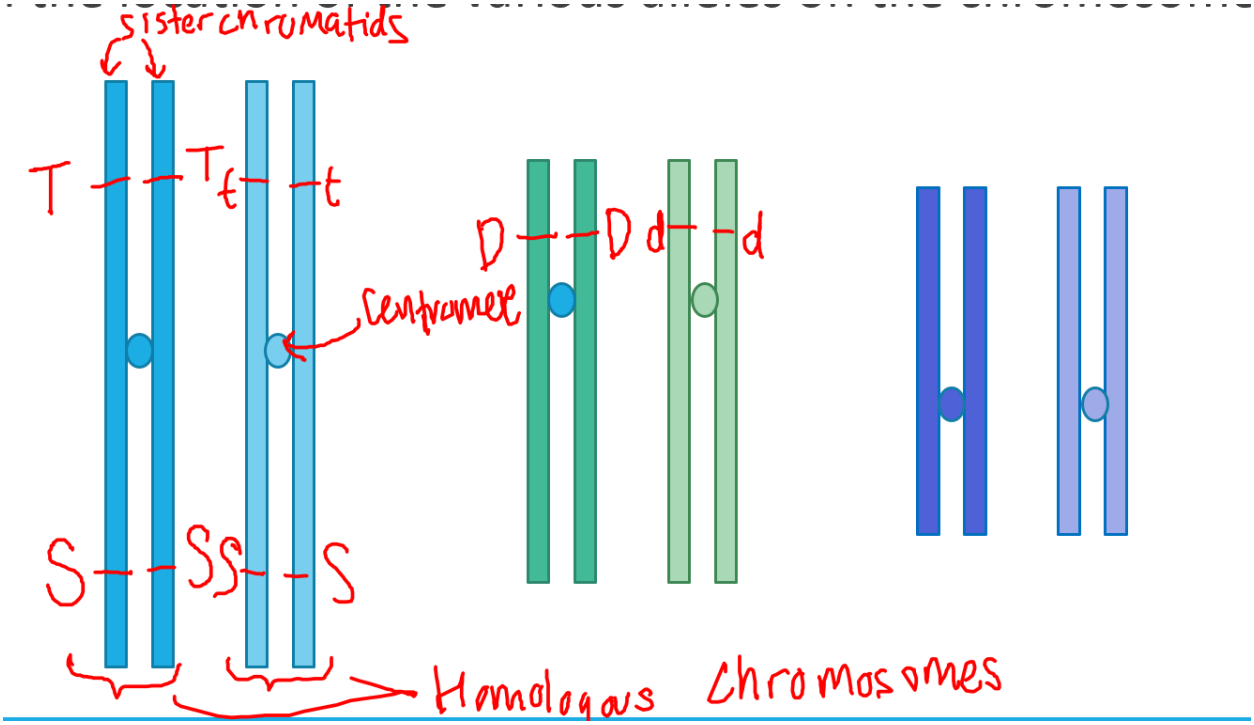
1: You are studying the C. elegans organism, a diploid organism with n = 3. You are interested in 3 genes, which dictate the external appearance of the worm. The worm you caught is heterozygous for the Tan gene (Tt), homozygous for the Smooth gene (SS) and heterozygous for the Dotted gene (Dd). The Tan gene and the Smooth gene are linked on the same chromosome.

a. **What does one of their somatic cells look like after DNA replication and condensation, but before mitosis? Label the location of the various alleles on the chromosomes.**
b. **Add the following labels to your diagram: Sister chromatids, Homologous chromosomes, Centromeres**



c. **At the end of meiosis I, how many chromosomes are present in each of the cells? Are these cells haploid or diploid?**

3 chromosomes

Haploid

2: You have isolated this DNA molecule in the lab:

5’ AGGATGCAATACTTCGCGTAACCATGCTT 3’
3’ TCCTACGTTATGAAGCGCATTGGTACGAA 5’

a. **How many open reading frames are present in this molecule?**

1

b. **What are the protein products created by the open reading frames in the previous question?**

Met Gln Tyr Phe Ala

c. **A point mutation is introduced in the DNA strand as follows. What effect does this have on the length of the mRNA product?**

5’ AGGATGCAATACTTCGCGTCACCATGCTT 3’
3’ TCCTACGTTATGAAGCGCAGTGGTACGAA 5’

No discernible effect on the length of the mRNA product! Changing a stop codon will affect the length of the **protein** product, not the mRNA.

3: The lin-14 gene, which codes for the LIN-14 protein, controls the timing of development in C. elegans. The lin-14 gene is inactive during infancy, and gradually becomes active when C. elegans begins to mature. The lin-14 gene remains highly expressed and active throughout adulthood.

a) **You isolate a sample of cells from a C. elegans organism and inspect the cells under a high-powered microscope. This microscope allows you to observe DNA structure. How would you describe the chromatin structure in the region of the lin-14 gene in an <u>infant</u> C. elegans? What about a <u>mature</u> C. elegans?**

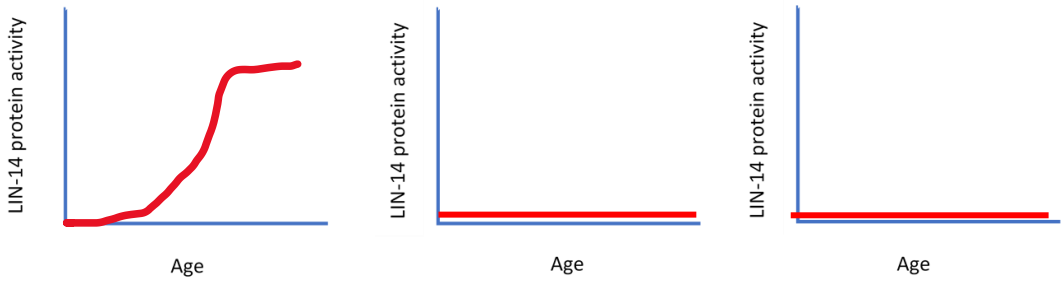
Infant = heterochromatin (gene is inactive)

Mature = euchromatin (gene is actively expressed)

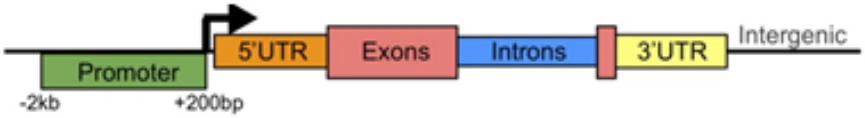
Animals that are homozygous wild-type for the lin-14 gene develop normally. The presence of at least one copy of the mutant allele codes for a defective protein that completely inactivates all LIN-14 activity.

b) **Using vocabulary learned during the lectures, how would you describe this type of mutation?**
Dominant negative, loss of function

c) **Draw a graph depicting LIN-14 protein activity throughout the lifespan (from birth to adulthood) of C. elegans in:**
   a. **An organism that is homozygous for wild-type alleles**
   b. **An organism that is homozygous for mutant alleles**
   c. **An organism that contains one copy of a mutant allele and a wild-type allele**



4: A geneticist is studying the effect of random mutations on the srab-12 gene (depicted below) in C. elegans.



He uses mutagenesis to create introduce a mutation at a random site along the srab-12 gene sequence, creating a mutant organism. The problem is, he does not know which region of the gene contains the mutation. He performs a set of experiments and obtains the following information to help him figure out where he could have inserted the mutation:

Organism	Quantification of srab-12 mRNA levels	Quantification of srab-12 protein levels	Protein sequence	srab-12 Protein function
Wildtype	100%	100%	Met-Ala-Thr-Asp	Normal
Mutant	0%	0%	---	---

A. **In which region of the gene did the mutation likely occur? (Promoter, UTR, Exon, Intron). Explain your answer using data from the table.**

Promoter. If there is no mRNA produced, it is likely that there was a significant transcription error. The promoter region is responsible for the initiation of transcription. Thus, it is likely that this significant transcription error was the cause of a mutation in the promoter region.

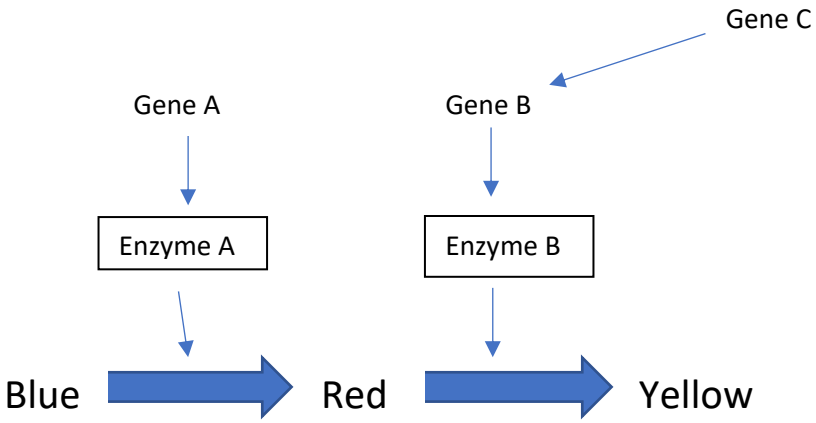
B. **The srab-12 gene is responsible for encoding the Class AB serpentine receptor protein. This receptor is only expressed the surface of neurons, and is not found on the surface of in any other cell type. The geneticist says that it is because neurons possess the srab-12 gene, while other cells do not possess the gene. Is this a valid explanation? If not, propose an alternate explanation that explains why the receptor cannot be found on other types of cells.**

No, because all cell types contain the exact same DNA. THe reason that some cells may not express the receptor, is that the conditions of gene expression vary per cell.

Possible explanations:

- Only neurons possess the transcription factor required for the expression of this gene. Other cells do not express the transcription factor, so the gene cannot be expressed
- Other cells express an inhibitory protein that inhibits the expression of this gene. Neurons do not express this inhibitory factor, so gene expression is allowed to proceed.
- Other answers may include: differences in post-transcriptional processing, differences in mRNA regulation and degradation, differences in post-translational modifications, etc.

5: The determination of eye colour in Drosophilia is determined by a multi-step biochemical pathway, as shown below. The red phenotype displays complete dominance to the blue phenotype, and the yellow phenotype displays incomplete dominance to the red phenotype. The C gene is haploinsufficient in completely inhibiting the function of the B gene.



A. **What would the eye colour of an organism that is AABBCc look like?** Yellow
B. **What would the eye colour of an organism that is AABbcc look like?** Orange

7: An individual's undergoes genetic testing after seeing signs of suspected sex reversal syndrome. Their karyotype indicates they are XY. Further genetic testing indicates that the individual is negative for the SRY gene.

a. **Considering the function of the SRY gene, describe the expected anatomy and physiology of this person.**

   SRY negative = the absence of the SRY gene results in the development of female gonads (ovaries). This individual will likely possess external female genitalia, instead of male genitalia.

b. **Describe an event that may occur during meiosis that can result in the loss of the SRY gene on the Y chromosome.**

   Crossing over between the X and Y chromosome during meiosis → can lead to the SRY gene being exchanged between the X and Y chromosome. The Y chromosome will lose the SRY gene while the X chromosome will possess the gene.