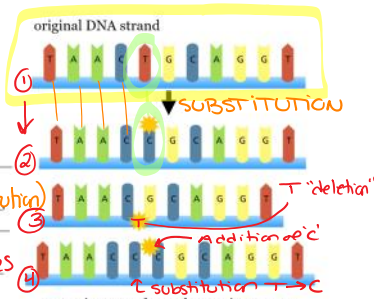


**Lesson 1.5 – Mutations**

**Mutations**

- Up to this point, we have considered DNA molecules to be permanent structures, but in nature the DNA of organisms is **always changing**
- Any change to the sequence of bases in a DNA molecule is called a **mutation**
- These changes can be small, such as **single base changes (substitution)**, **single base deletion**, or **addition**, or they can be large, causing **multiple base changes** and **deletions** as big as **whole chromosome**



Translocation  
create genetic variation

- mutations can change base pairs, remove base pairs, or add base pairs
- In most organisms the **rate of mutation is extremely small** perhaps as little as one or two changes per generation
- Mutation is **incredibly important for evolution** because it **creates new alleles**



- delayed growth + development
- intellectual impairment
- low muscle tone
- seizures

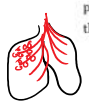
**Effects of Mutations**

- Positive Mutation** = any mutation that **improves** the function of a gene by causing it to create a **better protein**, and will give that organism an **advantage** over other members of its species

antibiotic resistance makes bacteria immune to an antibiotic, an example of a positive mutation

strain 1 is "resistant" to the antibiotic - bacteria grows/lives even after being treated with antibiotic

- Negative Mutation** = any mutation that **harms** the function of a gene by causing it to create a **less effective protein** or a protein that does not perform its job, and will give that organism a **disadvantage**



Normal airway vs Airway with CF

- ↑ risk of infection
- poor mucous membrane
- neg. affected muscles
- breathing difficult

cystic fibrosis is caused by a **mutation in gene CFTR**, an example of a negative mutation

- Neutral Mutation** = any mutation that **does not change** the function of a gene by causing it to create a protein that is **identical in structure + function** provides its organism with **no advantage or disadvantage**



**An Analogy:** If the **gene** was a **sentence**, and its **function** (meaning) was its protein: "The old dog sit"

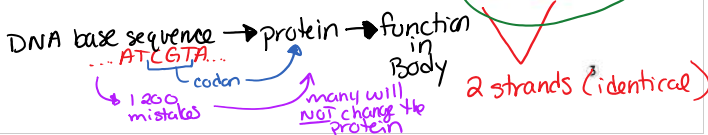
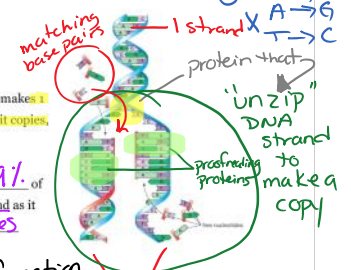
- Positive mutation: **The old dog sat**. The sentence has **improved function**
- Negative mutation: **The old dog sit**. The sentence has **reduced function**
- Neutral mutation: **The old cat sit**. The sentence has **identical function**

1 letter swaps  
1 base pair

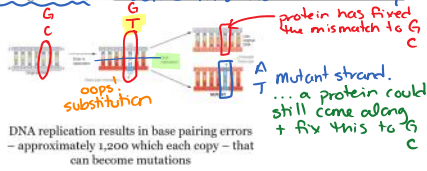
**Sources of Mutations**

1. **DNA replication**

- The protein that replicates DNA is **extremely accurate**; it only makes **1 base pairing mistake** for every **100,000 base pairs** it copies, but across all 46 chromosomes this creates **120,000 mistakes**
- The same protein that replicated DNA fixes **99%** of these mistakes by **proofreading** the new DNA strand as it goes, **leaves 1,200 mistakes**



- Other proteins look for these mismatches and repair them, removing almost all of the remaining base pairing mistakes, but some mistakes will always remain



## 2. Mutagen

- natural and man-made agents that can cause mutations are called "mutagen"

### Measuring Radiation's Effects

Activity	Millirads
Typical yearly dose, all sources	100.00
Full set of dental X-rays	40.00
Round-trip flight	2.00
Flying round-trip from D.C. to Los Angeles	5.00
Living outside nuclear power plant for a year	0.10

Health risk	Expected life lost
Smoking a pack of cigarettes a day	6 years
Being 15 percent overweight	3 years
Working in coal mines 40 years	2.2 years
Working in nuclear plant (1,000 mrem/yr)	51 days
Typical annual background radiation dose (100 mrem/yr)	18 days

- Nuclear radiation (alpha particles, gamma rays) and high energy light (X-rays, ultraviolet light) physically damage or break down cells.

↑  
The Sun

- Chemicals such as benzene attach themselves to DNA and cause errors during replication



benzopyrene from cigarettes inserted into the DNA double helix



interferes with DNA replication ... and causes errors to the code.

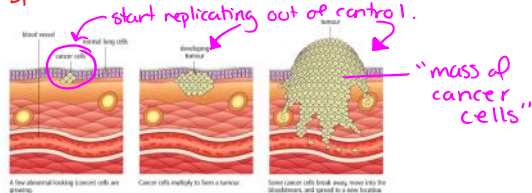
contain KNOWN carcinogens (also in "vape-juice")

4

- Heavy metals such as arsenic, cadmium, mercury also damage DNA and interfere with DNA repair
- Some viruses insert themselves into the DNA of the host cell, interrupting genes

### Mutations and Cancer

- While some cancers are inherited, almost all of them are caused by mutations to genes that alter a cell's identity and affect normal cell division
- Agents that cause cancer, called "carcinogens", are thus mostly mutagens
- Cancer mutations result in abnormal cells with uncontrolled growth that can spread or invade other tissues.



5

# Homework

Assignment #5: Complete the following worksheet in the space provided below

1. Identify each of the following examples as a positive, negative, or neutral mutation:

Example	Type of Mutation
a. Some plants carry a mutated gene that protects them from a fungus parasite called powdery mildew.	_____
b. Some people are born with a mutated gene that prevents the cells in their throat and lungs from making normal mucus, as a result the thick mucus accumulates and clogs the lungs (cystic fibrosis).	_____
c. The Spirit Bears of coastal British Columbia have a mutation that makes their fur white instead of black, but does not affect their survival.	_____
d. Some people have a mutation that prevents the virus HIV from infecting their cells.	_____
e. Some people have a mutation which causes blood protein involved in clotting to have a different shape, as a result they have internal and external bleeding (haemophilia).	_____

2. Which of the following can result from a DNA mutation?
- The protein will no longer function.
  - The protein will have reduced function.
  - The protein will have improved function.
  - All of the above can occur.
3. Which of the following correctly describes a mutation?
- a change in the base pair sequence of a DNA molecule
  - a change in the proteins that wrap around the DNA molecule
  - a change in the order of sugars and phosphates in a protein
  - a change in the structure of a protein
4. A man is sitting outside of a restaurant on a park bench smoking a cigarette, reading the newspaper. Which of the following is a mutagen that he is exposing himself to?
- the newspaper
  - the cigarette
  - the oxygen he is breathing
  - the park bench
5. The three kinds of mutations are called:
- neutral, negative, and carcinogenic
  - positive, negative, and neutral
  - mutagens, carcinogens, and mutations
  - complete, incomplete, co-mutant

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6. A woman breaks her arm riding motocross. After checking into the hospital, she waits in the emergency room and drinks a glass of water. Eventually the doctor identifies the break with an X-ray and casts the arm. Which of the following is a mutagen that she is exposing herself to?
- the glass of water
  - the hospital waiting room
  - the chair she sits on
  - the X-ray exam
7. Cancer results when mutations cause body cells to lose their identity and divide uncontrollably. Cancer is an example of
- a negative mutation.
  - a positive mutation.
  - a neutral mutation.
8. A mutation in a gene that creates proteins which carry fat in blood was discovered in people living in a small village in northern Italy. Blood fat can accumulate inside the walls of blood vessels, blocking blood flow and causing heart attacks and strokes. The mutation greatly reduces the accumulation of fat inside the walls of blood vessels, and so is an example of
- a neutral mutation.
  - a negative mutation.
  - a positive mutation.
9. A mutation which does not affect the function of the protein is called a
- positive mutation.
  - negative mutation.
  - neutral mutation.
10. Cats in the American Curl breed have ears that curl upwards at the tips. This does not appear to affect their hearing in any way. This mutation is an example of
- a neutral mutation.
  - a positive mutation.
  - a negative mutation.
11. The Huntingtin gene creates a protein which is found in many different body cells. In humans there is a mutant allele of this gene which results in a protein that kills many types of cells, especially brain cells. This mutation is an example of
- a negative mutation.
  - a neutral mutation.
  - a positive mutation.

7

## ANSWERS:

- positive
  - negative
  - neutral
  - positive
  - negative
- D
- A
- B
- B
- D
- A
- C
- C
- A
- A
- A

Original DNA Sequence:	TACACCTTGGCGAGACT	Type of Mutation
Mutated DNA Sequence #1:	TACA <b>T</b> CTTGGCGAGACT	substitution
Mutated DNA Sequence #2:	TAC <b>(added G)</b> ACCTTGGCGAGACT	addition
Mutated DNA Sequence #3:	TACACCTTAG <b>(missing G)</b> CGAGACT	deletion
Mutated DNA Sequence #4:	TACACCTTGGCGAG <b>A</b> CT	substitution
Mutated DNA Sequence #5:	TACACCTTGG <b>(missing C)</b> GAGACT	deletion

13. TEF ATC ATA TET HER AT.. does not make sense. This is a NEGATIVE mutation

12. Identify the type of mutation below (substitution, addition, or deletion). Fill in the table.

Original DNA Sequence:	TACACCTTGGCGACGACT	Type of Mutation
Mutated DNA Sequence #1:	TACATCTTGGCGACGACT	
Mutated DNA Sequence #2:	TACGACCTTGGCGACGACT	
Mutated DNA Sequence #3:	TACACCTTAGCGACGACT	
Mutated DNA Sequence #4:	TACACCTTGGCGACTACT	
Mutated DNA Sequence #5:	TACACCTTGGGACGACT	

13. Look at the following sequence: THE FAT CAT ATE THE RAT. Delete the first H and regroup the letters in groups of three- write out the new groups of three. Does the sentence still make sense? What type of mutation is this an example of?