# Lecture 1
# Advanced physiological genetics

## Gene Structure:

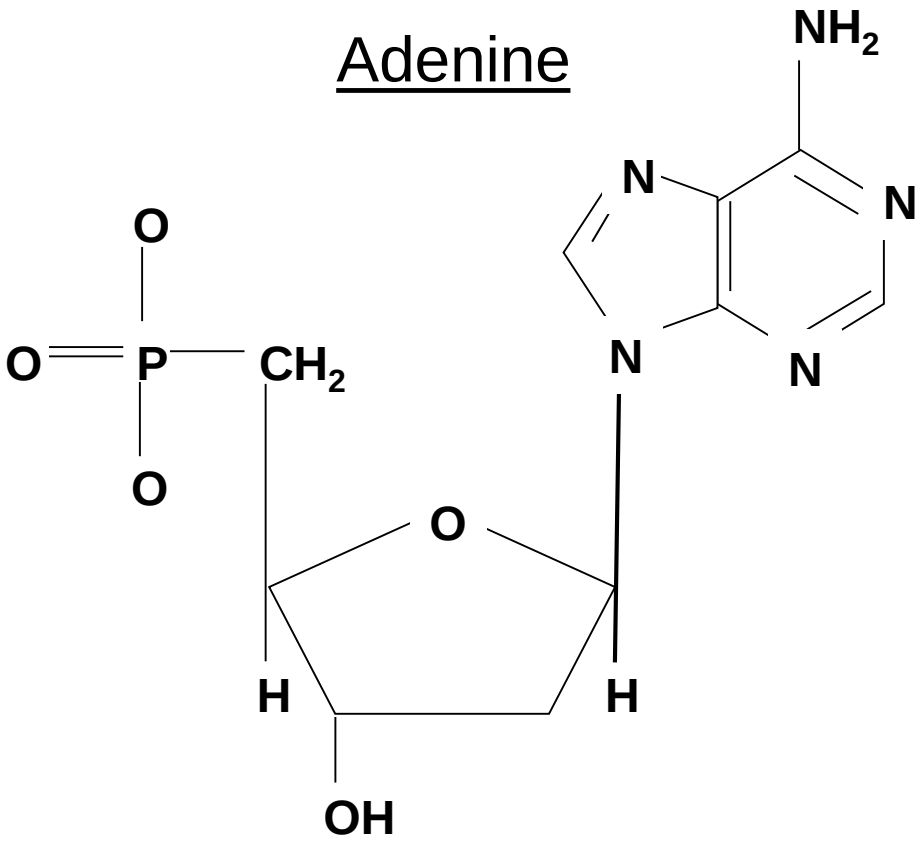
# Nucleic Acids

- Sequence of Nucleotides
- Nucleotide composed of:
  - Nitrogenous Base
    - Purine
    - Pyrimidine
  - Sugar
    - Ribose
    - Deoxyribose
  - Phosphate

# DNA

- Deoxyribonucleic Acid
- 4 Bases
  - Purines
    - Adenine
    - Guanine
  - Pyrimidines
    - Cytosine
    - Thymine*
- Sugar is Deoxyribose

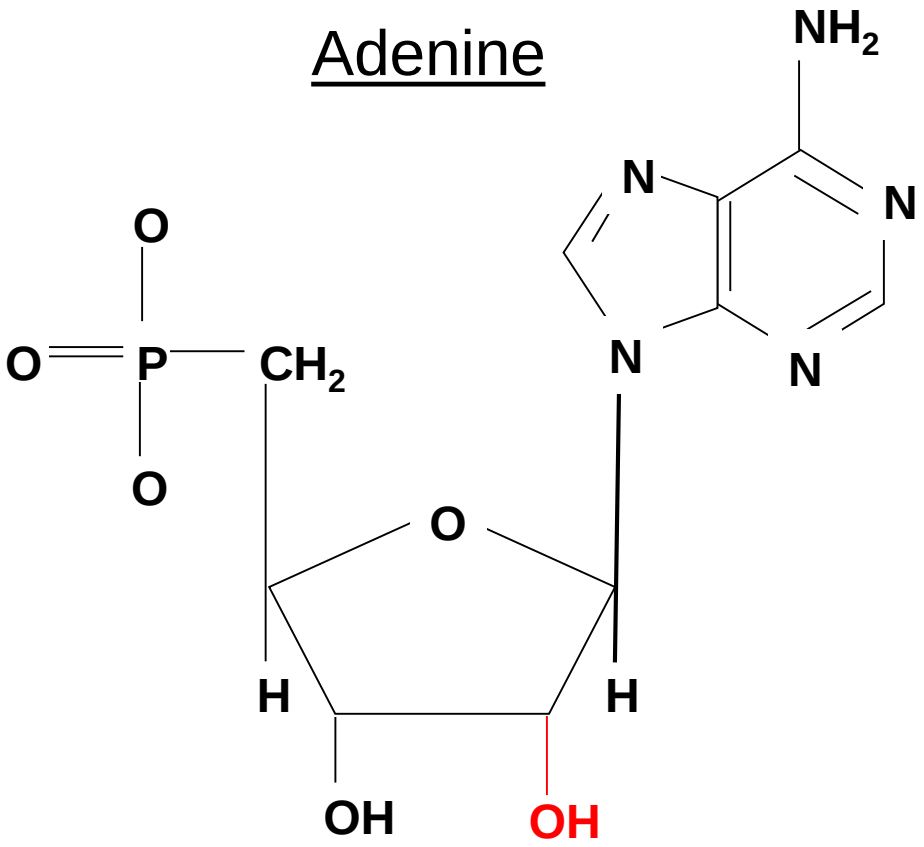
Adenine



# RNA

- Ribonucleic Acid
- 4 Nucleotides
  - Purine
    - Adenine
    - Guanine
  - Pyrimidines
    - Cytocine
    - Uracil*
- Sugar is Ribose

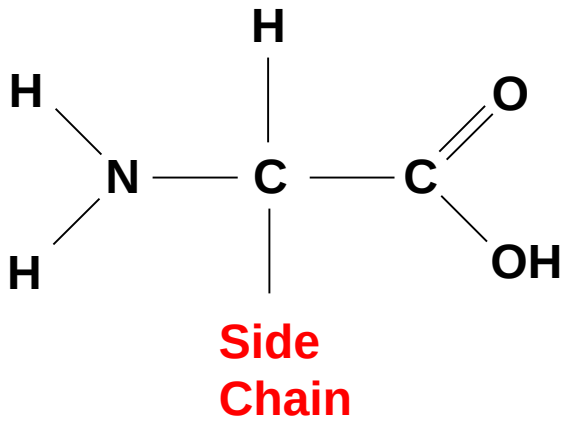
Adenine



# Proteins

- Polymer made of monomers – Amino Acids
- 20 Naturally occurring Amino Acids
- Grouped by Side Chain:
  - Hydophobic
  - Hydrophilic
    - Acidic
    - Basic

**Amino Acid**



# Proteins

- Special cellular components called ribosomes use the triplet genetic code to translate the nucleotides of a mRNA sequence into the amino acid sequence of a protein.

- There are 20 different amino acids. Proteins are created by linking amino acids together in a linear fashion to form polypeptide chains.

- Protein polypeptide chains fold into three-dimensional structures that can associate with other protein structures to perform specific functions.

# Central dogma of molecular biology:
# DNA ➔ RNA ➔ Protein

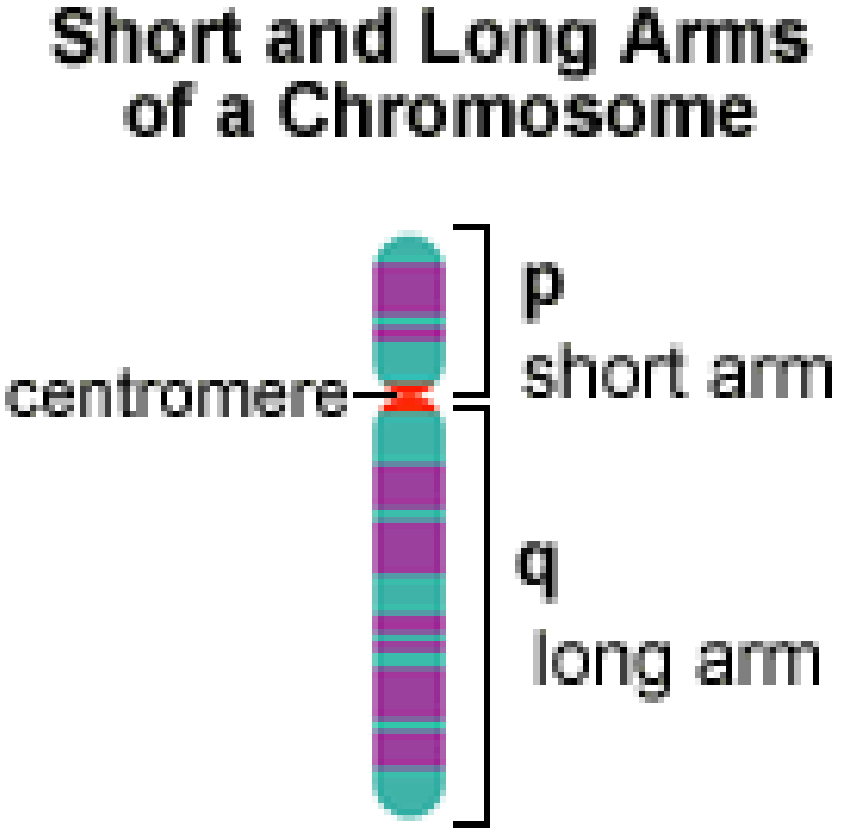
1. Genetic information is stored in DNA.
2. Segments of DNA that encode proteins or other functional products are called genes.
3. Gene sequences are transcribed into messenger RNA intermediates (mRNA).
4. mRNA intermediates are translated into proteins that perform most life functions.

# Genes

- Genes are the basic physical and functional units of heredity. Each gene is located on a particular region of a chromosome and has a specific ordered sequence of nucleotides (the building blocks of DNA).

# What is a locus?

- A locus describes the region of a chromosome where a gene is located. **11p15.5** is the locus for the **human insulin gene**. **11** is the chromosome number, **p** indicates the short arm of the chromosome, and **15.5** is the number assigned to a particular region on a chromosome. When chromosomes are stained in the lab, light and dark bands appear, and each band is numbered. The higher the number, the farther away the band is from the centromere.

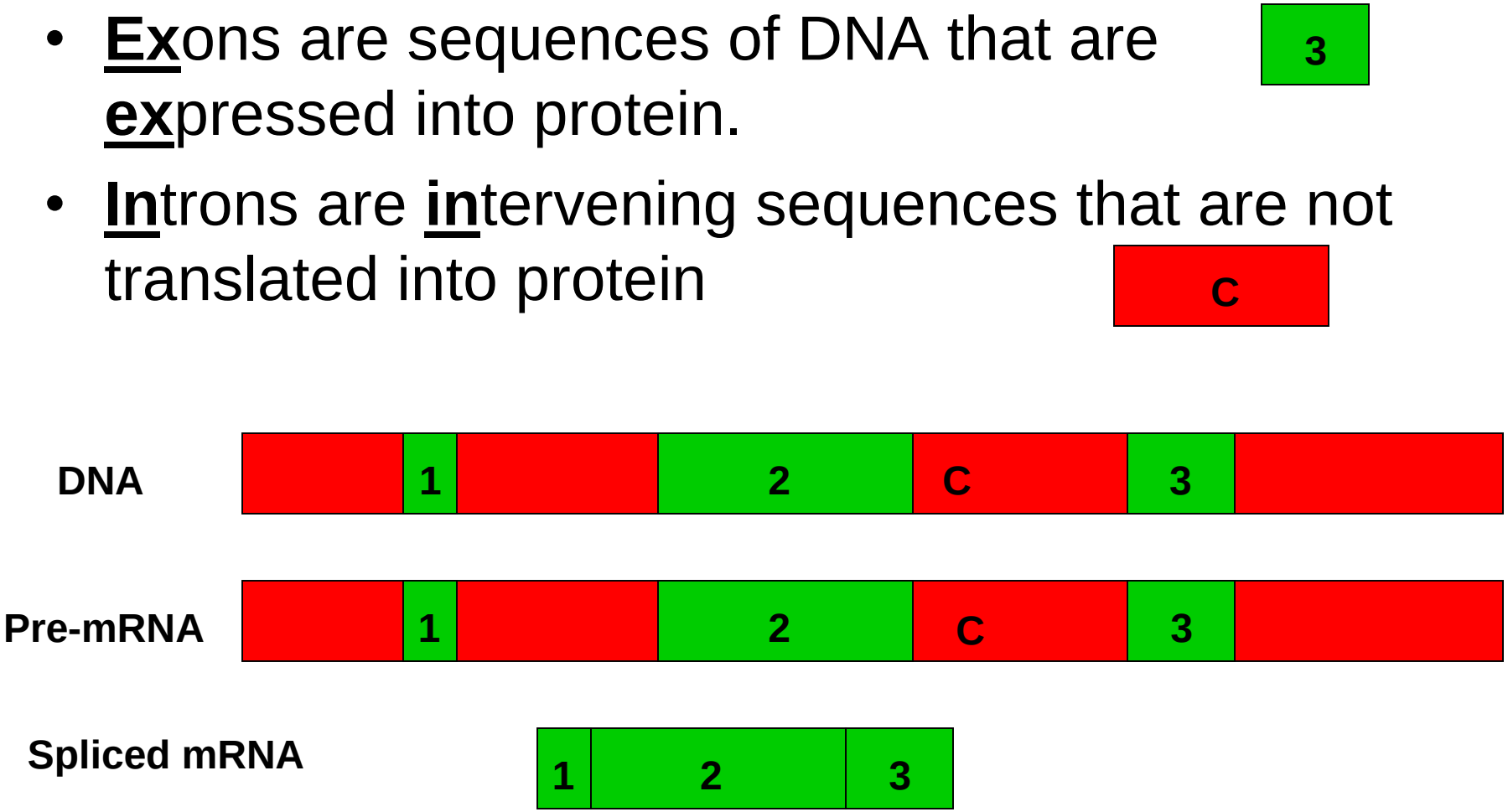


# Exons vs Introns

- Eukaryotic genes have introns and exons. Exons contain nucleotides that are translated into amino acids of proteins. Exons are separated from one another by intervening segments of junk DNA called introns. Introns do not code for protein. They are removed when eukaryotic mRNA is processed. Exons make up those segments of mRNA that are spliced back together after the introns are removed; the intron-free mRNA is used as a template to make proteins.

# Splicing

- **Ex**ons are sequences of DNA that are **ex**pressed into protein.
- **In**trons are **in**tervening sequences that are not translated into protein



# Exons and Coding

**What's the difference between exons and coding sequence?**

Exons often are described as short segments of protein coding sequence. This is a bit of an oversimplification. Exons are those segments of sequence that are spliced together after the introns have been removed from the pre-mRNA. Yes, the coding sequence is contained in exons, but it is possible for some exons to contain no coding sequence. Portions of exons or even entire exons may contain sequence that is not translated into amino acids. These are the untranslated regions or UTRs. UTRs are found upstream and downstream of the protein-coding sequence.