Week 11
Genetics: BIO-2306

The concepts this resource covers are the topics typically covered during this week of the semester. If you do not see the topics your particular section of class is learning this week, please take a look at other weekly resources listed on our website for additional topics throughout the semester.

We also invite you to look at the group tutoring chart on our website to see if this course has a group tutoring session offered this semester.

If you have any questions about these study guides, group tutoring sessions, private 30 minute tutoring appointments, the Baylor Tutoring YouTube channel or any tutoring services we offer, please visit our website www.baylor.edu/tutoring or call our drop in center during open business hours. M-Th 9am-8pm on class days 254-710-4135.

Keywords: Mutation, Mutagen, DNA Repair, Electrophoresis

Topic of the Week: ***DNA Mutations and Repair*** (18)

DNA Mutation: a change in DNA base sequence. May lead to genetic variation and diversity, but may also lead to genetic diseases, cell damage or cancer.
***note:** DNA mutations are often used to investigate molecular (DNA/RNA/protein) function in lab (ex. *Lac* operon)

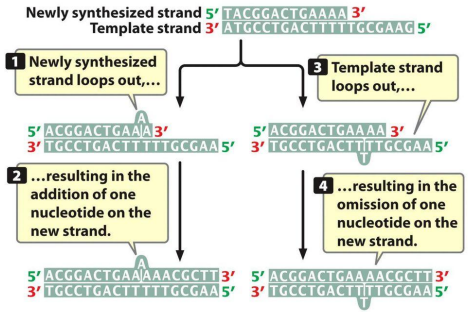
Somatic: mutation of a body cell that will not be passed to offspring
Germline: mutation of gametes which will affect an organism's *offspring*
→ genetic diseases

Causes of Mutations:

Spontaneous: natural changes in DNA

Strand Slippage: multiple variations of a sequence occur on a DNA molecule. This could cause the DNA strand to **slip** and form a loop. The *loop itself* will give a short deletion. However, the location of the loop will determine the overall change to DNA

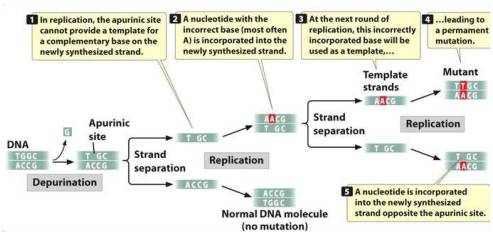
Template Strand: deletion mutation
New Strand: addition mutation



Short video: https://www.youtube.com/watch?v=E18Jl-LP4TU

Unequal Crossing Over: misalignment of chromosomes in **Prophase I** of meiosis causes an **insertion** on one chromosome and a **deletion** on another (**indel**).

Depurination: removal of a purine (A or G) base from DNA by cleavage of the attachment to the 1'C. During replication, this location on the **template strand** can't serve as a *template*, so the *DNA-pol* will synthesize an **A** nucleotide on the new strand. (**right**)



Induced: caused by a **mutagen** (something that is mutation-inducing)

Base Analogs: a molecule similar to a base that causes transition mutation

Intercalating Agents: molecules which intercalate (stick themselves between) the DNA helix; distorted DNA structure leads to ***indels*** → frameshift mutation

Ethidium Bromide: stain used to dye DNA in molecular bio because it intercalates DNA and gives off fluorescent red/purple color

Radiation: may change DNA sequence or helix/chromosomal structure

Ionizing Radiation: high energy rays penetrate tissue and damage DNA

Non-ionizing Radiation: causes bases to dimerizes, forming bulky lesions of DNA (ex. Thymidine dimers from UV radiation) → lesions prevent normal DNA replication

Types of Mutations:

Base Substitution: one base or type of base is substituted for another

Transition: Purine → Purine; Pyrimidine → Pyrimidine

Transversion: Purine → Pyrimidine; Pyrimidine → Purine

Indels: mutation that causes an insertion and a deletion

Frameshift Mutation: changes the entire reading frame; from mutation site downstream all other amino acids are affected

In-Frame Mutation: changes a particular amino acid without changing the reading frame; in multiples of 3

Expanding Nucleotide Repeats (ENR): when genes exist in many copies, DNA may exhibit *strand slippage* due to hairpin loops caused by complementarity. The number of copies of an ENR gene is directly correlated to how severe its effects are and how early it will onset.

Anticipation: Over generations, a particular **ENR** gene will get more and more severe and start sooner because of continued strand slippage

Protein Mutations:

General:

Missense: mutation results in coding for a <u>different</u> amino acid

Silent: changes in base(s) does <u>not</u> result in the change of amino acid due to the degeneracy of the code

Nonsense: turns an amino acid codon into a nonsense codon (**STOP**) → ends translation early!

Specific:

Neutral: change in protein sequence, but no change in protein function

Loss of Function: takes away a trait normally present (think of as recessive)

Gain of Function: gives a new trait *not normally* present (think of as dominant)

Conditional: a mutant phenotype that will *only* be expressed under a given [set of] condition(s).

Lethal Mutation: causes death in the organism which it affects

Suppressor Mutations: a mutation that hides the effect of another → restores 'wild type'

Intragenic: suppressor mutation is in the same gene as the mutation it is affecting

May alter the original mutation to restore the original amino acid

May cause a new mutation to give a *new and functional* amino acid

Intergenic: occurs in another gene affecting the *translation* of the mutant gene

Effect on Phenotype:

Forward: normal phenotype to mutant phenotype

Reverse: mutant phenotype to normal phenotype

DNA Repair:

Direct repair: Converts altered base back to its original without removal (chemically changes altered base)

Mismatch Repair: Mismatched bases cause a distorted lesion in the DNA which will be recognized by proteins

Lesion is excised by mismatch repair proteins and replace it with special polymerases and new nucleotides

Note: Damage to DNA repair mechanisms can cause diseases like *Xeroderma pigmentosum* or even cancer

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Highlight #1: Gel Electrophoresis (19.2)

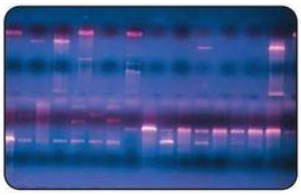
Gel Electrophoresis (GE): Separation of DNA due to its mass (ie molecular weight)

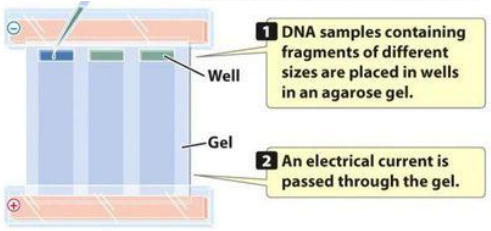
DNA moves down an electrophoresis gel due to its net negatively charged backbone

Gel: highly porous agarose gel allows DNA to pass through. The largest pieces will travel the furthest and the **smallest pieces** will travel the **furthest** and the **largest fragments** will travel the ***least far***. DNA is dyed to visualize under UV light **(above, right)**

Cathode (-): the negatively charged pole will repel the DNA towards the *anode*

Anode (+): the negatively charged DNA will be attracted to the positive charge

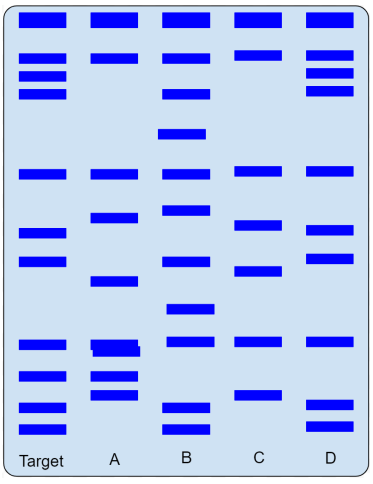




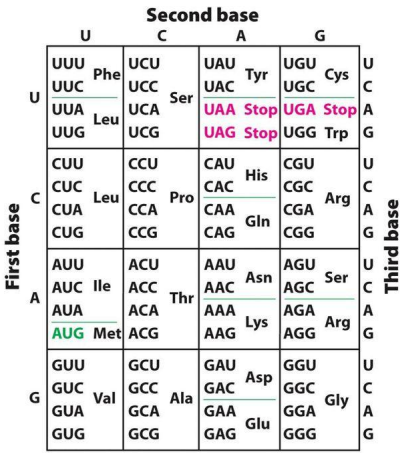
CHECK YOUR LEARNING

Concept Check: *(Answers found on the last page)*

1. What is the word for a DNA mutation that would alter the sequence of amino acids in a protein but would *not* change the protein identity?
    a. Frameshift
    b. Neutral
    c. Silent
    d. Nonsense
2. DNA of individuals who experienced high levels of radiation at the Chernobyl power plant disaster had severe damage to DNA and chromosomal structure. What type of radiation did they most likely encounter?
    a. Non-ionizing (UV)
    b. Neutral
    c. Slow
    d. Ionizing
3. A DNA molecule moves towards the _____ due to its _____ charge.
    a. Anode; negative
    b. Cathode; positive
    c. Anode; positive
    d. Cathode; negative
4. Bob's family has a long running genetic disease caused by a single mutated protein. However, Bob does not display this. After a DNA sequence was done, it was found that the disease-causing gene was intact and mRNA was transcribed, but there was another mutation that prevented cap-binding protein from attaching it to a ribosome. What type of mutation does he have?
    a. A neutral mutation
    b. A frameshift mutation
    c. An intergenic suppressor mutation
    d. An intragenic suppressor mutation
5. DNA evidence is collected at a crime scene. The "target" DNA collected is compared to 4 other samples of DNA from suspects. Which DNA profile *best* matches the target?
    a. A
    b. B
    c. C
    d. D



6. A DNA transition mutation of the following ***coding*** sequence could *not* have resulted in which of the following amino acids?
   5'-ATG-3'
   a. Met (start)
   b. Val
   c. Thr
   d. Ile



THINGS YOU MAY STRUGGLE WITH:

1. In unequal crossing over, there is no net gain or loss of genetic info; a larger part of a homolog is transferred to one chromosome and the smaller segment of the crossover is transferred to the other chromosome (an **indel**). The issue here in terms of change of DNA is twofold:
   a. The first is that a possible break in a gene could be initiated.
   b. The second is the *serial positioning effect*, where the actual order in which genes are expressed
2. ENR often includes specifically *tri*-nucleotide repeats, thus may be called expanding trinucleotide repeats. They are the same thing, but this is just specific to the number of repeats of a DNA within a given gene.
3. DNA fragment size can be determined by comparing it to a known "ladder." A ladder is given by a company to show bands of DNA of specific size. Any band horizontal to a ladder band will be the same size.
   a. Exception to this: circular DNA can change shape as it supercoils, so it may travel "further" than DNA with same # of BPs should because it is *very* condensed

CONGRATS: You made it to the end of the resource! Thanks for checking out these weekly resources! Don't forget to check out our website for group tutoring times, video tutorials and lots of other resources: www.baylor.edu/tutoring!

Answers to check your learning questions are below!

Answers:

1. B.
2. D.
3. A.
4. C.
5. D.
6. A.