

Dr. H. N. Sinha Arts And Commerce College, Patur

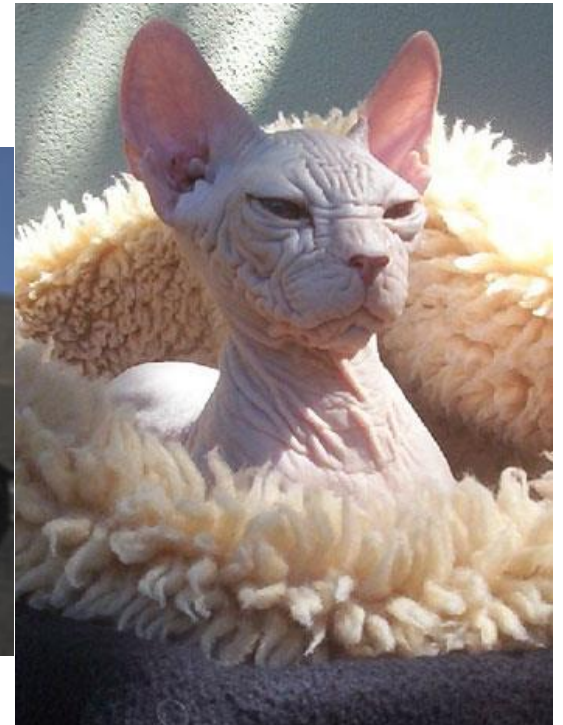
MUTATIONS

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- In biology, a **mutation** is an alteration of the **nucleotide sequence** of the genome of an organism
- May occur in **somatic cells** (aren't passed to offspring)
- May occur in **gametes** (eggs & sperm) and be passed to offspring

Mutations can:

- be bad, leading to cancer, aging, birth defects, self-aborted embryos



CACGTGGACTGAGGA**CTC**CTC

Codon for CTC =

glutamate



Normal
Blood Cell

CACGTGGACTGAGGA**CAC**CTC

Codon for CAC =

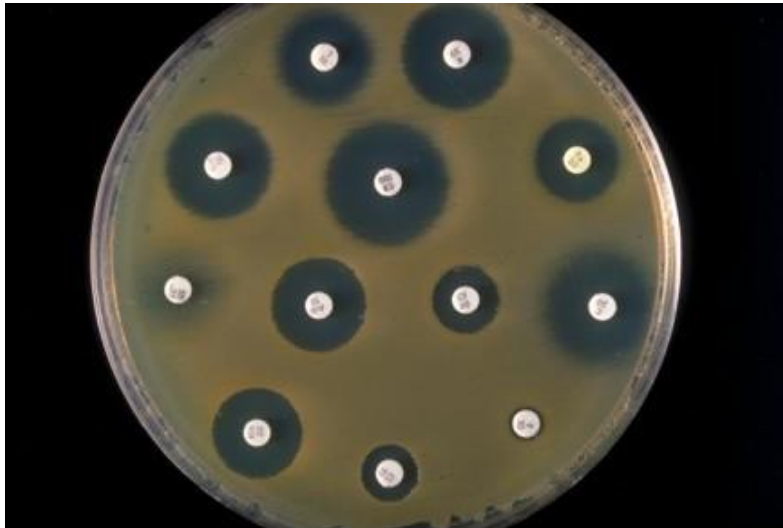
valine



Sickled Red
Blood Cell

What does it matter???

- be good, making an organism survive better in its environment
 - Example: bacteria becoming antibiotic-resistant



The ability to drink milk as an adult is a helpful mutation.

Not red hair

gacaggaactatggctgtgcagggatccccagagagacttctgggctccct
caactccacccccacagccaatccccagctgggctggctgccaaccag
caggaagccccgtgcttggaggtgtccatctctgacgggctcttccctcagc
ctggggctggctgagcttgggtggagagcgcgctgggtgggccaccaatgc
caagAACGGAAcctgcaactcaaccaatgtactgcttcaatctgctgctgg
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gcagctggacaaatgtcaattgacgtgactcaacctgacgctccaatgctgtcca
gctctgcttccctgggagccaatgcgcggtgggaccgctacatctccatcttc

taagcactgc **GCT** accacagcaaccgtgacccctgcccggggcggg

cgagccgttgcggccaatctgggtggccaagtgtcgtcttcagcagctctt
catcgccatctacgaccacgtggccgtccctgctgtgctctgctggctctt
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tcaatctacgcttccacagccaaggaagctccgcaggagcgtcaaggaaggtg
ctgacgtgctccctgggtg

Red hair

gacaggaactatggctgtgcagggatccccagagagacttctgggctccct
caactccacccccacagccaatccccagctgggctggctgccaaccag
caggaagccccgtgcttggaggtgtccatctctgacgggctcttccctcagc
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gctctgcttccctgggagccaatgcgcggtgggaccgctacatctccatcttc

taagcactgc **CCT** accacagcaaccgtgacccctgcccggggcggg

cgagccgttgcggccaatctgggtggccaagtgtcgtcttcagcagctctt
catcgccatctacgaccacgtggccgtccctgctgtgctctgctggctctt
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ctgacgtgctccctgggtg

My red hair gives me
super powers.



Types of Mutations

Gene Mutations

- Change in the **nucleotide sequence** of a **gene**
- May only involve a **single nucleotide**

Types of Gene Mutations

–Point Mutations

- a. Missense
- b. Nonsense
- c. Silent

–Frameshift

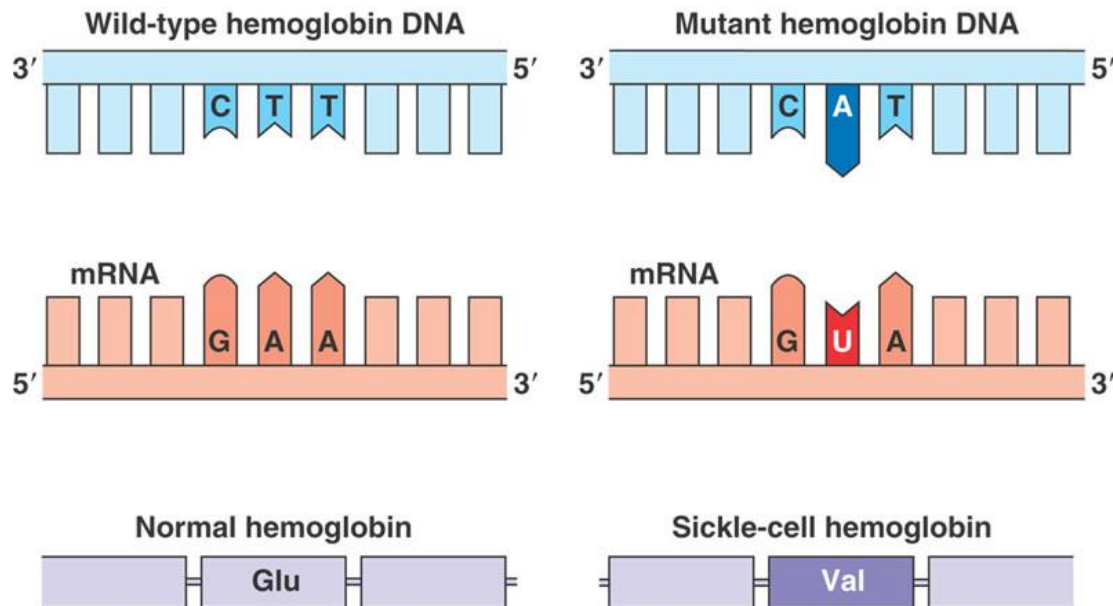
- a. Insertions
- b. Deletions

Point Mutations

- A **point mutation** or **substitution** is a genetic mutation where a **single nucleotide base is changed.**

a. Missense Mutation

This type of mutation is a **change in one DNA base pair** that results in the substitution of **one amino acid for another** in the protein made by a gene. Example is **Sickle cell anemia**



b. Nonsense Mutation

- A nonsense mutation is **also a change in one DNA base pair**. Instead of substituting one amino acid for another, however, the altered DNA sequence prematurely signals the cell to stop building a protein. This type of mutation results in a shortened protein that may function improperly or not at all. Examples include **Duchenne Muscular Dystrophy** and **Thalassemia**

Normal condition

DNA

5'ATG	ACT	CAC	CGA	GCG	CGA	AGC	TGA3'
3'TAC	TGA	GTG	GCT	CGC	GCT	TCG	ACT5'

mRNA

AUG	ACU	CAC	CGA	GCG	CGA	AGC	UGA
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Protein

<u>Met</u>	<u>Thr</u>	<u>His</u>	<u>Arg</u>	<u>Ala</u>	<u>Arg</u>	<u>Ser</u>	Stop
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Nonsense mutation

DNA

5'ATG	ACT	CAC	TGA	GCG	CGA	AGC	TGA3'
3'TAC	TGA	GTG	ACT	CGC	GCT	TCG	ACT5'

mRNA

AUG	ACU	CAC	UGA	GCG	CGA	AGC	UGA
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Protein

<u>Met</u>	<u>Thr</u>	<u>His</u>	Stop
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c. Silent Mutations

- **Silent mutations** are mutations in DNA that do not have an observable effect on the organism's phenotype.
- For example, if the codon **AAA** is altered to become **AAG**, the same amino acid – lysine – will be incorporated into the peptide chain

Frame Shift Mutations

1. Insertion Mutations

- An insertion changes the number of DNA bases in a gene by adding a piece of DNA. As a result, the protein made by the gene may not function properly.
- Example
- | | | | | |
|-----|-----|-----|-----|-----------------|
| UUU | UUA | UCU | | Phe-Leu-Ser |
| UUU | UUA | UUU | UCU | Phe-Leu-Phe-Ser |

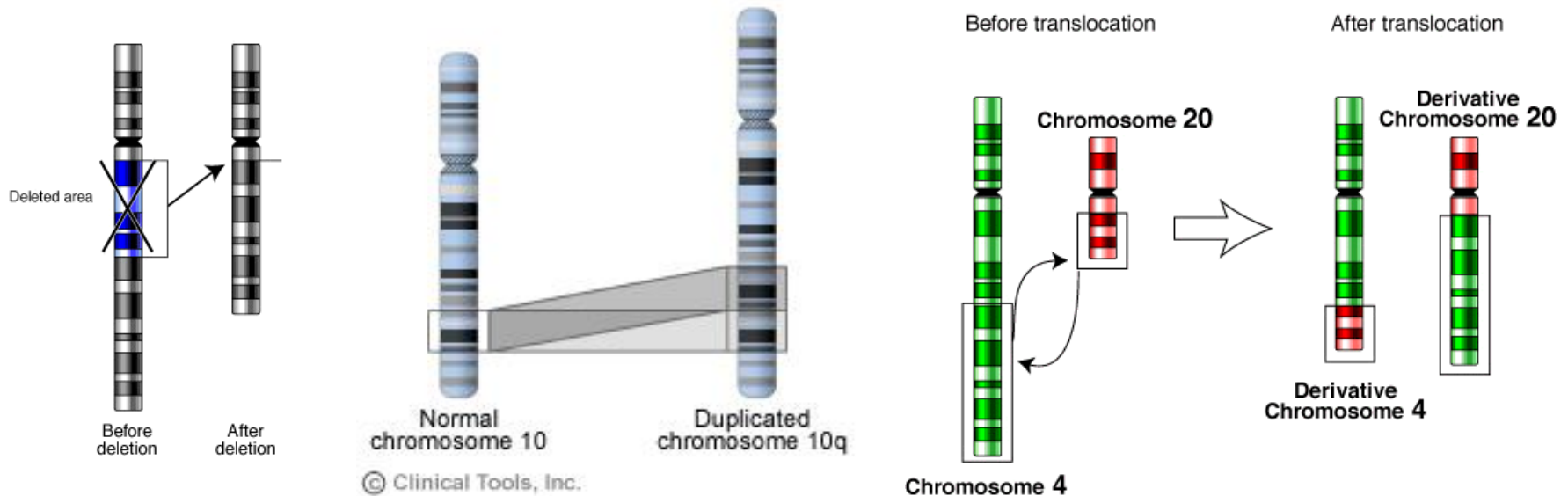
2. Deletion

- A deletion changes the number of DNA bases by removing a piece of DNA. Small deletions may remove one or a few base pairs within a gene, while larger deletions can remove an entire gene or several neighboring genes. The deleted DNA may alter the function of the resulting protein(s).
- UUU UUA UUU UCU Phe-Leu-**Phe**-Ser
- UUU UUA UCU Phe-Leu-Ser

Types of Mutations

2. Chromosomal mutation – may affect more than one gene

Examples: nondisjunction, translocation



Thank you