AP BIOLOGY

Big Idea 3
Part B

December 2012

www.njctl.org

Big Idea 3:

Living systems store, retrieve, transmit and respond to information essential to life processes.

Big Idea 3: Part B

Click on the topic to go to that section

- Cell Cycle Control
- Heredity and Meiosis
- Meiosis and Mendel
- A Closer Look at Chromosomes
- Probability, Statistics, & Genetics

Cell Cycle Control

Return to Table of Contents

Cell Cycle Control

Cells divide only when the correct set of internal and external signals are present, allowing the cell to enter the next phase of the cell cycle.

If a cell fails to recognize the proper conditions it will lead to divisions that should not occur, or no division when it is necessary.

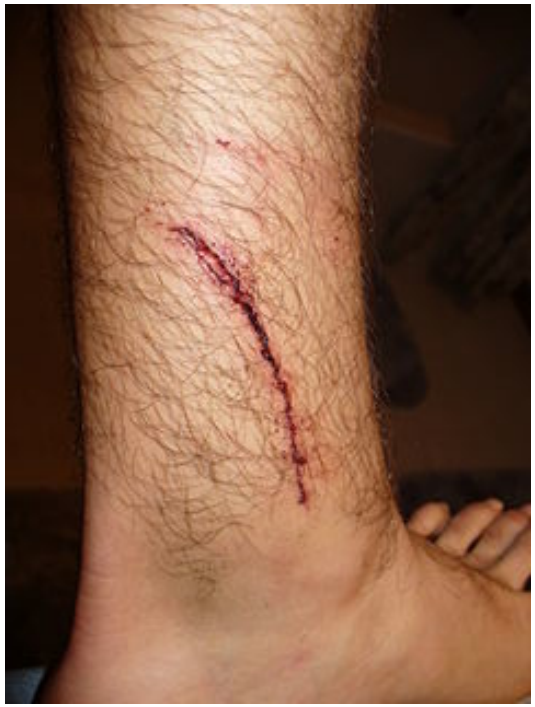
Cell Cycle Control

Example:
You accidentally cut your leg. Consider...
What has happened to the cells in the cut?

How will the cut be repaired by your body?

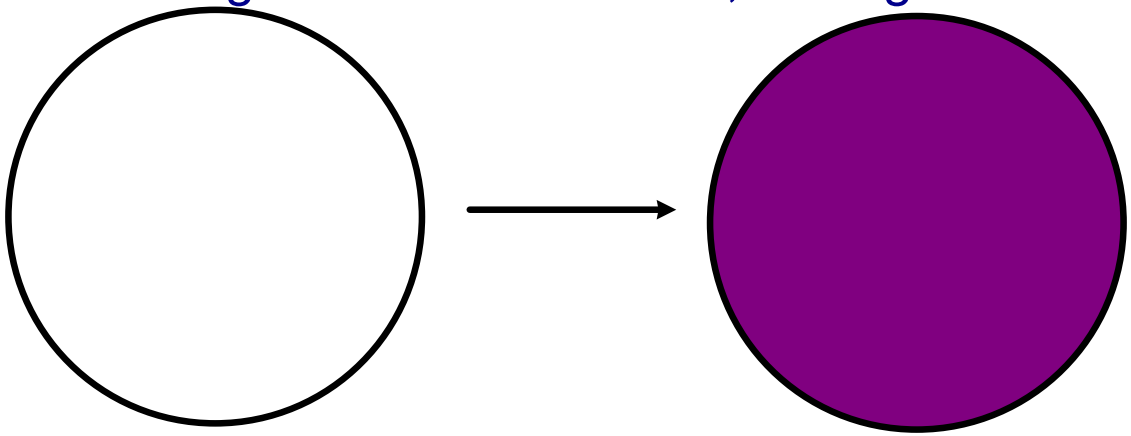
What does this have to do with cell cycle control?

Take a minute to talk this over with your group.



Density Dependent Inhibition

Cells are able to determine the density of the cells around them by chemical signals. The more cells, the higher the level of signal.

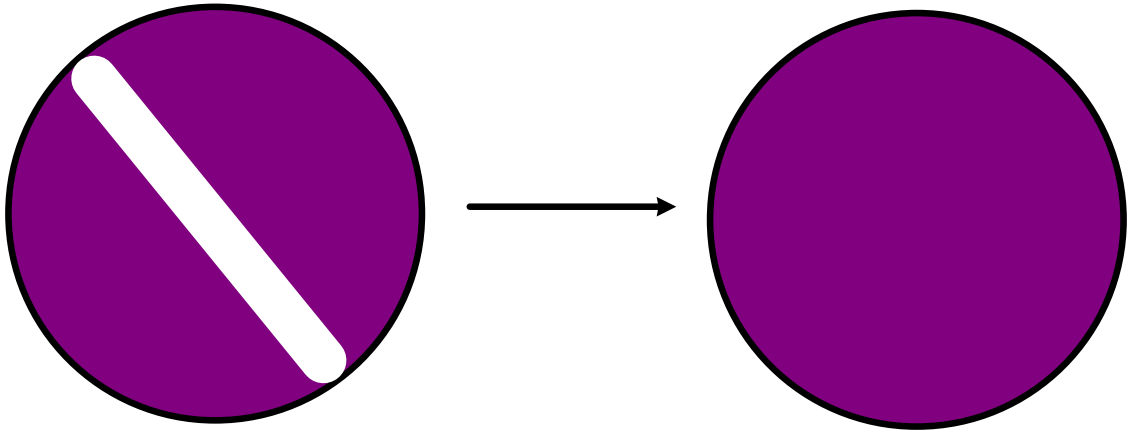


A few human cells are placed in a petri dish that contains all the necessary nutrients for survival and growth.

After a period of incubation allows the cells to divide, the cells form a single layer covering the entire petri dish and stop doing mitosis.

Density Dependent Inhibition

Cells are able to determine the density of the cells around them by chemical signals. The more cells, the higher the level of signal.

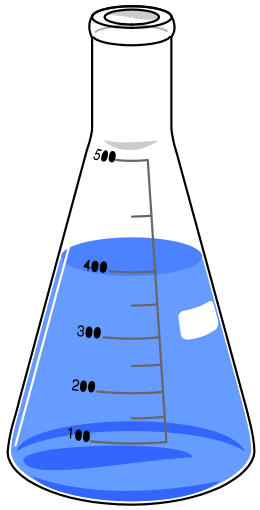


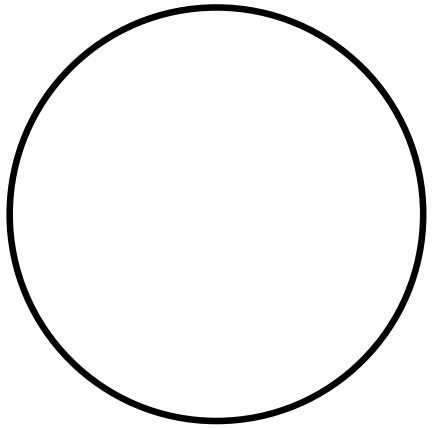
A scientist scrapes off a line of these cells in the center of the dish

After a period of incubation allows the cells to divide, the cells reform a single layer and stop doing mitosis.

Anchorage Dependence

Many human cells require a substrate (surface) to adhere to before they can divide. A signal from proteins in the plasma membrane enable the cell to progress through the cell cycle.





Human skin cells in a suspension don't divide.

Poured into a petri dish and allowed to adhere to a surface, they begin mitosis.

Growth Factors

A growth factor is a naturally occurring substance capable of stimulating cellular growth, division, and differentiation. Usually it is a protein or a steroid hormone. They are important for regulating a variety of cellular processes.

Growth factors typically act as signaling molecules between cells. Examples are cyclin and hormones that bind to specific receptors on the surface of their target cells.

Without the correct growth factors present, a normal cell cannot divide.

Control System Failure

When cells do not respond normally to external signals and begin to divide when they are not supposed to, serious problems arise.

They divide uncontrollably, they invade areas of the organism that should not be interfered with, and they could kill the organism. These rogue cells are known as **cancer** cells.

Cancer

Cancer is a general term for many diseases in multi-cellular organisms which is caused by uncontrolled cell division. Cancer cells and normal cells are identical, with the exception that cancer cells divide uncontrollably.

Cancer cells are non-responsive to the cell cycle control system.

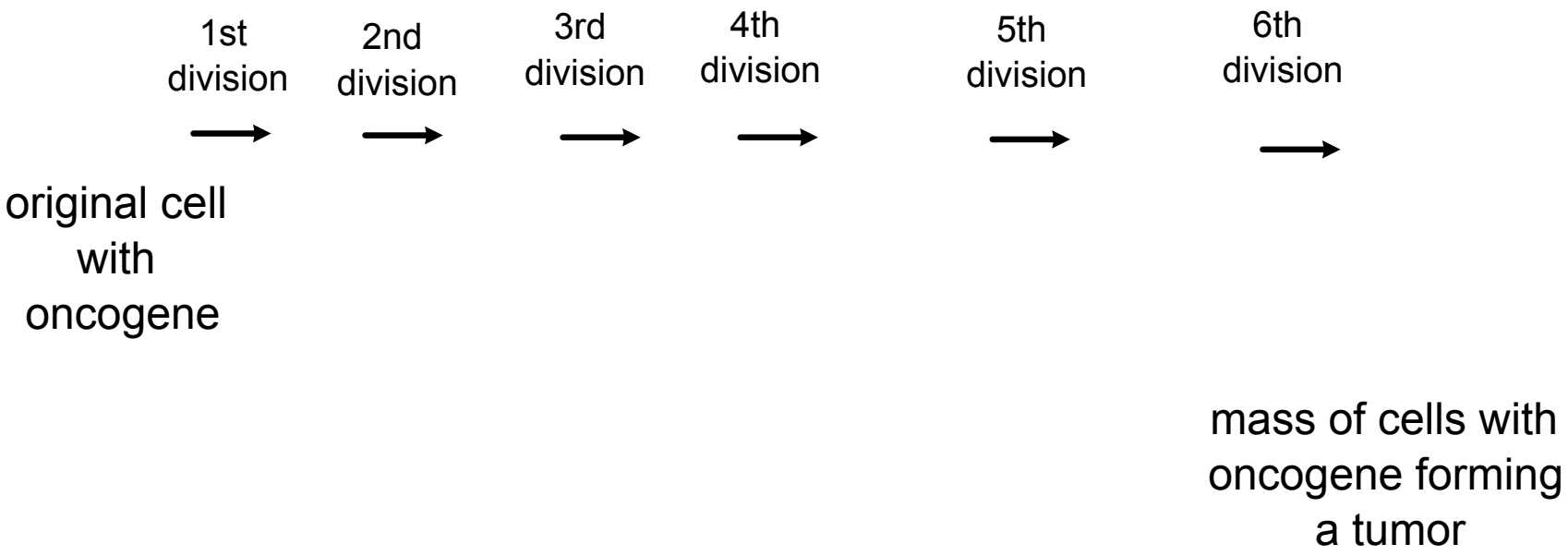
Cancer

The progression of cancer cells begins most often with a mutation in a **proto-oncogene**. These are the genes that promote normal cell division by producing growth factors.

When they become damaged and begin to cause too much cellular growth and division, then they are referred to as **oncogenes**, or tumor causing genes.

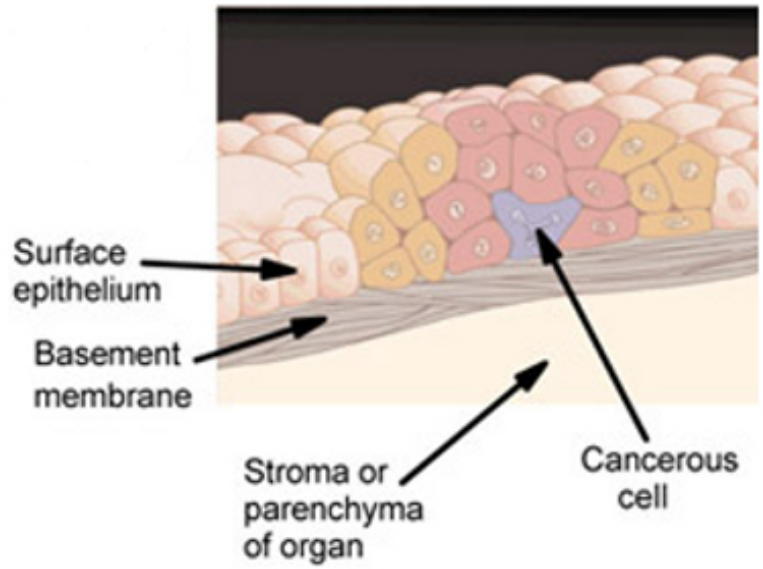
Cancer

An oncogene will cause a cell to ignore cell density or anchorage. This cell will proceed through the cell cycle. When it divides, the two daughter cells both have the oncogene and quickly proceed through their cell cycle.



Cancer

Once a tumor is formed it can be a serious problem for the organism. If the tumor remains localized and does not threaten the health of the overall organism it is called a **benign tumor**. However, if left unchecked most tumors will cause more sever problems.



If this tumor does not invade the basement membrane or other tissues it will remain benign.

1 All growth factors cause cancer.

- True
- False

2 Tumors are most often caused by a mutation in

- A a proto-oncogene
- B an oncogene
- C any density dependent cell
- D any achorage dependent cell

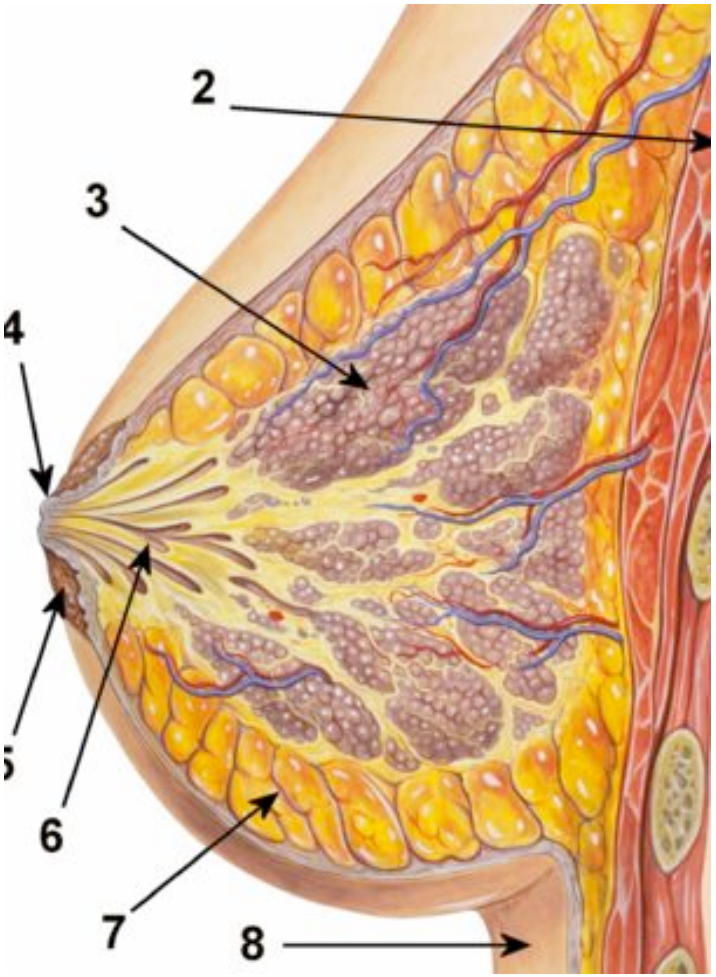
Progression of Breast Cancer

Breast cancer awareness is focussed on the idea of early detection. If a tumor in the mammary gland is detected early, it minimizes the damage caused by the diseased cells and may only require a simple surgery to remove the tumor.

Progression of Breast Cancer

This diagram represents a cross section of a healthy human female breast. 3 is the mammary gland where most breast cancers emerge.

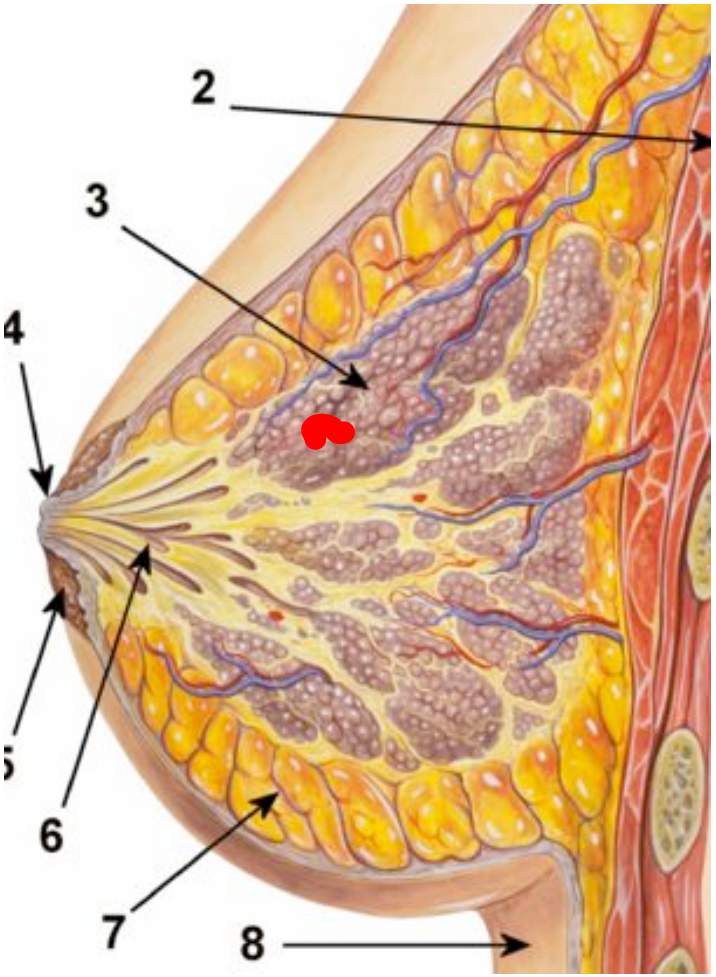
2 - muscle tissue
4 - nipple
5 - areola
6 - collecting ducts
7 - insulating fat for protection
8 - skin



Progression of Breast Cancer

The red area represents a recently formed tumor. If the lump is detected at this point it is possible that the lump can be removed in a surgical procedure known as a lumpectomy.

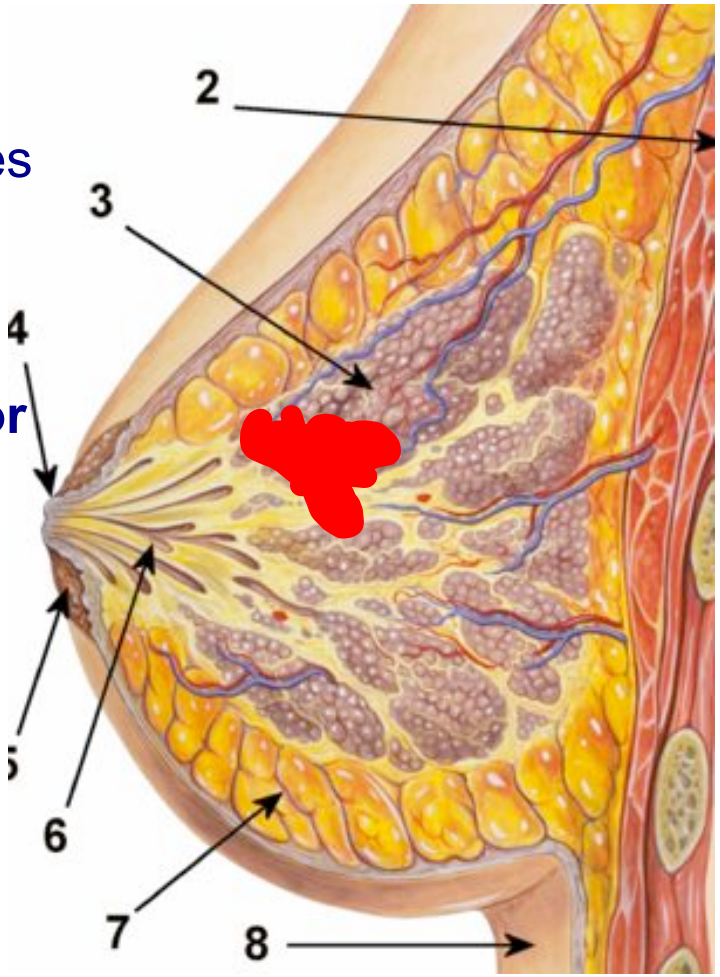
This is minimally invasive and the patient should return to normal quickly.



Progression of Breast Cancer

If the tumor grows larger and interferes with the tissue of the mammary gland then it may be necessary to completely remove the mammary gland. This is now a **malignant tumor** because it is interfering with normal organism functions.

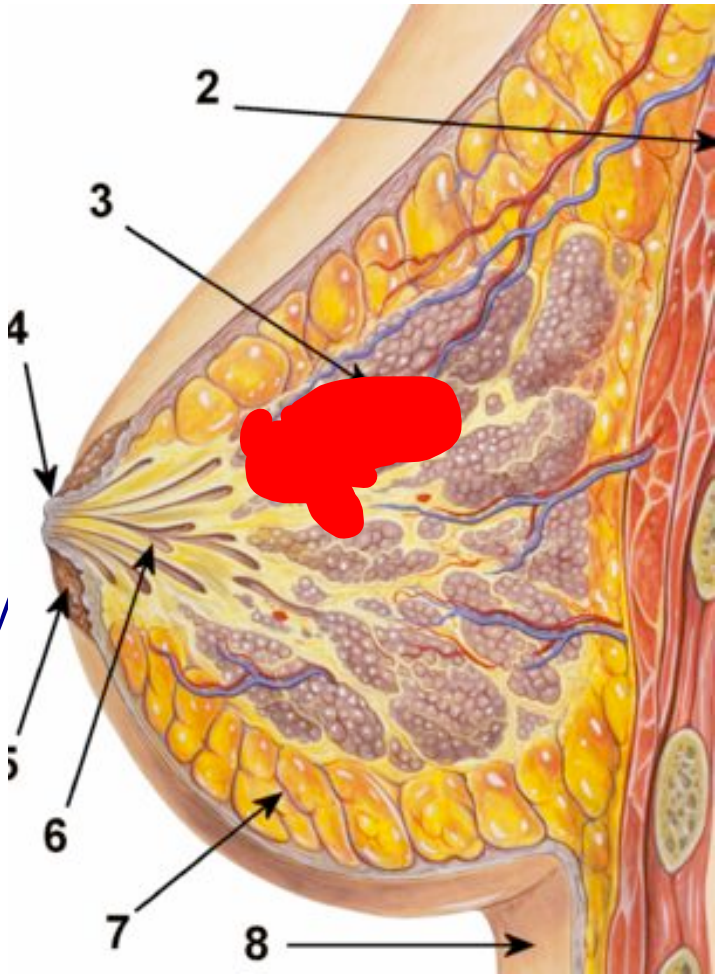
This mastectomy requires removal of the entire breast.



Progression of Breast Cancer

If the tumor escapes the immediate surroundings and cancerous cells enter the blood or lymph systems the cancer has **metastasized**. It may now lead to cancer spreading throughout the organism.

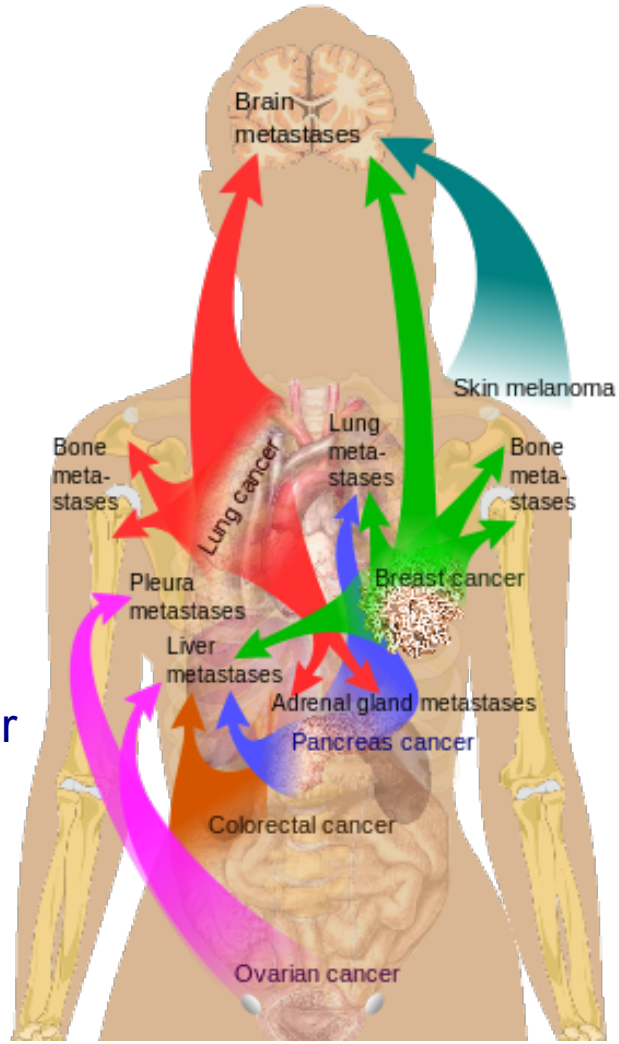
This patient will require a mastectomy as well as chemotherapy and radiation treatments to avoid tumors appearing in other organs.



Metastasis

Metastasis is extremely dangerous to the host organism. With breast cancer, the most likely place for new cancer to appear is in the brain. Many forms of brain cancer are incurable.

This diagram shows the likely metastases sites for common cancers.



Cancer and Heredity

It is also important to know family history. Since we inherit genes from our parents, grandparents and so on, certain people can be more likely to get cancers that their ancestors had. If a woman's mother had breast cancer she is more likely to get breast cancer in her life. Proper precautions could minimize the damage.

3 Rank the following in the order of severity from least to greatest.

I. malignant tumor

II. mutation in proto-oncogene

III. metastasis

IV. oncogene

- A I, II, III, IV
- B III, IV, I, II
- C II, IV, I, III
- D II, III, I, IV

Heredity and Meiosis

Return to Table of Contents

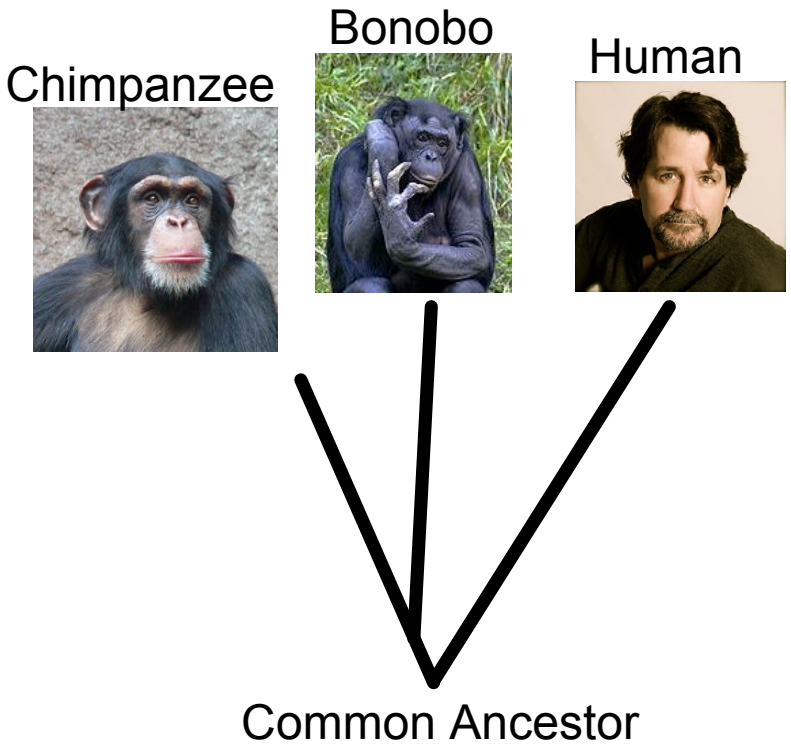
Heredity

Heredity is the passing of traits to offspring from parents. This is the process by which an offspring acquires, or becomes likely to have, the characteristics of its parent organisms. Through heredity, variations exhibited by individuals can accumulate and cause some species to evolve. **The study of heredity in biology is called genetics.**

Heredity

Two individuals have a genetic relationship if one is the **ancestor** of the other, or if they share a common ancestor. In evolutionary theory, species which share an evolutionary ancestor are said to be of **common descent**.

The gene pool of the chimpanzee and bonobo share 99.6% of the same genes. 98.7% of human genes are common with the bonobo.



Heredity

Fill in the blanks below as a reminder of what you know...

_____ is the molecule that is used to store information. _____s are the basic unit of heredity. A _____e is defined as a discrete package of genes that is used to transfer information. The _____ is where genes are kept in eukaryotic organisms.

Heredity and Reproduction

Reproduction is a requirement for heredity.
The 2 methods of reproduction that are most frequently used:

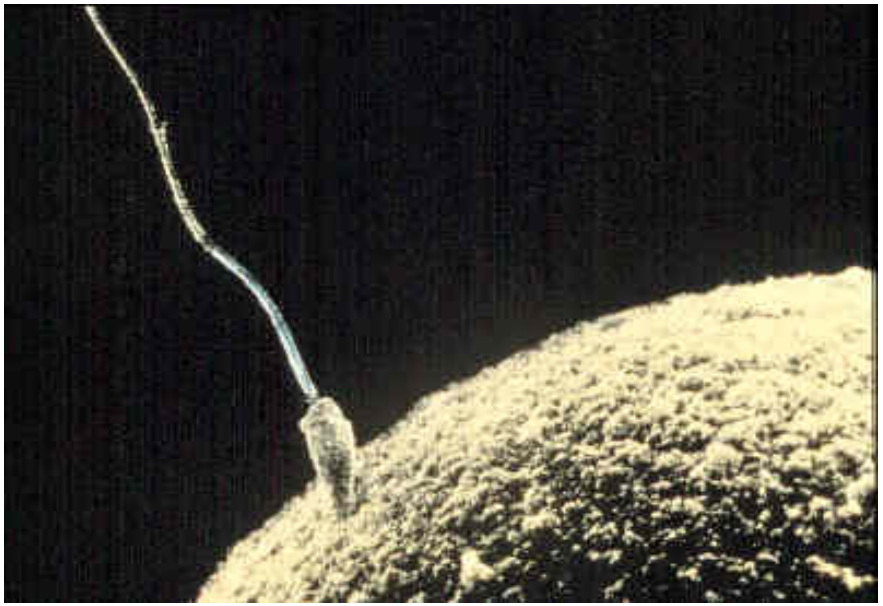
1) Asexual **binary fission** in prokaryotes.
This method is simple but produces minimal variation.

2) **Sexual reproduction** in multicellular eukaryotes
This method is highly complex requiring massive amounts of energy. The trade off is more variation.

Single celled eukaryotes can take advantage of asexual mitosis to produce offspring daughter cells.

Gametes

The sex cells of organisms are called **gametes.** Eggs in female animals, sperm in male animals; ovules and pollen, respectively in plants. In many eukaryotic organisms, the **somatic cells** (those that are not sex cells) have two sets of chromosomes (**diploid**).



Fusion of haploid gametes during fertilization results in a diploid offspring.

Gametes have one set of chromosomes (**haploid**) and they are produced by **meiosis**.

Sexual life cycles alternate between haploid and diploid phases.

\

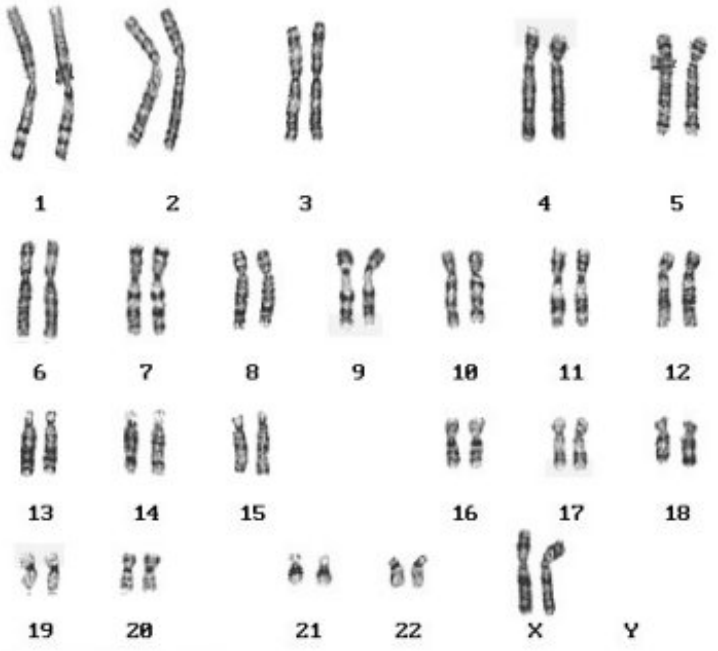
Homologous Chromosomes

The pairs of matching chromosomes in the somatic cells of diploid organisms are called **homologous chromosomes**. In humans, each somatic cell contains 46 chromosomes, which make up 23 homologous pairs.

Homologous chromosomes share shape and genetic loci, each pair controlling the same inherited characteristics. Each pair is inherited from the parents, one from mother, one from father (the sets are combined in the first cell following fertilization and then passed down by mitosis).

Karyotype

A **Karyotype** is a photographic inventory of chromosomes - the chromosomes are digitally separated and ordered.



A karyotype of a human female, showing 23 sets of homologous chromosomes

Alleles

Homologous chromosomes can carry different versions of the same gene. These "versions" are called **alleles**

2 examples: coat color and eye color in mice

Coat Color: Brown and White are different versions of the same gene for coat color.

Eye Color: Black eyes and Pink eyes are different alleles of the gene coding for eye color.

4 Two chromosomes in a nucleus that carry loci for the same traits in the same positions on the chromosome but can specify different versions of the same traits constitute a pair of:

- A homologous chromosomes
- B complimentary chromosomes
- C heterozygous chromosomes
- D none of these are correct

5A karyotype is analogous to which of the following examples?

- A a map of hidden treasure
- B a movie showing the reproductive cycle of a beetle
- C a photograph of every couple at the prom
- D the answer key for a test

6Which of the following statements is ***false***?

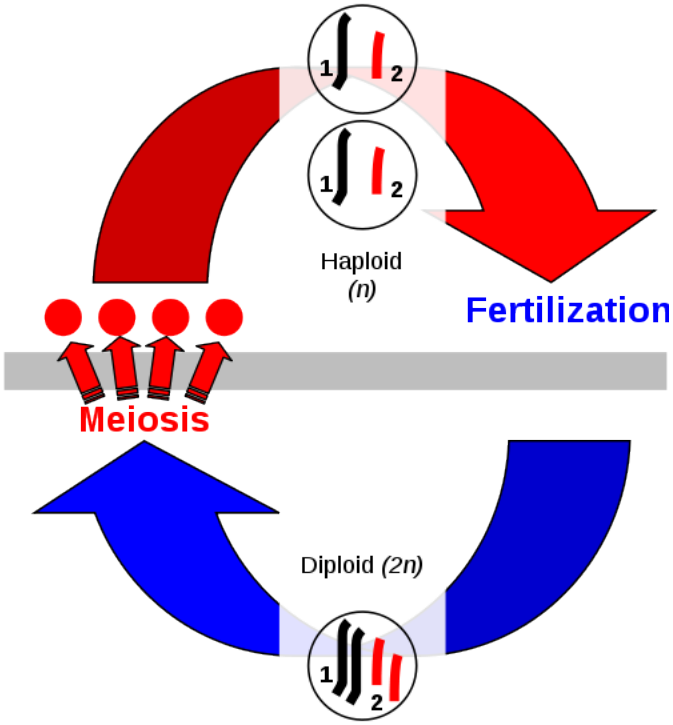
- A a typical body cell is called a somatic cell
- B gametes are haploid cells
- C somatic cells are diploid
- D gametes are made by mitosis
- E a zygote is an egg

Meiosis

Meiosis reduces chromosome numbers in diploid organisms to create sex cells.

Like mitosis, meiosis is begun by a single duplication of chromosomes.

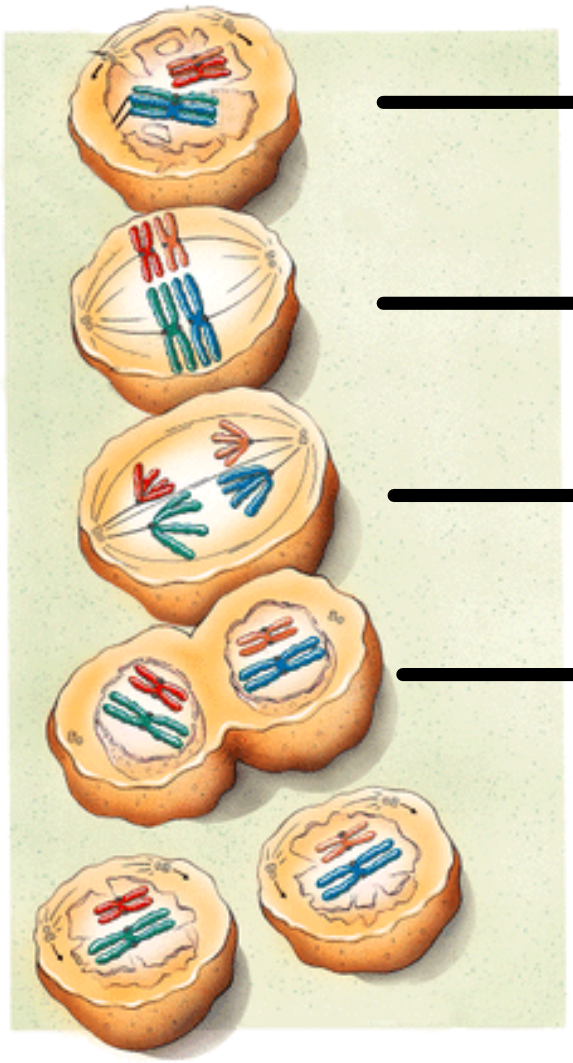
Unlike mitosis, the overall result of meiosis is 4 haploid daughter cells.



The Two Divisions of Meiosis

The process involves 2 consecutive divisions, simply called **Meiosis I** and **Meiosis II**.

Halving the actual chromosome number occurs in Meiosis I. Then, the sister chromatids separate in Meiosis II, resulting in 4 cells.



MEIOSIS I

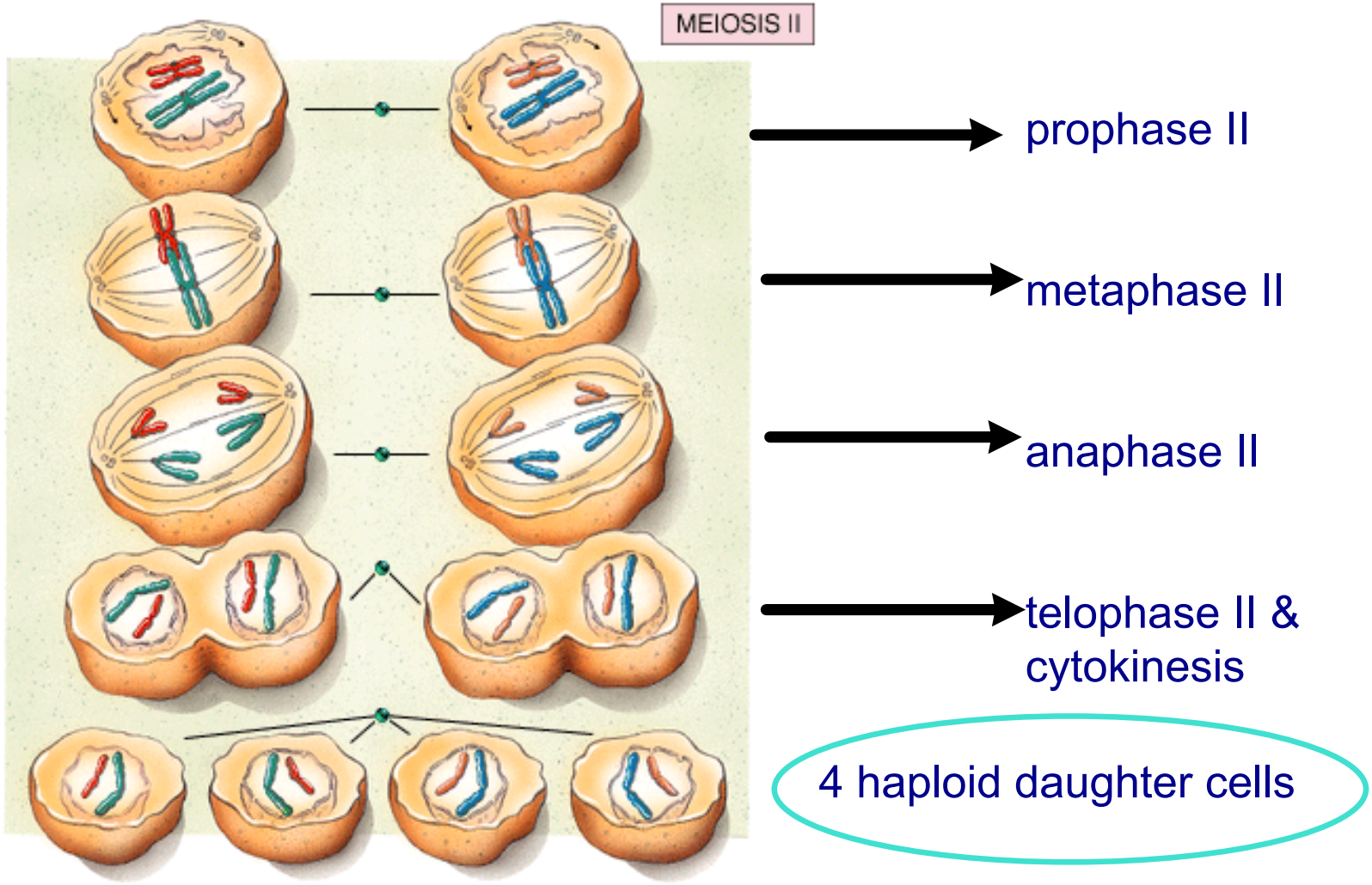
prophase I

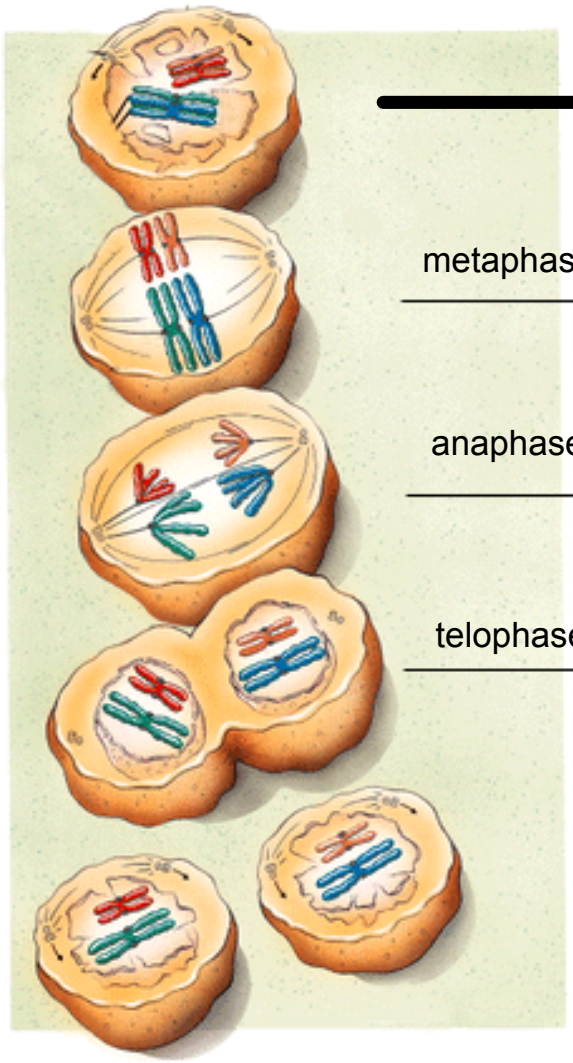
metaphase I

anaphase I

telophase I & cytokinesis

2 diploid daughter cells


MEIOSIS II
prophase II
metaphase II
anaphase II
telophase II &
cytokinesis
4 haploid daughter cells

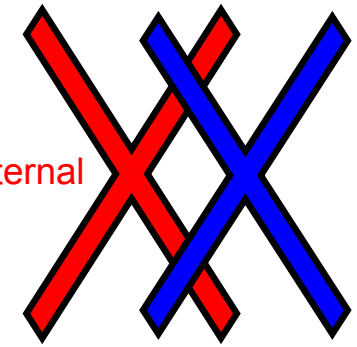


prophase I

The nuclear envelope disappears and the spindle begins to form.

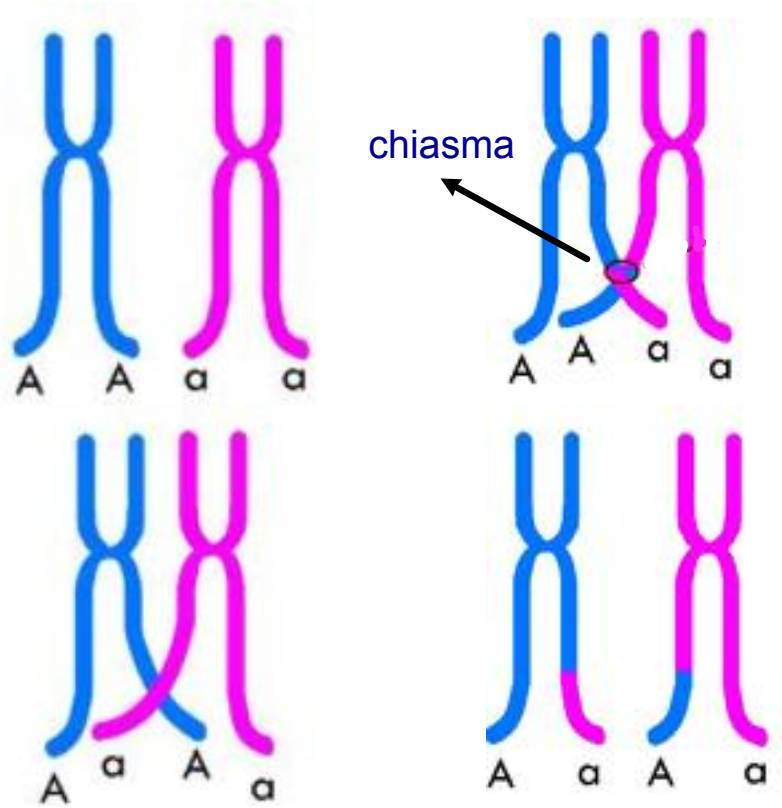
Homologous chromosomes (replicated during interphase) pair to form a **tetrad.**

2 copies of maternal chromosome 5



2 copies of paternal chromosome 5

Crossing Over



Crossing over occurs during prophase I. This is a genetic rearrangement between 2 homologous chromosomes that happens at a site called a **chiasma.**

Crossing over increases the genetic variation of the offspring. Since this can occur several times at variable locations in each tetrad, the variation which can occur between 2 parents is extremely large.

This is one of the reasons that, with the exception of identical twins, everyone is a unique genetic entity.

Video on crossing over

7 A genetic rearrangement between 2 homologous chromosomes is called:

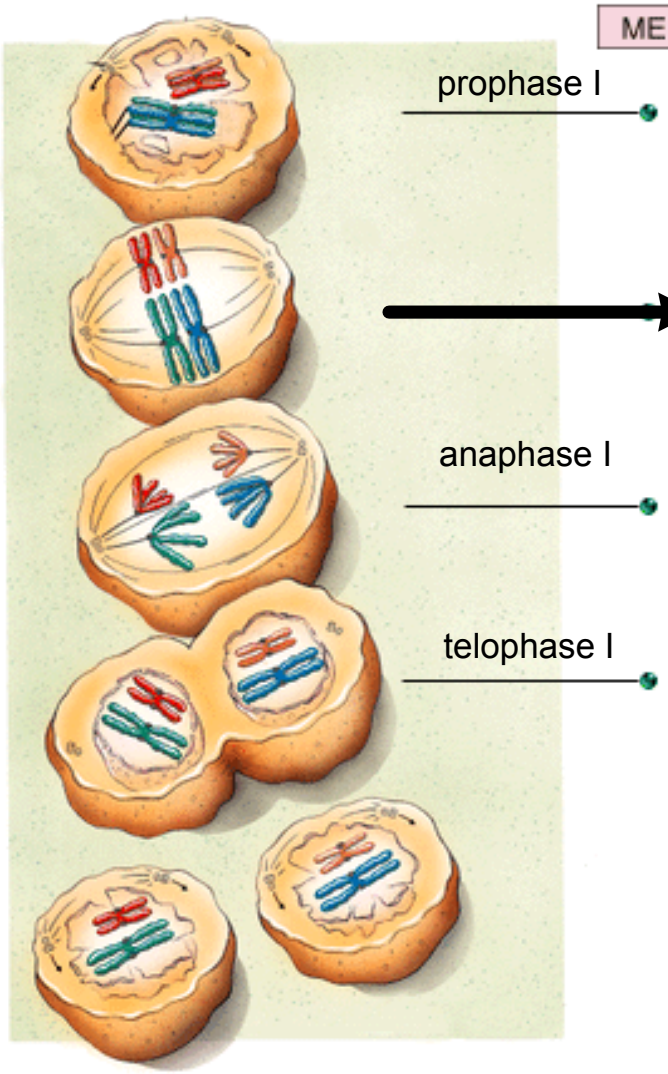
- A chiasma
- B homologous rearrangement
- C crossing over
- D haploid reduction

8 Which of these is NOT a component of meiosis?

- A crossing over
- B pairing of homologous chromosomes
- C random fertilization
- D production of gametes

9 Crossing over can occur many times on each homologous pair.

- True
- False



Video on independent assortment

metaphase I

The tetrads line up at the equator of the cell.

Independent Assortment

During prophase I of meiosis, each homologue pairs up with its match and the X and Y chromosomes behave as one homologous pair.

During anaphase I, maternally and paternally inherited genes on the homologues move to one pole or the other, independent of the other genes.

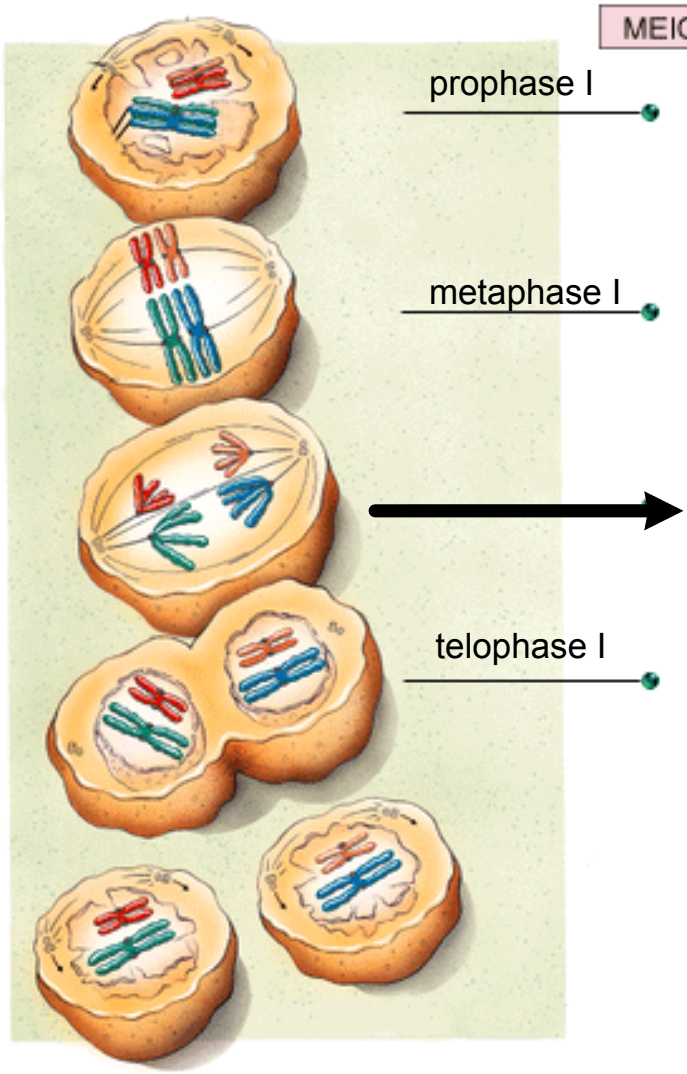
The way chromosomes line up during metaphase I gives each cell a unique combination of genes from each parent's chromosomes (an "independent assortment").

Independent Assortment

Given *n* pairs of chromosomes, there are 2^n ways in which chromosomes can line up during metaphase I.

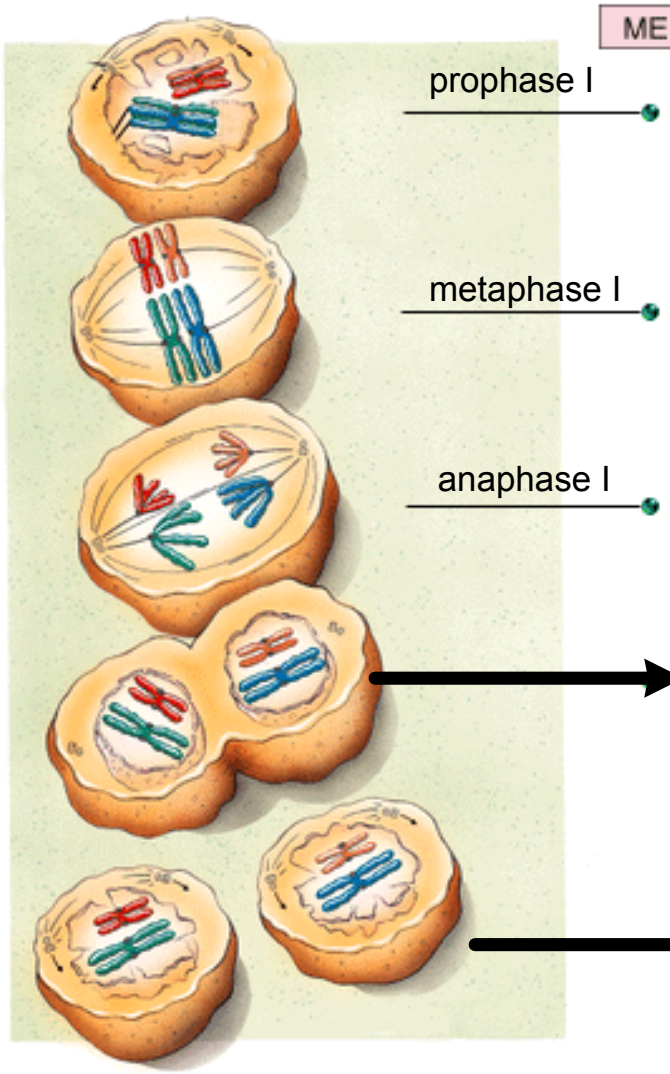
In humans there are 2^{23} *(8 million)* ways of combining homologues.

This means combining human gametes can produce 64 trillion combinations in the zygote!



anaphase I

The tetrad splits, maternally and paternally inherited chromosomes moving to one pole or the other, independent of other chromosomes.

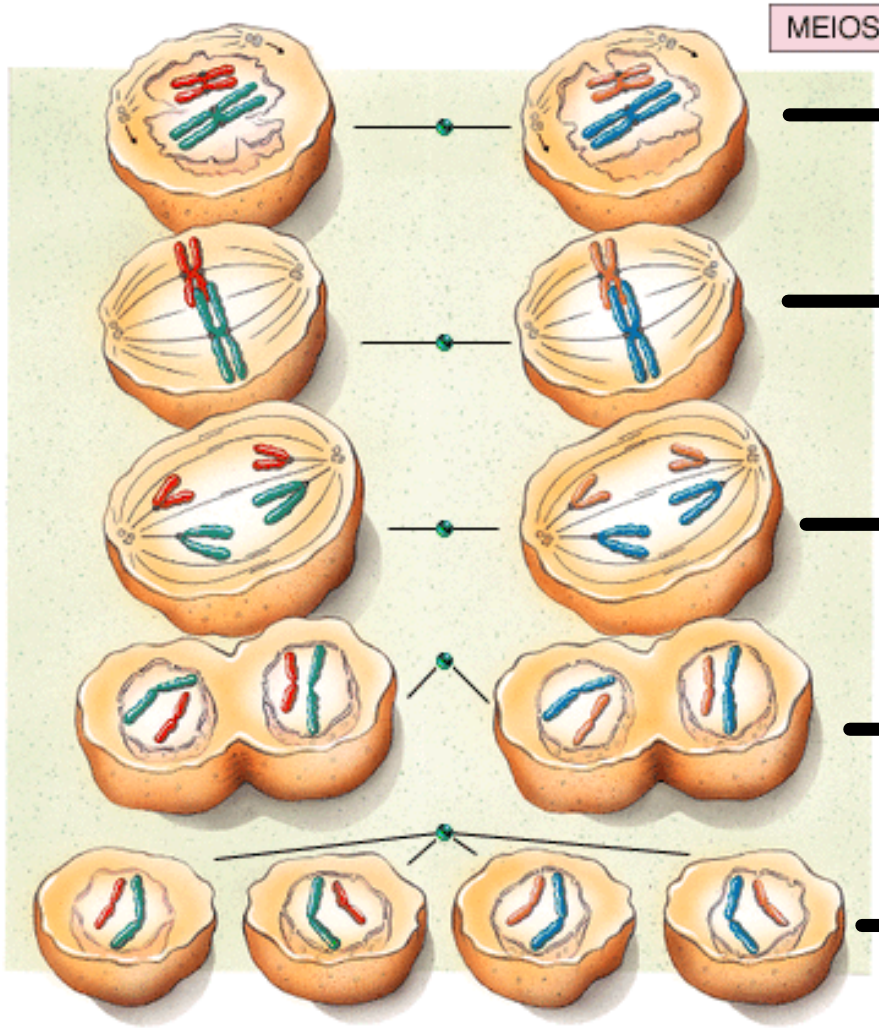


telophase I

Nuclear envelope reforms.
Nucleus is now haploid.

interkinesis

Division of the cytoplasm.
Similar to cytokinesis.



prophase II: nuclear envelope disappears; spindle forms

metaphase II: chromosomes with 2 sister chromatids line up at the equator

anaphase II: chromosomes split apart, one chromatid moving to each pole

telophase II: nuclear envelope reappears

cytokinesis: cytoplasm divides

4 haploid daughter cells

10 Independent assortment states that

- A each pair of gametes separate independently of each other during meiosis
- B genes sort independently in animals but not in plants
- C independent sorting produces polyploid individuals
- D individual genes from each parent sort independently of other during meiosis

11 Which of these is NOT a component of meiosis?

- A crossing over
- B pairing of homologous chromosomes
- C random fertilization
- D production of gametes

12 With the exception of identical twins, siblings with the same parents will likely look similar but not identical to each other because

A they have identical chromosomes

B they have identical genes but not chromosomes

C they have a similar but not identical combination of genes

D they have a small chance of having identical genes

Meiosis and Mendel

Return to Table of Contents

Meiosis and Mendel

Remember that Mendel's punnet squares were used to predict the probability that offspring would inherit a trait.

Mendel was discovering how inheritance works without knowing about DNA or chromosomes.

Meiosis and Mendel

Review Mendel's work by doing this cross:
Organism 1 mates with organism 2. Both are heterozygous for 2 traits. What is the probability that that an offspring will be born with the recessive phenotype of trait A and dominant trait B.

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

Answer:

When we look at this chart knowing modern biology we can explain in more depth than Mendel could.

Meiosis and Mendel

First step is to see all the possible outcomes of meiosis for both organisms to produce egg and sperm. In other words, the possible outcomes of assortment of chromosomes from metaphase 1.

haploid sperm possibilities

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

haploid egg possibilities

Answer:

Meiosis and Mendel

Next, look at all the possible combinations that could arise if each possible combination of chromosomes were to meet during fertilization.

haploid sperm possibilities

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

haploid egg possibilities

Possible zygote combinations for chromosomes containing trait A and B

Answer:

Meiosis and Mendel

What the answer tells us is the possibility, given all that occurs in the sexual life cycle, that the chromosome containing A and the chromosome containing B will wind up in the same individual.

haploid sperm possibilities

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

haploid egg possibilities

Possible zygote combinations for chromosomes containing trait A and B

Answer:

3/16 or .19

there is a 19% chance for the given combination

Meiosis and Mendel

Expanding on Mendel's Postulates...

Hypothesis # 1: Alleles

There are alternative forms of genes that account for variations in inherited characteristics.

Meiosis and Mendel

Hypothesis # 2: All Organisms have 2 alleles

For each characteristic, an organism inherits two alleles (one from each parent). The two alleles may be the same or they may be different.

Technically the offspring is inheriting a group of alleles for many different traits contained on a chromosome. Each gamete contributes a full set of haploid chromatids containing one allele for every possible trait.

Meiosis and Mendel

Hypothesis # 3: Dominant and Recessive

If the two alleles of a pair are different (heterozygous), one determines the appearance and is called the dominant allele.

We now understand that the "dominant" allele does not overcome the "recessive" allele. It simply masks the phenotype of the recessive. Both alleles are active in the diploid offspring. This often leads to **intermediate traits** that are a combination of the allele variants.

Meiosis and Mendel

Hypothesis # 4: Law of Segregation

A sperm or egg carries only one allele for each trait because allele pairs separate from each other during sex cell formation.

In meiosis, a complex shuffling of chromosomes and the production of 4 genetically unique sex cells from one diploid cell leads to many possible variations. This leads to a practically infinite number of trait combinations and a healthier population.

A Closer Look at Chromosomes

Return to Table of Contents

Mendelian Problems

When Mendel chose his pea plant traits he did so very carefully. In fact, sometimes he would not get the desired results and omit them from his studies. Explain in terms of modern understanding of genes why the following may happen:

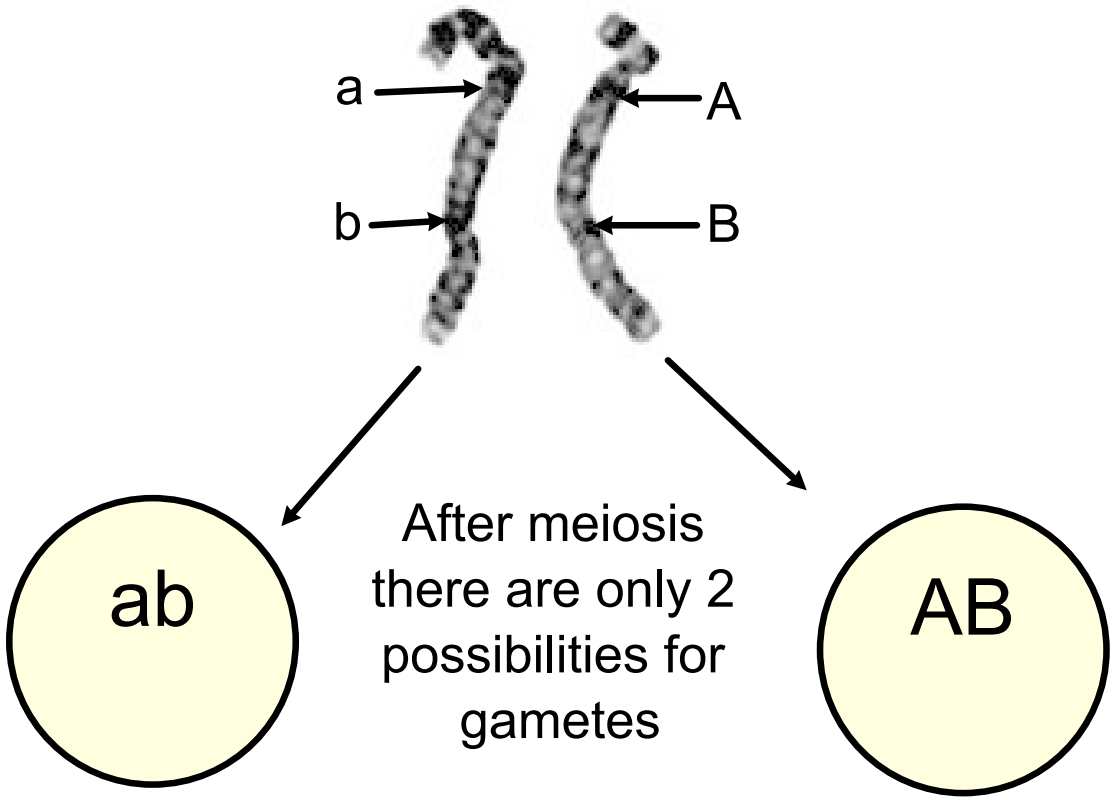
AaBb X AaBb Mendel would expect this cross to yield a phenotype ratio of...

9 both dominant:
3 A dominant B recessive:
3 A recessive B dominant:
1 both recessive

Actual Results...
3 both dominant :1 both recessive

Mendelian Problems

What Mendel didn't know was that alleles are grouped together on chromosomes. In this case the recessive are on one of the chromosome pairs of the parent and the dominants on the other.



Linked Genes

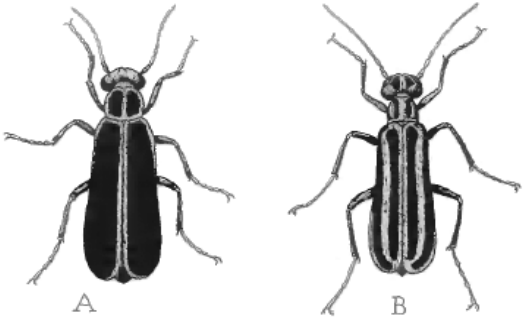
The previous slide illustrated the concept of linked genes. They turned out to be an important focus of study that lead to a more advanced understanding of chromosomes.

Take notes as you go through the next 4 slides so that you can answer a question about the practice experiment.

Beetle Genes

A geneticist is working with blister beetles and happens to find a new mutation in one individual. This individual is striped while the rest of the population is one color.

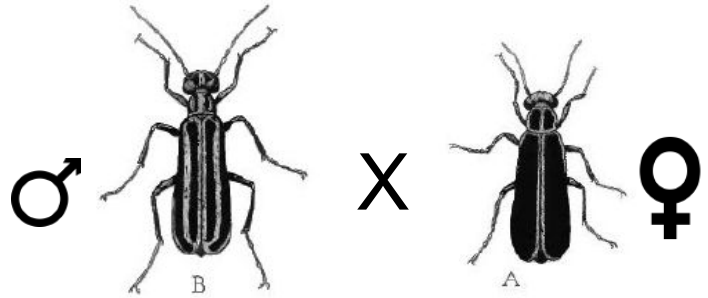
Beetle A represents the **wild type**, or the common variant found in nature.



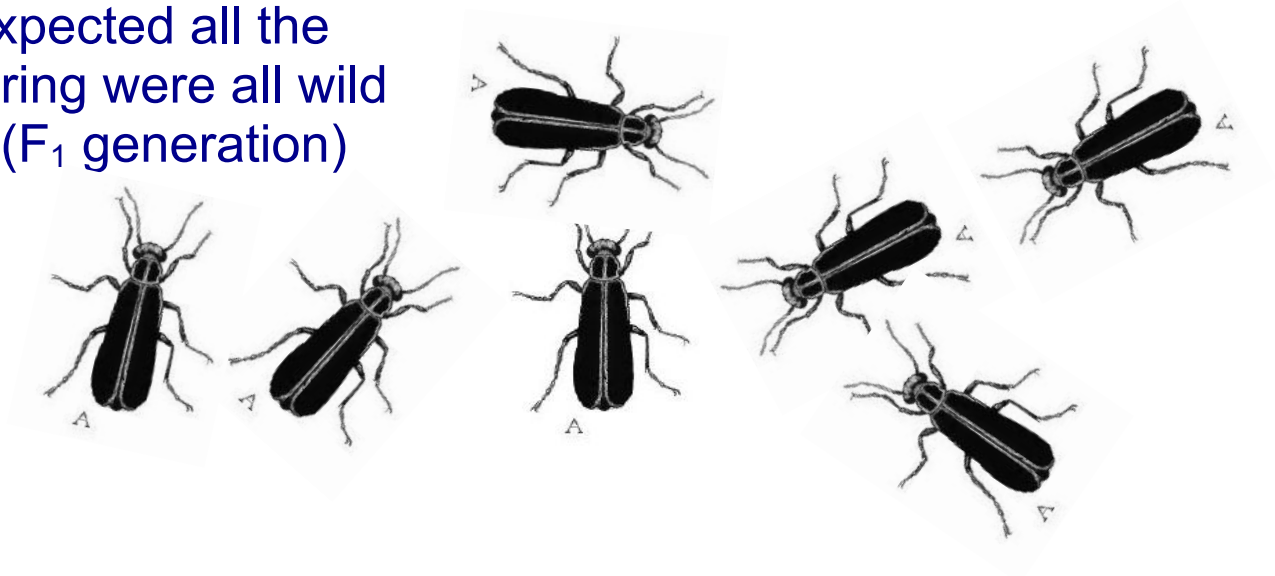
Beetle B represents the **mutant**, or the unusual variant.

Beetle Genes

Curious to see if the new allele was dominant or recessive he conducted a Mendelian style cross. The mutant was male so he crossed it with a female wild type.

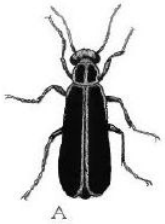
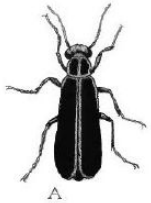


As expected all the offspring were all wild type (F_1 generation)

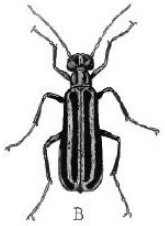


Beetle Genes

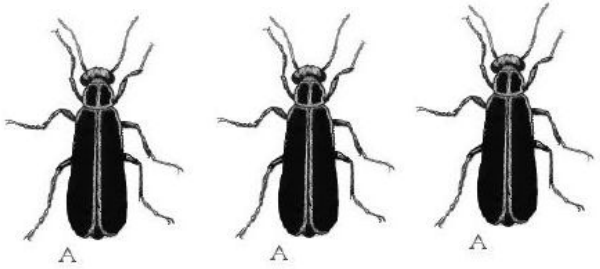
Next he took a male and female from the F_1 generation and crossed them.

♂  X  ♀

As expected he got a 3 to 1 ratio (F_2 generation).
However, out of more than 10,000 offspring, **all the striped individuals were male.**



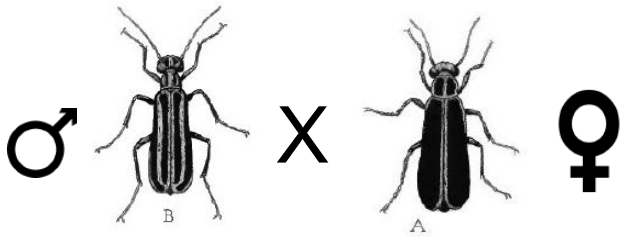
only ♂



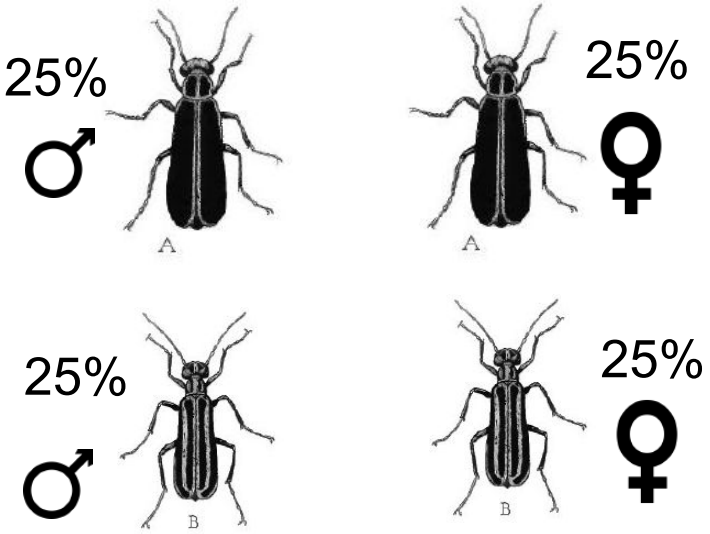
♂ and ♀

Beetle Genes

Finally he took a striped male and an F_2 generation female and crossed them. Take a look at his results. With a small group theorize why he got these results. Start with the original pairing and work to this cross.



F2



15,000 total offspring produced

Thomas Hunt Morgan

The previous experiment mimics the one done by Thomas Hunt Morgan in 1904. Look through the below slide show and compare Morgan's result to your own.

Click here for a synopsis of Morgan's experiment

Sex-linked Genes

The sex chromosomes have genes for many characteristics unrelated to sex. A gene located on eithersex chromosome is called a **sex-linked gene**.

Sex-linked genes follow specific patterns of inheritance. Many sex-linked disorders are located on the X chromosome. Typically these disorders are inherited through the mother, as she gives an X chromosome to any child she has. Sex-linked disorders show up in males more often than females because they only have 1 copy of an X chromosome.

X-linked Disorders

Some disorders caused by recessive alleles on the X chromosome in humans:

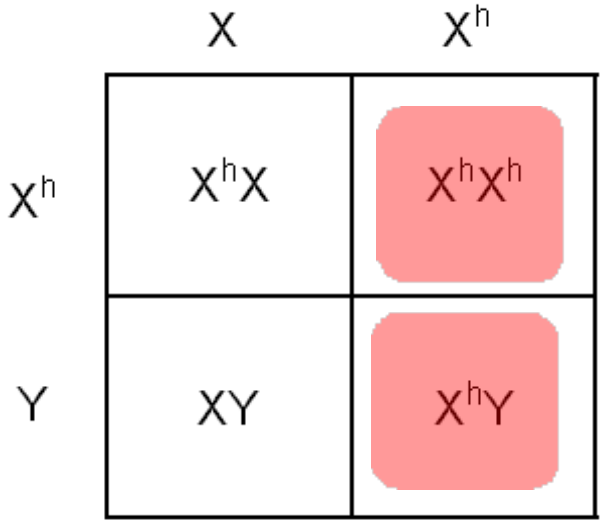
Color blindness

Duchenne muscular dystrophy

Hemophilia

Male pattern baldness

Sex-linked
H = normal & h = hemophilia
Cross: XX^h x X^hY



Genotypic ratio: 1:1:1:1
(X^hX =25% X^hX^h=25% XY=25% X^hY=25%)

Phenotypic ratio: 1:1:1:1
Female carrier =25% Female hemophilia =25%
Male normal =25% Male hemophilia =25%

13 X-linked disorders show up more often in males because in terms of sex chromosomes:

- A they are likely to inherited the recessive allele from their father
- B they only inherit one X-chromosome
- C the Y-chromosome from their mother carries the recessive allele
- D the father can pass the damaged allele on

14 In reference to the parental genotypes below, _____% of children are likely to be male.

- A 100
- B 25
- C 50
- D cannot be determined

father	mother
X^hY	X^hX

15 In reference to the parental genotypes below, _____% of children are likely to be female.

- A 100
- B 50
- C 25
- D cannot be determined

father	mother
XY	X^hX

16 In reference to the parental genotypes below, there is _____ likelihood that a child will have a hemophelia phenotype.

- A no
- B some

father	mother
X^hY	X^hX

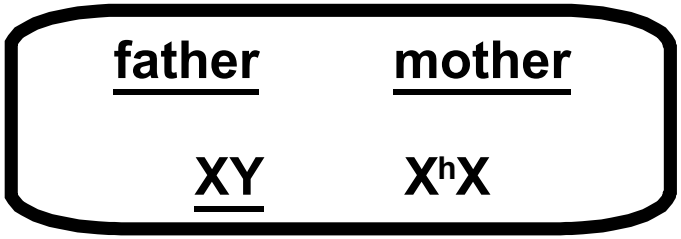
17 In reference to the parental genotypes below, there is _____ likelihood that a female child will have a hemophelia phenotype.

- A no
- B some

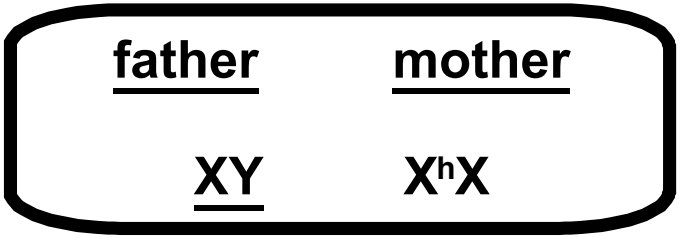
father	mother
XY	X^hX

18 In reference to the parental genotypes below, there is _____ likelihood that a male child will have a hemophelia phenotype.

- A no
- B some



19 The likelihood that a male child with hemophelia will be born is _____ %.



Genetic linkages

Take notes: Another researcher was studying a type of caterpillar. She noticed that a very few individuals in the population had blue spots. After careful screening, she also observed that if a caterpillar had blue spots it was also very likely to have long antennae.

Wild Type 

 Mutant

Genetic linkages

After extensive work she determined that the 2 mutant traits are **recessive** and **linked**. She notated the wild type genes as + and the recessives as a and s

Wild Type

Short Antennae = +

No spots = +

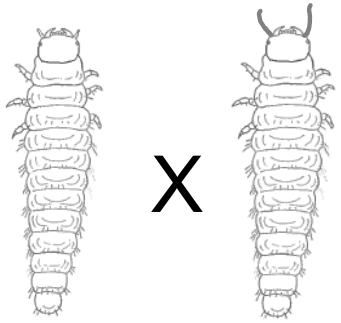
Mutant

Long Antennae = a

Spots = s

Genetic linkages

She preformed the following cross:



++, as
Heterozygous
wild type female

X

as, as
Mutant
Male

Determine
expected results:

	as	as
++	++, as	++, as
as	as, as	as, as

Genetic linkages

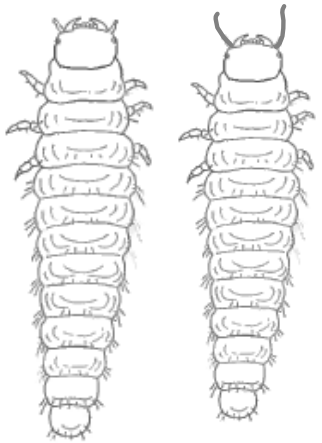
Her actual results differed from her expected.

Actual Results:

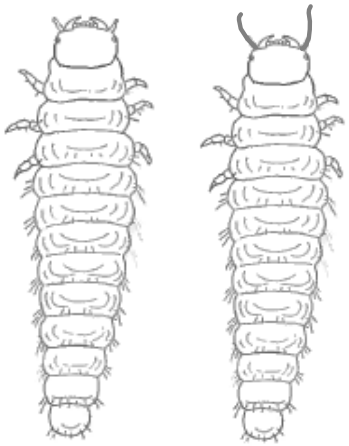
22,000 offspring produced

10,855 both traits wild type
10,930 both traits mutant
110 blue spots short antenae
105 no spots long antenae

What has taken place that caused these linked genes to recombine?



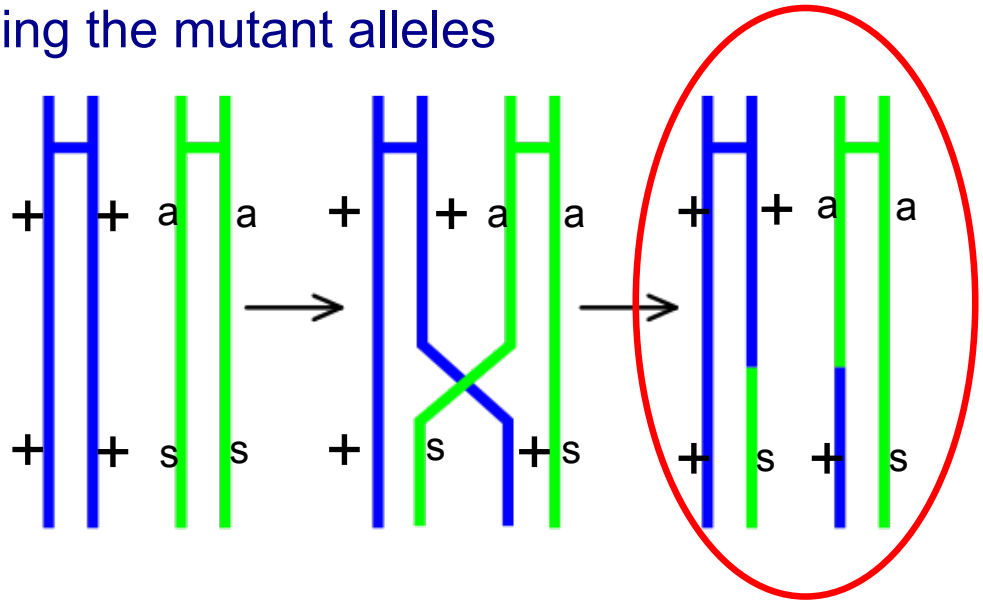
Parental type



Recombinations

Recombination Frequency

Crossing over can separate genes that are typically on the same chromosome. In this case the maternal chromosome containing the wild type crossed over with homologous pair containing the mutant alleles



The individuals that received the parental type genomes had no crossover event. The ones the received recombined traits received crossed over chromosomes.

Recombination Frequency

Recombination frequency is the average percentage of cross over events that occur between linked genes.

$$\frac{\text{number of recombinations}}{\text{number of offspring}} = \text{Recombination Frequency}$$

Use the actual results from the previous experiment to calculate the recombination frequency of the 2 linked genes.

22,000 offspring produced
10,855 both traits wild type
10,930 both traits mutant
110 blue spots short antenae
105 no spots long antenae

Genetic Linkage Maps

After further observation and experiments a third difference was recognized between the wild type and mutant caterpillars.

Wild Type

Short Antennae = +

No spots = +

Mutant

Long Antennae = a

Spots = s

Genetic Linkage Maps

When placed near a bright light source the wild type moved toward it. The mutant moved away.

Wild Type

Short Antennae = +

No spots = +

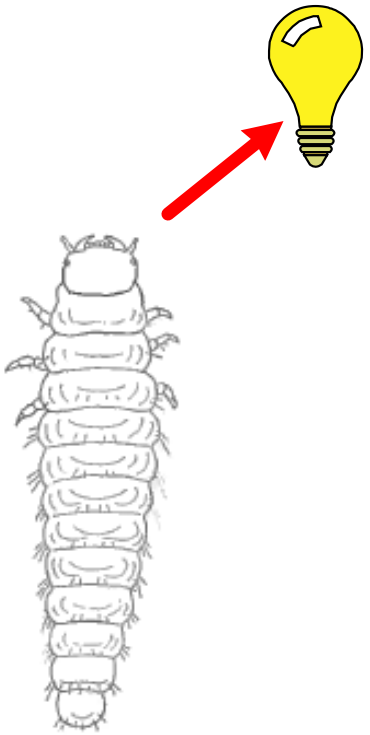
Mutant

Long Antennae = a

Spots = s

Genetic Linkage Maps

This behavioral trait was recognized as a third linked gene and new notation produced.

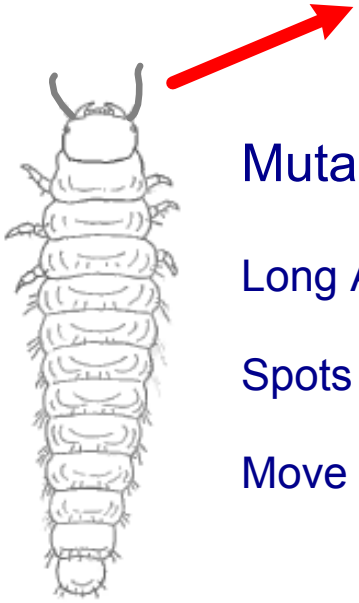


Wild Type

Short Antennae = +

No spots = +

Move to light = +



Mutant

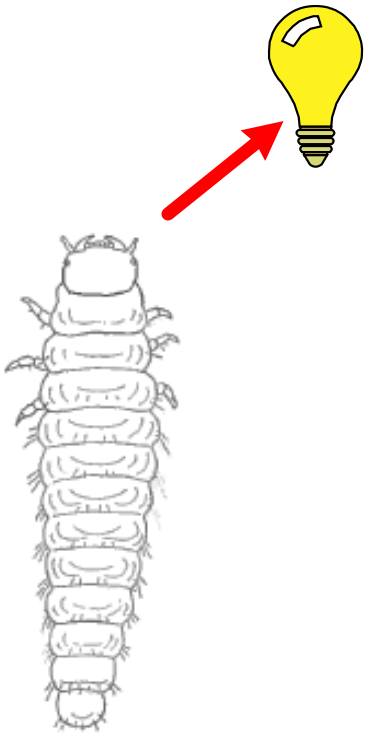
Long Antennae = a

Spots = s

Move from light = m

Genetic Linkage Maps

Using the previous experiment as a model, design experiments that will show the recombination frequency of the new trait and each of the previously tested mutant traits.

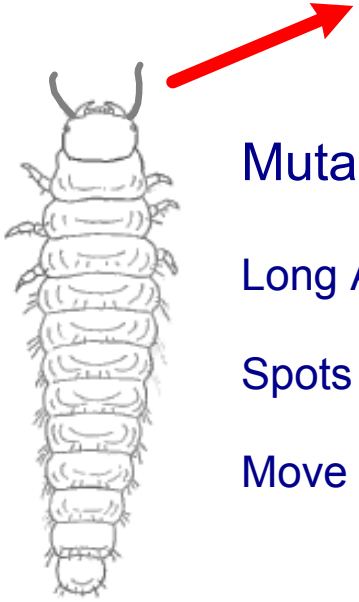


Wild Type

Short Antennae = +

No spots = +

Move to light = +



Mutant

Long Antennae = a

Spots = s

Move from light = m

Genetic linkage maps

After you run your experiments you get the following data:

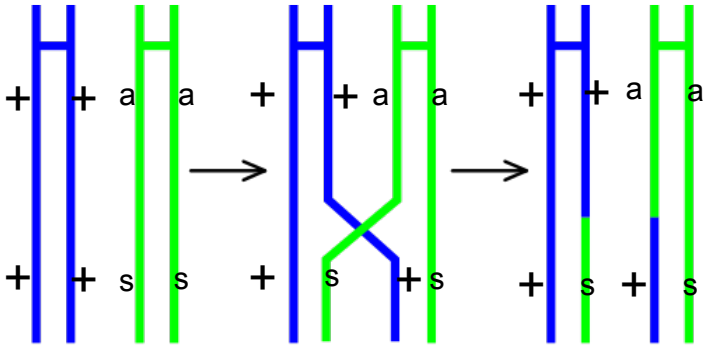
Rf	a	s	m
a	x	1%	3%
s	x	x	2%
m	x	x	x

What does this tell us about the position of these genes on the common chromosome?

Genetic linkage maps

Remember that the amount of times that a crossing over event happens **between 2 genes** correlates to the recombination frequency.

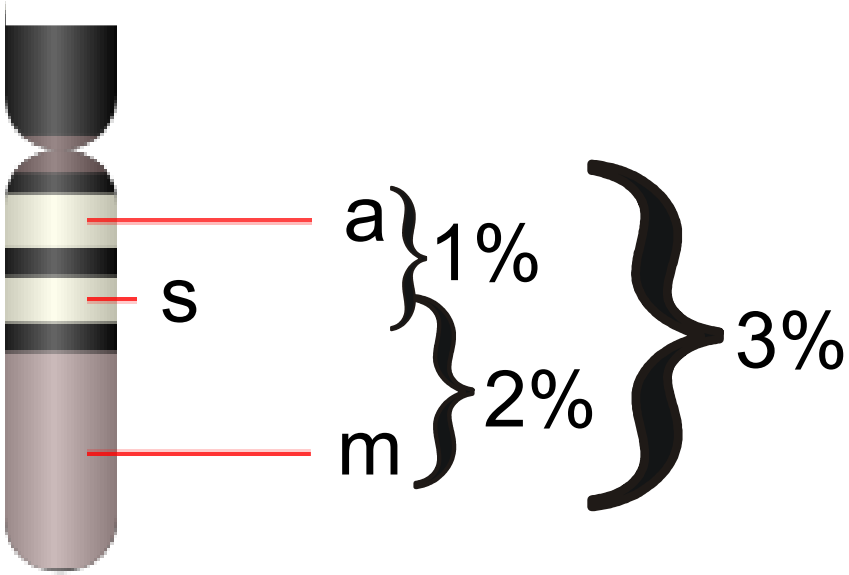
Rf	a	s	m
a	x	1%	3%
s	x	x	2%
m	x	x	x



Genetic linkage maps

Recombination frequency can show the relative positions of genes on a chromosome. The higher the Rf the further apart the genes. In this case...

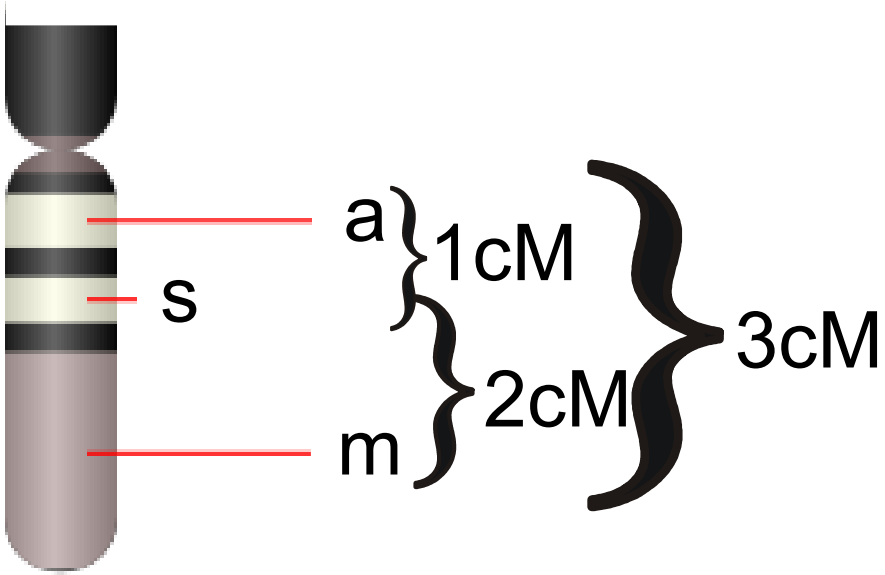
Rf	a	s	m
a	x	1%	3%
s	x	x	2%
m	x	x	x



Genetic linkage maps

Since recombination frequency directly relates to distance, Rf can be converted to a unit of distance known as a **centimorgan (cM)**, also called a **map unit (mu)**

Rf	a	s	m
a	x	1%	3%
s	x	x	2%
m	x	x	x



Genetic linkage maps

Construct a linkage map for the following table of map units. Start by drawing a line to represent a chromosome. Then use a ruler to create a scale (ex- 1mu = 1/2centimeter).

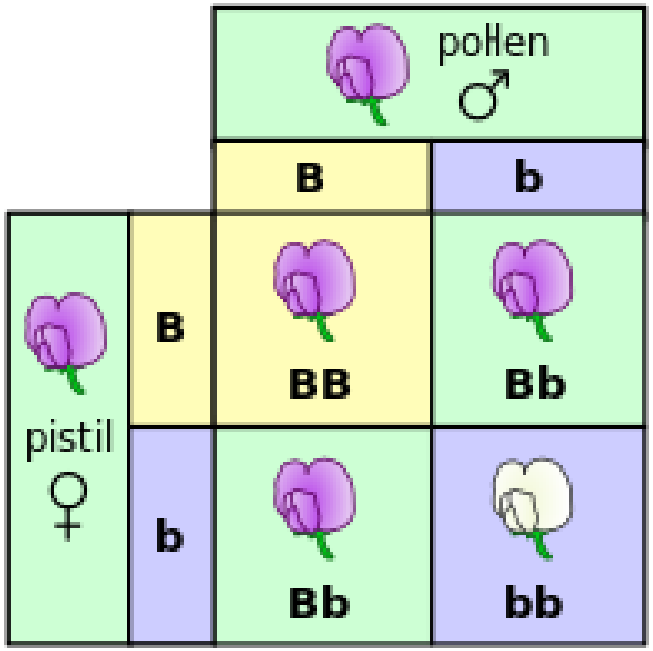
	Gr	*Rc*	*S*	*Y*	*P*	*oa*
Gr	—	25	1	19	7	20
Rc	25	—	26	6	32	5
S	1	26	—	20	6	21
Y	19	6	20	—	26	1
P	7	32	6	26	—	27
oa	20	5	21	1	27	—

Probability, Statistics and Genetics

Return to Table of Contents

Genetics is based on prediction

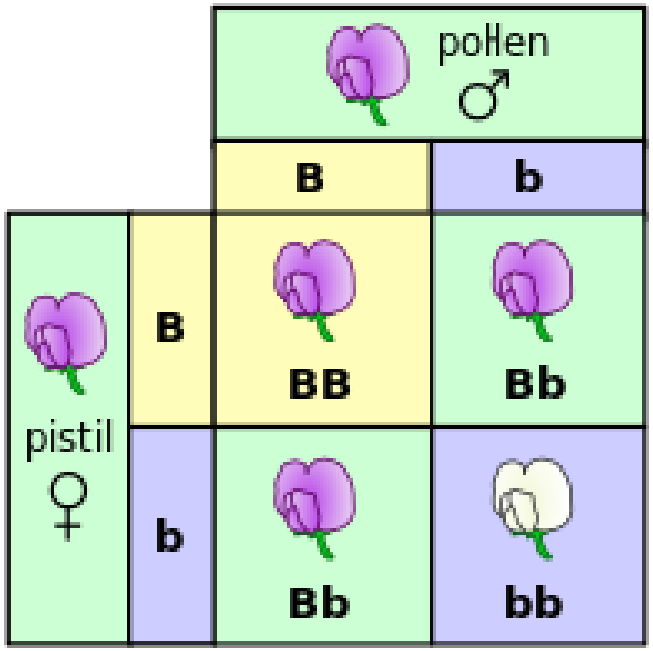
The study of genetics relies on the comparison of **expected values** and **observed values** based on actual experimental observation.



For example, if we cross 2 individuals with different phenotypes for a trait we **expect** to get a 3:1 phenotype ratio. This is because we have established a rule based on the observations of Mendel.

Genetics is based on prediction

But what if we don't get exactly 3:1? Did we prove Mendel wrong? Look at the numbers below that were generated by an actual experiment. What are the expected values? Do the observed results adhere to Mendel's rules?



10,000 offspring produced

6,860 purple flowers
3,200 white flowers

Chi Squared Test

The Chi-Squared Test is designed to help us decide if the difference between the observed results and expected results of an experiment are **statistically significant.**

In other words, it tells us if the difference is due to random chance or if it is due to some other factor that effected the results of our experiment.

Chi Squared Test

This is the formula:

$$\chi^2 = \sum \frac{(O - E)^2}{E}$$

χ^2 = Chi Squared $\sum$ = the sum of

O = Observed frequencies E = Expected frequencies

Chi Squared Test $X^2 = \sum \frac{(o-e)^2}{e}$

A simple example:

The laws of probability state that if we flip a coin 100 times we should get 50 heads and 50 tails. These are our expected results (e).

However, when we actually flip a coin 100 times we get (o) =

56

44

Chi Squared Test $X^2 = \sum \frac{(o-e)^2}{e}$

Is this difference because of chance? Or is it because something is effecting the outcome? For example, the way it is being flipped or weight imbalance causing more heads to be flipped.

56

44

Chi Squared Test $X^2 = \sum \frac{(o-e)^2}{e}$

This is where Chi Squared is used. The test is assessing the **null hypothesis** which states: **There is no significant difference between the observed and expected frequencies.**

56

44

Chi Squared Test $X^2 = \sum \frac{(o-e)^2}{e}$

The first step is to get a value for X^2.

$$X^2 = \frac{(56 - 50)^2}{50} + \frac{(44 - 50)^2}{50} = \frac{72}{50} = 1.44$$

56

44

Chi Squared Test $X^2 = \sum \frac{(o-e)^2}{e}$

1.44

No that we have our value for Chi Squared, we can compare it to a statistical analysis chart known as **critical values.**

VassarStats: Critical Values of Chi-Square

	Level of Significance			
df	0.05	0.025	0.01	0.005
1	3.84	5.02	6.63	7.88
2	5.99	7.38	9.21	10.60
3	7.81	9.35	11.34	12.84
4	9.49	11.14	13.28	14.86
5	11.07	12.83	15.09	16.75
6	12.59	14.45	16.81	18.55
7	14.07	16.01	18.48	20.28
8	15.51	17.53	20.09	21.95
9	16.92	19.02	21.67	23.59
10	18.31	20.48	23.21	25.19

Chi Squared Test $X^2 = \sum \frac{(o-e)^2}{e}$

1.44

First we must figure out our **degrees of freedom (df).** This is simply the amount of possible outcomes minus 1. For this example: **2-1 = 1df**

VassarStats: Critical Values of Chi-Square

	Level of Significance			
df	0.05	0.025	0.01	0.005
1	3.84	5.02	6.63	7.88
2	5.99	7.38	9.21	10.60
3	7.81	9.35	11.34	12.84
4	9.49	11.14	13.28	14.86
5	11.07	12.83	15.09	16.75
6	12.59	14.45	16.81	18.55
7	14.07	16.01	18.48	20.28
8	15.51	17.53	20.09	21.95
9	16.92	19.02	21.67	23.59
10	18.31	20.48	23.21	25.19

Chi Squared Test $X^2 = \sum \frac{(o-e)^2}{e}$

1.44

Next we decide our level of significance. Usually it is .05, which means that we are 95% certain of our outcome. If a higher standard is needed another level could be used.

VassarStats: Critical Values of Chi-Square

	Level of Significance			
df	0.05	0.025	0.01	0.005
1	3.84	5.02	6.63	7.88
2	5.99	7.38	9.21	10.60
3	7.81	9.35	11.34	12.84
4	9.49	11.14	13.28	14.86
5	11.07	12.83	15.09	16.75
6	12.59	14.45	16.81	18.55
7	14.07	16.01	18.48	20.28
8	15.51	17.53	20.09	21.95
9	16.92	19.02	21.67	23.59
10	18.31	20.48	23.21	25.19

Chi Squared Test $X^2 = \sum \frac{(o-e)^2}{e}$

1.44 <

VassarStats: Critical Values of Chi-Square

	Level of Significance			
df	0.05	0.025	0.01	0.005
1	3.84	5.02	6.63	7.88
2	5.99	7.38	9.21	10.60
3	7.81	9.35	11.34	12.84
4	9.49	11.14	13.28	14.86
5	11.07	12.83	15.09	16.75
6	12.59	14.45	16.81	18.55
7	14.07	16.01	18.48	20.28
8	15.51	17.53	20.09	21.95
9	16.92	19.02	21.67	23.59
10	18.31	20.48	23.21	25.19

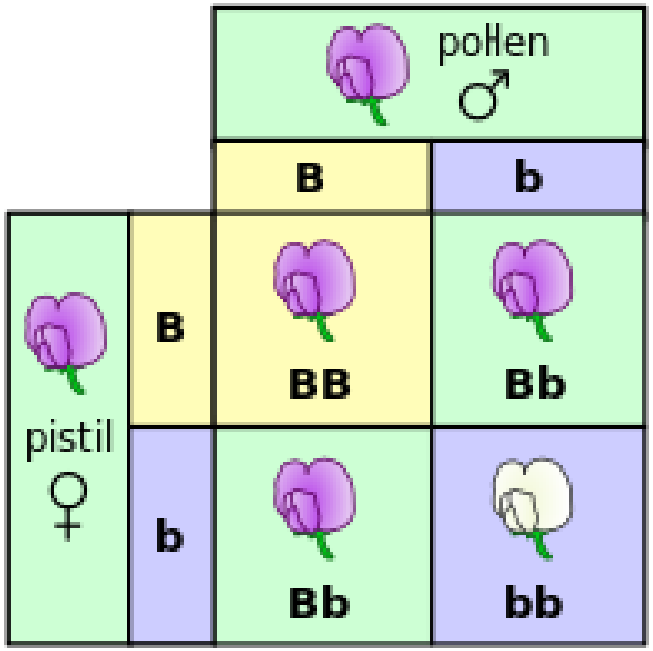
3.84 is our critical value. If X^2 is **less** than the critical value we **accept** the null hypothesis.

If it is **more** than we **reject** the null hypothesis

null hypothesis- There is no significant difference between the observed and expected frequencies.

Genetics is Based on Prediction

Now that you know chi squared, does this observed data meet with the expectations set by Mendel?

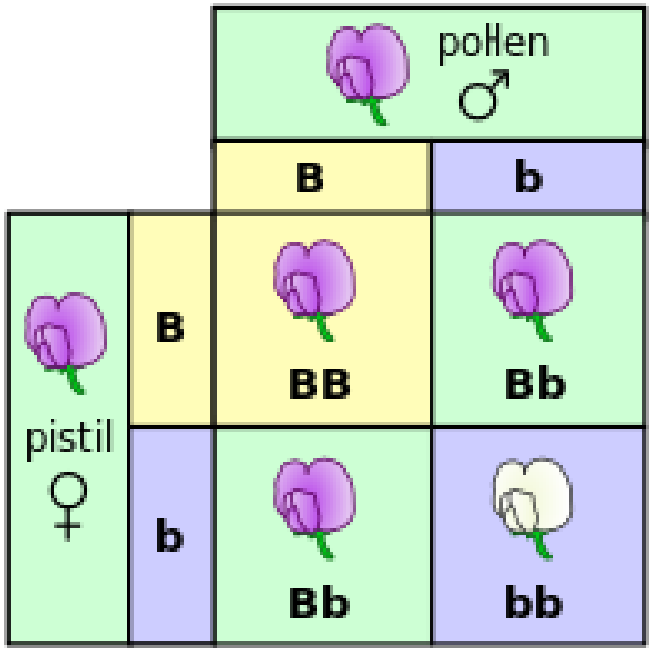


10,000 offspring produced

6,860 purple flowers
3,200 white flowers

Genetics is Based on Prediction

What if we look at genotype? Does this observed data coincide with Mendel's postulates?



10,000 offspring produced

2,930 Homozygous dominant
3,870 Heterozygous
3,200 Homozygous recessive

Using Probability Rules When Solving Genetics Problems

There are two rules of probability which are helpful when solving problems in genetics:

The Multiplication Rule

The Addition Rule

The Multiplication Rule

The **multiplication rule** states that the probability that two or more independent events will occur together is the product of their individual probabilities

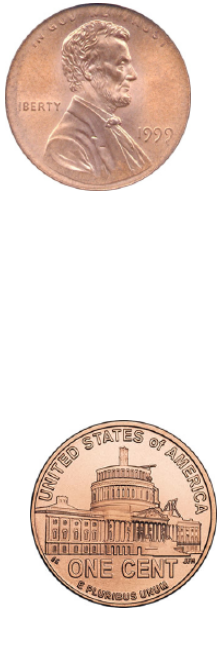
Probability in an F_1 monohybrid cross can be determined using the multiplication rule. Segregation in a heterozygous plant is like flipping a coin: Each gamete has a 1/2 chance of carrying the dominant allele and a 1/2 chance of carrying the recessive allele

The Multiplication Rule

The **multiplication rule** states that the probability that two or more independent events will occur together is the product of their individual probabilities

Probability in an F_1 monohybrid cross can be determined using the multiplication rule. Segregation in a heterozygous plant is like flipping a coin: Each gamete has a 1/2 chance of carrying the dominant allele and a 1/2 chance of carrying the recessive allele

The Multiplication Rule



20 If we toss two coins at the same time, what is the chance that both coins will land heads up?

21 In an F_1 cross between two pea plants that are heterozygous (Ww) for purple flowers, what is the probability that a particular offspring of this cross will have the ww genotype (white flowers)?

22 Mendel stated that each pair of alleles segregates independently of other pairs of alleles during gamete formation. What is this law known as?

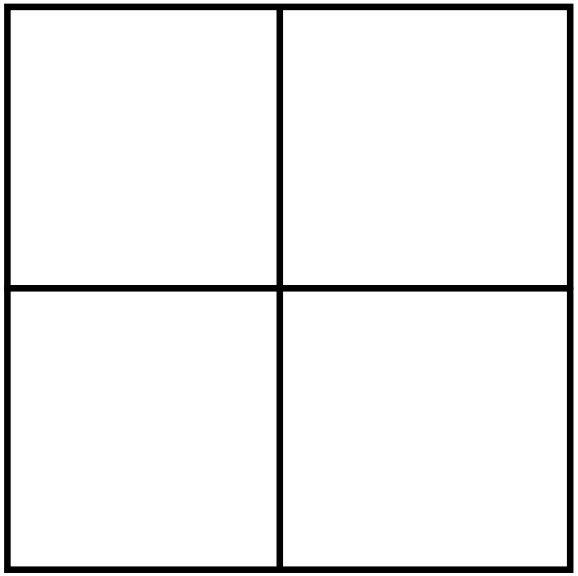
- A Law of Segregation.
- B Law of Independent Assortment.
- C Law of Probability.
- D Law of Pea Plant Genetics.

23An organism heterozygous for two characteristics is a _____.

- A dihybrid
- B monohybrid
- C homozygote
- D double dominant

Heterozygous Offspring

Now let us consider the probability that an F_2 pea plant will be heterozygous (Ww) for flower color. There are two ways in which F_1 gametes can combine to produce heterozygous offspring.
Draw a Punnett square to confirm this ...



The Rule of Addition

As we can see from our Punnett square, there are two ways in which F_1 gametes can combine to produce heterozygous offspring.

The dominant allele can come from the egg and the recessive allele from the sperm,

or ...

The dominant allele can come from the sperm and the recessive allele can come from the egg.

The rule of addition states the probability that an event can occur in two or more alternative ways is the sum of the separate probabilities of the different ways.

24 Use the rule of addition to calculate the probability of an F_2 heterozygote.

Trihybrid Crosses

We can use the rules of probability to solve complex genetics problems.

If we crossed two organisms both having the genotype AaBbCc, what is the probability that an offspring of this cross will have the genotype aabbcc? Since each allele pair assorts independently, we can treat this trihybrid cross as three separate monohybrid crosses:

Aa X Aa: Probability of aa offspring = _____

Bb X Bb: Probability of bb offspring = _____

Cc X Cc: Probability of cc offspring = _____

aabbcc

Because the segregation of each allele pair is an independent event, we can use the multiplication rule to calculate the probability of an offspring being aabbcc.

What is the probability?

25 If you cross two F_1 individuals both having the genotype AaBbCc, what is the probability that an F_2 offspring will have the genotype AabbCc?

26A plant with genotype AABbCC is crossed with a plant having genotype AaBbCc. What is the probability of an offspring having the genotype AABBCC?