









- 1  **Chapter 18**
Gene Mutations & DNA Repair
- 2  **Nature of Mutation**
 - ▶ Importance
 - ▶ Diversity
 - ▶ Knockout mutants
 - ▶ Biochemistry, developmental biology, physiology, behavioral science
- 3 
 - ▶ Categories
 - ▶ Somatic
 - ▶ Cancer
 - ▶ Germ-line
 - ▶ Gene mutations
 - ▶ Chromosome mutations
- 4 
 - ▶ Types
 - ▶ Base Substitutions
 - ▶ Transition (purine → purine or pyrimidine → pyrimidine)
 - ▶ Transversion (purine → pyrimidine)
 - ▶ Insertions & deletions
 - *sam the cat ate joe rat
 - ▶ Frameshift mutations
 - sam fth eca tat ejo era t
 - ▶ In-frame insertions/deletions
 - Sets of 3 added or removed
 - sam ftl the cat ate joe rat / sam_cat ate joe rat
 -
- 5 
 - ▶ Expanding trinucleotide
- 6 
 - ▶ Phenotypic effects
 - ▶ Forward or reverse mutation
 - ▶ Missense
 - ▶ Nonsense
 - ▶ Silent
 - ▶ Neutral (missense)
- 7 
 - ▶
 - ▶ Loss-of function
 - recessive
 - ▶ Gain of function
 - dominant
 - ▶ Conditional
 - ▶ Lethal
 - ▶ Suppressor mutations
 - Double mutations
 - Same gene or another location
- 8 
 - ▶ Mutation Rate
 - ▶ Frequency wild-type changes to mutant
 - ▶ # mutations per biological unit
 - ▶ Cell division, gamete, round of replication

This is only a guideline topics discussed in-class as well as the assigned pages from the text and supplemental material may also be on the exam.

- ▶ How often a mutation arises
- ▶ Affected by
 - ▶ Frequency of primary change
 - ▶ Probability of repair
 - ▶ Probability mutation is recognized
- ▶ Adaptive mutation?
 - ▶ stress

9  **Causes of Mutations**

- ▶ Spontaneous Replication Errors
 - ▶ Tautomeric shift
 - ▶ Change in position of protons in the bases
 - ▶ Nonstandard pairings (wobble)

10 

- ▶ Incorporated error
 - ▶ Replication error
 - ▶ Strand slippage
 - ▶ Unequal crossover

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- ▶ Spontaneous Chemical Changes
 - ▶ Depurination
 - ▶ Loss of purine
 - ▶
 - ▶
 - ▶
 - ▶ Deamination
 - ▶ Loss of amino group

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- ▶ Chemically Induced Mutations
 - ▶ Base analogs
 - ▶ CG→TA & CG→TA
 - ▶
 - ▶
 - ▶
 - ▶
 - ▶

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- ▶ Chemically Induced Mutations
 - ▶
 - ▶ Alkylating agents
 - ▶ CG→TA & CG→TA
 - ▶ Deamination
 - ▶ CG→TA & CG→TA
 - ▶
 - ▶ Hydroxylamine
 - ▶ CG→TA
 - ▶
 - ▶

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- ▶ Oxidative reactions
 - ▶ CG→TA

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- ▶ Interclalating agents
 - ▶ frameshifts
- ▶
- 15 ▶ Radiation
 - ▶ Prymidine dimers
 - ▶ thymine dimers (most common)
- 16 ▶ Ames Test
 - ▶ Determine if chemicals are hazardous (carcinogenic)
- 17 ▶ Radiation Exposure in Humans
 - ▶ Effects depend on duration and intensity of exposure
 - ▶ Hiroshima & Nagasaki (Japan)
 - ▶ Techa River (Russia)
 - ▶ Chernobyl (Ukraine)
- 18 **Chernobyl**
 - ▶ April 26, 1986
- 19
- 20
- 21
- 22
- 23
- 24 **DNA Repair**
 - ▶ Redunancy
 - ▶ Damage can be corrected by more than one pathway
 - ▶ Proofreading
 - ▶ Polymerase 3'→5' exonuclease activity
 - ▶
- 25 ▶ Mismatch Repair
 - ▶ E. coli
- ▶
- 26 ▶ Direct Repair
 - ▶ Changes altered nucleotides back to their original structures
- 27 ▶ Base-Excision Repair
 - ▶ Specific glycosylase
- 28 ▶ Nucleotide-Excision Repair
 - ▶ Removes lesions that distort helix
 - ▶ Pyrimidine dimers
- 29 ▶ Genetic Diseases & Faulty DNA Repair

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