

Genetic Mistakes: Mutations

Remember...

- ▶ DNA → gene → mRNA → amino acid order → protein shape → protein function → trait
- ▶ So what happens if you change the DNA sequence?

Genetic Mistakes

- ▶ **Mutation** – a change in DNA, mRNA, or protein.
- ▶ Not all mutations are bad; some are good!
- ▶ This is how populations get NEW traits!

Point Mutations

- ▶ **Substitution** – when a base is incorrectly paired; affects only ONE nucleotide in the codon.
- ▶ Ex.
 - Normal: THE FAT CAT
 - Mutated: THA FAT CAT
 - May or may not code for a different amino acid.

Frame Shift Mutations

- ▶ **Deletion** – when a nucleotide is skipped during replication or transcription.
 - Normal: THE FAT CAT
 - Mutated: THF ATC AT
- ▶ **Insertion** – an extra nucleotide is added
 - Normal: THE FAT CAT
 - Mutated: THA EFA TCA T
- ▶ Changes every codon AFTER the mutation

What are the Effects of Mutations?

1. No new amino acids – no effect
2. Premature stop
3. New amino acids
4. Missing stop

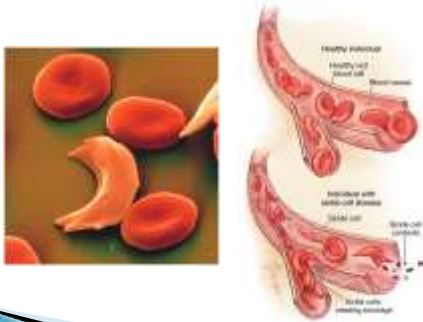
Consequences...

- ▶ If a mutation happens in a gene, and it is passed on...a person could have a dysfunctional protein (ENZYMES???)

Genetic Disorders

- ▶ Genetic disorders are caused by mutated genes that are passed on...
- 1. **Sickle cell anemia**
 - **mutation:** substitution
 - **protein effected:** hemoglobin in red blood cells
 - Decreases circulation and causes pain.

Sickle Cell



Genetic Disorders

- 2. **cystic fibrosis**
 - **Mutation:** deletion (70% of cases)
 - **Protein effected:** cell transport protein
 - Patient produces lots of mucous that houses infections (pancreas, lungs, etc.) and malnutrition

Cystic Fibrosis



Genetic Disorders

- 3. **PKU**
 - **Mutation:** many different types
 - **Protein effected:** enzyme that breaks down the amino acid phenylalanine
 - IF the person eats food with the amino acid → builds up → causes brain damage
 - **All newborns are screened for PKU

PKU Warning Labels

Supplement Facts

Serving Size: 1 Packet
 Servings Per Container: 30

Amount per Serving		% Daily Value*
Calories	10	
Total		
Sodium	150 mg	6%
Glucosamine Sulfate	1500 mg	

* Daily Value not established.
 † Percent Daily Values are based on a diet of 2,000 calories. †† Your daily values may be higher or lower depending on your calorie needs.

Other Ingredients (Dose powder): Aspartame (sweetener), Citric Acid, Sorbitol (sweetener), Phosphates, Caffeine, phenylalanine.

Genetic Disorders

4. **Tay-sachs**

- **Mutation** – insertion
- **Protein affected** – enzyme that breaks down lipids
- Lipids build up in infants brain → brain dead → death by age 7 (in all cases)

Tay Sachs