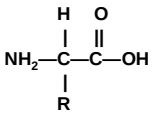
AMINO ACIDS

amine group –NH_2
carboxyl group –COOH
alpha carbon
side chain



levo- and dextro-rotatory
L-isomers

AMINO ACIDS (2)

aromatic: contains benzine
aliphatic: benzineless

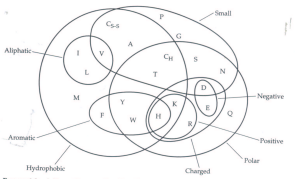


FIGURE 1.9 A Venn diagram showing the division of the 20 primary amino acids into overlapping categories according to size, structure of the side chain, polarity, charge, and hydrophobicity. Note the placement of cysteine in two places, as reduced cysteine (C_H) and as cystine (C_{S-S}). See Table 1.2 for the one-letter abbreviations of the amino acids. Modified from Taylor (1986).

PROTEINS

peptide bonds

amino or N terminus
carboxyl or C terminus

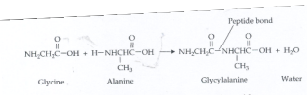


FIGURE 1.10 Formation of a dipeptide from two amino acids.

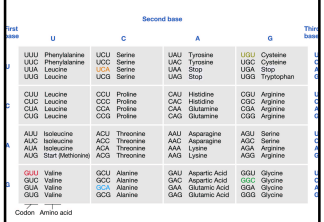
PROTEINS (2)

Structure

- **primary**
- **secondary**
  - **α helix**
  - **β-pleated sheet**
- **tertiary**
- **quaternary**

TRANSLATION

- **transfer RNA (tRNA)**
- **codons, anticodons**
- **start and stop codons**
- **reading frames**
- **standard genetic code**
- **sense codons**
- **synonymous codons**
- **codon family**



MUTATION

- replication or repair errors
- length
  - point & segment
- change
  - substitution
  - recombination
  - deletions
  - insertions
  - inversions

FIGURE 1.11 Types of mutations. (a) Original sequence. (b) Transition from C to T. (c) Transversion from G to C. (d) Recombination, the exchange of the sequence GTCTT by CAAAC. (e) Deletion of the sequence ACCTA. (f) Insertion of the sequence AAAGC. (g) Inversion of 5′—GCAAAC—3′ to 5′—GTTTGC—3′.

SUBSTITUTION

- transition & transversion
  - 4 & 8
- non- & synonymous
- silent
- missense & nonsense

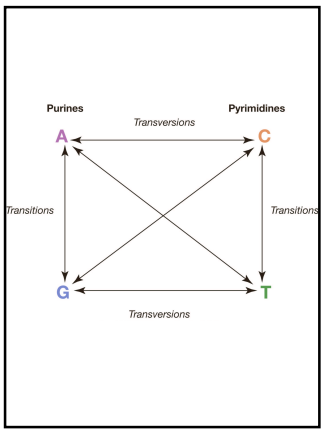


TABLE 1.5 Relative frequencies of different types of mutational substitutions in a random protein-coding sequence

Substitution	Number	Percent
Total in all codons	549	100
Synonymous	134	25
Nonsynonymous	415	75
Missense	392	71
Nonsense	23	4
Total in first codons	183	100
Synonymous	8	4
Nonsynonymous	175	96
Missense	166	91
Nonsense	9	5
Total in second codons	183	100
Synonymous	0	0
Nonsynonymous	183	100
Missense	176	96
Nonsense	7	4
Total in third codons	183	100
Synonymous	126	69
Nonsynonymous	57	31
Missense	50	27
Nonsense	7	4

RECOMBINATION

crossing over
(reciprocal)

gene conversion
(nonreciprocal)

generalised & site-specific

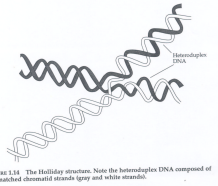


FIGURE 1.14 The Holliday structure. Note the heteroduplex DNA composed of mismatched chromatid strands (gray and white strands).

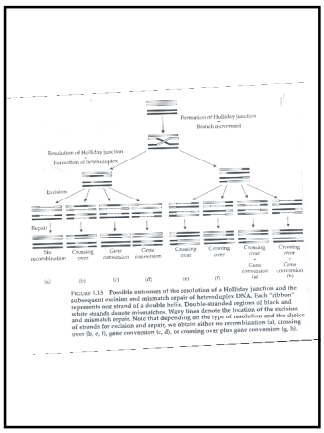


FIGURE 1.15 Possible outcomes of the resolution of a Holliday junction and the subsequent excision and mismatch repair of heteroduplex DNA. Each "ribbon" represents one strand of a double helix. Dash-bounded regions of black and white strands denote mismatches. Wavy lines denote the location of the excision and mismatch repair. Note that depending on the type of resolution and the choice of strands for excision and repair, we obtain either no recombination (a), crossing over (b, c, f), gene conversion (c, d), or crossing over plus gene conversion (g, h).

INSERTION/DELETION

indels

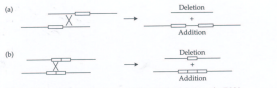


FIGURE 1.16 (a) Unequal crossing over resulting in the deletion of a DNA sequence in one of the daughter strands and the duplication of the same sequence in the other strand. (b) When a DNA segment is duplicated in tandem, the chance of misalignment increases, as does the chance of unequal crossing over. A box denotes a particular stretch of DNA. Modified from Li (1997).

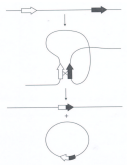
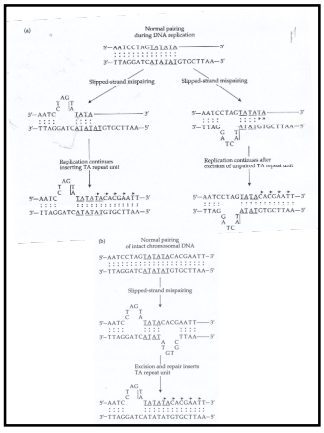


FIGURE 1.17 Generation of a deletion by the intrastrand deletion process. The repeated sequences (arrows) that are oriented in the same direction recombine (c) to produce a genomic deletion (left) and an extrachromosomal element (right).



INVERSION

chromosome breakage,rejoining

intrachromosomal crossing over

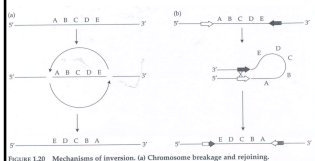


FIGURE 1.20 Mechanisms of inversion. (a) Chromosome breakage and rejoining. (b) Crossing over (×) between homologous segments (arrows) on the same chromosome that are oriented in opposite directions results in an inversion involving the DNA sequence between the homologous inverted repeats.

MUTATION RATE

replication or repair errors

length
- **point & segment**

change
- **substitution**
- **recombination**
- **deletions**
- **insertions**
- **inversions**