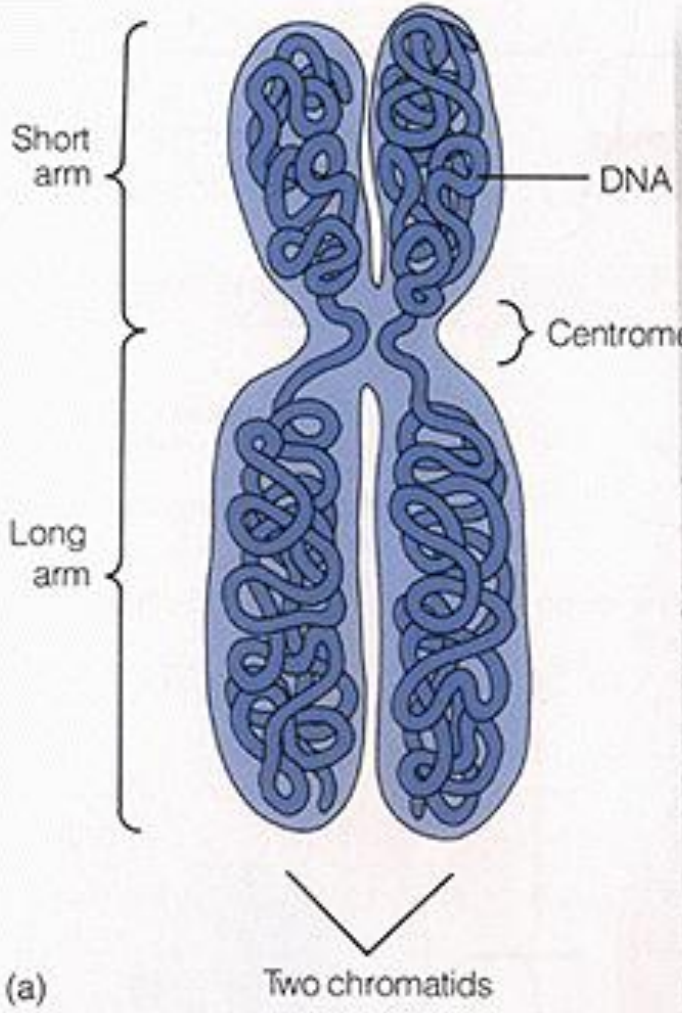
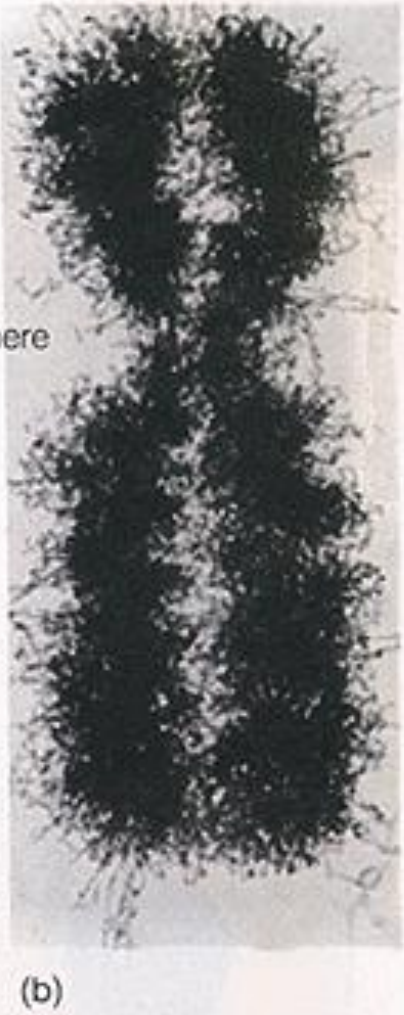
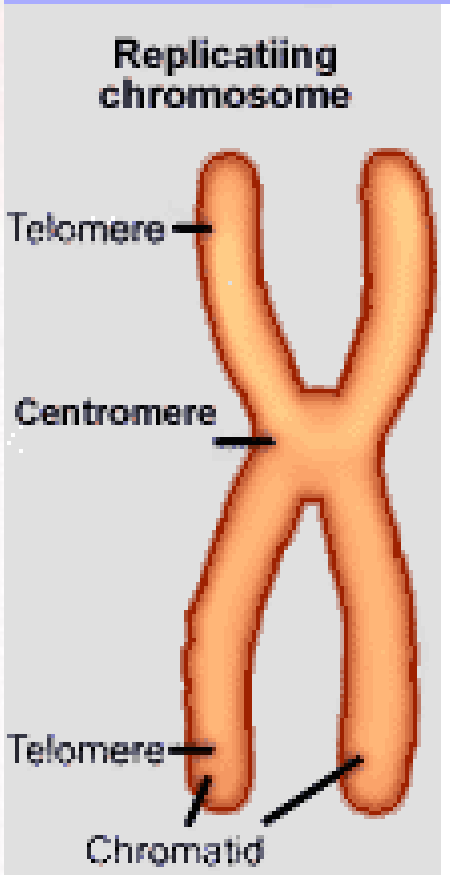
Homologous Chromosomes? Sister Chromatids? Centromere?

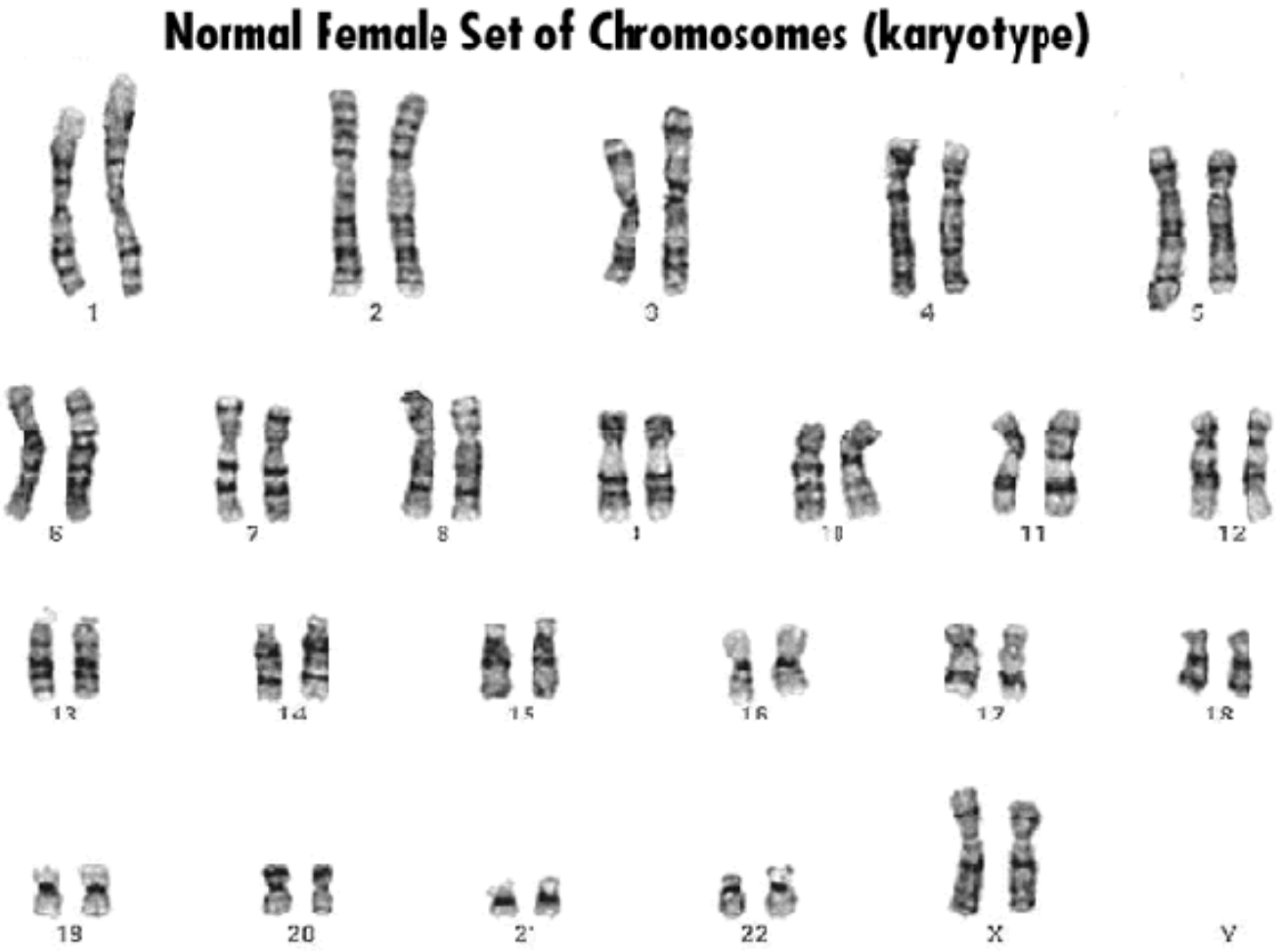




When chromosomes are preparing to divide the DNA replicates itself into two strands called chromatids



Autosomes and Sex Chromosomes



- 22 pairs of homologous autosomes and 1 pair sex chromosomes

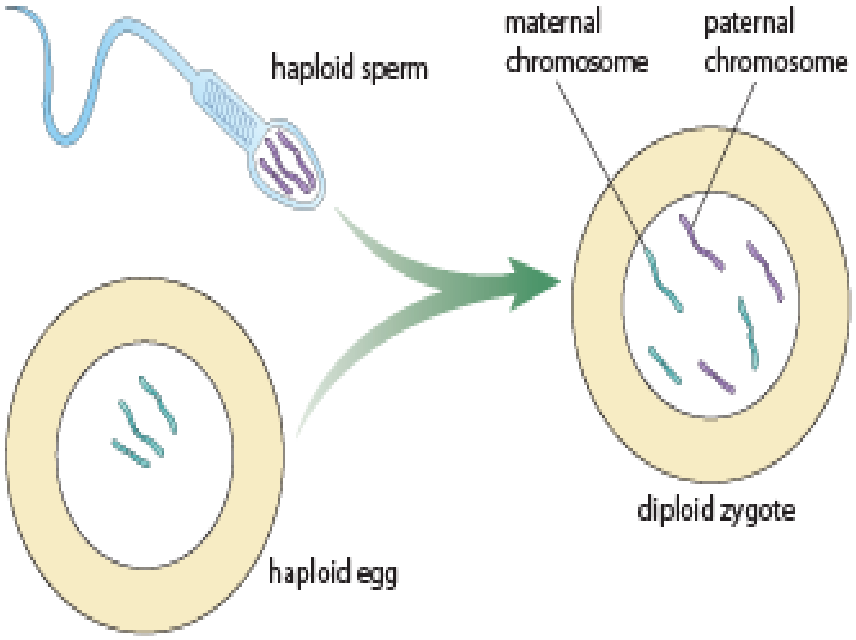
Cancer

- Uncontrolled Cell growth forms tumours
- Little time in interphase
- Unspecialized cells - non-functional
- Metastasize as no adhesion to other cells

Cloning

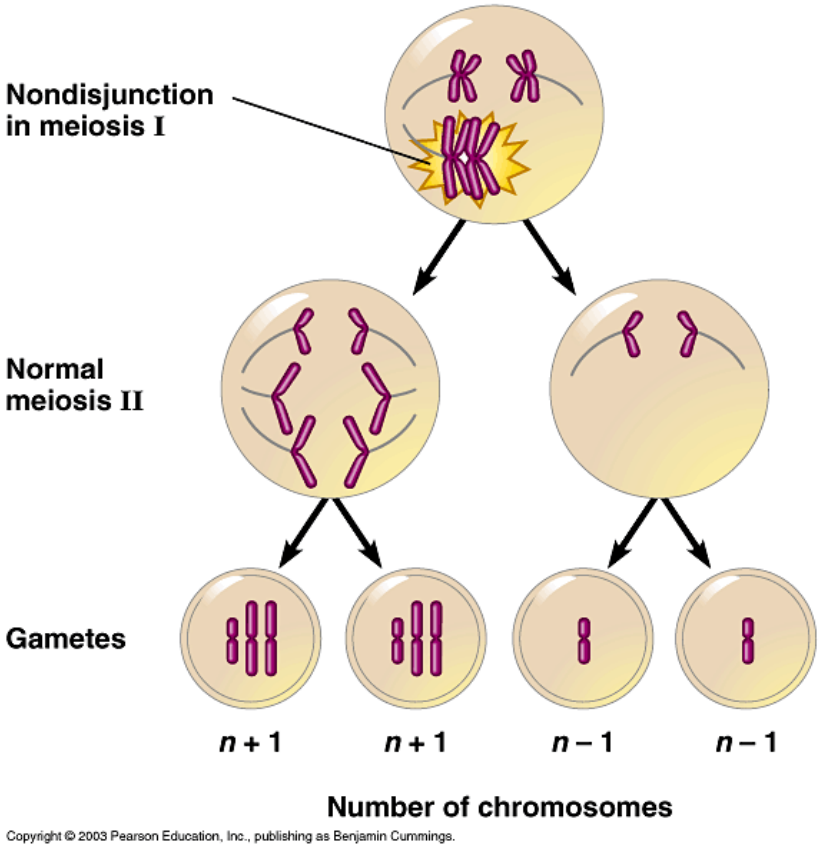
- Mitosis
- Budding (yeast)
- Vegetative propagation (plants runners)
- Identical twins (monozygotic)
- Adult Stem cells e.g.: bone marrow produces blood cells
- Embryonic Stem Cells from blastula inner mass are Totipotent (produce entire organism)
- Blastula nucleus transplanted
- Parthenogenesis

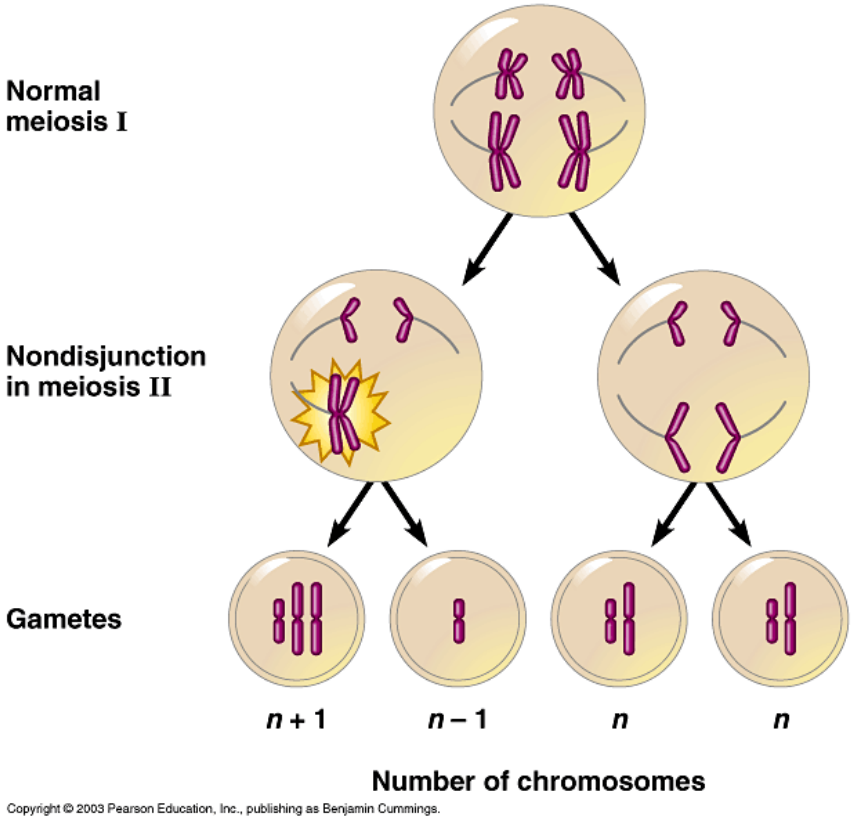
Why form gametes? (nonsomatic cells)



- Diploid somatic (2n) → Haploid Gamete (n)
- Union produces zygote
- Genetic recombination of genes from both parents for natural selection

Nondisjunction





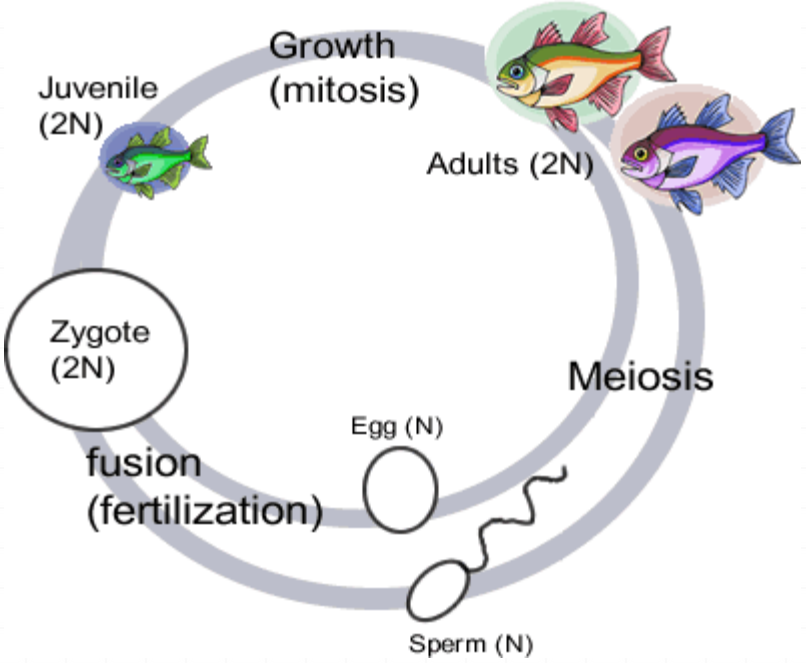
- Trisomy – 1 extra chromosome examples?
- Monosomy – 1 less chromosome example?

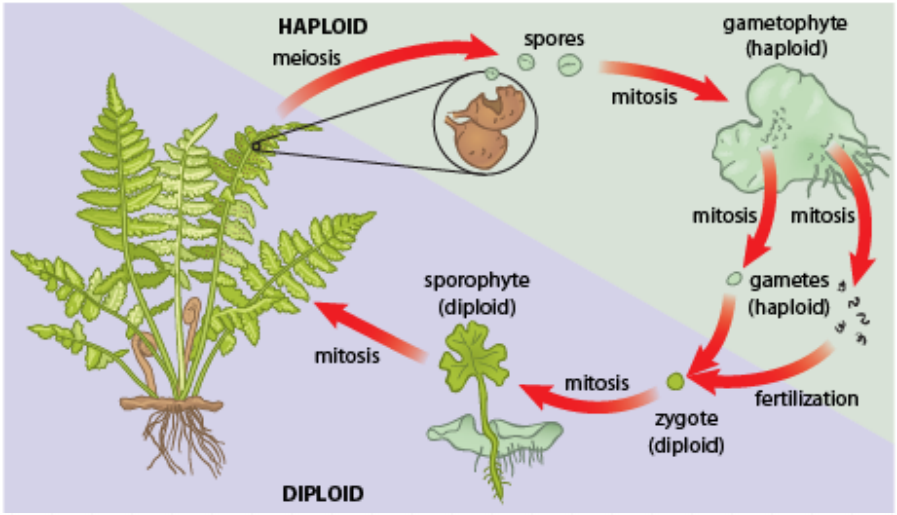
Comparison

- **Mitosis**
  - Somatic cells
  - 2 identical cells produced
  - 2n→2n **or** n→n
  - No pairing or crossing over of homologs
- **Meiosis**
  - Gonads
  - 4 different cells produced
  - 2n→n
  - Pairing of homologous chromosomes
  - Crossing over

Alternation of Generations

- Some organisms alternate between sexual and asexual reproduction
- Sporophyte (diploid) produces spores by Meiosis
- Gametophyte (haploid) produces gametes by Mitosis
- Compare processes in animals





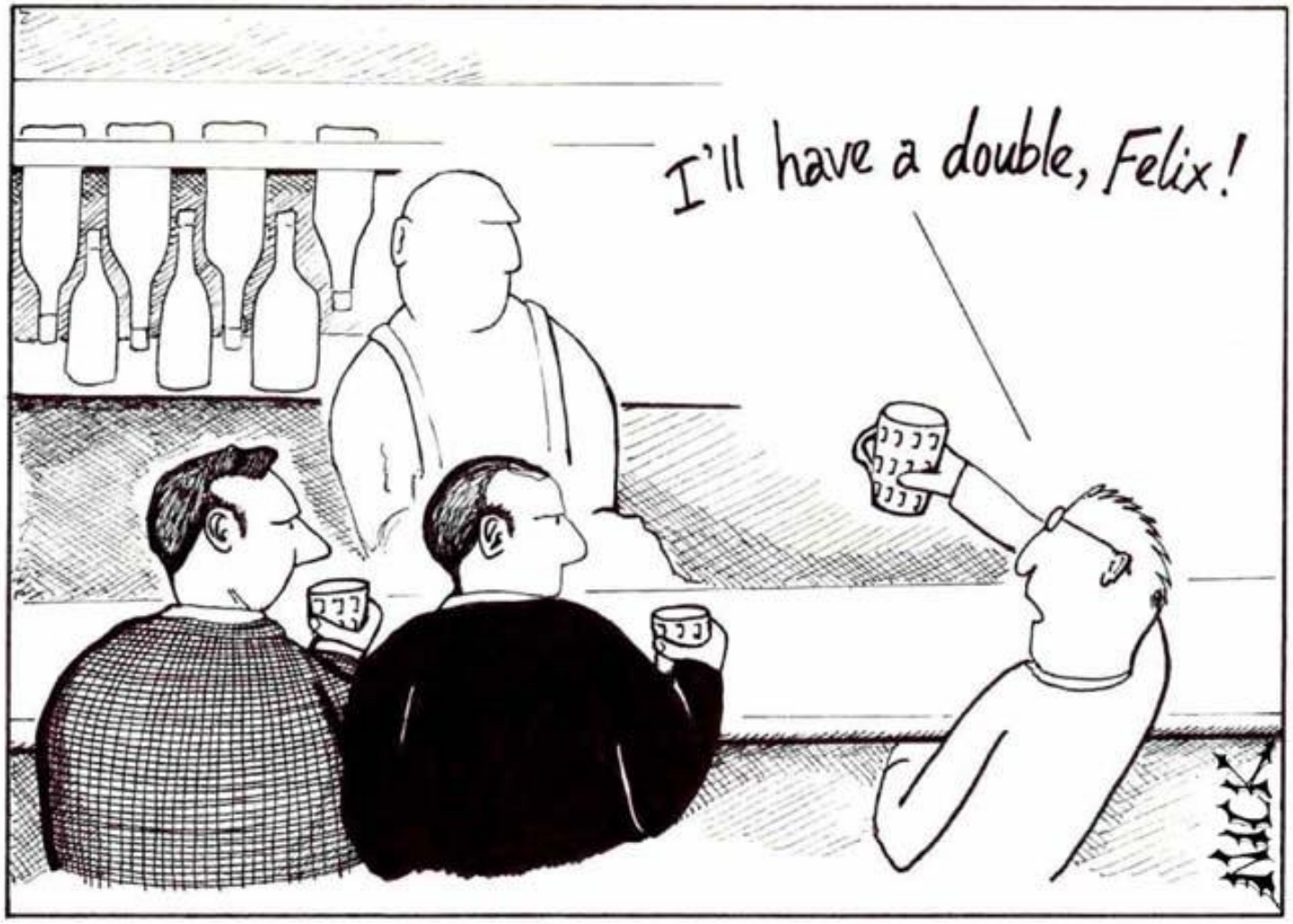
Multiple Choice #14

The sex and the condition of the individual whose karyotype is shown above are given in row

Row	Sex	Condition
A.	Female	Patau syndrome
B.	Female	Down syndrome
C.	Male	Edward syndrome
D.	Male	Normal

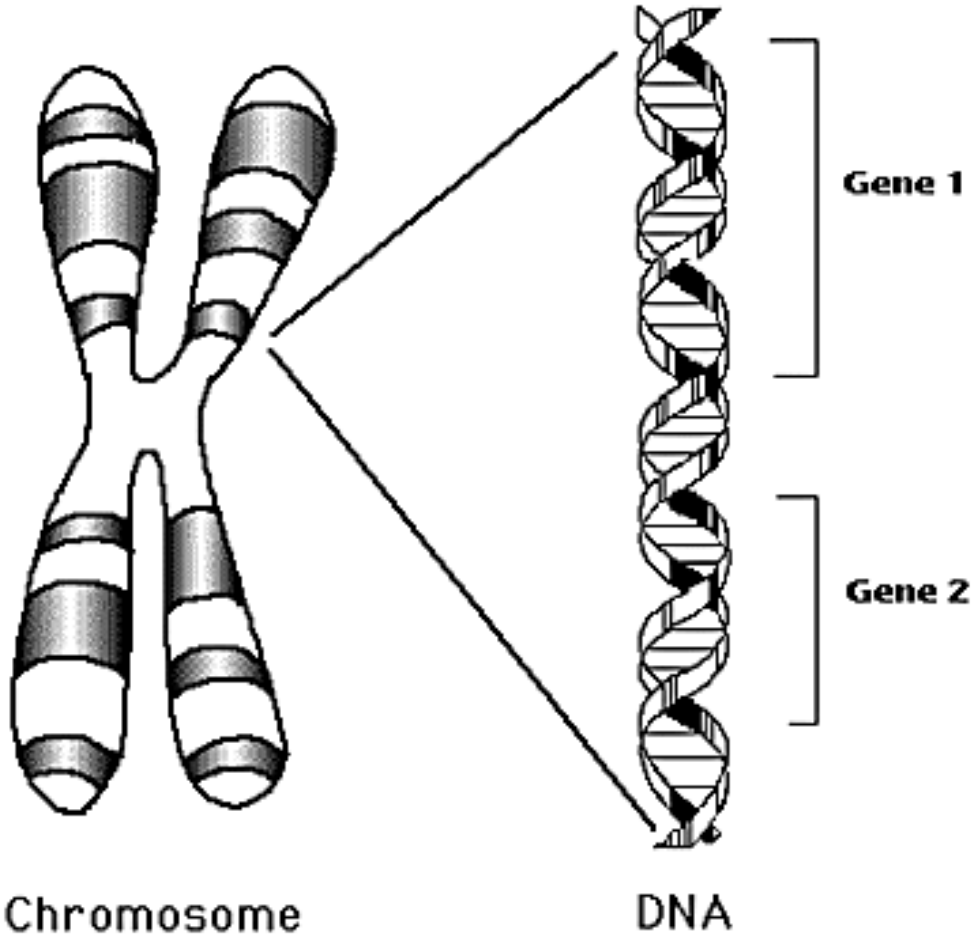
Part II…

DNA and Molecular Genetics



Cambridge, 1953. Shortly before discovering the structure of DNA, Watson and Crick, depressed by their lack of progress, visit the local pub.

DNA Structure

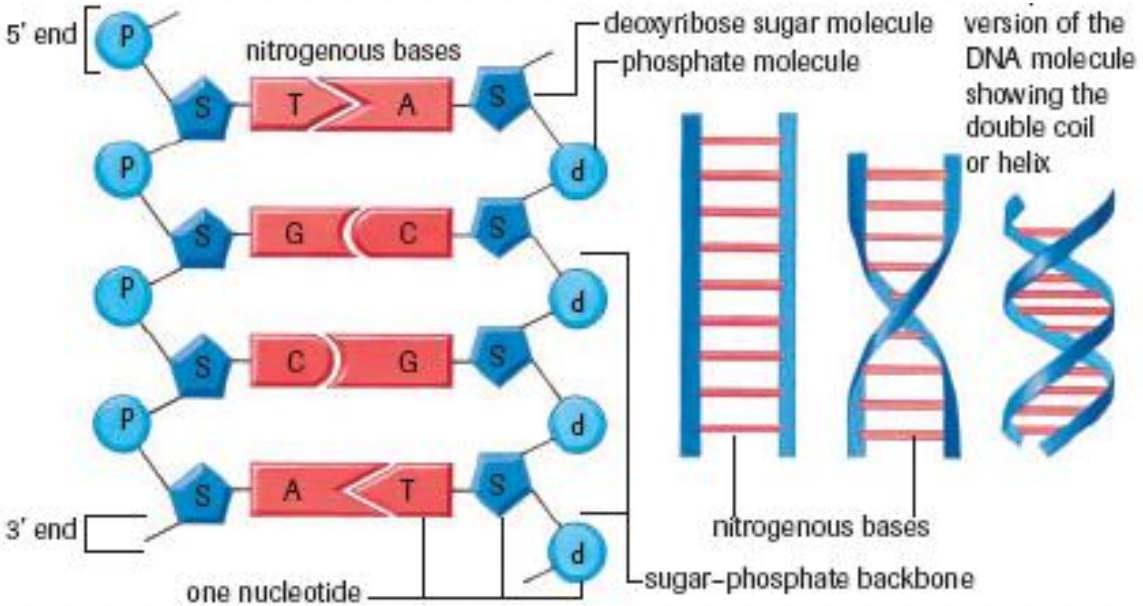


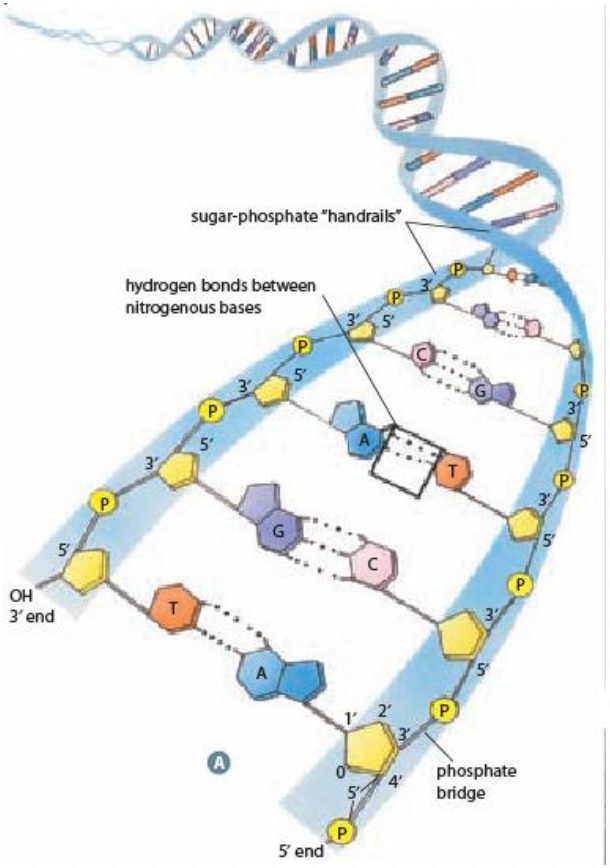
Genes

A Little History...

Year	Scientist	Experimental results
late 1860s	Friedrich Miescher	• isolated nonprotein substance from nucleus of cells; named this substance nuclein
1928	Frederick Griffith	• experimented using mice and two different strains of pneumococcus bacteria (virulent and nonvirulent); observed that when heat-treated virulent pneumococcus was mixed with nonvirulent pneumococcus and was injected into healthy mice, death resulted • discovered the process of transformation
1943	Joachim Hammerling	• experimented using green alga *Acetabularia;* observed that regeneration of new appendages was driven by the nucleus-containing "foot" of the alga • hypothesized that hereditary information is stored in the nucleus
1944	Oswald Avery, Maclyn McCarty, and Colin MacLeod	• demonstrated that DNA was the transforming principle of pneumococcus bacteria
1949	Erwin Chargaff	• discovered that in the DNA of numerous organisms the amount of adenine is equal to the amount of thymine, and the amount of guanine is equal to that of cytosine
1952	Alfred Hershey and Martha Chase	• used radioactively labelled viruses, infected bacterial cells; observed that the infected bacterial cells contained radioactivity originating from DNA of the virus, suggesting that DNA is hereditary material
1953	Rosalind Franklin	• produced an X-ray diffraction pattern of DNA that suggested it was in the shape of a double helix
1953	James Watson and Francis Crick	• deduced the structure of DNA using information from the work of Chargaff, Franklin, and Maurice Wilkins

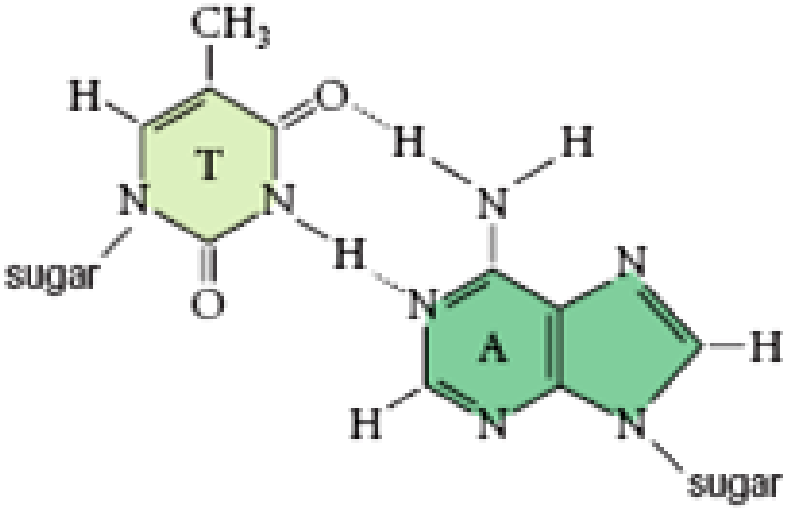
DNA Composition

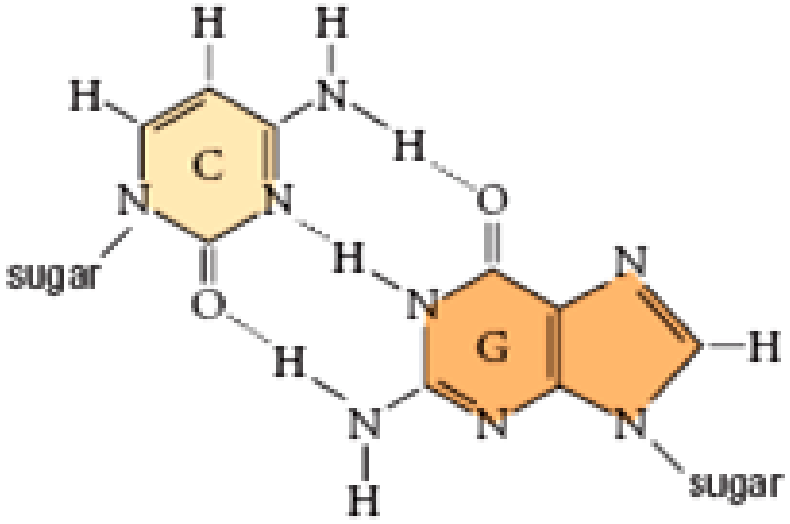




- Nucleotide = sugar, phosphate & nitrogen base
- Adenine, Thymine, Guanine, Cytosine
- Antiparallel strands
- Sequence of bases on one strand determines sequence on other

Base Pairing Rule





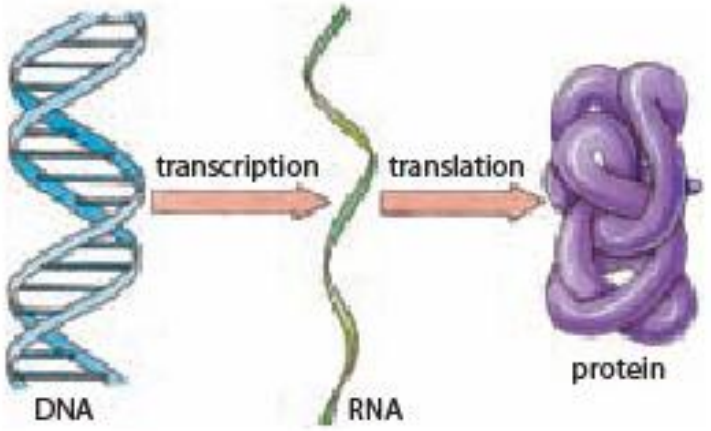
Size Matters…

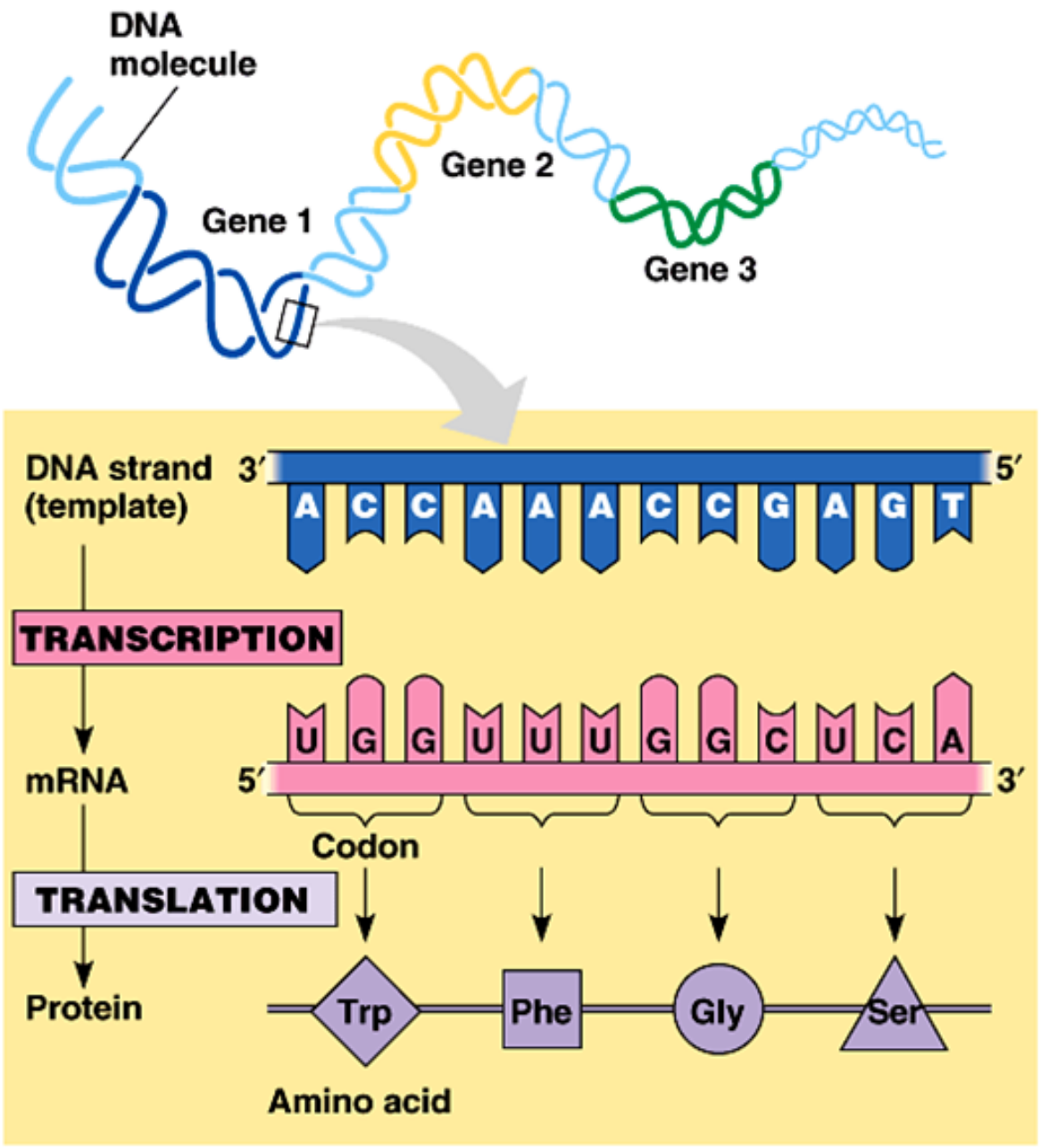
Information About Nitrogen Bases

Nitrogen Base	Classification	Abbreviation
Adenine	Purine	A
Guanine	Purine	G
Cytosine	Pyrimidine	C
Thymine	Pyrimidine	T
Uracil	Pyrimidine	U

Central Dogma of Molecular Genetics…Basics

- **Replication**
  - DNA is copied (DNA → DNA)
- **Transcription**
  - A message is made from the DNA template (the gene is transcribed) (DNA → mRNA)
- **Translation**
  - mRNA is "read" and an amino acid sequence (protein) is made (mRNA → protein)

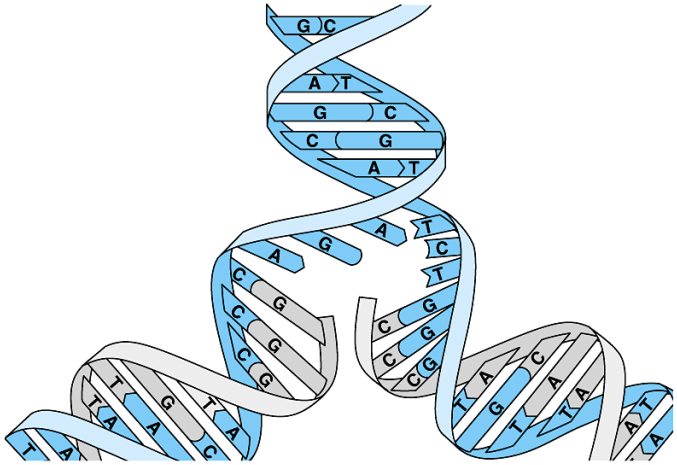


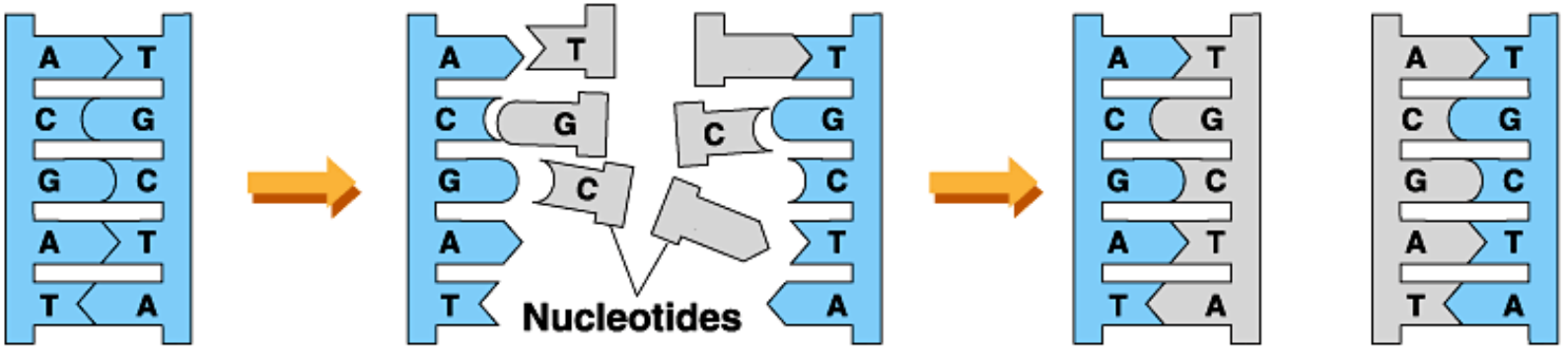


DNA Replication

- Each chromosome = long DNA strand, must be replicated precisely
- During what phase of cell cycle?
- Location?
- Enzymes involved: **Helicase**, **DNA polymerase**
- **Semi-conservative**

DNA Replication is Semi-conservative



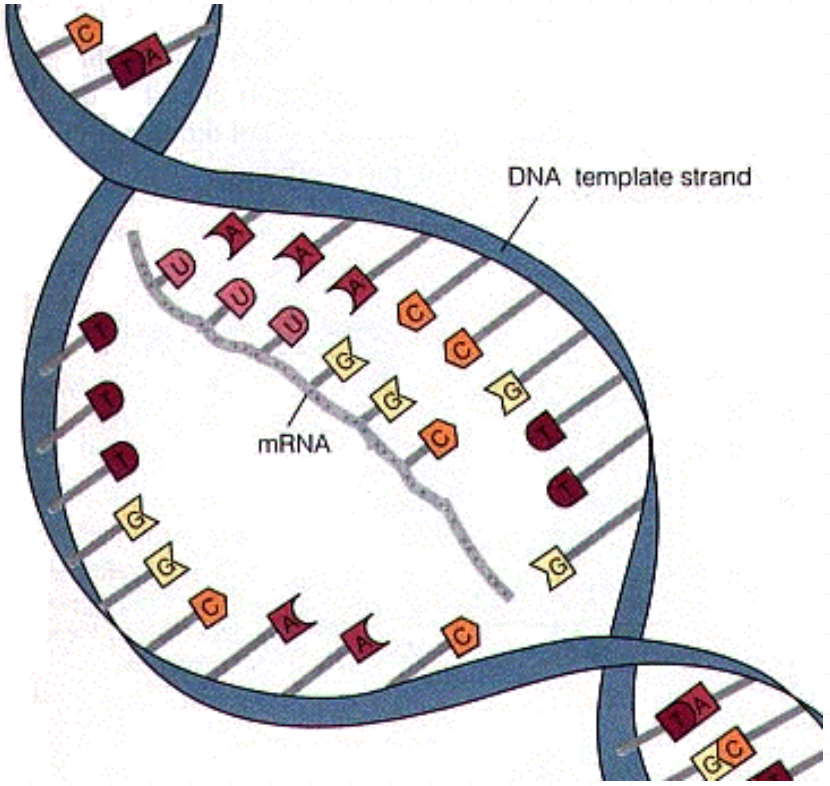


Biotechnologies: PCR

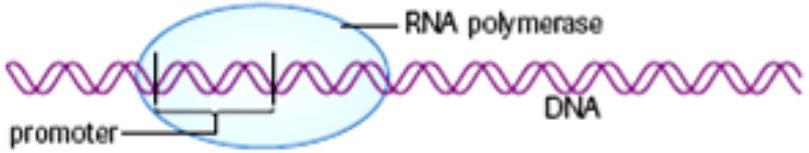
- Polymerase Chain Reaction
- Making millions of copies of DNA sample using laboratory equipment, heat resistant polymerase enzyme and nucleotide supply
- Used for DNA analysis

Transcription

- Nucleus
- One side of DNA
- DNA → mRNA
- RNA – **uracil**, ribose, single stranded
- RNA polymerase



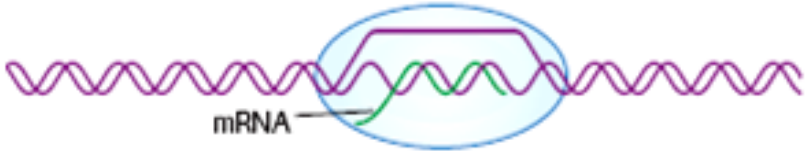
(a) Initiation
RNA polymerase binds to DNA at a promoter.



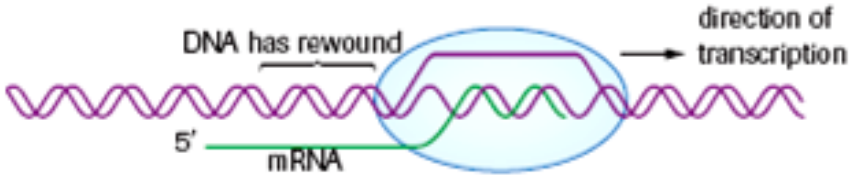
Details...

(b) DNA double helix is unwound, exposing the template strand.

(c) Elongation
mRNA is synthesized using one strand of DNA as a template. mRNA is synthesized in the 5′ to 3′ direction.

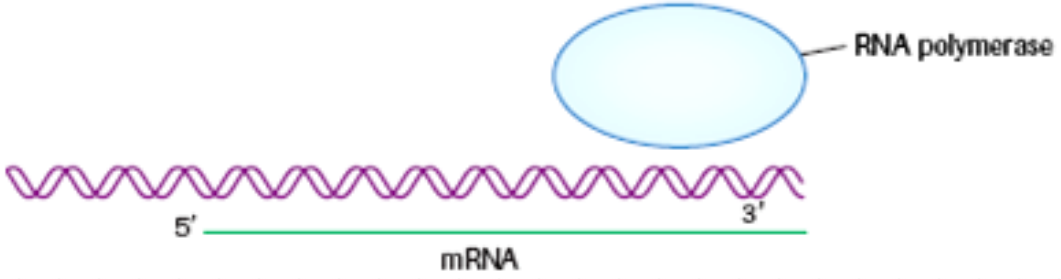


(d) As elongation proceeds, RNA polymerase moves along DNA, synthesizing mRNA. DNA that has already been transcribed rewinds into a double helix.



(e) RNA polymerase reaches the termination sequence at end of gene.

(f) Termination
Transcription stops. mRNA and RNA polymerase are released.



Initiation

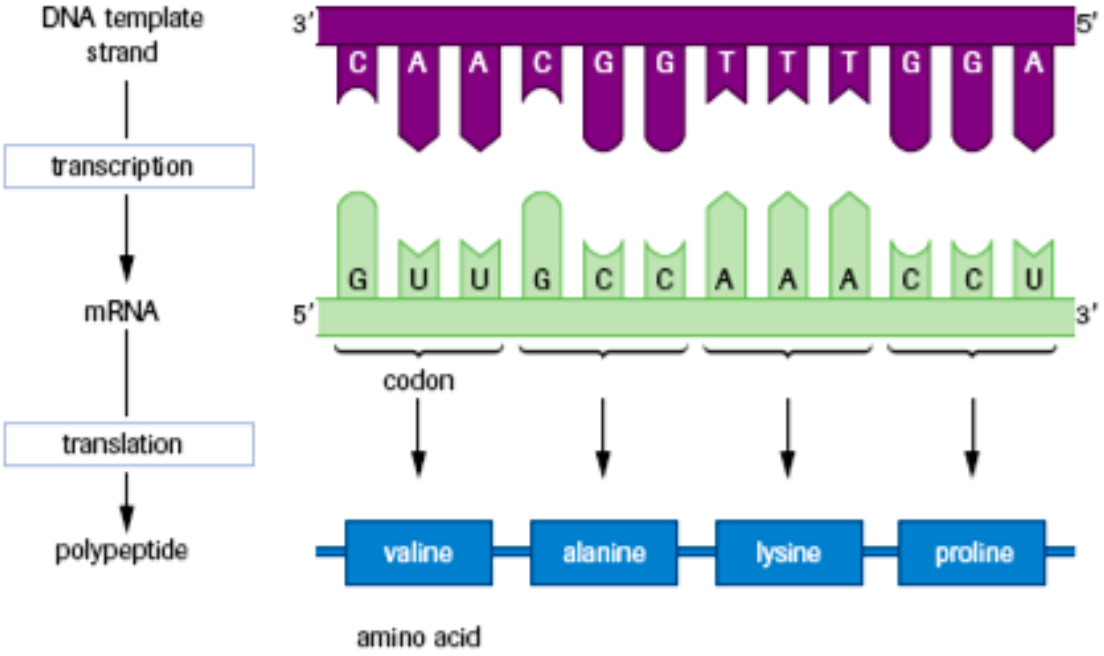
- Initiation of transcription starts when the RNA polymerase binds to the promoter region of the gene to be transcribed.
- The DNA is unwound and the double helix is disrupted.

Elongation

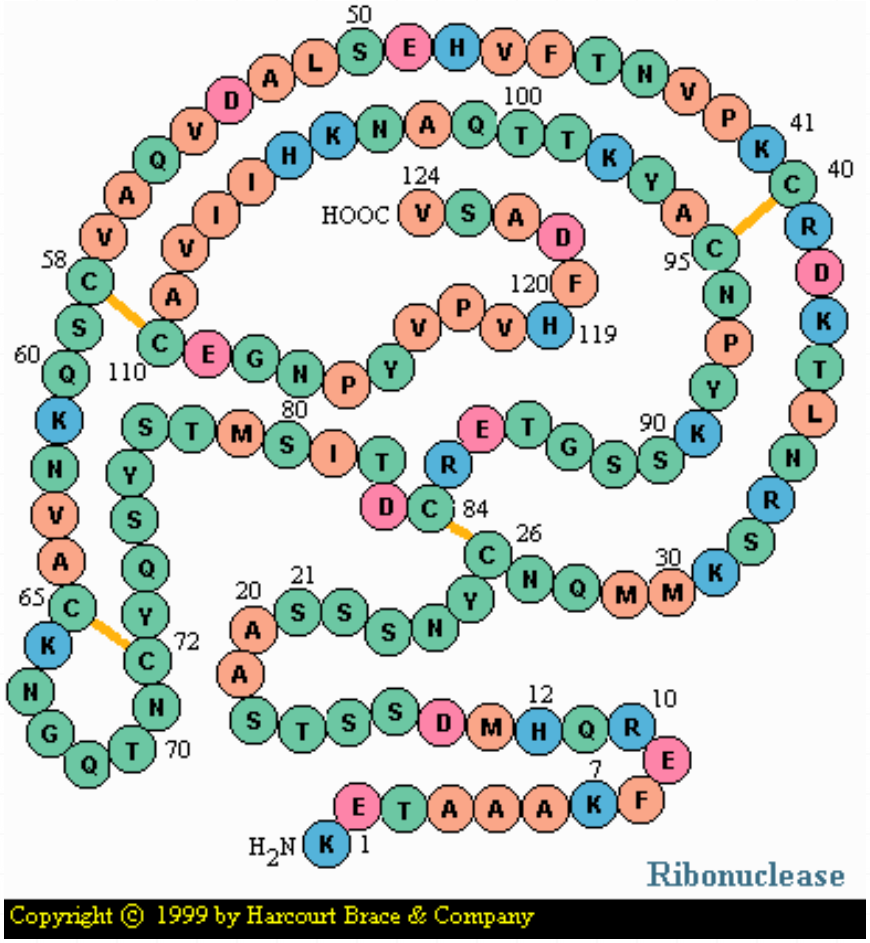
- A complementary messenger RNA (mRNA) molecule is synthesized in the 5′ to 3′ direction, using one strand of DNA as a template.
- Adenine (A) bases in the DNA are paired with uracil (U) in the mRNA.
- Transcription continues until the RNA polymerase reaches a termination sequence.

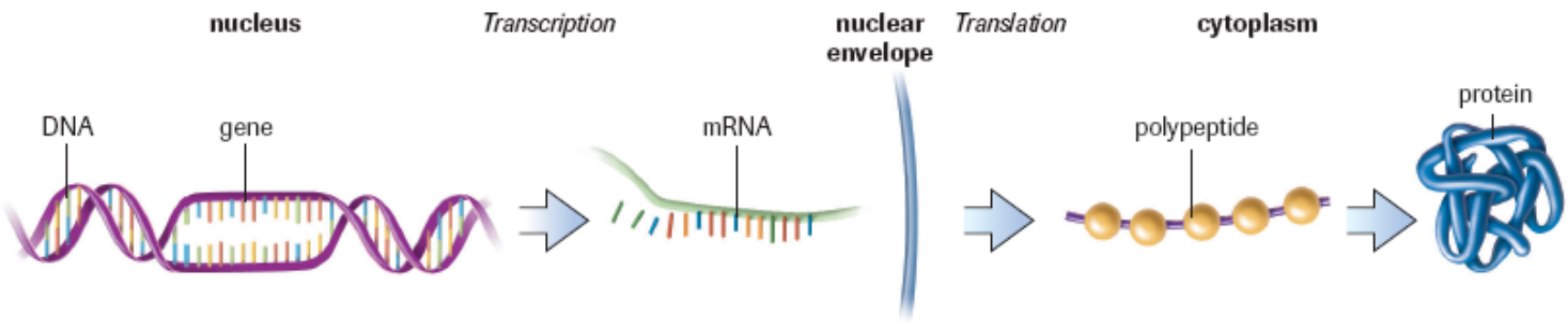
Termination

- When the RNA polymerase comes to a termination sequence, it falls off the DNA molecule.
- The mRNA separates from the DNA.



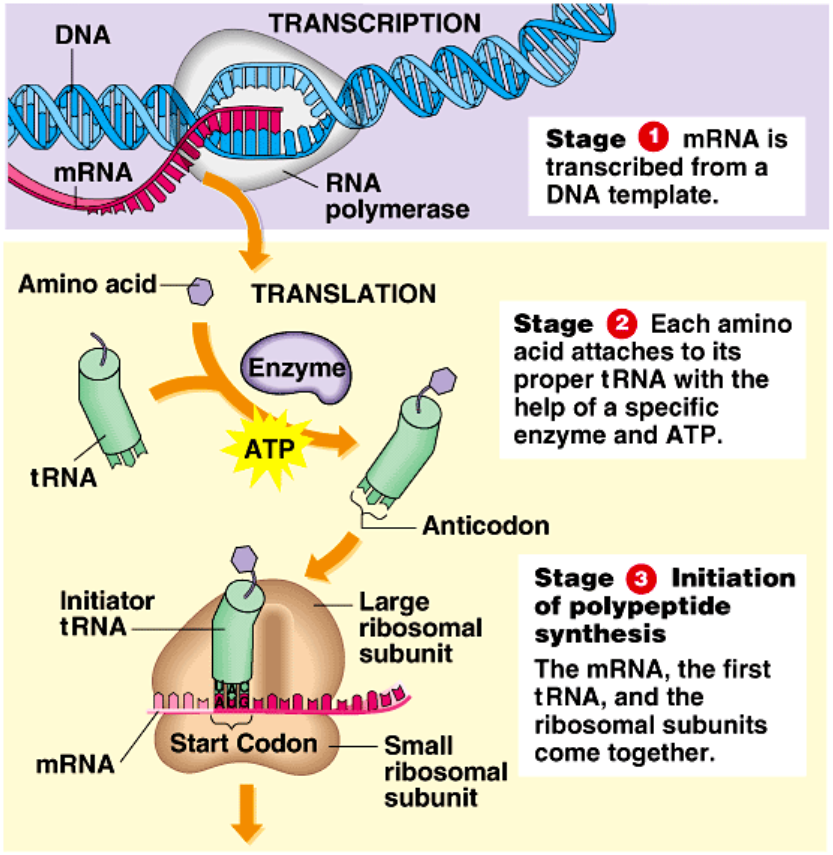
Amino Acid Chain = Polypeptide Chain = Protein

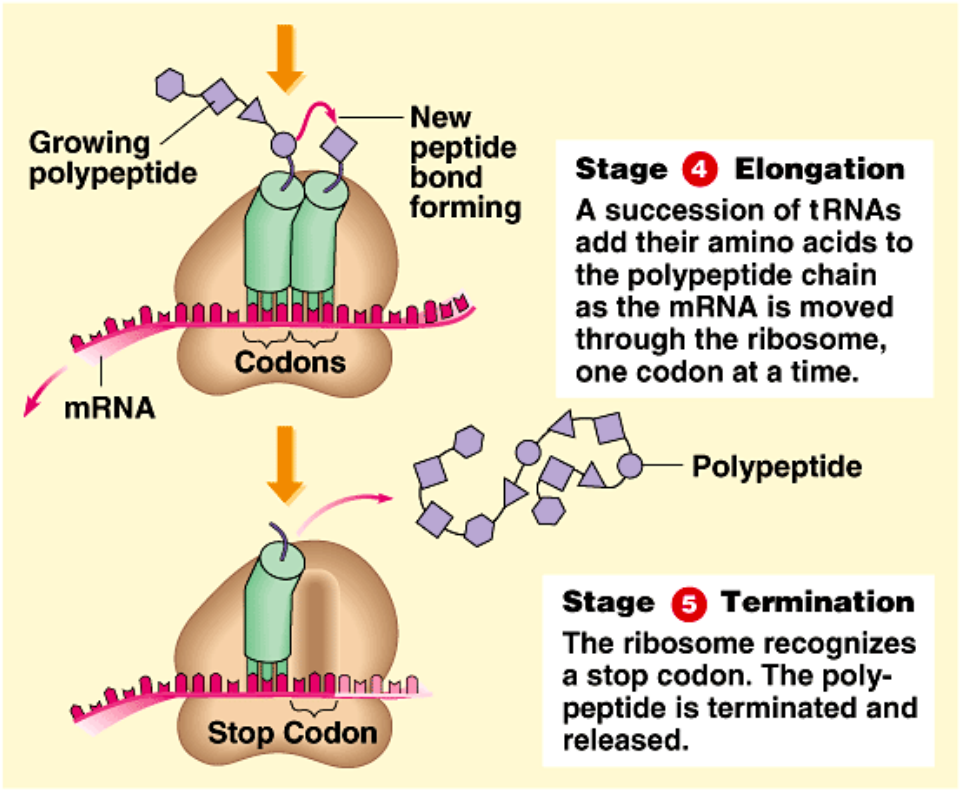




- Proteins functions: hormones, enzymes, antibodies, structures
- Function of a protein as a structural component or as an enzymatic tool is COMPLETELY dependent on its **shape.**
- Shape determined by the **SEQUENCE** of amino acids
- Sequence of amino acids is determined by the **SEQUENCE** of **mRNA CODONS**
- Sequence of CODONS determined by the base sequence in the DNA (also CODONS) in the Gene
- Defective (mutant) or missing gene → altered protein function

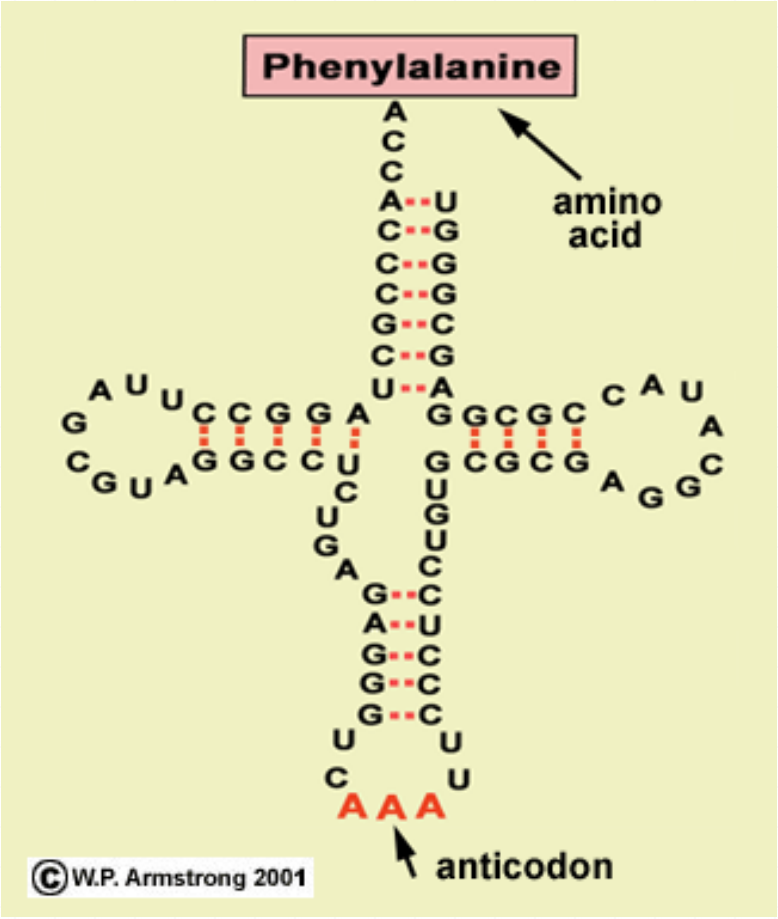
Translation (Cytoplasm) - One gene → one protein





RNA

- Messenger - mRNA
- Ribosomal - rRNA
- Transfer - tRNA



Messenger RNA Codons and Their Corresponding Amino Acids

First Base	Second Base				Third Base
	U	C	A	G	
U	UUU phenylalanine	UCU serine	UAU tyrosine	UGU cysteine	U
	UUC phenylalanine	UCC serine	UAC tyrosine	UGC cysteine	C
	UUA leucine	UCA serine	UAA stop**	UGA stop**	A
	UUG leucine	UCG serine	UAG stop**	UGG tryptophan	G
C	CUU leucine	CCU proline	CAU histidine	CGU arginine	U
	CUC leucine	CCC proline	CAC histidine	CGC arginine	C
	CUA leucine	CCA proline	CAA glutamine	CGA arginine	A
	CUG leucine	CCG proline	CAG glutamine	CGG arginine	G
A	AUU isoleucine	ACU threonine	AAU asparagine	AGU serine	U
	AUC isoleucine	ACC threonine	AAC asparagine	AGC serine	C
	AUA isoleucine	ACA threonine	AAA lysine	AGA arginine	A
	AUG methionine*	ACG threonine	AAG lysine	AGG arginine	G
G	GUU valine	GCU alanine	GAU aspartate	GGU glycine	U
	GUC valine	GCC alanine	GAC aspartate	GGC glycine	C
	GUA valine	GCA alanine	GAA glutamate	GGA glycine	A
	GUG valine	GCG alanine	GAG glutamate	GGG glycine	G

* Note: AUG is an initiator codon and also codes for the amino acid methionine.

** Note: UAA, UAG, and UGA are terminator codons.

Process of Translation

- As ribosome moves along mRNA, **tRNAs** sequentially bring in the appropriate amino acids which are then bonded together in a polypeptide chain = protein
- The tRNAs are then free to pick up another amino acid in cytoplasm
- When ribosome reads a stop codon on the mRNA, the amino acid chain is released as a protein and the ribosome releases the mRNA

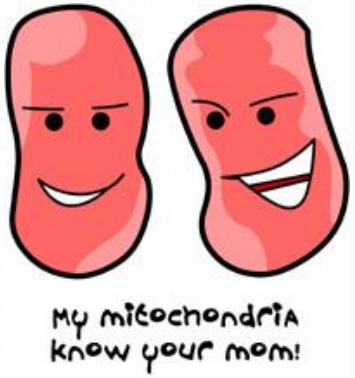
For the details…

Table 4 Summary of Translation

Initiation
• Ribosome subunits (large and small) bind to the mRNA transcript, sandwiching the mRNA between them.
• The ribosome moves along the mRNA, reading the codons.
• Translation begins when the ribosome reaches the start codon, AUG.
Elongation
• Through the genetic code, each codon specifies a particular one of the 20 amino acids that make up polypeptides.
• Transfer RNA (tRNA) molecules have an anticodon that is complementary to the codon in the mRNA. The tRNA carries the amino acid specified by the codon.
• The ribosome contains two sites, the A (aminoacyl) site and the P (peptidyl) site.
• When the start codon is in the P site, the first tRNA delivers methionine. Since the start codon codes for methionine, all polypeptides initially start with this amino acid.
• The second codon of the mRNA is exposed at the same time in the A site. When the tRNA delivers the second amino acid, a peptide bond is formed between the two amino acids.
• The ribosome shifts over one codon. The tRNA that delivered the methionine is released from the P site.
• When the ribosome shifts, the tRNA containing the growing polypeptide moves to the P site. A third amino acid, specified by the third codon, is brought in to the A site by the next tRNA. A peptide bond is formed between the second and third amino acid.
• Amino acids continue to be added to the polypeptide until a stop codon is read in the A site.
Termination
• The stop codons are UAG, UGA, and UAA. At this point the ribosome stalls.
• A protein known as the release factor recognizes that the ribosome has stalled and causes the ribosome subunits to disassemble, releasing the mRNA and newly formed polypeptide.

Mitochondrial DNA

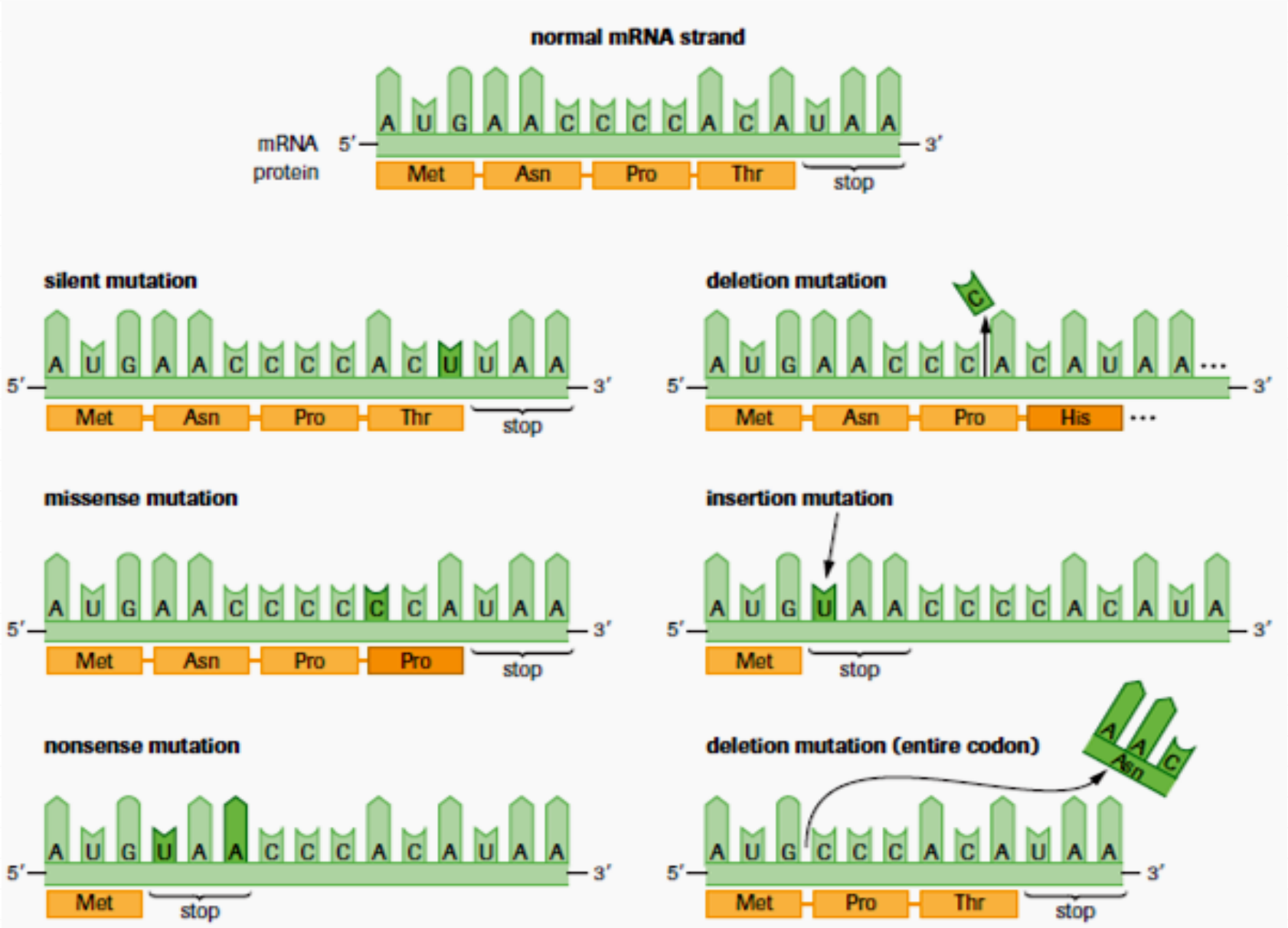
- **mtDNA**
  - Small number of bases (70 000 vs. 6 billion in genome)
  - Mitochondria from **mother** exclusively, except ICSI where sperm is injected
  - Trace lineage **maternally**



Mutations…

- Gene mutation - DNA sequence altered
- Chromosome – missing portions or entire chromosome lead to huge alterations
- Mutagens – spontaneous error, radiation, chemicals
- Substitution, Deletion, Insertion…

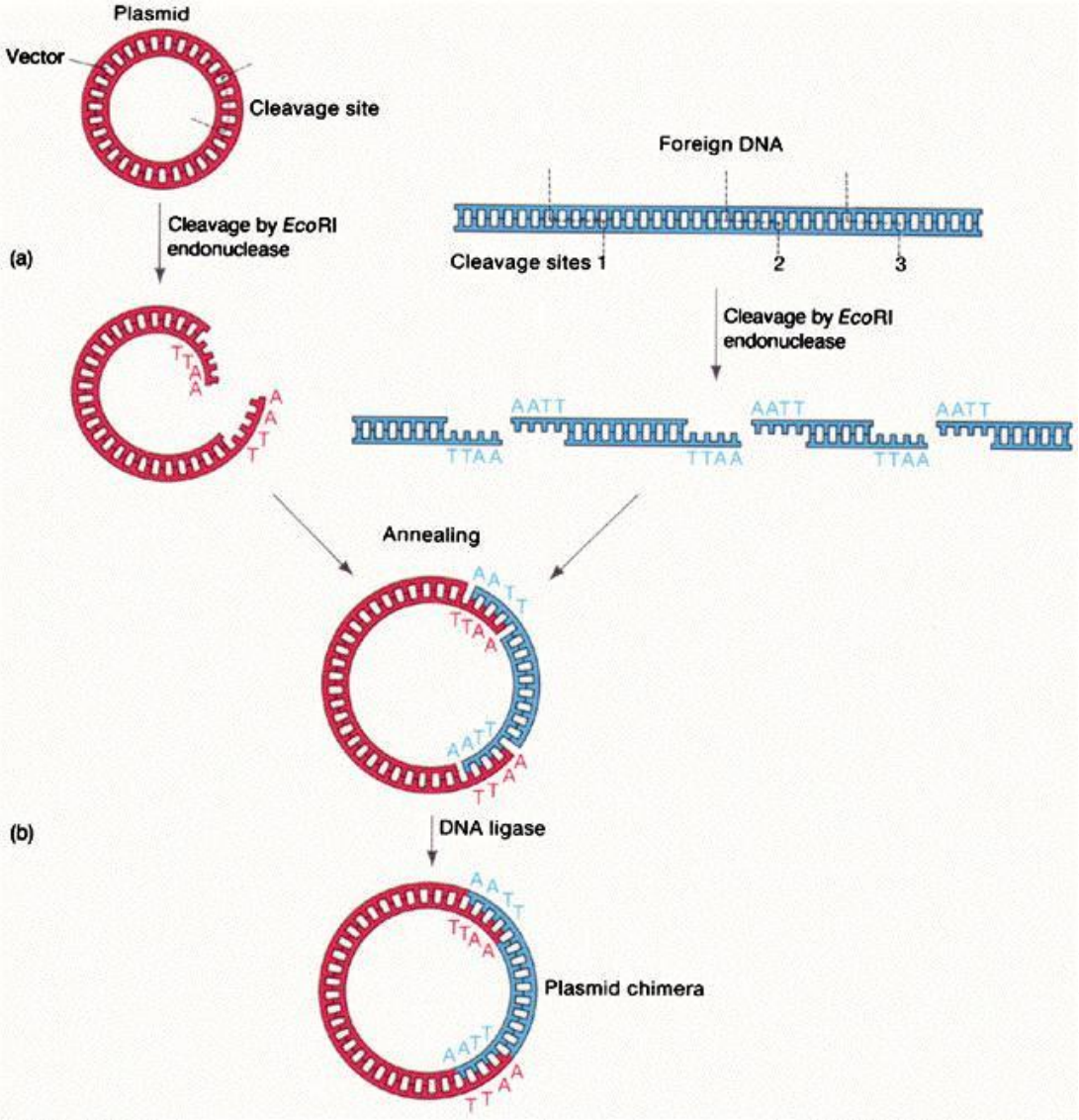
Types of Mutations



Genetic Engineering

- DNA can be artificially recombined in the lab to have specific gene combinations and characteristics
- Requires
  - **Restriction Enzymes** – cut DNA (scissors)
  - **DNA Ligase** – glues DNA backbone back together
- Can produce transgenic organisms that contain foreign DNA. Why?...
- Bacteria can make human proteins (insulin)
- Modify plants to withstand disease, harsh conditions
- Animals with increased growth or serve as organ donors

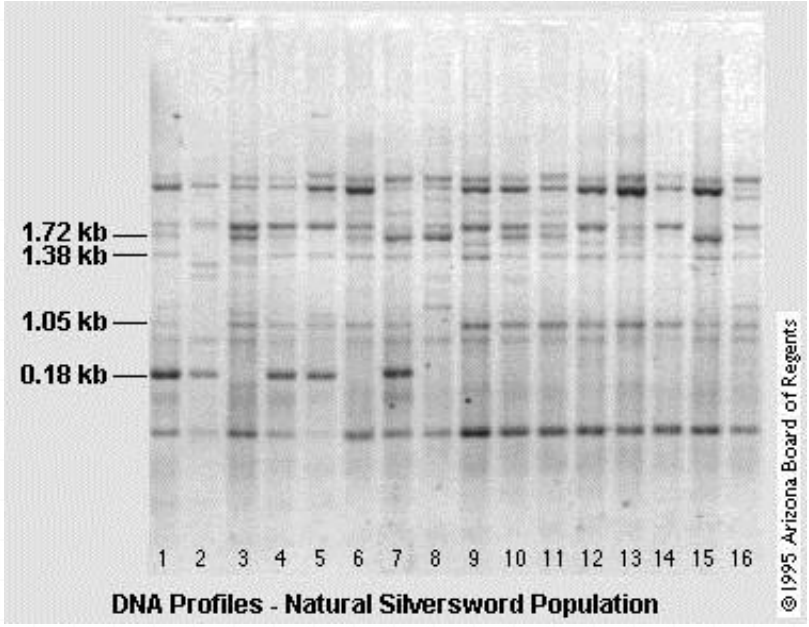
Biotechnology-Recombinant DNA



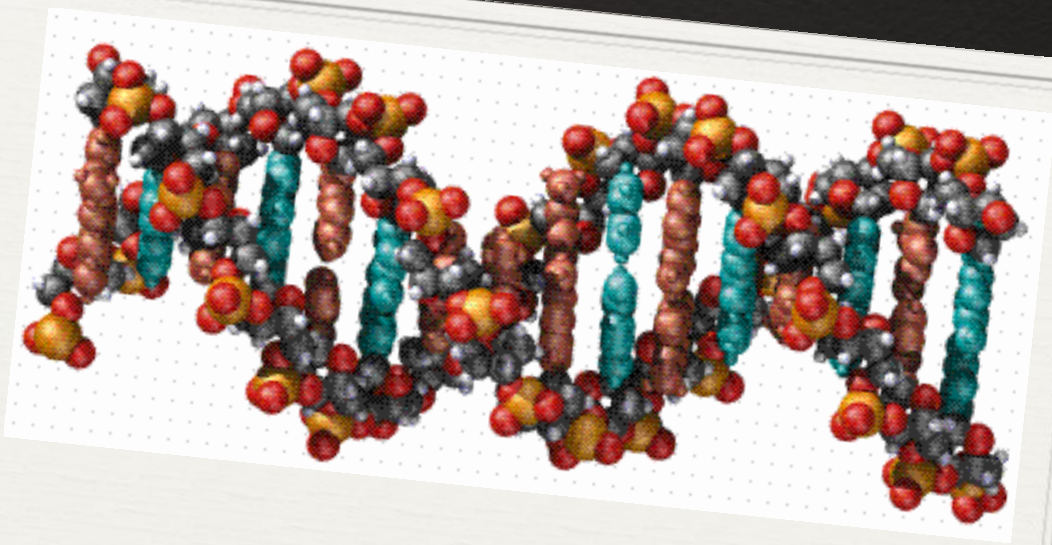
- Cut donor gene with **restriction enzyme**
- Cut bacterial plasmid with same enzyme
- Use **ligase** to join new DNA into place
- Insert into bacterium
- All new bacteria contain foreign DNA and are transgenic

Gel Electrophoresis

- Separation of DNA fragments based on size
- Restriction enzymes cut DNA
- Apply to gel, electric current
- Smaller fragments move farther/faster
- Paternity testing, gene presence, fingerprint court evidence



Let's see what you can do...



DNA practice questions

Multiple Choice #22

Use the following information to answer the next question.

> The San Diego Zoo is preserving DNA from hundreds of species by freezing cell samples in its Centre for the Reproduction of Endangered Species. The cell specimens are often from individuals in wild populations that are chosen for distinctive characteristics.
>
> —from *Vedantam*

To obtain all the representative DNA of an organism, it would be necessary to collect **only**

A. an egg
B. a sperm
C. a body cell
D. a cell from each type of body tissue

Multiple Choice #23

As cells age, there is an increase in DNA damage and a decrease in DNA repair processes. The **initial** effect is

A. a decrease in ATP synthesis
B. an increase of cancerous cells
C. the production of altered proteins
D. the production of abnormal mRNA

Multiple Choice #24

Use the following information to answer the next question.

Portion of Insulin Protein

Phenylalanine–Valine–Asparagine–Glutamine–Histidine

What is the strand of DNA that would code for this portion of insulin?

A. AAG CAA TTA GTT GTA
B. AAA CAA TTC CAC CTA
C. CAC GAG AAC GTA TTC
D. TTC GTA AAC GAG CAC

Multiple Choice #25

DNA is structurally different from RNA in that DNA

A. contains uracil and is composed of double strands
B. contains adenine and is composed of single strands
C. contains guanine and is composed of single strands
D. contains thymine and is composed of double strands

Multiple Choice #26

Analysis of a DNA sample showed that 15% of the nitrogen-base molecules present were adenine molecules. This sample would likely contain

A. 15% thymine
B. 15% uracil
C. 85% thymine
D. 85% uracil

Multiple Choice #27

Use the following information to answer the next two questions.

In DNA replication, the two strands of the double helix separate and a new strand forms along each old one. Each new DNA molecule has one old and one new strand.

The name of the old DNA strand and the site of DNA replication are identified in row

Row	Name of Old Strand	Site
A.	a template	nucleus
B.	a template	cytoplasm
C.	haploid	nucleus
D.	haploid	cytoplasm

Multiple Choice #28

The backbone of a DNA molecule and the composition of A, C, T, and G are identified in row

Row	DNA Backbone	A, C, T, and G
A.	phosphate groups	deoxyribose sugars
B.	purines	deoxyribose sugars
C.	pyrimidines	nitrogen-containing bases
D.	sugars and phosphate groups	nitrogen-containing bases

Multiple Choice #35

Use the following information to answer the next two questions.

A deletion mutation in mitochondrial DNA causes Kearns–Sayre syndrome (KSS). A large sample of different types of somatic cells was removed from a male with KSS, tested, and found to contain the deletion. The only type of mitochondrial DNA that was found in somatic cells from the man's mother was mitochondrial DNA that did not have the KSS deletion.

A reasonable hypothesis to explain these results is that the mutation in the mitochondrial DNA that caused KSS in the man first occurred in the

A. mother's oocytes
B. man's somatic cells
C. man's spermatocytes
D. mother's somatic cells

Multiple Choice #36

Both males and females can be affected by mitochondrial mutations, but only females can transmit genetic mutations to their offspring. For this inheritance pattern, which of the following rows identifies the contributions to the zygote made by the sperm and by the egg?

Row	Sperm Contribution	Egg Contribution
A.	Nuclear contents only	Both nuclear and cytoplasmic contents
B.	Both nuclear and cytoplasmic contents	Nuclear contents only
C.	Neither nuclear nor cytoplasmic contents	Both nuclear and cytoplasmic contents
D.	Both nuclear and cytoplasmic contents	Neither nuclear nor cytoplasmic contents

Multiple Choice #37

Use the following information to answer the next question.

Over time, mitochondrial DNA accumulates non-lethal mutations at a constant rate. There is a higher degree of variation in mitochondrial DNA in earlier populations than in more recent populations. Scientists have taken samples of mitochondrial DNA from people living on different continents and compared the number of mitochondrial DNA mutations in these samples. They used this data as evidence to determine the order in which Earth's continents were populated.

In this study, the manipulated variable was the

A. amount of mitochondrial DNA tested
B. time of migration from one continent to another
C. amount of variation in mitochondrial DNA base sequences
D. geographic location of subjects whose sample of mitochondrial DNA was tested

Multiple Choice #42

Use the following information to answer the next question.

In the late 1980s, some molecular biologists found evidence in human genes suggesting that all people share a common female ancestor who lived in Africa about 200 000 years ago. These biologists studied DNA transmitted only through mitochondria, and they used a "molecular clock" that assumes mutations occur at a known, constant rate. Another researcher used DNA possessed and transmitted only by males to trace humans to a common male ancestor who lived 188 000 years ago.

Which part of the human cell would the researcher have studied in order to trace the ancestry of DNA unique to males?

A. The X chromosome
B. The Y chromosome
C. RNA in the ribosomes
D. DNA in the mitochondrion

Multiple Choice #43

Use the following additional information to answer the next question.

In an individual with KSS, part of the coding strand of mitochondrial DNA that has been deleted has the following base sequence.

ACC TCC CTC ACC AAA

The third amino acid coded for by this segment of mitochondrial DNA is

A. lysine
B. threonine
C. glutamate
D. phenylalanine

Multiple Choice #44

Which enzymes would a geneticist use to cut DNA into fragments?

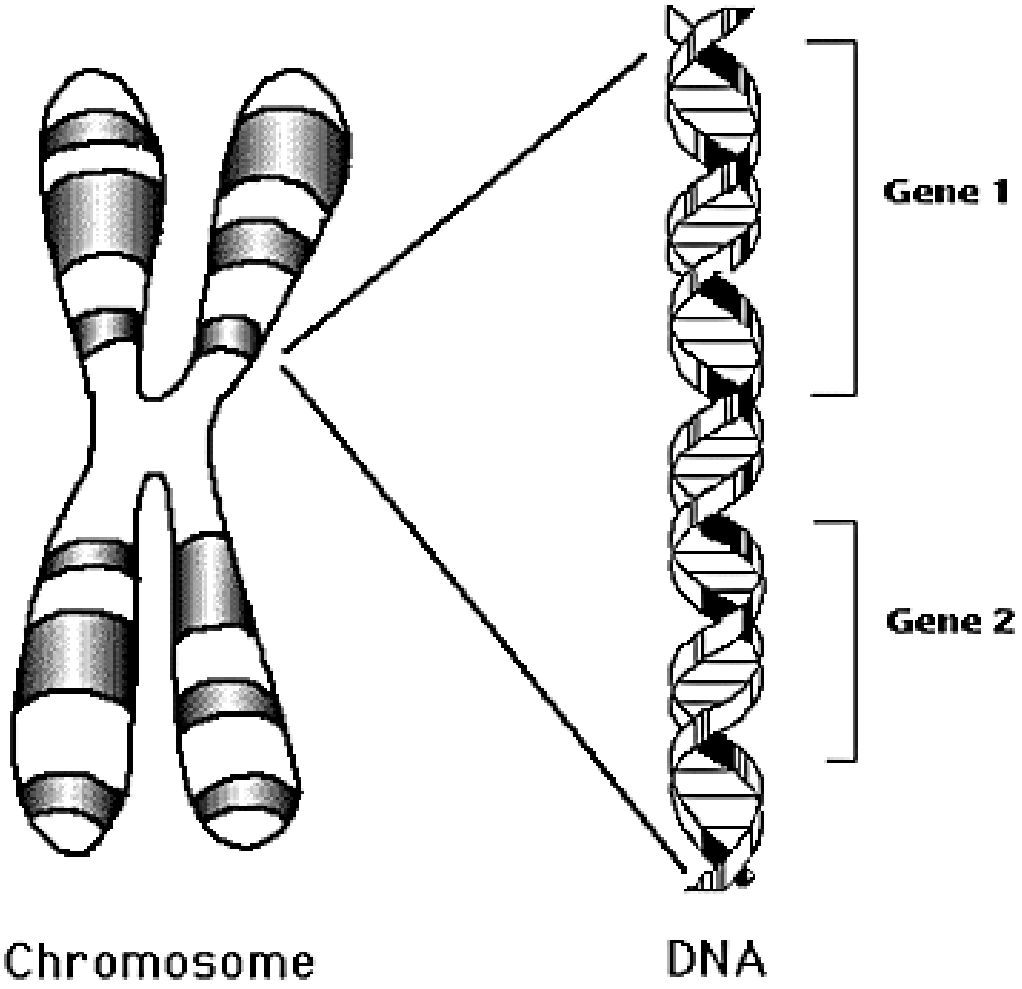
A. Ligase enzymes
B. RNA polymerase
C. DNA polymerase
D. Restriction enzymes

Part III.

CLASSICAL GENETICS


So you see, the wizard gene is "recessive", while the muggle gene is "dominant"...
M+M=M
W+W=W
W+M=M
Do we tell him it's just a story?...

Genes - Units of Heredity



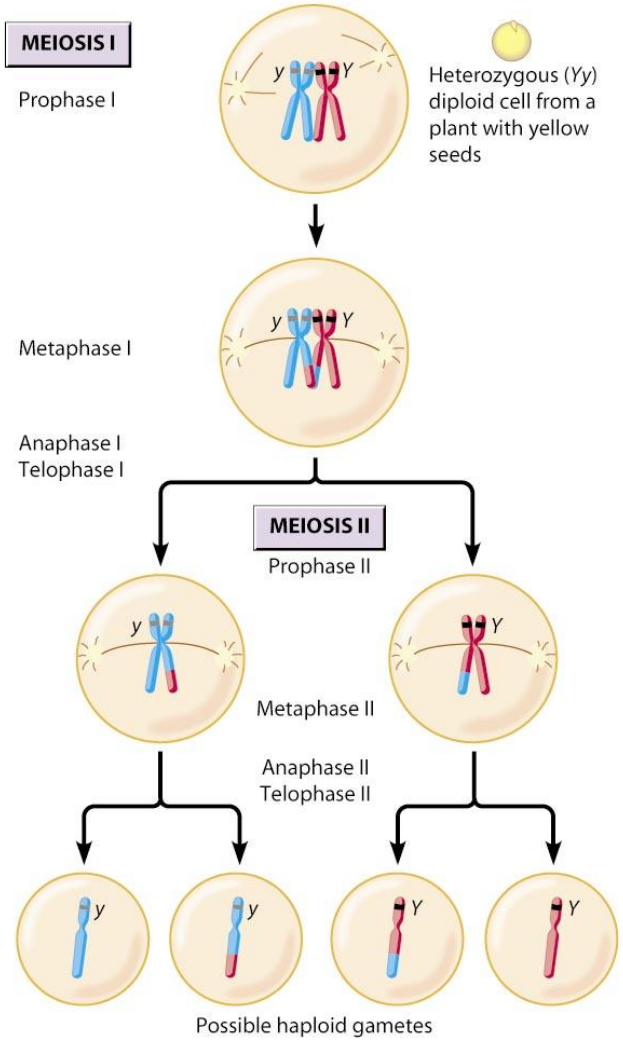
Genes

Heredity

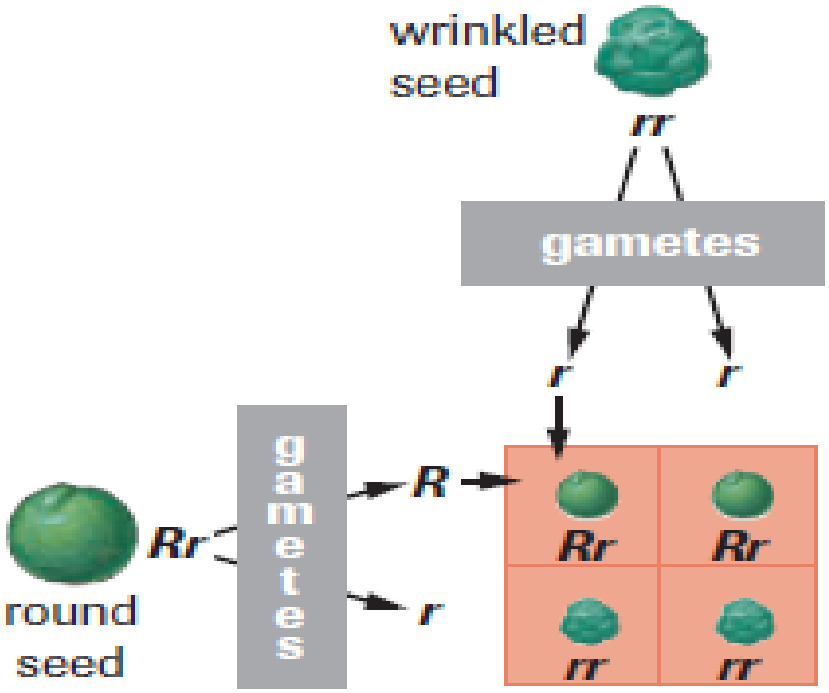
- **Genes** – determine a protein and hence a trait
- **Polygenic** – 2 or more genes determine some traits e.g.: height, intelligence
- **Genome** – all the genes of an organism in DNA
- **Alleles** – Forms of a gene
- Law of Dominance
  - Dominant
  - Recessive
  - Homozygous (Pure)
  - Heterozygous (Hybrid, Carrier)

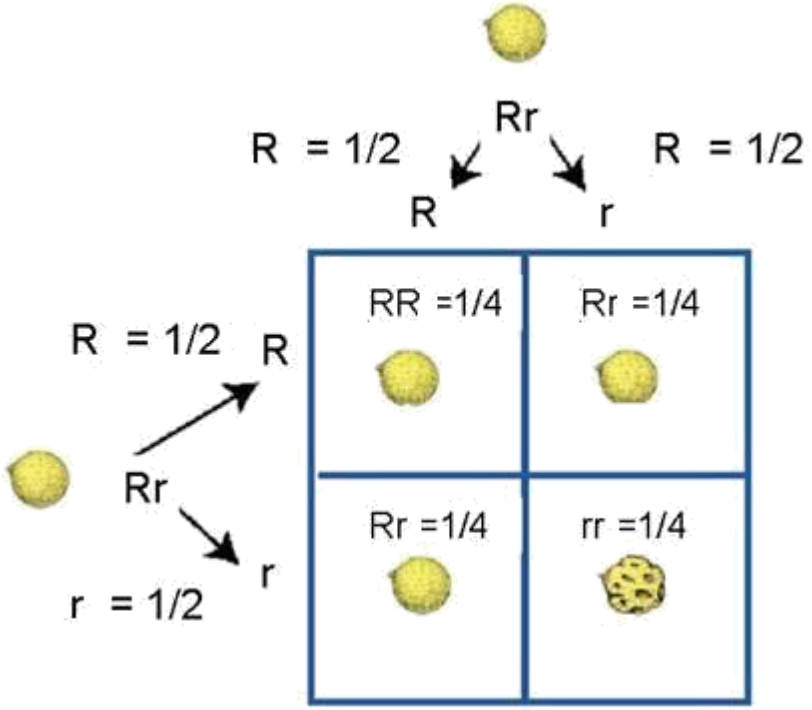
Law of Segregation

- pairs of alleles separate during meiosis



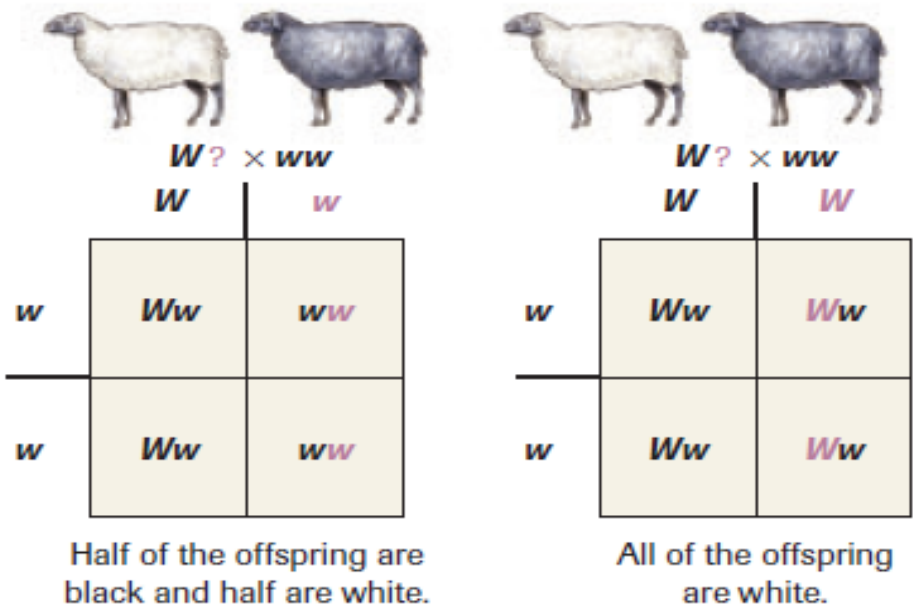
Phenotype and Genotype





Testcross

- To determine the genotype of a dominant phenotype
- Unknown phenotype mated with **HOMOZYGOUS RECESSIVE**
- If all offspring are dominant the parent is pure = homozygous dominant
- If any offspring are recessive the parent is heterozygous



Multiple Choice #45

Mendel's principle of segregation states that alternate forms of a gene separate during

A. fertilization
B. seed dispersal
C. cross-pollination
D. gamete formation

Multiple Choice #46

An organism is heterozygous for two pairs of genes. The number of different combinations of alleles that can form for these two genes in the organism's gametes is

A. 1
B. 2
C. 4
D. 8

Multiple Choice #47

Use the following information to answer the next two questions.

An inheritable mutation in the DNA that codes for an enzyme that breaks down a neurotransmitter in the brain has been found to be associated with abnormally aggressive behaviour. Females who are heterozygous for the mutation associated with abnormally aggressive behaviour do not have an aggressive phenotype.

The reason that heterozygous females do **not** express the mutation is that

A. nondisjunction occurs
B. gene dominance occurs
C. gene segregation occurs
D. independent assortment occurs

Multiple Choice #48

Use the following additional information to answer the next question.

The mutation associated with abnormally aggressive behaviour results in the normal codon for glutamine becoming a stop codon.

Which of the following mutations would change a normal codon for glutamine into a stop codon?

A. CAA to ATT
B. GAA to UAA
C. GAG to UAG
D. CAG to UAG

Multiple Choice #49

Use the following information to answer the next two questions.

Tay-Sachs disease is a hereditary disease that kills 1 in 360 000 individuals in the general population. Children who are homozygous for Tay-Sachs disease die at an early age. Genetic screening can be done to determine if an individual is a carrier of the Tay-Sachs allele.

What type of inheritance is demonstrated in Tay-Sachs disease?

A. Autosomal recessive
B. Autosomal dominant
C. Sex-linked recessive
D. Sex-linked dominant

Multiple Choice #50

Use the following additional information to answer the next question.

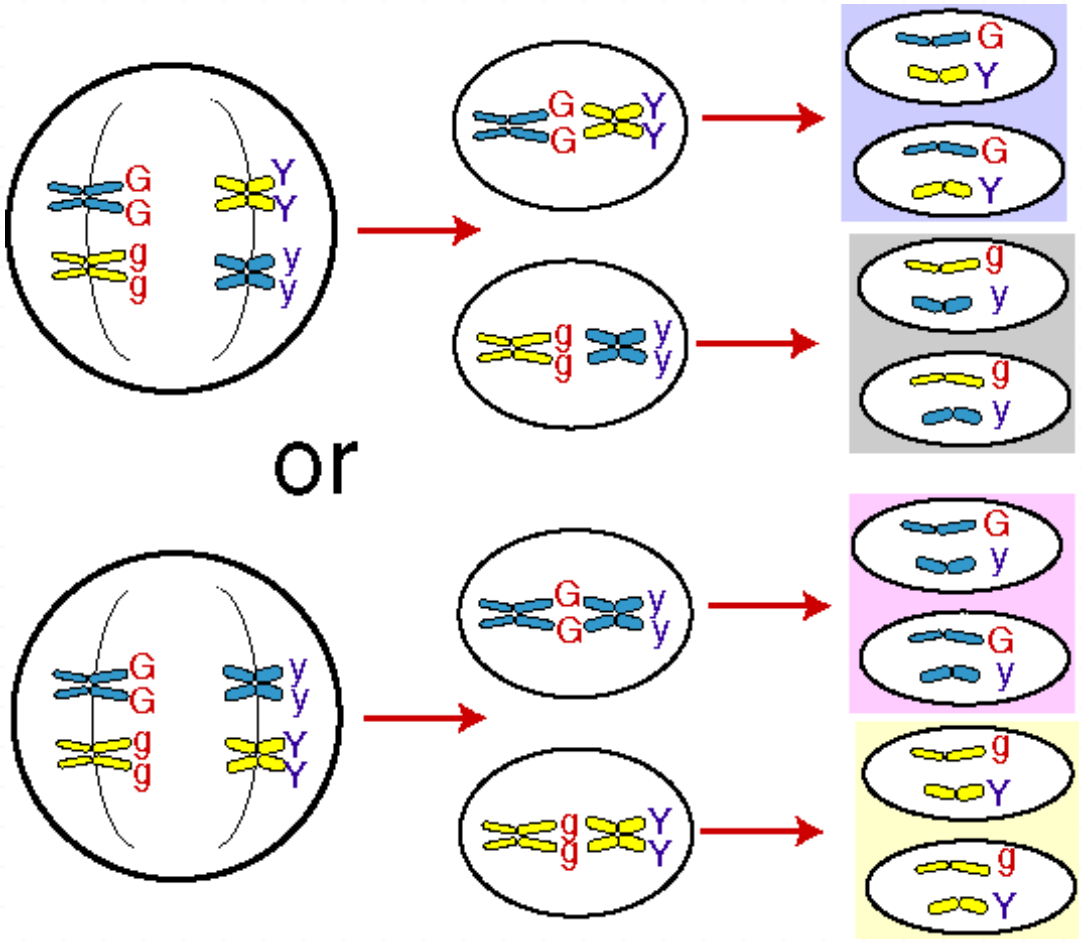
Genetic screening can involve producing complementary DNA probes of a gene's alleles and determining if these bind to an individual's DNA sample.

Genetic screening results show that an individual is a carrier of Tay-Sachs disease if the individual's DNA binds to

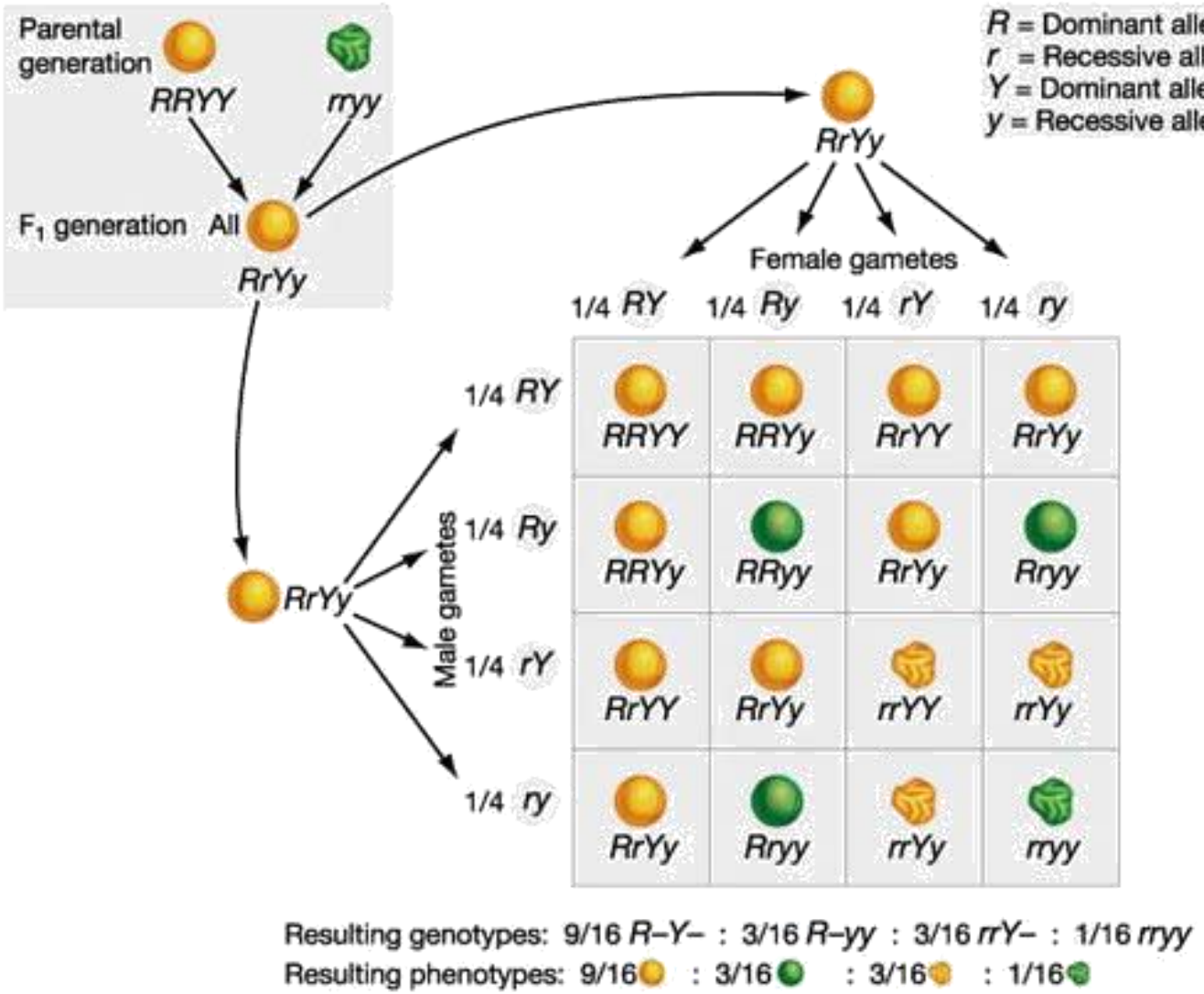
A. none of the DNA probes
B. two of the normal allele DNA probes
C. two of the defective allele DNA probes
D. one of the normal allele DNA probes and one of the defective allele DNA probes

Law of Independent Assortment

- Genes on separate chromosomes assort independently (randomly) during gamete formation



Dihybrid Cross



Resulting genotypes: 9/16 *R–Y–* : 3/16 *R–yy* : 3/16 *rrY–* : 1/16 *rryy*

Resulting phenotypes: 9/16 : 3/16 : 3/16 : 1/16

Predict Gametes First!!!

Steps to solving a dihybrid Question:

1) Determine the mode of inheritance (autosomal or sex-linked) for each gene
2) Write out the gamete combinations for BOTH parents
   - Examples: BbCc → BC, Bc, bC, or bc
     Bbcc → Bc or bc
3) Create a Punnet Square that allows you to predict outcomes
   - Example: If we cross the two individuals above, then:

	BC	Bc	bC	bc
Bc				
bc				

   - Determine the probability that the offspring would be homozygous recessive for both traits.

Alternatively…

- You may also separate the genes and treat them as two monohybrid crosses, then use the probability rules (ex. Product rule) to determine probabilities in the offspring.
- Example using previous parents (BbCc X Bbcc). The probability of having an offspring that is homozygous recessive for both traits is?

	B	b
B		
b		bb

1/4

	C	C
C	Cc	Cc
c	cc	cc

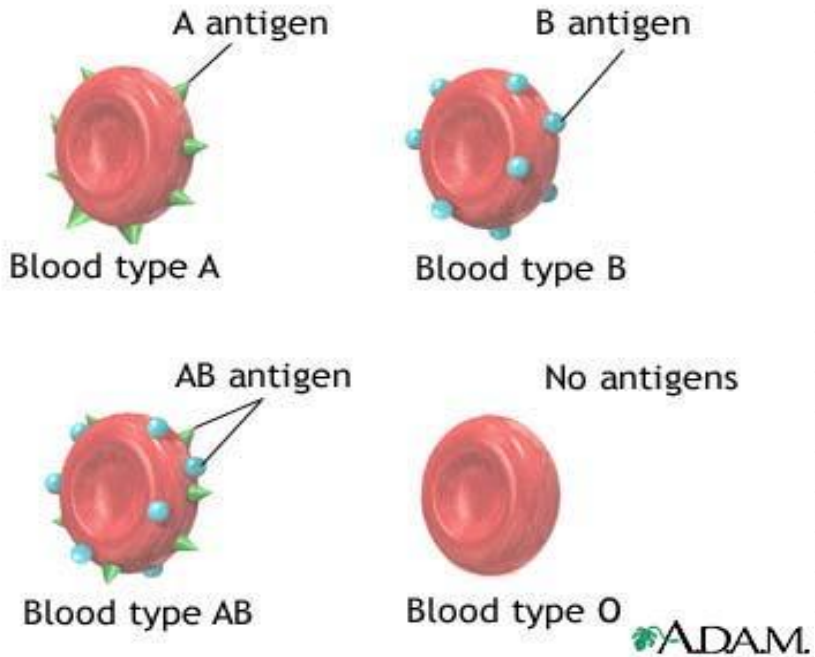
1/2

So the probability would be ¼ X ½ = 1/8

- **Incomplete Dominance**
  - alleles are equally dominant and create a blend
  - What symbols should you use?

- **Codominance**
  - both alleles are expressed in the heterozygote
  - Use $R^{+}R^{-}$ instead of capital and small letters

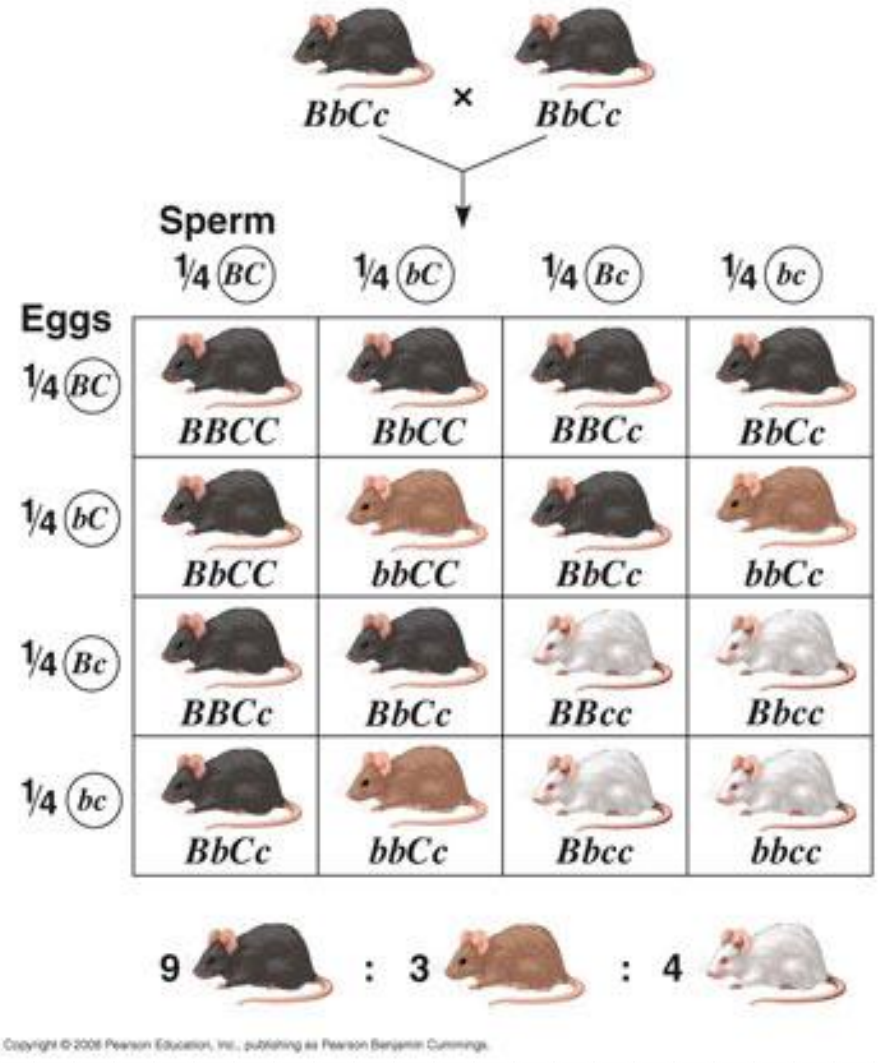
Multiple Alleles - Blood Groups



Phenotype (Blood Group)	Genotypes	RBC
O	ii	
A	$I^A I^A$ or $I^A i$	
B	$I^B I^B$ or $I^B i$	
AB	$I^A I^B$	

Epistasis

- One gene affects the expression of another gene
- E.g.: Coat color



Multiple Choice #51

Use the following additional information to answer the next two questions.

A different variety of homozygous *M. jalapa* produces flowers that are light crimson. Pure-breeding genotypes and phenotypes are:

R^PR^P – deep crimson
RR – light crimson
rr – yellow

When two pure-breeding P_1 plants are cross-pollinated, only scarlet-red-flowered offspring (R^Pr) are produced.

When another pair of pure-breeding P_1 plants are cross-pollinated, only orange-flowered offspring (Rr) are produced.

—from *Engels, 1975*

The likely genotypes of the P_1 plants for these two crosses is represented in row

Row	P_1 genotypes scarlet-red-flowered offspring	P_1 genotypes orange-flowered offspring
A	$R^PR \times rr$	$RR \times rr$
B	$R^PR^P \times rr$	$RR \times rr$
C	$R^Pr \times R^Pr$	$Rr \times Rr$
D	$R^PR^P \times RR$	$R^PR \times Rr$

Multiple Choice #52

Use the following information to answer the next two questions.

In pea plants, tall (*T*) is dominant over short (*t*), and round seed (*R*) is dominant over wrinkled seed (*r*). The Punnett square below shows a cross between a heterozygous tall–heterozygous round-seed pea plant and a short–heterozygous round-seed pea plant. Different types of offspring are represented by numbers.

	TR	*Tr*	*tR*	*tr*
tR	**1**	**2**	**3**	**4**
tr	**5**	**6**	**7**	**8**

Which two types of offspring are pure breeders for both plant height and seed shape?

A. 1 and 6
B. 2 and 5
C. 3 and 8
D. 4 and 7

Multiple Choice #53

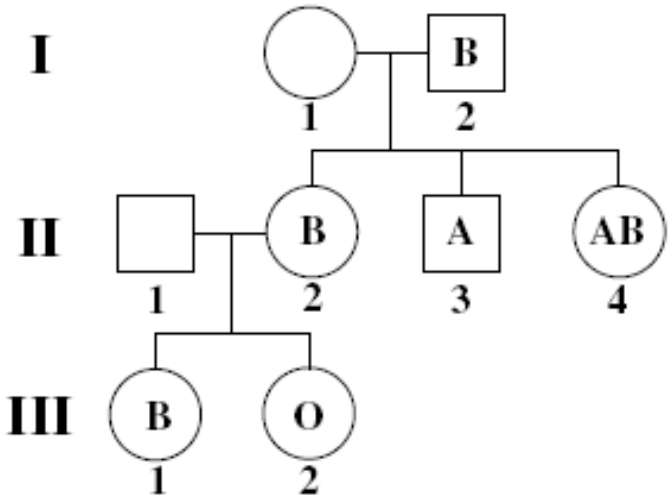
Which two types of offspring, when crossed, could be expected to produce a population in which 50% of their offspring would be tall and 100% would produce round seeds?

A. 1 and 8
B. 2 and 4
C. 3 and 7
D. 5 and 6

Multiple Choice #54

Use the following information to answer the next question.

Pedigree of Human ABO Blood Types



The genotype of individual **I-1** is

A. ii
B. $I^A i$
C. $I^B i$
D. $I^A I^A$

Numerical Response #4

Use the following information to answer the next question.

In addition to the ABO system, human blood may be typed as Rh^+ or Rh^-. The blood types Rh^+ and Rh^- are controlled by the dominant allele *R* (Rhesus positive) and the recessive allele *r* (Rhesus negative).

If a woman with the genotype $I^A I^B Rr$ and a man with the blood type O Rh^- have a child, what is the probability that the child will have blood type A Rh^-?

(Record your **answer as a value from 0 to 1, rounded to two decimal places**, in the numerical-response section on the answer sheet.)

Answer: _____

Multiple Choice #55

Use the following information to answer the next two questions.

Feather colour in parakeets is controlled by two genes. For one pigment gene, the *B* allele produces blue colour and the *b* allele does not produce any colour. For the other pigment gene, the *Y* allele produces yellow colour and the *y* allele does not produce any colour. Any genotype containing at least one *B* allele and one *Y* allele will produce a green parakeet.

Which of the following parental genotypes could produce offspring with the **four** different colour patterns?

A. $BBYy \times BbYy$
B. $BbYY \times Bbyy$
C. $bbYY \times bbyy$
D. $Bbyy \times bbYy$

Multiple Choice #56

What is the probability of obtaining a blue parakeet when two green heterozygous parakeets are crossed?

A. 0

B. $\frac{3}{16}$

C. $\frac{1}{4}$

D. $\frac{9}{16}$

Numerical Response #5

Use the following information to answer the next question.

The coat colour of Labrador retrievers is determined by two alleles. The black allele, *B*, is dominant to the brown allele, *b*. A second pair of alleles, *E* and *e*, affects the expression of the coat colour: the homozygous recessive condition, *ee*, prevents the expression of black or brown and produces a pup with a yellow coat.

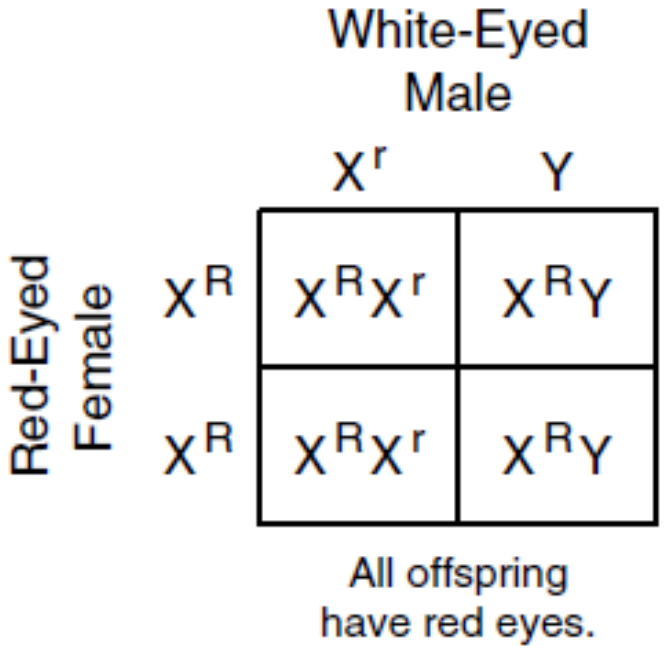
Genotype	Phenotype
B _ E _	Black
bbE _	Brown
_ _ ee	Yellow

If two Labrador retrievers with the genotype *BbEe* were to be crossed, what phenotypic ratio would be expected in their offspring?

Ratio: _____ : _____ : _____

Phenotype: Black Brown Yellow

Sex-linked Punnett



Note: males and females are already considered so don't multiply by half when answering question!

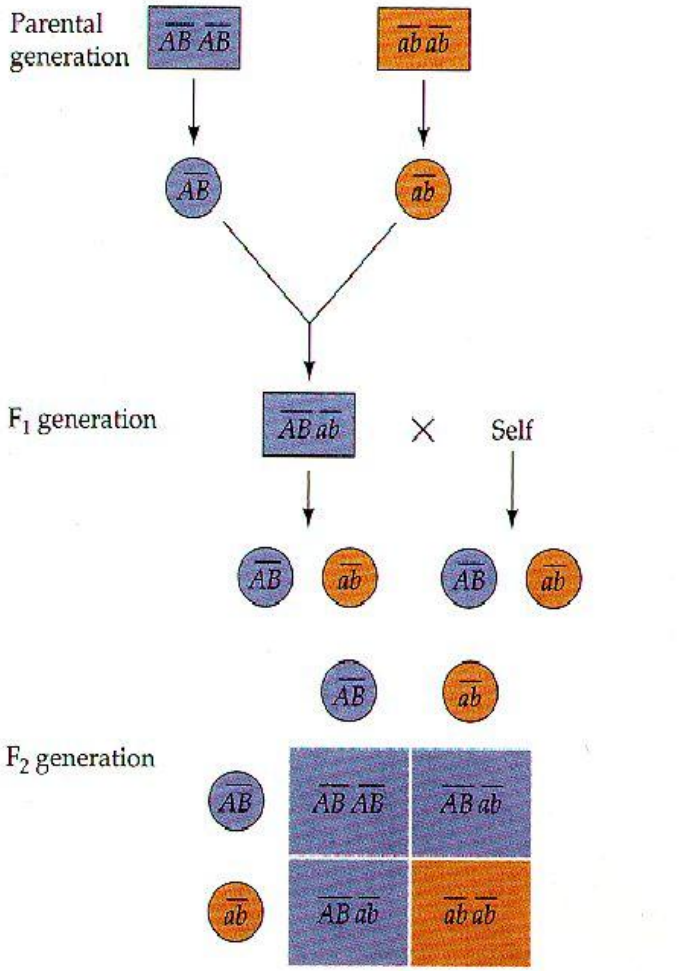
- If traits are carried on the X chromosome (most):
- Only female carriers
- Never passed from father to son

- If carried on the y chromosome (rarely):
- only affect males
- Passed from father to son

Genetic Relationships

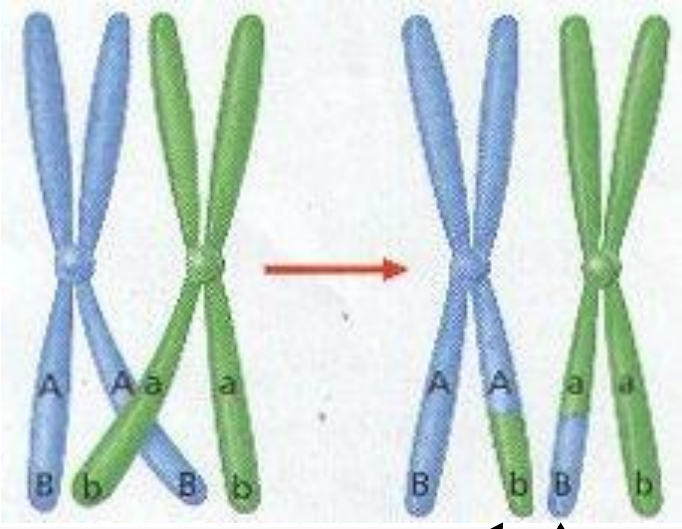
Dominant/Recessive	2 phenotypes, 3 genotypes
Multiple alleles	3+ phenotypes, many genotypes
Incomplete dominance	3 phenotypes (blend), 3 genotypes
Codominance	Both phenotypes show in the heterozygote genotype
Epistasis	Not as many phenotypes as expected for the genotypes because one gene affects expression of another
Pleiotropy	One genotype may have numerous phenotypes
Sex linkage	Allele is carried on a sex chromosome, and phenotype depends on gender
Autosomal inheritance	Males and females show same ratios

Linked Genes



Linked Genes Do Not Assort Independently

Will not produce 4 different gametes…only 2.

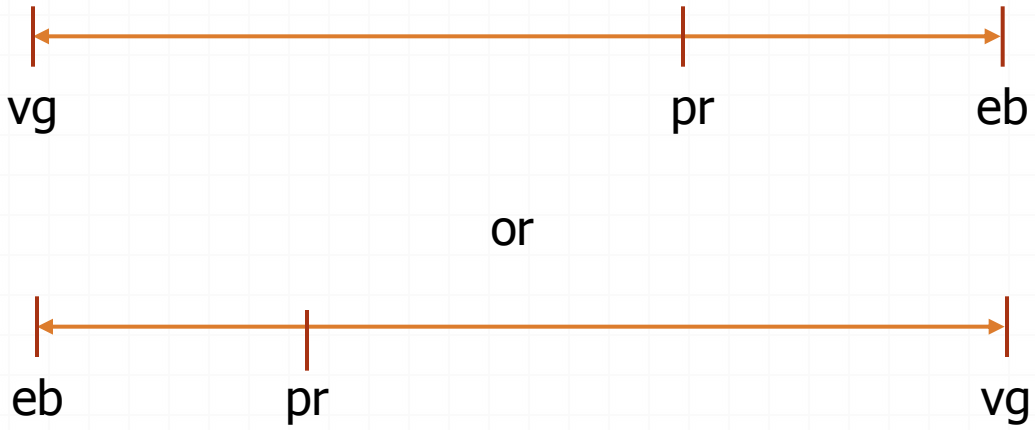


Except **crossing-over** produces 2 new recombinants.

Gene Mapping

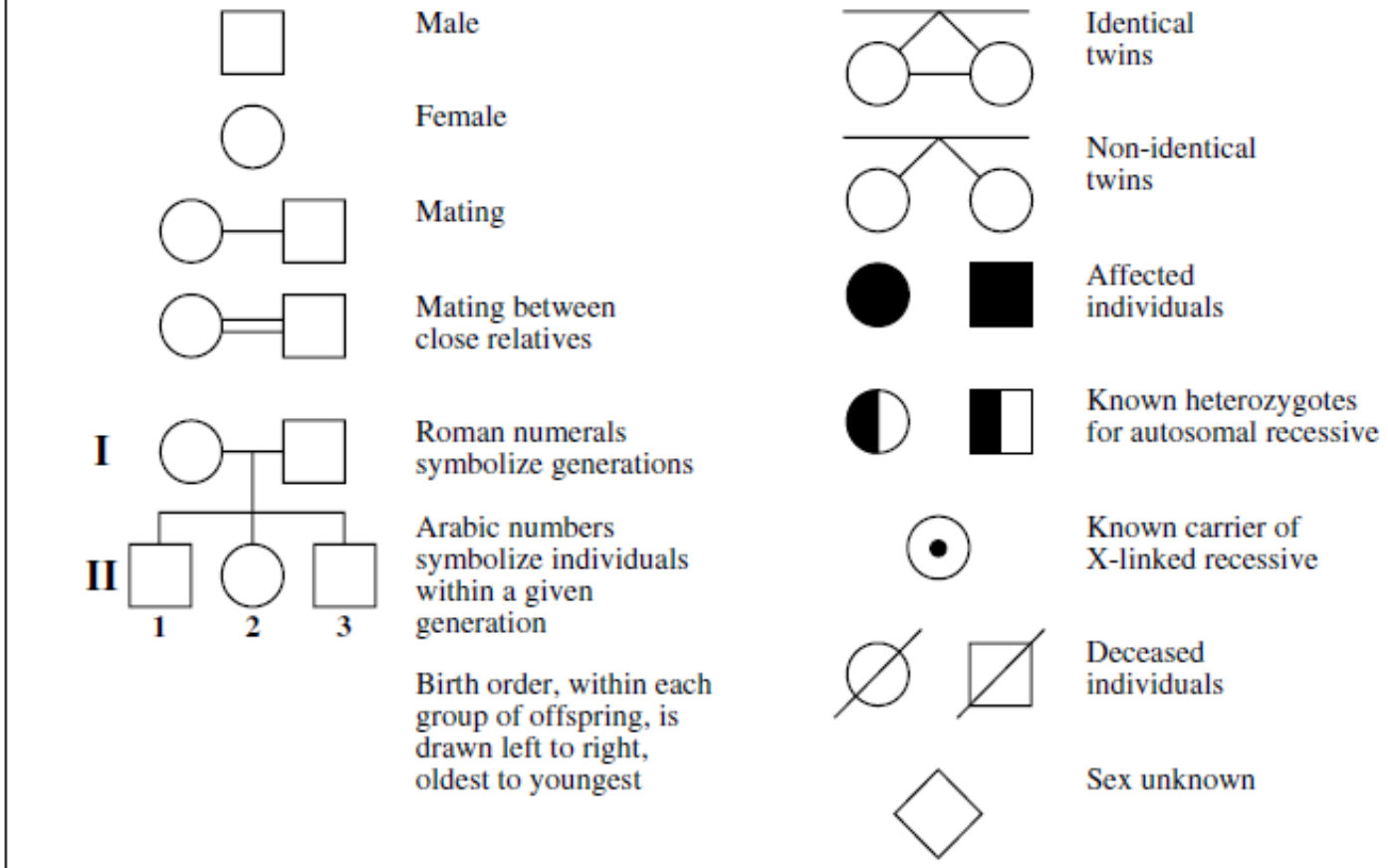
% Crossover= # recombinants x 100= map units
total offspring

- Genes that are **close** together are more likely to be **inherited** together.
- In Drosophila, genes for eye color(pr), wing shape(vg), and body color (eb) are on the same chromosome. Use the following frequencies to map the chromosome.
- pr and vg 12.5%
- pr and eb 6.0%
- vg and eb 18.5%



Pedigree Symbols

Pedigree Symbols

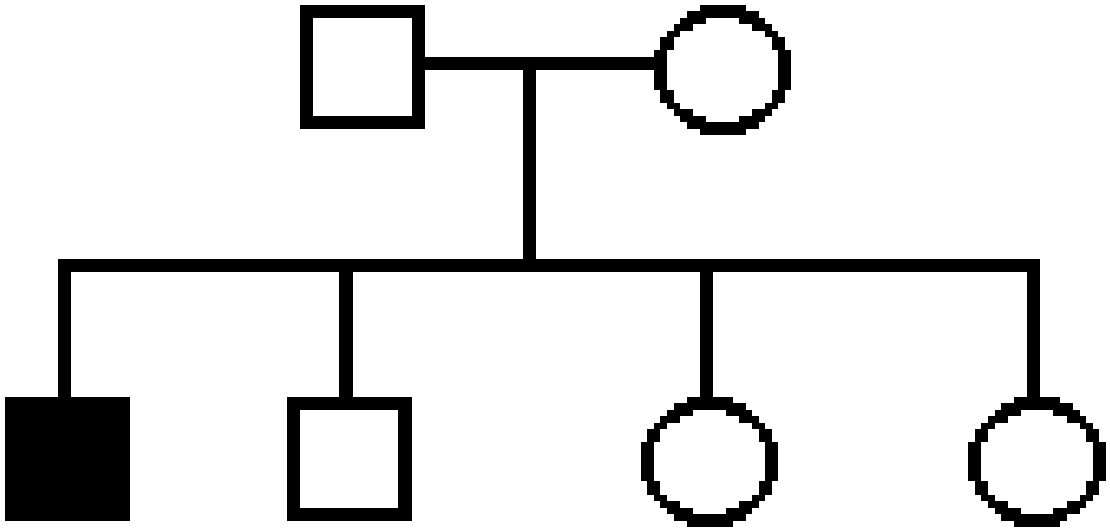


Note: Pedigrees may or may not show individuals who are carriers for a particular trait.

Pedigree Analysis

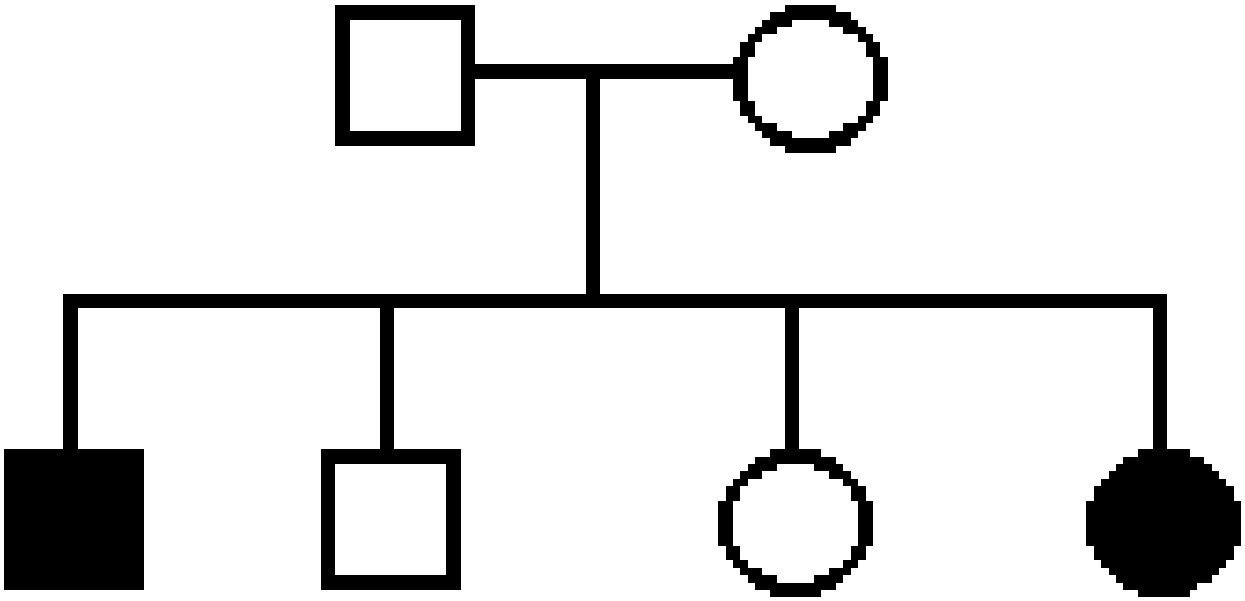
Possible modes of inheritance you need to identify, interpret, calculate probabilities:

- **Autosomal** – equal male female
  - **Dominant** – affected child has affected parent
  - **Recessive** – 2 unaffected parents have affected child
- **X-linked** – more males
  - **Dominant** – no transmission from father to son, affected males produce only affected daughters
  - **Recessive** - affected female has affected father
- **Y-linked** – only males, from father to son



A. For each pedigree, determine modes of inheritance that are POSSIBLE and then use the probabilities to select the one that is most probable:
Could this be Autosomal Dominant?
Could this be X-linked Dominant?
Could this be Autosomal Recessive?
Could this be X-linked Recessive?

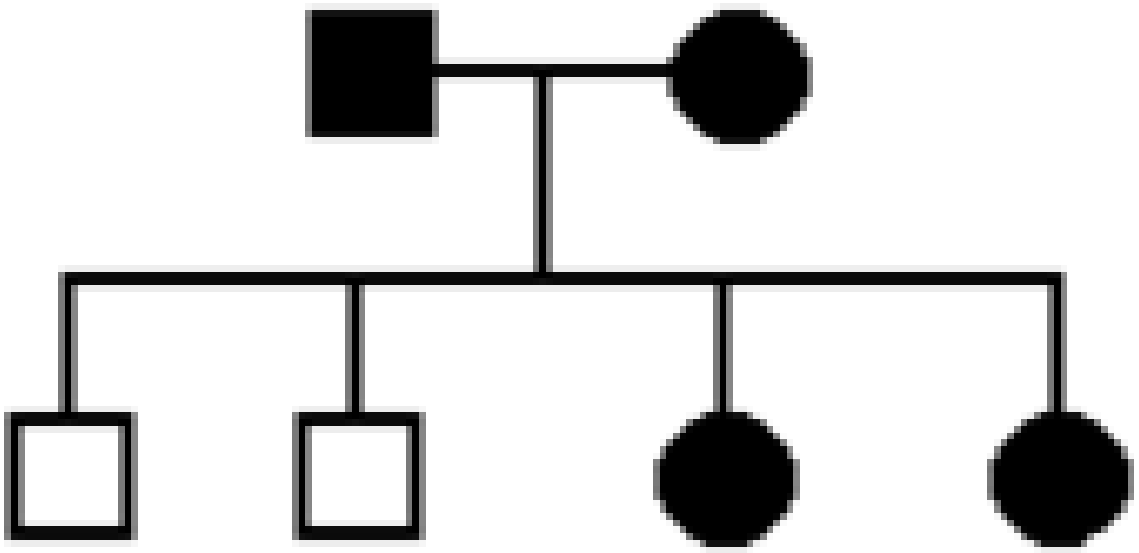
Autosomal or X-linked Recessive



B.

Could this be Autosomal Dominant?
Could this be X-linked Dominant?
Could this be Autosomal Recessive?
Could this be X-linked Recessive?

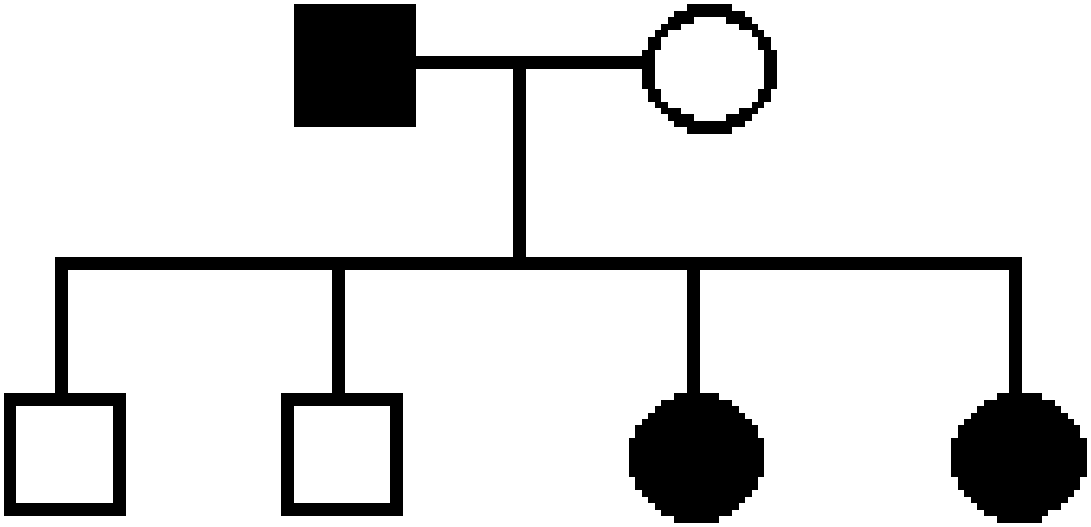
Autosomal Recessive



C.

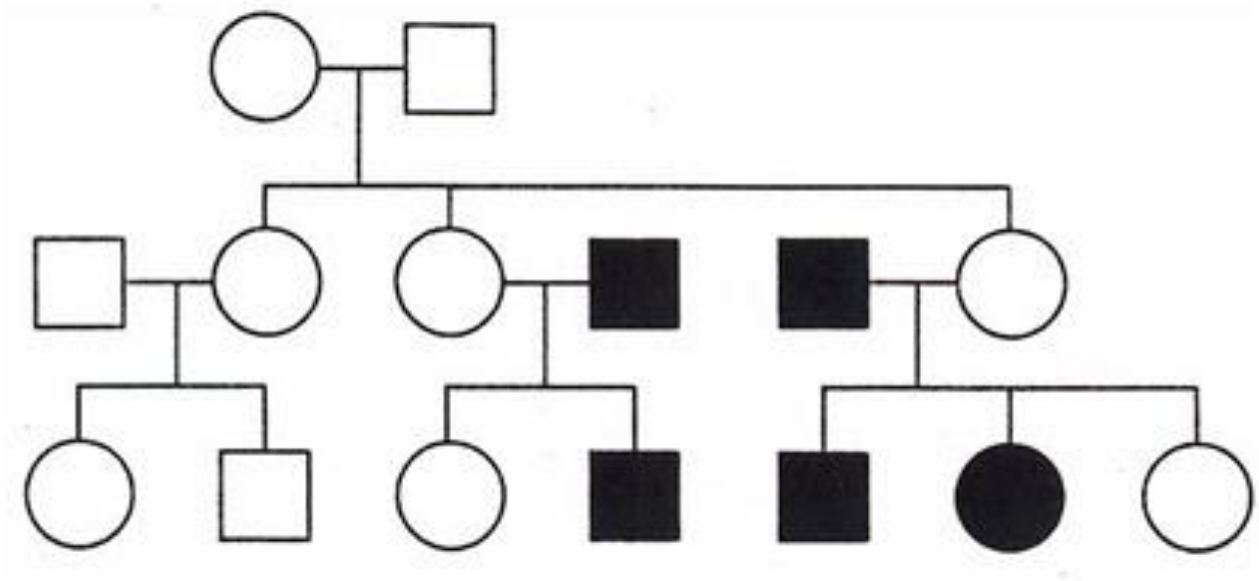
Could this be Autosomal Dominant?
Could this be X-linked Dominant?
Could this be Autosomal Recessive?
Could this be X-linked Recessive?

X-linked or autosomal Dominant



D.
Could this be Autosomal Dominant?
Could this be X-linked Dominant?
Could this be Autosomal Recessive?
Could this be X-linked Recessive?

Could be all 4!



E.
Could this be Autosomal Dominant?
Could this be X-linked Dominant?
Could this be Autosomal Recessive?
Could this be X-linked Recessive?

X-linked or autosomal recessive

Ok, what do you know now?

Genetics Practice Questions

Multiple Choice #57

Use the following information to answer the next two questions.

The gene for a light-sensitive protein found in red cones and the gene for a light-sensitive protein found in green cones lie side by side on the X chromosome. A third gene for a light-sensitive protein found in blue cones was discovered on chromosome 7. Mutations to any of these genes result in the common forms of colourblindness. The mutant alleles for these disorders are recessive.

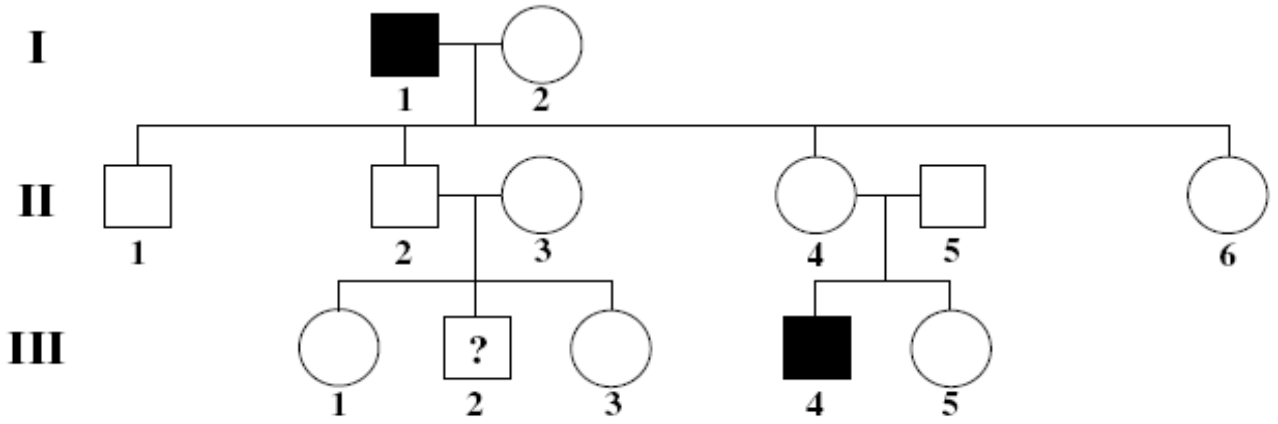
A valid assumption based on this information is that

A. all types of colourblindness are sex-influenced
B. males may be carriers for all types of colourblindness
C. only females may be carriers for blue colourblindness
D. blue colourblindness occurs in males and females with equal frequency

Multiple Choice #58

Use the following additional information to answer the next question.

Pedigree of Red–Green Colourblindness in Humans



Note: Heterozygous individuals have not been identified. The phenotype of III-2 is unknown.

Based on this pedigree,

A. the probability that individual II-4 is a carrier is 50%

B. it is impossible to determine whether individual II-6 is a carrier

C. if individual III-5 is a carrier, all of her female children will have red–green colourblindness

D. if individual II-3 is a carrier, there is a 50% chance that her male child will have red–green colourblindness

Numerical Response #6

Use the following information to answer the next two questions.

Marfan syndrome is an autosomal-dominant disorder of humans. Affected individuals tend to be tall and thin. They have defects in the lens of the eye and weak connective tissue around the aorta. Often, affected individuals excel in sports like volleyball or basketball, but it is not uncommon for people with this syndrome to die suddenly.

A man, heterozygous for Marfan syndrome, and a homozygous recessive woman have a child. What is the probability that the child will be affected by Marfan syndrome?

(Record your **answer as a value from 0 to 1, rounded to two decimal places,** in the numerical-response section on the answer sheet.)

Answer: _____

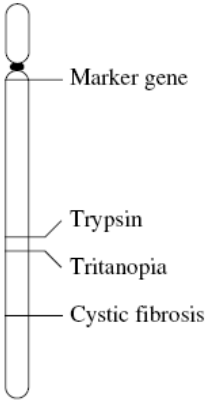
Multiple Choice #59

The use of marker genes and the analysis of crossover frequencies of genes have enabled geneticists to map the location of many genes on human chromosomes. Blue colour vision and blue colourblindness (tritanopia) are controlled by a gene on chromosome 7. The gene for the production of trypsin (a digestive enzyme) and the gene responsible for cystic fibrosis are also found on chromosome 7. Some crossover frequencies of these genes are shown below.

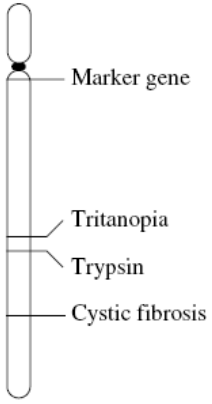
Pair of Genes	Crossover Frequency
Marker gene — cystic fibrosis	18%
Marker gene — tritanopia	13%
Cystic fibrosis — trypsin	6%
Trypsin — tritanopia	1%

Which of the following gene maps shows the correct sequence of these genes on chromosome 7?

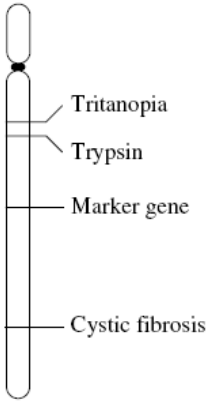
A.



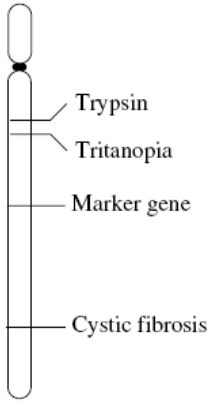
B.



C.



D.



Multiple Choice #60

Use the following information to answer the next three questions.

Cystic fibrosis is the most common genetic disorder among Caucasians, affecting one in 2 000 Caucasian children. The cystic fibrosis allele results in the production of sticky mucus in several structures, including the lungs and exocrine glands. Two parents who are unaffected by the disorder can have a child with the disorder.

Which term **best** describes the allele for cystic fibrosis?

A. X-linked
B. Recessive
C. Dominant
D. Codominant

Multiple Choice #61

Use the additional information to answer the next two questions.

A girl and both her parents are unaffected by the disease. However, her sister is affected by cystic fibrosis.

The genotypes of the mother and father are

A. both homozygous
B. both heterozygous
C. homozygous and heterozygous, respectively
D. heterozygous and homozygous, respectively

Numerical Response #7

These parents, who are unaffected by cystic fibrosis, are planning to have another child. What is the percentage probability that their next child will be affected by cystic fibrosis?

(Record your **answer as a whole number percentage** in the numerical-response section on the answer sheet.)

Answer: _____%

Multiple Choice #62

Use the following information to answer the next question.

> Hypophosphatemia is one of the few genetic diseases caused by a dominant allele carried on the X chromosome. It causes a severe deficiency of phosphate ions in the blood.
>
> —from *Rimoin, et al., 1996*

A female with hypophosphatemia whose father had the disease but whose mother did not will likely transmit the disorder to

A. her sons only
B. her sons and her daughters equally
C. all of her daughters but none of her sons
D. all of her daughters and 50% of her sons

Answer KEY to Multiple Choice

- Try the questions BEFORE you check the answer!!!!!
- Ok, here are the answers:

1. B
2. C
3. D
4. D
5. C
6. B
7. C
8. A
9. D
10. A
11. D
12. B
13. C
14. C
15. D
16. B
17. C
18. C
19. D
20. C
21. A
22. C
23. D
24. A
25. D
26. A
27. A
28. D
29. A
30. A
31. C
32. C
33. D
34. D
35. A
36. A
37. D
38. D
39. D
40. B
41. A
42. B
43. C
44. D
45. D
46. C
47. B
48. D
49. A
50. D
51. B
52. C
53. A
54. B
55. D
56. B
57. D
58. D
59. A
60. B
61. B
62. B

Answers to Numerical Response:

1. 1342
2. 1243
3. 3142
4. 0.25
5. 934
6. 0.50
7. 25