Genetics

- **Genetics:** Is the study of heredity.
- Biologists study Genetics to find out what controls we can have on disease and traits that are passed on though the generations.
- **Traits** are the characteristics that may be passed on some may be visible and others may be not or difficult to see.

- **TWO TYPES OF TRAITS**: Some traits may not be expressed at all.
- **Phenotypic traits** : These are the physical characteristic or what the individual looks like.
- Ex. Color, Height, etc.

- **Genotypic traits**: What the actual genes are like.
- This may give a clue to what traits an individual organism carries regardless of their Phenotype.
- **Genes**: These are factors that control the traits.
- They are located on a chromosome and are made up of DNA.

- **Alleles**: Are different forms of a gene.
- There are Dominant and Recessive alleles

Alleles may be….

- 1.)**Homozygous** or alleles that are the same. This means that both parents gave the same gene to their offspring. (DD, dd)
- 2.) **Heterozygous** or alleles that are opposite, not the same. (Dd)

- **Dominant:** These are alleles that will be expressed when present.
- Always use a capitol letter.
- **Recessive:** Alleles are expressed only when homozygous.
- Always use a lowercase letter.

Punnet Square: this is a way of showing all the possible allele combinations.

It Gives a **probability** of what the offspring from each cross may look like.

Chromosomal Theory of Heredity

- Genes are located on the chromosomes and each occupies a specific place.
- Genes and chromosomes are inherited together. These are linked genes.
- Some genes can move or trade places to another chromosome due to crossing over

- The human chromosome has about 70,000 genes per chromosome.

Gregor Mendell

- *The Father of Genetics.* 1850's

He worked with pea plants and noticed that if he crossed peas with different characteristics that some would be passed on to the next generation.

- *Used true breeding plants that would only produce a certain trait such as color
- **He did not know** how this happens only that it did.
- Did not know about alleles, genes or chromosomes

Gregor Mendell

- **He did know:**
- **1.** That each individual had 2 chromosomes for each trait because they had to parents.
- Each gene was passed on to the next generation.
- **2**. Gametes are separate cells that have only 1 chromosome and that there must be a process that breaks the pair in two.

Gregor Mendell

- **3**. alleles for each gene are *segregated independent*ly.
- **4.** In the cases of when there are to or more forms of a single trait some forms of the gene may be dominant or recessive.

Gregor Mendell

- **Purebred:** If self - pollinated, the offspring will have the same traits as the parents. (AKA: Homozygous) (T T)
-
- **Hybrids:** Organisms produce by crossing parents with different
- characteristics. (T t) (AKA: Heterozygous)
- **Genes:** The heredity material that determines a trait. (Found on the
- Chromosomes) (DNA = the chemical found in the genes)

Gregor Mendell

- **<u>Dominance:</u>** Some alleles are dominant. Tall alleles are dominant
- over short alleles. (T / t). (Dominance does NOT apply to all genes).
- **<u>Incomplete Dominance / Codominance:</u>** Neither allele is dominant. (A red flower parent and a white flower parent = a pink flower) (Rr = pink).

Incomplete dominance: the heterozygote is intermediate in phenotype between the 2 homozygotes.

Red crossed with white makes pink.

Codominance: the heterozygote shows some aspect of the phenotypes of both homozygotes.

Black crossed with white makes gray.

Gregor Mendell

- **Recessive:** The allele that is not dominant. If two recessive allele are
- present, then and only then will that trait be present.
- **P generation:** The parents.
-
- **F1:** The offspring of the P generation.
-
- **F2:** The offspring of the F1 generation.

Gregor Mendell

- Mendel's Experiment:
- Purebred Tall (TT) Crossed with Purebred Short (tt)
-
- F1 All tall plants (Tt) (Hybrids)
- F2 Three tall One short. (3:1 ratio)
- TT, Tt, Tt, tt

Thomas Morgan

1900's He expanded on the principles of Mendel by working with animals. Drosophila or Fruit Flies.

- He Proved that mieotic division works in animals.

Monohybrid Crosses

Monohybrid cross: a cross to study only 2 variations of a single trait

Mendel produced true-breeding pea strains for 7 different traits

-each trait had 2 alternate forms (variations)

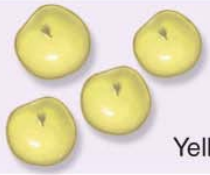
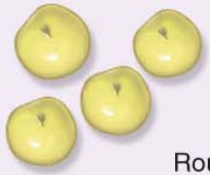
-Mendel cross-fertilized the 2 true-breeding strains for each trait

Monohybrid Crosses

F_1 generation (1st filial generation): offspring produced by crossing 2 true-breeding strains

For every trait Mendel studied, all F_1 plants resembled only 1 parent

-no plants with characteristics intermediate between the 2 parents were produced

Dominant	Recessive	F_2 Generation
1. Flower Color		
 Purple	X  White	705 Purple: 224 White 3.15:1
2. Seed Color		
 Yellow	X  Green	6022 Yellow: 2001 Green 3.01:1
3. Seed Texture		
 Round	X  Wrinkled	5474 Round: 1850 Wrinkled 2.96:1
4. Pod Color		
 Green	X  Yellow	428 Green: 152 Yellow 2.82:1

Dominant	Recessive	F_2 Generation
5. Pod Shape		
 Inflated	X  Constricted	882 Inflated: 299 Constricted 2.95:1
6. Flower Position		
 Axial	X  Terminal	651 Axial: 207 Terminal 3.14:1
7. Plant Height		
 Tall	X  Short	787 Tall: 277 Short 2.84:1

Monohybrid Crosses

F_1 generation: offspring resulting from a cross of true-breeding parents

F_2 generation: offspring resulting from the self-fertilization of F_1 plants

dominant: the form of each trait expressed in the F_1 plants

recessive: the form of the trait not seen in the F_1 plants

F_2 plants exhibited both forms of the trait in a very specific pattern:

¾ plants with the dominant form

¼ plant with the recessive form

The dominant to recessive ratio was 3 : 1.

Mendel discovered the ratio is actually:

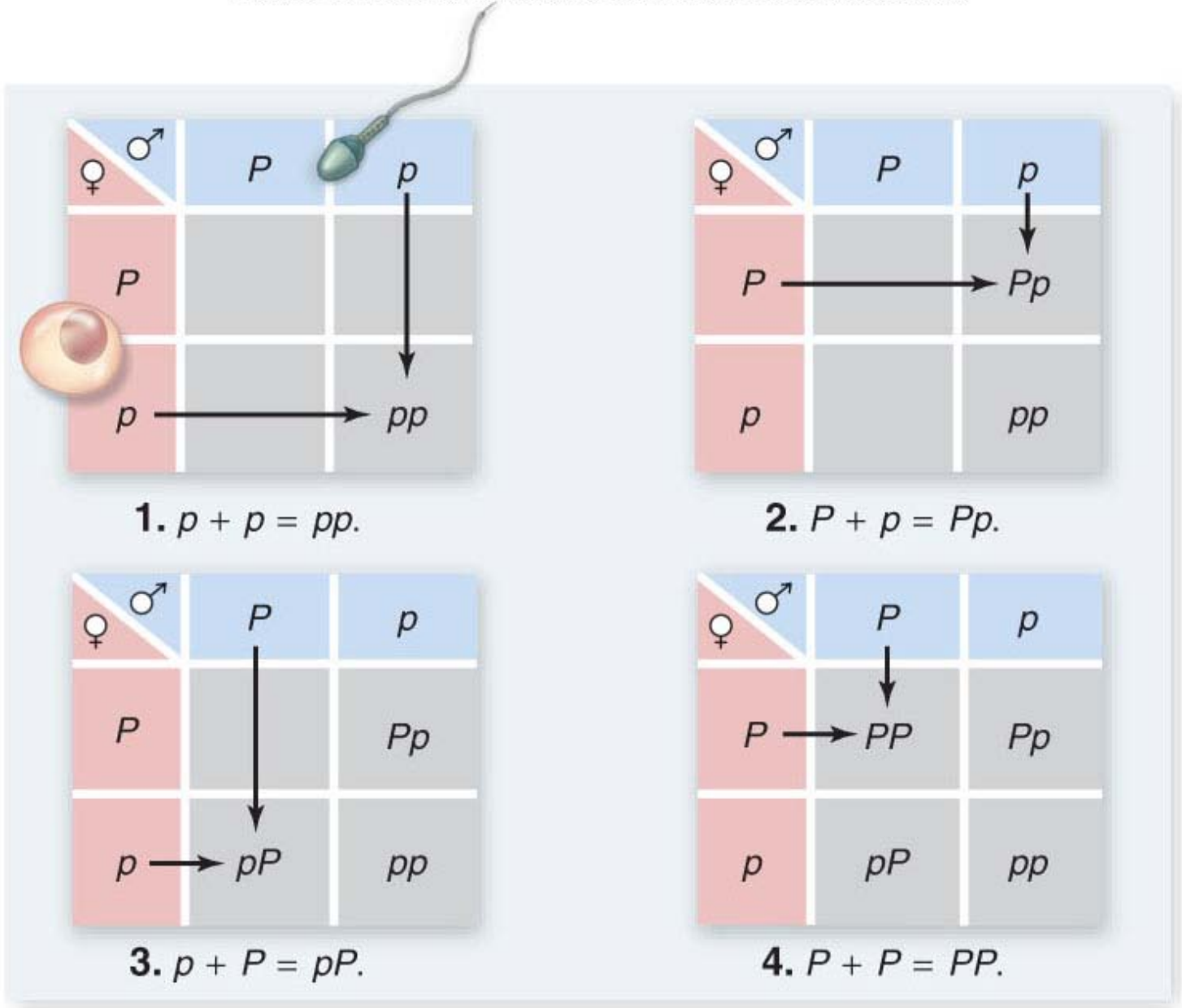
1 true-breeding dominant plant

2 not-true-breeding dominant plants

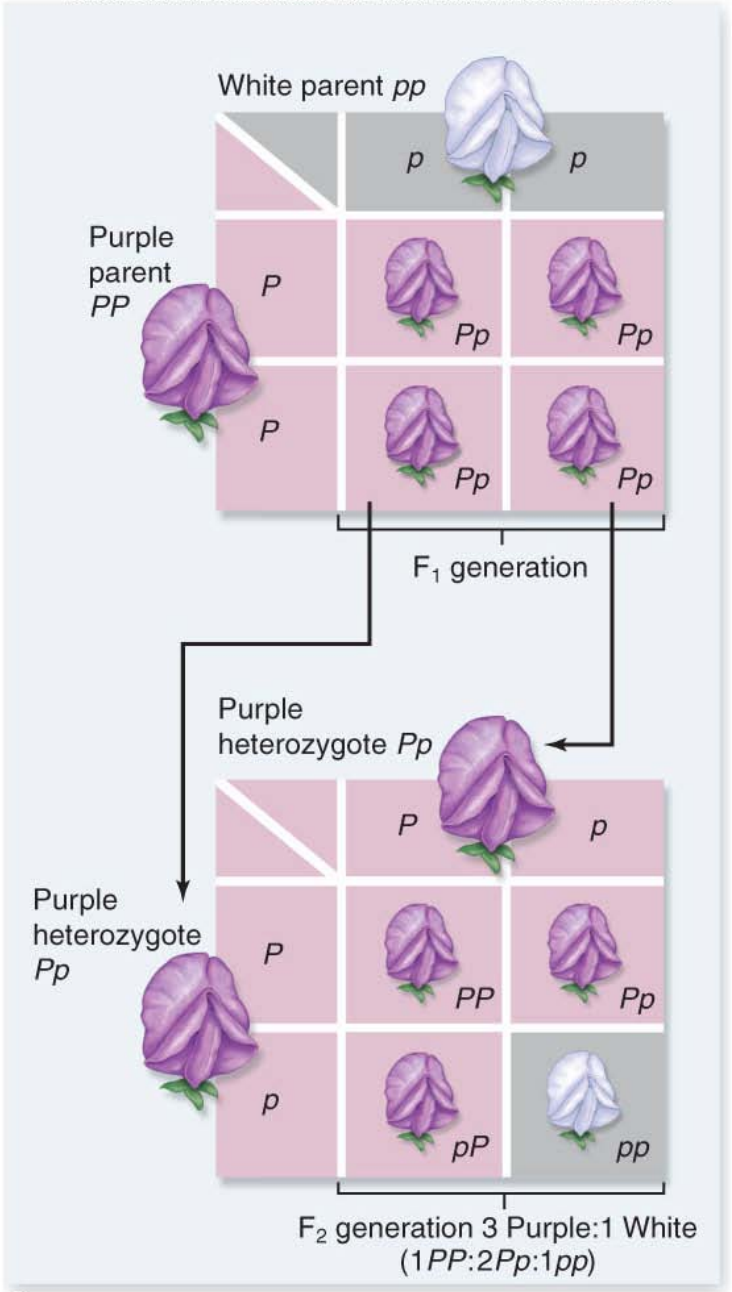
1 true-breeding recessive plant

Principle of Segregation

Two alleles for a gene segregate during gamete formation and are rejoined at random, one from each parent, during fertilization.



a.



b.

Monohybrid Crosses

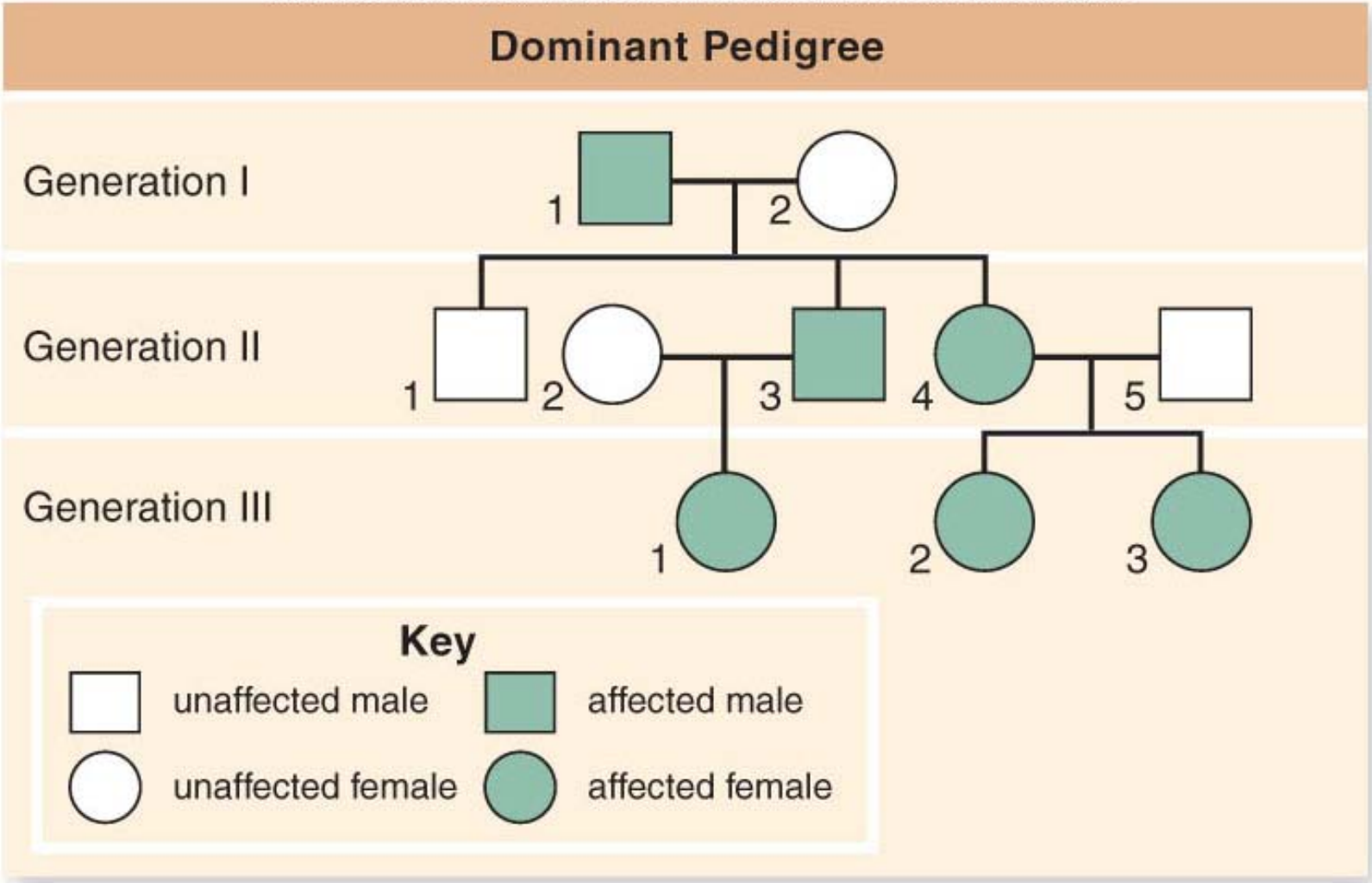
Some human traits are controlled by a single gene.

-some of these exhibit dominant inheritance

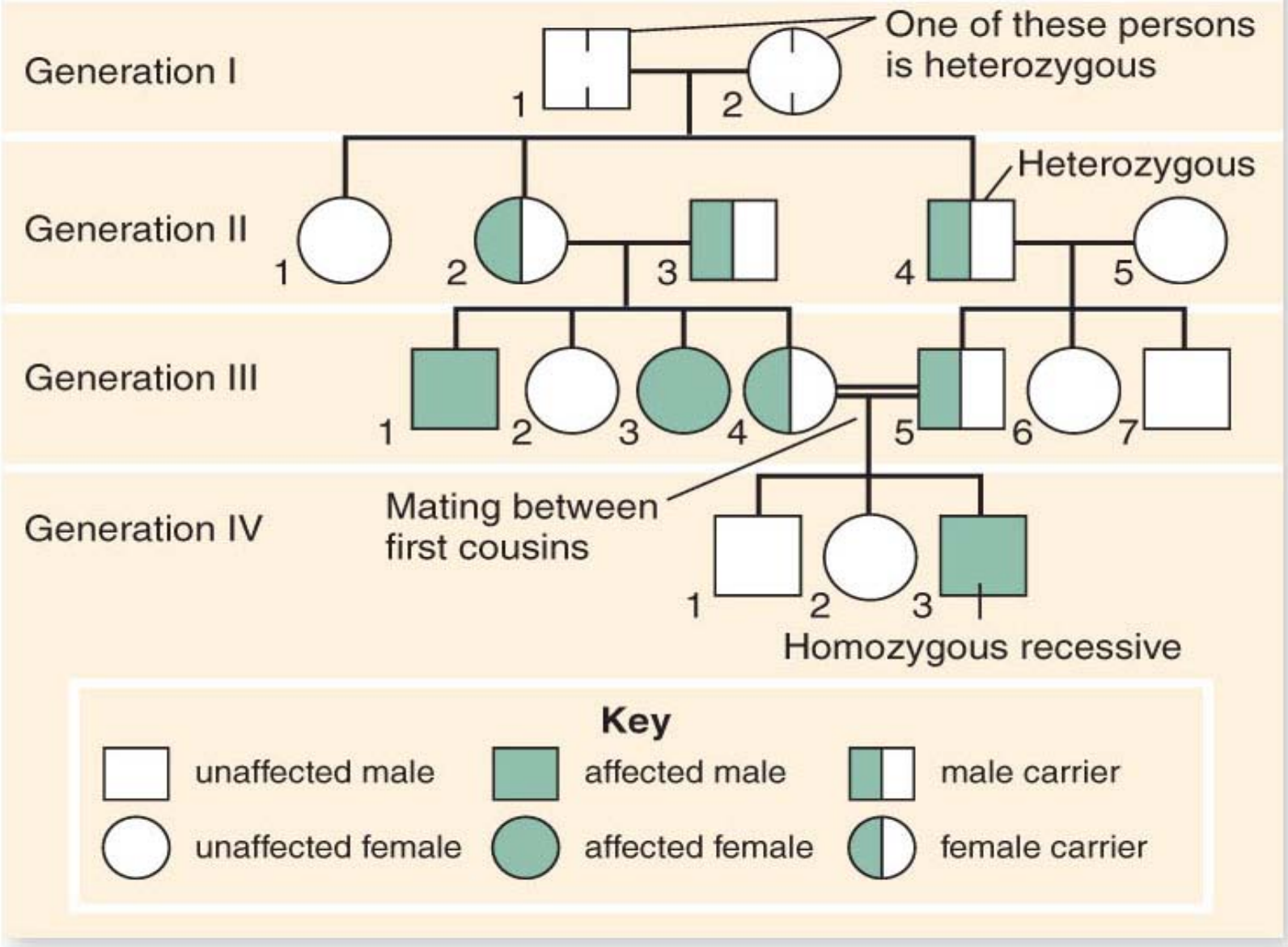
-some of these exhibit recessive inheritance

Pedigree analysis is used to track inheritance patterns in families.

Dominant Pedigree



Recessive Pedigree



Dihybrid Crosses

Dihybrid cross: examination of 2 separate traits in a single cross

-for example: RR YY x rryy

The F_1 generation of a dihybrid cross (RrYy) shows only the dominant phenotypes for each trait.

Dihybrid Crosses

The F_2 generation is produced by crossing members of the F_1 generation with each other or allowing self-fertilization of the F_1.

-for example RrYy x RrYy

The F_2 generation shows all four possible phenotypes in a set ratio:

9 : 3 : 3 : 1

CELL DIVISION

- **A cell divides and produces two identical cells.** (a mother cell splits in half to make two daughter cells that are **genetically identical).**

CELL DIVISION

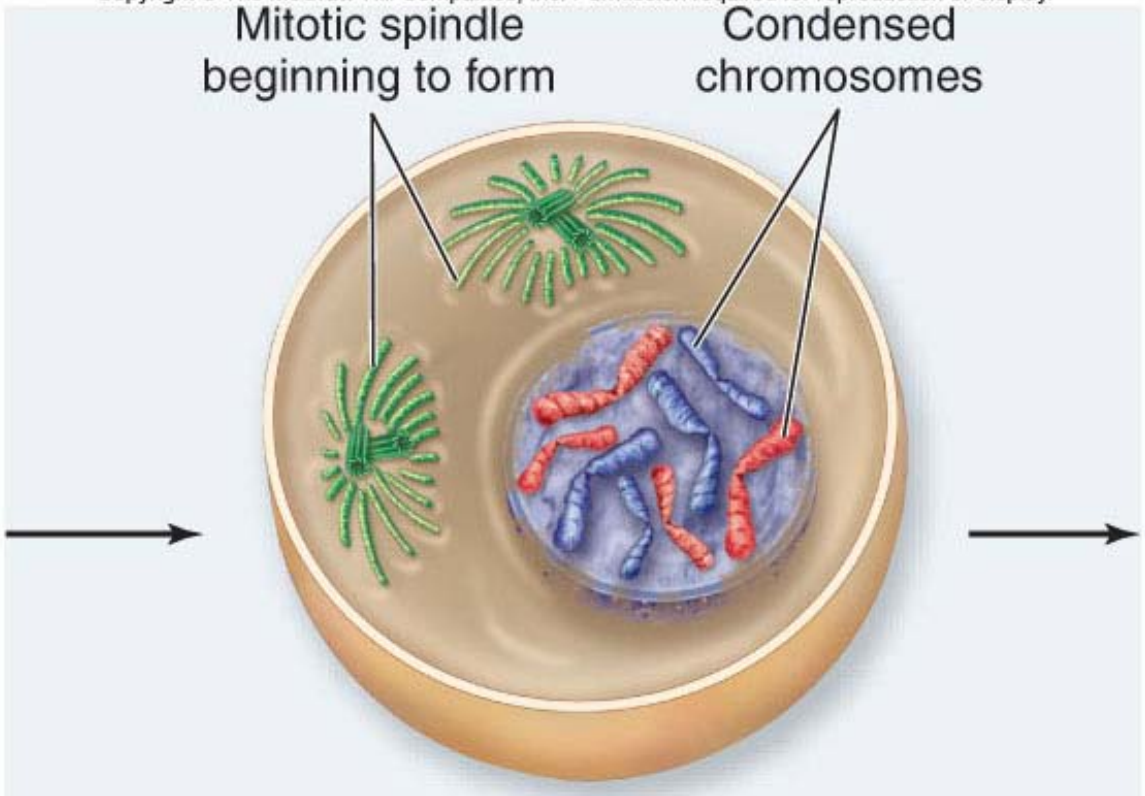
- **Mitosis:** The process in which the nucleus divides to produces two identical nuclei.
- **Cytokinesis:** The process in which the cytoplasm divides to produces two new cells.

Mitosis: (five phases)

- **1. Interphase:** The period between cell division. (can be a long time).
- The nucleus is active in producing messenger RNA. Proteins are made, and DNA is copied.

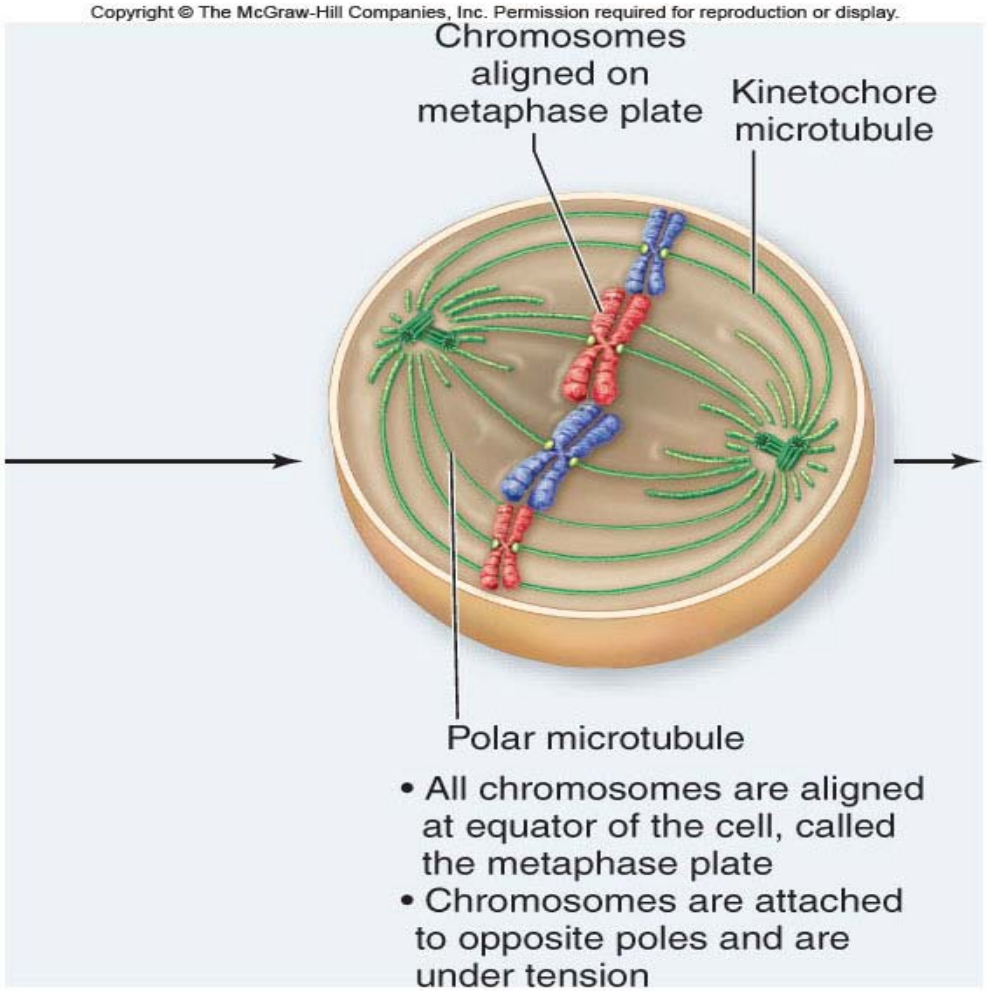
Mitosis

- **2. Prophase:** Nuclear envelope disappears. chromosomes become visible. Centrioles move to opposites sides of the cell and form spindles (threads). Chromosomes replicate to form two chormatids connected in the middle by centromere.

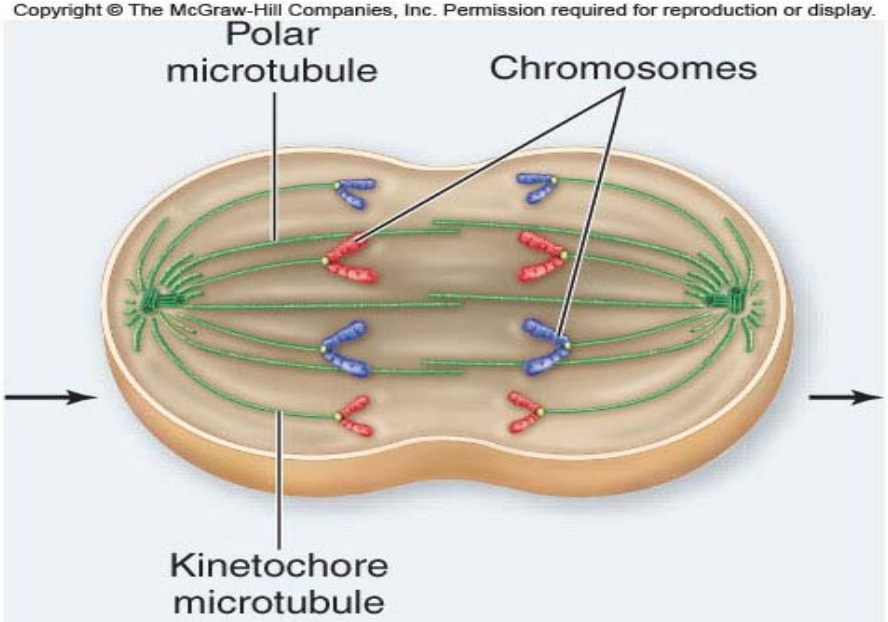


- Chromosomes condense and become visible
- Chromosomes appear as two sister chromatids held together at the centromere
- Cytoskeleton is disassembled: spindle begins to form
- Golgi and ER are dispersed
- Nuclear envelope breaks down

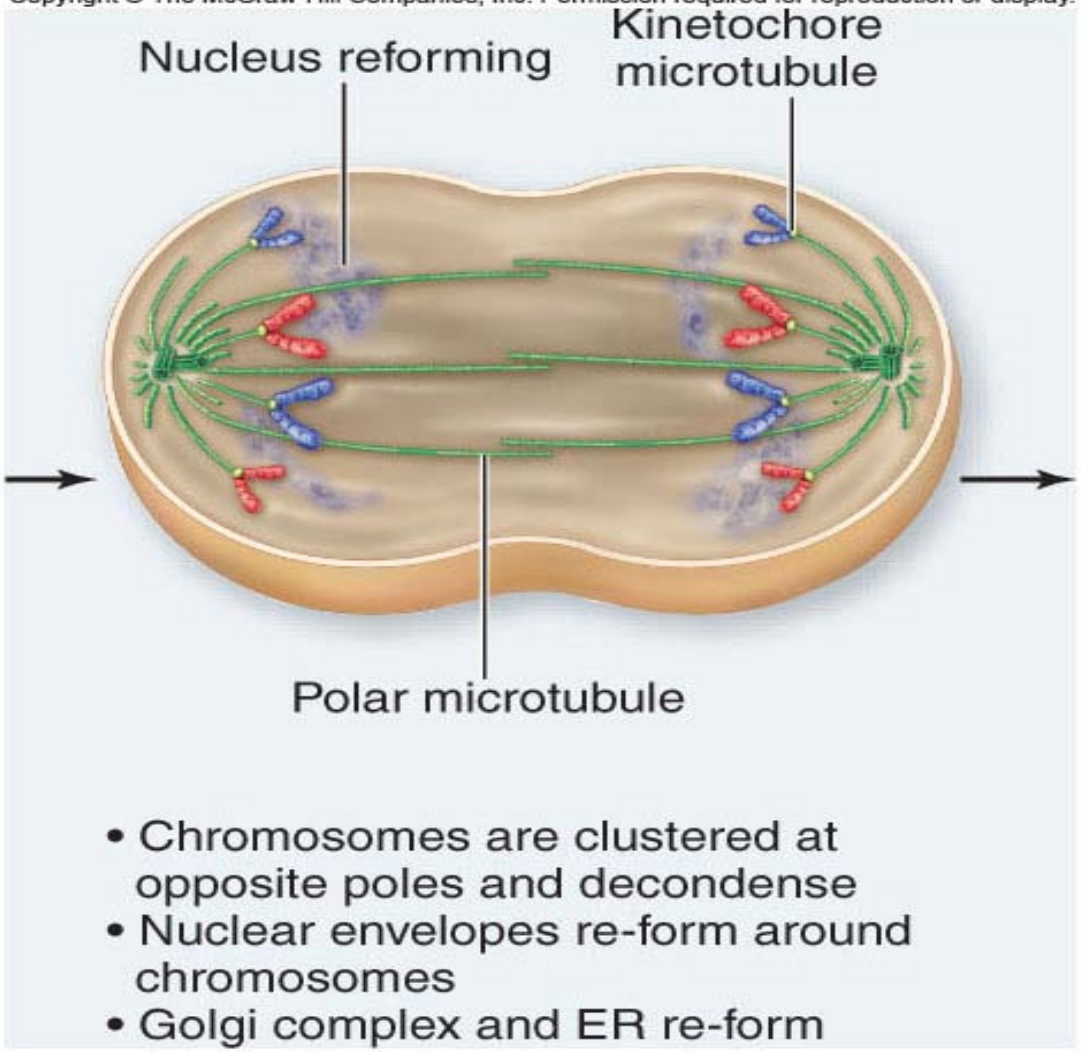
- **3. <u>Metaphase:</u>** chromososmes line up in the middle of the cell. the spindles from the centrioles connect to the centromeres.



- **4. <u>Anaphase:</u>** the two chromatids are pulled apart to opposites sides of the cell by the spindles. (once the chromatids are pulled apart, they are called chromosomes).



- **5. Telephase:** The final phase of mitosis. The chromosomes located at both sides of the cell uncoil and nuclear envelope reforms.



- **<u>Cytokinesis:</u>**
- Once the genetic material is divided in two (mitosis), the cell (Cytoplasm) now splits into two daughter cells.
- In animals:; The cell membrane pinches in the middle to form two new cells.
- In plants: A cell plates forms in the middle of the cell. A cell wall will form at the cell plate to form two new cells.

Cell Growth and Division

- Cells can grow at astonishing rates.
- Some cells, like E-coli, can double their volume in 30 minutes and divide in 30 minutes
- Ideal conditions of cell growth can never be maintained for very long.
- **Cells grow until they come into contact with other cells.*

Cell Growth and Division

- Cell Division can be turned *off* and *on*.
- Turns *on* when we have an injury, cut or bone break.
- Cells at edge of injury grow fast - growth slows down as the injury becomes healed.

Cell Growth and Division

- Some cells divide and grow very rapidly and some rarely divide.
- Skin cells & cells of the Digestive Tract grow and divide throughout our life.
- Cells of our Heart and Nervous System will do this only rarely.

Uncontrolled Cell Growth

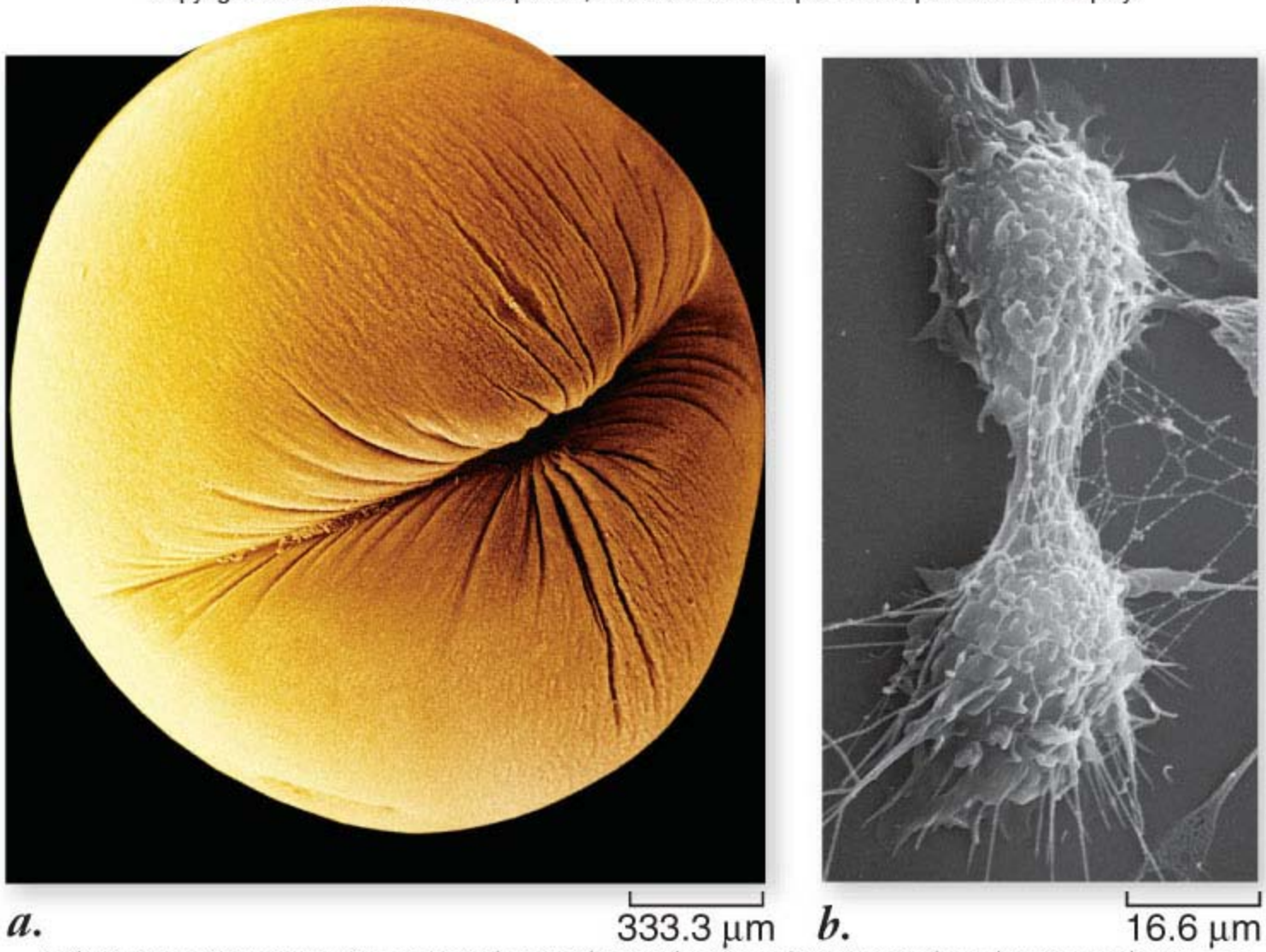
- Cancer is caused by uncontrolled cell growth.
- The cell growth is controlled by DNA.
- The DNA is changed by viruses in many cases, causing the cell's chromosomes to be changed, altering their normal functions.
- - These cells do not stop dividing when they come in contact with each other.

Chromosomes

- Chromosomes = Structures that contain genetic information.
- - Means colored body because when dye is added the Chromosomes pick up the color so we can see them.
- - Made of material called Chromatin which is made up of DNA and Protein.
- - Humans contain 46 chromosomes - 23 from each parent.

Chromosome Structure

- Made up of two Chromatids. these are the large thread structures. Each Chromosome has 2.
- The Chromatids are attached at an area called the Centromere.



(left): © Dr. David M. Phillips/Visuals Unlimited; (right): Guenter Albrecht-Buehler, Northwestern University, Chicago

Protein Synthesis

- The nitrogenous bases in DNA contains
- information that directs Protein Synthesis.

Protein Synthesis

- Proteins are very versatile molecules and most enzymes are proteins
- 1. They control biochemical pathways in the cell
- 2. Make lipids (fats)
- 3. Make carbohydrates
- 4. Make nucleotides
- 5. Responsible for cell movement and cell structure

Protein Synthesis

- So the genetic code must have a four-letter alphabet. More than one nucleotide must make up
- the code word, for each amino acid.
- The code words of the DNA nucleotides are copied onto a strand to **mRNA**.

- Each combination of 3 nucleotides on **mRNA** is called a codon.
-
- Each codon specifies an amino acid that is placed on polypeptides.

Protein Synthesis

Translation

- Translation is the decoding of a messenger RNA message into a polypeptide chain. (Protein)
- 2 other types of RNA are involved here.
- They are transfer RNA (mRNA) and ribosomal RNA (r RNA)

Protein Synthesis

Translation

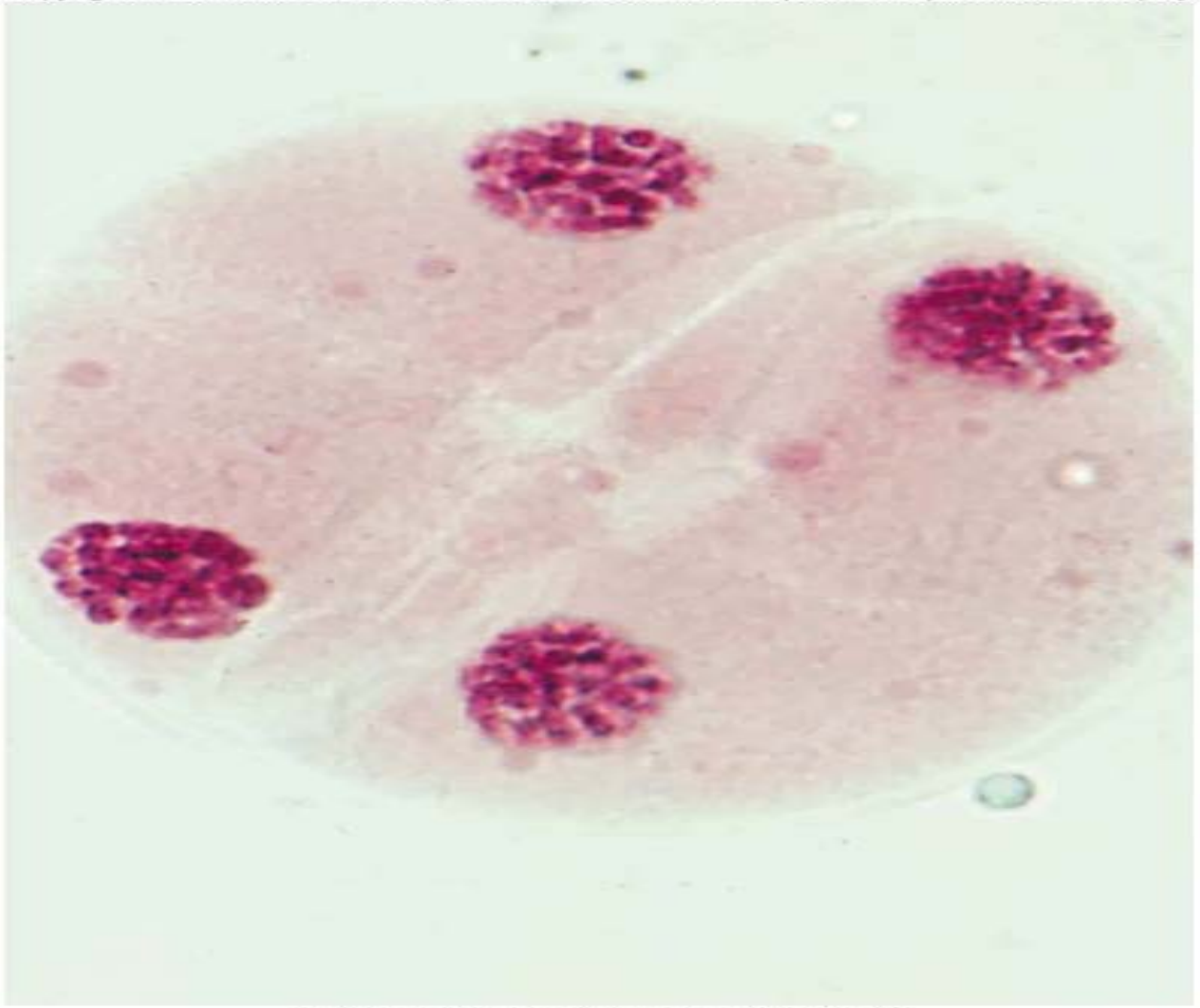
- **t RNA** carries the amino acids to the ribosomes.
- r RNA makes up most of the ribosome.
- The role of transfer RNA = can form a loop.
- The 3 exposed bases on t RNA will pair up with the 3 base pairs of mRNA.

Protein Synthesis

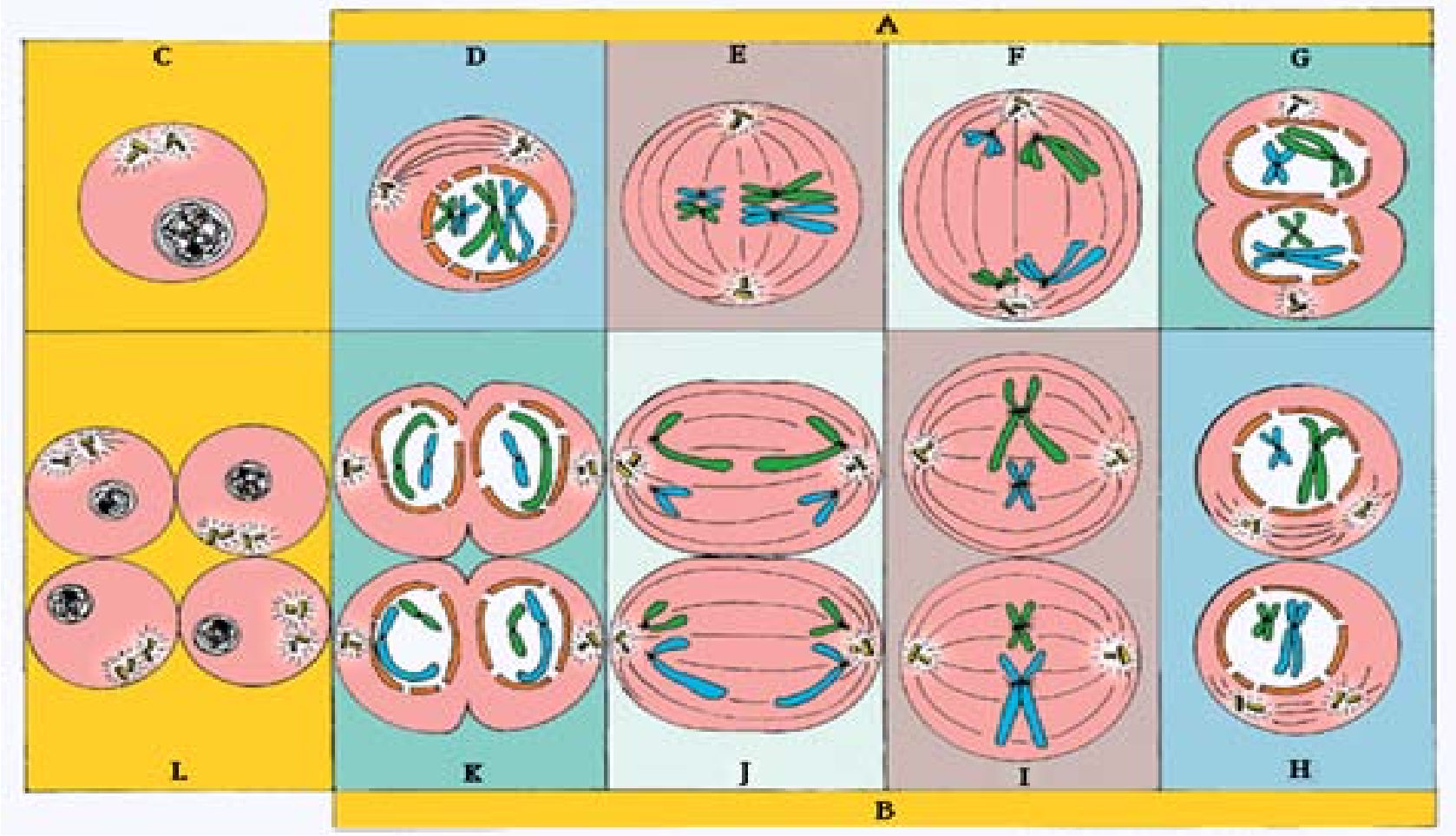
Translation

- 3 bases of **tRNA** are called **anticodons**.
- 3 bases of **mRNA** are called **codons.**
- Once the mRNA bases are paired with the t RNA bases the proper amino acid is attached to the t RNA.
- Role of Ribosomes - Acts as a cite for **protein synthesis.**
- Each ribosome is made up of **r RNA** and **proteins**.

Sexual Reproduction and Meiosis



© Clare A. Hasenkampf/Biological Photo Service.



A meiosis I, *B* meiosis II, *C* interphase, *D* prophase I, *E* metaphase I, *F* anaphase I, *G* telophase I, *H* prophase II, *I* metaphase II, *J* anaphase II, *K* telophase II, *L* four haploid cells

Dihybrid Crosses

Principle of Independent Assortment

In a dihybrid cross, the alleles of each gene assort independently.

The Process of Meiosis

Prophase I:

-chromosomes coil tighter

-nuclear envelope dissolves

-homologues become closely associated in synapsis

-crossing over occurs between non-sister chromatids

The Process of Meiosis

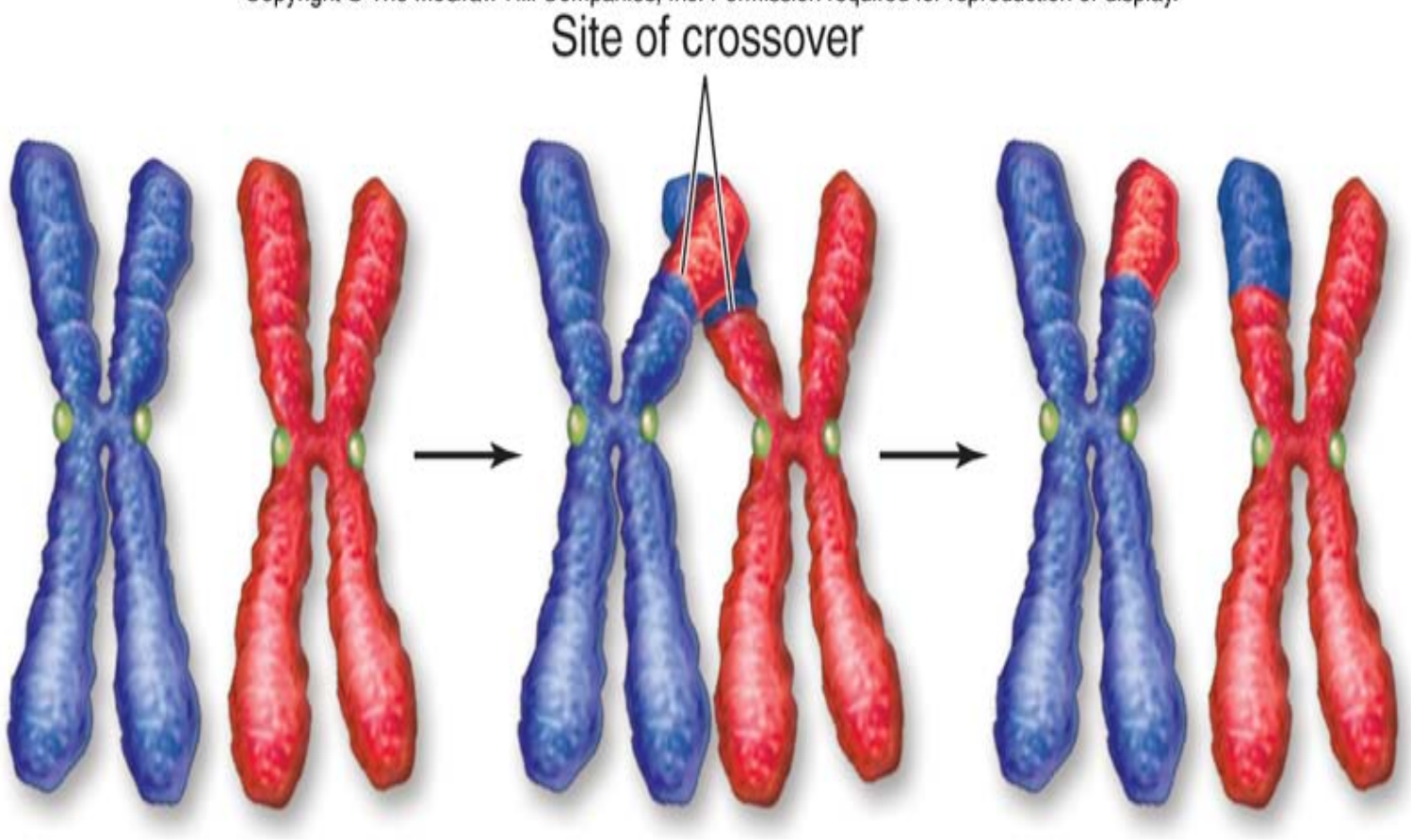
Prophase I:

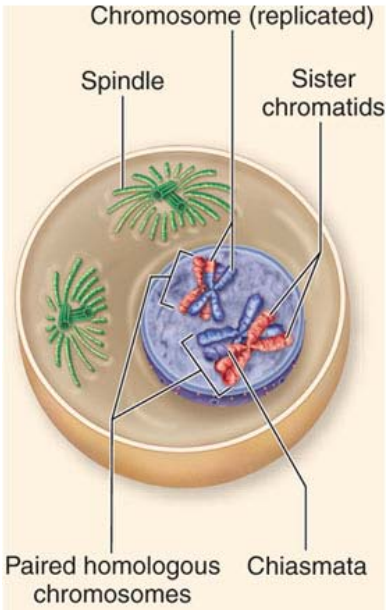
-chromosomes coil tighter

-nuclear envelope dissolves

-homologues become closely associated in synapsis

-crossing over occurs between non-sister chromatids





In prophase I of meiosis I, the chromosomes begin to condense, and the spindle of microtubules begins to form. The DNA has been replicated, and each chromosome consists of two sister chromatids attached at the centromere. In the cell illustrated here, there are four chromosomes, or two pairs of homologues. Homologous chromosomes pair up and become closely associated during synapsis. Crossing over occurs, forming chiasmata, which hold homologous chromosomes together.

The Process of Meiosis

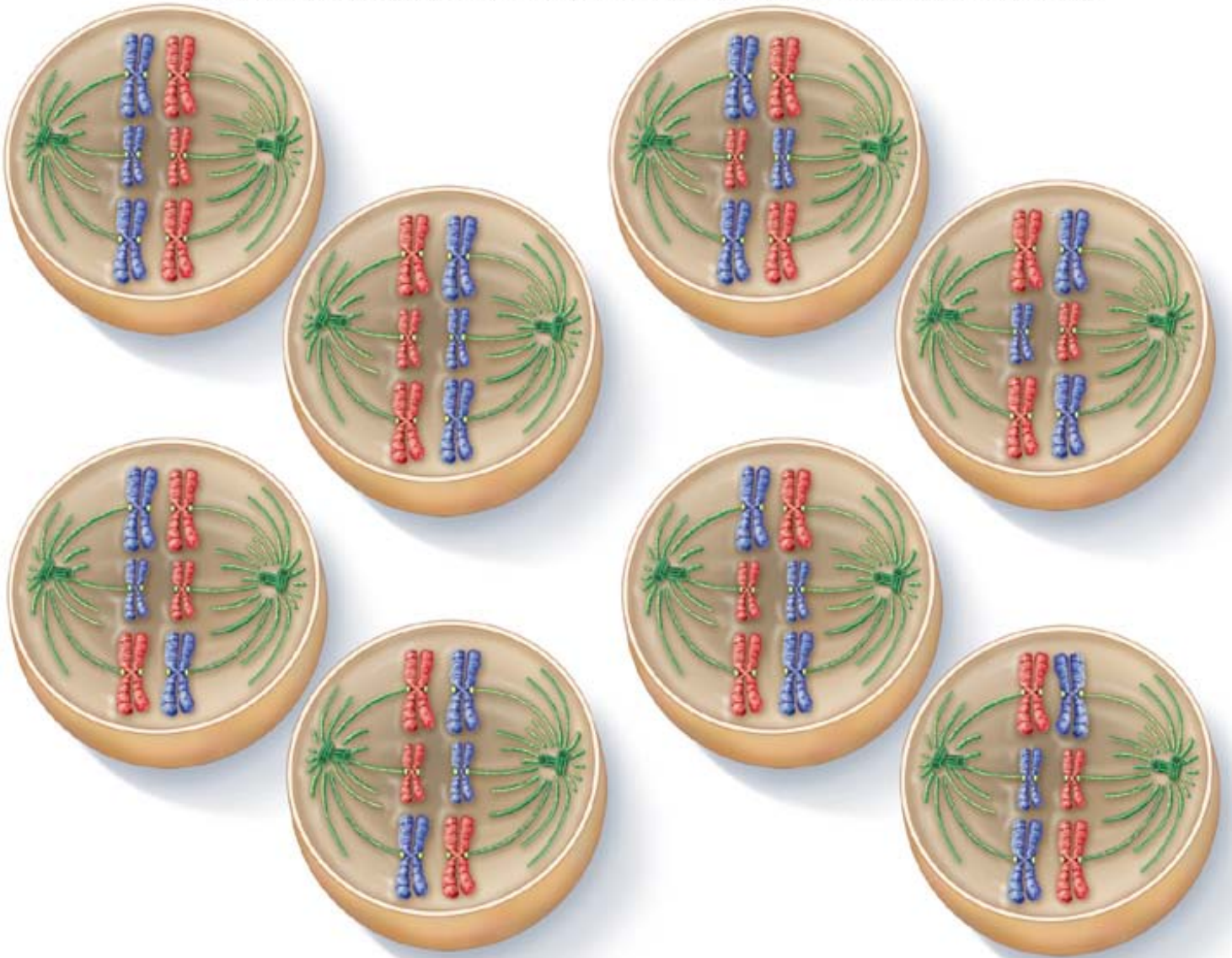
Metaphase I:

-terminal chiasmata hold homologues together following crossing over

-microtubules from opposite poles attach to each *homologue*, not each sister chromatid

-homologues are aligned at the metaphase plate side-by-side

-the orientation of each pair of homologues on the spindle is random



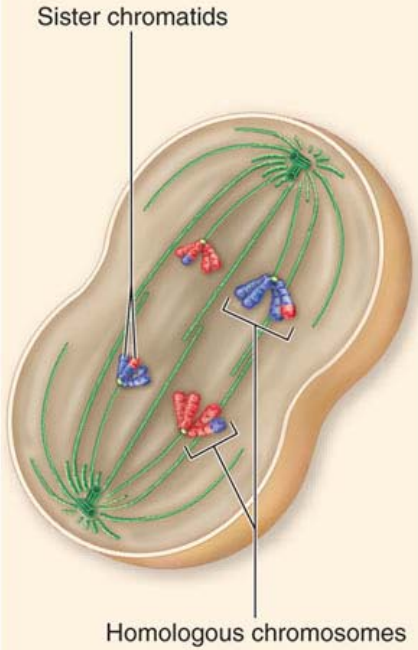
The Process of Meiosis

Anaphase I:

-microtubules of the spindle shorten

-homologues are separated from each other

-sister chromatids remain attached to each other at their centromeres



In anaphase I, kinetochore microtubules shorten, and homologous pairs are pulled apart. One duplicated homologue goes to one pole of the cell, while the other duplicated homologue goes to the other pole. Sister chromatids do not separate.This is in contrast to mitosis, where duplicated homologues line up individually on the metaphase plate, kinetochore microtubules from opposite poles of the cell attach to opposite sides of one homologue's centromere, and sister chromatids are pulled apart in anaphase.

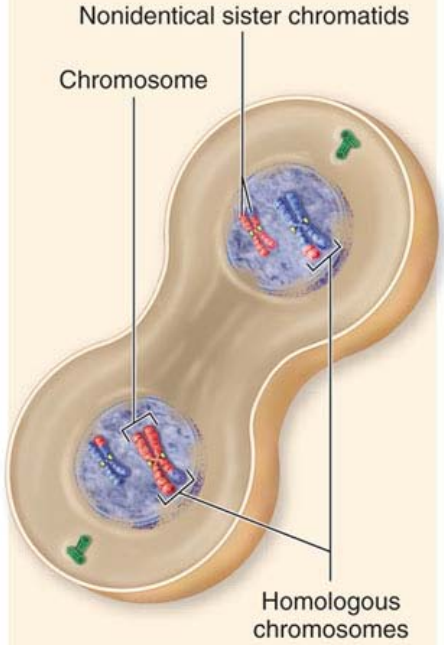
The Process of Meiosis

Telophase I:

-nuclear envelopes form around each set of chromosomes

-each new nucleus is now haploid

-sister chromatids are no longer identical because of crossing over



In telophase I, the separated homologues form a cluster at each pole of the cell, and the nuclear envelope re-forms around each daughter cell nucleus. Cytokinesis may occur. The resulting two cells have half the number of chromosomes as the original cell: In this example, each nucleus contains two chromosomes (versus four in the original cell). Each chromosome is still in the duplicated state and consists of two sister chromatids, but sister chromatids are not identical because crossing over has occurred.

Meiosis II resembles a mitotic division:

-prophase II: nuclear envelopes dissolve and spindle apparatus forms

-metaphase II: chromosomes align on metaphase plate

-anaphase II: sister chromatids are separated from each other

-telophase II: nuclear envelope re-forms; cytokinesis follows

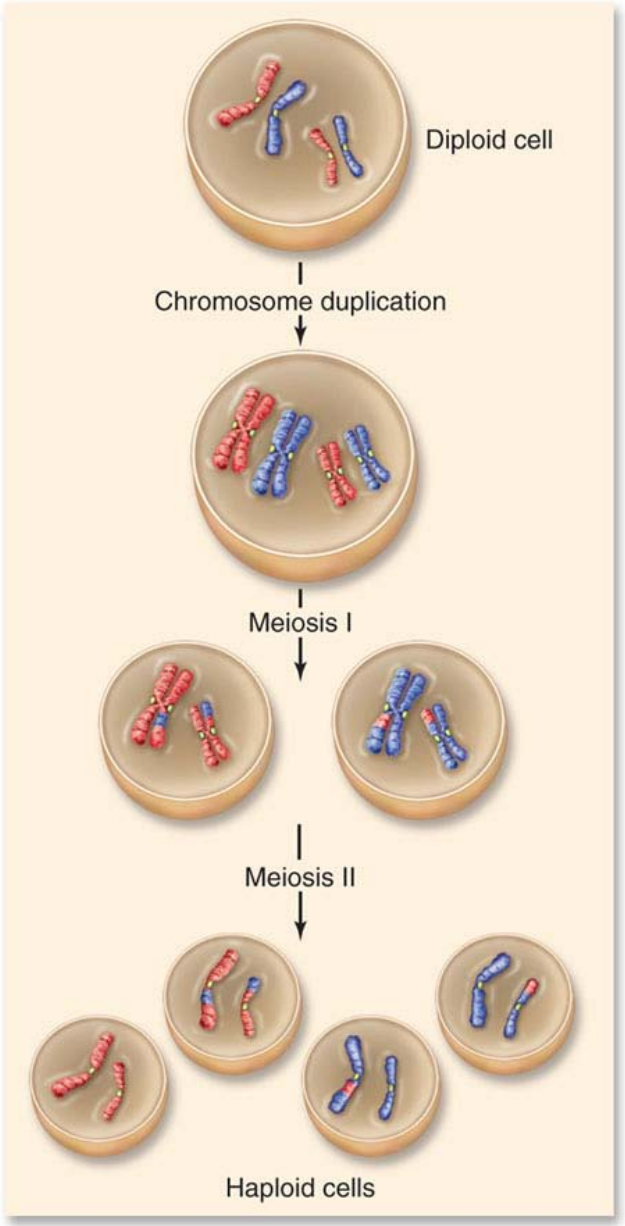
Meiosis is a form of cell division that leads to the production of **gametes**.

gametes: egg cells and sperm cells

-contain half the number of chromosomes of an adult body cell

Adult body cells (**somatic cells**) are **diploid**, containing 2 sets of chromosomes.

Gametes are **haploid**, containing only 1 set of chromosomes.

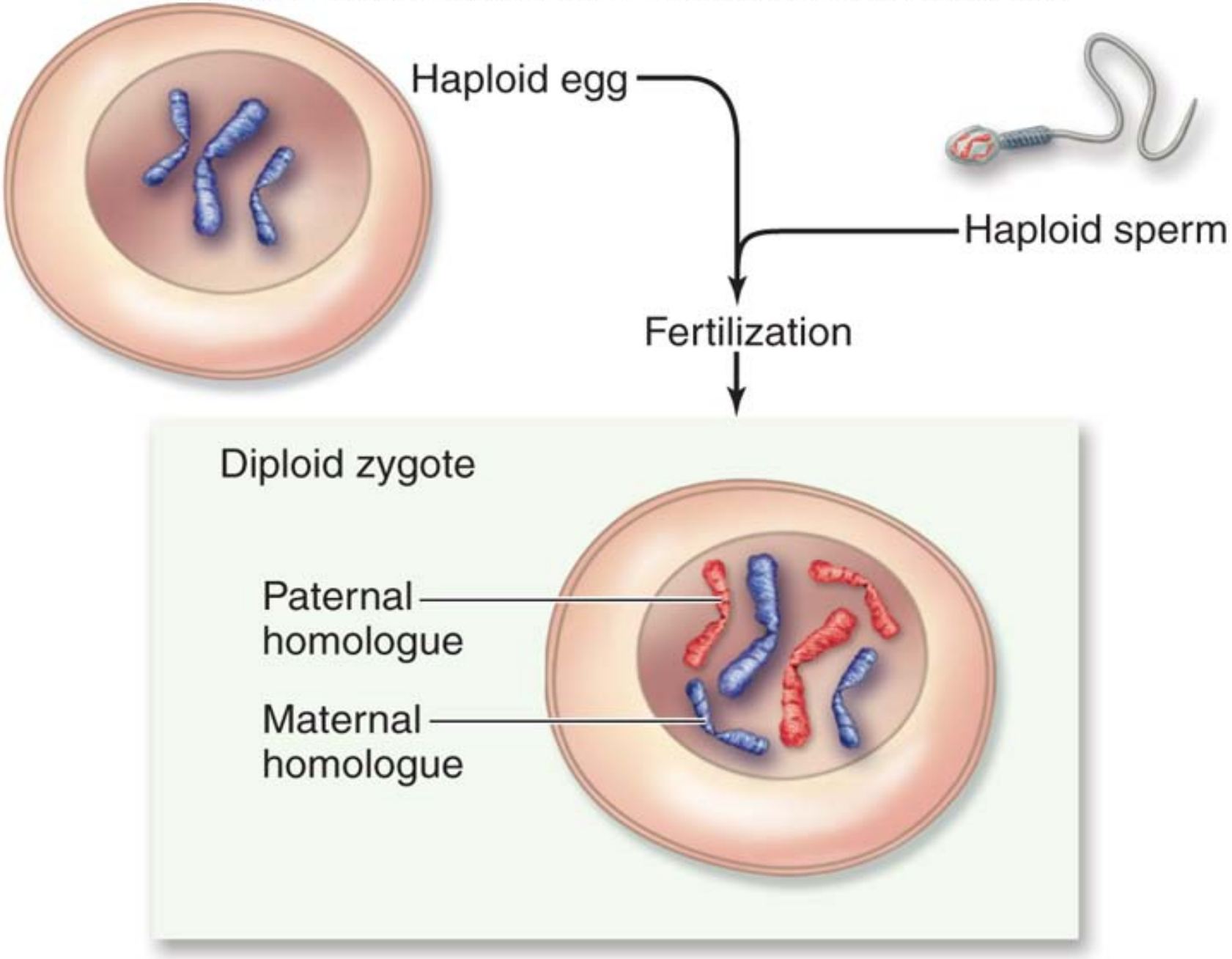


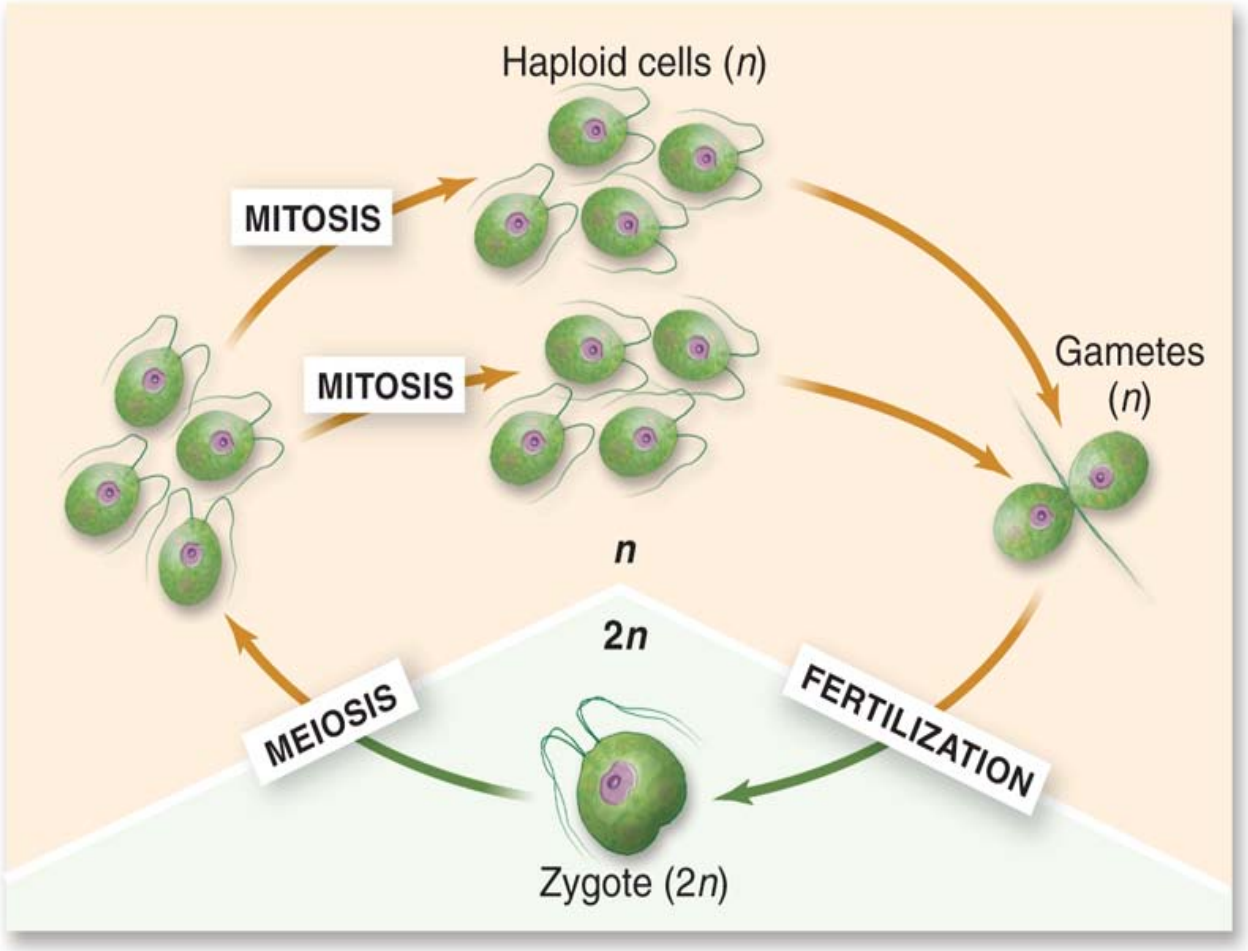
c.

Sexual reproduction includes the fusion of gametes (**fertilization**) to produce a diploid **zygote**.

Life cycles of sexually reproducing organisms involve the alternation of haploid and diploid stages.

Some life cycles include longer diploid phases, some include longer haploid phases.





a. Algae and fungi

Mutations

- **Mutations:** Changes that occur to the chromosomes.
-
- -These can be both good or bad.
-
- -Most Mutations are never shown.
-
- -These can occur in any cell that divides.
-
- -Mutations can eventualy lead to changes in the entire population over many years.

Chromosomal Mutations

- Change in the number or structure of chromosomes.
-
- -These are mutations that can involve the entire chromosome, on part or even pairs of chromosomes.

Four types of Chromosomal Mutations:

- 1. Deletion: A loss of part of the chromosome.
-
- 2. Duplication: Segment of chromosome is repeated.
-
- 3. Inversion: Part of the chromosome is orientated in reverse of its usual direction.
-
- 4.Translocation: One part of the chromosome breaks off and attaches to another chromosome

Nondisjunction:

- This is a failure for a chromosome to separate during Meiosis.
-
- -Extra chromosome results in one cell and a loss of a chromosome occurs in the other cell.

Gene Mutations

- These mutations involve individual genes.
-
- -Any chemical change that affects the DNA can cause this type of mutation to happen.
-
- -Some may cause a change to 1 nucleotide, while some may change many.

Point Mutation

- Affects only one nucleotide.

- **Frameshift Mutation:** May change the entire polypeptide or protein chain produced by the gene.

- **Germ Mutations**: Mutations that affect the reproductive cells.
-
- **Somatic Mutations**: These do not affect the reproductive cells.
-
- -They are not inherited.
-
- (Both can occur at the level of Chromosomal and Gene Mutations)
-
- Sex Linked Genes:

- Remember **Nondisjunction** can be caused by a failure of the chromosomes to seperate duing meiosis.
-
- -This can cause a great increase in the numbers of chromosomes.
-
- This is called **Polyploidy**: Triploidy (3n)
  tetraploidy(4n)
-
- -This is almost always fatal in animals.

- **Aneuploidy**: Not true multiples of chromosomes.
-
- Only one half of a pair is given off.

Probability – Predicting Results

Rule of addition: the probability of 2 mutually exclusive events occurring simultaneously is the sum of their individual probabilities.

When crossing Pp x Pp, the probability of producing Pp offspring is

probability of obtaining Pp (1/4), PLUS

probability of obtaining pP (1/4)

¼ + ¼ = ½

Probability – Predicting Results

Rule of multiplication: the probability of 2 independent events occurring simultaneously is the PRODUCT of their individual probabilities.

When crossing Rr Yy x RrYy, the probability of obtaining rr yy offspring is:

probability of obtaiing rr = ¼

probability of obtaining yy = ¼

probability of rr yy = ¼ x ¼ = 1/16

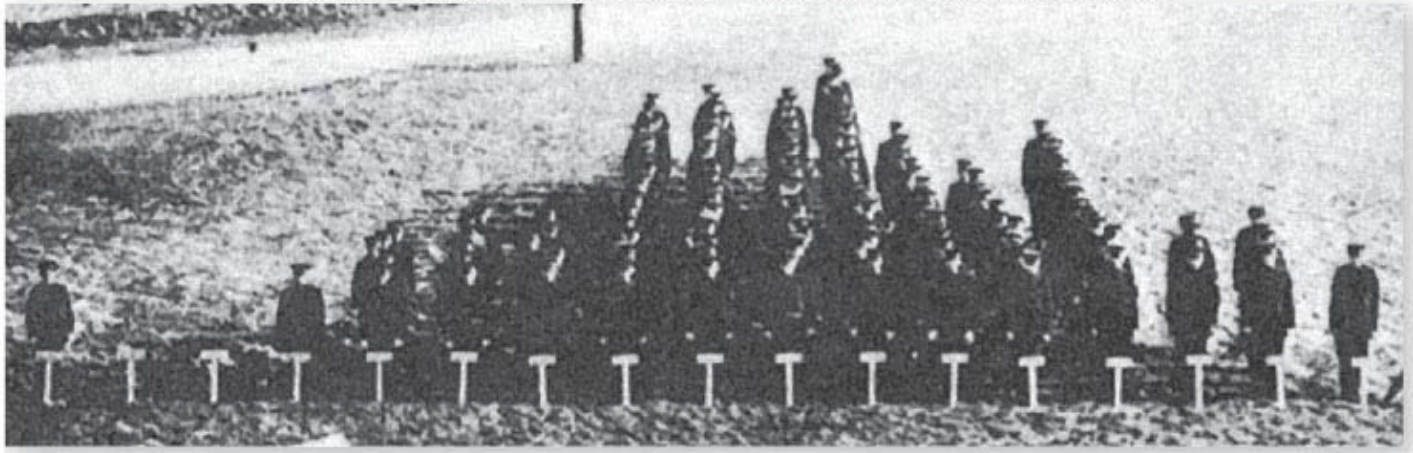
Extensions to Mendel

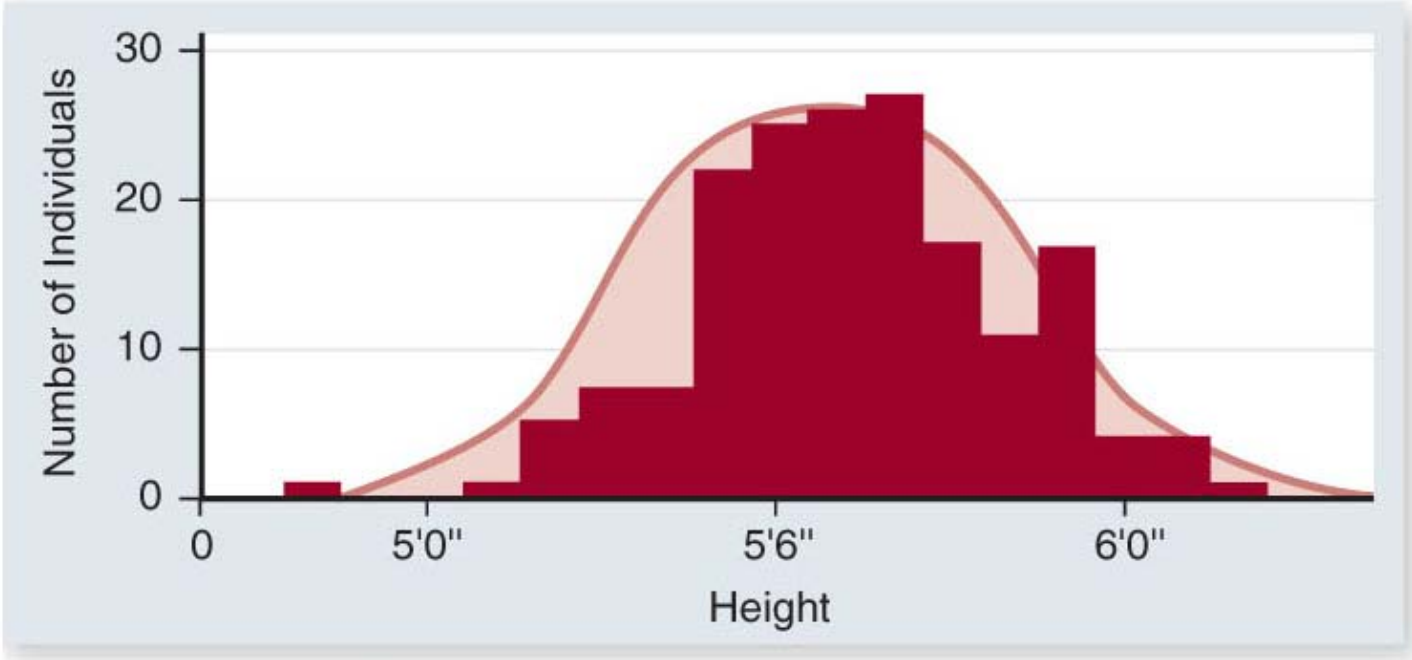
Polygenic inheritance occurs when multiple genes are involved in controlling the phenotype of a trait.

The phenotype is an accumulation of contributions by multiple genes.

These traits show **continuous variation** and are referred to as **quantitative traits**.

For example – human height





From Albert & Blakeslee Corn and Man Journal of Heredity, Vol. 5, pg 511, 1914, Oxford University Press

Extensions to Mendel

Pleiotropy refers to an allele which has more than one effect on the phenotype.

This can be seen in human diseases such as cystic fibrosis or sickle cell anemia.

In these diseases, multiple symptoms can be traced back to one defective allele.