

#ref #ret

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## 0.1 | Questions

You may discuss the following questions in groups but please produce your own individual responses here based on your own synthesis of the concepts. These questions are based on today's learning and **will be assessed** (reassessments are okay if needed). Submit your answers to this assignment.

*Siblings from the same parents are related but not identical.*

- 1) What are all the mechanisms that create this genetic variation between “full” siblings? Describe these processes in as much detail as possible.
- 2) Do you expect there to be any genetic variation between identical twins (from the same fertilized egg, which split into two separate embryos early in development)? Explain your answer.

## 0.2 | Answers!

### 0.2.1 | One

**Crossing Over** During meiosis 1, DNA segments are swapped between homologous chromosomes. These homologs are aligned on the meiotic plates and attached with the synaptonemal complex, where segments of each are broken then recombined with the appropriate nucleotide sequence. Mutations can also occur in the DNA synthesis that fills gaps created by this process.

**Independent Assortment** During metaphase 1, homologs align randomly and independently to form gametes. This results in significant variation of genetic information in each daughter cell. There are  $2^n$  combinations of chromosomes, with  $n$  being the number of unique chromosomes.

**S Phase** Cell division requires the replication of DNA. This replication occurs during the S phase, where ribosomal errors that go undetected lead to genetic variation.

**Environmental / Damage** Environmental factors like UV rays or smoking can damage DNA, leading to genetic variation. Errors can also result from the process of repairing or replacing this damaged DNA. Environmental factors can also induce epigenetic change.

**Viruses** Viruses inject their own genetic information into its host's cells. While this foreign genetic information may not be permanent, it is still genetic variation.

### 0.2.2 | Two

Yes, I do. Identical twins occur when a single egg splits into multiple embryos. Thus, at the very least, genetic variation arising from cell division will occur, not to mention environmental factors and viruses.

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### 0.3 | **More notes!**

#### 0.3.1 | **Trait vs Phenotype**

T: characteristics influenced by genes but can also have nurture component P: Collection of traits

#### 0.3.2 | **Mutations**

- point mutation: single base substitution
  - silent mutation: no effect, doesn't impact codon sequence
  - missense: changes amino acid structure
  - nonsense: inserts a stop codon
- frameshift: insertion/deletion of n amount of bases
  - deleting two shifts the entire sequence to the right
  - break alot of things
  - so frameshift mutation != frameshift, and frameshift mutation sometimes leads to frameshift
- mutagens are like carcinogens for mutations

**Mutation Think Through** thinking through mutations: - Can you think of scenarios in which the insertion or deletion of bases in the above sequence would not result in a frameshift? - deleting or inserting multiples of three that are not in junctions - not true! will fix itself – delete three means 1 and 2, combine, back to three. - delete what would get frameshifted - delete from the end - A silent mutation has no effect on protein sequence. Could a silent mutation ever affect an organism's phenotype? Explain. - no... it shouldnt be able to - could be on some binding site that would break? - mutates protein coding sequences - What functional predictions would you make for a nonsense mutation that occurs very early vs. very late in a gene's sequence? - very early would make it not get created, middle would cause a strange protein, late would make little impact

**Large scale changes** chromosomal rearrangements are a thing. generally not called mutations deletion, duplicatio, inversion, ect. of large sections

#### **Impact**

- **Loss of function**
  - complete loss of protein of function
  - reduction of function
  - -function
- **Gain of function**
  - increase in function
  - new function
  - new expression time
  - +function but, most proteins are like links in a chain