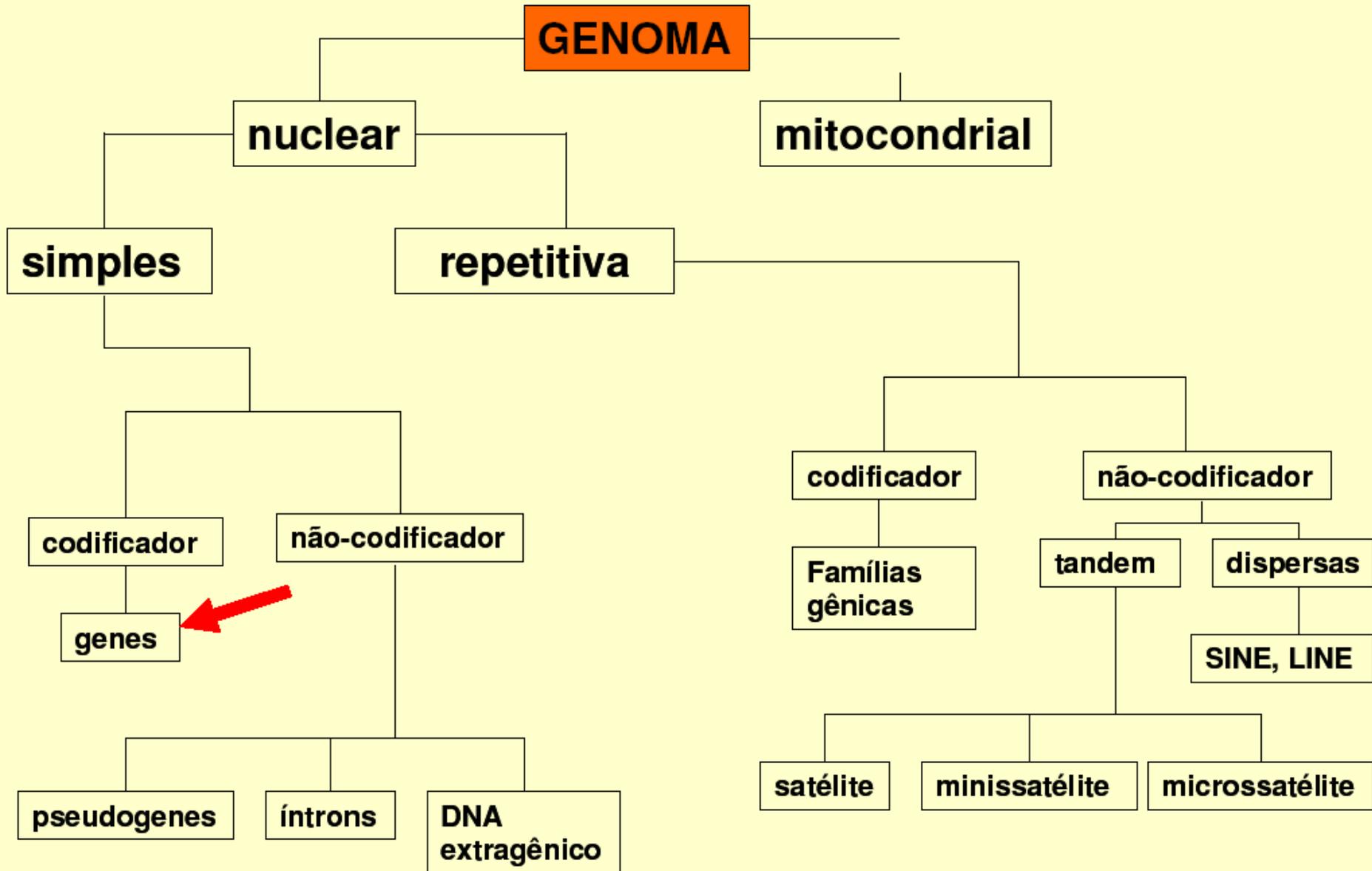
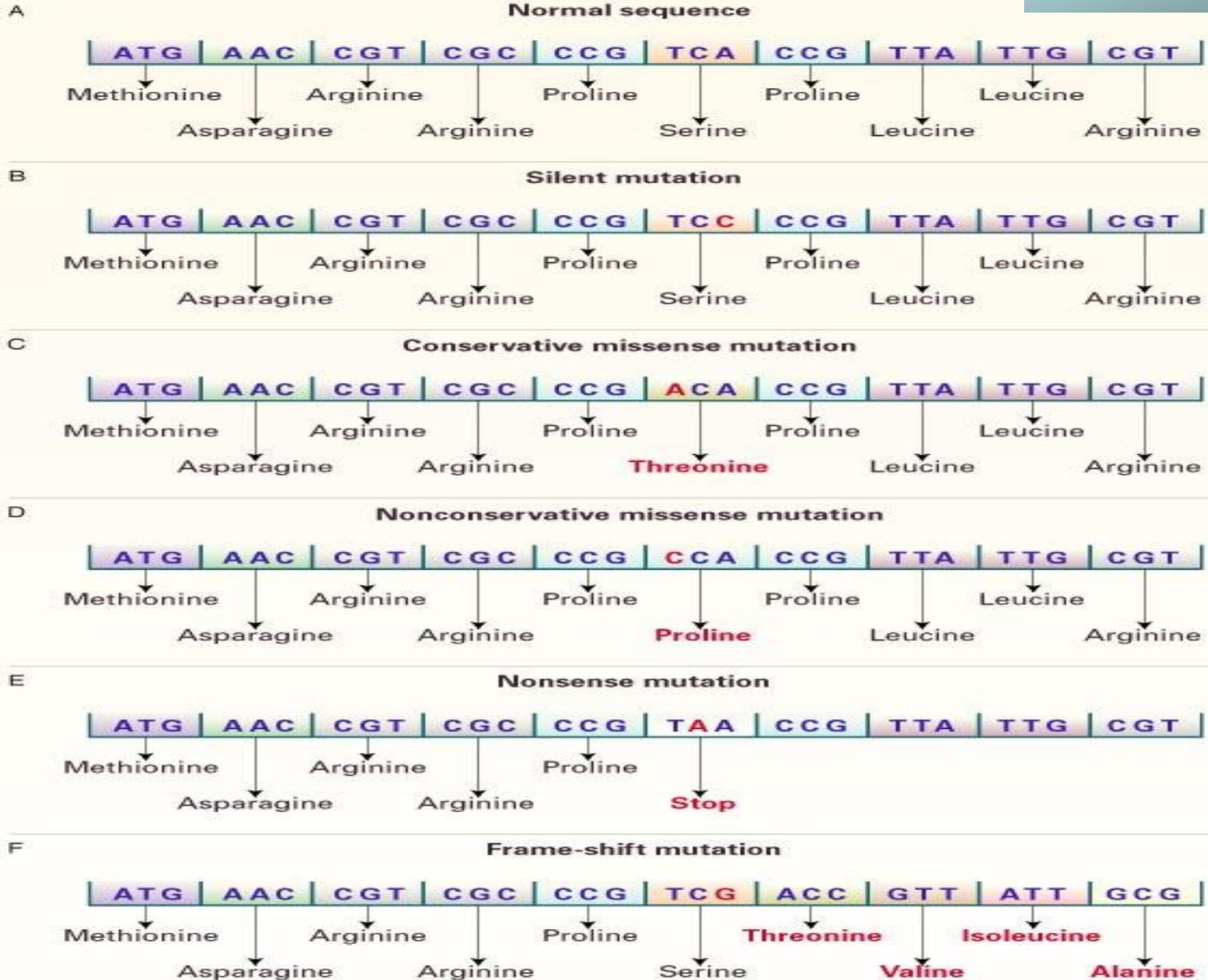


# VARIABILIDADE GENÉTICA



# 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

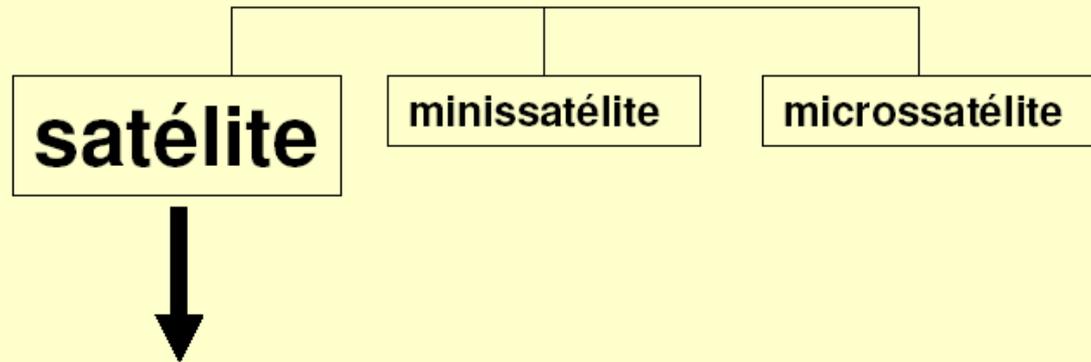
167	168	169	170
His	Ser	STOP	



Associated with:  
Inhibitory to metabolise  
debrisoquine (Declinax)

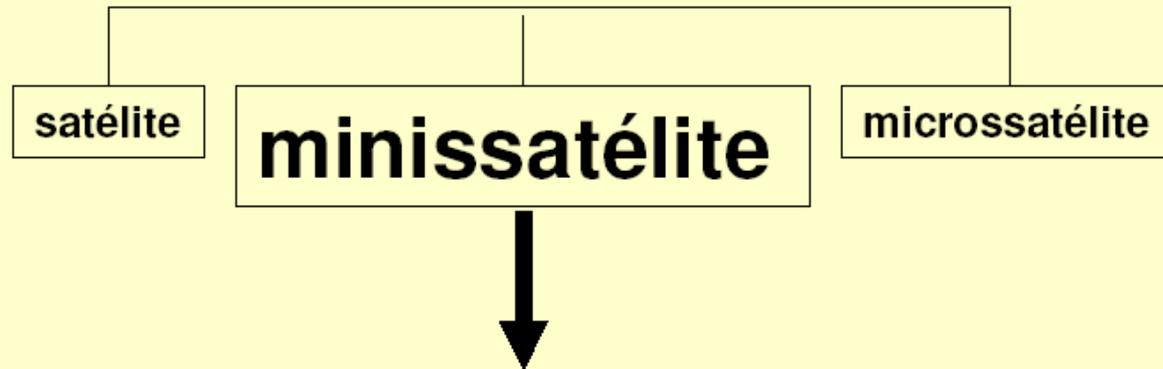
Droga neurobloqueadora adrenérgica

# Organização do Genoma



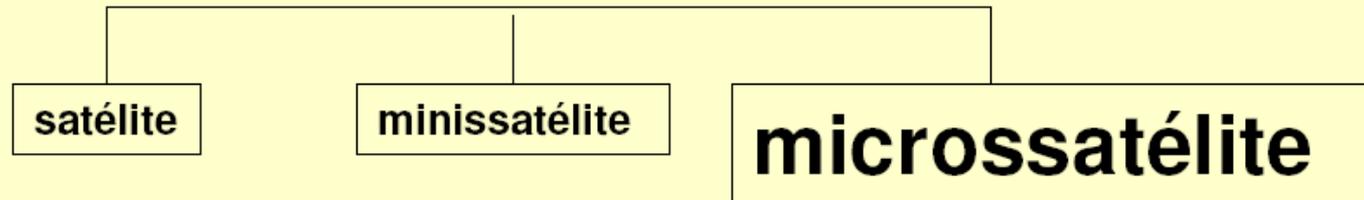
**Seqüências longas de DNA altamente repetitivas (>10<sup>4</sup> cópias), de 100 a 2000 pares de bases, que constituem uma fração importante do genoma dos eucariontes superiores, variando de 25% em algumas espécies até 50% em outras.**

# 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

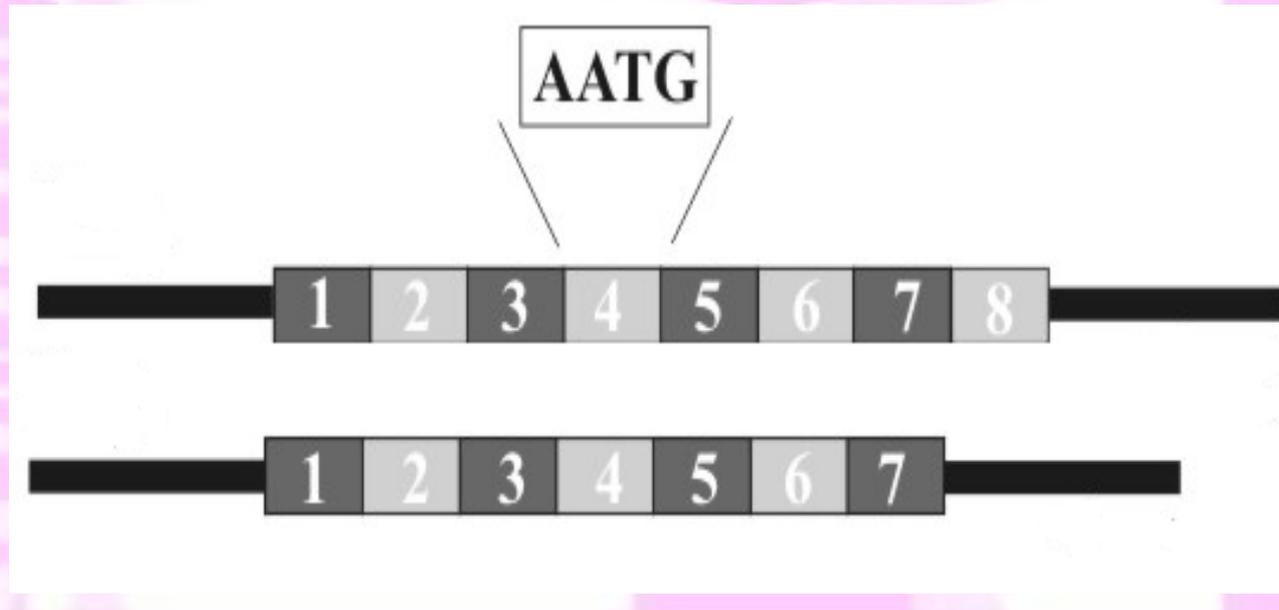
(AG)<sub>9</sub>

AGAGAGAGAGAGAGAGAG

TC TC TC TC TC TC TC TC TC

STR

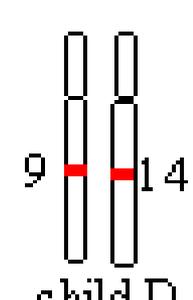
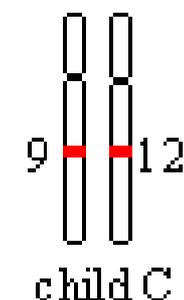
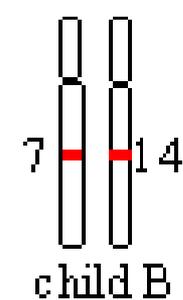
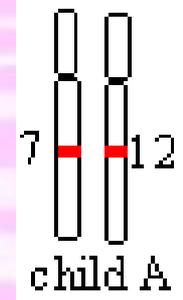
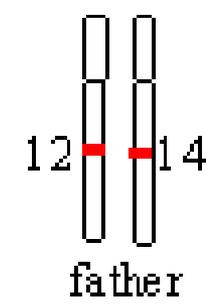
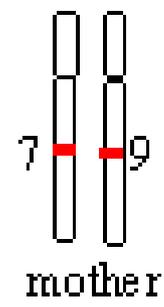
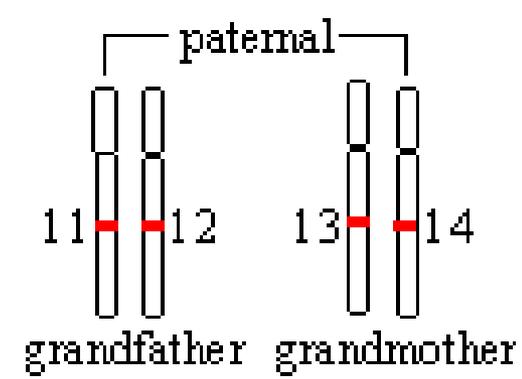
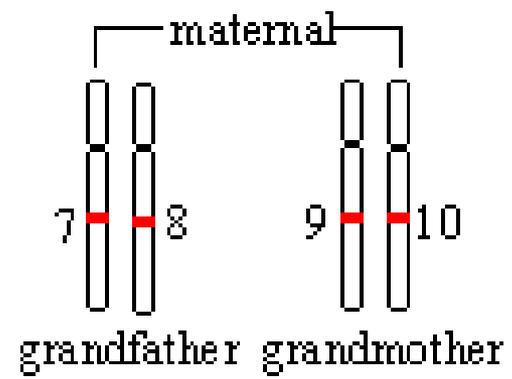
(Short Tandem  
Repeats)



# Microssatélites

→ STRs

## Herança?



# Herança dos STRs ?

Exemplo:

STR **5D123** (4pb)

Alelos = 2 a 9

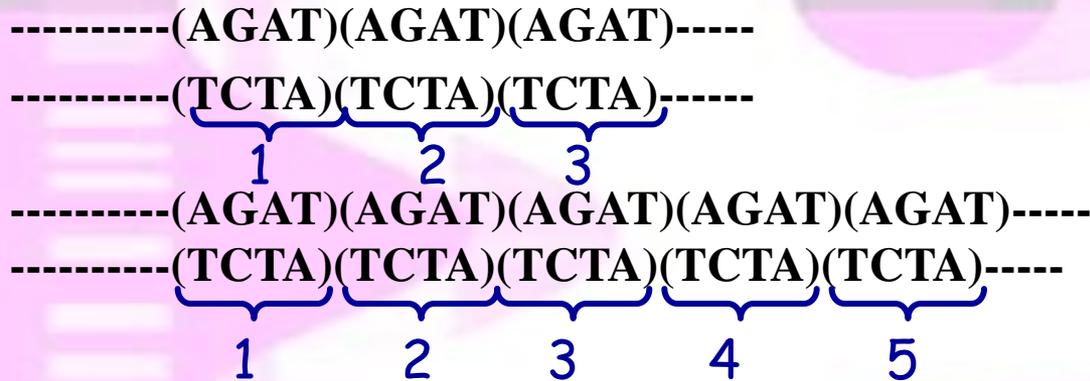
**Microssatélites =  
STRs**



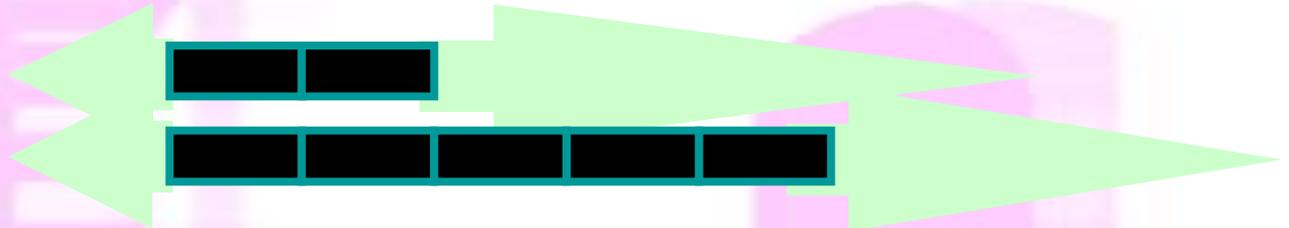
**MÃE**



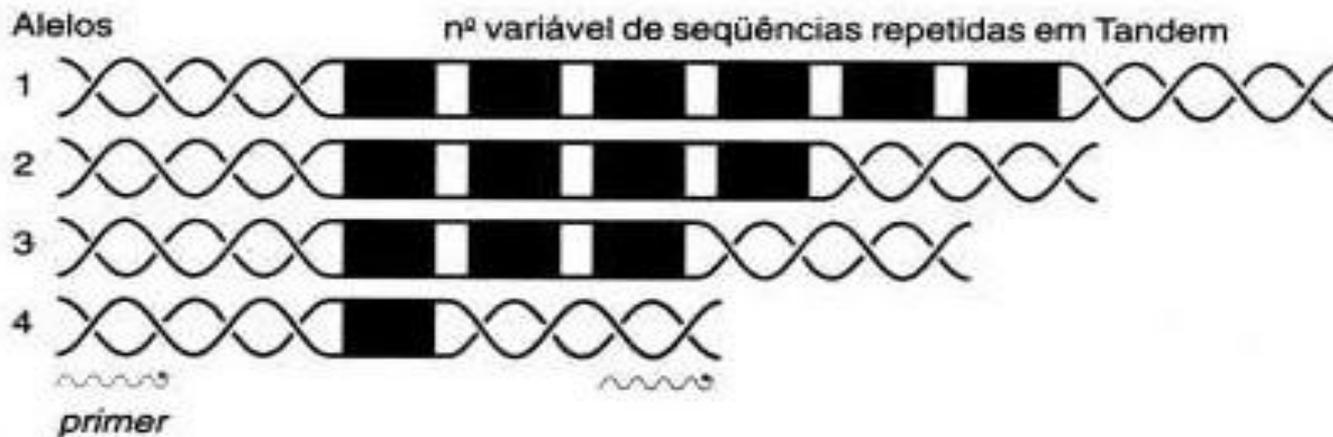
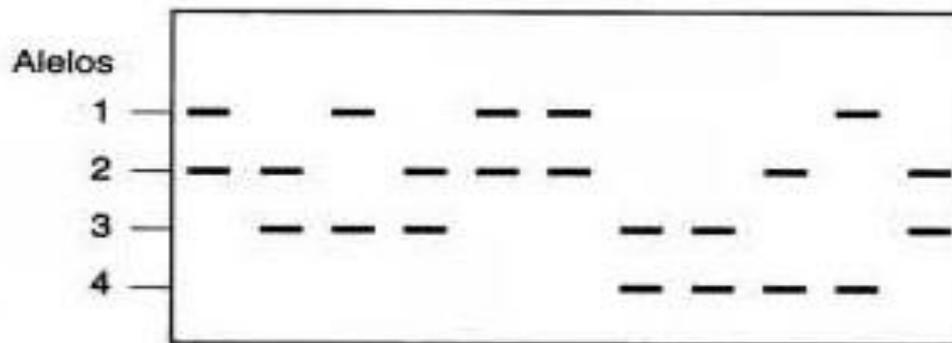
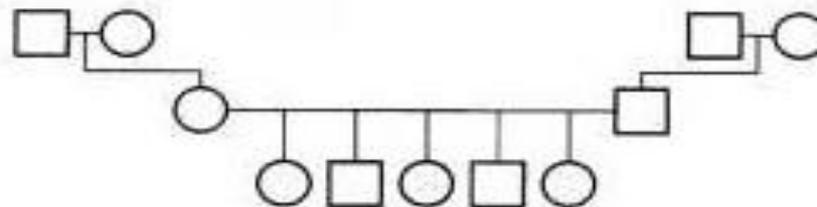
**PAI**



**materna**  
**paterna**



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



# Variabilidade do DNA

SNP

MICROSATELLITE



Male 1 GTACT**T**AGACTACTACTACTACTACTACTGGTG...  
5 repeats

Male 2 GTAC**A**AGACTACTACTACTACTACTACTACTGGTG...  
6 repeats

Male 3 GTAC**G**AGACTACTACTACTACTACTACTACTACTGGTG...  
7 repeats