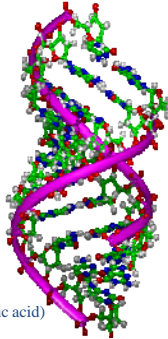


Genetic Control of Protein Structure and Function (F)



DNA
(Deoxyribonucleic acid)

AICE Biology
Ch. 5 Jones; Ch 14 Raven

F Genetic control

Content

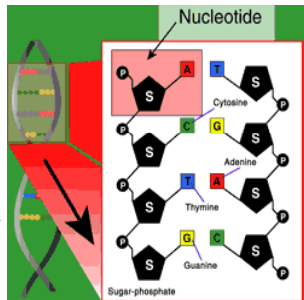
- Structure and replication of DNA
- Role of DNA in protein synthesis

Learning Outcomes

- describe the structure of RNA and DNA and explain the importance of base pairing and the different hydrogen bonding between bases;
- explain how DNA replicates semi-conservatively during interphase;
- state that a gene is a sequence of nucleotides as part of a DNA molecule, which codes for a polypeptide and state that a mutation is a change in the sequence that may result in an altered polypeptide;
- describe the way in which the nucleotide sequence codes for the amino acid sequence in a polypeptide with reference to the nucleotide sequence for HbA (normal) and HbS (sickle cell) alleles of the gene for the β -haemoglobin polypeptide;
- describe how the information on DNA is used during transcription and translation to construct polypeptides, including the role of messenger RNA (mRNA), transfer RNA (tRNA) and the ribosomes;
- use the knowledge gained in this section in new situations or to solve related problems.

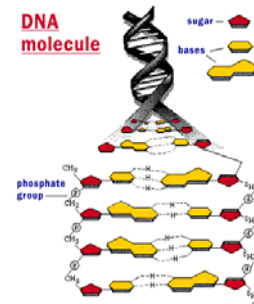
DNA (Deoxyribonucleic acid)

- polynucleotides
- made up of smaller subunits of nucleic acid called *nucleotides*
- there are four possible nucleotides each containing one of four bases



Components of DNA

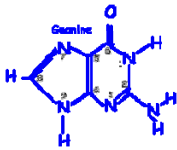
- Deoxyribose sugar
- Phosphate group
- Nitrogen base
 - Organic ring structure that contains one or more atoms of nitrogen



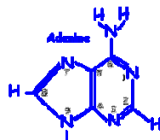
The Nitrogen Bases

Purines (double-ring, larger)

- Triple bond \equiv



Guanine

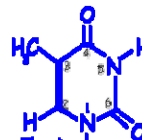


Adenine

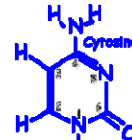
The Nitrogen Bases

Pyrimidines (single ring, smaller)

- Double bond $=$



Thymine



Cytosine

Which proteins a cell makes is determined by genes that are organized in series of 3 base pairs, called "codons"

GCCTA **GTT** ACTGC
CGGAT **CAAT** GACG

codon

Every codon codes a specific amino acid in a specific order

amino acid

Protein

Transcription to RNA

- Making of a single copy of a DNA strand

Codon – RNA code; set of three bases coding for an amino acid (AAA, CGC, etc.)

DNA Strand

5' TGCCTAAGGACCTTATCGAACCTCCTTTAAA 3'
3' ACGGATTCCTGGAATAGCTTGGAGAAATTT 5'

RNA Strand

5' UGCCUAAGGUCCUUAUCGCAACCUCCUUUAAA 3'

Translation (The synthesis of proteins)

- tRNA
- Ribosomes
- Aminoacyl-tRNA synthases

amino acids

Polypeptide

Ribosome

tRNA with amino acid attached

tRNA

Anticodon

mRNA

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Building a polypeptide (Translation)

- Initiation:**
 - brings mRNA, tRNA, and the two ribosomal subunits together
- Elongation:**
 - three-step cycle that adds amino acids one by one to the initial amino acid, requires cooperation of several
- Termination:**
 - release of the polypeptide chain from the complex.

Translation to Protein

- Converting mRNA sequence to amino acid sequence that makes up a protein

Me

UAC

5' [cap] AUGAGAUACCAAGAACCUACCAAGGUAGAGCUUUGCCCG | AAAAAAAAAAAAAA 3'

tRNA

- Interpreter between base sequence of mRNA and amino acid sequence of protein
- 45 different types
- About 80 nucleotides long
- Anticodon base pairs with codon of mRNA

Amino acid attachment site

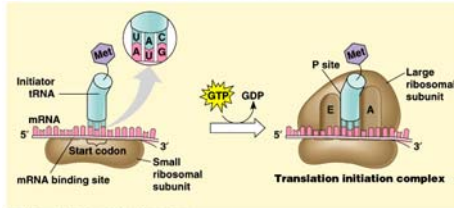
Anticodon

(b) Two-dimensional structure

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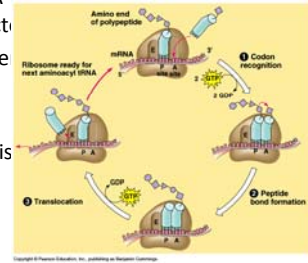
Initiation

- 5' cap attaches to small ribosome subunit
- tRNA carrying methionine attaches to mRNA codon
- Large ribosomal subunit attaches



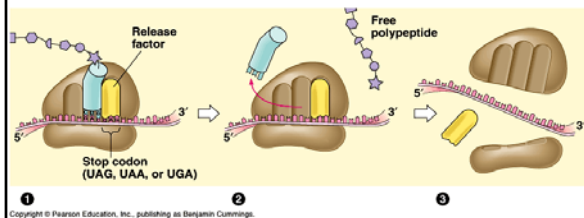
Elongation

- **Codon recognition:**
 - tRNA directed into the A site by an elongation factor
- Peptide bond forms between adjacent amino acids
- **Translocation:**
 - amino acid in the A site is moved to the P site
- mRNA moves through the ribosome 5'→3' direction

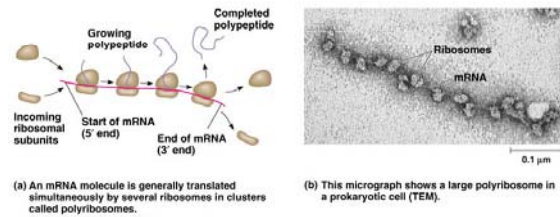


Termination

- Termination sequence is encountered
- Release factor binds to sequence
- Release factor separates polypeptide and tRNA



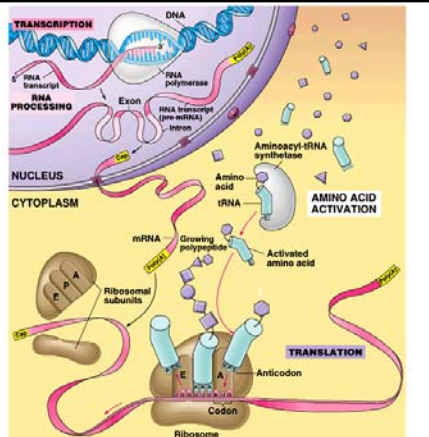
- Typically a single mRNA is used to make many copies of a polypeptide simultaneously.
- Multiple ribosomes, **polyribosomes**, may trail along the same mRNA.
- A ribosome requires less than a minute to translate an average-sized mRNA into a polypeptide.



(a) An mRNA molecule is generally translated simultaneously by several ribosomes in clusters called polyribosomes.

(b) This micrograph shows a large polyribosome in a prokaryotic cell (TEM).

Protein Synthesis



Mutations in DNA

- Frameshift mutation – a single base is added or deleted from DNA

Original Strand

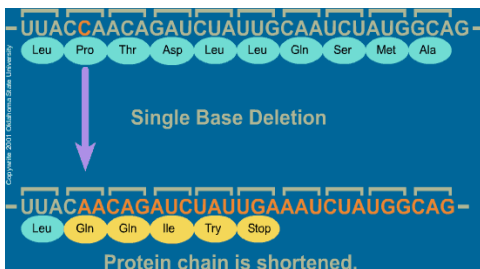
AUG AAG UUU GGC GCA
Met Lys Phe Gly Ala

New Strand

AUG AAG UUG GCG CA..
Met Lys Leu Ala

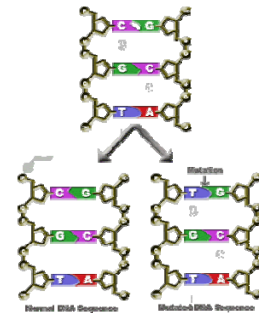
Mutations in DNA

- Point Mutation – a change in a single base pair in DNA



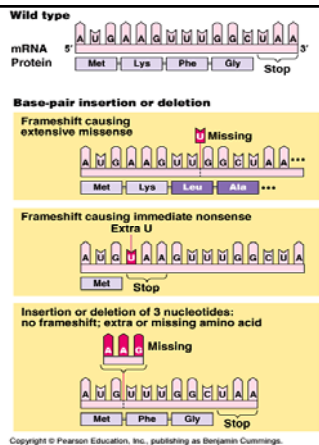
Causes of mutation

- Spontaneous mutation – mutations that occur at random
- Environmental
 - Exposure to X-rays, UV light, radioactive substances and other chemicals



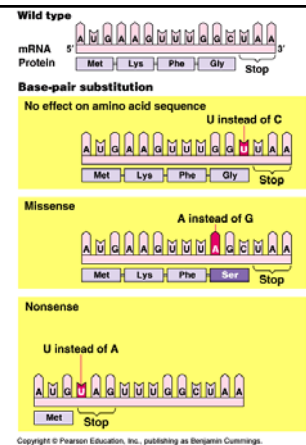
Point Mutations

- Insertions or deletions:
 - add or subtract base pair(s)
- May cause frameshift
- Mutations



Point Mutations: *substitutions*

- Replacement of one base pair with another
- Types:
 - silent
 - conservative
 - missense
 - nonsense
- One wrong letter (8 ½ min)

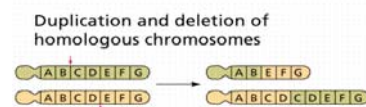
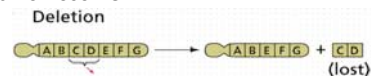


Mutations in Chromosomes

- Changes happen to the chromosome itself
- Affect the distribution of genes to gametes during meiosis

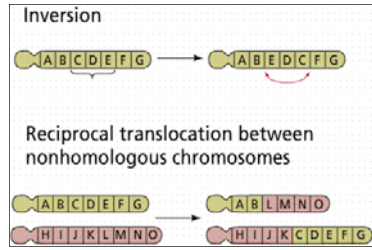
Types of Mutations

- Deletion – part of a chromosome is left out
- Insertion – a chromatid breaks off and attaches to sister chromatid
 - Results in duplication of genes on the same chromosome



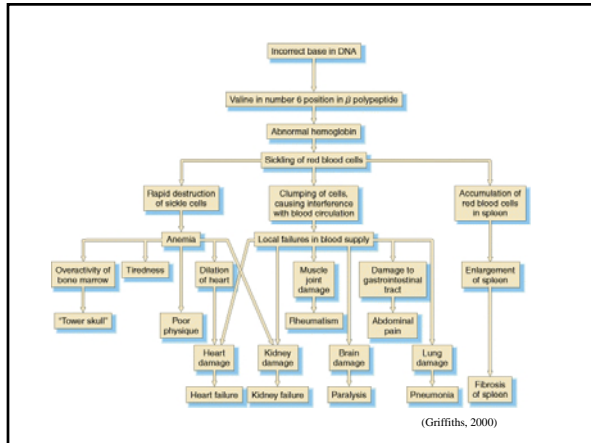
Types of Mutations

- Inversion – part a chromosome breaks off and is reinserted backwards
- Translocation – part of one chromosome breaks off and added to a different chromosome
- [clip](#)



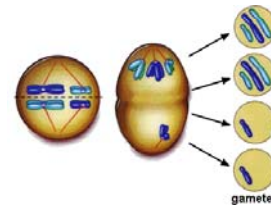
Sickle Cell

- Of all the amino acids known to make up a hemoglobin molecule, a substitution of valine for glutamic acid at just one point, position 6 in the β chain, is all that is needed to produce the defective hemoglobin
- <http://www.ncbi.nlm.nih.gov/books/NBK21811/#A1638>



Nondisjunction

- The failure of homologous chromosomes to separate properly during meiosis



Trisomy

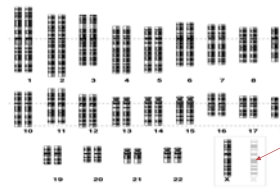
- A gamete with an extra chromosome is fertilized by a normal gamete
- Zygote has an extra chromosome
- Ex. Down syndrome (Trisomy 21)



Extra chromosome

Monosomy

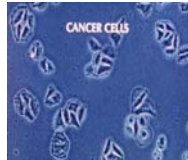
- A gamete with a missing chromosome is fertilized by a normal gamete
- Zygote lacks a chromosome
- Ex. human females with only a single X-chromosome



Missing chromosome

Meiosis & Mitosis ??

- If mutation occurs in gametes
 - Leads to birth defects
- If mutation occurs in body cells
 - Cancer



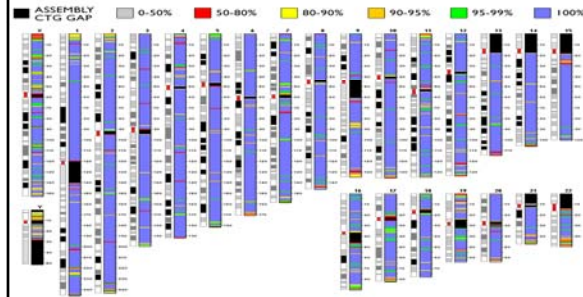
Oncogene: Cancer



[Oncogene clip](#)

The Human Genome Project

- Working to determine the sequences of nucleotide bases for the human species.



Resource

Griffiths, A., Miller, J., Suzuki, D., et al. (2000). *An Introduction to Genetic Analysis*. 7th edition. New York: W. H. Freeman and Company.

Jones, M., Fosbery, R., Taylor, D., & Gregory, J. (2007). *AS Level and A Level Biology*, 2nd ed. Cambridge, UK: Cambridge University Press.