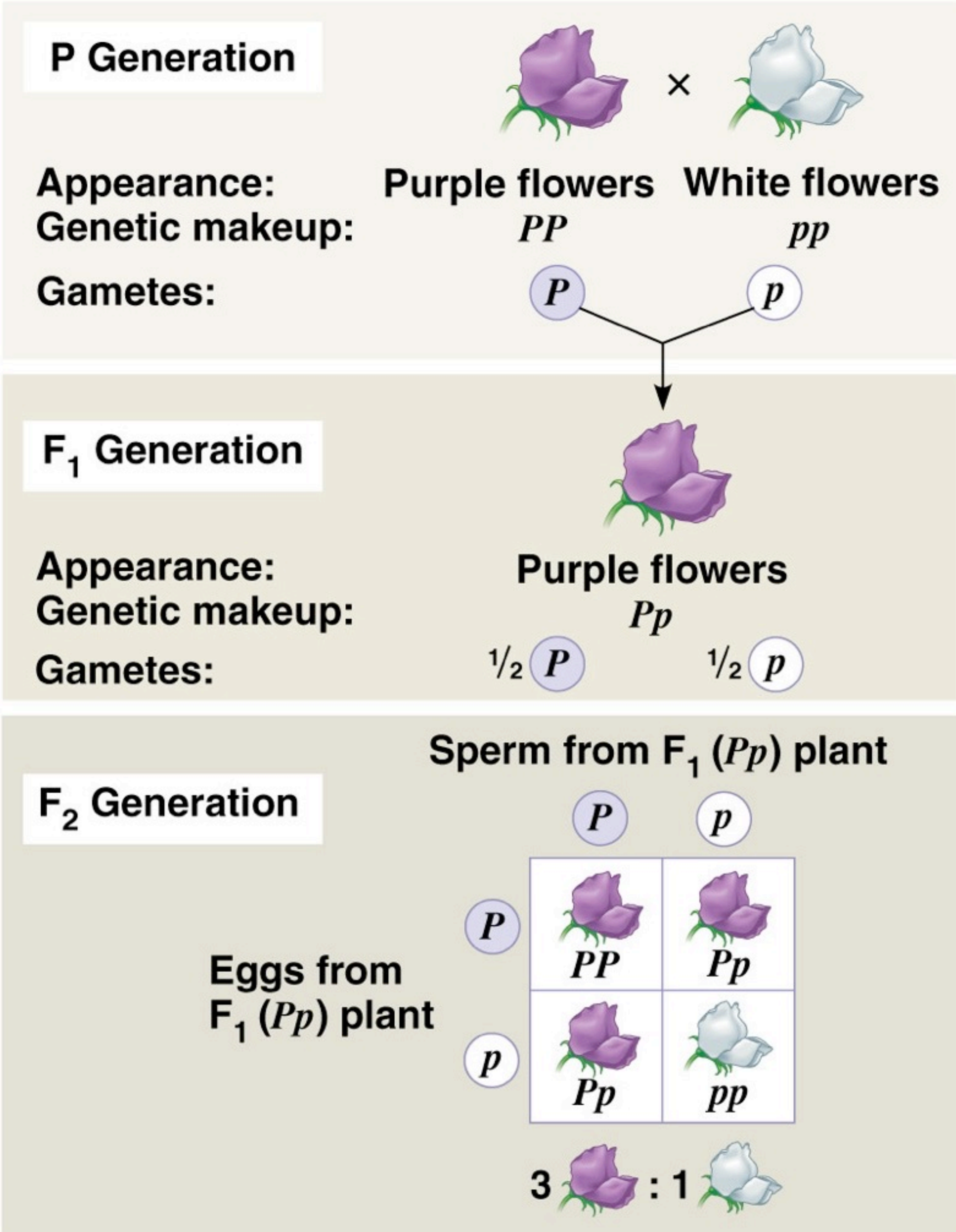
Mendel's Model

- 1) Trait variation is due to alternative variations (alleles) of "heritable factors" (genes)
- 2) For each character an organism inherits two alleles, one from each parent
- 3) Dominant alleles mask recessive alleles
- 4) Two alleles for a heritable character segregate (separate) during gamete formation and end up in different gametes (= Law of Segregation)



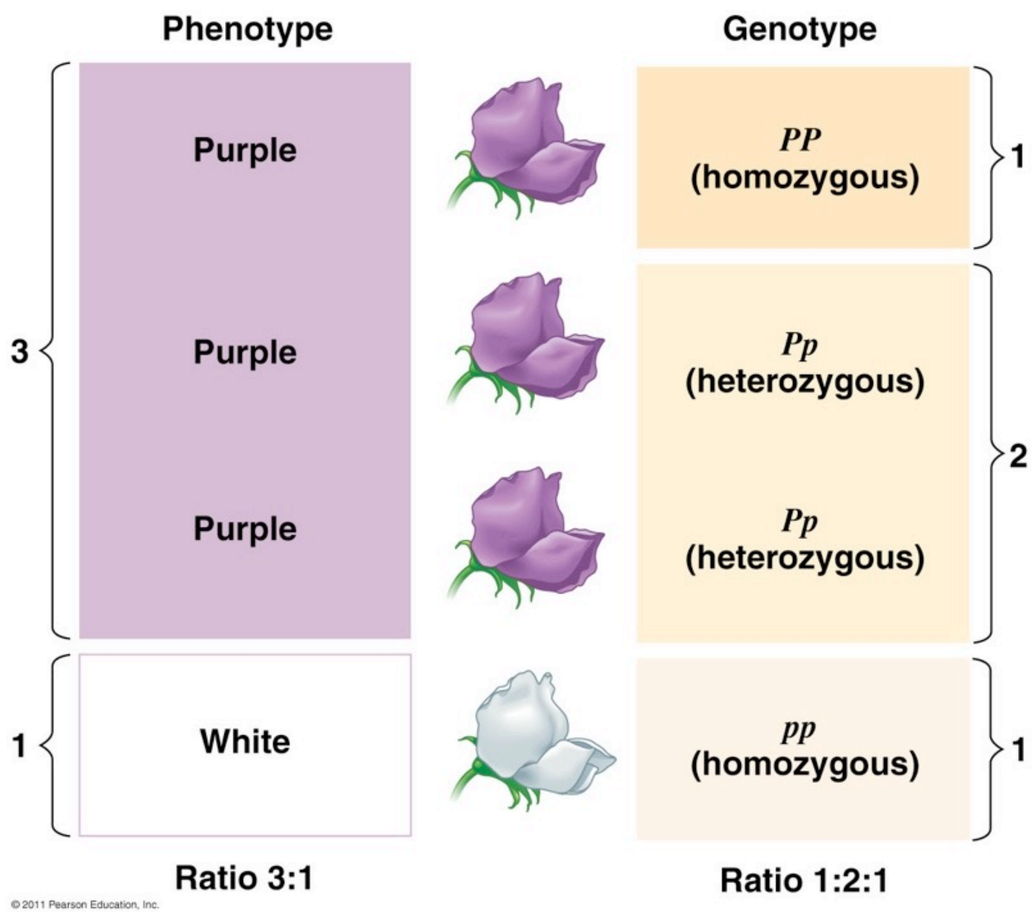
The allele for purple color is dominant (P), white is recessive (p)

Genes occur in pairs in most cells, but are separated in gametes i.e. gametes are haploid whereas somatic cells are diploid

Punnett square explains 3:1 ratio in F2

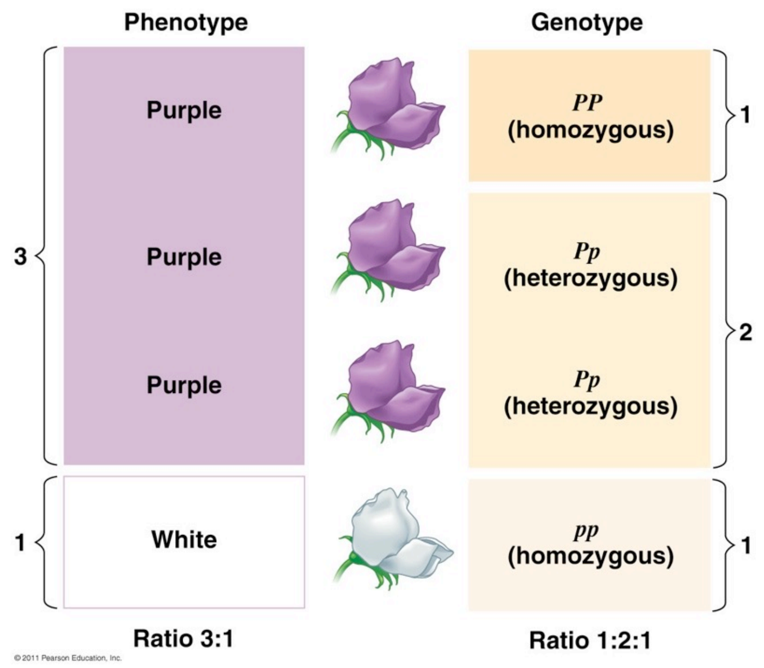
Mendel's study of dominant and recessive traits

Genotype vs phenotype



Note: many other phenotypic traits affected by environment as well as genetics

Genotype : homozygous vs heterozygous (remember; for one trait)



Homozygous: the organism has two identical alleles

Heterozygous: the organism has two different alleles

Note: heterozygous individuals will show the trait of the dominant allele

Genotype vs phenotype

Genotype: the genetic makeup of an organism

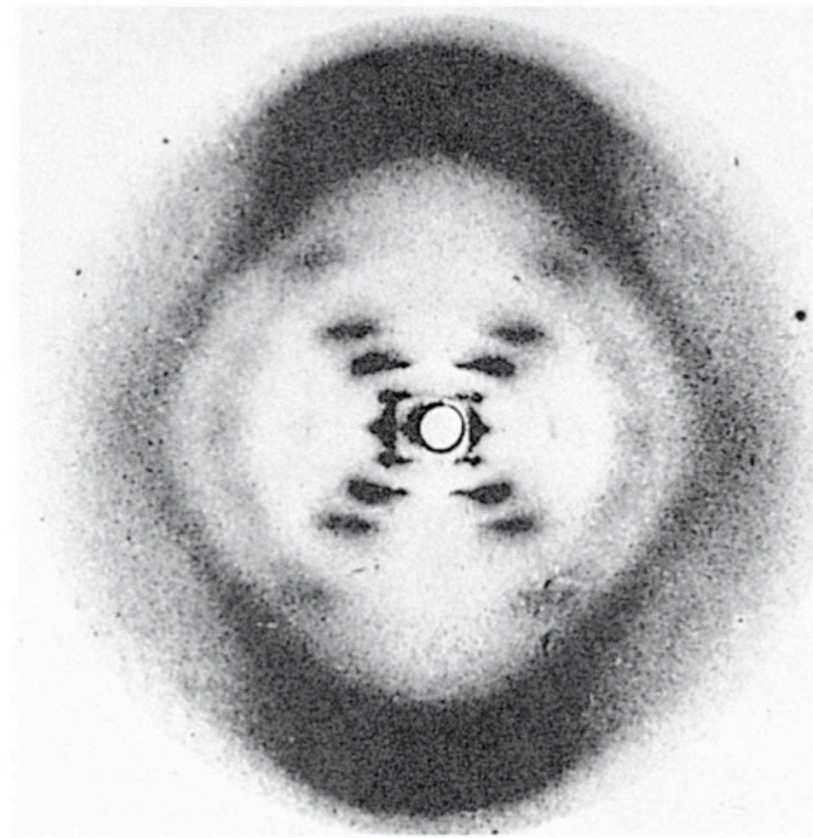
Phenotype: the physical traits as observed in the living organism

Note: many phenotypic traits affected by environment as well as genetics e.g. how tall you are

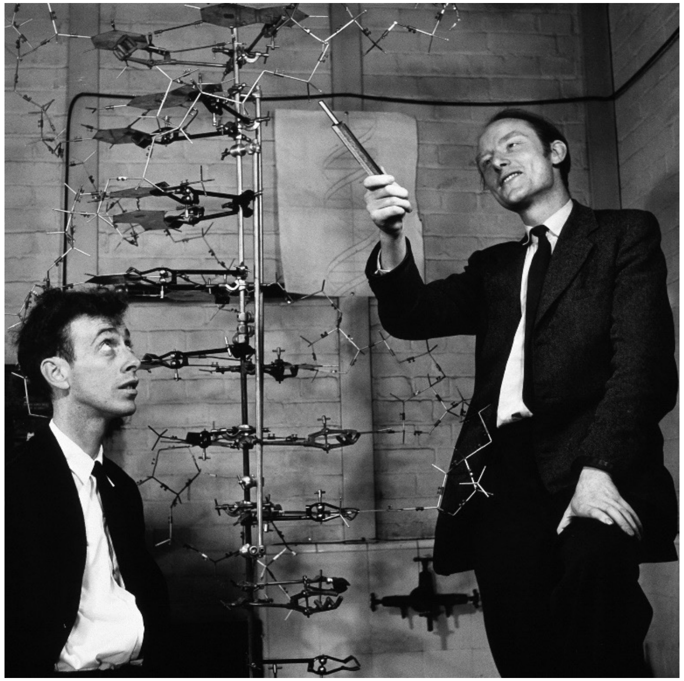
Other phenotypic traits can be directly related to genotype only; e.g. eye color

But what is the structure of nucleic acids?

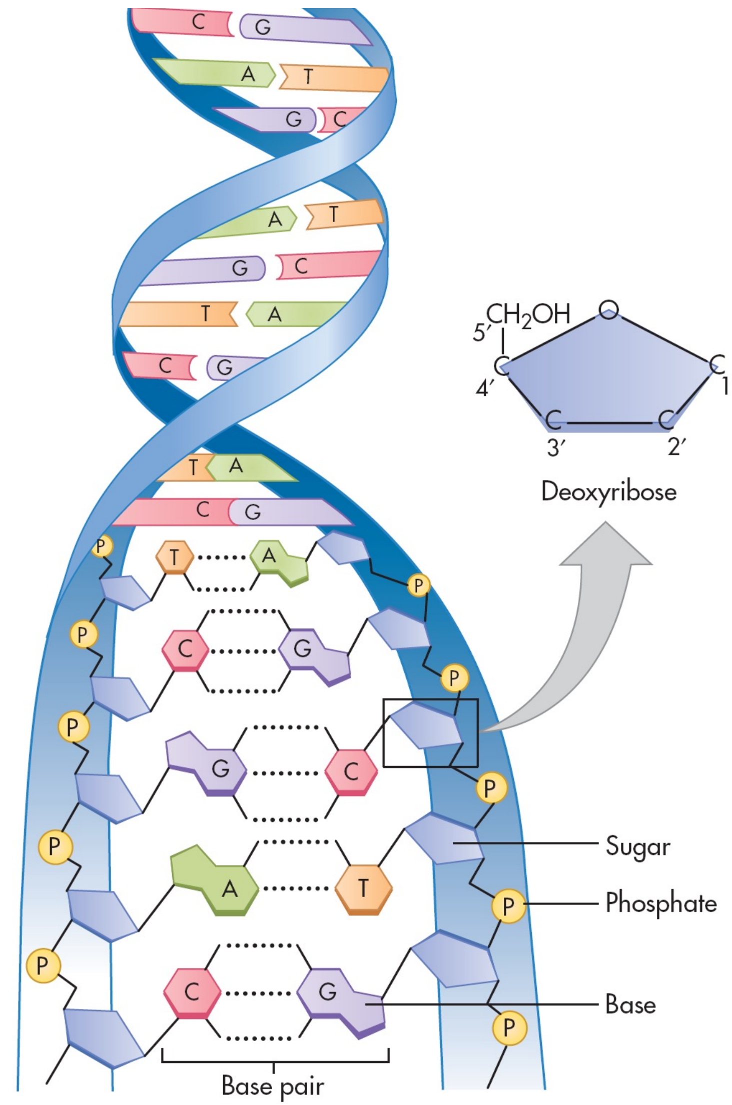
(a) Rosalind Franklin



(b) Franklin's X-ray diffraction photograph of DNA



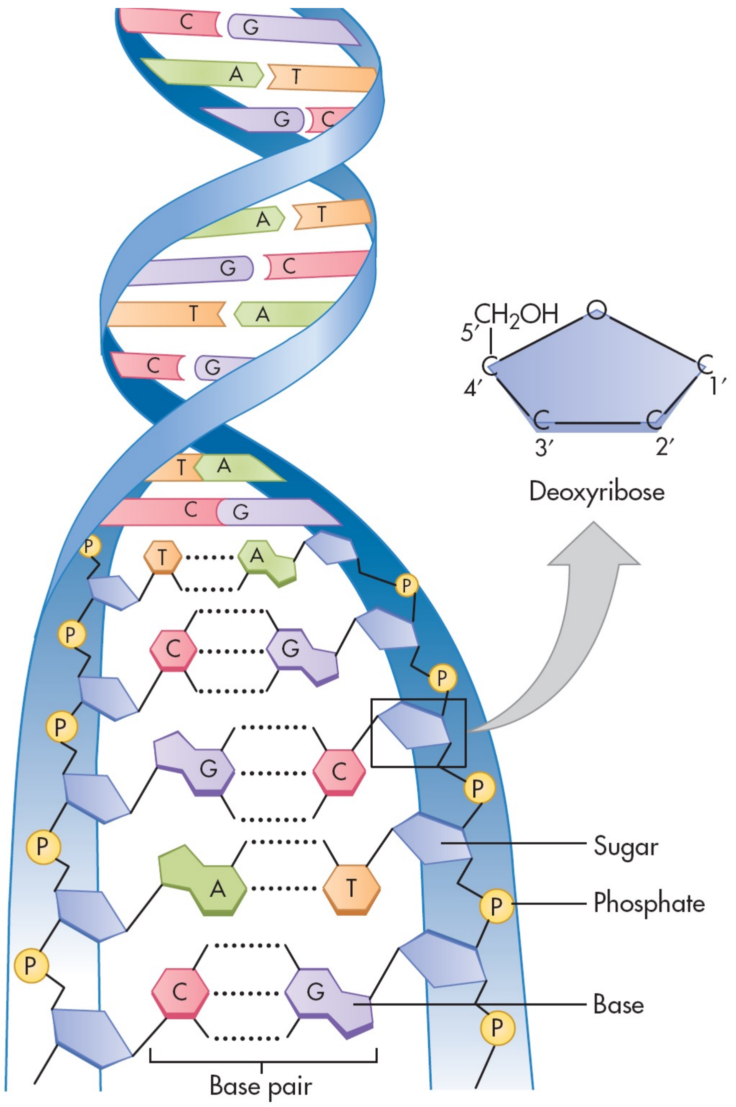
DNA



- Structure described by Watson and Crick 1962 using images by Rosalind Franklin's lab
- A double helix, antiparallel strands

DNA

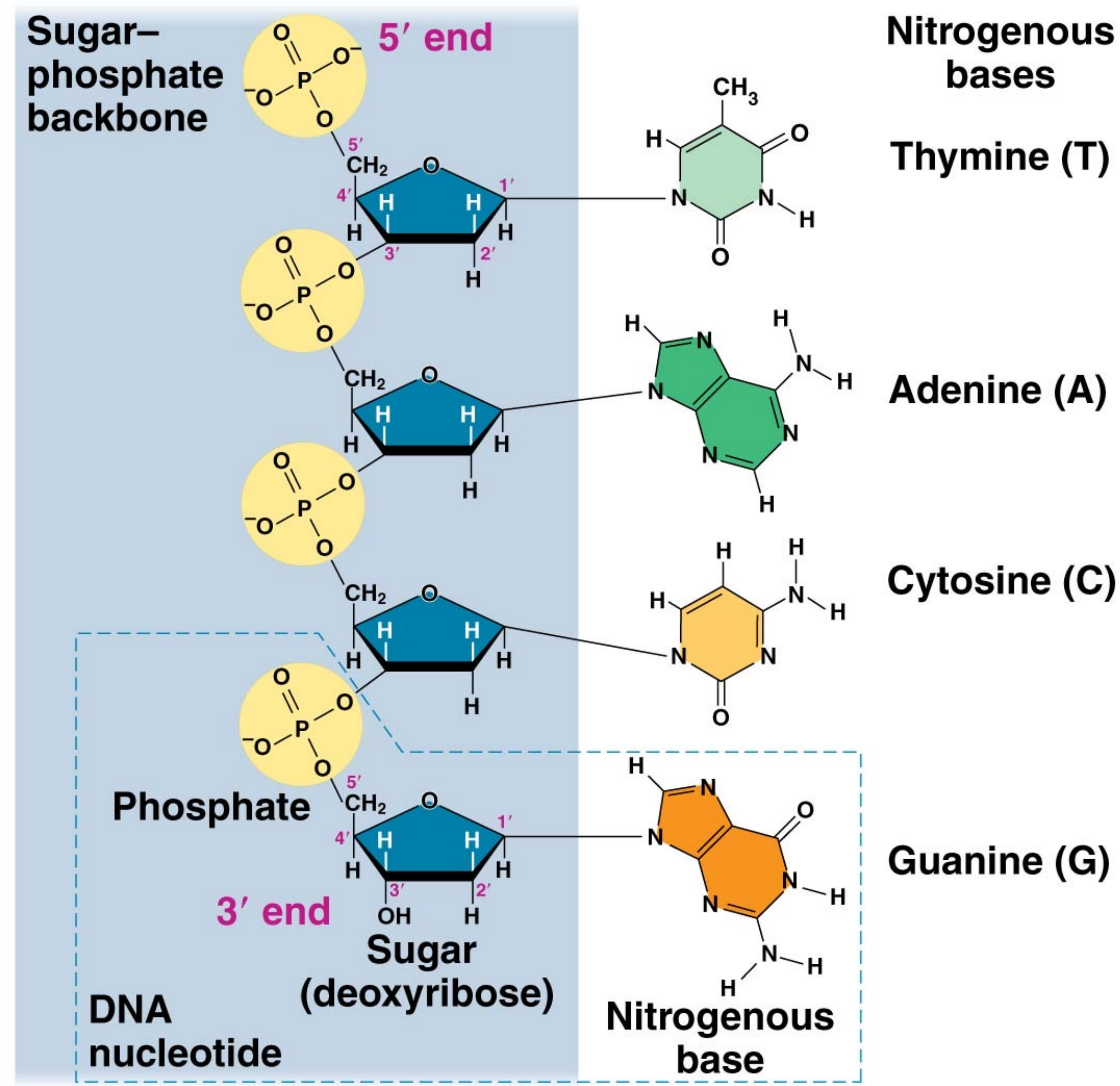
- **DNA= Deoxyribonucleicacid**
  - **Sugar part consists of deoxyribose**
  - **Bases on the two strands form base pairs: Adenine with Thymine, Guanine with Cytosine ; bound with relatively weak hydrogen bonds**

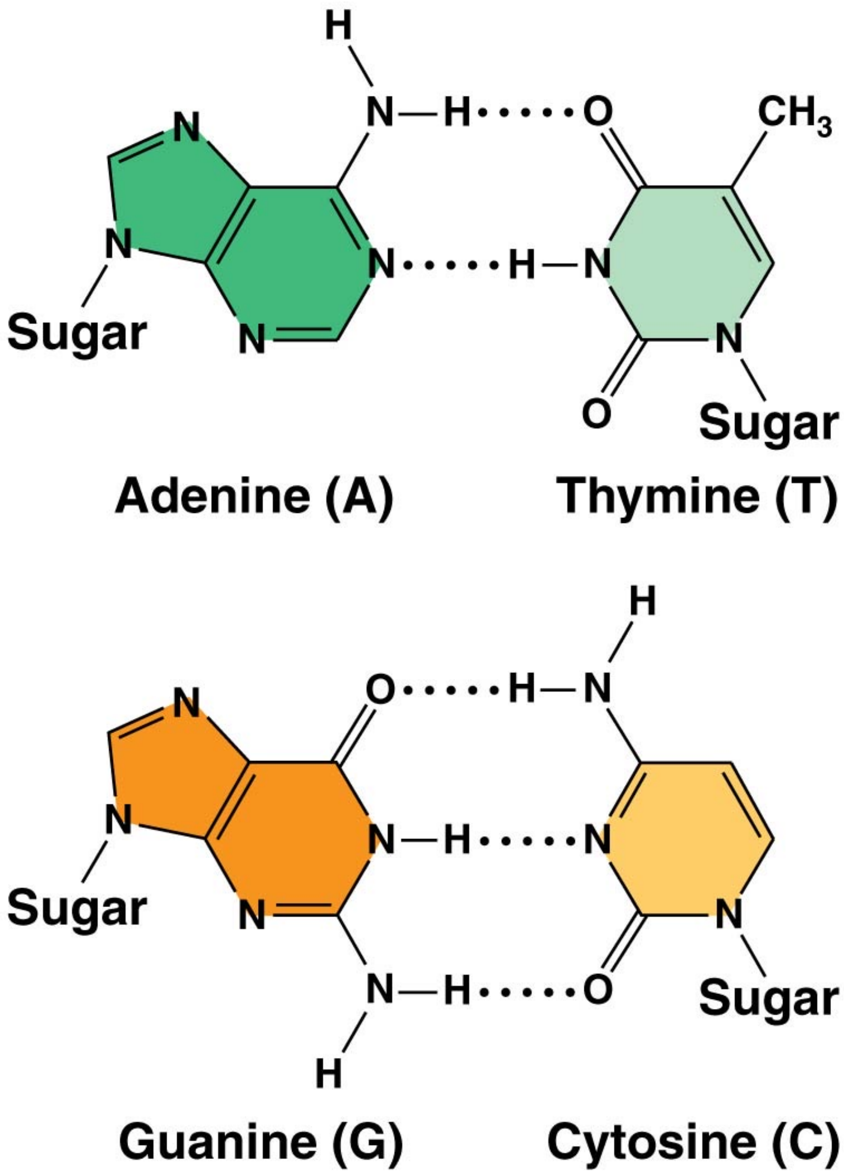


- The pentose sugar (deoxyribose or ribose) and phosphate groups make up the “backbone” of a nucleic acid. Formed with covalent bonds

-Each nucleotide also contains nitrogenous bases. (H are slightly positive and O, N slightly negative)

-Notice the numbering of the carbons in the pentose sugar (1-5): each strand has a 5’ (5 prime) end, and 3’ end





Purines (2 rings)

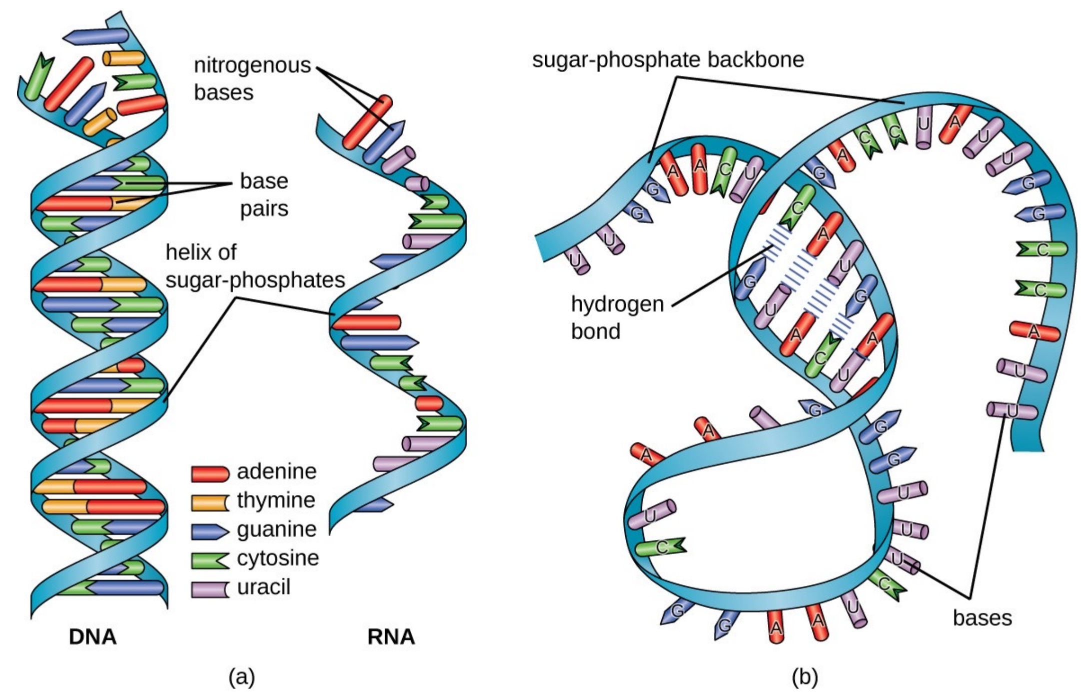
Pyrimidines (1 ring)

The four nitrogenous bases that comprise DNA form the **genetic code**

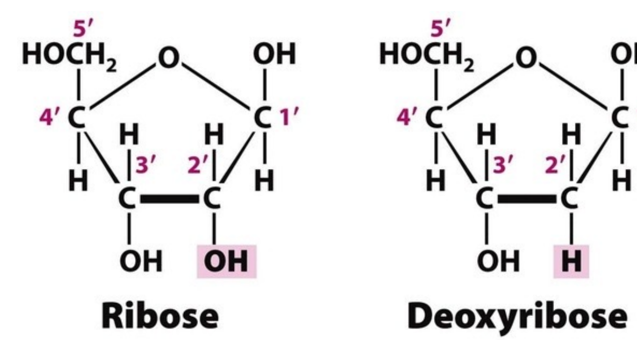
A DNA molecule's sequence of bases contains the instructions for the production of proteins (through mRNA intermediate)

Nucleic acids: DNA vs RNA

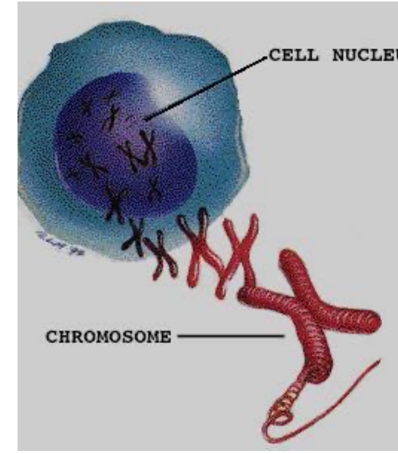
- **DNA= Deoxyribonucleicacid**
  - **Sugar part consists of deoxyribose**
  - **Adenine, Thymine, Guanine, Cytosine (ATGC)**
  - **Genetic code in the cell nucleus**
  - **Antiparallel double helix**



- **RNA = Ribonucleicacid**
  - **Sugar part consists of ribose**
  - **Adenine, Uracil, Guanine, Cytosine (AUGC)**
  - **Can function as an enzyme as well as storing genetic code**
  - **Not a double helix: usually single strand, complex shape**



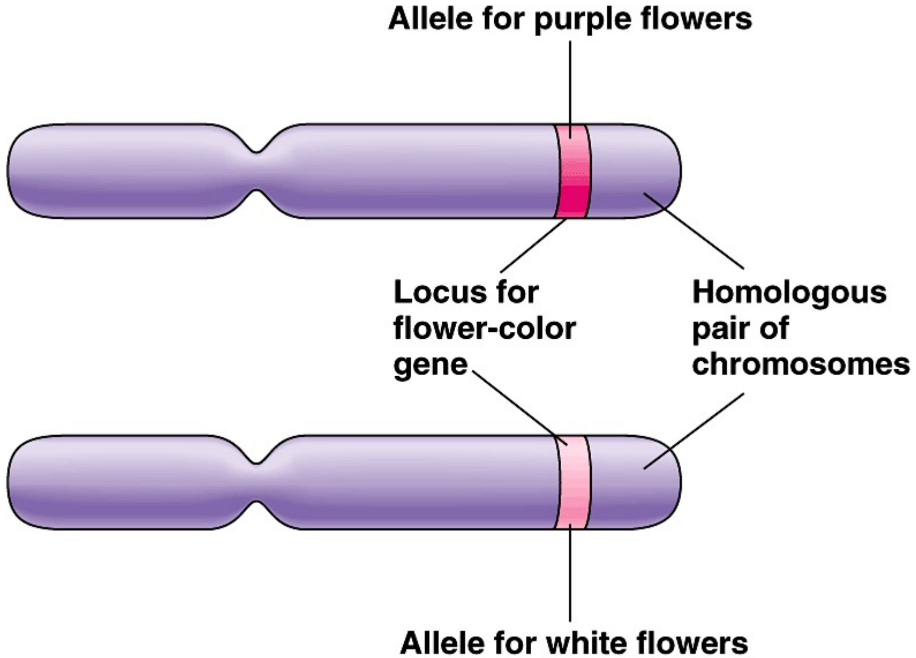
Chromosomes



- Long string of DNA plus proteins
- 23 pairs (2n) in humans (including 1 pair of sex chromosomes)
- Paired (2n) in <u>diploid</u> cells:
    - <u>Haploid</u> cells (sperm, egg) have only one set (n)
- Therefore: most cells have 2 copies of each gene

Gene

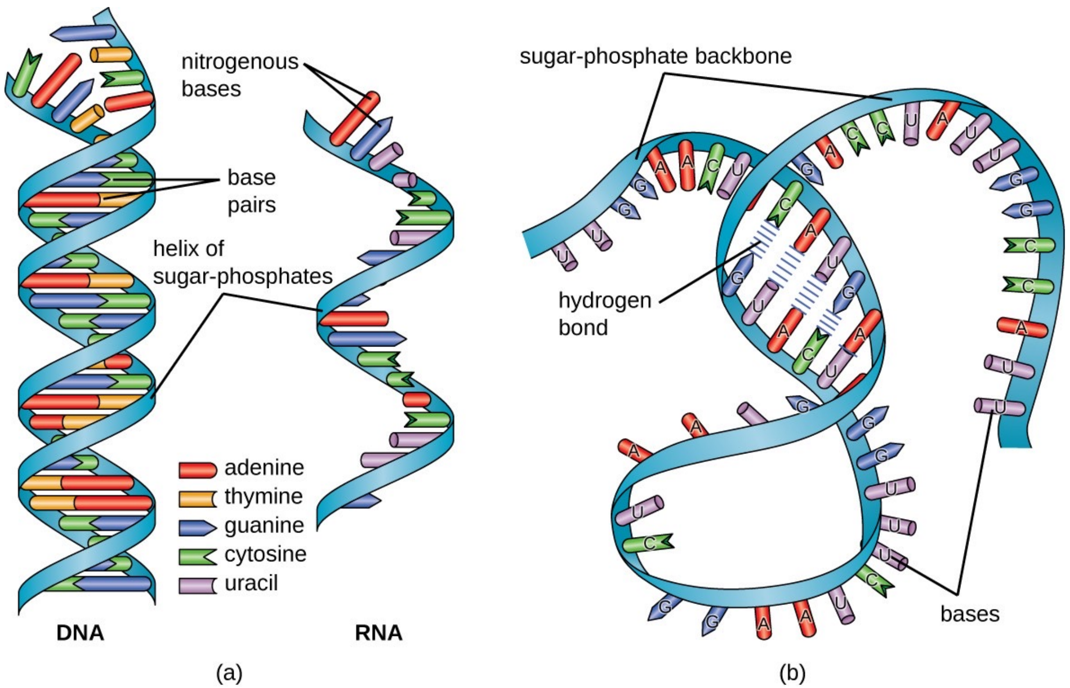
- Gene = fundamental unit of heredity on the DNA (humans: about 25 000 genes)
  - Sequence of DNA that codes for particular combination of amino acids (polypeptide, protein)
  - Much of the DNA is non-coding
  - Different versions of the same gene are called alleles
  - Locus = position of gene in genome



How do genes code for characteristics?

-most genes are a “recipe” for the production of protein

-Genes on DNA are transcribed to mRNA, which is then translated to protein through the use of tRNA (mRNA=messenger RNA; tRNA = transfer RNA)

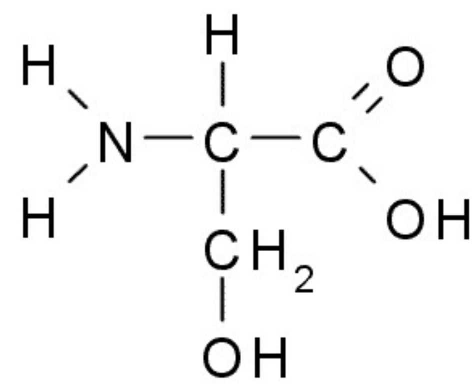


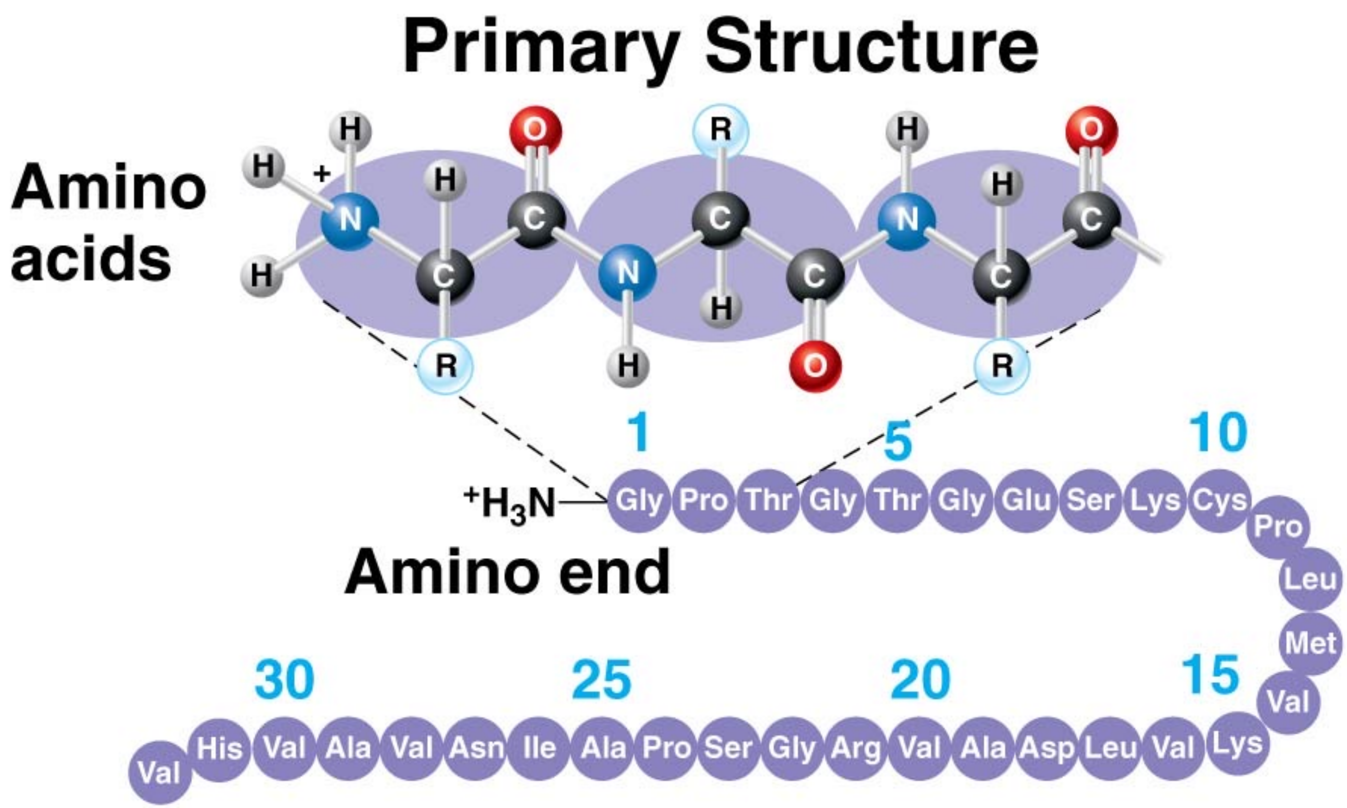
How do genes code for characteristics?

Genes code for proteins

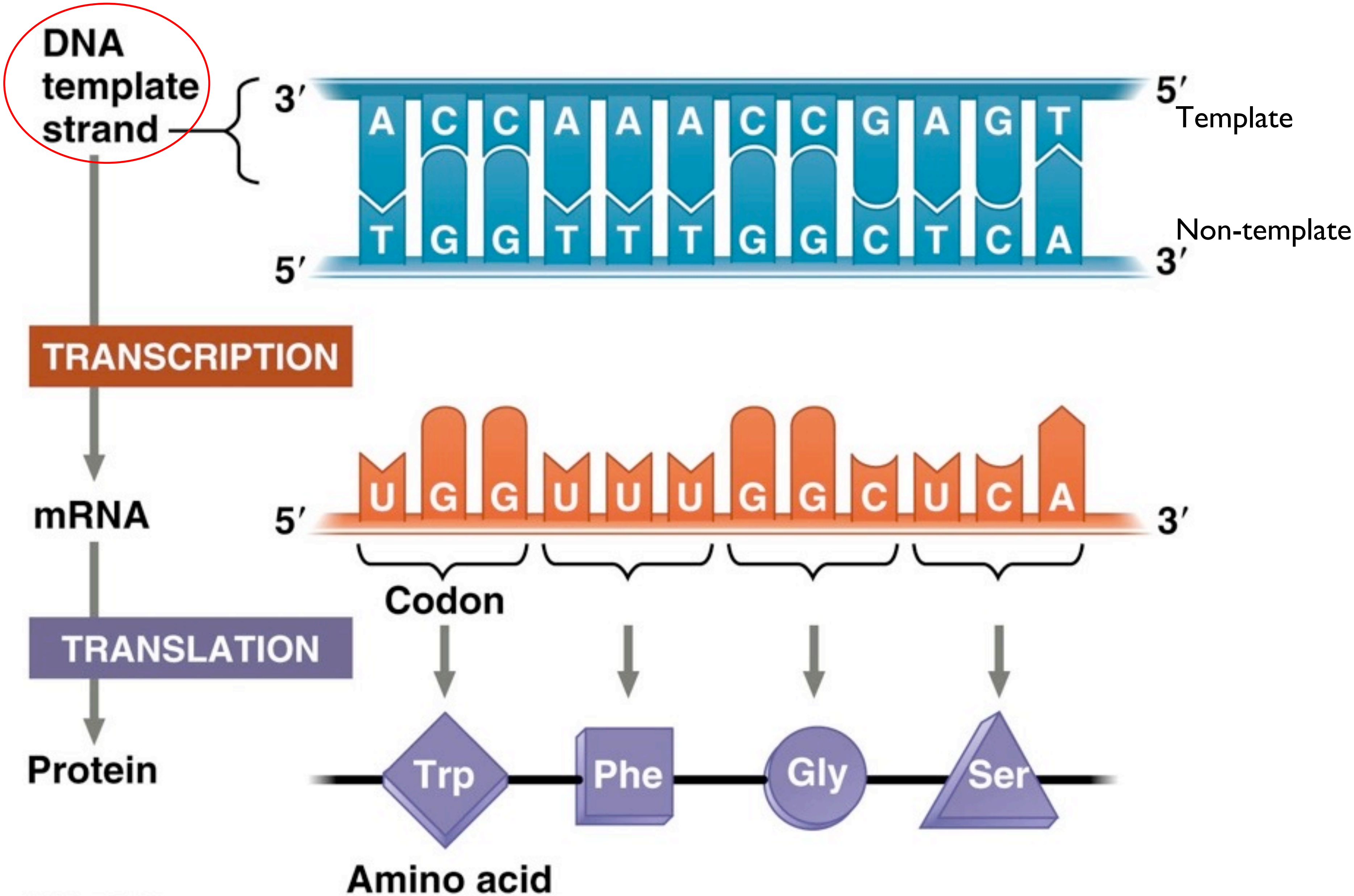
= Polymers of amino acids = polypeptides
- constructed from 20 amino acids

Example amino acid: serine



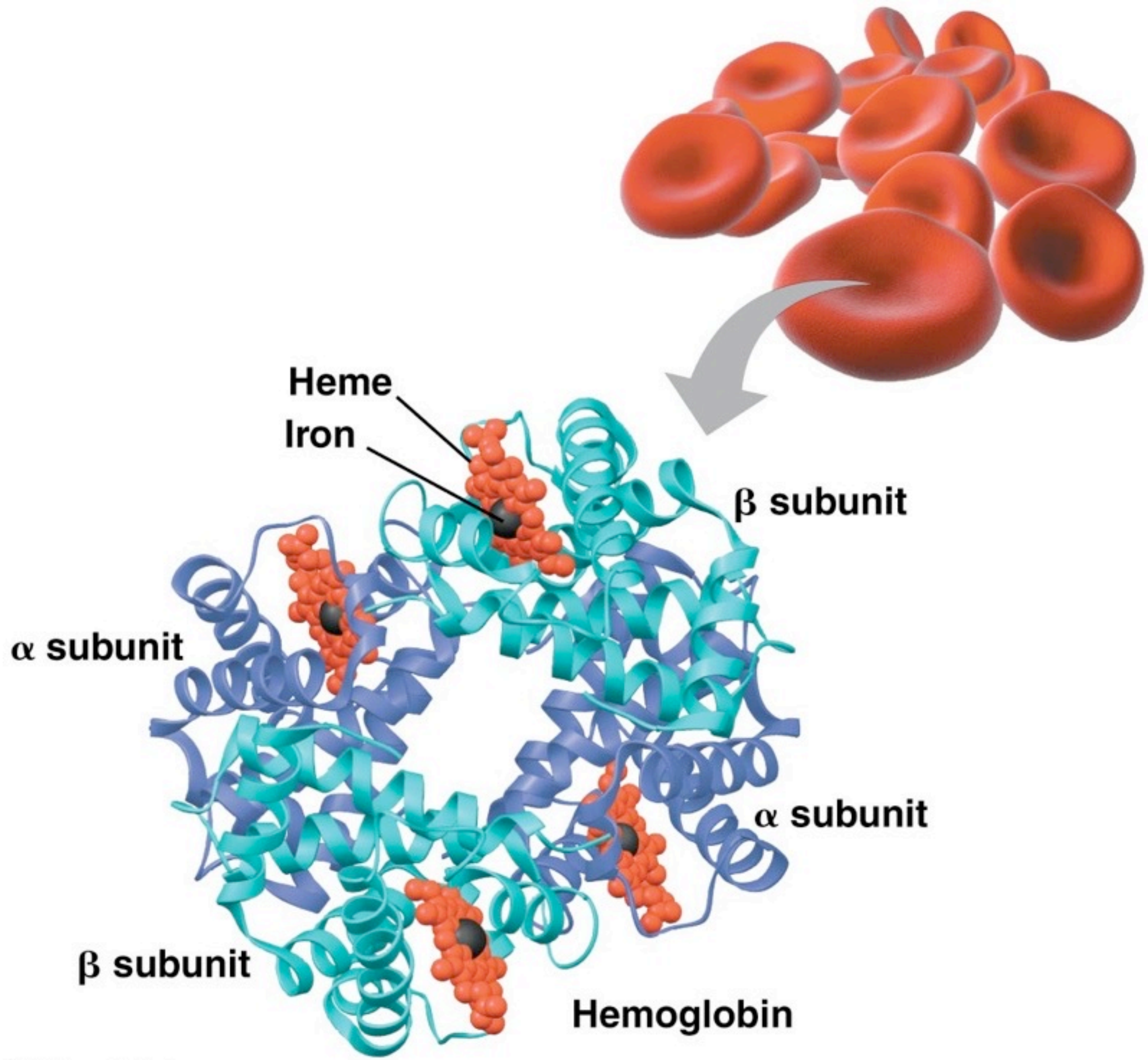



DNA
RNA
Protein


DNA template strand
3′
A C C A A A C C G A G T
5′
Template
T G G T T T G G C T C A
5′
Non-template
3′
TRANSCRIPTION
mRNA
5′
U G G U U U G G C U C A
3′
Codon
TRANSLATION
Protein
Trp
Phe
Gly
Ser
Amino acid

Proteins are the most structurally complex molecules known...

- vary extensively in structure, enabling their diverse functions
- each protein has a unique three-dimensional shape



Nearly every dynamic function of a living being depends on proteins

Enzymatic proteins

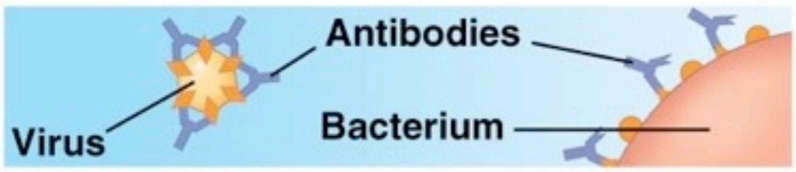
Function: Selective acceleration of chemical reactions

Example: Digestive enzymes catalyze the hydrolysis of bonds in food molecules.

Defensive proteins

Function: Protection against disease

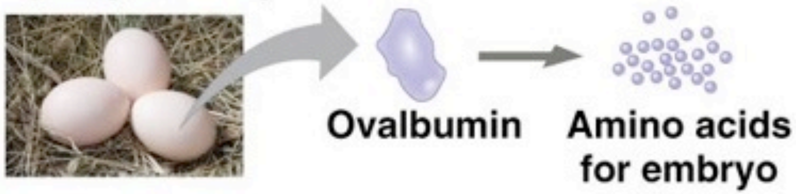
Example: Antibodies inactivate and help destroy viruses and bacteria.



Storage proteins

Function: Storage of amino acids

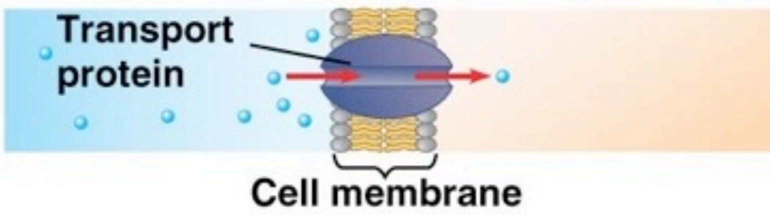
Examples: Casein, the protein of milk, is the major source of amino acids for baby mammals. Plants have storage proteins in their seeds. Ovalbumin is the protein of egg white, used as an amino acid source for the developing embryo.



Transport proteins

Function: Transport of substances

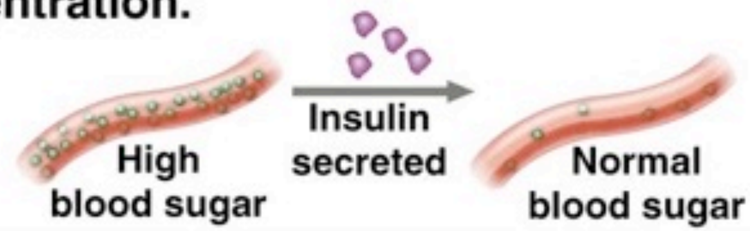
Examples: Hemoglobin, the iron-containing protein of vertebrate blood, transports oxygen from the lungs to other parts of the body. Other proteins transport molecules across membranes, as shown here.



Hormonal proteins

Function: Coordination of an organism's activities

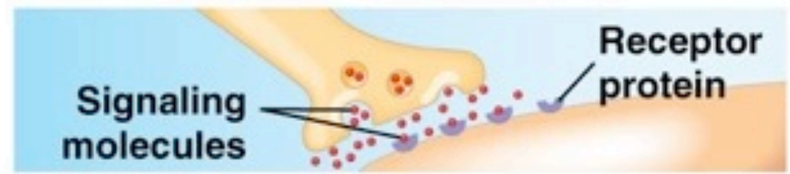
Example: Insulin, a hormone secreted by the pancreas, causes other tissues to take up glucose, thus regulating blood sugar concentration.



Receptor proteins

Function: Response of cell to chemical stimuli

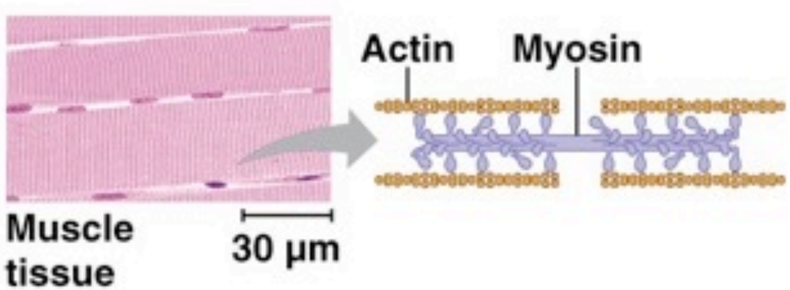
Example: Receptors built into the membrane of a nerve cell detect signaling molecules released by other nerve cells.



Contractile and motor proteins

Function: Movement

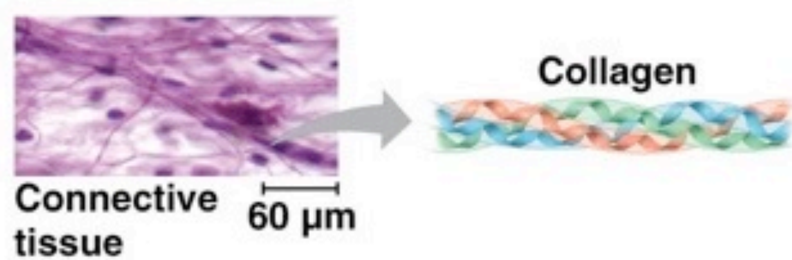
Examples: Motor proteins are responsible for the undulations of cilia and flagella. Actin and myosin proteins are responsible for the contraction of muscles.



Structural proteins

Function: Support

Examples: Keratin is the protein of hair, horns, feathers, and other skin appendages. Insects and spiders use silk fibers to make their cocoons and webs, respectively. Collagen and elastin proteins provide a fibrous framework in animal connective tissues.



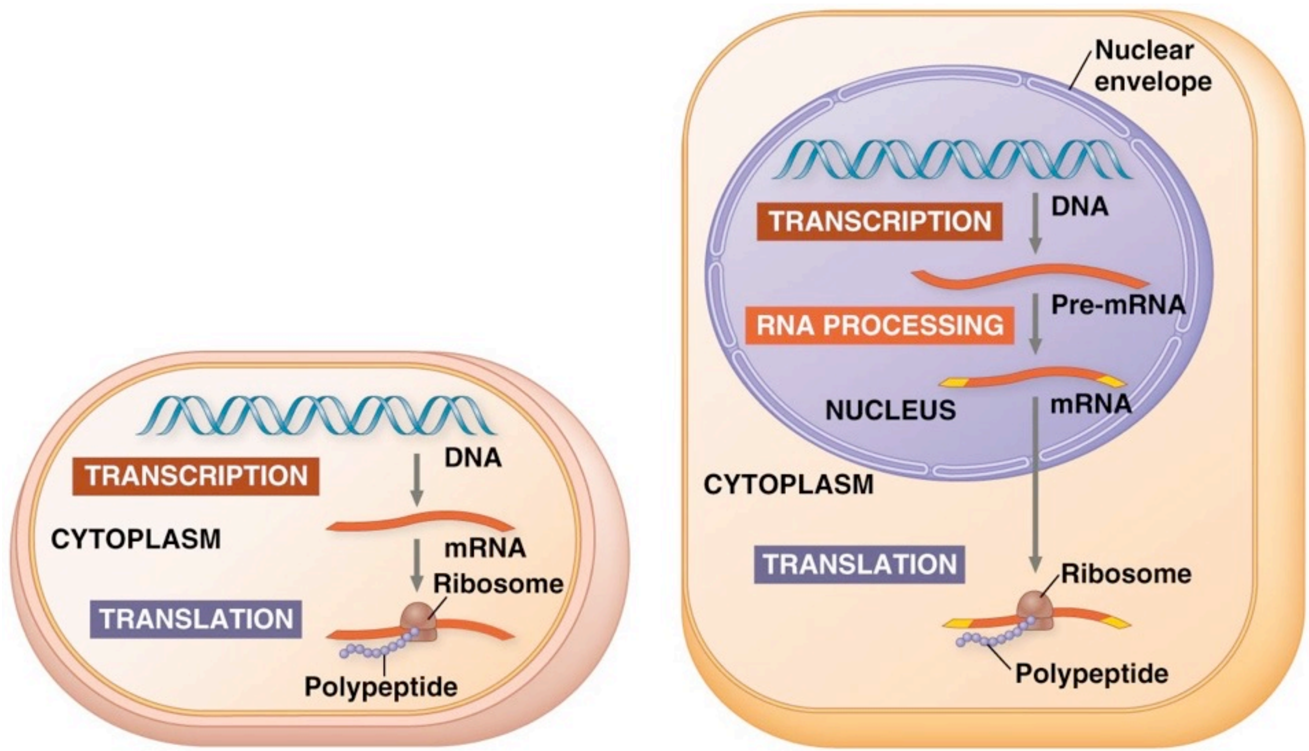
mRNA is translated into amino acids (includes a processing step in eukaryotic organism)

The genetic code is based on nitrogenous base triplets

The way the genetic code is shown in literature is usually the mRNA

Example start and stop codes of a gene: AUG (start), UGA (stop)

Example amino acid: AGU = serine



(a) Bacterial cell (b) Eukaryotic cell

mRNA is translated into amino acids

The flow of information from gene to protein is based on a triplet code

Every three bases (a triplet) codes for a single, specific amino acid (but each amino acid is associated with several codes)

Proteins are made by transcribing and translating this code

Certain codes stand for “start” (AUG) or “stop” (e.g. UGA)

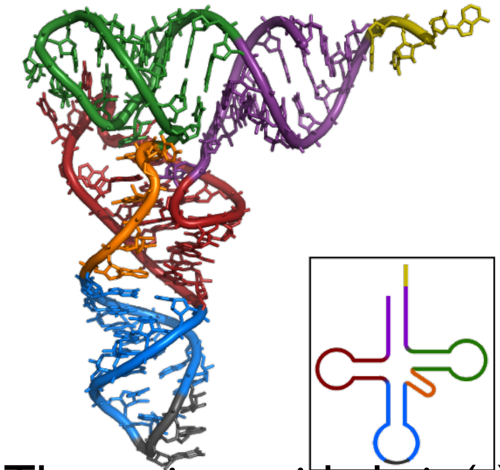
(amino acids are abbreviated; e.g. Ser=serine, Met=methionine)

First mRNA base (5′ end of codon)	Second mRNA base: U		C		A		G		Third mRNA base (3′ end of codon)
U	UUU	Phe	UCU	Ser	UAU	Tyr	UGU	Cys	U
	UUC		UCC		UAC		UGC		C
	UUA	Leu	UCA		UAA	Stop	UGA	Stop	A
	UUG		UCG		UAG	Stop	UGG	Trp	G
C	CUU	Leu	CCU	Pro	CAU	His	CGU	Arg	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	Gln	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	Ile	ACU	Thr	AAU	Asn	AGU	Ser	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	Lys	AGA	Arg	A
	AUG	Met or start	ACG		AAG		AGG		G
G	GUU	Val	GCU	Ala	GAU	Asp	GGU	Gly	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	Glu	GGA		A
	GUG		GCG		GAG		GGG		G

Transcription and translation summary

Transcription in nucleus: DNA is opened and the code is transcribed to an mRNA (messenger RNA) molecule (with some enzyme processing)

Translation in ribosome: done with the help of tRNA (transfer RNA) molecules which bind to specific amino acids



The amino acid chain(s) (polypeptide) forms a protein