

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

point mutation

WILD-TYPE
DNA

ATGCATGCATGC
TACGTACGTACG

| change in one
| base

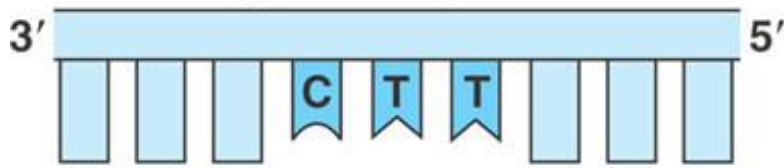
MUTANT
DNA

ATGC**T**TGCATGC
TACG**A**ACGTACG

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

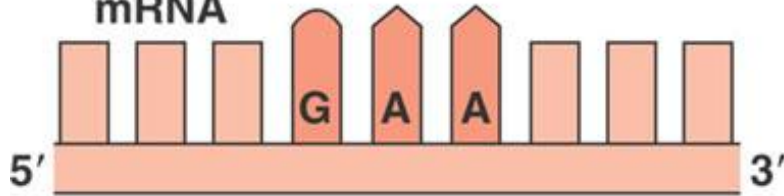
Wild-type hemoglobin DNA



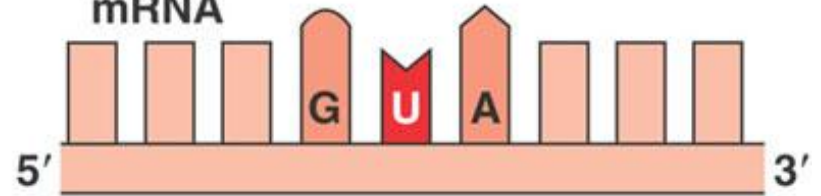
Mutant hemoglobin DNA



mRNA



mRNA



Normal hemoglobin



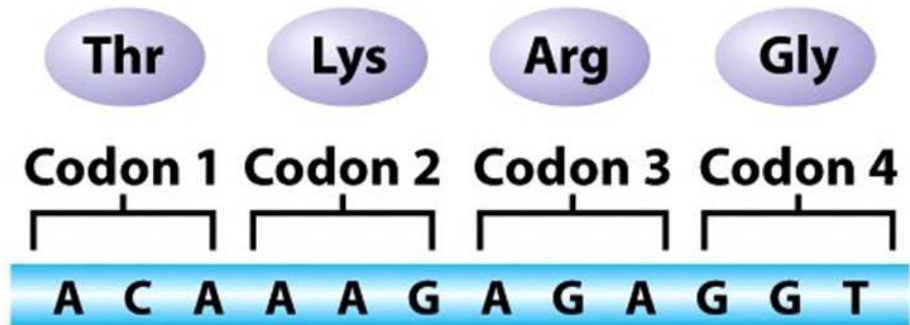
Sickle-cell hemoglobin



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

Point mutations

No mutation

Silent

Nonsense

Missense

conservative

non-conservative

DNA level

TTC

TTT

ATC

TCC

TGC

mRNA level

AAG

AAA

UAG

AGG

ACG

protein level

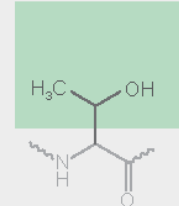
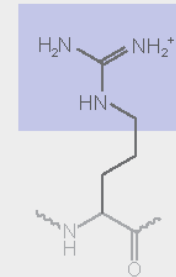
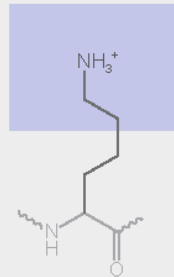
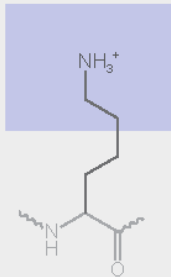
Lys

Lys

STOP

Arg

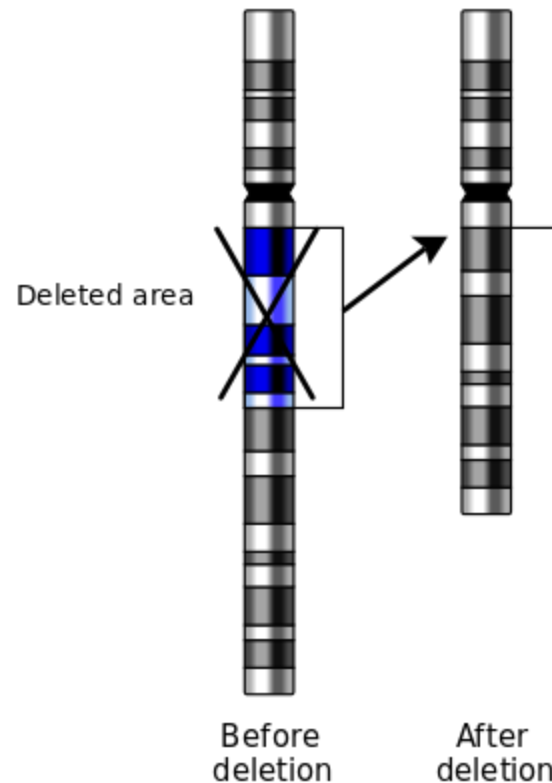
Thr



basic 
polar 

Chromosome Mutations

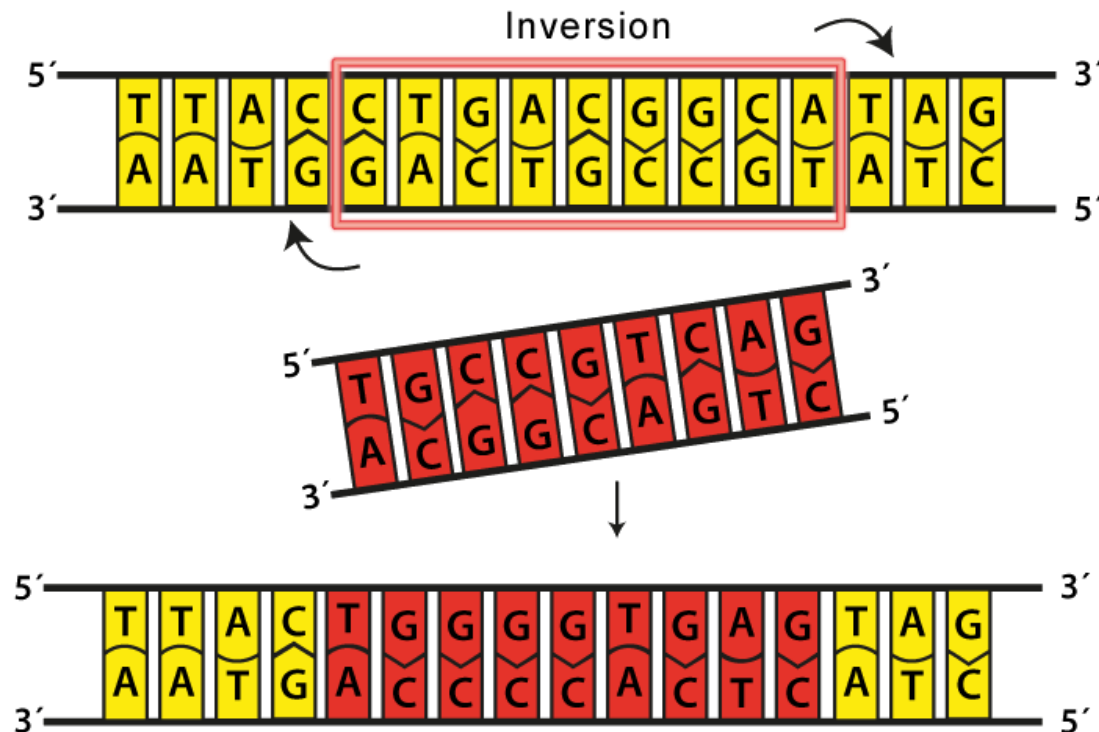
- Deletion
 - Removal of large sections of the chromosome



Chromosome Mutations

- Inversion

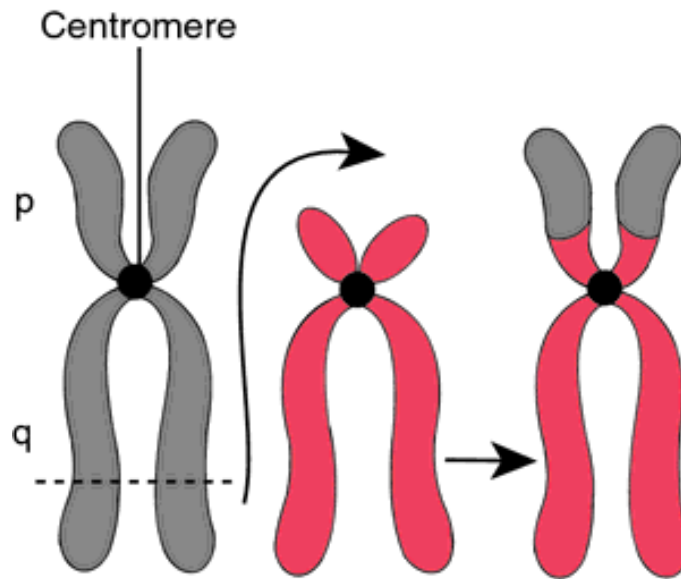
- When a piece of a chromosome breaks off and reattaches in the reverse order



Chromosome Mutations

- Translocation

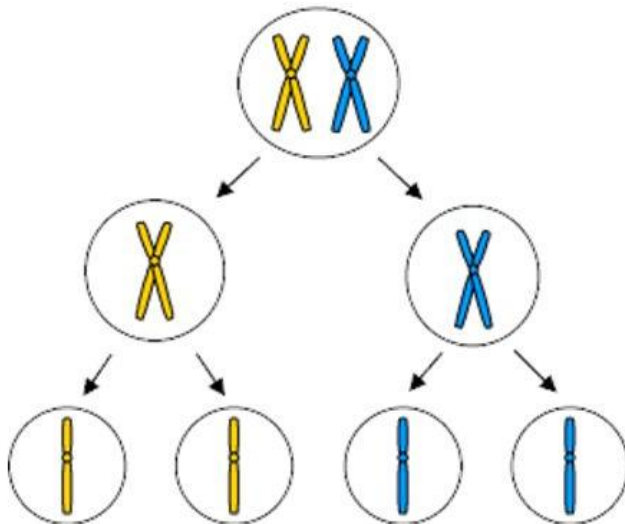
- When a piece of a chromosome breaks off and attaches to another non-homologous chromosome (a different piece of DNA)



Chromosome Mutations

- Non-disjunction
 - Failure of a chromosome to separate in meiosis (making of the sex cells)

What should happen



Nondisjunction

