



Life in the Northwoods

Mutations





Mutations



- ▶ Mutations - heritable changes in the DNA sequence
 - ▶ Happen when a change occurs in nucleotide bases
 - ▶ Are a source of genetic diversity
 - ▶ Are random events



Mutations: good or bad?

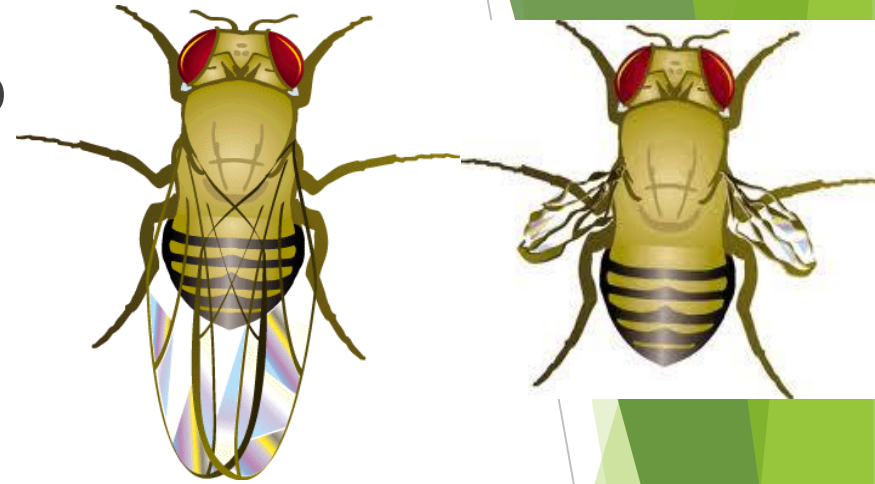
- Positive Mutations- produce proteins with new or altered functions that can be useful to organisms in different or changing environments

ex. adaptations

- Negative Mutations- when proteins are dramatically changed in structure or function; disrupts normal biological activities

ex. Cancer, sickle cell anemia

- Neutral Mutations- little or no effect on function of proteins



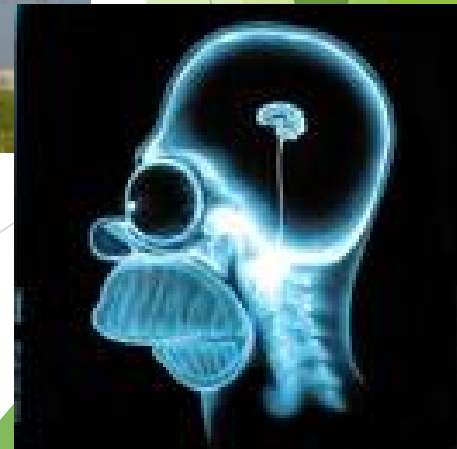
NORMAL

MUTATION



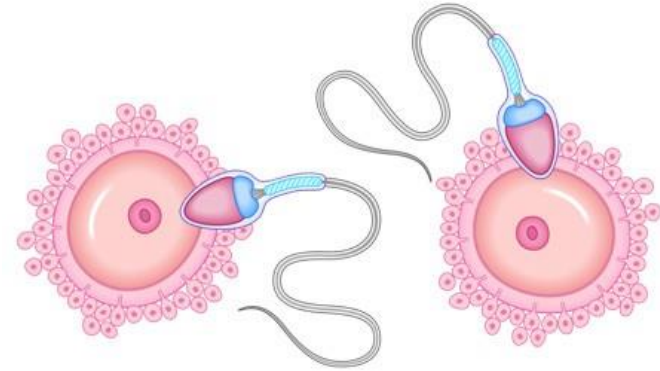
Causes of mutations

- ▶ Mistake made during DNA replication or transcription during protein synthesis.
- ▶ Mutagens: chemical or physical agents in the environment EX: X rays, UV light, nuclear radiation, asbestos, cigarette smoke, viruses



Can you give a mutation (mistake) to your kids?

► YES, if a mutation occurs in a sperm or egg cell



► NO, if a mutation occurs in a body cell (example skin cell)



Types of Mutations: Point Mutations

Point Mutations - mutations that occur at a single point in the DNA sequence and changes only one or a few nucleotides. This affects a single gene.

1. Substitutions - one base is changed to a different base

Ex. TAC GC T AGA → TAC GT AGA

2. Frame shift mutations - change the reading frame for the rest of the DNA sequence

1. Insertion - one base is added

Ex. TAC GCT AGA → TA CGC TAG A

2. Deletion - one base is removed

Ex. TA C GCT AGA → TCG CTA GA

Substitution vs. Frameshift

▶ THE FOX WAS RED (original/ correct protein)

▶ TTE FOX WAS RED
(Substitution)

▶ TAH EFO XWA SRE D Longer sentence!
(Insertion)

▶ TEF OXW ASR ED Shorter sentence!
(Deletion)

Substitution Affects:

- Missense - changes the amino acid and protein expressed, usually harmful

CAC = histidine → CC = proline

- Nonsense - Codes for a stop, stops the production of the protein, usually harmful

UAU = tyrosine → UAG = STOP

- Silent - does not change amino acid, not harmful (alters DNA sequence but has no effect on phenotype or function)

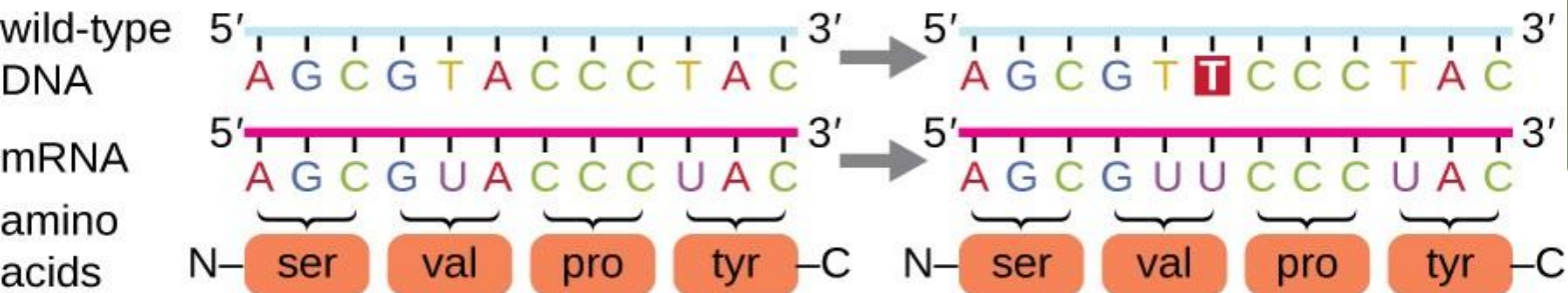
CAC = histidine →

CAU = histidine

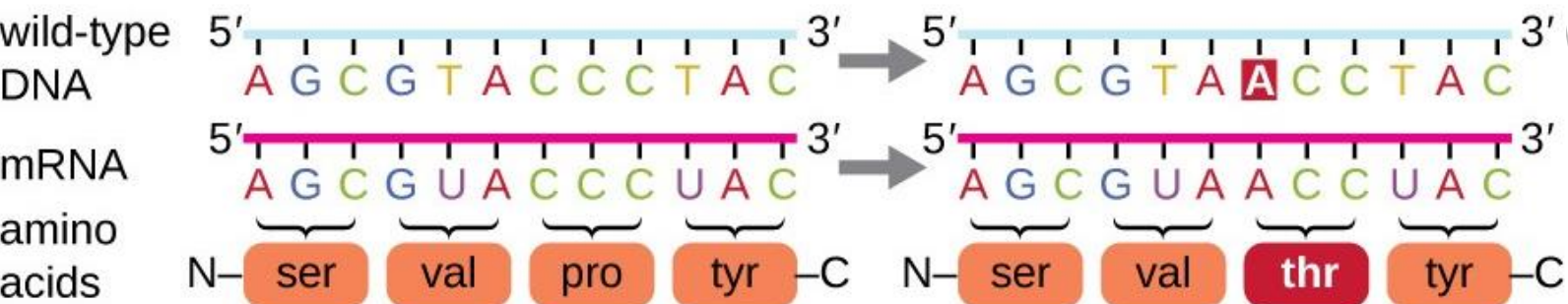
		Second Position									
		U		C		A		G			
		code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid		
First Position	U	UUU	phe	UCU	ser	UAU	tyr	UGU	cys	U	
		UUC		UCC		UAC		UGC		C	
		UUA	leu	UCA		UAA	STOP	UGA	STOP	A	
		UUG		UCG		UAG	STOP	UGG	trp	G	
	C	CUU	leu	CCU	pro	CAU	his	CGU	arg	U	
		CUC		CCC		CAC		CGC		C	
		CUA		CCA		CAA	CGA	A			
		CUG		CCG		CAG	CGG	G			
	A	AUU	ile	ACU	thr	AAU	asn	AGU	ser	U	
		AUC		ACC		AAC		AGC		C	
		AUA		ACA		AAA	lys	AGA	arg	A	
		AUG		ACG		AAG		AGG		G	
	G	GUU	val	GCU	ala	GAU	asp	GGU	gly	U	
		GUC		GCC		GAC		GGC		C	
		GUA		GCA		GAA	glu	GGA		A	
		GUG		GCG		GAG		GGG		G	

point mutation: substitution of a single base

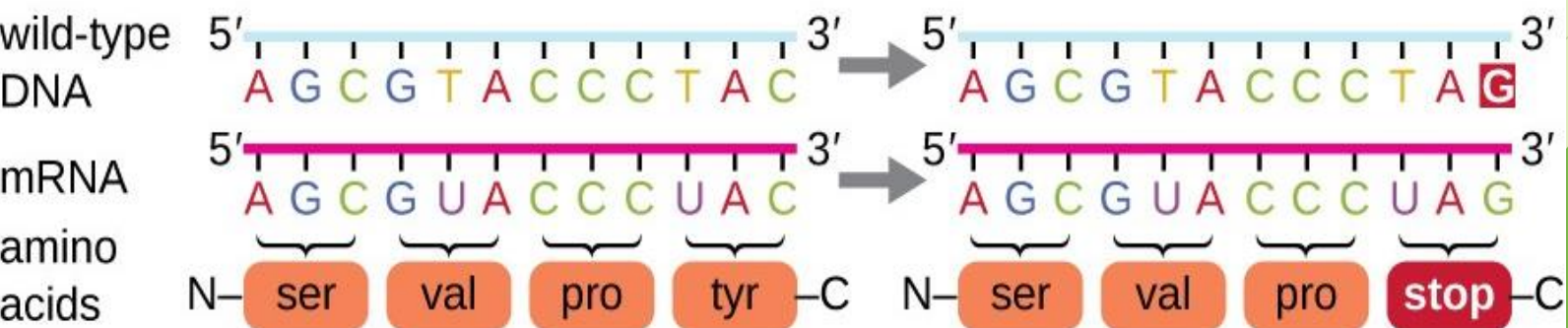
silent: has no effect on the protein sequence



missense: results in an amino acid substitution



nonsense: substitutes a stop codon for an amino acid

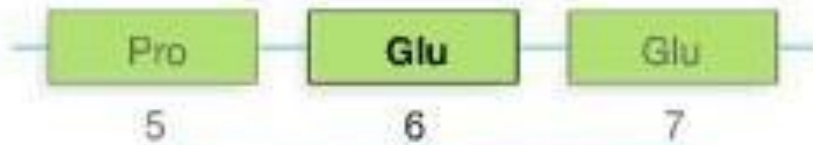


Substitution Examples:

1. Sickle cell anemia
2. Color blindness
3. Albinism



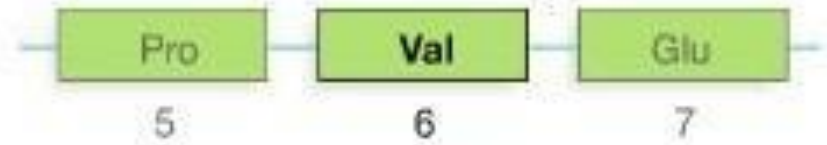
(a) Normal amino acid sequence



Normal red blood cells



(b) Single change in amino acid sequence



Sickled red blood cell



Frameshift Affects:

- ▶ Bases are inserted (put in) or deleted (taken out)
- ▶ Very harmful because a mistake in DNA is carried into mRNA and results in many wrong amino acids

- ▶ For example, read the following sentence:

Original: The fat cat ate the wee rat.

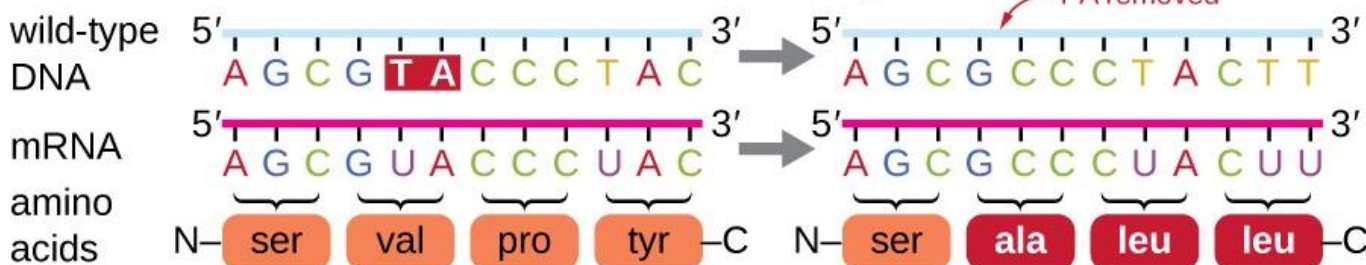
Frame Shift: The fat **caa tet hew eer at.**



***The “t” in cat was deleted causing most of the sentence to be wrong!

frameshift mutation: insertion or deletion of one or more bases

Insertion or **deletion** results in a shift in the reading frame.



Practice Point Mutations...

▶ DNA: **GTA GTA GTA**

▶ What type of single base change is the following mutation?

GTA GAG TA Frameshift/Deletion!

▶ What type of point mutation is the following mutation?

GTA GA GTA Substitution!

Types of mutations: Chromosomal mutations (*not just a base)

- ▶ Chromosomal Mutations: produces change in whole chromosomes
- ▶ Chromosomes break or are lost
- ▶ Broken chromosomes may rejoin incorrectly
- ▶ Almost always lethal (kills) when it occurs in a zygote (fertilized egg that will become a baby)
- ▶ Results in major changes to proteins produced



Chromosomal Mutations

- ▶ Results in major changes to proteins produced
 - ▶ Deletion - loss of all or part of a chromosome
 - ▶ Duplication - extra copies of a chromosome
 - ***Also called polyploidy
 - ▶ Inversion - reverse the direction of chromosomes
 - ▶ Translocation - when part of a chromosome breaks off and attaches to another



Chromosomal Mutation



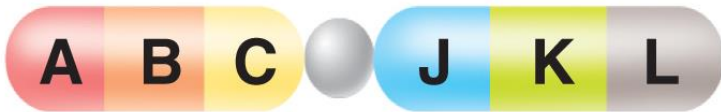
Original chromosome



Deletion



Duplication



Translocation



Inversion