

Name: \_\_\_\_\_

Hour: \_\_\_\_\_

# Gene Mutations

Mutations = permanent changes in the sequence or number of nucleotides in DNA

1) Point Mutations = mutations that involves a single nucleotide

- Substitutions = changing one nucleotide with a different one

o May be:

- Silent mutation = has NO effect on the amino acid sequence → still codes for same amino acid

• Ex. ACCGCGUCG → ACCGCCUCG  
Thr - ala - ser      Thr - ala - ser

- missense mutation = codes for and inserts a different amino acid

• May affect protein structure and function

• Ex. ACCGCGUCG → ACCGGGUCG  
Thr - ala - ser      Thr - gly - ser

- Nonsense mutation = changes a sense codon to a STOP codon

• Ex. ACCGCGUCG → ACCGCCUAG  
Thr - ala - ser      Thr - ala - STOP

2) Frameshift Mutations = a deletion or addition of nucleotides may cause a shift in the "reading frame" of mRNA

1) Deletion = one or more nucleotides are lost from the sequence

• Ex. ACCGCGGUCG → ACCCUCG  
Thr - ala - ser      Thr - leu - ?

2) Insertion = one or more extra nucleotides are added to the sequence

• Ex. ACCGCGUCG → ACGCGCGCCUCG  
Thr - ala - ser      Thr - arg - ala - ser

o changes the codon triplets read from that point on

▪ Ex. THE RED FOX ATE THE FAT RAT

▪ almost always BAD



Syndrome	What type of mutation? Where is the mutation?	What happens if you have this syndrome? Symptoms?	What treatment options are available?
Sickle Cell Anemia			
Cystic Fibrosis			
Tay-sachs			
Crohn's Disease			
Smith-magenis Syndrome			
Hypertrophic Cardiomyopathy			
Cri-du-chat Syndrome			