

SPEC MODULE 6

6.1.1 - Cellular Control

(a) types of gene mutations and their possible effects on protein production and function. To include substitution, insertion or deletion of one or more nucleotides
AND the possible effects of these gene mutations: (i.e. beneficial, neutral or harmful).

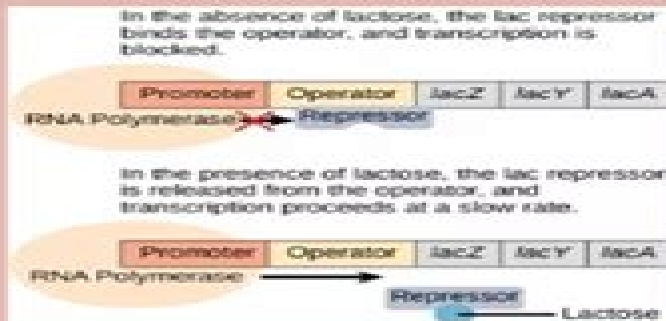
- **Nonsense mutation:** becomes a stop codon instead of coding for an AA
- **Missense mutation:** incorporation of the wrong AA

Mutations: Changes to base nucleotide sequence of DNA

- **Substitution:** 1 or more bases are swapped for another base
- **Deletion:** 1 or more bases are removed → causes frame-shift
- **Insertion:** 1 or more bases are added → causes frame-shift

Effects of mutations:

- **Silent:** no effect because genetic code is degenerate → may code for the same AA (1) sequence of AA is not altered (1)
- **Beneficial:** gives survival adv. (e.g antibiotic resistance)
- **Harmful:** reduces survival adv. (e.g CF)
- E.g insertion/ deletion causes frame-shift (1) → all triplets downstream will be different (1) different sequence of AAs (1) different tertiary structure of protein (1)



(b) the regulatory mechanisms that control gene expression at the transcriptional level, post-transcriptional level and post-translational level.

To include control at the transcriptional level: lac operon, and transcription factors in eukaryotes
 post-transcriptional level: the editing of primary mRNA and the removal of introns to produce mature mRNA
 post-translational level: the activation of proteins by cyclic AMP.

1) TRANSCRIPTIONAL LEVEL

Lac Operon:

3 **structural genes** (lacZ, lacY, lacA) which code for specific enzymes + transcribed onto mRNA

Regulatory gene (lacI) codes for repressor

- 1) **Repressor** binds to the **operator** so that RNA polymerase can't bind to it, so no transcription
- 2) When lactose is present, it binds to the repressor, changing its **shape** (no longer complementary)
- So RNA polymerase can bind to promoter and transcribes
- 3) cAMP binds to cAMP receptor protein (CRP) to increase rate of transcription

Transcription factors (activator/ repressor) start or stop transcription

Regulatory genes code for TFs + controls expression of structural Gs

2) POST-TRANSCRIPTIONAL LEVEL

- After transcription, mRNA is edited
- **Introns:** DON'T code for amino acids
- **Exons:** DO code for amino acids

How gene expression is regulated after transcription. (3)

- [Transcription creates the **primary mRNA** with introns and exons]
- **Primary mRNA** is modified (1) **Splicing:** introns are removed are to form mature mRNA (1)
- [Mature mRNA leaves the nucleus for translation]
- [mRNA may be edited by deletion/ insertion/ substitution → so range of proteins can be produced from a single mRNA]
- **Alternative splicing** produces different versions of mRNA (1)

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