Gene structure

Gene Structure

Introduction

- A gene is a segment of DNA that carries the information necessary to produce a functional product — usually a protein, but sometimes a functional RNA such as tRNA or rRNA.
- The structure of genes is organized in such a way that ensures proper expression, regulation, and protein synthesis.
- Additionally, the genetic code is the universal system that defines how the information stored in a gene (in the form of nucleotide sequences) is translated into a sequence of amino acids in a protein.
- This code has been elucidated through landmark experiments in molecular biology.
- **To understand how genes work, we need to examine:**
    1. The structure and organization of genes in both eukaryotes and prokaryotes
    2. The organization of the genetic code
    3. The elucidation (discovery) of how this code functions

Gene Structure in Eukaryotes

- Eukaryotic genes (in organisms like animals, plants, fungi) are complex and include both coding and non-coding regions.
- These components regulate when and how genes are expressed and how the RNA is processed into a protein.

A. Regulatory Elements

- Before the coding sequence begins, there are regulatory DNA sequences that control whether the gene is turned on or off, and how strongly it is expressed.
- **Promoters**: Located immediately upstream of the gene, promoters serve as the binding site for **RNA polymerase** — the enzyme that synthesizes RNA.
- A common motif is the TATA box, which helps RNA polymerase bind at the correct site.
- **Enhancers and Silencers**: These elements can be located far upstream or downstream of the gene.
    - **Enhancers** bind transcription factors that **increase gene expression**.
    - **Silencers** bind repressors that **decrease or suppress expression**.
- These regions do not code for proteins but are essential for controlling gene activity.

B. Transcription Start Site (TSS)

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- The TSS is the specific point on DNA where RNA polymerase begins transcribing the gene. It marks the beginning of the RNA transcript.

C. 5' Untranslated Region (5' UTR)

- Located between the TSS and the coding region, the 5' UTR is transcribed into RNA but is not translated into protein.
- It plays roles in:
  - Regulating **translation efficiency**
  - Facilitating **ribosome binding**
  - Controlling **mRNA stability**

D. Coding Region: Exons and Introns

- This region contains the actual instructions to make a protein.
- **Exons**: These are the coding sequences that remain in the mature mRNA after processing and are translated into protein.
- **Introns**: These are non-coding sequences between exons. They are transcribed but removed from the RNA during splicing.
- A key feature of introns is that they allow for alternative splicing, which enables a single gene to produce multiple different proteins depending on which exons are retained.

E. 3' Untranslated Region (3' UTR)

- This region lies downstream of the stop codon and is transcribed but not translated. It influences:
  - **mRNA stability**
  - **mRNA localization**
  - **Translational efficiency**

F. Termination Signal and Polyadenylation Site

- At the end of the gene, there are termination sequences that stop transcription. Following this:
  - A **polyadenylation signal** (e.g., AAUAAA) is transcribed.
  - This leads to the addition of a **poly-A tail** to the mRNA.
- The poly-A tail protects the mRNA from degradation and helps with:
  - **Nuclear export**
  - **Stability**
  - **Translation initiation**

Gene Structure in Prokaryotes

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Prokaryotic organisms (such as bacteria) have a simpler gene structure, suited for their fast growth and limited cellular complexity.

A. No Introns

- Most prokaryotic genes are continuous, meaning they do not contain introns.
- The gene is transcribed into a single, uninterrupted RNA molecule that is ready for translation.

B. Operons

- Prokaryotic genes are often arranged in operons — groups of related genes controlled by a single promoter and transcribed into a single polycistronic mRNA (an mRNA that encodes multiple proteins).
- **Example:** The lac operon in *E. coli* controls the breakdown of lactose using several enzymes, all coded by genes transcribed together.

C. Promoters and Regulatory Regions

- Prokaryotic promoters include specific sequences (e.g., -35 and -10 regions, like the Pribnow box) that guide RNA polymerase to start transcription.
- Regulatory proteins can bind to operators near the promoter to turn genes on or off.

D. Coupled Transcription and Translation

- In prokaryotes, transcription and translation happen simultaneously because there is no nuclear membrane. As the mRNA is being synthesized, ribosomes immediately begin translating it into protein.

Organization of the Genetic Code

- The genetic code is the set of rules that translates the sequence of nucleotides in mRNA into a sequence of amino acids in a protein.

Key Features of the Genetic Code:

- It is a **triplet code**: Each codon is made of 3 nucleotides.
- There are **64 possible codons** ($4^{43} = 64$).
  - **61 codons** code for amino acids.
  - **3 codons** are **stop signals**: UAA, UAG, UGA.
- **The code is degenerate:** Most amino acids are coded by more than one codon (e.g., leucine has 6 codons).
- **It is non-overlapping and comma-less:** Codons are read one after another, without gaps or overlap.
- **It is almost universal:** The same codons specify the same amino acids in nearly all organisms.
- It includes a start codon (AUG) which codes for methionine and signals the start of translation.

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Elucidation of the Genetic Code

The genetic code is the set of rules by which nucleotide sequences are translated into amino acids in proteins.

A. Codons

- A codon is a sequence of three nucleotides in mRNA that specifies an amino acid.
- Example: AUG = methionine; UUU = phenylalanine.
- There are 64 possible codons (4^3 combinations of A, U, G, C).
  - 61 code for amino acids.
  - **3 are stop codons:** UAA, UAG, UGA.

B. Key Features of the Genetic Code

- **Triplet nature**: Each amino acid is encoded by a triplet of nucleotides.
- **Degeneracy**: Multiple codons can code for the same amino acid.
  - Example: Leucine has six codons.
- **Unambiguous**: Each codon codes for only one amino acid.
- **Universal (mostly)**: The code is nearly universal across organisms.
- **Non-overlapping**: Codons are read in order without overlapping.
- **Comma-less**: No punctuation between codons.

C. Start and Stop Codons

- **Start codon**: AUG (methionine).
  - In prokaryotes, codes for formyl-methionine (fMet).
- **Stop codons**:
  - UAA: ochre.
  - UAG: amber.
  - UGA: opal.
  - Signal the end of translation.

D. tRNA and Anticodons

- tRNA molecules have anticodons that pair with codons on mRNA.
- Each tRNA carries a specific amino acid.
- Example:
  - mRNA codon: AUG.
  - tRNA anticodon: UAC.
  - Amino acid: Methionine.

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E. Wobble Hypothesis

- Some tRNAs can pair with more than one codon due to flexibility at the third base position.
- This explains the degeneracy of the code.

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