#ref #ret

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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.

2 | Answers!

2.1 | One

2.1.1 | 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.

2.1.2 | 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. Their are 2^n combinations of chromosomes, with n being the number of unique chromosomes.

2.1.3 | 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.

2.1.4 | Environmental / Damage

Enviromental 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.

2.1.5 | 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.

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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3 | More notes!

3.1 | Trait vs Phenotype

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

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 seqence to the right
  - break alot of things
  - so frameshift mutation != frameshift, and frameshift mutation sometimes leads to frameshift
- mutagens are like carcinogens for mutations

3.2.1 | 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 shoudnt be able to - could be on some binding site that would break? - mutates protein coding seqences - 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

3.2.2 | Large scale changes

chromosonal reanrangrments are a thing. generally not called mutations deletion, duplicatio, inversion, ect. of large sections

3.2.3 | Impact

- **Loss of function**
  - complete loss of protein of function
  - reduction of function
  - -function
- **Gain of function**
  - increanse in function
  - new function
  - new expression time
  - +function but, most proteins are like links in a chain. Jehnna's term is "pathway" which she seems to like

germline vs. somatic

4 | And the following questions

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> /Red blood cells have various carbohydrate molecules attached to proteins on their surfaces (see diagram below). Human A-B-O blood types are determined by the presence or absence of two particular carbohydrate modifications, "A" and "B."/ /One gene with three main alleles controls the A-B-O trait; it encodes a glycosyltransferase (an enzyme that can attach carbohydrates to other molecules). The A and B alleles both encode a functional enzyme, but each version of the enzyme generates a different carbohydrate modification, "A" or "B." The O allele encodes a non-functional enzyme./

4.0.1 | What two alleles could a person with blood type A have? With type

B? With type AB? With type O? :CUSTOM$_{ID}$: what-two-alleles-could-a-person-with-blood-type-a-have-with-type-b-with-type-ab-with-type-o

- **A**: *AA, AO*
- **B**: *BB, BO*
- **AB**: *AB*
- **O**: *OO*

4.0.2 | If a person with type AB blood had a child with a type O person,

what possible blood types could their child have? What would be the likelihood of each type? :CUSTOM$_{ID}$: if-a-person-with-type-ab-blood-had-a-child-with-a-type-o-person-what-possible-blood-types-could-their-child-have-what-would-be-the-likelihood-of-each-type A + B, O + O: - A + O -> A - B + O -> B

50% A, 50% B

4.0.3 | If the offspring from the previous question grew up to have a child

with a type AB person, what blood types could their child potentially have? :CUSTOM$_{ID}$: if-the-offspring-from-the-previous-question-grew-up-to-have-a-child-with-a-type-ab-person-what-blood-types-could-their-child-potentially-have O+A||B, A+B: - O + A -> A - O + B -> B - A + A -> A - B + B -> B - A + B -> A+B - B + A -> A+B

1/3 A, 1/3 B, 1/3 A + B

5 | More notes pt 2..

- **differnet inheritence patterns**
- mendelian: dominant vs recesive
  - alleles, alt versions of traits, are responsible for varitations in inherited traits
- incomplete dominance
  - both alles are visible, neither are completely dominant
- codomidance
  - both alleles are visible in distinguishible ways
- polygnenic inheritance
  - single phenotype determined the the addition of multiple
- epistatis
  - one gene alters another!
- S-linked
  - linked by sex