Review Questions
Mutations

1. **What is a mutation?**
   A mutation is a change in the sequence of bases in a DNA molecule. A mutation can occur in any cell but the most important ones happen in the gamete-making cells because they are passed onto the next generation.

2. **What causes mutations?**
   Many mutations are caused by mutagens. Common mutagenic sources include radiation and DNA interacting-chemicals. Ultraviolet light has a shorter wavelength than visible light and has more energy. UV has enough energy to damage biological molecules like DNA. Ionizing radiation, such as x-rays, gamma rays, and the decay products of radioactive substances are even more damaging due to their shorter wavelengths and/or higher energies. All chemical carcinogens cause mutations: industrial compounds (e.g. benzene, formaldehyde, PCB’s), heavy metals, petroleum products, dyes, tobacco, asbestos, pesticides, base analogs. Many mutagens are products of the industrial revolution but there are also many that naturally occur. We are surrounded by mutagens.

   Another source of mutations is the replication machinery of a cell itself. Replication is not perfect. On average replication has an error rate of 1 out of every 10,000-100,000 base pairs. These mistakes are quickly fixed by special proofreading enzymes called exonucleases. For example, UV light damages DNA by making thymine-thymine dimers. UV causes adjoining thymine bases to bond together making a lump in the strand. Exonucleases recognize the error, remove the dimer, and replace it with correct DNA. Xeroderma pigmentosum is a genetic disorder in which a person is born without the exonuclease that repairs thymine-thymine dimers. The affected are highly susceptible to skin cancers if they are exposed to UV light. During the day they must remain indoors at all times and can only go outside at night.

![Diagram of UV-induced thymine dimer and repair process](image-url)
The exonucleases are not perfect and do not catch every mistake. On average, for every 10 billion nucleotide pairs replicated there is one error. So every time a DNA molecule replicates there is a 33% chance of a mutation. Now add onto this the effects of everyday exposure to mutagens and the mutation rate rises. Mutations are fairly common.

3. **Describe some common chromosomal mutations: inversions, deletions, duplications, fusions, fissions, and translocations.**
   Mutations range from a change in a single nucleotide pair up to a change in vast regions of a chromosome. Chromosomal mutations are detected by comparing the banding pattern of chromosomes. Chromosomal mutations cause several genetic disorders but also are extremely useful in tracing evolutionary change in a related group of organisms. Below are some examples of the most common types of chromosomal mutations.

4. **Name the two basic kinds of point mutations.**
   Point mutations usually involve a change in just a single nucleotide pair. The first kind is called a base substitution. As its name implies, a base is replaced by an incorrect base. The second kind is actually two kinds: insertions and deletions (the addition or subtraction of nucleotide pairs).
5. **What is the difference between a missense mutation and a nonsense mutation?**

Point mutations can be classified as a base substitution or an insertion/deletion but they are also classified on whether the change produces a premature stop in the gene. Nonsense mutations are mutations that result in a premature stop. They are called “nonsense” because all the codons following the stop codon are never translated, they are nonsense. If there is no premature stop, the mutation is said to be missense. So when classifying a mutation, you have to be specific. For example, a mutation might be described as a base substitution-missense, or a base substitution-nonsense, or an insertion-missense, or a deletion-nonsense, and so on.

6. **What is a frameshift mutation?**

Picture the genetic code as being composed of a language of three-letter words without any spaces in between. An insertion or deletion will cause every “word”
downstream of the change to be different. We call this a change in the “reading frame”. A frameshift mutation is like taking a scantron test. If you accidentally get one question off every answer from then on will probably be wrong. Frameshift mutations radically change proteins. Most of the time they are quite deleterious.

We can add to our classification of mutations. Anytime you have a deletion or insertion mutation you also have a frameshift. The only time an insertion or a deletion is not a frameshift is if a whole codon (or groups of codons) is inserted or deleted. Let’s say you have an insertion mutation that produces a premature stop. We would classify that as an insertion nonsense frameshift mutation. How about a deletion mutation? Deletion missense frameshift. You get the picture?

7. Why are mutations so important to living organisms?
Most mutations are harmful to an organism. Random changes in the gene sequence may result in the malformation and subsequent loss of function of a protein. Humans suffer from over 3000 genetic diseases. Every one of these is caused by a mutation. Some mutations however are neutral. The protein may be identical or, if changed, works equally well. A very few mutations are beneficial to an organism. A different protein may alter or create a trait that better adapts an organism to its niche in life. In the big picture, all living organisms today are the result of the accumulation of beneficial mutations over the past 3.5 billion years.

8. What is a “sport”? Give an example.
A sport is an organism deviating abruptly from type. That sounds kind of vague. What it means is that if a breeder finds in her fields or flocks an individual organism that has a novel trait as the result of a random mutation, it is called a sport. Many of our varieties of domesticated plants and animals originated as sports. Take apples for instance. The golden delicious variety originated as a sport in 1912 in West Virginia. A random mutation of a red delicious changed the color and flavor and a new variety was born. Sports are great examples of beneficial mutations.
9. **Describe the purpose and process of mutation breeding?**
   Since many new varieties of domesticated plants and animals originate as sports, breeders, anxious for new varieties, discovered that they could increase the number of sports by exposing their flocks and fields to mutagens. In agriculture, more than 3000 new varieties of crops were created in the 20th Century by exposing seeds to non-lethal amounts of radiation. Of course, most plants exposed to the mutagens would die, or produce useless traits, but occasionally a novel organism would appear. Mutation breeding is still used, but not to the extent it once was, because of advances in genetic engineering.