



DNA MARKERS & THEIR ROLE IN DNA TYPING

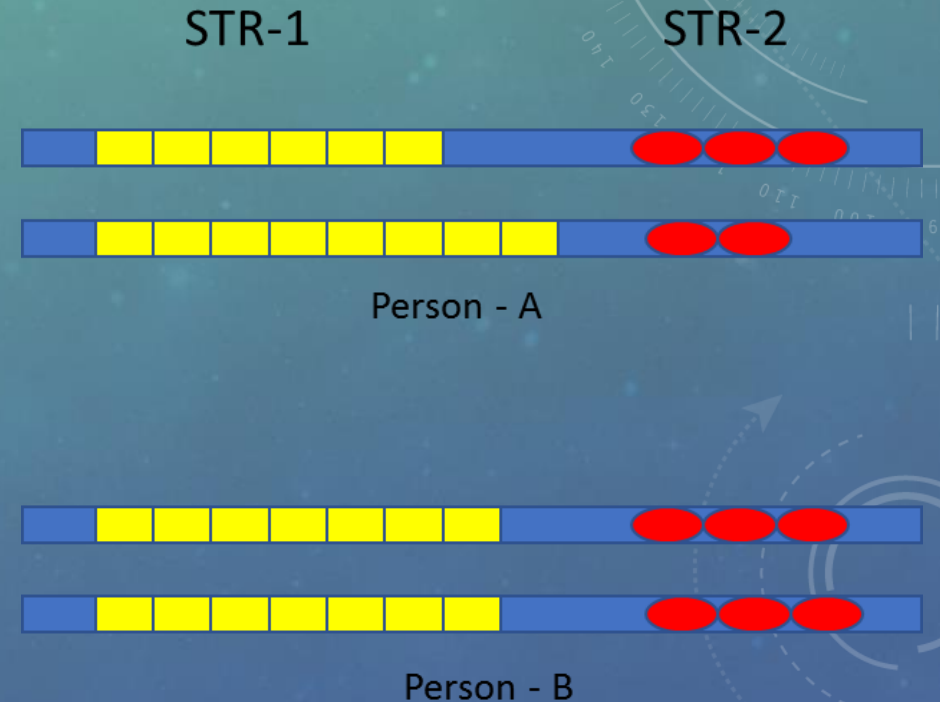
GARGI DESHMUKH
SCIENTIST IN-CHARGE
GENOME FOUNDATION

MARKERS

- A **marker** is a gene or DNA sequence with a known location on a chromosome that can be used to identify individuals or species.
- Markers used for human identity testing are found in the non-coding regions either between genes or within genes (i.e., introns).
- The most recent and probably most rapidly accepted forensic DNA markers are short tandem repeats (STRs)

SHORT TANDEM REPEATS (STRS)

- **Short Tandem Repeats - STRs**
 - 2-7 nucleotide bases per repeat
 - Results in DNA segments of about 500-600 bases in length
 - Shorter segments make it less susceptible to degradation
- Homozygote = both alleles are of the same length
- Heterozygote = alleles differ in length and can be resolved from one another

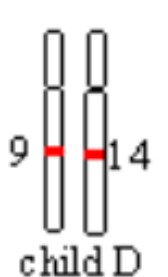
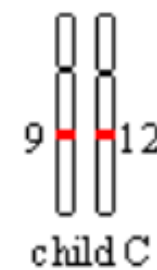
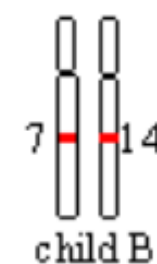
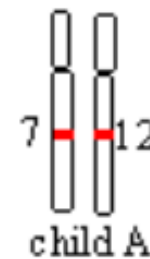
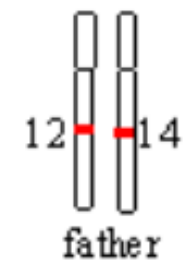
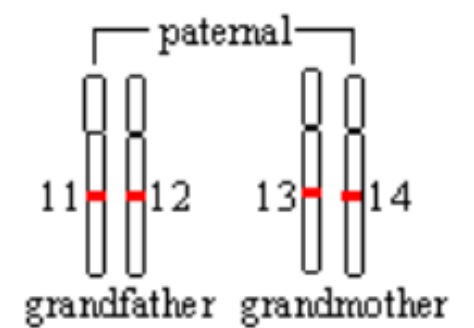
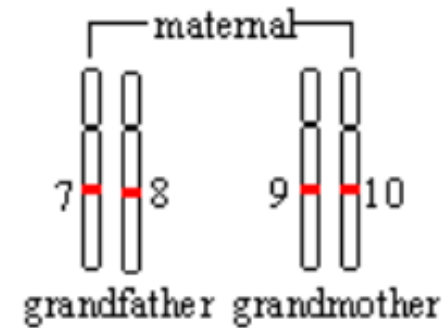


- Currently the most used of all forensic markers
- Individual identification possible
- Type of data used in the FBI CODIS database
- People differ in length at these loci
- Are located in the nuclear DNA (chromosomes)

Length of Repeat Unit	Example
Dinucleotide	(CA) (CA) (CA) (CA)
Trinucleotide	(GCC) (GCC) (GCC)
Tetranucleotide	(ACTG) (ACTG) (ACTG)
Pentanucleotide	(ATATG) (ATATG) (ATATG)
Hexanucleotide	(ATATGA) (ATATGA) (ATATGA)

INHERITANCE OF ALLELES

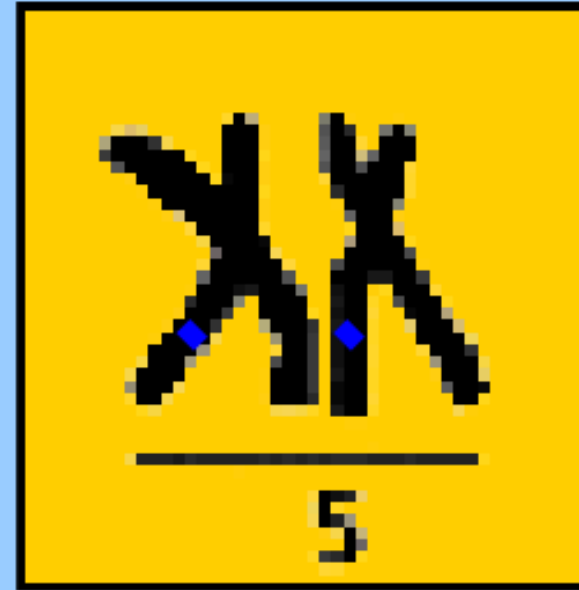
- Every individual inherits half the genetic material from each of the parents.
- The genetic material inheritance for every STR marker is similar.



Example

Locus: D5S818

Alleles: 7,9



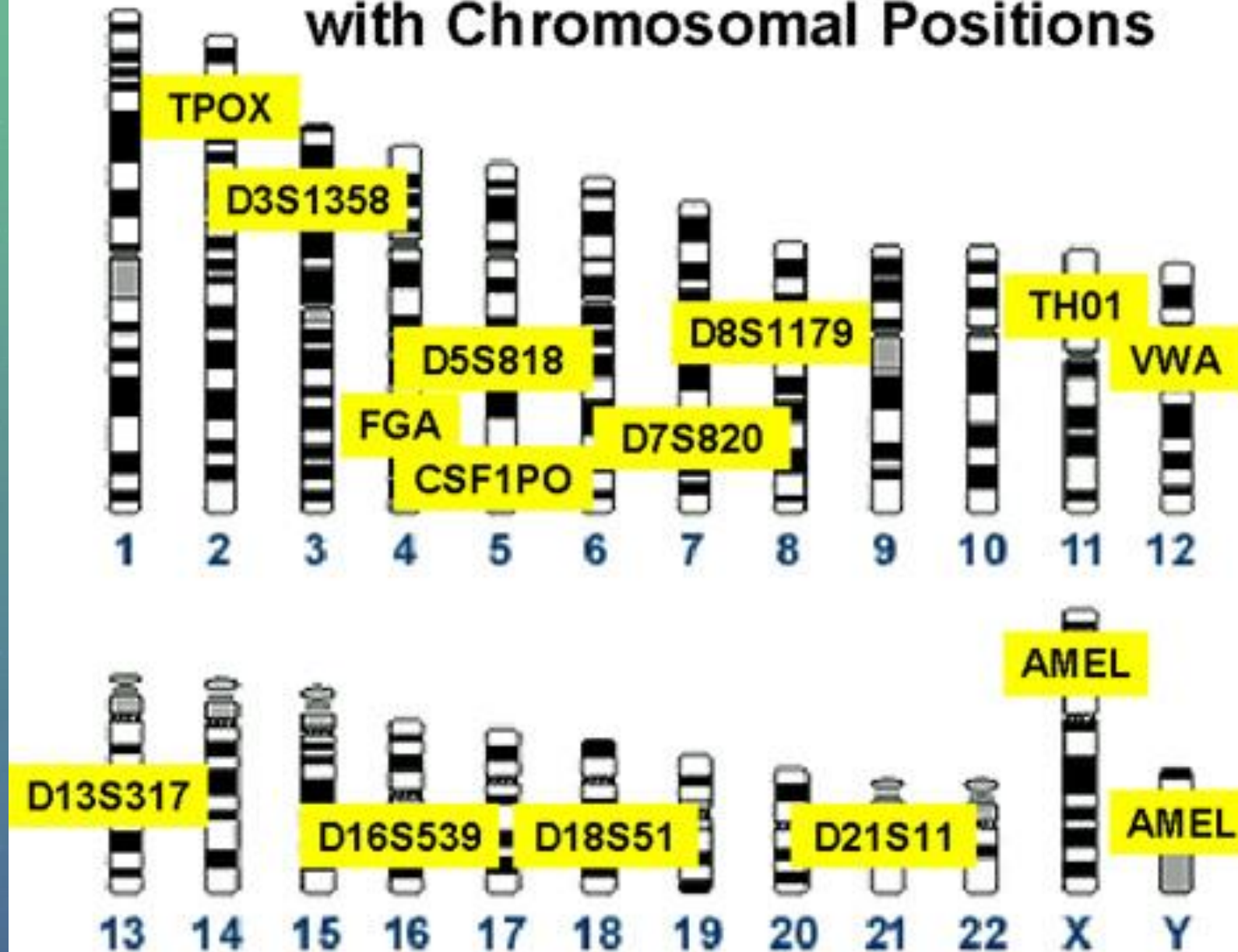
Paternal chromosome 5

CCAGATAGATAGATAGATAGATAGATAGATCC

Maternal chromosome 5

CCAGATAGATAGATAGATAGATAGATAGATAGATAGATCC

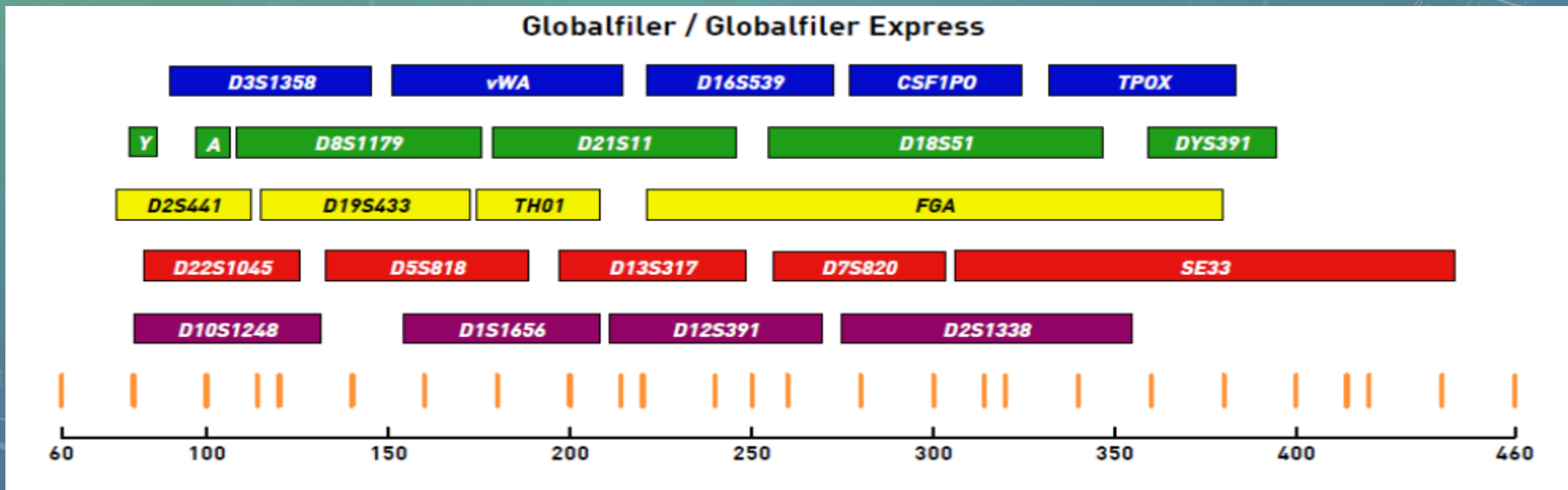
13 CODIS Core STR Loci with Chromosomal Positions

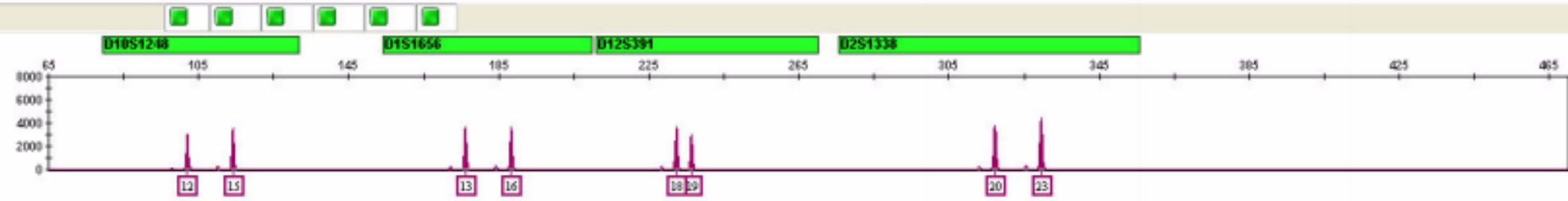
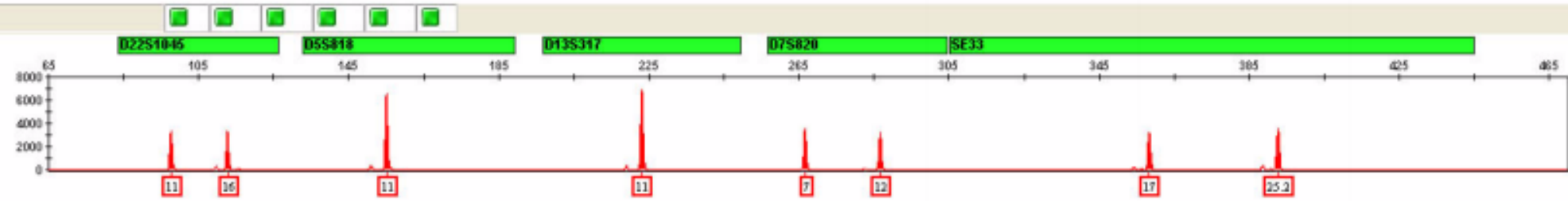
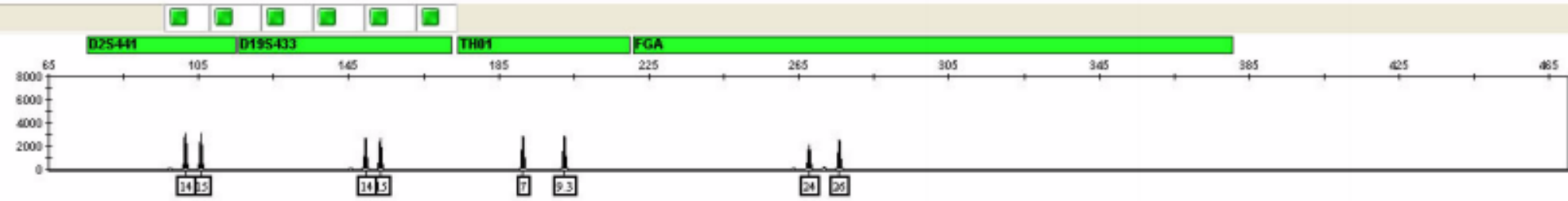
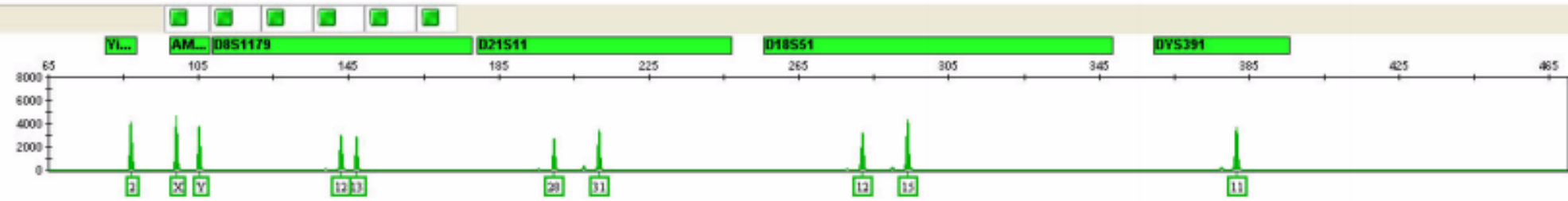
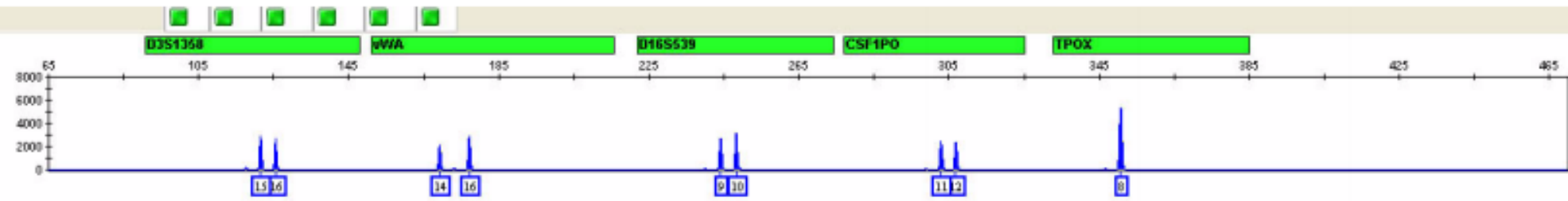


STR Marker	Chromosome	Repeat Sequence	Repeat units	Other Alleles
TPOX	2	AATG	6 - 14	
CSF 1PO	5	AGAT	6 - 15	10.3
D5S818	5	AGAT	7 - 15	
D7S820	7	GATA	6 - 14	
D8S1179	8	TATC	8 - 19	
D13S317	13	TATC	7 - 15	
D16S539	16	GATA	5, 8 - 15	
D3S1358	3	TCTA*	9, 11- 20	15.2, 16.2
FGA	4	CTTT*	15 - 30	16.2 -30.2
				22.3, 34.2, 46.2
TH01	11	AATG*	3, 5 - 12	8.3, 9.3, 10.3, 13.3
VWA	12	TCTA*	11 - 22	15.2
D18S51	18	AGAA*	8 - 27	13.2, 14.2, 15.2
				17.2, 19.2
D21S11	21	TCTA*	24 - 38	24.2 - 35.2

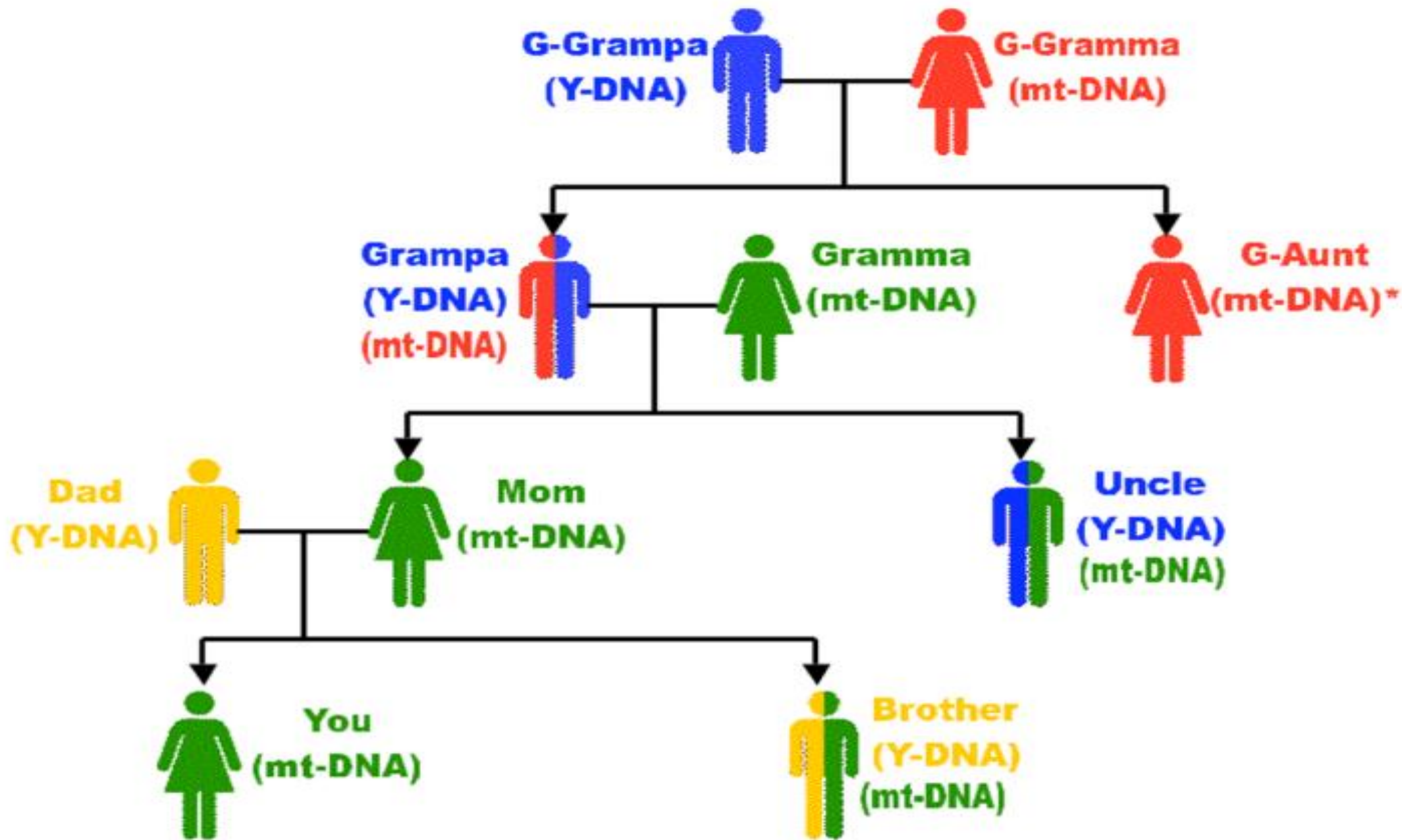
GLOBALFILER KIT

- Uses 6-dye chemistry
- Provides high power of discrimination
- Rapid analysis is seen, with run time of 40 to 120 minutes



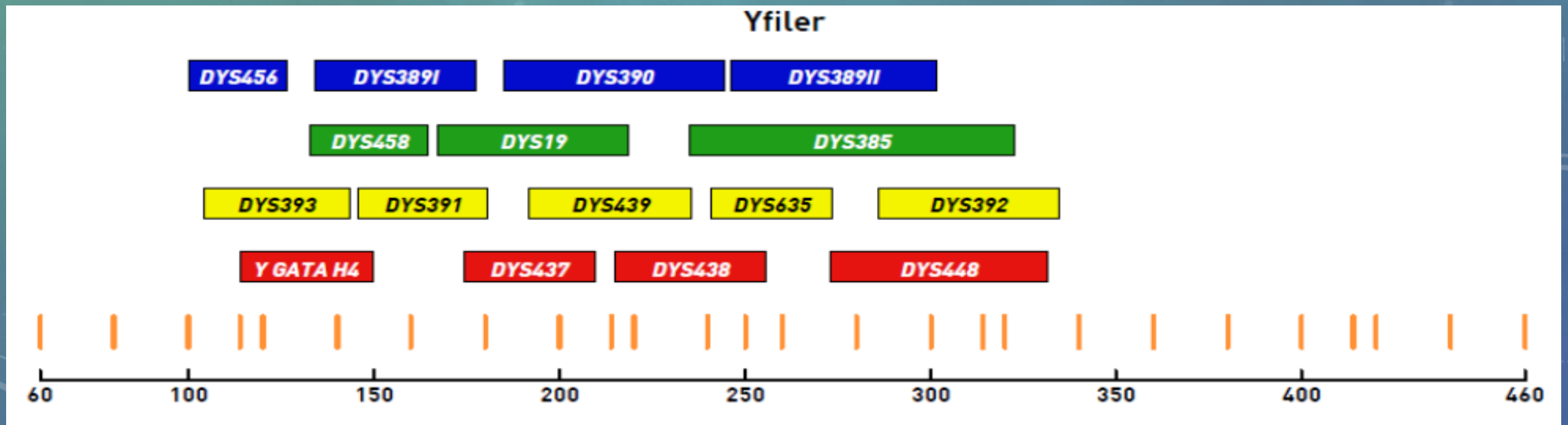


AUTOSOMAL STR LOCI	Male	Female
1. D3S1358	16, 17.2	16, 18.2
2. vWA	15, 17	19, 20
3. D16S539	9, 12	9, 9
4. CSF1PO	10.3, 11.3	9, 10.3
5. TPOX	10.3, 10.3	7.3, 10.3
6. Y indel	1	N.A.
7. Amelogenin	X, Y	X, X
8. D8S1179	13, 14	10, 16
9. D21S11	31, 32.2	30, 32.2
10. D18S51	20, 20	12.2, 14.2
11. DYS391	10	N.A.
12. D2S441	9, 10	11, 13.3
13. D19S433	14, 16	13, 15.2
14. TH01	6, 6	7, 8
15. FGA	21, 23	22, 25
16. D22S1045	12, 12	12, 14
17. D5S818	12, 15	11, 12
18. D13S317	13, 13	10, 13
19. D7S820	10, 12	11, 12
20. SE33	21, 21	22, 30.2
21. D10S1248	15, 15	13, 14
22. D1S1656	11, 14	11, 13

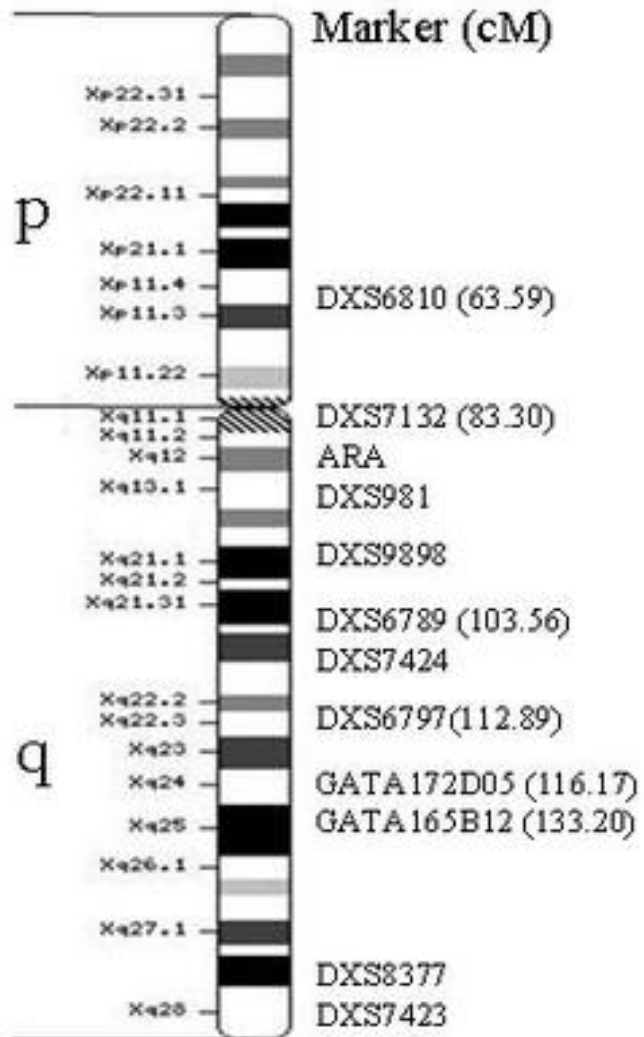


Y-FILER KIT

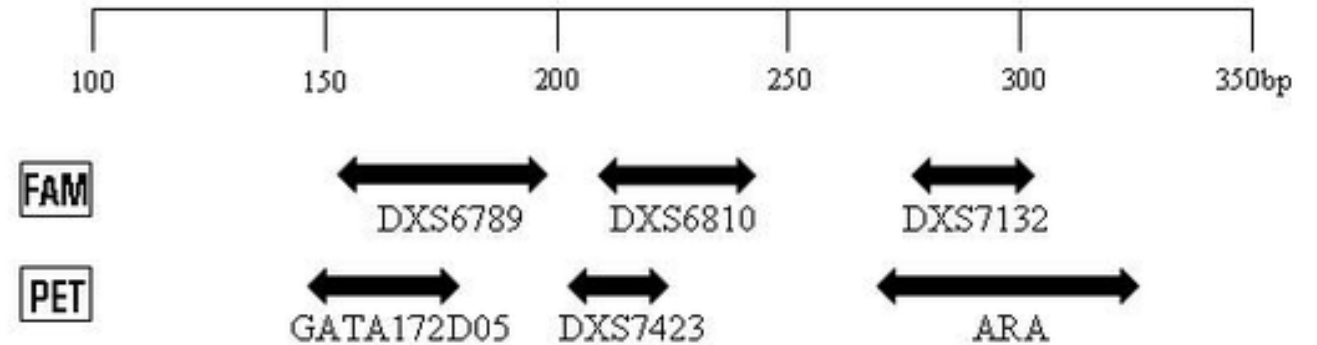
- Uses 5-dye chemistry
- Power of discrimination is good
- Run time of greater than 180 minutes



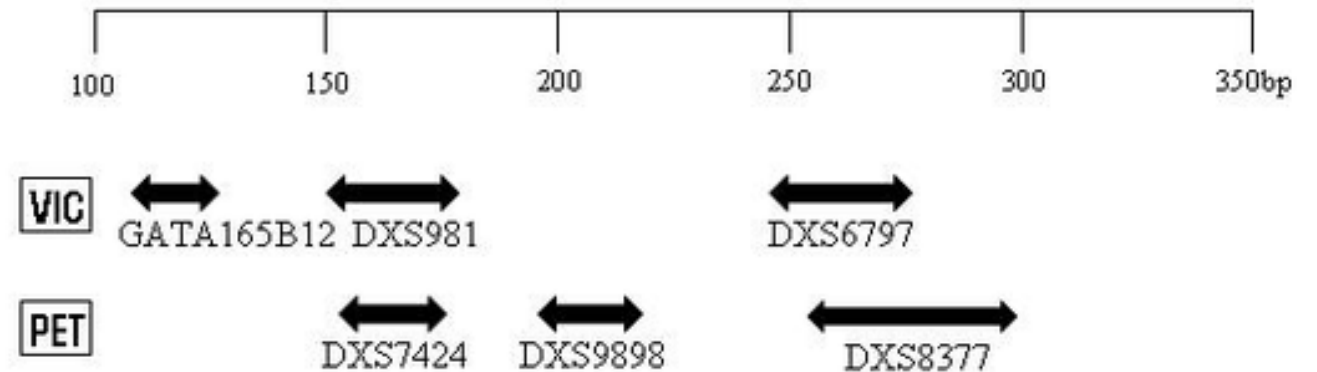
X-CHROMOSOME MARKERS



Hexaplex - A

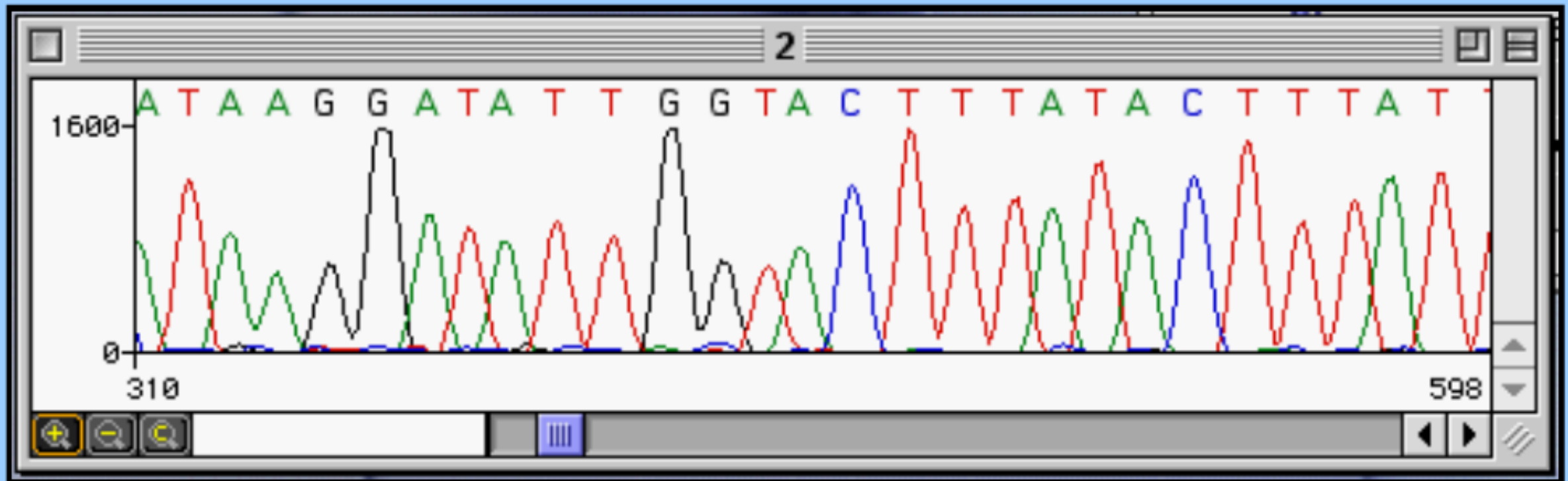


Hexaplex - B



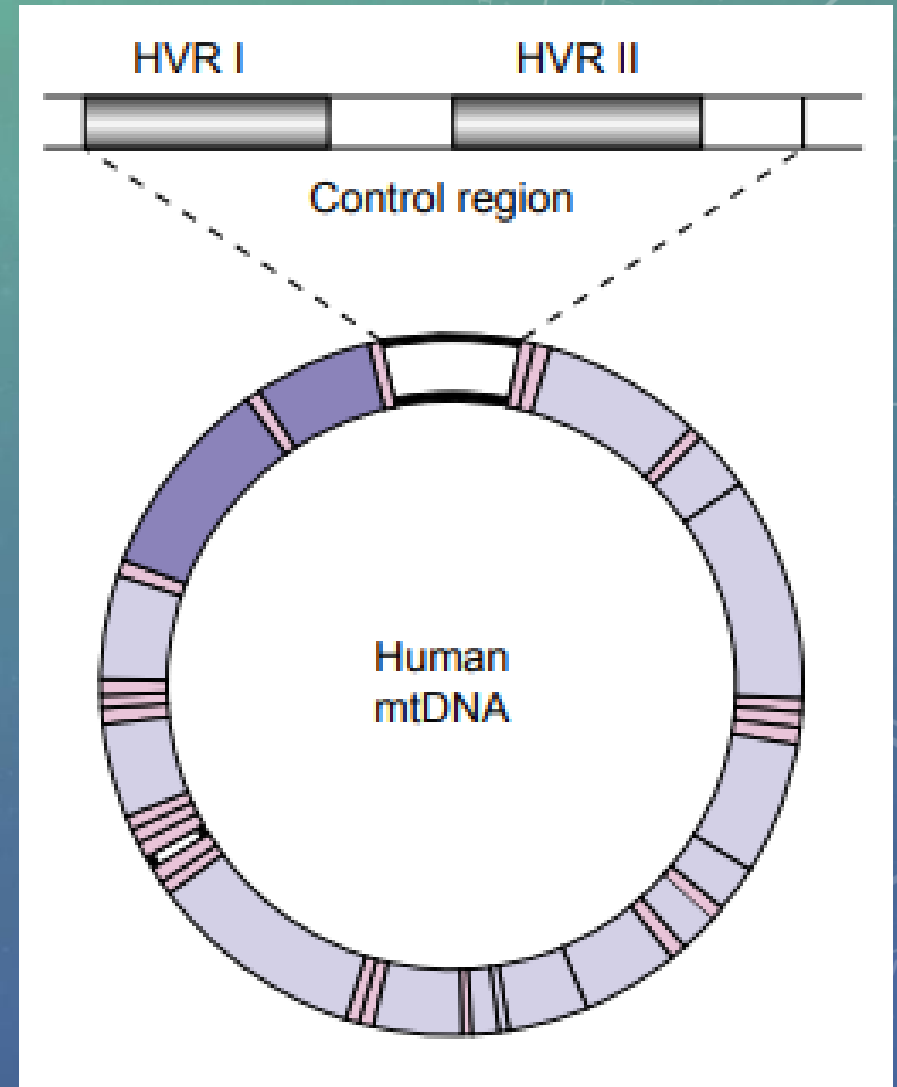
For nuclear DNA: Length is measured

For mtDNA: Sequence is examined



MITOCHONDRIAL DNA

- Mitochondrial DNA (mtDNA) is present in every cell
- Multiple copies of mtDNA is present in every cell
- It is circular in structure
- Human mtDNA is 16569bp long
- The mitochondrial DNA in every child is identical to his mother's
- mtDNA is indistinguishable in the same maternal line
- mtDNA has 2 major parts:
 - Coding region
 - Control region



MITOCHONDRIAL DNA

- The control has 2 regions where maximum variations are found in humans: Hypervariable region 1 (HV1) & Hypervariable region 2 (HV2)
- HV1, HV2 & coding regions are tested during mtDNA analysis
- **mtDNA mismatch** = not full siblings, but can have the same father
- **mtDNA match** = related through maternal line & could be siblings but have different father

AGCTAGATCGTTATTCCGAG

AGCTAGATCGTTATTCCGAG

Identical mtDNA

AGCTAGATTGTTATTCCGAG

AGCTAGATCGTTATTCCGAG

Unidentical mtDNA

