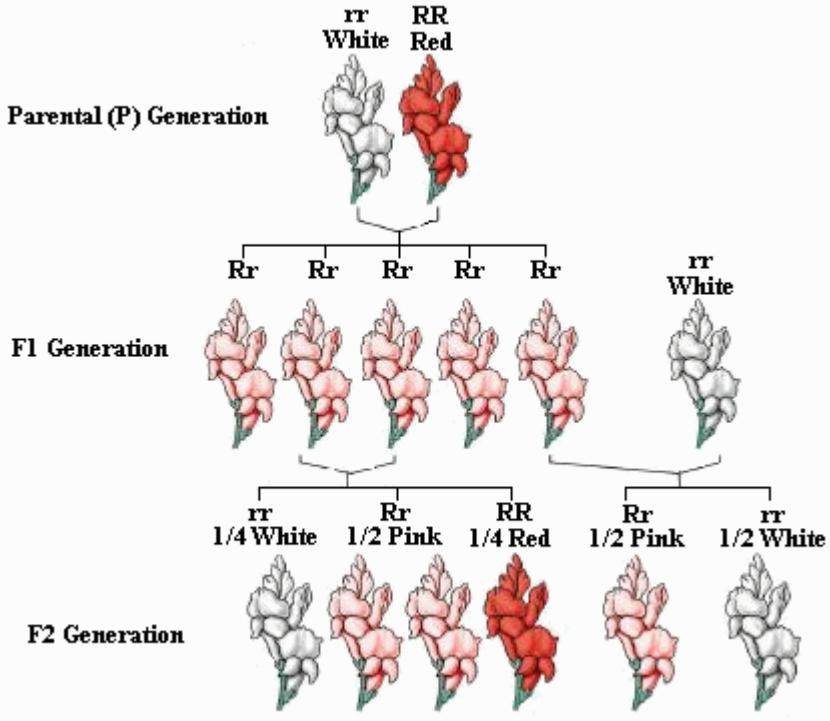
Genetics Part I

Biology 40S

Mrs. Kornelsen



What is Biology?

What is Alive?

We are alive, we talk about living things all the time. We also use the word life regularly, but what do we really mean by these words life, alive and living?

We all know what a plant is, what an animal is, and that the two are not the same. If we are pushed for definitions we may say that plants sit still and get energy from the sun via photosynthesis, while animals move around and get their energy from eating other living things, either dead or alive.

This is relatively true and most living things are either animals, plants or something else such as fungi or bacteria. However there are animals that don't move, there are living things which get there energy both from the sun via photosynthesis and by eating other organisms, and some plants wave there limbs around slowly and even move around as a whole organism, either floating in water or blown on the breeze.

join host BOB MCDONALD
QUIRKS & QUARKS

January 23, 2010

Solar Powered Sea Slug


courtesy Nick Curtis and Ray Martinez

Here's a joke. What's green and photosynthetic? Well, a sea slug, of course. OK, that's not very funny, but it is pretty interesting. **Dr. Sidney (Skip) Pierce** of the Department of Integrative Biology at the University of South Florida has discovered that a sea slug has somehow developed the ability to photosynthesize - and live out its life as a solar-powered animal. Apparently, the slug has stolen essential genes from a marine algae, and integrated them into its own genome. These genes allow it to make chlorophyll and important enzymes that are essential for photosynthesis. It can't do it all on its own, though, so early in life, it has to eat algae and steal their solar-energy producing chloroplasts - the energy factories of the plant cell. It then can supply these chloroplasts with the raw materials for photosynthesis, and bask in the sun - and a life of leisure.

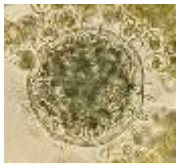
http://www.cbc.ca/quirks/archives/09-10/qq-2010-01-23.html

Characteristics of living things:

One of the big questions in biology is defining what life is. This is hard to do, so biologists often focus on describing the key characteristics of life.

- _____
- _____
- _____
- _____
- _____
- _____
- _____
- _____
- _____
- _____

All the things that science currently accepts as living exist as either single cells, or as a collection of cells working together (unicellular life or multicellular life). These cells exist as an area of cytoplasm enclosed by a membrane. All use DNA as there means of storing information concerning the mechanics and methods of reproduction. All contain proteins of some sort.



Your task: to create a life form; a brand new living thing.

It must fit into one of the following classifications for life scientists have roughly distinguished.

Bacteria – bacteria like the strep throat or E. coli
Fungi – fungus like mushrooms or yeast
Plantae – trees, flowers, or moss
Animalia - animals

Some questions you might want to think about are listed below in the chart. The organism you create must make sense; for example, an animal with wings wouldn't live underwater. You might choose to make a hybrid animal (characteristics from two different animals) like a snat…a snake/rat.

Put your ideas below in point form. List all the possibilities for each column to help you brainstorm unique ideas. For example, 'how does your life form obtain energy', it eats, it decomposes other life, it converts chemicals, photosynthesis, etc.

There is a version of the chart along with a rubric for you to hand in as a group for marks. The good copy of your chart only needs to include the final decision for your life form (for example, photosynthesis as a way to obtain energy). Your chart must include explanations for why your organism has a specific adaption, why it looks the way it does, why it eats what it does, or why it reproduces the way it does. The more info you give me the better I will get to know your organism.

Where does your organism live?	How does your life form obtain energy?	How does your life form reproduce?	What does your life form look like?	What special features does your life form have to help it adapt to its living environment?

Finally, build your new life form. Use any materials you would like, and have fun. The more creative the better.

What I need from you (checklist):

1) A life form
2) Good copy of your hand-in assignment page with explanations
3) Your organism's new scientific name

What do we remember from grade 9 science?

1. What is the difference between the following terms:
    a. dominant and recessive?

    b. genotype and phenotype?

    c. gene and allele?

2. What is the genotype of a person with blue eyes? Explain.

3. Could a person with brown eyes have another genotype other than BB? Explain.

4. What is a…
    a. diploid cell?

    b. haploid cell?

    c. gamete?

    d. Zygote?

Learning checklist – Genetics Part I

Learning increases when you have a goal to work towards. Use this checklist as guide to track how well you are grasping the material. In the center column, rate your understand of the topic from 1-5 with 1 being the lowest and 5 being the highest. Be sure to write down any questions you have about the topic in the last column so that you know what you have yet to learn.

Outcomes	Understanding	Questions
Outline Mendel's principles of inheritance, stating their importance to the understanding of heredity. Include: principles of segregation, dominance, independent assortment		
Explain what is meant by the terms heterozygous and homozygous.		
Distinguish between genotype and phenotype and use these terms appropriately when discussing the outcomes of genetic crosses.		
Use Punnett squares to solve a variety of autosomal inheritance problems and justify the results by using appropriate terminology. Include: monohybrid cross, dihybrid cross, testcross, P generation, F_1 generation, F_2 generation, phenotypic ratio, genotypic ratio, dominant alleles, recessive alleles, purebred, hybrid, carrier		
Describe examples and solve problems involving the inheritance of phenotypic trais that do not follow a dominant-recessive pattern. *Examples: co-dominance, incomplete dominance, multiple alleles, lethal genes ...*		
Explain the basis for sex determination in humans. Include: XX and XY		
Describe examples and solve problems involving sex-linked genes. *Examples: red-green colour-blindness, haemophilia, Duchenne*		

muscular dystrophy		
Use pedigree charts to illustrate the inheritance of genetically determined traits in a family tree and to determine the probability of certain offspring having particular traits. Include: symbols and notation used		
Discuss ethical issues that may arise as a result of genetic testing for inherited conditions or disorders.		
Discuss the role of meiosis and sexual reproduction in producing genetic variability in offspring. Include: crossing-over, randomness		
Explain how chromosome mutations may arise during meiosis. Include: non-disjunction		
Identify monosomy and trisomy chromosome mutations from karyotypes. *Examples: Down syndrome, Turner syndrome, Klinefelter syndrome*		

Sort and Predict

Unit _____ Topic _____

Directions:

Read the list of words on the left and sort them into four different categories by placing them in the boxes. For the words that you are unsure of, predict which category each would belong to. When selecting categories, try to make the fourth category different than any category that the rest of the class would think of. Use your creativity; be original! You may use **one** word in more than one category.

- Carrier - Incomplete dominance - Punnett square - Duchenne muscular dystrophy - Dominant - Recessive - Phenotype - Genotype - Red colour-blindness - Hybrid - Hemophilia - Co-dominance - Gene - Allele - Trait	1. _____	2. _____
	3. _____	4. _____

Sort and Predict Frame: Used by permission of Lynda Matchullis and Bette Mueller, Nellie McClung Collegiate, Pembina Valley S.D. No. 27, Manitoba.

Introduction to Genetics

For millennia, people have known that offspring inherit traits (characteristics) from parents. This knowledge has been used to improve the likelihood of breeding the best animals and plants. However, the underlying mechanisms that control heredity were unknown until the middle of the 1800s. While people knew that many traits bred true and could be consistently expected in offspring, there were many examples of traits that disappeared over time. There were also situations where offspring had a trait not held by either parent (for example, a blue-eyed child born to brown-eyed parents).

Acquired or Inherited Traits?

Think of traits that a person can inherit from his or her parents and write them down on the first list ("Inherited"). Then, make another list of traits that are not inherited from parents. For example, think about the way you behave. Is the way you behave inherited from your parents or is it something you learn and acquire?

The traits in the second list (that are not inherited from your parents) are called acquired traits. These characteristics have not been passed on to you from your parents. Rather, you have acquired them because of your environment or some other factor.

Inherited	Acquired

The "nature versus nurture" debate has raged for a long time. Exactly which traits are inherited, and to what degree they are inherited is still not clear in the case of certain traits. For example, there have been efforts to find a gene for criminality or addictive personalities, but none have been proven to date.

Sometimes we blame genetics for something that in reality has nothing to do with genes. Parents sometimes want to take credit for passing on good qualities or talents to their children, when in reality the talent is the result of a nurturing and supportive environment as opposed to genetic endowment. Think about the following questions:

If someone is considered "accident prone," is this characteristic inherited or is it something the person acquires?

If one parent decides to get into shape by lifting weights and develops large muscles, will the children inherit large muscles without having to lift weights?

A child has a wonderful musical ability but neither parent even plays an instrument. Is this an inherited or acquired trait?

Inherited traits PowerPoint.

Questions

1. If both parents have brown eyes, what colour of eyes will the children inherit (best guess)?

2. If one parent had an accident and lost a finger (has only nine fingers), how many fingers will the children inherit?

3. If one parent loves the outdoors, will all of the children be "outdoors types?"

4. If a blue-eyed man and a brown-eyed woman have four children with brown eyes, will their fifth child also have brown eyes? Explain as best you can.

5. Is it possible for a person to have one brown eye and one blue eye (glass eyes or coloured contacts notwithstanding)?

Meiosis and Genetic Variation

How is it possible that genes from two parents unite to form a new life that still has the critically correct number of chromosomes? To understand the transfer of traits from parents to offspring, a clear concept of the nature of DNA, chromosomes, and genes is vital.

We will begin by looking at a special kind of cell division called meiosis. Meiosis is from the Greek word for "diminution," which means to make smaller. This is the process that results in egg and sperm cells with one half of the normal number of chromosomes. Egg and sperm unite to form a new individual with the correct amount of genetic material.

Most cells in the human body have 46 chromosomes. This is called a **diploid** cell. When new cells are made, they also have 46 chromosomes. The process of mitosis describes how this is done. Forty-six is the normal number of chromosomes found in human cells; this is called the diploid number. Any more or less causes abnormalities.

If a sperm has 46 chromosomes and fertilizes an egg with 46 chromosomes, you would get a cell with 92 chromosomes. This cell would not survive and therefore, there would be no new life. This is because each cell must have 46 chromosomes--no more, no less.

A special type of cell division produces sperm and egg cells with half the diploid number. Having half the normal number of chromosomes is called **haploid**. In effect, a sperm cell ends up with 23 chromosomes and fertilizes an egg with 23 chromosomes. This new cell is the beginning of life and it has 46 chromosomes--the normal number. The special type of cell division that ensures that each sperm and egg has the haploid number of chromosomes is called **meiosis**.

Meiosis Resources:

1) *Comparing Mitosis and Meiosis - http://www.pbs.org/wgbh/nova/baby/divi_flash.html*
2) *Mitosis - http://www.teachersdomain.org/asset/lsps07_int_celldivision/*
3) *Great Meiosis animation http://www.biostudio.com/d_%20Meiosis.htm*
4) *http://www.johnkyrk.com/meiosis.html*

Homologous Chromosome: In a biological cell, a chromosome pairs with another chromosome during meiosis. This pairing happens between two chromosomes that are homologous, i.e. chromosomes having the same genes at the same loci but possibly different alleles. For example, two chromosomes may have genes encoding eye color, but one may code for brown eyes, the other for blue.

Sister chromatid - Two identical strands joined by a common centromere as a result of a chromosome that duplicated during interphase.

Centromere - The constricted region joining the two sister chromatids that make up an X-shaped chromosome.

Online Activity:

Go to the location written on the board. Follow the steps of meiosis and answer the following questions:

1) With the help of your textbook (p. 199-205), write down what happens in the following steps. At each step, include the number of chromosomes as well as whether the cell is haploid or diploid.

Interphase: Before meiosis begins, _____.

First division of meiosis (Meiosis I)

a. **Prophase 1:**

Number of Chromosomes:

b. **Metaphase 1**:

Number of Chromosomes:

c. **Anaphase 1**:

Number of Chromosomes:

d. **Telophase 1**:

Number of Chromosomes:

2) **Second division of meiosis: Gamete formation** (Meiosis II)

a. **Prophase 2**:

Number of Chromosomes:

b. **Metaphase 2**:.

Number of Chromosomes:

c. **Anaphase 2**:

Number of Chromosomes:

d. **Telophase 2**:

Number of Chromosomes:

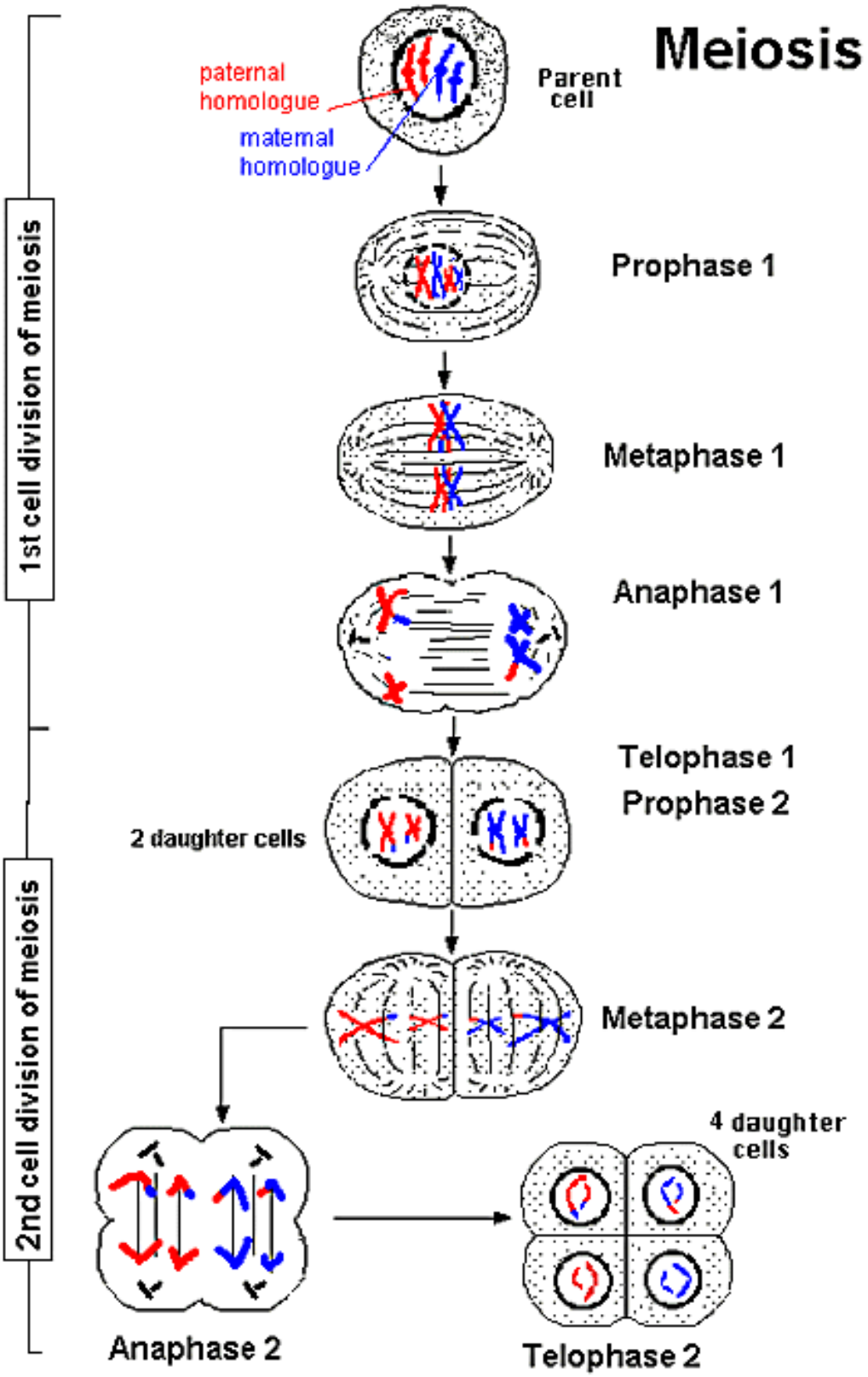
3) Using the website *http://www.pbs.org/wgbh/nova/baby/divi_flash.html,* outline the differences and similarities between meiosis and mitosis.

Mitosis	Meiosis
Similarities:	
Differences:	

4) How do you think meiosis adds to the genetic variability in human offspring?

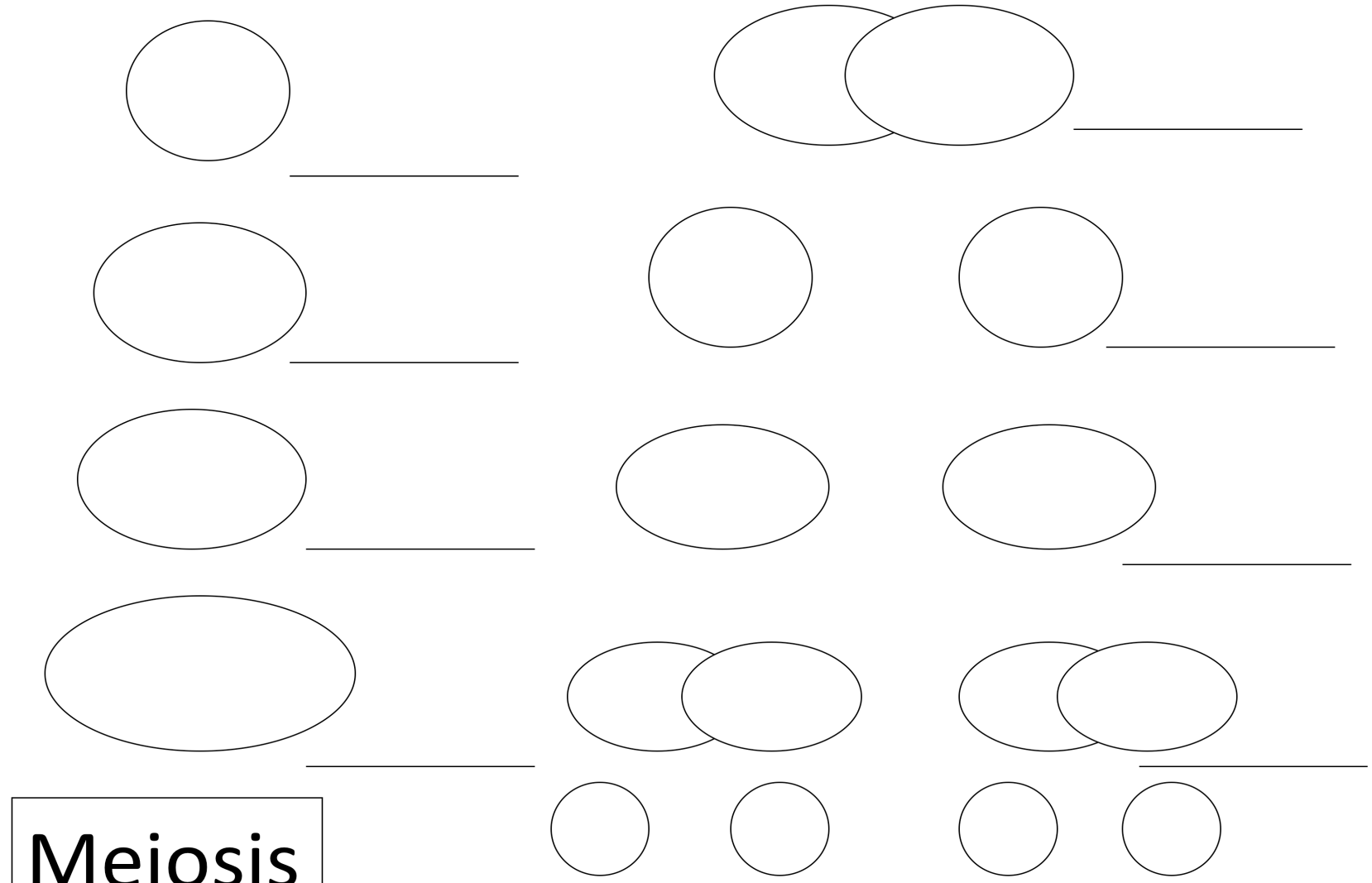
5) When does crossing-over occur? How does this add to the genetic variability in human offspring?

6) When you are finished, draw the steps on meiosis on the diagram provided.


Meiosis
paternal homologue
maternal homologue
Parent cell
Prophase 1
Metaphase 1
Anaphase 1
Telophase 1
Prophase 2
2 daughter cells
Metaphase 2
4 daughter cells
Anaphase 2
Telophase 2
1st cell division of meiosis
2nd cell division of meiosis

Please label the following:

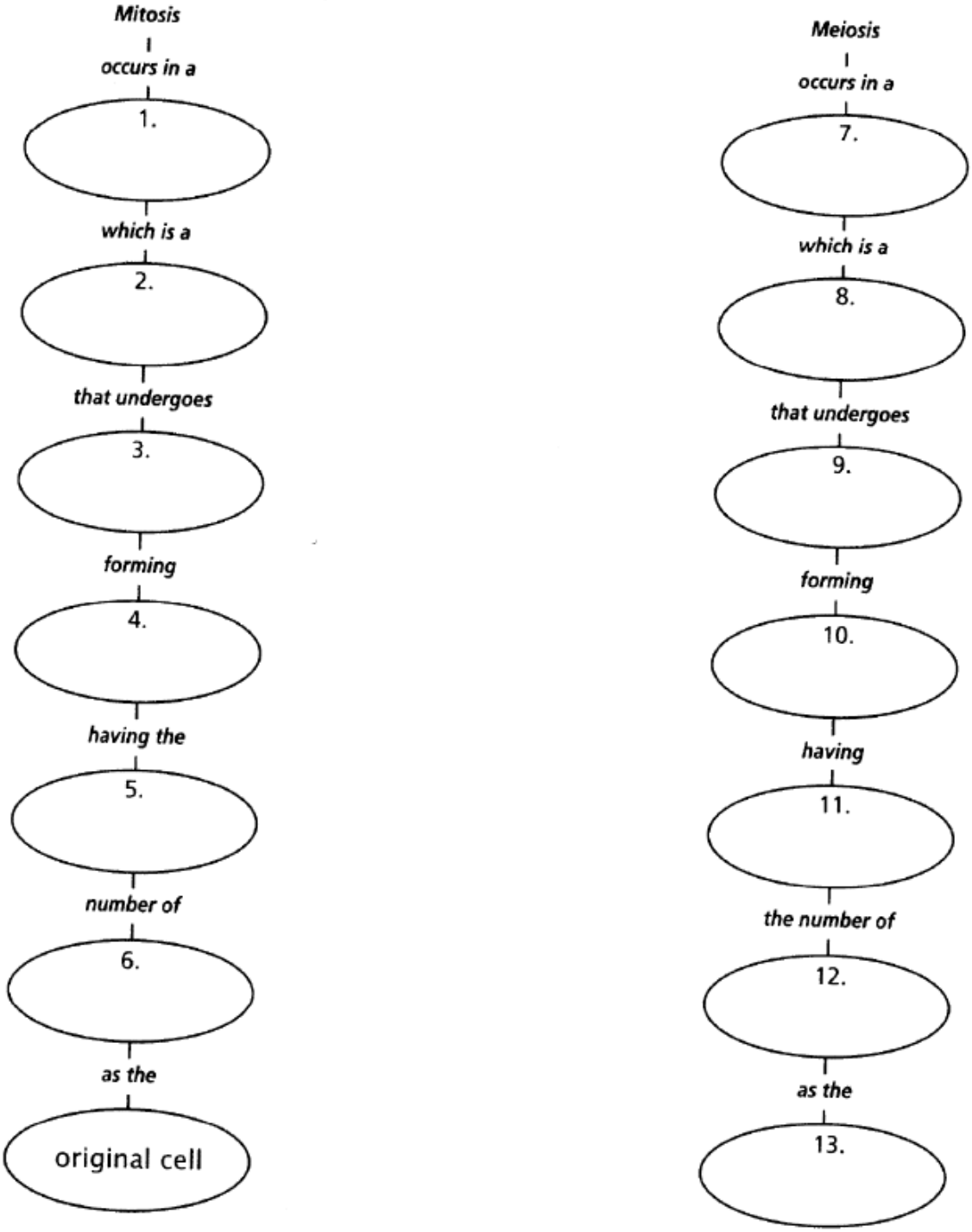
Homologous pair, All of the phases, Centromere, Gametes, Meiosis I, Meiosis II, Diploid, Haploid



Meiosis

Mitosis/Meiosis

Complete the concept map comparing mitosis and meiosis. Use these words or phrases one or more times: *diploid cell, one cell division, four haploid cells, original cell, two cell divisions, body cell, same, chromosomes, gamete-producing cell, half, two diploid cells.*



Genetic Variability and Meiosis

The reason we learn about meiosis is to understand how people can have such great diversity.

Great Resource for Meiosis on YouTube:
Khan's Academy
Example:
http://www.youtube.com/watch?v=ijLc52LmFQg&feature=channel

- ☐ Based on only the random separation of homologous chromosomes during meiosis, a person with _____ _____ can theoretically produce about 8 million different haploid gametes.
- ☐ Add in _____ and the chance of any two gametes being the same is highly _____
- ☐ When two _____, the newly formed _____ contains a full set of chromosomes, or the diploid number of chromosomes. Thus, meiosis allows _____ _____ to take place

Alleles

Every somatic (body) cell has sets of homologous chromosomes. In human cells, there are _____ chromosomes in the nucleus of each cell. There are _____ pairs of chromosomes.

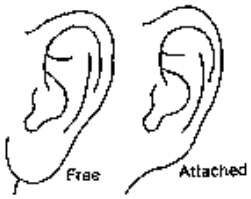
Great Online Resources for Genetics:

1) DNA from the Beginning
www.dnaftb.org

One out of the 23 pairs is an exception--the pair of _____. This pair is different because they do not appear homologous; in other words the chromosomes of this pair are _____ _____.

However, each of the other 22 pairs (_____) are considered to be homologous chromosomes because _____ _____.

There are alternate forms of a gene. For instance, the gene that determines whether the earlobe is free or attached has two forms: one form for free earlobes; another for attached earlobes.



These alternate forms of a gene are called alleles. They occupy the _____ _____ _____. The following diagram shows the location of the gene on a pair of homologous chromosomes.

Although there are two alleles for the trait of having free or attached earlobes, you will only show one trait. That is, you will either have free earlobes or you will have attached earlobes. You cannot have both types. This means that if the alleles are different, only one of them will express itself. The one allele that _____. The other allele, that _____.

In the case of the trait of having free or attached earlobes, the allele for free earlobes will dominate over the allele for attached earlobes. In other words, free earlobes are dominant and attached earlobes are recessive. Most human traits can be explained this way.

Genotype and Phenotype

The genetic make-up of an organism is its genotype. We indicate the genotype by letters of the alphabet. An _____ represents a dominant allele and a _____ _____ represents a recessive allele.

The phenotype is the way the trait actually appears in the organism.

The letters are written in pairs (e.g., GG or Gg) because the alleles on homologous chromosomes also occur in pairs (one on each chromosome).

The lower case *cc* labelling on homologous chromosomes indicates that the individual is _____.

The homologous chromosomes labelled in upper case *CC* indicate that the individual's genotype is _____.

If one allele is dominant and the other is recessive (labelled *Cc*) then we say the individual's genotype is _____.

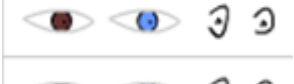
Using the ear lobe example, if someone is homozygous dominant CC, what phenotype is displayed? How about homozygous recessive cc? or heterozygous Cc?

Which of the following genotypes are homozygous or heterozygous?

Genotype	Homozygous	Heterozygous
Aa	○	○
AA	○	○
RR	○	○
Rr	○	○
rr	○	○
Yy	○	○
YYgg	○	○
YyGg	○	○

For each genotype, choose the matching phenotype. (on the right)

C - dominant brown eye
c - recessive blue eye
E - dominant free lobe
e - recessive attached lobe

Genotype	Phenotype
CCEE	
CcEE	
CCEe	
CcEe	
cCEE	
ccEE	
cCEe	
ccEe	
CCeE	
CceE	
CCee	
Ccee	
cCeE	
cceE	
cCee	
ccee	

It is important to note that it is not possible to directly observe an individual's genotype. We can only observe the phenotype and infer the genotype. This learning activity is opposite to real life. However, we do it this way to learn how to decode genotypes.

If we treat each allele independently, there are 16 possible genotypes but only four different phenotypes. Thus when studying inheritance, it is important to know both the phenotypes and genotypes.

Questions

1. If an animal has 20 as its diploid (2n) number, how many chromosomes would be present in a sperm (haploid, n)? An egg (haploid, n)?

2. Describe the process of synapsis. What is it, when does it happen, and why does it happen?

3. What is the difference between genotype and phenotype?

4. Use the letter 'H' to represent the following genotypes: homozygous dominant, homozygous recessive, and heterozygous.

5. If brown eyes are dominant over blue eyes and represented by the letter 'C', and free earlobes are dominant over attached earlobes and represented by the letter 'E', what does the genotype CCEe represent? CcEe? CCee? ccEE?

What do you think?

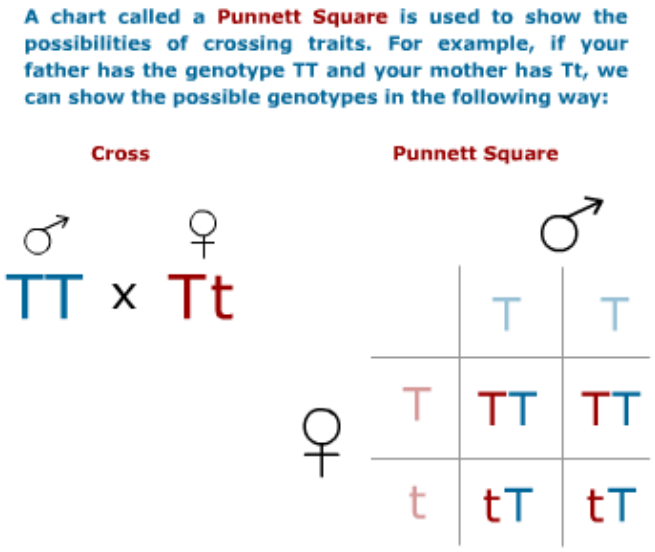
Members of a family show many similarities in appearance, but are not identical to each other (except in the case of identical twins). Why do offspring inherit certain characteristics from their parents, but not others?

- Write as detailed of an explanation as you can! Don't worry about being right. Imagine no one had discovered heredity. What would you come up with?

Punnett Squares

1875-1967

A Punnett square (it is capitalized because it is named after a genetics professor) is used to show the possibilities of genotypes and phenotypes of parents and offspring. The example below demonstrates how to create and use a Punnett square.



Each chromosome carries one allele or version of a gene. Homologous chromosomes separate so each egg or sperm has one allele (recall meiosis). Each allele comes from one of a person's parents. The alleles in the egg and sperm are shown on the side and top of the square. By completing the square, we can see the possible genotypes of the offspring.

Virtual Punnett Square Lab:

http://glencoe.mcgraw-hill.com/sites/dl/free/0078695104/383934/BL_05.html

Mendel's Laws and Punnett Square Animation:
http://www.teachersdomain.org/resource/hew06.sci.life.gen.mendelinherit/

Punnett Square Questions:

1. In summer squash, white fruit colour (W) is dominant over yellow fruit colour (w). If a squash plant homozygous for the dominant trait is crossed with a plant homozygous for the recessive trait, what would be the phenotypic and genotypic ratios of the offspring?

2. In horses, black colour (B) is dominant over chestnut (b). If a homozygous chestnut horse horse (bb) is crossed with a homozygous black horse (BB), what are the possible genotypes and phenotypes of the offspring?

3. In humans, dimpled cheeks (D) are dominant over smooth cheeks (d). What are the genotypes and the phenotypes of a heterozygous father and a heterozygous mother? What are the possible genotypes and phenotypes of their children?

Introduction to Gregor Mendel

Imagine this!

- ☐ You don't know what chromosomes, DNA, or genes are.
- ☐ You have knowledge of Math and the study of plants
- ☐ You decide to do experiments on pea plants to find out how traits are inherited.

What would YOU do with the pea plants to find out how traits are passed on from one generation to the next?

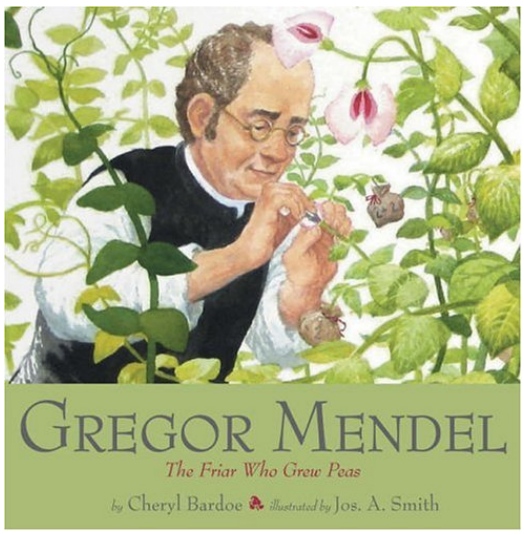
Gregor Mendel who is often called the "Father of Genetics" was born on July 22, 1822 to a relatively poor peasant family in a small town in Austria, called Heinzendorf (this town is now a part of Czech Republic.)

He entered the Augustinian monastery in Brunn, which at that time was known for being one of the best centers of learning in the scientific field.

He studied at the University of Vienna from 1851 to 1853. He went to study science and mathematics at the Univeristy of Vienna but failed his tests to receive a teaching degree. Later, in the same site as the university, Gregor Mendel Institute of Molecular Plant Biology was founded in the year 2000.

He later returned to the monastery where he became an abbot and spent the rest of his life. At the monastery, he started investigations of variation, heredity and evolution of plants at the monastery's experimental garden. Because he knew other scientists had done experimental crossings between peas, he already knew that he could observe the traits of the different pea generations.

Mendel was fortunate to have all the needed materials at the monastery. There were a large amount of true-bred pea plants available to him. Mendel raised and tested over 28,000 pea plants between the years 1856 and 1863, carefully analyzing seven pairs of seed and plant characteristics. He specifically studied plant height, pod shape, pod color, flower position, seed color, seed shape and flower color.



He made two very important generalizations from his pea experiments, known today as the *Laws of Heredity* and coined the present day terms in genetics: *recessiveness and dominance*. (you may remember these from Grade 9 Science)

In 1866 he published his work but it didn't take effect in the science field until 1900, years after his death. He named his paper "Experiments on Plant Hybridization."

Many other biologists used Mendel's research as a basis for their own, and Mendelian genetics is studied and taught throughout the world. Gregor Mendel died in Brunn on January 6, 1884.

Mendel's Laws animation:

http://www.dnaftb.org/2/concept/index.html

Law of Segregation

_____

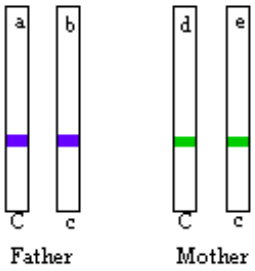
_____

_____

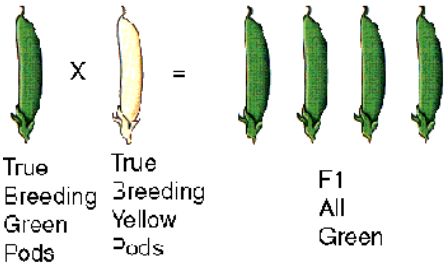
_____

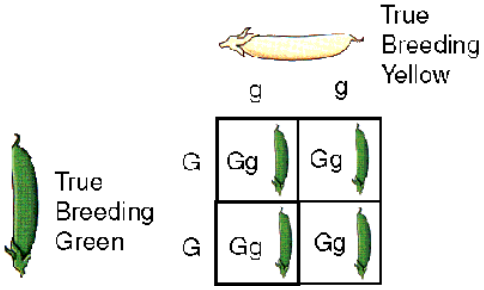
When human gametes (sperm and eggs) are produced, all 23 pairs of chromosomes in the cell separate or segregate. Only one of each pair goes into a sperm or egg cell. Thus, each sperm contains 23 different chromosomes and each egg cell contains 23 different chromosomes.

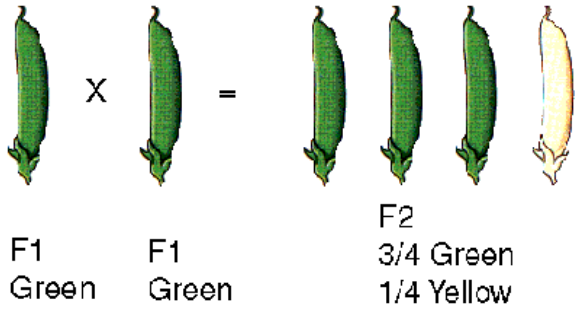
Example: We have two parents who are heterozygous eye colour. What are the possible genotypes for the sperm and eggs?

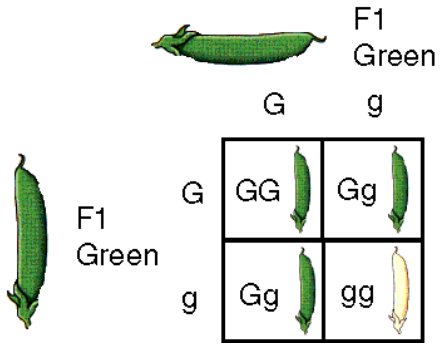


Therefore, half of the sperm produced by the male illustrated above will have chromosome "a" and half will include chromosome "b" along with 22 other chromosomes. Likewise, half of the egg cells produced by the female will have chromosome "d" and half will include chromosome "e" along with 22 other chromosomes.









Law of Dominance

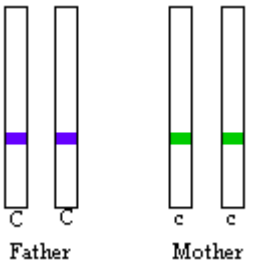
_____

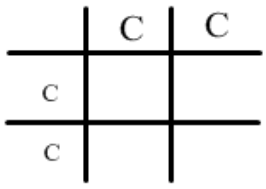
_____

_____

Mendel discovered, during his study of pea plants, that when an individual plant was heterozygous (based on knowing the characteristics of the parents), possessing contrasting expressions of a trait, _____. He called the manifested expression a dominant trait and the hidden expression a recessive trait.

Example: The father is homozygous for brown eyes (CC) and the mother is homozygous for blue eyes (cc). That means, the father can only contribute to his offspring an allele for brown eyes and the mother can only contribute to her offspring an allele for blue eyes. What eye colour will a child of these parents have?





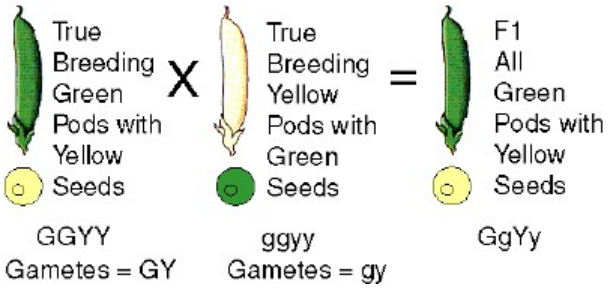
Law of Independent Assortment

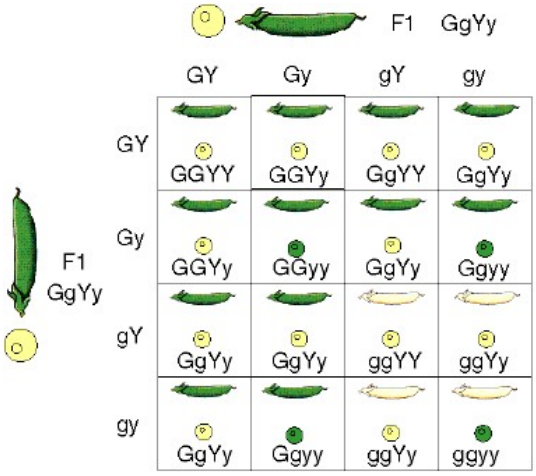
_____

_____

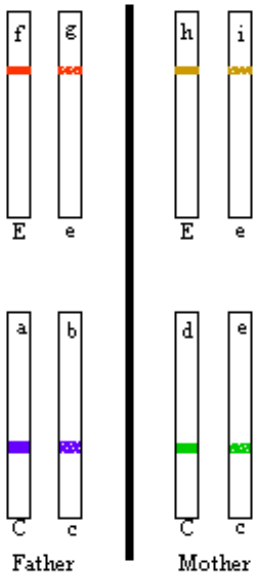
_____

Example: 'C' represents eye colour and 'E' represents earlobe shape. These genes are found on different chromosome pairs. The male is heterozygous for both eye colour and earlobe shape. His genotype is CcEe and his phenotype is brown eyes and free earlobes. The female has exactly the same genotype and phenotype as the male.





F2 Genotypes	F2 Phenotypes
GGYY, GGYy, GgYY, GgYy	Green pod, Yellow seeds
GGyy, Ggyy	Green pod, Green seeds
ggYY, ggYy	Yellow pod, Yellow seeds
ggyy	Yellow pod, Green seeds



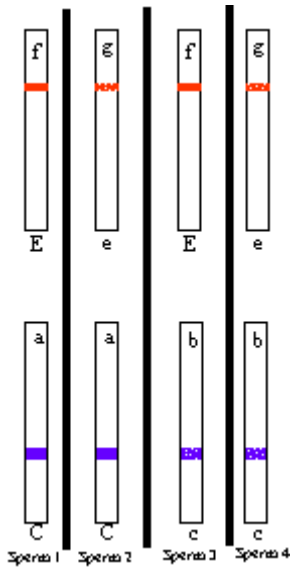
According to the Law of Independent Assortment, each chromosome becomes part of a sperm independently of other chromosomes.

This means that:
chromosomes "a" and "f'" could produce sperm 1 (CE), or

chromosomes "a" and "g" could produce sperm 2 (Ce), or

chromosomes "b" and "f" could could produce sperm 3 (cE), or

chromosomes "b" and "g" could could produce sperm 4 (ce).



The same applies for the female…

Questions

1. Which of Mendel's laws best fits the following description?
   In humans, the ability to taste a chemical called PTC is coded for by a single pair of genes. (PTC has a pronounced bitter taste to those who are able to taste it.) A man who can taste PTC, and whose parents can also taste PTC, has several children with a woman who is unable to taste PTC. The children are all tasters. State Mendel's law that applies to this situation.

2. Which of Mendel's laws best fits the following scenario?
   Dark hair is dominant over light-coloured hair in humans. It is possible, however, for dark-haired parents to produce a light-haired offspring even if most of their children are dark-haired. If so, how? Which of Mendel's laws underlies an explanation for this?

3. Eye colour, hair colour, dimples, freckles, widow's peak (hairline), eyelash length, and nose size make up a partial list of human traits. It is possible to see any combination of these traits in individuals. For example, people with black hair can have blue eyes. State Mendel's law that accounts for this phenomenon.

4. Use a Punnett square to show the probabilities of the phenotypes and genotypes of offspring if the parents are Tt and Tt. The letter 'T' represents the gene that causes the tongue to roll, and it is dominant over not being able to roll the tongue. Why are phenotypic ratios different from genotypic ratios?

5. Why are children not identical copies of their parents? And, why are children of the same parents not identical to each other? Use your knowledge of genetics principles to answer this question.

Genetic Crosses

Over time, plant and animal breeders can develop organisms that consistently produce offspring with the desired traits that are the same as their parents. In other words, they breed true. We call these _____.

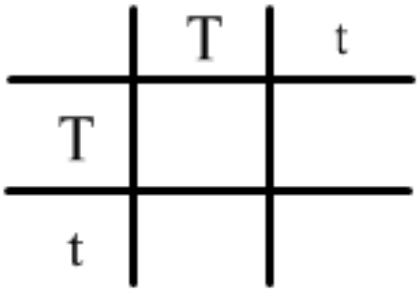
If we could see the genotypes of these organisms, we would see that they are almost always _____, either _____ or _____. Each generation of offspring will inherit only certain traits and not show variation.

A cross between two true-breeding individuals with different versions of the trait produces - _____ offspring. These offspring are called _____.

To help keep things organized, we refer the parents as _____ or _____ generation. The first generation of offspring from this cross is referred to as the _____ or _____ generation. The next generation produced by crossing the F_1 is called _____ or _____ generation.

Recall: Probabilities of Genotypes and Phenotypes

Let's examine a cross of Tt and Tt (a _____ cross), that is, a situation in which both parents can roll their tongues. Because they have both the dominant and recessive form of the gene, they are said to be heterozygous. Another way to describe this genotype is to say that the parents are _____ of the recessive trait.



It is important to notice the difference between the probabilities in genotypes compared to phenotypes. There are several versions of gene combinations, but there are only two expressions of the genes.

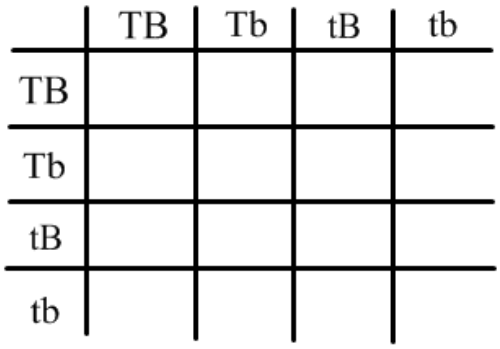
The probabilities of the genotypes are:

The probabilities of the phenotypes are:

Probabilities with Two Traits

A _____ cross involves parents that are hybrids or heterozygous for two different traits. If brown eyes are dominant over blue eyes, the letters "B" and "b" can be used. Keep in mind that eye colour is not as simple as shown.

If both parents are heterozygous for both traits, the genotype is TtBb. After meiosis, the possible gametes are TB, Tb, tB, and tb. Study the following Punnett square that shows the offspring of these parents.



	TB	Tb	tB	tb
TB				
Tb				
tB				
tb				

After analyzing the 16 possibilities:

- _____
  _____
- _____
  _____
- _____
  _____
- _____
  _____
  _____
  _____

Test Cross

A breeder wants to find out if an organism is homozygous dominant or heterozygous for a trait. Remember that the genotype can't be known by simple observation because the dominant trait will be expressed but a recessive trait may be present and not expressed. The breeder may do a test cross in which the unknown genotype is bred (crossed) with an individual that is _____ _____ _____ for the trait. When writing the genotype, the unknown gene is represented by the question mark, "?". If a red pea plant is crossed with a white plant, then we express the test cross as R**?** x rr.

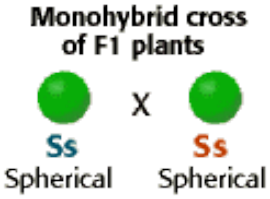
Practice:

1. What are the genotypic and phenotypic ratios of the following test crosses?
   a. RR x rr

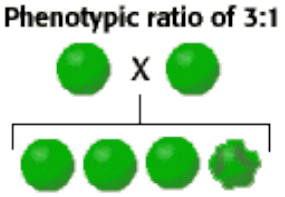
   b. Rr x rr

Problems - Monohybrid and Dihybrid Crosses #1

1. In pea plants, spherical seeds (S) are dominant to dented seeds (s). In a genetic cross of two plants that are heterozygous for the seed shape trait, what fraction of the offspring should have spherical seeds?
   a) none
   b) ¼
   c) ½
   d) ¾
   e) All



2. A phenotypic ratio of 3:1 in the offspring of a mating of two organisms heterozygous for a single trait is expected when:
   a) the alleles segregate during meiosis.
   b) each allele contains two mutations.
   c) the alleles are identical.
   d) the alleles are incompletely dominant.
   e) only recessive traits are scored.



3. In one of Mendel's experiments true-breeding pea plants with spherical seeds were crossed with true-breeding plants with dented seeds. (Spherical seeds are the dominant characteristic.) Mendel collected the seeds from this cross, grew F1-generation plants, let them self-pollinate to form a second generation, and analyzed the seeds of the resulting F2 generation. The results that he obtained, and that you would predict for this experiment are:

   a) 1/2 the F1 and 3/4 of the F2 generation seeds were spherical.
   b) 1/2 the F1 and 1/4 of the F2 generation seeds were dented.
   c) All of the F1 and F2 generation seeds were spherical.
   d) 3/4 of the F1 and 9/16 of the F2 generation seeds were spherical.
   e) All the F1 and 3/4 of the F2 generation seeds were spherical.

4. A genetic cross between two F1-hybrid pea plants for spherical seeds will yield what percent spherical-seeded plants in the F2 generation? (Recall, spherical-shaped seeds are dominant over dented seeds.)
   a) 100%
   b) 75%
   c) 50%
   d) 25%
   e) 0%

5. When true-breeding tall stem pea plants are crossed with true-breeding short stem pea plants, all of the _____ plants, and 3/4 of the _____ plants had tall stems. Therefore, tall stems are dominant.
   a) F1, F2.
   b) G1, G2.
   c) parental, F2.
   d) F2, parental.
   e) P1, P2

6. To identify the genotype of yellow-seeded pea plants as either homozygous dominant (YY) or heterozygous (Yy), you could do a test cross with plants of genotype _____.
   a) y
   b) Y
   c) yy
   d) YY
   e) Yy

7. A pea plant is heterozygous for both seed shape and seed color. S is the allele for the dominant, spherical shape characteristic; s is the allele for the recessive, dented shape characteristic. Y is the allele for the dominant, yellow color characteristic; y is the allele for the recessive, green color characteristic. What will be the distribution of these two alleles in this plant's gametes?

   a) 50% of gametes are Sy; 50% of gametes are sY
   b) 25% of gametes are SY; 25% of gametes are Sy; 25% of gametes are sY; 25% of gametes are sy
   c) 50% of gametes are sy; 50% of gametes are SY
   d) 100% of the gametes are SsYy
   e) 50% of gametes are SsYy; 50% of gametes are SSYY.

8. A phenotype ratio of 9:3:3:1 in the offspring of a mating of two organisms heterozygous for two traits is expected when:

   a) the genes reside on the same chromosome
   b) each gene contains two mutations
   c) the gene pairs assort independently during meiosis
   d) only recessive traits are scored
   e) none of the above

9. Which of the following genetic crosses would be predicted to give a phenotypic ratio of 9:3:3:1?

   a) SSYY x ssyy
   b) SsYY x SSYy
   c) SsYy x SsYy
   d) SSyy x ssYY
   e) ssYY x ssyy

10. The gametes of a plant of genotype SsYy should have the genotypes:

    a) Ss and Yy
    b) SY and sy
    c) SY, Sy, sY, and sy
    d) Ss, Yy, SY and sy
    e) SS, ss, YY, and yy

11. Which of the following genotypes would you not expect to find among the offspring of a SsYy x ssyy test cross:

    a) ssyy
    b) SsYy
    c) Ssyy
    d) ssYy
    e) SsYY

12. The expected phenotypic ratio of the progeny of a SsYy x ssyy test cross is:

    a) 9:3:3:1
    b) 3:1
    c) 1:1:1:1
    d) 1:2:1
    e) 3:1:1:3

13. In a dihybrid cross, AaBb x AaBb, what fraction of the offspring will be homozygous for both recessive traits?

    a) 1/16
    b) 1/8
    c) 3/16
    d) 1/4
    e) 3/4

14. Following a SsYy x SsYy cross, what fraction of the offspring are predicted to have a genotype that is heterozygous for both characteristics?

    a) 1/16
    b) 2/16
    c) 3/16
    d) 4/16
    e) 9/16

15. In a dihybrid cross, SsYy x SsYy, what fraction of the offspring will be homozygous for both traits?

    a) 1/16
    b) 1/8
    c) 3/16
    d) 1/4
    e) 3/4

http://www.biology.arizona.edu/mendelian_genetics/mendelian_genetics.html

More Questions…

1. What does F_1 and F_2 mean?

2. In mice, black fur is dominant to white fur. Cross a heterozygous black mouse with a white mouse. Give the genotypic and phenotypic ratios for the F_1 generation.

3. Explain how a test cross is used. Why is it necessary?

4. In a cross between Bb and Bb, about what percentage of the progeny will have the same genotype as their parents?

5. In humans, brown eyes are dominant over blue eyes. If a blue-eyed man marries a brown-eyed woman whose father was blue-eyed, what proportion of their children do you expect to be blue-eyed?

6. If a brown-eyed man marries a blue-eyed woman and they have eight children, all brown-eyed, can you be certain that the man it homozygous? If the couple has a ninth child and it is also brown-eyed, what will that show about the father's genotype?

7. In corn plants, yellow seed (Y) is dominant to red seed (y). Round seed (R) is dominant to wrinkled seed (r). What will be the phenotypic ratio of the offspring when a red and heterozygous round is crossed with a plant that is homozygous yellow and heterozygous round?

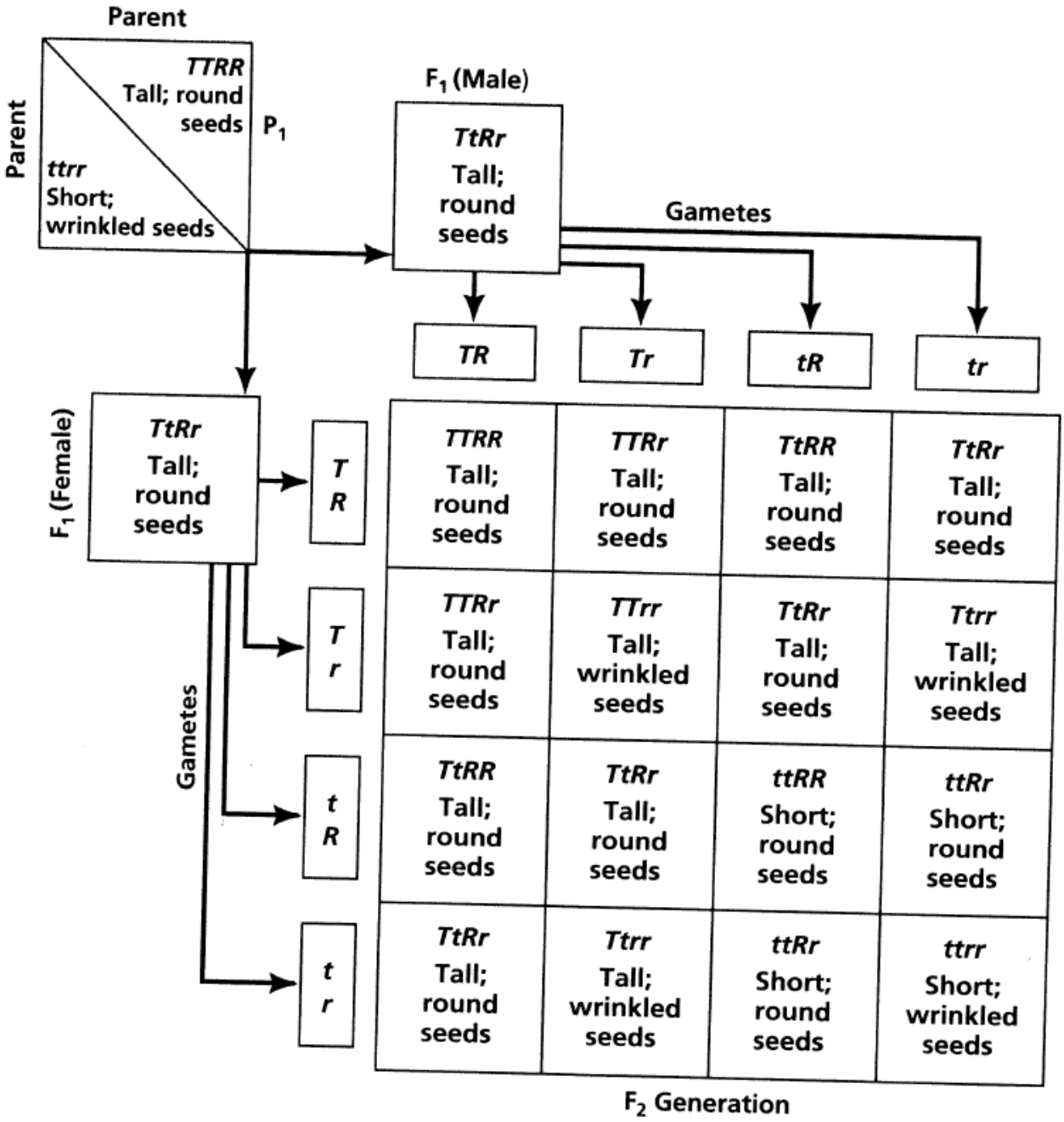
8. In mice, the ability to run is determined by a dominant gene, while those that are homozygous recessive are termed "waltzing mice" because of their pronounced limp. If a black waltzing mouse that is heterozygous for black fur is crossed with a black running mouse that is heterozygous for both traits, what types of offspring could they produce and in what ratios?

9. A brown-eyed man whose father was brown-eyed and whose mother was blue-eyed married a woman whose father and mother were both brown-eyed. The couple has a blue-eyed son. For which of the individuals mentioned can you be sure of the genotypes? What are their genotypes? What genotypes are possible for the others?

Prepare for Quiz

Human Inheritance Lab

Dihybrid Cross



Dihybrid Cross Questions – Use the diagram on the previous page to answer the following questions:

1. Describe the P_1 dihybrid cross shown in the transparency.

2. Describe the F_1 dihybrid cross shown in the transparency.

3. Identify the combinations of alleles that are contained in the gametes of the F_1 plants.

4. Complete the Punnett square for the F_2 generation. Each box should contain four alleles, two for each gene. One allele for each gene came from the female parent. The other allele for each gene came from the male parent. Each box should also identify the phenotype of each offspring. How many *different* genotypes are there in your completed square? Identify them.

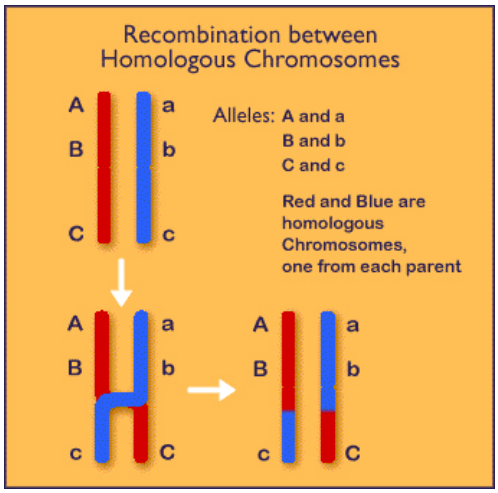
5. What are the proportions of the different genotypes of the F_2 generation?

6. What are the phenotypic proportions?

7. In studying the dihybrid crosses, Mendel found that the inheritance of one trait does not influence the inheritance of another trait. What would be another way of expressing this fact, using the phenotypes of the F_2 generation on the transparency?

Genetic Variation and Crossing Over

There are three major sources of genetic variation between the offspring of two individuals: crossing over, independent assortment of chromosomes and fusion of gametes.

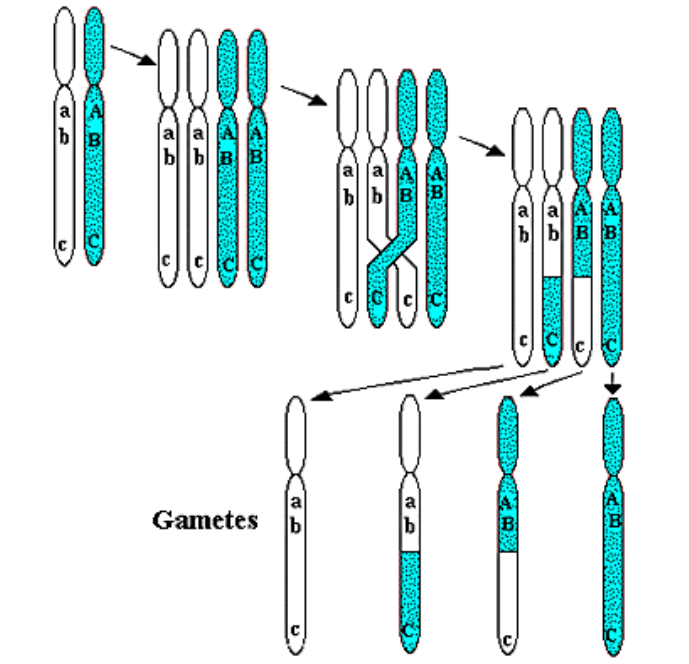


Crossing over allows certain traits carried on the same chromosome to assort independently sometimes, contrary to expectations. Normally, if genes are on the same chromosome, the resultant traits are _____ _____.

However, even though the parents show the gene linkage, _____.

In other cases, new linkages seemed to have formed. These are relatively rare events but it happens often enough for us to realize there must be a mechanism that causes this change in gene linkage.

It turns out that during synapsis of meiosis, while chromosomes are lined up, there are times when _____ _____. This is known as _____. The result is a recombination of alleles. The new combination creates new gene linkages and eliminates previous ones.



Crossing-over and recombination during meiosis

The implications are rather interesting. Crossing over during the formation of gametes results in egg and sperm that are not exact copies of the parental chromosomes. Without crossing over, offspring would have exact copies of parental chromosomes. Crossing over

_____

_____

_____

Frequencies of crossing over are related to the distance genes are apart on a chromosome. The _____, the less likely genes are to be separated by crossing over. Crossing over frequencies have been used to map relative locations of genes on chromosome.

Independent assortment and fusion of gametes at random, even without-crossing over, creates a huge number of possible genotypes for the offspring of two individuals. A man or woman has the potential for 2^{23} genetically different sperm or eggs. When a zygote is formed there are 2^{46} genetically different possibilities, more than all the people on earth.

Other Patterns of Inheritance

When Gregor Mendel crossed a purple-flowered pea plant with a white flowered pea plant, the flowers in the next generation are all purple.

However, the cross between a red-flowered snapdragon and a white-flowered snapdragon produces pink-flowered snapdragons.

THINK

1) What is the dominant flower colour in pea plants?

2) How does the snapdragon cross differ from the pea plant cross?

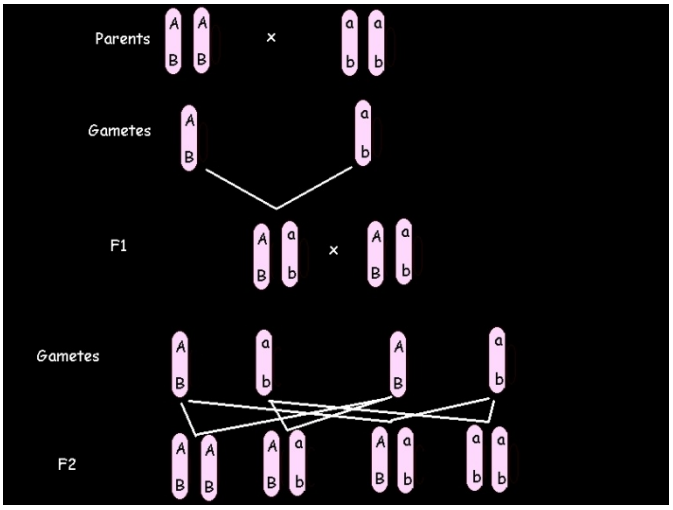
3) Can you come up with an explanation for the results of the snapdragon cross?

1) Linkage

We have been working with traits carried on separate chromosomes. _____

_____

_____. In this case, the Law of Independent Assortment _____.
Particular alleles for these traits tend to be inherited dependently instead of independently. However, with crossing over new linkages can be formed.



The latest estimates from the Human Genome Project work is that a human has about 30 to 35 thousand genes (as recently as 1999 it was thought that humans had at least 100,000 genes). Given the sheer number of genes and the behaviour of chromosomes, predicting genotypes and phenotypes gets complicated indeed. Geneticists actually use the probabilities of crossing over and linkage to create a map of most of the genes in the human genome.

2) Multiple Alleles and Codominance

Genotype	*Phenotype*
AA	Type A
AO	Type A
BB	Type B
BO	Type B
AB	Type AB
OO	Type O

In some traits, there are more than two alleles for its expression.

_____

_____

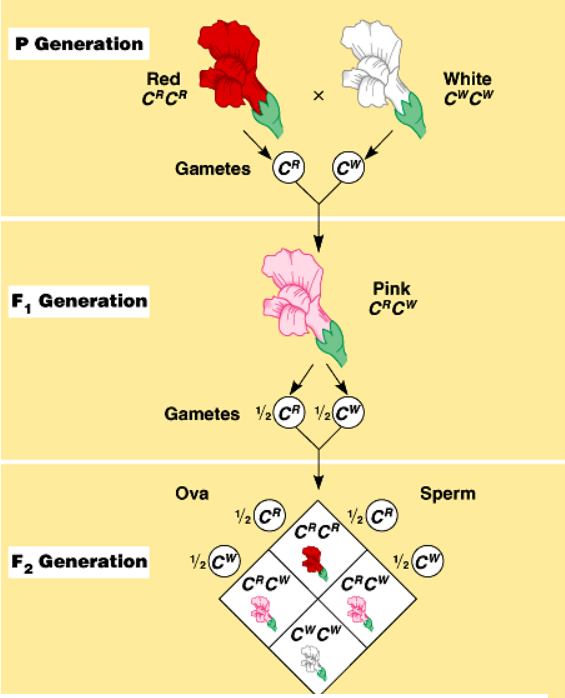
_____

Regardless of how many alleles a gene has, only two alleles can be present in a particular individual.

Blood type is an example of multiple alleles. There are three alleles for blood type: A, B, and O. A person can only have two of these. The chart shows all the possible genotypes and phenotypes for blood type.

In the Type AB, _____. In this heterozygous condition, an individual has both types of proteins in their blood. This condition is known as _____. (Both A and B are dominant of O.)

3) Incomplete Dominance and Polygenic Inheritance



http://io.uwinnipeg.ca/~simmons/cm1503/dihybrid.htm

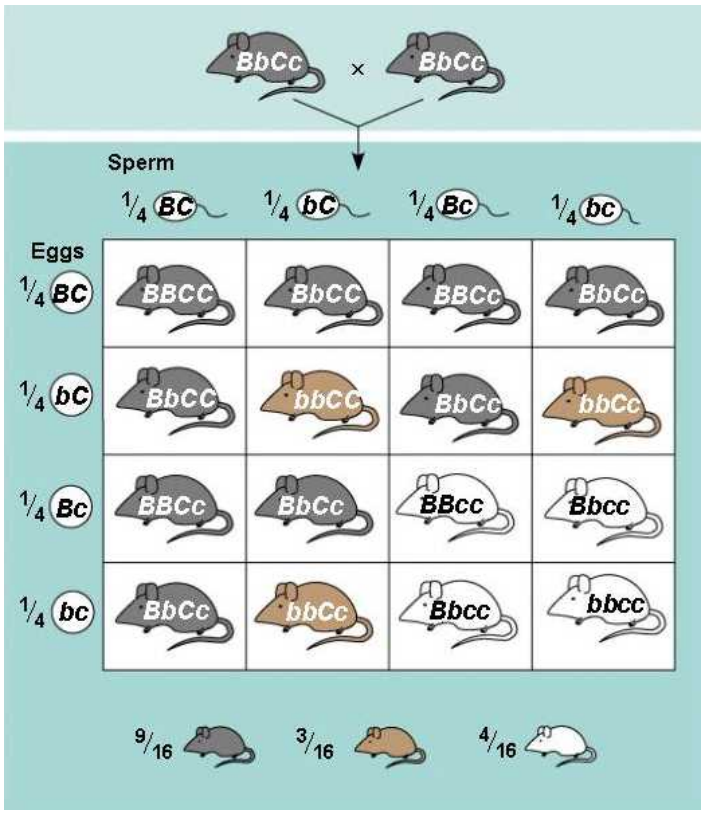
Another condition exists where neither allele is dominant. This is called _____ _____ _____ _____. For example, if red is usually dominant over white in a type of plant, incomplete dominance means that the effects of the two genes seem to blend and produce a pink colour. Here, all heterozygous plants are pink.

We see a more complicated form of incomplete dominance in humans when we examine skin colour or height. In these traits and others, _____ _____ _____. There are many degrees of skin colour just like there are many variations of intelligence. These are examples of _____ _____.

_____ is a condition where a gene on a different chromosome can _____ _____ the effect of the other genes. Genes that interfere with the operation of other genes are said to be **epistatic.**

Let us use the colour of mice as an example. Imagine that the dominant gene B produces black colour and the recessive gene b produces brown. There are other genes on another chromosome that also affect coat colour.

These genes can be called C and c. If the dominant C gene is present, normal colouration occurs. The recessive gene c prevents normal colouration to occur (it stops normal pigmentation).



Questions

1. Earlier in this lesson there was an example of what is known as an epistatic effect of genes. In the example, mice colour was described as dependent on dominant (B = black) and recessive (b = brown) genes. However, a second pair of genes interacts with the genes for coat colour. This gene also has a dominant form (C = normal colour) and a recessive form (c = inhibits colour). Show the genotypes and phenotypes of a cross between an albino mouse and a heterozygous black mouse. The cross is: Bbcc x BbCc.

2. Chickens have different shapes of combs (the thick, red, fleshy crest on top of the heads).
    - A rose comb looks as though it has multiple rows and has rounded edges, and peaks into a point at the back. R_pp
    - A pea comb is smaller with rounded edges. rrP_
    - A walnut comb has no feathered edges; it looks like a walnut. R_P_
    - A single comb is made of one fleshy lobe that has well defined edges. rrpp

    With the knowledge about chickens' combs, determine the phenotypes if you cross a chicken with a rose comb, RRpp with a chicken with a pea comb, rrPP. Show the F1 generation and the F2 generation. What type of gene interaction does this represent?

3. Shortly after the births of three babies in the same hospital, it was feared that the name tags of the babies had been mixed up. One way to determine if errors had been made was to take a blood sample from all the parents and babies and then see if the puzzle could be sorted out. The blood test result are as follows:

    - Mr. Jones: type A; Mrs. Jones: type O
    - Mr. Livingstone: type O; Mrs. Livingstone: type O
    - Mr. Peters: type A; Mrs. Peters: type B
    - Baby X: type A; Baby Y: type O; Baby Z: type AB

    Match the babies with the parents.

4. When a plant with red flowers was crossed with a plant with white flowers, all of the offspring had pink flowers. What is the name for this type of inheritance? Show the resulting progeny of a cross between two pink parents. What is the phenotypic ratio?

DO BRAINY BUNNIES ACTIVITY

Problems - Monohybrid and Dihybrid Crosses #2

1. A test cross is used to determine if the genotype of a plant with the dominant phenotype is homozygous or heterozygous. If the unknown is homozygous, all of the offspring of the test cross have the _____ phenotype. If the unknown is heterozygous, half of the offspring will have the _____ phenotype.

   A. dominant, incompletely dominant
   B. recessive, dominant
   C. dominant, epistatic
   D. codominant, complimentary
   E. dominant, recessive

2. In Mendel's experiments, if the gene for tall (T) plants was incompletely dominant over the gene for short (t) plants, what would be the result of crossing two Tt plants?

   A. 1/4 would be tall; 1/2 intermediate height; 1/4 short
   B. 1/2 would be tall; 1/4 intermediate height; 1/4 short.
   C. 1/4 would be tall; 1/4 intermediate height; 1/2 short.
   D. all the offspring would be tall.
   E. all the offspring would be intermediate.

3. A genetic cross of inbred snapdragons with red flowers with inbred snapdragons with white flowers resulted in F1-hybrid offspring that all had pink flowers. When the F1 plants were self-pollinated, the resulting F2-generation plants had a phenotypic ratio of 1 red: 2 pink: 1 white. The most likely explanation is:

   A. pink flower color is epistatic to red flower color.
   B. pink flowers are the result of a blending of the red and white genotypes.
   C. flower color is due to 2 or more complementary genes.
   D. heterozygous plants have a different phenotype than either inbred parent because of incomplete dominance of the dominant allele.
   E. flower color inheritance in snapdragons does not behave as a Mendelian trait.

4. Human blood type is determined by codominant alleles. There are three different alleles, known as I^A, I^B, and i. The I^A and I^B alleles are co-dominant, and the i allele is recessive. The possible human phenotypes for blood group are type A, type B, type AB, and type O. Type A and B individuals can be either homozygous (I^AI^A or I^BI^B, respectively), or heterozygous (I^Ai or I^Bi, respectively). A woman with type A blood and a man with type B blood could potentially have offspring with which of the following blood types?

   A. type A
   B. type B
   C. type AB
   D. type O
   E. all of the above

5. What are the possible blood types of the offspring of a cross between individuals that are type AB and type O? (Hint: blood type O is recessive)
   A. AB or O
   B. A, B, or O
   C. A or B
   D. A, B, AB, or O
   E. A, B, or AB

6. If Mendel's crosses between tall, spherical-seeded plants and short, dented-seeded plants had produced many more than 1/16 short, dented-seeded plants in the F2 generation, he might have concluded that:

   A. the dented seed and short traits are unlinked.
   B. he would not have concluded any of the above.
   C. all traits in peas assort independently of each other.
   D. the spherical seed and tall traits are linked.
   E. all traits in peas are linked.

7. In Mendel's experiments, the spherical seed character (SS) is completely dominant over the dented seed character (ss). If the characters for height were incompletely dominant, such that TT are tall, Tt are intermediate and tt are short, what would be the phenotypes resulting from crossing a spherical-seeded, short (SStt) plant to a dented-seeded, tall (ssTT) plant?

   A. all the progeny would be spherical-seeded and tall.
   B. 1/2 would be spherical-seeded and intermediate height; 1/2 would be spherical-seeded and tall.
   C. all the progeny would be spherical-seeded and short.
   D. you cannot predict the outcome.
   E. all the progeny would be spherical-seeded and intermediate height.

8. Two unlinked genes affect mouse hair color. **CC** or **Cc** mice are pigmented. Mice with genotype **cc** are albino because all pigment production and deposition of pigment in hair is blocked. At the second locus, the **B** allele (black coat) is dominant to the **b** allele (brown coat). A mouse with a black agouti coat is mated with an albino mouse of genotype **bbcc**. Half of the offspring are albino, one quarter are black, and one quarter are brown. What is the genotype of the black parent?
   A. BBCC
   B. BbCc
   C. bbCC
   D. BbCC
   E. BBcc

9. Two unlinked genes effect mouse hair color. **CC** or **Cc** mice are pigmented. Mice with genotype **cc** are albino because all pigment production is blocked, regardless of the phenotype at the second locus. At the second locus, the **B** allele (black coat) is dominant to the **b** allele (brown coat). What would be the result of a cross between two mice of genotype AaBb?
   A. 4 brown: 4 black: 8 albino
   B. 9 brown: 3 black: 3 albino: 1 grey
   C. 9 brown: 3 black: 4 albino
   D. 8 brown: 4 black: 4 albino

Sex Determination in Humans

Henry VIII of England married six times in an attempt to have a legitimate male heir to the English throne. Recalling your knowledge of Senior 1 Science, was Henry correct in blaming has wives for their inability to produce a son? (Henry did eventually have on son, who inherited the kingdom after his father's death.)

_____

_____

_____

_____

Sex Chromosomes

Humans have ____ chromosomes - ____ pairs of homologous chromosomes. 22 of these pairs are called _____ and 1 pair is known as the _____. The sex chromosomes are responsible for determining sex and are known as the X and Y chromosomes. A female has two X chromosomes and the male has one X chromosome and one Y chromosome. The X chromosome is larger and has genes required by both sexes. The Y chromosome carries genes related to maleness and male fertility.

Using a Punnett square, what are the chances of a couple having a boy or a girl?

	X	**Y**
X		
X		

Girl?

Boy?

Sex Linked Traits

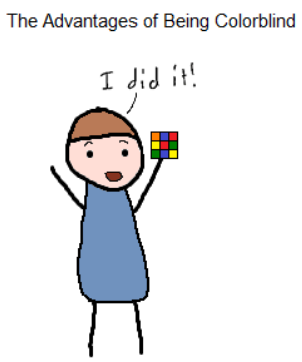
Some traits are controlled by genes on the sex chromosomes. Such traits, as well as the genotypes and phenotypes these genes control, are called sex-linked.

In humans, sex-linked traits are on the X chromosome, sometimes they are called X-linked. Because males have only one X chromosome, they always show the allele on that X chromosome, whether it is dominant or recessive. Females must have two recessive alleles for a recessive trait to be expressed. So most cases of sex-linked traits are in males.

There are a number of sex-linked traits that have been studied in humans. Two well-known examples are colour-blindness and hemophilia. Colour-blindness is the inability to discern certain colours. Red-green is one example. To a person who has this condition, those colours look alike. Hemophilia is a potentially life-threatening condition in which sufferers do not have the ability to clot their blood. In some cases bleeding into the joints and internal organs can occur. Even a slight injury can cause lethal bleeding if not treated quickly.

Let's look at an example:

The gene for red-green colour blindness in linked to the X chromosome.
Let's denote B = dominant (regular vision)
b = recessive (red-green colour blindness)
Because that gene is linked to the X chromosome we denote it like this:
X^B and X^b



Suppose a woman with regular vision (X^BX^B) mates with a colour blind man (X^bY). What are the F1 genotypes and phenotypes?

	X^b	Y
X^B		
X^B		

Genotypes:

Phenotypes:

We see here that the males produced would _____the recessive gene. The females however, though they are not colour blind, do carry the recessive gene. We call them _____ _____.

Suppose now that an F1 female mates with a male with normal vision. What are the resultant genotypes and phenotypes?

	X^B	Y
X^B		
X^b		

Genotypes:

Phenotypes:

Here we see that though the mother was a carrier, the male offspring have a chance of being colour blind and the female offspring have no chance.

Questions

1- Hemophilia is a sex linked recessive trait (more specifically X-linked). A female carrier mates with a hemophilic male. What are the resulting phenotypes and genotypes?

2- A non-hemophilic male mates with a non-hemophilic female. They have 2 non-hemophilic daughters and 1 hemophilic son. What are the genotypes of the parents? Are the daughters carriers or not?

Eye Colour in Fruit Flies

In fruit flies, red is the dominant trait for eye colour and white eye is the recessive trait. Experiments have shown that eye colour in fruit flies is a sex-linked trait (X-linked).

Example: We will begin with a parent cross: a red-eyed female and a white-eyed male.

X^RX^R *x* X^rY

We want to know the following information:
What are the phenotypes of the females in F_1?
What are the phenotypes of the males?
Are the females in this generation carriers of the recessive gene?

Predict the genotypes and phenotypes of the F_2 generation: X^RX^r *x* X^RY

Questions

1. Colour-blindness is a sex-linked trait. If a woman with normal vision whose father was colour-blind has children with a man with normal vision, what are the possibilities of phenotypes for any children they may have?

2. In cats, black colour is incompletely dominant over white. The heterozygous condition produces a gray coat. A furry coat is dominant to straight hair. Cross two cats that are heterozygous for both conditions. What are the possible phenotypes and ratios of the offspring?

3. In fruit flies, red eyes are dominant over white but the trait is sex-linked. Two flies mate and produce 236 red-eyed and 233 white-eyed offspring. What are the probable genotypes of the parents? Explain how you arrived at your answer.

4. A woman who is a carrier for colour-blindness has children with a colour-blind male. What are the probabilities that their male and female children will be colour-blind?

5. In cats a pair of genes for coat colour is sex linked. A heterozygous genotype is expressed as a tortoise coat colour. If a homozygous yellow female mates with a black male, what are the expected phenotypic ratios of the offspring? Why can't the male cats be tortoise coloured?

6. A man who is AB for blood type is married to a woman who is type O. Their first child has blood type O. The husband sues for divorce on the grounds that his wife has been unfaithful. You are called as an expert witness to give genetic evidence. What would you say?

Sex-Linked Questions

1. In a cross between a white-eyed female fruit fly and red-eyed male, what percent of the female offspring will have white eyes? (White eyes are X-linked, recessive)
   A. 100%
   B. 25%
   C. 50%
   D. 75%
   E. 0%

2. A female Drosophila of unknown genotype was crossed with a white-eyed male fly, of genotype X^rY (r = white eye allele is recessive, R= red-eye allele is dominant.) Half of the male and half of the female offspring were red-eyed, and half of the male and half of the female offspring were white-eyed. What was the genotype of the female fly?
   A. X^RY
   B. X^RX^R
   C. X^rX^r
   D. X^rY
   E. X^RX^r

3. In a cross between a pure bred, red-eyed female fruit fly and a white-eyed male, what percent of the male offspring will have white eyes? (white eyes are **X**-linked, recessive)
   A. 100%
   B. 75%
   C. 50%
   D. 25%
   E. 0%

4. What is the genotype of a red-eyed, yellow-bodied female fruit fly who is homozygous for the eye color allele? Red eyes (W) and tan bodies (Y) are the dominant alleles. (Both traits are **X** chromosome linked).
   A. $X^{WY}X^{WY}$
   B. $X^{WY}Y$
   C. $X^{wY}X^{wY}$
   D. $X^{wY}Y$
   E. $X^{Wy}X^{WY}$

5. A white-eyed female fruit fly is crossed with a red-eyed male. Red eyes are dominant, and **X**-linked. What are the expected phenotypes of the offspring?
   A. All of the females will have red eyes; half of the males will have red eyes, and half of the males will have white eyes.
   B. All of the females and all of the males will have white eyes.
   C. All of the females will have red eyes; all of the males will have white eyes.
   D. All of the females and all of the males will have red eyes.
   E. All of the females will have white eyes; half of the males will have red eyes, and half of the males will have white eyes.

6. Hemophilia in humans is due to an **X**-chromosome mutation. What will be the results of mating between a normal (non-carrier) female and a hemophilac male?

A. half of daughters are normal and half of sons are hemophilic.
B. all sons are normal and all daughters are carriers.
C. half of sons are normal and half are hemophilic; all daughters are carriers.
D. all daughters are normal and all sons are carriers.
E. half of daughters are hemophilic and half of daughters are carriers; all sons are normal.

7. A human female "carrier" who is heterozygous for the recessive, sex-linked trait causing red-green color blindness (or alternatively, hemophilia), marries a normal male. What proportion of their male progeny will have red-green color blindness (or alternatively, will be hemophiliac)?
   A. 100%
   B. 75%
   C. 50%
   D. 25%
   E. 0%

8. Women have sex chromosomes of **XX**, and men have sex chromosomes of **XY**.
   Which of a man's grandparents could not be the source of any of the genes on his **Y**-chromosome?
   A. Father's Mother.
   B. Mother's Father.
   C. Father's Father.
   D. Mother's Mother, Mother's Father, and Father's Mother.
   E. Mother's Mother.

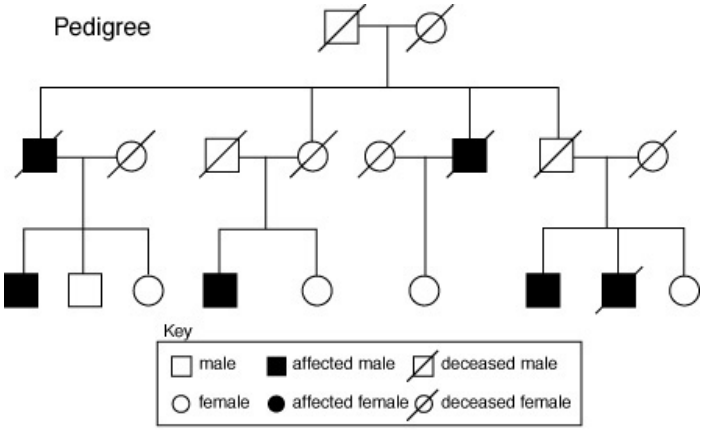
9. Women have sex chromosomes of **XX**, and men have sex chromosomes of **XY**.
   Which of a women's grandparents could not be the source of any of the genes on either of her **X**-chromosomes?
   A. Mother's Father.
   B. Father's Mother.
   C. Mother's Mother.
   D. Father's Father.
   E. Mother's Mother and Mother's Father.

10. A human female "carrier" who is heterozygous for the recessive, sex-linked trait red color blindness, marries a normal male. What proportion of their female progeny will show the trait?
    A. All
    B. 1/2
    C. 1/4
    D. 0
    E. 3/4

Pedigree Charts

What are they and why are they used?

- Pedigree charts are diagrams that show the inheritance of traits within a biological family.
- Pedigree charts were used to find the likelihood of certain traits in offspring before alleles were known.
- Pedigree charts are used to find the genotype of animals and people using the trait expression of others in the biological family.



How to make a Pedigree chart?

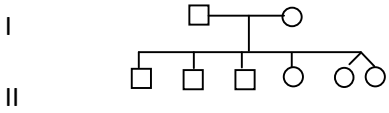
- Squares represent males.
- Circles represent females.
- Diamonds represent unknown sex.

- Mating is designated as a line connecting the circle and the square.

- Offspring is a line from the connecting mating line (two boys on left).

- Fraternal twins are shown the same as other siblings (one boy and one girl in middle).

- Identical twins are shown with diagonal lines from the same point (two girls on left).

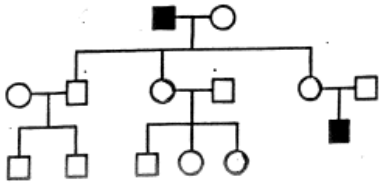


- Generations are marked using roman numerals.
- Affected traits are designated by a solid circle or square.
- Include a title
- ALWAYS USE A LEGEND TO DESIGNATE TRAITS

Example: Huntington's disease is a dominant trait that causes a deterioration of brain tissue and develops late in middle age. A man inherited Huntington's disease from his mother. He marries a healthy woman whose parents are healthy. This couple has 5 children, two healthy girls and 3 boys, one with the disease. Create a pedigree chart of this family.

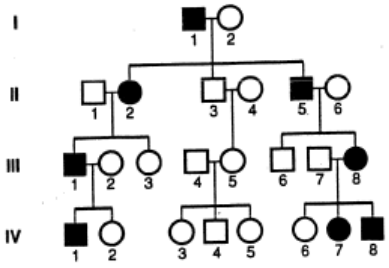
Autosomal or X-linked? Dominant or Recessive?

1. Determine if the pedigree chart shows an autosomal or X-linked disease.
   - If males are more affected than females, the disorder is likely X-linked. Remember that a male has only one allele for each X-linked gene.

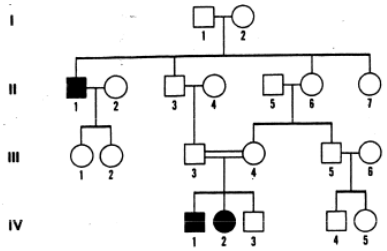


   - If males and females are about equally affected, the disorder is likely autosomal.

2. Determine whether the disorder is dominant or recessive.
   - For dominant disorders, if it appears in a child, one of the parents must have the disorder.



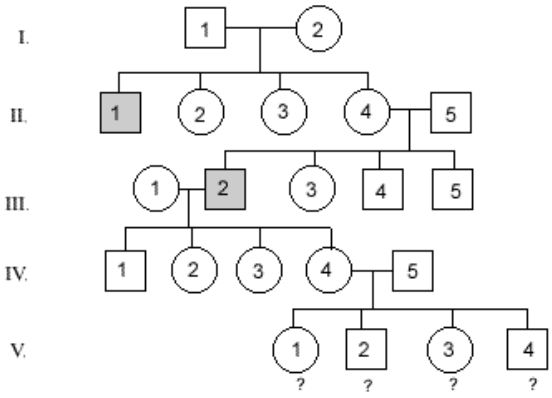
   - For recessive disorders, it can appear in a child when neither parent has the disorder because they can be heterozygous.



Problems

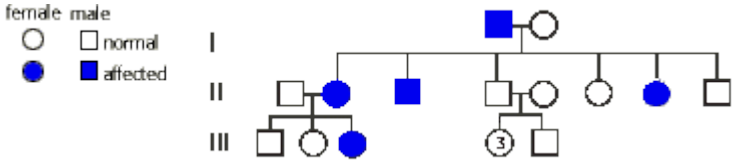
1. From the pedigree chart showing the inheritance of hemophilia (a sex-linked, recessive trait), determine the genotype of each individual listed below. Explain how you determined the genotype for each.

a) I-2
b) II-1
c) What are the probabilities of various genotypes for the following: V-1, V-2, V-3, V-4?

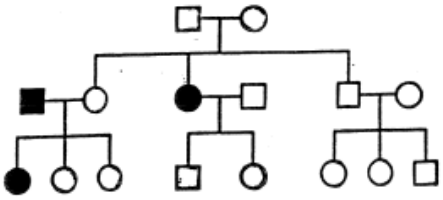


2. A human geneticist determined the pedigrees shown in the diagrams below. Filled symbols show the affected individuals. For each pedigree, indicate whether the pattern of inheritance is autosomal or X-linked, and dominant or recessive.

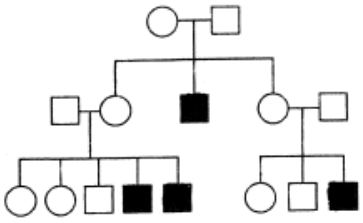
a.



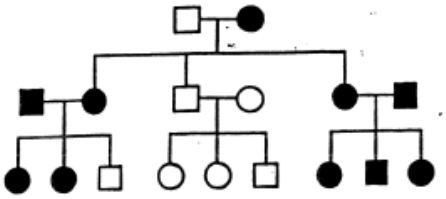
b.



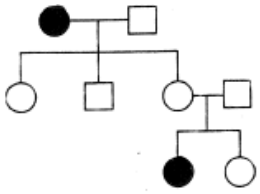
c.



d.



e.

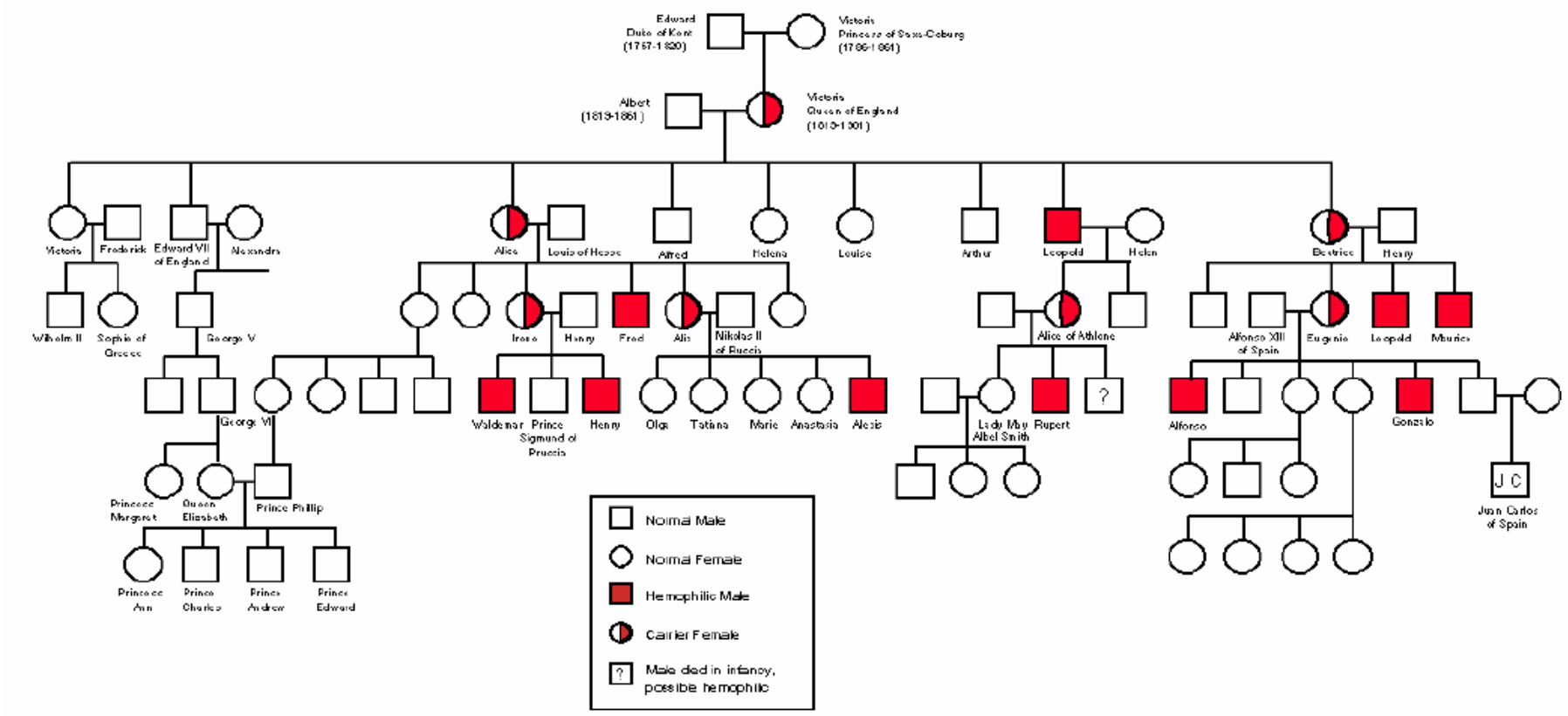


3. A man who had purple ears came to the attention of a human geneticist. The human geneticist did a pedigree analysis and made the following observations:

   In this family, purple ears proved to be an inherited trait due to a single genetic locus. The man's mother and one sister also had purple ears, but his father, his brother, and two other sisters had normal ears. The man and his normal-eared wife had seven children, including four boys and three girls. Two girls and two boys had purple ears.

   Draw the pedigree for the family and determine whether the pattern of inheritance of purple ears.

Appendix 5: Pedigree of Queen Victoria and her Descendents (BLM)



Challenge question:

In groups of two or three, hypothesize how Queen Victoria became a carrier of hemophilia when neither of her parents was hemophiliac, nor was there any previlous history of hemophilia in their families. Everyone must record their predictions below

Causes of Genetic Disorders

There are three main causes of genetic disorders. Gene mutations are changes in the nucleotide sequence of a gene. Chromosome mutations are changes in the structure of a chromosome. Non-disjunction is the incorrect assortment of chromosome during meiosis.

1. Gene Mutation

A change, even slight, to the chemical structure of a gene is called a mutation. The rate at which a given gene mutates is normally _____. But the number of genes that an individual carries is _____ and when including a whole species, the total number of genes is vast. For these reasons, mutations are _____. Pure chance dictates which gene in which individual mutates.

A mutation can cause a very subtle phenotypic effect or it can be dramatic. When a mutation causes the death of an organism, it is said to be _____. Many mutations are _____ _____and are not expressed unless the individual is _____.
_____ will quickly eliminate a harmful mutation from a natural population. Natural selection operates on the phenotype and only indirectly on the genotype. If a mutation is recessive, it can persist in a population masked by a dominant gene. Natural selection will not eliminate the mutation quickly if there is no harmful phenotype. _____ increases homozygosity and can greatly increase the likelihood of recessive genes being expressed.

In certain situations the harmful effect of the gene can help the individual. People homozygous for sickle-cell

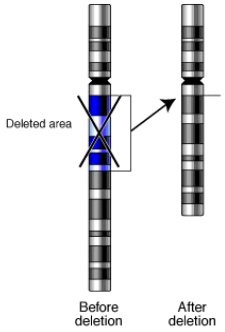
anemia (a condition where red blood cells are improperly shaped) have a reduced life expectancy. People with the heterozygous genotype have a _____, but they also have an _____ to malaria and therefore there is natural selection pressure to maintain the gene in the population. In this case, the benefit outweighs the cost to the individual. In regions where malaria is not a threat, selection pressure operates against the mutation. For example, the descendants of African blacks living in the United States show a steady decrease in the incidence of sickle-cell anemia.

Only rarely is a mutation beneficial to an organism resulting in natural selection propagating the gene.

2. Chromosome Mutations

Keep in mind that chromosomes have genetic information or instructions to produce _____ _____. Sometimes during meiosis, chromosomes break and rejoin incorrectly. If the genetic information is not in the correct place or is lost from the chromosomes, the cells will not _____. As a result, the corresponding proteins will not be made and the cell will not be able to perform all its functions.

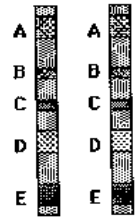
Deletions



When chromosomes break, the pieces can become lost. The genetic information in these pieces will never be expressed in the individual. This type of error is called a _____ and it can be _____, depending on which genes are lost.

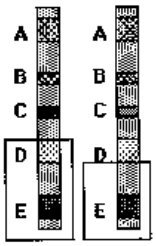
One well-known condition caused by a deletion is called the _____ _____syndrome. In this condition, a portion of the chromosome number 5 is deleted. The *cri du chat* syndrome is a French term describing how these children cry: they sound like a cat mewing. These children are _____ handicapped and do not live very long.

Duplications

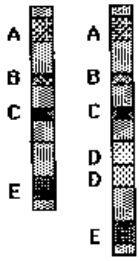


Another mutation is called a _____. Notice below the pair of chromosomes showing five genes. (Five genes are shown for the sake of simplicity--keep in mind that a chromosome has many other genes between the five labeled genes, A to E.)

The next diagram shows the same pair of chromosomes with boxes around segments of genes that will break away.



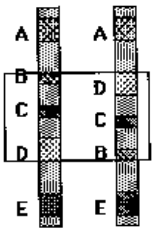
After breaking, these segments rejoin as shown below. Notice that one chromosome has a duplicate gene D.



Not all duplications cause harm. Some duplications may actually be _____, because the chromosome has a _____ for the cell.

Inversions

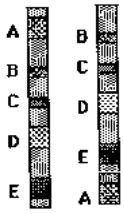
The next diagram shows an _____. Here a segment of a chromosome breaks off and rejoins the same chromosome but in a different orientation, or in a different order.



Noticed how B, C, and D have rearranged themselves.

Inversions may affect gene expression but usually do not have very harmful effects unless it happens in gametes. Gametes with inverted chromosomes _____ with its homologous chromosome and _____.

Translocations



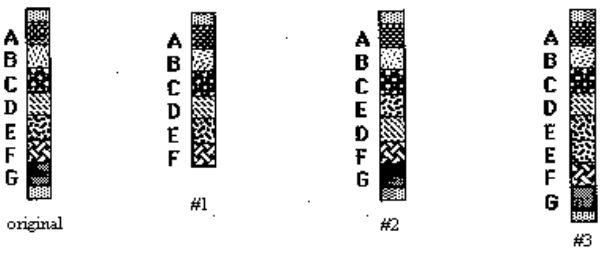
Sometimes, a broken piece of chromosome is added to a different part of the same or another chromosome. This mutation is called an _____.

In this example, the segment with gene A on the left has broken off and been added to the _____ of the same chromosome. Translocation is the cause of one form of _____.

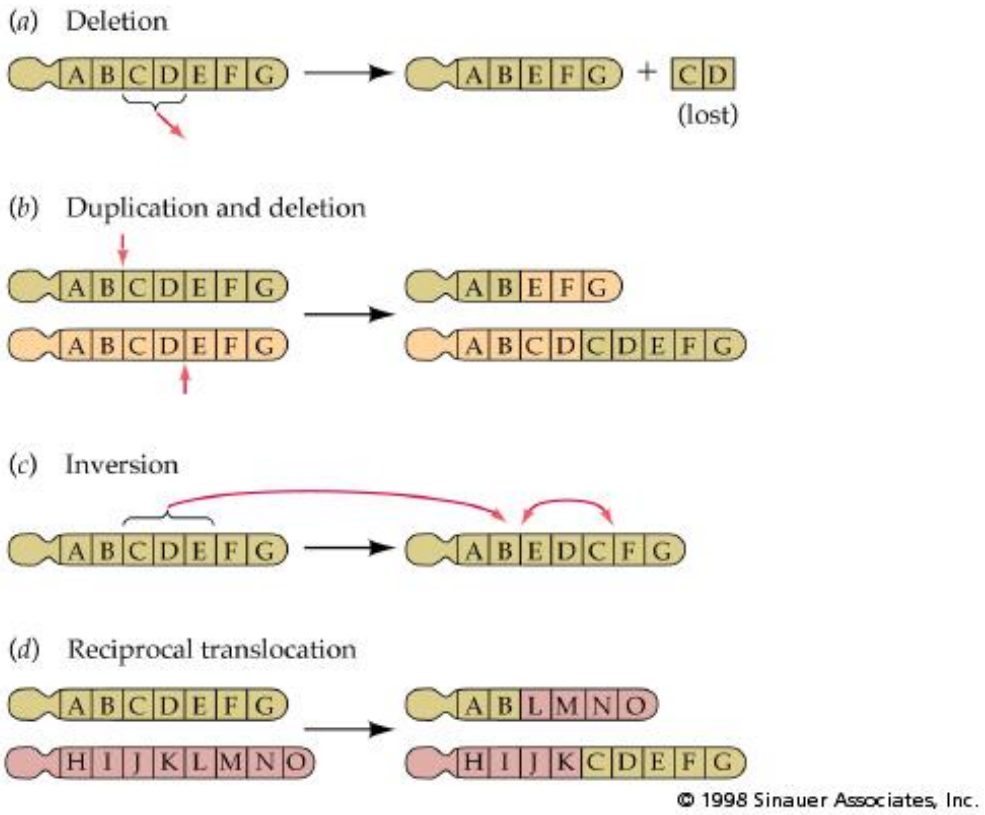
<u>Question</u>:

The diagram to the right shows a normal chromosome with genes A to G.
Label the type of mutation for each of the following errors.

1.
2.
3.



Summary:


(a) Deletion
A B C D E F G
A B E F G
+ C D
(lost)
(b) Duplication and deletion
A B C D E F G
A B C D E F G
A B E F G
A B C D C D E F G
(c) Inversion
A B C D E F G
A B E D C F G
(d) Reciprocal translocation
A B C D E F G
H I J K L M N O
A B L M N O
H I J K C D E F G
© 1998 Sinauer Associates, Inc.

3. Nondisjunction in Meiosis

Make a diagram that shows normal meiosis in a cell that has two chromosomes

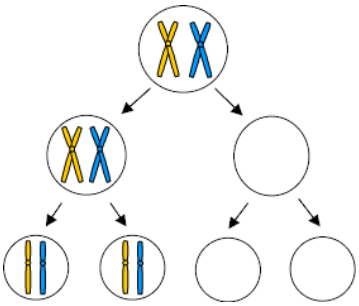
Nondisjunction:

_____

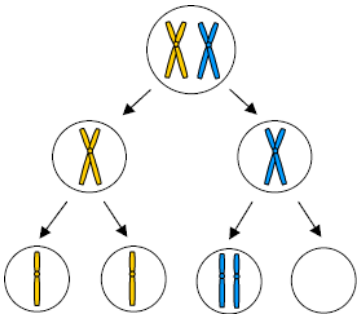
_____

_____

Nondisjunction in Meiosis I



Nondisjunction in Meiosis II



In meiosis I nondisjunction occurs when:

_____

_____

_____

_____

In meiosis II nondisjunction occurs when:

_____

_____

_____

_____

Most embryos from these gametes are inviable and abort spontaneously. The following combinations are viable in human embryos.

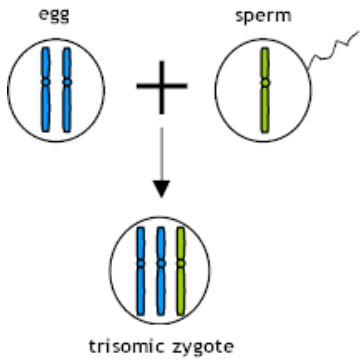
- XO, XXY, XXX and XYY (but not Y-)
- 3 copies of chromosome 13, 18 or 21

These embryos will have an extra chromosome in every cell of his/her body.

Trisomy: _____

When a normal egg and sperm fuse to create a human zygote, how many chromosomes exist?

_____



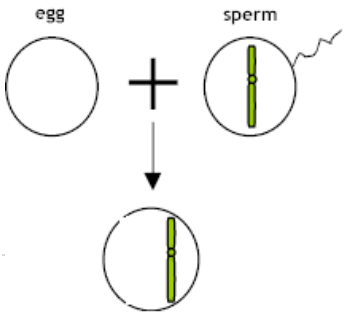
If a human sperm or egg carries an extra copy of a chromosome (due to nondisjunction), how many chromosomes will be present in the reproductive cell?

_____

If this sperm or egg fused with another normal sperm or egg, what will be the total number of chromosomes?

_____

Monosomy: _____



If a human sperm or egg carries one less copy of a chromosome (due to nondisjunction), how many chromosomes appear to be in the reproductive cell?

_____

If this sperm or egg fused with another normal sperm or egg, what will be the total number of chromosomes?

_____

Common Syndromes Caused by Nondisjunction:

1. Down syndrome (aka trisomy 21):
   -All persons with syndrome have extra genetic material associated with the 21st chromosome.
   -People with Down syndrome therefore have 47 chromosomes.
   -Symptoms include distinctive physical characteristics and varying degrees of mental retardation.
   -1 in 800 children born in Canada.

2. Turner syndrome:
   - All girls with syndrome are missing or have a damaged X chromosome (XO).
   - Individual has 45 chromosomes instead of 46.
   - Symptoms include underdeveloped sexual characteristics and infertility.
   - 1 in every 2500 girls born in Canada.

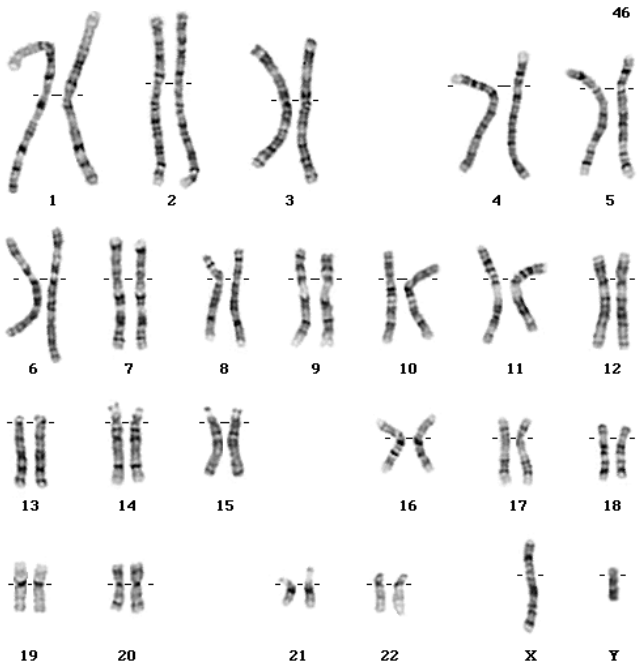
3. Klinefelter syndrome:
   - All boys with syndrome carry an extra X chromosome (XXY).
   - Is a condition where there is also a trisomy, but a trisomy of sex chromosomes.
   - These individuals are males with 47 chromosomes.
   - Symptoms include some mixed sexual characteristics after puberty.
   - 1 in every 1000 boys born in Canada.
   - infertility

4. Edwards syndrome:
   - Caused by trisomy 18 – three copies of chromosome 18
   - Individuals have 47 chromosomes in total
   - Symptoms include severe mental retardation, an elongated skull and a narrow pelvis
   - Individuals normally die in early infancy
   - Occurs in 1 out of 5000 live births

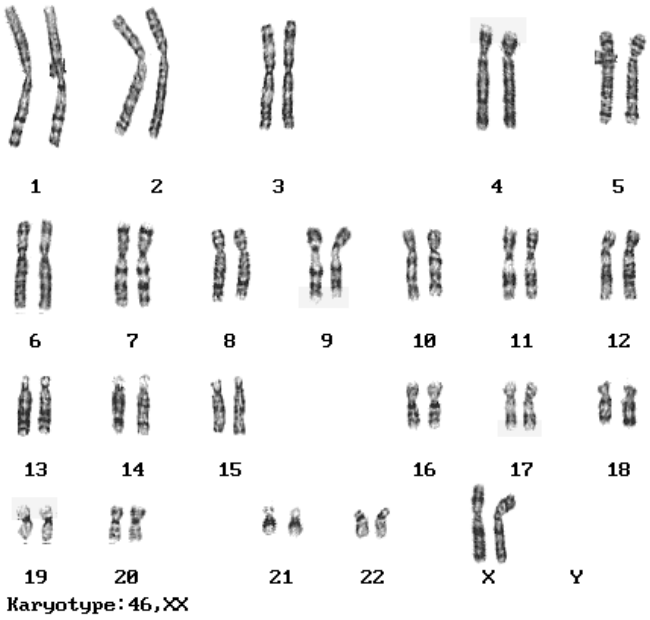
Karyotypes:

Karyotypes are used to diagnose genetic disorders due to chromosome mutations and non-disjunction. A karyotype is a preparation of the chromosomes from a cell of an individual during metaphase. The condensed chromosomes are stained to show bands in the DNA. Then a photograph is taken and a machine arranges the chromosomes in order of their size, largest to smallest. The photo then shows the numbers, sizes and shapes of all the chromosomes. Any abnormalities can be clearly seen.

Normal male karyotype:



Normal female karyotype:



Bioethics Decision Making

You will be assigned a case study in groups of three. In your group, decide which members will take the following roles: *doctor, counsellor, individual.*

Debate the bioethical issue and make a decision regarding the situation

1) Huntington Disease
   You and your older brother (20) have just found out that your father has been diagnosed with Huntington Disease. This is an incurable disorder that causes a slow progressive deterioration of the brain, resulting in death. Symptoms show in the affected individual around age 30 – 50. It is an autosomal dominant disorder, for which a genetic screening test has been developed. A DNA test can reveal with 100% certainty as to whether or not one will develop the disease.

2) Down Syndrome
   You have two healthy children from a previous marriage, but now you and your second husband would like to have a child together. You are 40 years old, and are concerned about the higher chance of having a child with Down Syndrome. This condition is caused by the presence of an extra chromosome 21, which leads to intellectual disability and health problems in the affected individual. Amniocentesis is available for pre-natal Down Syndrome diagnosis.

3) Hemophila
   You and your partner are thinking of starting a family. However, you have hemophilia, a sex-linked recessive bleeding disorder. You are being successfully treated with injections of Factor VIII, the blood-clotting enzyme your body lacks. No pre-natal screening tests are available for this disorder.

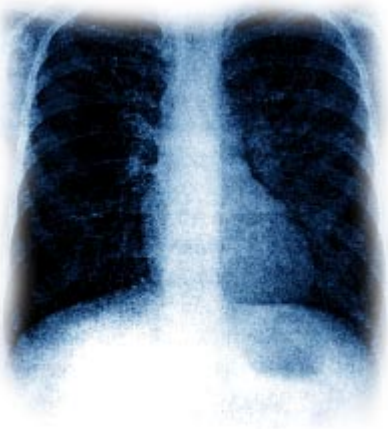
4) Cystic Fibrosis
   You and your spouse have just found out that your 14 month old daughter has cystic fibrosis. This is a fatal autosomal recessive disorder affecting the lungs and digestive tract. People with CF live shorter lives, and require daily medication and physical therapy. Your wife is pregnant again. Your doctor has informed you that genetic screening is available for pre-natal CF diagnosis.

5) Tay-Sachs
   When you were a child, you had a sister who died of Tay-Sachs disease. Now you and your husband want to start a family, but you have concerns about the risk of passing this recessive autosomal disorder to your children. Children born with Tay-Sachs suffer from progressive brain deterioration and loss of motor function. There is no treatment or cure, and death occurs in early childhood. Carriers of Tay-Sachs can be identified through a blood test, and amniocentesis can be used for pre-natal Tay-Sachs diagnosis.

6) Turner Syndrome
As a result of the information you have learned in this Biology course, you think you may have Turner Syndrome. Females with Turner Syndrome are usually short in stature, tend to be weak in mathematics and do not menstruate. They cannot have children. Your parents have never heard of this condition, caused by a missing X chromosome, and diagnosed with a blood test.

7) Sickle-cell Anemia
Sickle-cell anemia is present in both you and your partner's families. Blood tests have revealed that you are both carriers of the disease. Individuals with the disease tend to live shorter lives and suffer chronic pain, swelling in the joints and increased risk of infection, stroke and heart attack. There is no cure for the disease, and treatment involves the use of drugs and blood transfusions. Amniocentesis can be used to diagnose sickle-cell anemia in fetuses.

Sometimes it *is* All in the Genes



Part I—"The Genetic Test"

by
Anne Galbraith and **David Howard***
Department of Biology
University of Wisconsin—La Crosse

"Your pregnancy seems to be progressing just fine, but we'll do some blood work to be on the safe side. As long as we're drawing blood from you today, Nancy, would you consent to participate in one of the genetics studies we're conducting here at People's Best Hospital?" Dr. Kwin prided herself on putting her patients at ease, but her question caught Nancy a little off guard.

"What would that entail, Dr. Kwin?" While Nancy believed Dr. Kwin wouldn't ask her to do anything that wasn't a good idea, she never said yes to anything until she had all the information.

Dr. Kwin began to explain, "Well, as you know, PBH is a research and teaching hospital. One research team is trying to determine the frequency of the gene for the genetic disorder cystic fibrosis in the U.S. population. And since you are having blood drawn anyway as you enter your second trimester...."

Genetic disorder? Cystic fibrosis? The phrases made Nancy feel panicky. She interrupted Dr. Kwin mid-sentence. "You don't think I have that disease, do you? Isn't cystic fibrosis serious? How could I have caught it?" The questions came out in a rush.

Hearing the anxiety in Nancy's voice, Dr. Kwin quickly reassured her. "No, no. I'm quite sure that you do not have cystic fibrosis. You can't 'catch' it; you can only inherit it. If you did have cystic fibrosis, you would have been diagnosed when you were a toddler. You have absolutely nothing to worry about. However, let me emphasize that if you feel uncomfortable about participating in this study in any way, just say 'No.' My feelings won't be hurt. There is no pressure to participate."

"If it's inherited and I don't have it, why would they want to test me?" The situation still didn't make sense to Nancy.

"First, let me explain some basics. Cystic fibrosis, or CF, is a disease that is caused by defects in a particular gene. Actually because CF is genetic, 'disorder' is a better term than 'disease.' The word disease should really be used for illnesses caused by bacterial or viral infection. However, people use both words for CF.

"The reason that they need to include people who don't have CF in the study is that cystic fibrosis is a recessive disorder. With a recessive disorder, a person has to have two defective versions of the CF gene to have CF. Therefore you can be a carrier of one defective version but not have CF at all. It is precisely that which has inspired the study. They want to know what percentage of people in the United States carries one defective gene for cystic fibrosis. That percentage is called the gene frequency and can provide a lot of information for people working on CF."

Dr. Kwin went on to explain, "CF is one of the most common genetic disorders among Americans of European descent. Although it has many symptoms, the worst are severe respiratory problems that typically lead to death around the age of 30. One of the first reliable tests for any genetic disorder was the one developed for CF in 1990. Because CF is one of the first inherited illnesses that researchers have been able to get a handle on, these sorts of genetic screenings are pioneering work not only for CF research but also for understanding other genetic ailments."

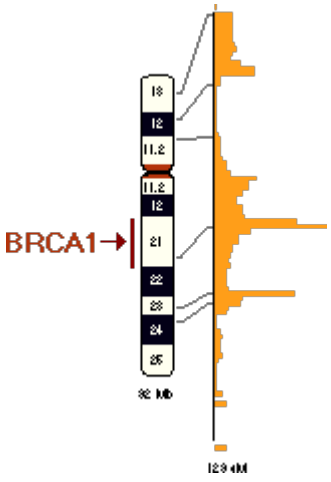
Nancy considered the flood of information for a few seconds. "I can't see what harm it would do. You run so many tests on me anyway. What's one more?"

"I'll have one of the staff bring you the consent forms and explain the procedure to you further. *If* you do consent, your biggest decision will be whether or not you want to have the results reported to you. You need to think carefully about that question before signing the consent form."

As Dr. Kwin left the room, Nancy thought to herself, *what are the pros and cons of knowing the results?*

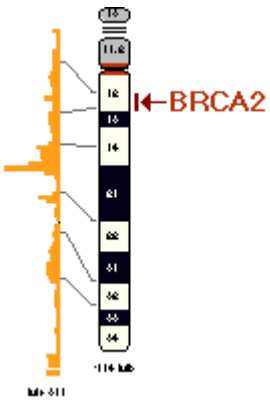
Questions:

1. Discuss why Nancy might or might not want to know the results of her blood test for CF.
2. Dr. Kwin told Nancy that she has "absolutely nothing to worry about." Although Nancy cannot get CF, is Dr. Kwin's statement entirely correct?
3. Did Dr. Kwin provide Nancy with enough information about cystic fibrosis and the test to make a good decision?
4. Should Nancy consent to the test? Provide the reasoning for your answer.



Genetic Testing and Breast Cancer: Is a Little Knowledge a Dangerous Thing?

by
Charlotte R. Zales, Education Department
Moravian College, Bethlehem, PA
Joseph C. Colosi, Biology Department



DeSales University, Center Valley, PA

Kathy was late for her book club meeting. She had had her yearly mammogram and the results had been negative—all was well. However, she had to wait, because she wanted to talk to Dr. Benjamin, the radiologist.
She raced into the restaurant where the book club was meeting, sat down with her friends, and burst into tears. Her friends gathered around her, concerned.
Mary spoke first. "Kathy, why are you so upset?"
Kathy calmed herself and looked at her friends. "I've just come from Dr. Benjamin's office."
Sarah interjected, "Please don't tell us that he found a lump."
"No, no," said Kathy. "My mammogram was negative. But we had the most upsetting conversation afterwards. I told him that my cousins are encouraging me to get tested for the breast cancer gene. They say that I should find out if I'm at risk."
Peter jumped in. "Of course you're at risk. Your daughter, Rachel, survived breast cancer—you know you are at risk. What's the matter with them?"
John added, "I guess they figure that since Rachel had breast cancer, she might have the gene, and she might have gotten it from you."
"Dr. Benjamin was explaining the gene to me, but I really don't get it. You know, the whole thing scares me," said Kathy. "I can't see what good can come from knowing if I have it."
John interjected, "Susan and I had ourselves tested for Tay Sachs. Knowledge is power, Kathy. Why wouldn't you get tested? The more you know, the better informed decisions you can make."
"Knowledge may not always be an ally," said Sarah. "Some years ago, when I was fired from my job, I suspected it was because I mentioned that my Dad had Huntington's disease."
"I'll tell you something good you can do," said Peter. "Eat healthy and exercise. That'll do you more good than all the worry and stress about testing."
Martha jumped in. "The knowledge from the Human Genome Project is exciting, Kathy. Every day at work we move closer to finding the causes of disease by our knowledge of genes."
"My parishioners are often telling me of good things accomplished by genetic testing," said Mary.
Kathy started crying again. "This is just too confusing for me."
Martha put her arm around Kathy. "We're here for you, Kathy. We'll sort this out. It sounds like each of us has some information that may help. Let's think about what we know that might be useful, and then we can share that with you. I'm sure there's a good answer for you."
Go to Role-Play Sheets for:
Martha John Sarah Peter Mary

Image Credit: The BCRA1 and BCRA2 genes map, respectively, to chromosomes 17 and 13. Illustrations from the National Center for Biotechnology Information.

Role-Play Sheet for Martha

Bioinformatics Researcher at a Pharmaceutical Company (Expertise—genetic and DNA analysis)

Bioinformatics is a new field of study that combines biology and information technology. It is used to analyze large databases of information, such as those resulting from the Human Genome Project.

- Breast cancer facts
  - 180,000 women a year are diagnosed with breast cancer.
  - 5%-10% of these are hereditary (caused by known breast cancer genes).

- Two breast cancer genes have been identified: BRCA1 and BRCA2.

- The normal functioning of both genes is to inhibit the development of cancerous cells. They are tumor suppressor genes.
  - A mutation in either gene raises a woman's lifetime chance of getting cancer from 12% to 85%.
    - A mutation in either gene also raises the risk for ovarian cancer.
    - A BRCA2 mutated gene raises the risk for breast cancer in males.
    - More than 600 different mutations that would increase the risk of breast cancer have been found in each gene.
    - There are many more mutations that have been found and are being studied. They may be harmless, and their connection to breast cancer is not yet known.
  - Every person has two copies of each gene. A mutation in one copy will increase the risk of breast cancer.
    - A person with one good copy and one mutated copy has a 50-50 chance of passing the mutated gene on to his/her children.
    - Virtually all individuals with a mutation in BRCA1 or BRCA2 have inherited it from a parent.

- Benefits of genetic information to research
  - The Cancer Family Registry for Breast Cancer Studies has information and lab specimens from more than 5,000 families with breast and/or ovarian cancer.
    - The Registry makes this information available to researchers.
    - The Registry provides information that would take researchers years to gather by themselves.
    - The information in the Registry is key to a speedy solution to breast cancer. The greater the number of families that provide information to the Registry, the more we will know about detecting and treating breast cancer and the faster we will learn how to prevent it.

---

1. **Would Martha encourage Kathy to get the test? Why or why not?**
2. **What other advice would Martha give Kathy?**
3. **How might these decisions affect Kathy cognitively, psychologically, emotionally?**

Role-Play Sheet for John

Father of Three Adopted Children
(Expertise—genetic testing as a means to information)

John and his wife, Barbara, decided to be tested for the Tay Sachs gene. John and Barbara both tested positive for the mutated form of the Tay Sachs gene. John and Barbara decided to marry anyway, but to adopt children rather than run the risk of having a child with Tay Sachs disease. (The book club members do not know this story—you may tell it to them.)

- Genetic testing
  - The test only requires a blood sample to be drawn.
  - The cost of genetic testing for breast cancer varies from several hundred to several thousand dollars, depending on the type and levels of testing required.

- Tests are available that can screen for cystic fibrosis, Duchenne muscular dystrophy, fragile X syndrome, Gaucher's disease, Huntington's disease, Lou Gehrig's disease, Marfan syndrome, Tay Sachs, and many others.

- Tay Sachs disease—a genetic disease that can be identified by genetic testing.
  - Children born with Tay Sachs, a genetic disease, die by the age of two.
  - Tay Sachs is a recessive disease—you need two copies, one from each parent, to get the disease. Each natural child of two carriers has a one in four chance of having the disease.
  - A massive government genetic screening program during the 1970s reduced the number of babies dying per year from 50-100 to less than 15.

- Law
  - The Americans with Disabilities Act (ADA) forbids employers from discriminating against disabled individuals who can do their job.
  - In March 1995, the Equal Employment Opportunities Commission (EEOC) said that people with genetic predisposition to disease are perceived as disabled and protected by the ADA.
  - The Health Insurance Portability and Accountability Act (HIPAA) of 1996 prohibits group health plans from denying coverage based on health status. An individual cannot be excluded just because of genetic information.

- News—Government genetic screening program recommendations address cystic fibrosis.
  - Cystic fibrosis (CF) is a genetic disease that usually affects the lungs and digestive system. How sick a person gets varies widely. There is no cure; most die by age 30.
  - CF is a recessive disease. A person who is a carrier has no symptoms.
    - While over 900 mutations have been found, about 25 are the major contributors.
    - The presence of the disease varies widely among ethnic groups.
  - Screening is recommended for individuals with a family history of CF, for partners of people with CF, for couples planning a pregnancy, and for couples seeking prenatal care. Testing for the general population is not recommended.

- Screening for the 25 mutations is more cost effective than for 900 mutations.
- Screening for the 25 mutations will identify carriers more accurately for ethnic groups that have the 25 mutations, and less accurately for ethnic groups with rare mutations.

---

1. **Would John encourage Kathy to get the test? Why or why not?**
2. **What other advice would John give Kathy?**
3. **How might these decisions affect Kathy cognitively, psychologically, emotionally?**

Role-Play Sheet for Sarah

Daughter of a Man who had Huntington's Disease
(Expertise—insurance/employment risks as a result of genetic testing)

Sarah's father died of Huntington's disease. She has a 50% chance of having inherited the disease. She mentioned at a staff meeting that she was the primary caretaker of her father who had Huntington's disease. One week later, she was fired, although her previous evaluations showed outstanding performance on the job.

- Employment risk example
  - Burlington Northern Santa Fe Railroad story—In 2000, this company drew blood samples during a physical exam from employees who had developed carpal tunnel syndrome on the job. The blood was sent for genetic testing, without employee knowledge or consent, in an attempt to find a gene that is in some way connected to carpal tunnel syndrome, so the employers could absolve themselves of their responsibility. One employee refused testing and was threatened with termination.

- Health insurance risk example
  - Danny's story—Danny is 7 years old and in perfect health. Genetic tests revealed that he has a gene predisposing him to a heart disorder. He takes medication to lower his risk of heart attack. His insurance company canceled his health insurance, stating that since he had the gene from birth, it was a pre-existing medical condition.

- Testing implications
  - Many surveys of physicians and patients have been conducted and revealed hundreds of cases of genetic discrimination. Examples:
    - In 1996 Georgetown University polled 332 families with perceived genetic risks:
      - 22% were refused health insurance.
      - 13% were fired from their jobs based on their risk.
    - A 1999 Yale Cancer Center survey of 296 genetic counselors, when asked how they would deal with genetic screening for themselves, reported:

- 68% said they would not bill their insurance company for the cost of genetic screening for fear of discrimination.
- 26% said they would give a false name.
- 57% said they would seek psychological counseling to cope with the results.

- Legislation
  - President Clinton signed an executive order prohibiting the federal government from using genetic testing results in any decision to hire, fire, or promote its employees. This bill covers 2 million federal employees. It does not apply to the private sector.
  - In 2001, a bill sponsored in the House of Representatives (H.R. 602) by Slaughter and Morella, and in the Senate (S. 318) by Kennedy and Daschle, would protect all Americans from genetic discrimination. In 2002, it is still in committee.
  - There are existing laws as of 2001 against genetic discrimination pertaining to:
    - health insurance: 42 states, plus 5 states with very minimal protection
    - employment: 21 states
    - disability insurance: 8 states
    - life insurance: 7 states
    - privacy of genetic information: 23 states

---

1. **Would Sarah encourage Kathy to get the test? Why or why not?**
2. **What other advice would Sarah give Kathy?**
3. **How might these decisions affect Kathy cognitively, psychologically, emotionally?**

Role-Play Sheet for Peter

Yoga Instructor
(Expertise—healthy eating, aerobic exercise, stress reduction)

More than 90% of women who get breast cancer do not have the gene. Every woman has a 12% chance of getting cancer.

Things you should know to reduce the risk of cancer:

- Diet
  - Alcohol increases the risk of cancer, including breast cancer. A few drinks a week is related to an increased risk.
  - Vegetables and fruits reduce cancer risk.
    - Normal metabolism can damage tissues, and this damage is related to an increased risk for cancer. Vegetables and fruits contain antioxidants that may protect tissues.
    - Antioxidants include: vitamin C, vitamin E, selenium, carotenoids, and phytochemicals.

- Obesity is related to increased cancer risk, including cancer of the breast among postmenopausal women.
- Saturated fats may increase cancer risk.
- Fats containing omega-3 fatty acids may reduce cancer risk.
  - Fish (such as salmon) are high in omega-3 fatty acids.
- Too little folic acid increases risk of cancer, including breast cancer.

- Exercise
  - Physical activity, weight control, and diet may delay or prevent cancer in people with genetic risk factors.
  - Moderate to vigorous levels of physical activity lowers risk of cancer, including breast cancer.

- Life style
  - Use of hormone replacement therapy after menopause for more than 5 years increases the risk of breast cancer.
  - Smoking is linked to cancer. Teenage smoking increases the risk of breast cancer.

- Psychological factors
  - When genetic testing reveals having the gene, psychological outcomes vary, and sometimes depend on the personality of the person tested.
    - Carriers expressed anger and worry.
    - Persons who had cancer reacted with anger, worry, discouragement, and various concerns upon finding that they had positive genetic test results.
    - Persons who underestimated the impact of testing were still distressed six months later.
    - Some persons wanted to join support groups.
    - Some no longer exhibited six months after testing.
  - When genetic testing reveals not having the gene:
    - There is psychological relief.
    - False negative results are possible and can lead to complacency.

- Medical support includes: regular mammograms, monthly self examination, and annual gynecological exam.

---

1. **Would Peter encourage Kathy to get the test? Why or why not?**
2. **What other advice would Peter give Kathy?**
3. **How might these decisions affect Kathy cognitively, psychologically, emotionally?**

Role-Play Sheet for Mary

Minister
(Expertise—making ethical decisions)

- Ethical responsibilities
  - Genetic testing allows you to give useful information to other family members.
    - All blood relatives have similar genetic inheritances.
    - If you are tested, you may feel responsible to share the information with other family members.
    - Some family members may want information, some may not.
  - Genetic testing now provides data for future studies that may help future generations, such as Kathy's grandchildren.

- Ethical worries
  - There is no guarantee that test results will remain private.

- Psychological factors
  - How does the possibility that a person has breast cancer affect their outlook?
    - How do you see yourself? How do you see the world?
  - Which aspects of your health do you regard with personal guilt?

- Questions, when a test for a disease is available:
  - Who should be screened?
  - At what age?
  - Who will explain the test beforehand?
  - Who will explain the results?
  - What about results of a test that is known to not be 100% accurate?

- Quote and comment from Dr. Francis Collins, Director of the National Center for Human Genome Research:
  - "It is not inconceivable that every woman in America may want to be screened for this [breast cancer] gene. The economic, ethical, and counseling issues will be very daunting."
  - In the near future, physical examinations for 18 year olds will include DNA testing for diseases with genetic components, and that physicians in the interest of preventive medicine will make risk-based recommendations for a healthy life style.

1. **Would Mary encourage Kathy to get the test? Why or why not?**
2. **What other advice would Mary give Kathy?**
3. **How might these decisions affect Kathy cognitively, psychologically, emotionally?**

Bioethics – Genetic Testing

Recent advances in genetics have raised ethical questions regarding the screening of individuals for inherited conditions and disorders. DNA screening, biochemical tests, amniocentesis and family pedigree analysis are all tools that are used by genetic counsellors. Some tests may be performed on individuals, others on a fetus.

These tools are used by genetic counsellors to analyze the risk to individuals for developing a disorder, or to parents of bearing a child with a known inherited disorder or condition. Genetic counsellors can present options to parents so that potential risks can be avoided or reduced.

There are many issues to be considered in genetic testing:

- Are tests equally available to all Manitobans, or only to those with the money to pay for the tests, or to those who live in larger urban centers?
- Should genetic testing be performed on individuals for disorders of which there is no available treatment (e.g. Huntington disease)?

Letter to the Editor Assignment:

Overview
Find and read two current articles on the topic of genetic testing. When you have completed the readings, they are to express their own point of view in a letter to the editor.

Resources
The following websites contain information about genetic testing and inherited conditions and disorders. You may use these websites or other websites from a ***credible source.***

Canadian Association of Genetic Counsellors (www.cagc-accg.ca)
Canadian Cystic Fibrosis Foundation (www.cysticfibrosis.ca)
Huntington Society of Canada (www.hsc-ca.org)
Canadian Hemophilia Society (www.hemophilia.ca)
Sickle Cell Association of Ontario (www.sicklecellontario.com)
Canadian Society for Mucopolysaccharide and Related Diseases (including Tay-Sachs) (www.mpssociety.ca)
The Genetic Science Learning Center website http://gslc.genetics.utah.edu contains a section called the Genetic Disorder Corner. Information on genetic disorders, their causes, genetic screening and the role of genetic counselors is provided in tutorials and interactive animations.

The Task:

As you read about various issues in newspapers or magazines, you are presented with different points of view. The more well read you are, the better you are able to formulate your own opinion on an issue. Your task it to find and read two current articles on the topic of genetic testing. A current piece should be written in the last two years. Try to find articles with a Canadian focus. If you wish, you may certainly read or view more articles on the topic to increase your understanding. Once you have completed your readings, you are to express your point of view in a Letter to the Editor.

Before you begin:

a) Consider what you have been learning in class on the topic. What are your responses to some of the issues raised?

b) As you read over the articles that you have selected, highlight the statements you wish to react to. Address yourself to the arguments outlined in each article. You may want to summarize the arguments briefly before refuting or reacting in the letter.

c) Look at sample letters to the editor from various newspapers. Consider what makes them powerful (or not).

Drafting the letter:

a) Have a strong opening. You must catch the editor's attention in order to be published. Put your introduction and main claim in the first paragraph.

b) Be persuasive. You are trying to convince someone of your point of view by reacting to the material in the articles you read. Refer to point from those articles in your letter and reference appropriately.

c) Make your points clearly and concisely. There is little space in most newspapers for letters; the briefer you are, the more likely you letter will be published. Make your letter 200 – 300 words in length.

d) You may use rhetoric to make your point. A rhetorical question is asked for effect with no answer expected.

e) Have a strong ending by leaving your readers with the most important thought.

Assessment

Research

Student / Teacher

_____ _____ /5 submitted two current articles dealing with the issue

_____ _____ /3 key statements were highlighted

_____ _____ /3 material from each article is used/reacted to in the letter

_____ _____ /2 material used is referenced in the letter (the author or article title is referred to)

Drafting Format

_____ _____ /3 followed guidelines (length, typed, proper referencing, layout)

_____ _____ /2 attention was paid to content, organization, style and mechanics

Expressing Your Opinion

_____ _____ /2 introduction and main claim are in the first paragraph

_____ _____ /2 clear point of view is expressed

_____ _____ /3 evidence is given from that point of view

_____ _____ /3 points are made clearly and concisely

_____ _____ /2 has a strong ending and leaves the reader with the most important thought

Total

____ ____ /30

Comments:

Study Guide: Genetics & Inheritance

* Be sure you do the learning checklist at the beginning of the unit *

SHORT ANSWER QUESTIONS:

A. MEIOSIS, MENDEL'S LAWS, CHROMOSOMES, & SEX CELLS (Gametes)

1. The term "meiosis" comes from Greek. It means _____
2. Non-sex chromosomes are called _____
3. Two copies of a particular chromosome in a diploid cell, each copy being derived from a different parent are called _____.
4. The twin copies of the fully replicated chromosome that are separated completely from each other at meiotic anaphase II to become individual chromosomes are called _____.
5. During which stage of meiosis do chromatids separate completely? _____
6. List and describe Mendel's laws.

7. Gregor Mendel concluded that traits are…
   a) Not inherited by offspring
   b) Inherited through the passing of factors from parents to offspring
   c) Determined by dominant factors only
   d) Determined by recessive factors only

8. Unlike mitosis, meiosis results in the formation of…
   a) Diploid cells
   b) Haploid cells
   c) Body cells
   d) None of the above

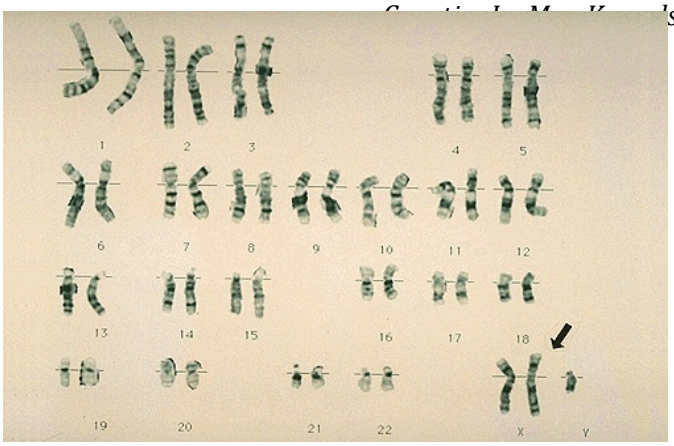
9. During which phase of meiosis do homologous chromosomes segregate?
   a) Metaphase 1
   b) Anaphase 1
   c) Metaphase 2
   d) Anaphase 2

10. During which phase of meiosis does synapsis occur?
   a) Prophase 1
   b) Metaphase 1
   c) Anaphase 1
   d) Telophase 1

11. In the diagram of the cell, how many chromatids are present in the cell?
   a) 8
   b) 16
   c) 4
   d) 2

12. How many chromosomes are found in every human body cell? _____
13. How many **pairs** of chromosomes are found in every human body cell? _____
14. Name the 2 types of gametes: _____& _____
15. How many chromosomes are found in every human gamete? _____
16. What process forms the sex cells? _____
17. What chromosomes determine the (normal) male gender? _____
18. What chromosomes determine the (normal) female gender? _____
19. On a chromosome, genetic information that relates to a specific trait is called: _____
20. Where is the extra chromosome located on a Downs Syndrome patient? _____
21. Where is the extra chromosome located on an Edward Syndrome patient? _____
22. A person with these sex chromosomes, XXY, suffers from: _____
23. A person with just an X-chromosome suffers from: _____
24. If a human cell has a DIPLOID number of chromosomes, there would be this many chromosomes in each cell: _____
25. If a human cell has a HAPLOID number of chromosomes, there would be this many chromosomes in each cell: _____
26. Cri du Chat is a disorder affecting this chromosome: _____

27. The Karyotype to the right shows
    a) Normal Female
    b) Male with Klinefelter's syndrome
    c) Female with Turner's Syndrome
    d) Normal Male



B. PUNNET SQUARE PROBLEMS:

Monohybrid Crosses

1. Suppose 5 digits (D) are dominant over 6 digits (d). If a heterozygous male mates with a homozygous recessive female, determine the genotypic and phenotypic ratio of the F_1 offspring

2. In Horses, black hair is dominant over white hair. If two heterozygous horses mate what is the genotypic and phenotypic ratio of the F1 generation?

3. In the above question, suppose a member of the F_1 generation mates with a horse that is homozygous dominant. What is the probability of having a white horse in the F_2 generation?

4. Suppose a green seeded plant (g) and a yellow seeded plant (G) produced plants with all yellow seeds. What are the genotypes of the P-generation?

5. In Golden Retrievers, short hair (S) is dominant over shaggy hair (s). Khrystyna sold Margo a purebred Golden Retriever with short hair (or so she claims). How can Margo find out if Khrystyna was telling the truth? Explain.

6. In an F_1 generation, there exist a total of 2500 pea plants. 1250 pea plants were discovered to have a heterozygous genotype. 630 pea plants had a homozygous dominant genotype and 620 had a homozygous recessive genotype. What genotype must the parents have?

DIHYBRID CROSSES

7. How many traits are involved in a dihybrid cross? _____

8. In humans, blonde hair (b) is recessive to brown hair (B) and Huntington Disease (n) is recessive to the normal (N) condition. If a homozygous normal blonde haired person marries a heterozygous brunette with Huntington Disease, what percentage of their children will:
    a) Be normal with blonde hair?
    b) Be normal with brown hair?
    c) Have Huntington Disease?
    d) Have Huntington Disease with blonde hair?
    e) Have Huntington Disease with brown hair?

CO-DOMINANT CROSSES

9. How many traits are involved in a co-dominant cross? _____

10. In a court case involving a paternity dispute, a man claims that a male child with a **BO-blood type** is his biological son. However the mother claims that he is not the father.

    If the man has type **O-blood** and the woman has type **AB-blood**, could the child be his son? Show your work using a Punnett square and state the probability.

11. Brenda has two dominant alleles for B-type blood. She marries Conner who is A-type blood, but it is unknown whether he his AA-type or AO-type. Conner and Brenda had 4 children. Half of them had AB-type blood and the other half had BO-type blood. What is Conner's exact genotype?

INCOMPLETE DOMINANCE

12. How many traits are involved in an incomplete dominant cross? _____

13. How many phenotypes may arise from a single incomplete dominance cross? ____

14. Suppose an exotic flower shows incomplete dominance for pedal colour. Assume that blue (B) is incompletely dominant over yellow (Y). If a blue flower pollinates with a yellow flower, what are the genotypic and phenotypic ratios?

15. In Snapdragons, red (R) and white (W) are incompletely dominant. If a pink snapdragon pollinates with another pink snapdragon, what is the probability of having:
    a) Pink snapdragons in the F1 generation?
    b) Red snapdragons in the F1 generation?
    c) White snapdragons in the F1 generation?

EPISTASIS

16. In a certain breed of dog, the dominant, B, is required for black fur; it's recessive, b, produces brown fur. However, the dominant, I is the epistatic to the color locus and can inhibit pigment formation. The recessive allele, i, on the other hand, permits pigment deposition in the fur. What would be the phenotypes of the following sets of parents and what would be the results of the mating?

    a. bbii x Bbli b. bbii x Bbii c. bbli x BBli

C. GENETIC DISORDERS:

1. What **kinds of disorders** may occur as a result of **trisomy**? Give 3 possible disorders and describe the characteristics of each condition.

2. Explain why mutations are necessary to our survival as a species.

3. Explain how mutations may cause harmful effects in our bodies.

D. Pedigrees

On a pedigree:

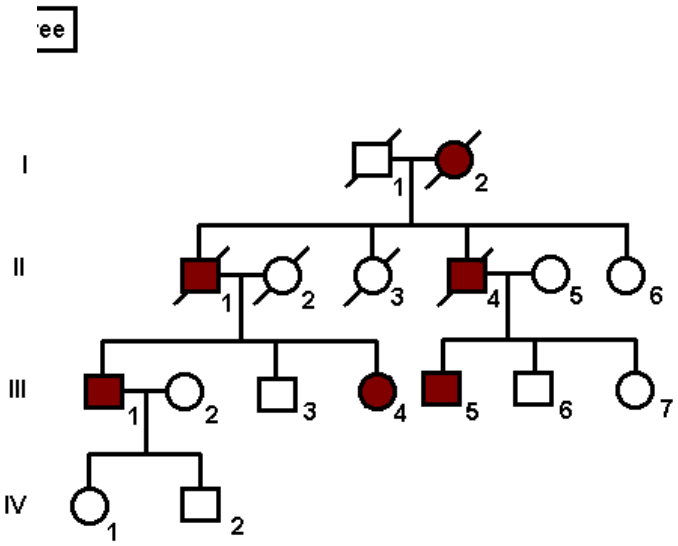
1. What does a square represent? _____
2. What does a circle represent? _____
3. What does a shaded circle or square represent? _____
4. What does a diagonal line crossing through a square represent? _____
5. What does a horizontal line joining a circle and square represent? _____
6. What does a vertical line represent? _____

Draw the correct pedigree based on the following information:

- This pedigree follows the recessive disorder sickle-cell anemia.
- There are 4 generations in this pedigree.
- The P-generation appear to be unaffected by sickle cell anemia. They sire 2 children: a boy and then a girl who both appear to be unaffected by sickle cell anemia.
- The F_1 generation boy marries a girl who is a carrier. They have 4 children: a boy, boy, girl and boy. Only the girl has sickle cell anemia.
- The F_1 generation girl marries a homozygous normal man. They have 2 girls who are not affected with sickle-cell anemia
- Individual III-4 marries a woman who is heterozygous normal. They have 2 girls and 2 boys (in that order). Only the eldest boy and eldest girl have sickle-cell anemia.

Determine all the genotypes of each individual in the pedigree.

Determine all the genotypes of each individual. Every shaded shape represents a person who has green eyes. Green eye colour (b) is a recessive trait while brown eyes are dominant (B)



I-1 _____ I-2_____

II-1 _____ II-2_____ II-3_____ II-4_____ II-5_____ II-6_____

III-1_____ III-2_____ III-3_____ III-4_____ III-5_____ III-6_____ III-7_____

IV-1_____ IV-2_____

Terms & Concepts to Know:

- □ Chromosomes
- □ Gene
- □ Allele
- □ Dominant
- □ Recessive
- □ Homozygous
- □ Heterozygous
- □ Genotype
- □ Mitosis
- □ Meiosis
- □ Klinefelters
- □ Turners Syndrome
- □ Carrier
- □ Red-Green Colorblindness
- □ Phenotype
- □ Punnett Square
- □ Filial (F_1, F_2, …)
- □ Test Cross
- □ Segregation
- □ Independent Assortment
- □ Inheritance
- □ Mendel
- □ Co-Dominance
- □ Incomplete Dominance
- □ Mutations
- □
- □ Gametes
- □ Monohybrid Cross
- □ Dihybrid Cross
- □ Purebred
- □ Hybrid
- □ Pedigree
- □ Autosome
- □ Sex Chromosome
- □ Marriage Line
- □ Downs Syndrome
- □ Cri du Chat
- □ Hemophilia
- □ Gametes
- □ Karyotype
- □ Diploid
- □ Haploid
- □ Sex-Linked Trait
- □ Non-disjunction
- □ XX
- □ XY
- □ Trisomy 21
- □ Edwards Syndrome

MEIOSIS WORKSHEET – KNOWING THE STEPS IN CREATING YOUR GAMETES!

Instructions: Below are drawings in the stages of meiosis. Cut these out and put them in the proper order for meiosis on the next sheet provided. You will also need to record the main events that are happening at each stage.