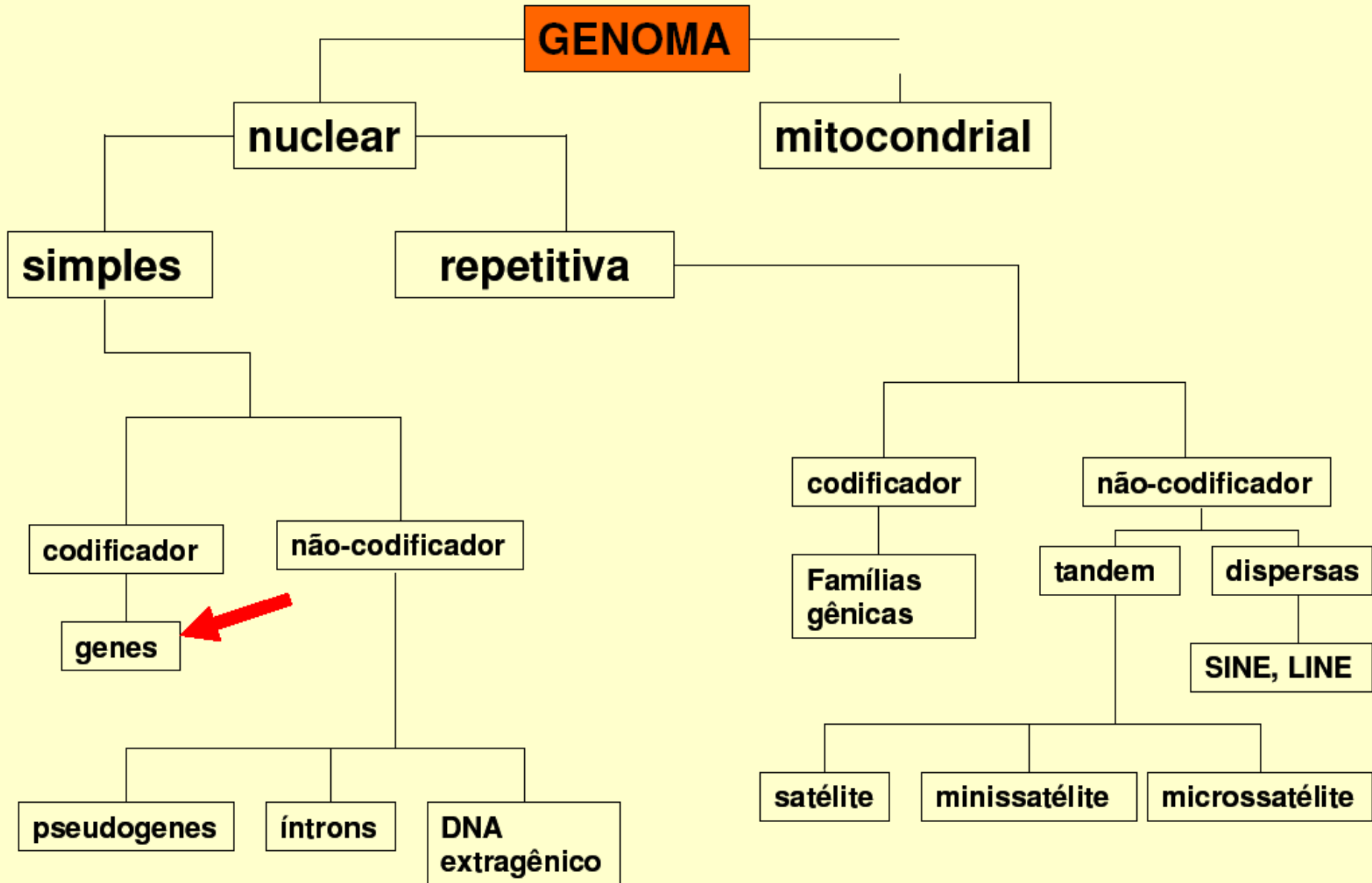
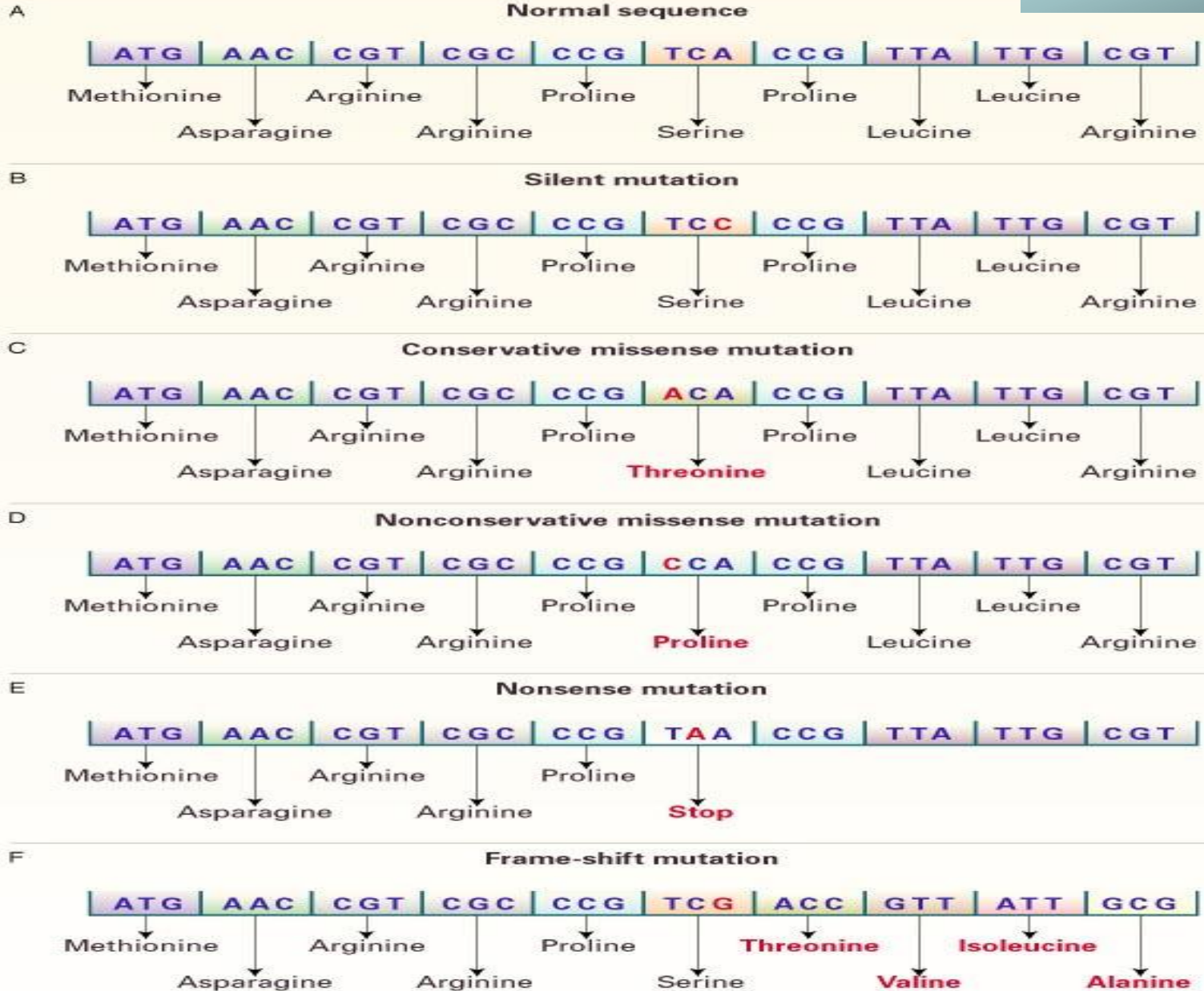


VARIABILIDADE GENÉTICA



SEQUENCIA CODIFICANTE



Nonsynonymous Coding SNP involved in Drug Metabolism

Wild-type
CYP2D6 gene

... C A C T C C G G A C G C ...

SNP variant in
CYP2D6 gene

... C A C T C C T G A C G C ...

Wild-type
CYP2D6 gene

167	168	169	170
His	Ser	Gly	Arg

SNP variant in
CYP2D6 gene

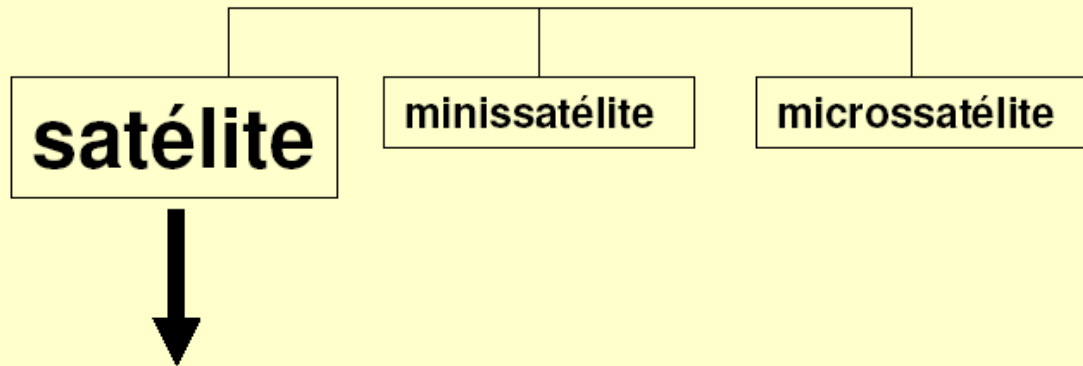
167	168	169	170
His	Ser	STOP	



Associated with:
Inhibitory to metabolise
debrisoquine (Declinax)

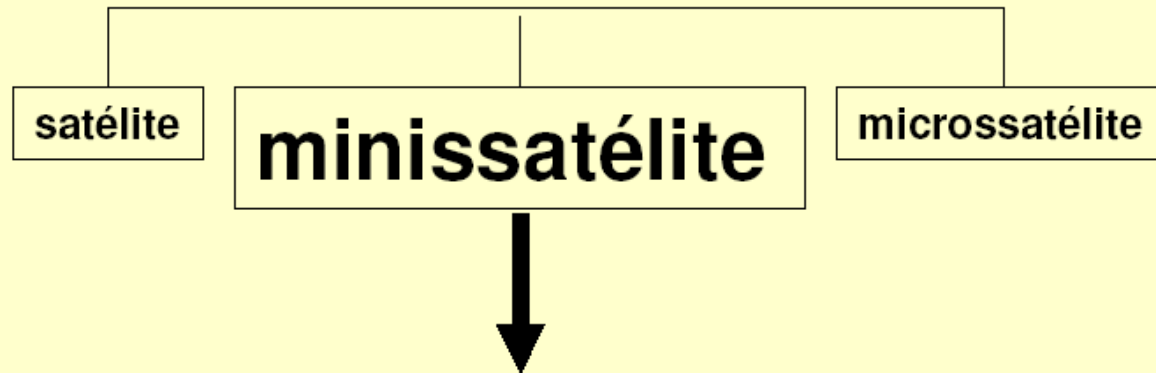
Droga neurobloqueadora adrenérgica

Organização do Genoma



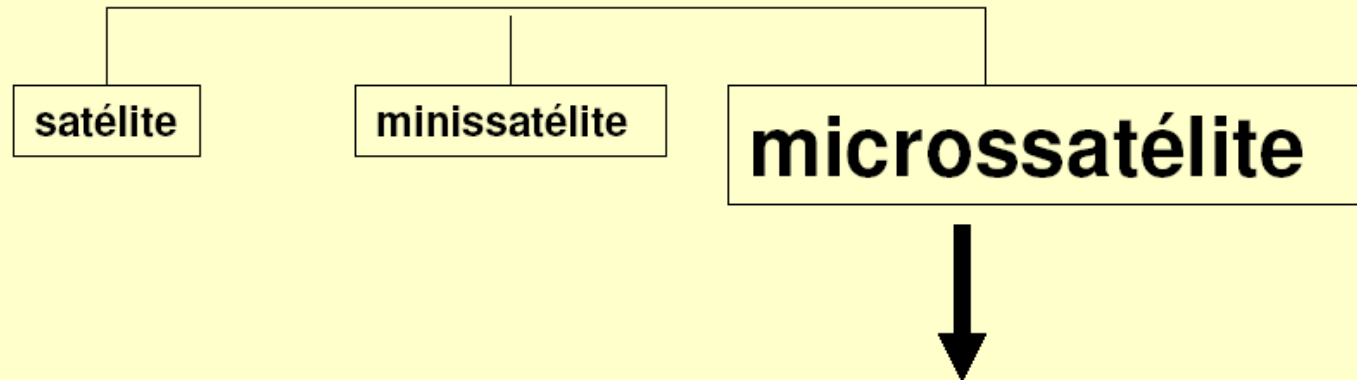
Seqüências longas de DNA altamente repetitivas (>10⁴ cópias), de 100 a 2000 pares de bases, que constituem uma fração importante do genoma dos eucariontes superiores, variando de 25% em algumas espécies até 50% em outras.

Organização do Genoma



Seqüências de DNA moderadamente repetitivas, de 10 a 100 pares de base.

Organização do Genoma



Seqüências de DNA moderadamente repetitivas de 2 a 9 pb. Altamente polimórficas. Arranjos com menos de 300 pb

MICROSSATÉLITE

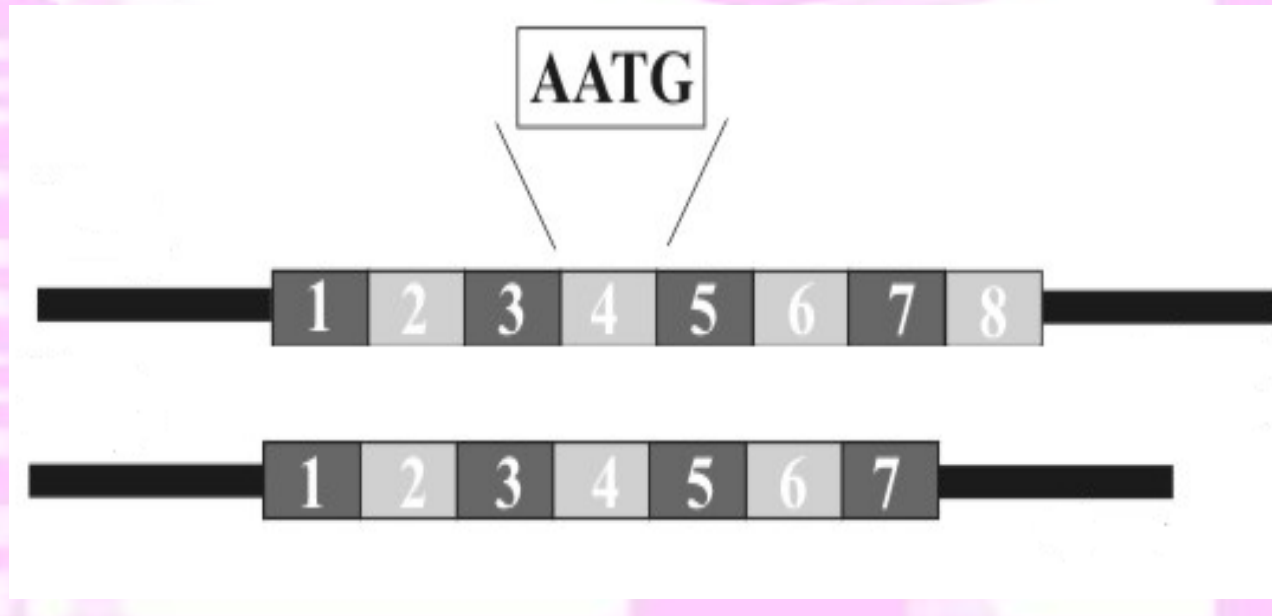
(AG)₉

AGAGAGAGAGAGAGAGAGAG

TC TC TC TC TC TC TC TC TC

STR

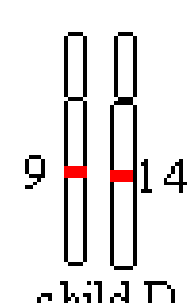
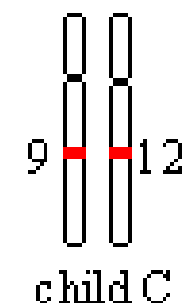
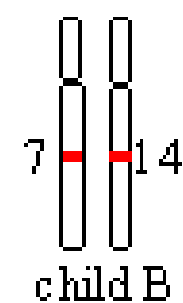
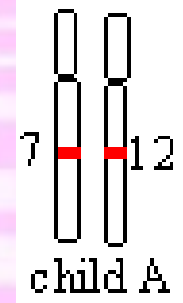
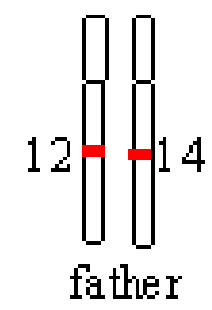
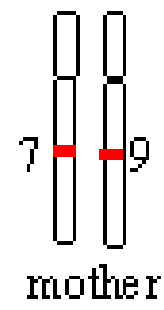
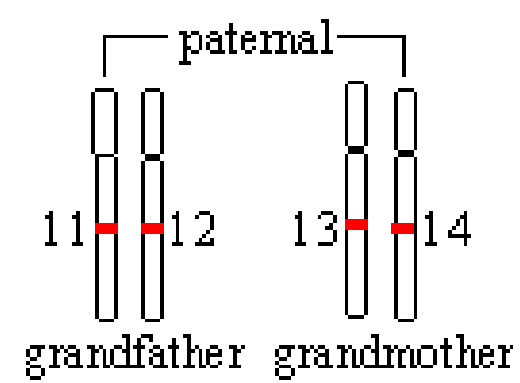
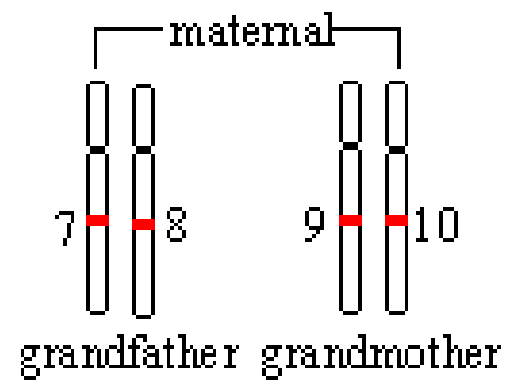
(Short Tandem Repeats)



Microssatélites

→ STRs

Herança?



Herança dos STRs ?

Exemplo:

STR **5D123** (4pb)

Alelos = 2 a 9

**Microssatélites =
STRs**

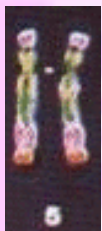


MÃE

1 2
-----**(AGAT)(AGAT)**-----

-----**(TCTA)(TCTA)**-----

1 2 3 4
-----**(AGAT)(AGAT)(AGAT)(AGAT)**-----
-----**(TCTA)(TCTA)(AGAT)(AGAT)**-----



PAI

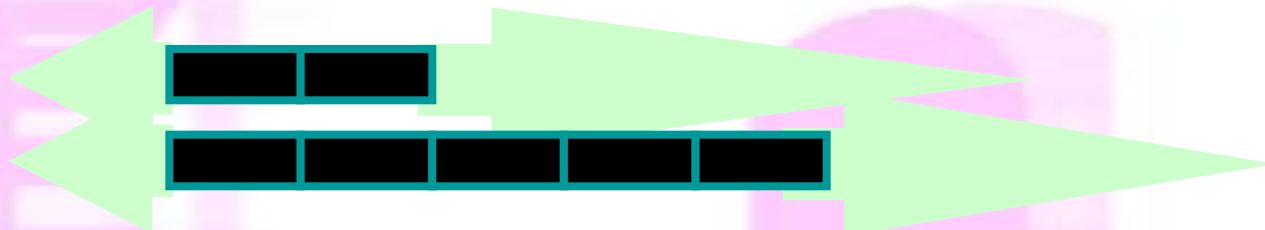
-----**(AGAT)(AGAT)(AGAT)**-----

-----**(TCTA)(TCTA)(TCTA)**-----

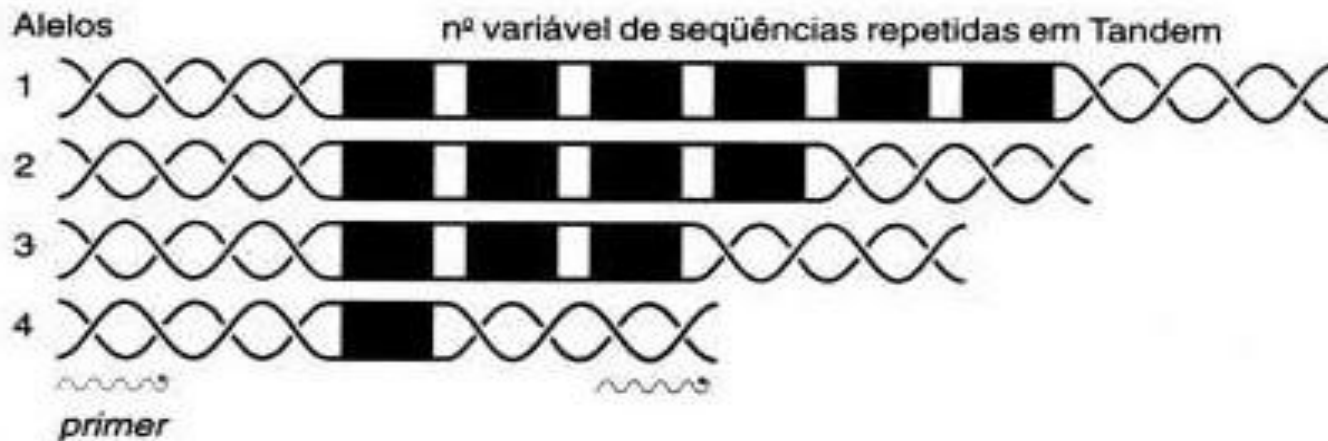
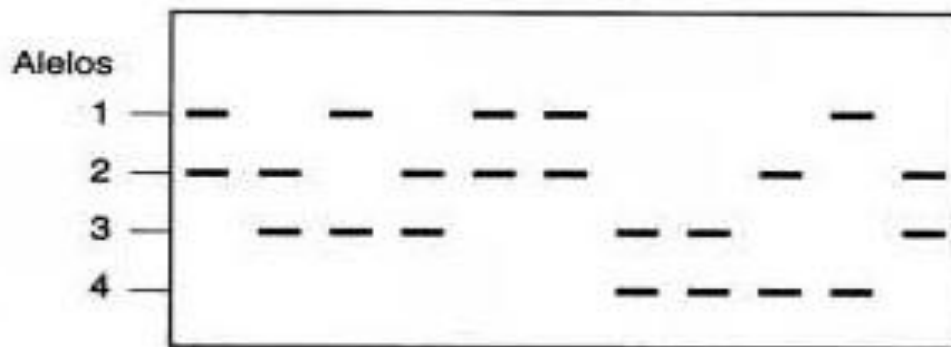
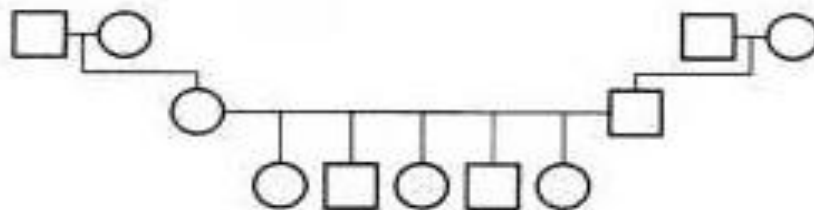
1 2 3
-----**(AGAT)(AGAT)(AGAT)(AGAT)(AGAT)**-----

-----**(TCTA)(TCTA)(TCTA)(TCTA)(TCTA)**-----
1 2 3 4 5

materna
paterna



VISUALIZAÇÃO DOS GENÓTIPOS (DNA) DOS INDIVÍDUOS DO HEREDOGRAMA EM GEL



Variabilidade do DNA

SNP

MICROSATELLITE



Male 1 GTACTAGACTACTACTACTACTACTGCGTG...
5 repeats

Male 2 GTACAAGACTACTACTACTACTACTACTGCGTG...
6 repeats

Male 3 GTACGAGACTACTACTACTACTACTACTACTGCGTG...
7 repeats