

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).

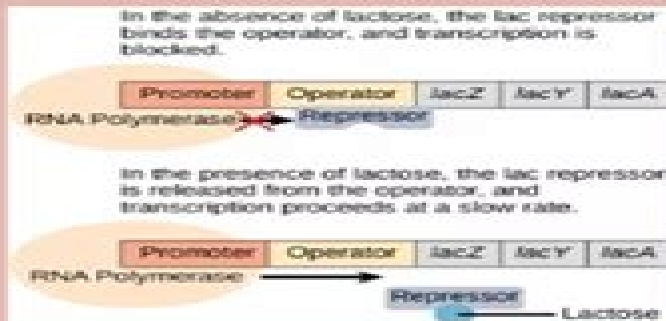
- **Nonsense mutation:** becomes a stop codon instead of coding for an AA
- **Missense mutation:** incorporation of the wrong AA

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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Table of Contents Ocr Biology Mark Schemes Jan 2013

1. Understanding the eBook Ocr Biology Mark Schemes Jan 2013
 - The Rise of Digital Reading Ocr Biology Mark Schemes Jan 2013
 - Advantages of eBooks Over Traditional Books
2. Identifying Ocr Biology Mark Schemes Jan 2013
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Ocr Biology Mark Schemes Jan 2013
 - User-Friendly Interface
4. Exploring eBook Recommendations from Ocr Biology Mark Schemes Jan 2013
 - Personalized Recommendations
 - Ocr Biology Mark Schemes Jan 2013 User Reviews and Ratings
 - Ocr Biology Mark Schemes Jan 2013 and Bestseller Lists

5. Accessing Ocr Biology Mark Schemes Jan 2013 Free and Paid eBooks
 - Ocr Biology Mark Schemes Jan 2013 Public Domain eBooks
 - Ocr Biology Mark Schemes Jan 2013 eBook Subscription Services
 - Ocr Biology Mark Schemes Jan 2013 Budget-Friendly Options
6. Navigating Ocr Biology Mark Schemes Jan 2013 eBook Formats
 - ePub, PDF, MOBI, and More
 - Ocr Biology Mark Schemes Jan 2013 Compatibility with Devices
 - Ocr Biology Mark Schemes Jan 2013 Enhanced eBook Features
7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Ocr Biology Mark Schemes Jan 2013
 - Highlighting and Note-Taking Ocr Biology Mark Schemes Jan 2013
 - Interactive Elements Ocr Biology Mark Schemes Jan 2013
8. Staying Engaged with Ocr Biology Mark Schemes Jan 2013
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Ocr Biology Mark Schemes Jan 2013
9. Balancing eBooks and Physical Books Ocr Biology Mark Schemes Jan 2013
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Ocr Biology Mark Schemes Jan 2013
10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine Ocr Biology Mark Schemes Jan 2013
 - Setting Reading Goals Ocr Biology Mark Schemes Jan 2013
 - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of Ocr Biology Mark Schemes Jan 2013
 - Fact-Checking eBook Content of Ocr Biology Mark Schemes Jan 2013
 - Distinguishing Credible Sources
13. Promoting Lifelong Learning

- Utilizing eBooks for Skill Development
- Exploring Educational eBooks

14. Embracing eBook Trends

- Integration of Multimedia Elements
- Interactive and Gamified eBooks

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