

# Mutations

Tuesday, December 11, 2018

8:26 AM

## Gene mutations

↳ change in a single base pair  
- can be harmful, beneficial (rare) or have no effect.

## Types of mutations:

① Point/substitution mutation  
↳ one nucleotide is replaced with another

a. missense mutation → DNA codes for the wrong amino acid

ie. sickle cell anemia

normal: ATG GTG CAC CTG ACT CCT GAG  
mutated: ATG GTG CAC CTG ACT CCT **GTG**

This mutation causes sickle cell anemia. ↑

↳ codes for Val amino acid instead of Glu amino acid.

b. nonsense mutation

→ single change causes a protein to stop being made

② Frameshift mutation → one nucleotide is added or deleted

a. Insertion - Addition of one extra

nucleotide

ie. ATG GTG CAC CTG ACT  
Insertion: ATG GTA GCA CCT GAC T

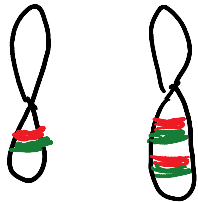
b. Deletion - the removal of a nucleotide

ie. ATG GTG CAC TGA  
Deletion: ATG TGC ACT GA

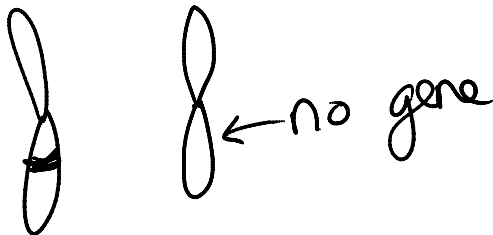
& can be much more harmful as they can change many amino acids instead of just one.

## Chromosomal mutations

① Duplication: extra copies of genes are generated



② Deletion: A loss of a gene



③ Inversion: broken chromosome segment gets put ~~at~~ back on upside down





④ Translocation: Fragment from one chromosome detaches and adds to another

⑤ nondisjunction: when chromosomes don't split completely in meiosis so you get 3 copies of ~~so~~ one or more chromosomes