Lesson 1

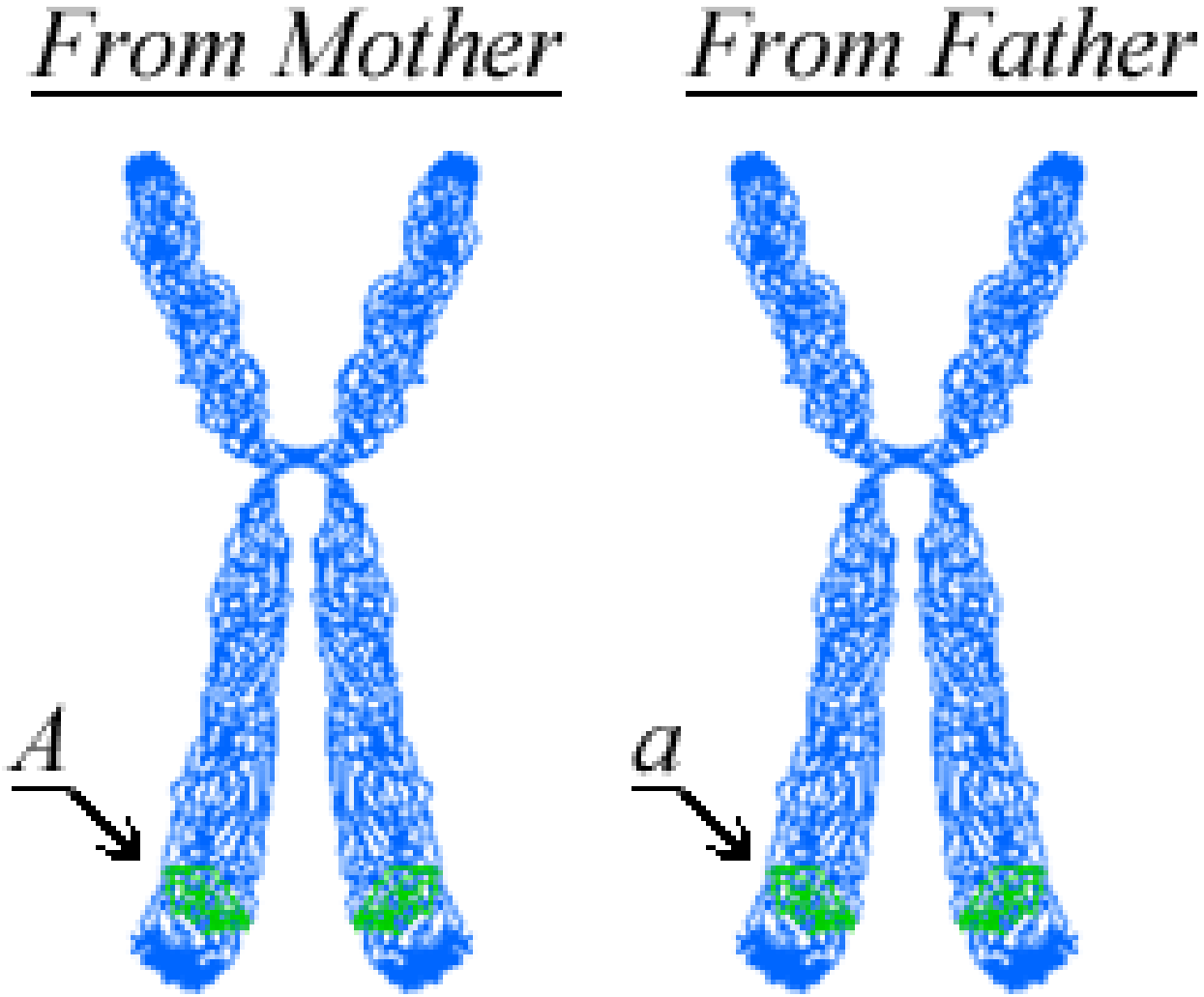
Intro to Genetics

Mendel

Important Vocabulary

Dominance

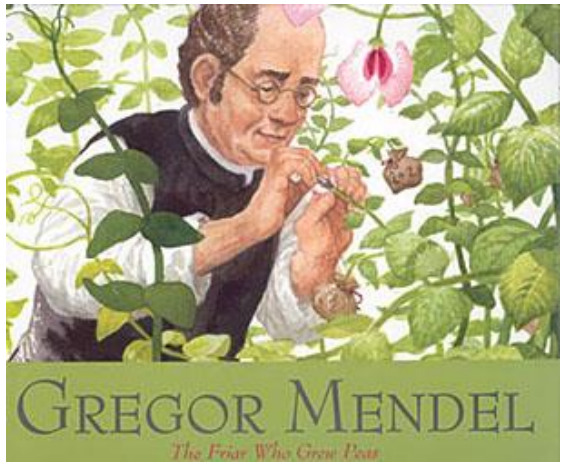
Basics of Heredity and Genetics

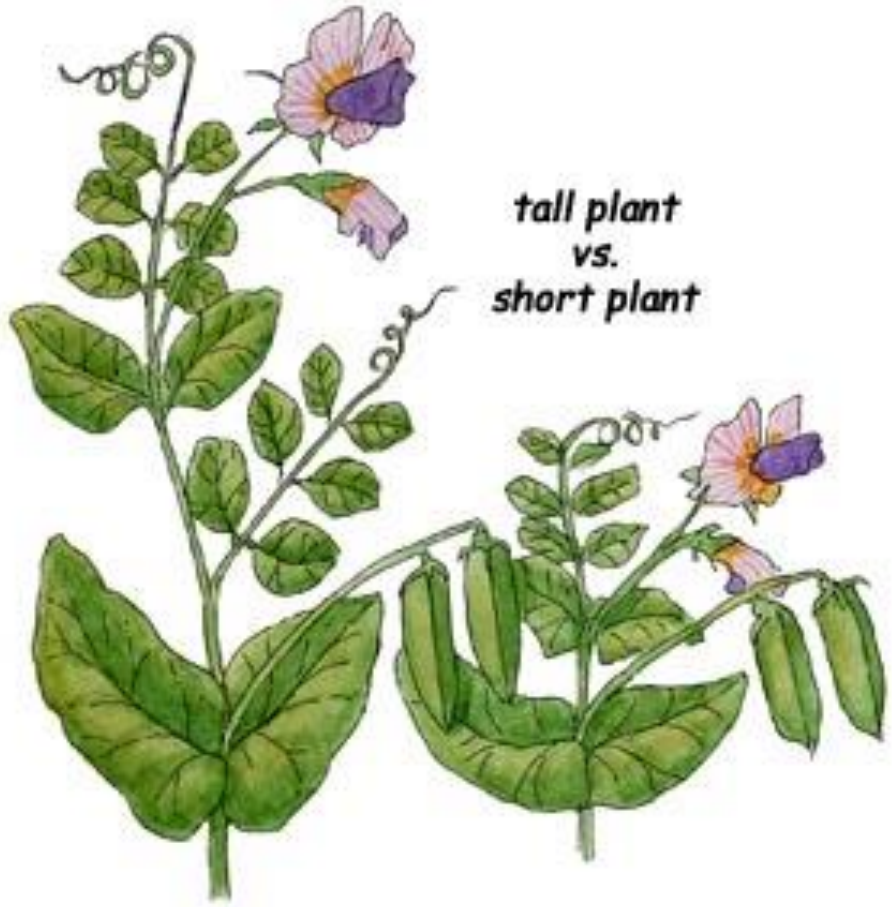


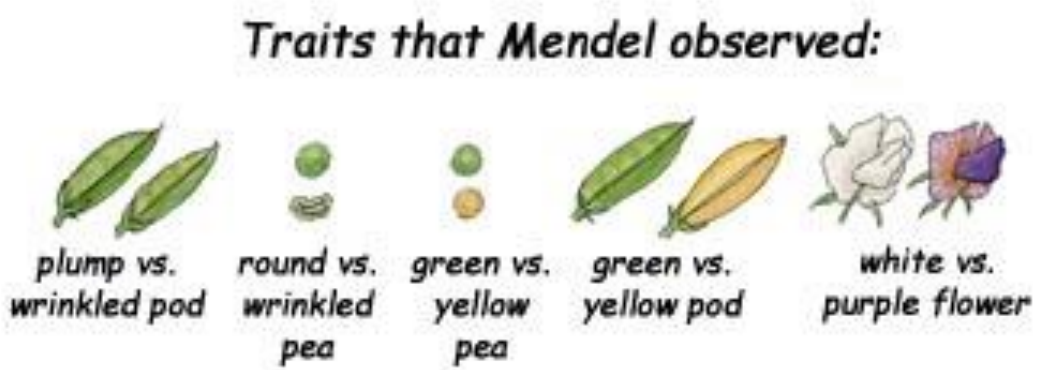
Historical Genetics

Gregor Mendel (1822-1884)

- Established principles of heredity using pea plants
- Inheritance is determined by individual units (we now know as genes)
- Had no knowledge of genes or chromosomes



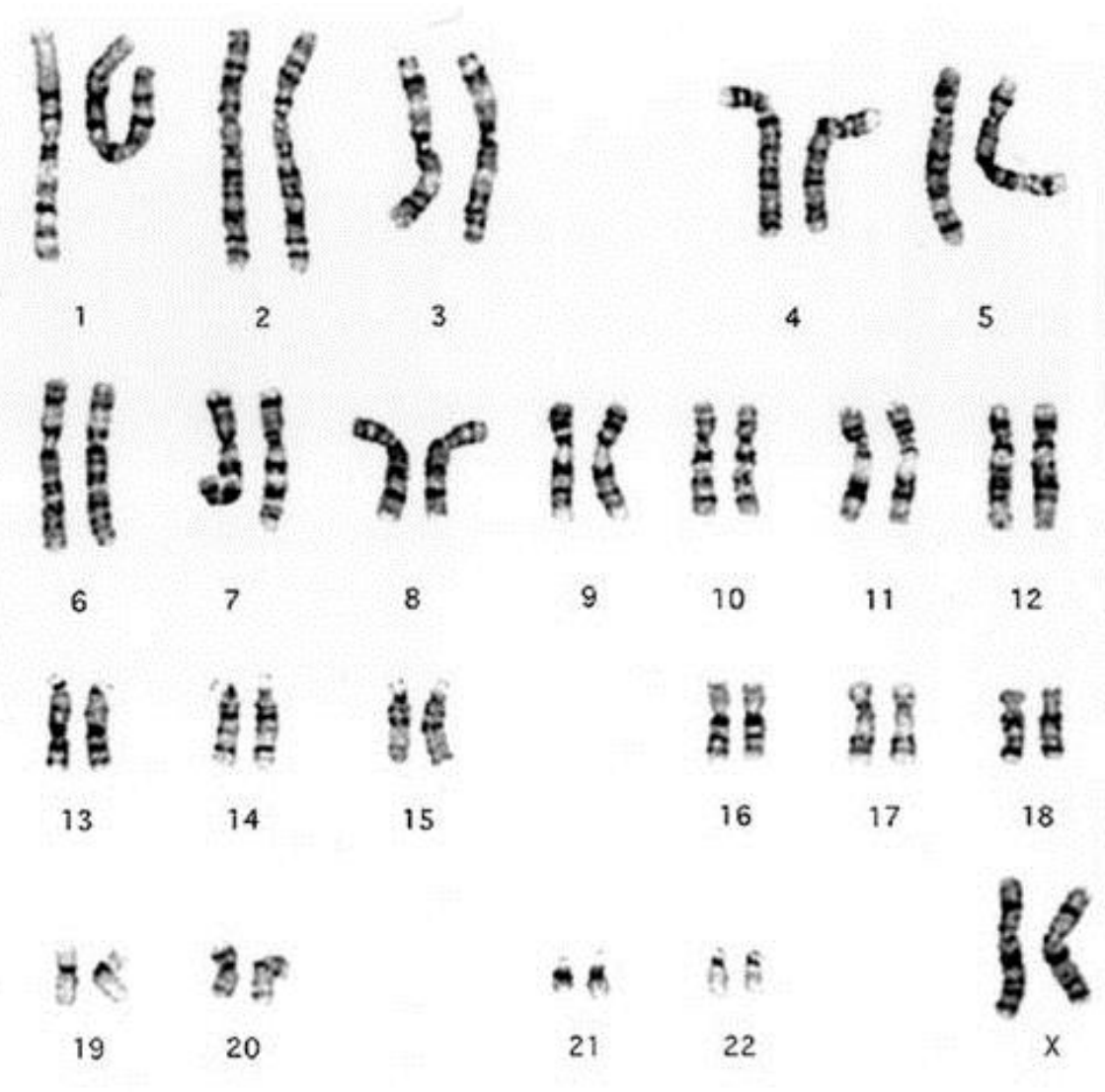




Basics of Genetics

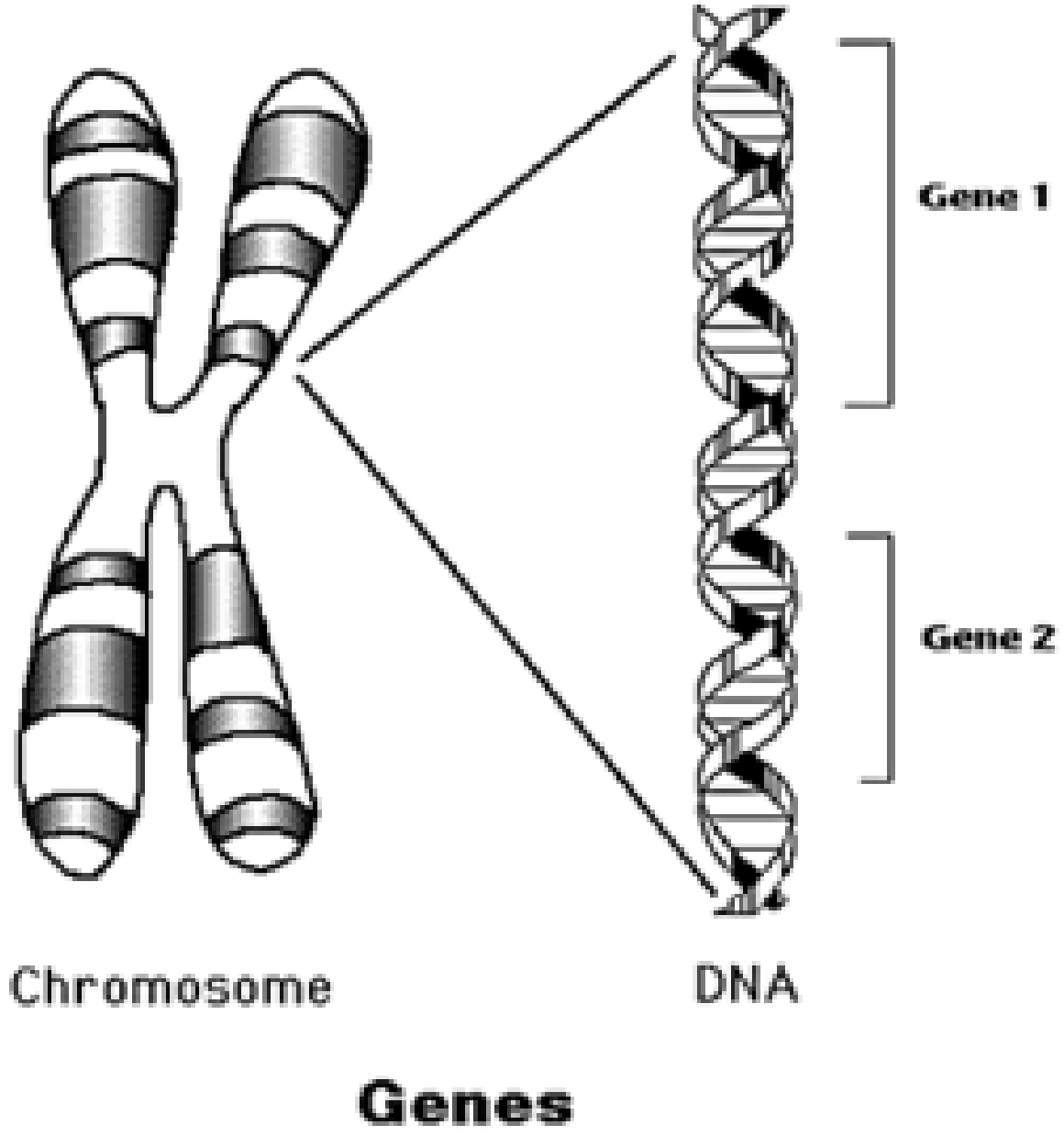
Homologous chromosomes

- pairs of chromosomes, one from each parent (23 pair in humans), with similar length, gene position, centromere location
- Autosomes are chromosomes # 1 - 22
- Sex chromosomes are X and Y (in humans)



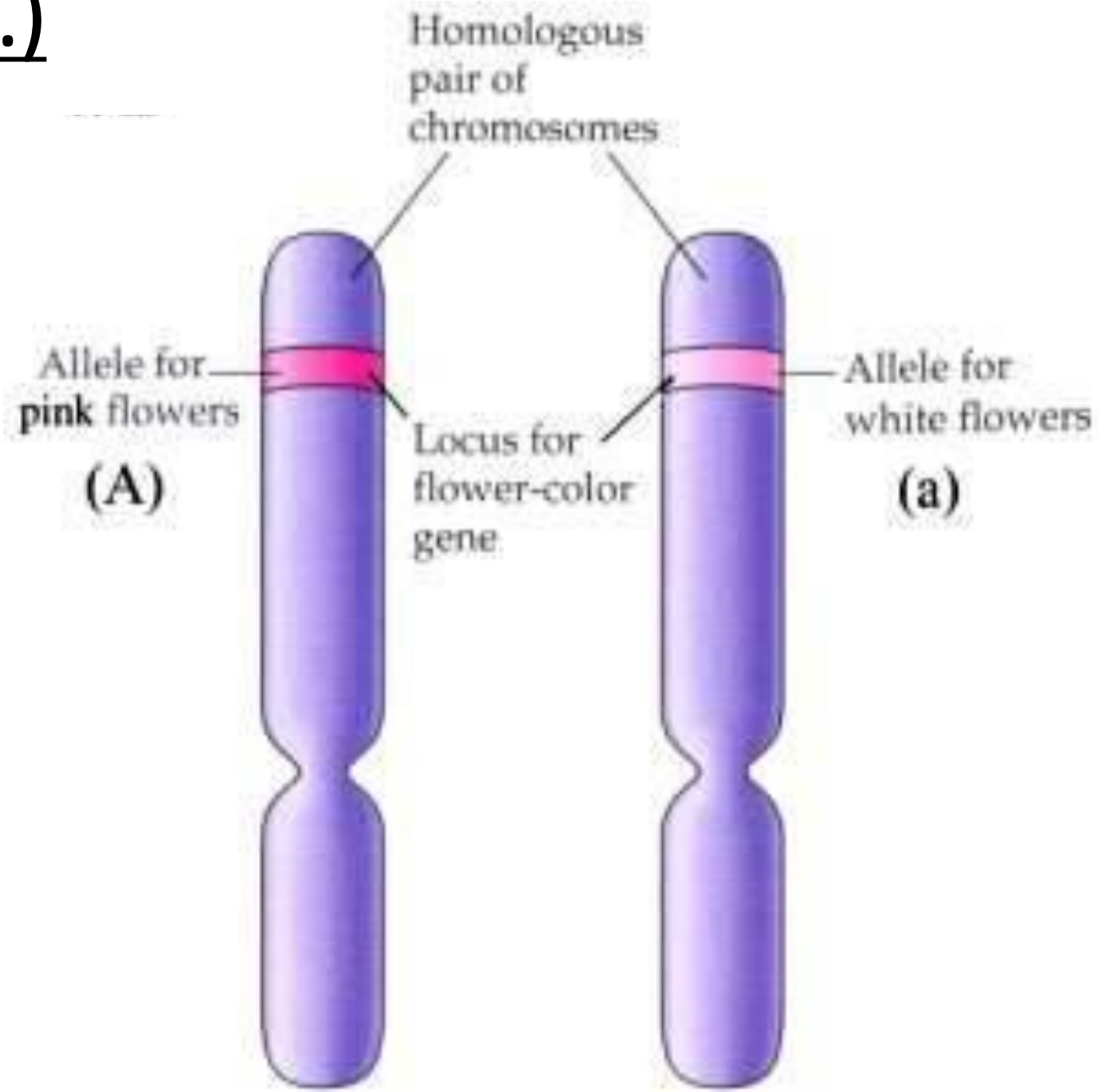
Basics of Genetics (cont.)

Gene - a sequence of DNA that codes for making certain protein which determines traits



Basics of Genetics (cont.)

Allele - different forms of a gene, one from each parent (ex. Tall / Short in pea plants)



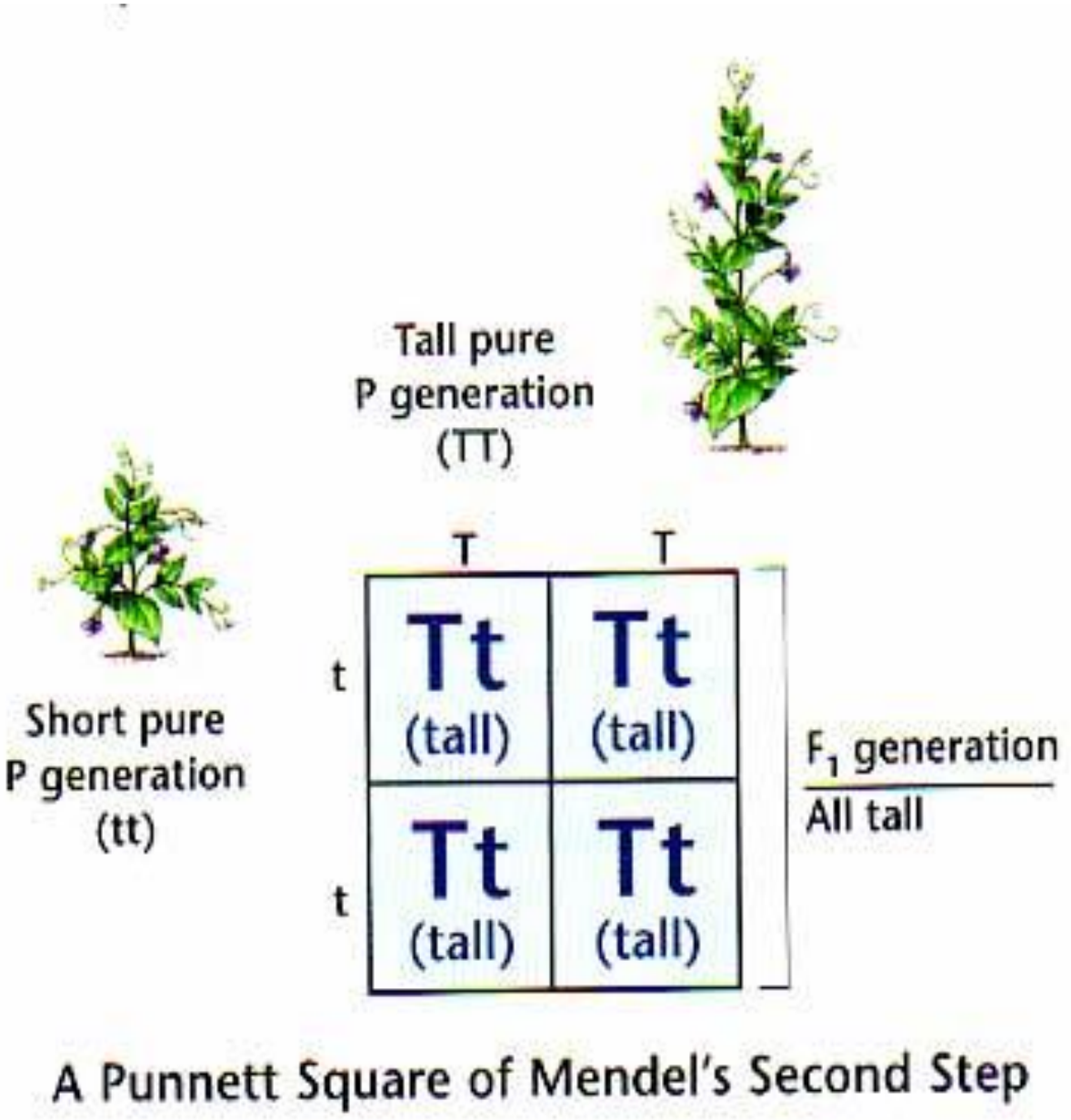
Principle of Dominance

Dominant

- allele that is expressed with 1 copy of the gene
- **CAPITAL** letter

Recessive

- requires 2 copies of the gene to be expressed
- **lower case** letter



A Punnett Square of Mendel's Second Step

Genotype

- genetic makeup of an organism
- represented by combination of capital and/or lower case letters

Phenotype

- Physical appearance of an organism

Ex. genotypes & phenotypes of height in pea plants

TT (pure or homozygous dominant) = Tall

Tt (hybrid or heterozygous) = Tall

tt (pure or homozygous recessive) = short

Dominance

- Example - height in pea plants
- 1st cross: homozygous tall (TT) and short (tt)

Genotypes:

0 % homozygous dominant (pure, TT)

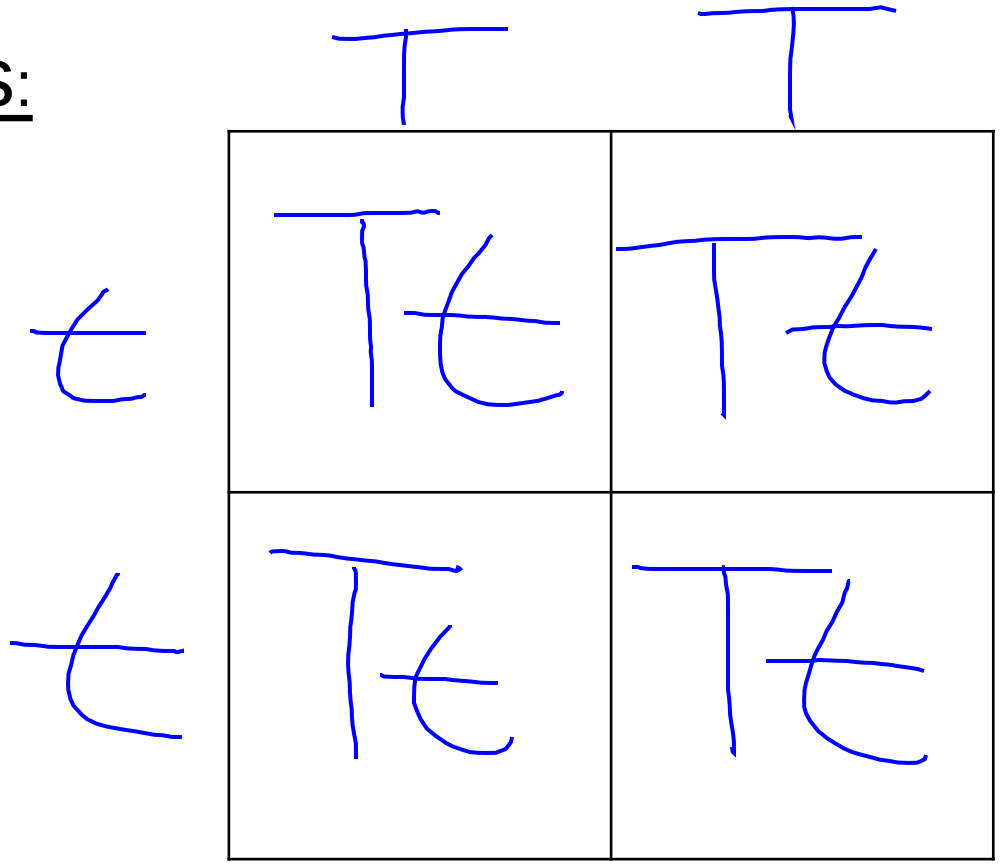
0 % homozygous recessive (tt)

100 % heterozygous

Phenotypes:

100 % Tall

0 % Short



2nd cross: Both Heterozygous Tall (Tt)

Genotypes:

25 % homozygous dominant

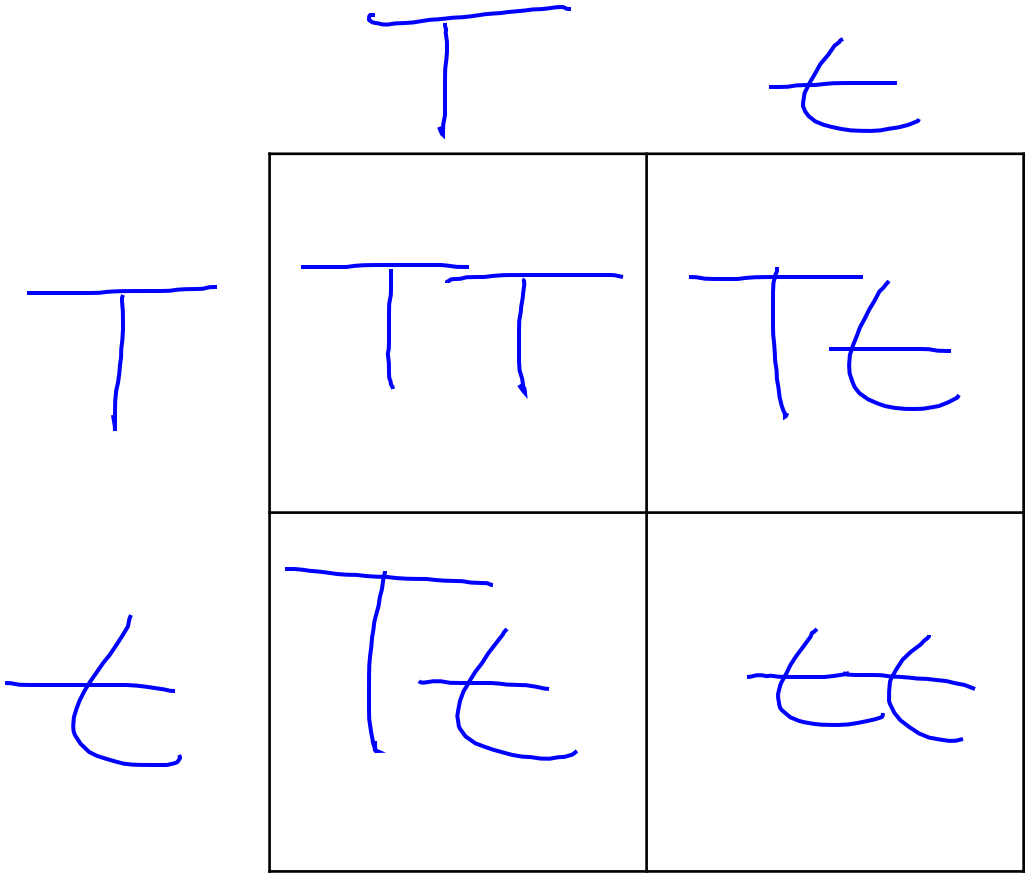
25 % homozygous recessive

50 % heterozygous

Phenotypes:

75 % Tall

25 % Short



Check Your Work!

In a certain species of animal, black fur (B) is dominant over brown fur (b). Using the following Punnett square, predict the genotypes and phenotypes of the offspring whose parents are both Bb or have heterozygous black fur.

	B	b
B	BB	Bb
b	Bb	bb

Genotypes:
25 % homozygous black fur (BB)
50 % heterozygous black fur (Bb)
25 % homozygous brown fur (bb)

Phenotypes:
75 % black fur
25 % brown fur

Check Your Work!

Now do the same when one parent is homozygous black and the other is homozygous brown.

	B	B
b	Bb	Bb
b	Bb	Bb

Genotypes:
- 0 % homozygous black fur (BB)
- 100 % heterozygous black fur (Bb)
- 0 % homozygous brown fur (bb)

Phenotypes:
- 100 % black fur
- 0 % brown fur

Check Your Work!

Repeat this process again when one parent is heterozygous black and the other is homozygous brown.

	B	b
b	Bb	bb
b	Bb	bb

Genotypes:
0 % homozygous black fur (BB)
50 % heterozygous black fur (Bb)
50 % homozygous brown fur (bb)

Phenotypes:
50 % black fur
50 % brown fur

Lesson 2

Intermediate Inheritance

Incomplete Dominance

Codominance

Multiple Alleles (blood typing)

Intermediate Inheritance

- traits are not clearly dominant or recessive

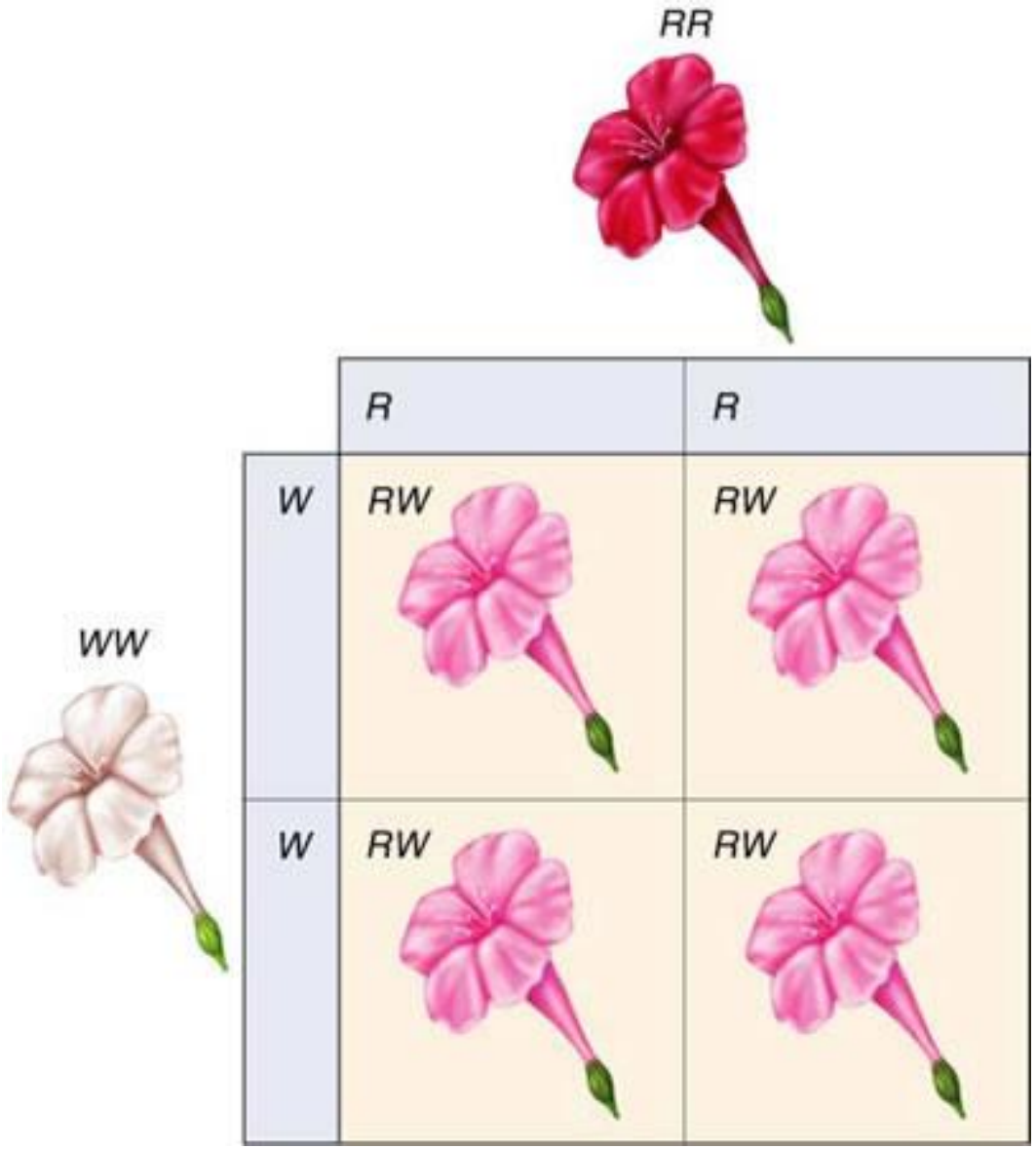
1. Incomplete Dominance

- exhibits a phenotype in between both parents

ex. If a pure white parent (WW) flower is crossed with a pure red parent (RR)…

pink (RW) offspring result!

- (Four o'clock flowers)



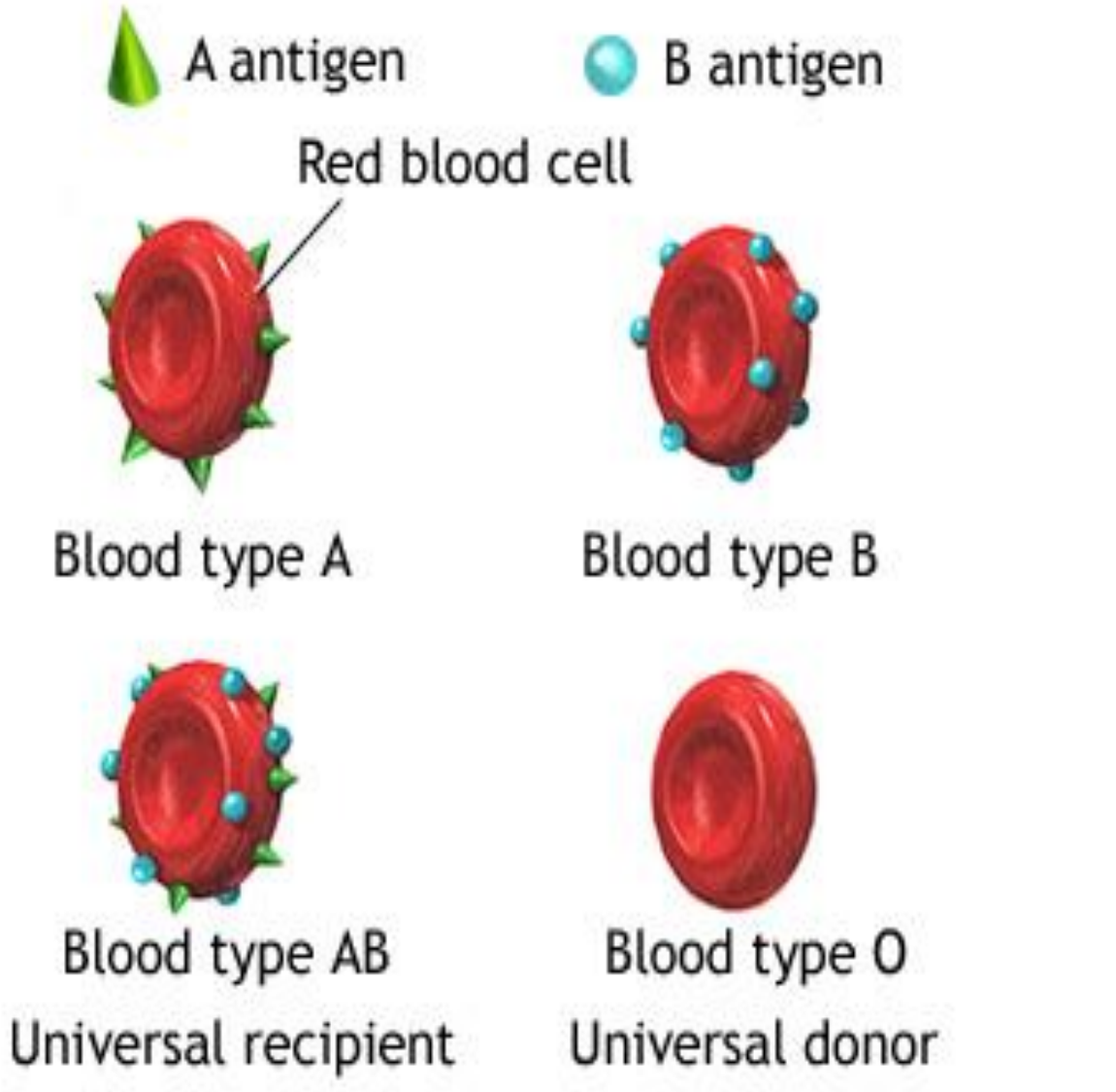
2. Co-dominance

- both alleles are expressed
- ex: Coat color in roan cattle (both red and white hairs)

3. Multiple Alleles

3 or more alleles of the same gene exist

- ex. A, B, and O alleles
- type A and B are co-dominant
- type O is recessive to both A and B



Multiple Alleles (cont.)

Crosses:

Type A and Type B (homozygous)

	A	A
B	AB	AB
B	AB	AB

Phenotypes:

0 % O

0 % A

0 % B

100 % AB

Type A and B (hybrid/heterozygous)

	A	O
B	AB	BO
O	AO	OO

Phenotypes:

25 % O

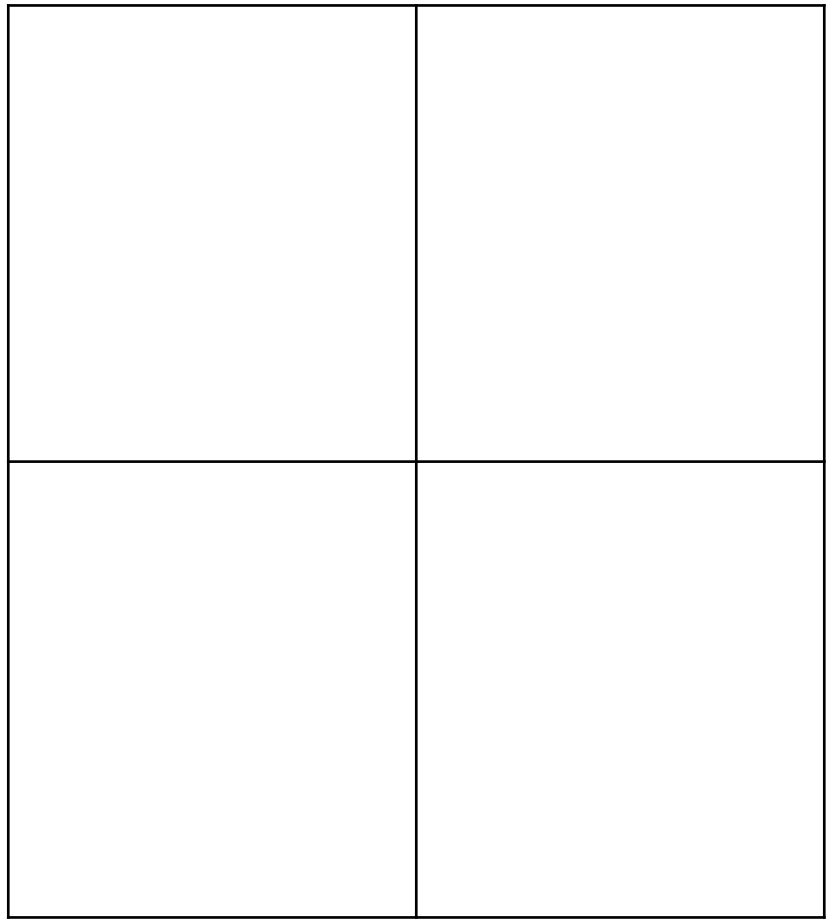
25 % A

25 % B

25 % AB

Worksheet Problem #1

Father and Mother Type O



Phenotypes:

100 % O

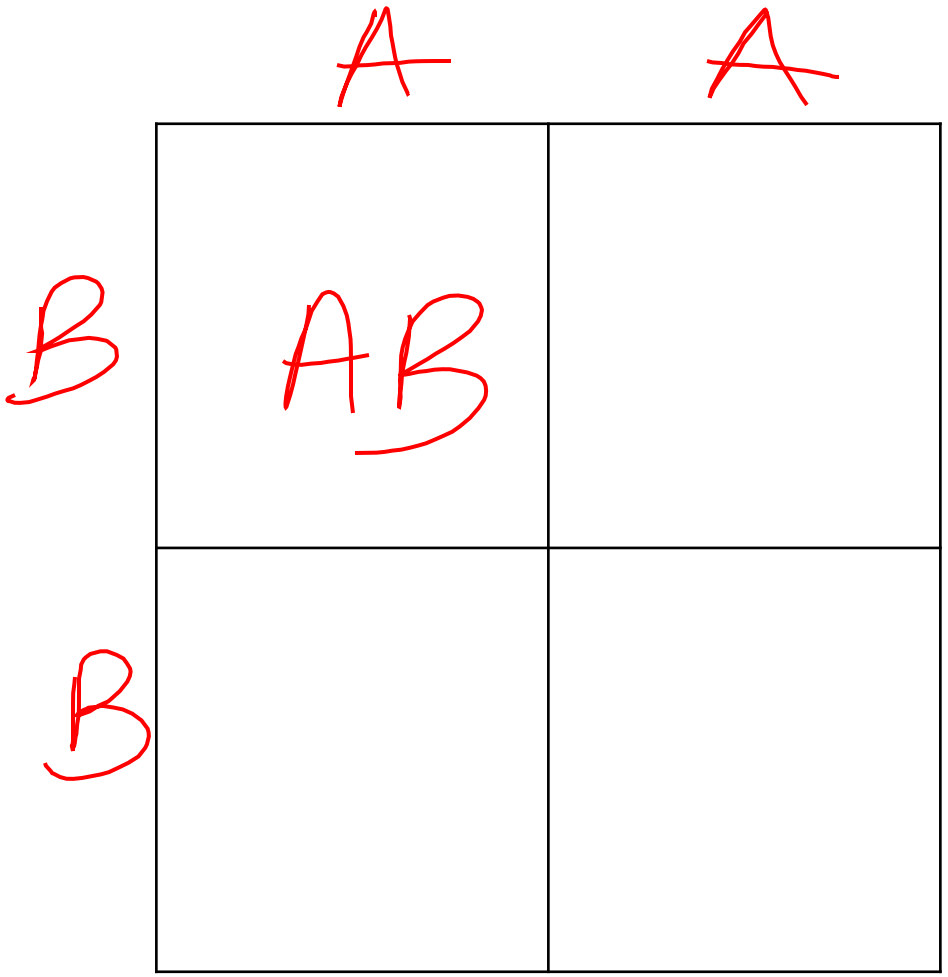
0 % A

0 % B

0 % AB

Worksheet Problem #2

Father A (homozygous) Mother B (homozygous)



Phenotypes:

_____ % O

_____ % A

_____ % B

100 % AB

Worksheet Problem #3

Father A (heterozygous) Mother B (heterozygous)

	A	O
B	AB	BO
O	AO	OO

Phenotypes:

25 % A

_____ % B

_____ % AB

_____ % O

Worksheet Problem #4

Father O, Mother AB

	O	O
A	AO	AO
B	BO	BO

Phenotypes:

50 % A

50 % B

% AB

% O

Worksheet Problem #5

Father AB and Mother AB

	A	B
A	AA	AB
B	AB	BB

Phenotypes:

0 % O

25 % A

25 % B

50 % AB

Practice Punnet Square Word Problems

1) Let's say that in seals, the gene for the length of the whiskers has two alleles. The dominant allele (W) codes long whiskers & the recessive allele (w) codes for short whiskers.

a) What percentage of offspring would be expected to have short whiskers from the cross of two long-whiskered seals, one that is homozygous dominant and one that is heterozygous?

	W	W
W	WW	WW
w	W w	W w

50% Homozygous dominant

50% Heterozygous

100% long-whiskers

0% short-whiskers

b) If one parent seal is pure long whiskered and the other is short whiskered, what percent of offspring would have short whiskers?

	W	W
w	W w	W w
w	W w	W w

100% Heterozygous

100% long-whiskers

0% short-whiskers

2) In purple people eaters, one-horn is dominant and no horns is recessive. Draw a Punnet Square showing the cross of a purple people eater that is hybrid for horns with a purple people eater that does not have horns. Summarize the genotypes & phenotypes of the possible offspring.

	H	**h**
h	**H h**	**h h**
h	**H h**	**h h**

50% Heterozygous

50% Homozygous recessive

50% horns

50% no horns

I^A = A allele

I^B = B allele

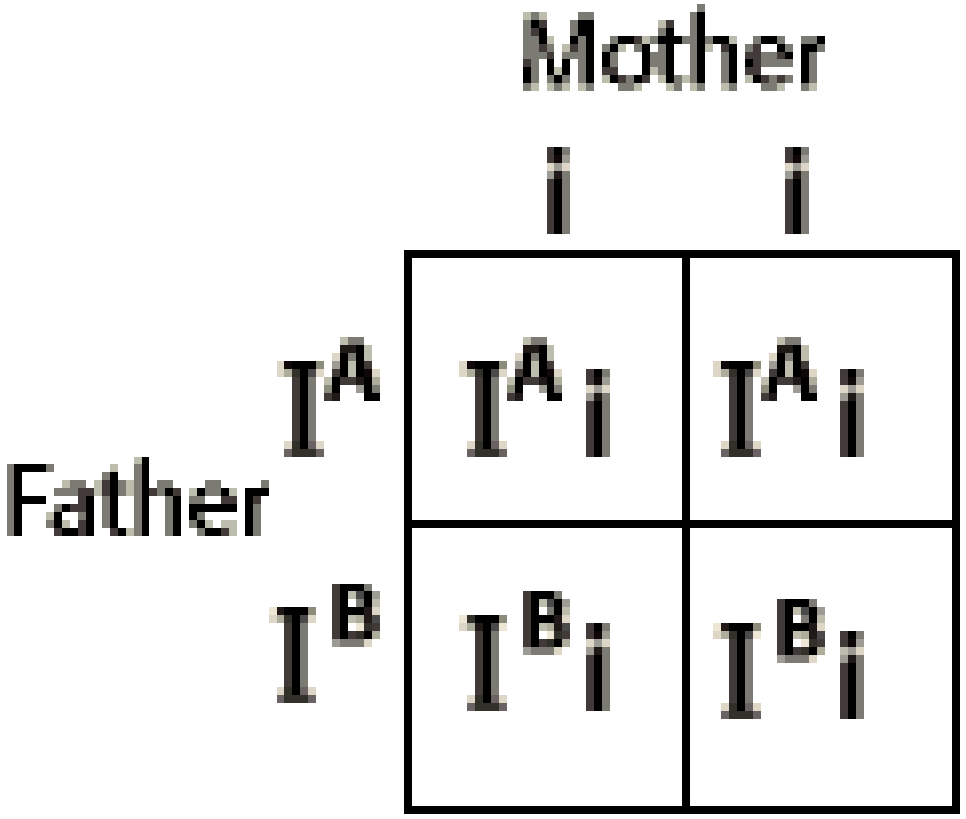
i = O allele

What blood type is the father?

Type AB

What blood type is the mother?

Type O



Half of the children predicted to be **Type A**, and half **Type B**.

Lesson 3

- DNA structure

Oh, we love DNA, made of Nucleotides
Phosphate, Sugar and a Base bonded
down the side!

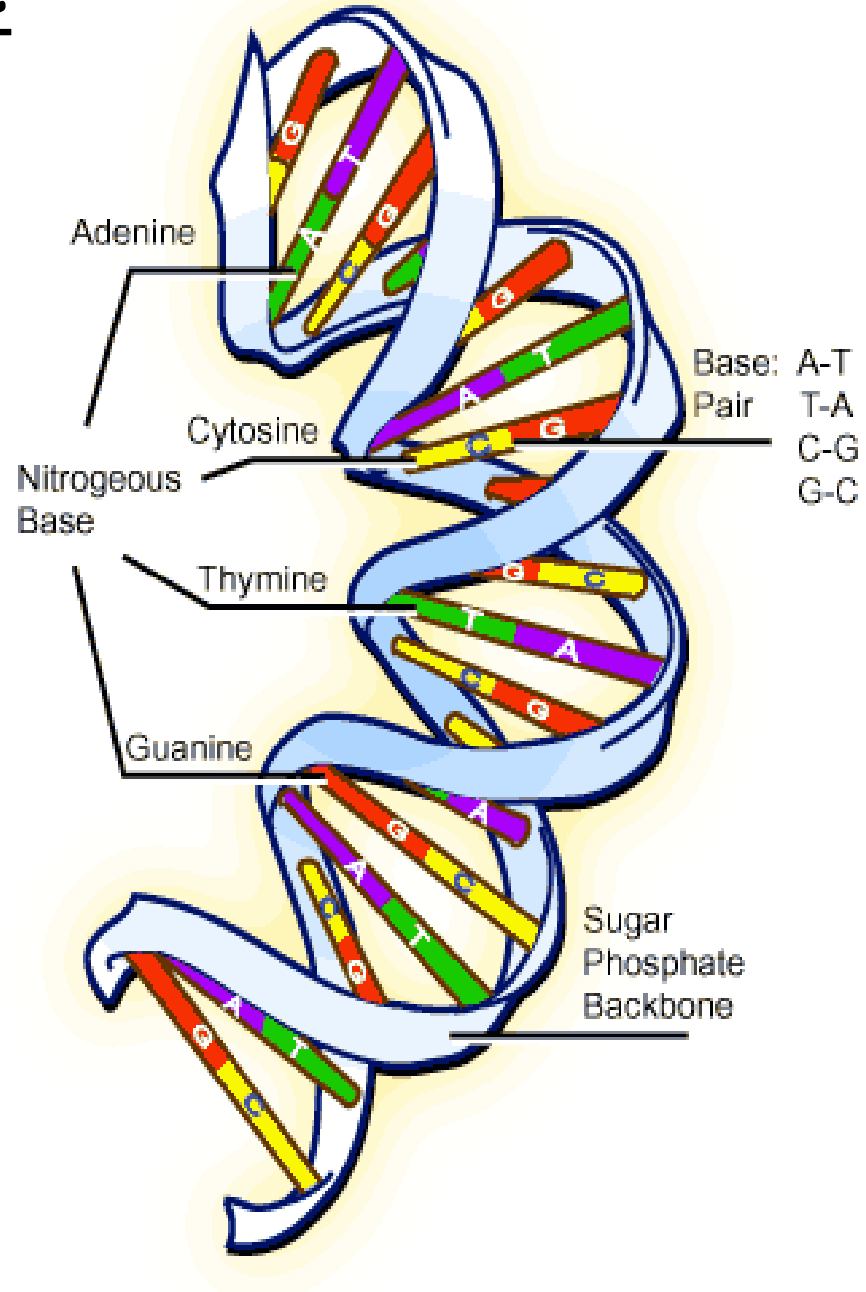
Adenine and Thymine make a lovely pair,
Cytosine without Guanine would feel very bare,

Shaped like a twisted ladder,
it controls your traits,
Watson and Crick discovered it,
DNA is GREAT!!!

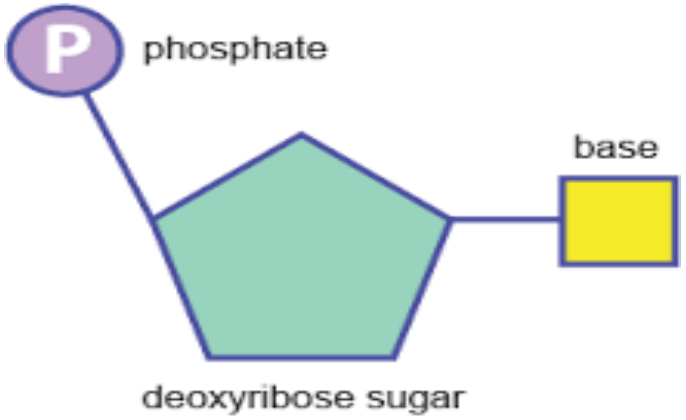
DNA Structure

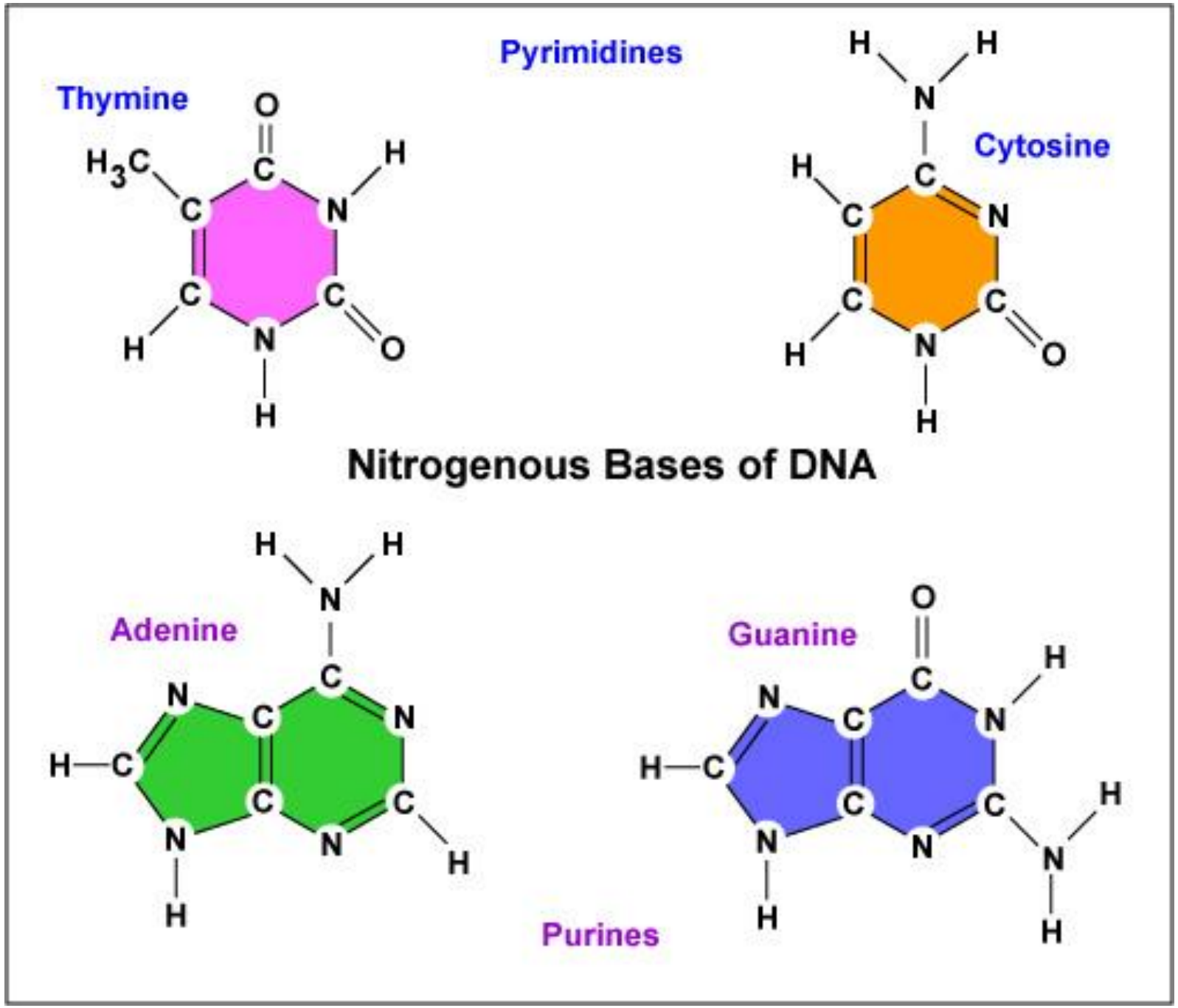
Deoxyribo**n**ucleic **A**cid

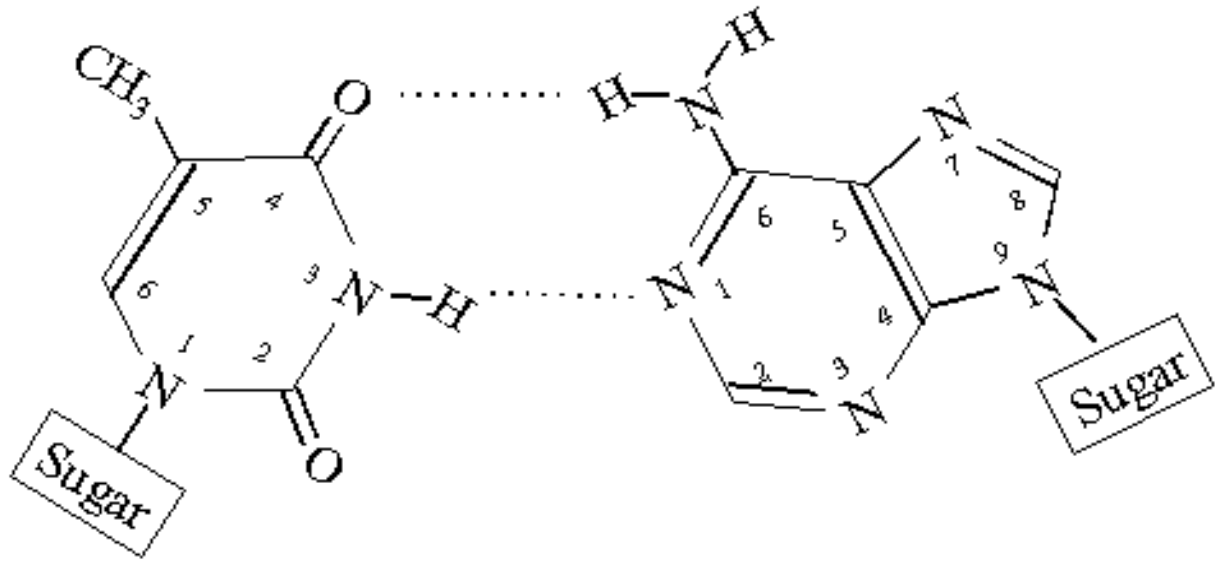
- genetic material passed from generation to generation
- found in the nucleus of cells (mitochondria & chloroplasts too)
- consists of thousands of smaller repeating units called nucleotides



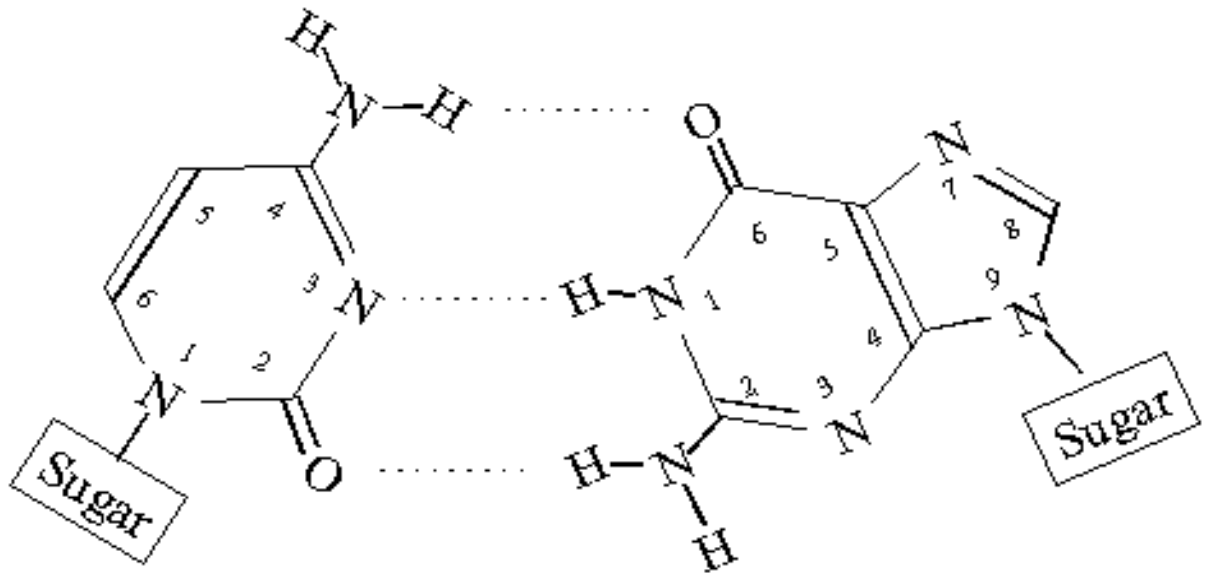
- A nucleotide is composed of 3 parts:
  - phosphate group made up of elements O, H, P
  - deoxyribose (5 carbon sugar) made up of C, H, O
  - Nitrogenous base made up of C, H, O, N
    - **4 different nitrogenous bases:**
      - **Adenine (A) pairs with Thymine (T)**
      - **Guanine (G) pairs with Cytosine (C)**




Pyrimidines
Thymine
Cytosine
Nitrogenous Bases of DNA
Adenine
Guanine
Purines



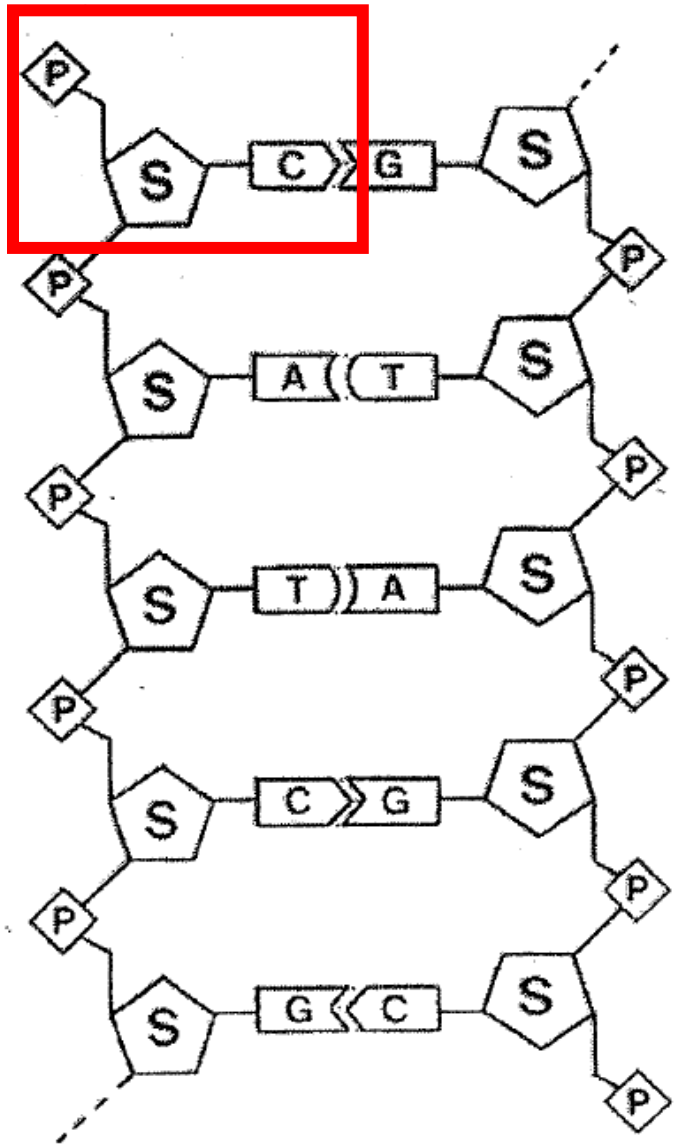
Thymine (T) Adenine (A)



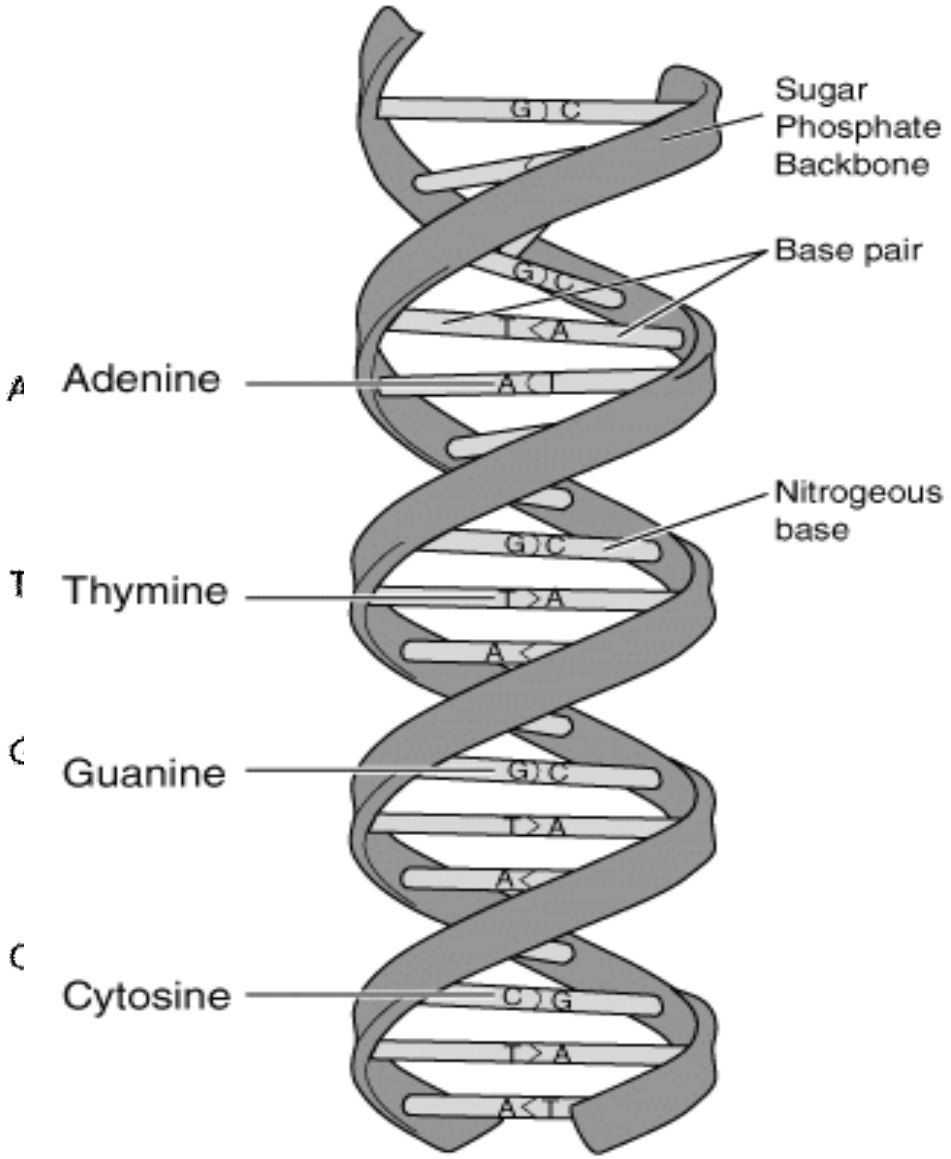
Cytosine (C) Guanine (G)

DNA Structure

1 nucleotide

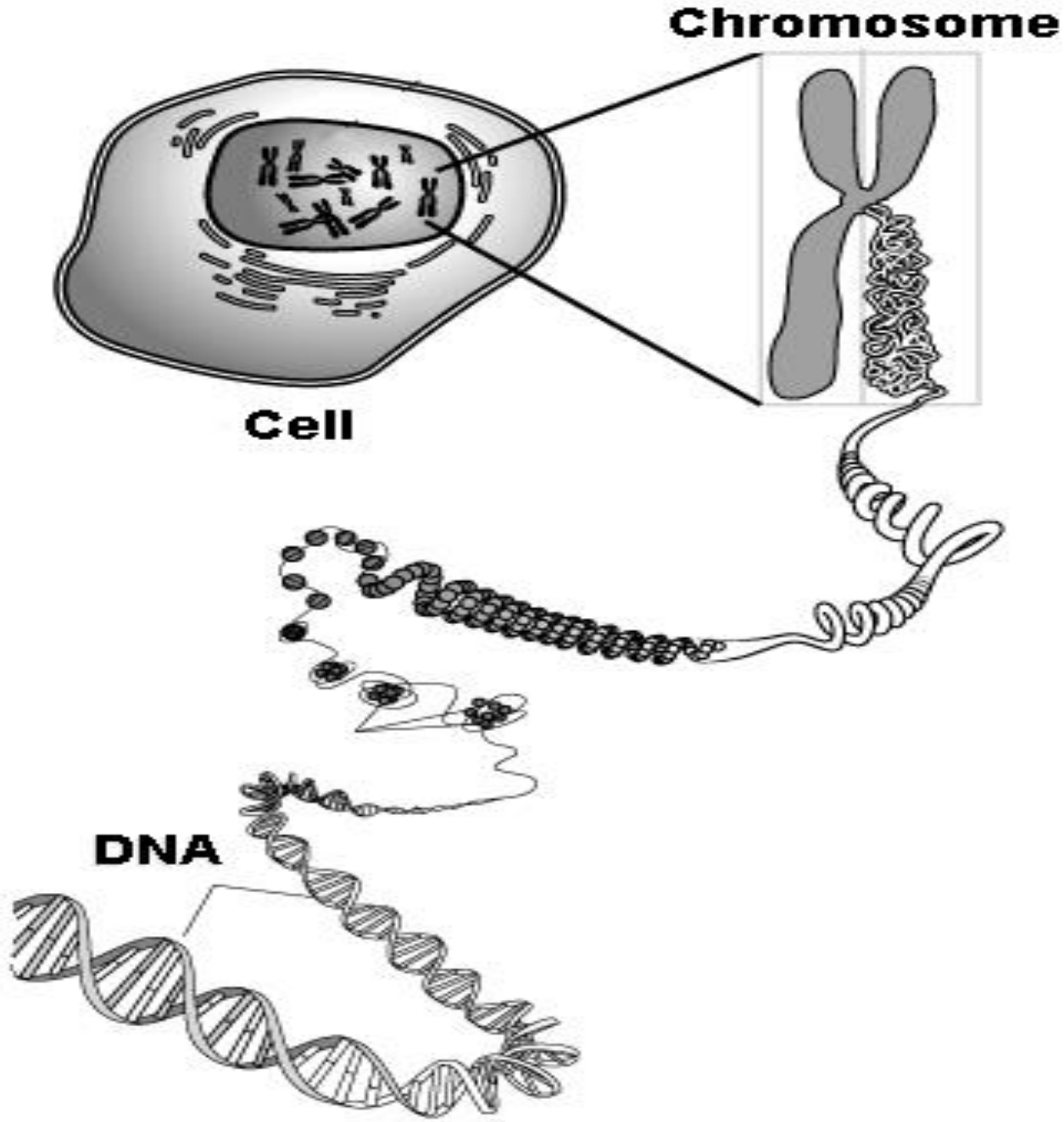


(untwisted diagram of DNA)

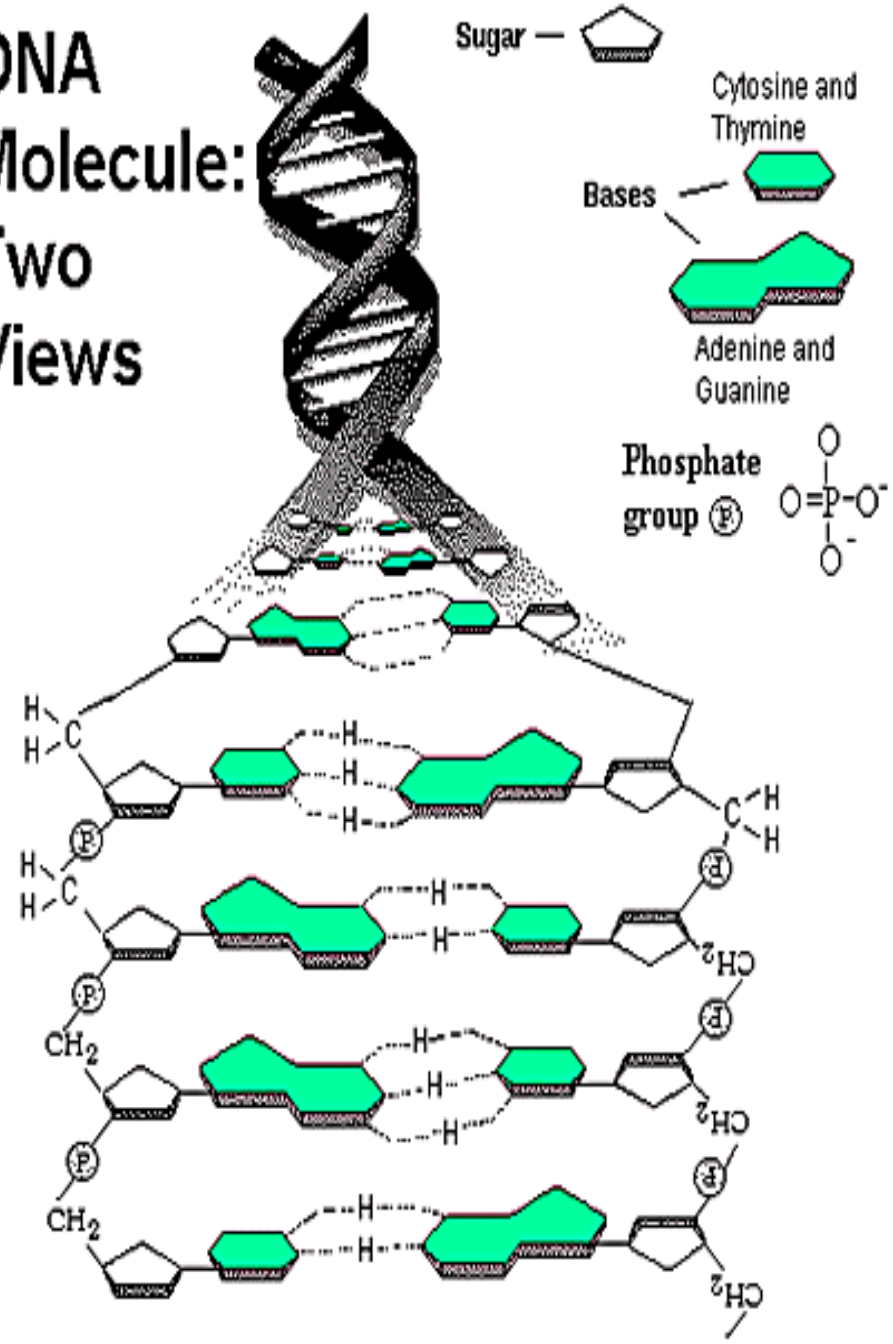


The Double Helix

- Discovered by James Watson and Frances Crick in 1953
- 2 complementary strands of nucleotides
- forms a twisted “ladder” shape
- “ladder” rungs are made of nitrogenous base pairs
- held together by weak hydrogen bonds



DNA Molecule: Two Views



Chromatin and Condensed Chromosome Structure

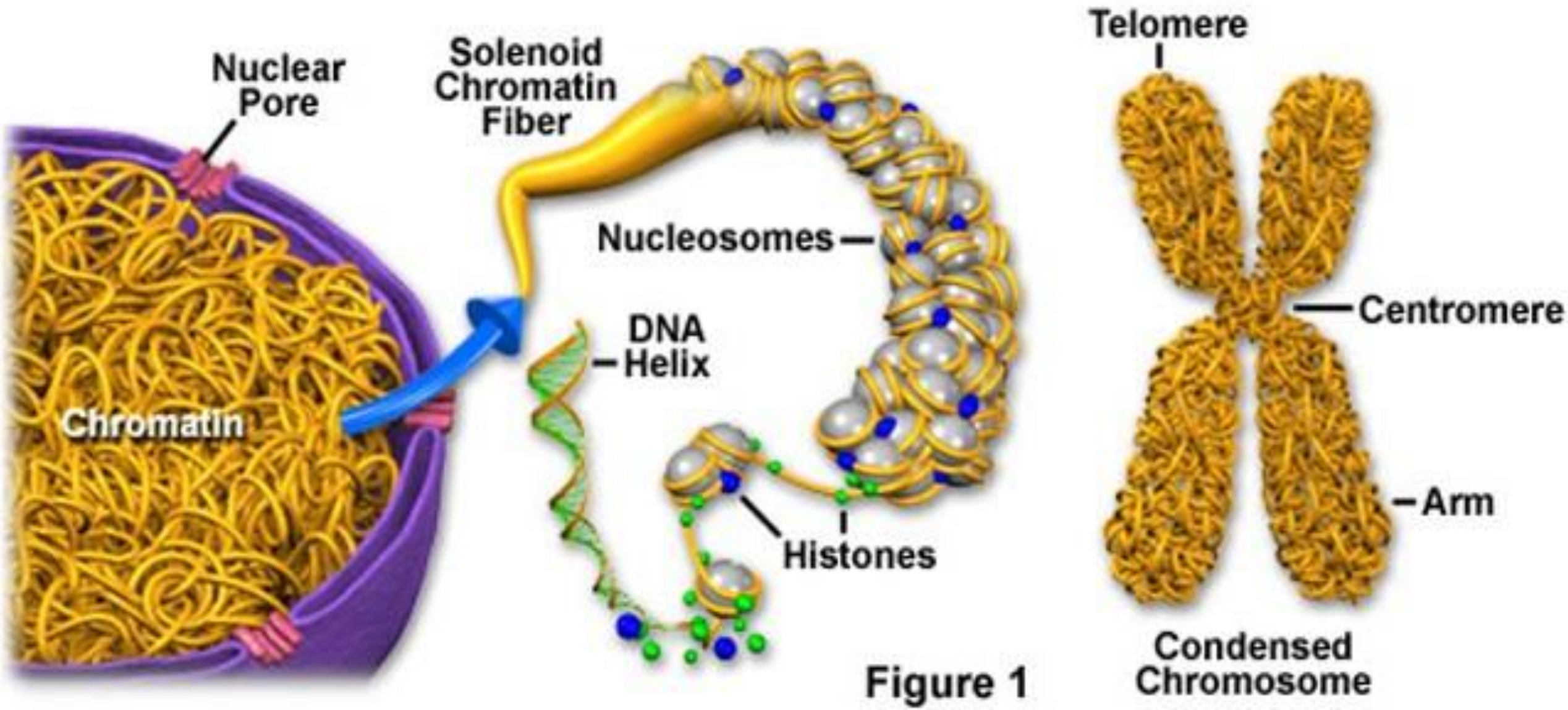
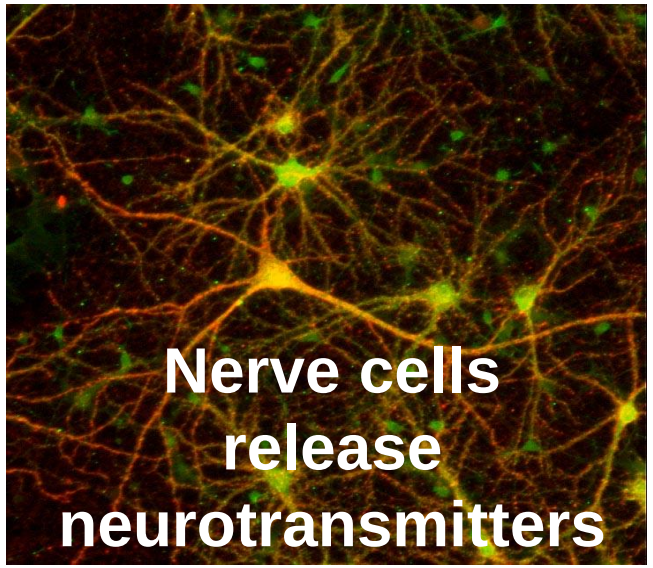
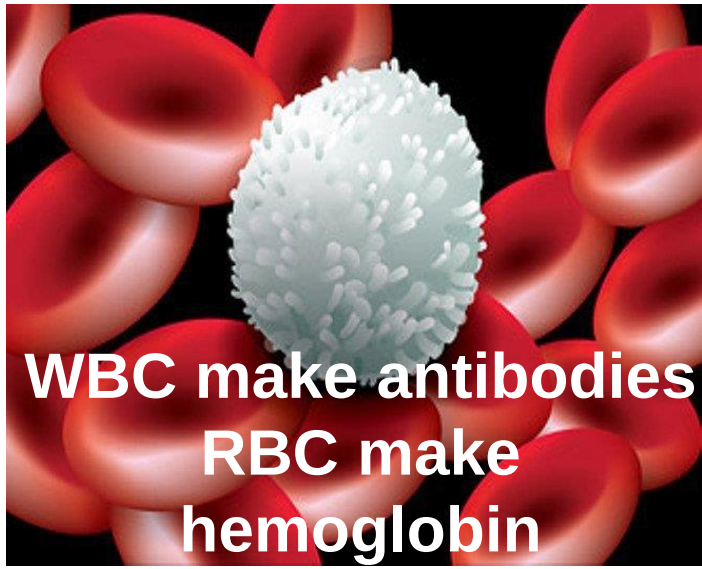


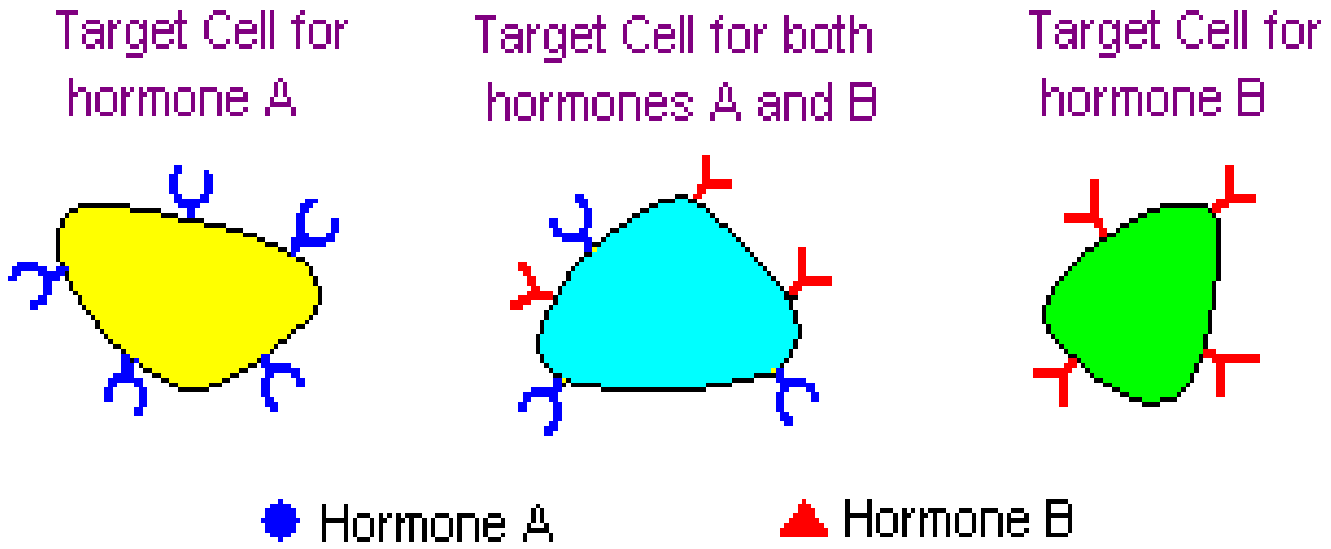
Figure 1

How do cells with identical DNA function so differently?



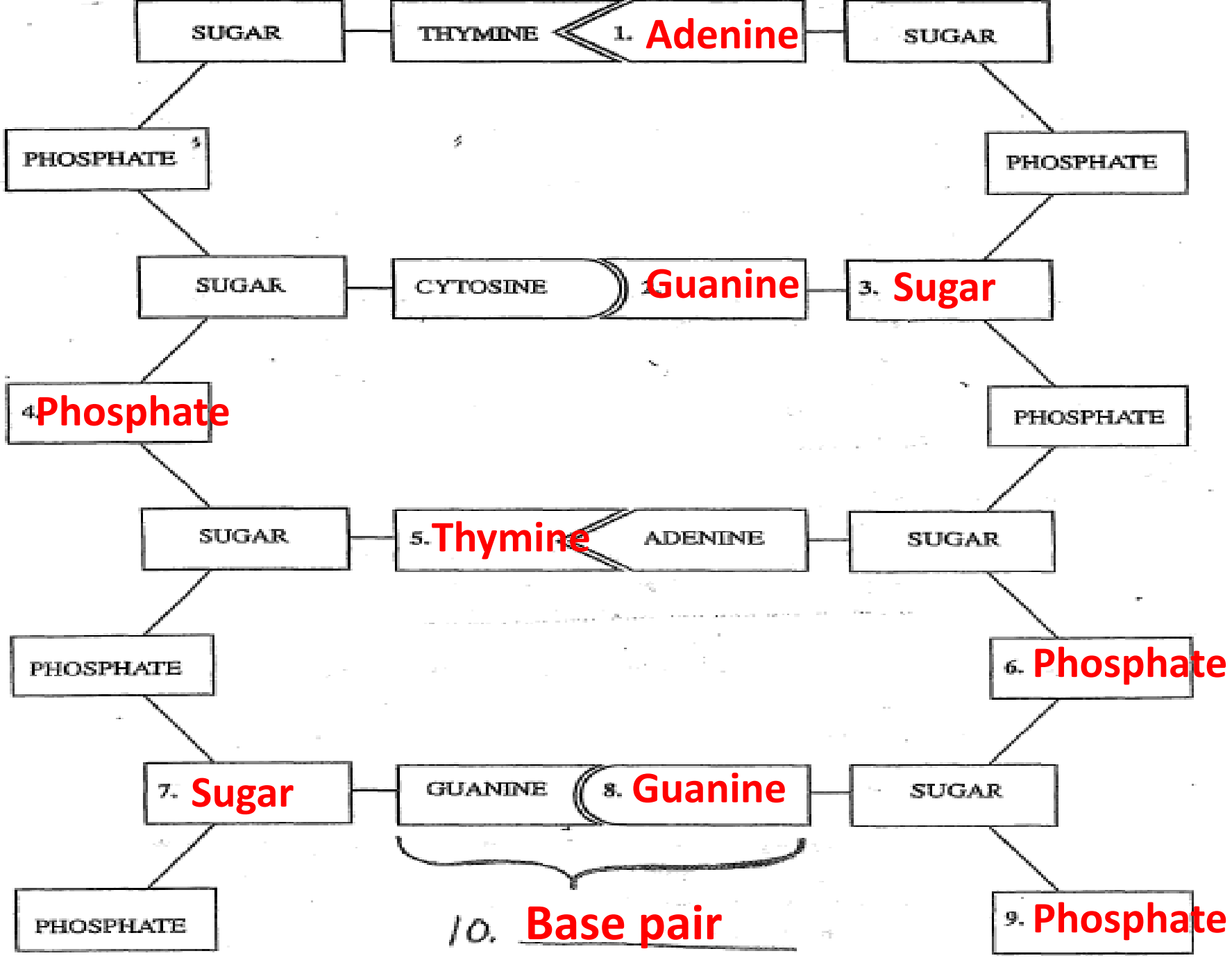
Receptor molecules & hormones detected





How do cells with identical DNA function so differently?

- **Cells only use (express) certain parts of the DNA code**
- **Cells are able to "turn off" genes they don't need to use**


SUGAR
THYMINE
1. Adenine
SUGAR
PHOSPHATE
PHOSPHATE
SUGAR
CYTOSINE
2. Guanine
3. Sugar
4. Phosphate
PHOSPHATE
SUGAR
5. Thymine
ADENINE
SUGAR
PHOSPHATE
6. Phosphate
7. Sugar
GUANINE
8. Guanine
SUGAR
PHOSPHATE
10. Base pair
9. Phosphate

DNA Practice Questions

b

1. Which type of compound is found in every DNA molecule?
   a. Starch
   b. Nitrogenous base
   c. Lipid
   d. Amino acid

c

2. The "ribo" part in the name deoxyribonucleic acid refers to the
   a. Rungs of the spiral ladder
   b. Bonds that hold two strands together
   c. Sugar component of DNA
   d. Type of helical arrangement

c

3. A molecular group consisting of a sugar molecule, a phosphate group, and a nitrogen base is a
   a. Nucleoprotein
   b. Nucleic acid
   c. Nucleotide
   d. Nucleolus

c

4. A nucleotide of DNA could contain
   a. Adenine, ribose and phosphate
   b. Nitrogenous base, phosphate and glucose
   c. Phosphate, deoxyribose and thymine
   d. Uracil, deoxyribose and phosphate

a

5. A nucleotide would *least* likely contain the element
   a. Sulfur
   b. Carbon
   c. Nitrogen
   d. Phosphorous

d

6. In a DNA molecule, a base pair normally could be composed of
   a. Adenine and guanine
   b. Adenine and cytosine
   c. Thymine and guanine
   d. Guanine and cytosine

Lesson 4

- DNA replication

When does DNA “unzip”?

1) When replicating before cell division (mitosis/meiosis)
2) When providing the instructions for creating a protein in the cell (protein synthesis)

DNA Replication Animation

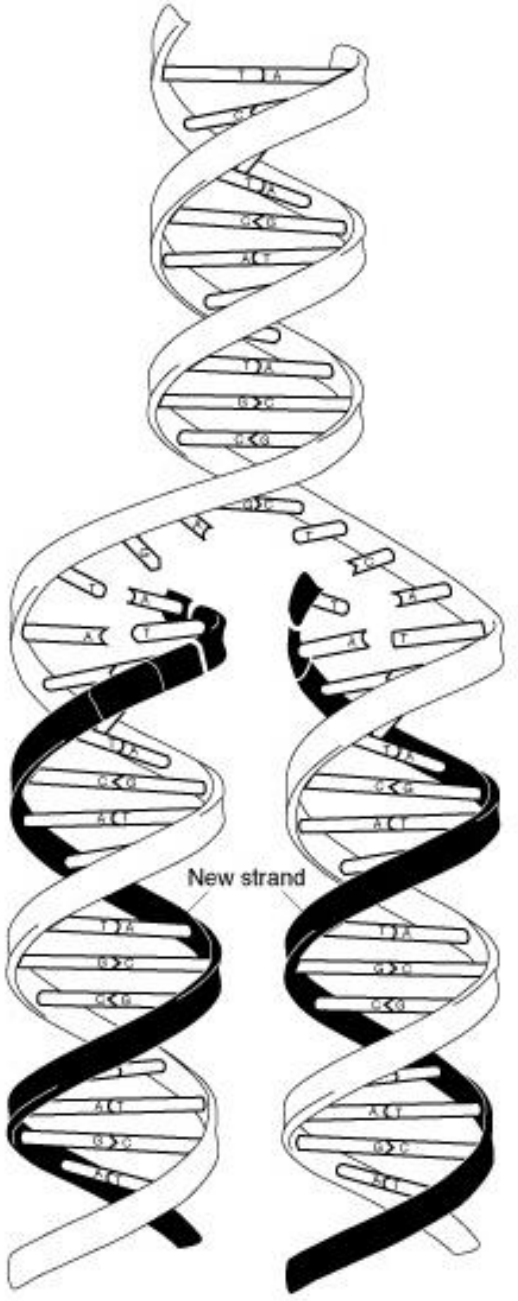
Replication Steps:

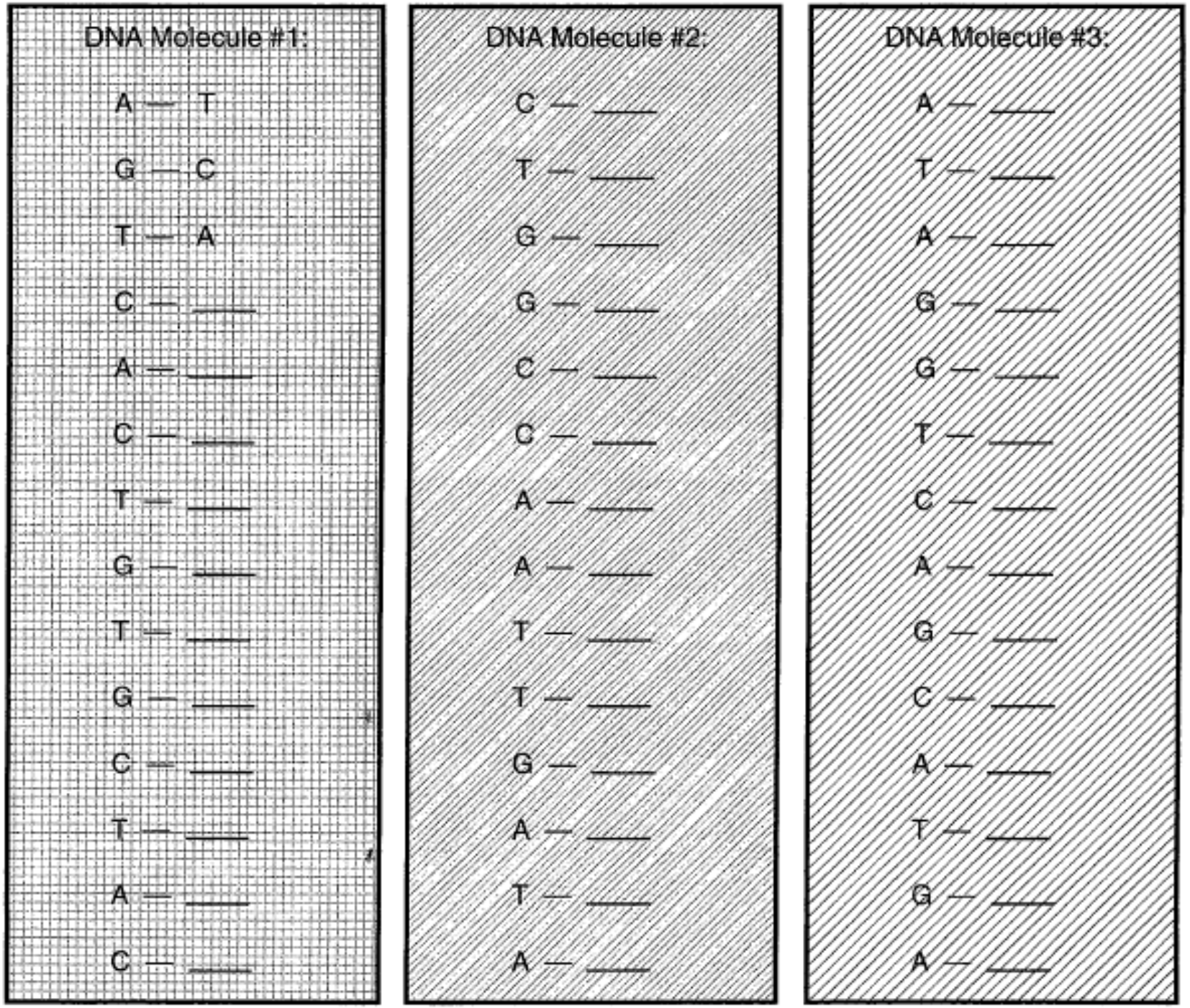
1) DNA unwinds & “unzips” w/ enzyme Helicase (breaks weak hydrogen bonds between bases)

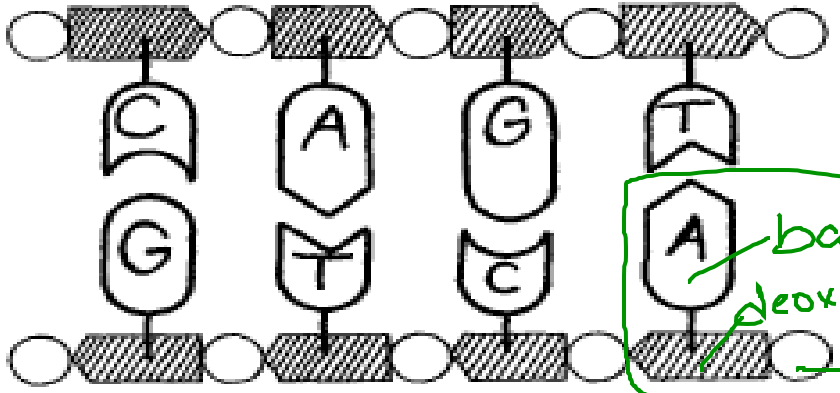
2) Free nucleotides from cytoplasm enter nucleus

3) They bond to their complementary bases on the unzipped DNA strands w/ help of enzyme DNA Polymerase

4) Produces 2 identical double stranded DNA molecules, each having 1 original strand and 1 new strand




DNA Molecule #1:
A — T
G — C
T — A
C — ____
A — ____
C — ____
T — ____
G — ____
T — ____
G — ____
C — ____
T — ____
A — ____
C — ____
DNA Molecule #2:
C — ____
T — ____
G — ____
G — ____
C — ____
C — ____
A — ____
A — ____
T — ____
T — ____
G — ____
A — ____
T — ____
A — ____
DNA Molecule #3:
A — ____
T — ____
A — ____
G — ____
G — ____
T — ____
C — ____
A — ____
G — ____
C — ____
A — ____
T — ____
G — ____
A — ____



A = Adenine

T = Thymine

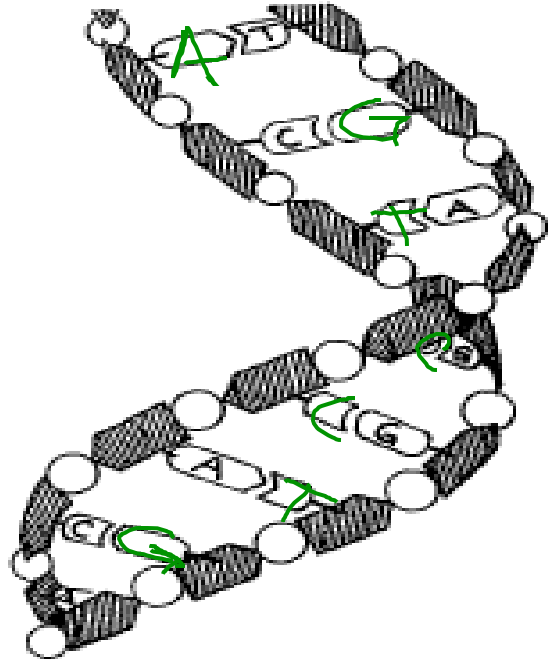
G = Guanine

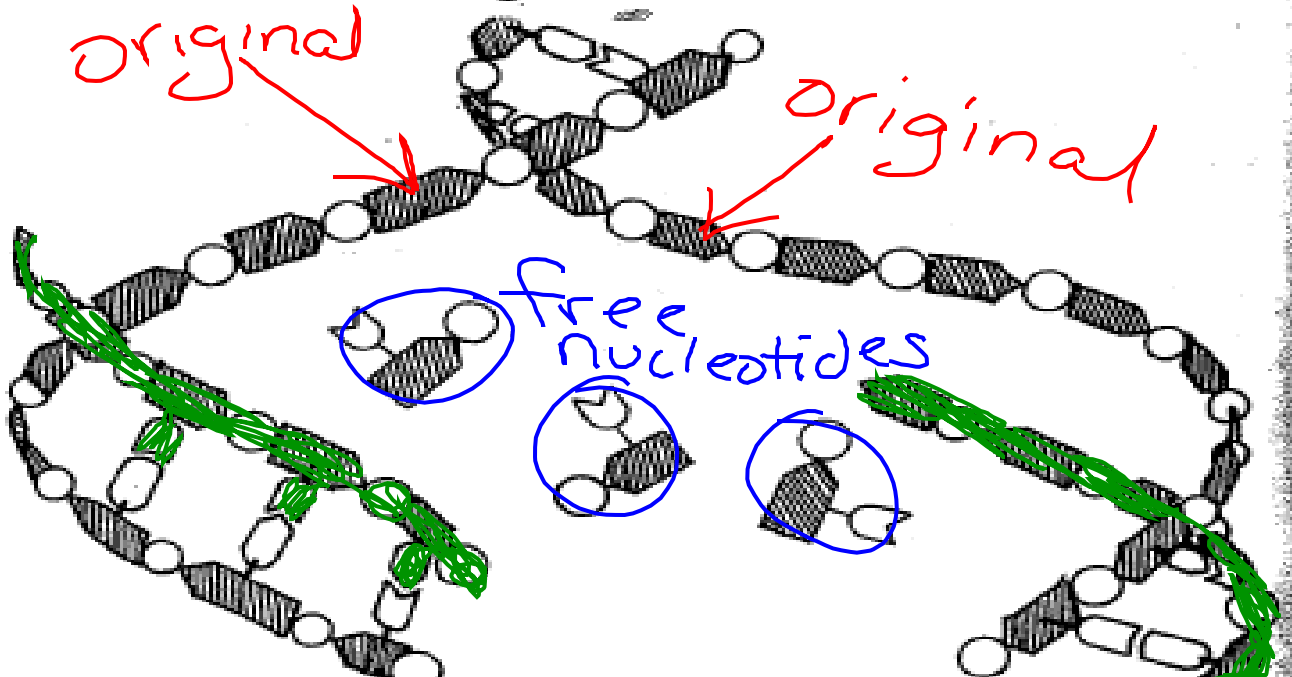
C = Cytosine

The DNA molecule has a double helix shape. Two strands of DNA are coiled around each other and attached by bonds between the nitrogenous bases of each chain. Adenine always bonds with thymine, and cytosine bonds with guanine.

In the illustration at the left below, label a phosphate and a deoxyribose sugar. Fill in the symbol for each base depending on its complementary base in the opposite strand.

The diagram at the right shows the replication of DNA. Fill in the symbol for each base. Label the original strand, a new strand and a free-floating nucleotide.





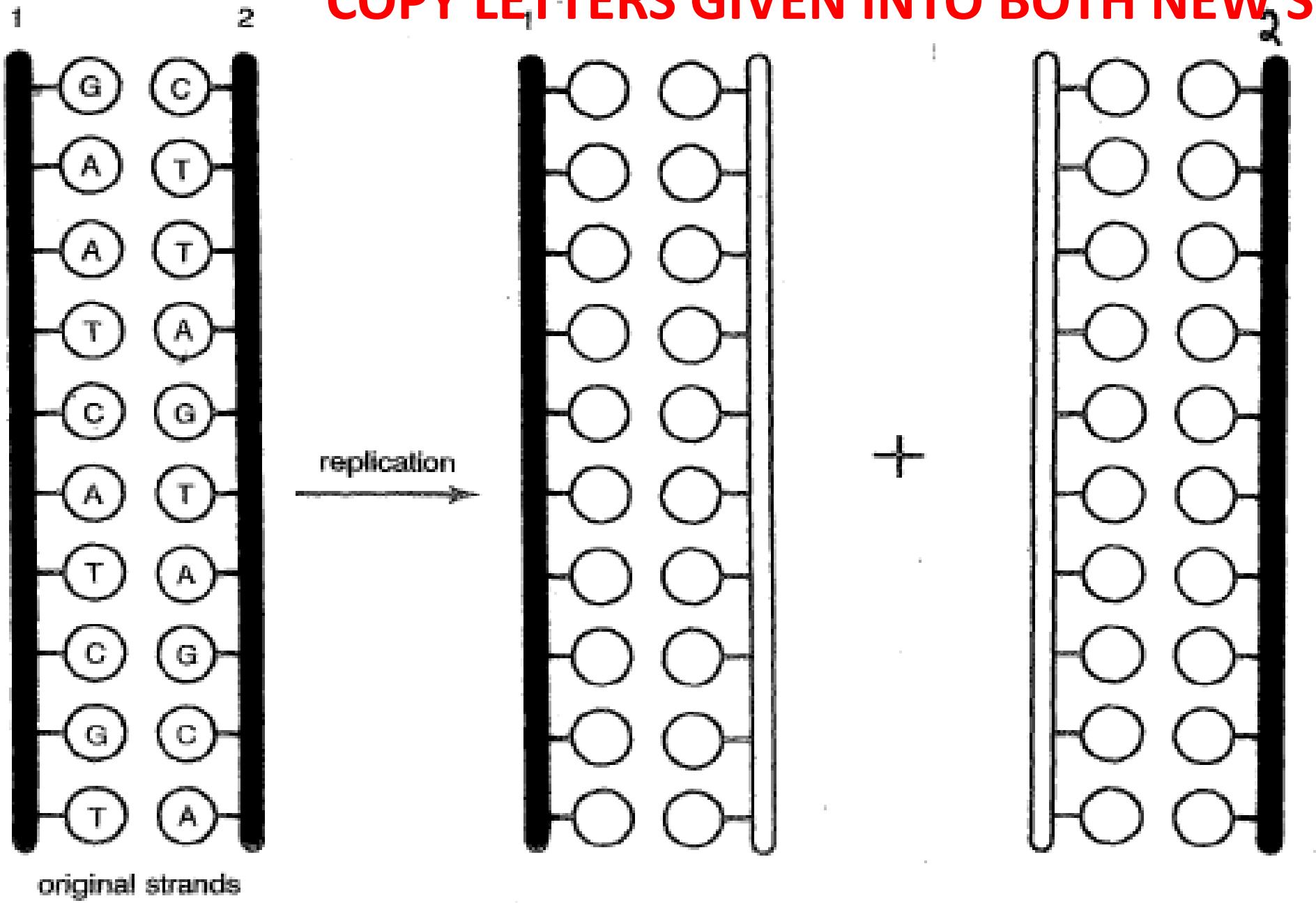
Fill in the blanks with the correct answers.

The structure of DNA was determined by Watson (person's name) and Crick (person's name).

They described the shape of the DNA molecule as a double helix.

After replication, two (number) identical molecules of DNA (name of molecule) are produced. A gene is a sequence of nucleotides (DNA building blocks) in a DNA molecule.

COPY LETTERS GIVEN INTO BOTH NEW STRANDS



QUESTIONS:

1- Which bonds are broken during DNA replication? hydrogen bonds

2- What determines the linear sequence of nucleotides in the new strands?

The sequence of bases in the original strand

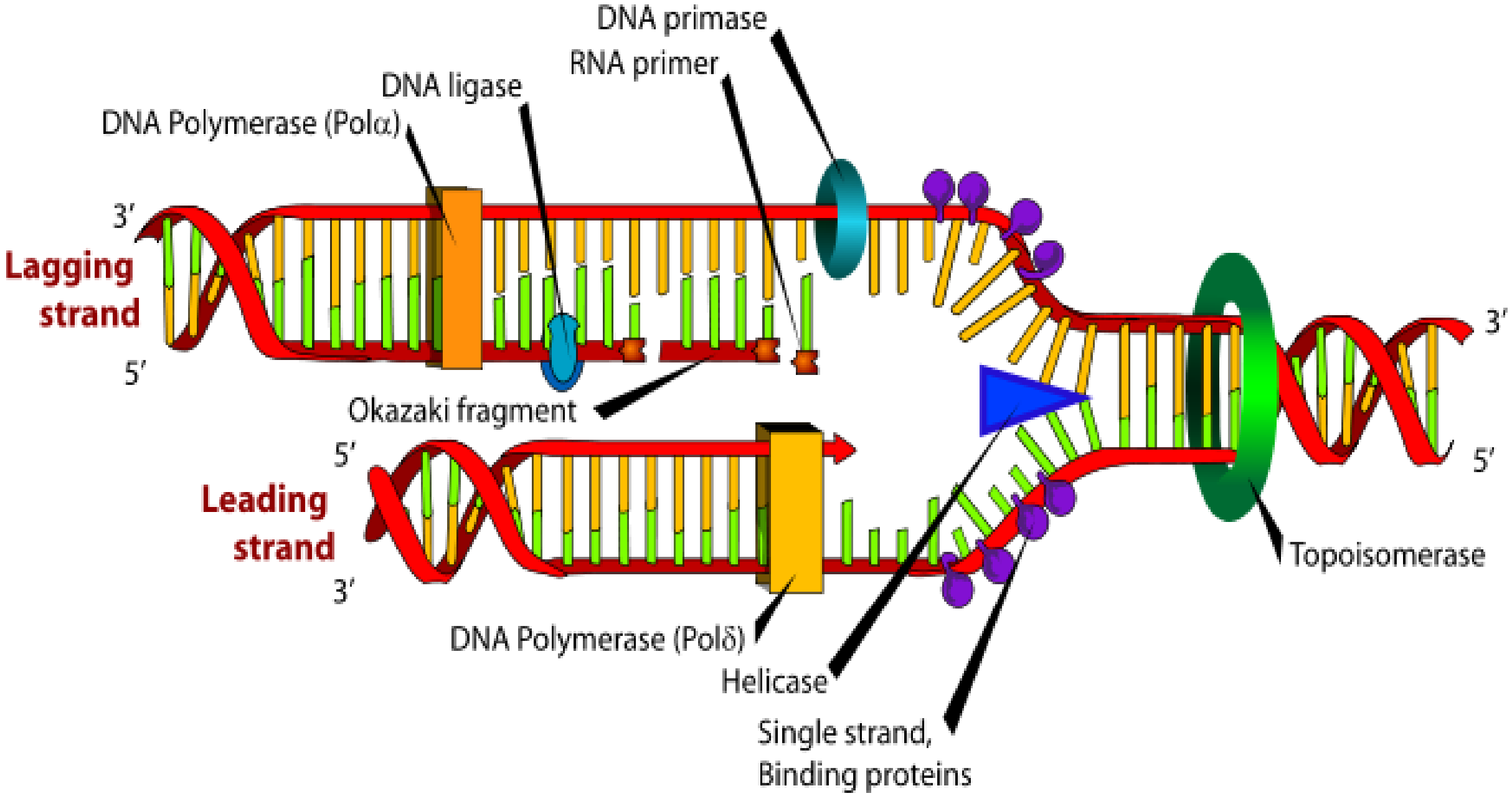
3- When replication is complete, how many double-stranded DNA molecules are formed?

2

4- How do the two new DNA molecules compare to the original one?

They are identical

5- The process by which new molecules of DNA are formed is called replication



[Video - DNA Replication (Advanced)]

Video clips

Heredity Crash Course (10min)

Video - Human Genome 3 sad findings (3 min)

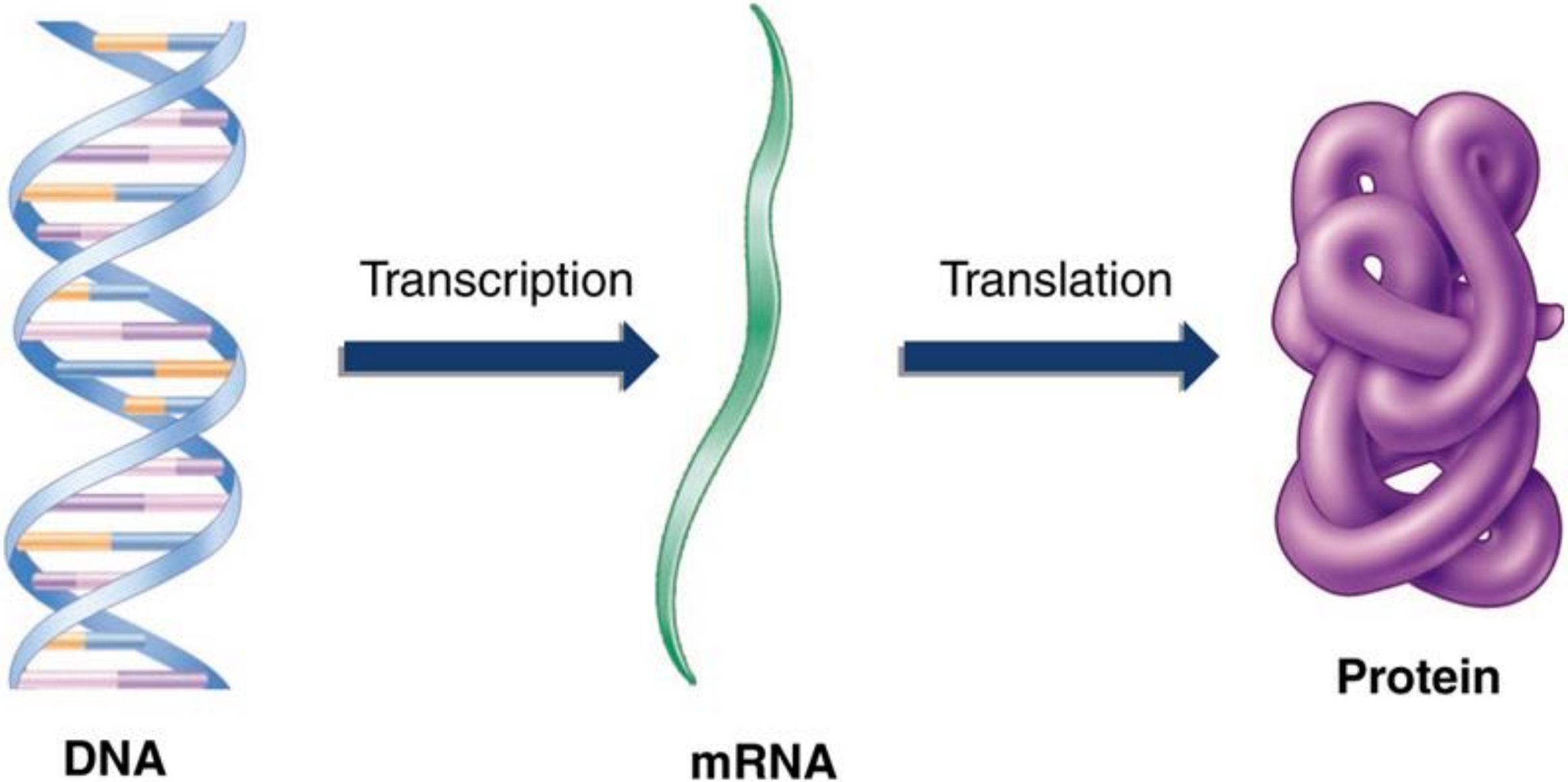
DNA Data Storage (partnership with Microsoft) (1.5 min)

Lesson 5

QUIZ

Protein Structure & Function

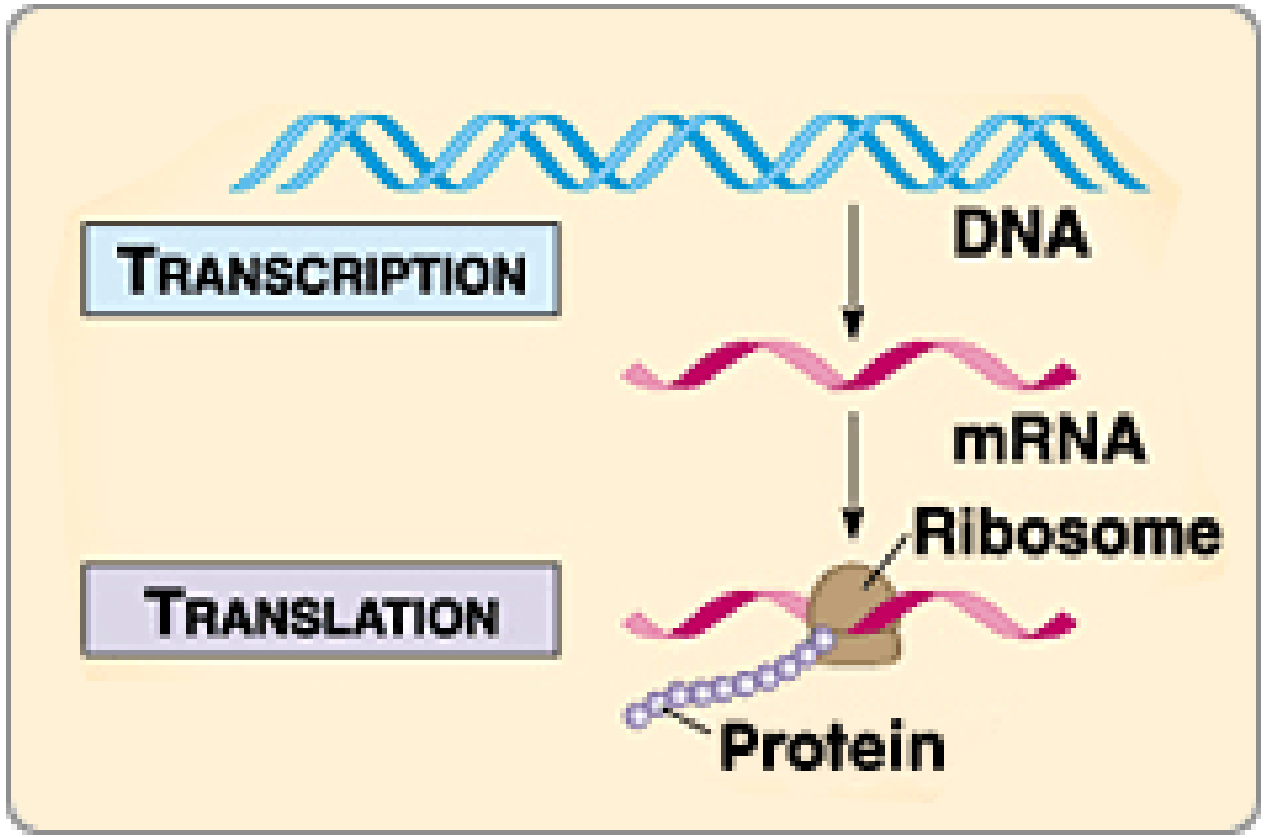
Comparing DNA & RNA



[Video - Protein Synthesis (Advanced)]

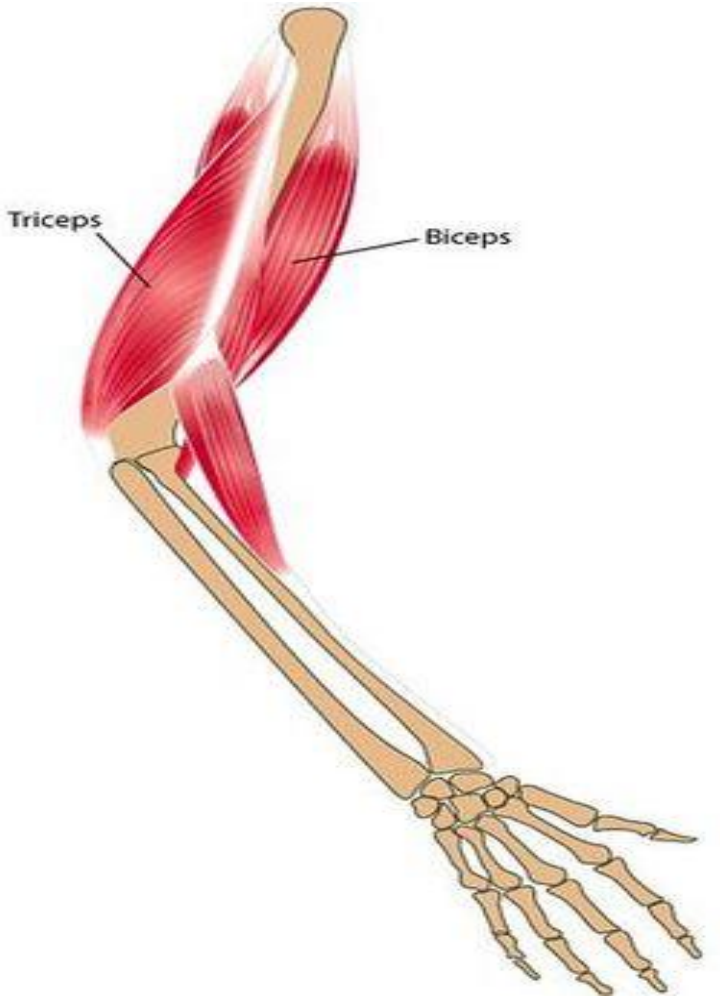
DNA's Main Function is to direct Protein Synthesis

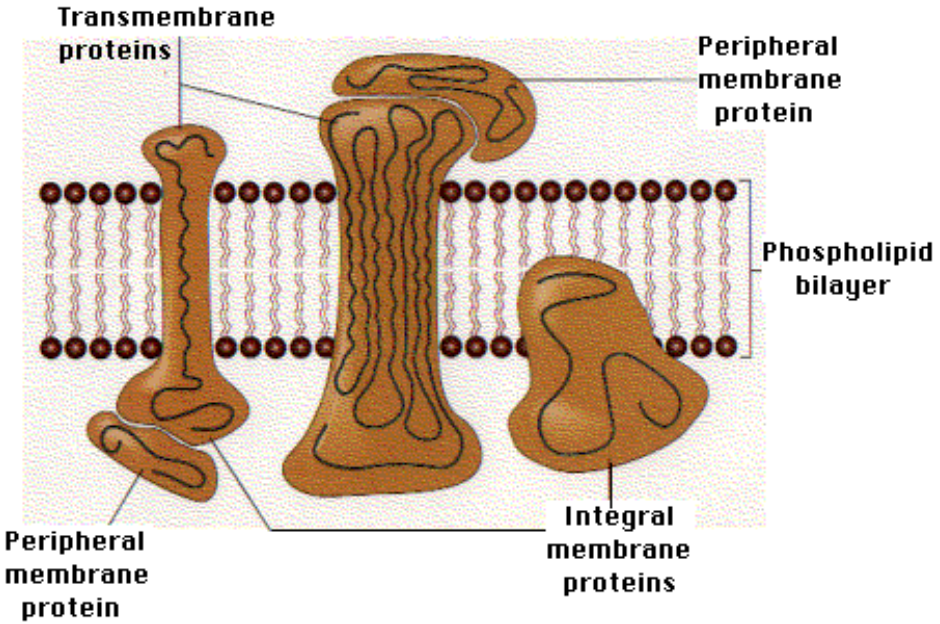
- The sequence of nitrogenous bases in DNA serve as a template (instructions or code) for building proteins

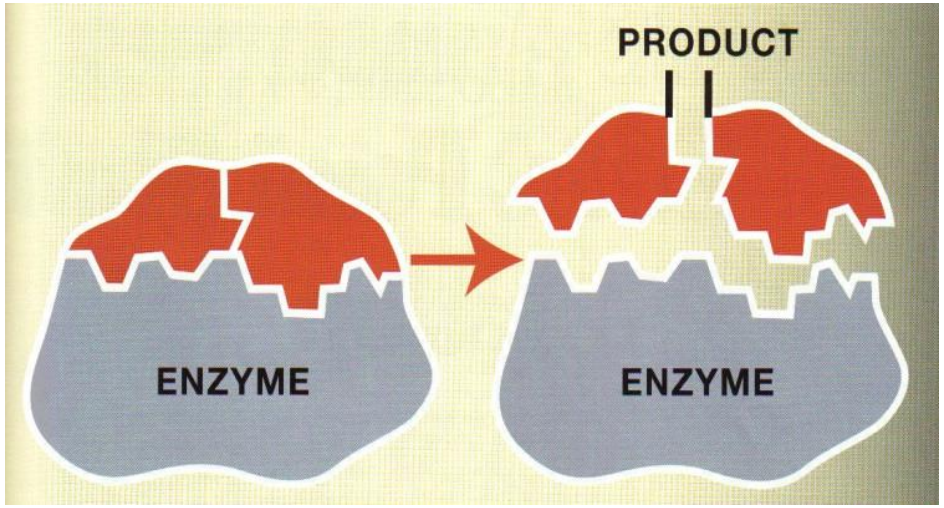


BrainPOP RNA

Examples of Important Proteins



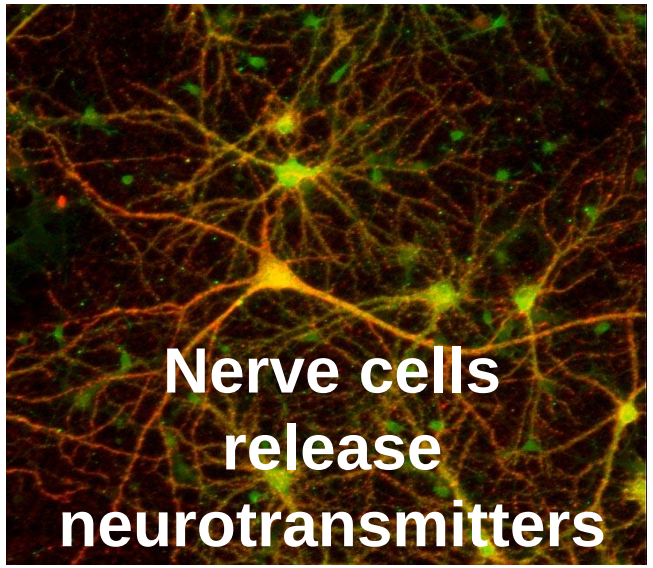




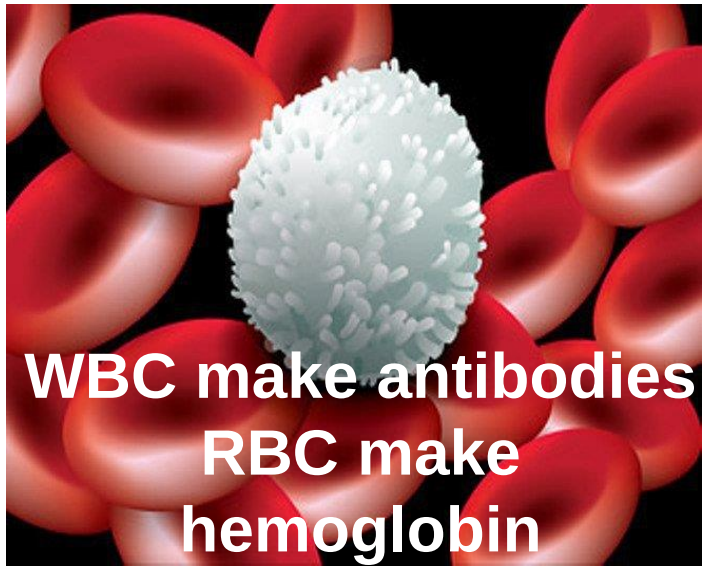
Examples of Important Proteins

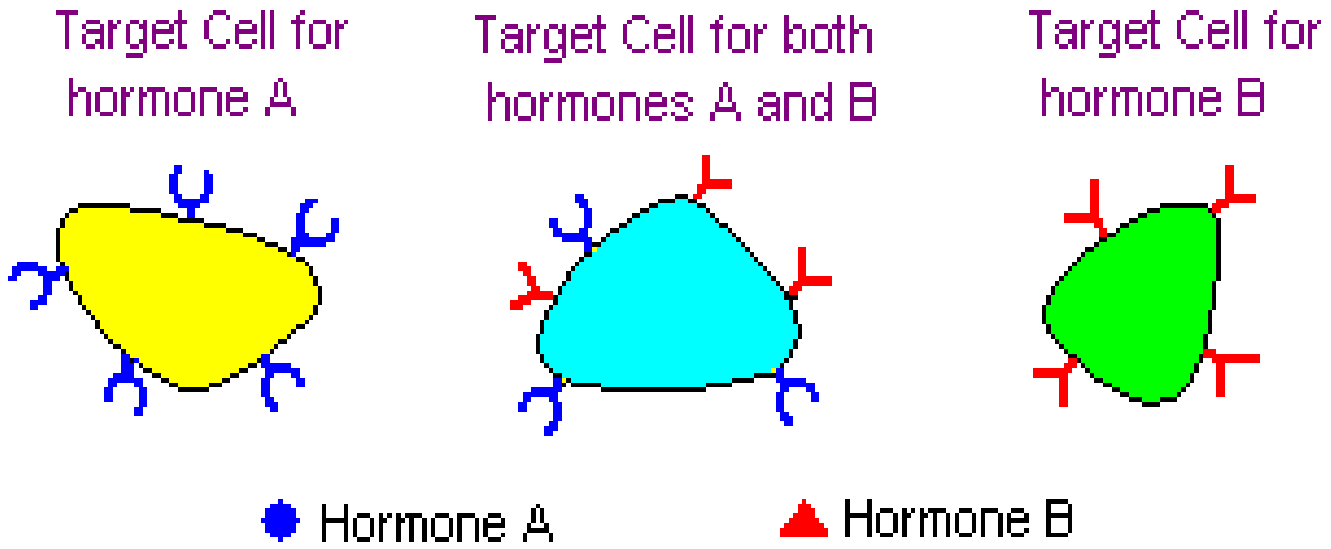
- Enzymes
- Hormones
- Neurotransmitters
- Receptor molecules
- Hemoglobin
- Membrane transport proteins
- Antibodies
- Antigens

How do cells with identical DNA function so differently?



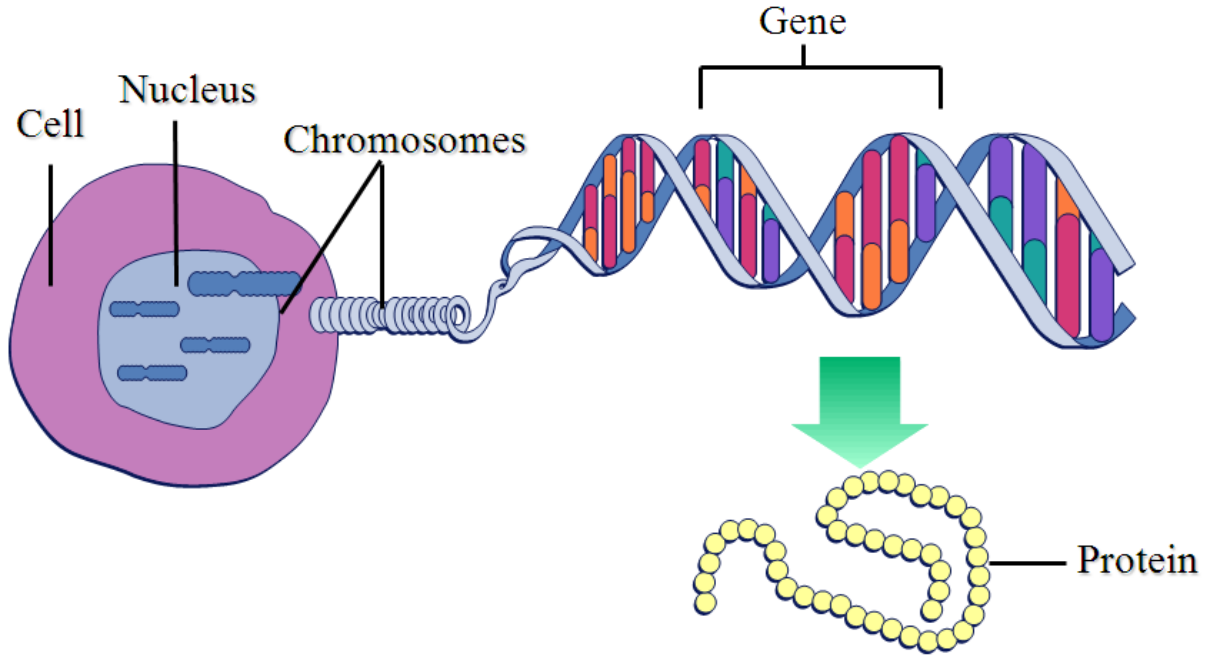
Receptor molecules & hormones detected





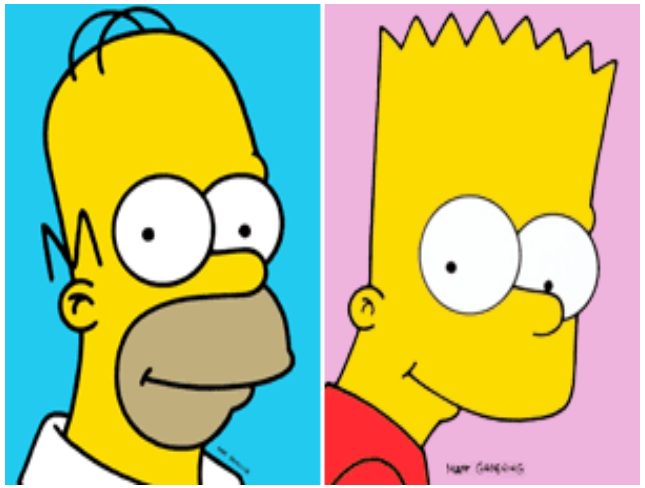
Cells with identical DNA function differently because…

- **Cells only use (express) certain parts of the DNA code OR**
- **Not all cells express (use) the same genes, allowing them to produce different proteins**



Why do offspring resemble their parents?

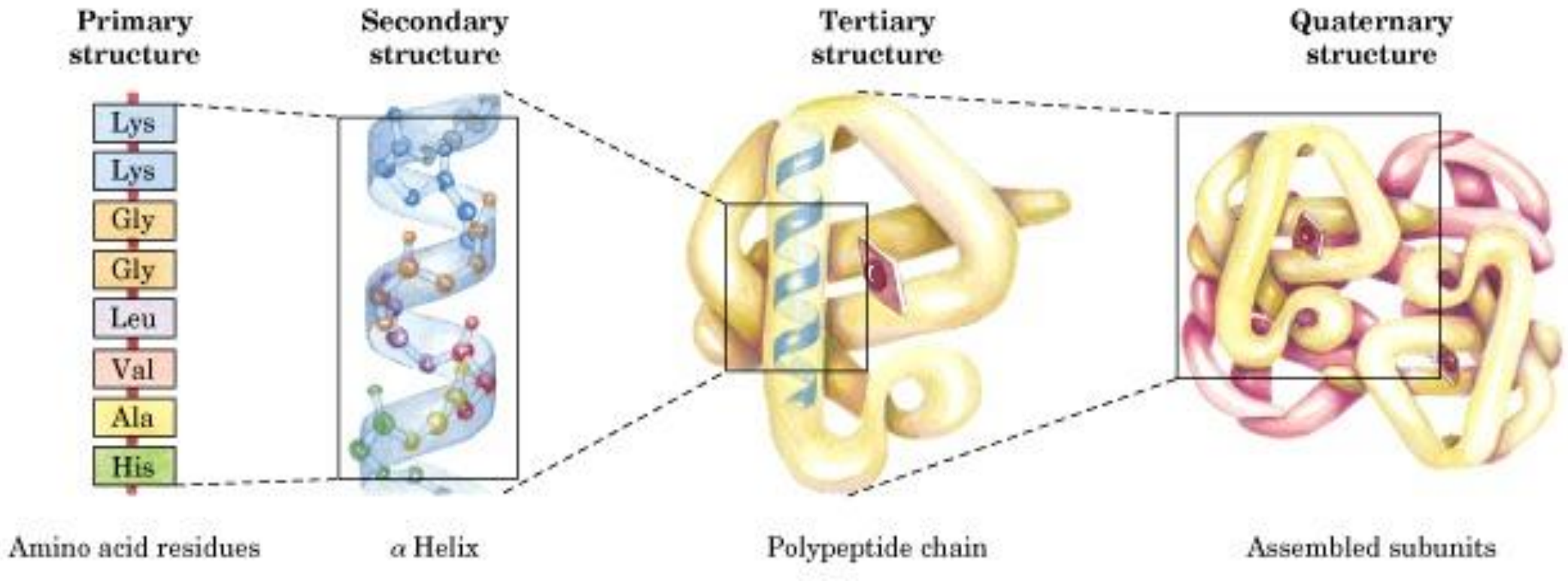
Offspring resemble their parents because…



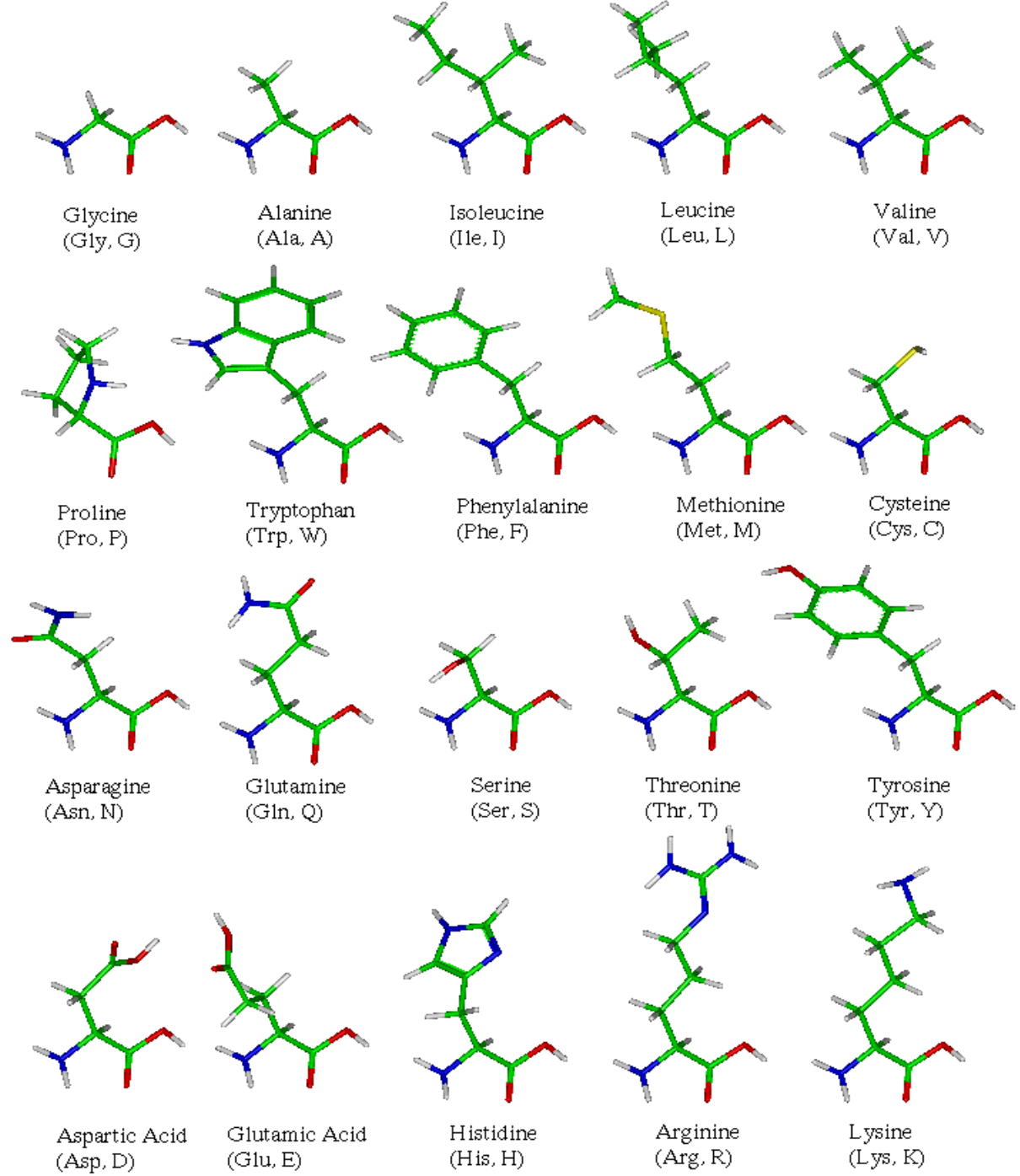
- **Genes inherited from parents are instructions for making proteins**
- **We have more proteins in common with parents and siblings than with non-relatives**

Proteins

- long folded chain of amino acids in a specific sequence
- Specifically shaped to perform a certain function



20 amino acids coded for by human genes



The Importance of Sequencing in Building Proteins

How do you spell this? →

C A T

What other words can be formed using those same letters?

ACT TAC

The order of letters determines the pronunciation & meaning of a word, just like…

The order of amino acids determines the shape and function of a protein!

DNA needs help making the proteins! It's too big to get out of the nucleus to give the message to the ribosome! What to do?

RNA to the rescue!

Comparing DNA and RNA

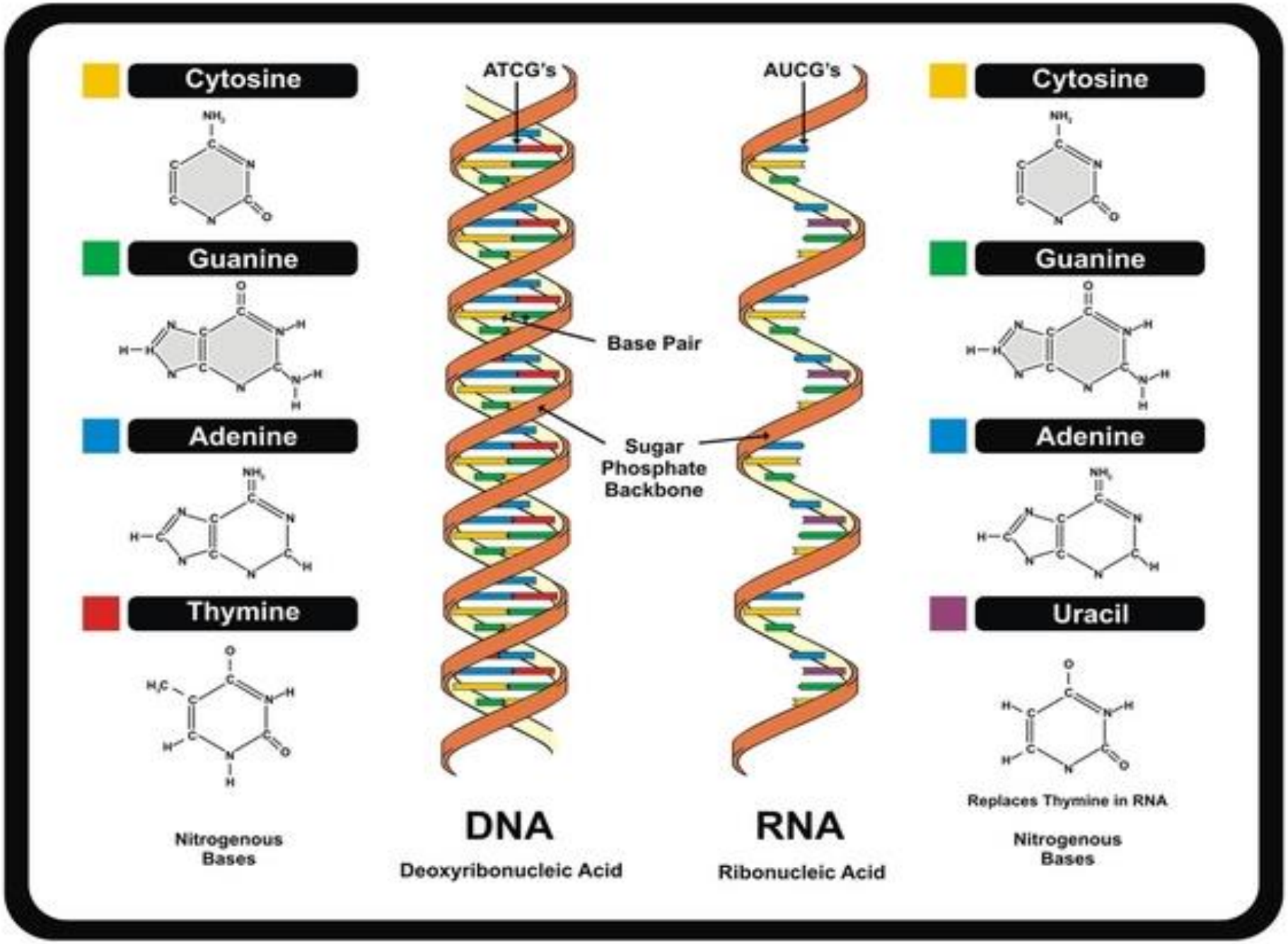
DNA

- Four bases: A T C G
- Sugar = Deoxyribose
- Double stranded
- One type of DNA

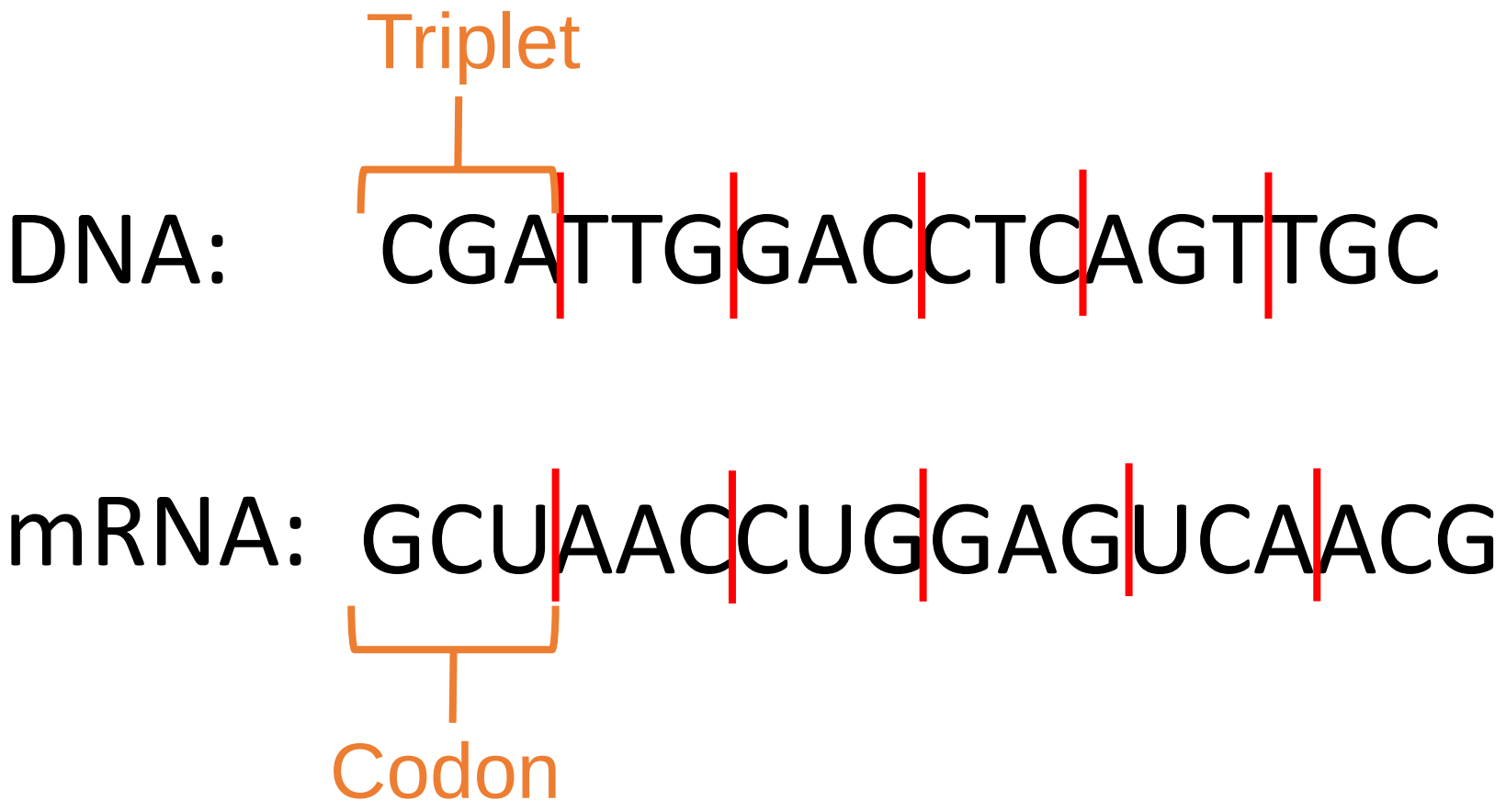
RNA (Ribonucleic Acid)

- Four bases: A U C G
  Uracil (U) instead of Thymine (T)
- Sugar = Ribose
- Single stranded
- 3 types of RNA

Video - Why RNA is just as cool as DNA (Amoeba Sisters)


Cytosine
Guanine
Adenine
Thymine
Nitrogenous Bases
ATCG's
Base Pair
Sugar Phosphate Backbone
DNA
Deoxyribonucleic Acid
AUCG's
RNA
Ribonucleic Acid
Cytosine
Guanine
Adenine
Uracil
Replaces Thymine in RNA
Nitrogenous Bases

What would the complementary mRNA strand look like?



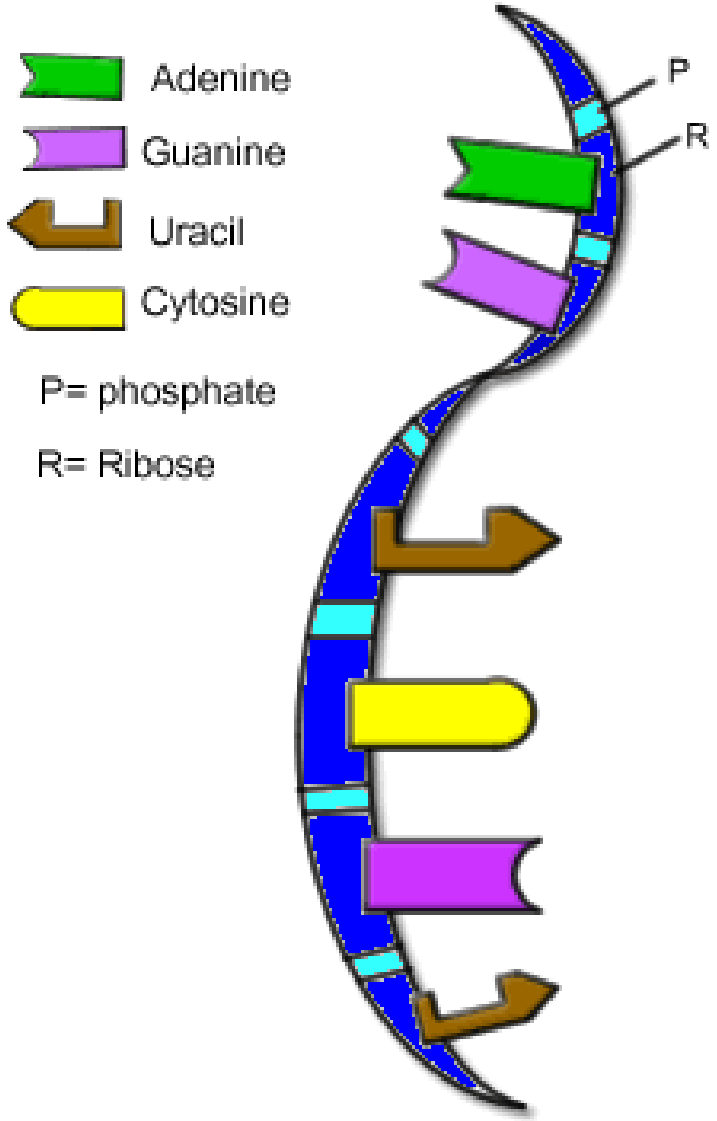
Lesson 6
(two periods)

Protein Synthesis

3 types of RNA

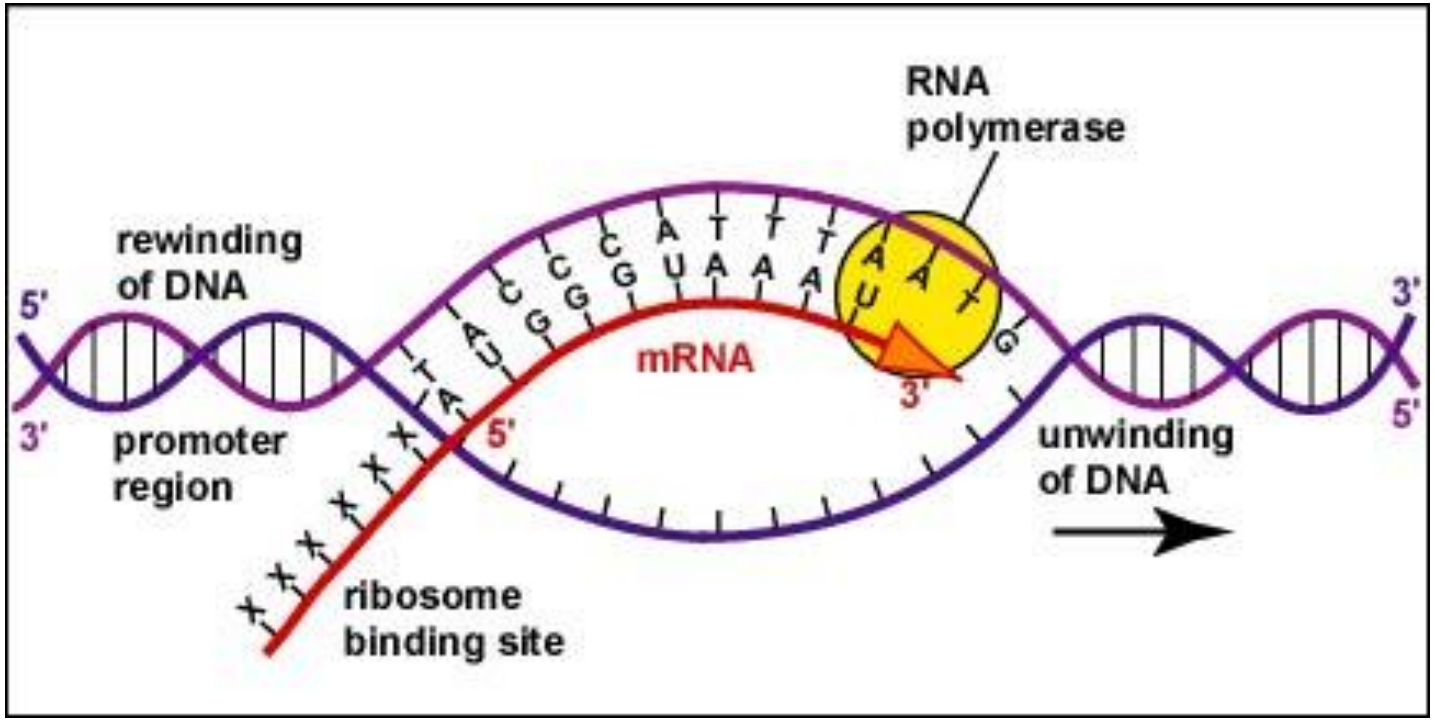
Transcription & Translation

Three types of RNA



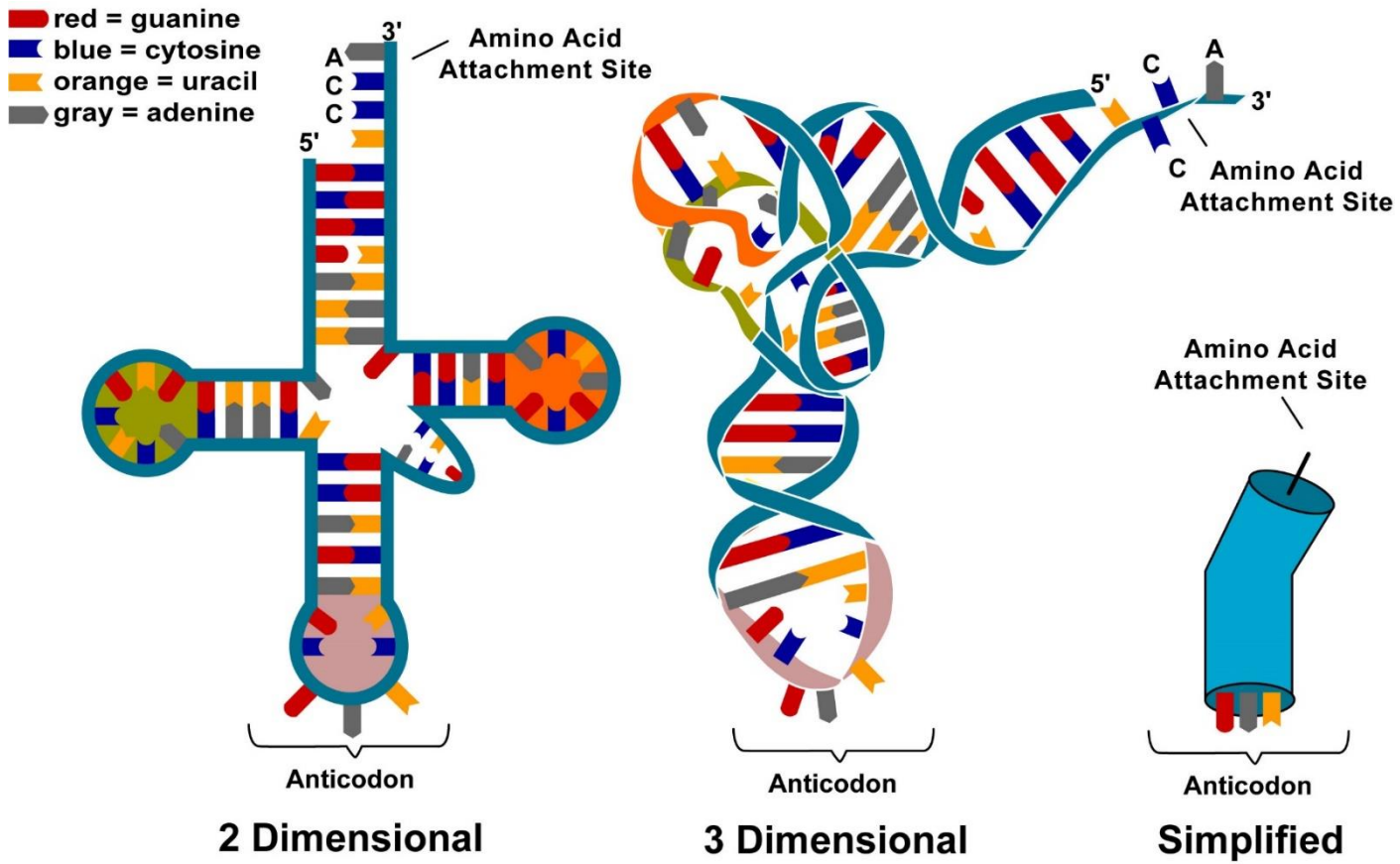
1) mRNA (messenger RNA)

- Forms in nucleus to copy DNA's code
- Carries code from the nucleus to the ribosome (site of protein synthesis)
- Sequence of 3 bases is a codon



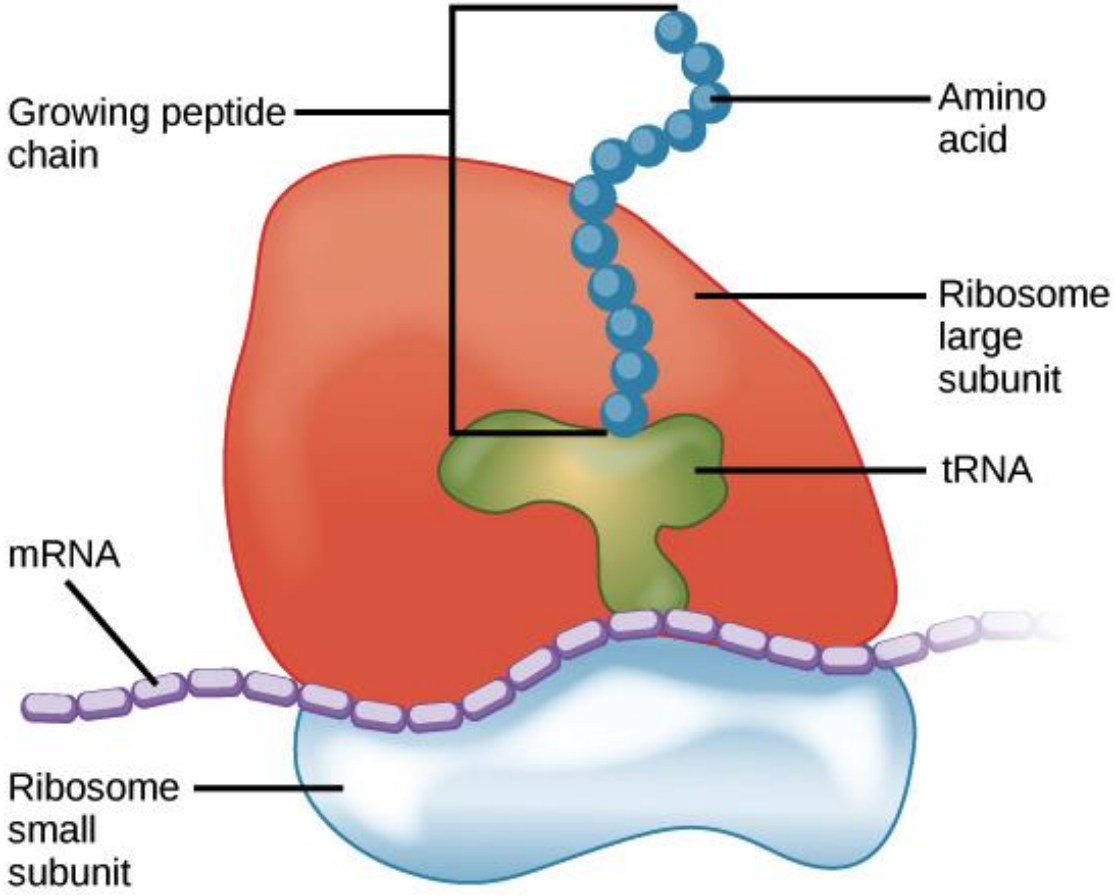
2) tRNA (transfer RNA)

- brings amino acids from cytoplasm to ribosome
- sequence of 3 bases complementary to mRNA codons are called anticodons



3) rRNA (ribosomal RNA)

- Part of ribosome, codes for which protein is made there



2 _____1. In the synthesis of proteins, what is the function of messenger-RNA molecules?

(1) They act as a template for the synthesis of DNA.
(2) They carry information that determines the sequence of amino acids.
(3) They remove amino acids from the nucleus.
(4) They carry specific enzymes for dehydration synthesis.

1 _____2. A sequence of three nitrogenous bases in a messenger-RNA molecule is known as a

(1) codon
(2) gene
(3) polypeptide
(4) nucleotide

2 _____3. Which base is normally used in the synthesis of RNA but *not* in the synthesis of DNA?

(1) adenine
(2) uracil
(3) cytosine
(4) guanine

4 _____4. If a portion of a messenger RNA molecule contains the base sequence A-A-U, the corresponding transfer RNA base sequence is

(1) A-A-U
(2) G-G-T
(3) T-T-C
(4) U-U-A

3 _____5. The code of a gene is delivered to the enzyme-producing region of a cell by a

(1) hormone
(2) nerve impulse
(3) messenger RNA molecule
(4) DNA molecule

_____6. Which statement best describes the relationship between cells, DNA, and proteins?

(1) Cells contain DNA that controls the production of proteins.
(2) DNA is composed of proteins that carry coded information for how cells function.
(3) Proteins are used to produce cells that link amino acids together into DNA.
(4) Cells are linked together by proteins to make different kinds of DNA molecules.

1

mRNA AND TRANSCRIPTION

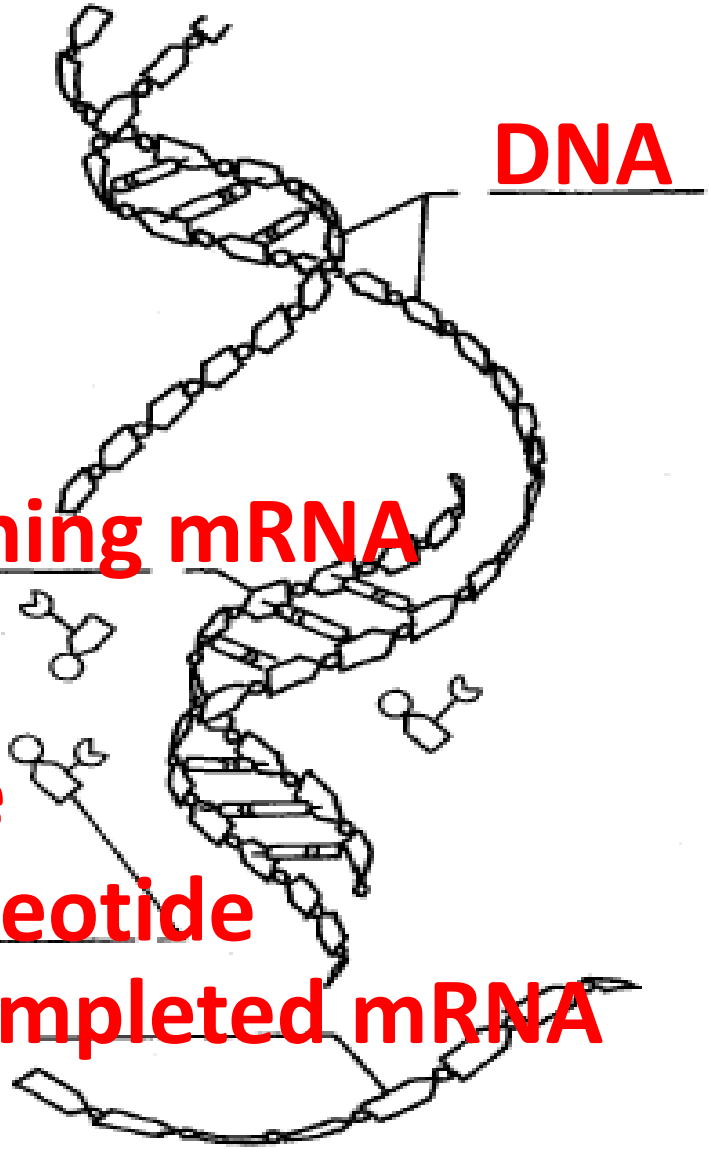
Name _____

Transcription

Fill in the blanks below. On the illustration of transcription, label the DNA, the newly-forming mRNA, the completed strand of mRNA and a free nucleotide.

Messenger RNA (mRNA) carries the instructions to make a particular **protein** from the DNA in the **nucleus** to the ribosomes. The process of producing mRNA from instructions in the DNA is called **transcription**.

During transcription, the DNA molecule unwinds and separates, exposing the nitrogenous bases. Free RNA **nucleotides** pair with the exposed bases. There is no **thymine** (T) in RNA. **Uracil** (U) pairs with adenine (A) instead. RNA contains the sugar **ribose** instead of deoxyribose. The mRNA molecule is completed by the formation of **bonds** between the RNA **nucleotides**, and it then separates from the DNA. The mRNA molecule is a **single** strand, unlike DNA.



Codons

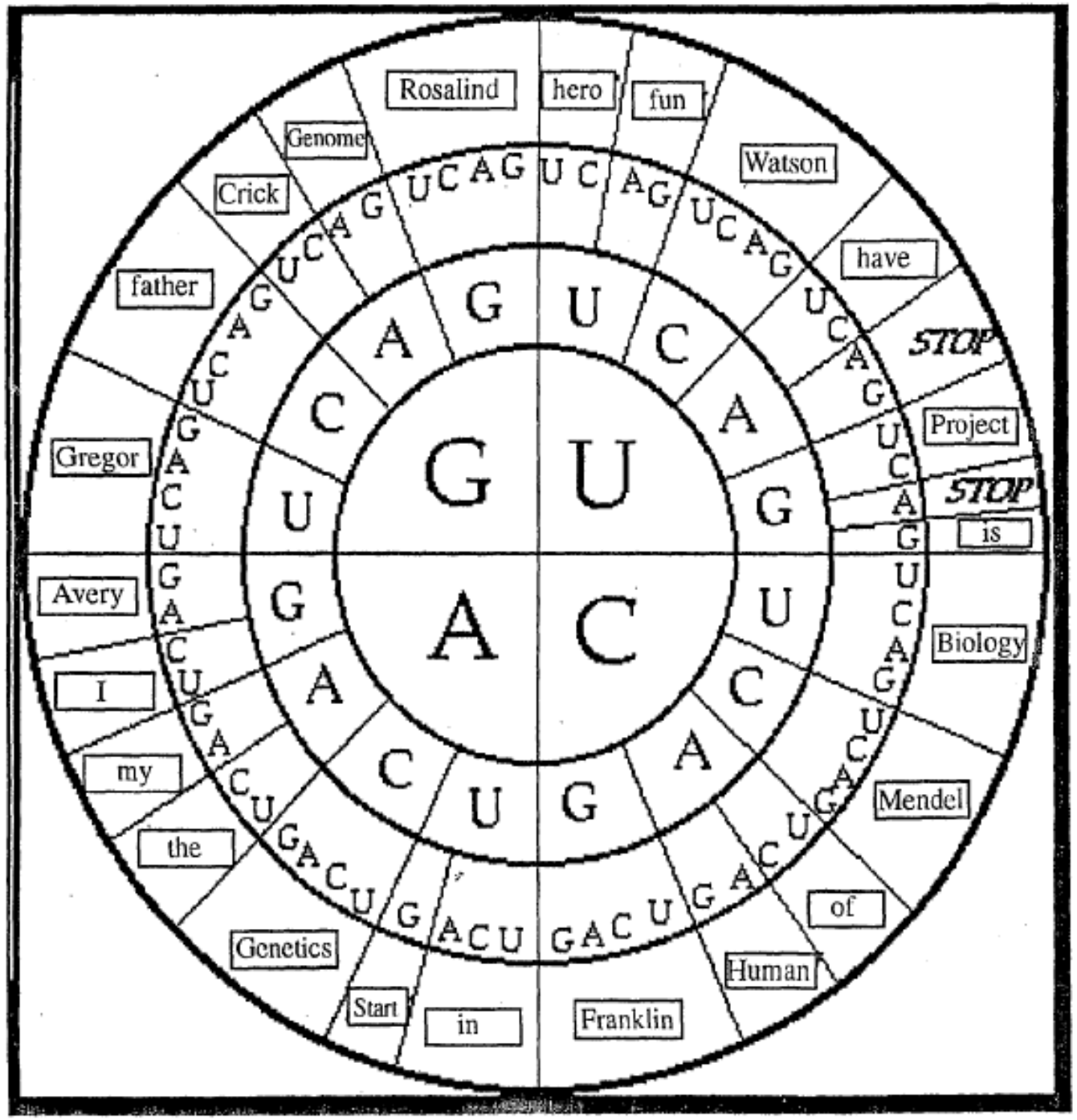
Each combination of three nitrogenous bases on the mRNA molecule is a codon, a three-letter code word for a specific amino acid. The table below shows the mRNA codon for each amino acid. Use the table to answer the questions below.

1. The codon for trytophan is UGG
2. For leucine, there are 6 different codons.
3. The codon GAU is for Aspartic acid.
4. In a stop codon, if the second base is G, the first and third bases are U and A.

First Base in Code Word	Second Base in Code Word: A	Second Base in Code Word: G	Second Base in Code Word: U	Second Base in Code Word: C	Third Base in Code Word
A	Lysine	Arginine	Isoleucine	Threonine	A
	Lysine	Arginine	Methionine	Threonine	G
	Asparagine	Serine	Isoleucine	Threonine	U
	Asparagine	Serine	Isoleucine	Threonine	C
G	Glutamic Acid	Glycine	Valine	Alanine	A
	Glutamic Acid	Glycine	Valine	Alanine	G
	Aspartic Acid	Glycine	Valine	Alanine	U
	Aspartic Acid	Glycine	Valine	Alanine	C
U	"Stop" codon	"Stop" codon	Leucine	Serine	A
	"Stop" codon	Trytophan	Leucine	Serine	G
	Tyrosine	Cysteine	Phenylalanine	Serine	U
	Tyrosine	Cysteine	Phenylalanine	Serine	C
C	Glutamine	Arginine	Leucine	Proline	A
	Glutamine	Arginine	Leucine	Proline	G
	Histidine	Arginine	Leucine	Proline	U
	Histidine	Arginine	Leucine	Proline	C

The RNA Wheel Game

Using mRNA codons, start in the center and work your way out to translate the message.



Procedure :

Transcribe the DNA codes provided below into mRNA code. Then use the RNA Translation Wheel to reveal the secret messages.

1. DNA Segment:

TAC TCG ATA AAT TAT GAT ACT

mRNA Transcribed Code:

AUG AGC UAU UUA AUA CUA UGA

Translated Message:

START I have fun in biology STOP

2. DNA Segment:

TAC CAA GGA ACC TTG CGT GTG

TGA ACT

mRNA Transcribed Code:

AUG GUU CCU UGG AAC GCA CAC

ACU UGA

Translated Message:

START Gregor Mendel is the father of genetics STOP

3. DNA Segment:

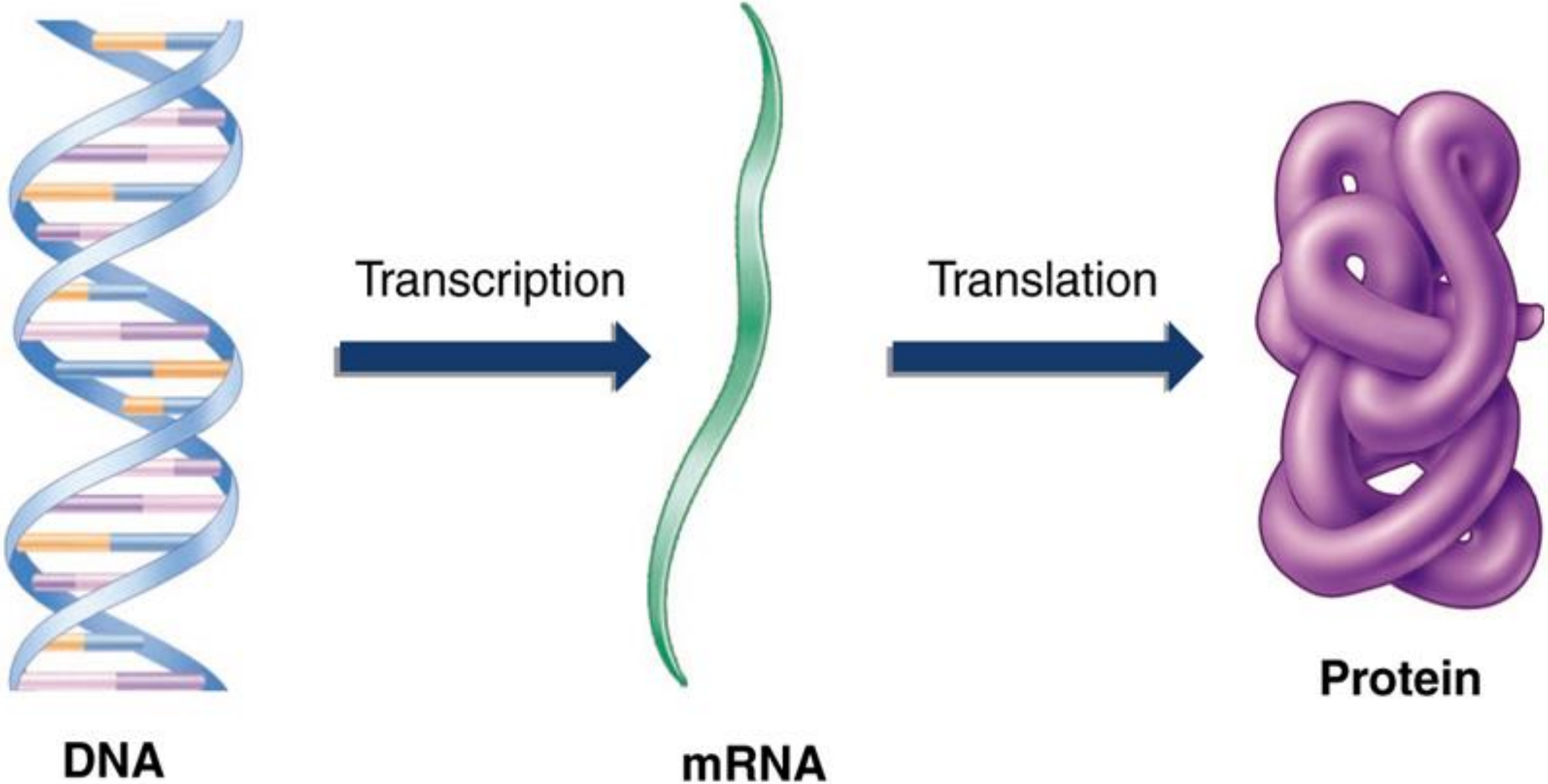
TAC CCG GCC ACC TTT AAA ACT

mRNA Transcribed Code:

AUG GGC CGG UGG AAA UUU UGA

Translated Message:

START Rosalind Franklin is my hero STOP

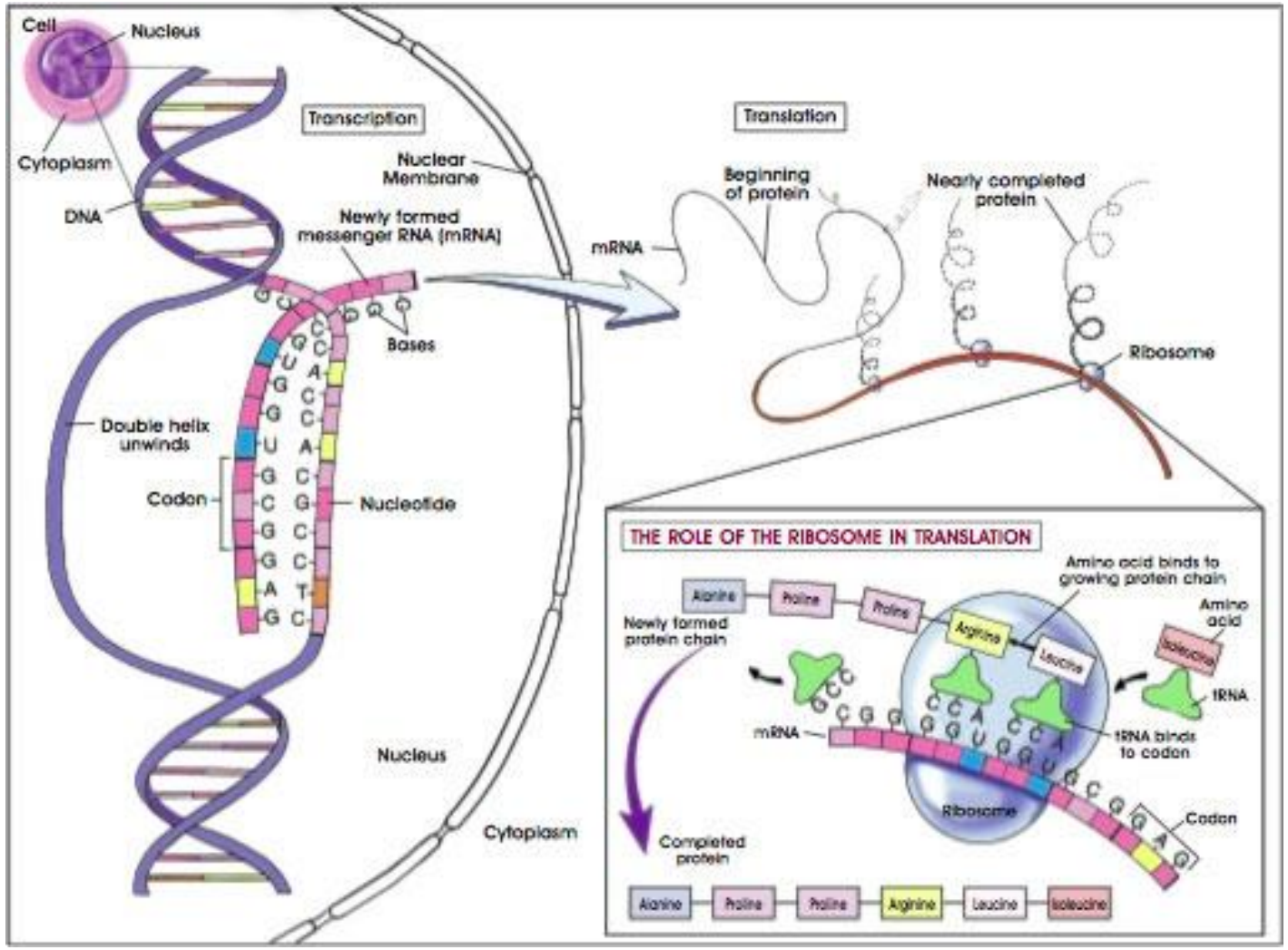


[Video - Protein Synthesis (Transcription & Translation)]

How are proteins made? (Protein Synthesis)

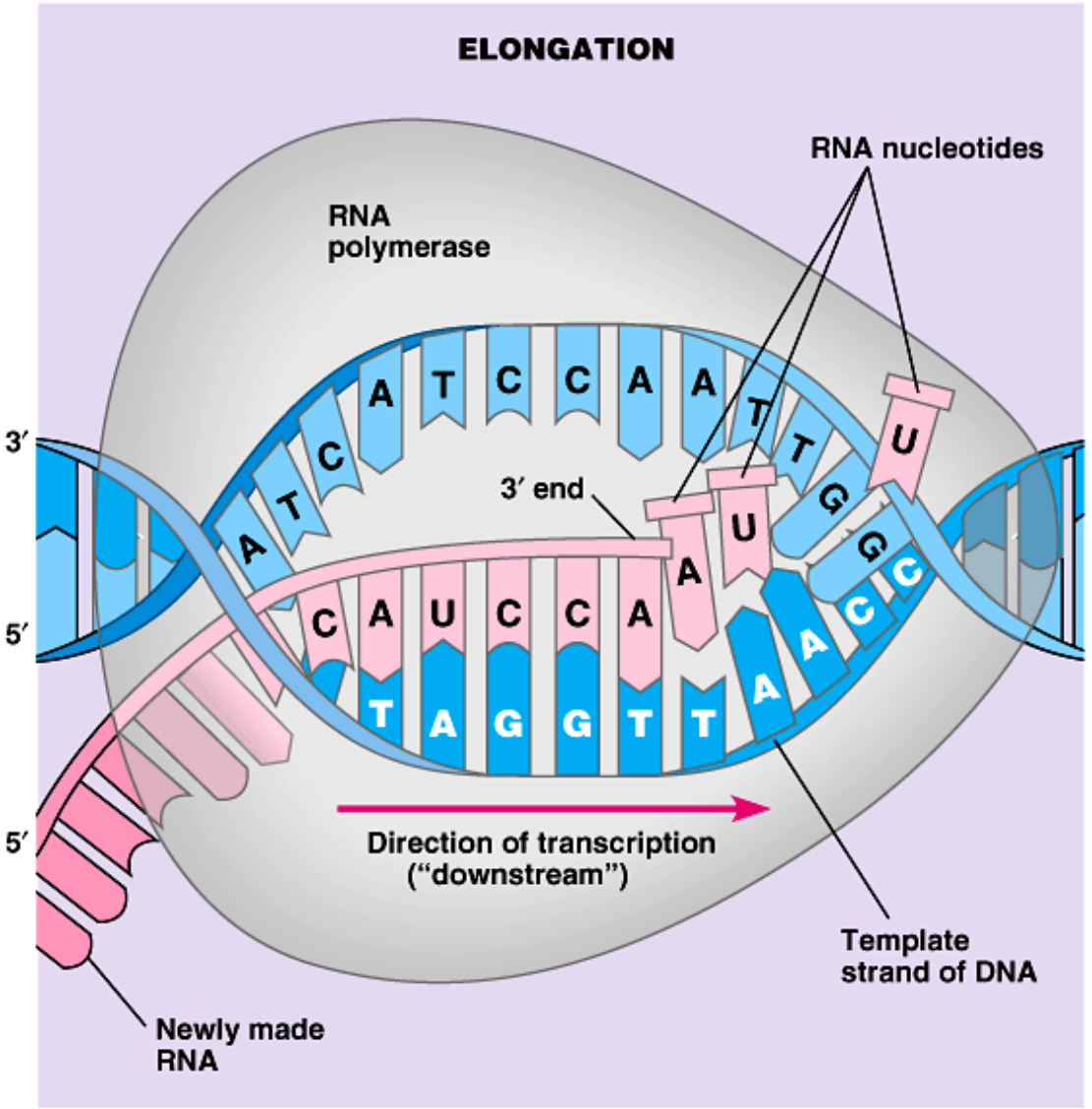
Step 1 – Transcription (in nucleus)

Step 2 – Translation (at ribosome)



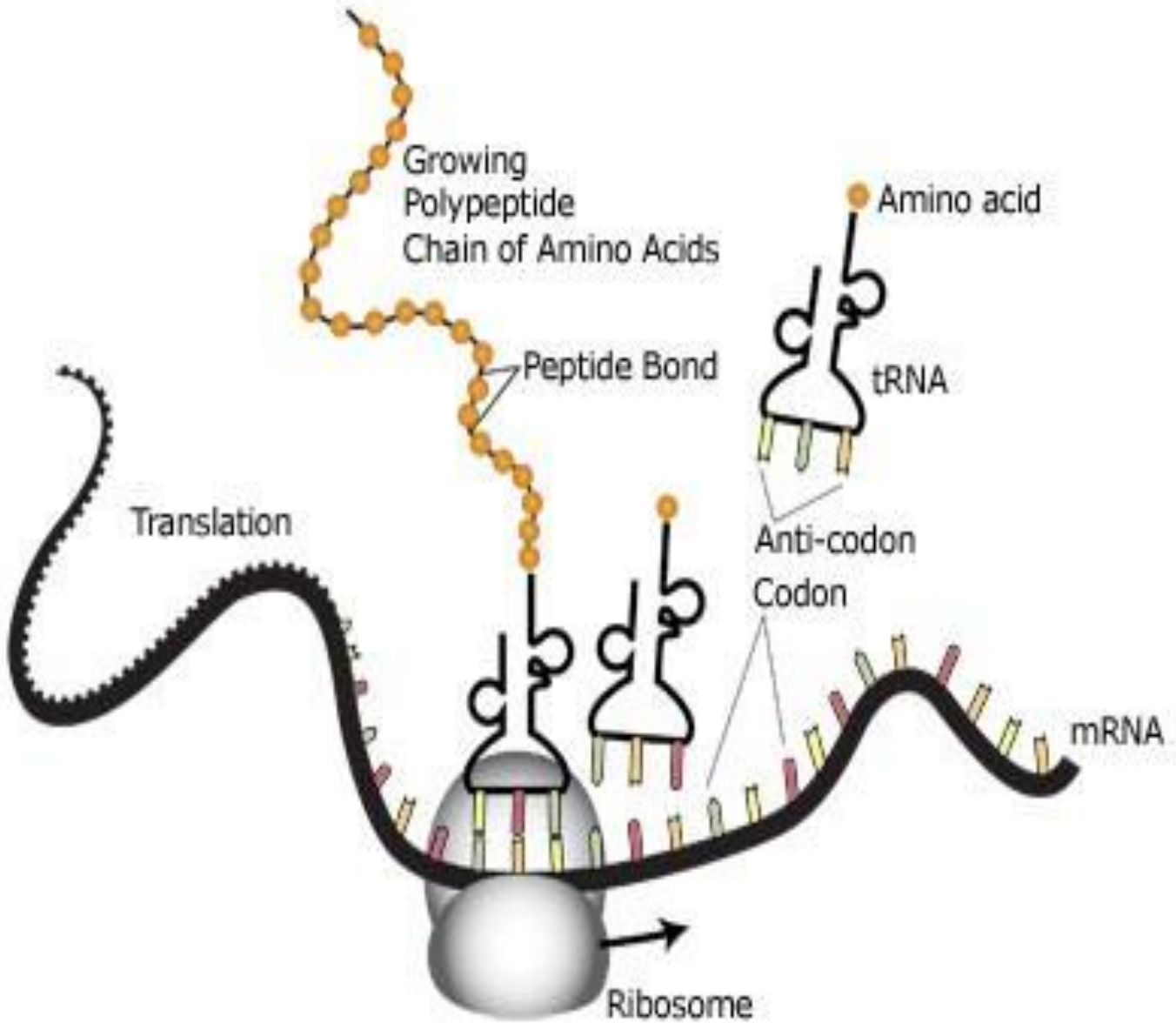
Protein Synthesis - Step 1: Transcription

- occurs in the nucleus
- RNA Polymerase copies (transcribes) the nucleotide (base) sequence of a specific gene, forming mRNA
- similar to DNA replication (except U binds with A, not T)
- mRNA leaves the nucleus to bring the code to the ribosome for translation



Protein Synthesis - Step 2: Translation

- Occurs at the ribosome
- mRNA codons are translated into a specific amino acid sequence
- tRNA brings the correct amino acids (in specific order) to the ribosome to make a protein

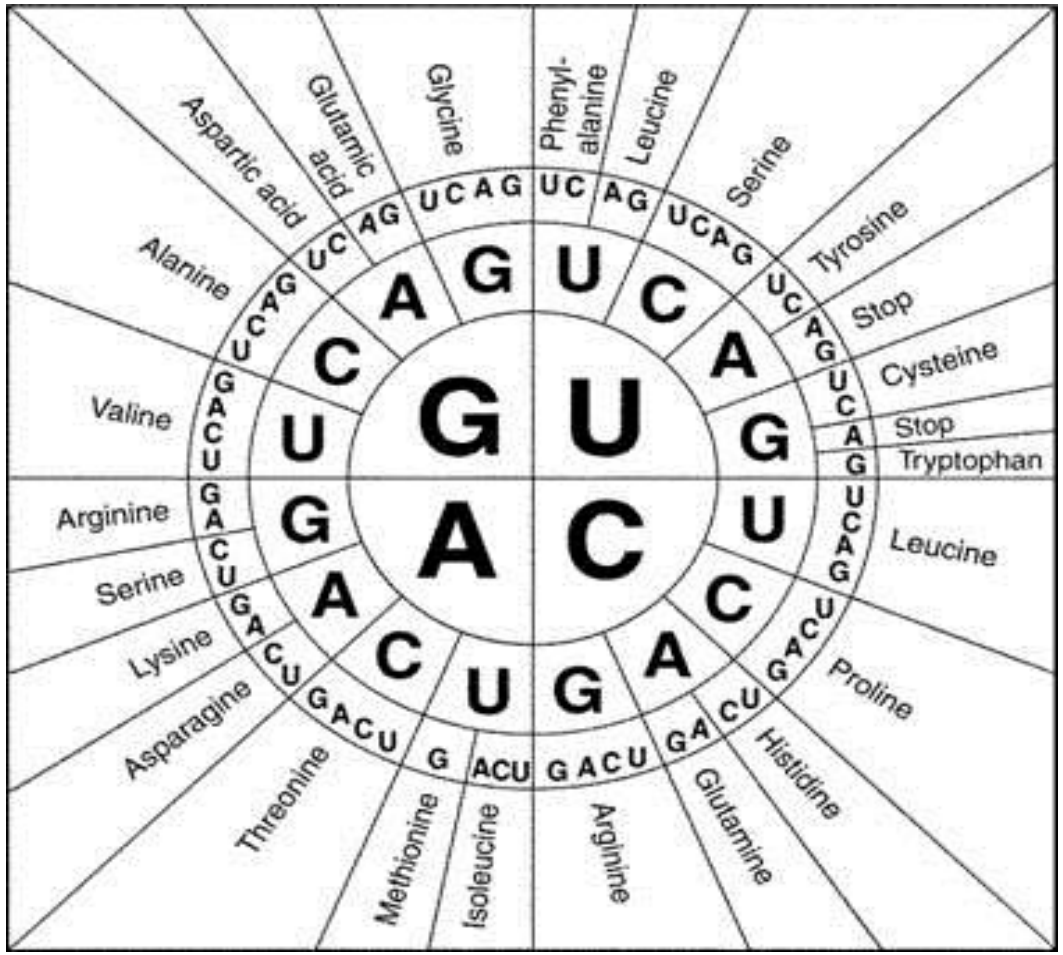


DNA Sequence: CGA TTG GAC CTC AGT TGC

Transcribe (mRNA): GCU AAC CUG GAG UCA ACG

Translate (a.a.): Ala Asp Leu Glu Ser Thr

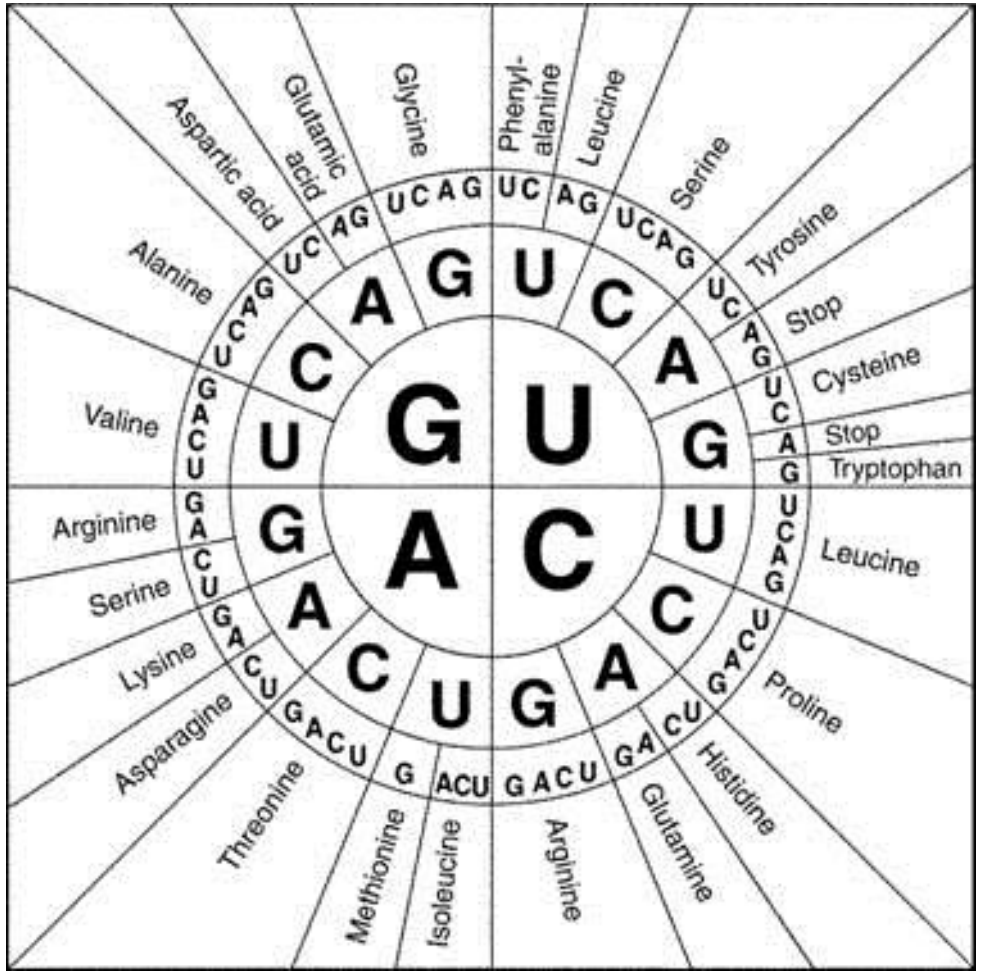
You may write the first 3 letters of each amino acid rather than the full name.

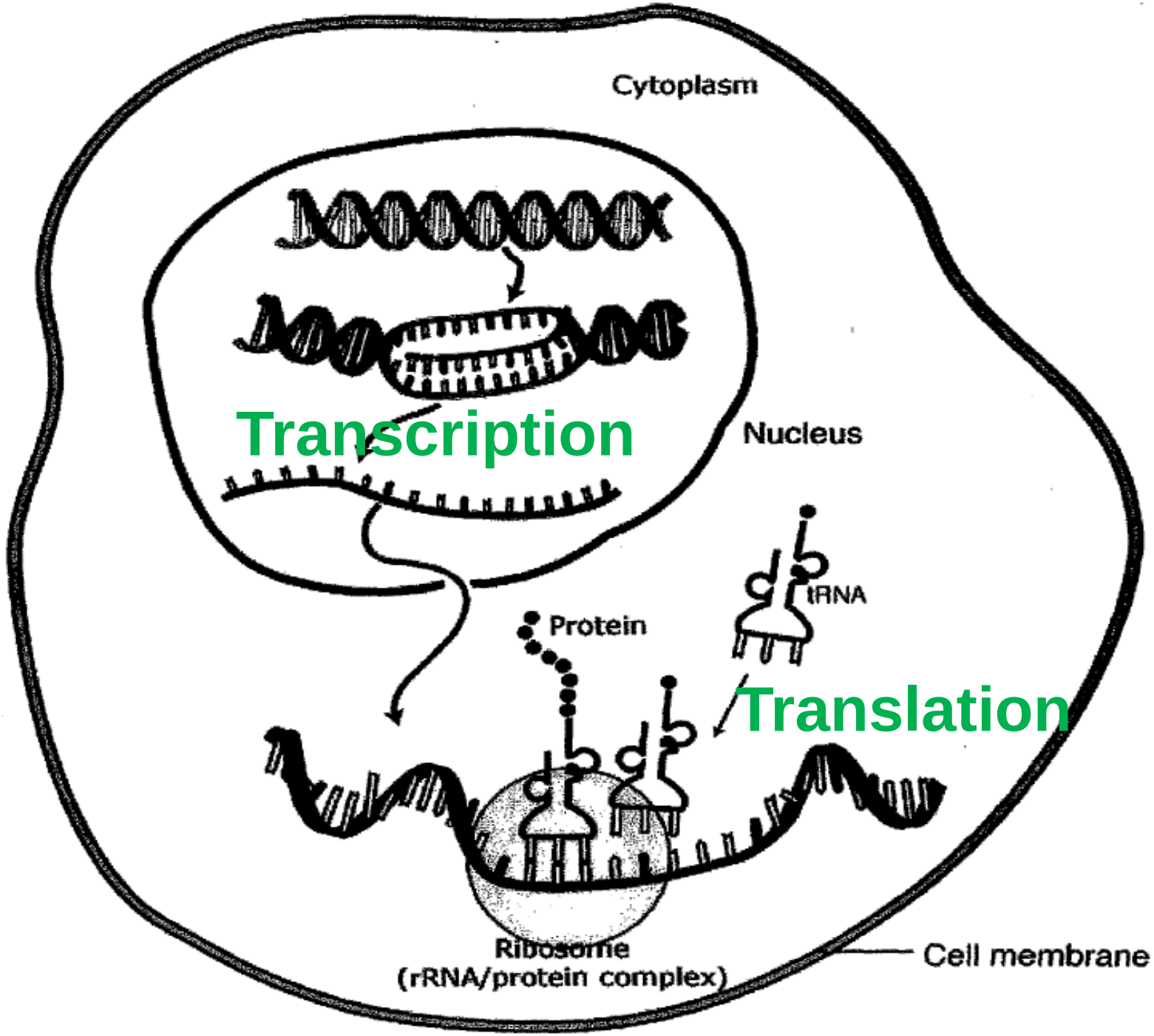


DNA: AUG TTT TAC CCA CGG GTC

mRNA: UAC AAA AUG GGU GCC CAG

AA: Tyrosine, Lysine, Methionine, Glycine, Alanine, Glutamine




Cytoplasm
Transcription
Nucleus
tRNA
Protein
Translation
Ribosome
(rRNA/protein complex)
Cell membrane

Protein Synthesis
(Transcription and Translation)

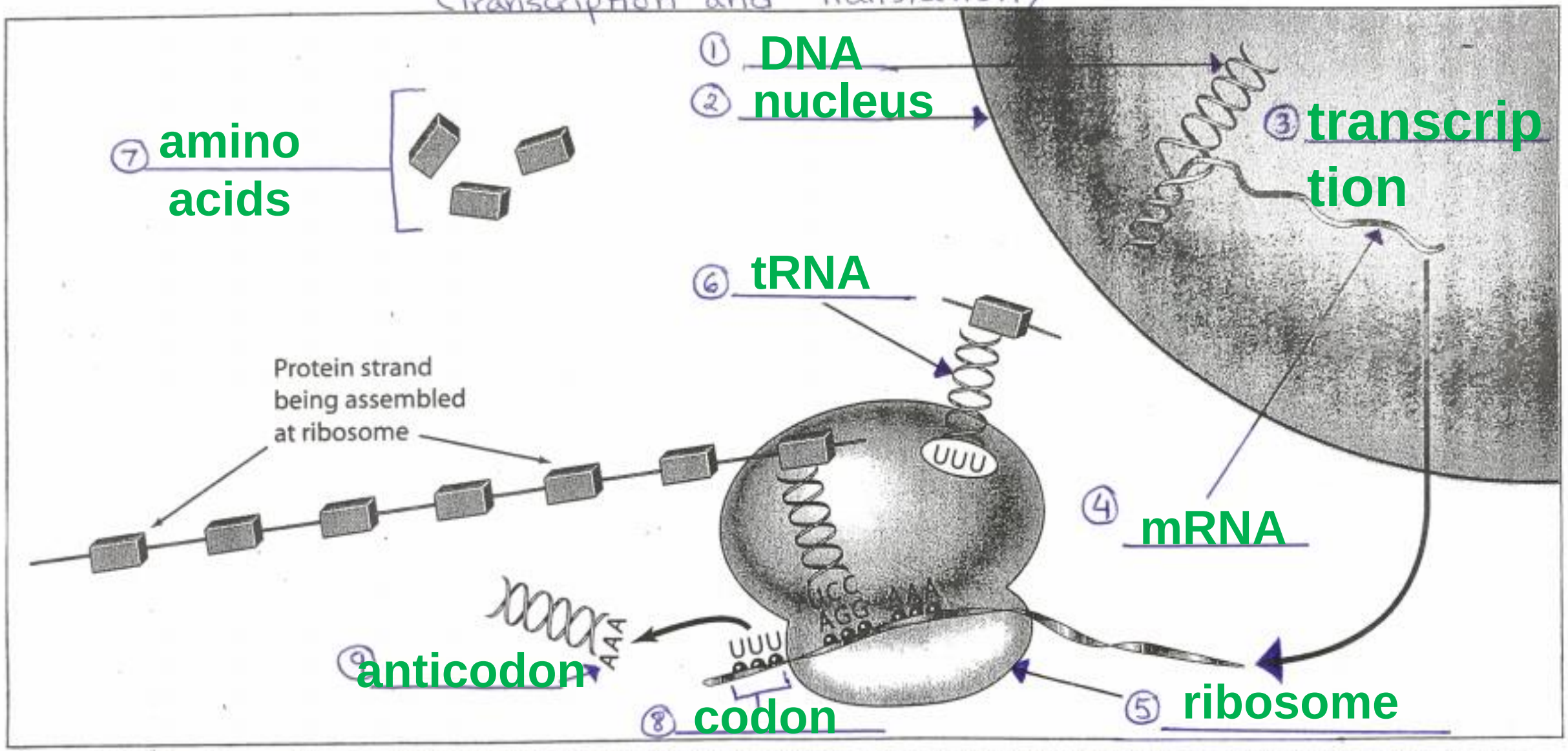


Figure 3-5. Protein synthesis: Notice that the DNA in the nucleus supplies the instructions for how to assemble the protein to the messenger molecule. The transfer molecules help assemble amino acids. The whole assembly occurs at a ribosome.

Lesson 7

Mutations

Race to Transcribe!

You have 30 seconds to write down this DNA sequence and transcribe it to form mRNA

DNA:

TCG TCC AGT AGC TAG CGT TAC CAG

mRNA:

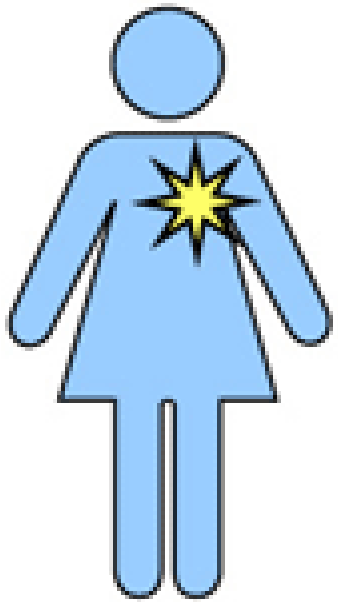
AGC AGG UCA UCG AUC GCA AUG GUC

MAKE ANY MISTAKES?

Mutations

- a change in genetic material
- May occur randomly or be caused by exposure to mutagens
  - Examples: UV rays, X-rays, asbestos
- May be harmful, may have no effect, or can be beneficial depending on the environment
- Most are recessive (hidden by a dominant / healthy allele)

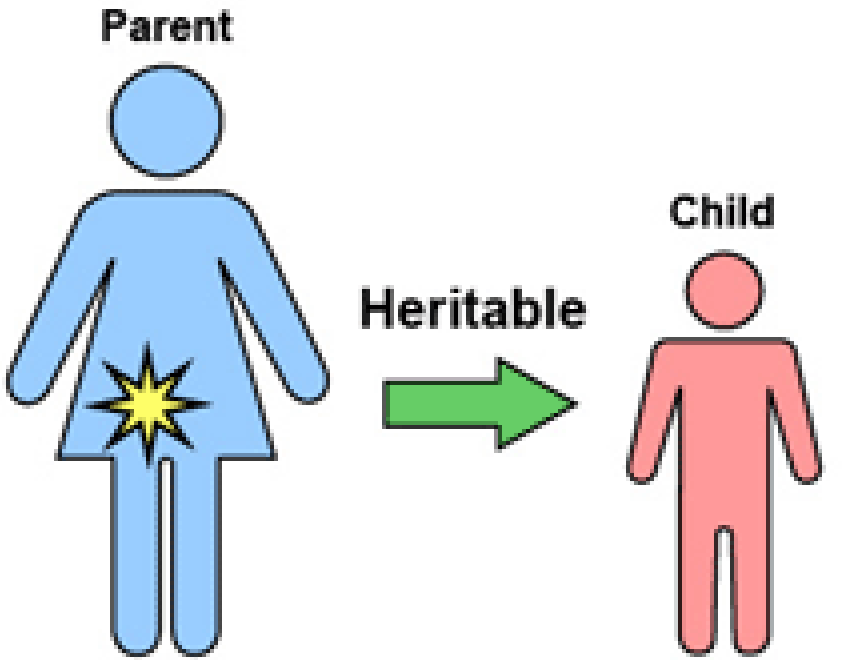
Mutations in a somatic (body) cell CANNOT be passed to offspring!



Nonheritable

Mutation in tumor only
(for example, breast)

Mutations in **gametes CAN** be passed to offspring!



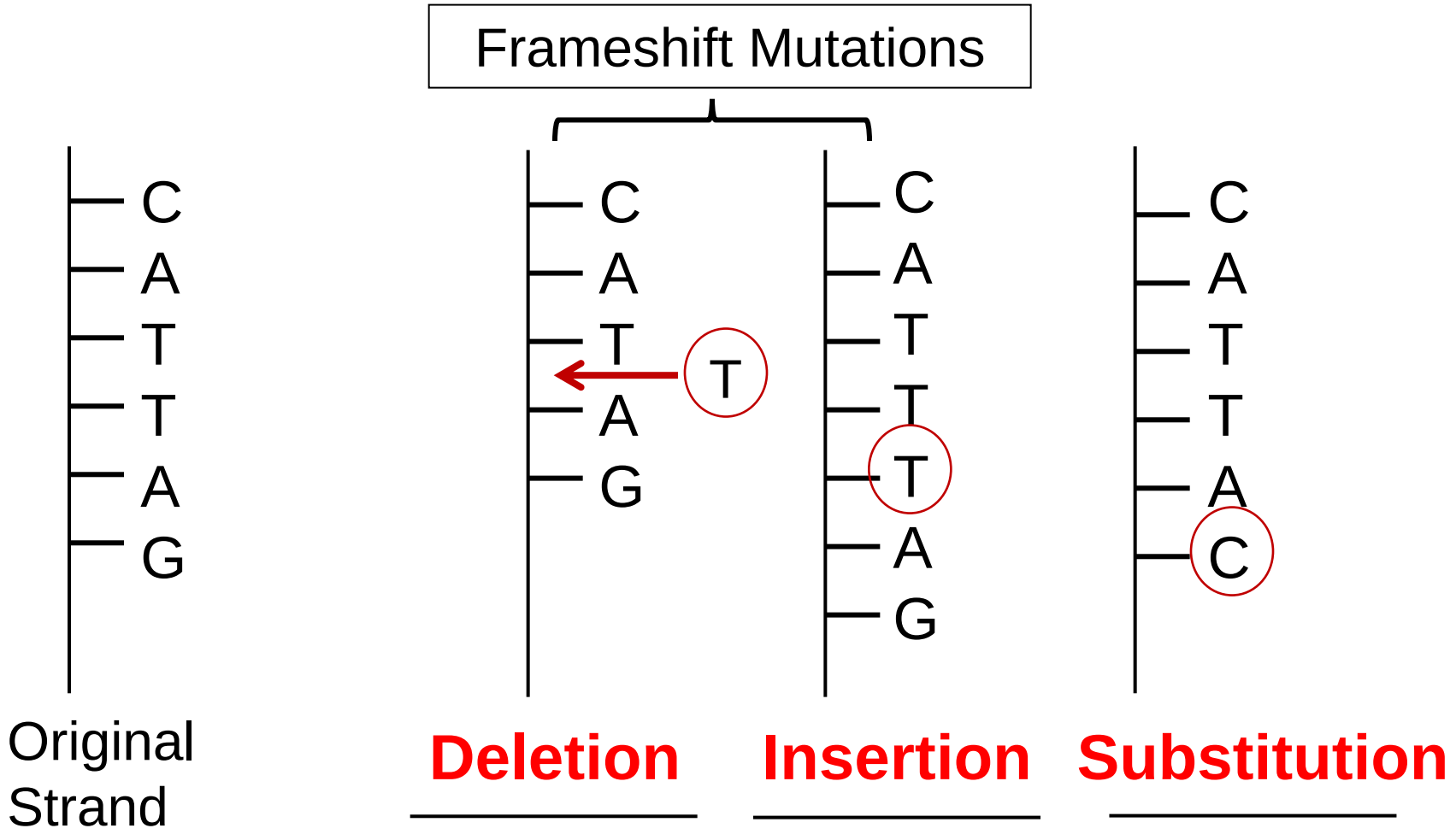
Mutation in
egg or sperm

All cells
affected in
offspring

Adapted from the National Cancer Institute and the American Society of Clinical Oncology

Types of Gene Mutations:

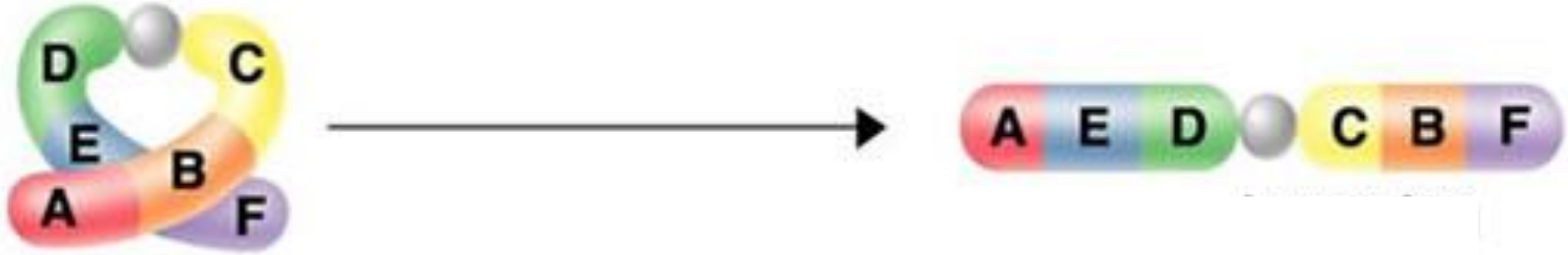
- change in nitrogenous base sequence
- May occur during Replication or Transcription



Types of Chromosomal Mutations

Deletion: loss of all or part of a chromosome

Duplication: a segment of a chromosome is repeated



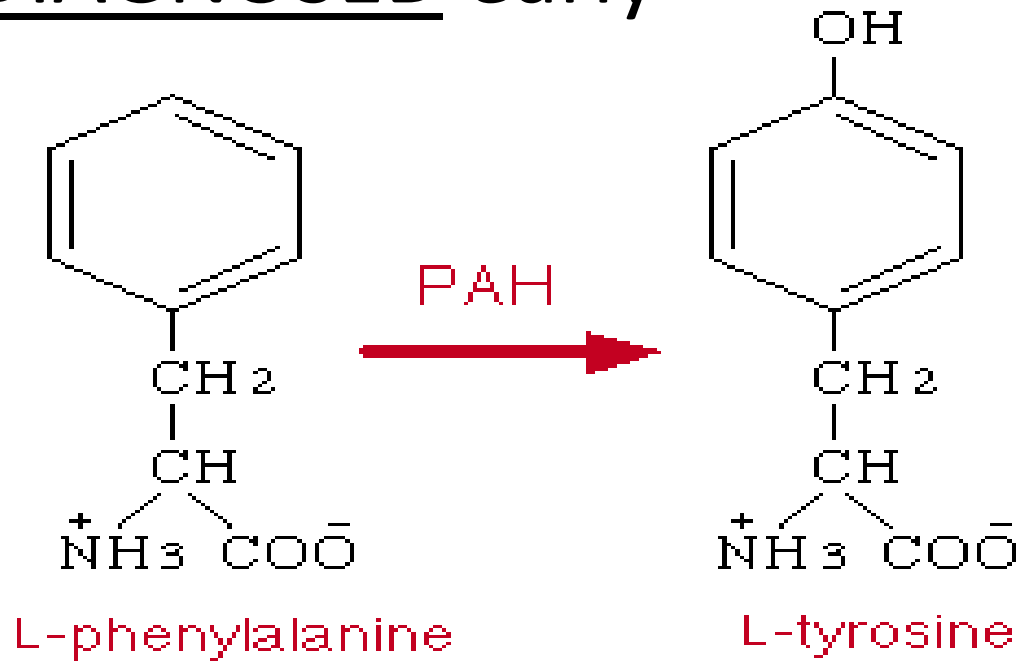
Inversion: Chromosome sections become disoriented

Translocation: part of one chromosome breaks off and attaches to a NON-homologous chromosome

Some genetic disorders:

1. PKU (phenylketonuria)

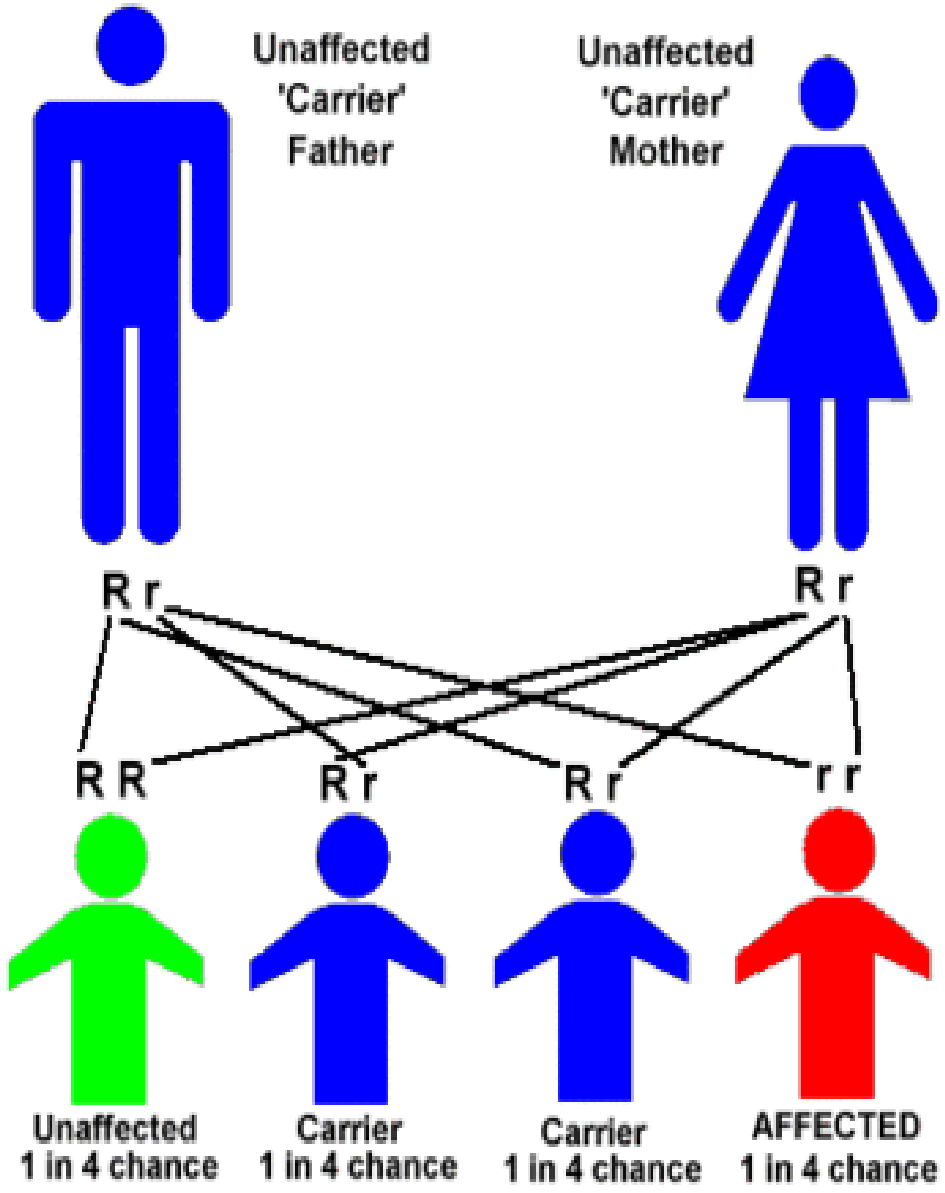
- Buildup of amino acid phenylalanine in tissue due to missing enzyme (brain damage, mental retardation)
- can be CONTROLLED by diet if DIAGNOSED early



The enzyme phenylalanine hydroxylase converts the amino acid phenylalanine to tyrosine.

2. Tay Sach's Disease

- Lipid buildup in brain cells
- causes deterioration of mental and physical abilities
- usually results in death by the age 5

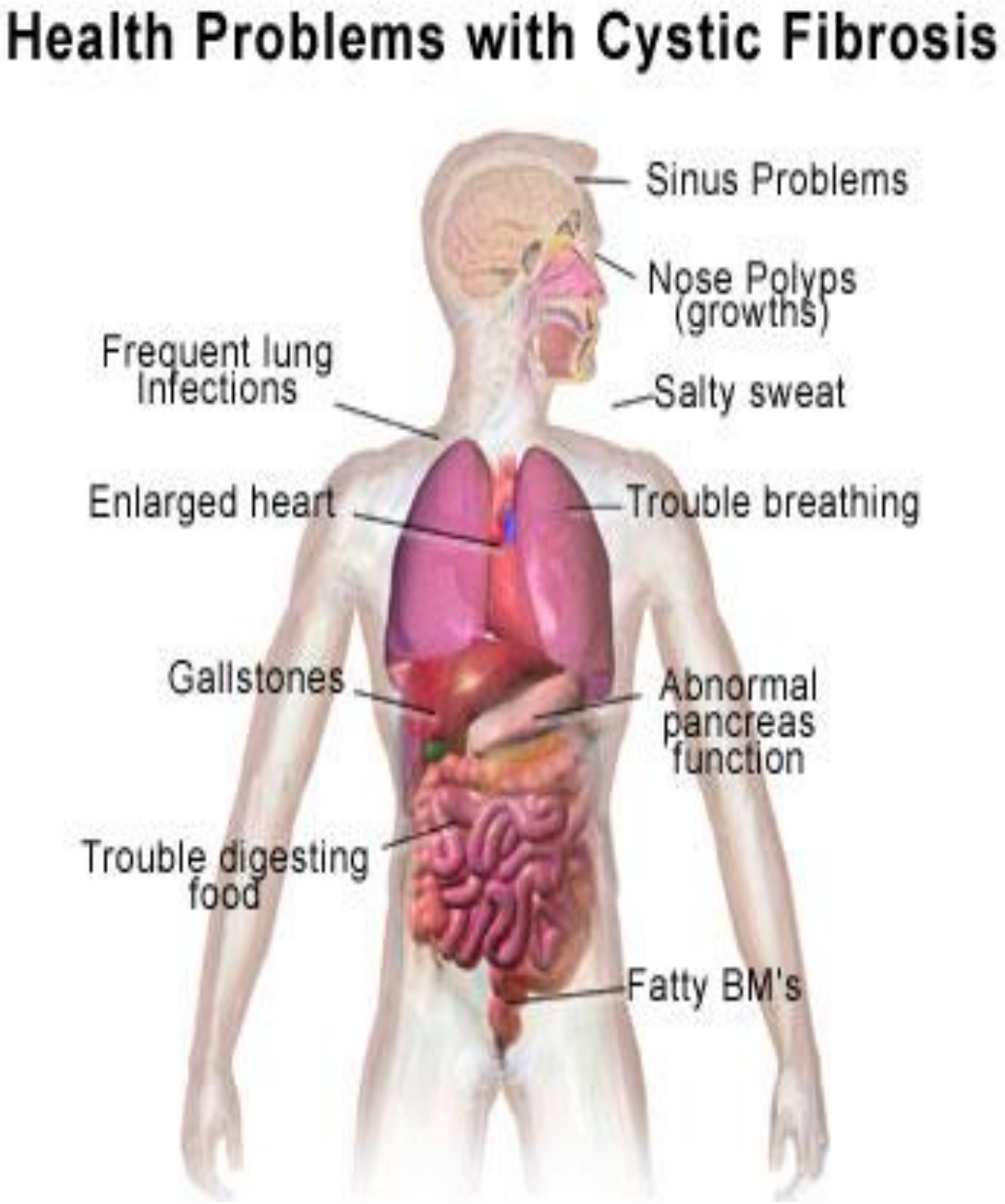


mutations on chromosome 15 in the *HEXA* gene

3. Cystic Fibrosis

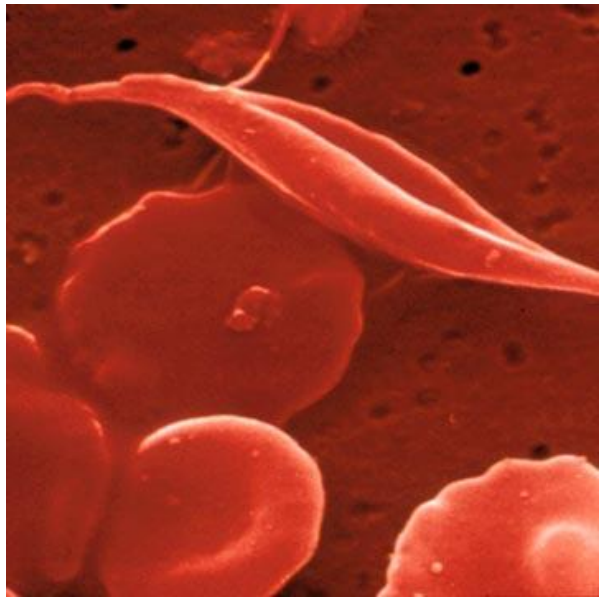
- thick, sticky mucus buildup in the lungs and digestive tract
- Death in young adulthood

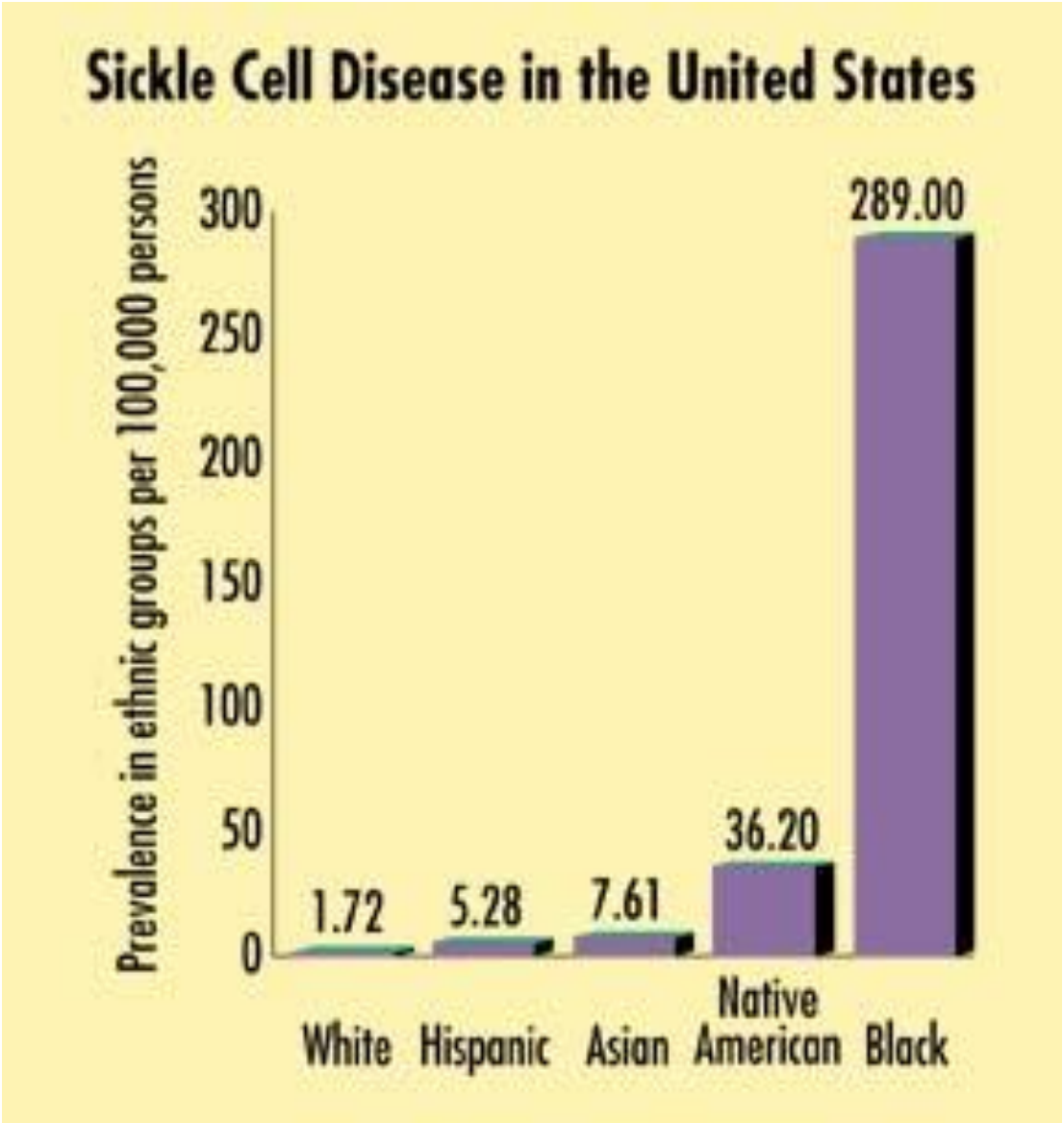
CF is caused by a mutation in the gene cystic fibrosis transmembrane conductance regulator (CFTR). The most common mutation, ΔF508, is a deletion (Δ) of three nucleotides that results in a loss of the amino acid phenylalanine (F) at the 508th (508) position on the protein.



4. Sickle Cell Anemia

- Sickle-shaped red blood cells are fragile and prone to rupture
- can block blood vessels causing tissue and organ damage and pain





Lesson 8

Detection of Disorders

Screening (blood & urine)

Amniocentesis

Karyotypes

Chromosomal Disorders

- Down Syndrome
- Kleinfelters
- Turner Syndrome

Detection of Genetic Disorders

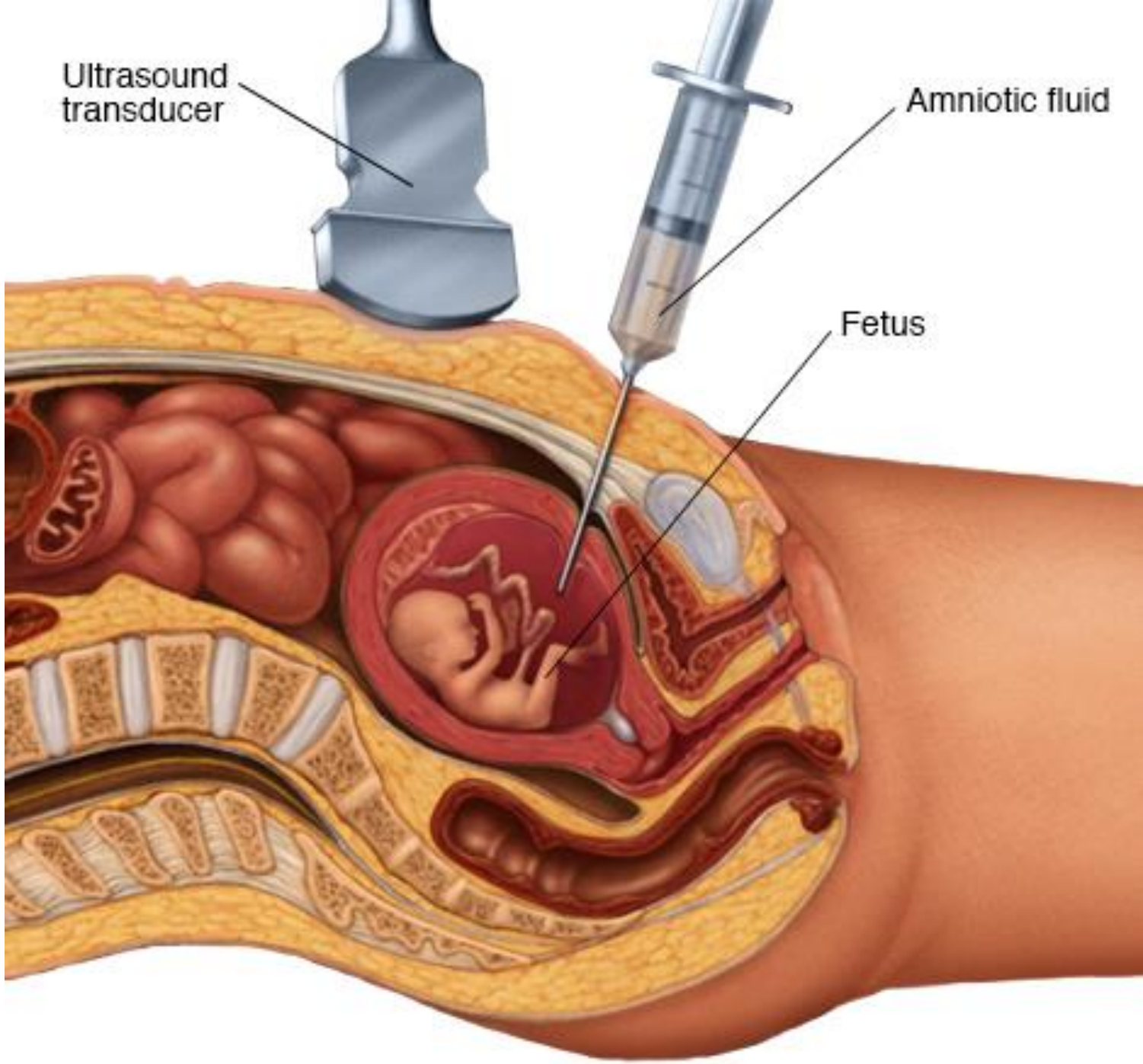
1. Screening

- Chemical analysis of body fluids (ex. Urine and blood)
- Indicates presence of chemicals that are associated with genetically related disorders

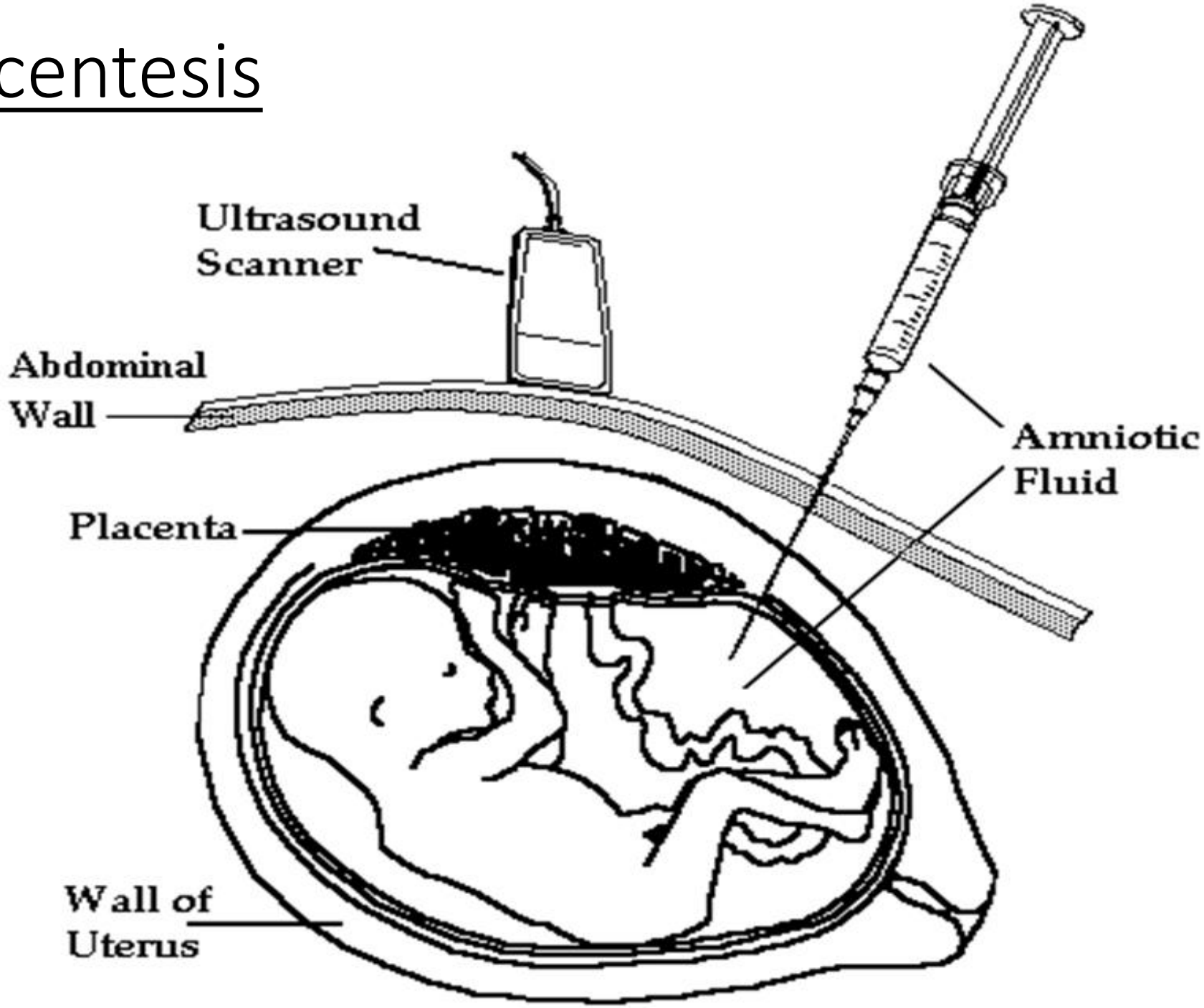
2. Amniocentesis:

removal of amniotic fluid from the uterus of a pregnant female for genetic analysis of fetal cells

Amniocentesis Procedure
(animation)



Amniocentesis

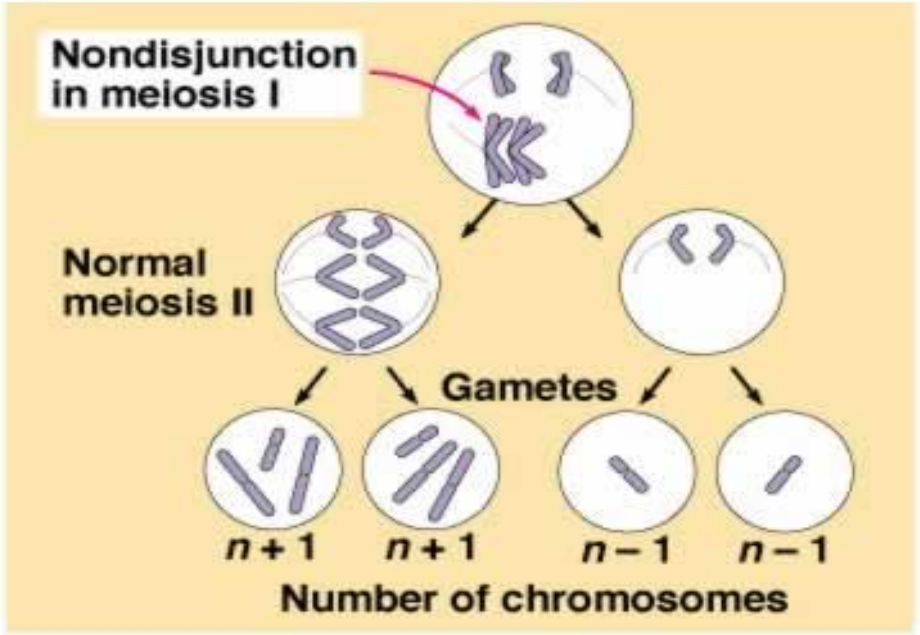


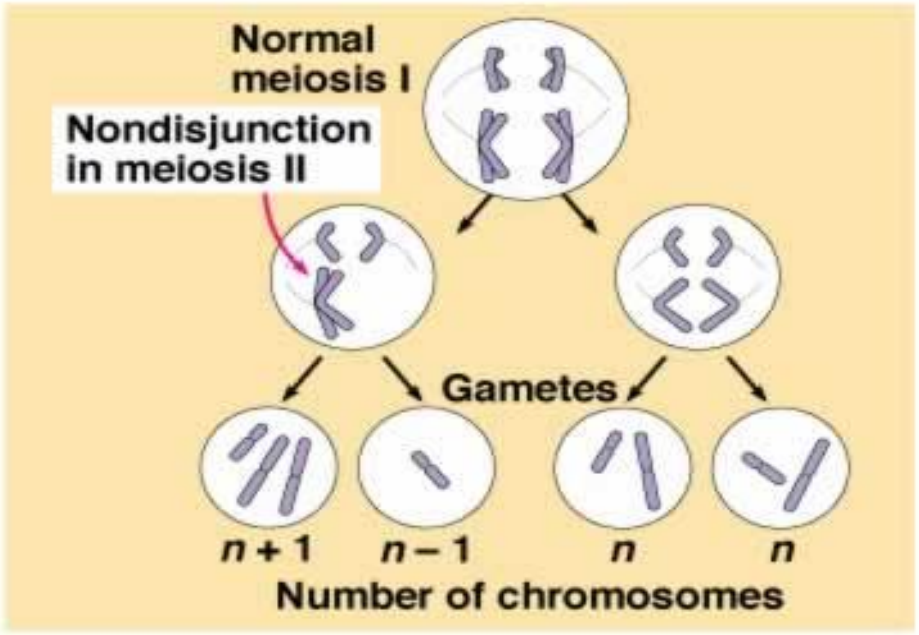
3. Karyotyping:

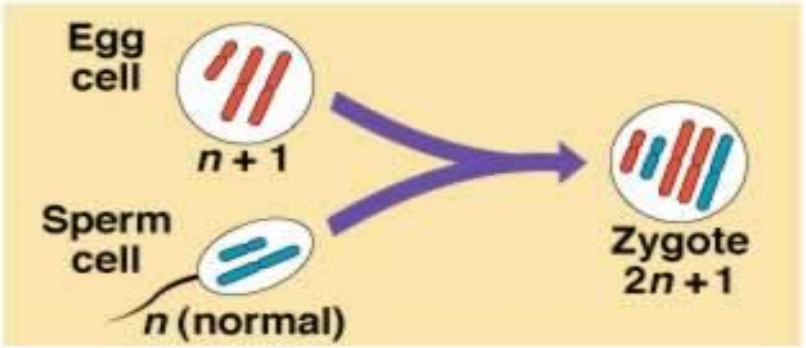
- Arrangement of chromosomes in homologous pairs in descending order by size
- Can identify some chromosomal abnormalities & sex

How can extra/missing chromosomes occur?

Nondisjunction







Chromosomal Genetic Disorders

1. Turner Syndrome

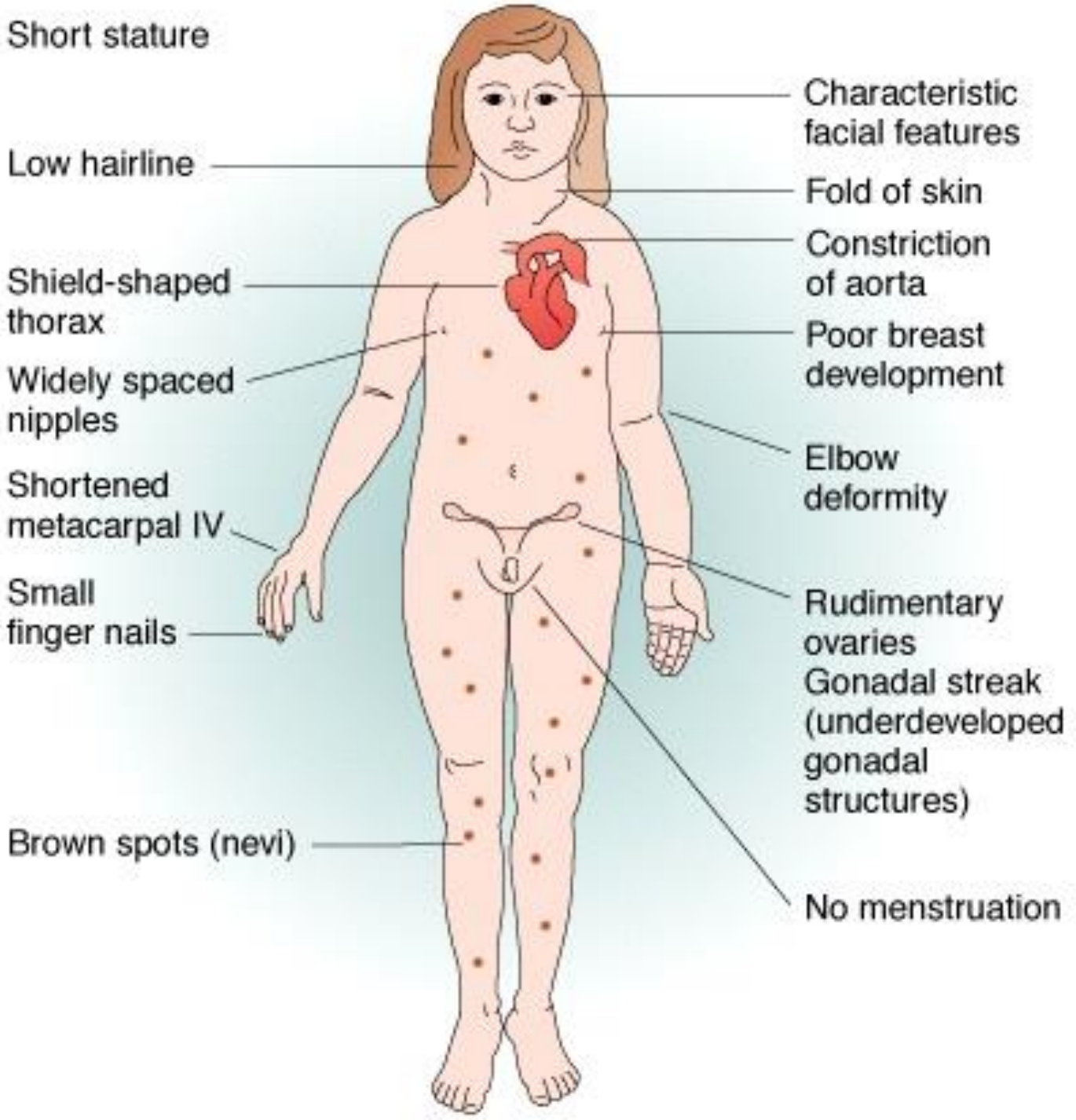
Female missing one X chromosome

Genotype 45, X0

1 in 2,500 female births

Characteristics of Turner Syndrome

- Short
- No puberty
- Child-like body
- Infertile

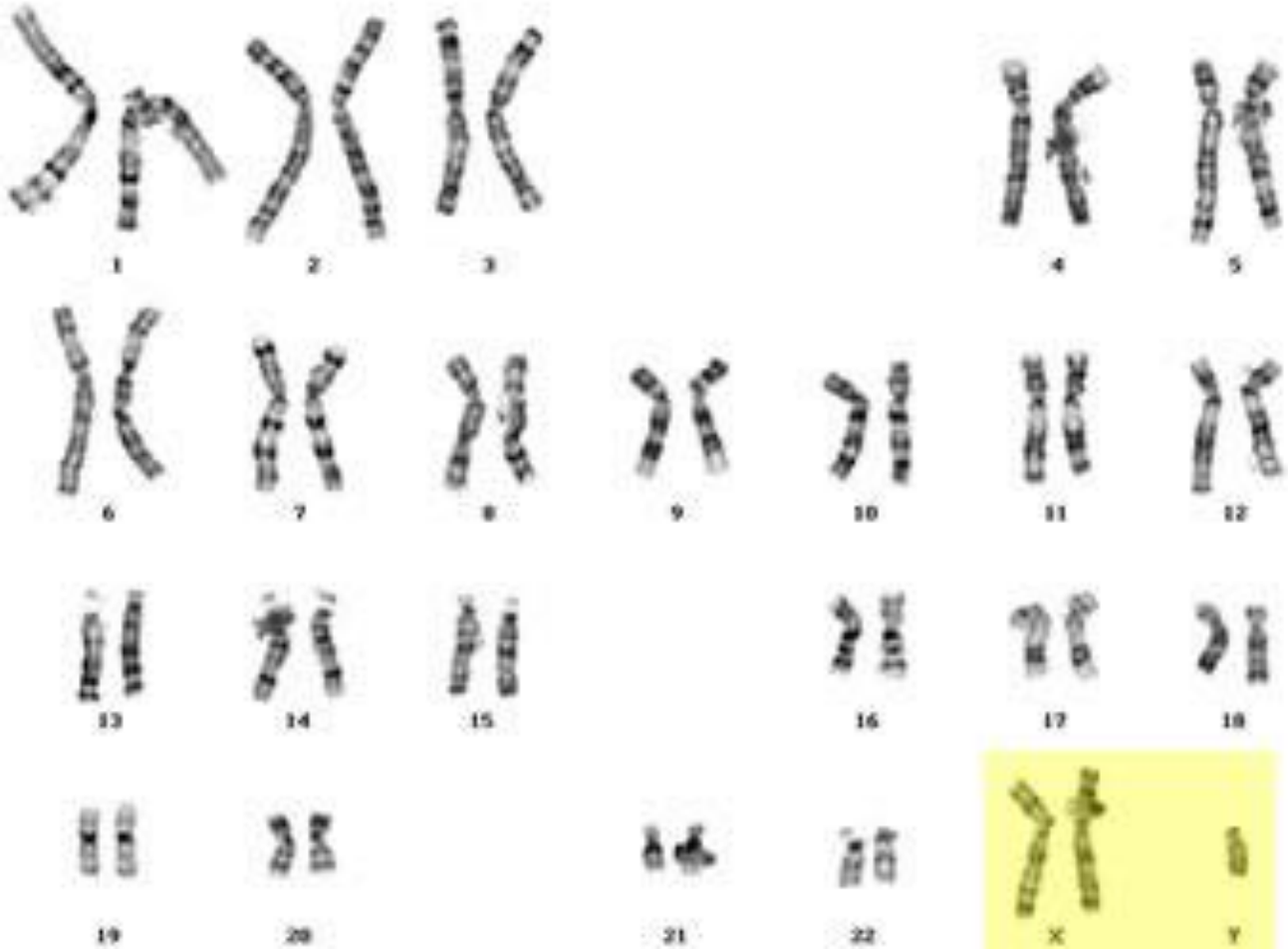


2. Klinefelter Syndrome

Male with an extra X chromosome

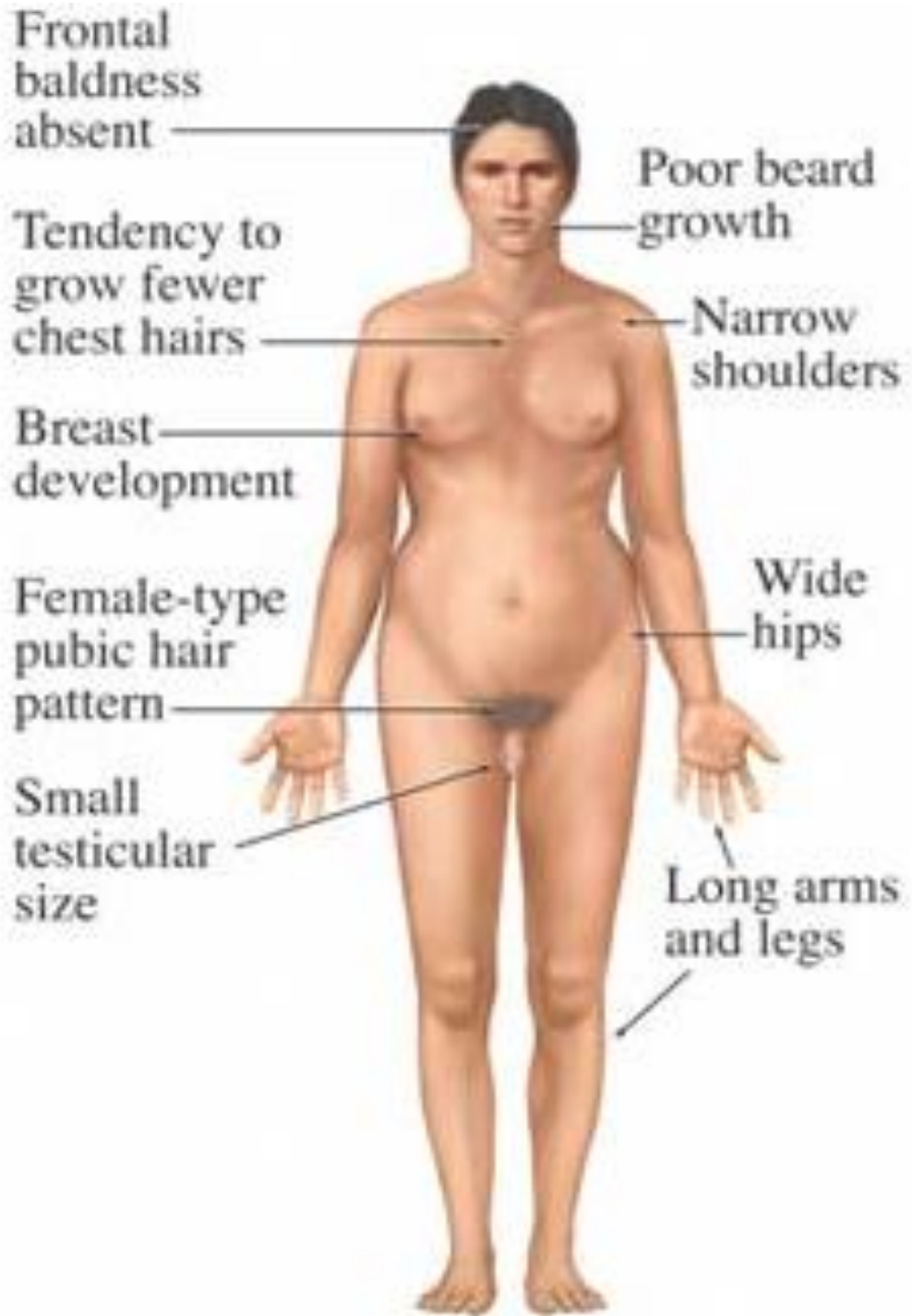
Genotype – 47, XXY

1 in 500 male births



Characteristics of Klinefelter Syndrome

- Infertile
- Breast development
- Narrow shoulders
- Wide hips
- Less body hair

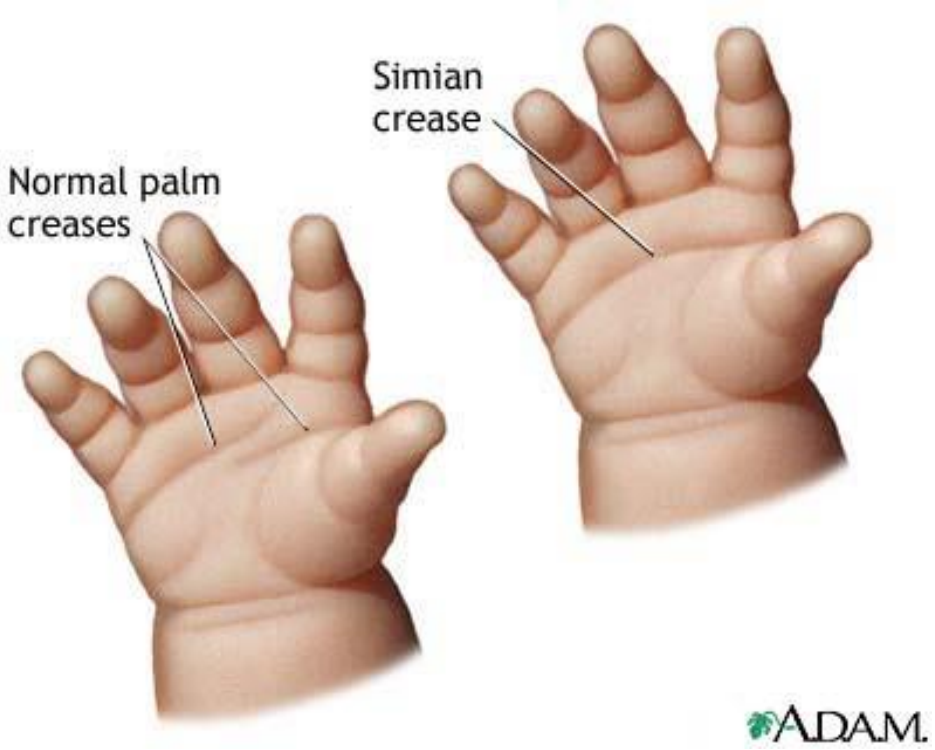


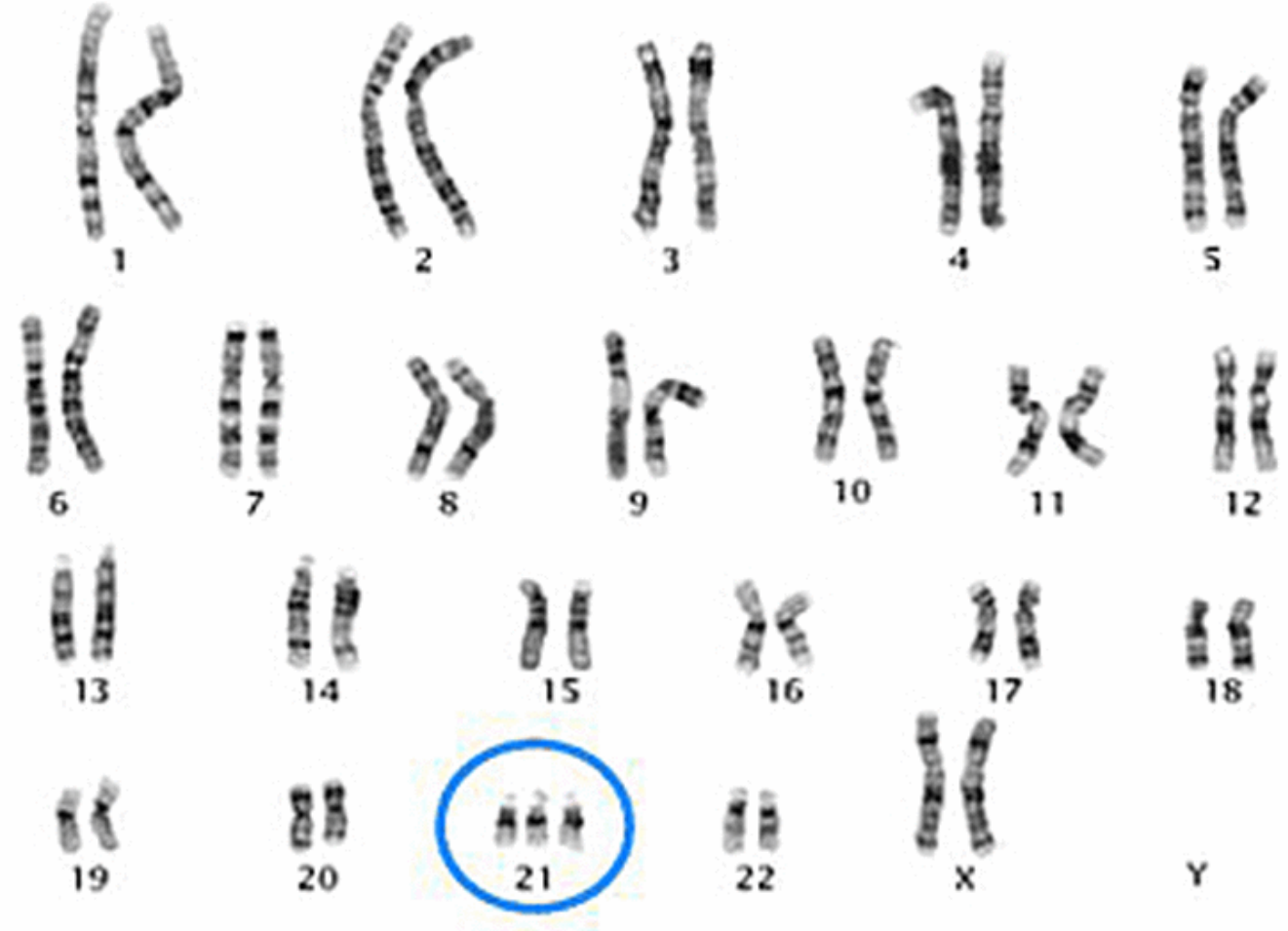
3. Down Syndrome

Trisomy 21 (Extra chromosome #21)

Genotype – 47 XX or XY

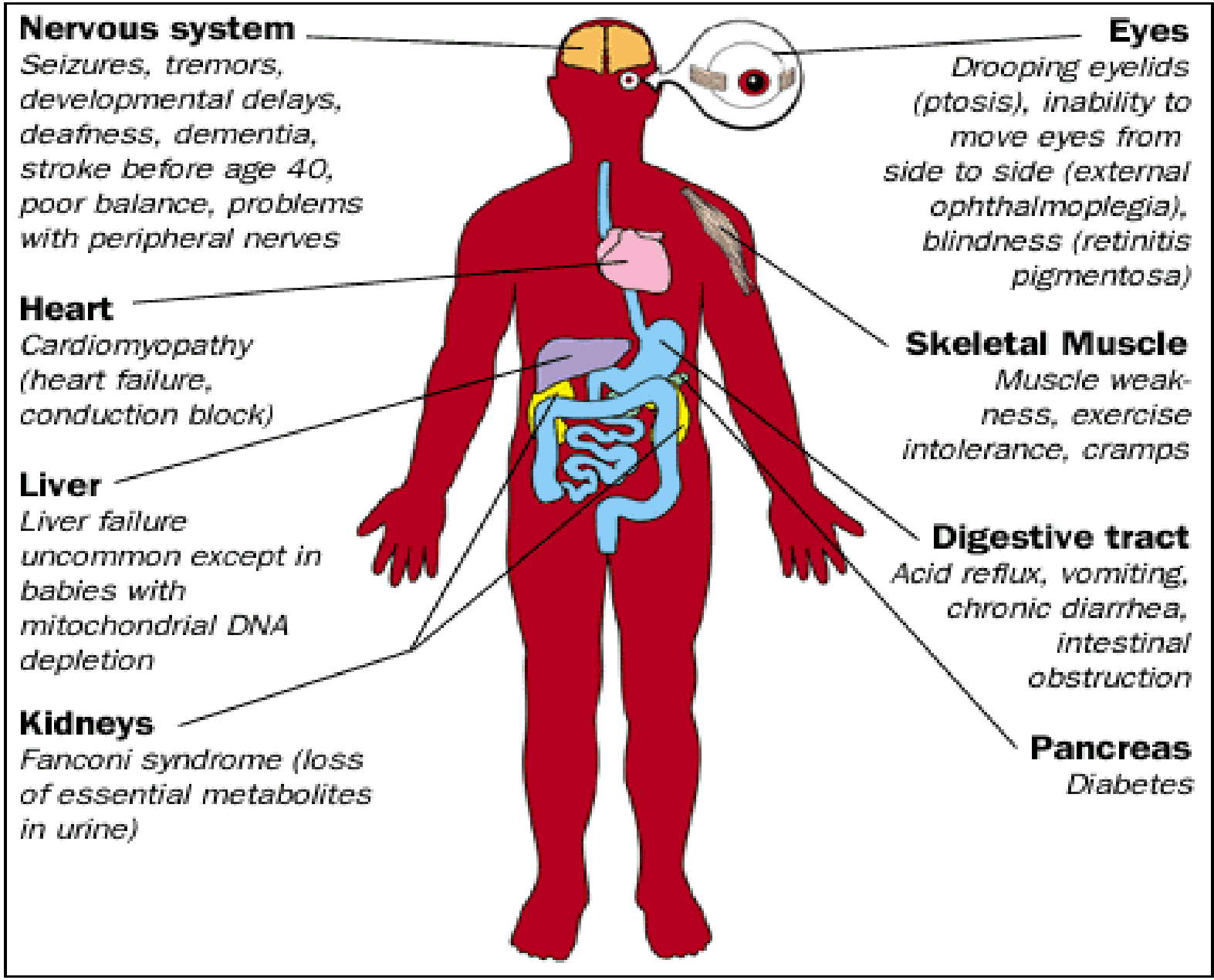
1 in 700 births



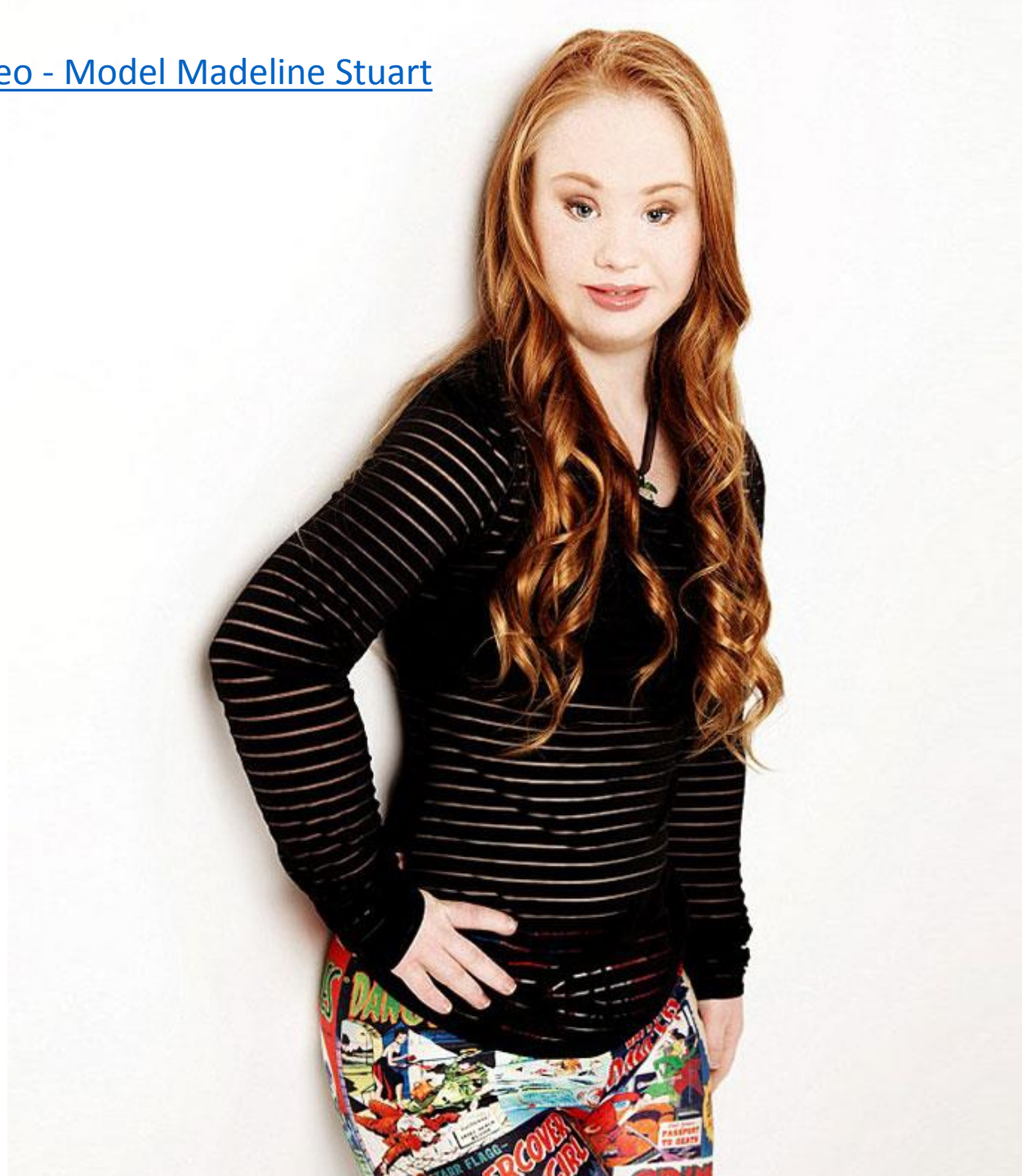


Characteristics of Down Syndrome

- Drooping eyelids
- Flat facial features
- Deep palm crease
- Enlarged tongue
- Higher chance of heart defects



[Video - Model Madeline Stuart]



Lesson 9

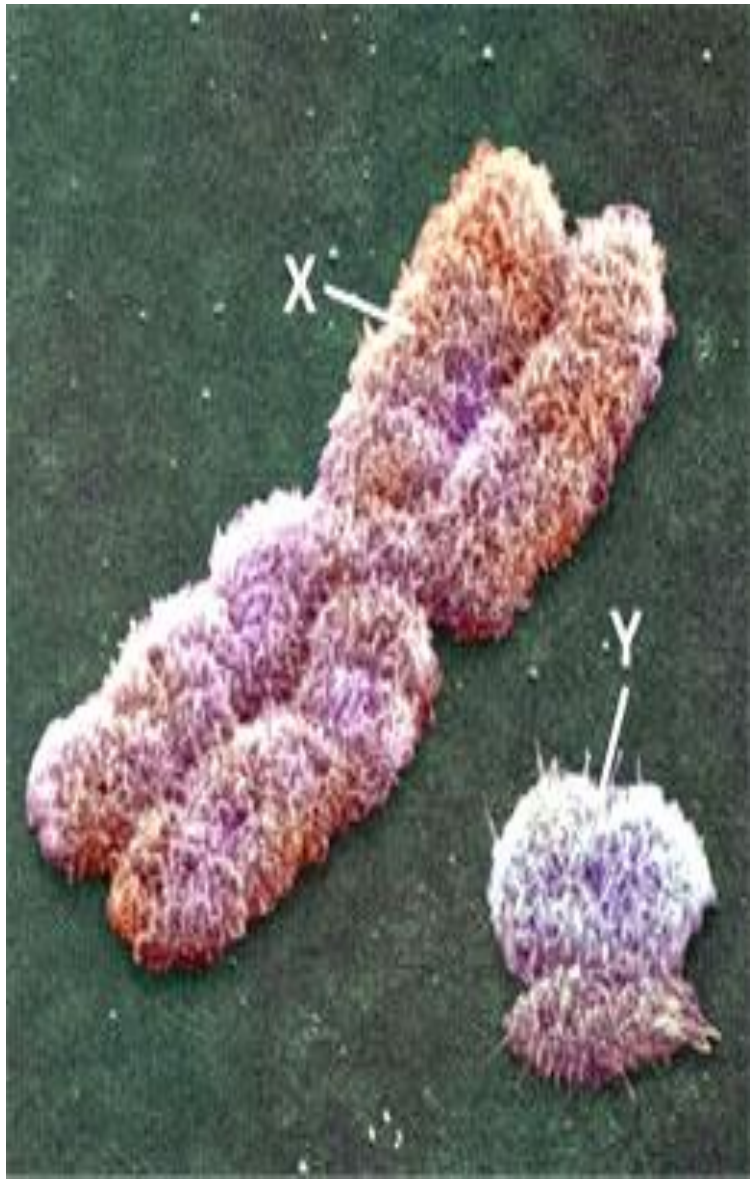
Sex-linked genes & disorders

Genetic counseling

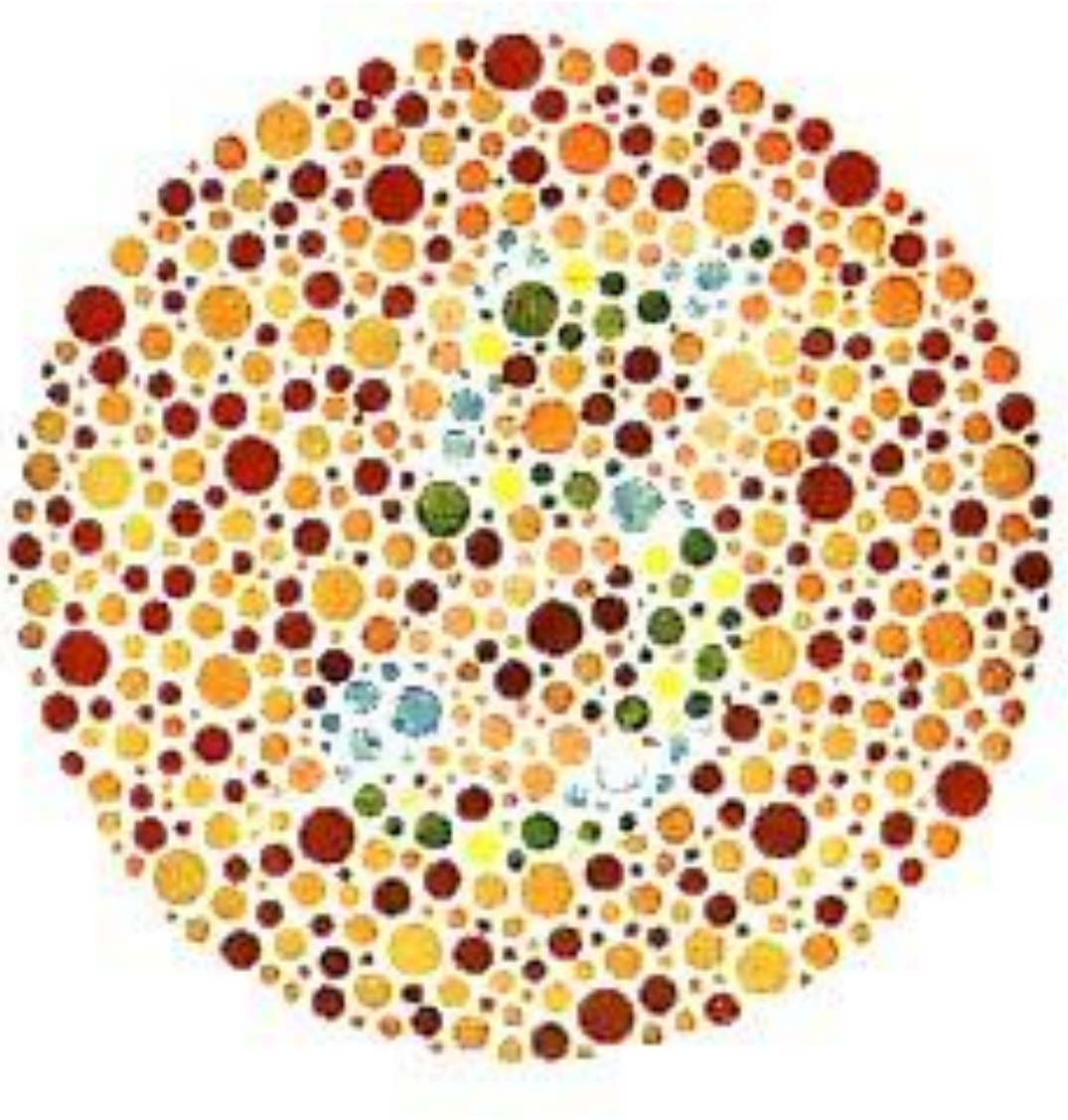
Pedigrees

Sex Linked Traits / Disorders

- Coded for by genes located on the X and Y chromosomes (most on X)
- More than 100 sex-linked disorders
- More often expressed in males because they only have one X chromosome
  - Ex. Colorblindness, hemophilia, baldness



Colorblindness - mutations on the X chromosome



Video - Color Blindness test

Possible Colorblindness Cure?

- Mapping of the human genome has shown there are many causative mutations for colorblindness. Mutations capable of causing color blindness originate from at least 19 different chromosomes and 56 different genes!

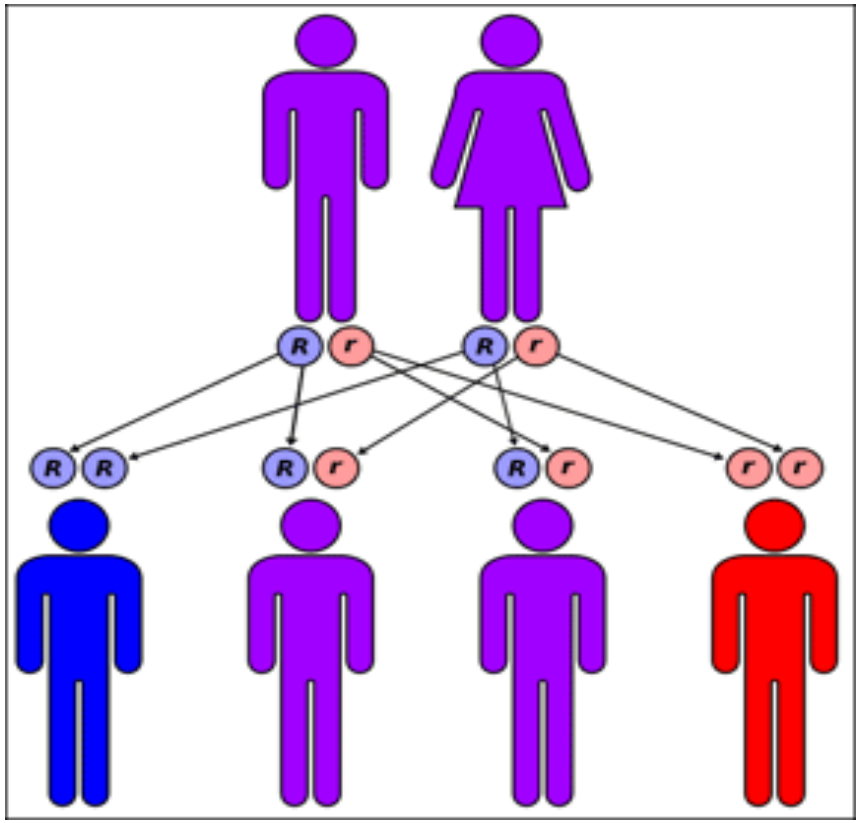
153 million base pairs

Short stature, idiopathic familial
Leri-Weill dyschondrosteosis
Langer mesomelic dysplasia
Leukemia, acute myeloid, M2 type
Chondrodysplasia punctata
Kallmann syndrome
Ocular albinism, Nettleship-Falls type
Oral-facial-digital syndrome
Nance-Horan cataract-dental syndrome
Heterocellular hereditary persistence of fetal hemoglobin
Pyruvate dehydrogenase deficiency
Glycogen storage disease
Coffin-Lowry syndrome
Mental retardation
Spondyloepiphyseal dysplasia tarda
Paroxysmal nocturnal hemoglobinuria
Infantile spasm syndrome
Aicardi syndrome
Deafness, sensorineural
Simpson-Golabi-Behmel syndrome, type 2
Adrenal hypoplasia, congenital
Dosage-sensitive sex reversal
Deafness, congenital sensorineural
Retinitis pigmentosa
Wilson-Turner syndrome
Cone dystrophy
Aland island eye disease (ocular albinism)
Optic atrophy
Night blindness, congenital stationary, type 1
Erythroid-potentiating activity
Arthrogryposis multiplex congenita
Night blindness, congenital stationary, type 2
Brunner syndrome
Wiskott-Aldrich syndrome
Thrombocytopenia
Dent disease
Nephrolithiasis, type I
Hypophosphatemia, type III
Proteinuria
Anemia, sideroblastic/hypochromic
Cerebellar ataxia
Renal cell carcinoma, papillary
Diabetes mellitus, insulin-dependent
Sutherland-Haan syndrome
Cognitive function, social
Mental retardation, nonspecific
Menkes disease
Occipital horn syndrome
Cutis laxa, neonatal
FG syndrome
Immunodeficiency, moderate and severe
Miles-Carpenter syndrome

Hodgkin disease susceptibility, pseudoautosomal
Ichthyosis
Microphthalmia, dermal aplasia, and sclerocornea
Episodic muscle weakness
Mental retardation
Ocular albinism and sensorineural deafness
Amelogenesis imperfecta
Charcot-Marie-Tooth disease, recessive
Keratosis follicularis spinulosa decalvans
Hypophosphatemia, hereditary
Partington syndrome
Retinoschisis
Gonadal dysgenesis, XY female type
Mental retardation, non-dysmorphic
Agammaglobulinemia, type 2
Craniofrontonasal dysplasia
Opitz G syndrome, type I
Pigment disorder, reticulate
Melanoma
Duchenne muscular dystrophy
Becker muscular dystrophy
Cardiomyopathy, dilated
Chronic granulomatous disease
Snyder-Robinson mental retardation
Norrie disease
Exudative vitreoretinopathy
Coats disease
Renpenning syndrome
Retinitis pigmentosa, recessive
Mental retardation, nonspecific and syndromic
Dyserythropoietic anemia with thrombocytopenia
Chondrodysplasia punctata, dominant
Autoimmunity-immunodeficiency syndrome
Renal cell carcinoma, papillary
Faciogenital dysplasia (Aarskog-Scott syndrome)
Chorioathetosis with mental retardation
Sarcoma, synovial
Prieto syndrome
Spinal muscular atrophy, lethal infantile
Migraine, familial typical
Androgen insensitivity
Spinal and bulbar muscular atrophy
Prostate cancer
Perineal hypospadias
Breast cancer, male, with Reifenstein syndrome
Ectodermal dysplasia, anhidrotic
Alpha-thalassemia/mental retardation
Juberg-Marsidi syndrome
Sutherland-Haan syndrome
Smith-Fineman-Myers syndrome
Hemolytic anemia
Myoglobinuria/hemolysis

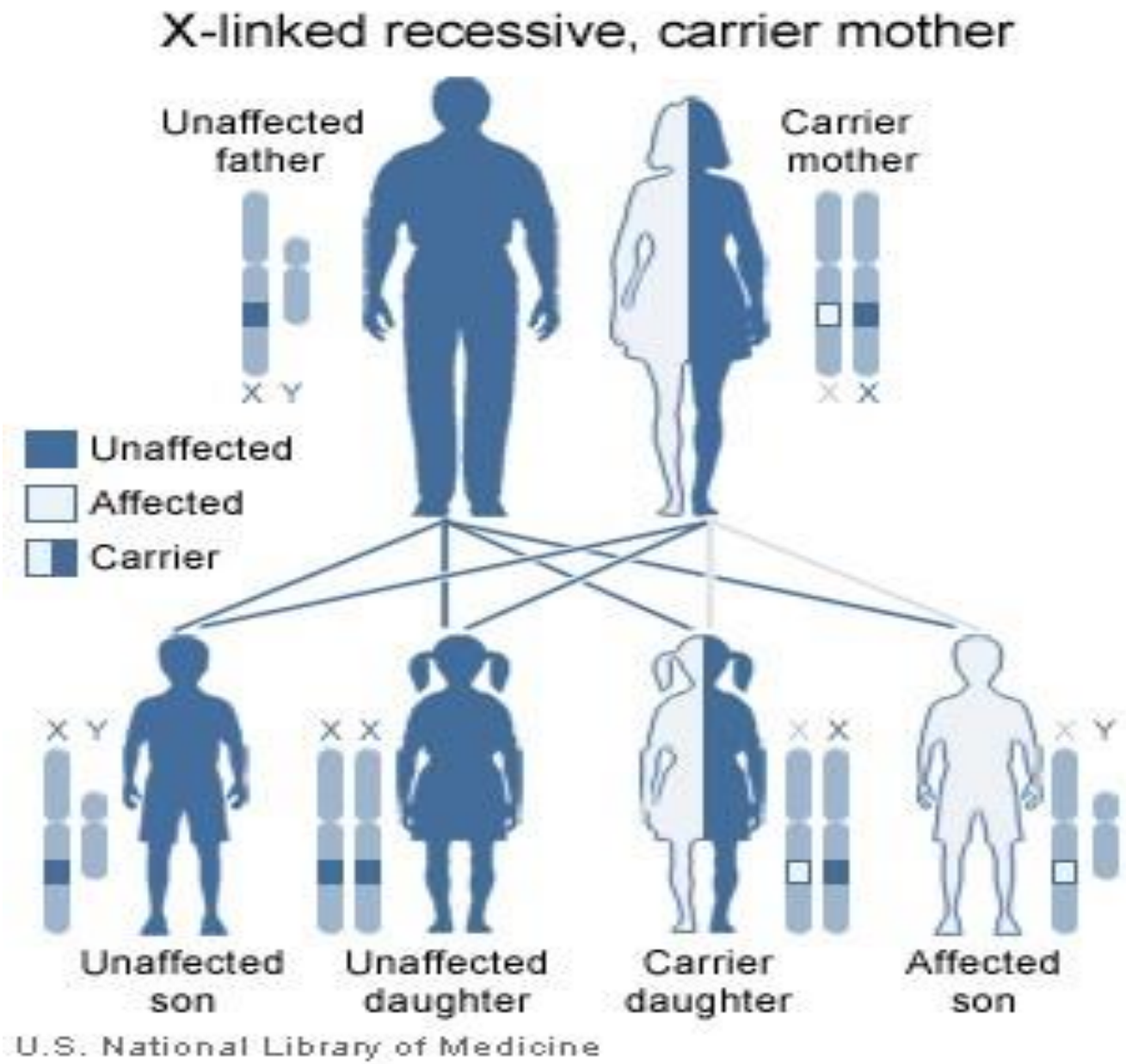
Medical Genetic Research

- diagnosing, preventing, treating, and controlling genetic disorders



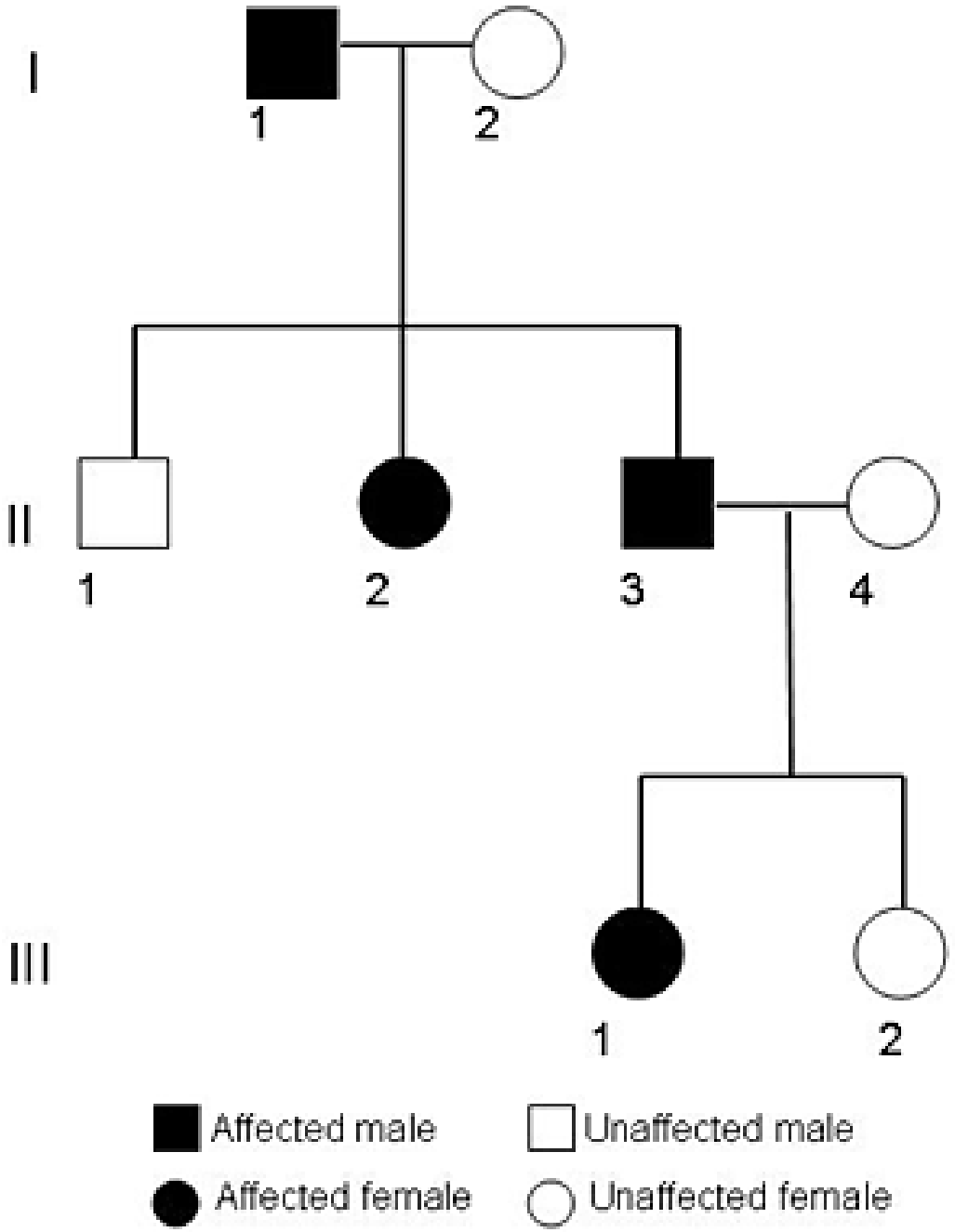
Genetic Counseling

- Involves genetic testing and prediction of disorders
- Family history is applied
- Uses pedigree charts



Pedigree Charts

- A genetic family tree
- Shows family traits over generations
- Helps counselors predict possibilities of passing on diseases to offspring

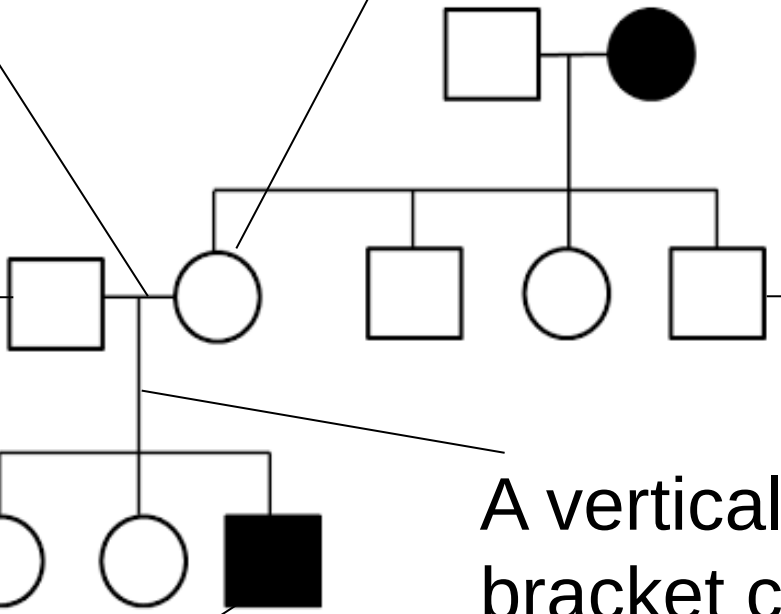


A horizontal line connecting a male and female represents a **marriage**

A circle represents a **female**

A half-shaded circle or square indicates that a person is a **carrier** (not always shown on pedigree)

A square represents a **male**

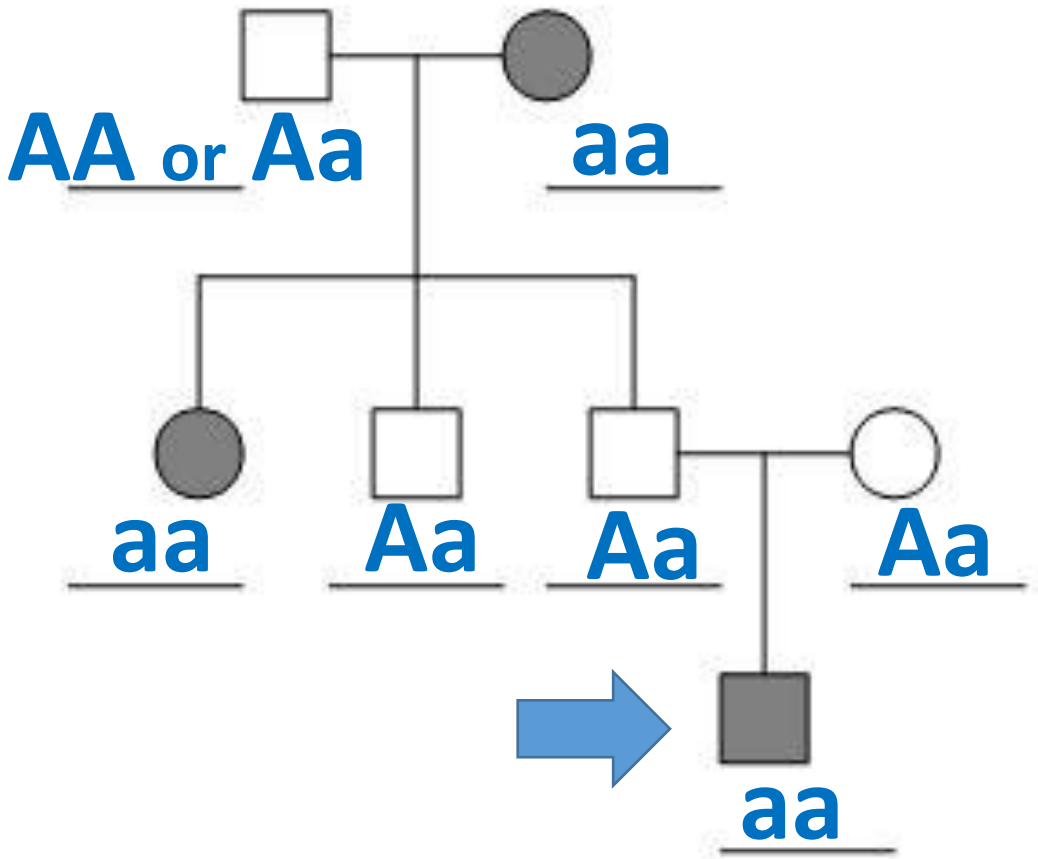


not shaded = person does **not express the trait**

A vertical line and a bracket connect the **parents to children**

shaded = person **expresses the trait**

Understanding Pedigrees



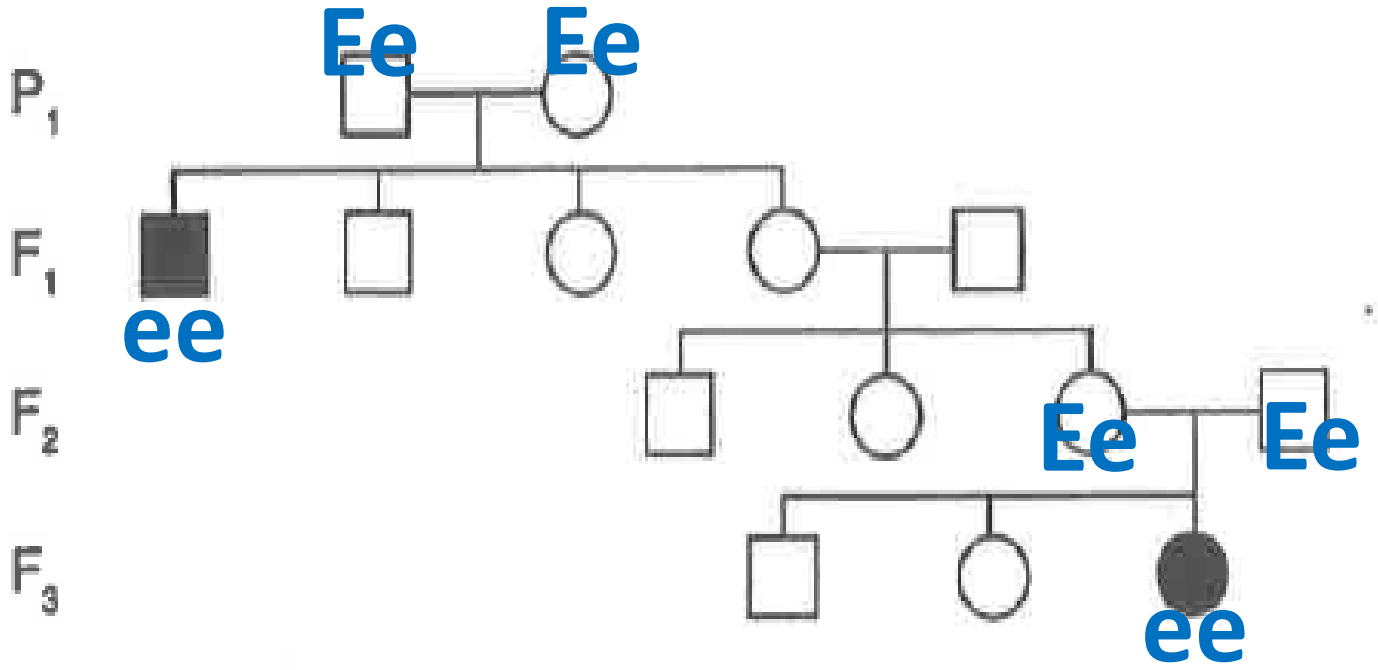
How many children does the couple in generation 1 have? **3**

How many grandchildren do they have? **1**

What is the sex of their oldest child? **female**

What is the sex of the grandchild? **male**

The pedigree shows the inheritance of attached earlobes for four generations.

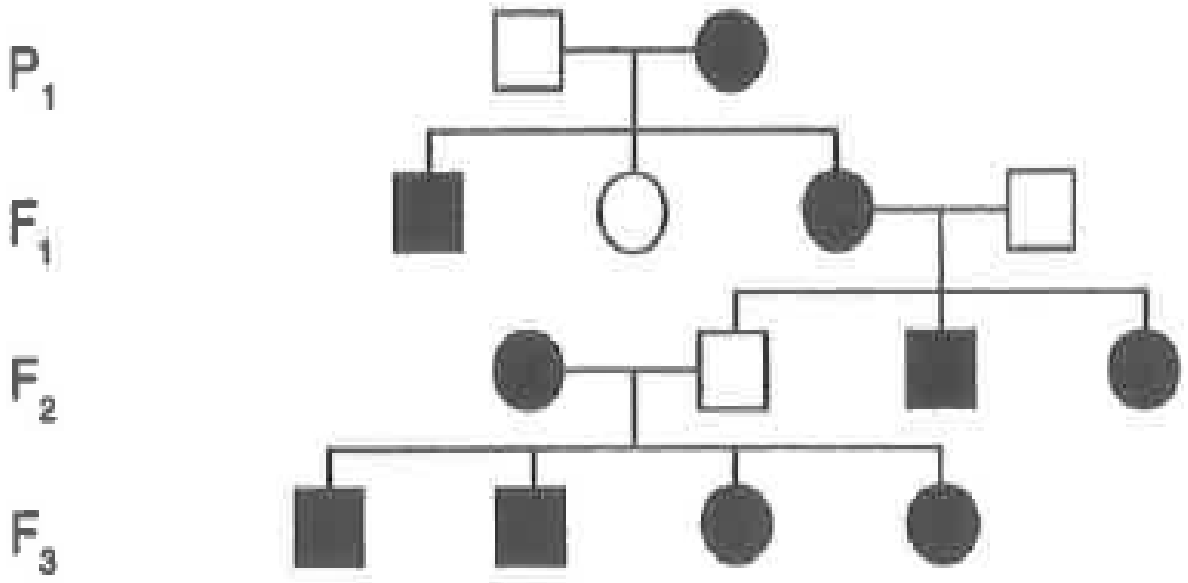


Is the trait for attached earlobes, versus free earlobes, dominant or recessive?

recessive How do you know?

If it were dominant then at least 1 parent of a child who has the trait would also have the trait

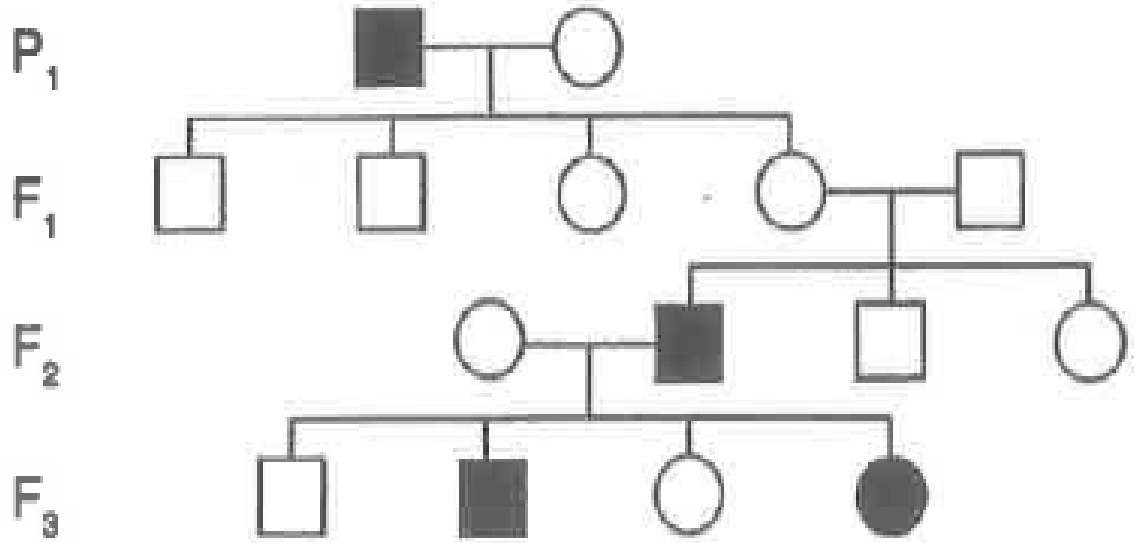
The pedigree shows the inheritance of tongue rolling.



Is this trait dominant or recessive? dominant Explain. _____

All affected individuals have parents that are also affected.

This pedigree shows the inheritance of colorblindness, a sex-linked trait.



(F2 male has it but not parents)

Is this trait dominant or recessive? recessive Is the mother of the colorblind girl in the F_3 generation colorblind, a carrier, or a person with normal color vision?

Mother of the color blind girl in F3 generation must be a carrier for colorblindness (heterozygous)