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?**

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?**

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

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’

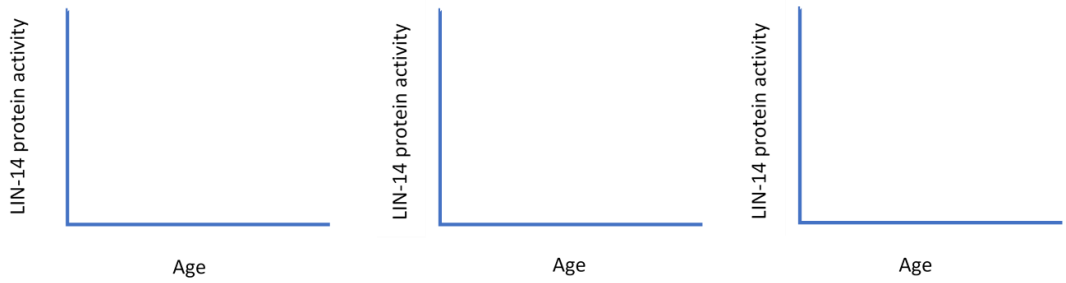
3: The lin-14 gene, which codes for the LIN-14 protein, controls the timing of development in C. elegans. The lin-14 gene is inactive during infancy, and gradually becomes active when C. elegans begins to mature. The lin-14 gene remains highly expressed and active throughout adulthood.

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?**

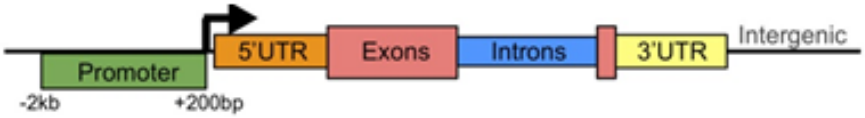
Animals that are homozygous wild-type for the lin-14 gene develop normally. The presence of at least one copy of the mutant allele codes for a defective protein that completely inactivates all LIN-14 activity.

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

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.



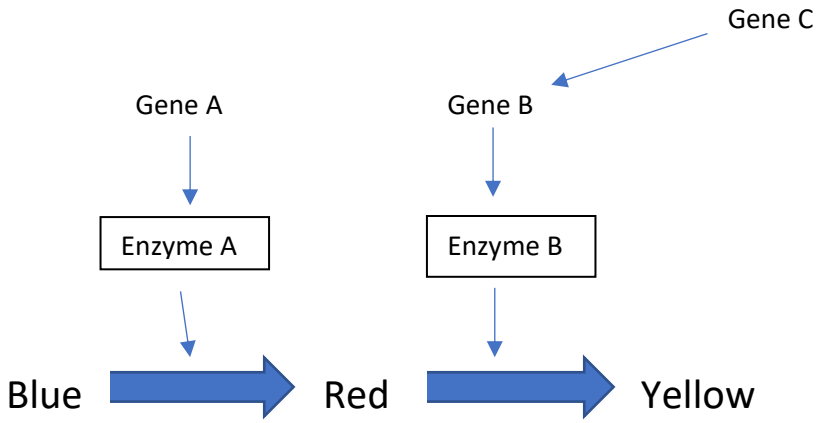
He uses mutagenesis to create introduce a mutation at a random site along the srab-12 gene sequence, creating a mutant organism. The problem is, he does not know which region of the gene contains the mutation. He performs a set of experiments and obtains the following information to help him figure out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

Organism	Quantification of KCN1 mRNA levels	Quantification of KCN1 protein levels	Protein sequence	KCN1 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.**

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.**

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

6: Phenylketonuria is a metabolic disorder caused by a deficiency in the enzyme phenylalanine hydroxylase (PAH). It is an autosomal recessive condition, meaning that individuals who are homozygous for the mutant variant of the gene will be deficient in PAH.

A couple brings their newborn baby to a genetic counselor after observing severe PKU symptoms. Further genetic testing reveals that the baby is homozygous wild-type for the PAH gene. Briefly discuss 2 possible reasons that could explain why the baby has PKU.

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.**

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