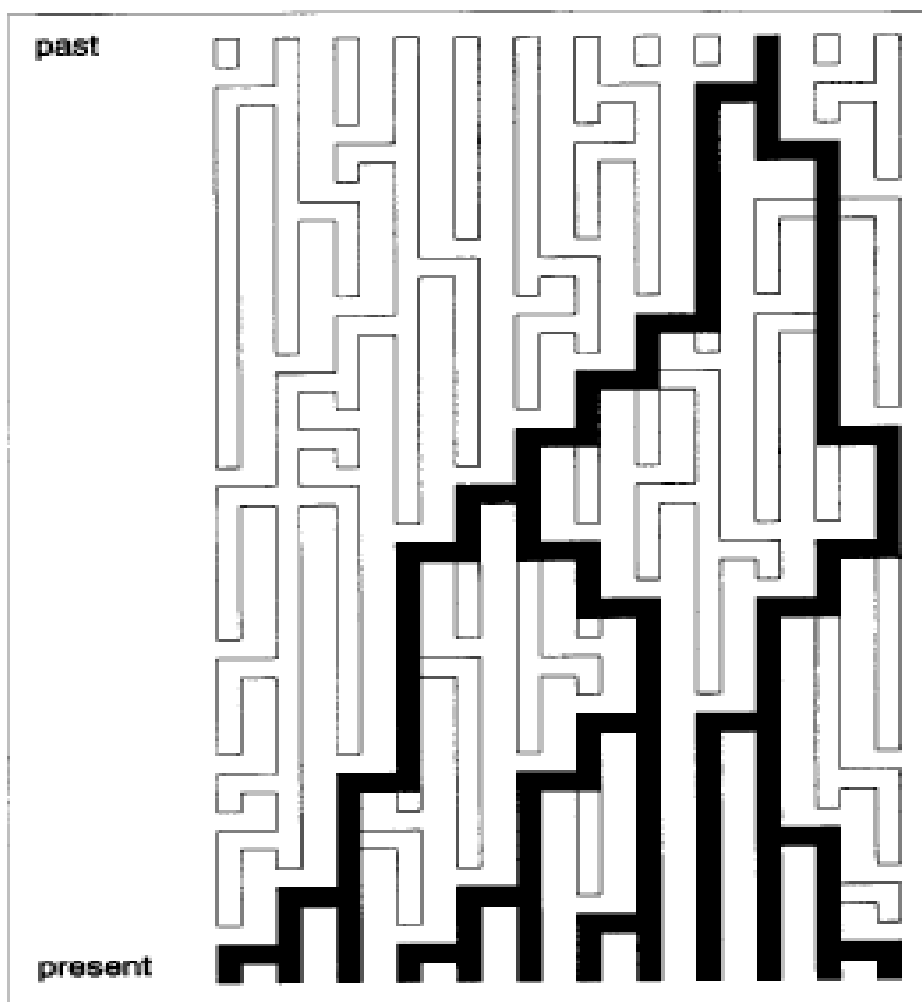


# *Humán evolúció- és populációgenetika*

## *- a magyar népesség populációgenetikája*



<https://genetics.elte.hu>

user/pw: genetika2016

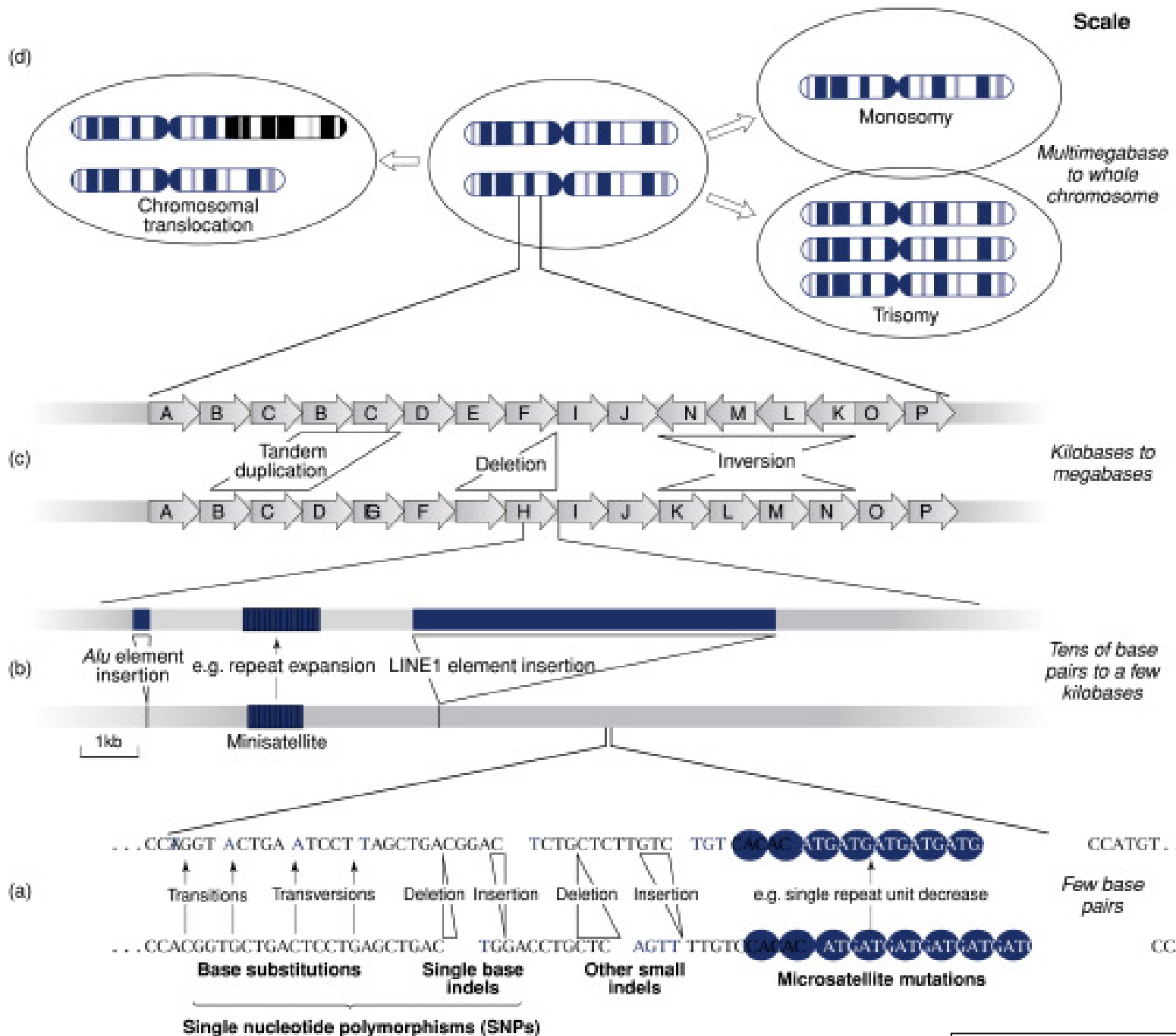
*Egyed Balázs*

*ELTE TTK Genetikai Tanszék*

*begyed@caesar.elte.hu*

## *Genetikai variációk, hasonlóságok és különbségek*

- Az egyének között megfigyelt genetikai variációk tanulmányozása felhasználható-e az egyénekből álló csoportok elkülönítésére?
- Megtudhatunk-e valamit az emberi népeiséget alkotó csoportok (populációk) egymás közötti kapcsolatáról?
- Hogyan dönthető el a genetikai variációk vizsgálata alapján, hogy a humán populációk egymással milyen szintű rokonságban vannak?
- A populációk genetikai rokonsága milyen járulékos „következménnyel” jár?
- Meg tudjuk-e határozni az egyes egyénről és/vagy csoportról, hogy ugyanazt a leszármazási vonalat képviselik, vagy ugyanahhoz a genetikailag definiált csoporthoz tartoznak? (population history)



# *AB0 vércsoport allélgyakoriság*

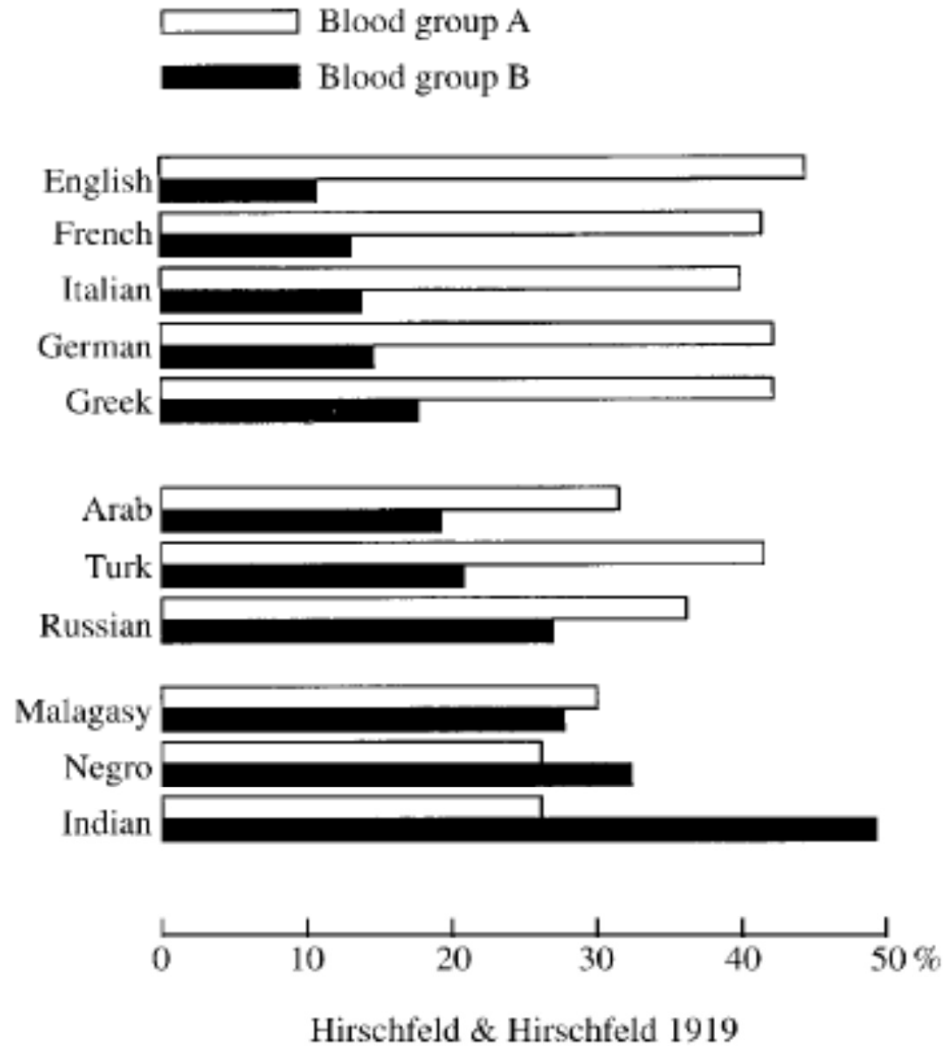


Figure 1. Frequencies of blood groups A and B from different ethnic groups. Data from Hirschfeld & Hirschfeld (1919).

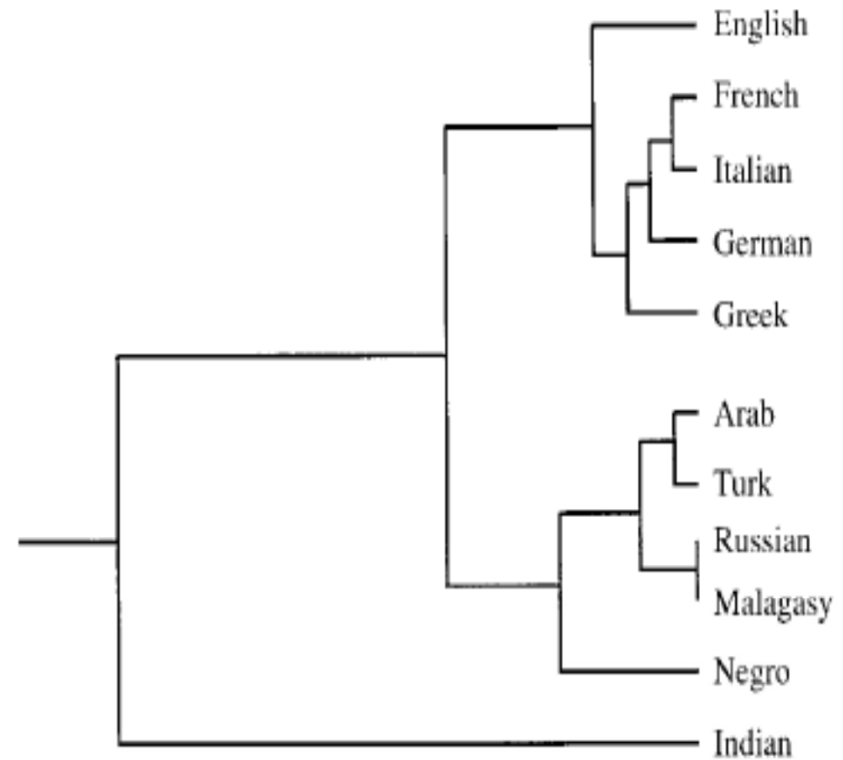


Figure 2. UPGMA diagram constructed from blood group B data from figure 1.



## *Multi-lokusz allélfrekvencia adatok*

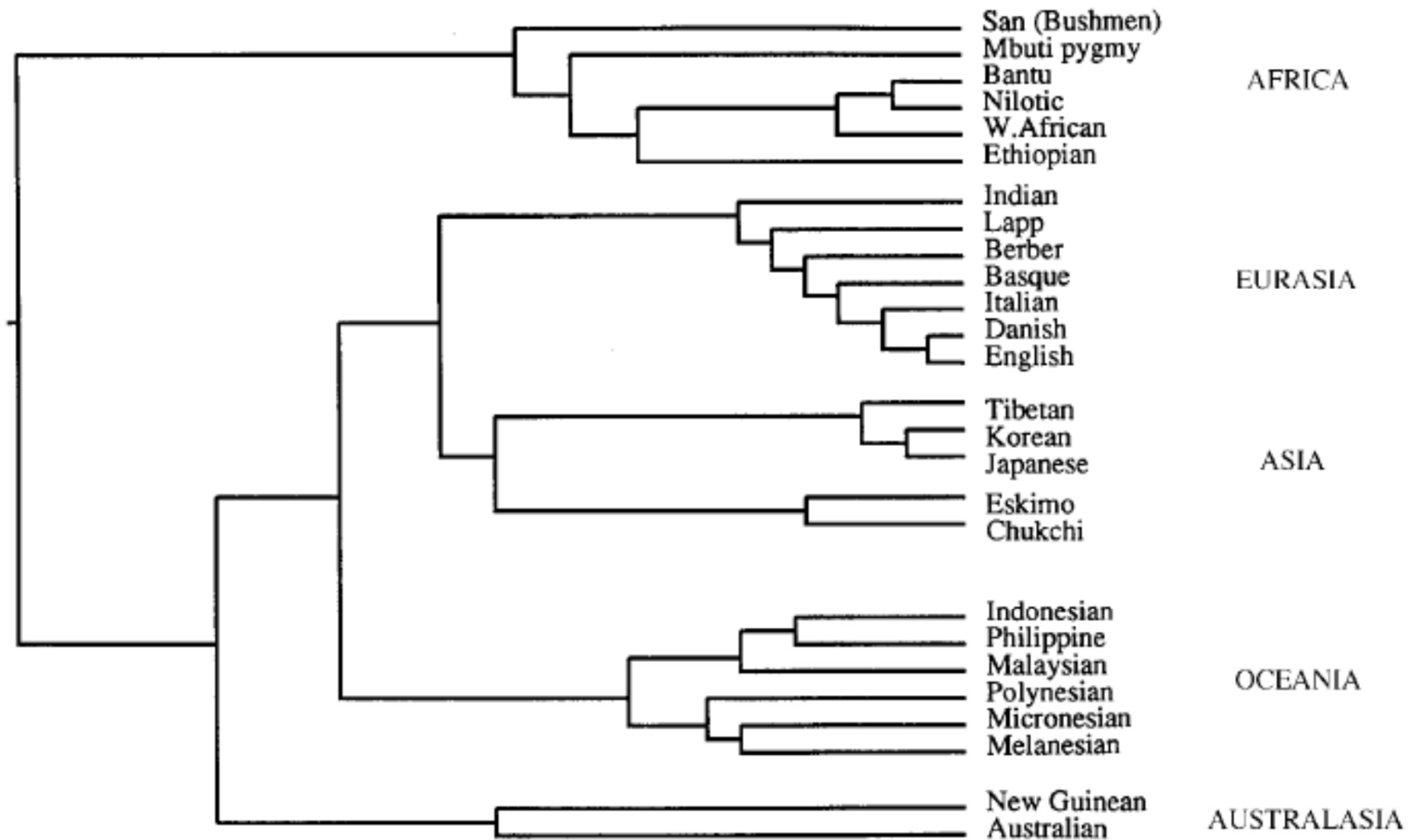


Figure 3. Genetic tree of worldwide populations (redrawn from Cavalli-Sforza *et al.* (1994)).

*Admixture*

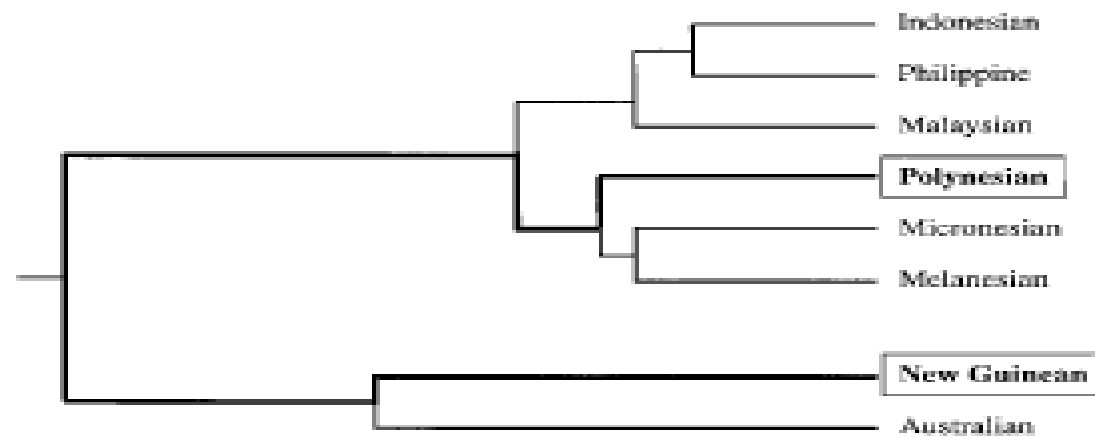
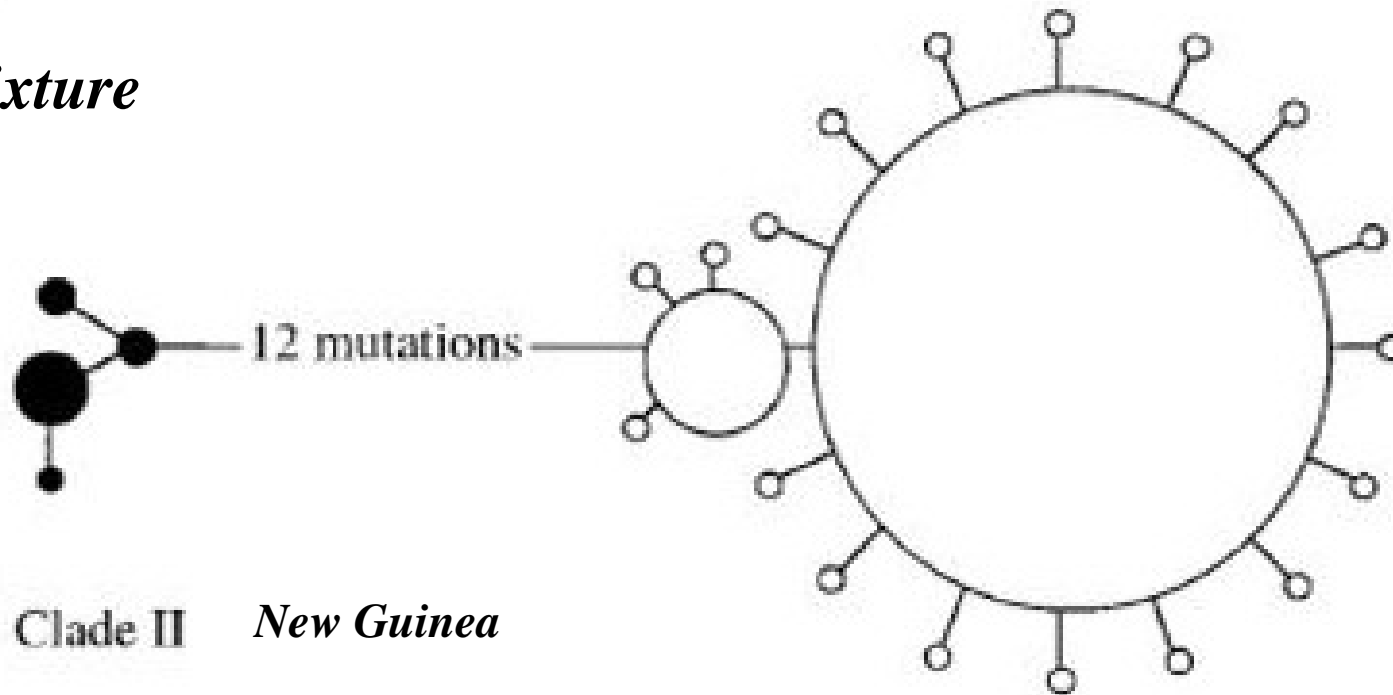
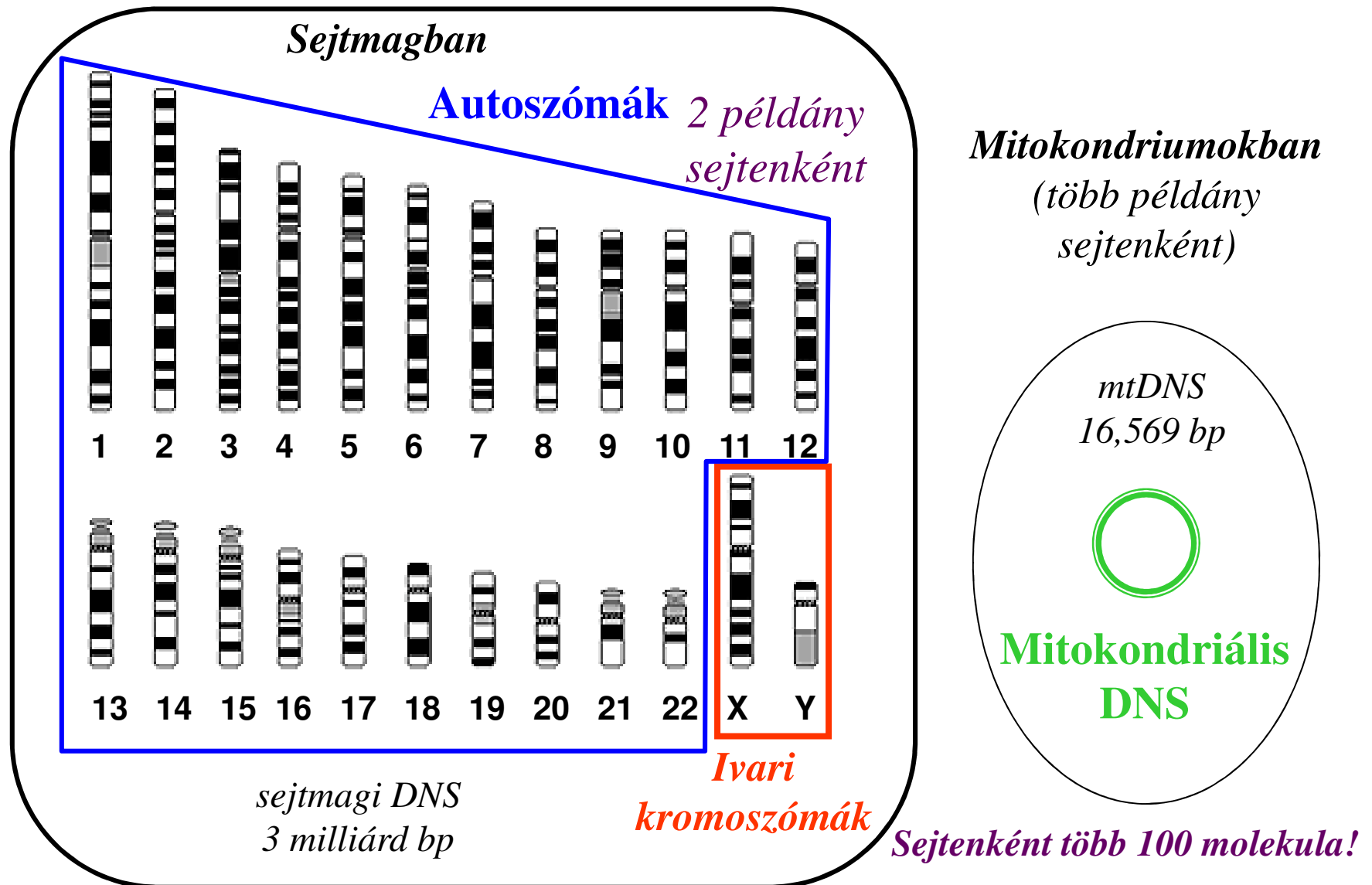


Figure 5. The positions of Polynesian and New Guinean populations on the genetic tree from figure 3.

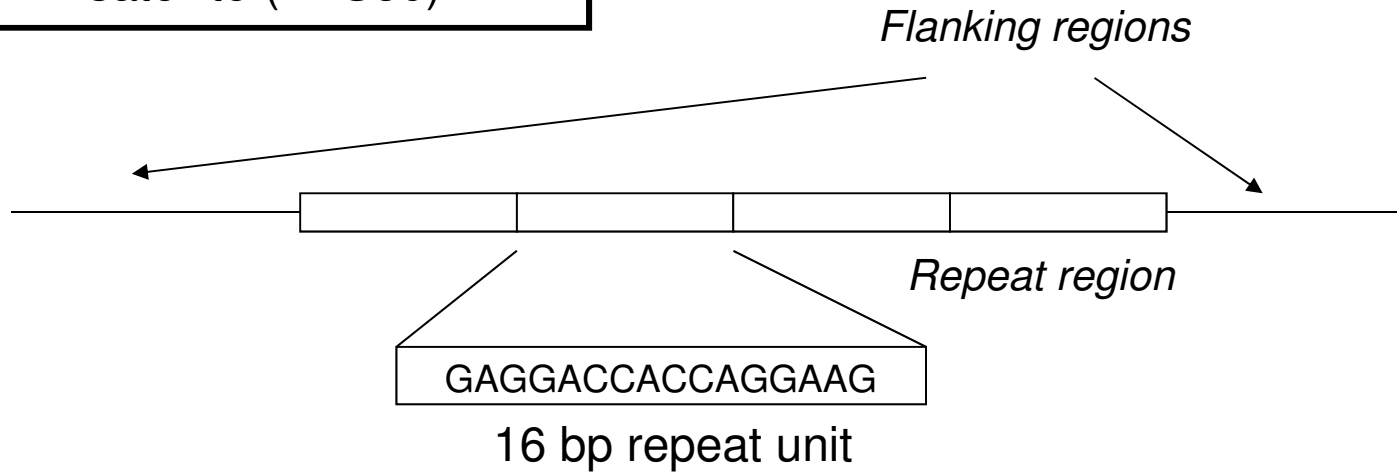
# Az emberi DNS

23 pár kromoszóma + mtDNS



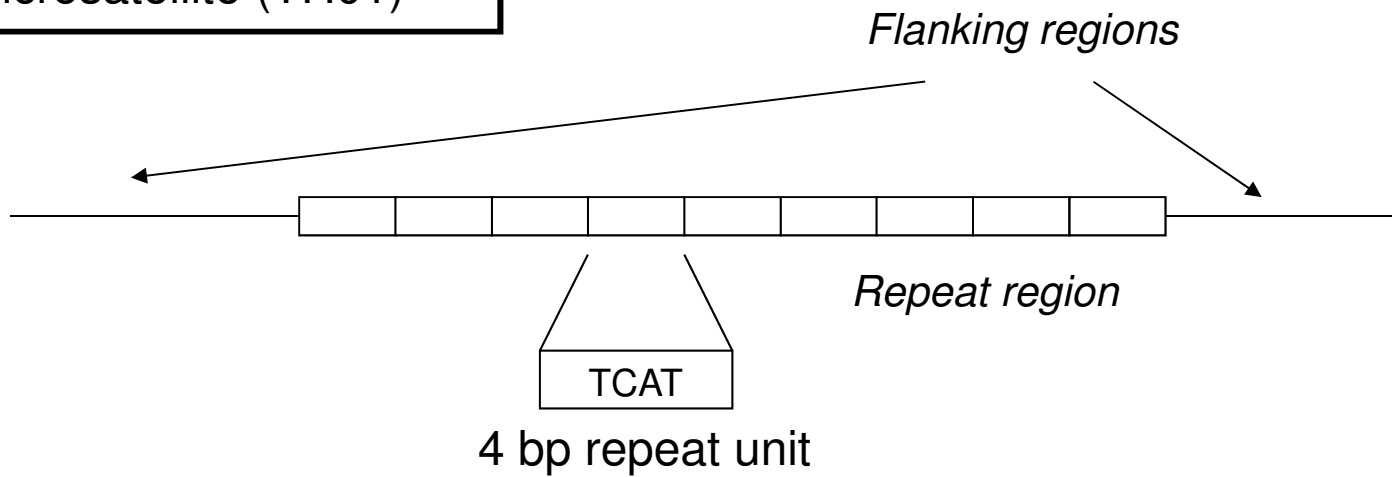
Minisatellite (D1S80)

VNTRs



Microsatellite (TH01)

STRs (SSRs)

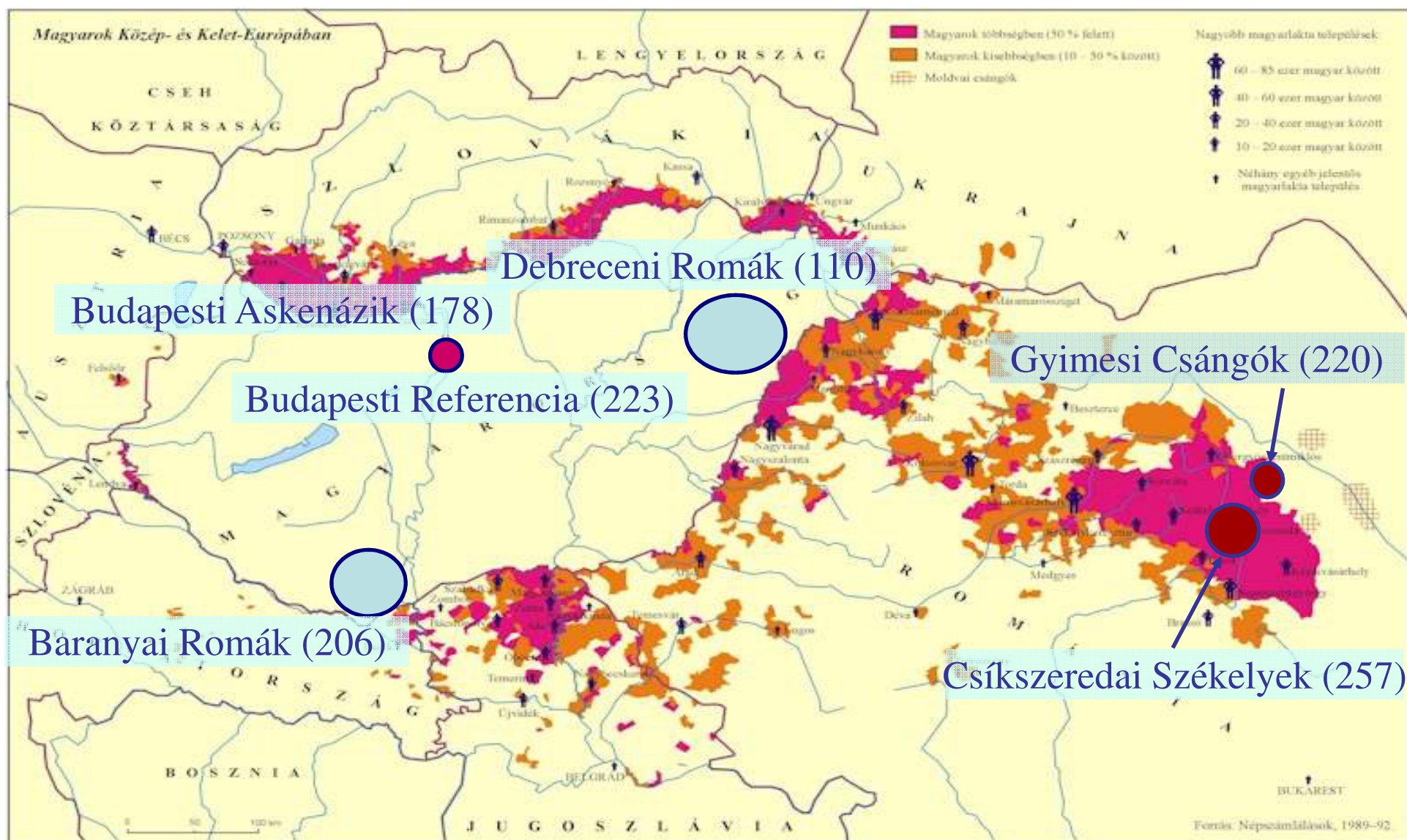




## *A hazai népesség populációgenetikai felmérése*

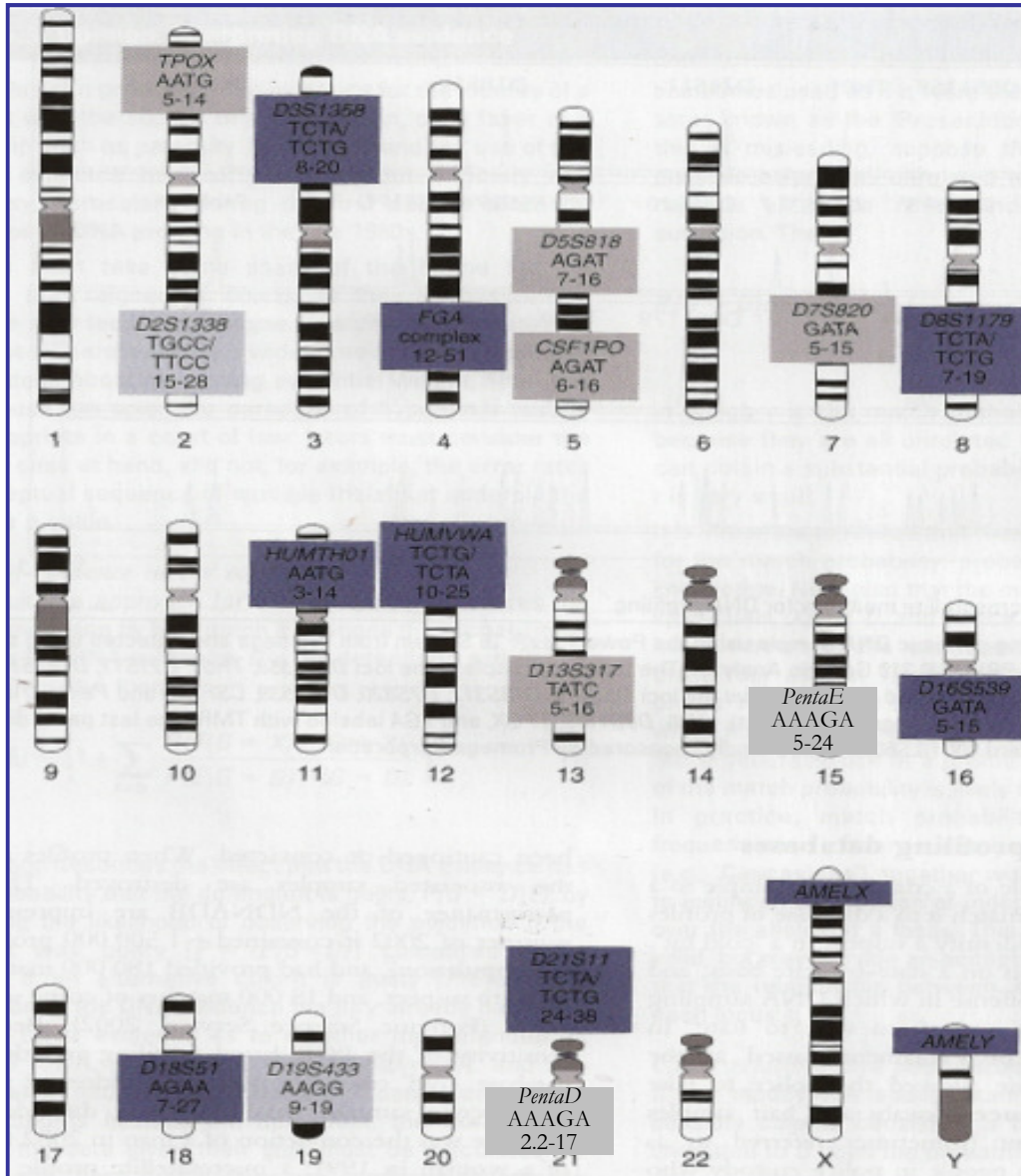
- Humán multilókuszos autoszómás és Y-kromoszómás STR-profilok, valamint mitokondriális DNS kontroll régió haplotípusok generálása és eloszlásának analízise a magyar populációban.
- A magyar népesség genetikai heterogenitásának felmérése 17 autoszómás és Y-kromoszómás mikroszatellita marker és a mitokondriális DNS kontroll régió polimorfizmusai alapján.
- Magyar populációs referencia adatbázis(ok) generálása genetikai profilok populációstatisztikai becsléséhez.
- A megfigyelt allél- és profilgyakoriságok összevetése egyéb európai populációs adatokkal, a belső és külső genetikai struktúráltság megállapítása.

# *A magyar népesség heterogenitásának felmérésébe bevont populációs minták*



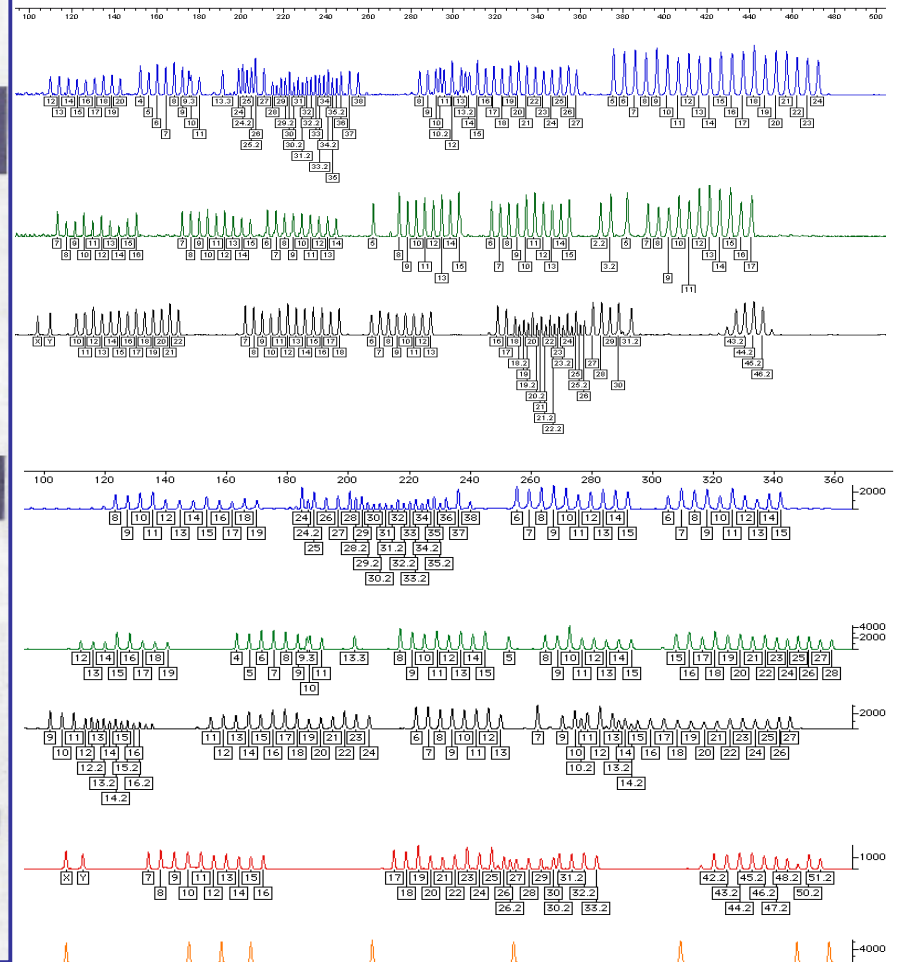


# Populációs felmérésbe bevont autoszómás mikroszatelliták



15 tetramer, 2 pentamer STR

Multiplex PCR és fluoreszcens detektálás

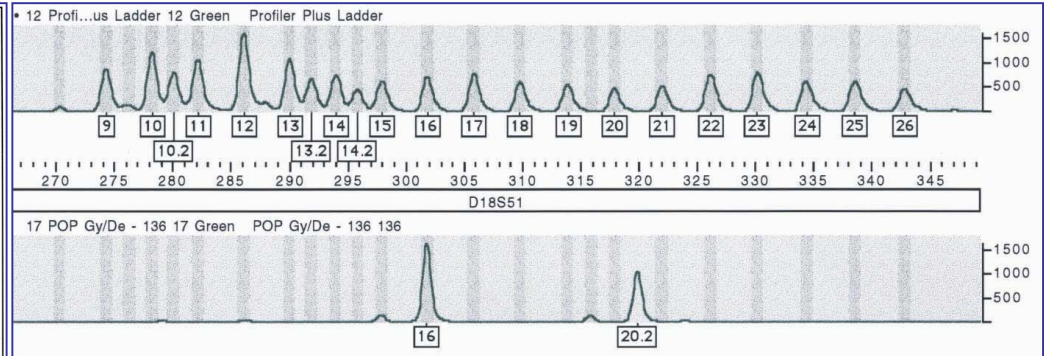
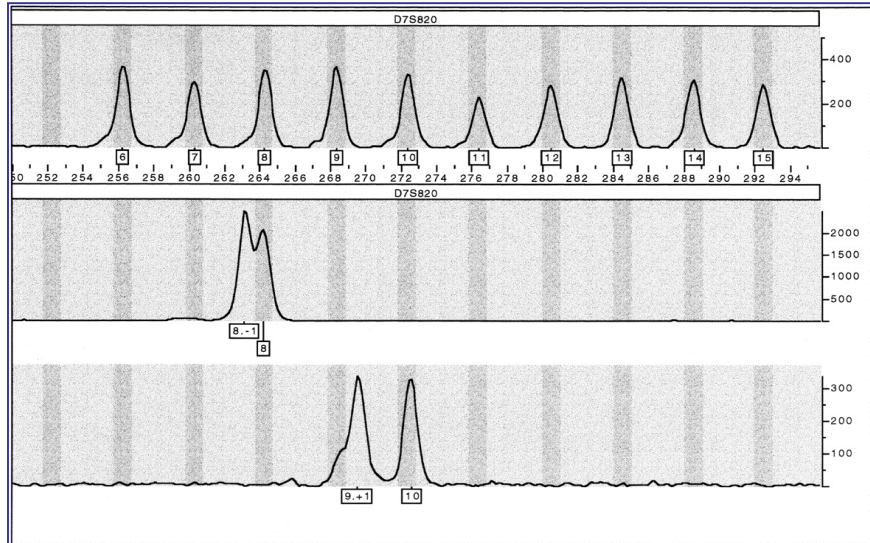




# *Autszómás STR lokuszok allél- és profiltáblája*

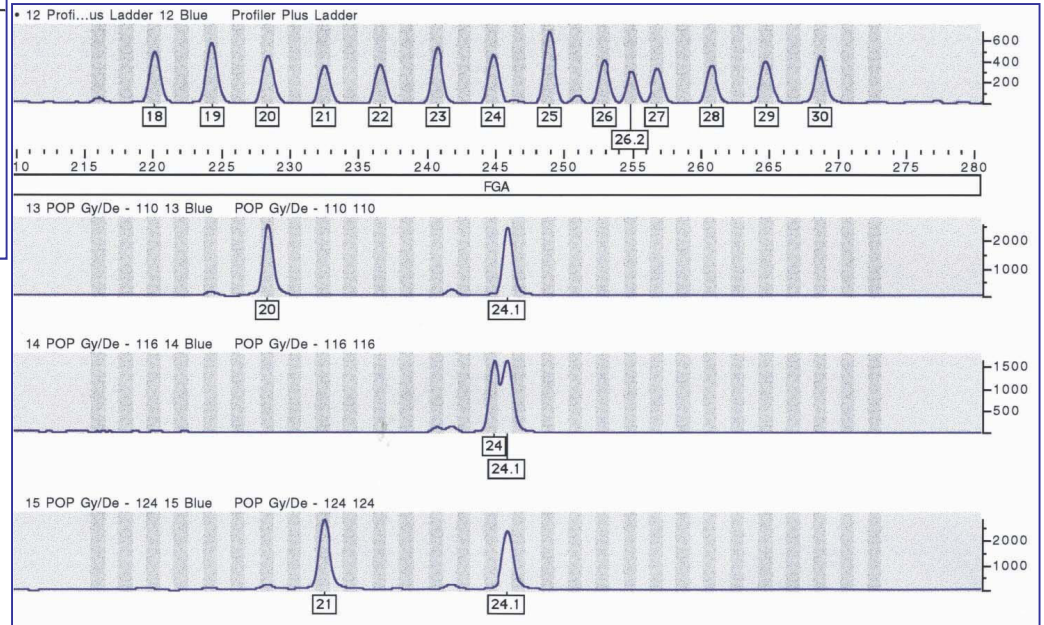
Minta	AMEL	AMEL	D8S11D8S11D21S1D21S1D7S82D7S82	CSF1	CSF1	FD3S13D3S13	TH01	TH01	D13S2	D13S2	D16S5D16S5	vWA	vWA	TPOX	TPOX	D18S5D18S5	D5S81D5S81	FGA	FGA	21338	11338	23433	13433	2	Penta	Penta	Penta	Penta								
BuCa-001	X	X	14	14	28	30,2	11	11	10	10	16	16	7	9	12	12	12	14	14	17	8	11	13	14	11	12	20	22	17	20	13	15,2	12	17	10	12
BuCa-002	X	Y	12	14	29	30	8	11	11	12	15	18	9	9,3	8	11	13	13	16	17	8	10	14	17	12	12	19	24	18	25	13	14	11	13	9	9
BuCa-003	X	Y	13	14	28	29	11	11	10	12	16	18	6	9	10	11	13	13	16	18	8	8	14	15	11	13	21	22	24	13	16	5	15	11	14	
BuCa-004	X	Y	13	13	32,2	32,2	11	12	12	13	16	17	7	9,3	8	13	9	14	17	18	8	9	12	14	12	12	22	23	18	19	13	14	7	12	9	13
BuCa-005	X	Y	13	13	28	30	8	11	11	12	16	18	6	9	8	11	12	13	16	16	8	8	13	18	11	13	20	26	17	25	12	13	7	12	9	15
BuCa-006	X	Y	13	14	28	30	8	11	11	12	14	17	9	9,3	10	12	8	10	14	15	8	8	12	18	9	13	20	25	17	17	12	14	13	13	12	13
BuCa-007	X	X	10	13	30	30	8	11	11	12	15	16	6	7	11	11	11	15	17	8	8	12	18	11	12	21	22	17	24	14	15	14	18	12	13	
BuCa-008	X	Y	12	14	29	31,2	9	10	9	10	16	19	6	9	12	12	11	12	16	18	9	12	11	16	11	13	21	22	25	13,2	15	10	10	9	10	
BuCa-009	X	Y	13	14	28	29	10	12	10	12	15	17	9	9,3	8	9	9	13	15	18	8	12	13	14	12	13	20	23	17	23	14	14	11	12	12	13
BuCa-010	X	Y	14	14	27	31	11	12	13	13	16	18	6	8	9	11	10	12	16	19	8	11	12	21	11	12	23	24	20	25	14	14	5	13	9	11
BuCa-011	X	Y	14	14	28	30	10	11	11	11	15	18	6	9,3	11	11	11	13	16	17	8	8	15	17	11	13	20	23	19	24	13	14	7	7	11	12
BuCa-012	X	X	12	14	28	32,2	8	10	11	12	17	18	6	9,3	11	12	11	12	14	16	8	11	17	17	10	13	20	23	17	24	14	14	7	16	10	10
BuCa-013	X	Y	12	14	30	31	10	10	12	12	17	18	7	9,3	11	12	9	13	14	19	8	8	13	20	12	12	22	25	20	23	14	14	10	10	9	12
BuCa-014	X	Y	13	14	27	32	10	10	9	9	16	17	6	9	10	14	12	13	16	19	8	11	12	20	12	12	21	22	19	20	14	15	7	10	12	13
BuCa-015	X	Y	13	15	30	33,2	8	10	11	12	15	17	8	9	11	12	11	13	14	16	8	8	18	20	11	13	21	24	20	21	14	16	13	13	8	9
BuCa-016	X	Y	11	15	27	28	8	11	10	10	16	17	7	9,3	9	12	11	12	15	18	8	8	17	19	10	11	22	22	17	20	14	14	5	12	11	12
BuCa-017	X	Y	11	13	28	30	9	10	10	12	16	18	8	9	13	13	11	13	14	20	8	8	15	16	11	13	20	21	25	26	13	14	7	16	10	11
BuCa-018	X	Y	14	14	30	33,2	10	12	10	12	17	18	6	9,3	12	14	9	11	14	16	8	8	13	20	10	12	23	24	19	20	13	14	7	10	12	13
BuCa-019	X	Y	10	12	28	32,2	10	11	10	11	17	17	9	9,3	11	11	12	13	15	17	8	8	10	15	9	13	19	23	17	25	13	14	12	14	11	13
BuCa-020	X	X	12	15	29	29	10	11	12	12	13	16	9	9	9	11	8	12	16	17	8	8	19	19	12	12	20	22	23	23	15	15	14	16	9	13
BuCa-021	X	X	13	16	28	32,2	10	10	10	11	15	17	8	9,3	11	11	10	11	17	19	11	12	17	18	11	12	22	24	17	20	14	14	7	10	9	12
BuCa-022	X	Y	12	14	28	30,2	10	11	10	13	15	15	7	9,3	12	12	9	12	17	17	8	10	15	16	12	13	19	20	24	24	14	15	5	13	9	13
BuCa-023	X	X	9	14	28	28	8	10	11	11	14	17	7	9	12	13	11	12	16	17	8	12	14	15	12	12	20	21	19	20	15	15	7	13	9	12
BuCa-024	X	Y	13	13	29	29	10	10	11	12	14	17	9,3	9,3	8	8	10	11	17	19	8	11	15	19	12	12	20	21	19	20	12	13	5	7	13	15
BuCa-025	X	X	14	16	30	31,2	8	13	10	13	15	16	6	9	12	12	9	13	17	18	8	8	17	20	12	12	20	21	18	22	13	15	11	13	9	12
BuCa-026	X	Y	11	14	30	32,2	8	12	10	11	15	19	6	9	9	12	11	12	14	19	8	8	12	13	11	13	23	24	17	25	14	15	7	7	12	13
BuCa-027	X	X	12	13	31	31,2	8	8	10	10	16	18	7	9,3	8	8	11	13	16	17	8	9	16	18	12	13	21	22	16	20	12	15	7	10	11	16
BuCa-028	X	X	13	14	29	33,2	8	10	10	12	14	17	9	9,3	11	13	11	13	16	17	8	11	15	22	11	12	20	21	17	23	12	14	5	6	9	10
BuCa-029	X	Y	12	13	27	32,2	8	12	10	11	14	15	9	9,3	11	12	11	13	17	17	8	9	12	14	10	12	19	21	26	26	14	15	12	17	11	12
BuCa-030	X	X	13	15	29	29	8	11	11	11	14	16	9,3	9,3	11	11	11	12	17	18	8	8	12	14	11	12	23	24	20	20	13	15	10	12	10	13
BuCa-031	X	X	10	13	30	31,2	10	10	11	11	14	15	6	9	8	12	11	13	17	19	9	9	14	18	11	13	21	23	17	17	13	14	12	16	11	13
BuCa-032	X	Y	12	13	28	29	8	11	10	11	16	17	7	8	11	12	9	12	15	15	8	9	12	14	11	12	19	20	20	23	15	15	7	17	9	13
BuCa-033	X	X	12	15	32,2	32,2	8	9	10	12	15	17	9	9,3	11	13	11	13	15	17	9	11	17	18	11	12	21	21	17	20	13	14,2	5	5	9	12
BuCa-034	X	X	12	13	30,2	32,2	9	10	10	13	14	17	9	9,3	11	12	12	12	14	18	8	11	14	18	11	12	20	22	17	19	15	15,2	16	17	11	12
BuCa-035	X	X	10	12	28	28	9	13	10	10	15	16	8	8	10	11	12	13	17	18	8	10	18	18	10	12	19	22,2	17	19	12	13	7	13	9	11
BuCa-036	X	X	13	17	30	32,2	9	11	11	11	14	15	9	9,3	11	13	9	11	16	16	8	12	13	17	12	12	21	22	17	23	12	14	7	17	12	13
BuCa-037	X	X	14	15	28	29	10	11	12	14	16	17	7	9	8	9	12	14	16	17	8	11	16	17	10	11	21	21	23	25	13	14	7	12	9	14
BuCa-038	X	X	13	14	30	33,2	8	12	9	12	14	14	6	9	11	12	12	12	16	17	8	10	16	18	11	12	18	18	21	23	12	15	11	13	10	12
BuCa-039	X	Y	11	12	29	30	8	9	12	13	16	17	6	9	8	11	9	11	15	16	8	8	16	20	13	13	21	24	20	23	12	14	11	15	9	11
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# Mikrovariáns mikroszatellita allélek



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

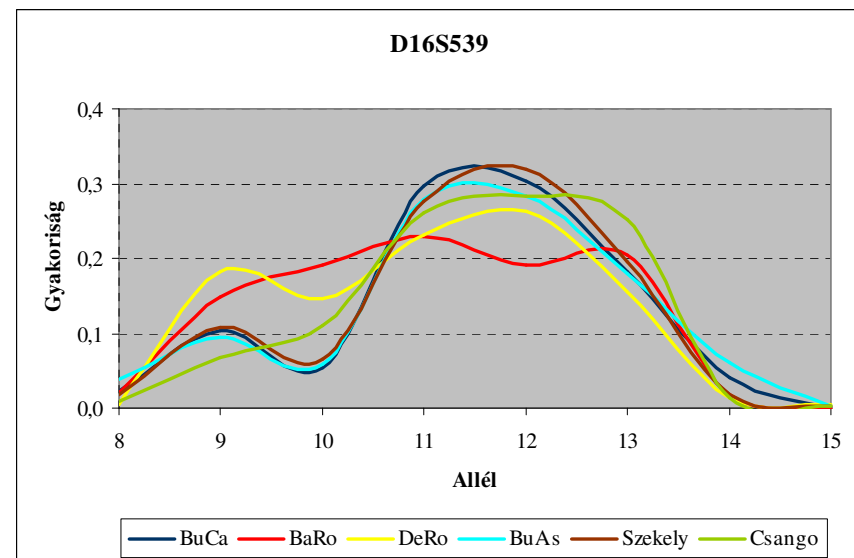
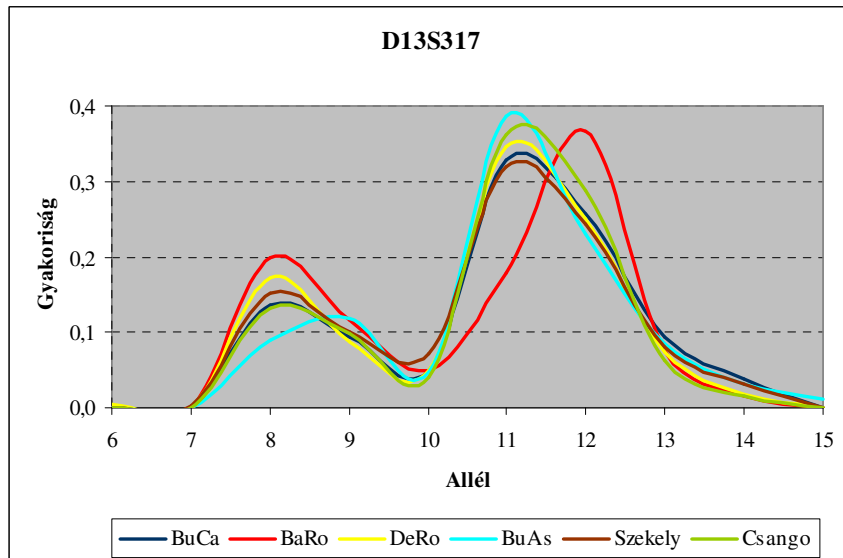
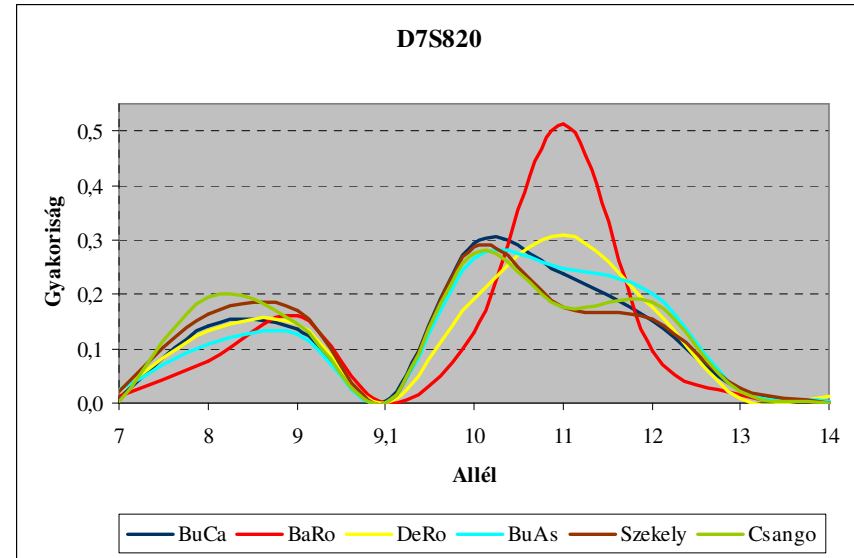
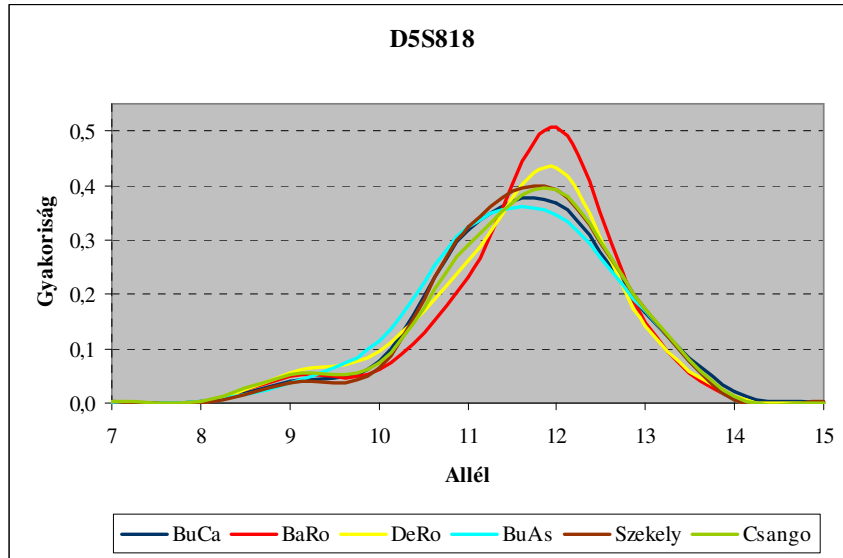
Allél	Fragmens méret	5' Flanking régió	Repeat régió	3' Flanking régió
8.-1	205 bp	██████████	- (GATA) <sub>8</sub> -	██████████ - (T) <sub>8</sub> ATCT -
9.+1	211 bp	██████████	- (GATA) <sub>9</sub> -	██████████ - (T) <sub>10</sub> ATCT -
10	214 bp	██████████	- (GATA) <sub>10</sub> -	██████████ - (T)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



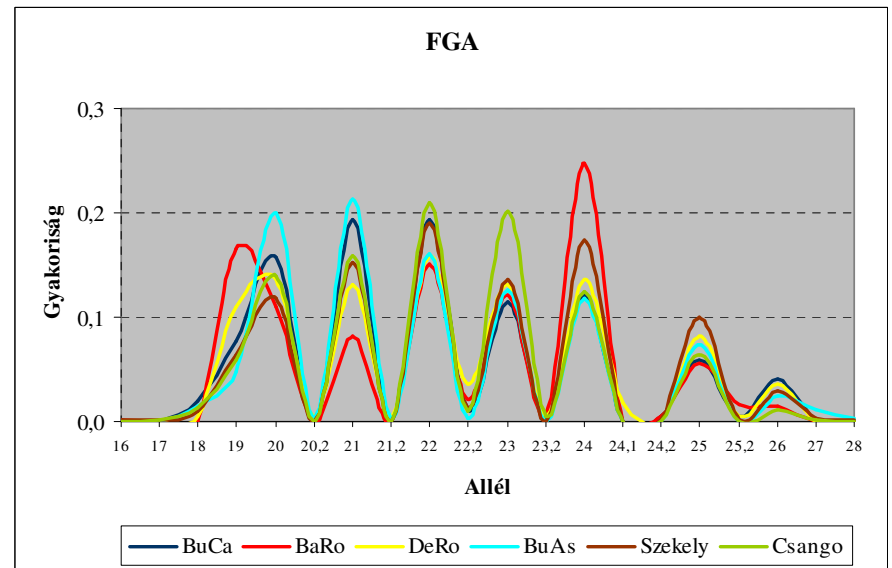
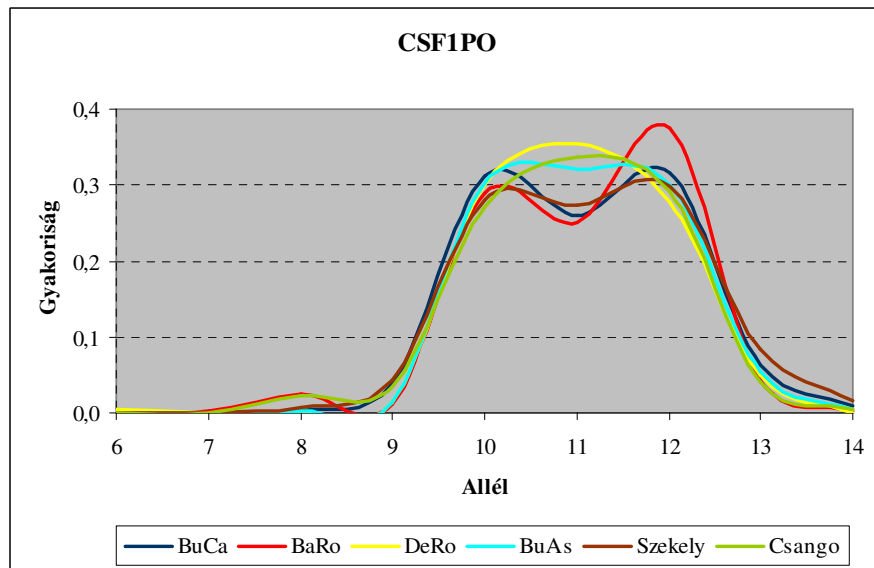
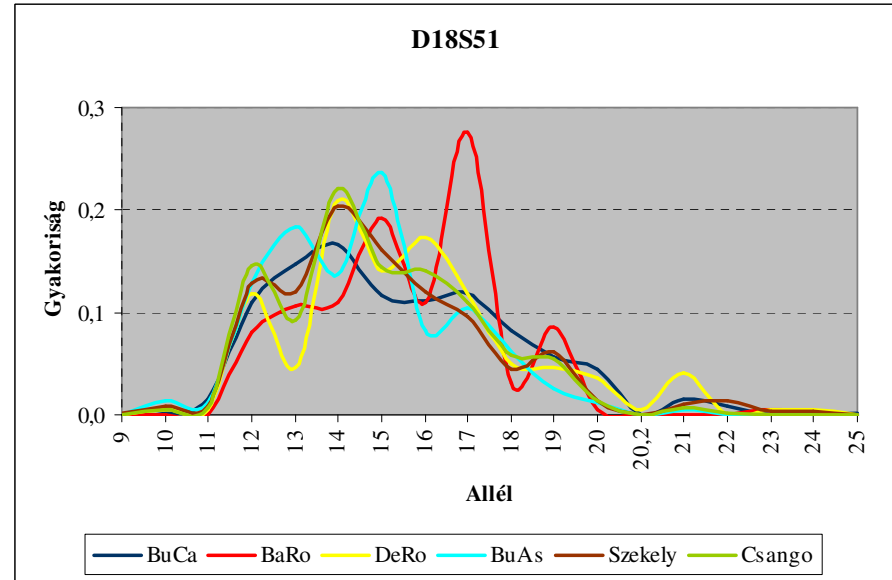
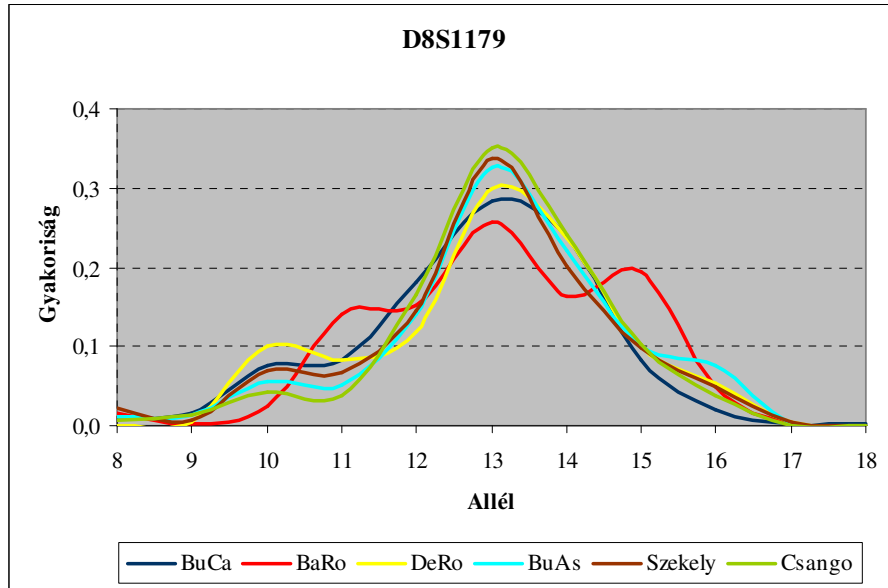
**STR Base adatbázis**

<http://www.cstl.nist.gov/strbase/>

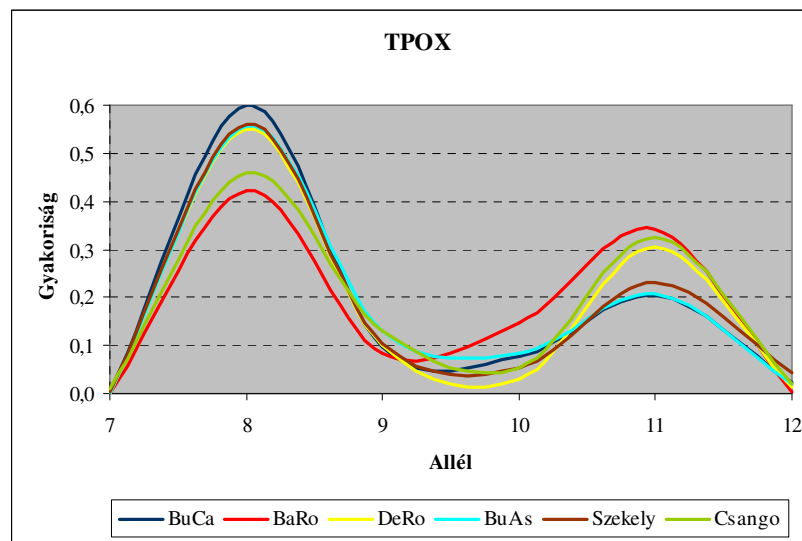
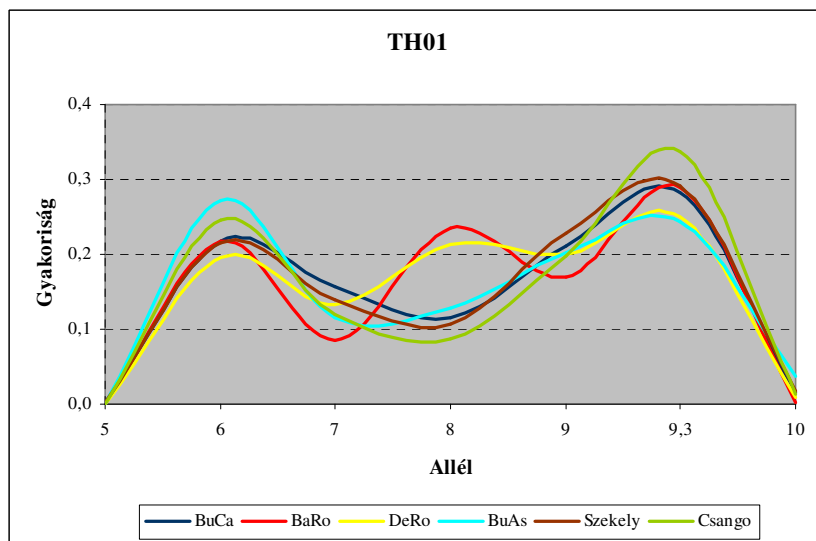
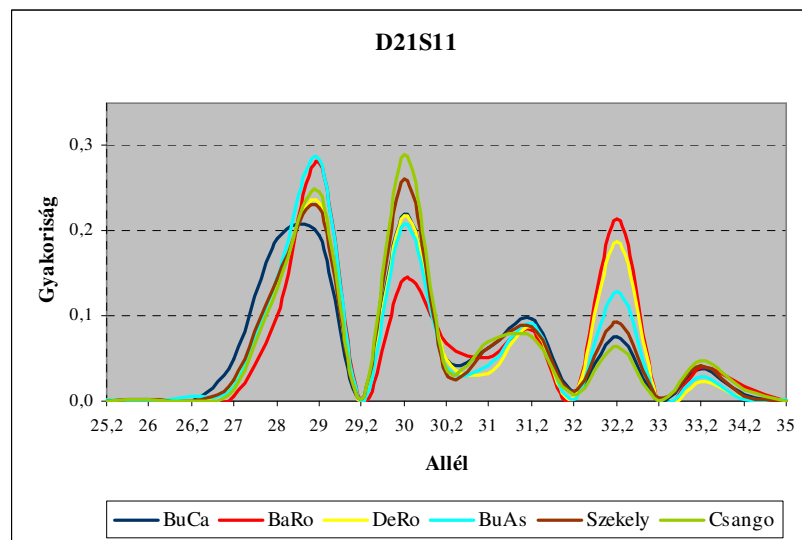
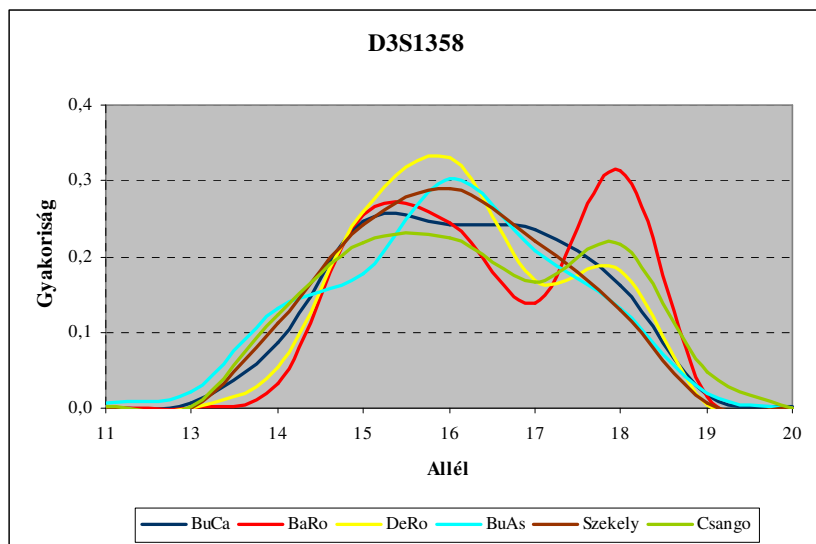
# *Autoszómás mikroszatellita lokuszok allélfrekvencia diagramjai*



# *Autoszómás mikroszatellita lokuszok allélfrekvencia diagramjai*



# *Autoszómás mikroszatellita lokuszok allélfrekvencia diagramjai*



## *A magyar populációk HWE tesztelése: Bonferroni korrekció*

Marker	BuCa	BaRo	DeRo	BuAs	Székely	Csángó
D3S1358	0,850	0,033*	0,765	0,202	0,223	0,518
TH01	0,255	0,577	0,378	0,471	0,731	0,122
D21S11	0,694	0,337	0,112	0,726	0,566	0,031*
D18S51	1,000	0,296	0,144	0,108	0,208	0,163
PentaE	0,057	0,669	0,507	0,769	0,190	0,357
D5S818	0,643	0,736	0,998	0,415	0,074	0,033*
D13S317	0,874	0,235	0,016*	0,194	0,279	0,632
D7S820	0,341	0,011*	0,169	0,062	0,234	0,342
D16S539	0,222	0,242	0,550	0,437	0,349	0,944
CSF1PO	0,932	0,008*	1,000	0,597	0,491	0,323
PentaD	0,132	0,009*	0,550	0,625	0,481	0,610
VWA	0,098	0,512	0,264	0,496	0,484	0,435
D8S1179	0,364	0,004*	0,054	0,068	0,162	0,087
TPOX	0,899	0,045*	0,858	0,412	0,105	0,465
FGA	0,997	0,831	0,576	0,668	0,220	0,322
D2S1338	0,714	$\alpha \equiv \acute{a}/L$	-	-	0,859	0,658
D19S433	0,328	-	-	-	0,393	0,092





# *locus-by-locus AMOVA*

\*\*\*\*\*  
 Locus by locus AMOVA:  
 \*\*\*\*\*

List of loci with only one allele:  
 -----

Results for polymorphic loci only:  
 -----

Among Populations:					Within Populations:				Fixation indices:		
Locus	SSD	d.f.	Va	% variation	SSD	d.f.	Vb	% variation	FST	P-value	
0	7.75764	3	0.00625	1.57629	557.66144	1430	0.38997	98.42370	0.01576	0.00000	
1	3.74656	3	0.00243	0.61035	565.29877	1430	0.39531	99.38965	0.00610	0.00000	
2	5.49025	3	0.00401	0.94814	599.47559	1430	0.41921	99.05186	0.00948	0.00000	
3	9.69658	3	0.00797	1.81185	617.26575	1430	0.43165	98.18815	0.01812	0.00000	
4	5.61727	3	0.00404	0.88904	644.59894	1430	0.45077	99.11096	0.00889	0.00000	
5	4.78144	3	0.00352	0.98429	507.02957	1430	0.35457	99.01571	0.00984	0.00000	
6	8.45171	3	0.00691	1.74893	555.01764	1430	0.38812	98.25107	0.01749	0.00000	
7	15.26867	3	0.01339	3.37880	547.42377	1430	0.38281	96.62120	0.03379	0.00000	
8	6.03527	3	0.00459	1.14319	567.88104	1430	0.39712	98.85681	0.01143	0.00000	
9	2.33228	3	0.00119	0.33057	513.23395	1430	0.35890	99.66943	0.00331	0.03324	
10	13.52616	3	0.01166	2.77988	583.30298	1430	0.40790	97.22012	0.02780	0.00000	
11	4.75754	3	0.00340	0.86127	559.32263	1430	0.39113	99.13873	0.00861	0.00000	
12	4.79482	3	0.00339	0.82593	581.83771	1430	0.40688	99.17406	0.00826	0.00000	
13	7.80187	3	0.00651	2.04652	445.60049	1430	0.31161	97.95348	0.02047	0.00000	
14	7.29035	3	0.00569	1.30329	615.84143	1430	0.43066	98.69671	0.01303	0.00000	

-----  
 AMOVA design and results (average over 15 loci):  
 -----



# Overall AMOVA

Distance method: Sum of squared size difference (RST)

-----  
AMOVA design and results :  
-----

Reference: Weir, B.S. and Cockerham, C.C. 1984.  
Excoffier, L., Smouse, P., and Quattro, J. 1992.  
Weir, B. S., 1996.

Source of variation	d.f.	Sum of squares	Variance components	Percentage of variation
Among populations	3	796.462	0.61601 Va	1.24
Within populations	1430	69928.155	48.90081 Vb	98.76
Total	1433	70724.616	49.51682	
Fixation Index	FST :	0.01244		

-----  
Significance tests (1023 permutations)  
-----

Va and FST : P(rand. value > obs. value) = 0.00000  
P(rand. value = obs. value) = 0.00000  
P(rand. value >= obs. value) = 0.00000+-0.00000

## *Genetic structure: F-statistics and AMOVA*

Conventional  $F_{ST}$  and AMOVA ( $\Phi_{ST}$ ) values for each analysed STR loci among the four Hungarian population samples

	D3S1358	VWA	FGA	D8S1179	D21S11	D18S51	D5S818	D13S317
$F_{ST}$	0.016	0.009	0.013	0.008	0.009	0.018	0.010	0.017
$\Phi_{ST}$	0.015	0.009	0.013	0.008	0.009	0.018	0.009	0.017
$P_{AMOVA}$	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$
	D7S820	TH01	TPOX	CSF1PO	D16S539	PentaE	PentaD	Overall loci
$F_{ST}$	0.033	0.006	0.020	0.003	0.011	0.009	0.027	0.014
$\Phi_{ST}$	0.033	0.006	0.020	0.003	0.011	0.009	0.028	0.012
$P_{AMOVA}$	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$	0.03	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$	$<10^{-3}$

$P_{AMOVA}$ : significance value of the AMOVA test.

Table 3

Conventional  $F_{ST}$  and AMOVA ( $\Phi_{ST}$ ) values between the Hungarian population pairs for the overall 15 STR loci

	BuCa/BaRo	BuCa/EHRo	BuCa/ASK	BaRo/EHRo	BaRo/ASK	EHRo/ASK
$F_{ST}$	0.023	0.006	0.003	0.012	0.025	0.007
$\Phi_{ST}$	0.014	0.002	0.010	0.009	0.019	0.017
$P_{AMOVA}$	$<10^{-3}$	0.100	$<10^{-3}$	0.002	$<10^{-3}$	$<10^{-3}$

$P_{AMOVA}$ : significance value of the populations' pair-wise AMOVA; EHRo, ASK, BuCa, BaRo: see Tables 1 and 2.

# Genetic substructure: $F$ -statistics and AMOVA

a,

$F_{ST}$	$P$	BuCa															BaRo															DeRo															BuAs														
		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
BuCa																																																													
BaRo																																																													
DeRo																																																													
BuAs																																																													

b,

$\Phi_{ST}$	$P$	BuCa															BaRo															DeRo															BuAs														
		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
BuCa																																																													
BaRo																																																													
DeRo																																																													
BuAs																																																													

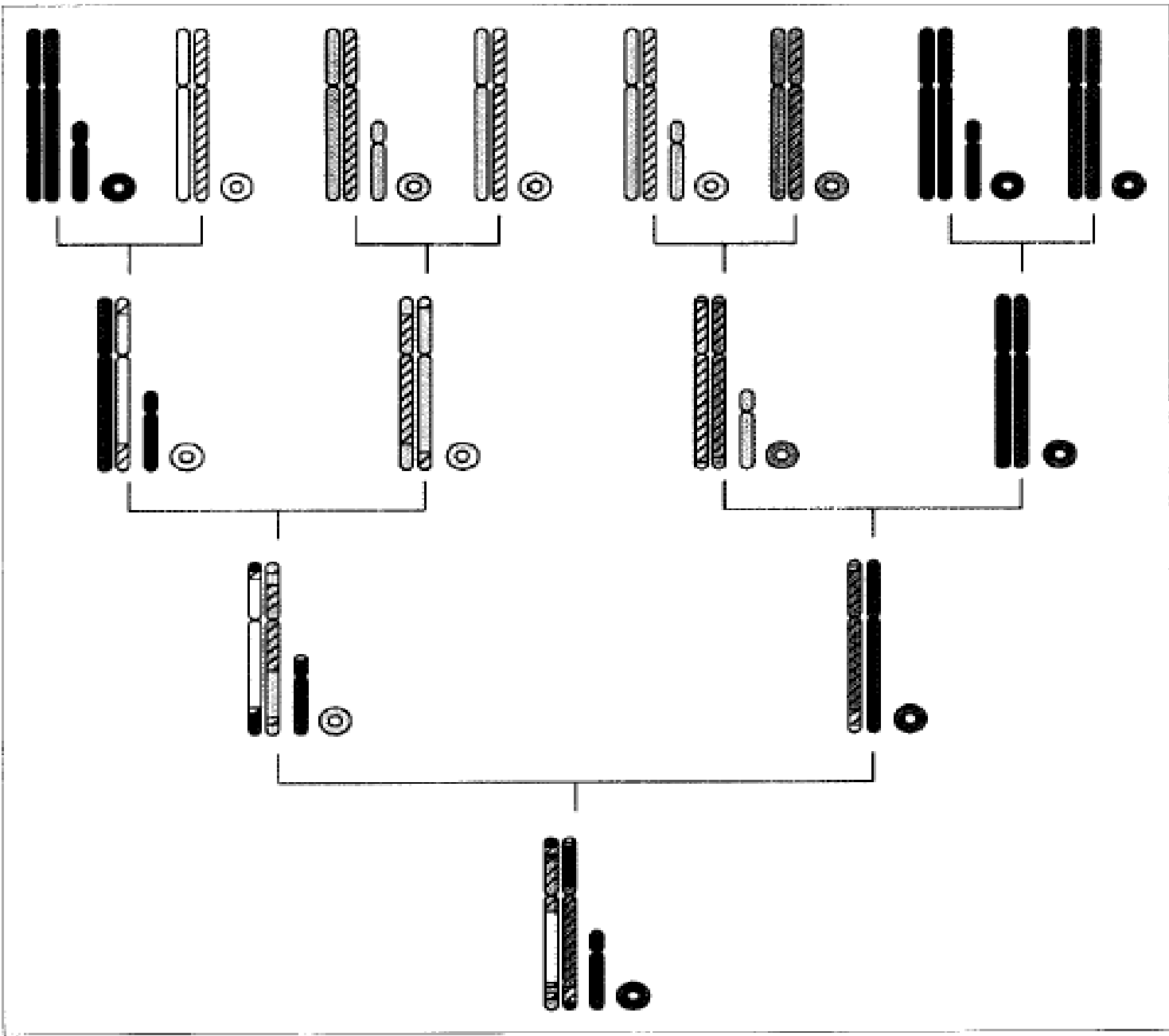
a, Jelölések: ■  $(F_{ST}, \Phi_{ST}) > 0,02; P < 0,1$     ▒  $0,02 > (F_{ST}, \Phi_{ST}) > 0,01; 0,01 < P < 0,05$     □  $(F_{ST}, \Phi_{ST}) < 0,01; P > 0,05$

$F_{ST}$	$P$	BuCa																	Szekely																	Csango																
		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17
BuCa																																																				
Szekely																																																				
Csango																																																				

b,

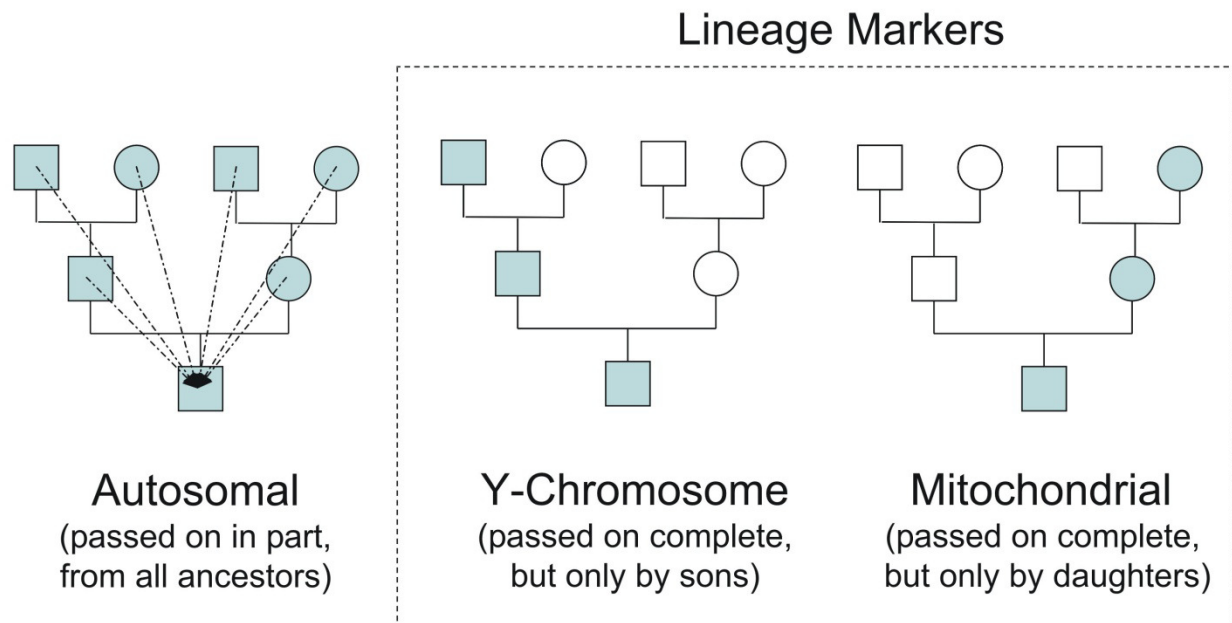
$\Phi_{ST}$	$P$	BuCa																	Szekely																	Csango																
		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17
BuCa																																																				
Szekely																																																				
Csango																																																				

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



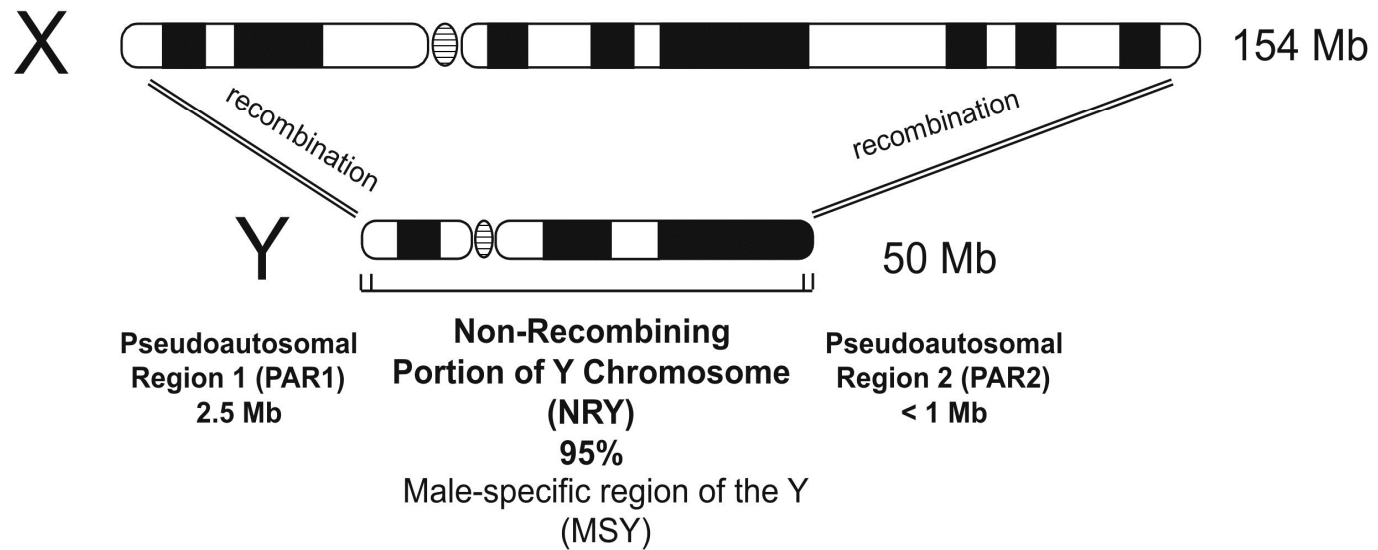
# Lineage Markers

- Y chromosome and mitochondrial DNA
  - passed down from generation-to-generation without changing (except for mutations)
  - patrilineal and matrilineal inheritance
  - haplotype: only a single allele per individual
  - linked markers (no recombination during meiosis)
  - product rule cannot be applied in statistical calculations



# Y Chromosome Structure

(A)



(B)

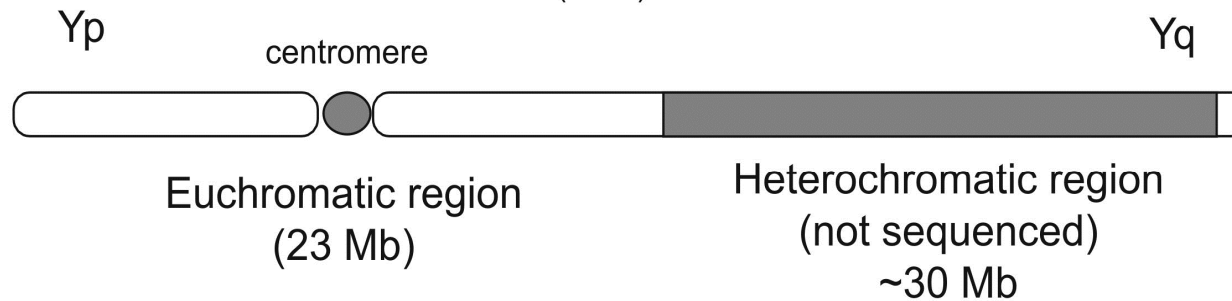
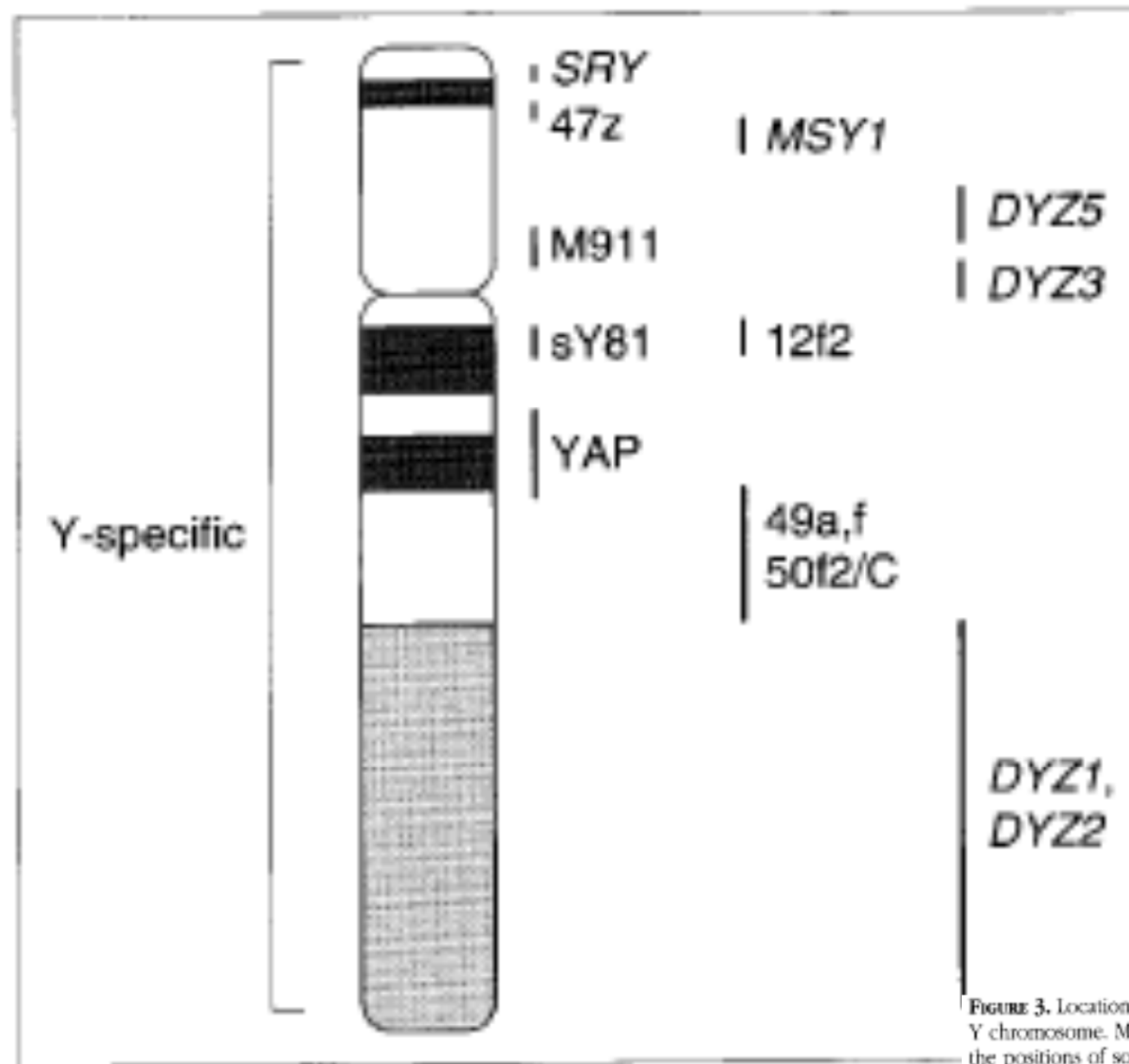


Figure 9.4, J.M. Butler (2005) *Forensic DNA Typing*, 2<sup>nd</sup> Edition © 2005 Elsevier Academic Press



**FIGURE 3.** Location of selected polymorphic loci on the Y chromosome. Most of the chromosome is Y-specific (left) and the positions of some polymorphic loci are shown (right). The ideogram of the chromosome shows the light giemsa-stained (G) bands (white), the dark G bands (dark grey) and the heterochromatic region (grey).

TABLE 2. Y polymorphisms that can be typed using PCR<sup>a</sup>

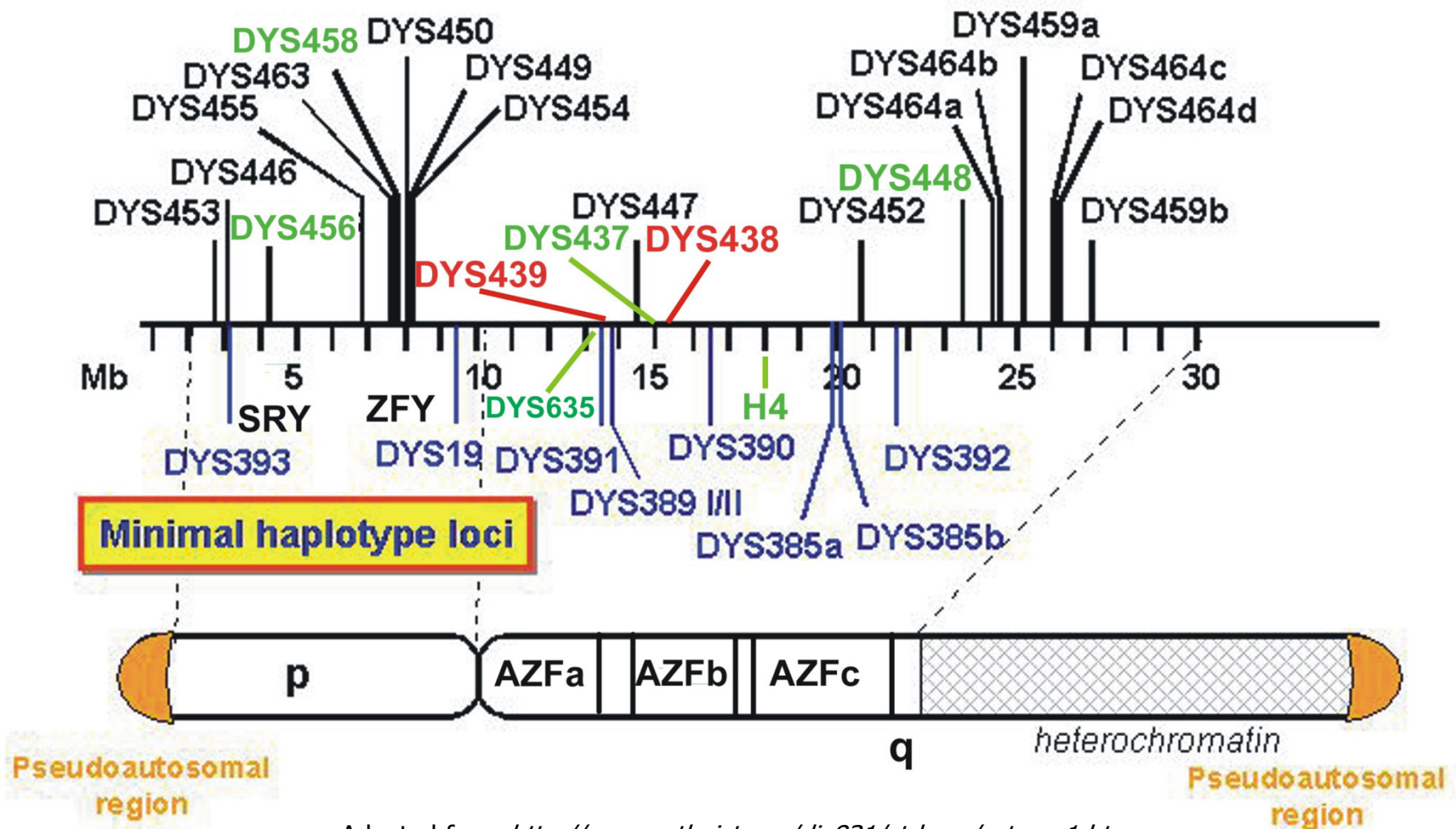
Locus	Name	Primers (5'→3')		Comments	Ref.
<i>DYS287</i>	YAP	CAGGGGAAGATAAAGAAATA	ACTGCTAAAAGGGGATGGAT	Alu insertion <sup>b</sup>	25
<i>DYS271</i>	sY81	AGGCACTGGTCAGAATGAAG	AATGGAAAATACAGCTCCCC	<i>Nla</i> III detects point mutation <sup>b</sup>	9
<i>SRY</i>		TCCTTAGCAACCATTAATCTGG	AAATAGCAAAAATGACACAAGGC	Point mutation	c
<i>YRRM2</i>		CTTTGAAAACAATTCCTTTTCC	AGAGATGCACTTCAGAGG	Product present or absent	31
<i>DYZ3</i>	Yα1	TCTGAGACACTTCTTTGTGGTA	CGCTCAAAATATCCACTTTCAC	<i>Hind</i> III cleavage (partial) indicates 6.0 kb unit	d
<i>DYF155S2</i>	50f2/C	CTCAAGCTAGGACAAAGGGAAAGG	GAGGTAGATGCTGAAGCGGTATAG	196 bp fragment present or absent	e
<i>DYS288</i>		CATTACAAATACCTGGACACTG	TTGCTTTGCTTGTCAATTCAGA	Dinucleotide repeat, 1 Y locus <sup>f</sup>	GDB8
-	YCAI	CCCATGCCTGTTCTCCAGATT	GAGAGTGTGACACATCAGGTA	CA repeat, 2 Y loci	198
-	YCAII	TATATTAATAGAAGTAGTGA	TATCGATGTAATGTTATATTA	CA repeat, 2 Y loci	19
-	YCAIII	CCACATTGTGTGAATGTGTGA	TCCTCAGAGAAGGAGAAACTA	CA repeat, 2 Y loci	19
<i>DYS388</i>		GTGAGTTAGCCGTTTAGCGA	CAGATCGCAACCCTGCG	Trinucleotide repeat, 1 Y locus <sup>f</sup>	GDB8
<i>DYS392</i>		TCATTAATCTAGCTTTTAAAAACAA	AGACCCAGTTGATGCAATGT	Trinucleotide repeat, 1 Y locus <sup>f</sup>	GDB8
<i>DYS19</i>	27H39LR	CTACTGAGTTTCTGTTATAGT	ATGGCATGTAGTGAGGACA	GATA repeat, 1 Y locus <sup>f</sup>	18
<i>DYS390</i>		TATATTTTACACATTTTGGGCC	TGACAGTAAAATGAACACATTGC	Tetranucleotide repeat, 1 Y locus <sup>f</sup>	GDB8
<i>DYS391</i>		CTATTCATTCAATCATAACCCCA	GATTCTTGTGGTGGGTCTG	Tetranucleotide repeat, 1 Y locus <sup>f</sup>	GDB8
<i>DYS393</i>		GTGGTCTTCTACTTGTGTCAATAC	AACTCAAGTCCAAAAAATGAGG	Tetranucleotide repeat, 1 Y locus <sup>f</sup>	GDB8
<i>DYS385</i>		AGCATGGGTGACAGAGCTA	TGGGATGCTAGGTAAGCTG	Tetranucleotide repeat, 2 Y loci	GDB8
<i>DYS389</i>		CCAACCTCATCTGTATTATCTATG	TCTTATCTCCACCCACCAGA	Tetranucleotide repeat, 2 Y loci	GDB8
<i>DXYS156Y</i>		GTAGTGGTCTTTTGCCTCC	CAGATACCAAGGTGAGAATC	TAAAA repeat on X and Y	32



# Y STR Positions along Y Chromosome

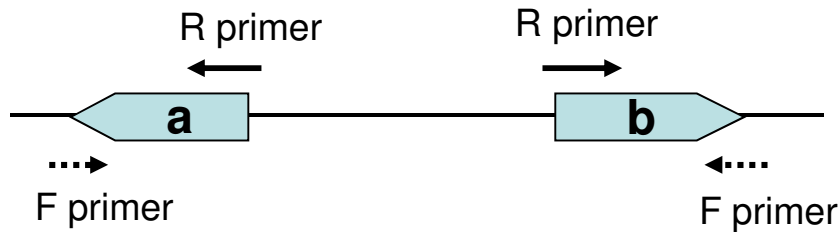
Extended **haplotype loci**

ABI AmpF/STR **Yfiler loci**



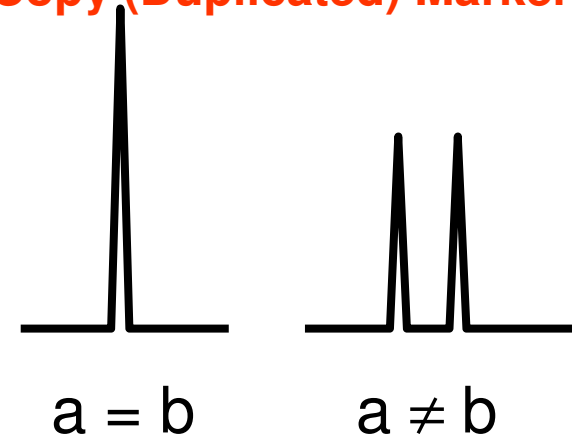
Adapted from <http://www.cstl.nist.gov/div831/strbase/ystrpos1.htm>

**(A) DYS385 a/b**

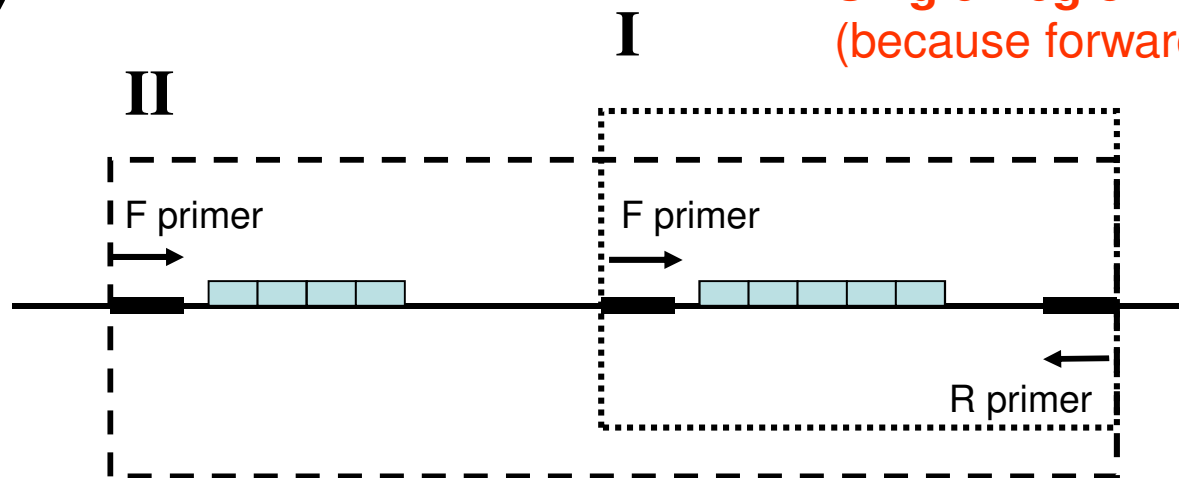


*Duplicated regions are 40,775 bp apart and facing away from each other*

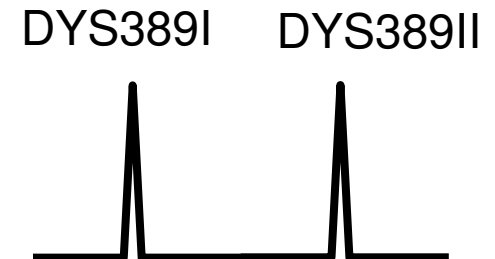
**Multi-Copy (Duplicated) Marker**



**(B) DYS389 I/II**

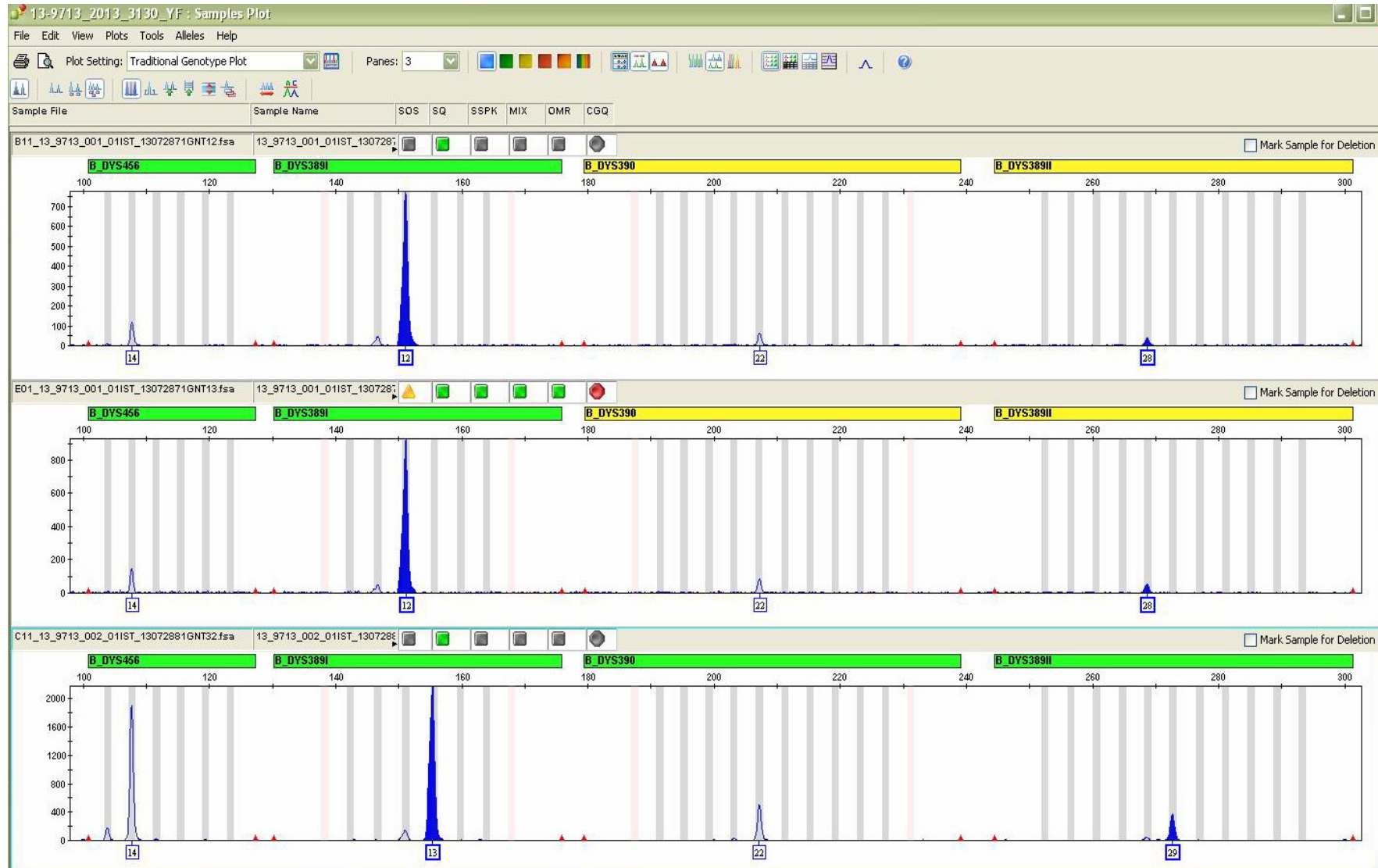


**Single Region but Two PCR Products**  
(because forward primers bind twice)



# *DYS389 I-II meiotikus mutáció*

## *- két testvér Y kromoszómás haplotípusa*



Twelve-locus Y-STR haplotype diversity values in the analysed Transylvanian populations (Szekler, Csango) as well as conventional  $F$ -statistic ( $F_{ST}$ ) and AMOVA ( $\Phi_{ST}$ ) values for the European minimal Y-STR haplotypes calculated for the Transylvanian and neighbouring population pairs

	Szekler ( $0.9987 \pm 0.005$ ) <sup>a</sup>		Csango ( $0.9883 \pm 0.016$ ) <sup>a</sup>	
	$F_{ST}$ <sup>b</sup>	$\Phi_{ST}$ <sup>b</sup>	$F_{ST}$ <sup>b</sup>	$\Phi_{ST}$ <sup>b</sup>
Szekler, Corund [17]	0.002 <sup>c</sup>	0.051 <sup>c</sup>	0.011 <sup>c</sup>	0.083 <sup>c</sup>
Romanian, Bukarest [18]	0.000	0.032 <sup>c</sup>	0.009 <sup>c</sup>	0.011
Bulgarian [19]	0.003 <sup>c</sup>	0.022 <sup>c</sup>	0.010 <sup>c</sup>	0.020 <sup>c</sup>
Bulgarian Turks [19]	0.002	0.012	0.009 <sup>c</sup>	0.031 <sup>c</sup>
Serbian [20]	0.003 <sup>c</sup>	0.040 <sup>c</sup>	0.012 <sup>c</sup>	0.000

[ ]: numbers in brackets are the same in references.

<sup>a</sup> Haplotype diversity ( $\pm$ S.E.).

<sup>b</sup> Genetic structure.

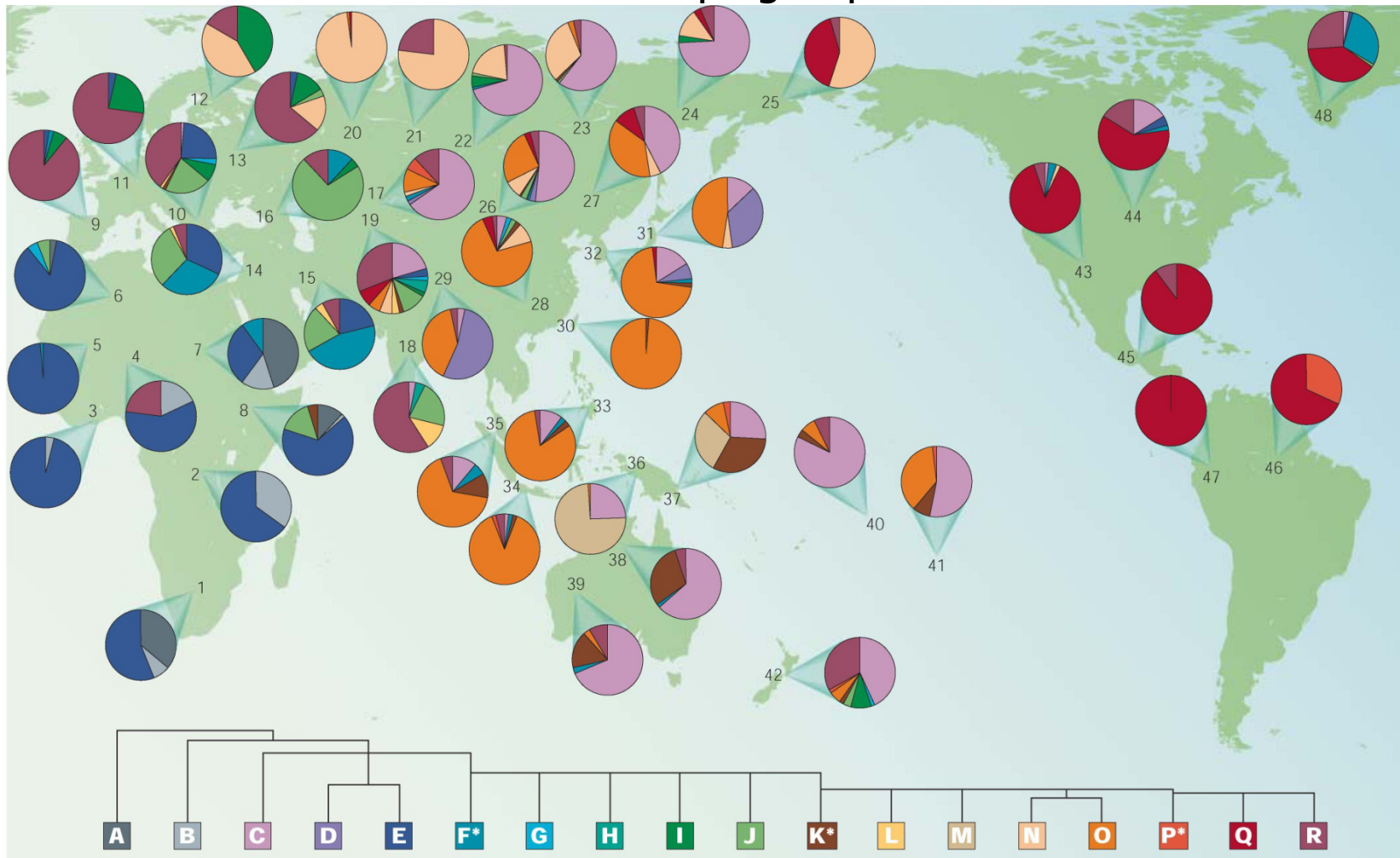
<sup>c</sup> The  $F$ -statistic value represents statistically significant difference at the  $P = 0.05$  level.





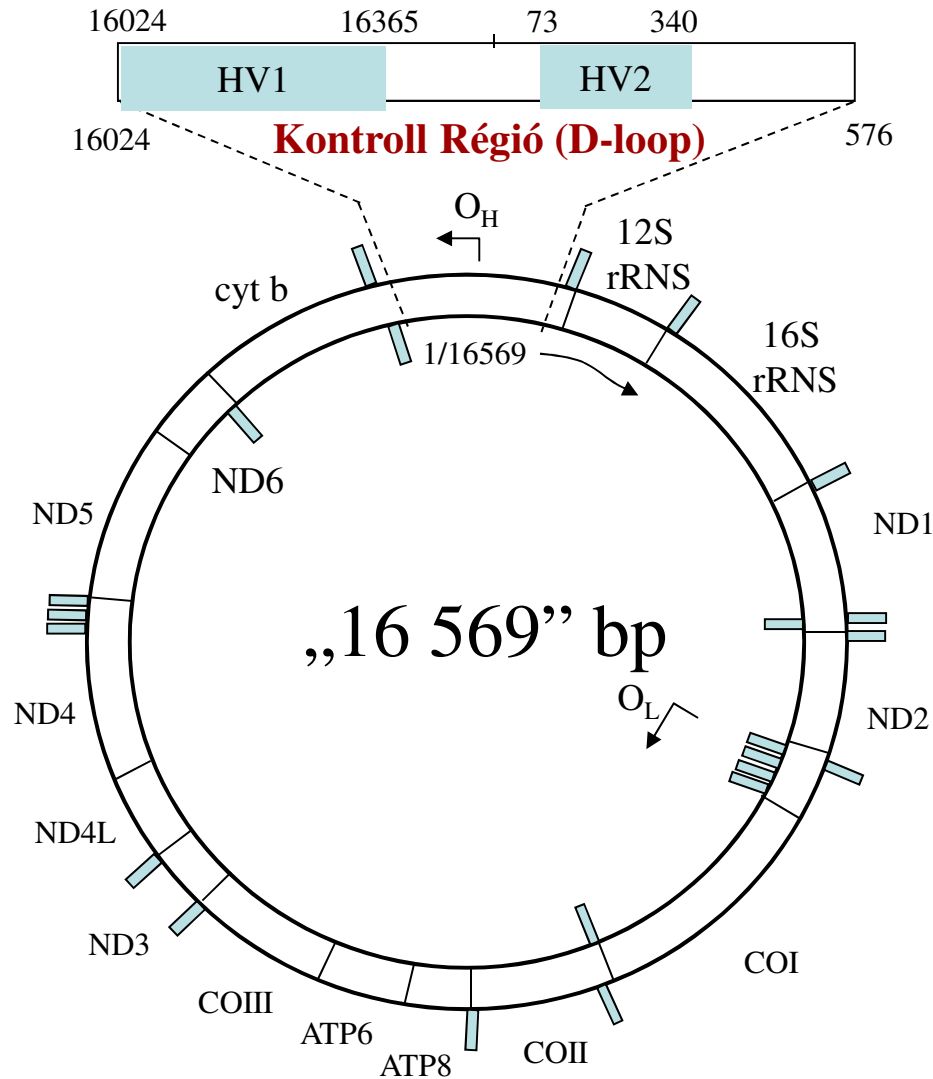
# V. Y-SNP and Bi-Allelic Markers

- Global Distribution of Y Haplogroups



*Y Chromosome Consortium (2003) Nat Rev Genet 4:598-612*

# Mitokondriális DNS



Sejten belüli nagy kópiaszám (~1000)

Rekombináció hiánya:

Maternális öröklésmenet

Magasabb mutációs ráta

Kódoló Régió: 37 gén

Kontroll Régió - D-loop (~1120 bp):

Hipervariábilis szakaszok (HV1, HV2)

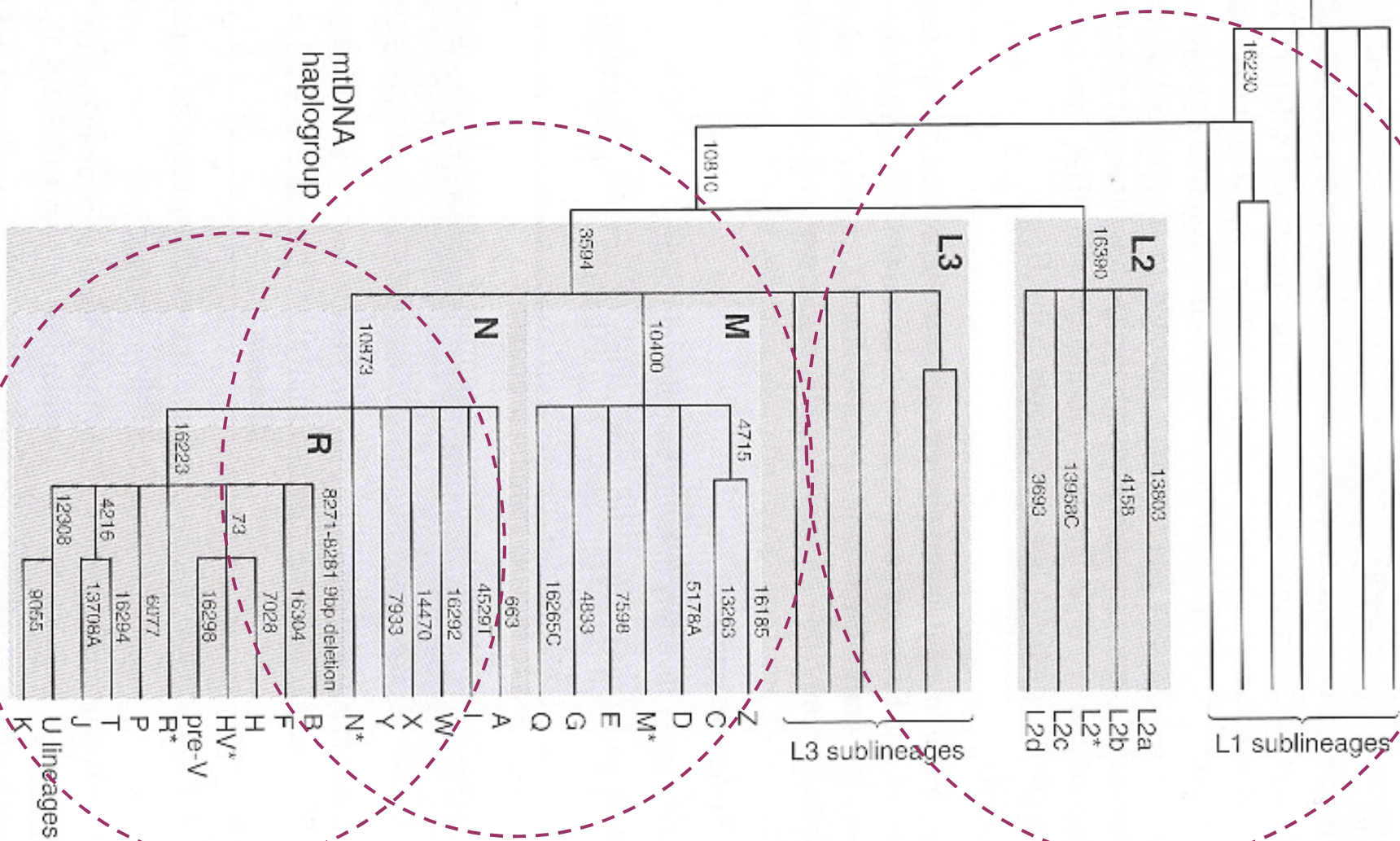
Cambridge referencia szekvencia

Heteroplazmia

Szekvencia-adatbázisok: EMPOP

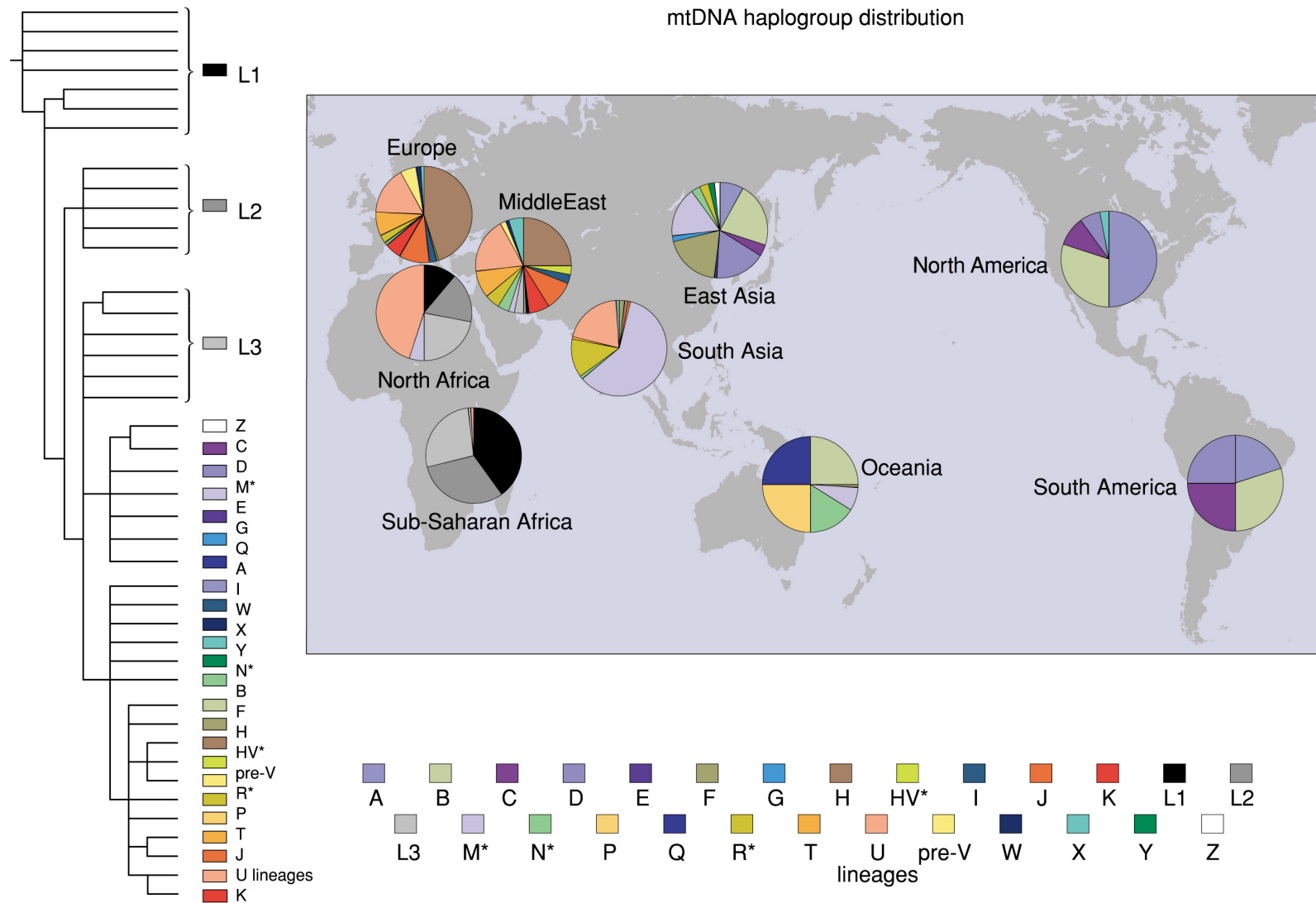


# A főbb mt DNS leszármazási vonalak

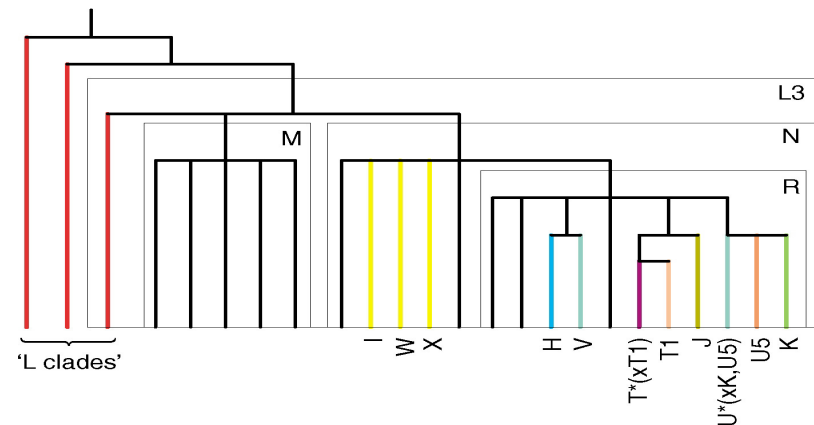
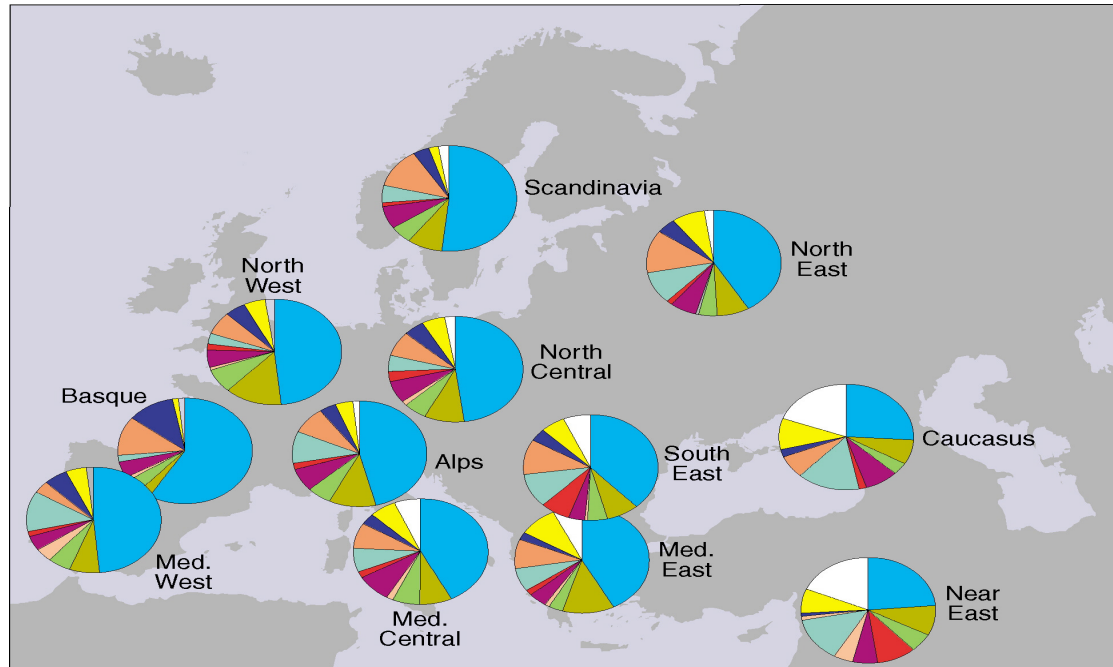




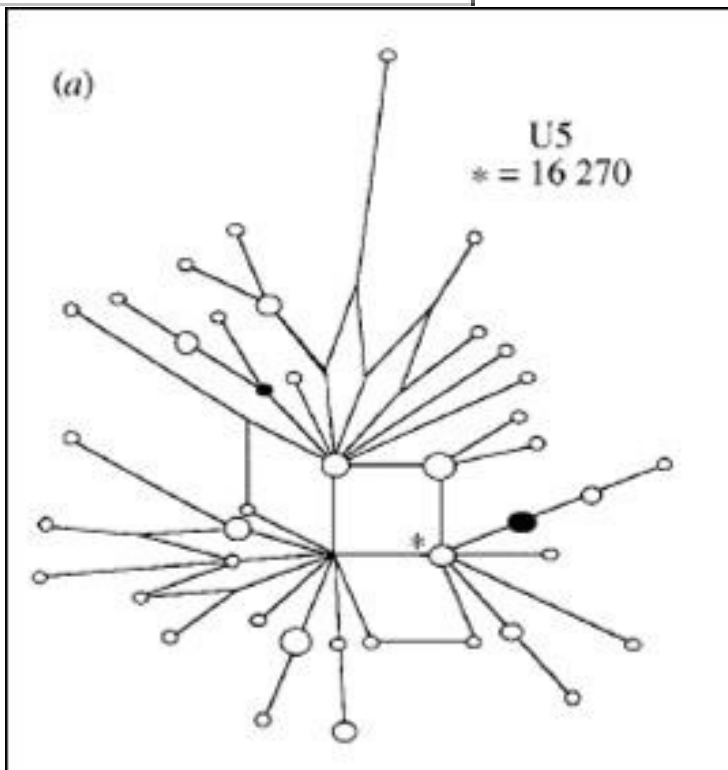
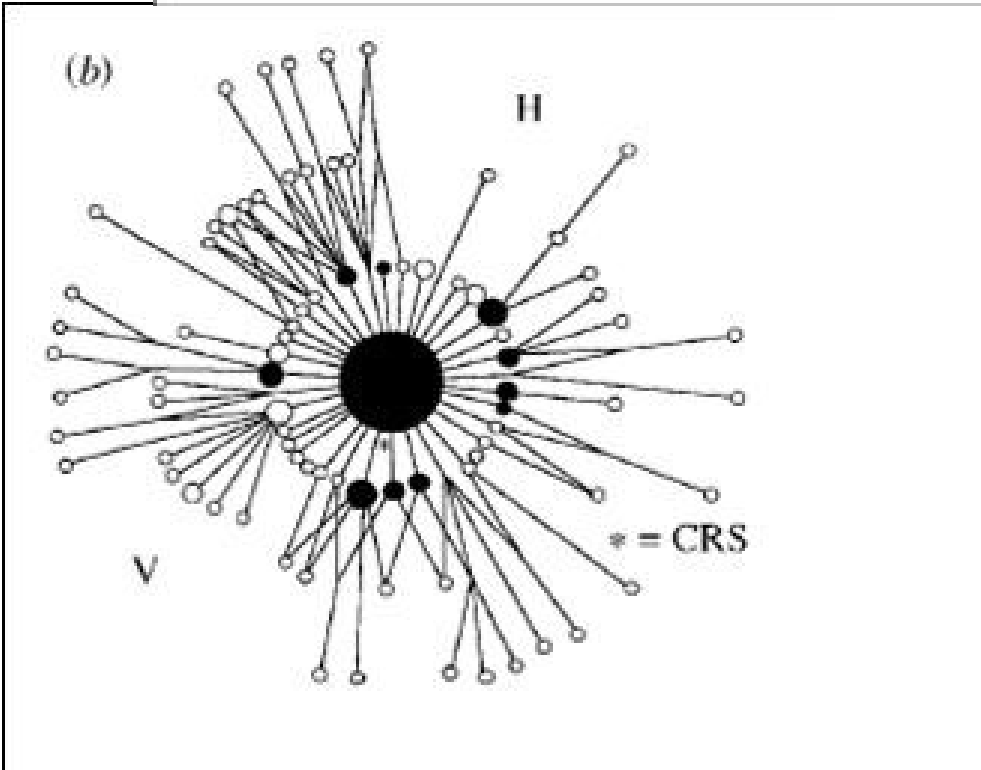
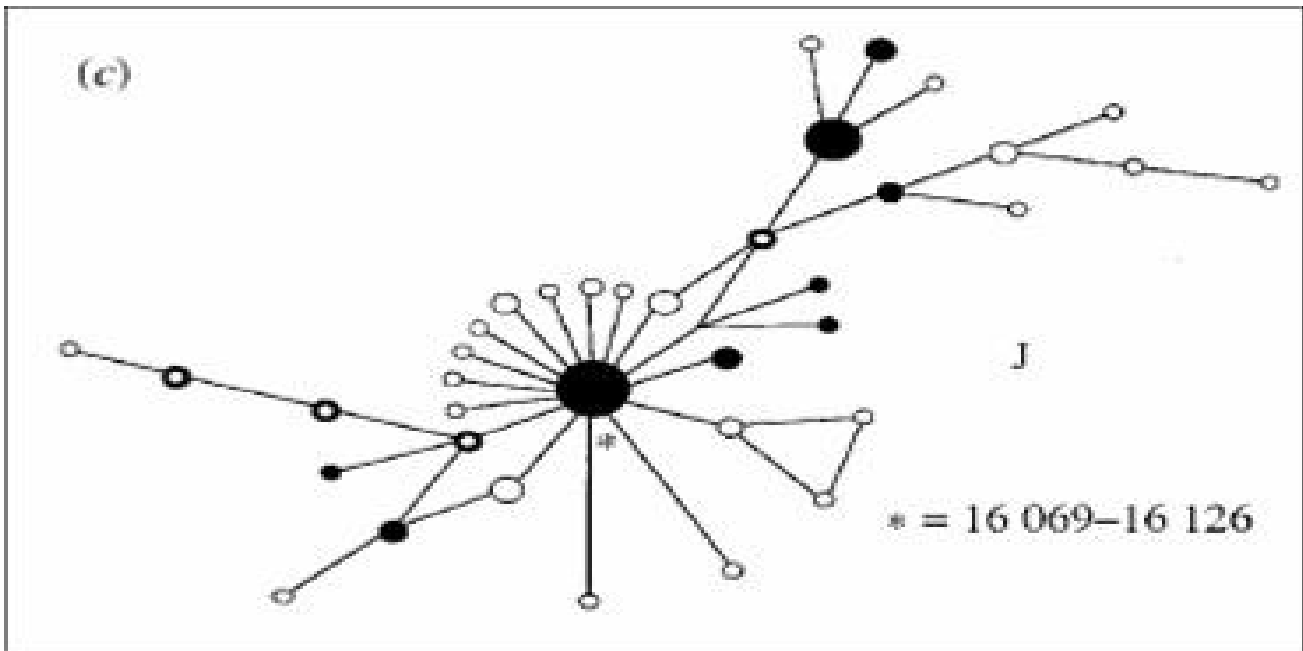
# A főbb mt DNS haplocsoportok eloszlása



# *A főbb mt DNS haplocsoportok eloszlása a nyugat-eurázsiai kontinensen*







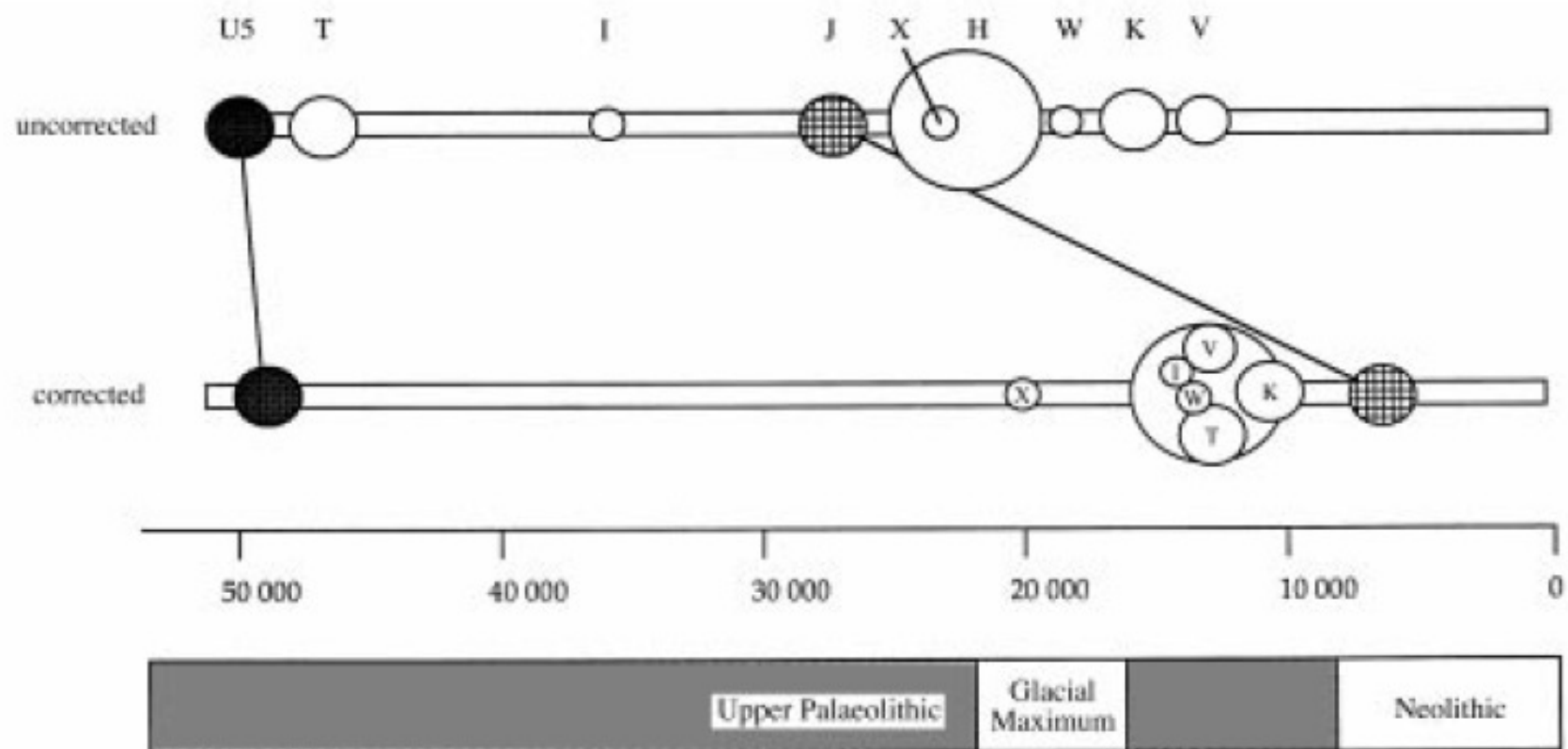


Figure 10. Uncorrected and corrected divergence times for the European clusters. Areas of circles are proportional to cluster frequencies.

component	dates (BP)	main associated clusters	contribution to modern gene pool
Neanderthal	300 000	unclassified	0%
Early Upper Palaeolithic	50 000	U5	10%
Late Upper Palaeolithic	11 000–14 000	H, V, I, W, T, K	70%
Neolithic	8500	J (+ more of H, T, K?)	20%

# Magyarországi előzmények I.: első populációs felmérések a HVR-I és HVR-II régióra

Table 3. *MtDNA haplogroup frequencies*

	Finns <sup>1</sup>	Skolt Saami	Inari Saami	Erzas	Csángós	Hungarians
H	0.42(44)	0	0.02(2)	0.36(21)	0.37(25)	0.37(29)
I	0.01(1)	0	0	0.07(4)	0.01(1)	0
J	0.08(9)	0	0.04(5)	0.14(8)	0.10(7)	0.17(13)
K	0.03(3)	0	0	0.03(2)	0.06(4)	0.04(3)
M	0.03(3)	0.10(5)	0.05(6)	0.02(1)	0	0
T	0.05(5)	0	0	0.09(5)	0.12(8)	0.03(2)
U	0.22(23)	0.38(19)	0.66(84)	0.19(11)	0.22(15)	0.17(13)
V	0.04(4)	0.52(26)	0.17(21)	0.05(3)	0.06(4)	0.04(3)
W	0.06(6)	0	0	0	0.03(2)	0.05(4)
X	0.04(4)	0	0	0.02(1)	0.01(1)	0.01(1)
Others or?	0.04(4)	0	0.07(9)	0.02(2)	0.01(1)	0.12(9)
Total	106	50	127	58	68	77

Lahermo et al., 2000.

Béres Judit és mtsai

Az mtDNS preV- és V-változatainak gyakorisága az ismert európai és néhány egyéb minták

Európai minták	Esetszám	V	preV
Albán	199	0,5	1,5
Angol	293	2,4	1,0
Baszk	97	12,4	0,0
Bolgár	81	0,0	1,2
Csángó	68	0,0	4,4
Cseh	89	4,5	1,1
Magyar	194	3,1	3,6*
Erra (mordvin)	58	3,4	0,0
Észt	148	0,7	0,0
Finn	236	2,5	0,0
Francia	56	7,1	0,0
Görög	208	1,4	0,5
Holland	21	4,8	0,0
Horvát (szigetek)	447	5,6	1,1
Ír, nyugati	88	5,7	1,1
Német, bajor	33	3,0	3,0
Német, szász	632	2,7	1,4
Német, kevert	157	4,5	0,6
Olasz	240	1,7	0,8
Orosz	144	3,5	1,4
Portugál	54	3,7	0,0
Skót	733	3,4	0,1
Spanyol	293	2,0	2,7
Számi (lapp, skolt)	50	52,0	0,0
Számi (lapp, inari)	127	7,1	0,0

Czeizel, 2003.



## *Előzmények II.: magyar etnikumú populációs minta felmérése a kódoló régió polimorf pontjain*

*Table 1*  
Haplogroup frequencies in the Hungarian, Finnish, Swedish, Tuscan and Italian populations

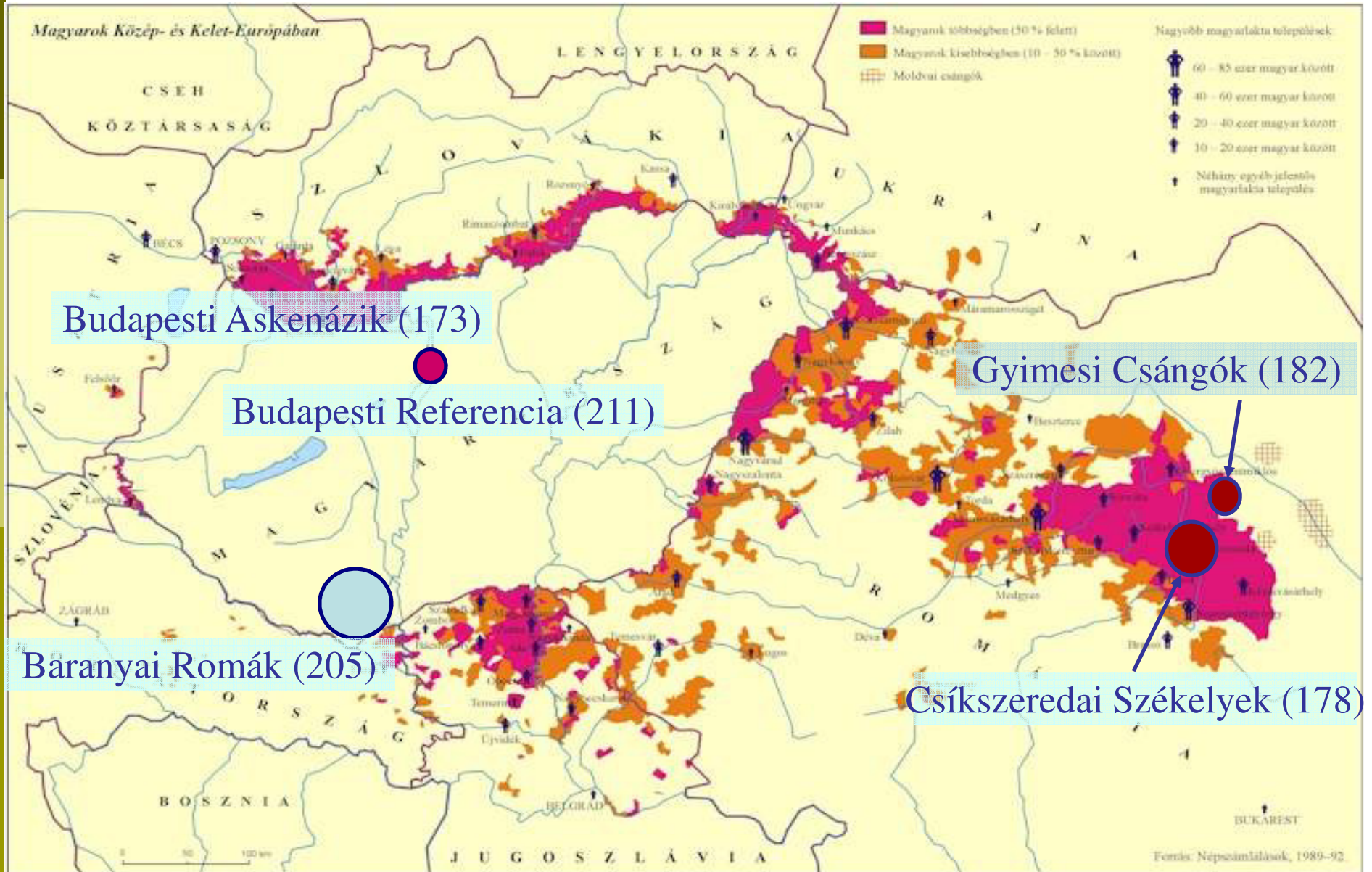
Haplo group	Hungarians (%)	Finnish* (%)	Swedish* (%)	Tuscans* (%)	Italians** (%)
H	18 (32.8)	20 (40.8)	15 (40.5)	20 (41.7)	33 (33.3)
I	1 (1.8)	1 (2.0)	0	2 (4.2)	4 (4.0)
J	9 (16.5)	7 (14.3)	1 (2.7)	7 (14.6)	7 (7.1)
K	8 (14.6)	2 (4.1)	5 (13.5)	3 (6.3)	8 (8.1)
T	1 (1.8)	3 (6.1)	8 (21.6)	5 (10.4)	9 (9.1)
U	2 (3.6)	8 (16.3)	6 (16.2)	5 (10.4)	22 (22.2)
V	1 (1.8)	2 (4.1)	2 (5.4)	0	5 (5.1)
W	2 (3.6)	2 (4.1)	0	1 (2.1)	2 (2.0)
X	4 (7.2)	2 (4.1)	0	4 (8.3)	3 (3.0)
M	3 (5.5)	1 (2.0)	0	0	NA
B	2 (3.6)	0	0	0	NA
Other	4 (7.2)	1 (2.0)	0	1 (2.1)	6 (6.1)
Total	55 (100.0)	49 (100.0)	37 (100.0)	48 (100.0)	99 (100.0)

\* Data from Torroni et al. 1996.

\*\* Data from Torroni et al. 1997.

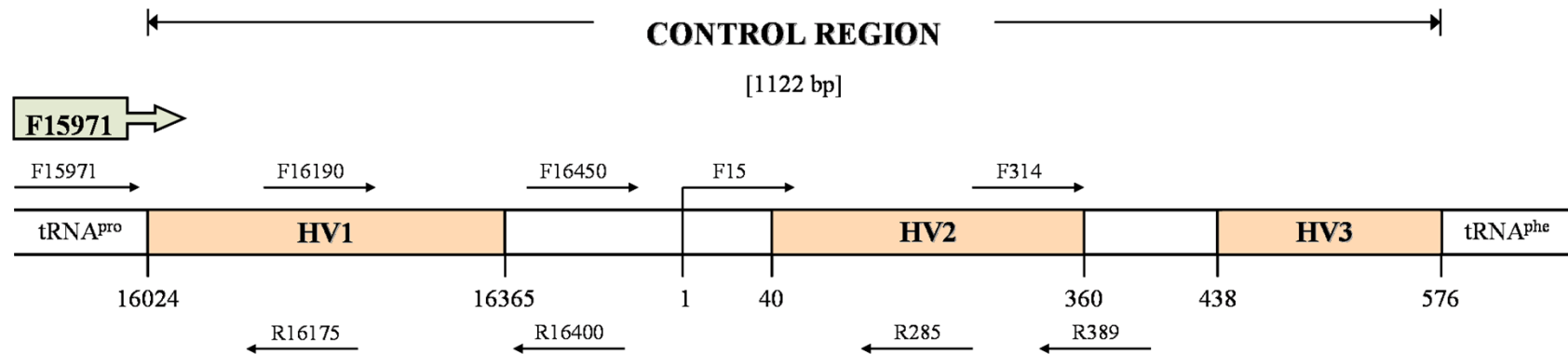
NA: not available

# *Igazságügyi genetikai aspektusú felmérés: a felmérésbe bevont populációs minták*

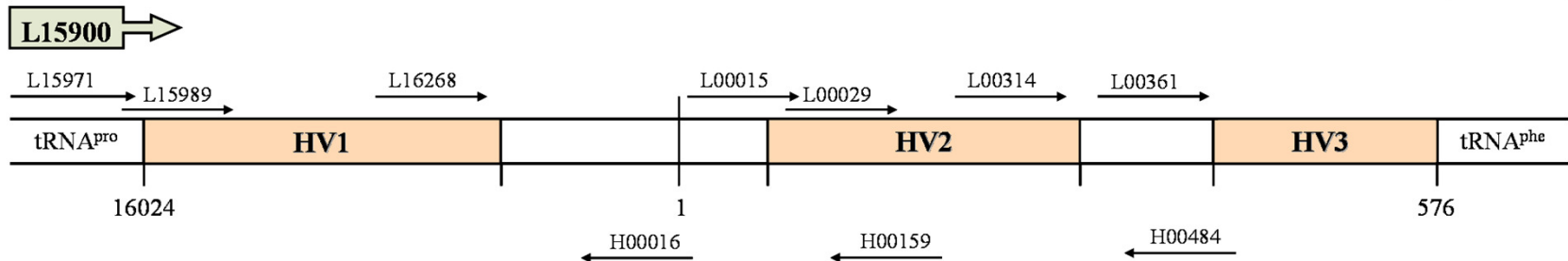




# Alkalmazott PCR amplifikálás és szekvenálási stratégiák a populációs felmérés során



A/ AFDIL, Genbank: DQ359478 – DQ359688, DQ359273 – DQ359477;



B/ Innsbruck, Genbank: EF185421 – EF185780;

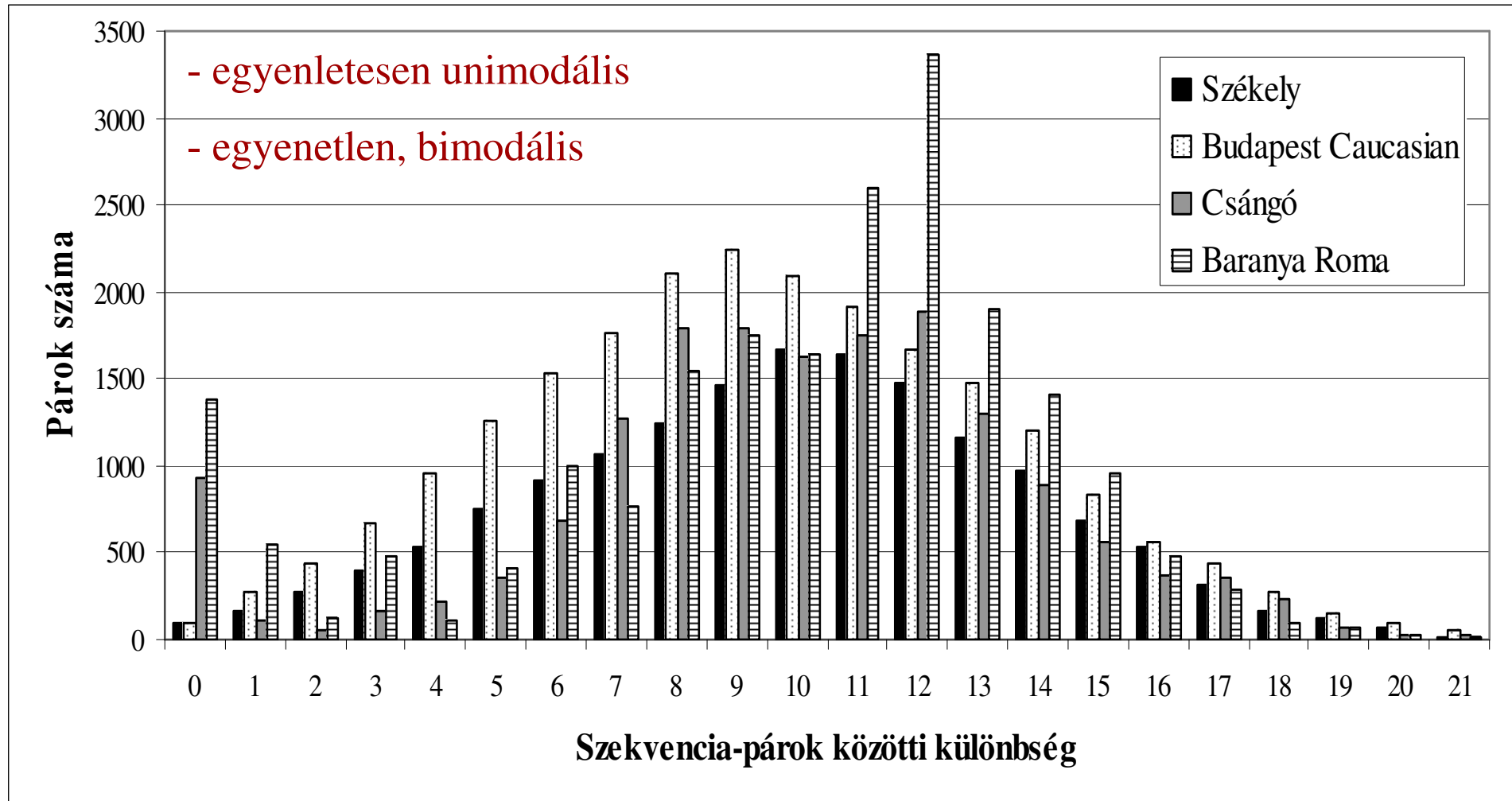
- mtDNS kontroll régió szekvenciák és haplotípusok minőség ellenőrzése
- Megbízható mtDNS szekvencia adatbázis felépítése
- EMPOP (<http://www.empop.org>); Genbank (<http://www.ncbi.nlm.nih.gov/genbank>)

## *Mitokondriális DNS Kontroll Régió diverzitási paraméterek a magyar populációban*

<b>Statisztikai paraméter</b> (1121 bp)	<b>BuCa</b> (n=211)	<b>BaRo</b> (n=205)	<b>Székely</b> (n=178)	<b>Csángó</b> (n=182)
Polimorf pontok	183	109	176	123
Tranzíciók	158	96	152	104
Transzverziók	12	3	11	7
Megfigyelt haplotípusok	180 (167 egyedi)	57 (33 egyedi)	135 (105 egyedi)	84 (59 egyedi)
Véletlen egyezési valószínűség (RMP)	1,00%	8,97%	1,04%	5,55%
Haplotípus-párok átlagos eltérése	9,5	9,8	10,8	10,7
Genetikai diverzitás	0,995	0,915	0,995	0,949

\* A policitozin szálak C inzercióit a 16193, 309 és 573 nukleotid pozíciókban kihagyva a számításokból

# *Szekvencia-párok között megfigyelt eltérések száma az egyes populációkban*



## *Leggyakoribb mtDNS haplotípusok a magyar populációs mintákban*

---

### Budapest Caucasians (211):

263G-315.1C-16519C (14, 6.6 %; H1)

### Csíkszeredai Székelyek (178):

1. 263G-315.1C-16519C (7, 3.9 %; H1)

2. 263G-315.1C-16172C-16173T-16519C (7, 3.9 %; H\*)

### Gyimesi Csángók (182):

1. 73G-146C-152C-263G-315.1C-498del-16224C-16311C-16519C (35, 19.2%; K1c)

2. 73G-185A-189G-263G-295T-315.1C-462T-489C-16069T-16126C (15, 8.2 %; J1)

### Budapesti Askenázi (173):

73G-114T-263G-315.1C-497T-16224C-16234T-16311C-16519C (19, 11%; K1a)

### Baranya Roma (205):

73G-263G-315.1C-489C-524.1A-524.2C-16129A-16223T-16291T-16298C-16519C  
(M5a; 23.9 %)

## *mtDNA haplogroup designation based on HVS-I and HVS-II*

TABLE 1—*List of the most important sites and states that identify major Caucasian haplogroups.*

Haplogroup	Polymorphism 1	Polymorphism 2	Polymorphism 3	Polymorphism 4	Polymorphism 5
H*	73A				
T	16126C	16294T			
J	16069T	16126C	295T		
K	16224C	16311C			
U5*	16270T				
I	16223T	199C	204C	250C	
V	16298C	72C			
W	16223T	189G	195C	204C	207A
M	16223T	16298C			
X	16189C	16223T	16278T	195C	

\* The most common Haplogroup, H, is usually defined based on the absence of these other variants and often it is associated with 73A (3). Subcluster U5 is listed as the most common form of the U haplogroup. For a more detailed discussion of additional informative polymorphisms see the text.

# mtDNS haplocsoportok

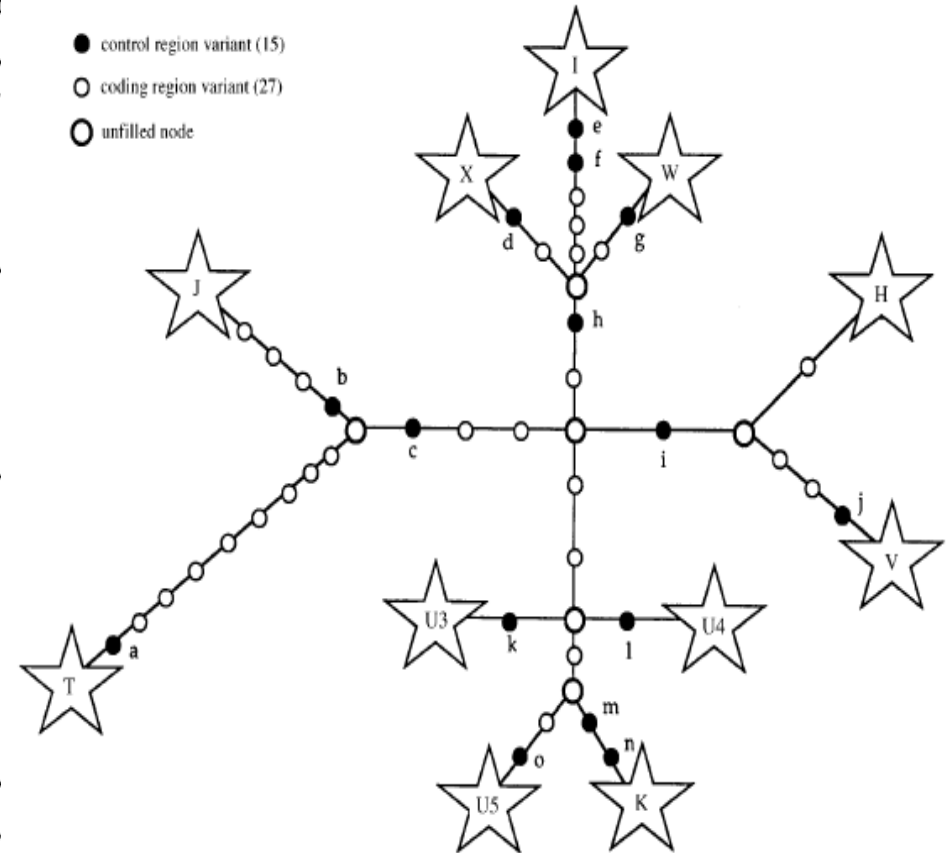
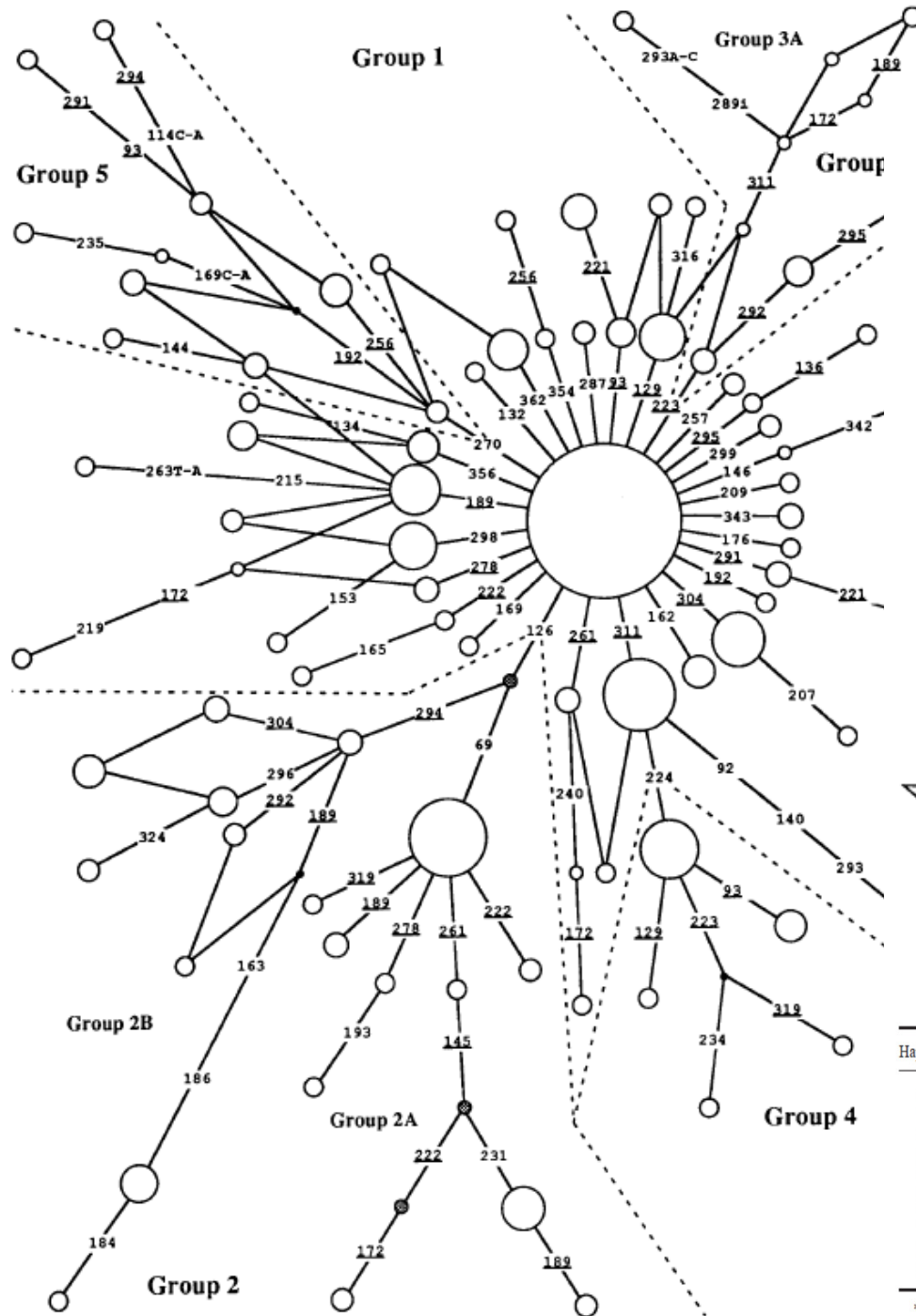
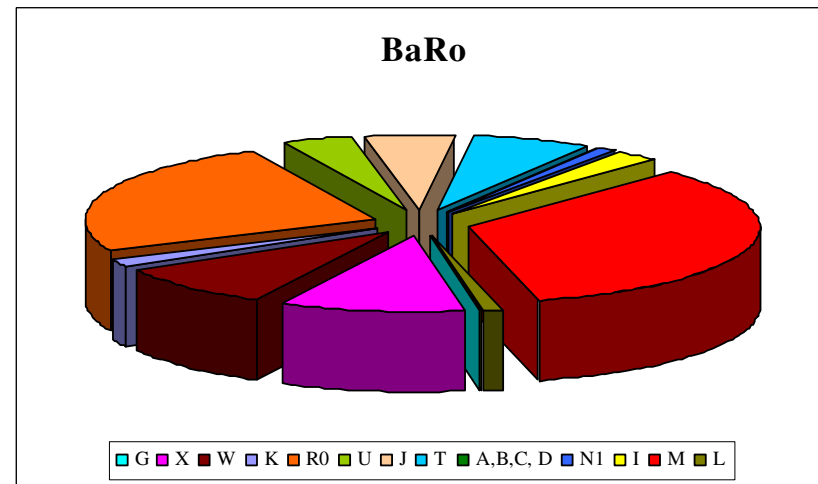
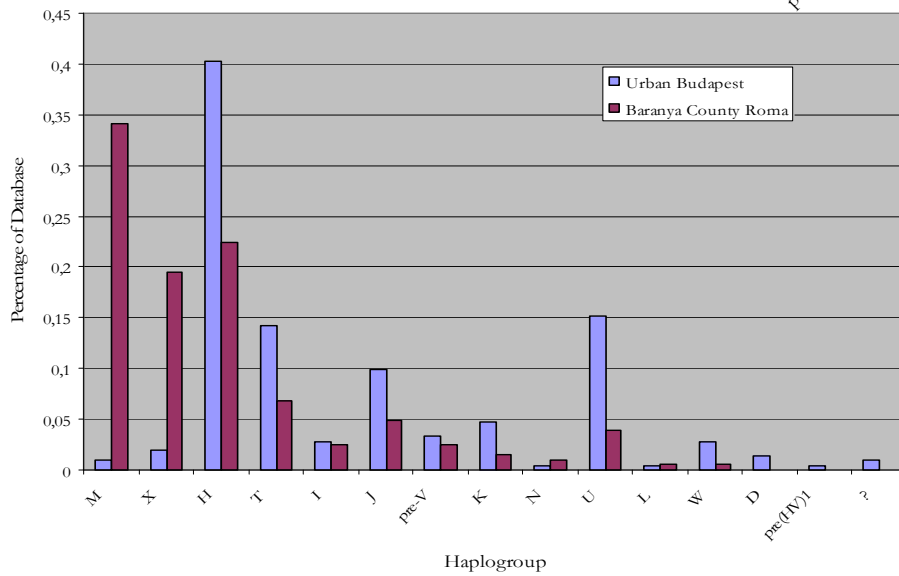
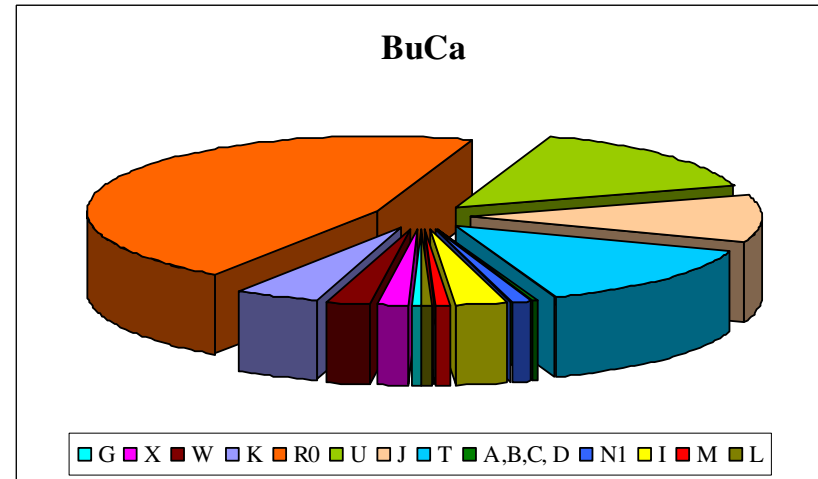
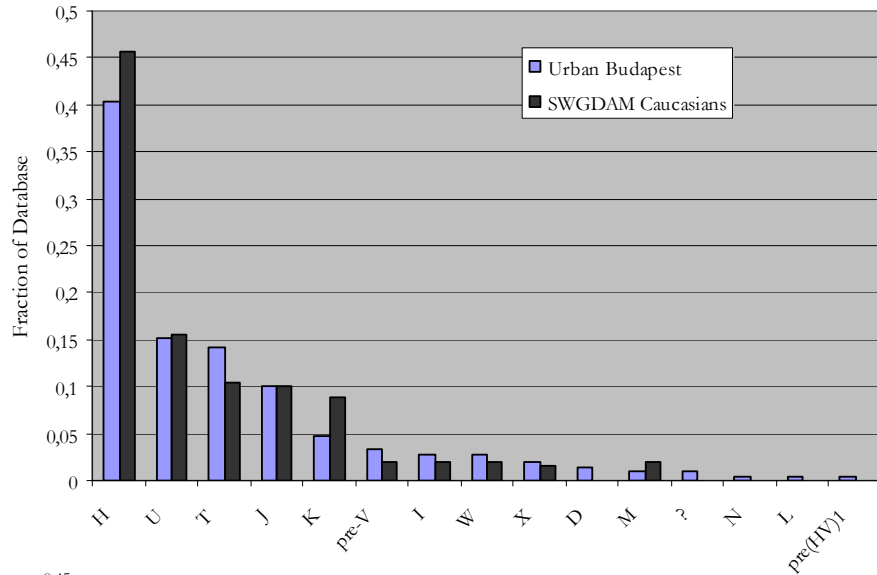


TABLE 1—List of the most important sites and states that identify major Caucasian haplogroups.

Haplogroup	Polymorphism 1	Polymorphism 2	Polymorphism 3	Polymorphism 4	Polymorphism 5
H*	73A				
T	16126C	16294T			
J	16069T	16126C	295T		
K	16224C	16311C			
U5*	16270T				
I	16223T	199C	204C	250C	
V	16298C	72C			
W	16223T	189G	195C	204C	207A
M	16223T	16298C			
X	16189C	16223T	16278T	195C	

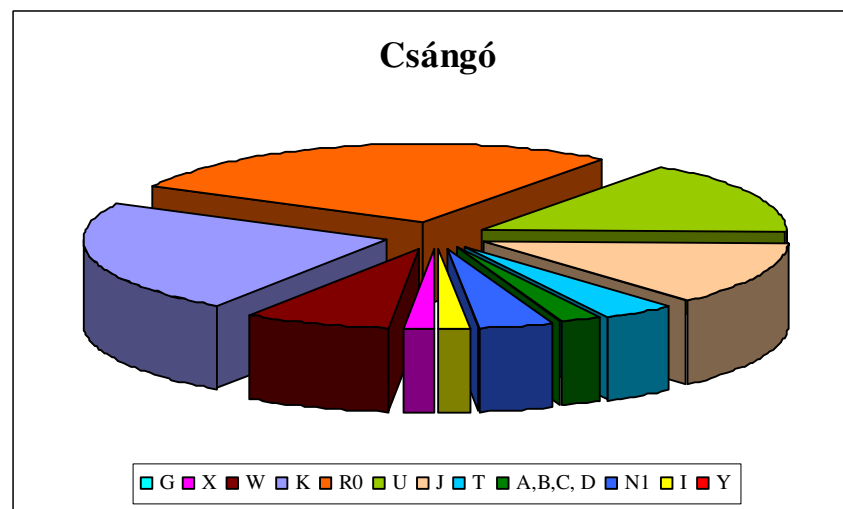
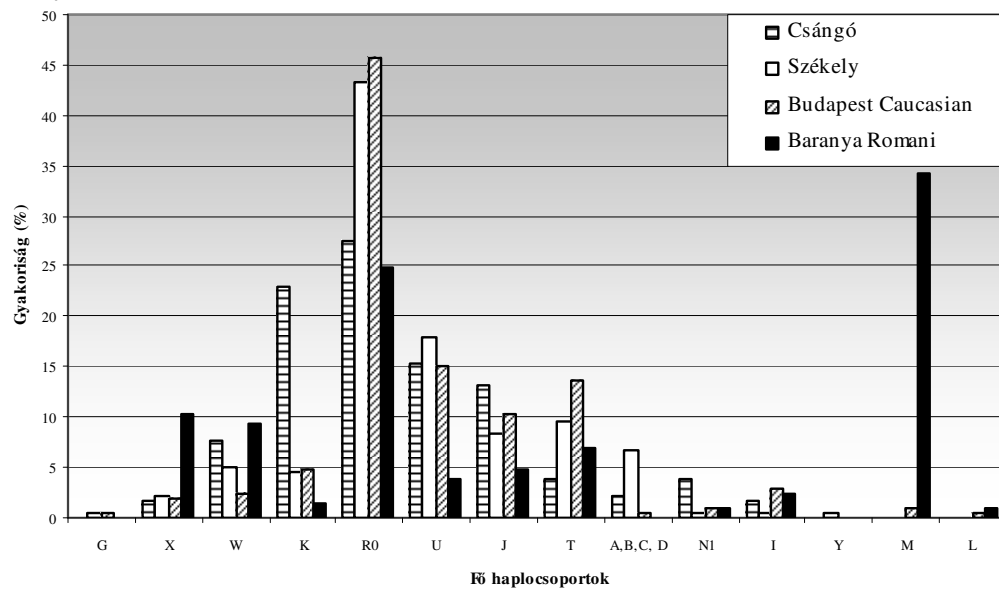
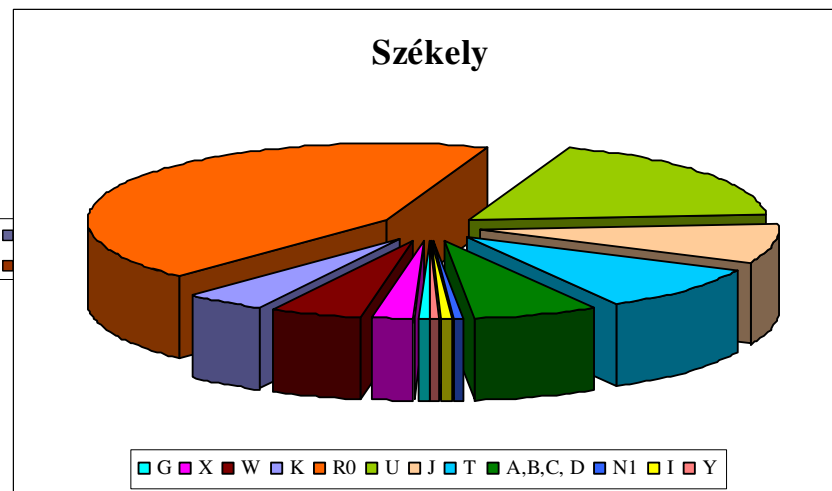
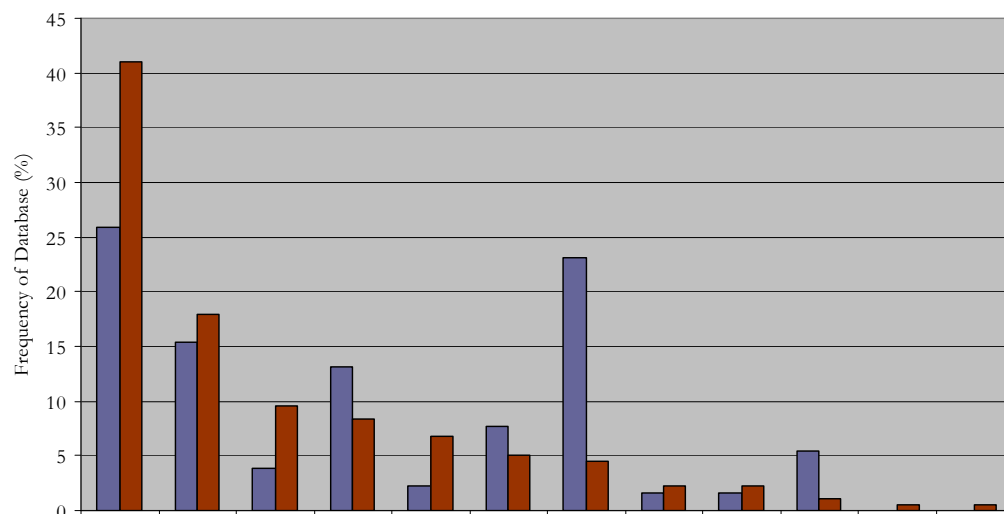
\* The most common Haplogroup, H, is usually defined based on the absence of these other variants and often it is associated with 73A (3). Subcluster U5 is listed as the most common form of the U haplogroup. For a more detailed discussion of additional informative polymorphisms see the text.

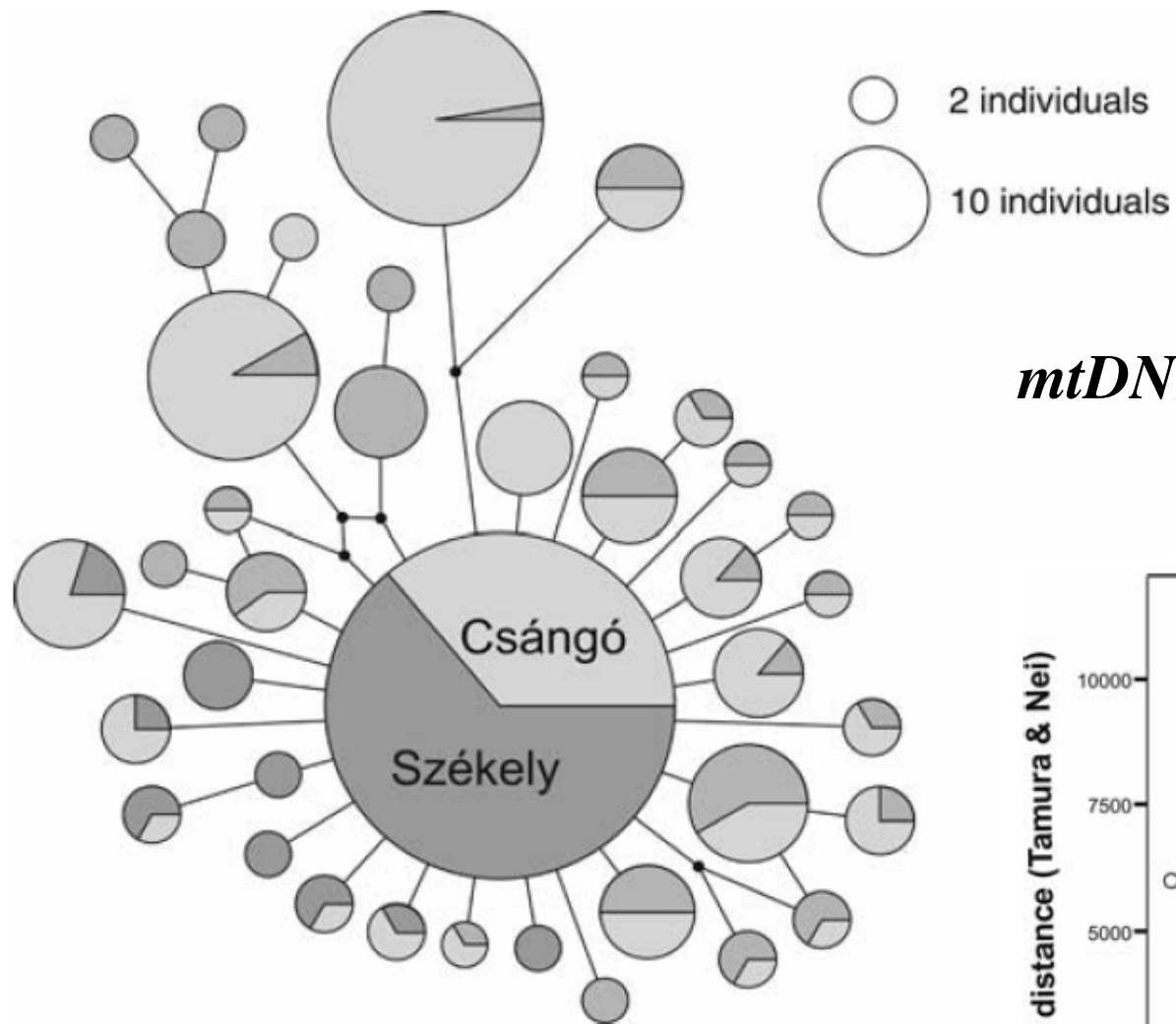
# Mitokondriális DNS haplocsoport eloszlások



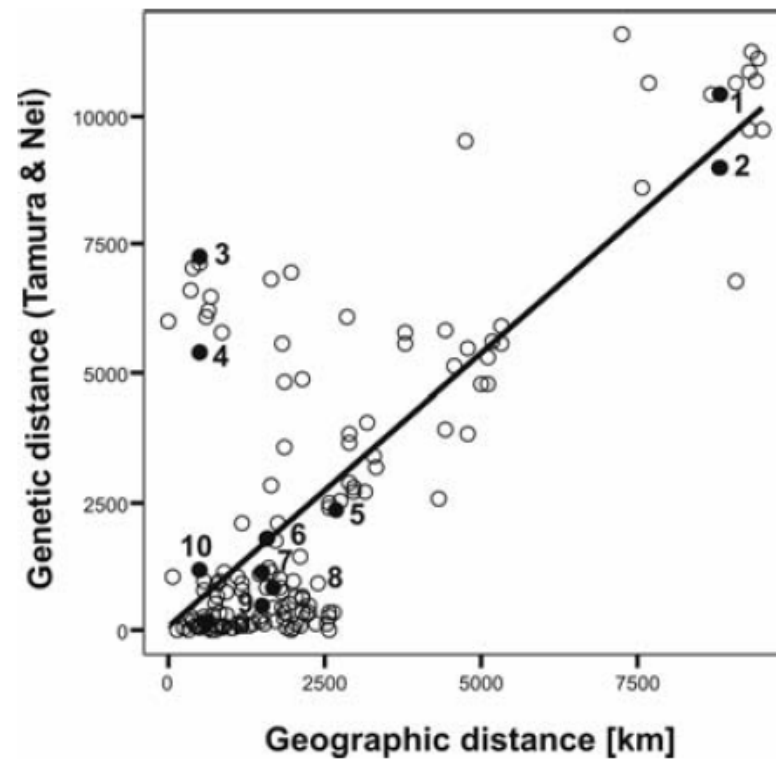


# Mitokondriális DNS haplocsoport eloszlások





*mtDNS genetikai távolságok*



## *Genetikai struktúráltság: mtDNS szekvenciák AMOVA analízise a magyar populációkban*

<i>P</i> \ $F_{ST}$	Gyimesi Csángó	Csíkszereda Székely	Budapesti Referencia	Baranya Roma
<b>Gyimesi Csángó</b>	-	0,024*	0,025*	0,101*
<b>Csíkszereda Székely</b>	0,000	-	0,001	0,070*
<b>Budapesti Referencia</b>	0,000	0,240	-	0,081*
<b>Baranya Roma</b>	0,000	0,000	0,000	-

\* A populáció-párok között megfigyelt  $F_{ST}$  értékek szignifikáns eltérést mutatnak a  $P = 0,05$  szignifikancia szinten.

Teljes variancia: 5,2 % vezethető vissza populációk közötti eltérésekre.

## *Genetikai struktúráltság: mtDNS szekvenciák AMOVA analízise európai roma populációkban*

	Baranya Roma	Kalderash (bolgár)	Monteni (bolgár)	Lom (bolgár)	Turgovzi (bolgár)	Kalaidjii (bolgár)	Spanyol Roma
Baranya Roma	-	0,021*	0,088*	0,013*	0,035*	0,038*	0,100*
Kalderash		-	0,033*	0,004	0,001	0,034	0,085*
Monteni			-	0,058*	0,044*	0,095*	0,12*
Lom				-	0,011*	0,009	0,084*
Turgovzi					-	0,011*	0,068*
Kalaidjii						-	0,066*
Spanyol Roma							-

\* A populáció-párok között megfigyelt  $F_{ST}$  értékek szignifikáns eltérést mutatnak a  $P = 0,05$  szignifikancia szinten.

Teljes variancia: 5,15 % vezethető vissza populációk közötti eltérésekre.

***Genetikai struktúráltság: az erdélyi mtDNS szekvenciák AMOVA analízise dél-kelet európai populációs adatokkal***

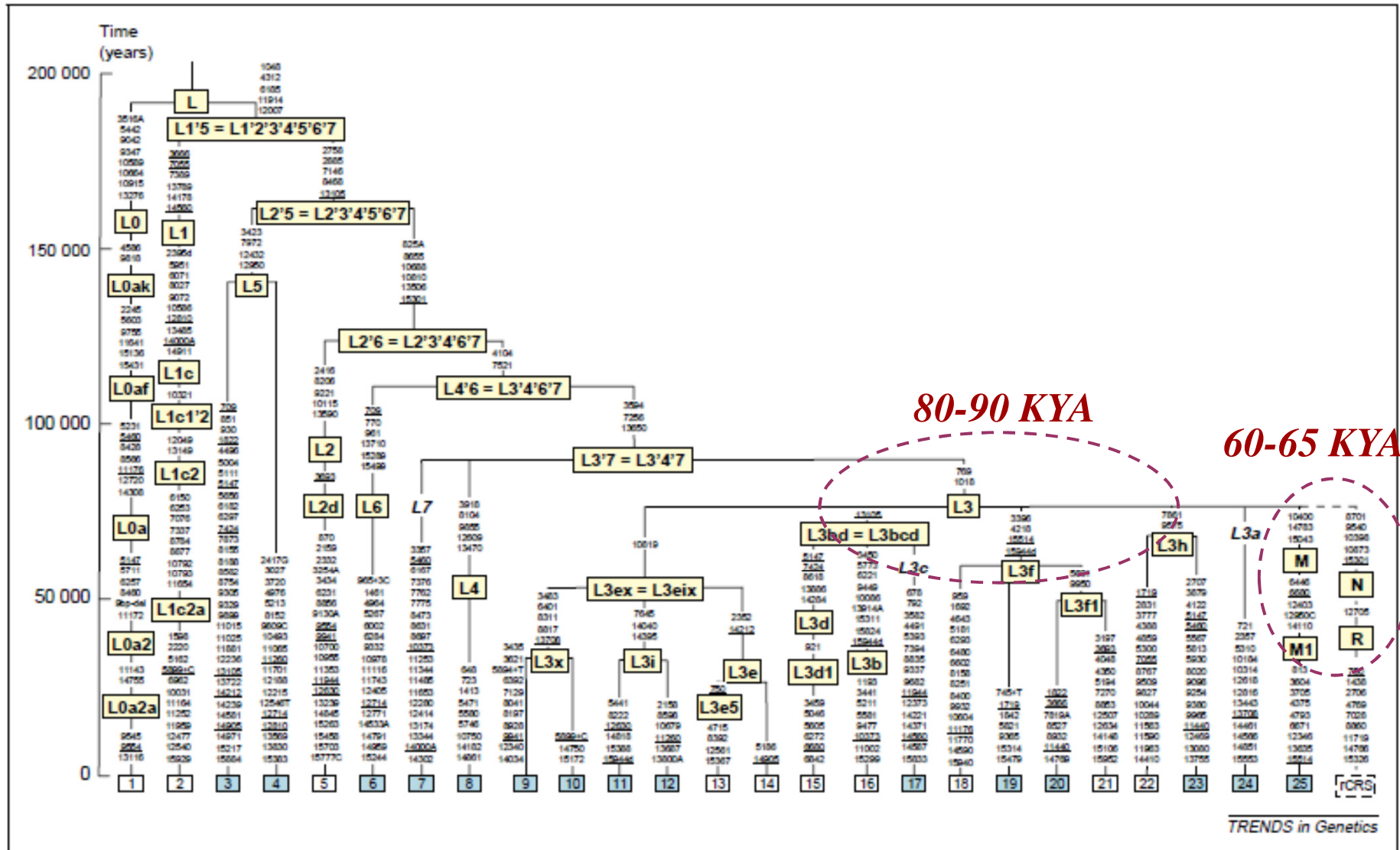
<i>P</i> \ $F_{ST}$	Csángó	Székely	Görög	Macedón
<b>Csángó</b>	-	0,026*	0,027*	0,027*
<b>Székely</b>	0,000	-	0,001	0,001
<b>Görög</b>	0,000	0,131	-	0,000
<b>Macedón</b>	0,000	0,178	0,373	-

\* A populáció-párok között megfigyelt  $F_{ST}$  értékek szignifikáns eltérést mutatnak a  $P = 0,05$  szignifikancia szinten.

Teljes variancia: 3,6 % vezethető vissza populációk közötti eltérésekre.

- *mtDNA control region (D-loop)*  
– *population genetics*
  
- *entire mtDNA molecule*  
– *phylogeny, evolutionary genetics*

# African mtDNA phylogeny (Hgs)

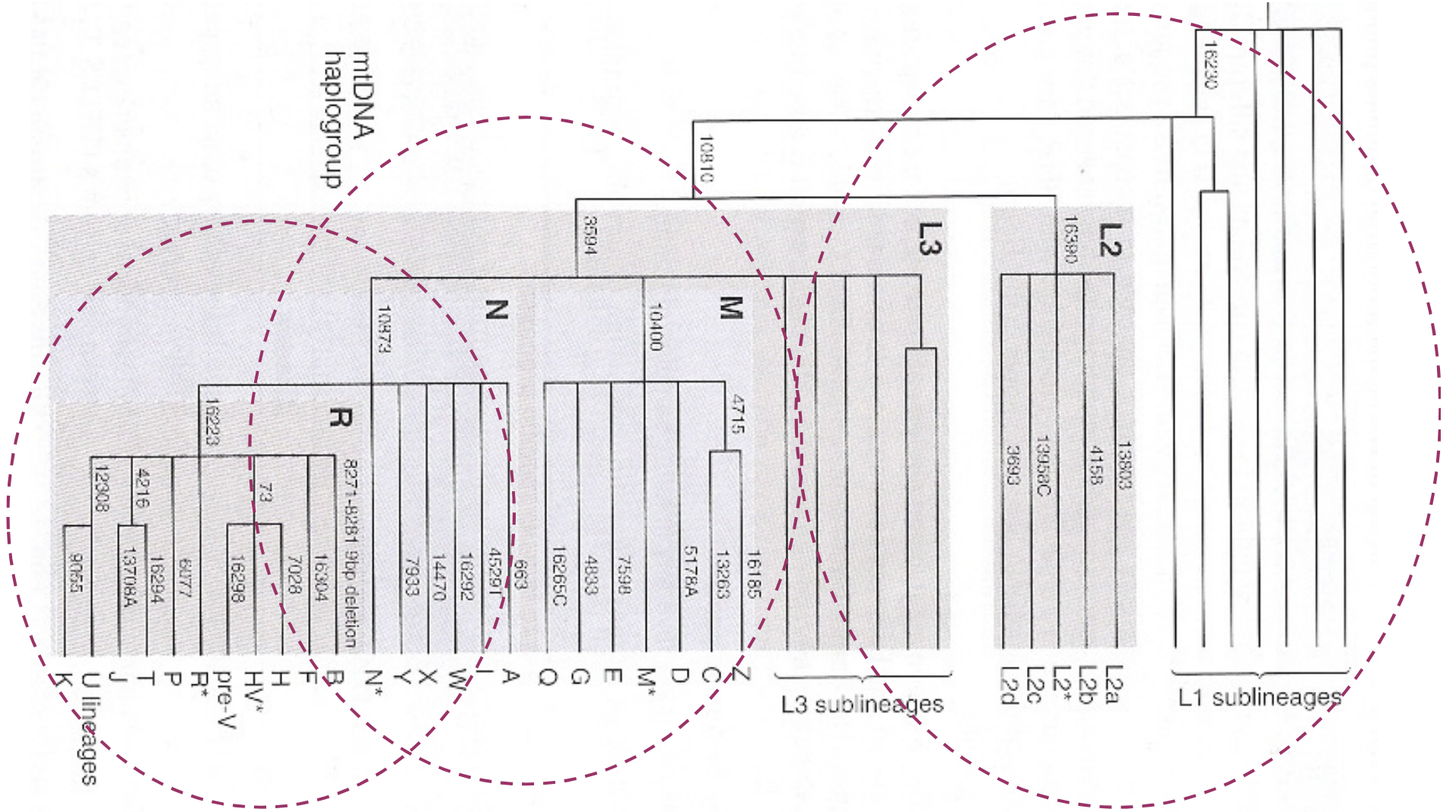


*entire mitochondrial genome sequence*

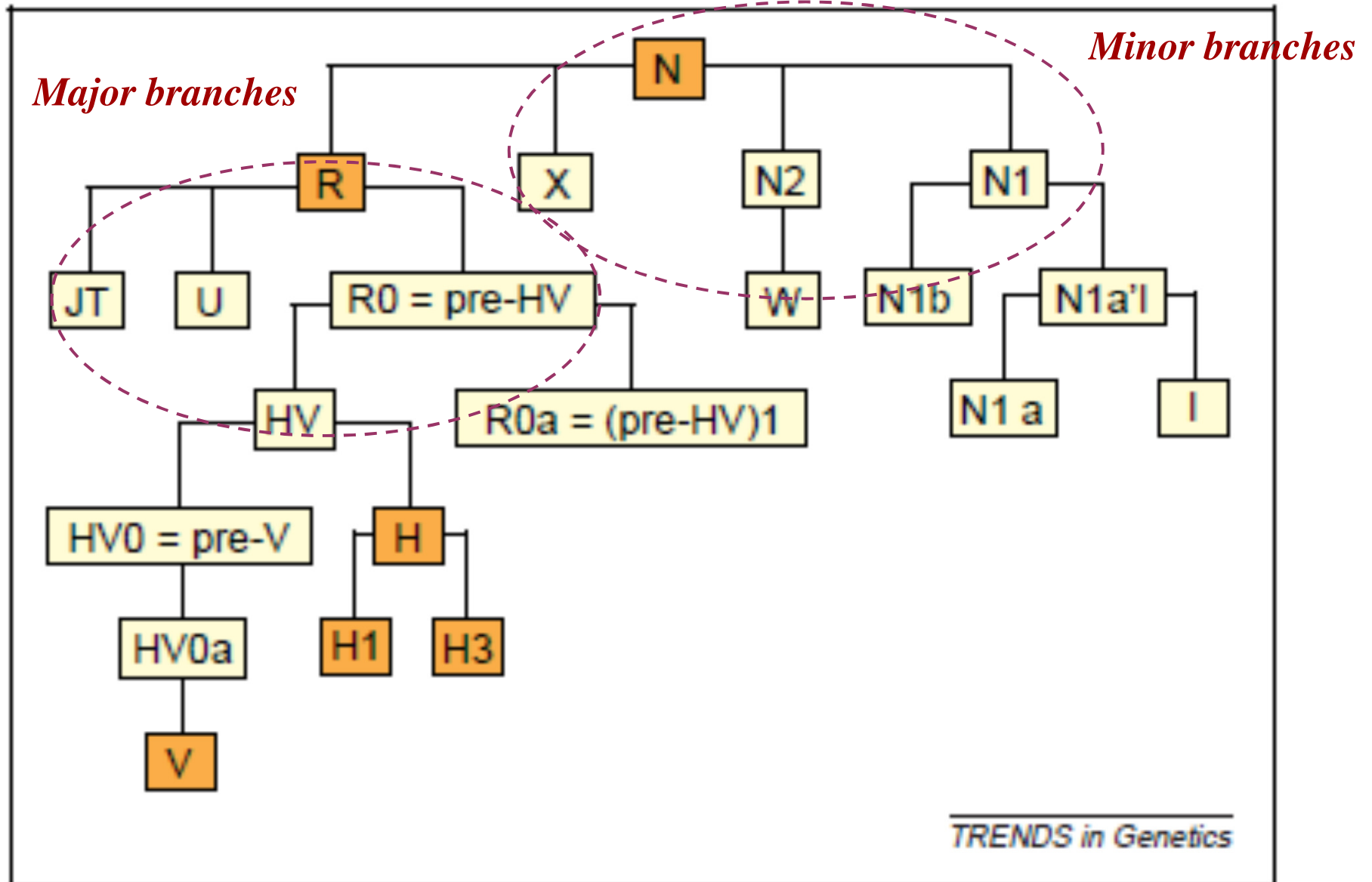
Torrioni et al, Trends Genet, 2006

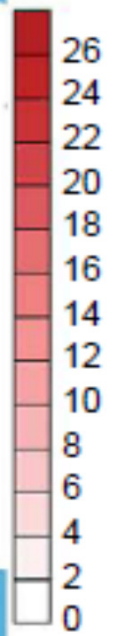
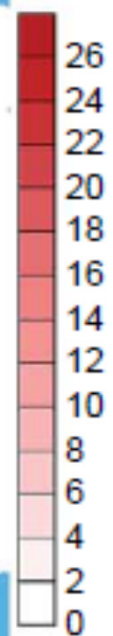
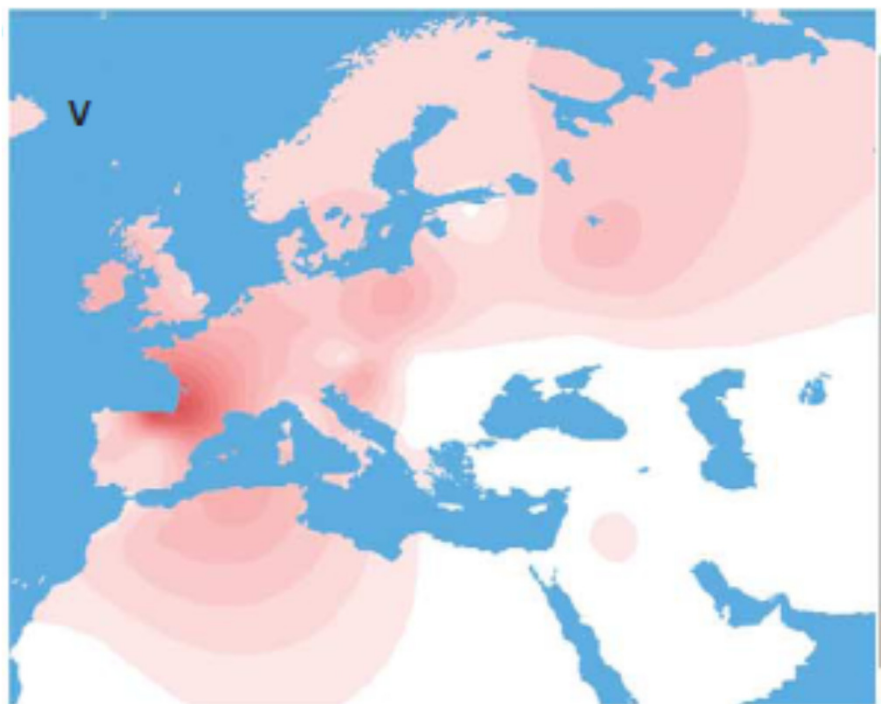
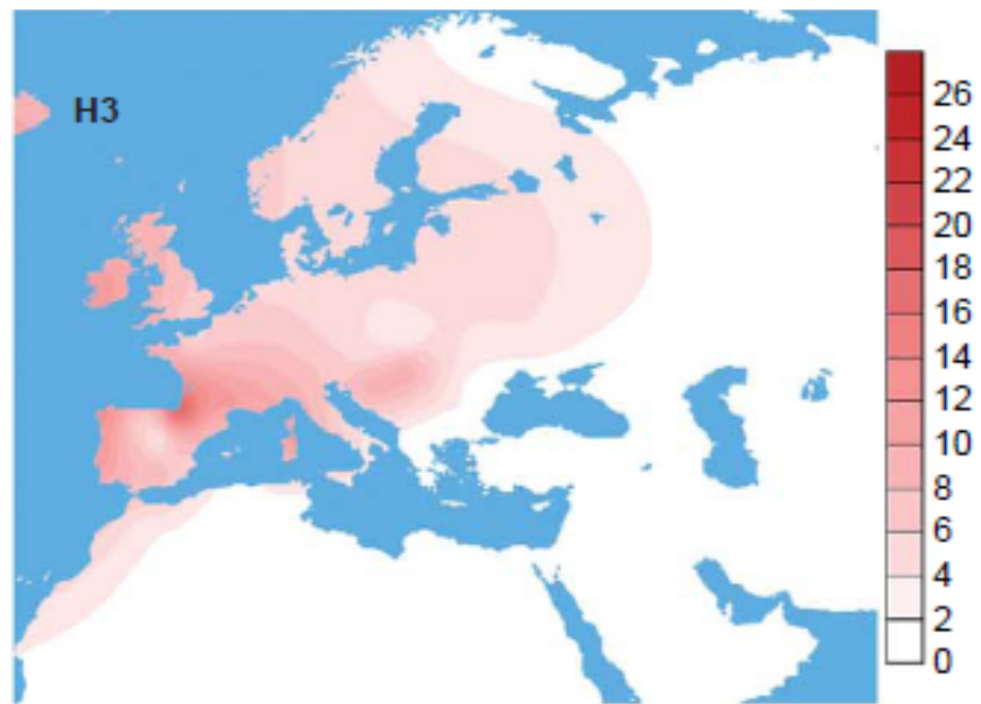
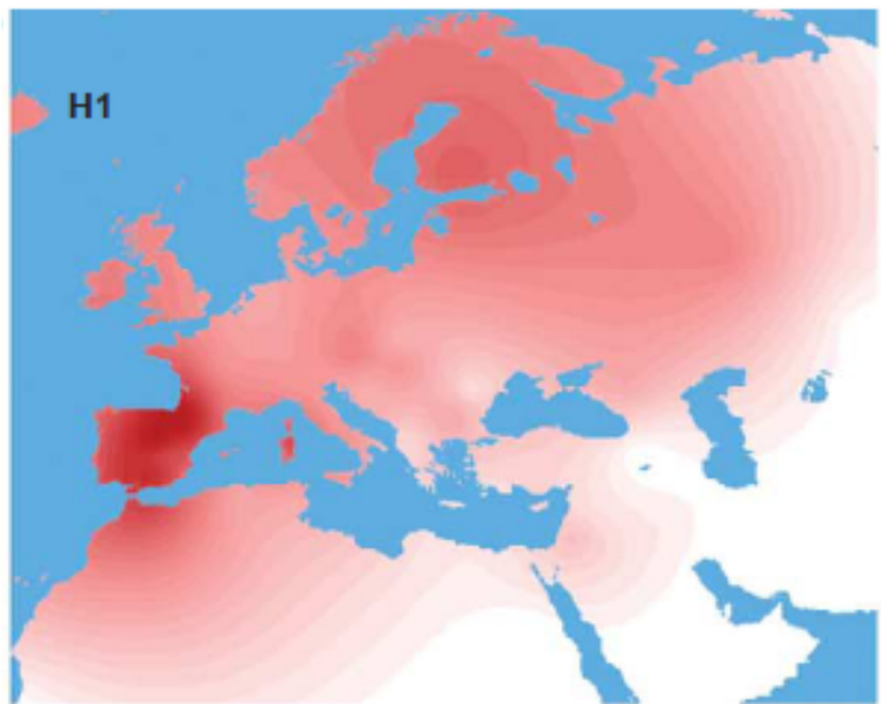


# *mitochondrial DNA lineages (Hgs)*



# European mtDNA lineages (Hgs)







# *West-Eurasian mtDNA phylogeny*

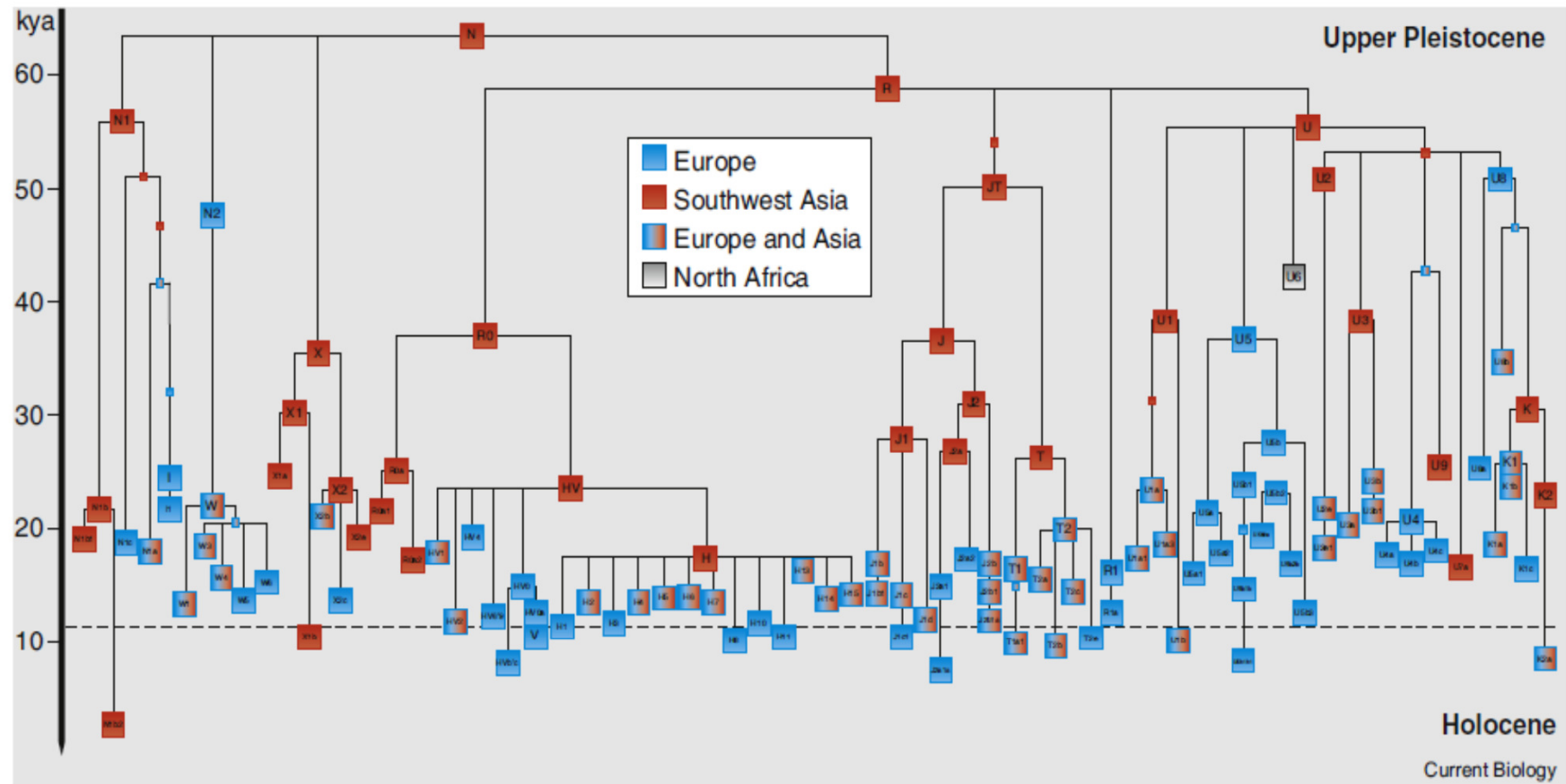
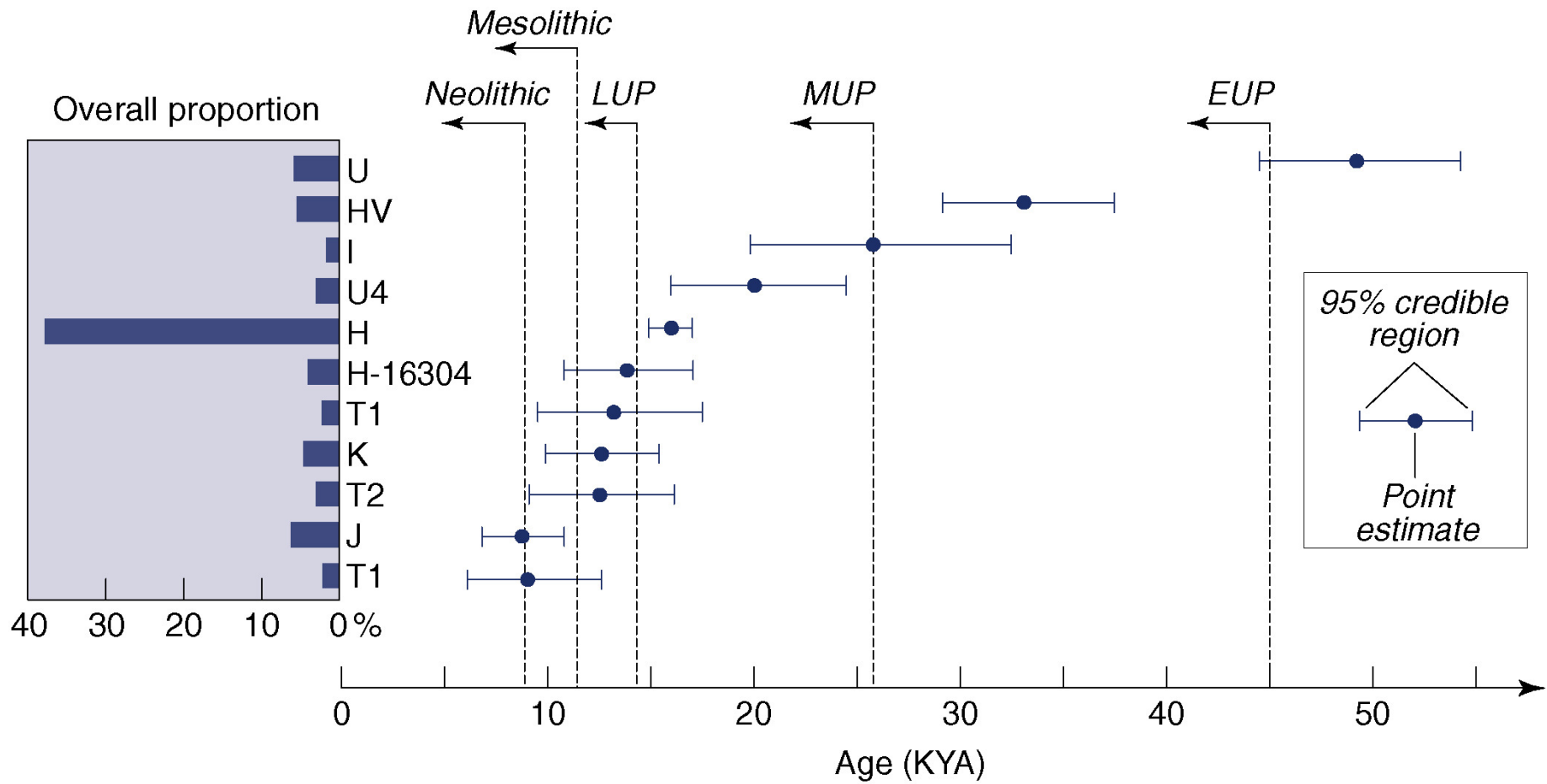


Figure 1. Phylogenetic tree of human mitochondrial DNA haplogroups commonly observed in Europeans, southwest Asians and North Africans. The timescale is based on maximum likelihood (ML) estimates and a time-dependent complete mtDNA genome clock [9]. Haplogroups are colour-coded according to their region of origin (with minor gene flow ignored), although caution is needed given the large sample sizes needed for reliable phylogeographic inferences. Haplogroup nomenclature and an updated dataset can be found at <http://www.phylotree.org> [136].

# *Time-scale of European mtDNA haplogroups*



# *Time-scale of European mtDNA sub-haplogroups*

Table 1. Age estimates with 95% confidence intervals for mtDNA haplogroups referred to in the text.

Clade	ML complete sequence <sup>a</sup>	$\rho$ estimates			
		Complete sequence <sup>a</sup>	Synonymous <sup>b</sup>	Synonymous transitions <sup>c</sup>	Coding region <sup>d</sup>
U	55.8 (48.5; 63.3)	50.7 (40.9; 60.9)	52.7 (37.8; 67.6)	50.4 (36.0; 64.9)	54.4 (41.9; 66.9)
U8	51.3 (44.0; 58.8)	49.1 (30.4; 68.8)	52.9 (22.2; 83.5)	50.5 (20.8; 80.2)	55.3 (31.2; 79.5)
K	31.6 (24.2; 39.2)	25.1 (16.9; 36.3)	22.0 (15.4; 28.6)	20.5 (14.1; 26.9)	28.3 (17.0; 39.6)
K2a	8.8 (5.4; 12.3)	6.8 (3.7; 9.9)	7.5 (3.1; 11.8)	7.2 (3.0; 11.4)	6.2 (3.5; 8.9)
U5	36.9 (27.5; 46.6)	31.3 (21.4; 41.5)	31.7 (13.0; 50.4)	30.3 (12.2; 48.5)	29.7 (17.9; 41.4)
U5b1 (+16189)	20.4 (14.9; 26.1)	17.4 (9.9; 25.2)	25.6 (6.3; 44.9)	24.6 (5.9; 43.3)	19.6 (8.2; 31.0)
U5b1b1	8.3 (4.4; 12.3)	9.0 (4.8; 13.3)	6.1 (0.7; 11.5)	5.9 (0.7; 11.4)	7.3 (3.6; 11.1)
U5b3	13.0 (9.7; 16.3)	13.2 (8.2; 18.2)	9.0 (5.5; 12.3)	8.1 (4.9; 11.3)	10.2 (6.2; 14.2)
U4	20.8 (15.8; 26.0)	17.8 (12.3; 23.4)	12.7 (7.9; 17.5)	12.2 (7.6; 16.8)	20.4 (13.1; 27.8)
I	24.7 (19.0; 30.5)	25.1 (16.9; 33.6)	22.0 (15.4; 28.6)	20.5 (14.2; 26.9)	28.3 (17.0; 39.6)
H	17.5 (15.6; 19.4)	15.7 (13.3; 18.0)	16.2 (12.9; 19.5)	15.2 (12.1; 18.4)	18.1 (14.6; 21.6)
H1	11.1 (9.3; 12.8)	10.8 (8.8; 12.8)	9.5 (7.2; 11.7)	8.8 (6.7; 11.0)	11.0 (9.1; 12.9)
H3	11.5 (8.9; 12.9)	10.8 (8.5; 13.2)	9.5 (6.3; 12.6)	8.8 (5.8; 11.7)	11.1 (8.4; 13.8)
H5	13.9 (10.7; 17.1)	12.4 (7.3; 17.7)	15.1 (3.0; 27.2)	14.6 (2.9; 26.4)	16.1 (7.7; 24.6)
V	11.0 (8.5; 13.5)	10.2 (7.9; 12.5)	10.7 (7.6; 13.8)	10.1 (7.1; 13.1)	13.2 (9.7; 16.7)
J2a1a	7.7 (1.7; 13.9)	7.9 (2.7; 13.3)	5.3 (0; 13.4)	5.1 (0; 13.0)	9.8 (1.8; 16.6)

Age estimates are given in thousands of years and were estimated from a complete mtDNA genome database of 956 West Eurasian samples for ML and 1898 for  $\rho$ .

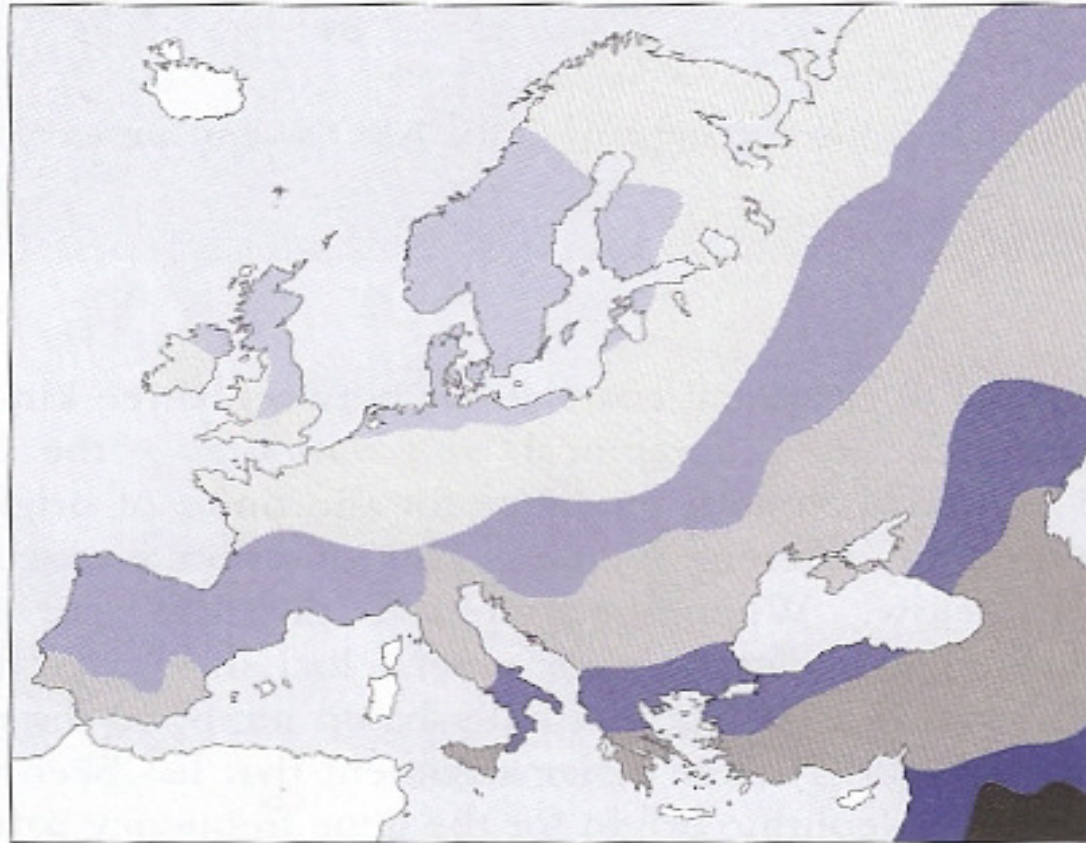
<sup>a</sup> Using the time-dependent clock of Soares *et al.* [9], with ML and  $\rho$ .

<sup>b</sup> Using the synonymous clock of Soares *et al.* [9].

<sup>c</sup> Using the synonymous clock of Kivisild *et al.* [142] rescaled by Perego *et al.* [143].

<sup>d</sup> Using the coding-region clock of Mishmar *et al.* [144] rescaled by Perego *et al.* [143].

*Demic diffusion – synthetic map based on PCA of classical genetic data*



**Figure 10.11:** Synthetic map of Europe and Western Asia obtained using the first principal component of classical genetic data.

Redrawn from Cavalli-Sforza *et al.* (1994).



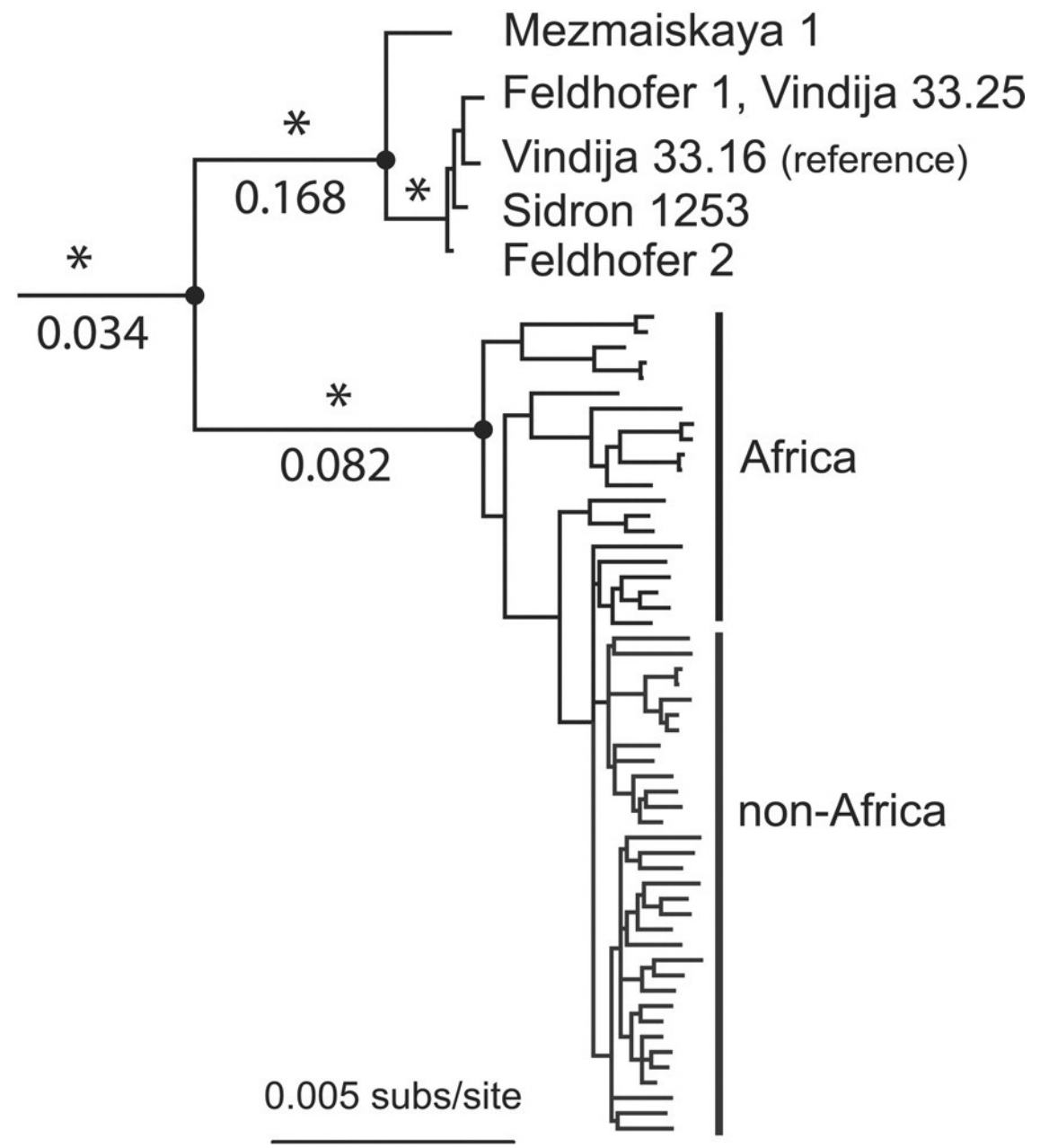
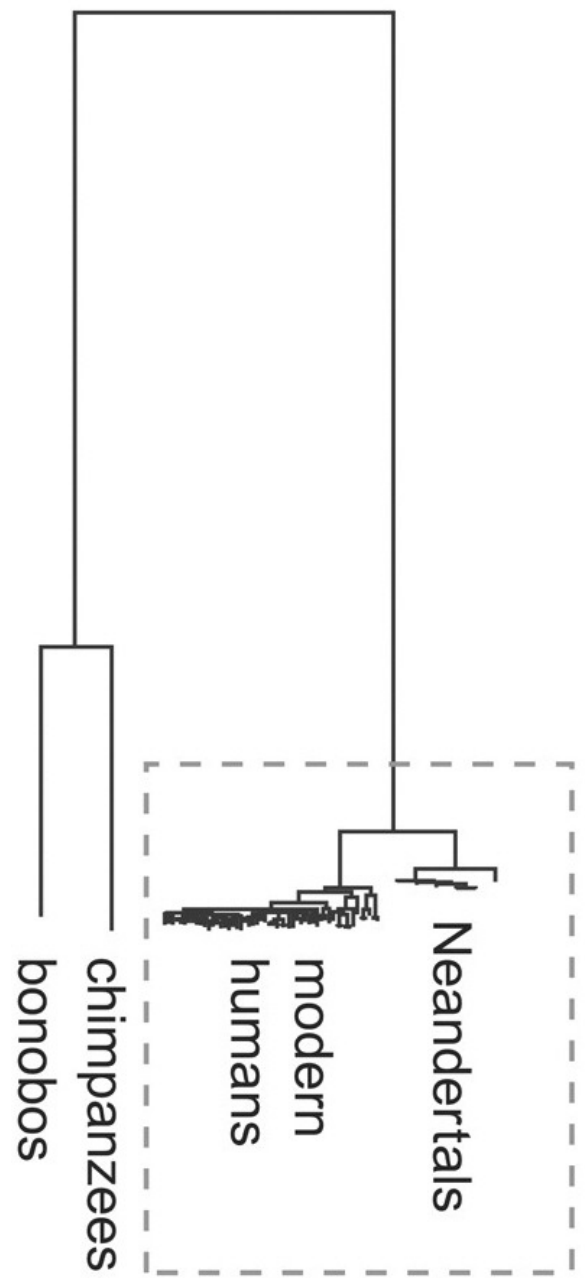
*mt genome sequenced Neanderthal remains*

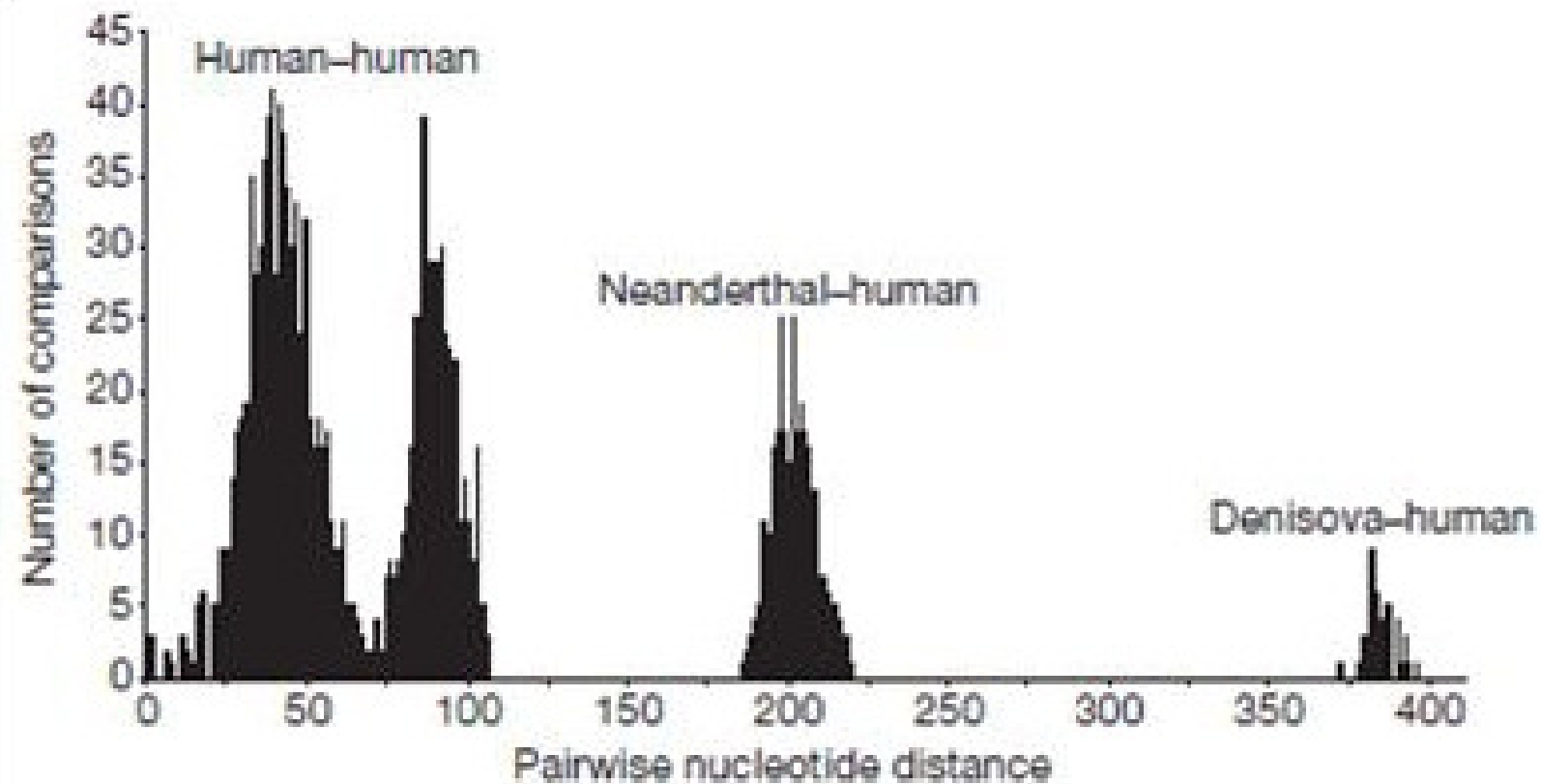


**Table 1.** Mitochondrial DNA variation in Neandertals and modern humans.

Sample	Length*	<i>N</i> †	Hap‡	§§	MPWD	$\theta_{\pi}$ (%)¶
		<i>All sites</i>				
Neandertals	16,565	6	5	55	20.4	0.123
Modern humans	16,547	54	54	658	60.5	0.365
African	16,556	21	21	365	76.5	0.462
Non-African	16,554	33	33	364	38.1	0.230
European	16,566	9	9	87	23.7	0.143
European (expanded)#	16,565	30	29	260	32.3	0.195
		<i>Third codon positions**</i>				
Neandertals	3,575	6	5	22	8.3	0.231
Modern humans	3,575	54	52	283	23.8	0.667
African	3,575	21	19	164	32.6	0.911
Non-African	3,575	33	33	134	13.5	0.378
European	3,575	9	9	23	5.9	0.165
European (expanded)#	3,575	30	29	91	10.5	0.293

\*Number of aligned positions excluding alignment gaps. †Number of sequences. ‡Number of distinct mtDNA sequences (haplotypes). §Number of variable sites. ¶Mean number of pairwise differences. ¶Average percentage of pairwise differences per site. #Expanded to include additional, previously published complete mtDNA genomes. See SOM for details. \*\*Based on a 10,725-bp concatenated alignment of the 12 mtDNA genes coded on the same strand, excluding regions with overlapping reading frames.

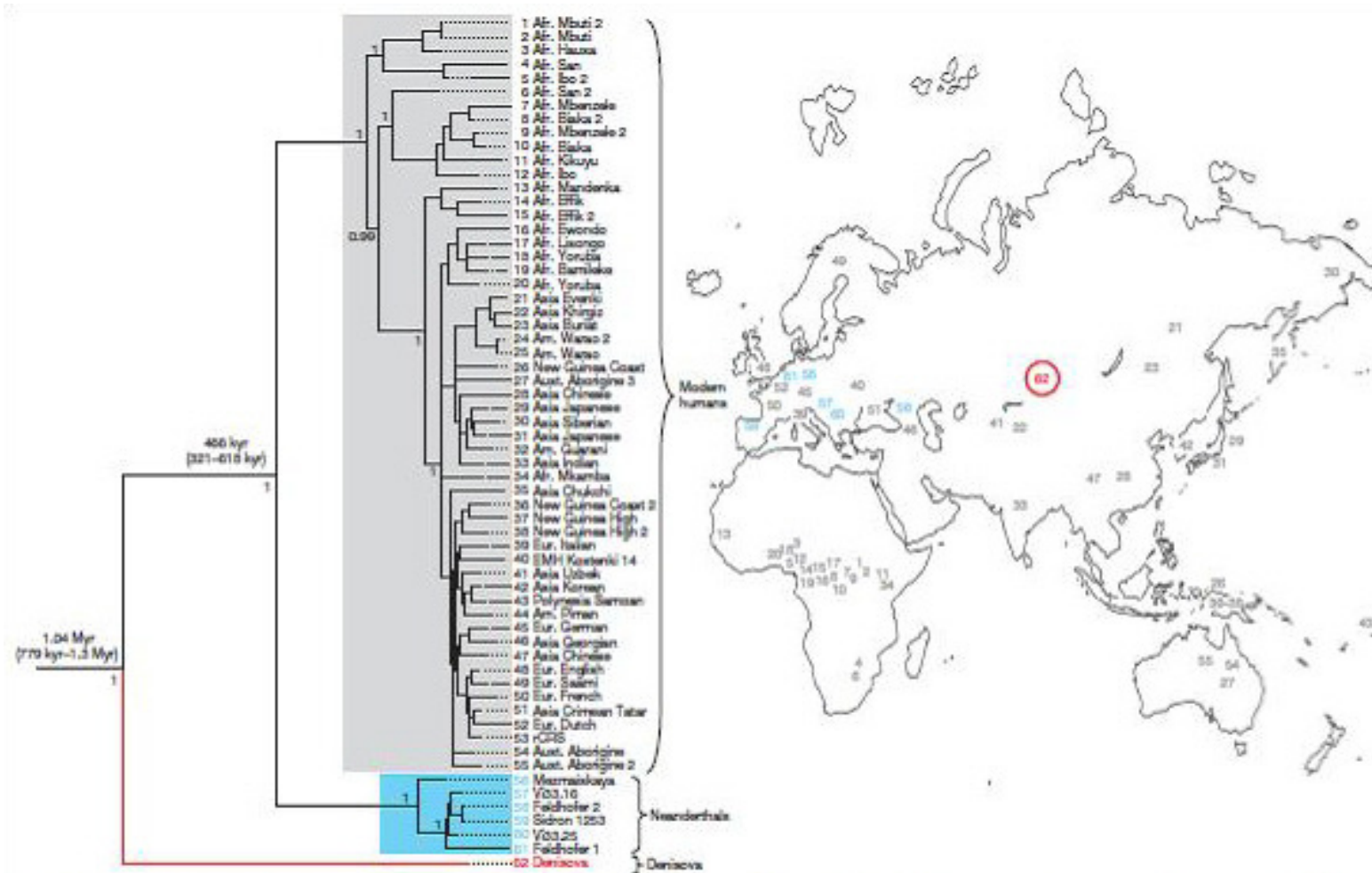




**Figure 2 | Distribution of pairwise nucleotide differences.** Pairwise nucleotide differences from all pairs of complete mtDNAs from 54 present-day and one Pleistocene modern human, six Neanderthals and the Denisova hominin are shown.

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# Denisova mt genome phylogeny



**Figure 3 | Phylogenetic tree of complete mtDNAs.** The phylogeny was estimated with a Bayesian approach under a GTR+I+ $\Gamma$  model using 54 present-day and one Pleistocene modern human mtDNA (grey), 6 Neanderthals (blue) and the Denisova hominin (red). The tree is rooted with a chimpanzee and a bonobo mtDNA. Posterior probabilities are given for

each major node. The map shows the geographical origin of the mtDNAs (24, 25, 32, 44 are in the Americas). Note that two partial mtDNAs sequenced from Teshik Tash and Okladnikov Cave in Central Asia fall together with the complete Neanderthal mtDNAs in phylogenies<sup>4</sup> (not shown).

- Oktatási segédanyag: <https://genetics.elte.hu>
- Felkészülési info: **Emberi evolúció- és humángenetika**
- Felhasználói név: **genetika2013**
- Jelszó: **genetika2013**
  
- M.A. Jobling, M.E. Hurles, C. Tyler-Smith:  
**Human Evolutionary Genetics (Origins, People & Disease)**

Brian Sykes: Éva 7 lánya

Spencer Wells: Az ember útja