

1) A regulator gene produces a repressor for an inducible operon. A geneticist isolates several constitutive mutations affecting this operon, meaning the operon is always transcribed. Where might these constitutive mutations occur? How would the mutations cause the operon to be constitutive?

A inducible operon is normally not being transcribed, meaning that the repressor is active & binding domain of trans-acting repressor is active & binds to the operator, inhibiting transcription. Transcription element changed, no longer able to bind to repressor. Takes place when the inducer binds to the repressor, making it unable to bind to the operator. Constitutive mutations cause transcription to take place at all times, whether the inducer is present or not. Constitutive mutations might occur in the regulator gene, altering the repressor so that it is never able to bind the operator. Alternatively, constitutive mutations might occur in the operator, altering the binding site for the repressor so that the repressor is unable to bind under any conditions.

2) Can you imagine what role RNA stability plays in gene regulation? What controls RNA stability in eukaryotic cells?

The total amount of protein synthesized depends on the amount of mRNA available for translation. Stability of expression

The amount of available mRNA depends on translation

the rates of mRNA synthesis and degradation. Less stable mRNAs degrade faster than stable mRNAs, and so fewer copies of the mRNA are available as templates for translation.

Stability affected by 5' cap & 3' poly(A) tail, 5'UTR, 3'UTR. coding region is an mRNA (secondary and tertiary structure of RNA). Poly(A) binding proteins bind at 3' poly(A) tail.

3) What will be the effect on sexual development in newly fertilized *Drosophila* embryos if the following genes are deleted?

a. *sex lethal (Sxl)*

→ SRY like
If Sxl is off, the embryo develops towards the male phenotype. If it is an XY zygote it will become a normal healthy male. If it is an XX zygote it will die because it's two X chromosomes will be hypertranscribed and too much of X chromosome product result.

b. *transformer (tra)*

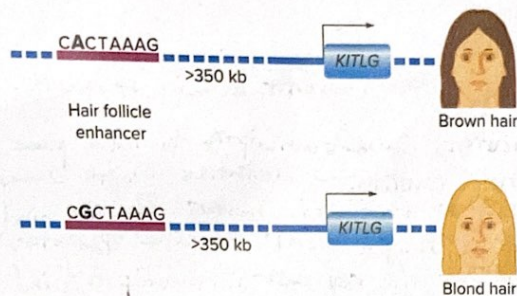
All animals will develop into males. But XX male will be sterile.

c. *male specific lethal 2 (msl-2)*

doublesex: both XX and XY animals survive and will show a development somewhere between female and male (intersex)

These proteins contribute to the stability of the tail and protect the 5' cap through direct interaction. When a critical number of adenine nucleotides have been removed from the tail, the protection is lost and the 5' cap is removed. The removal of the 5' cap enables 5' to 3' nucleases to degrade the mRNA.

4) As shown in the following diagram, a single nucleotide difference in a hair follicle enhancer of a human gene called *KITLG* contributes to the trait of hair color. People with an A-T base pair in the enhancer tend to have dark hair, while people with a G-C base pair at the same position tend to have blond hair. The base pair difference affects the level of *KITLG* transcription: The blond-associated allele is transcribed only 80% as frequently as the dark hair-associated allele. Explain how a single base pair difference in an enhancer sequence can have this effect.



↓
enhancer protein binding capacity
in expression of the pigment

The site of the point mutation is critical for high affinity of the enhancer binding protein (activation protein) to the DNA. If this affinity is reduced then the transcription rate is usually reduced.