

14.5

(26)

## MUTATION

- change to the genetic information of a cell or virus

### Large-scale mutations

- changes to an entire chromosome

### Small-scale mutation

⇓  
changes to one or several nucleotide pairs

example →

POINT Mutation is change to a single nucleotide pair

Genetic Disorder

or HEREDITARY Disease  
is when the change has an ADVERSE effect on the phenotype of the organism

## Small-scale mutations:

- 1) single nucleotide-pair substitution
- 2) nucleotide-pair insertions  
or deletions (one or more pairs)

## Substitution:

### ① SILENT MUTATION

- change in just one pair still produces the same amino acid.

### ② Missense Mutation

- change one AA into another

a) could have no effect on the protein

b) could be detrimental to function of protein - especially if in vital area affecting sec, tert, or quat structure

c) could improve the protein or lead to novel capabilities

evolution works on this

### (3) Non-sense Mutation

- changed a codon for an AAA into a "STOP" codon

⇒ translation stops prematurely

⇒ almost all non-sense mutations lead to a non-functional protein

Question ⇒ what might be an exception?

### INSERTIONS + DELETIONS

- DISASTROUS !!

⇒ alter remaining frame

⇒ called "Frameshift Mutations"

⇒ everything downstream of mutation gets improperly grouped into codons

MUTAGEN - chemical or physical agents that cause mutations

CARCINOGENS - cancer-causing chemicals  
⇒ most are also mutagenic.