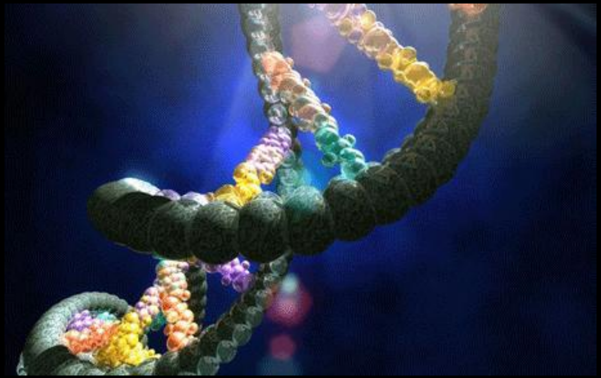
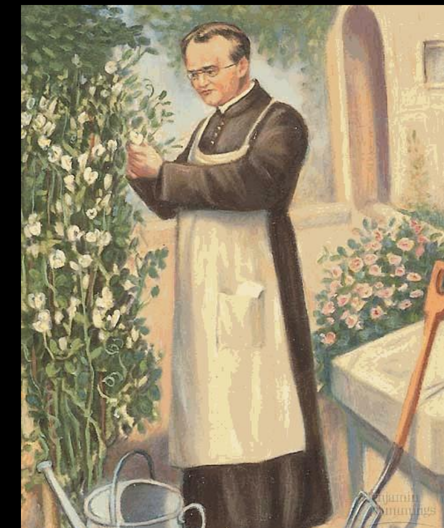
Introduction to Genetics

The Work of Gregor Mendel

B.1.21, B.1.22, B.1.29



Genetic Inheritance

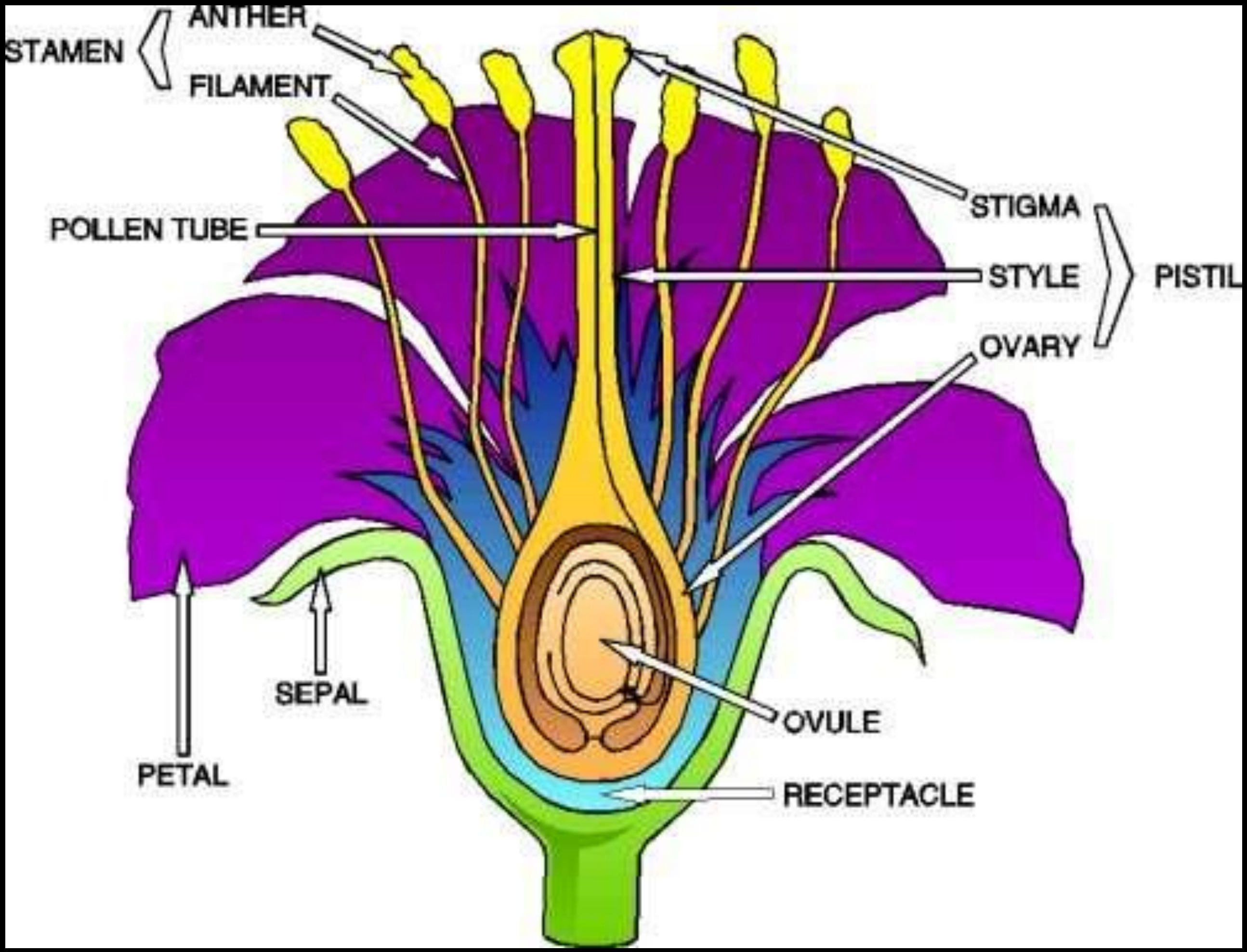


- **Heredity:** the transmission of characteristics from parent to offspring
- The study of heredity in biology is called **“Genetics”**
- **Gregor Mendel:** Austrian monk who studied science and mathematics in Vienna in the mid 1800’s. He was also in charge of the monastery garden. Began the new science of genetics with his study of inheritance in pea plants

Genetic Inheritance

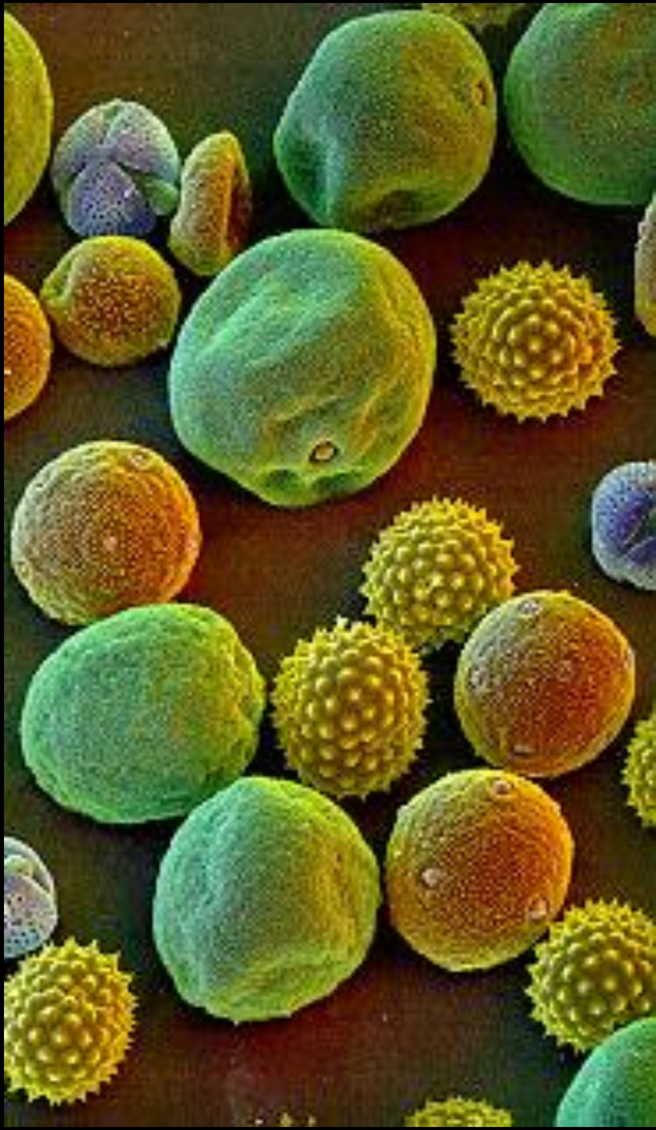
- **Fertilization: The process in sexual reproduction in which male (sperm) and female (egg) reproductive cells join to form a new cell.**
- **The Stamen is the male reproductive part of plants that produce pollen which holds the sperm cells. The Pistil contains the female reproductive organs (ovary, egg cell).**

Genetic Inheritance



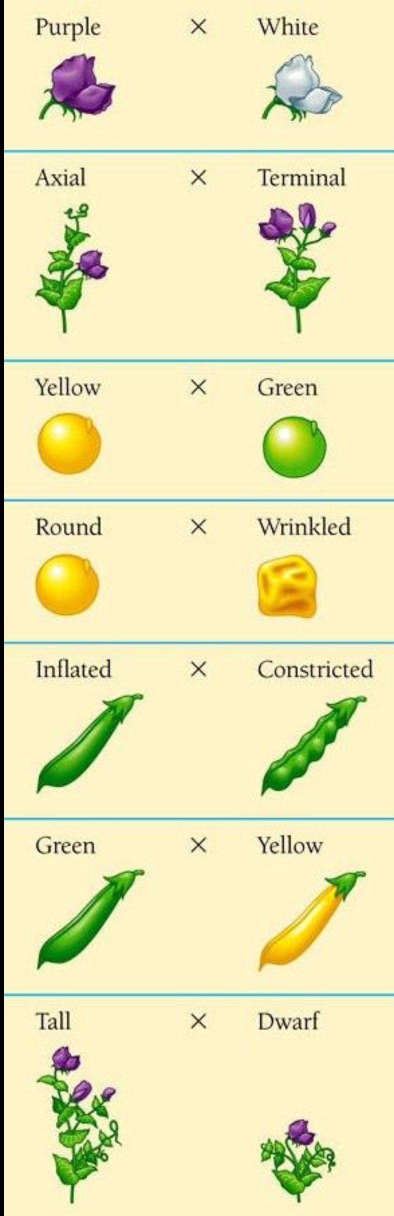
Genetic Inheritance

- **Pollination** is the fertilization of a plant.
- **Self-pollination** can occur when a plant fertilizes itself. Plants can also be fertilized by another plant.
- **True-Breeding:** Term used to describe an organism that produces offspring identical to itself if allowed to self-fertilize



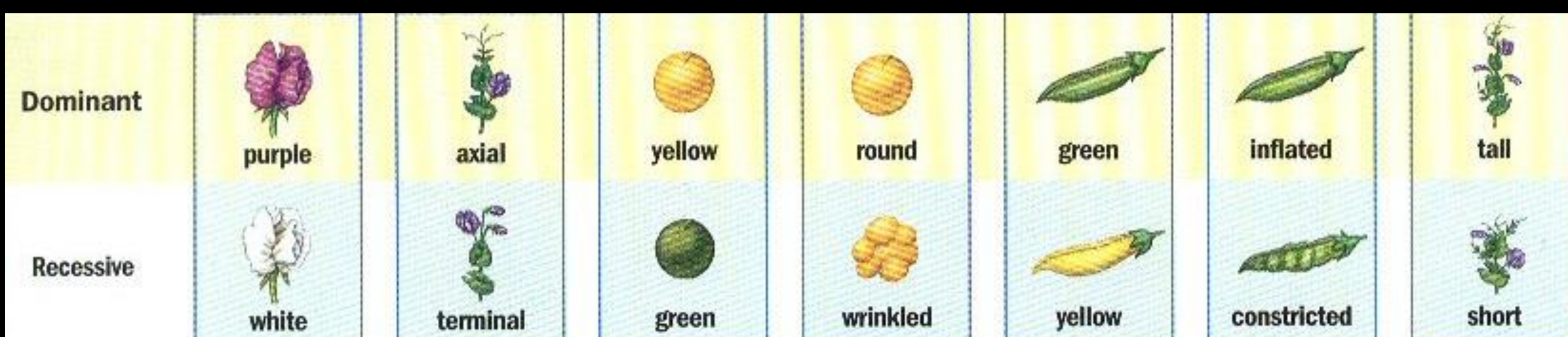
Genetic Inheritance

- True-Breeding plants were the basis of Mendel's experiments.
- He could control what traits he wanted to study by selecting plants that only produce specific traits. (tall, short, flower color)
- By removing the stamens from flowers, he could prevent self-pollination



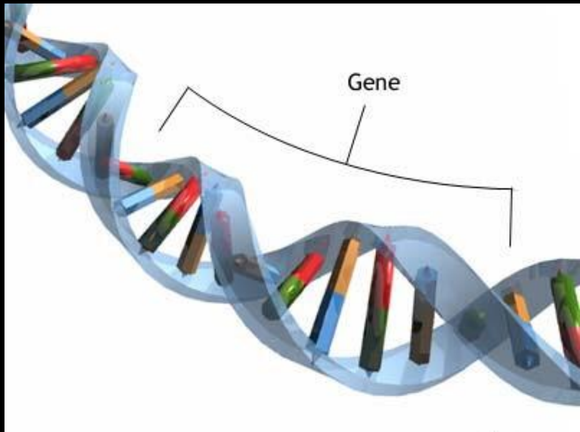
Genes and Dominance

- **Trait:** A trait is a specific characteristic that varies from one individual to another. (seed color, height)
- The traits Mendel studied had only <u>two</u> contrasting characters. (either short or tall, green or yellow seed)



Genes and Dominance

- Genes – Sequence of DNA that determines a trait
- Allele – Different forms of a gene.
  ex. allele for purple flower and allele for white flower: both determine flower color
- Alleles can either be Dominant or Recessive.



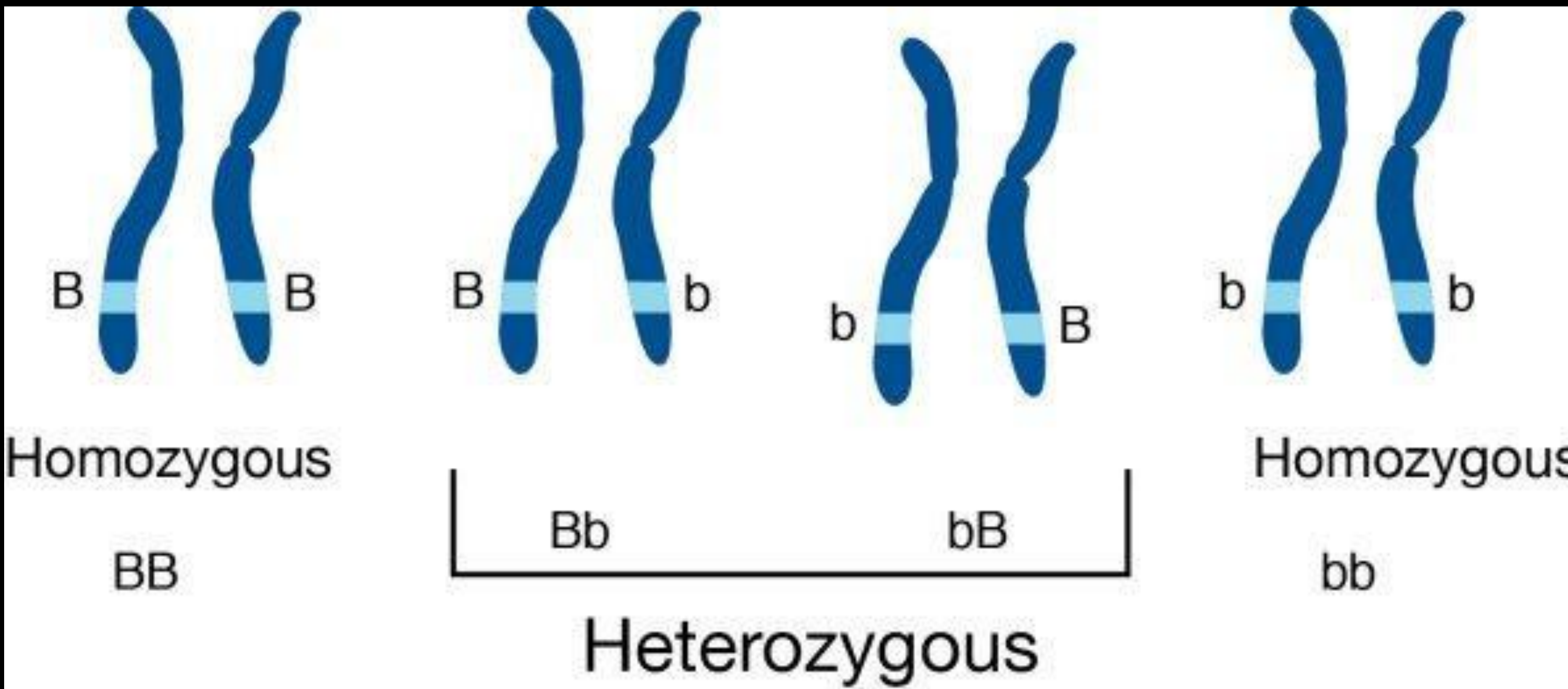
Genes and Dominance

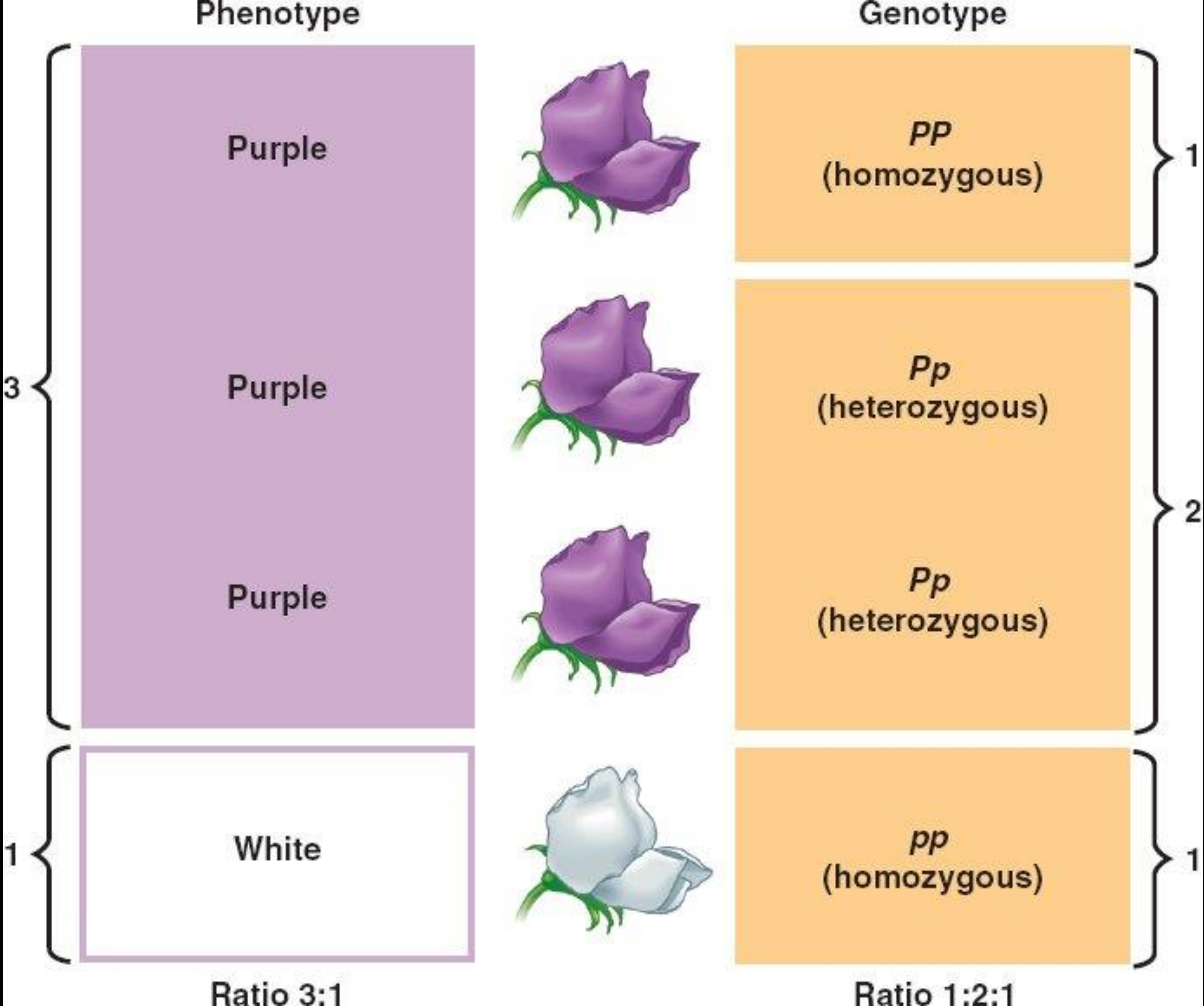
- Dominant Allele – Codes for a trait that is always seen in an organism (Capitol letter)
- Recessive Allele – Codes for a trait that is masked by the other trait (lower case)
- Ex.

Seed Color	Plant Height
Yellow = Y	Tall = T
Green = y	Short = t

Homozygous Vs. Heterozygous

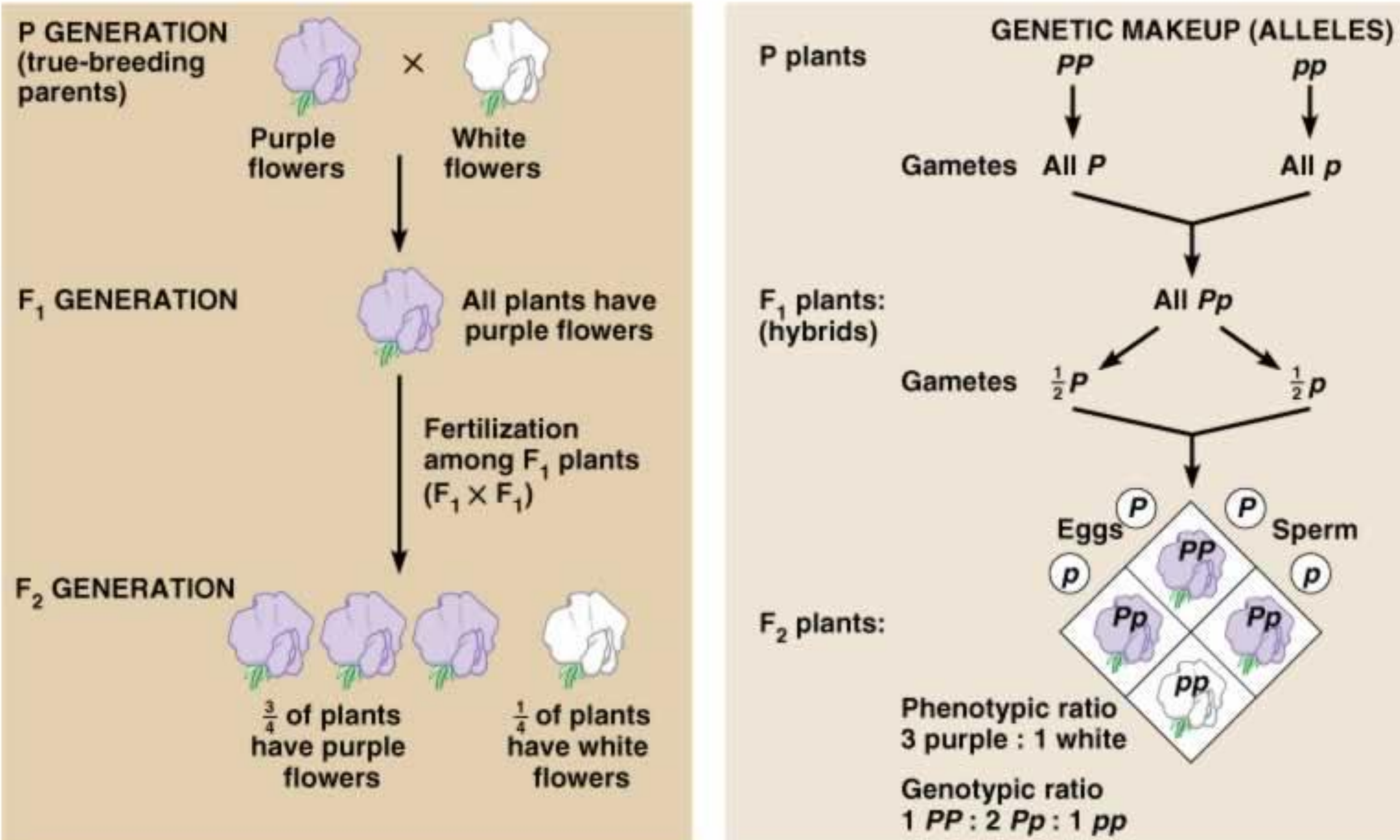
- Homozygous organisms are true-breeding for a particular trait
- Heterozygous organisms are hybrid for a particular trait
- **Genotype** = The genetic makeup of an organism (ex. Tt, PP, yy)
- **Phenotype** = The observable or physical characteristics of the organism
(ex. Tall, purple flower, green seeds)


B
B
B
b
b
B
b
b
Homozygous
BB
Bb
bB
Heterozygous
Homozygous
bb


Phenotype
Genotype
Purple
PP
(homozygous)
1
3
Purple
Pp
(heterozygous)
2
Purple
Pp
(heterozygous)
1
White
pp
(homozygous)
1
Ratio 3:1
Ratio 1:2:1

Genes and Dominance

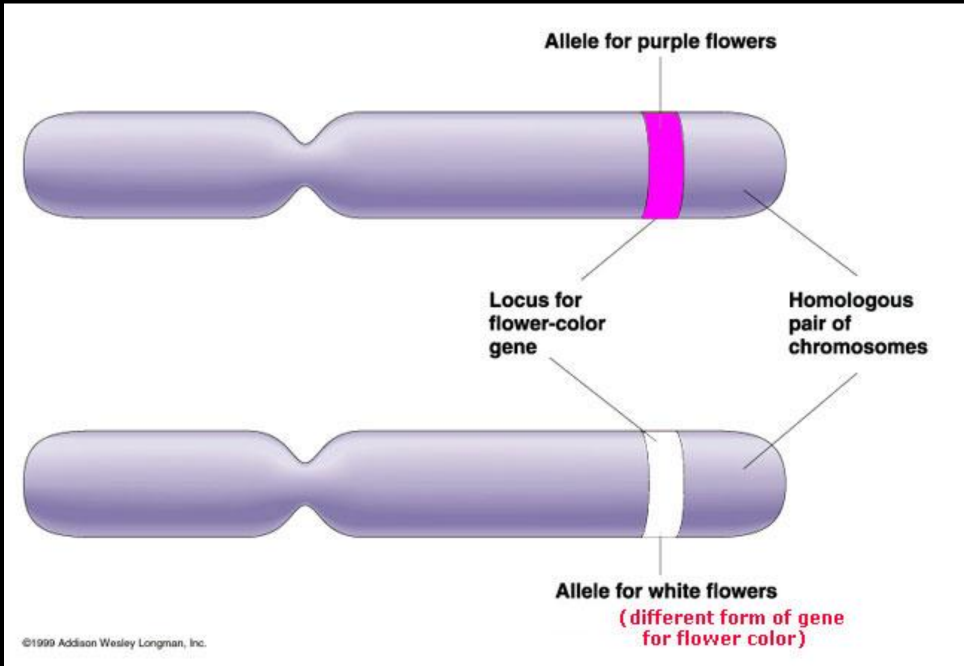
- **P -** Parental generation (The cross of the original pair of plants)
- **F_1 -** Offspring of the parental cross (stands for “first filial)
- **F_2 -** Offspring produced from crossing the F_1 generation.
- Each offspring receives one factor from each parent for every trait



P GENERATION
(true-breeding
parents)
×
Purple
flowers
White
flowers
F1 GENERATION
All plants have
purple flowers
Fertilization
among F1 plants
(F1 × F1)
F2 GENERATION
3/4 of plants
have purple
flowers
1/4 of plants
have white
flowers
GENETIC MAKEUP (ALLELES)
P plants
PP
pp
Gametes
All P
All p
F1 plants:
(hybrids)
All Pp
Gametes
1/2 P
1/2 p
Eggs
Sperm
P
P
p
p
PP
Pp
Pp
pp
F2 plants:
Phenotypic ratio
3 purple : 1 white
Genotypic ratio
1 PP : 2 Pp : 1 pp

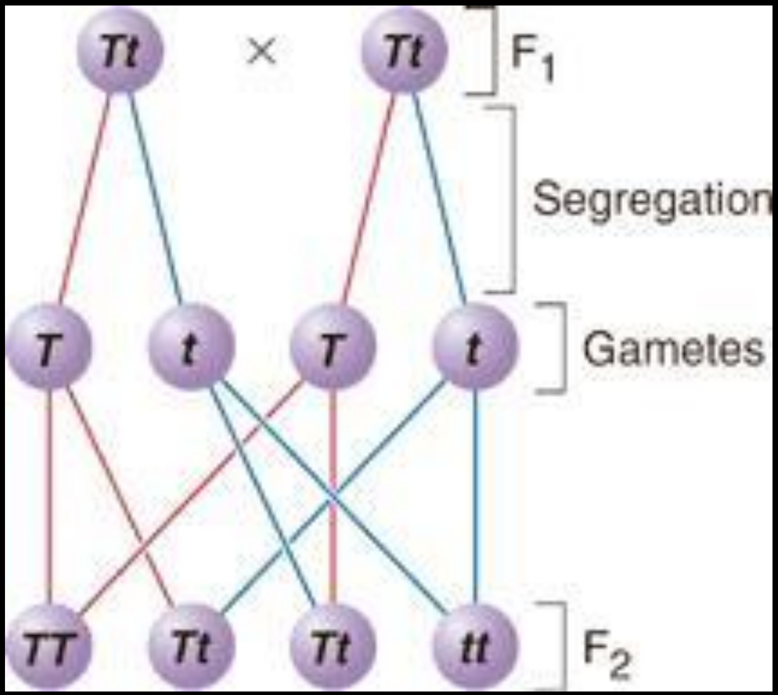
Segregation of Alleles

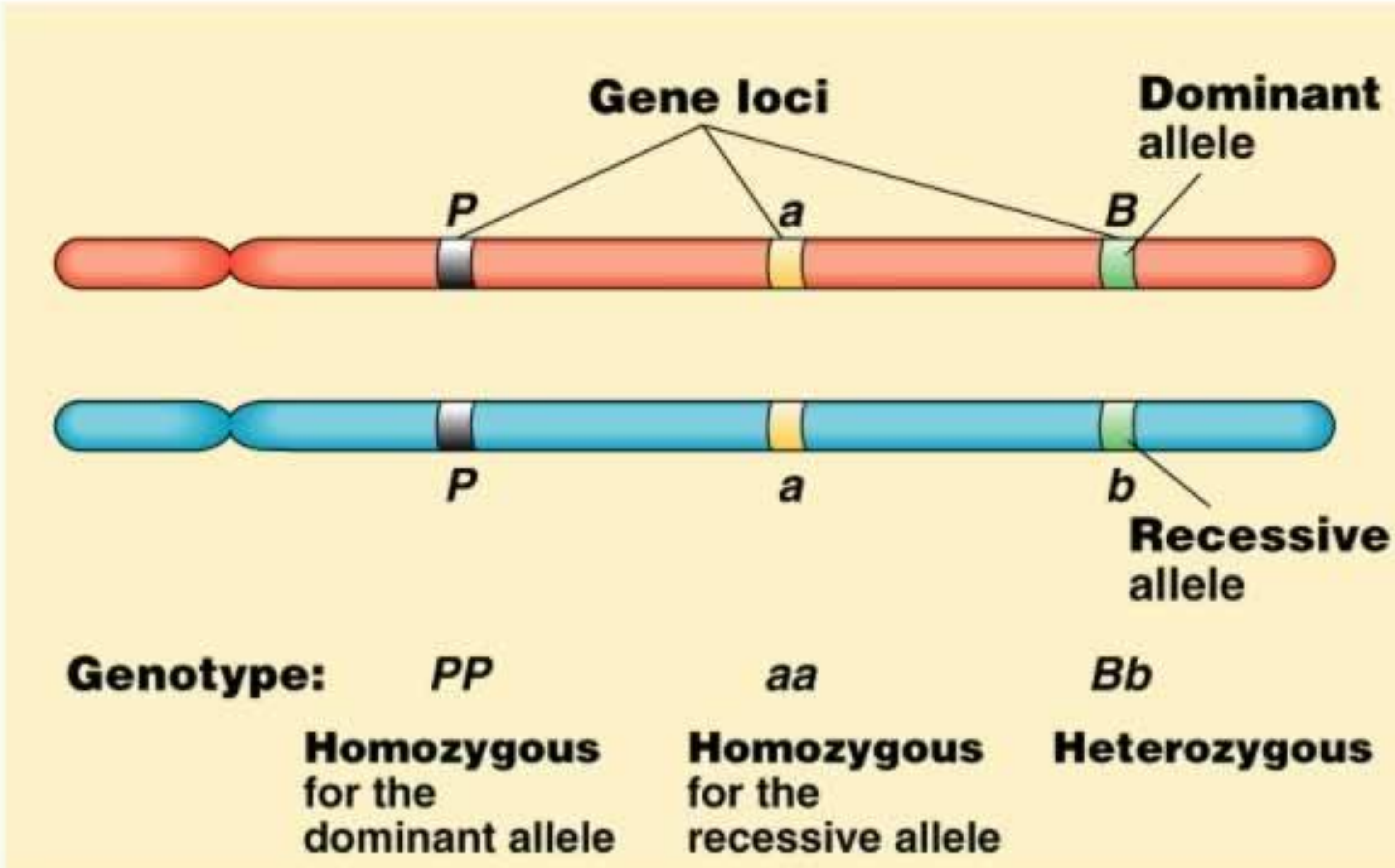
- **Segregation –** The separation of alleles during the formation of gametes.
- **Gametes –** Specialized cell involved in sexual reproduction.
  Ex. sperm and egg



Segregation of Alleles

- **Mendel's Law of Segregation:** When an organism produces gametes, the copies of a gene separate so that each gamete receives only one copy.

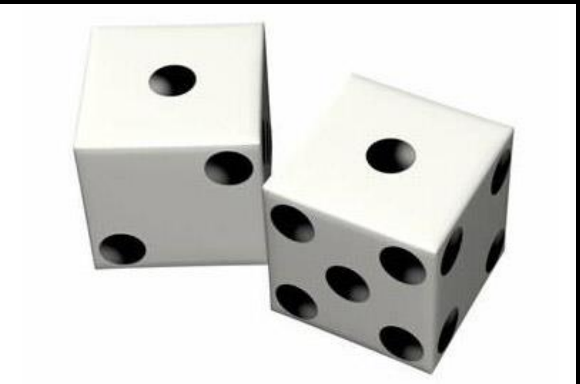




Gene loci
Dominant allele
P
a
B
P
a
b
Recessive allele
Genotype:
PP
aa
Bb
Homozygous for the dominant allele
Homozygous for the recessive allele
Heterozygous

Genetics and Probability

- **Probability –** The likelihood that a particular event will occurr.
  (ex. the flip of a coin, rolling dice)
- Alleles segregate at random when forming gametes, just like the flip of a coin
- Probability can be used to predict the outcomes of genetic crosses



Probability

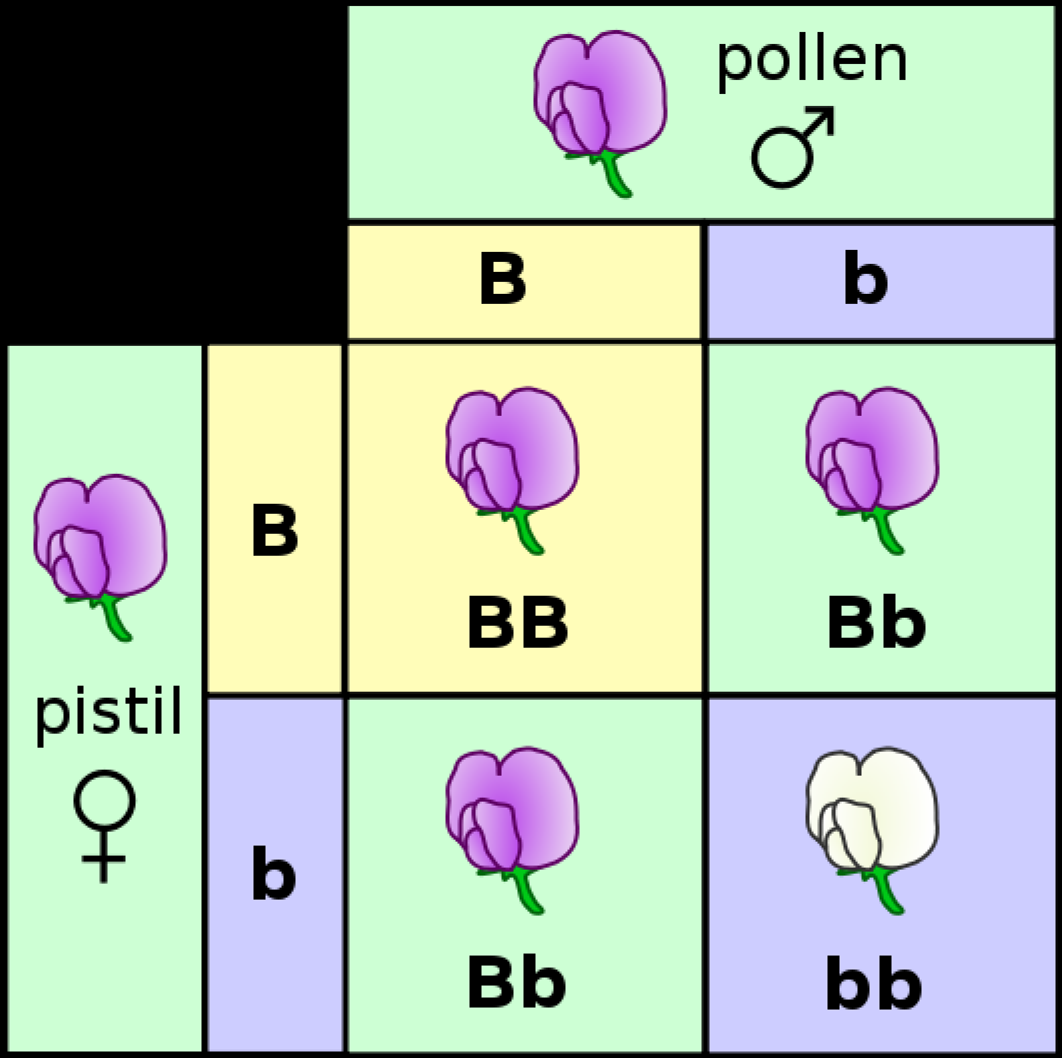
- What is the probability of flipping three heads in a row?

$$\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = 1/8$$

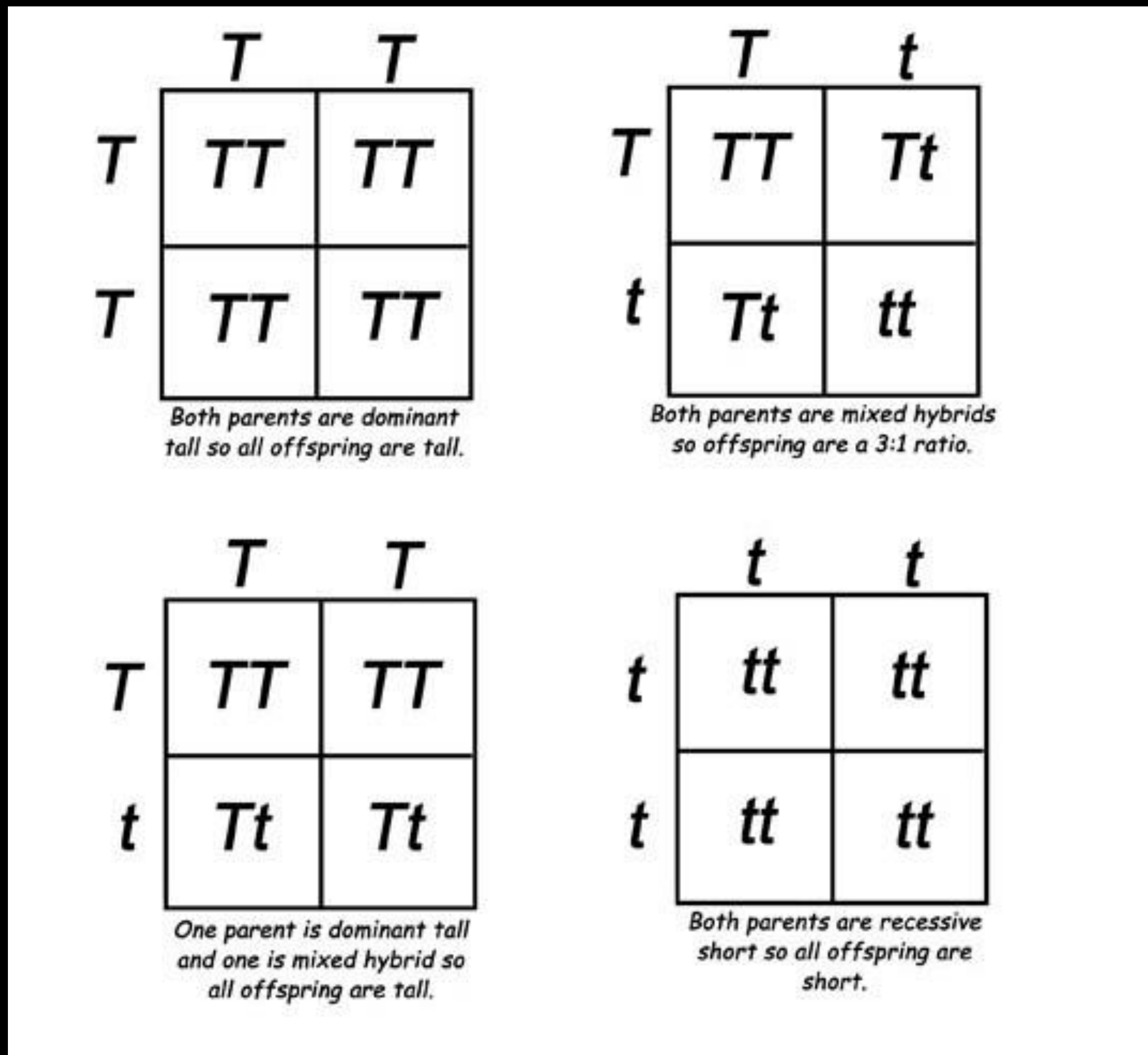
Probability and Punnett Squares

- Punnett Square – a diagram used to predict and compare the genetic variations that will result from a cross.
- Punnet squares show all the possible genetic combinations that offspring can inherit.

Punnett Square



Punnett Squares



Probability and Punnett Squares

- Mendel's assumptions about segregation proved to be correct.
- His predicted ratios (3 dominant to 1 recessive) showed up consistently
- For each of his seven crosses, about ¾ of the plants showed the trait controlled by the dominant allele.
- Around ¼ showed the trait controlled by the recessive trait.
- Punnett squares help support Mendel's assumptions about segregation

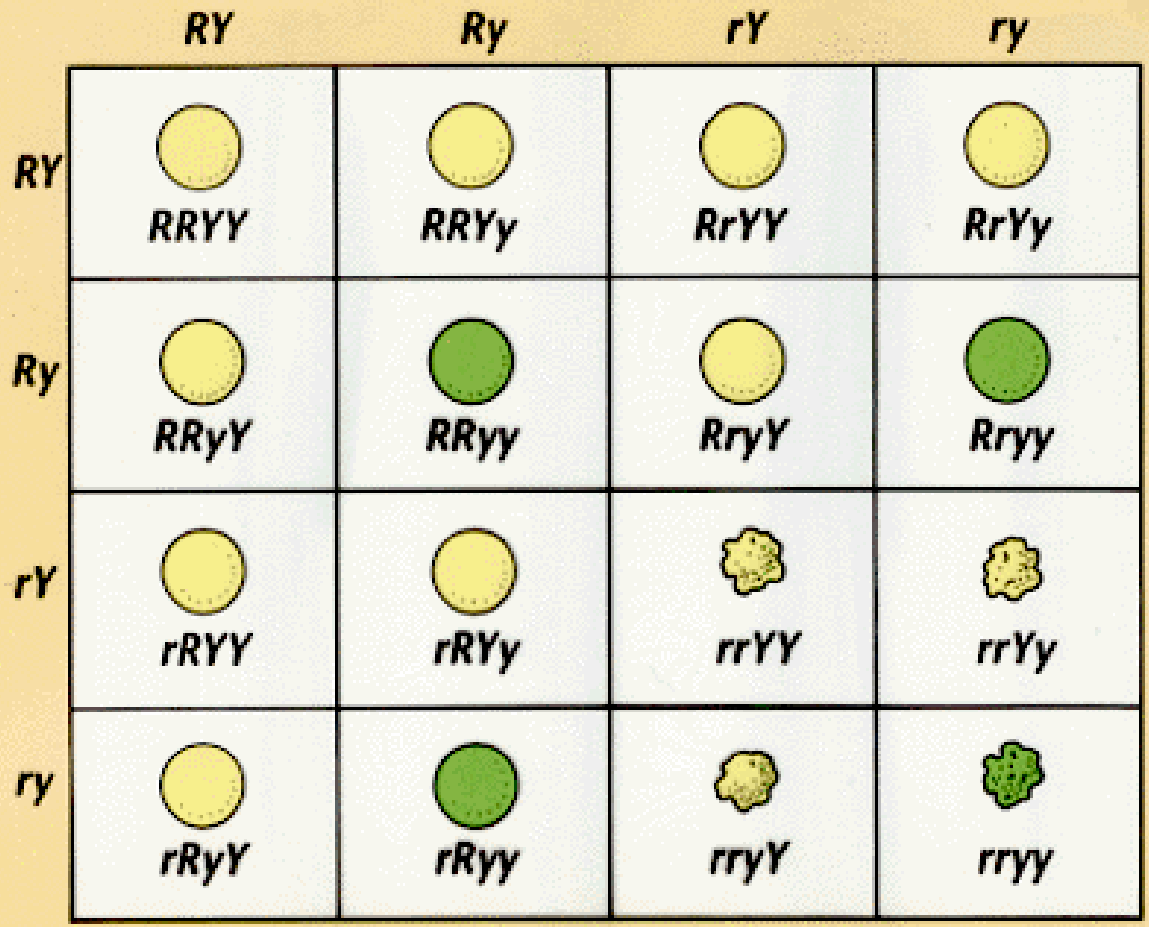
Probabilities Predict Averages

- Probabilities predict the average outcome of a large number of events.
- Probability cannot predict the precise outcome of an individual event
- Large number of events more accurately predicts the expected ratios.
  Ex. Flipping a coin 100 times gets you closer to the predicted 50:50 ratio

Exploring Mendelian Genetics

- Mendel showed that alleles segregate during gamete formation, but wondered if genes for different traits were independent or not. Question: Does Segregation of one pair of alleles affect the segregation of another pair of alleles?
- Mendel performed an experiment to test this. A Two-Factor Cross:
- Independent Assortment: Genes that segregate independently do not affect one another's inheritance

Parents (F_1) : *RrYy x RrYy*

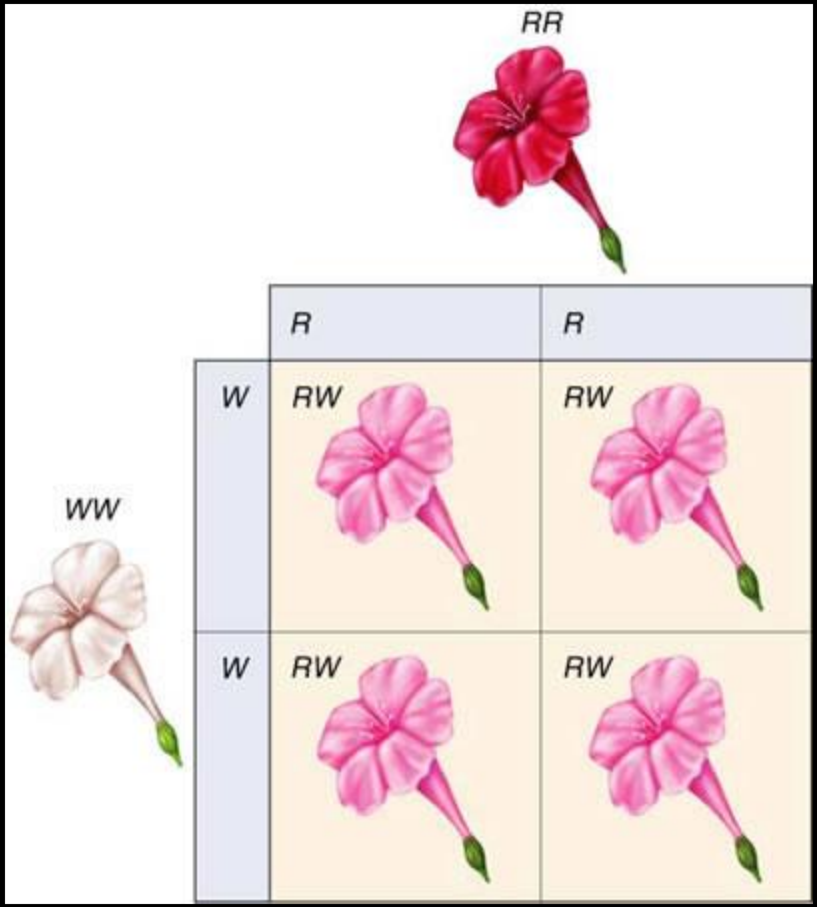


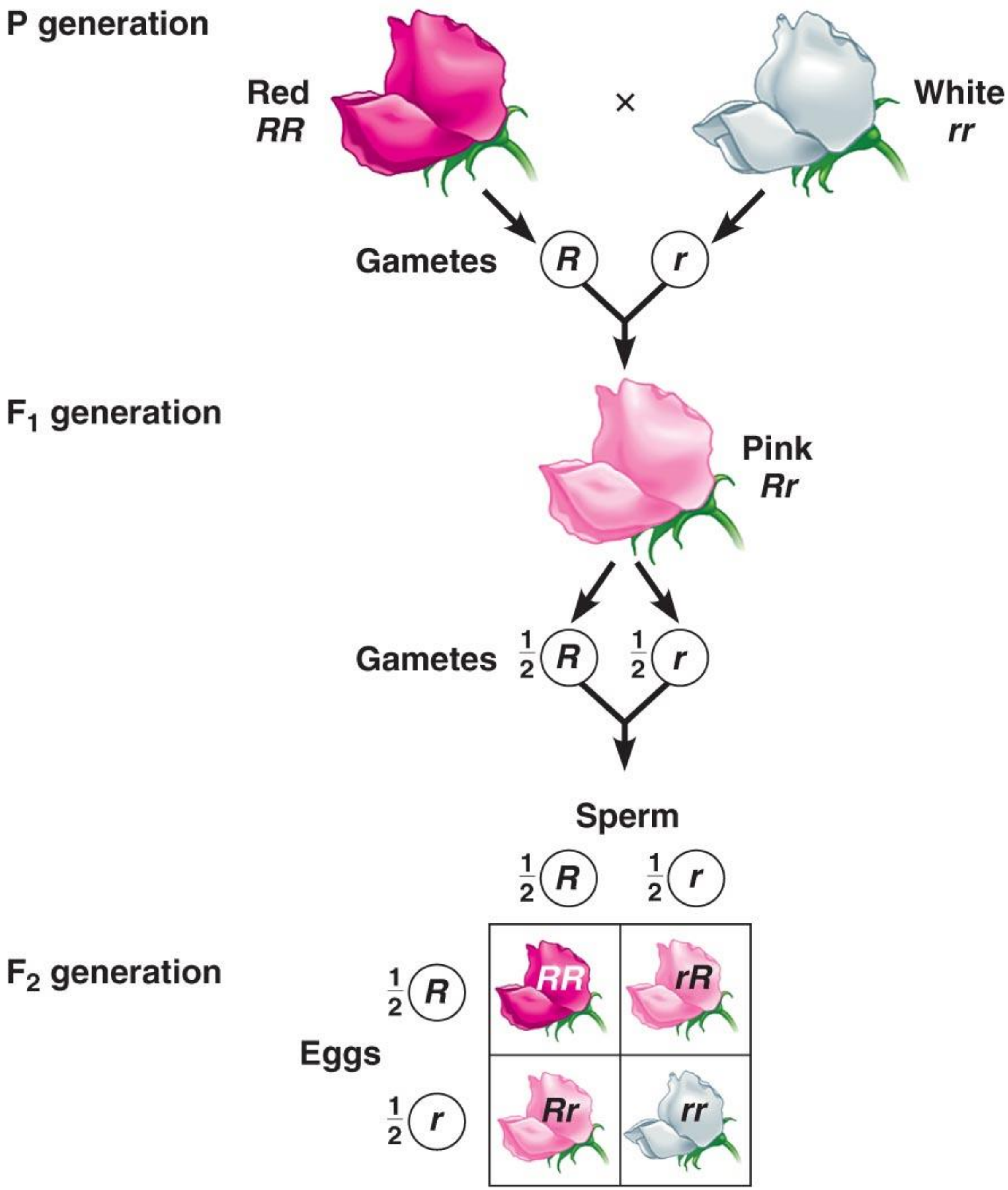
9/16 yellow-round
3/16 yellow-wrinkled
3/16 green-round
1/16 green-wrinkled

Summary of Mendel's Principles

- The inheritance of biological characteristics is determined by individual units know as genes. Genes are passed from parent to offspring.
- In cases in which two or more forms (alleles) of the gene for a single trait exist, some forms of the gene may be dominant and others recessive.
- In most sexually reproducing organisms, each adult has two copies of a gene -one from each parent. These genes are segregated from each other when gametes are formed.
- The alleles for different genes usually segregate independently of one another.

Incomplete/ Codominance




P generation
Red
RR
×
White
rr
Gametes
R
r
F1 generation
Pink
Rr
Gametes
1/2 R
1/2 r
Sperm
1/2 R
1/2 r
F2 generation
Eggs
1/2 R
1/2 r
RR
rR
Rr
rr


Wayne Jenkins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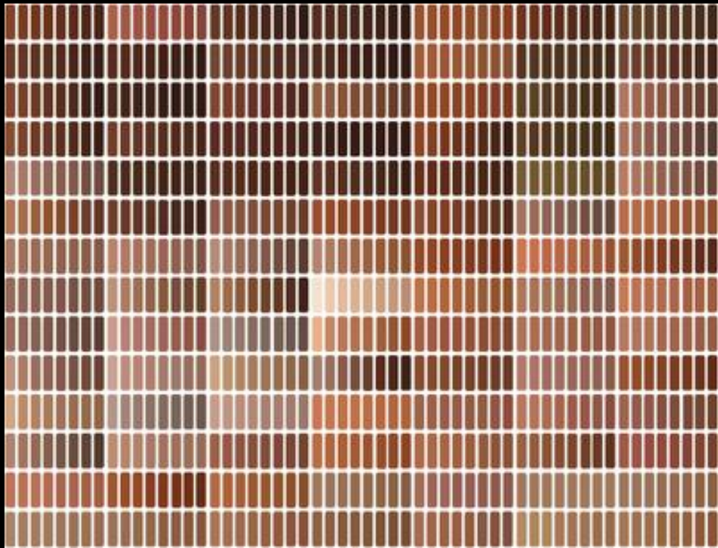
Beyond Dominance and Recessive

- Genetics is more complicated because most genes have more than two alleles.
- Many Important traits are controlled by more than one gene.
- **Incomplete Dominance:** One allele is not completely dominant over the other.
- **Codominance:** Both alleles contribute to the phenotype

Polygenic Traits

- Many traits are produced by the interaction of several genes.
- Traits controlled by two or more genes are said to be polygenic traits ("having many genes")
- EX: Wide range of skin tones in humans



GENETIC INHERITANCE: FACE LAB

- Determine sex of child:
  Male: XY Female: XX
- There are two sex chromosomes: X and Y
- The mother can only pass an X chromosome to the child
- The father can pass an X or a Y
- Have the “father” flip the coin. “Heads” is a male, “Tails” is a female.

GENETIC INHERITANCE: FACE LAB

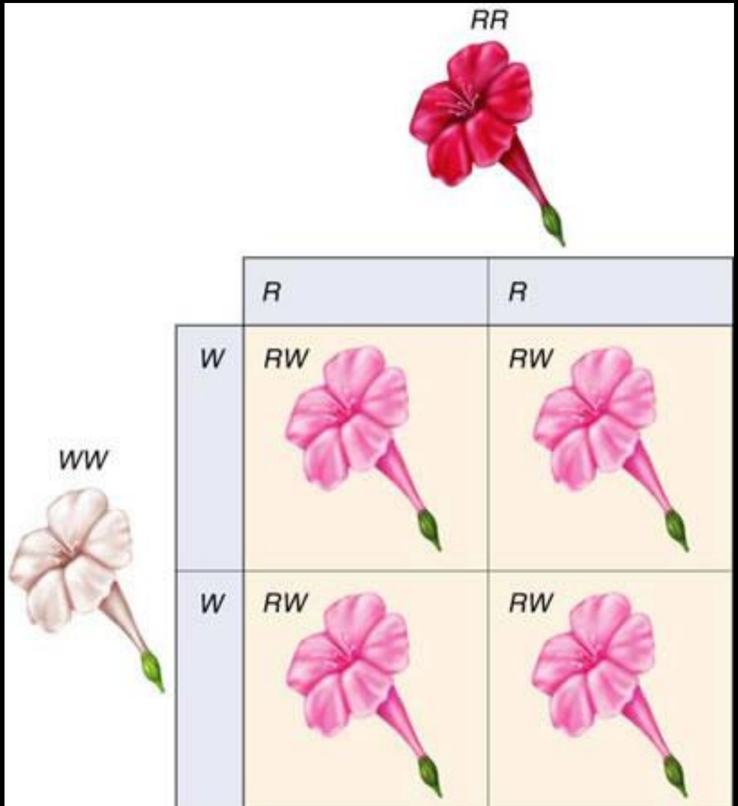
- For each trait, heads will be the dominant allele and tails will be the recessive allele.
- Flip your coins and check which trait get on the list.
- After you have gone through all the traits, make a sketch of what your child looks like on the last page using colored pencils.

GENETIC INHERITANCE: FACE LAB

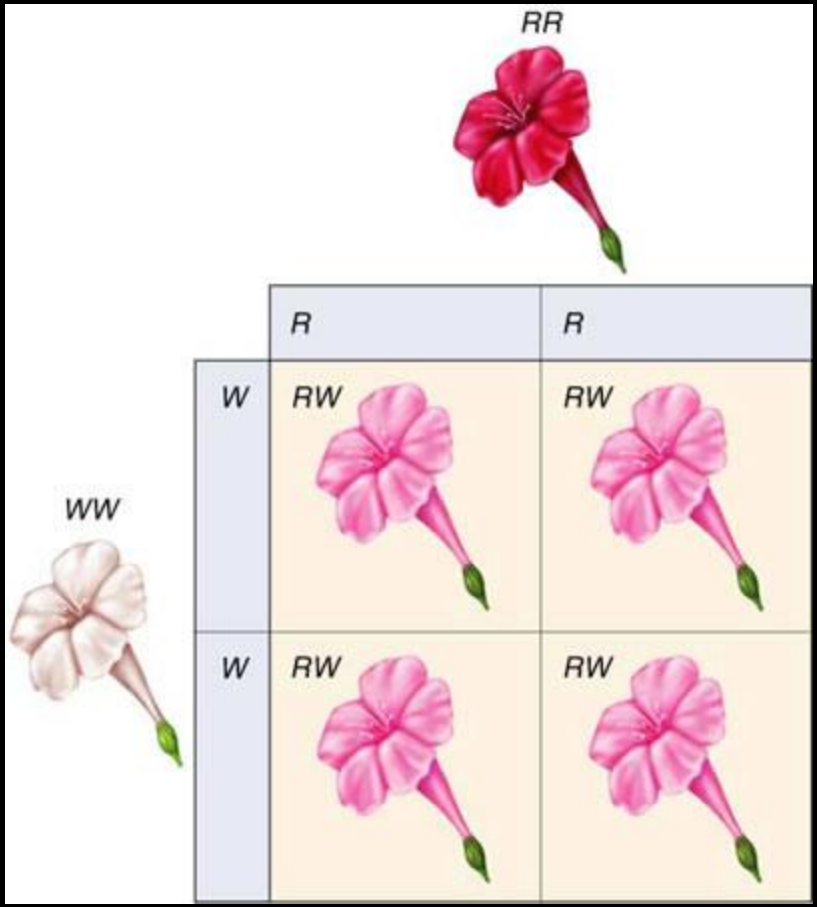
- Skin color and hair color are polygenic traits.
- You will flip the coins for each gene (A, B, C, D)
- The phenotype is determined by the number of DOMINANT alleles obtained.

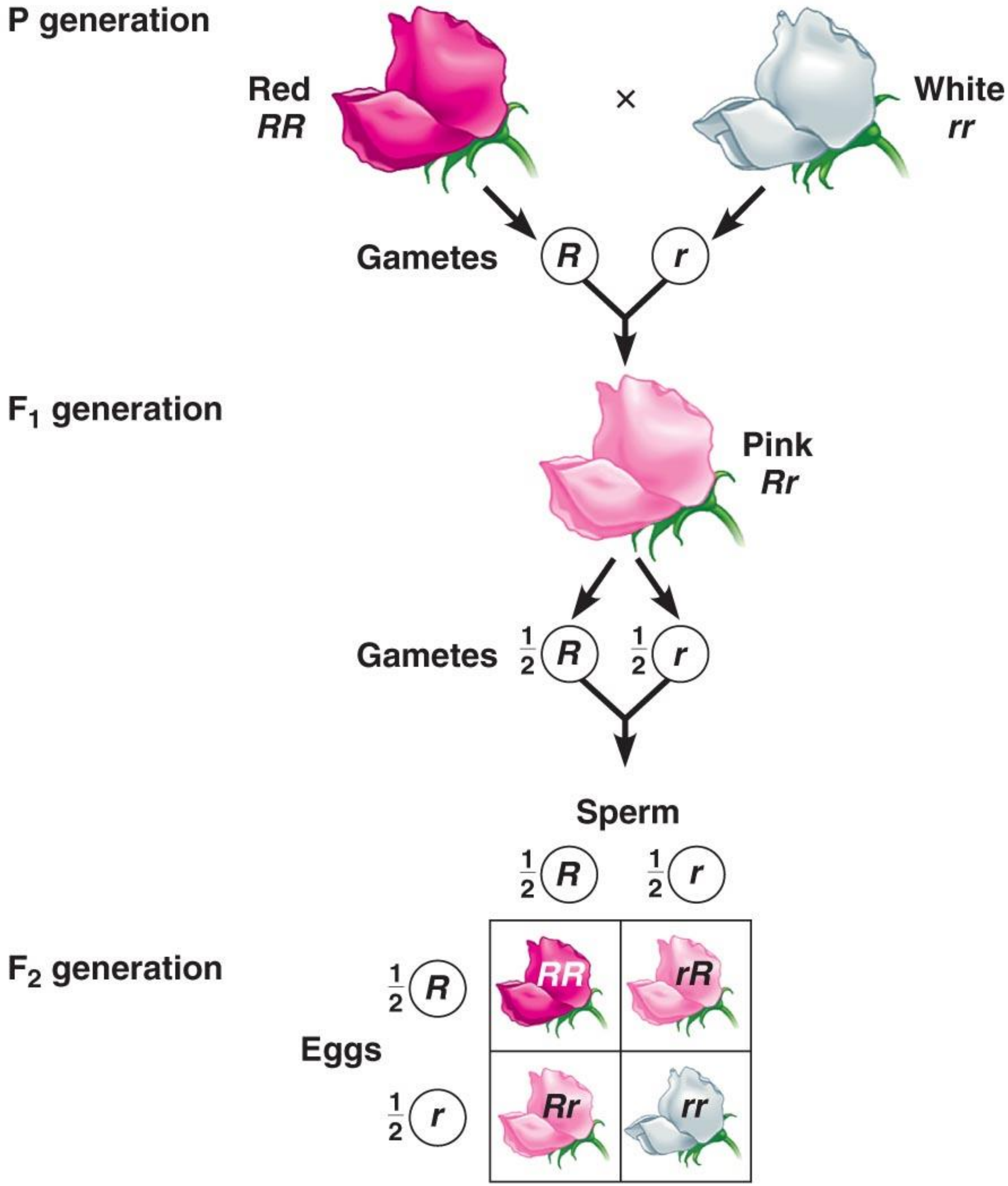
Trait	Genes from Mother	Genes from Father	Genotype	Phenotype
Face Shape	R	r	Rr	Round

Explain why the offspring of this genetic cross are neither white nor red using what you know about dominant and recessive traits.



Incomplete/ Codominance




P generation
Red
RR
×
White
rr
Gametes
R
r
F1 generation
Pink
Rr
Gametes
1/2 R
1/2 r
Sperm
1/2 R
1/2 r
F2 generation
1/2 R
Eggs
1/2 r
RR
rR
Rr
rr


Wayne Jenkins


62

Beyond Dominance and Recessive

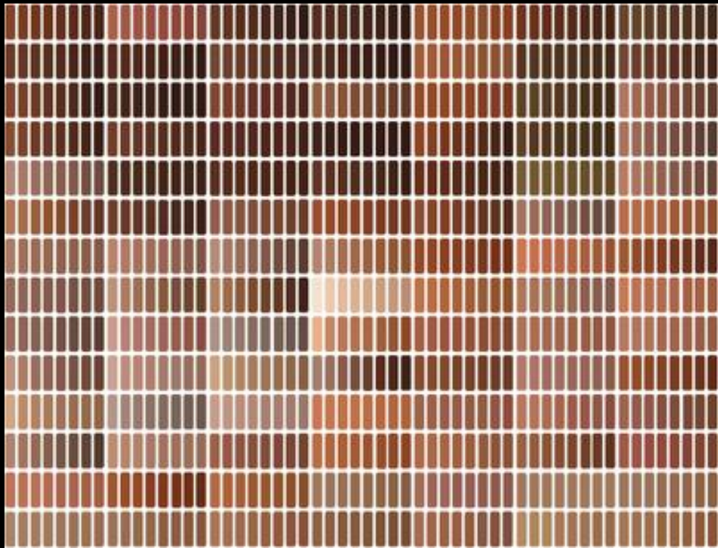
- Genetics is more complicated because most genes have more than two alleles.
- Many Important traits are controlled by more than one gene.
- **Incomplete Dominance:** One allele is not completely dominant over the other.
- **Codominance:** Both alleles contribute to the phenotype

Beyond Dominant & Recessive

- Multiple Alleles: Many genes have more than two alleles.
- This does not mean that an individual can have more than two alleles.
- It only means that more than two possible alleles exist in a population.
  Ex: Blood type in humans
  I^A, I^B, I^O

Polygenic Traits

- Many traits are produced by the interaction of several genes.
- Traits controlled by two or more genes are said to be polygenic traits ("having many genes")
- EX: Wide range of skin tones in humans



Applying Mendel’s Principles

- Thomas Hunt Morgan contributed to our understanding of genetics by studying fruit flies.
- He chose fruit flies because they were animals, easy to keep in the lab, had large offspring sizes, and had short generation times (reproduce quickly).

- Hunt discovered traits linked on sex chromosomes (X,Y)

11-4 Meiosis

- Key Terms:
- Meiosis- Process by which the number of chromosomes in each cell are cut in half through the separation of homologous chromosomes.
- Homologous Chromosomes - A pair of similar chromosomes, one from each parent.
- Diploid – (“two sets”) cells that contain both sets of homologous chromosomes
- Haploid – (“one set’) cells that only contain one set of homologous chromosomes

Meiosis

- Gregor Mendel did not know where the genes he had discovered were located in the cell. He did know that traits followed mathematical models of probability.
- Mendel’s contributions to genetics helped biologists discover what genes were and where they were located (on chromosomes)
- **Remember:** Genes are segment of DNA that code for a trait

Chromosome Number

- A cell that contains both sets of homologous chromosomes is said to be diploid, which means “two sets”
- The number of diploid chromosomes in a diploid cell is represented by the symbol 2N.
- Ex: The diploid number in humans is 46

  2N = 46

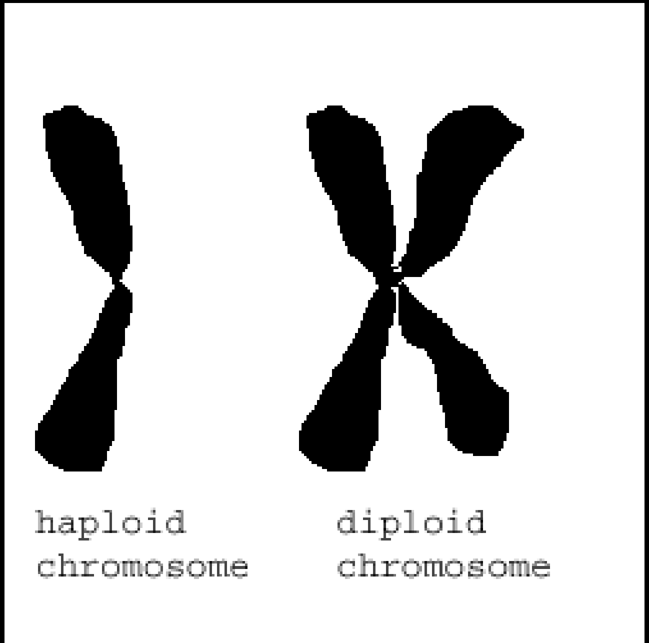
Chromosome Number

- By contrast, gametes of sexually reproducing organisms contain only a single set of chromosomes, therefore only a single set of genes.
- Such cells with only one set of chromosomes are said to be haploid.
- For humans, the haploid number is:

N = 23

Chromosome Number

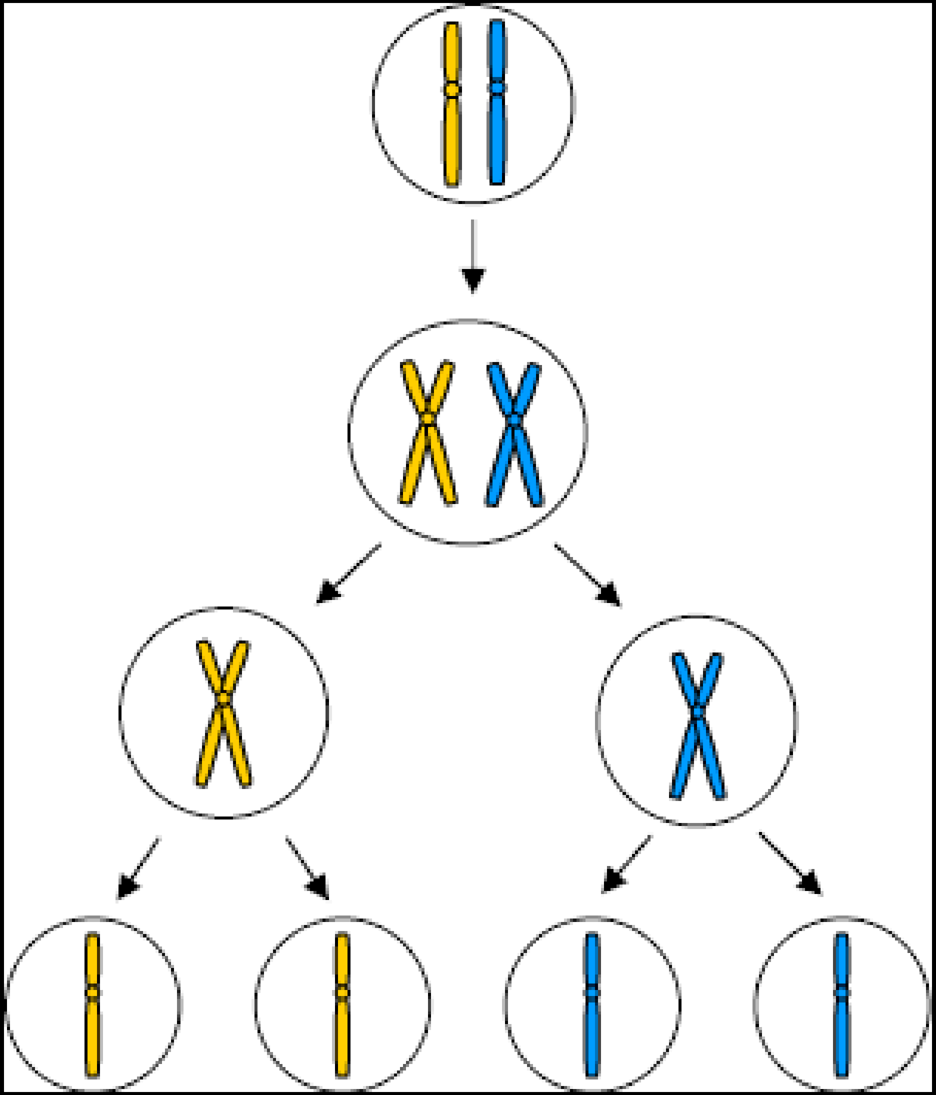
- Diploid – (“two sets”) cells that contain both sets of homologous chromosomes
- Haploid – (“one set’) cells that only contain one set of homologous chromosomes



Bell Ringer

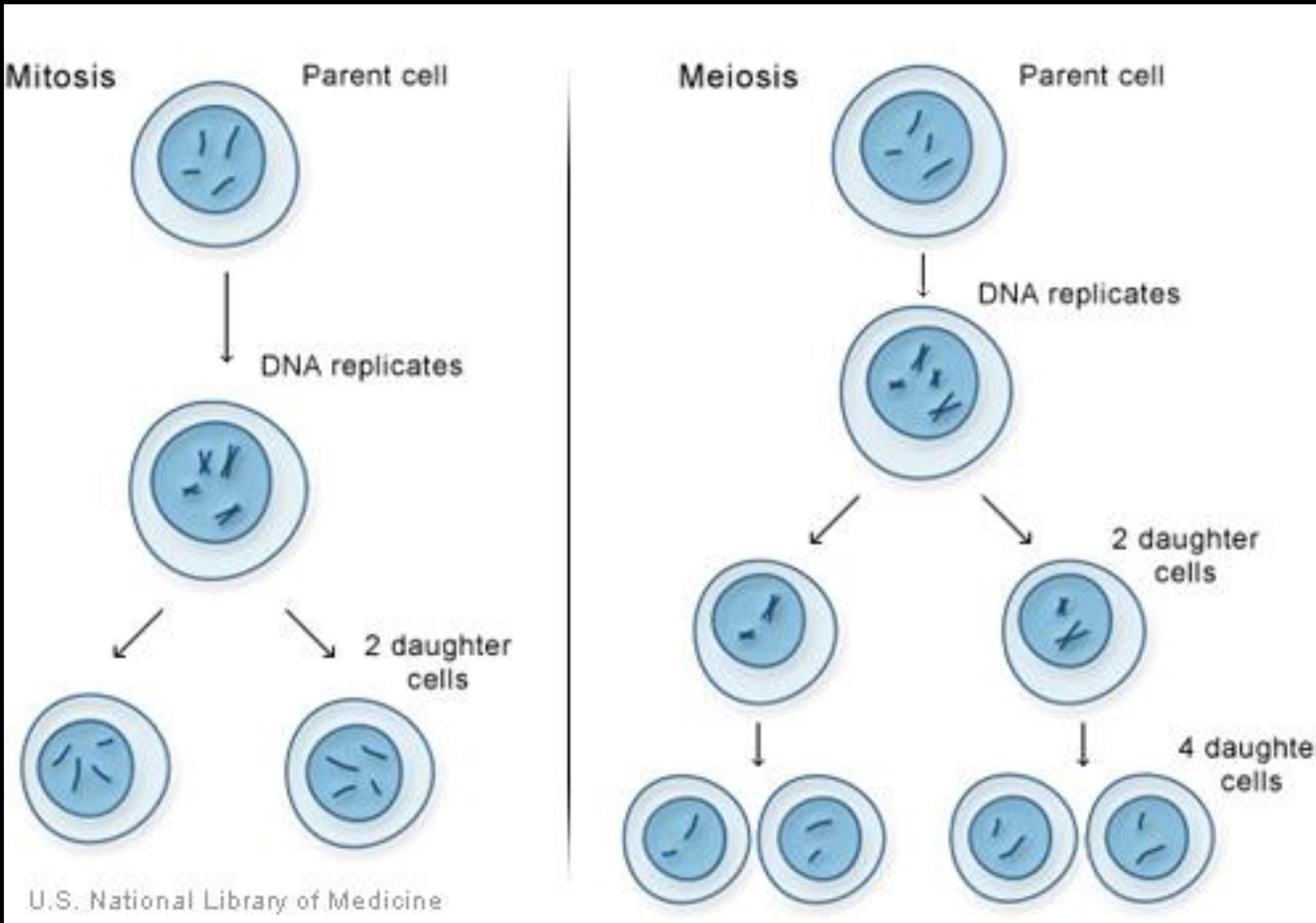
- What is the main difference between Mitosis and Meiosis.

Meiosis



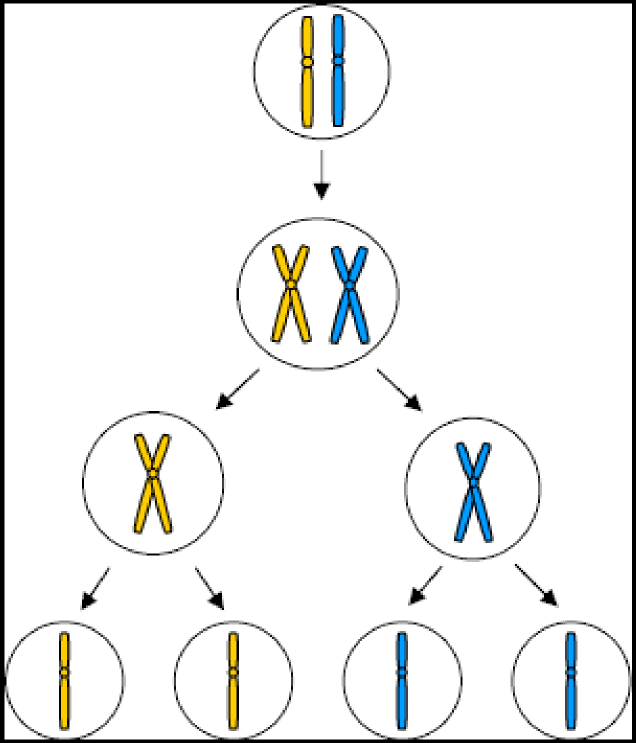
Phases of Meiosis

- Meiosis – The process of reductive division in which the number of chromosomes per cell is cut in half through a separation of homologous chromosomes in a diploid cell
- Meiosis is how haploid (N) reproductive cells are produced from diploid (2N) cells.
- In contrast, Mitosis is a process of cell division which results in the production of two identical daughter cells from a single parent cell.


Mitosis
Parent cell
DNA replicates
2 daughter cells
Meiosis
Parent cell
DNA replicates
2 daughter cells
4 daughter cells
U.S. National Library of Medicine

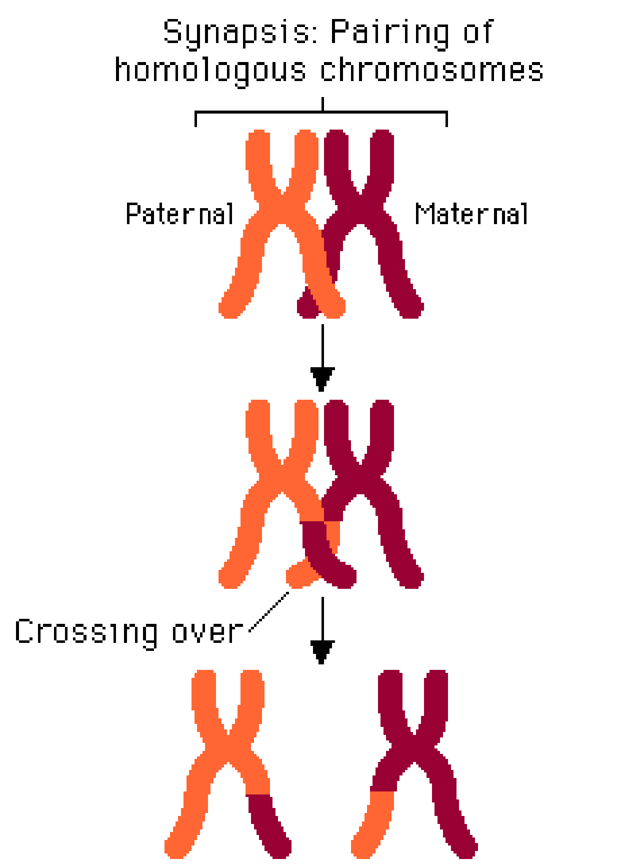
Phases of Meiosis

- Meiosis involves two distinct divisions, called Meiosis I and Meiosis II.
- During meiosis I, genetic material (DNA) is replicated and the cell divides, similar to mitosis

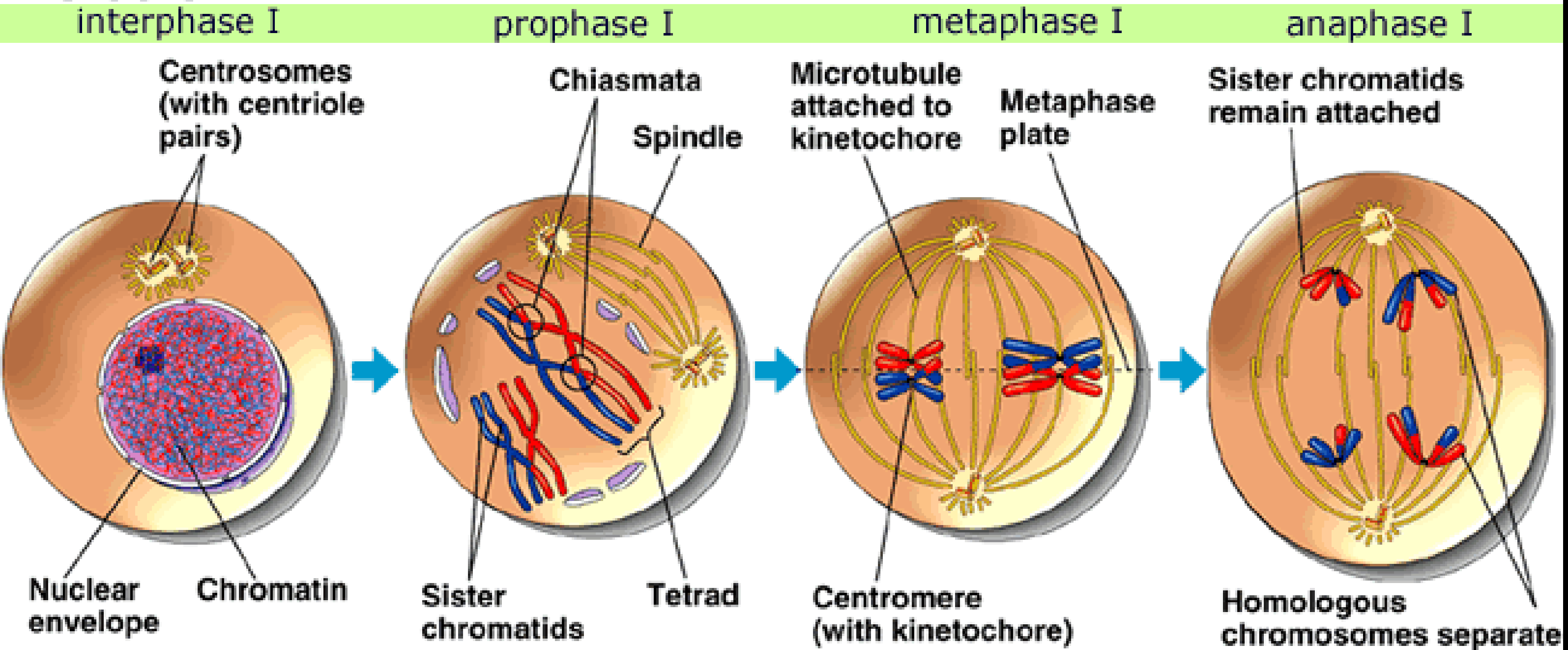


Phases of Meiosis

- In Prophase I of Meiosis I, two homologous pairs of chromosomes line up against each other, forming a tetrad.
- While the chromosomes are lined up next to each other, the they exchange portions of themselves in a process we call crossing over.
- Each chromosome is now different, due to crossing over



Meiosis I



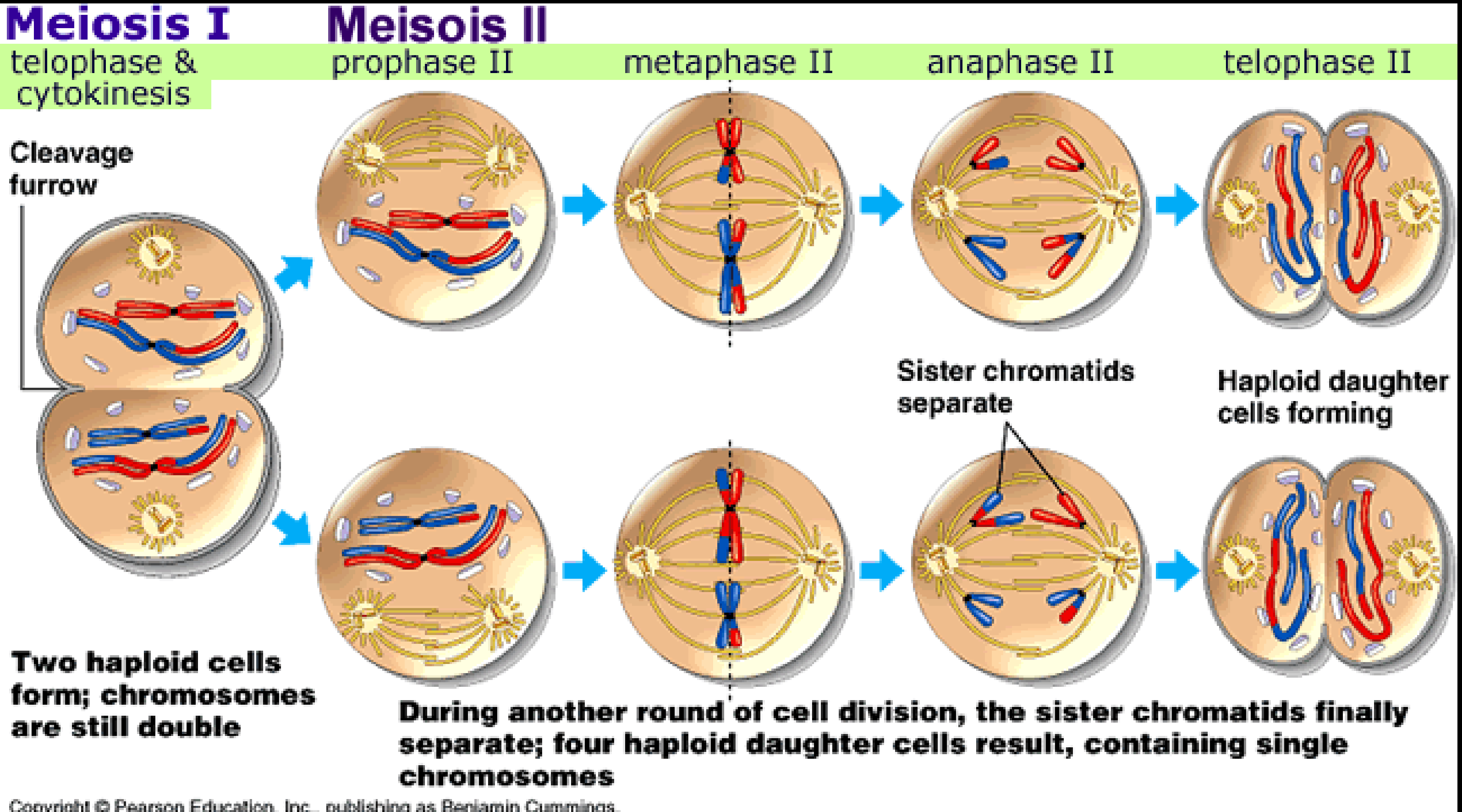
Chromosomes duplicate

Homologous chromosomes pair and exchange segments

Synapsis - pairing of homologs to form tetrad

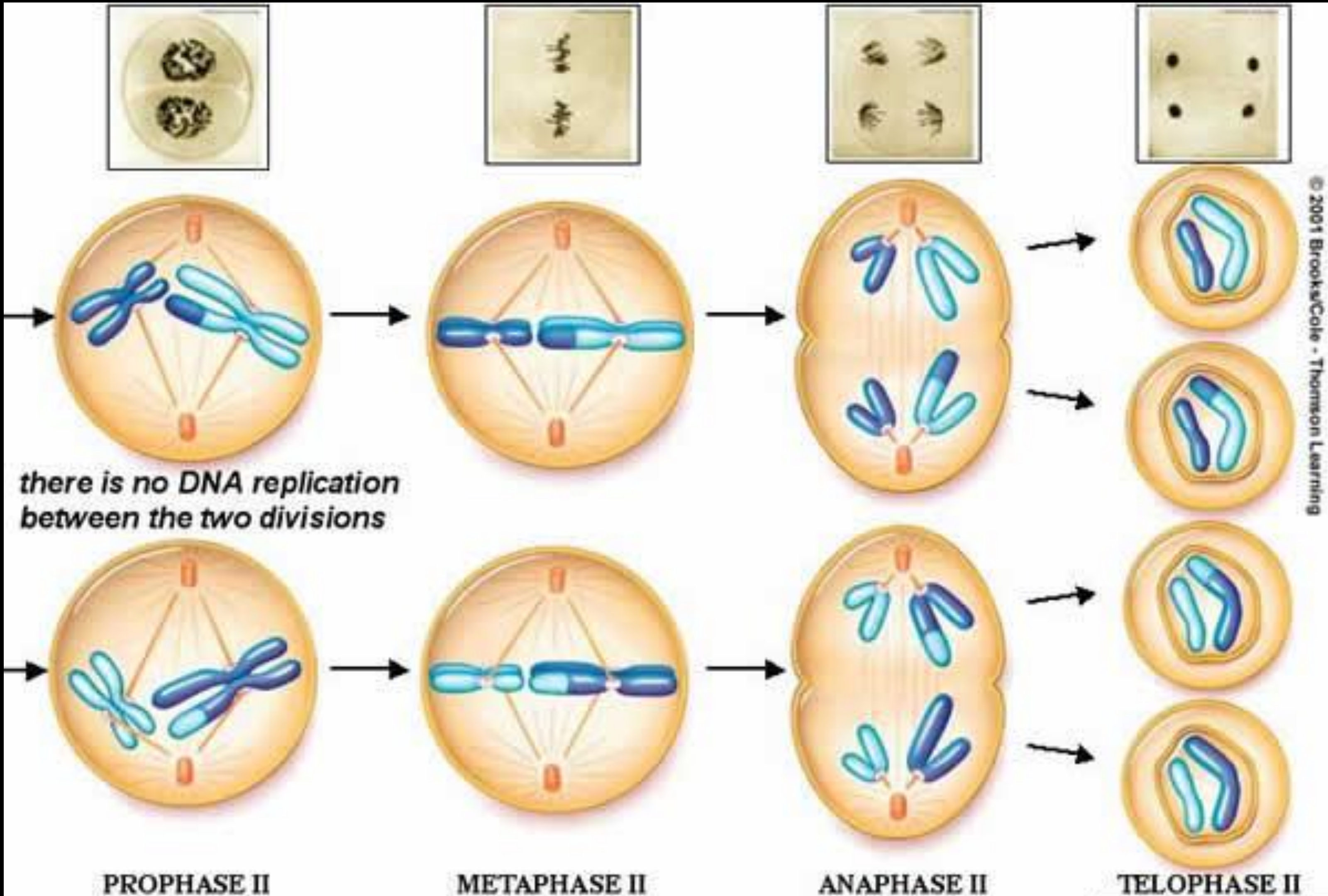
Tetrads line up

Pairs of homologous chromosomes split up


Meiosis I
Meisois II
telophase & cytokinesis
prophase II
metaphase II
anaphase II
telophase II
Cleavage furrow
Sister chromatids separate
Haploid daughter cells forming
Two haploid cells form; chromosomes are still double
During another round of cell division, the sister chromatids finally separate; four haploid daughter cells result, containing single chromosomes
Copyright © Pearson Education, Inc., publishing as Benjamin Cummings.

Phases of Meiosis

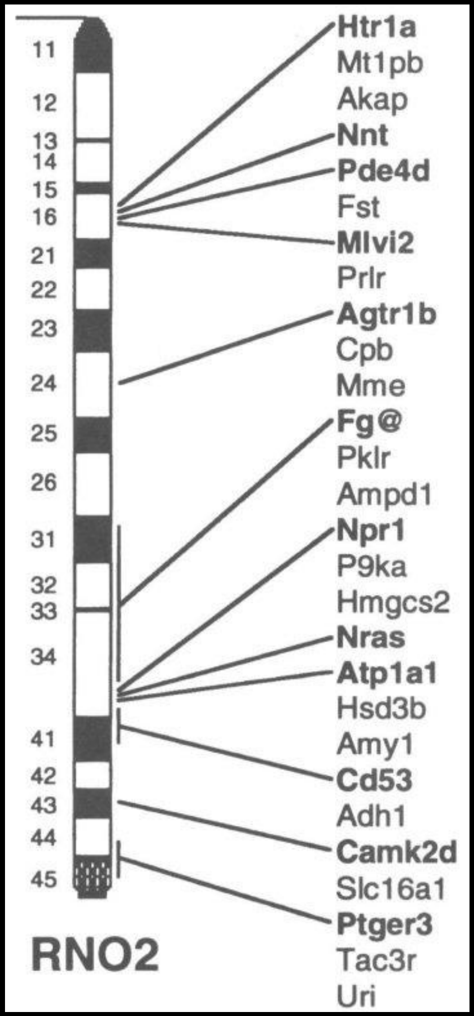
- DNA does not replicate during Meiosis II
- During Meiosis II, the two daughtor cells will divide again
- Chromosomes line up in the middle of the cells and split apart (just like Meiosis I)
- The end result is four haploid cells.
- Each sell is different from the original parent cell.



MEIOSIS II: Separate the Sister Chromatids (by mitosis)

Linkage and Gene Maps

- What structures actually assort independently?
- What are gene maps?
- What do gene maps tell us about the likelihood of independent assortment?

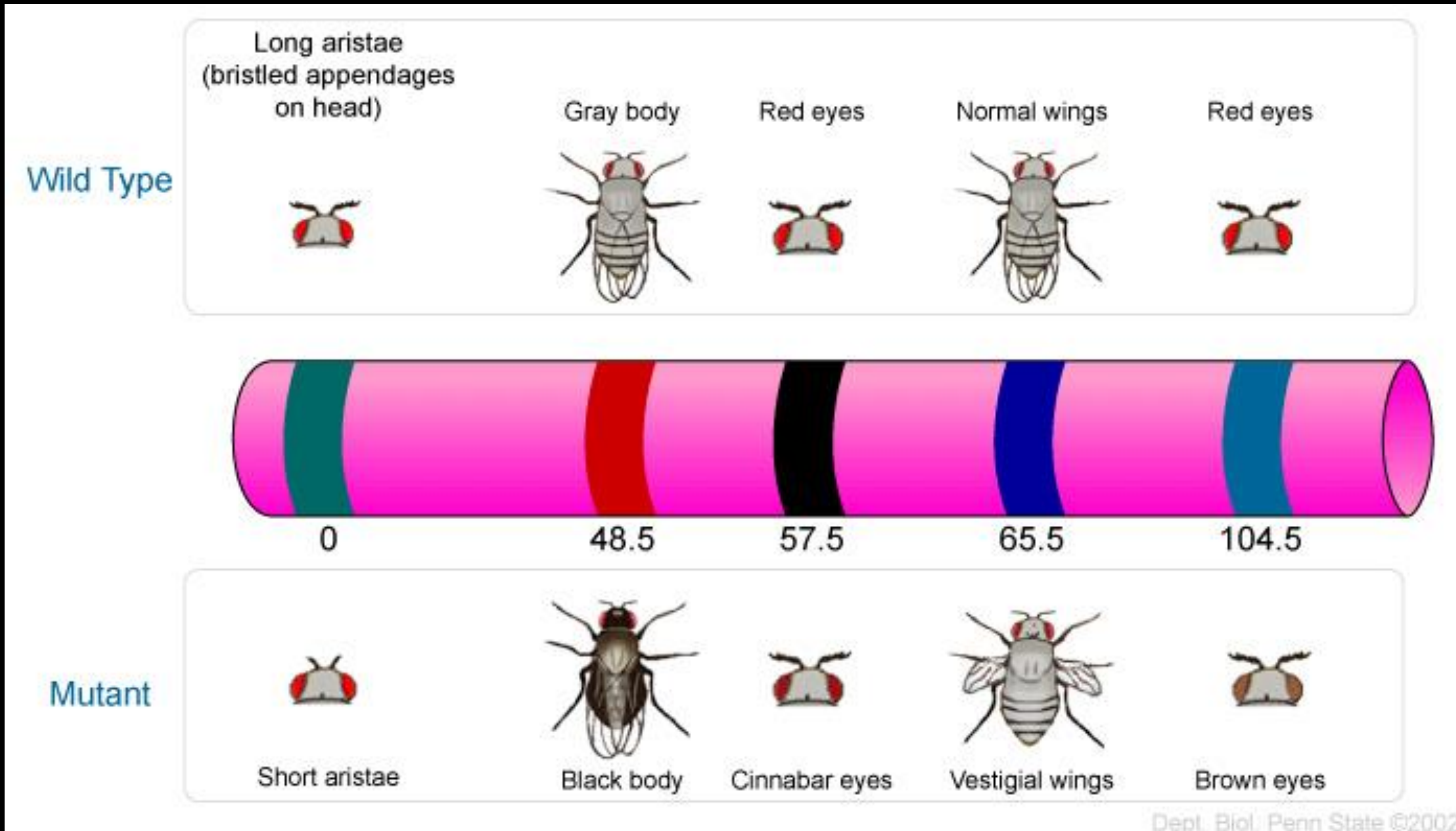


Linkage and Gene Maps

- Do genes on the same chromosome assort independently?
- Thomas Hunt Morgan’s research on fruit flies led him to the principal of linkage.
- He discovered that many genes appeared to be linked together, violating the law of independent assortment.
- He found that flies with orange eyes and minature wings almost always inherited these two traits together.

Linkage and Gene Maps

- These findings led Thomas Hunt Morgan to discover that each chromosome is a group of linked genes.
- It is chromosomes that assort independently of one another, not individual genes.
- Genes on the same chromosome, however, can still separate due to crossing over.


Wild Type
Long aristae (bristled appendages on head)
Gray body
Red eyes
Normal wings
Red eyes
0
48.5
57.5
65.5
104.5
Mutant
Short aristae
Black body
Cinnabar eyes
Vestigial wings
Brown eyes
Dept. Biol. Penn State ©2002

Linkage and Gene Maps

- Two genes on the same chromosome will assort independently depending on how far apart they are (map units).
- The farther apart two genes are, the more likely they are to be separated by crossing over during meiosis
- The rate at which genes separated and recombined is used to create a “map” of how far apart they are.

Bell-Ringer

- Write out a multiple choice or true/false question over material from chapter 11 to be used on the test on Thursday

Science News:

- Marine biologists have, for the first time, found a whale skeleton on the ocean floor near Antarctica, giving new insights into life in the sea depths. The discovery was made almost a mile below the surface in an undersea crater and includes the find of at least nine new species of deep-sea organisms thriving on the bones.

Chapter 11 Review

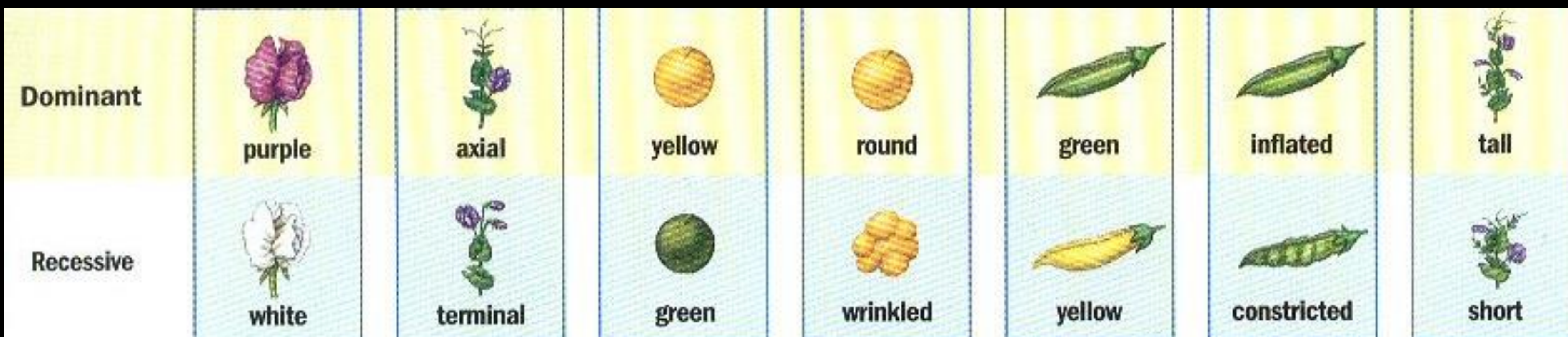
- Where does an organism get its unique characteristics?
- How are different forms of a gene distributed to offspring?
- How can we use probability to predict traits?
- How do alleles segregate when more than one gene is involved?

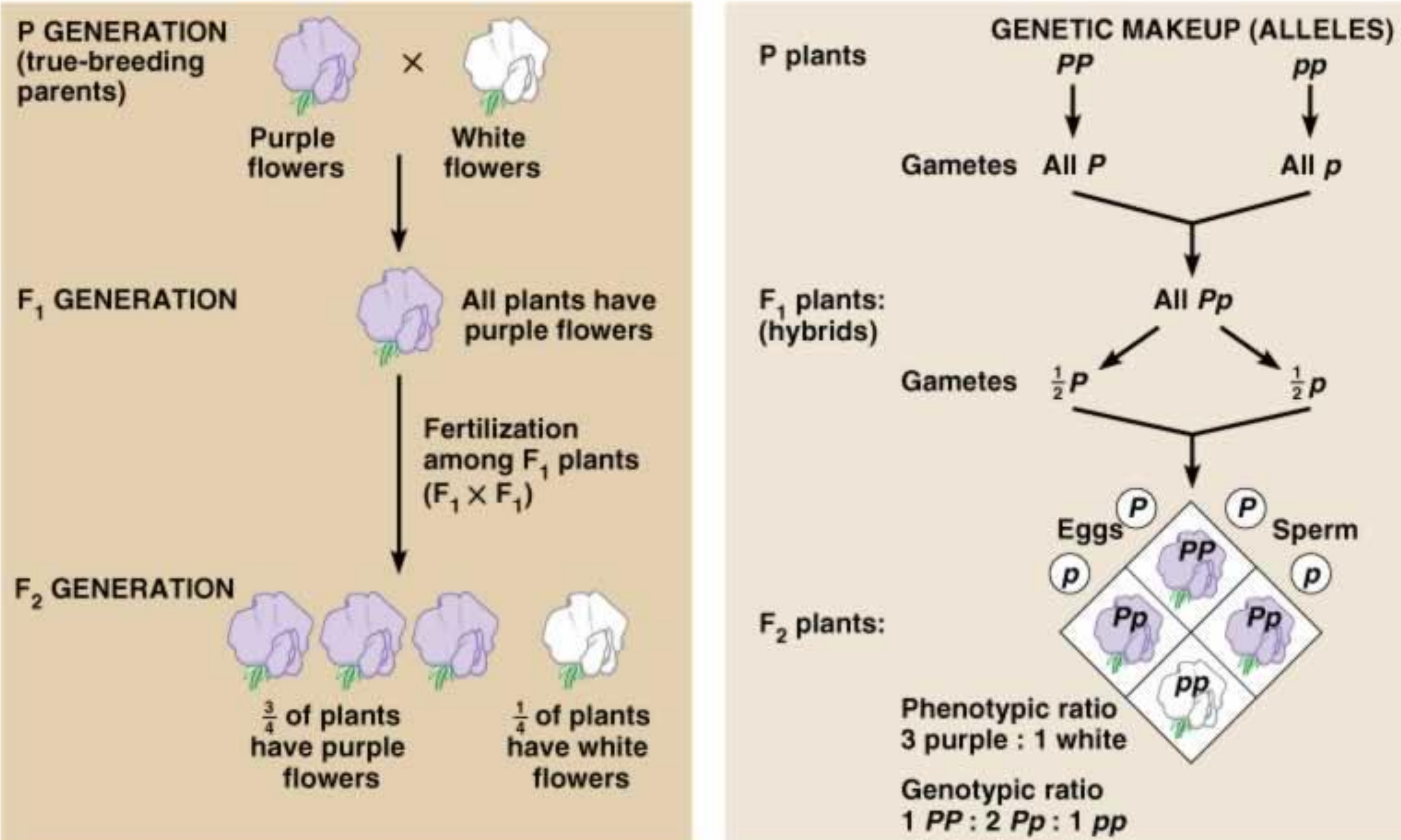
Chapter 11 Review

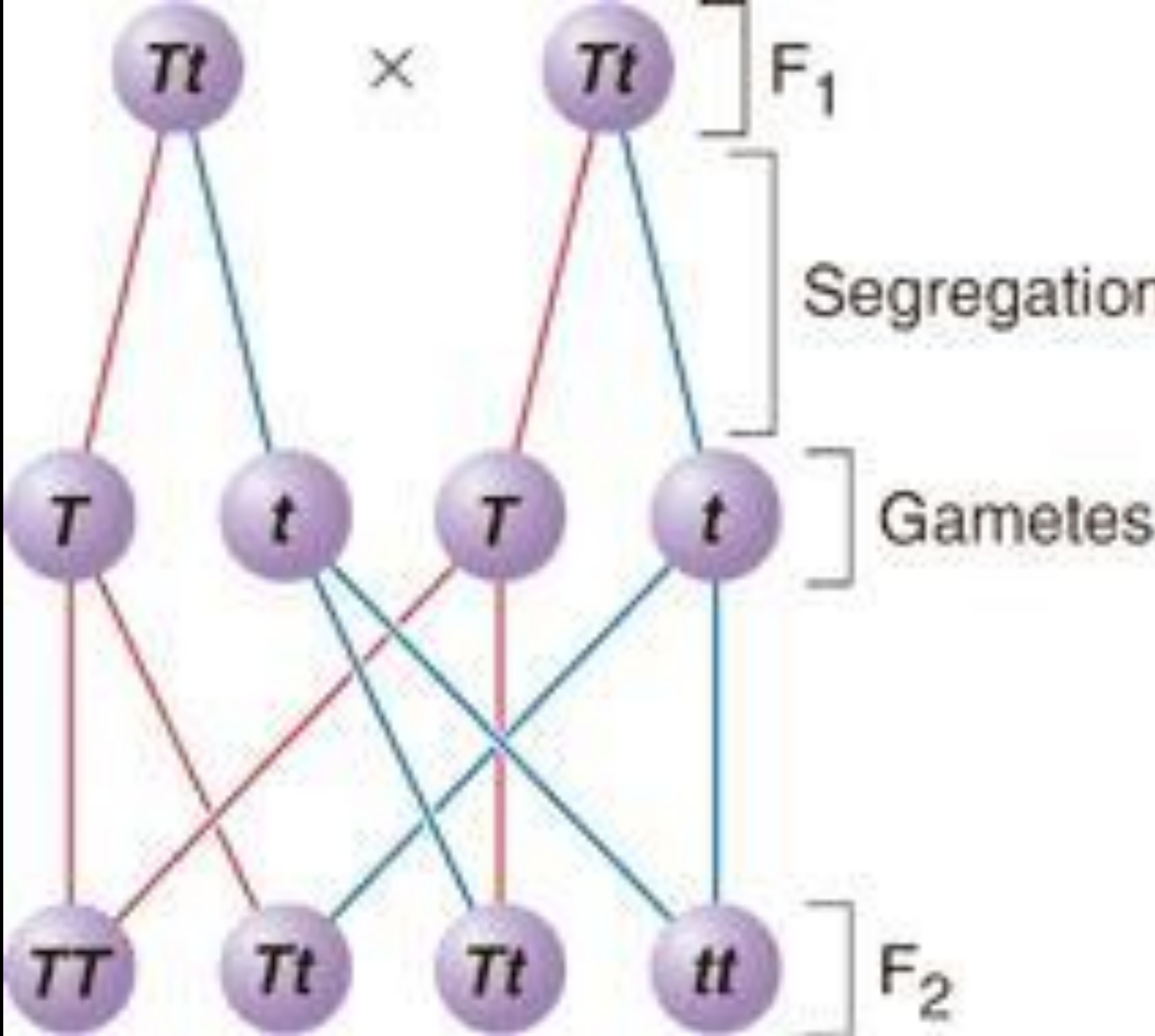
- What did Mendel contribute to our understanding of genetics?
- What are some exceptions to Mendel’s principles?
- Does the environment have a role in how genes determine traits?
- How many sets of genes are found in most adult organisms?

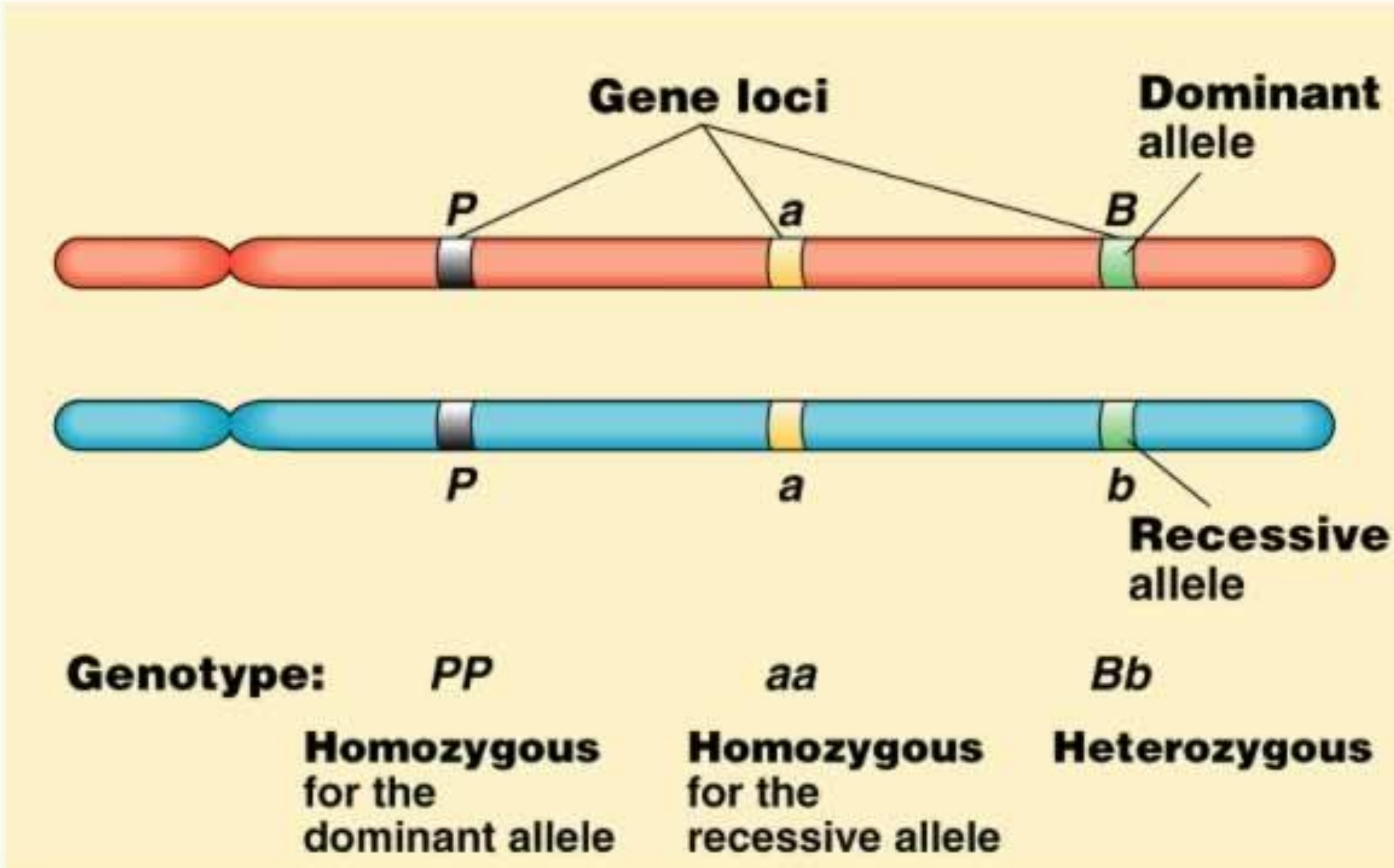
Chapter 11 Review

- What events occur during each phase of meiosis?
- How is meiosis different from mitosis?
- How can two alleles from different genes be inherited together?

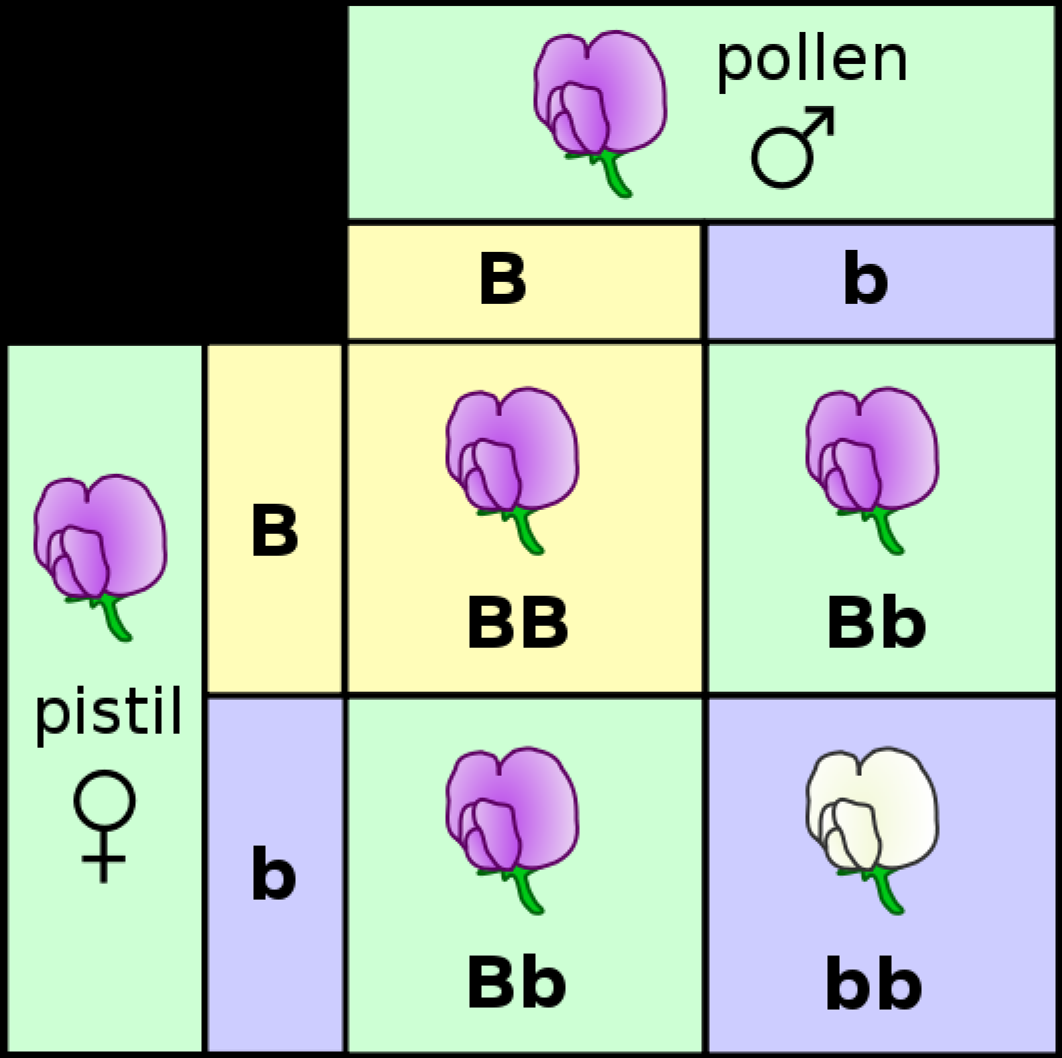

Dominant
purple
axial
yellow
round
green
inflated
tall
Recessive
white
terminal
green
wrinkled
yellow
constricted
short


P GENERATION
(true-breeding parents)
×
Purple flowers
White flowers
F1 GENERATION
All plants have purple flowers
Fertilization among F1 plants
(F1 × F1)
F2 GENERATION
3/4 of plants have purple flowers
1/4 of plants have white flowers
GENETIC MAKEUP (ALLELES)
P plants
PP
pp
Gametes
All P
All p
F1 plants: (hybrids)
All Pp
Gametes
1/2 P
1/2 p
Eggs
Sperm
P
P
p
p
PP
Pp
Pp
pp
F2 plants:
Phenotypic ratio
3 purple : 1 white
Genotypic ratio
1 PP : 2 Pp : 1 pp

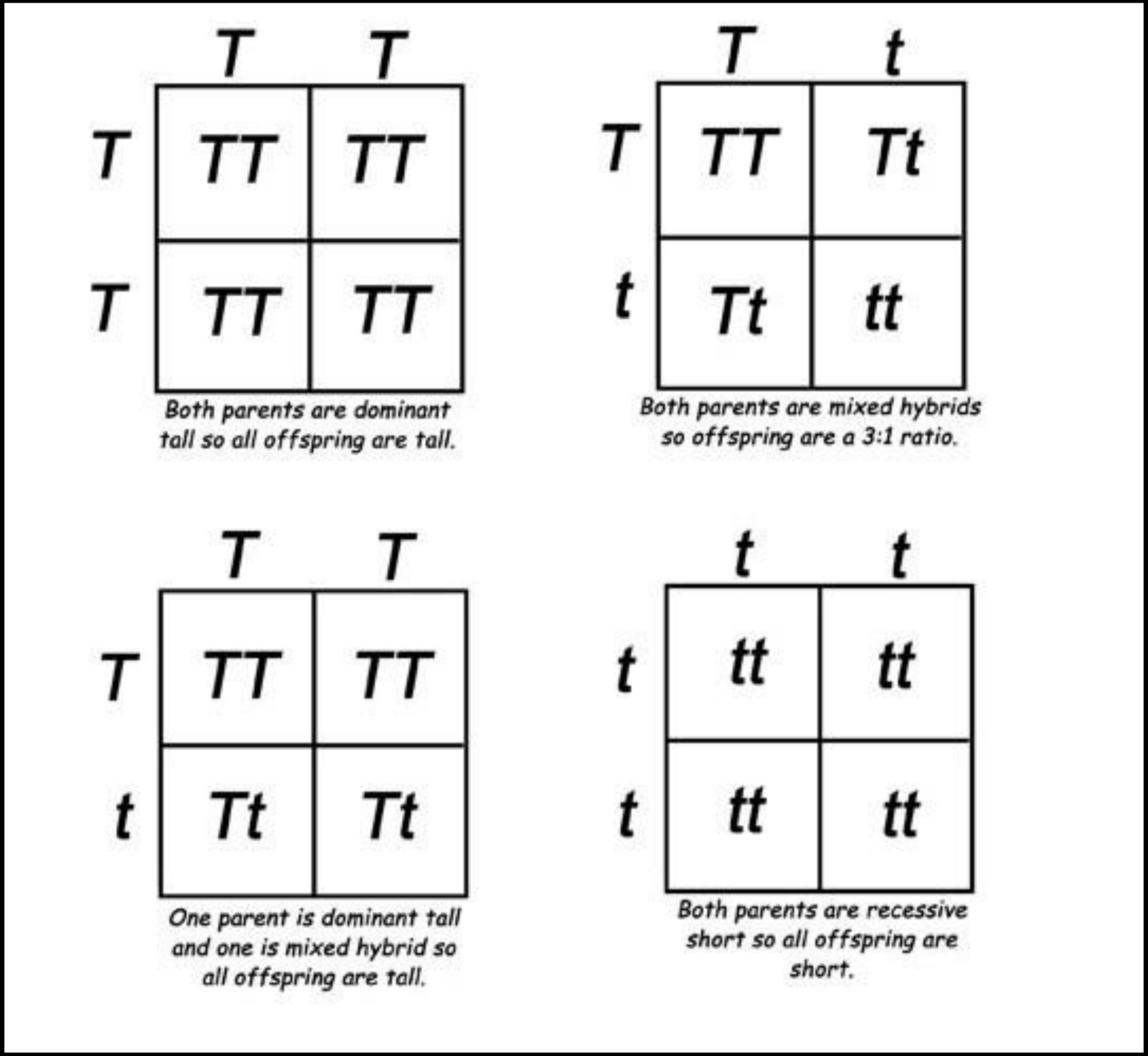

Tt
×
Tt
F1
Segregation
T
t
T
t
Gametes
TT
Tt
Tt
tt
F2


Gene loci
Dominant allele
P
a
B
P
a
b
Recessive allele
Genotype:
PP
aa
Bb
Homozygous for the dominant allele
Homozygous for the recessive allele
Heterozygous

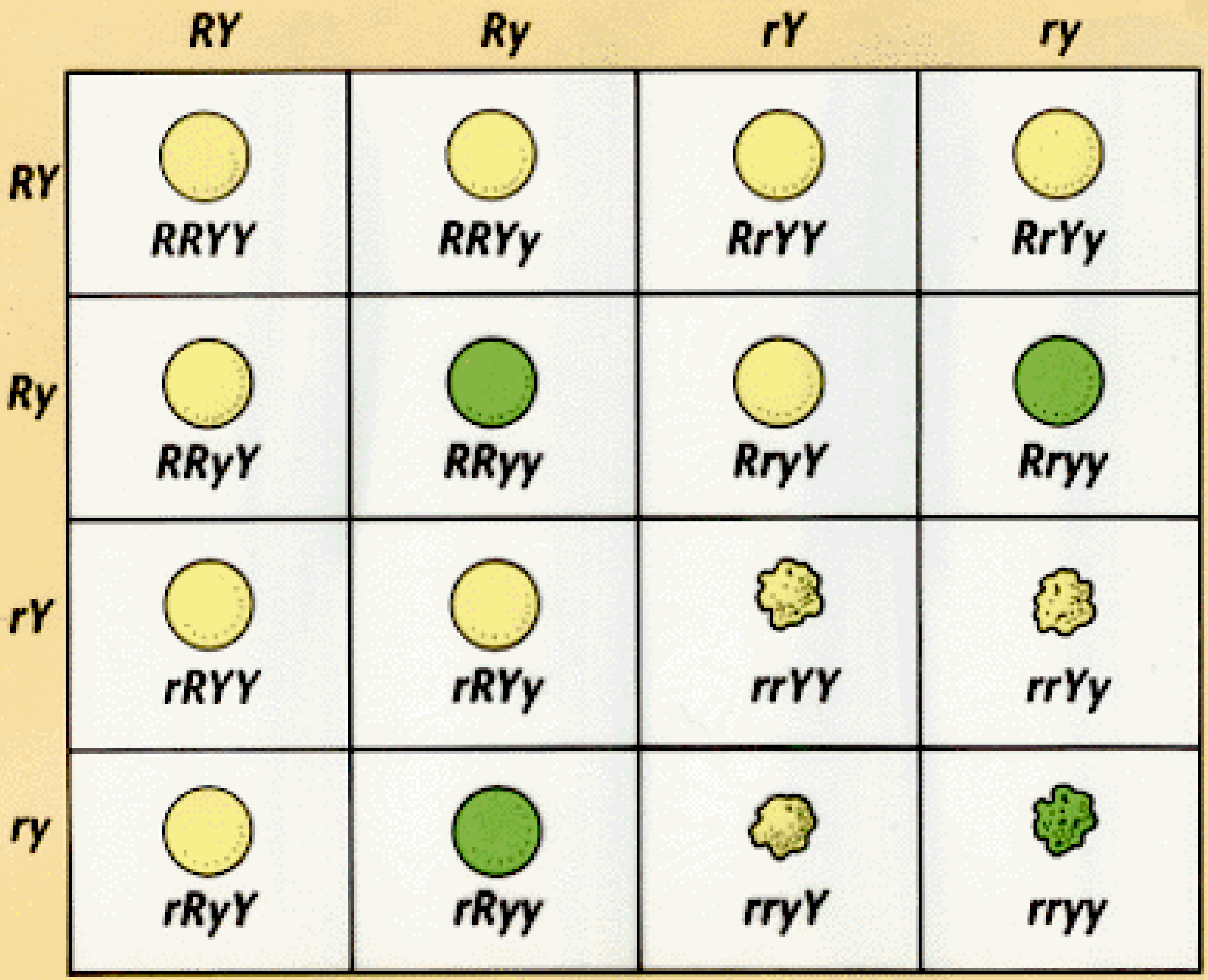
Punnett Square



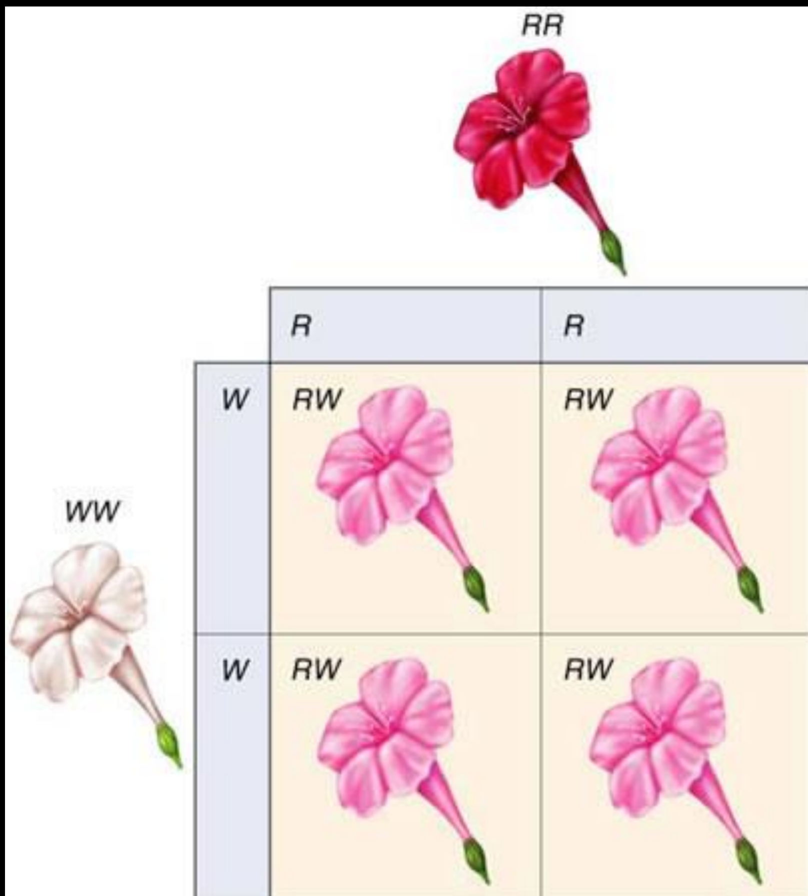
Punnett Squares

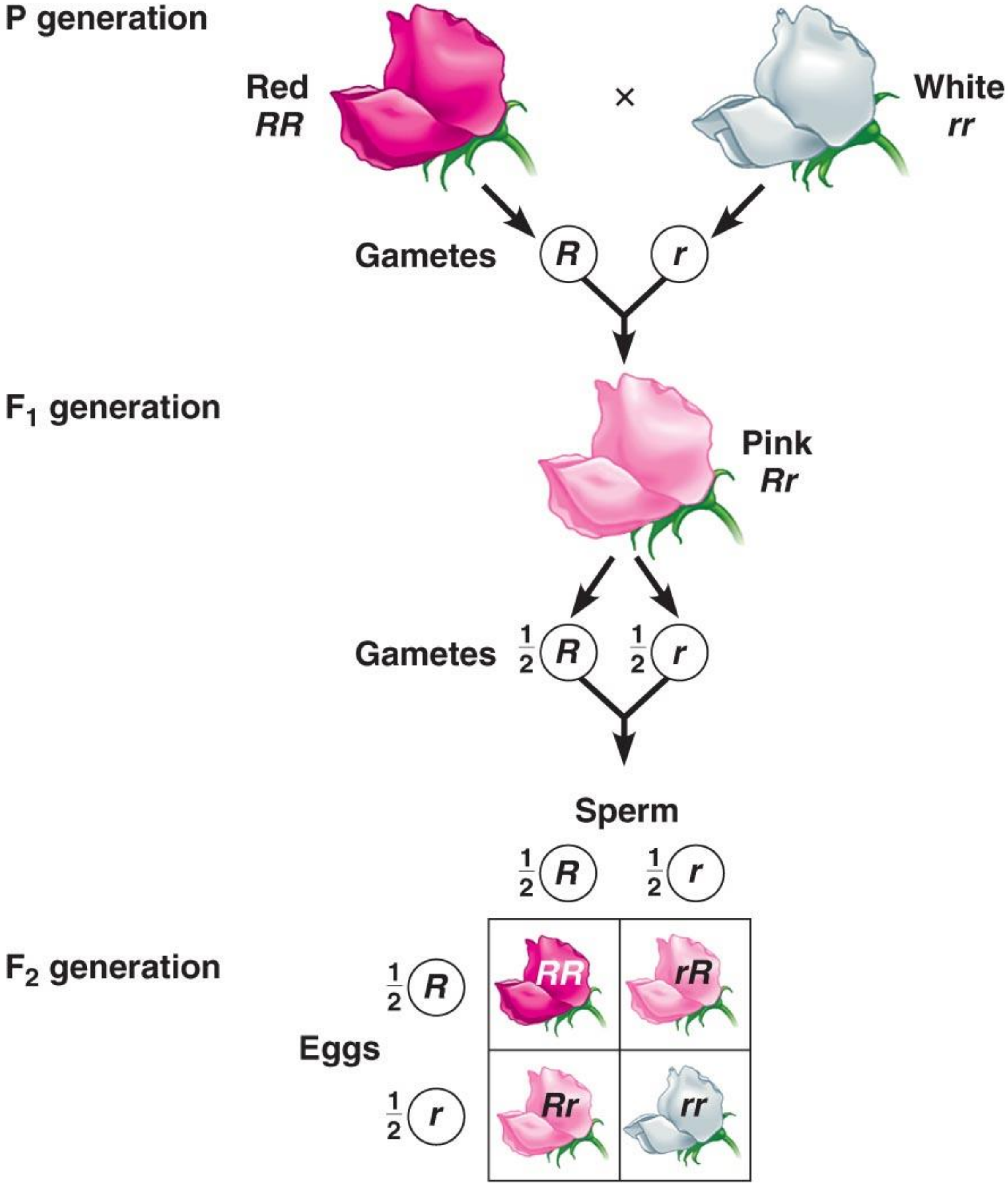


Parents (F_1) : *RrYy* x *RrYy*



9/16 yellow-round
3/16 green-round
3/16 yellow-wrinkled
1/16 green-wrinkled


RR
R
R
W
RW
RW
WW
W
RW
RW


P generation
Red
RR
×
White
rr
Gametes
R
r
F1 generation
Pink
Rr
Gametes
1/2 R
1/2 r
Sperm
1/2 R
1/2 r
F2 generation
1/2 R
Eggs
1/2 r
RR
rR
Rr
rr

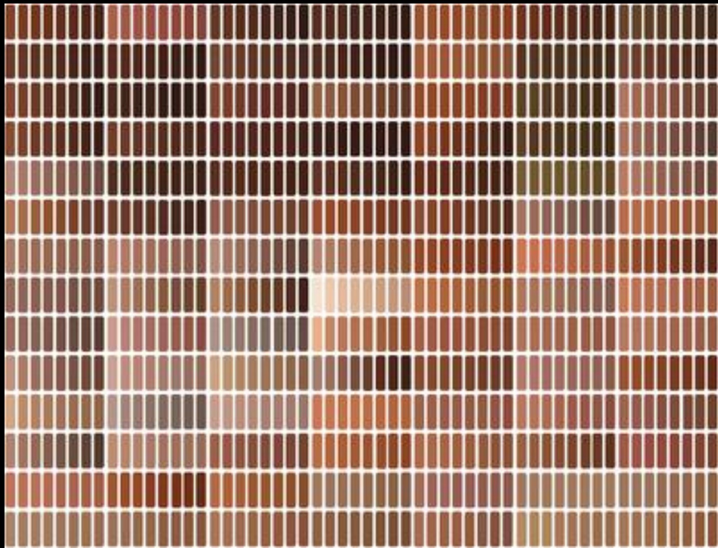
Beyond Dominant & Recessive

- Multiple Alleles: Many genes have more than two alleles.
- This does not mean that an individual can have more than two alleles.
- It only means that more than two possible alleles exist in a population.
  Ex: Blood type in humans

I^A, I^B, I^O

Polygenic Traits

- Many traits are produced by the interaction of several genes.
- Traits controlled by two or more genes are said to be polygenic traits ("having many genes")
- EX: Wide range of skin tones in humans



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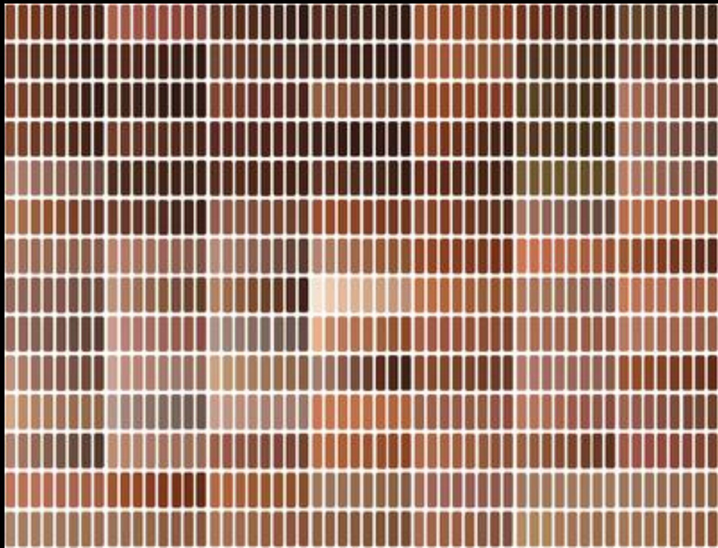
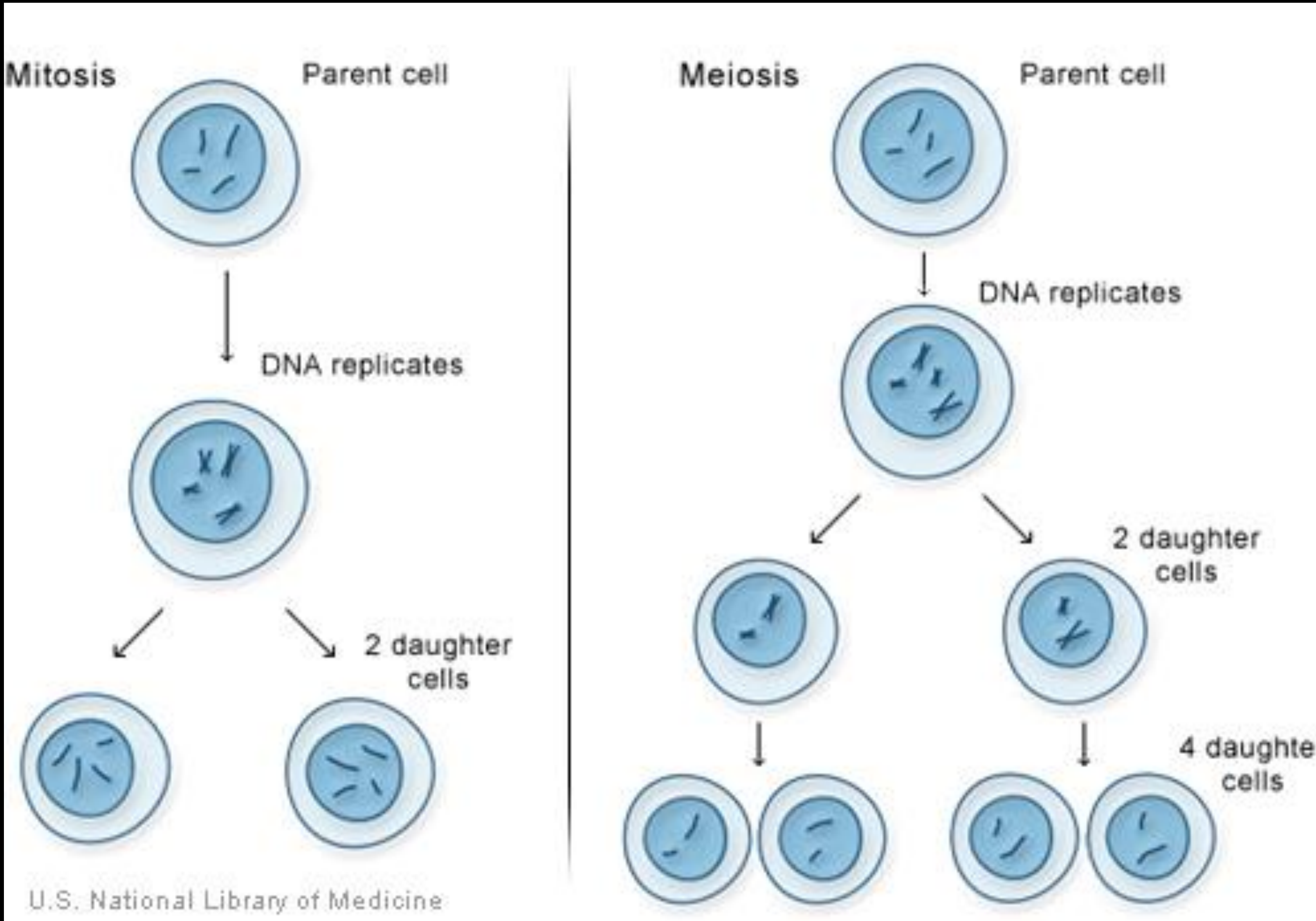
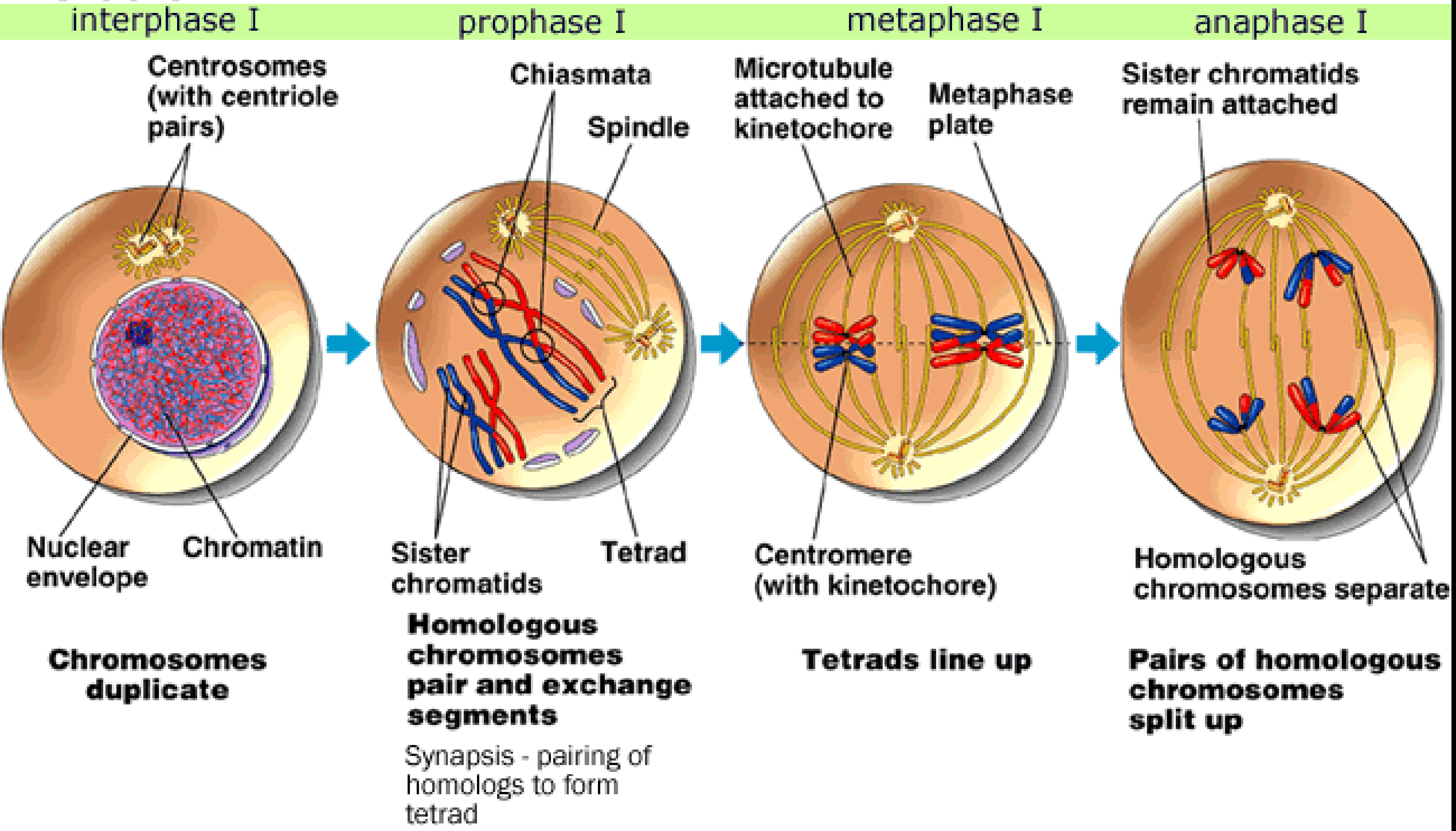
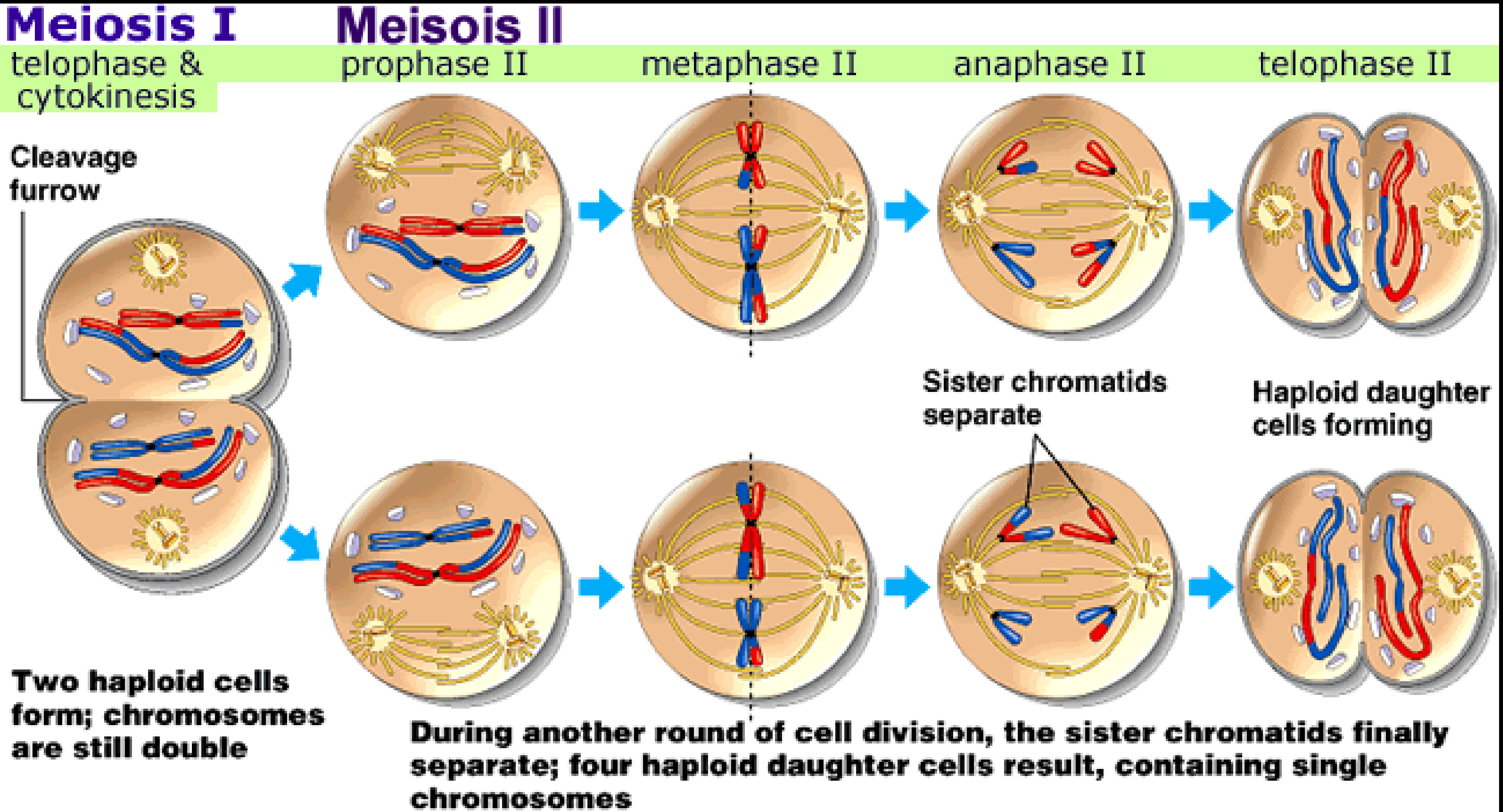




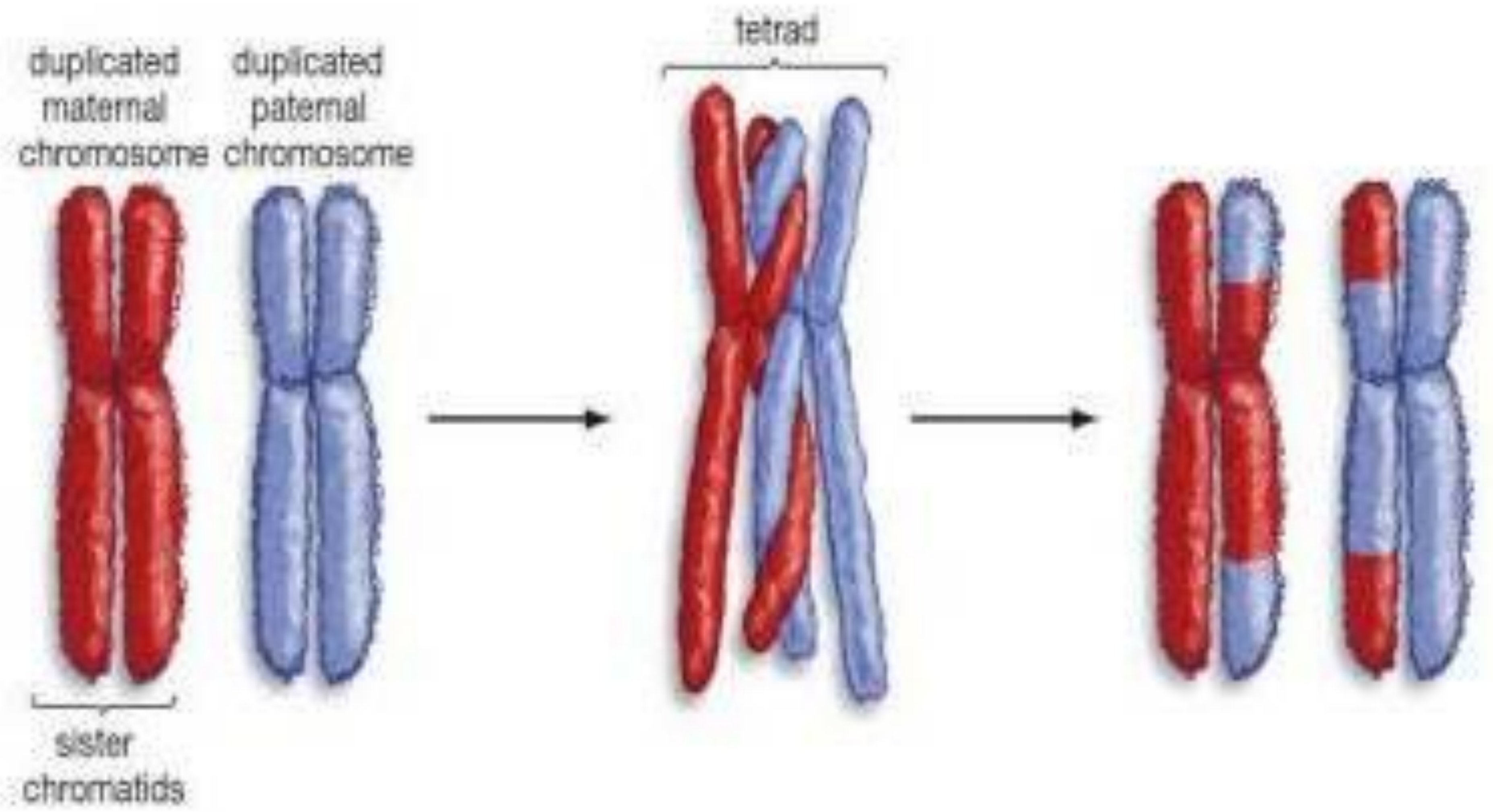
Mitosis
Parent cell
DNA replicates
2 daughter cells
Meiosis
Parent cell
DNA replicates
2 daughter cells
4 daughter cells
U.S. National Library of Medicine

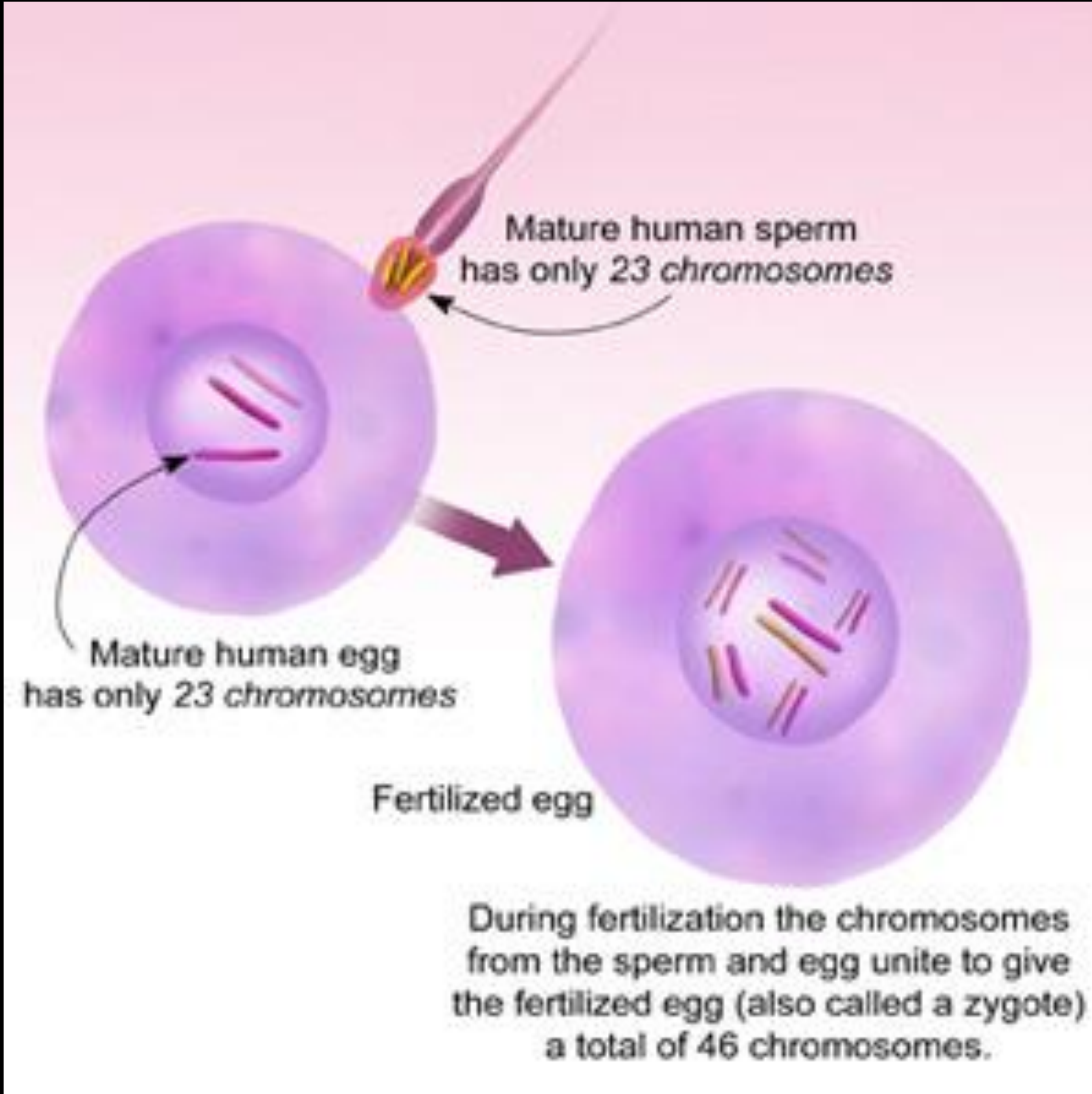
Meiosis I


interphase I
prophase I
metaphase I
anaphase I
Centrosomes (with centriole pairs)
Nuclear envelope
Chromatin
Chiasmata
Spindle
Sister chromatids
Tetrad
Microtubule attached to kinetochore
Metaphase plate
Centromere (with kinetochore)
Sister chromatids remain attached
Homologous chromosomes separate
Chromosomes duplicate
Homologous chromosomes pair and exchange segments
Synapsis - pairing of homologs to form tetrad
Tetrads line up
Pairs of homologous chromosomes split up



Meiosis I
Meisois II
telophase & cytokinesis
prophase II
metaphase II
anaphase II
telophase II
Cleavage furrow
Sister chromatids separate
Haploid daughter cells forming
Two haploid cells form; chromosomes are still double
During another round of cell division, the sister chromatids finally separate; four haploid daughter cells result, containing single chromosomes


duplicated maternal chromosome
duplicated paternal chromosome
tetrad
sister chromatids


Mature human sperm
has only *23 chromosomes*
Mature human egg
has only *23 chromosomes*
Fertilized egg
During fertilization the chromosomes from the sperm and egg unite to give the fertilized egg (also called a zygote) a total of 46 chromosomes.