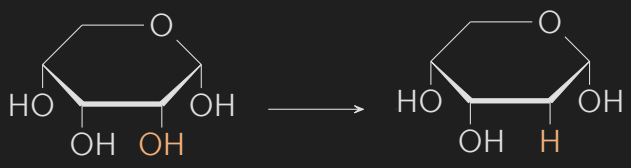
Human Genetics

DNA Structure and Function

This chapter was mostly basic review material. The portion on DNA was more of a test of my recent changed document class settings. The majority of the chapter was omitted. I might add more review content later if I find necessary.

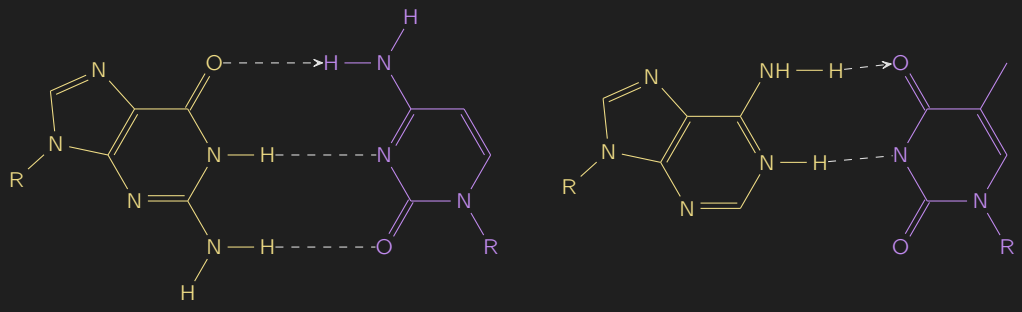
Deoxyribonucleic Acid

- **Deoxyribonucleic Acid (DNA)**: a double helix containing two polynucleotide chains that carries the genetic instructions for all known organisms and many viruses.
  - The bases are made of four bases:
    - The purine derivatives: adenine (A) and guanine (G).
    - The pyrimidine derivatives: thymine (T) and cytosine (C).
  - The backbone is made of alternating deoxyribose molecules (a ribose missing its 2' oxygen) connected to phosphodiester bonds from 5' → 3' positions—forming two antiparallel strands.



Ribose Deoxyribose

- Number of adenines = thymines. (A-T)
- Number of guanines = cytosines. (C-G)
  - Bonds between bases are noncovalent (no electron sharing, weak).
  - C—G pairs form three hydrogen bonds, while A—T forms two; making G—C slightly more stable.



Guanine Cytosine Adenine Thymine

Genetic Variation

Much of this chapter was once again mostly review. However, the mini-section on nomenclature was new to me and seemed useful, so I included this. This chapter provided several hooks for related topics, so I may add more content from elsewhere in this chapter if I find my understanding on basic material lacking.

Mutation Nomenclature

- **Level of mutational change**:
  - g = Genomic
  - c = Coding sequence
  - m = Mitochondrial sequence
  - r = RNA sequence
  - p = Protein sequence
- **Type of mutational change**:
  - > = Substitution in the DNA
  - _ = A range of affected bases
  - del = Deletion
  - dup = Duplication
  - ins = Insertion
  - inv = Inversion
- E+I = The last nucleotide of preceding exon (E) for genomic mutations at the 5' (+) and number of nucleotides into the intron (I).
- E-I = The first nucleotide of the next exon (E) for mutations at the 3' end of an intron (-) and the number of nucleotides into the intron (I).

Examples

- g.1346A>C: Change of A to C at position 1346 in the genomic DNA sequence.
- c.745delT: Deletion of T at position 745 in the coding sequence.
- g.1567_1568delAT: Deletion of AT at positions 1567-1568 in the genomic DNA sequence.
- c.145+1T: Change of splice donor (first position of intron after base 145 of preceding exon) to T
- p.Arg54Gly: Change of arginine at codon 54 to glycine.