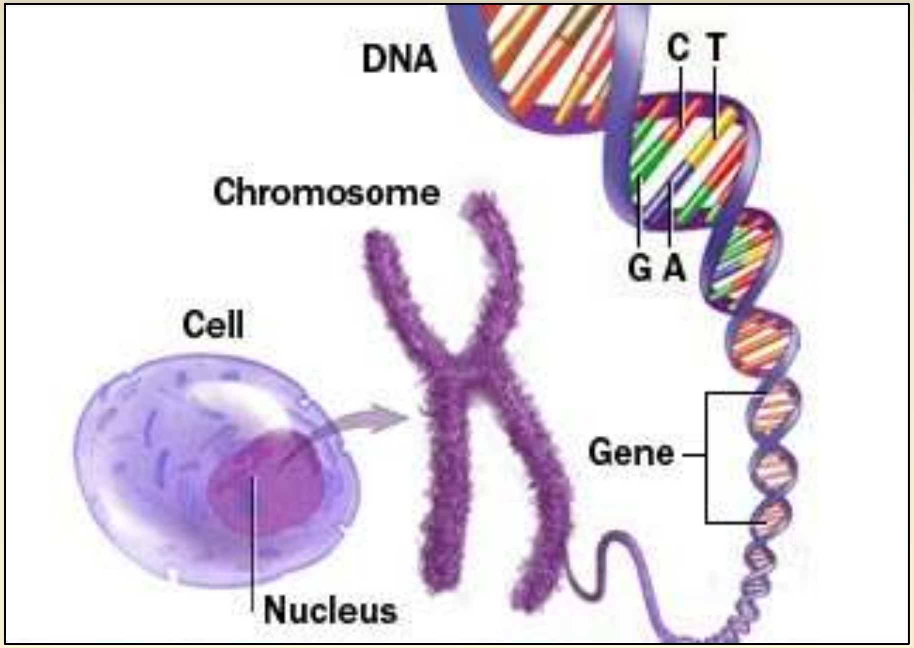
Genetics

the study of genes and inheritance

Figure 7.1 Gregor Mendel (1822–1884) is considered the father of genetics.
Courtesy of the National Library of Medicine



Mendel's work with peas led to discovery of genes and DNA as hereditary material
Gene – DNA coding for one polypeptide or protein. Several genes often involved in traits
Genetic variation - basis for evolution and plant selection

Summary

- Genetics is the study of genes.
- Inheritance is how traits, or characteristics, are passed on from generation to generation.
- Chromosomes are made up of genes, which are made up of DNA.
- Genetic material (genes, chromosomes, DNA) is found inside the nucleus of a cell.
- Gregor Mendel is considered “The Father of Genetics“, discovered laws of inheritance

Inheritance

Offspring appear to have mixture of parental traits.

Michelle, Malia, Barack, and Sasha Obama

John and Julian Lennon

Plants also have characters that vary, offspring also resemble parents

How are traits inherited?

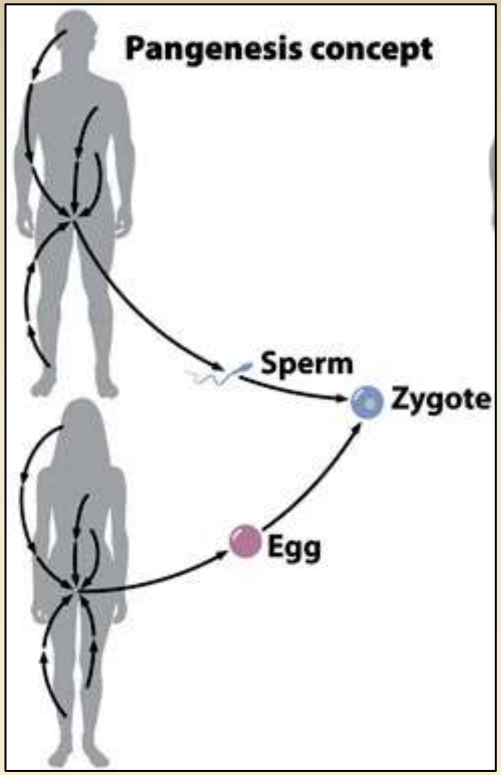
Old ideas

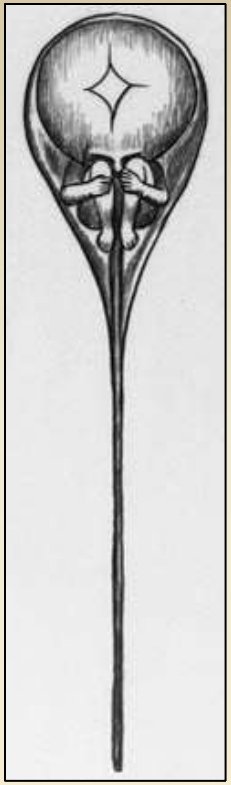
Pangenesis – genetic particles (pangenes) come from all over the body
Aristotle, Darwin

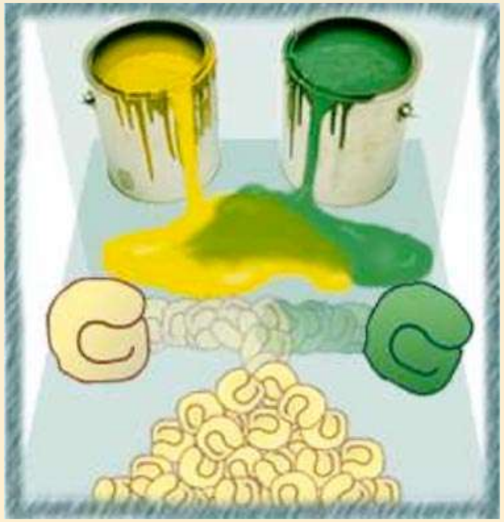
Homunculus – little person in sperm (Leeuwenhoek), woman serves only as incubator, or little human in eggs, sperm stimulates growth (deGraaf)

Blending Inheritance – both parents contribute
Traits blend into each other, like paint

Problems – some genes don’t blend, one is sometimes dominant. Other traits seem to disappear, but sometimes show up later.







Gregor Johann Mendel (1822—1884)

- Mendel discovered a mechanism of inheritance while conducting experiments on garden peas at a monastery in Brunn, Austria (now Brno, Czech Republic).

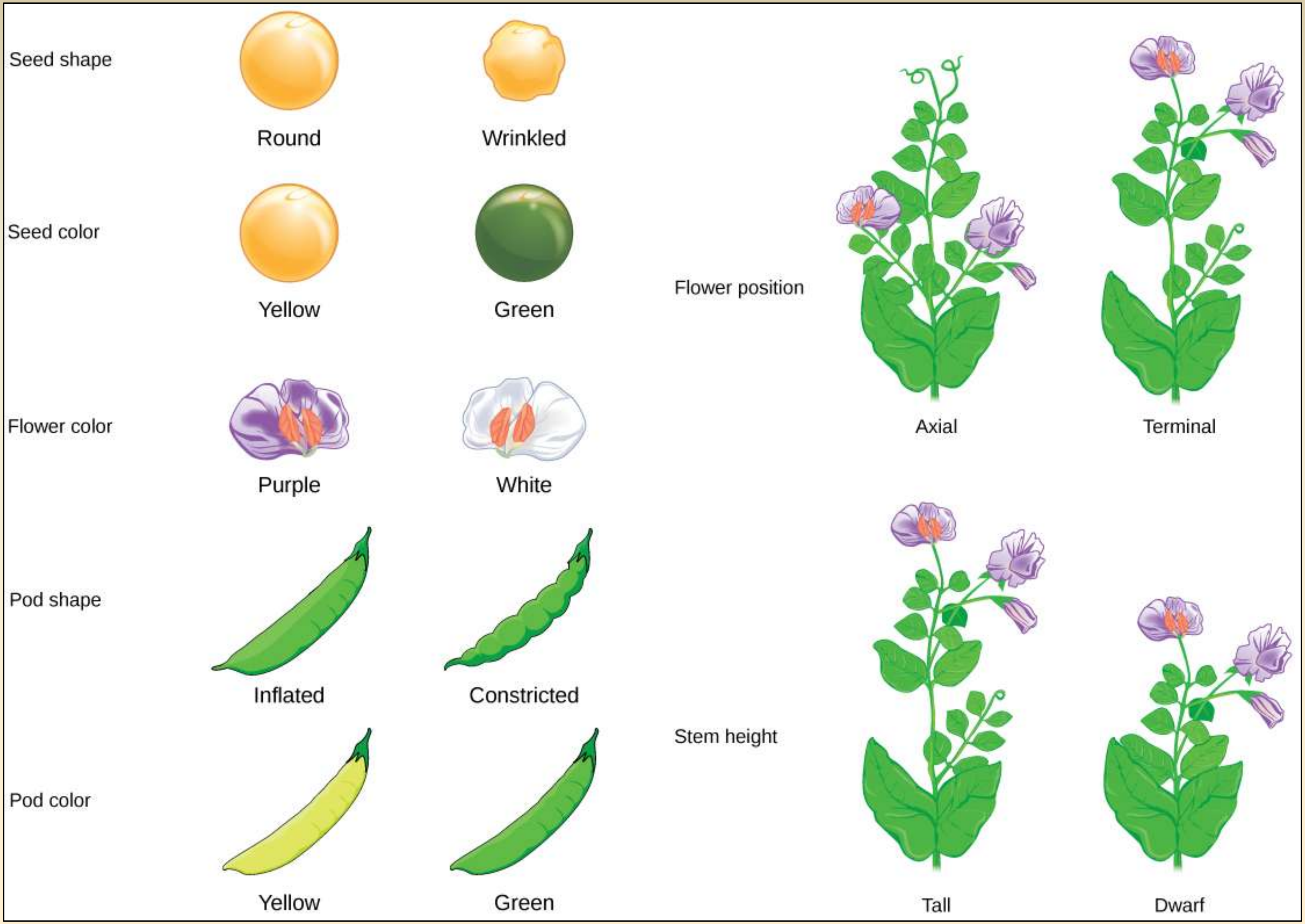
Gregor Johann Mendel

- Austrian Monk, born in what is now Czech Republic in 1822

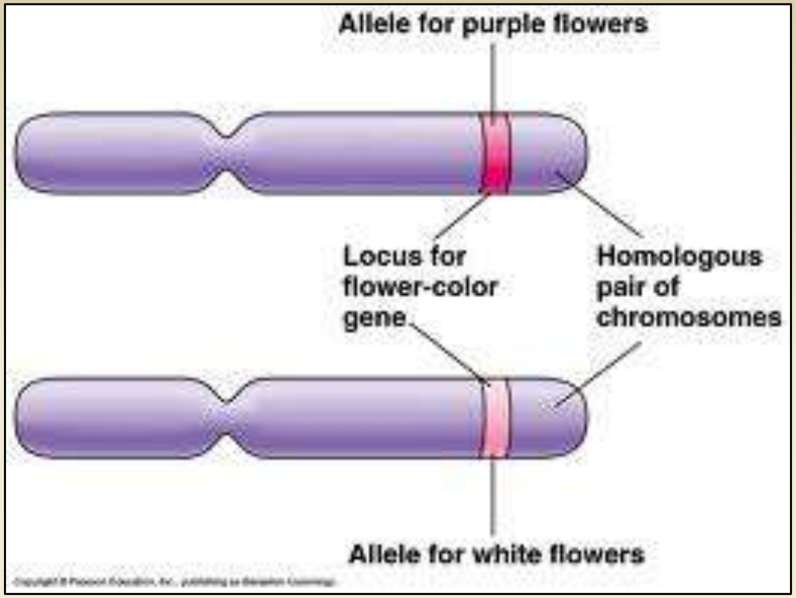
Gregor Mendel

- Son of peasant farmer, studied Theology and was ordained priest Order St. Augustine.
- Went to the university of Vienna, where he studied botany and learned the Scientific Method
- Worked with pure lines of peas for eight years
- Prior to Mendel, heredity was regarded as a "blending" process and the offspring were essentially a "dilution"of the different parental characteristics.

Mendel’s Pea Characters



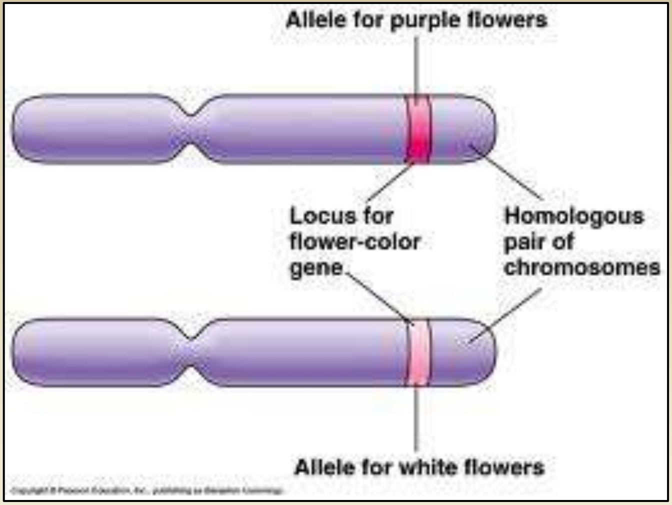
Gene – Locus - Alleles



Genes - sequence of nucleotides in a DNA molecule that code for specific traits.

Locus (Loci) - location of a gene on a chromosome.

Alleles - different molecular forms of a gene. (From the Greek "allelon" meaning "of each other").



Homozygous - alleles are the same

Heterozygous - alleles are different.

Dominant allele - when one allele masks the other.

Recessive allele - the trait that is masked by the dominant allele (this situation is often caused by a gene that codes for a non-functioning enzyme).

Homozygous dominant - when an individual has a pair of dominant alleles.

Homozygous recessive - has a pair of recessive alleles.

- **Genotype** – the genetic makeup of an organisms
- **Phenotype** – the physical appearance of an organism (Genotype + environment)

- **Monohybrid cross**: a genetic cross involving a single pair of genes (one trait); parents differ by a single trait.
- **P** = Parental generation
- **F_1** = First filial generation; offspring from a genetic cross.
- **F_2** = Second filial generation of a genetic cross

Mendel’s experimental method

True-breeding pea strains – one purple, one white

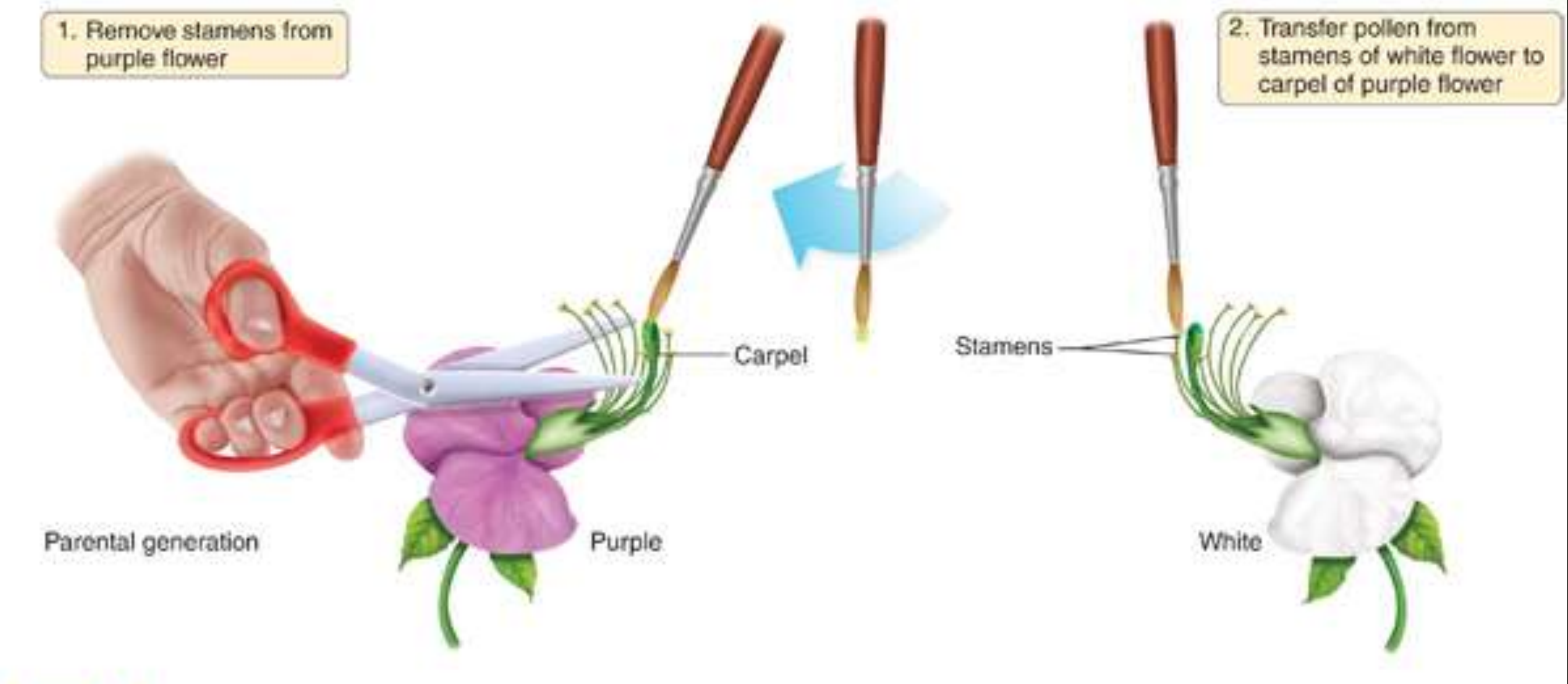
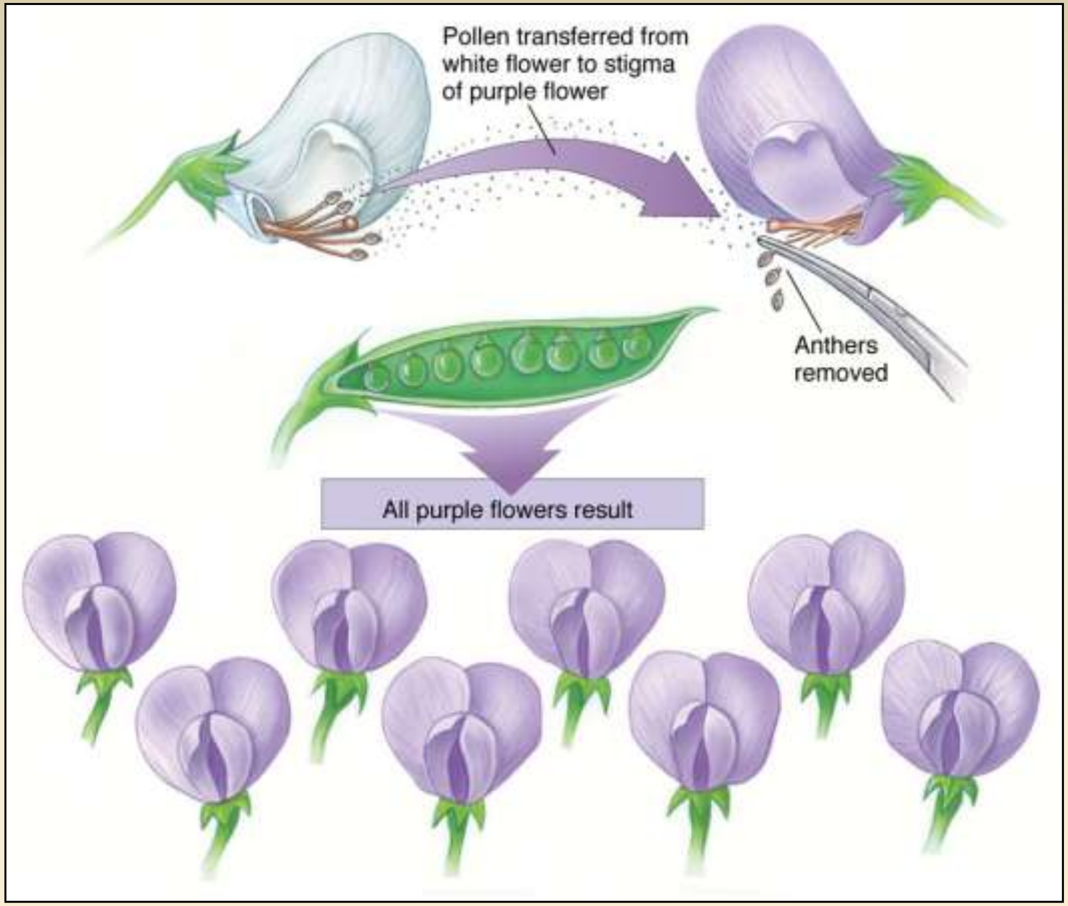


Figure 7.2 The flower of the garden pea. Although the garden pea is noarmally self-pollinated, experimental crosses can be made by opening the flower and removing the anthers before they mature. Pollen from another pea flower is then transferred.

Monohybrid Cross – mating individuals that differ in only one trait, in this case flower color

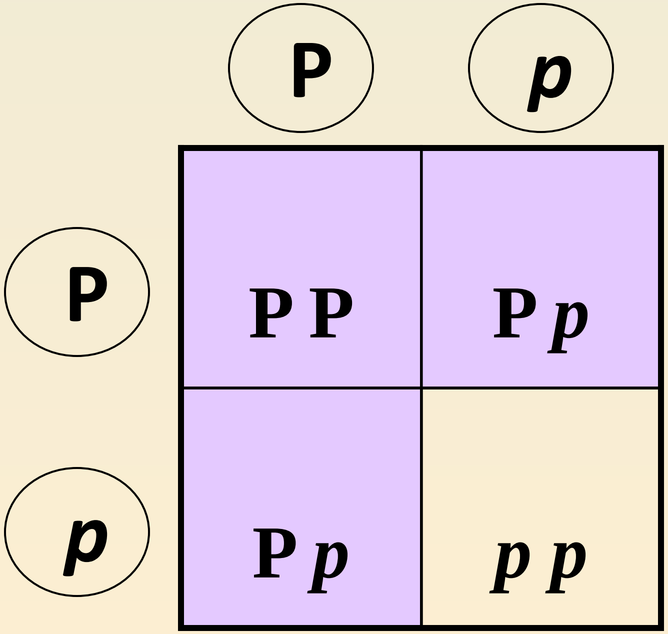
First Cross PP x pp – F1 all purple, Pp



- In this experiment of a cross between true breeding white- and purple-flowered plants, Mendel pried open the surrounding petals of the purple-flowered plant and removed the male part, thus preventing self-fertilization. Then he dusted the anther with pollen he had selected from the white-flowered plant. The resulting seeds were planted and grew, all producing purple flowers.

Punnett Square - diagram illustrating the possible offspring of a mating.

P *p* × P *p*

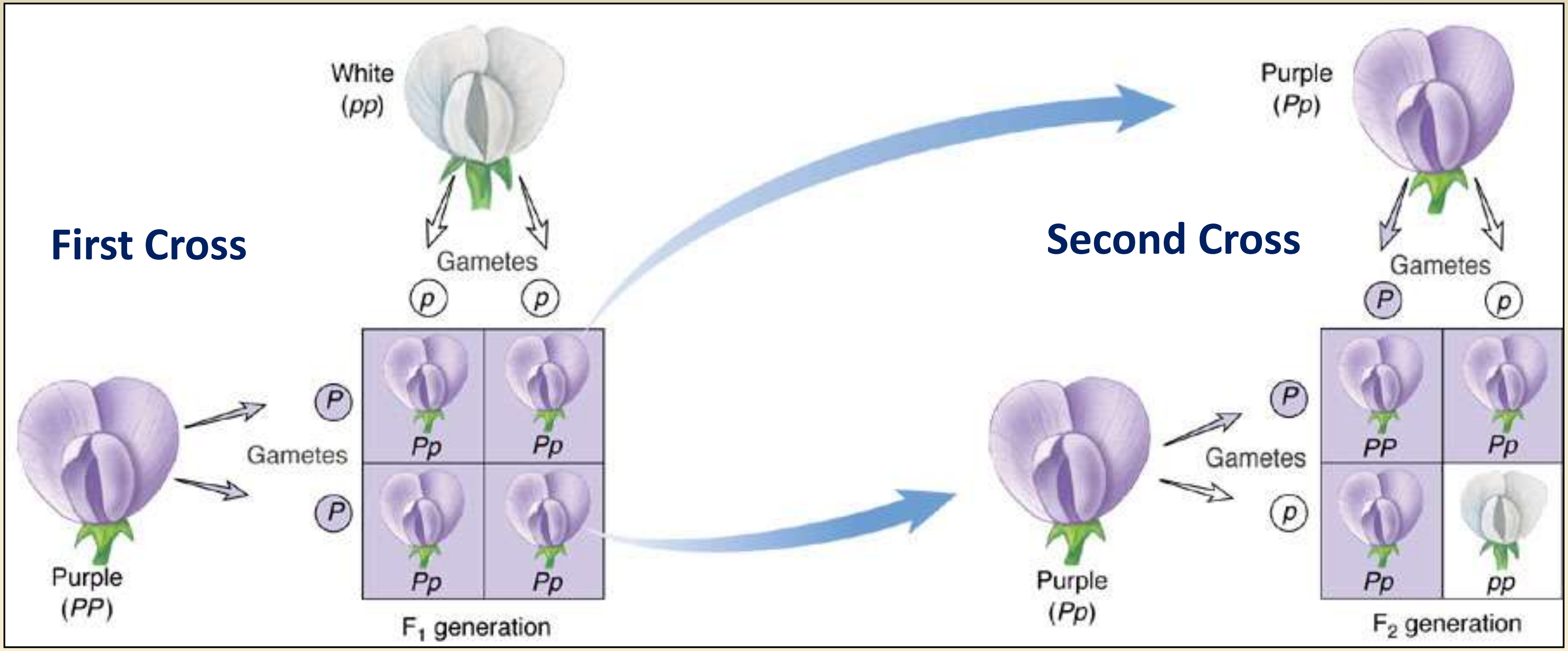


	P	***p***
P	**P P**	**P *p***
p	**P *p***	***p p***

Genotypes:
1 PP
2 Pp
1 pp

Phenotypes:
3 Purple
1 White

First Cross – PP x pp - all purple in F1, Pp
Second Cross – Pp x Pp – 3:1 ratio in F2



- Crossed true breeding white-flowered (recessive) and purple-flowered (dominant) plants. All F1 offspring of this cross were purple-flowered, and genetically heterozygous (Pp).
- Crossed F1 , the resulting F2 offspring averaged 3 purple- for every 1 white-flowered plant, a 3:1 phenotypic ratio. However, the ratio of genotypes is 1:2:1 (1PP: 2Pp : 1pp).

What did Mendel conclude?

- Inheritance is determined by pair of “*factors*” passed on from one generation to another.
- Mendel knew nothing about chromosomes, genes, or DNA. Why?

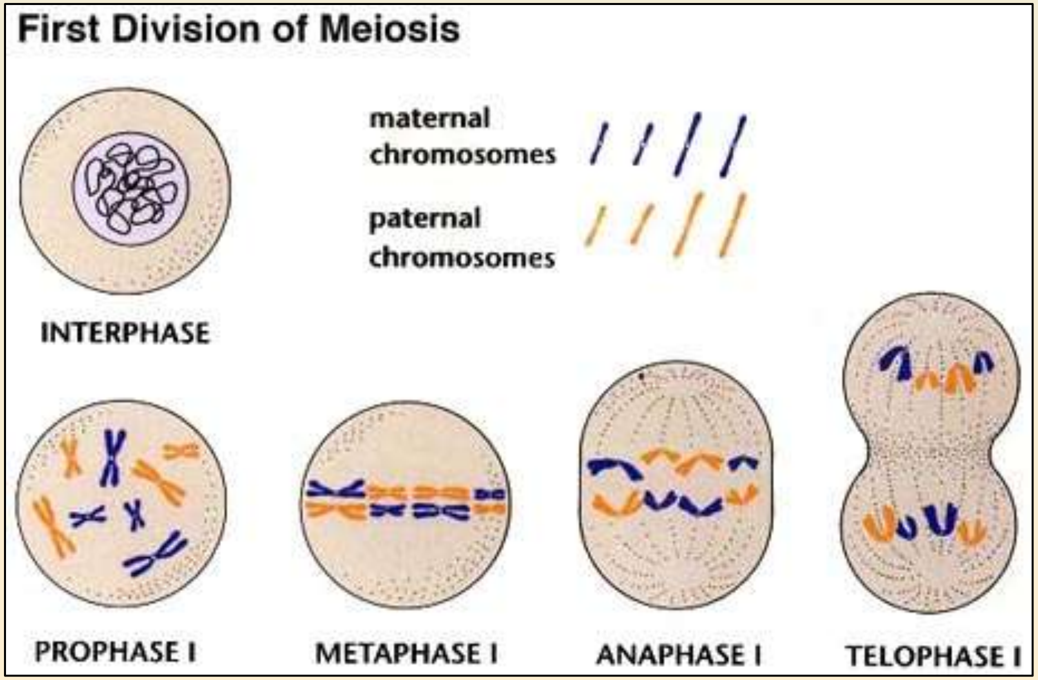
These terms hadn’t yet been defined.

What were Mendel’s “factors”

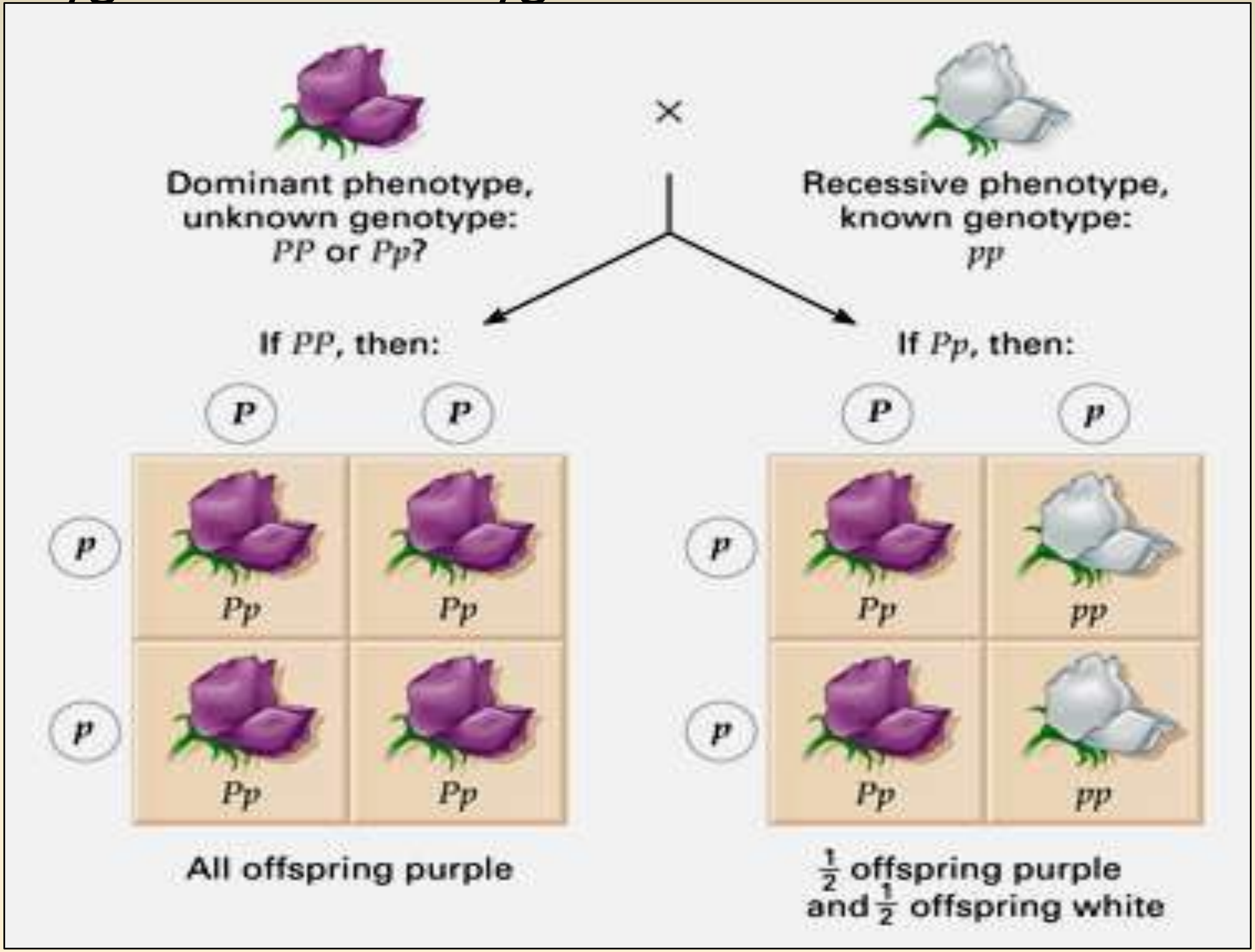
- The ‘factors” that Mendel mentioned were the genes.
- Each gene has different forms called *alleles.* Now we know each individual has two sets of chromosomes, one from each parent.
- Mendel also stated that some alleles are dominant and some are recessive.

Principle of Segregation

When gametes are formed, the pairs of hereditary factors (genes) become separated (segregated), so that each sex cell (egg/sperm) receives only one kind of gene.



Testcross - A testcross is designed to reveal whether an organism that displays the dominant phenotype is homozygous or heterozygous.



Dihybrid crosses

Matings that involve parents that differ in **<u>two</u>** genes (two independent traits)

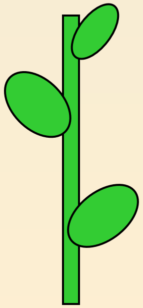
For example, flower color:

P = purple (dominant)

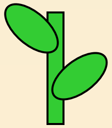
p = white (recessive)

and stem length:

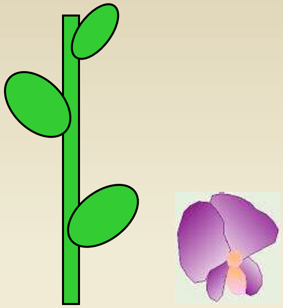
T = tall



t = short

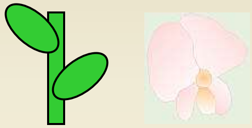


Dihybrid cross: flower color and stem length



TT PP × *tt pp*

(tall, purple) (short, white)



Possible Gametes for parents

T P and *t p*

	tp	*tp*	*tp*	*tp*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*

F1 Generation: All tall, purple flowers (T*t* P*p*)

Dihybrid cross F_2

If F_1 generation is allowed to self pollinate, Mendel observed 4 phenotypes:

T*t* P*p* × T*t* P*p*

(tall, purple) (tall, purple)

Possible gametes:

TP T*p* *t*P *tp*

	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

Four phenotypes observed

Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

Dihybrid cross

9 Tall purple

3 Tall white

3 Short purple

1 Short white

	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

Phenotype Ratio = 9:3:3:1

Dihybrid cross: 9 genotypes

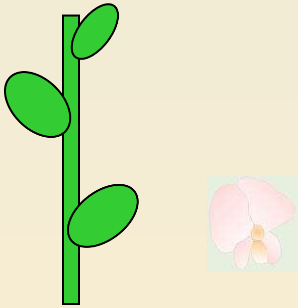
Genotype ratios (9):

1	TTPP	Tall, purple (9)
2	TTP*p*	
2	T*t*PP	
4	T*t*P*p*	
1	TT*pp*	Tall, white (3)
2	T*tpp*	
1	*tt*PP	Short, purple (3)
2	*tt*P*p*	
1	*ttpp*	Short, white (1)

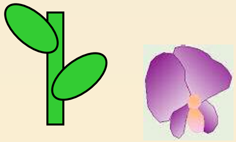
Four Phenotypes:

Tall, purple (9)

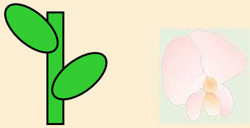
Tall, white (3)



Short, purple (3)

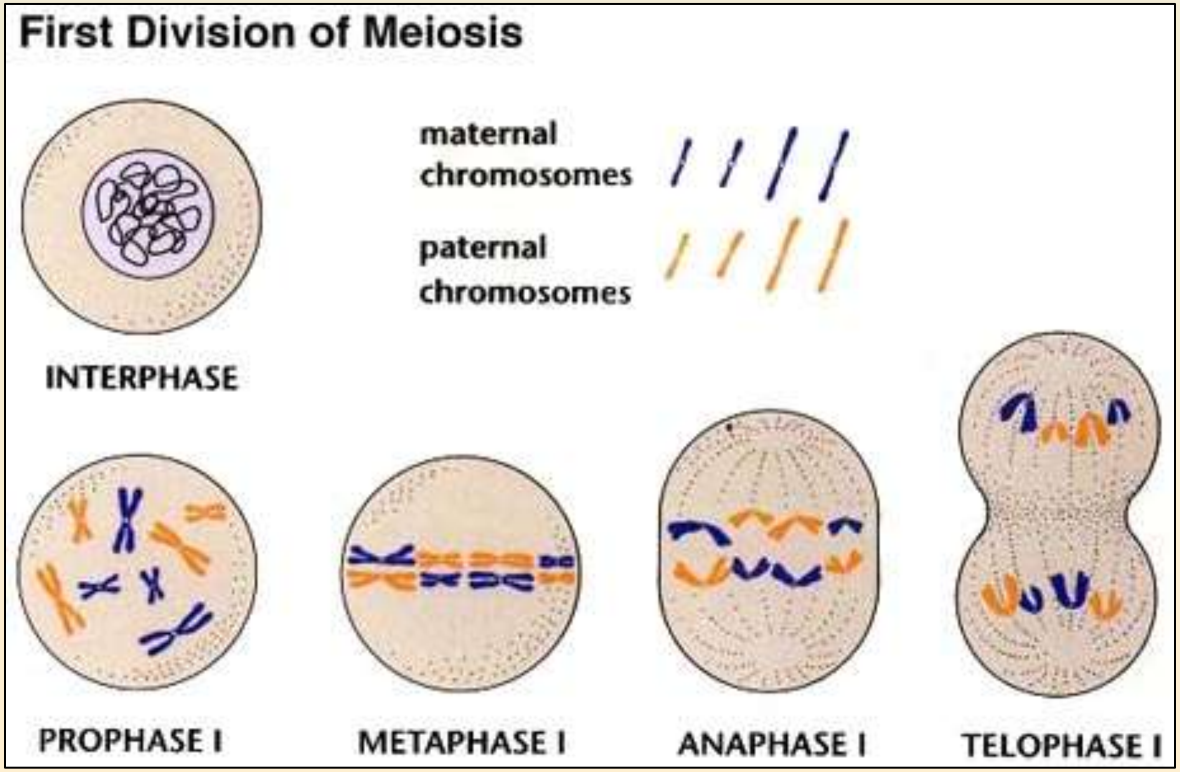


Short, white (1)



2. Principle of Independent Assortment

- Members of one gene pair segregate independently from other gene pairs during gamete formation
- Genes get shuffled – these many combinations are one of the advantages of sexual reproduction



Beyond Mendelian Genetics: Incomplete Dominance

Mendel was lucky!

Traits he chose in the pea plant showed up very clearly…

One allele was dominant over another, so phenotypes were easy to recognize.

But sometimes phenotypes are not very obvious…

Some exceptions to Mendel's principles:

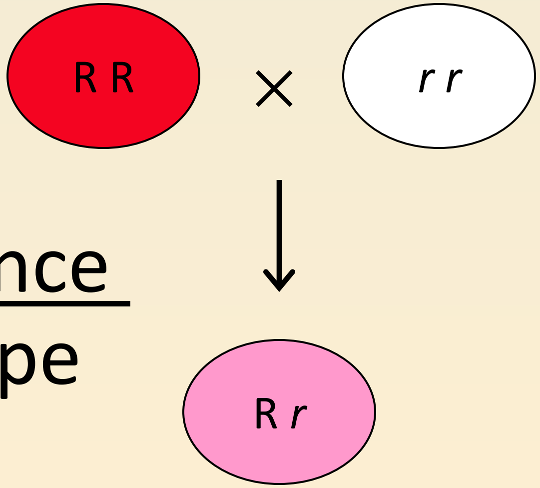
- Some alleles are neither dominant nor recessive (<u>incomplete dominance</u>).
- Many traits are controlled by more than one gene (<u>polygenic traits</u>)
- Genes are sometimes <u>linked</u> on the same chromosome

Incomplete Dominance

Snapdragon flowers come in many colors.

If you cross a red snapdragon (RR) with a white snapdragon (rr)

You get PINK flowers (Rr)!



R R × *r r*

↓

R *r*

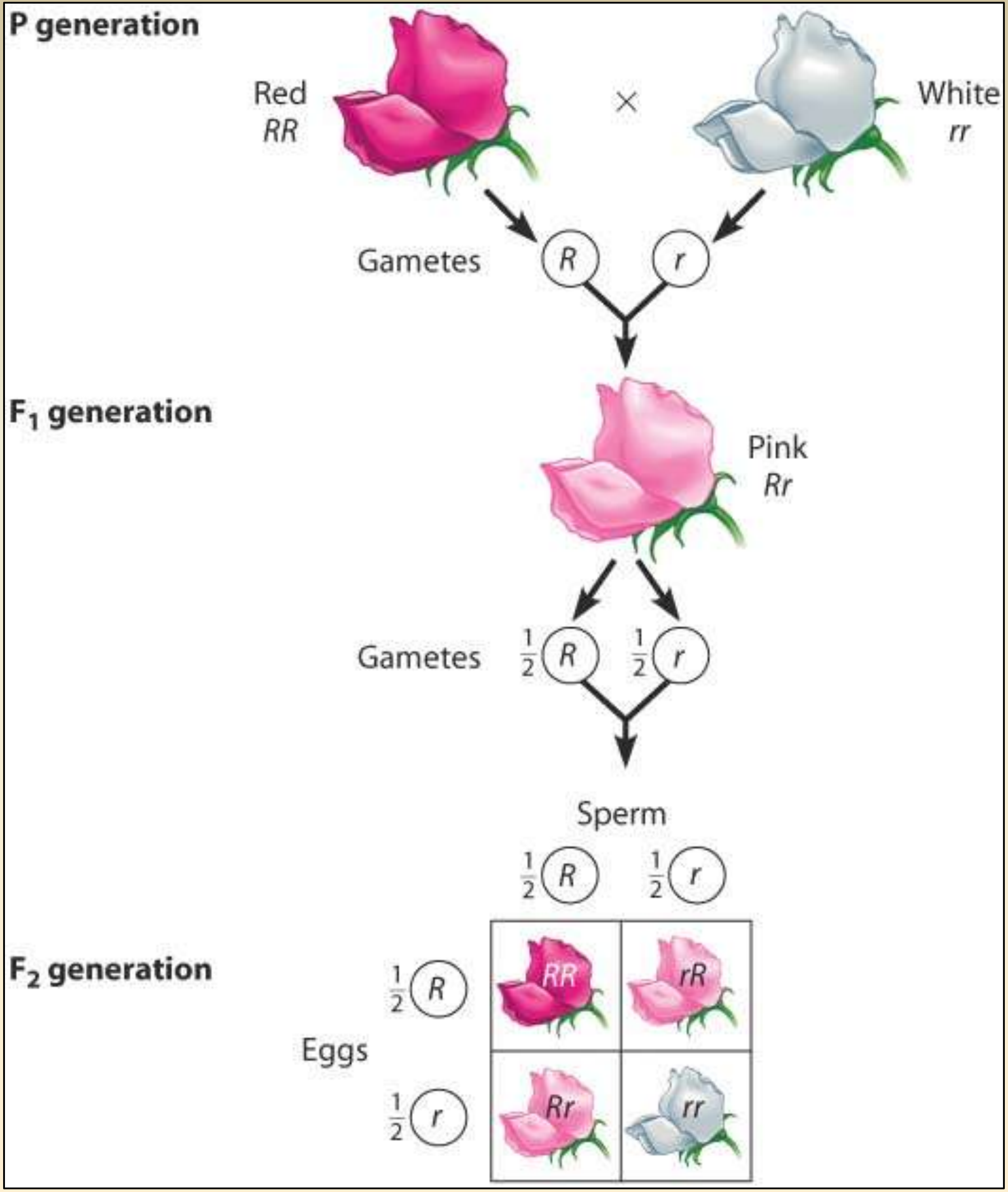
Genes show incomplete dominance when the heterozygous phenotype is intermediate.

Incomplete Dominance

Snapdragons

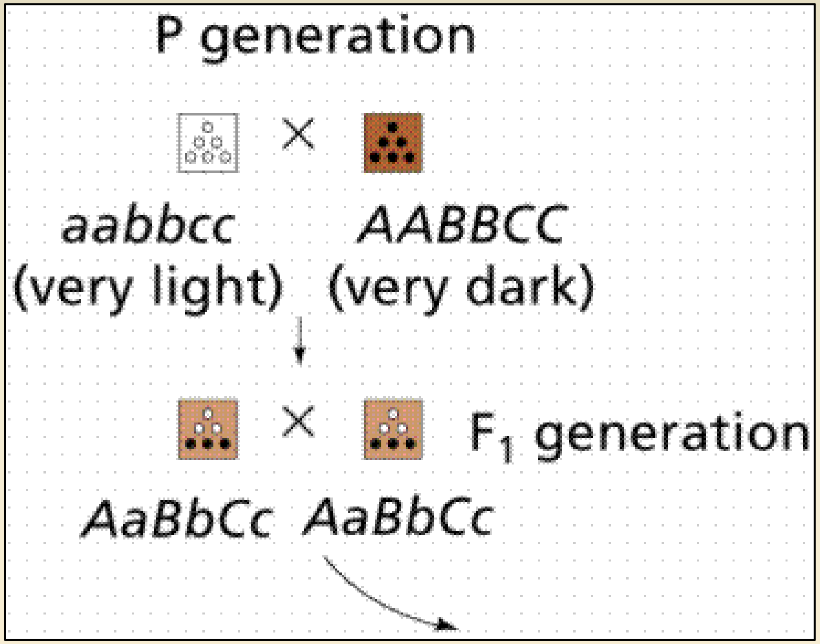
F1 is intermediate in color

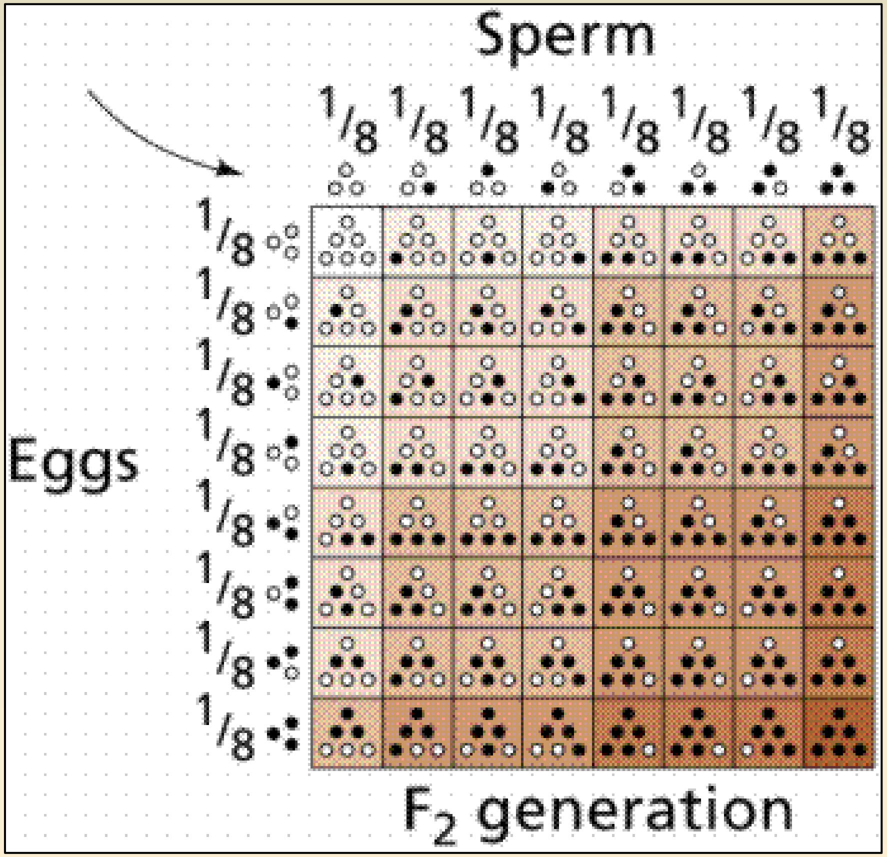
Neither allele is dominant, no masking

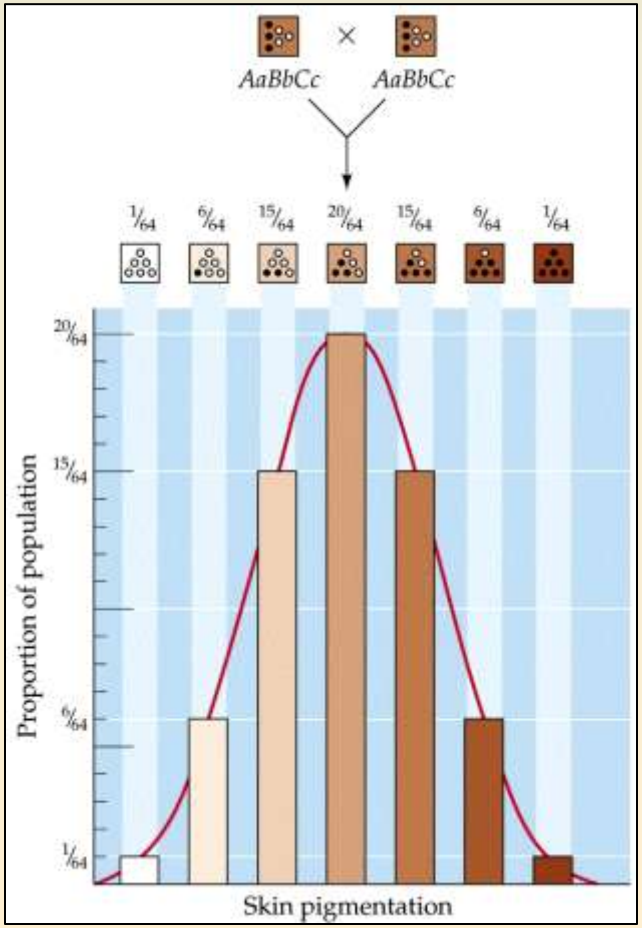


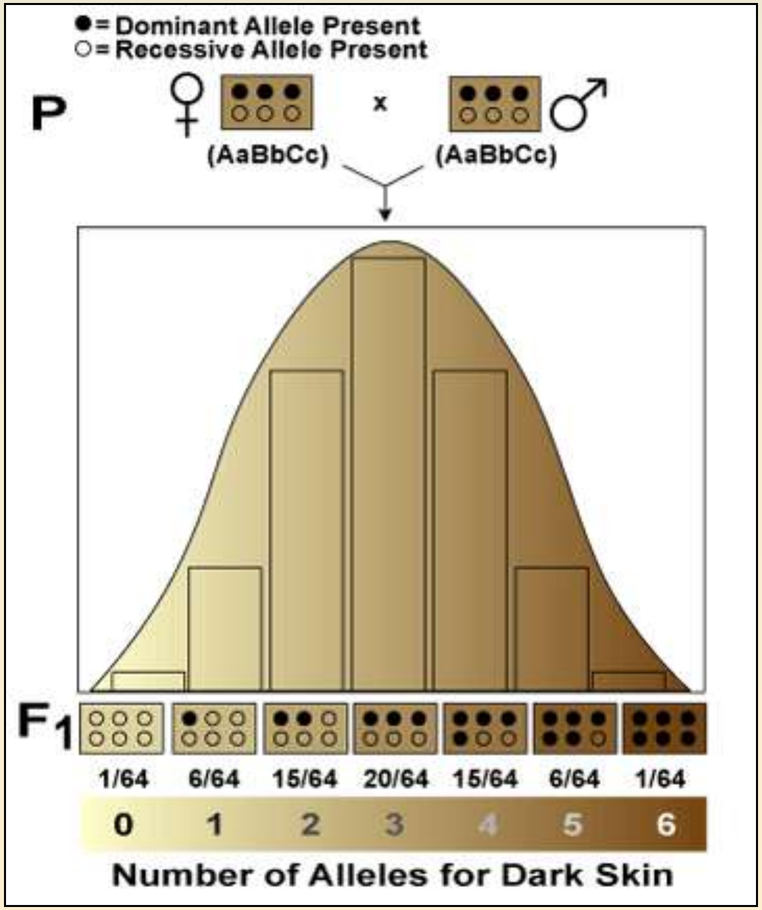
Polygenic trait – based on more than one gene (locus)

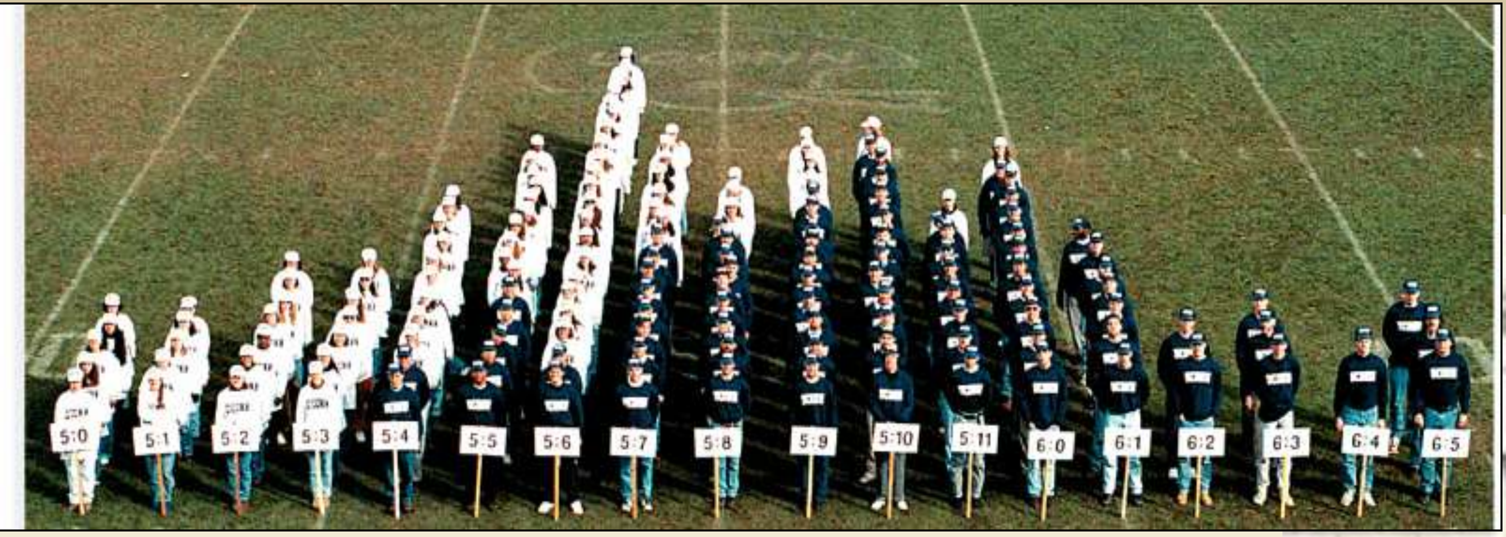
Human skin color - at least 3 loci that affect the production of melanin

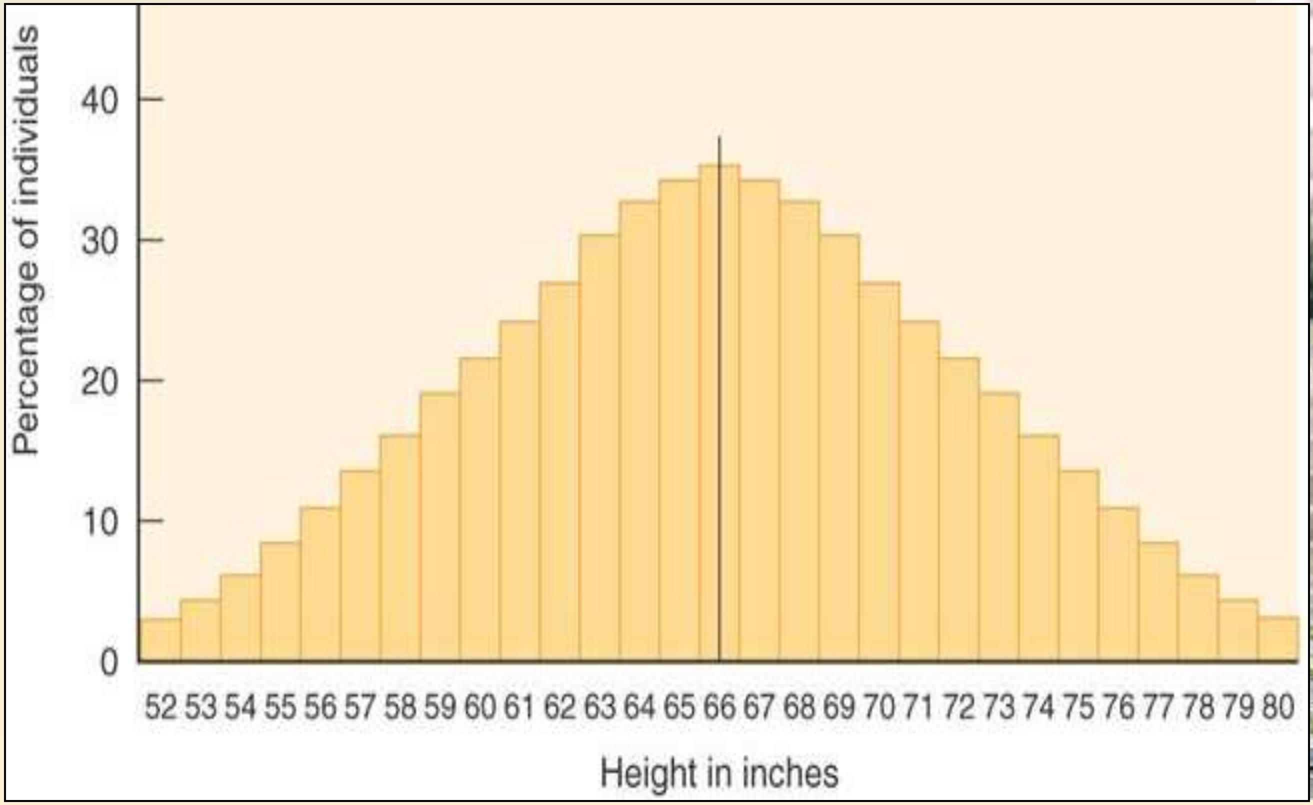





AaBbCc
×
AaBbCc
1/64
6/64
15/64
20/64
15/64
6/64
1/64
20/64
15/64
6/64
1/64
Proportion of population
Skin pigmentation


● = Dominant Allele Present
○ = Recessive Allele Present
P
♀
x
♂
(AaBbCc)
(AaBbCc)
F1
1/64
6/64
15/64
20/64
15/64
6/64
1/64
0
1
2
3
4
5
6
Number of Alleles for Dark Skin


5:0
5:1
5:2
5:3
5:4
5:5
5:6
5:7
5:8
5:9
5:10
5:11
6:0
6:1
6:2
6:3
6:4
6:5


Percentage of individuals
40
30
20
10
0
52 53 54 55 56 57 58 59 60 61 62 63 64 65 66 67 68 69 70 71 72 73 74 75 76 77 78 79 80
Height in inches

Wheat kernel color is an example of polygenic inheritance.

- There are two genes which control wheat kernel color. The phenotypes will vary from a dark red color to a light tan color (called white) .
- The darkest kernels are produced from a plant that has 4 dominant alleles.
- The lightest kernels are produced from a plant that has 4 recessive alleles.
- Intermediate color are produced from having 3 or 2 dominant alleles.

Inheritance of Seed Color in Wheat – controlled by 3 sets of alleles

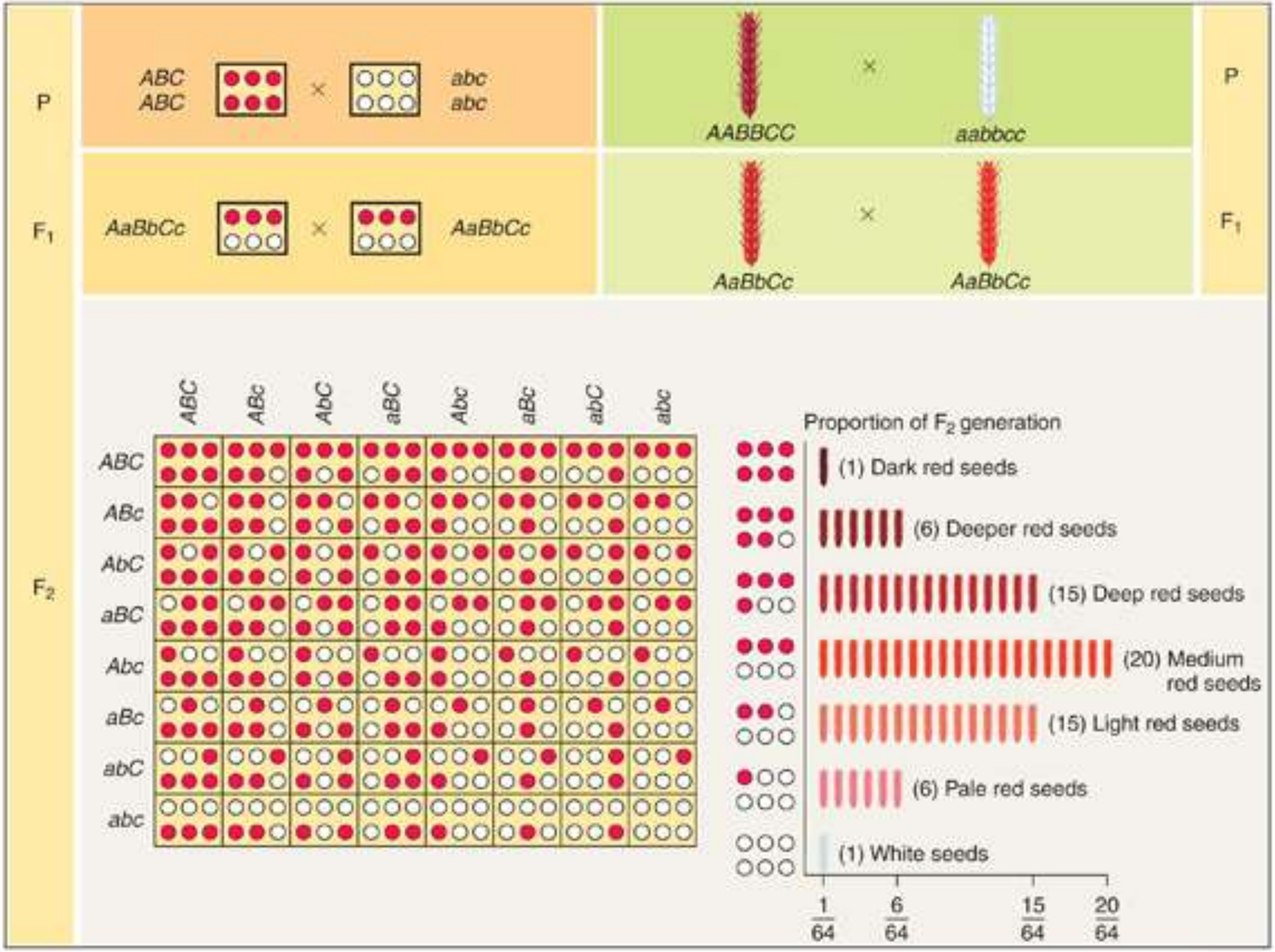
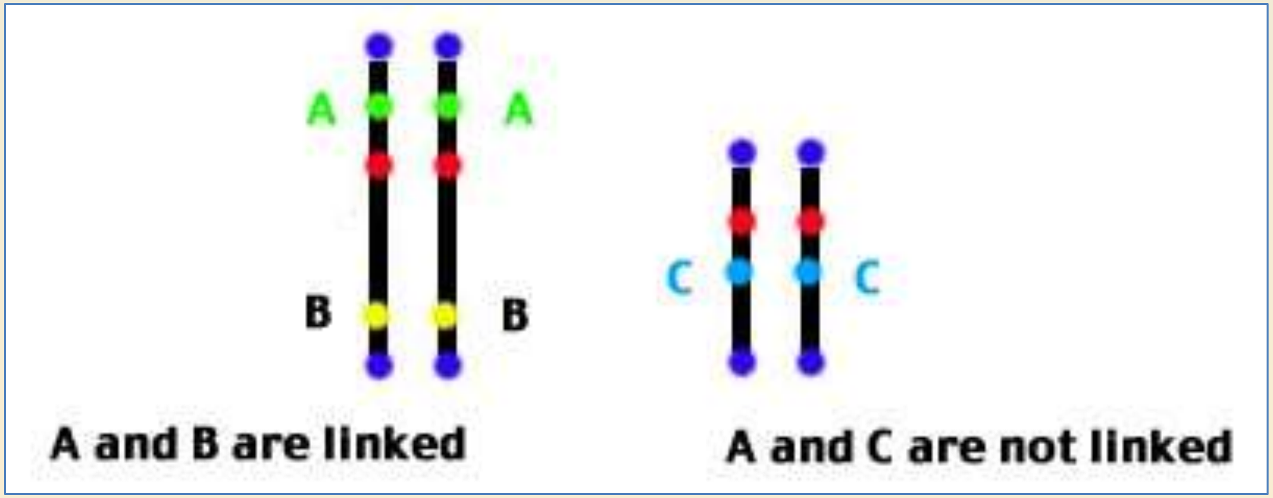


Figure 7.5 The inheritance of seed color in wheat is controlled by three sets of alleles, each of which has an additive effect on the phenotype.

Linked Genes

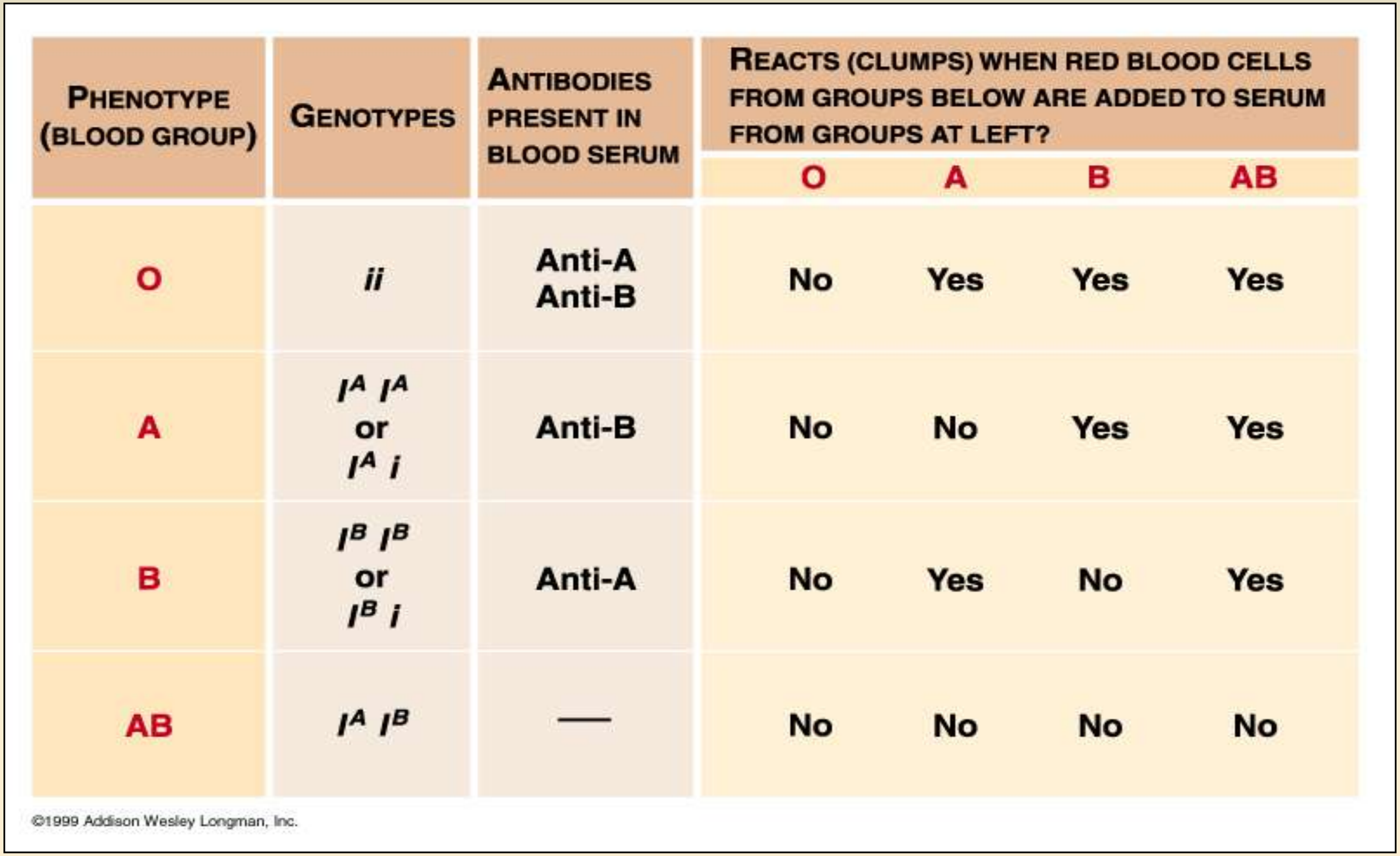
Genes that are close together on the same chromosome are often inherited together, not independently, and will yield a 3:1 ratio instead of 9:3:3:1 .



The only way linked genes will not be inherited together is if crossing over occurs in between them. The closer the loci, the less likely crossing over will occur

Multiple Alleles – more than 2 in the population

e.g. Human Blood types – A, B, O system



PHENOTYPE (BLOOD GROUP)	GENOTYPES	ANTIBODIES PRESENT IN BLOOD SERUM	REACTS (CLUMPS) WHEN RED BLOOD CELLS FROM GROUPS BELOW ARE ADDED TO SERUM FROM GROUPS AT LEFT?			
			O	A	B	AB
O	ii	Anti-A Anti-B	No	Yes	Yes	Yes
A	$I^A I^A$ or $I^A i$	Anti-B	No	No	Yes	Yes
B	$I^B I^B$ or $I^B i$	Anti-A	No	Yes	No	Yes
AB	$I^A I^B$	—	No	No	No	No

Summary of Genetics

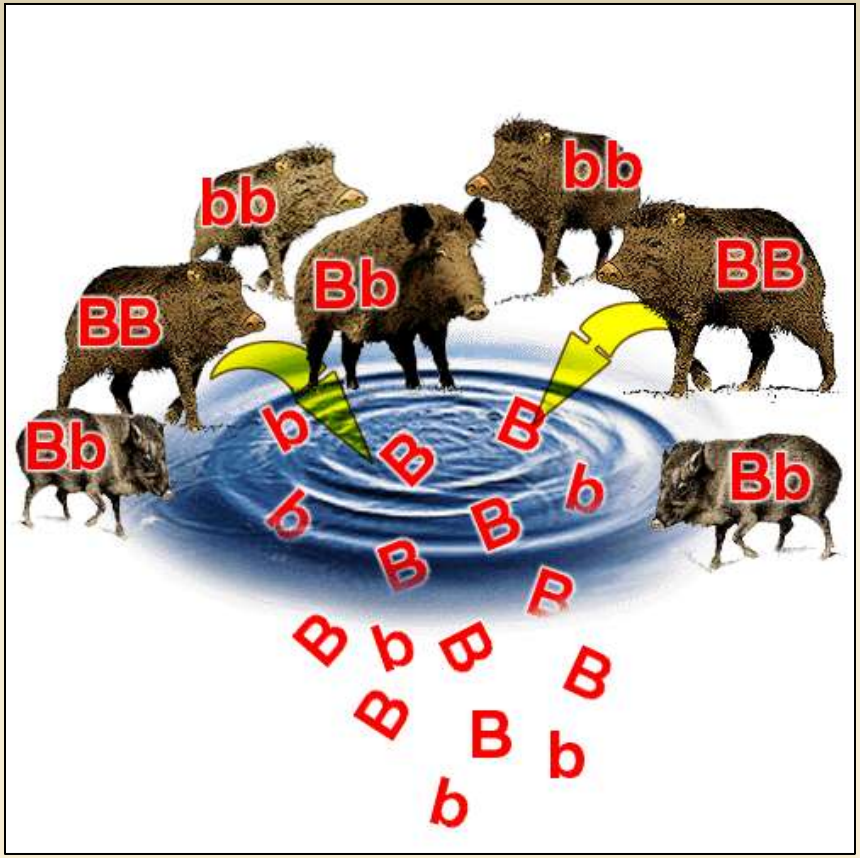
- Chromosomes carry hereditary info (genes)
- Chromosomes (and genes) occur in pairs
- New combinations of genes occur in sexual reproduction
- Monohybrid vs. Dihybrid crosses
- Mendel’s Principles:
  - Dominance: one allele sometimes masks another
  - Segregation: genes become separated in gamete formation
  - Independent Assortment: Members of one gene pair segregate independently from other gene pairs during gamete formation

Gene Pool – an important concept

All of the genes and different alleles in a population, group of interbreeding individuals

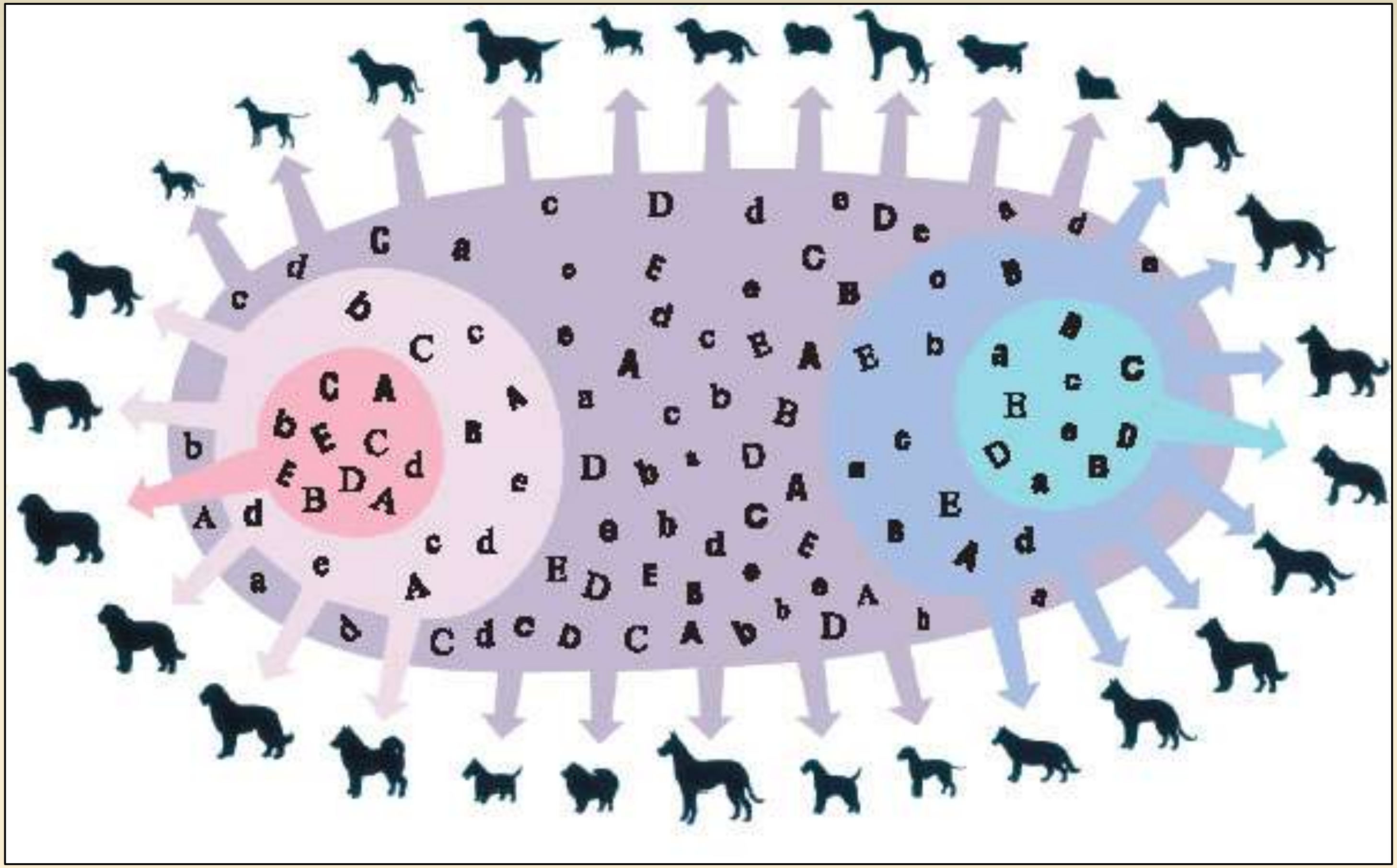
Crop genetic diversity
Wild relatives of crops

The Gene Pool



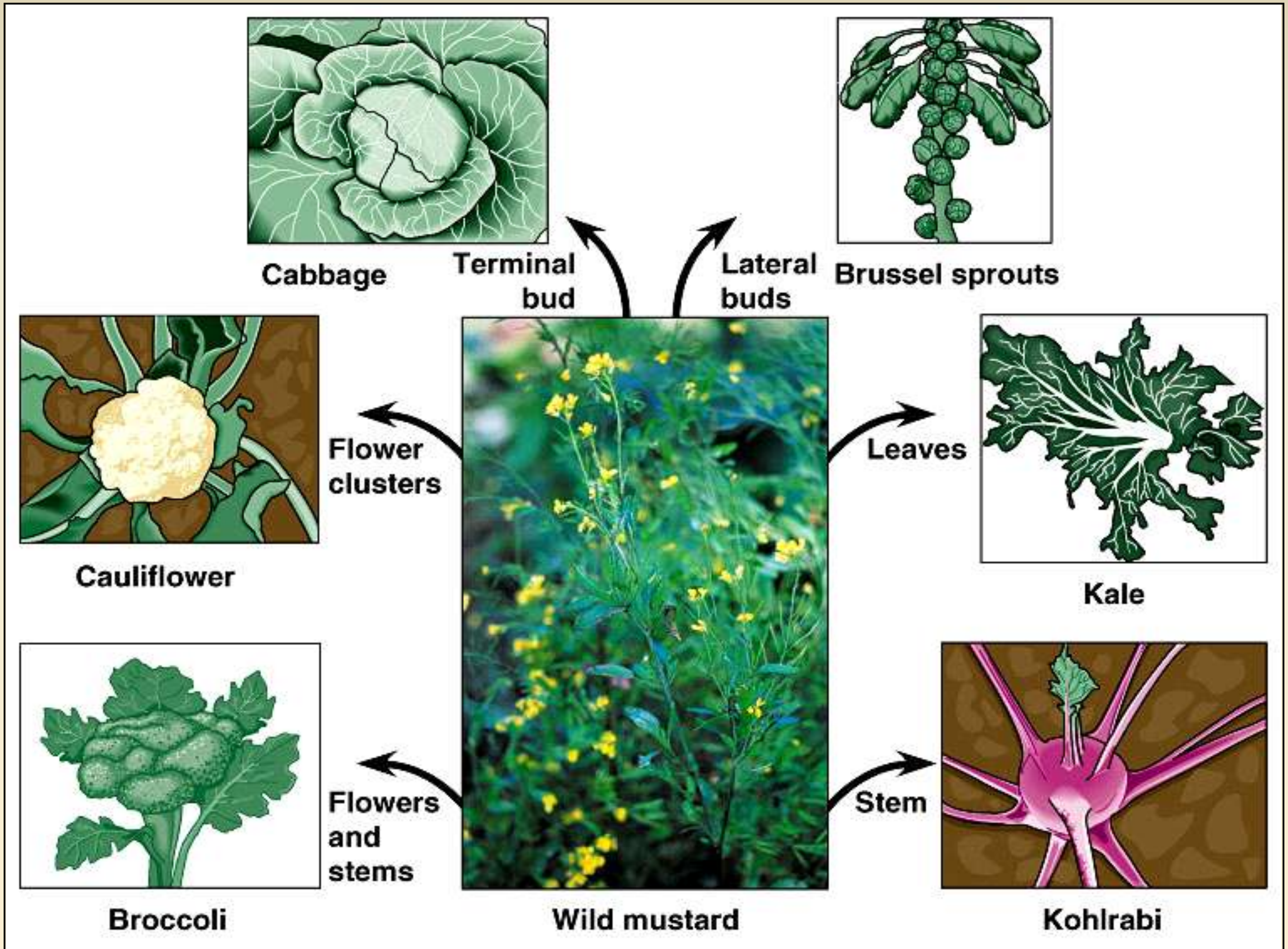
- Individual members of a population contribute their alleles to a common pool of genes.
- The forces of evolution shape and change the composition of this gene pool and thus the nature of the population.

Artificial Selection - Dogs

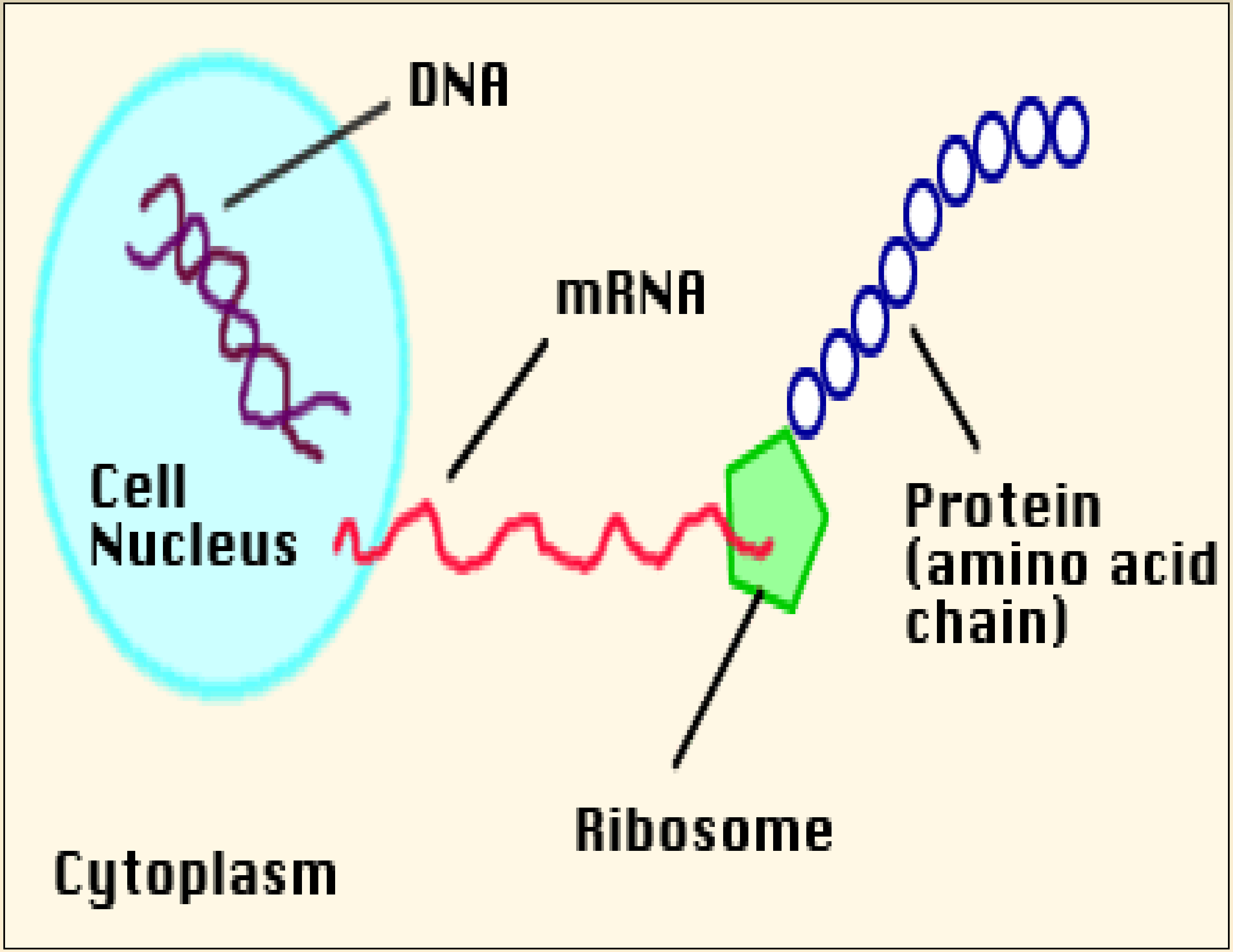


Artificial Selection –

Brassica oleraceae - Cole Crops



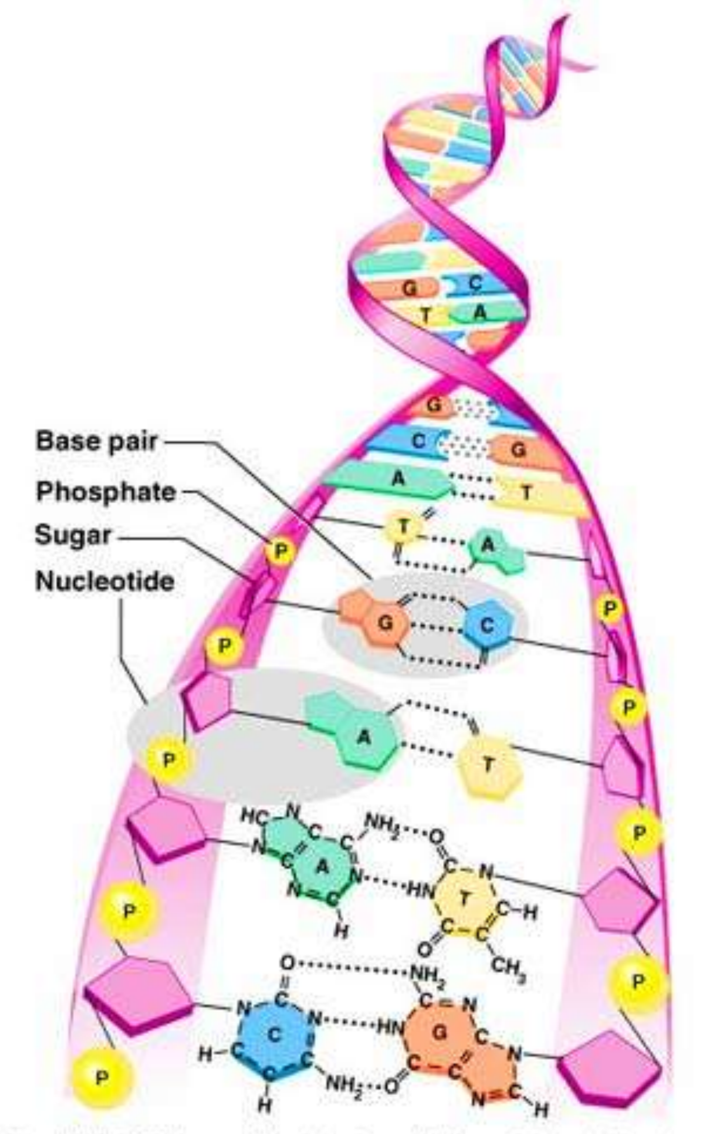
Molecular Genetics



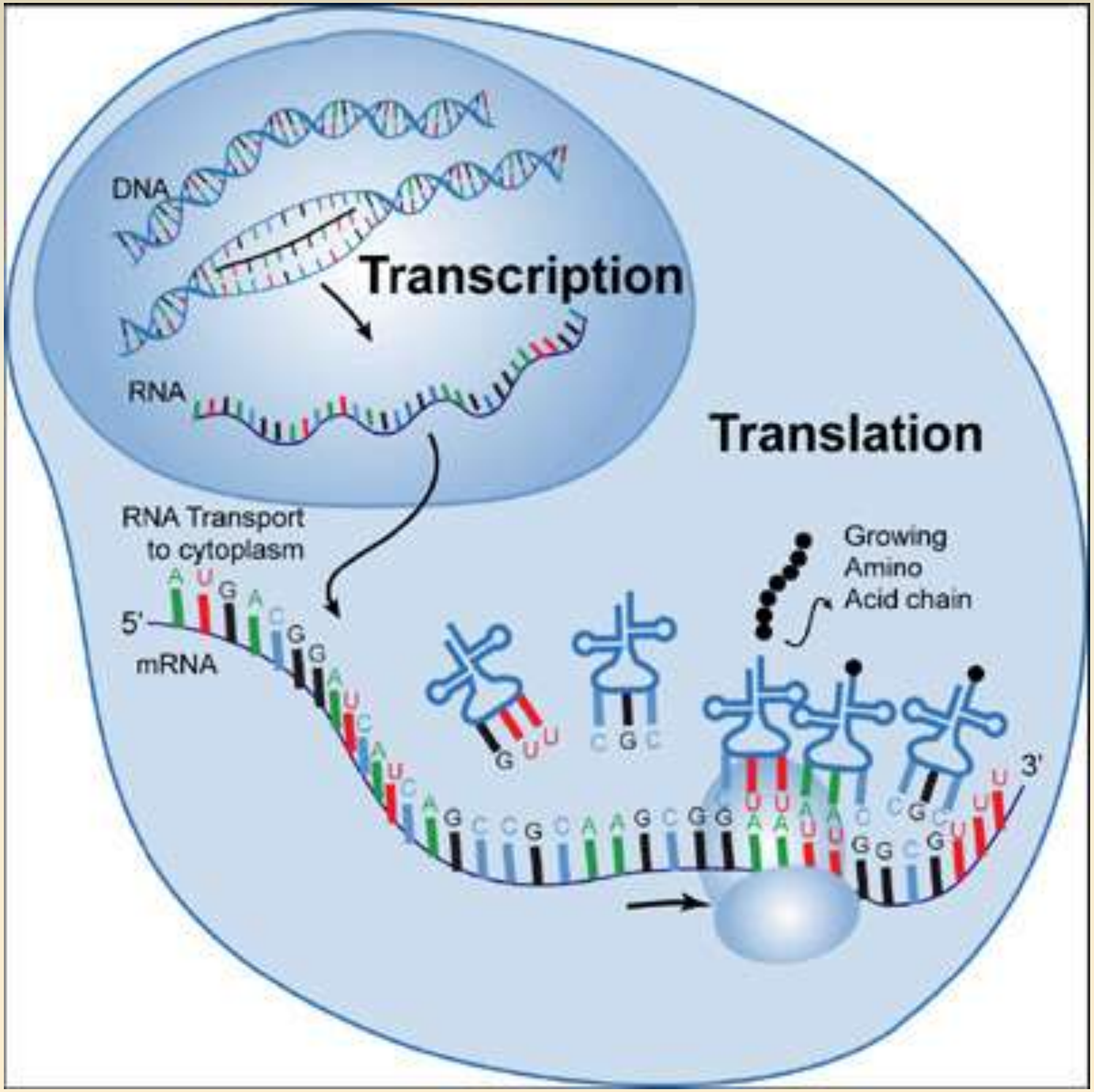
DNA Structure

- Double helix
- Paired strands are linked by bases
    - Adenine ("A")
    - Cytosine ("C")
    - Guanine ("G")
    - Thymine ("T")

  - A must bond with T
  - G must bond with C

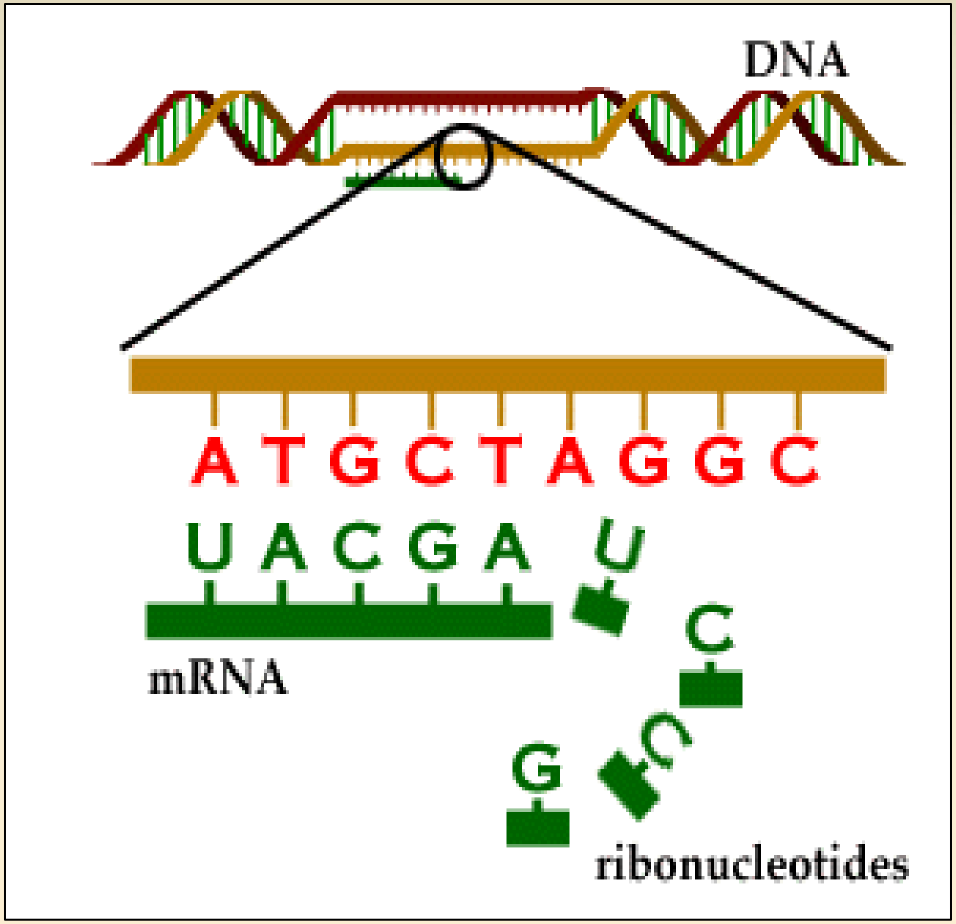


Transcription: DNA => RNA
Translation: RNA => Protein (enzymes)



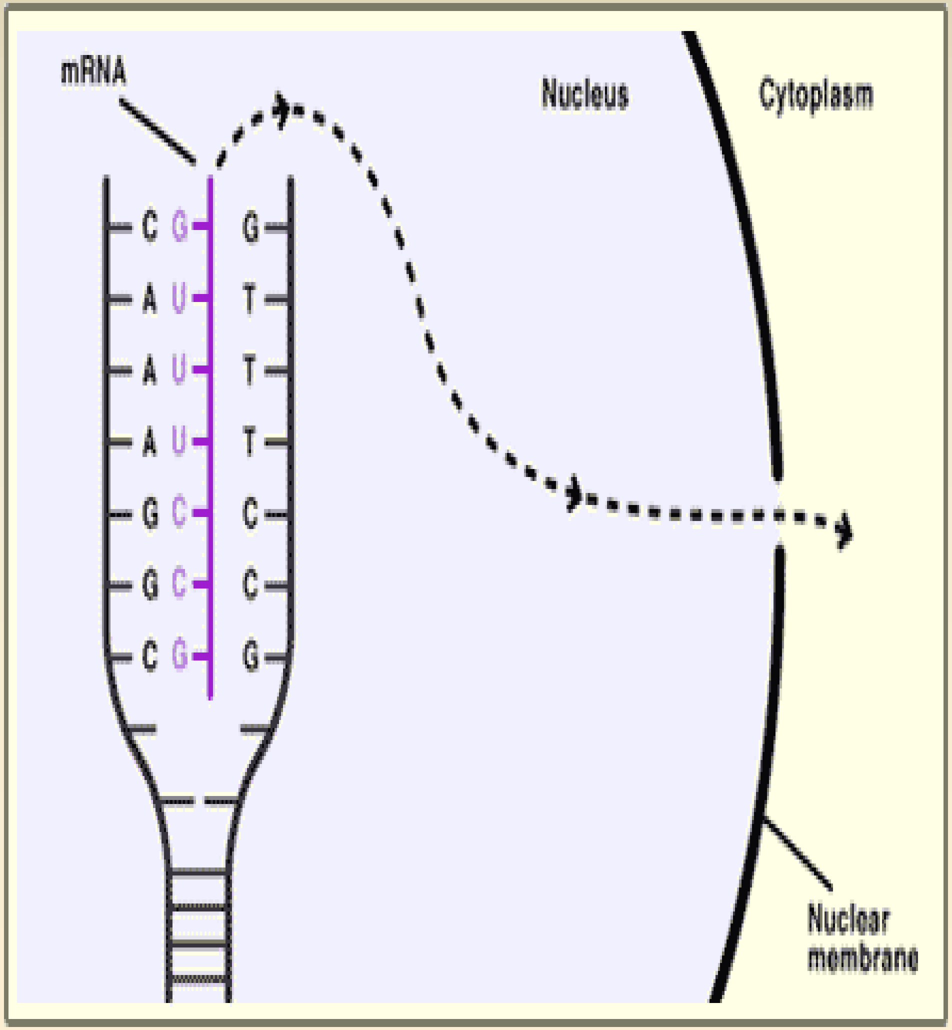
Transcription

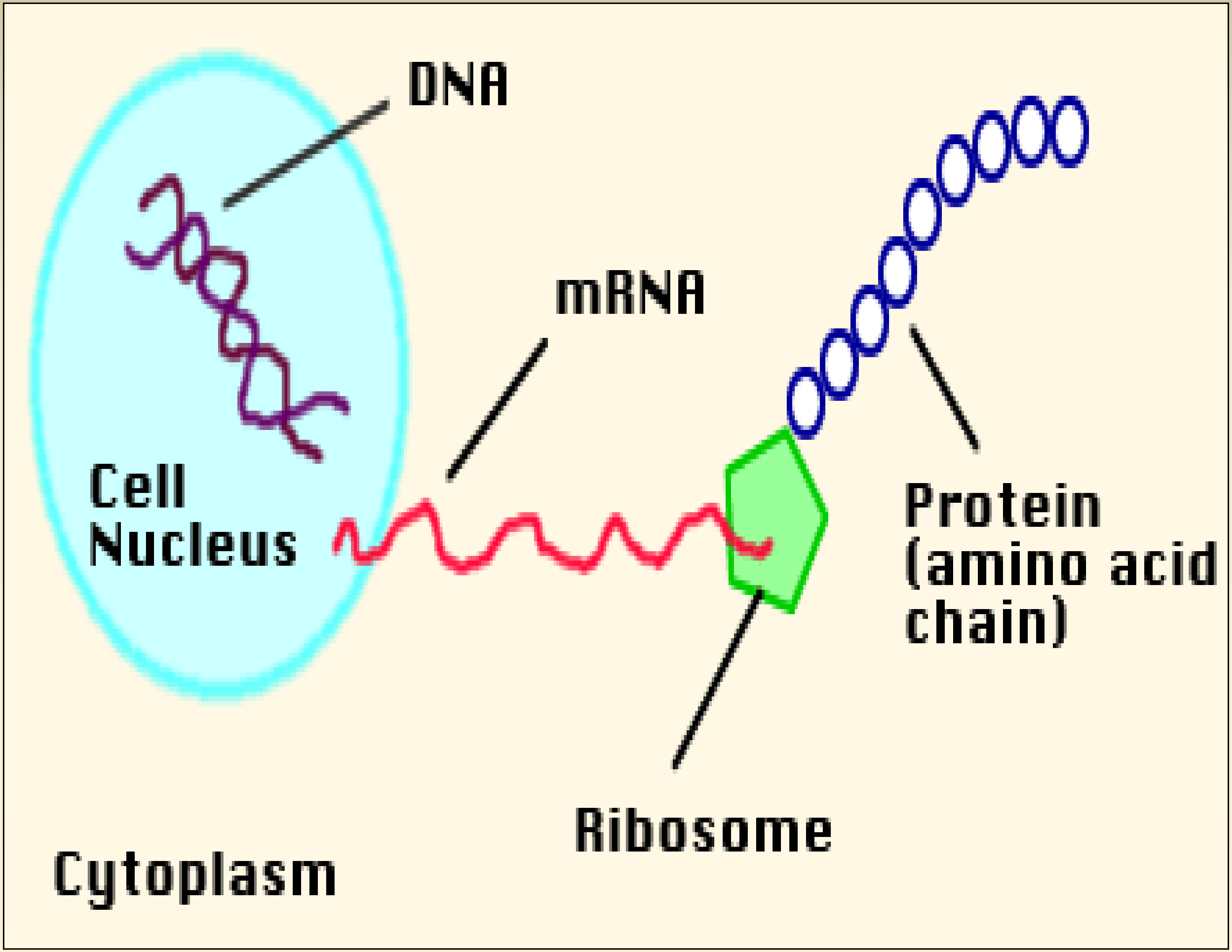
- Transcription- RNA is made from a DNA template in the nucleus.
- This type of RNA is called messenger RNA or **mRNA**



Transcription

- DNA is protected inside the nucleus.
- mRNA carries the message of DNA into the cytoplasm to the ribosome's




DNA
mRNA
Cell Nucleus
Protein (amino acid chain)
Ribosome
Cytoplasm

Translation – Protein Synthesis in the Ribosome

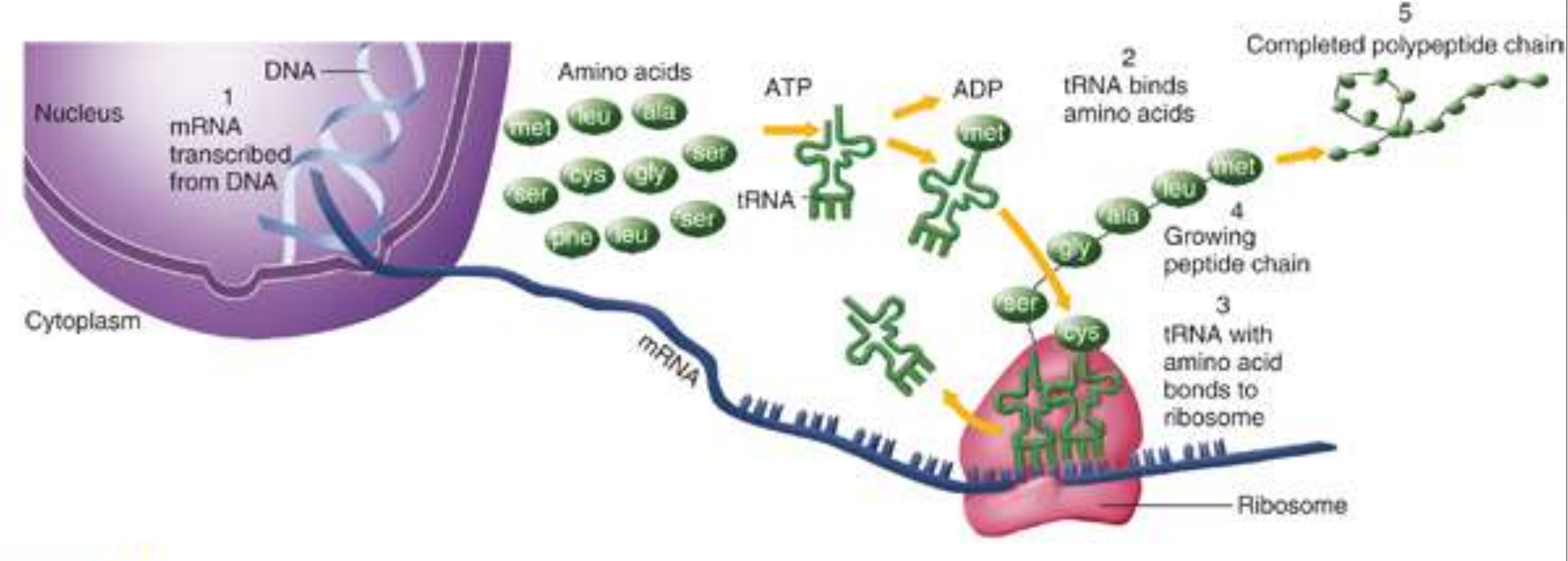
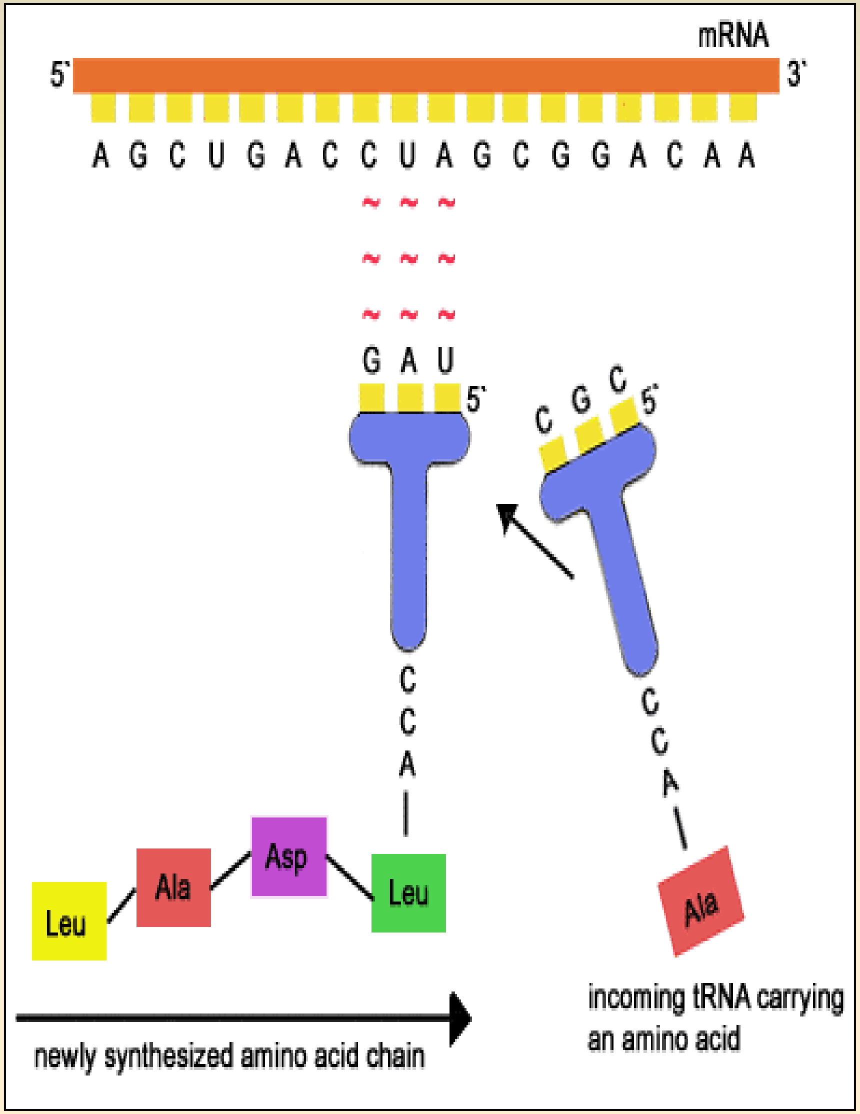


Figure 7.11 Summary of protein synthesis.

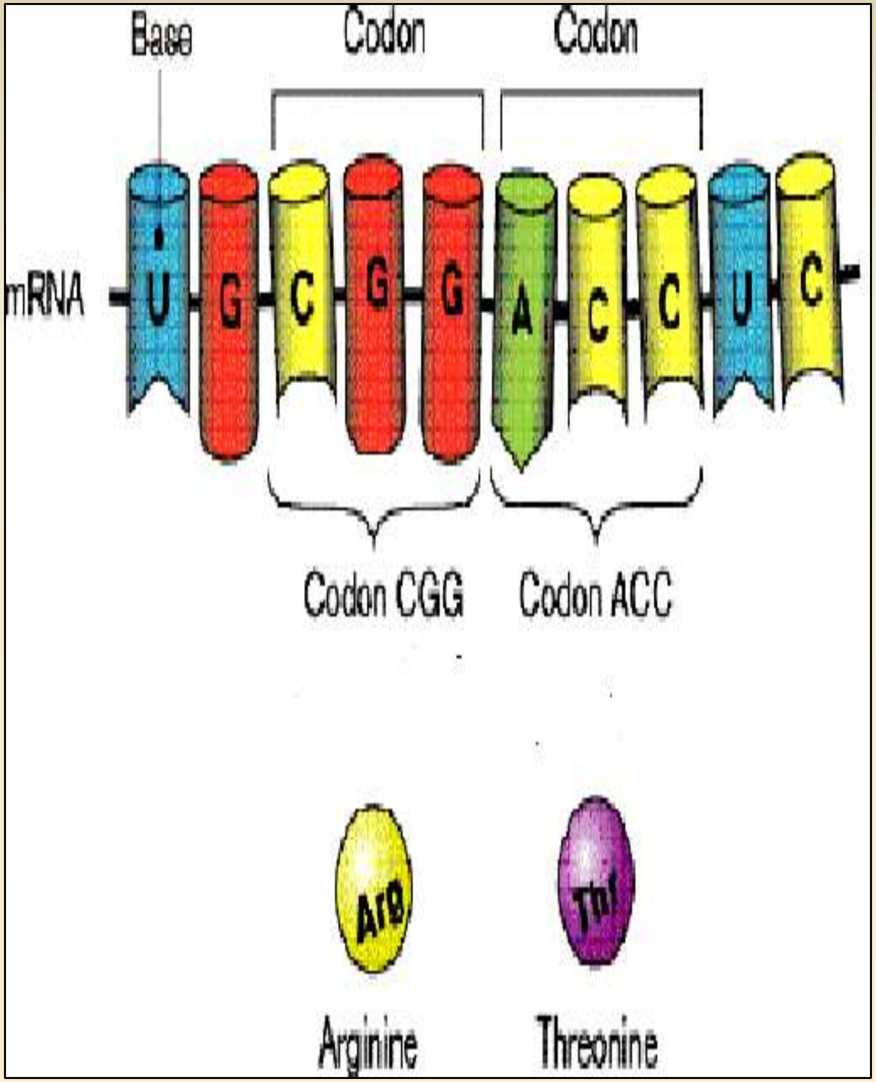
Translation

- Genetic translation converts nucleic acid language into amino acid language.

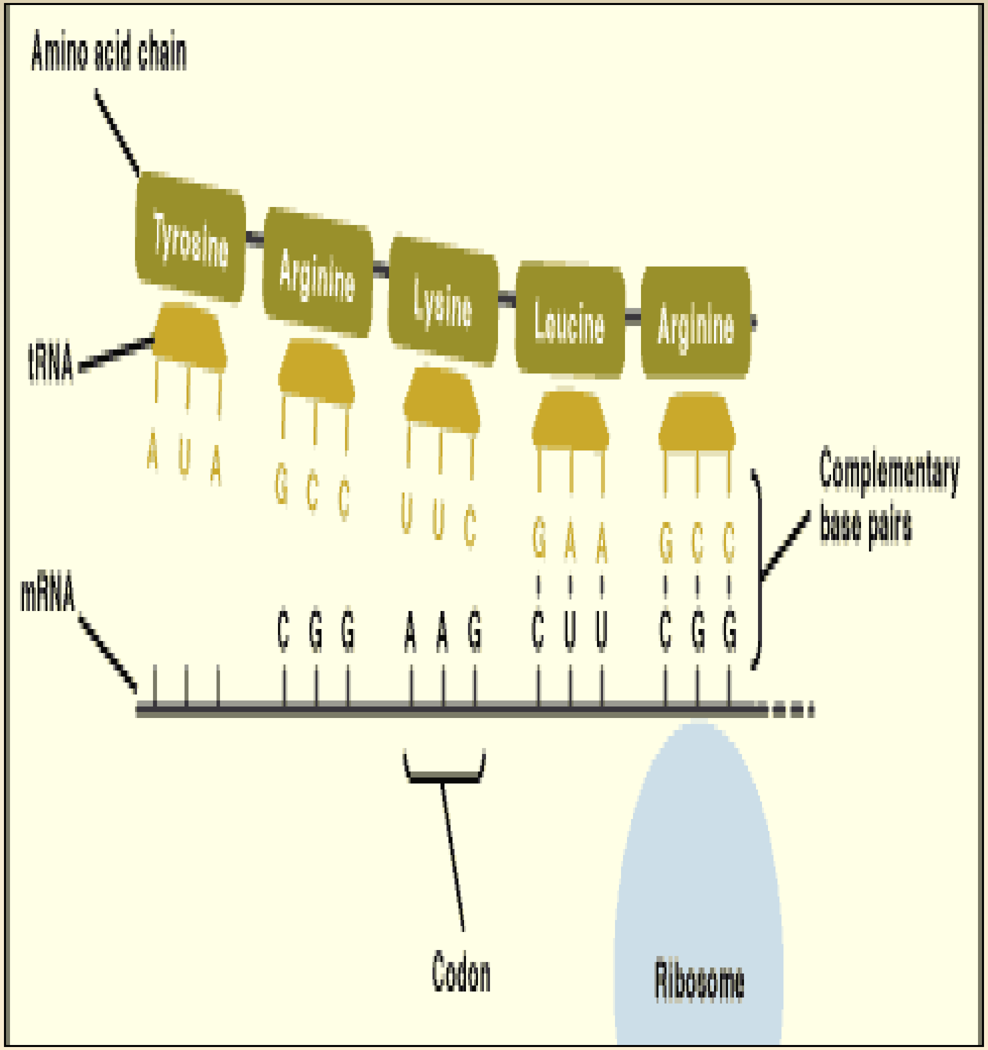


Codon

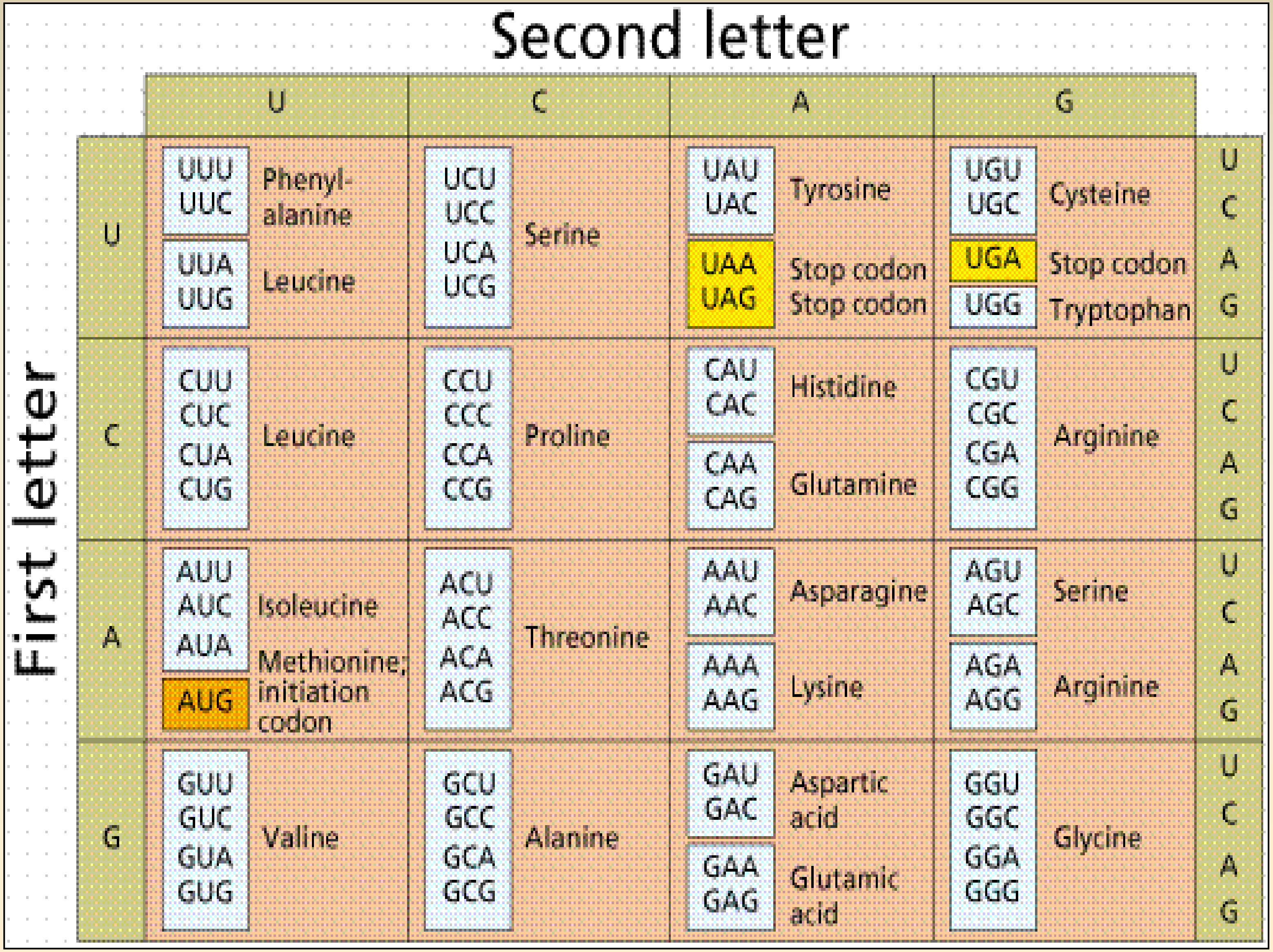
- The flow of information from gene to protein is based on codons.
- A codon is a three-base word that codes for one amino acid



- The flow of information from gene to protein is based on codons.



The Genetic Code – codons coding for specific amino acids in a protein



	Second letter				
First letter	U	C	A	G	
U	UUU UUC Phenylalanine UUA UUG Leucine	UCU UCC UCA UCG Serine	UAU UAC Tyrosine UAA Stop codon UAG Stop codon	UGU UGC Cysteine UGA Stop codon UGG Tryptophan	U C A G
C	CUU CUC CUA CUG Leucine	CCU CCC CCA CCG Proline	CAU CAC Histidine CAA CAG Glutamine	CGU CGC CGA CGG Arginine	U C A G
A	AUU AUC AUA Isoleucine AUG Methionine; initiation codon	ACU ACC ACA ACG Threonine	AAU AAC Asparagine AAA AAG Lysine	AGU AGC Serine AGA AGG Arginine	U C A G
G	GUU GUC GUA GUG Valine	GCU GCC GCA GCG Alanine	GAU GAC Aspartic acid GAA GAG Glutamic acid	GGU GGC GGA GGG Glycine	U C A G

Mutations

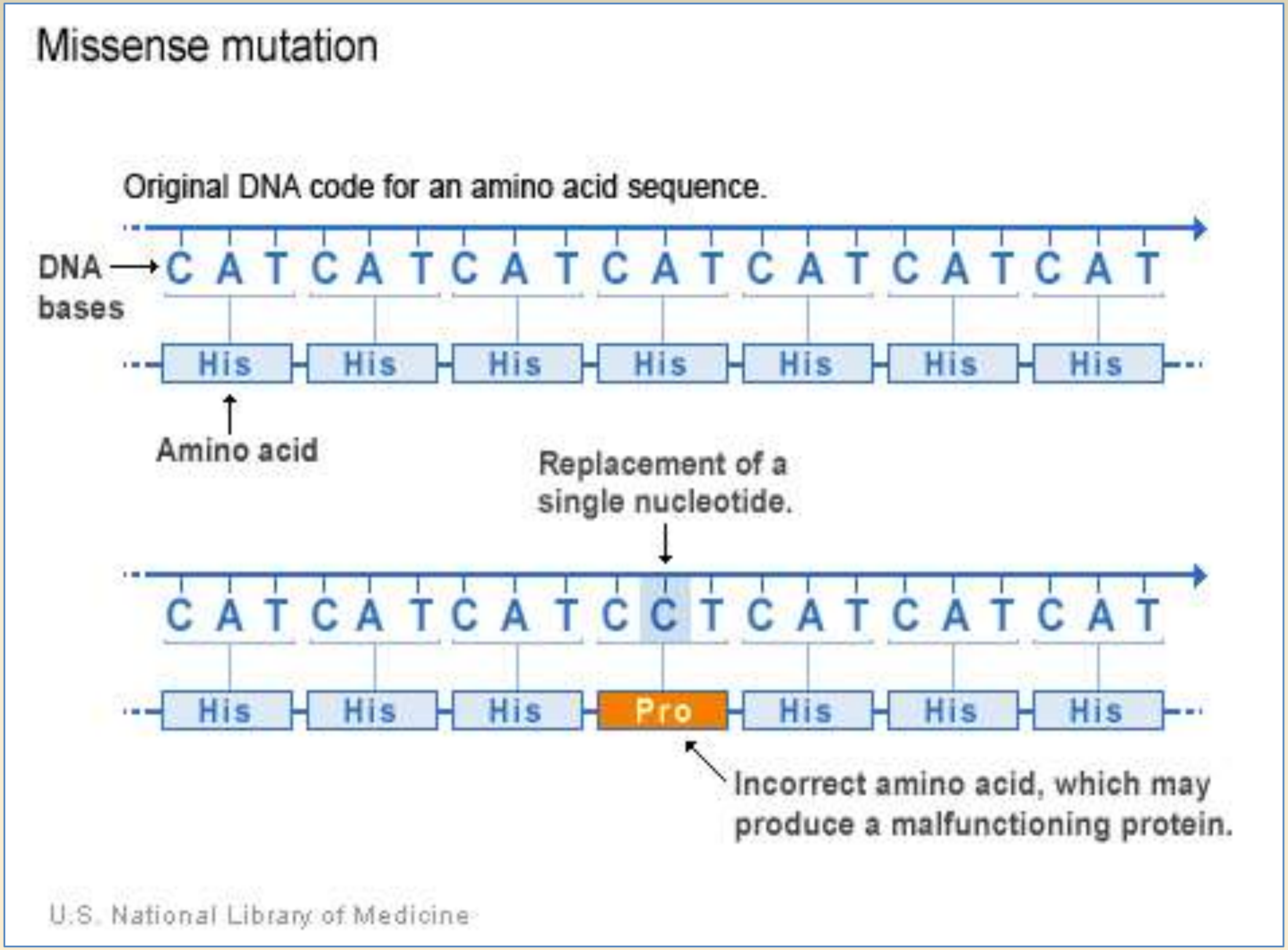
- happens randomly every few thousand cell divisions
- can be increased by exposure to *mutagens* like chemicals or radiation
- can be in somatic cells or in germ cells (gametes)
- can be POSITIVE, NEGATIVE, or NEUTRAL(silent)
- are the "raw material" for evolution by *natural selection*

Mutations – changes in DNA

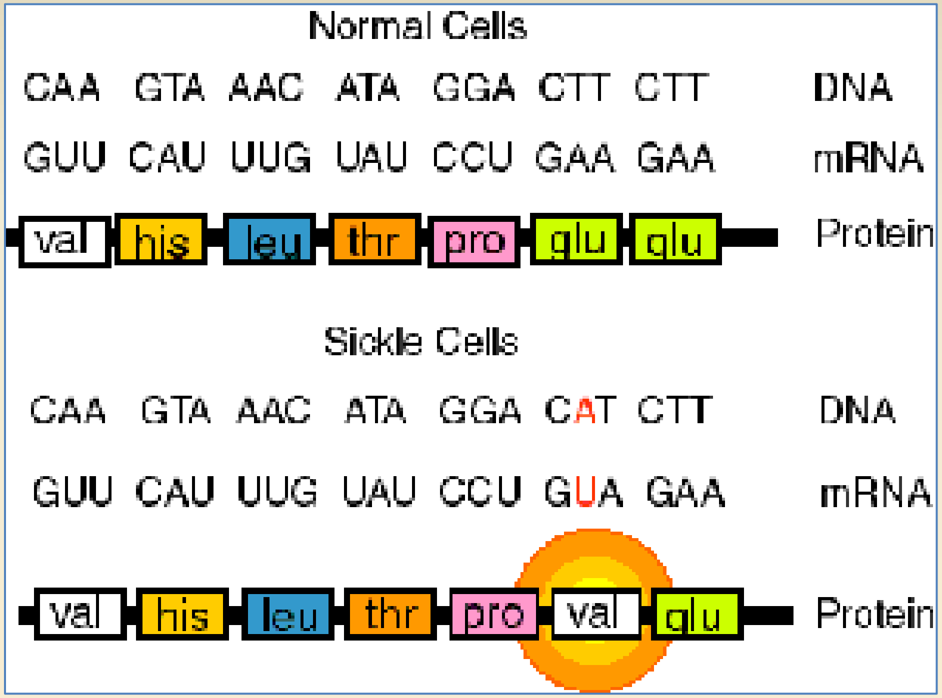
Mutation Types:
Point mutation – error in single base of DNA
Deletion – loss of a DNA segment
Insertion – added piece of DNA
Inversion
Frame-shift – error reading codons, amino acids wrong

Caused by:
Breakage of DNA
Chemicals, Sunlight, UV Radiation
Errors in replication

Important source of variation for new alleles, evolution


Missense mutation
Original DNA code for an amino acid sequence.
DNA bases
C A T C A T C A T C A T C A T C A T C A T
His His His His His His His
Amino acid
Replacement of a single nucleotide.
C A T C A T C A T C C T C A T C A T C A T
His His His Pro His His His
Incorrect amino acid, which may produce a malfunctioning protein.
U.S. National Library of Medicine

Sickle Cell Anemia



Normal Cells

CAA GTA AAC ATA GGA CTT CTT DNA

GUU CAU UUG UAU CCU GAA GAA mRNA

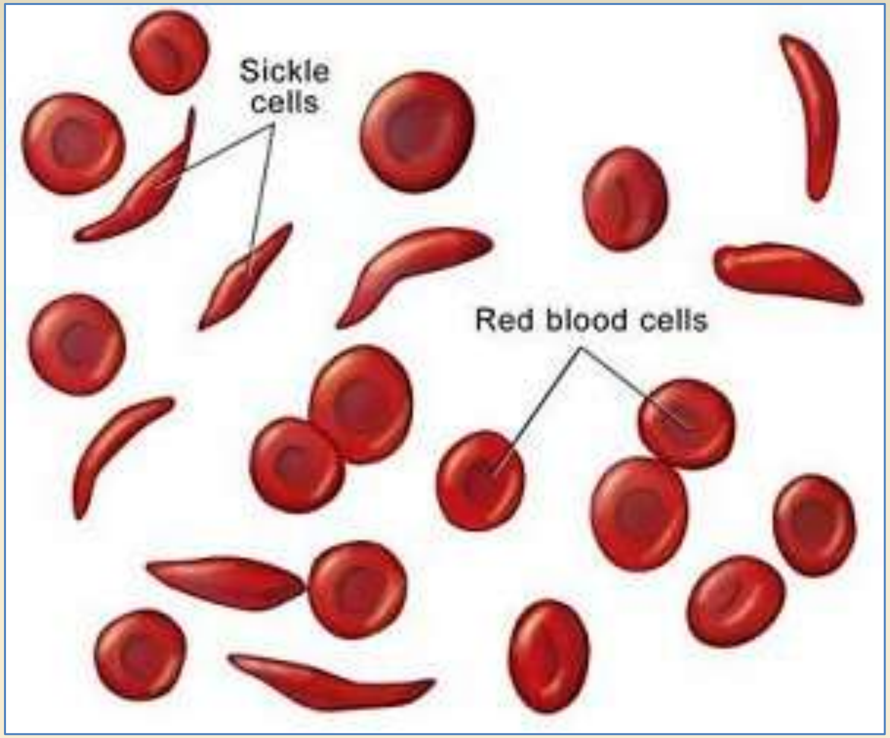
val his leu thr pro glu glu Protein

Sickle Cells

CAA GTA AAC ATA GGA CAT CTT DNA

GUU CAU UUG UAU CCU GUA GAA mRNA

val his leu thr pro val glu Protein



Swapping an A for a T in the gene for hemoglobin results in the insertion of valine instead of glutamine in the protein molecule causing the disease sickle cell anemia

Genetic Engineering - moving genes from one organism to another

Restriction Enzymes – used to cut and splice DNA, cut at specific sites in the DNA

Bacterial Plasmids – used as vectors to transfer DNA

Recombinant DNA – contains DNA from two organisms

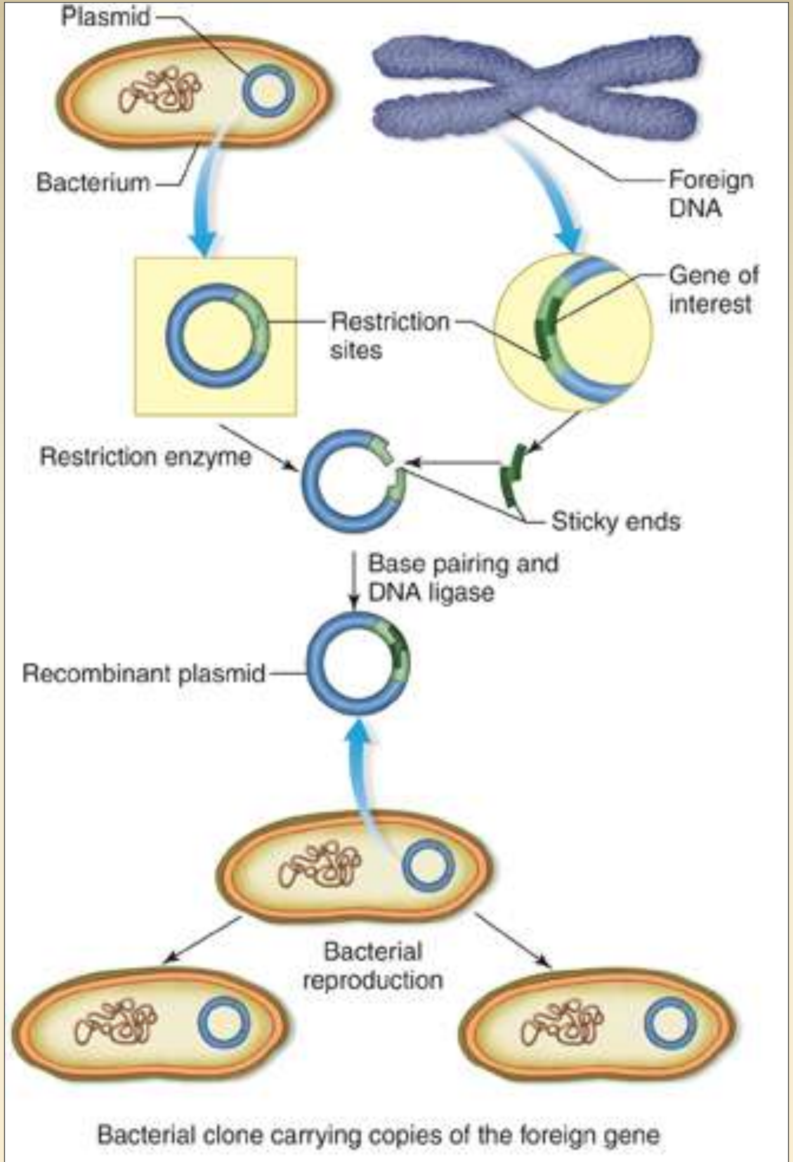


Figure 7.12 Recombinant DNA technology. A gene of interest from one organism is cut from the DNA molecule using a restriction enzyme. The same enzyme is used to cut a bacterial plasmid. The two DNA samples are mixed together and the DNA sealed with the enzyme DNA ligase. The recombinant plasmid is taken up by the bacterial cell, reproducing when the cell divides and expressing the foreign gene.

Genetically Modified Organisms (GMO) - refers to food crops that have been altered using a variety of molecular biology techniques in order to provide them with either new or enhanced characteristics.

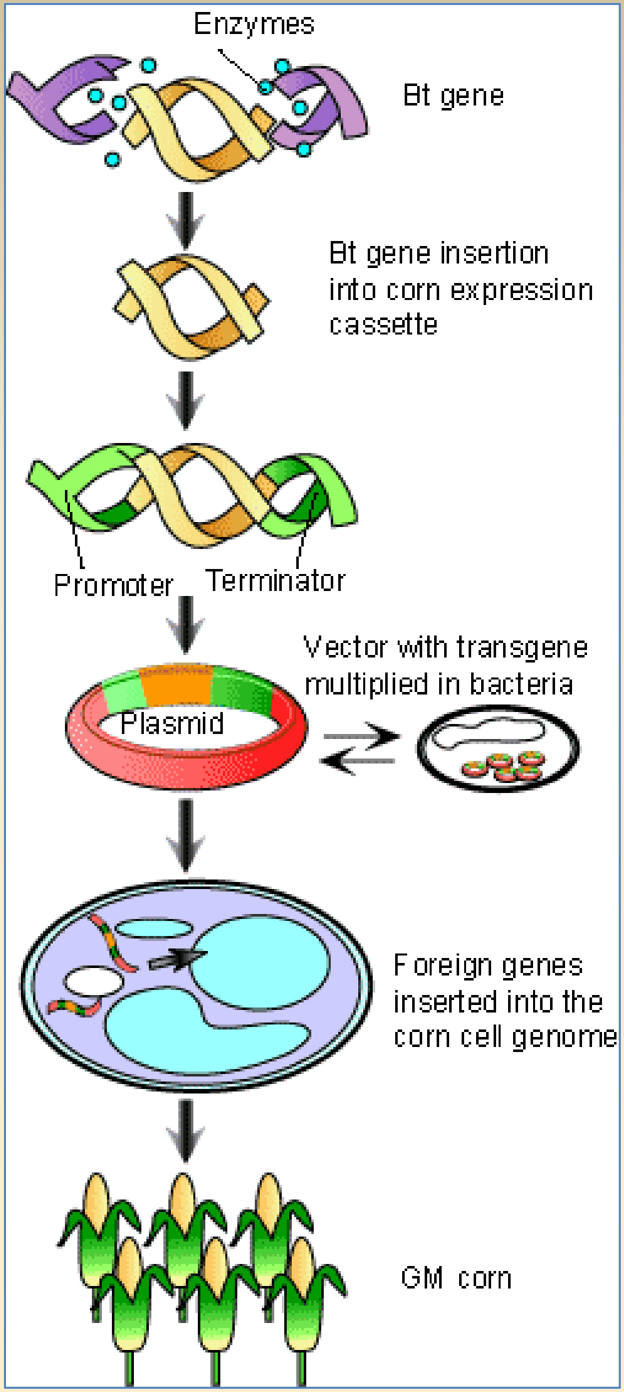
Examples:

- herbicide tolerance,
- pesticide resistance,
- greater nutritional content
- increased tolerance of cold temperatures

GMOs can also be referred to as <u>transgenic organisms</u>

Transferring the gene
Taking a gene from one organism and inserting it into another is essentially a process of cutting the gene which codes for the trait of interest from the foreign organism and pasting this gene into the genome of the organism that you want to alter.

Bacillus thuringiensis (Bt) gene from bacteria produces pesticide, transferred to corn genome.

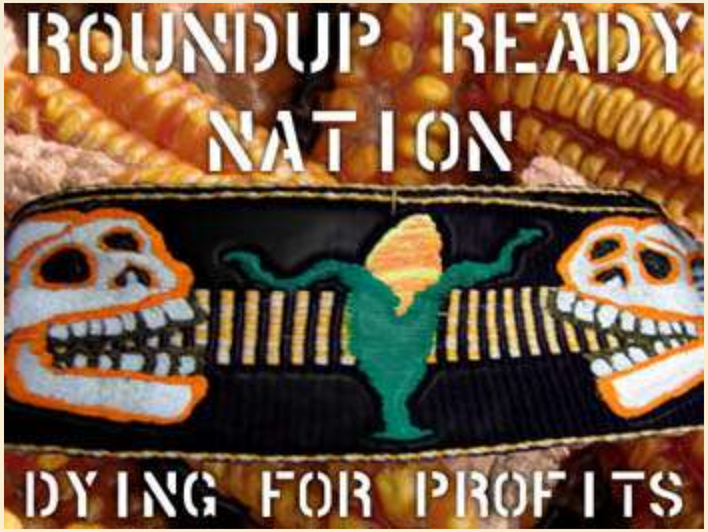


GMOs - Roundup Ready

Monsanto has created strains of soybeans, corn, canola and cotton that are resistant to the weed-killer Roundup®. The weed-killer can be sprayed over the entire crop, killing all plants except the transgenic crop intended to be grown.

Concerns

Ethics? Must buy license.
Environmental Concerns?
Bees, Pollen?
Super Weeds, Insects?
Human toxicity, allergies?
Labeling food?



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