DNA Mutations

Altering the genetic code

Causes of Mutations

- Environmental factors (sunlight, radiation, smoking) and errors during replication
  - Cause mutations in DNA, abnormal growth of cells (cancer), and cell death
- Mutagen – substance capable of causing a mutation

- Are all mutations harmful?
  - No, some are helpful and some are neutral (no effect)

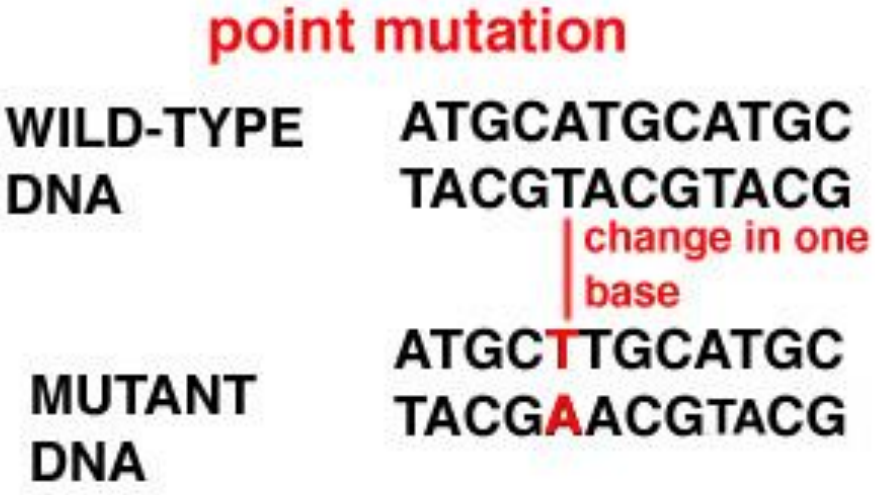
Causes of Mutations

- If mutations occur in somatic (body cells), it will not be passed on to offspring but can cause an increased risk of cancer.

- If mutations occur in reproductive (sex) cells, it may not harm the individual but could be passed on to the offspring

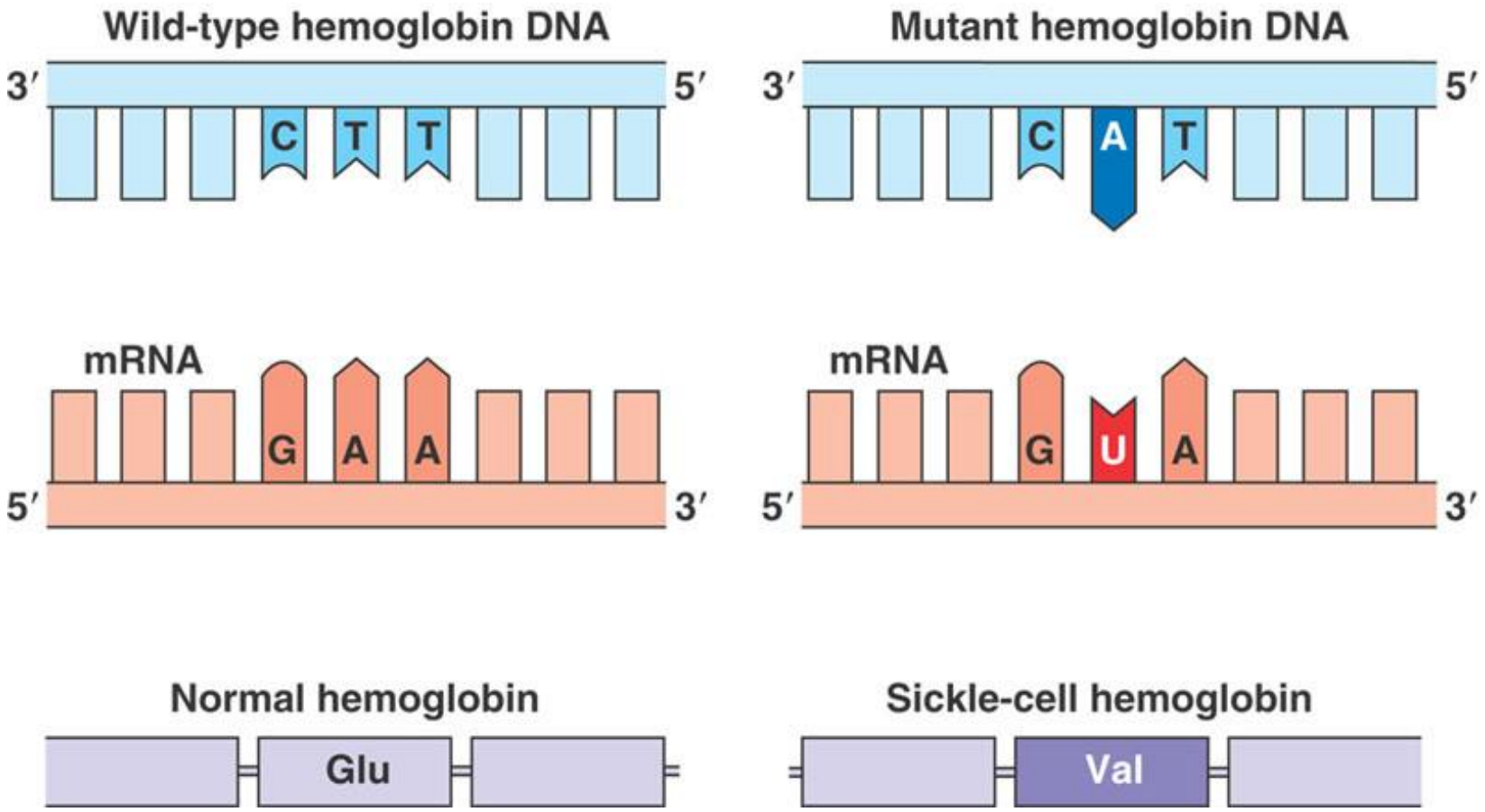
Point Mutation

- Change in a single DNA nucleotide



Examples of Point Mutations

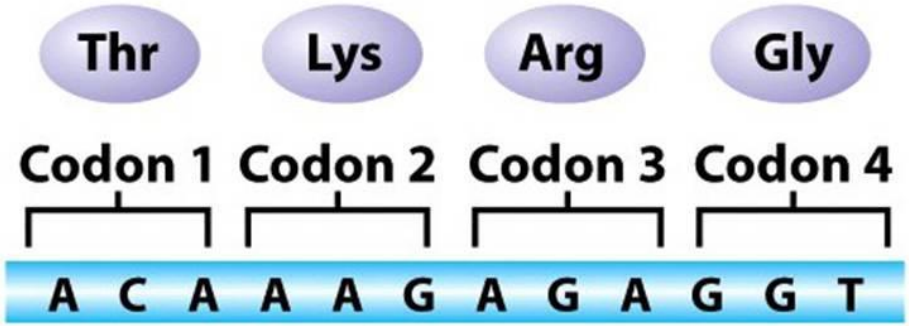
- Substitution
  - Change one nucleotide for another
  - The mutation only impacts one amino acid in the sequence
  - Ex: Sickle cell anemia: changes a Glutamine to a Valine


Wild-type hemoglobin DNA
3′
5′
C
T
T
Mutant hemoglobin DNA
3′
5′
C
A
T
mRNA
G
A
A
5′
3′
mRNA
G
U
A
5′
3′
Normal hemoglobin
Glu
Sickle-cell hemoglobin
Val

Examples of Point Mutations

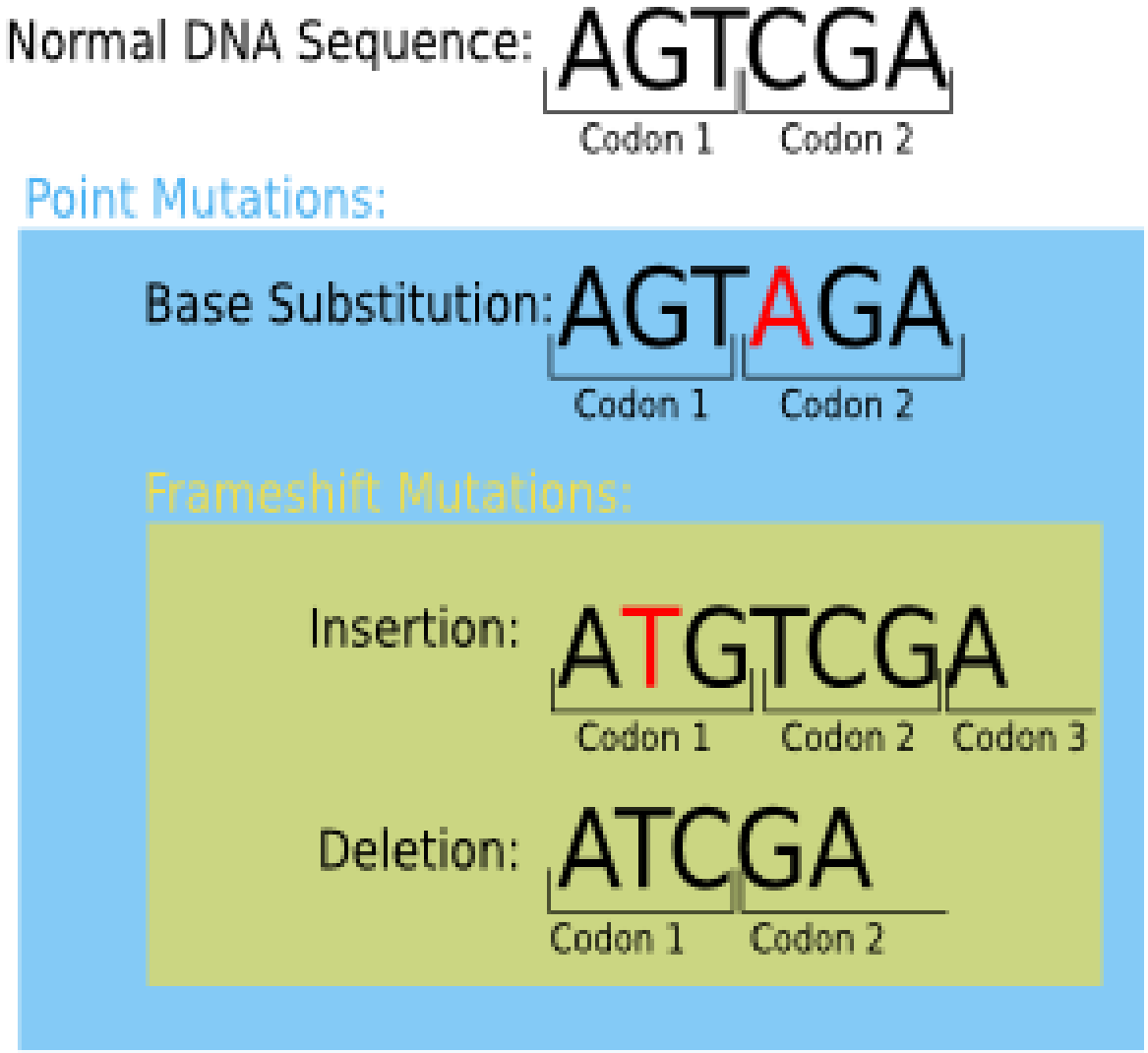
- Frameshift mutation: Addition or Deletion
  - Addition or removal of one nucleotide that changes the reading frame for the amino acids
  - This mutation changes the mRNA codon triplets which changes every amino acid after the mutation

Wild-type gene



Gene with insertion

Gene with deletion


Normal DNA Sequence: AGTCGA
Codon 1
Codon 2
Point Mutations:
Base Substitution: AGTAGA
Codon 1
Codon 2
Frameshift Mutations:
Insertion: ATGTCGA
Codon 1
Codon 2
Codon 3
Deletion: ATCGA
Codon 1
Codon 2

Results of Point Mutations

- Silent
  - Substitution does not change the resulting amino acid so there is no effect on the organism
- Missense
  - Substitution or frameshift causes some impact to the organism and changes one or many amino acids in the sequence
- Nonsense
  - Substitution or frameshift causes a STOP codon which causes the protein to not be completed

	No mutation	Point mutations			
		Silent	Nonsense	Missense	
				conservative	non-conservative
DNA level	TTC	TTT	ATC	TCC	TGC
mRNA level	AAG	AAA	UAG	AGG	ACG
protein level	**Lys**	**Lys**	**STOP**	**Arg**	**Thr**