

**BENG 100 Frontiers of Biomedical Engineering**  
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**Chapter 3**

**SUMMARY**

- Nucleic acids are linear polymers made up of monomer units called nucleotides. Each nucleotide is composed of a pentose sugar, and phosphate group, and a nitrogenous base. DNA nucleotides have deoxyribose sugars whereas RNA nucleotides have ribose sugars.
- DNA is the main information storage molecule of the cell. Information is stored in the form of a nucleotide base sequence. A gene is a sequence of DNA bases that code for RNA or a polypeptide chain.
- DNA is a double helix with two sugar phosphate backbones and nucleotide bases bridging the two chains.
- Base-pairing is accomplished by hydrogen bonding between purines and pyrimidines. Watson-Crick base-pairing dictates the base pairs in DNA are: A-T, C-G. In RNA, the thymine is replaced by a uracil, so base pairs in RNA are: A-U, C-G.
- DNA replication occurs every time a cell divides. DNA replication is semiconservative with each daughter cell receiving one parental strand and one newly synthesized strand. DNA polymerization only occurs in the 5' to 3' direction.
- Errors in replication or environmental factors can give rise to mutations.
- The central dogma of molecular biology dictates that DNA must first be transcribed into RNA, which is then translated into protein.
- Genes are transcribed into RNA by RNA polymerase. In eukaryotic cells, transcribed RNA must undergo processing to form an mRNA transcript.
- mRNA is translated into amino acids using the genetic code. Ribosomes and tRNA are involved in translation of codons to corresponding amino acids. The newly synthesized polymer of amino acids makes up a polypeptide chain. Proteins can be composed of more than one polypeptide chain.
- Recombinant DNA technology enables researchers to manipulate DNA for various purposes. Genes can be made to express in other species.
- PCR is used to amplify short segments of DNA.

- Plasmids and viruses are used as cloning vectors and expression vectors.
- Both non-viral and viral methods can be used to introduce a foreign gene into eukaryotic cells.

## KEY CONCEPTS AND DEFINITIONS

adenovirus - a group of DNA containing viruses, which cause respiratory disease, including one form of the common cold. Adenoviruses can also be genetically modified and used in gene therapy to treat cystic fibrosis, cancer, and potentially other diseases.

allele - One of two or more alternative forms of a gene located at the corresponding site (locus) on homologous chromosomes. Different alleles produce variation in inherited characteristics such as hair color or blood type. In an individual, one form of the allele (the dominant one) may be expressed more than another form (the recessive one).

Antibiotic selection – A method used to ascertain whether a particular plasmid has been successfully integrated into the DNA sequence of a bacterial cell.

Antiparallel – arranged in an opposite but parallel manner.

antisense RNA - RNA, with sequence complementary to a specific RNA transcript or mRNA, whose binding prevents processing of the transcript or translation of the mRNA

anneal – similar to hybridize, to bring to complementary strands of nucleic acids together.

bioinformatics - the science of managing and analyzing biological data using advanced computing techniques. Especially important in analyzing genomic research data.

bioremediation - the use of biological organisms such as plants or microbes to aid in removing hazardous substances from an area.

camptothecin - a chemotherapeutic agent that can inhibit protein synthesis by inhibiting the action of topoisomerase.

carcinogen – a substance capable of causing cancer in living tissues

Character – An feature that is transmissible from parent to offspring

clone – to create an exact copy made of biological material such as a DNA segment (e.g., a gene or other region), a whole cell, or a complete organism

cloning vector - DNA molecule originating from a virus, a plasmid, or the cell of a higher organism into which another DNA fragment of appropriate size can be integrated without loss of the vector's capacity for self-replication; vectors introduce foreign DNA into host

cells, where the DNA can be reproduced in large quantities

codon - sequence of three nucleotides in DNA or mRNA that specifies a particular amino acid during protein synthesis; also called *triplet*. Of the 64 possible codons, three are stop codons, which do not specify amino acids complementary DNA (cDNA) - DNA that is synthesized in the laboratory from a messenger RNA (mRNA) template

chromatin – a complex of DNA and proteins called histones

chromosome - the self-replicating genetic structure of cells containing the cellular DNA that bears in its nucleotide sequence the linear array of genes. In prokaryotes, chromosomal DNA is circular, and the entire genome is carried on one chromosome. Eukaryotic genomes consist of a number of chromosomes whose DNA is associated with different kinds of proteins.

daughter cells – two cells resulting from division of a single parental cell

denaturation - alteration in the conformation of a protein or nucleic acid due to disruption of various non-covalent bonds caused by heating or exposure to certain chemicals; usually results in loss of biological function

deoxyribonucleic acid – long linear polymer, composed of four kinds of deoxyribose nucleotides, that is the carrier of genetic information. In its native state, DNA is a double helix of two antiparallel strands held together by hydrogen bonds between complementary purine and pyrimidine bases

DNA binding proteins – protein molecules that help to stabilize the single-stranded DNA during DNA replication

DNA ligase - an enzyme that links together the 3' end of one DNA strand with the 5' end of another forming a continuous strand.

DNA polymerase - an enzyme that copies one strand of DNA (the template strand) to make the complementary strand, forming a new double-stranded DNA molecule. All DNA polymerases add deoxyribonucleotides one at a time in the 5' to 3' direction to a short pre-existing primer strand of DNA or RNA.

diploid - a full set of genetic material consisting of paired chromosomes, one from each parental set. Most animal cells except the gametes have a diploid set of chromosomes. The diploid human genome has 46 chromosomes

dominant - an allele that is almost always expressed, even if only one copy is present.

encode – in general use: to put into code; in genetics: to specify the genetic code for

exon - the protein-coding DNA sequence of a gene

expression vector - a modified plasmid or virus that carries a gene or cDNA into a suitable host cell and there directs synthesis of the encoded protein. Some expression vectors are designed for screening DNA libraries for a gene of interest; others, for producing large amounts of a protein from its cloned gene

gamete - mature male or female reproductive cell (sperm or ovum) with a haploid set of chromosomes (23 for humans).

gene - the fundamental physical and functional unit of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome that encodes a specific functional product (i.e., a protein or RNA molecule).

genetics - the study of inheritance patterns of specific traits

genetic code – the nucleotide triplets of DNA and RNA molecules, which carry genetic information in living tissue

genetic expression – the entire sequence that occurs in converting a DNA sequence into a protein genetically modified organism – plant or animal that has been modified using recombinant DNA technology.

genome – A complete set of genes or genetic material present in an organism's chromosome.

genotype - entire genetic constitution of an individual cell or organism

germ cell - sperm and egg cells and their precursors. Germ cells are haploid and have only one set of chromosomes (23 in all), while most other cells in humans have two copies (46 in all).

glycosylation - the addition of carbohydrates to proteins

haploid - a single set of chromosomes (half the full set of genetic material) present in the egg and sperm cells of animals and in the egg and pollen cells of plants. Human beings have 23 chromosomes in their reproductive cells

helicase - an enzyme that moves along a DNA duplex using the energy released by ATP hydrolysis to separate (unwind) the two strands. Required for the replication and transcription of DNA.

Heredity – the genetic transmission of characteristics from parent to offspring

heterozygous - referring to a diploid cell or organism having two different alleles of a particular gene

histone – A group of proteins packaged with DNA to form chromatin that play a role in

gene regulation

homologous – corresponding or similar in structure (or function)

homozygous - referring to a diploid cell or organism having two identical alleles of a particular gene

intron - DNA sequence that interrupts the protein-coding sequence of a gene; an intron is transcribed into RNA but is cut out of the message before it is translated into protein.

Incomplete dominance – A phenomenon whereby the presence of multiple alleles in a heterozygote leads to a trait that appears intermediate to the homozygous phenotypes.

karyotype - a photomicrograph of an individual's chromosomes arranged in a standard format showing the number, size, and shape of each chromosome type; used in low-resolution physical mapping to correlate gross chromosomal abnormalities with the characteristics of specific diseases.

knockout - inactivation of specific genes. Knockouts are often created in laboratory organisms such as yeast or mice so that scientists can study the knockout organism as a model for a particular disease.

Lagging strand – the DNA template of a double stranded DNA molecule opposite the leading strand that codes in the direction 5' to 3'

Leading strand – the DNA template of a double stranded DNA molecule opposite the lagging strand that codes in the direction 3' to 5'

liposome - spherical phospholipid bilayer structure with an aqueous interior that forms in vitro from phospholipids and may contain protein.

locus - the specific site of a gene on a chromosome. All the alleles of a particular gene occupy the same locus

meiosis - the process of two consecutive cell divisions in the diploid progenitors of sex cells. Meiosis results in four rather than two daughter cells, each with a haploid set of chromosomes.

messenger RNA – RNA that serves as a template for protein synthesis.

mitosis – the process of nuclear division in cells that produces daughter cells that are genetically identical to each other and to the parent cell.

mutagenesis – development of a mutation (change in DNA base)

mutant – individual, organism, or new genetic character arising or resulting from

mutation.

mutation – alteration or change in DNA base(s)

N-myristolation – the addition of fatty acids to proteins

nucleic acid hybridization - the process of joining two complementary strands of DNA or one each of DNA and RNA to form a double-stranded molecule.

nulliploid – devoid of a nucleus or nuclei

Okazaki fragments – short DNA fragments that are joined to form the lagging strand of DNA

oligonucleotide - a molecule usually composed of 25 or fewer nucleotides; used as a DNA synthesis primer

phenotype – the physical characteristics of an organism or the presence of a disease that may or may not be genetic

phosphodiester bond – a bond between the 5′-phosphate of one nucleotide and the 3′-hydroxyl of another

plasmid - autonomously replicating extra-chromosomal circular DNA molecules, distinct from the normal bacterial genome and nonessential for cell survival under nonselective conditions. Some plasmids are capable of integrating into the host genome. A number of artificially constructed plasmids are used as cloning vectors.

pleiotropy - one gene that causes many different physical traits such as multiple disease symptoms

polygenic – determined by more than one gene

polyploid - having multiple chromosome sets as a result of a genetic event that is abnormal (e.g., constitutional or mosaic triploidy, tetraploidy, etc.) or programmed (e.g. some plants and certain human body cells are naturally polyploid).

polymerase chain reaction (PCR) – a method for amplifying a region of DNA by repeated cycles of DNA synthesis *in vitro*.

post-translational modification – chemical change in the polypeptide chain after translation.

prenylation – the addition of lipids to proteins.

primer - short preexisting polynucleotide chain to which new deoxyribonucleotides can

be added by DNA polymerase.

promoter - a DNA site to which RNA polymerase will bind and initiate transcription.

proteolysis – the directed degradation of proteins or peptides by the actions of enzymes.

recessive gene - a gene which will be expressed only if there are 2 identical copies or, for a male, if one copy is present on the X chromosome

recombinant DNA technology – procedure used to join together DNA segments in a cellfree system (an environment outside a cell or organism). Under appropriate conditions, a recombinant DNA molecule can enter a cell and replicate there, either autonomously or after it has become integrated into a cellular chromosome.

Renaturation - The process by which proteins or complementary strands of nucleic acids re-form their native conformations.

replication origin – a site for the initiation of DNA replication

replication fork – a structure that forms when two parental DNA strands separates

restriction endonuclease - An enzyme that cleaves DNA at a specific sequence

retrovirus – a type of virus that contains RNA as its genetic material. The RNA of the virus is translated into DNA, which inserts itself into an infected cell's own DNA. Retroviruses can cause many diseases, including some cancers and AIDS

reverse transcriptase - an enzyme used by retroviruses to form a complementary DNA sequence (cDNA) from their RNA. The resulting DNA is then inserted into the chromosome of the host cell.

ribonucleic acid (RNA) – a chemical similar to a single strand of DNA. In RNA, the base uracil (U) is substituted for thymine (T). RNA delivers DNA's genetic message to the cytoplasm of a cell where proteins are made.

ribosome – A small molecule containing RNA and associated proteins, present within the cytoplasm of a living cells are is involved in protein synthesis by binding to mRNA and tRNA.

ribosomal RNA – a multi-component system of RNA synthesized in the nucleolus of cell and constitutes the central component of ribosome

RNA interference (RNAi) – short RNA sequences that are complementary to mRNA sequences and may interfere with translation.

RNA polymerase - an enzyme that catalyzes the synthesis of RNA

RNA primase – an enzyme that catalyzes the synthesis of RNA b

Semi-conservative - describes a form of replication whereby the newly formed DNA in the daughter cell is made up of an original parental strand and one newly synthesized strand.

single nucleotide polymorphism (SNP) - DNA sequence variations that occur when a single nucleotide (A, T, C, or G) in the genome sequence is altered.

small interfering RNA (siRNA) – a class of 20-25 nucleotide-long RNA molecules that play a number of roles in biology including RNA interference.

somatic cell - any cell in the body except gametes and their precursors

Southern blotting – transfer by absorption of DNA fragments separated in electrophoretic gels to membrane filters for detection of specific base sequences by radio-labeled complementary probes.

splice – the process of removing introns in an RNA transcript.

structural gene – any gene that codes for the amino acid sequences in a protein except the regulatory protein (non-regulatory gene).

systems biology – It is the study of interactions between the components of a biological system and how these interactions give rise the function and behavior of that system.

termination codon - any of three mRNA sequences (UGA, UAG, UAA) that do not code for an amino acid and therefore signal the end of protein synthesis.

topoisomerase - an enzyme that catalyzes the reversible breakage and rejoining of DNA strands

trait – a variation of a character.

transcription - the synthesis of an RNA copy from a sequence of DNA (a gene); the first step in gene expression.

transcription factors – A group of proteins involved in transcription that can regulate gene expression

transduction – the introduction of genetic materials into cells viral methods

transfection – the introduction of genetic material into eukaryotic cells.

transfer RNA – a class of RNA having structures with triplet nucleotide sequences that

are complementary to the triplet nucleotide coding sequences of mRNA. The role of tRNAs in protein synthesis is to bond with amino acids and transfer them to the ribosomes, where proteins are assembled according to the genetic code carried by mRNA.

transform – (bacteria) the genetic alteration of a cell caused by the uptake and expression of foreign DNA

transgenic – being an organism whose genome has been altered by the transfer of a gene or genes from another species or breed

translation - the process in which the genetic code carried by mRNA directs the synthesis of proteins from amino acids.

transcription factor - a protein that binds to regulatory regions and helps control gene expression

## QUESTIONS

1. Explain why DNA and RNA synthesis only occurs in the 5' to 3' direction.
2. What is a plasmid vector and what is its significance?
3. Are the following sequences located in a DNA or an RNA molecule? How can you tell?
  - a. ...CGCAGAAGGCAA...(sequence 1)
  - b. ...CGCTCTTCG...(sequence 2)
4. How many amino acids (just the number, not the identity) are specified in each of the sequences in Question 3?
5. The following sequence is from the transcribed strand of a DNA molecule. What is the sequence on the untranscribed strand?  
...GGGGATGCGAAA...
6. Explain the method behind gel electrophoresis and how it works to separate DNA into fragments.
7. Why was there interest in creating a gene map of the human genome? How could this information be used?
5. Explain how in certain situations, the same gene can be spliced differently and state an advantage of alternative splicing.

6. DNA replication takes place in which direction? What are Okazaki Fragments, why are they formed and how are they corrected?