


## Mutations



Sorry – NOT like this!!!! Note the FICTION part of Sci-Fi/Fantasy

### What is a Mutation?

- Change in DNA sequence
- May involve large segments of DNA (entire gene or chromosome) or a single nucleotide

### Mutations change DNA

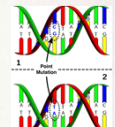
- $\Delta$  amino acid sequence
- $\Delta$  protein structure & function
- May  $\Delta$  characteristics

### Types of Mutations

- Gene Mutations**
  - Point Mutations
    - Missense
    - Nonsense
    - Silent
  - Frame Shift
    - Insertion
    - Deletion
- Chromosome Mutations**
  - Deletion
  - Duplication
  - Inversion
  - Translocation

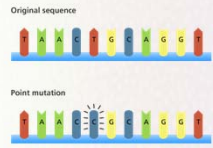
### Point Mutations

- The substitution, addition, or removal of a **single nucleotide** is called a **point mutation**



### Mutations: Substitution

- 1 nucleotide is replaced
- A new codon is formed



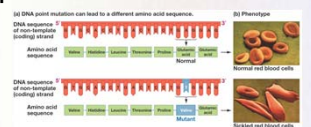
### Mutations: Substitution

- New codon may code for **different amino acid** & the protein may or may not be affected
- called a **missense mutation**

DNA	5' TCTCAA AATTACG 3'	5' TCTCAA AATTACG 3'
3'	AGAGTT TTTAAATGC 5'	3' AGAGTT TTTAAATGC 5'
mRNA	5' UCUCAAAAUUUACG 3'	5' UCUCAAAAUUUACG 3'
Protein	... Ser Gln Lys Phe Thr ...	... Ser Gln Gln Phe Thr ...

### Mutations: Substitution

- Example: **Sickle Cell Anemia**
- T instead of A in 1 codon
- Results in defective hemoglobin protein



### Mutations: Substitution

- If new codon changed to a **stop codon**, the protein is not made (shortened polypeptide)
- Known as a **nonsense mutation**

DNA	5' TCTCAA AATTACG 3'	5' TCTCAA AATTACG 3'
3'	AGAGTT TTTAAATGC 5'	3' AGAGTT TTTAAATGC 5'
mRNA	5' UCUCAAAAUUUACG 3'	5' UCUCAAAAUUUACG 3'
Protein	... Ser Gln Lys Phe Thr ...	... Ser Gln ...

### Mutations: Substitution

- It is possible for the mutation to be **silent** – new codon codes for **same amino acid** & the protein is unchanged

Met	Pro	Ser	Val
TAC	GGT	AGT	CAA
Met	Pro	Ser	Val
TAC	GGT	AGT	CAT

### Frame Shift Mutations

- Because a codon consists of 3 sequential nucleotides, adding or deleting 1 causes the rest to be **incorrectly grouped**

normal  
GGC GCT CCA AAG GTA

frame shift-mutation  
GTG CGC TCC AAA GGT A

### Mutations: Insertions & Deletions

- 1 or more nucleotides are **added to** or **taken from** a gene
- Tend to be more serious than substitutions, due to the frame shift – everything after is effected!

~~The fat cat sat~~  
hef atc ats at

### Chromosome Mutations

- Involves a change in a large portion of the DNA (1 or more genes)

### Chromosome Mutations

Deletion  
Duplication  
Inversion  
Translocation – involves more than 1 chromosome

### How often can this happen?

- Human cell copies its 6 billion bases & divide into daughter cells in only few hours
  - remarkably accurate
  - only ~1 error per 100 million bases
  - ~30 errors per cell cycle

### Possible Causes of Mutations

- Mutations in cells can be triggered by
  - UV radiation
  - chemical exposure
  - radiation exposure
  - Heat
  - Viruses
  - cigarette smoke
  - pollution
  - age
  - genetics

### Are all Mutations Bad?

- Some mutations can become a problem, causing cancer for example.
- Other mutations provide variations in a species \*important for evolution

### Questions? Comment?