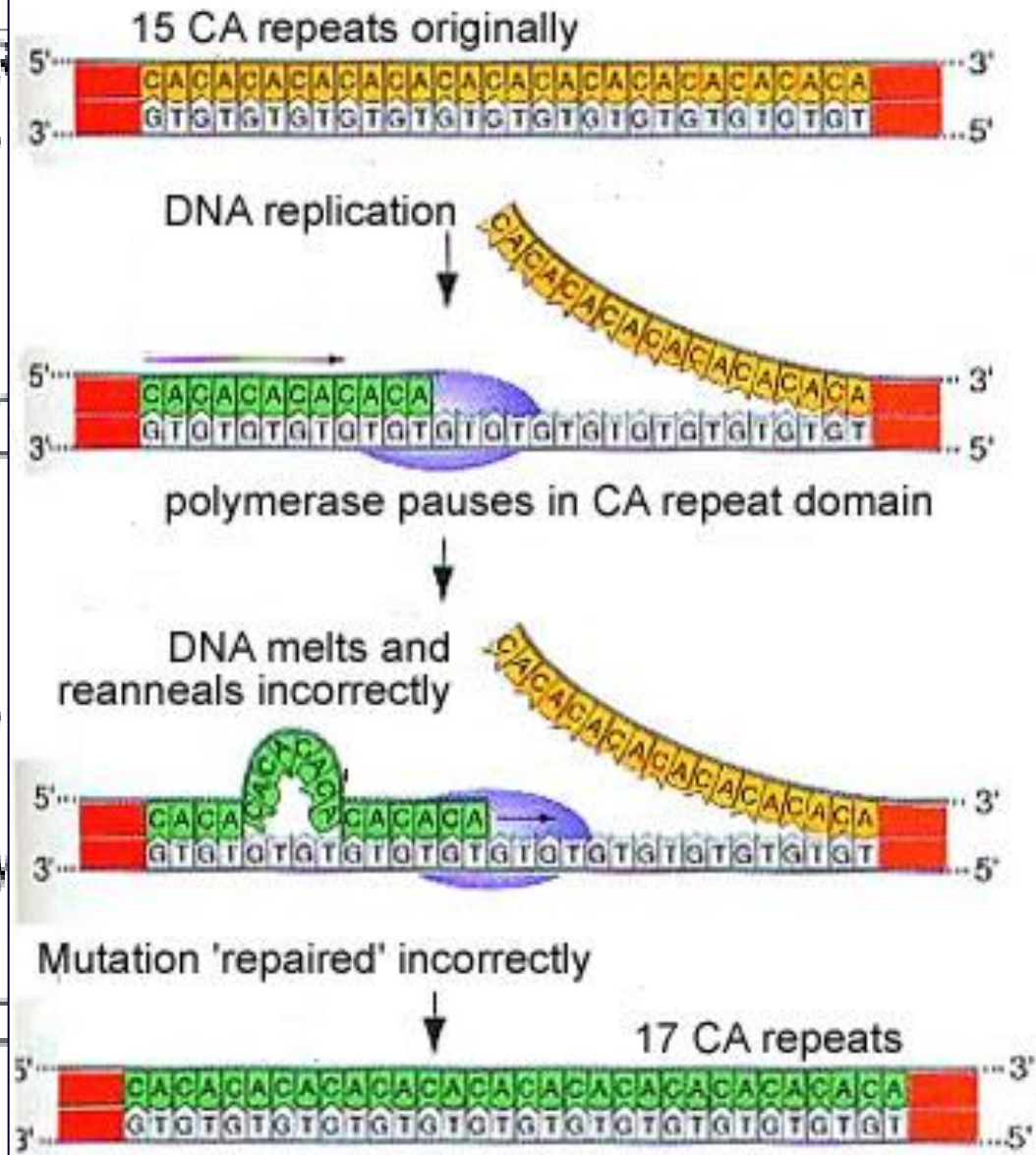
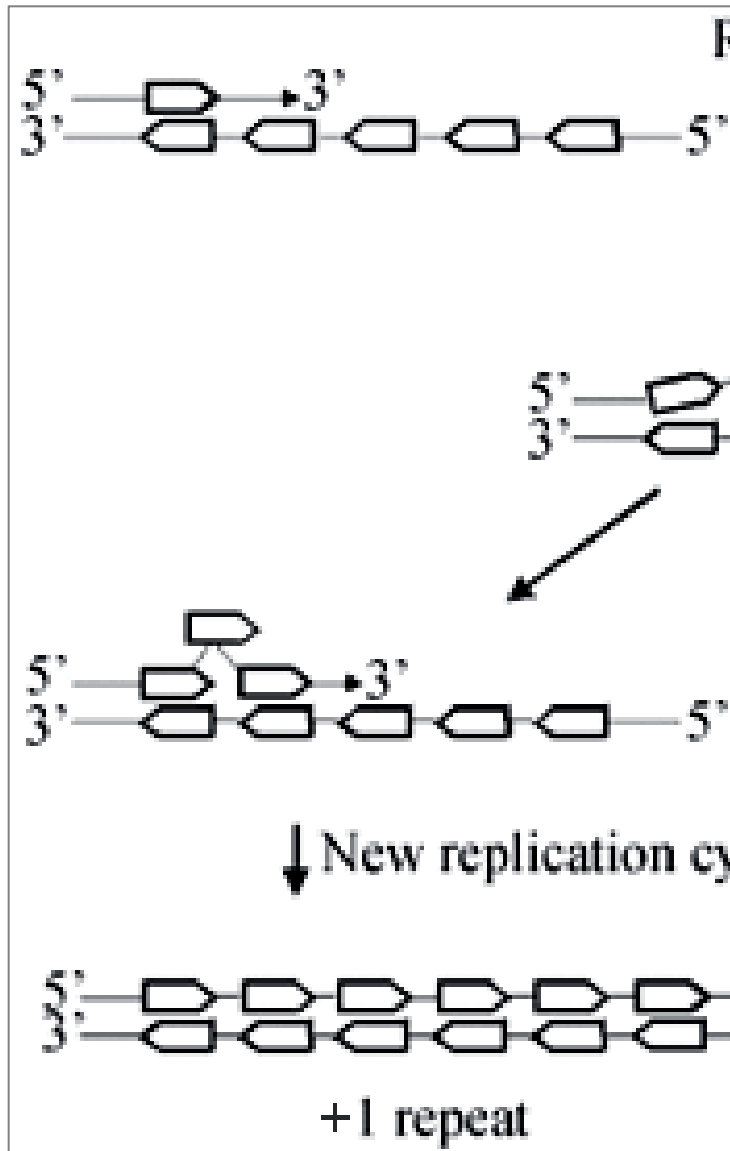
-----AGAC_TAGGCATT-----

Tandemly repeated sequence motifs: Fragment length polymorphism, satellite DNA (VNTRs, mini- and microsatellites, STRs)

----- (AATG)(AATG)(**AATG**) -----

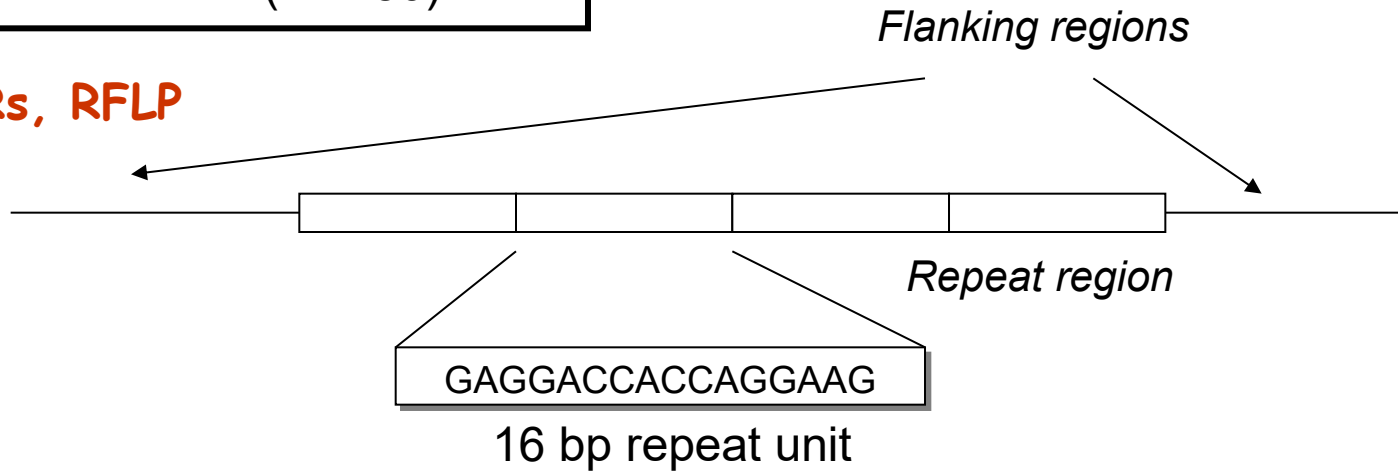
----- (AATG)(AATG) -----

Microsatellite evolution



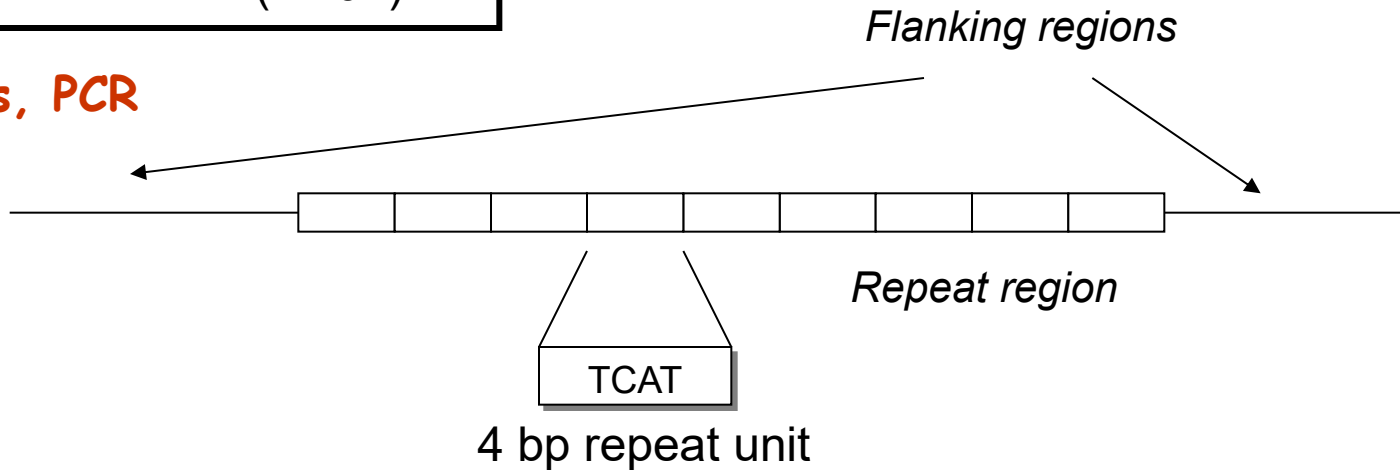
Minisatellite (D1S80)

VNTRs, RFLP

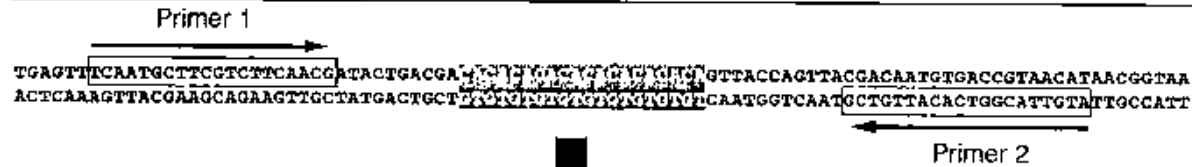


Microsatellite (TH01)

STRs, PCR



Genotyping microsatellites by PCR

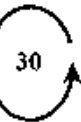


Note: the nucleotide composition of the most variable area is indicated in the enlarged region. Bases at variable positions are highlighted.

94°C

55°C

72°C



PCR

PCR = Polymerase chain reaction

Exponential amplification of a known DNA sequence using complementary primers, a thermostable DNA polymerase and a temperature cycling protocol

Allele length classes

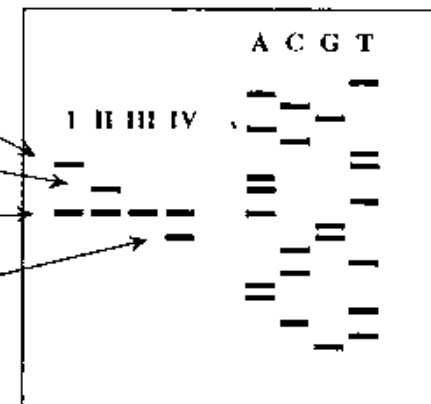
TCATGCTTCTGCTCTCAACGATACTGACGA **TACACACACACACACACACACAC** GTTACCGAGTTACGACAAATGTTACCGTAACAT
AGTTACGAGACAGAAGTGTCTATGACTGCT **CAGTGTGTGTGTGTGTGTGTGTGT** CAATGGTCAATGCTGTTACACTGGCATTGTA

TCAATGCTTCGTCTTCAACGTAFACTGACG**GAGCAGCAGCGTG**GTTACCAGTTACGACAATOTGACCOTARCAT
AGTTACGAAGCAGAAGTTGGCTATGACTGCT**GTGAGGATGATGCTGCCTGAT**CAATTGGTCAATGCTGTTAACTGCGCATTOTA

[illegible]

TCAATGCTTTCGTCTTTCAACGTAAGTAGCAGCGAAGGCATTGGTGCCGCTGTTACCAGGTACGACAATGTGACCGTAACAT
AGTTACGAAGCAGAAGTTGGCATGACTGCTGT

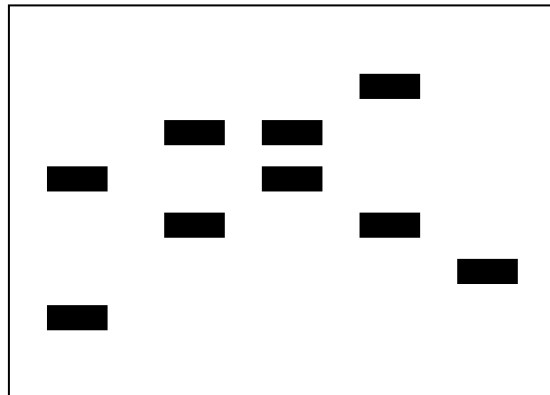
Fractionated on sequencing gel



Microsatellite locus genotyping: multiplex-PCR

TACAAAAAATACAAAAATTAGTTGGGCATGGTGGCACGTGCCTGTAGTCTCAGCTACT
TGCAGGGCTGAGGCAGGAGGAGTTCTTGAGCCAGAA [G→A] GTTAAGGCTGCAGTGAG
CCATGTTTCATGCCACTGCACTTCACTCTGAGTGACAAATTGAGACCTTGTCTC/agaa/
agaa/agaa/agaa/agaa/agaa/agaa/agaa/agaa/agaa/agaa/agaa/agaa/
/agaa/agaa/agaa/agaa/agaa/ [AAAGAGAGA]GGAAAGAAAGAGAAAAAGAAAA
GAAATAGTAGCAACTGTTATTGTAAGACATCTCCACACACCAGAGAAGTTAATTTTAAT
TTTAACATGTTAAGAACAGAGAGAAGCCAACATGTCCACCTTAGGCTGACGGTTTGTTT
ATTTGTGTTGTTGCTGGTAG*TC [G→A] GGTTTGTTATTTTTTAAAGTAGCTTATCCAAT
ACTTCATTAACAATTTTCAGTAAGTTATTTTCATCTTTCAACATAAATACGCACAAGGATT
TCTTCTGGTCAAGACCAAATAATATTAGTCCATAGTAG

Polyacrylamide Gel

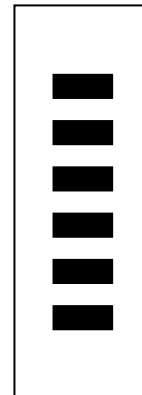


Find representative alleles
spanning population
variation

Combine



Re-
amplify



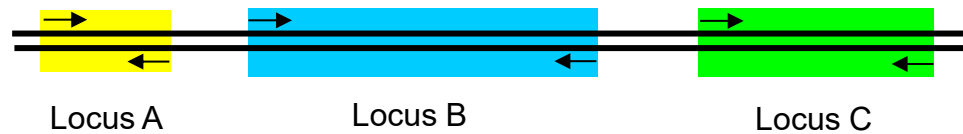
Separate PCR products from
various samples amplified with
primers targeted to a particular
STR locus

(a) Simultaneous amplification of three locations on a DNA template

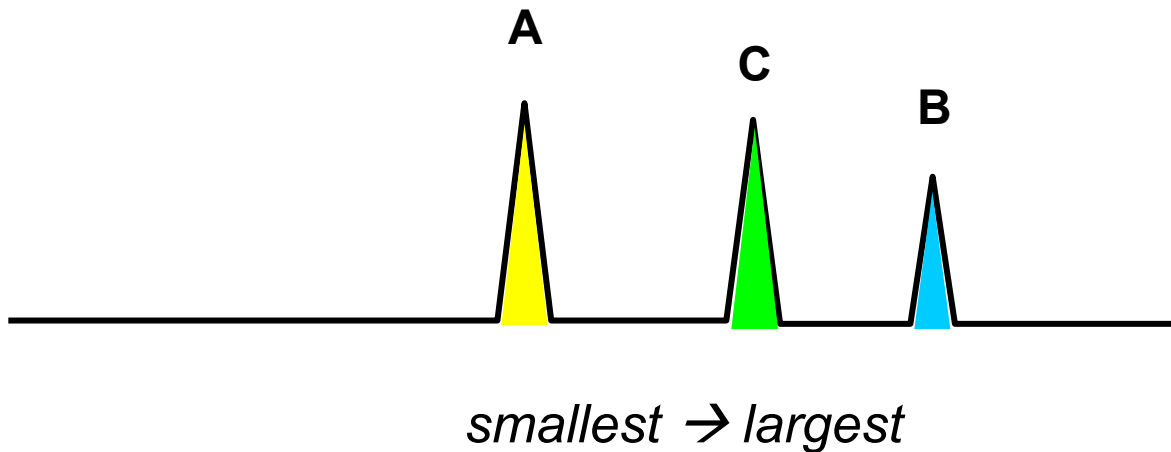
*Dye-labeled
PCR primers*

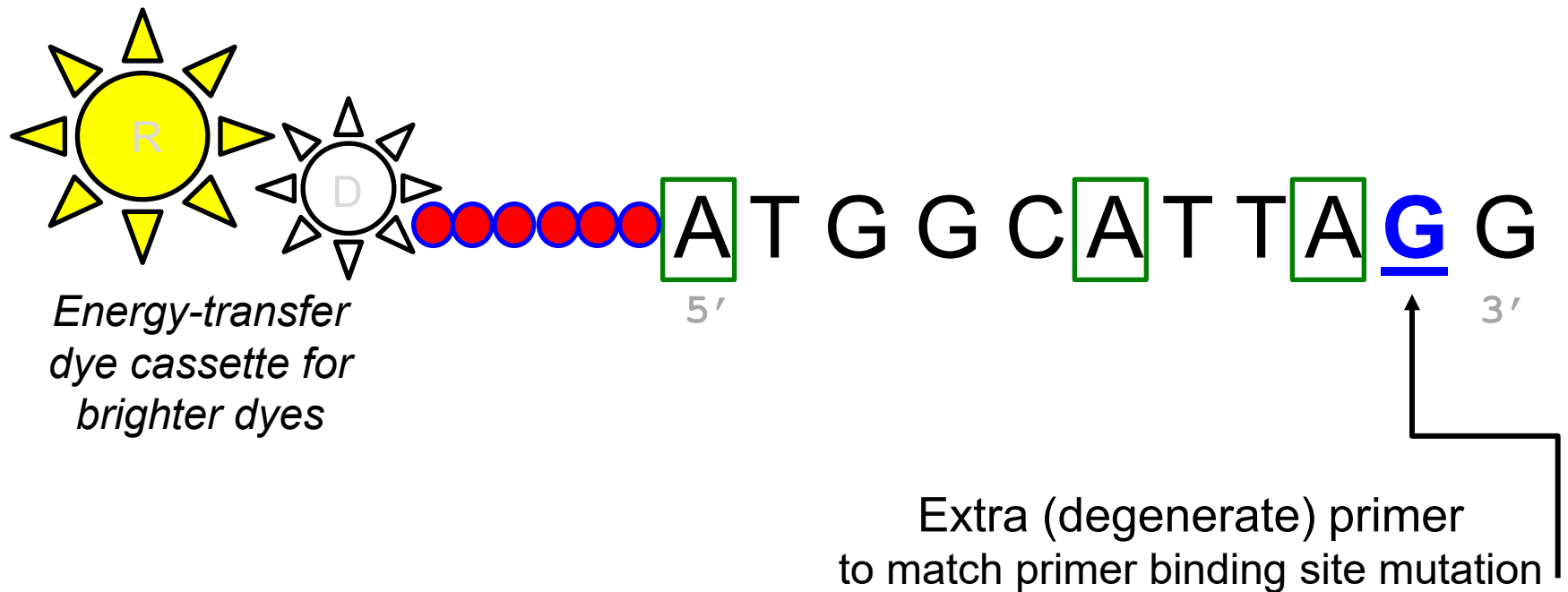
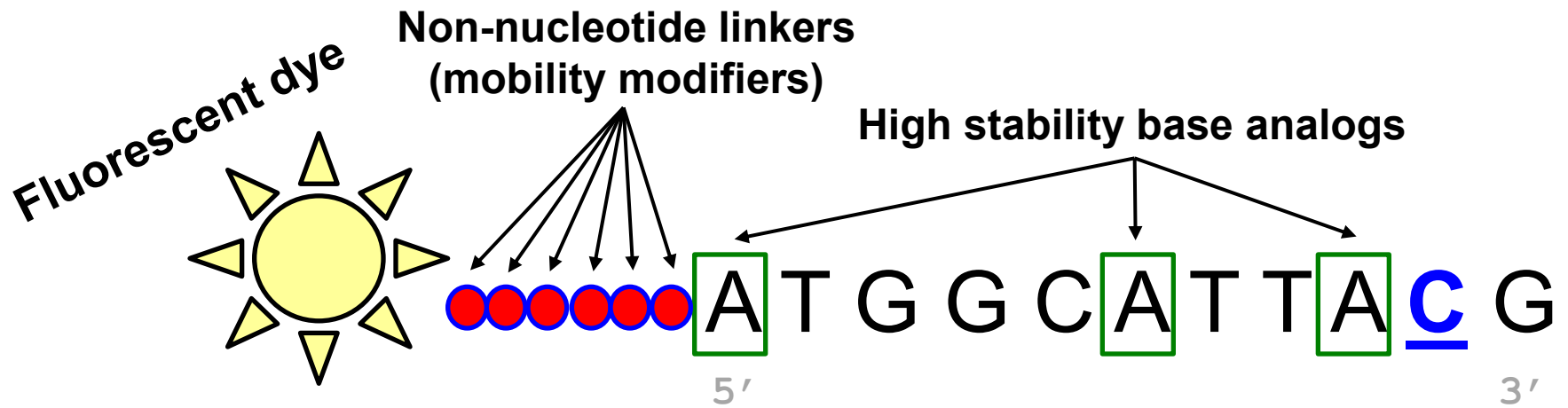


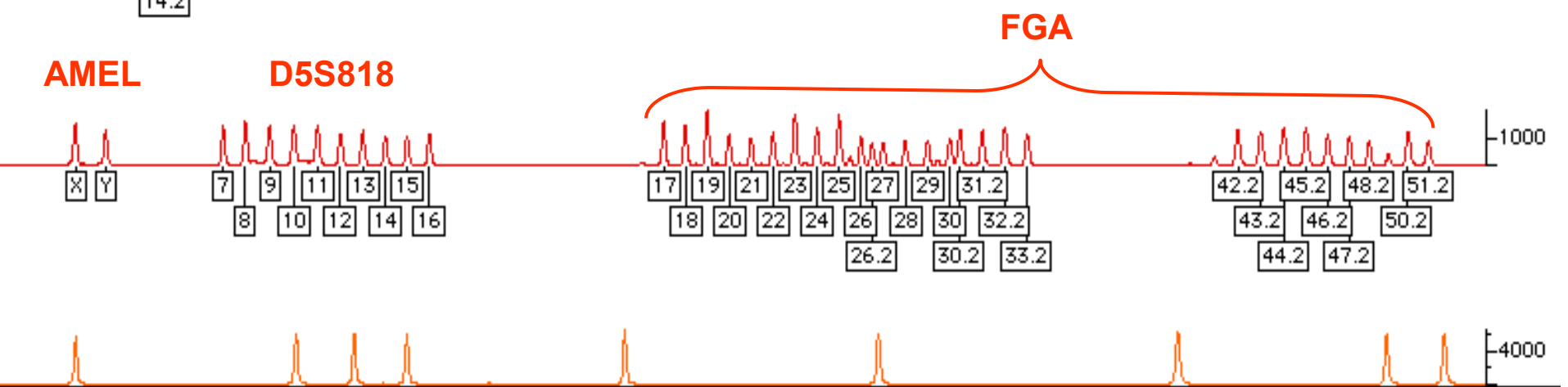
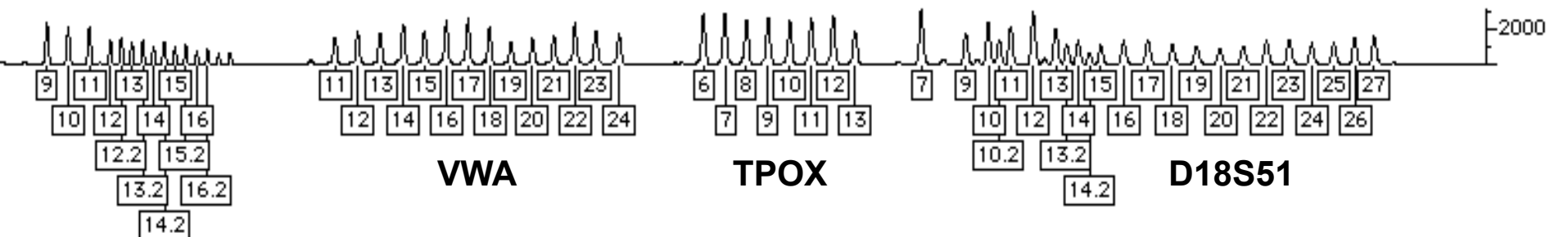
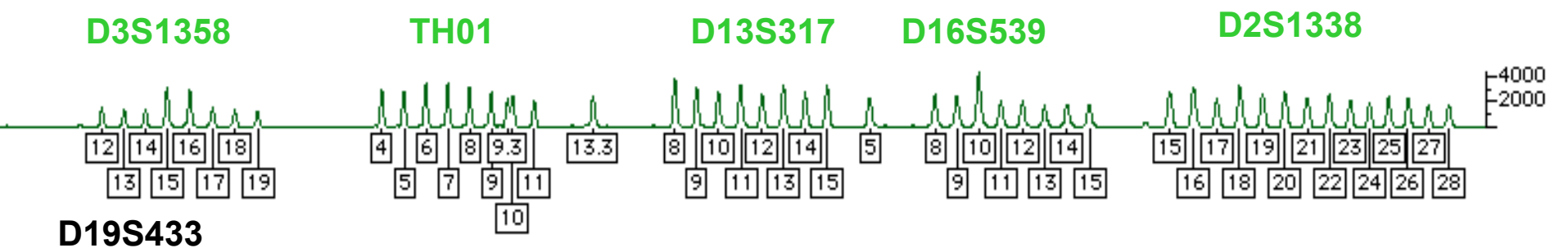
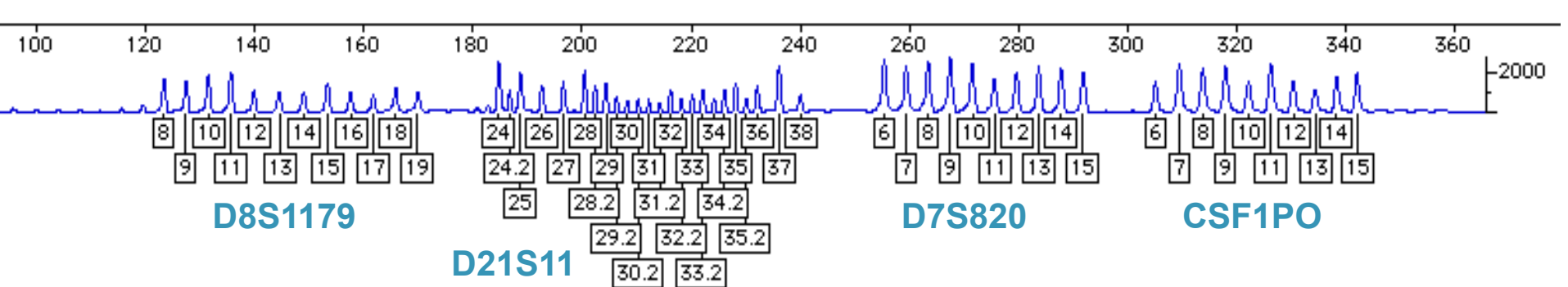
*Dye-labeled
PCR products*

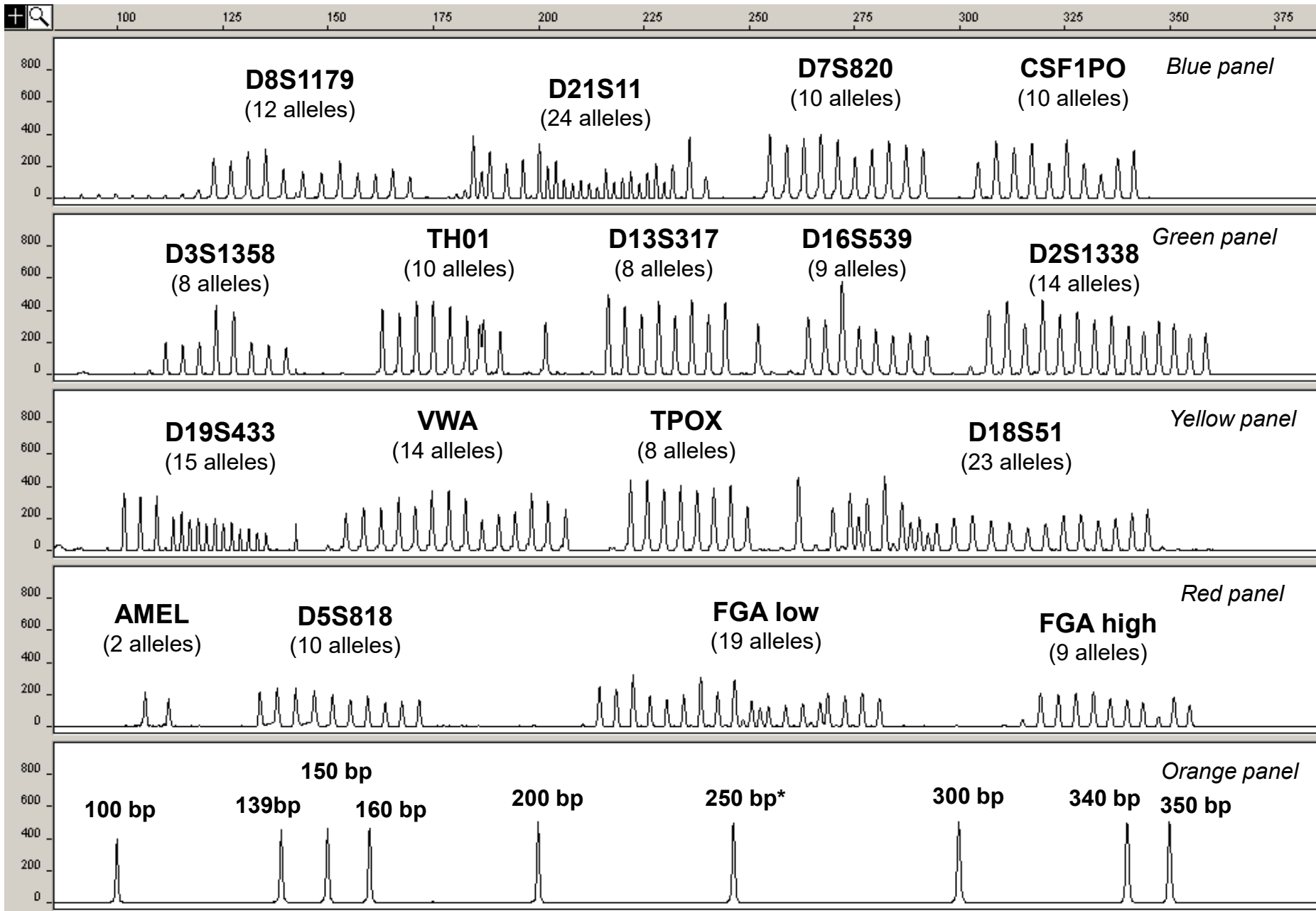


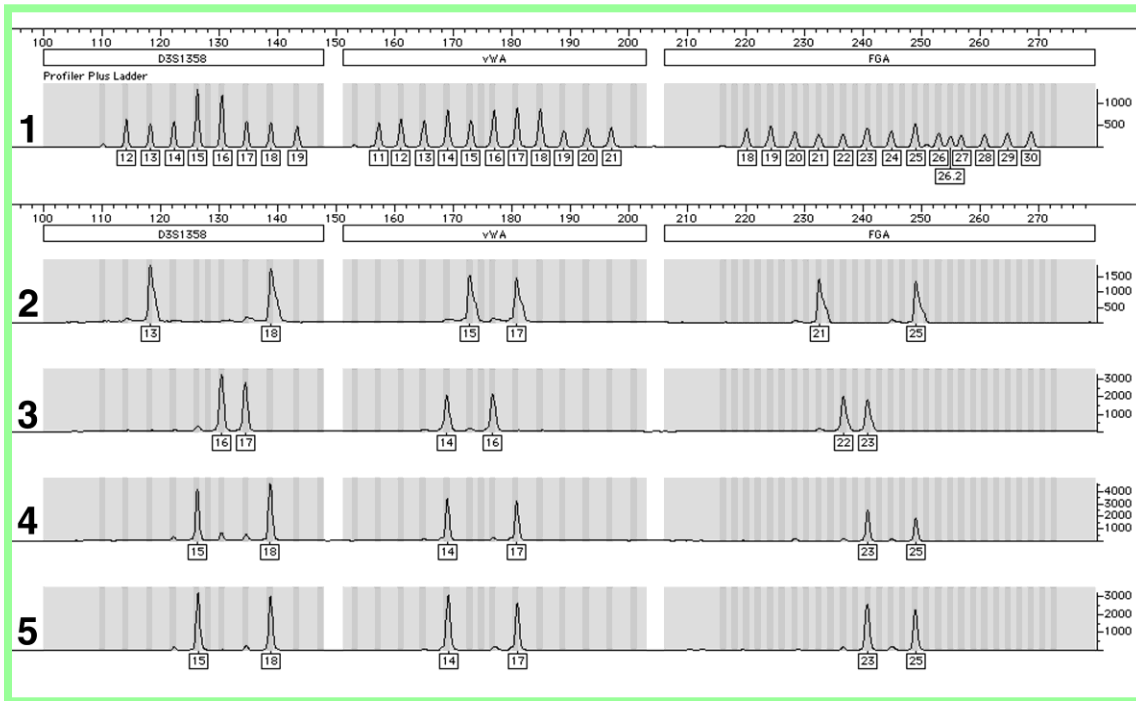
(b) Resolution of PCR products with size-based separation method











Multiplex STR analyses on three microsatellite loci

1: allelic ladder

2, 3, 5: DNA profile of three different persons (suspects)

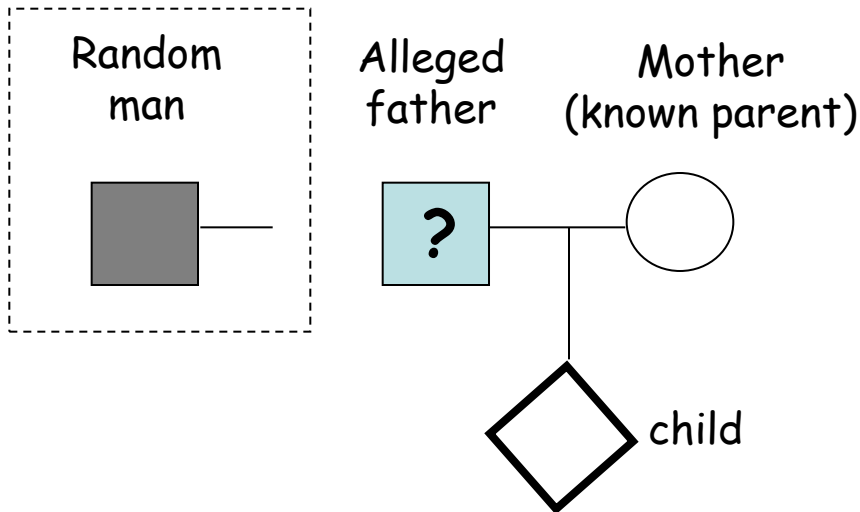
4: evidence DNA profile (cigarette butt)

	D3S1358	vWA	FGA
3: A person	16/17	14/16	22/23
4: Evidence	15/18	14/17	23/25
5: B person	15/18	14/17	23/25

Different origin = Exclusion

Identical? = Contribution?
Random match probability?

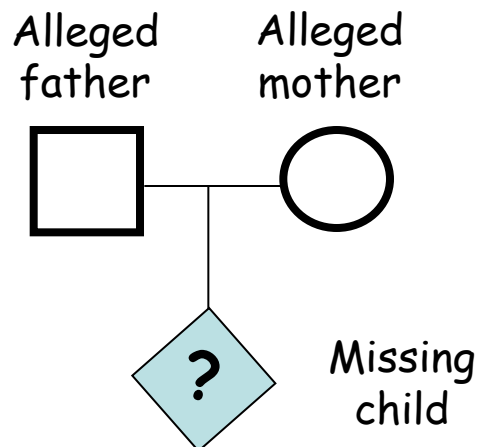
(a) Parentage (Paternity) Testing



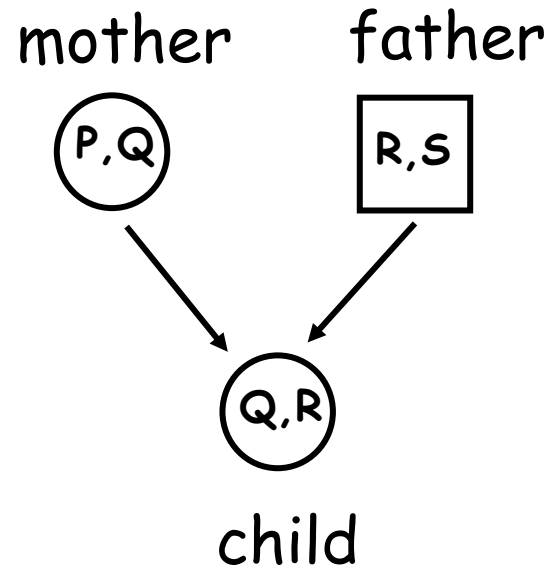
Rules of Inheritance

- 1) Child has two alleles for each autosomal marker (*one from mother and one from biological father*)
- 2) Child will have mother's mitochondrial DNA haplotype (barring mutation)
- 3) Child, if a son, will have father's Y-chromosome haplotype (barring mutation)

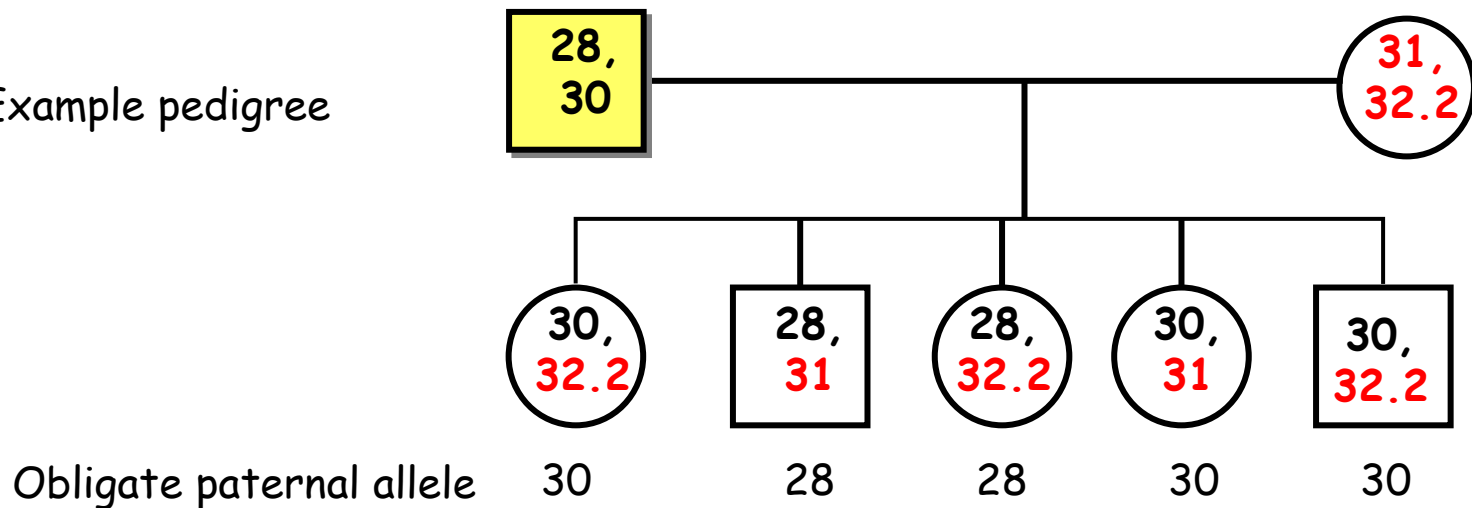
(b) Reverse Parentage Testing (Missing Persons Investigation)



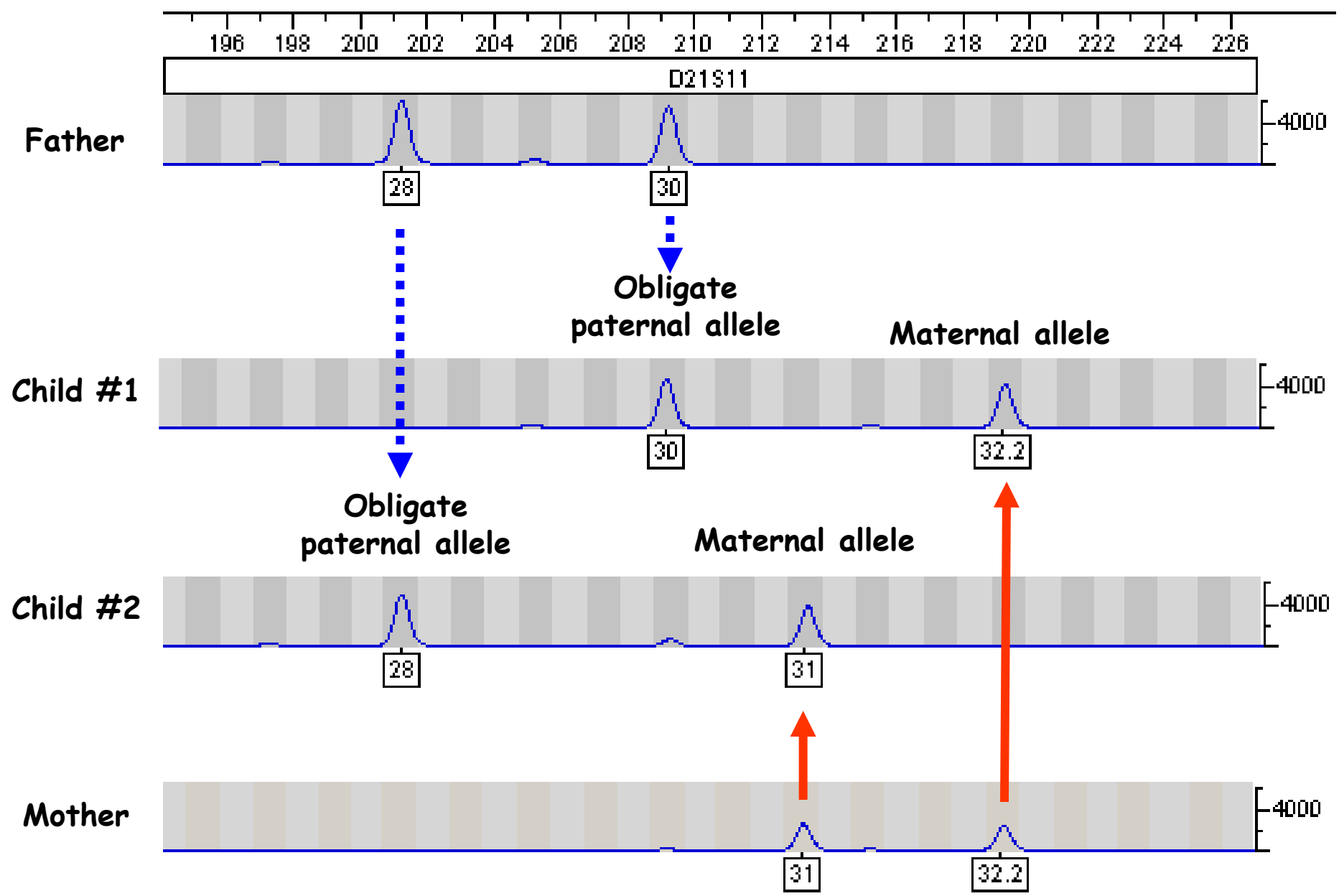
(a) Mendelian Inheritance



(b) Example pedigree



(c) Example data

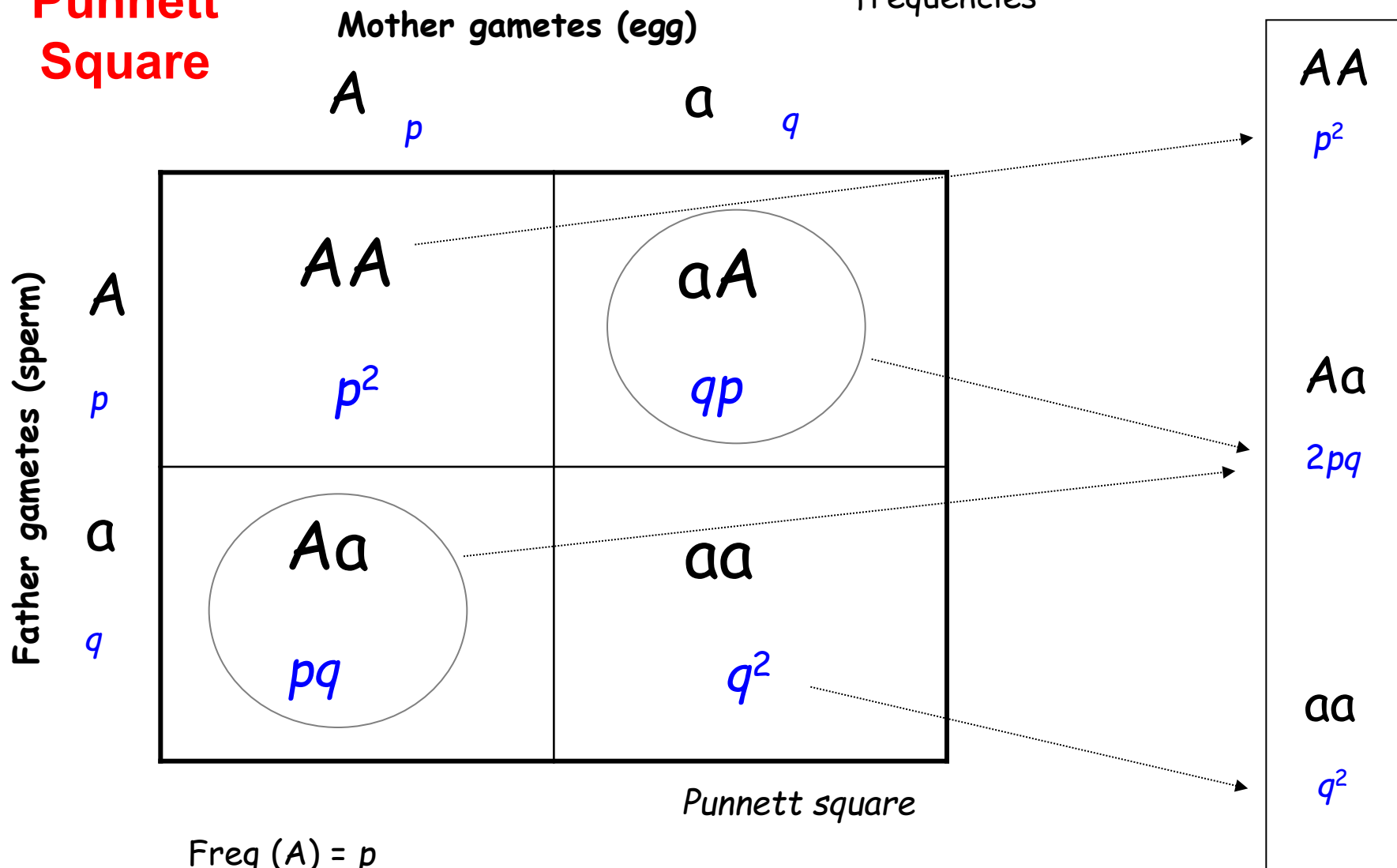


How Statistical Calculations are Made

- **Generate data** with set(s) of samples from desired population group(s)
 - Generally only 100-150 samples are needed to obtain reliable allele frequency estimates
- **Determine allele frequencies** at each locus
 - Count number of each allele seen
- Allele frequency information is used to **estimate the rarity of a particular DNA profile**
 - Homozygotes (p^2), Heterozygotes ($2pq$)
 - Product rule used (multiply locus frequency estimates)
 $PM = (P1)(P2)...(Pn)$

Punnett Square

Resulting genotype combinations and frequencies



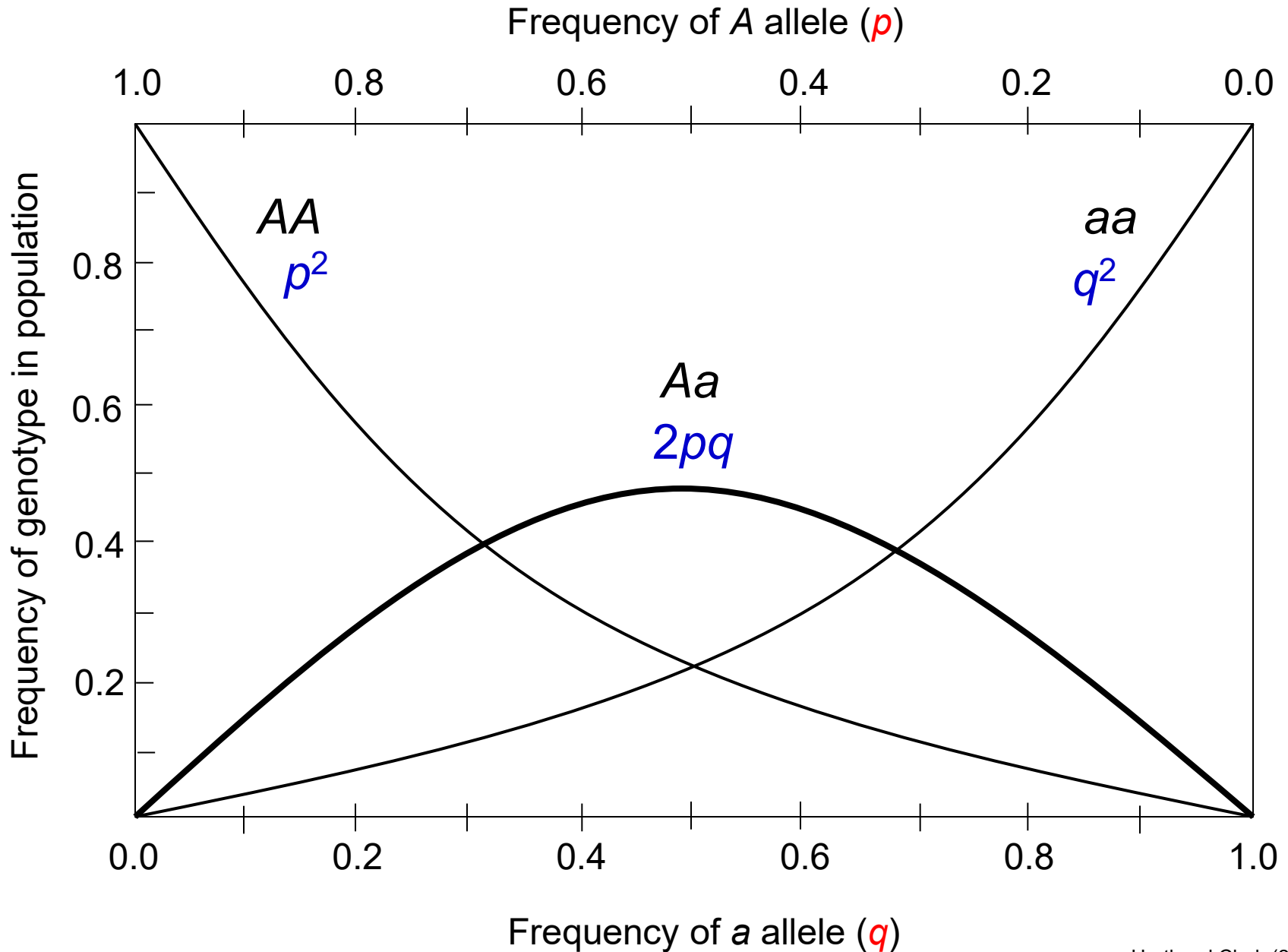
$$\text{Freq (A)} = p$$

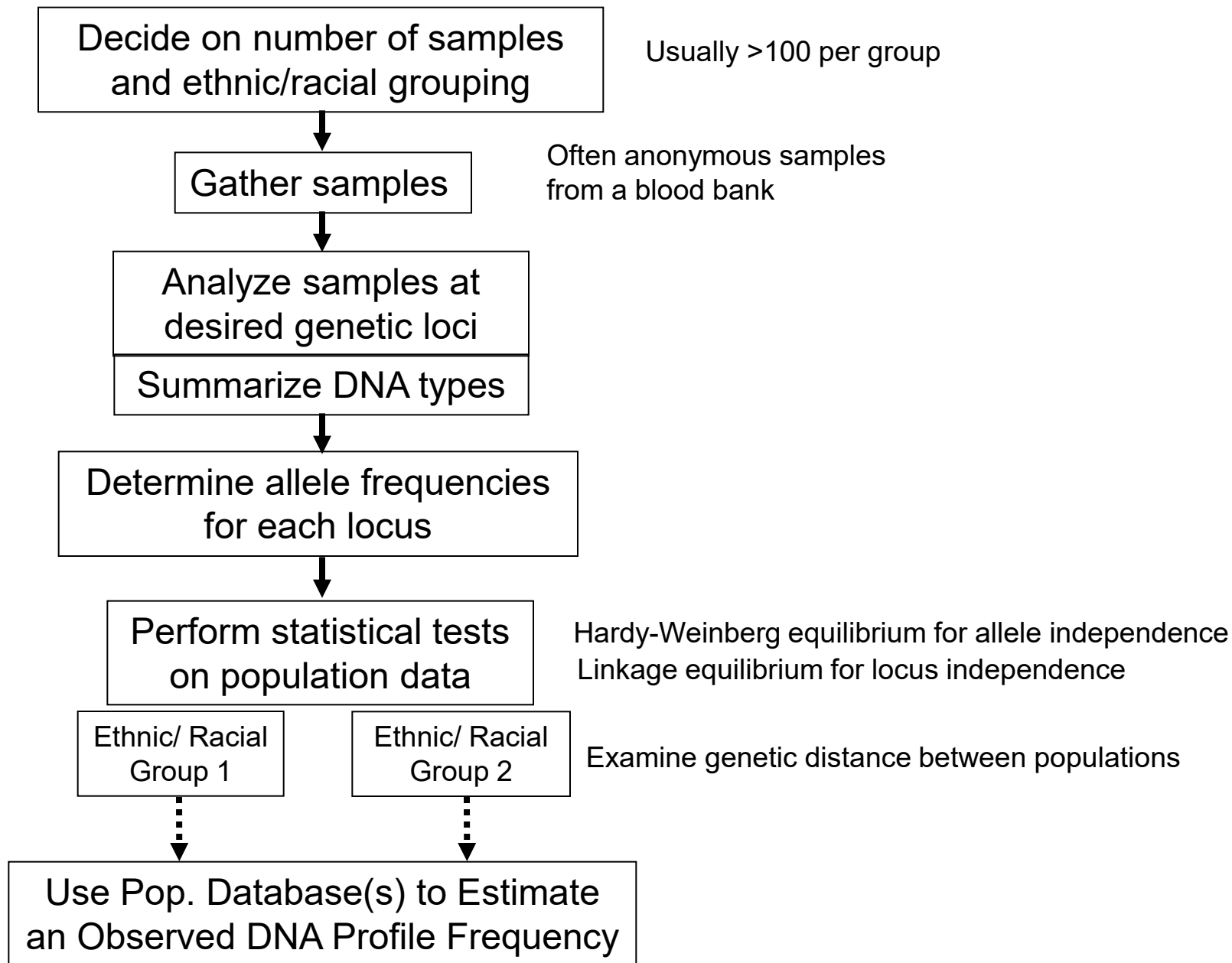
$$\text{Freq (a)} = q$$

$$p + q = 1$$

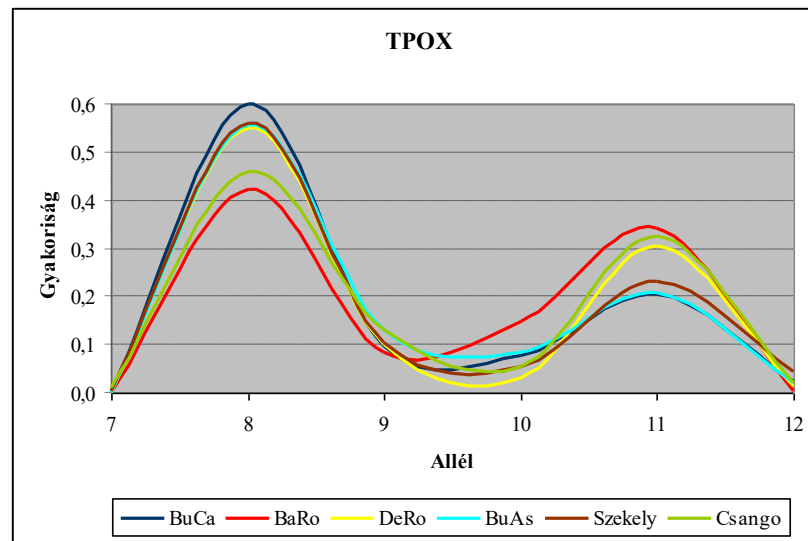
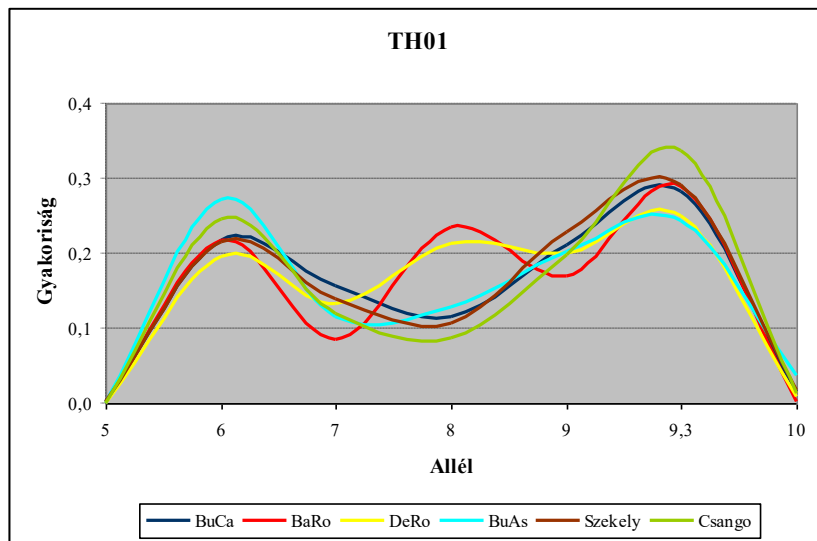
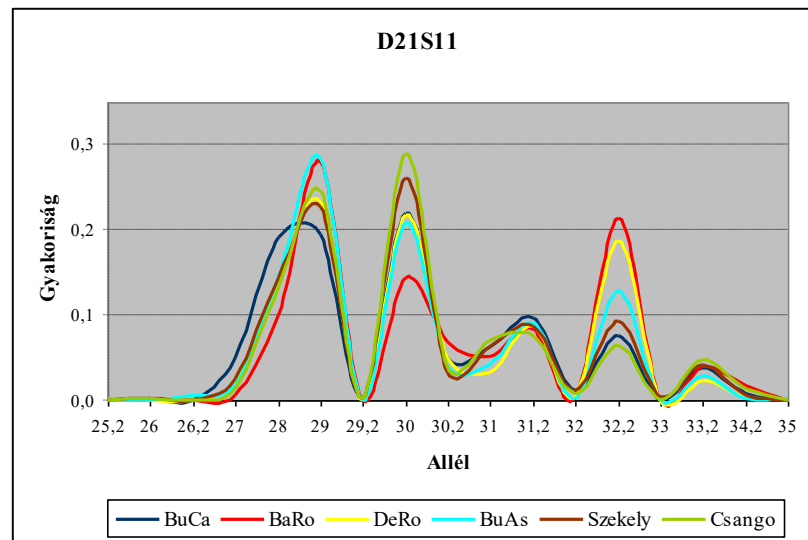
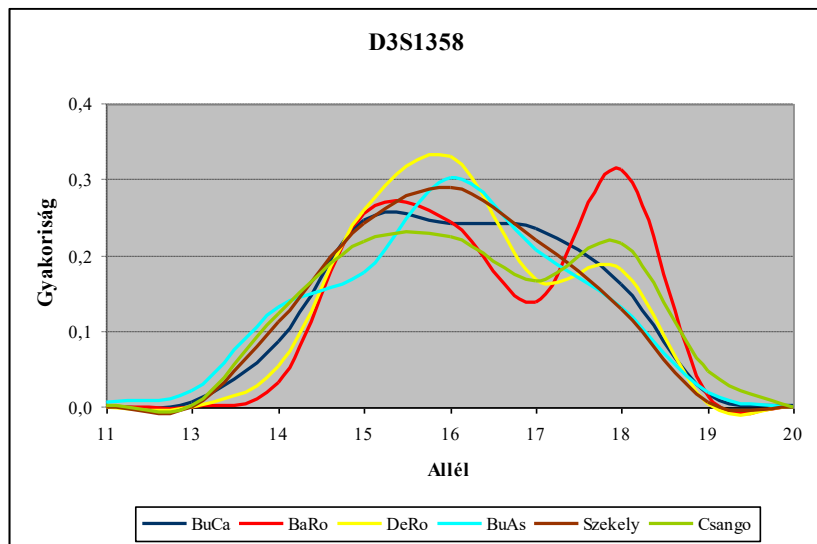
$$(p + q)^2 = p^2 + 2pq + q^2$$

Relationship between Allele Frequency and Genotype Frequency





Microsatellite allele frequency diagrams



Allele- and genotype frequencies

Genotype Array	8	9	10	11	12	13	14	15		Allele Count	Observed Frequency
D13S317	8,8	8,9	8,10	8,11	8,12	8,13	8,14	8,15			
8	9	9	1	17	13	10	0	0	8	68	0.11258
		9,9	9,10	9,11	9,12	9,13	9,14	9,15			
9		1	2	15	10	4	3	0	9	45	0.07450
			10,10	10,11	10,12	10,13	10,14	10,15			
10			2	12	6	3	2	1	10	31	0.05132
				11,11	11,12	11,13	11,14	11,15			
11				37	54	21	12	0	11	205	0.33940
					12,12	12,13	12,14	12,15			
12					21	18	7	0	12	150	0.24834
						13,13	13,14	13,15			
13						7	5	0	13	75	0.12417
							14,14	14,15			
14							0	0	14	29	0.04801
								15,15			
15								0	15	1	0.00166
										604	

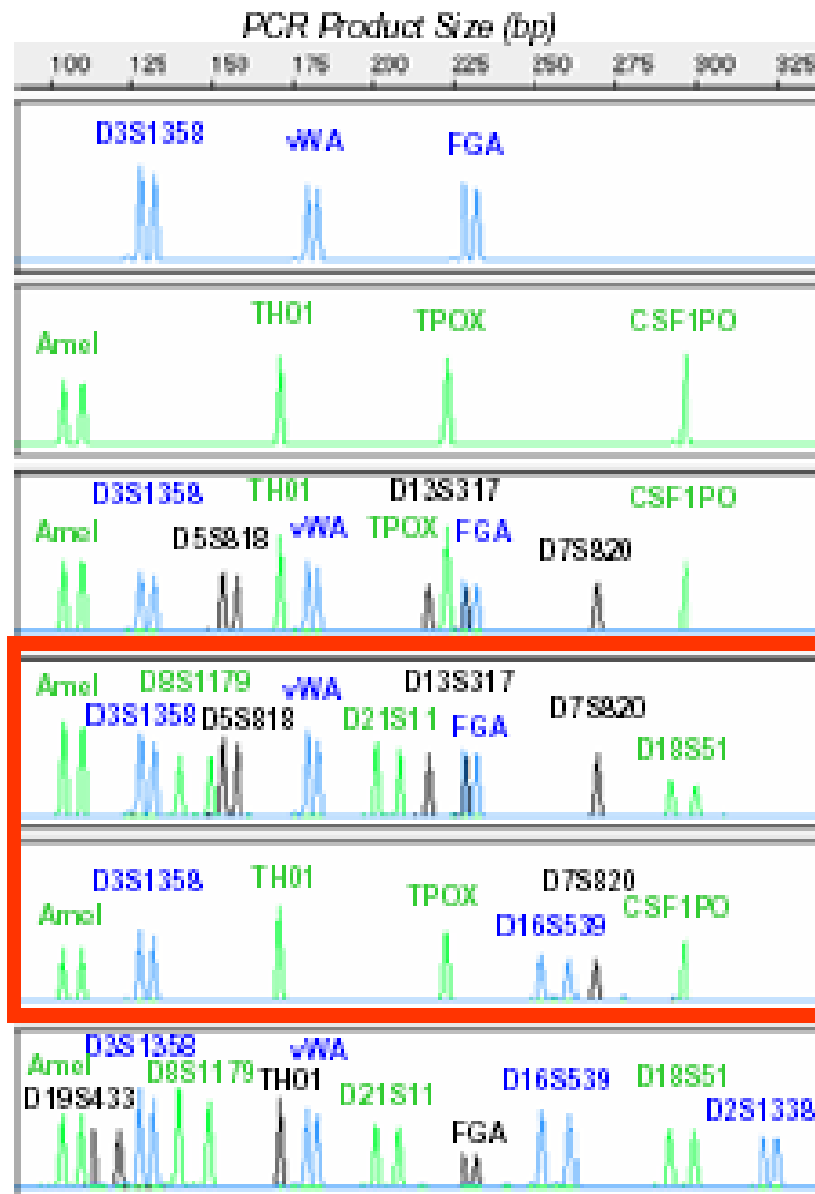
Genotype 11,12 was observed
54 times in 302 persons
(604 analysed chromosomes)

Int J Legal Med (2000) 113:272–275

ORIGINAL ARTICLE

B. Egyed · S. Füredi · M. Angyal · L. Boutrand
A. Vandenberghe · J. Woller · Z. Pádár

Analysis of eight STR loci in two Hungarian populations



Same DNA Sample Run with Each of the ABI STR Kits

Blue

Power of Discrimination
1:5000

Green I

1:410

Profiler™

$1:3.6 \times 10^9$

Profiler Plus™

$1:9.6 \times 10^{10}$

COfiler™

$1:8.4 \times 10^5$

SGM Plus™

$1:3.3 \times 10^{12}$

Likelihood ratio calculation /Bayes theorem/ Hypothesis testing

in case of matching DNA profiles:

$$LR = \frac{\Pr(E|H_1)}{\Pr(E|H_2)} = \frac{\Pr(\text{STR profile}|\text{the DNA comes from the suspect})}{\Pr(\text{STR profile}|\text{the DNA comes from a random person})}$$

The LR equals the probability of the evidence given the hypothesis of the prosecution (H_1 or H_p) divided by the probability of the evidence given the hypothesis of the defense (H_2 or H_d).

Example:

$$LR = \frac{\Pr(E|H_p)}{\Pr(E|H_d)} = \frac{1}{p^2} = \frac{1}{(0.139)^2} = \frac{1}{0.0193} = 51.8.$$

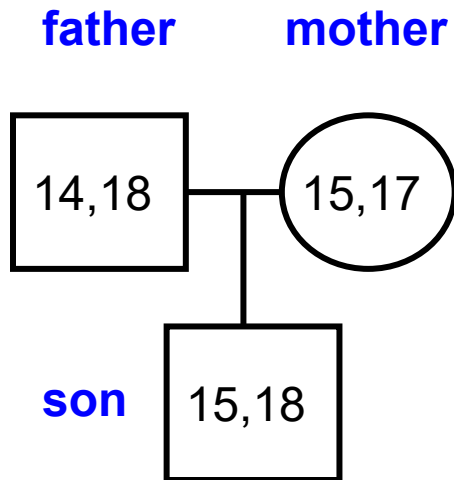
Homozygous
DNA profile

$$LR = \frac{\Pr(E|H_p)}{\Pr(E|H_d)} = \frac{1}{2pq} = \frac{1}{2(0.134)(0.147)} = \frac{1}{0.0394} = 25.4.$$

Heterozygous
DNA profile

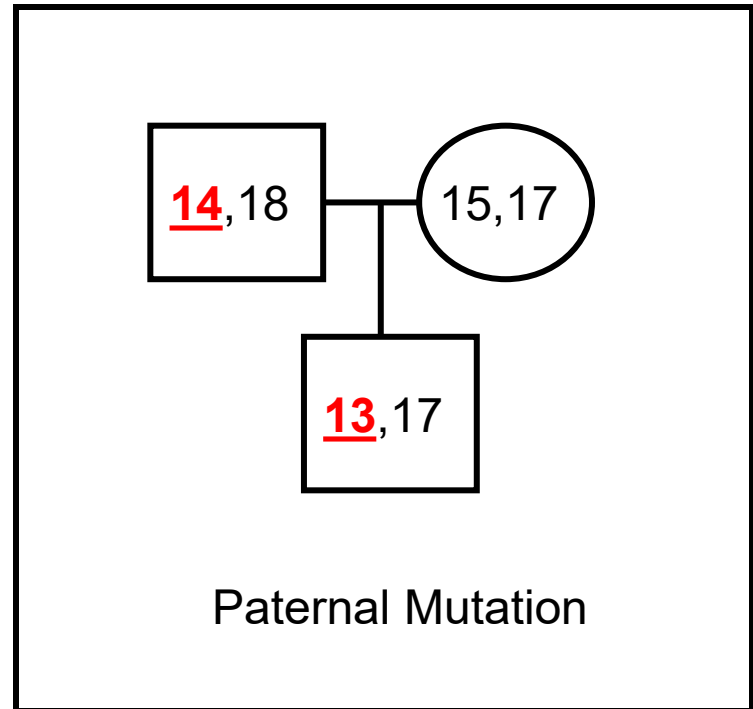
Mutation Observed in Family Trio

(a)



Normal Transmission of Alleles
(No Mutation)

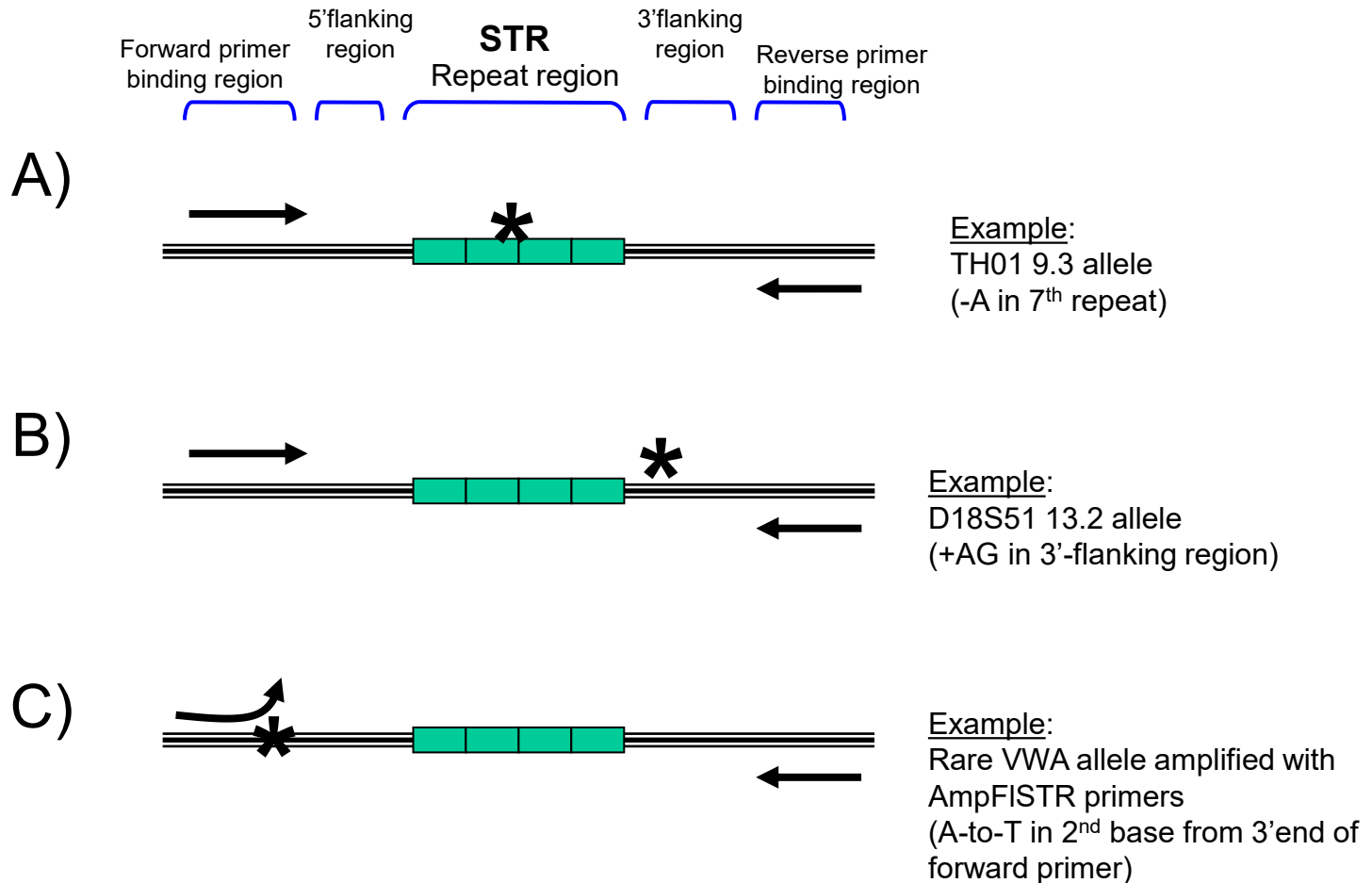
(b)



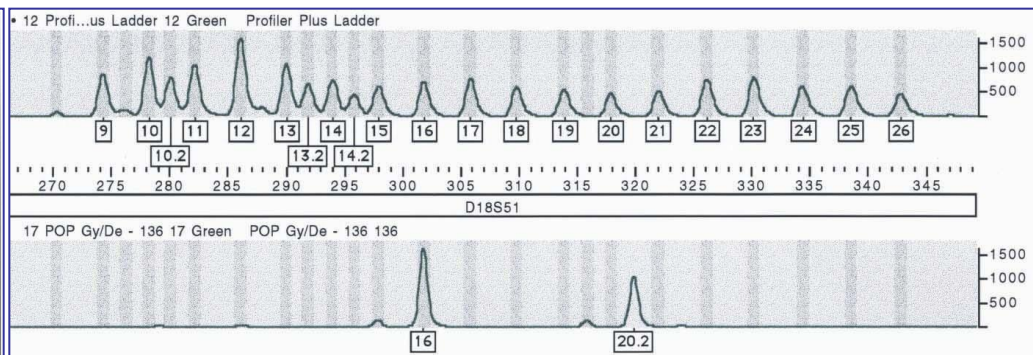
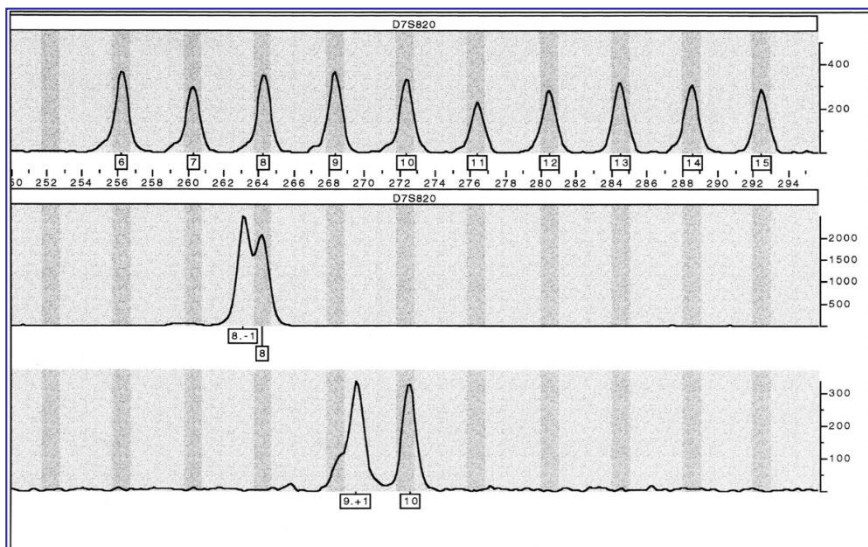
STR System	Maternal Meioses (%)	Paternal Meioses (%)	Number from either	Total Number of Mutations	Mutation Rate
CSF1PO	95/304,307 (0.03)	982/643,118 (0.15)	410	1,487/947,425	0.16%
FGA	205/408,230 (0.05)	2,210/692,776 (0.32)	710	3,125/1,101,006	0.28%
TH01	31/327,172 (0.009)	41/452,382 (0.009)	28	100/779,554	0.01%
TPOX	18/400,061 (0.004)	54/457,420 (0.012)	28	100/857,481	0.01%
VWA	184/564,398 (0.03)	1,482/873,547 (0.17)	814	2,480/1,437,945	0.17%
D3S1358	60/405,452 (0.015)	713/558,836 (0.13)	379	1,152/964,288	0.12%
D5S818	111/451,736 (0.025)	763/655,603 (0.12)	385	1,259/1,107,339	0.11%
D7S820	59/440,562 (0.013)	745/644,743 (0.12)	285	1,089/1,085,305	0.10%
D8S1179	96/409,869 (0.02)	779/489,968 (0.16)	364	1,239/899,837	0.14%
D13S317	192/482,136 (0.04)	881/621,146 (0.14)	485	1,558/1,103,282	0.14%
D16S539	129/467,774 (0.03)	540/494,465 (0.11)	372	1,041/962,239	0.11%
D18S51	186/296,244 (0.06)	1,094/494,098 (0.22)	466	1,746/790,342	0.22%
D21S11	464/435,388 (0.11)	732/526,708 (0.15)	580	1,816/962,096	0.19%
Penta D	12/18,701 (0.06)	21/22,501 (0.09)	24	57/41,202	0.14%
Penta E	29/44,311 (0.065)	75/55,719 (0.135)	59	163/100,030	0.16%
D2S1338	15/72,830 (0.021)	157/152,310 (0.10)	90	262/225,140	0.12%
D19S433	38/70,001 (0.05)	78/103,489 (0.075)	71	187/173,490	0.11%
SE33 (ACTBP2)	0/330 (<0.30)	330/51,610 (0.64)	None reported	330/51,940	0.64%

STR loci mutation rate: 10^{-3} - 10^{-4} / meiosis

Microsatellite point mutations

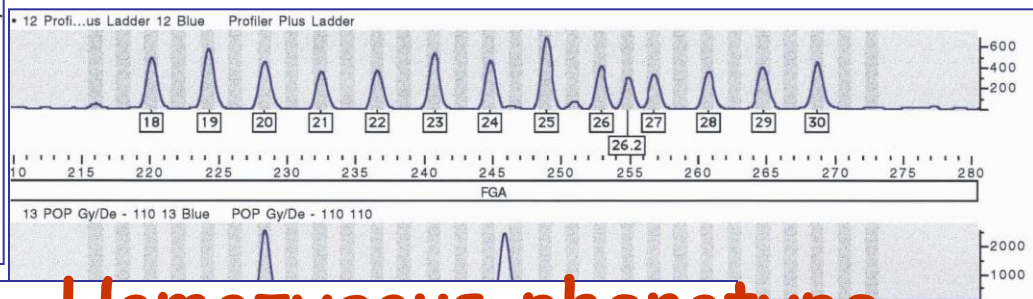


Variant microsatellite alleles: Null-alleles



Allél	Fragmenthossz	5' flanking régió	Repeat régió	3' flanking régió
16	299 bp	██████████	- (AGAA) ₁₆ -	AAAG AGAGAG - ██████████
20.2	317 bp	██████████	- (AGAA) ₂₁ -	AG AGAGAG - ██████████
15*	295 bp	██████████	- (ATAG) ₁₅ -	AAAG AGAGAG - ██████████

Allél	Fragments méret	5' Flanking régió	Repeat régió	3' Flanking régió
8.-1	205 bp	██████████	-(GATA) ₈ -	██████████ - (T) ₈ ATCT -
9.+1	211 bp	██████████	-(GATA) ₉ -	██████████ - (T) ₁₀ ATCT -
10	214 bp	██████████	-(GATA) ₁₀ -	██████████ - (T) ₈ AATCT -
12 (ref.)	222 bp	██████████	-(GATA) ₁₂ -	██████████ - (T) ₉ ATCT -
		24 bp		13 bp 124 bp



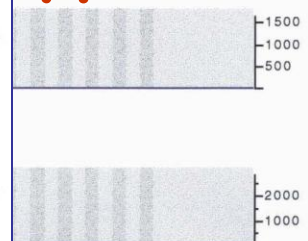
Homozygous phenotype

D13S317 allele 12

5'-gggttgctggacatggtatcACAGAAGTCTGGGATGTGGA---N82---(ATCT)₁₂ATCAATC(ATCT)₃TTCTGTCTGTCTTTTGGGC---N36---gaccaacaattcaagctctc-3'

D13S317 allele 7 (variant)

5'-gggttgctggacatggtatcACAGAAGTCTGGGATGTGGA---N82---(ATCT)₇ATCAATCAATC(ATCT)₃TTCTGTCTTTTGGGC---N36---gaccaacaattcaagctctc-3'



a,

b.

Jelölések: ■ $(F_{ST}, \Phi_{ST}) > 0,02; P < 0,1$ ■ $0,02 > (F_{ST}, \Phi_{ST}) > 0,01; 0,01 < P < 0,05$ □ $(F_{ST}, \Phi_{ST}) < 0,01; P > 0,05$

b,

Jelölések: $(F_{ST}, \Phi_{ST}) > 0,02; P < 0,1$ $0,02 > (F_{ST}, \Phi_{ST}) > 0,01; 0,01 < P < 0,05$ $(F_{ST}, \Phi_{ST}) < 0,01; P > 0,05$

Breed identification?

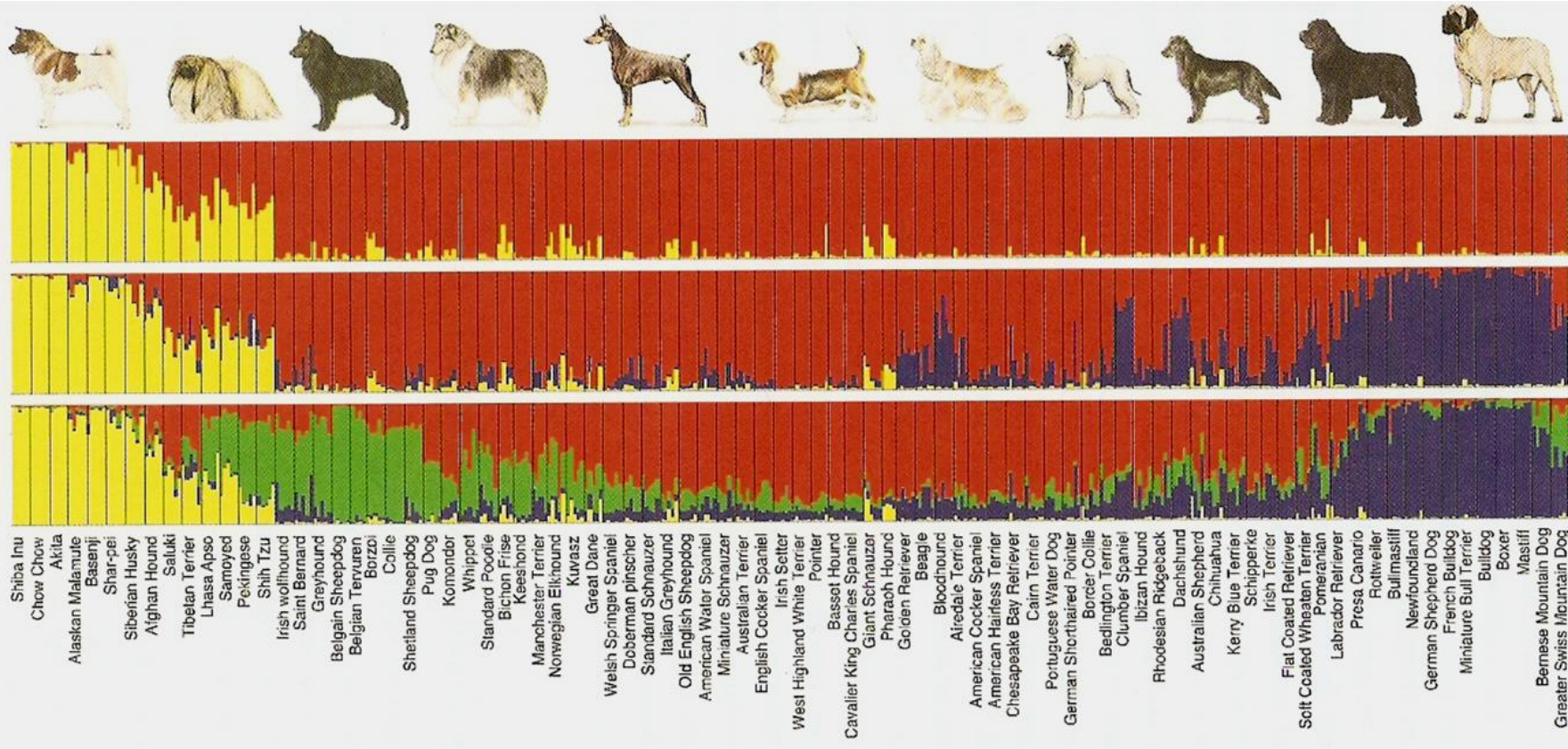
STRUCTURE statistics

1. Ancient

2. Sheperd

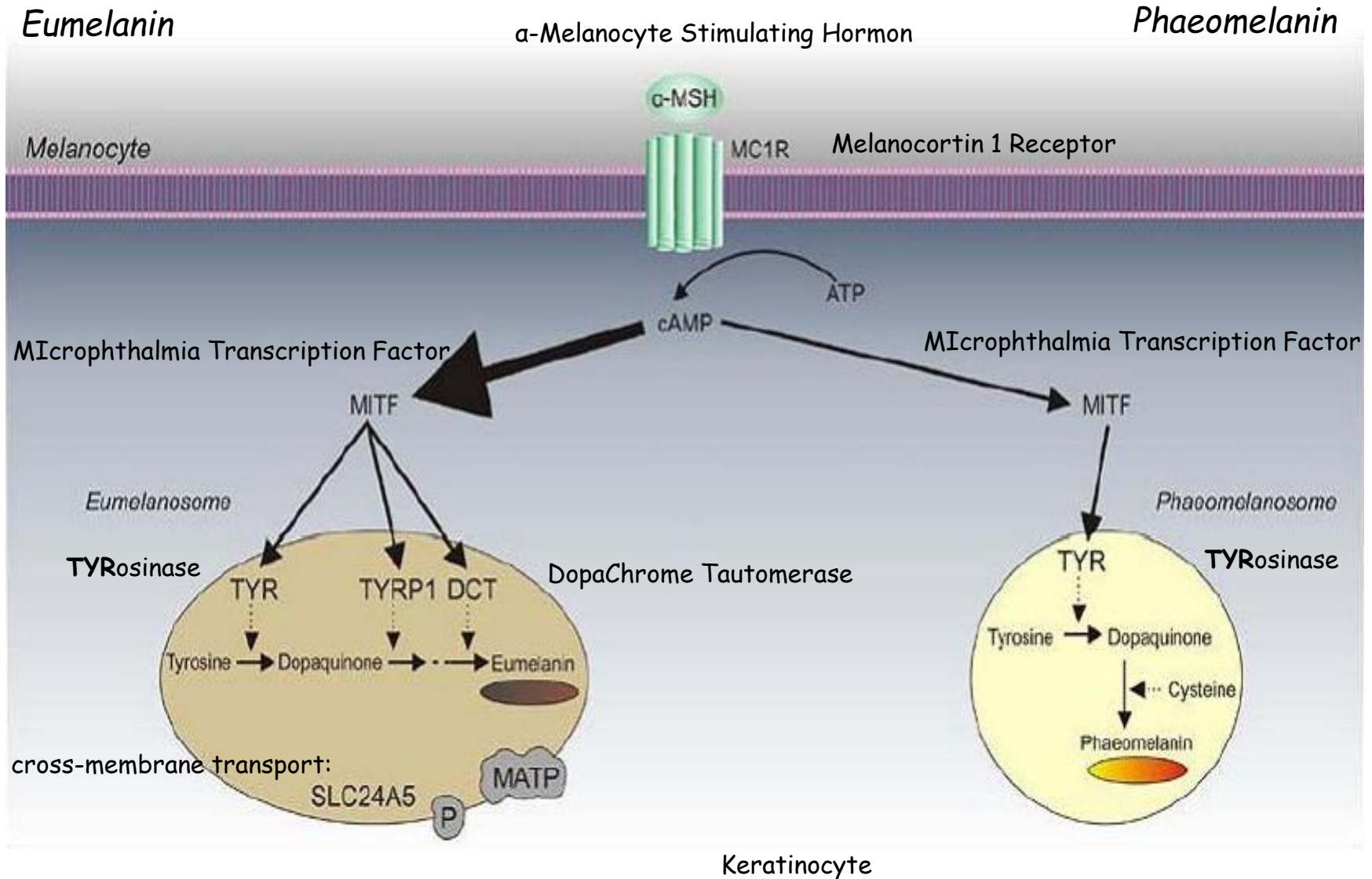
3. Hunting

4. Job



- 85 breeds (n=414)
- 95 microsatellites (dinucleotide repeat)
- 99% correct prediction!

Human melanogenesis



Genes responsible for skin pigmentation

Principal skin pigmentation candidate genes

Locus	Chromosome	Protein	Mut phenotype	Function
Melanosome proteins				
TYR	11q14-11q21	Tyrosinase	OCA1	Oxidation of tyrosine
TYRP1	9p23	Gp75, TRYP1	OCA3	DHICA-oxidase, TYR stabilisation
DCT	13q32	DCT, TRYP2		Dopachrome tautomerase
OCA2	15q11.2-15q12	P-protein	OCA2 (eye)	pH of melanosome
SLC45A2	5p14.3-5q12.3	MATP, AIM-1	OCA4 (skin)	Melansome maturation
SLC24A5	15q21.1	Cation exchanger		Melanosome precursor
Signal proteins				
ASIP	20q11.2-20q12	Agouti signal protein		MC1R antagonist
MC1R	16q24.3	MSH receptor	Red hair (skin)	G-protein coupled receptor
POMC	16q24.3	MSH receptor	Red hair	MC1R antagonist
OA1	Xp22.3	OA1 protein	OA1	G-protein coupled receptor
MITF	3p12.3-3p14.1	MITF	Waardenburg	Transcription factor
Proteins involved in melanosome transport or uptake by keratinocytes				
MYO5A	15q21	Myosin Va	Griscelli	Motor protein
RAB27A	15q15-15q21.1	Rab27a	Griscelli	RAS family protein
HPS1	10q23.1-10q23.3	HPS1	Hermansky-Pudlak	Organelle biogenesis and size
HPS6	10q24.32	HPS6	Hermansky-Pudlak	Organelle biogenesis

ACTH: adrenocorticotrophin hormone; DCT: dopachrome tautomerase; DHICA: 5,6-dihydroxyindole-2-carboxylic acid; MATP: membrane-associated transporter protein; MC1R: melanocortin-1 receptor; MITF: microphthalmia-associated transcription factor; MSH: melanocyte stimulating hormone; OCA: oculocutaneous albinism; POMC: pro-opiomelanocortin; TYRP1: tyrosinase-related protein 1.

MC1R gene mutations

Mutations in the MC1R gene, their penetrance and functional significance (where known)

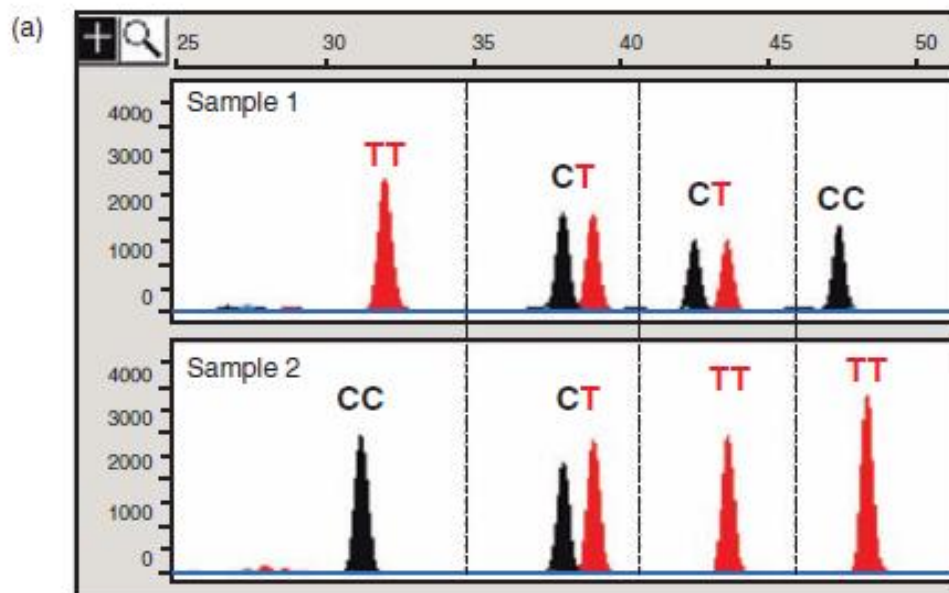
Mutation	Type	Designation	Penetrance (odds ratio)	Functional significance	References (for functional significance and penetrance)
R151C	Mis-sense	R	63.3	Altered cellular location	[16,26]
R160W	Mis-sense	R	63.3	Altered cellular location	[16,26]
D294H	Mis-sense	R	63.3	Impaired G coupling ability	[26,27]
D84E	Mis-sense	R	63.3	Altered cellular location	[16,26]
I155T	Mis-sense	Lack of statistical data—strong familial association		Altered cellular location	[16,26]
V92M	Mis-sense	r	5.1	Reduced α -MSH binding	[26,28,29]
V60L	Mis-sense	r	5.1		[26]
R163Q	Mis-sense	r	5.1	Slightly reduced α -MSH binding	[26,29]
R142H	Mis-sense	Lack of statistical data—strong familial association			[26]

- MC1R alleles possess different activity levels
- 317 AA and 7 transmembran domen
- SNPs: RHC phenotype - neanderthal pigmentation
- Phenotype prediction? Genetic tests?

SNaPshot: A Primer Extension Assay Capable of Multiplex Analysis

Minisequencing
(SNaPshot assay)

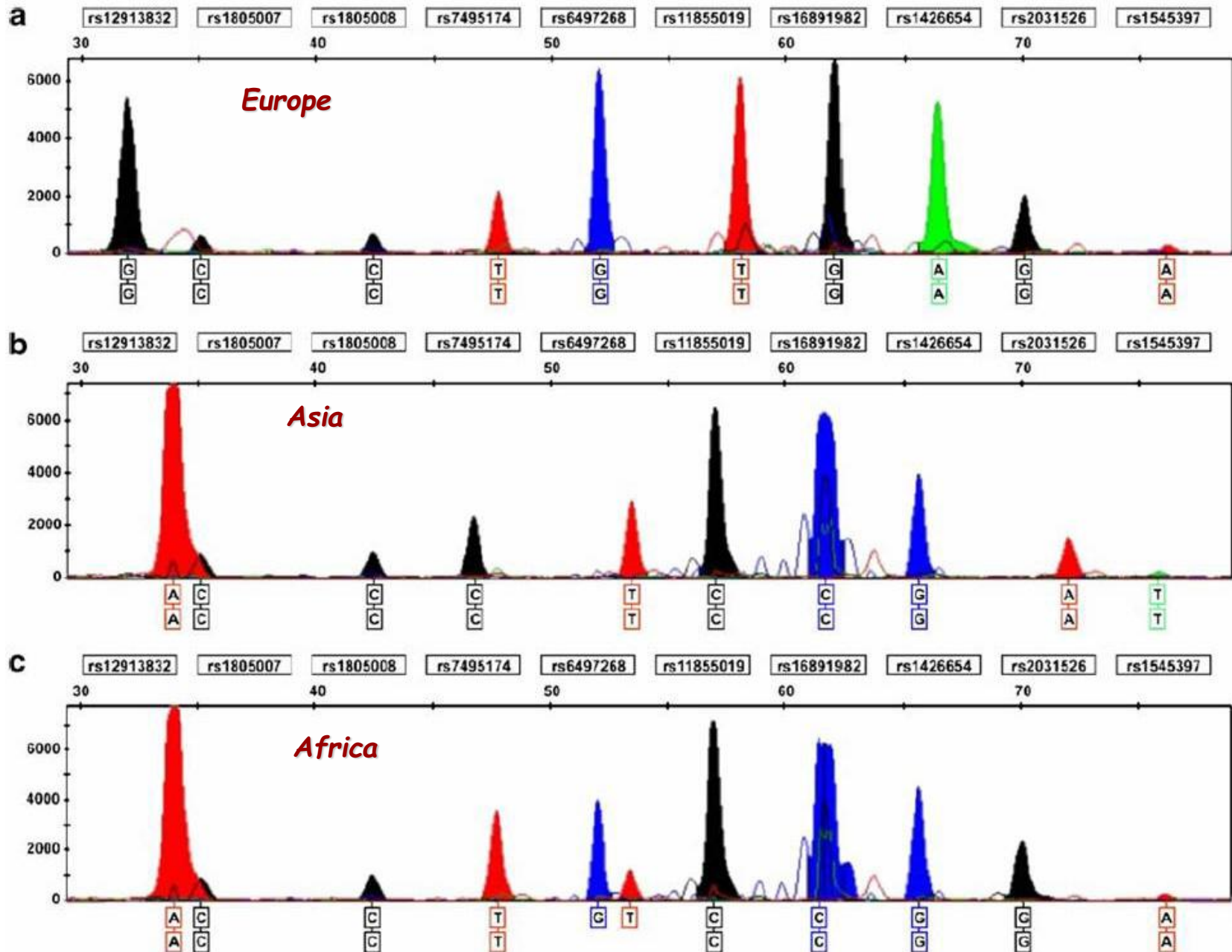
Allele-specific primer extension across the SNP site with fluorescently labeled ddNTPs; mobility modifying tails can be added to the 5'-end of each primer in order to spatially separate them during electrophoresis.



(b) (TTTTT)-**primer1** (chromosome 20)-**ddT/ddT**
 (TTTTT)-(TTTTT)-**primer2** (chromosome 6)-**ddC/ddT**
 (TTTTT)-(TTTTT)-(TTTTT)-**primer3** (chromosome 14)-**ddC/ddT**
 (TTTTT)-(TTTTT)-(TTTTT)-(TTTTT)-**primer4** (chromosome 1)-**ddC/ddC**

FIGURE 12.2 Allele-specific primer extension results using four autosomal SNP markers on two different samples (a). SNP loci are from separate chromosomes (1, 6, 14, and 20) and therefore unlinked. Electrophoretic resolution of the SNP primer extension products occurs due to poly(T) tails that are 5 nucleotides different from one another (b).

SNP genotyping of 10 pigmentation genes (SNaPshot)

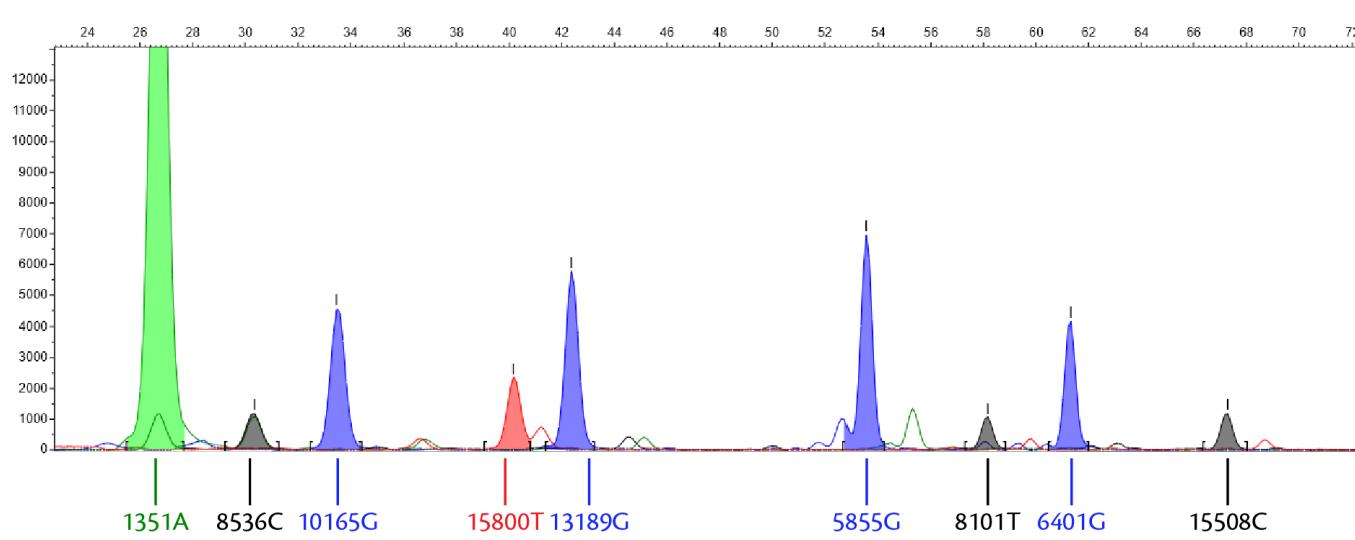


Sample	Self-reported pigmentary traits			rs12913832 HERC2	rs1805007 MC1R	rs1805008 MC1R	OCA2 diplotype ^a	rs16891982 SLC24A2	rs1426654 SLC24A5	rs2031526 DCT	rs1545397 OCA2	Inferred ancestry of individuals ^b		
	Eye color	Hair color	Skin color									European	Asian	African
E1	Blue	Red	Fair	<u>G/G</u>	C/C	C/T	<u>TGT/TGT</u>	G/G	A/A	G/G	A/A	0.963	0.012	0.024
E2	Green	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	A/G	A/A	0.954	0.021	0.025
E3	Blue	Blond	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	A/G	A/A	0.954	0.024	0.022
E4	Blue	Blond	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	A/G	A/A	0.960	0.020	0.020
E5	Blue/gray	Auburn	Fair	<u>G/G</u>	C/T	C/C	<u>TGT/TGT</u>	G/G	A/A	G/G	A/A	0.961	0.013	0.026
E6	Green/gray	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	C/G	A/A	G/G	A/A	0.787	0.038	0.175
E7	Green/hazel	Light brown	Fair	A/G	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	A/G	A/A	0.955	0.022	0.024
E8	Green/hazel	Dark brown	Fair	A/A	C/C	C/C	TGT/CTC	G/G	A/A	G/G	A/A	0.961	0.013	0.027
E9	Green/hazel	Dark brown	Fair	A/A	C/C	C/C	TTT/CTC	G/G	A/A	G/G	A/A	0.963	0.013	0.024
E10	Blue	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	C/G	A/A	G/G	A/A	0.789	0.049	0.163
E11	Green	Auburn	Fair	<u>G/G</u>	C/T	C/C	TGT/TGC	G/G	A/A	G/G	A/A	0.958	0.014	0.028
E12	Blue/hazel	Light brown	Fair	A/G	C/C	C/C	TGT/TTT	G/G	A/A	G/G	A/A	0.962	0.012	0.026
E13	Blue/hazel	Light brown	Fair	A/G	C/C	C/C	TGT/TTT	G/G	A/A	G/G	A/A	0.965	0.013	0.022
E14	Green	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	C/G	A/A	G/G	A/T	0.763	0.165	0.073
E15	Brown	Dark brown	Fair	A/G	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	A/G	A/A	0.957	0.022	0.021
E16	Brown	Dark brown	Fair	A/A	C/C	C/C	TGT/CTC	C/G	A/A	A/G	A/T	0.669	0.283	0.048
E17	Green/hazel	Dark brown	Medium	A/G	C/C	C/C	TGT/TTT	C/G	A/A	G/G	A/T	0.755	0.170	0.076
E18	Blue	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	G/G	A/T	0.935	0.045	0.021
E19	Brown	Red	Fair	A/G	C/T	C/C	<u>TGT/TGT</u>	G/G	A/A	G/G	A/A	0.964	0.013	0.022
E20	Green	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	C/G	A/A	G/G	A/A	0.792	0.047	0.161
E21	Green/gray	Blond	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	A/G	A/A	0.957	0.022	0.021
E22	Blue	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	G/G	A/A	0.959	0.014	0.026
E23	Green/hazel	Light brown	Fair	A/G	C/C	C/C	TGT/TTT	G/G	A/A	A/G	A/A	0.957	0.020	0.022
E24	Green	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	C/G	A/A	G/G	A/A	0.786	0.049	0.166
E25	Brown	Red	Fair	A/G	C/C	T/T	TGT/TGC	G/G	A/A	G/G	A/A	0.963	0.014	0.023
E26	Blue	Light brown	Fair	<u>G/G</u>	C/C	C/C	<u>TGT/TGT</u>	G/G	A/A	A/G	A/A	0.954	0.021	0.025
E27	Blue	Red	Fair	<u>G/G</u>	C/C	C/T	<u>TGT/TGT</u>	G/G	A/A	G/G	A/A	0.958	0.014	0.028
Af1	Brown	Black	Dark	A/A	C/C	C/C	TGC/TTC	C/C	G/G	A/G	A/A	0.028	0.094	0.878
Af2	Brown	Black	Dark	A/A	C/C	C/C	TGC/TTC	C/C	G/G	G/G	A/A	0.023	0.031	0.946
Af3	Brown	Black	Dark	A/A	C/C	C/C	TGC/TTC	C/C	A/G	G/G	A/A	0.164	0.041	0.795
As1	–	–	–	A/A	C/C	C/C	TTT/CTC	C/C	G/G	A/G	A/T	0.042	0.649	0.308
As2	–	–	–	A/A	C/C	C/C	CTC/CTC	C/C	G/G	A/G	T/T	0.020	0.921	0.060
As3	–	–	–	A/A	C/C	C/C	CTC/CTC	C/C	G/G	A/A	T/T	0.013	0.964	0.023
As4	–	–	–	A/G	C/C	C/C	TTT/CGC	C/C	A/G	A/A	A/T	0.212	0.708	0.080
As5	–	–	–	A/A	C/C	C/C	TTC/CGC	C/C	G/G	A/G	T/T	0.019	0.922	0.059
As6	–	–	–	A/A	C/C	C/C	CTC/CTC	C/G	G/G	A/A	T/T	0.119	0.858	0.023

E European modern sample, Af African modern sample, As Asian modern sample

^a OCA2 diplotypes correspond to markers rs7495174/rs6497268/rs11855019. OCA2 diplotypes and rs12913832 genotype predictive of blue eye color phenotype are underlined

^b Probability of being from European/Asian/African population determined using the STRUCTURE program. The greatest probability, most likely estimate of ancestry, is indicated in bold



**Rapid identification of
canine mitochondrial DNA
haplotypes and haplogroups**

based on 10-loci SNaPshot

Sample code	Identified peaks (positions and length of fragments)									Haplogroup
	1351	8536	10165	15800	13189	5855	8101	6401	15508	
	27	30	34	39	42	54	58	61	67	
Kim reference	A	C	G	T	G	G	C	G	C	A1a1
77	A	C	G	C	G	G	C	G	T	C
111	A	C	G	C	G	G	T	G	C	B1a
150	A	C	G	T	G	G	C	G	C	A1a1
198	A	C	G	C	G	G	T	G	C	B1a
202	G	C	G	T	G	G	C	A	C	A1b1a
236	G	C	G	T	G	G	C	A	C	A1b1a
240	A	T	A	T	G	G	C	G	C	A1a1b
286	A	C	G	T	A	G	C	G	C	A1a1a3
298	A	T	A	T	G	G	C	G	C	A1a1b
587	A	C	G	T	G	A	C	G	C	A1b2
610	A	C	G	C	G	G	T	G	C	B1a
655	A	C	G	T	G	G	C	G	C	A1a1

QUESTIONS ?

