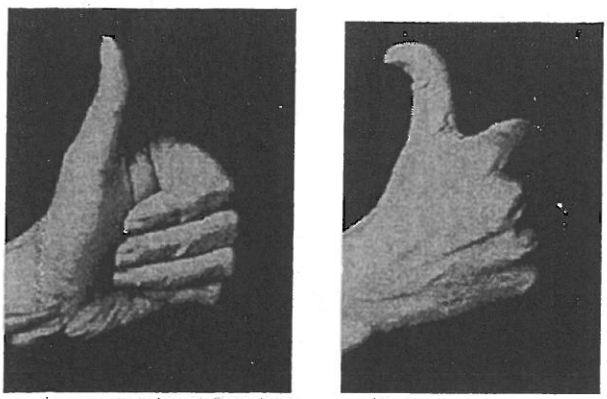
LaGuardia Community College City University of New York

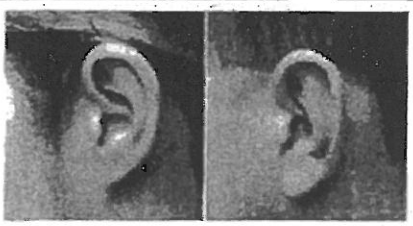
SCB 204: Human Anatomy & Physiology II

GENETICS - Lab 12

Which of these genetic traits do you have?



Hitchhiker's Thumb



attached free, or detached

Earlobe Attachment

widow's peak straight hairline

Forehead Hairline

Thumb = _____

Hairline = _____

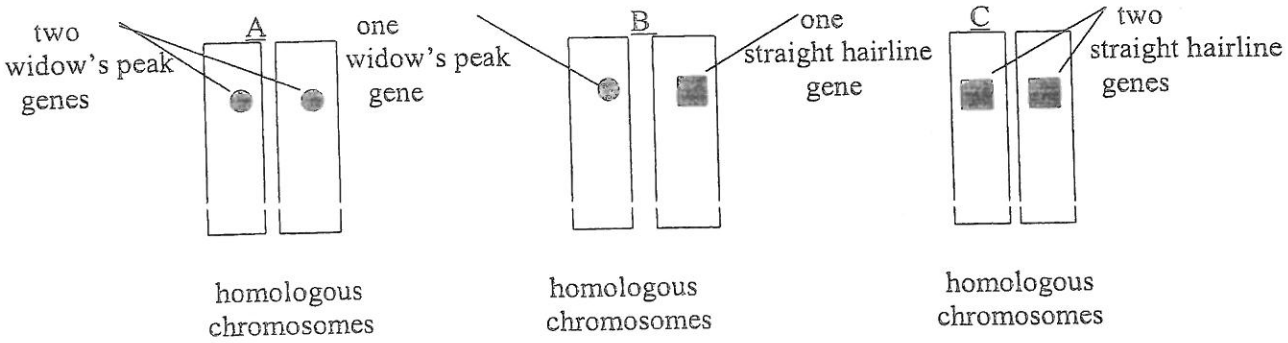
Earlobe Attachment = _____

Your genetic traits are determined by the genes that you carry on your chromosomes. These genes interplay with environmental factors to determine which characteristics (traits) you have. Genes (DNA) are made of pairs of nitrogenous bases lined up in a specific order. The sequence (order) in which your nitrogenous base pairs (adenine-thymine, cytosine-guanine) occur determines what a specific gene will be. A gene for an attached earlobe will have a different sequence of nitrogenous base pairs than a gene for a detached earlobe. The total human genome has approximately 3 billion (3,000,000,000) nitrogenous base pairs.

How do genes determine your characteristics?

Although you have 46 chromosomes, the chromosomes are not all different from each other. Chromosomes come in pairs. Thus, humans have 23 pairs of chromosomes (46 ÷ 2= 23). The paired chromosomes generally look alike and they carry genes for the same traits – we call them homologous chromosomes. Generally, every trait we have is controlled by at least 2 genes, one on each of the 2 homologous chromosomes.

For example, a person's hairline is determined by whether the homologous chromosomes which carry the information for hairline have a widow's peak gene or a straight-hairline gene. There are 3 possibilities.



(This illustration is considering only one pair of your 23 pairs of chromosomes. The other 22 pairs are not shown but they carry genes in the same manner.)

A person with two of the same genes (2 widow's peak or 2 straight - hairline genes – illustrations A and C) are said to be pure for that trait or homozygous for that trait. A person with 2 different genes (illustration B) for a trait is said to be heterozygous (hybrid) for that trait. Different versions of a gene are called alleles, so there are two alleles for forehead hairline (widow's peak and straight).

How will the three people (A, B and C) illustrated above look with regard to hairline?

Person A = _____

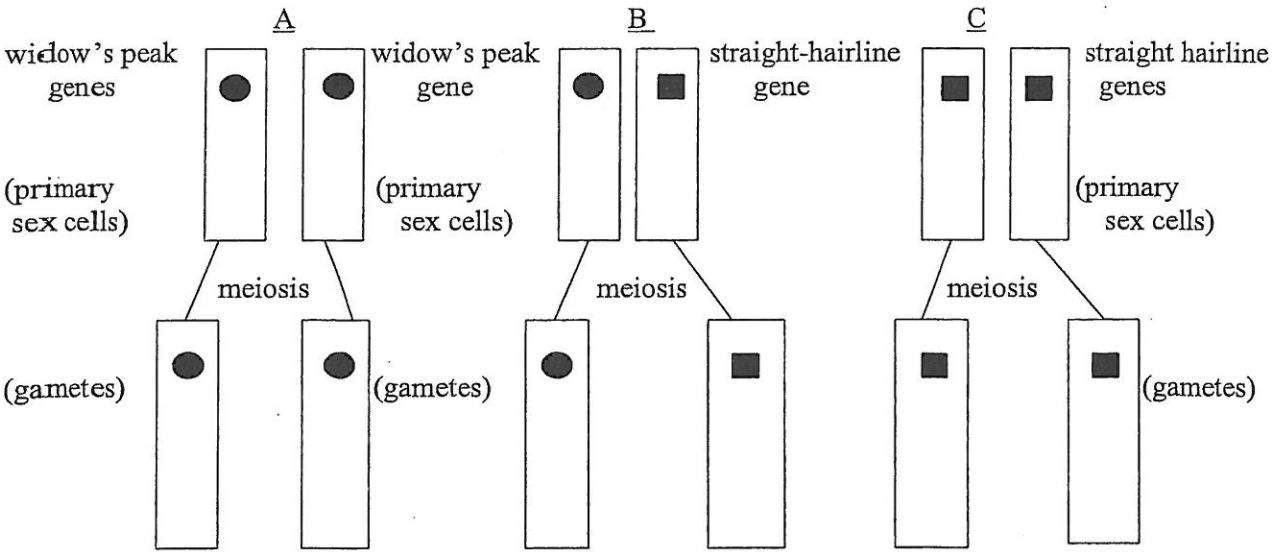
Person B = _____

Person C = _____

Person A will have a widow's peak. Person B will have a widow's peak and person C will have a straight hairline. Why will person B have a widow's peak? When a person is heterozygous for a trait he carries 2 different genes for that trait. Generally, one gene will be "stronger" than the other and will dominate; this is called the dominant gene. The gene that is not expressed ("weaker") is said to be the recessive gene. The widow's peak gene is dominant over the straight hairline gene; when they occur together a person will have a hairline with a widow's peak.

How are our children's (off-spring's) traits determined?

Homologous chromosomes separate during meiosis (when gametes form – egg and sperm). Thus, humans only have 23 chromosomes in each gamete (one chromosome from each homologous pair) and as a result only one gene for each trait. For example, our 3 people would each form gametes with the following distribution of genes.



(This illustration is showing only one of our 23 pairs of chromosomes; the other 22 pairs separate also and distribute themselves similarly.)

Thus, persons A and C each produce only one kind of gamete (A has widow's peak gametes and C has straight hairline gametes) but person B would produce two kinds of gametes (half with widow's peak genes and the other half with straight hairline genes).

If persons A and C mated, would their children have straight hairlines or widow's peaks?

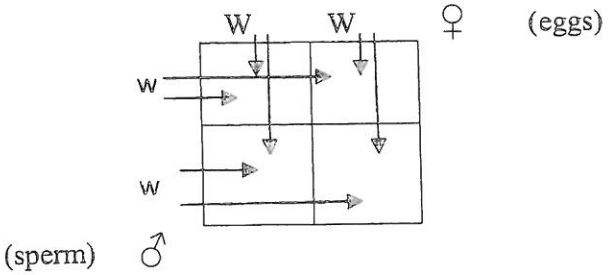
This is a genetic problem and when we do genetic problems we usually draw the traits we are considering as letters of the alphabet. The dominant trait is written with a capital letter and the recessive with a small letter. We call genetic problems – genetic crosses. When we do genetic crosses we ignore all the other traits (genes and chromosomes) a person may have except the one(s) we are working on. To find out the outcome of the mating of any 2 people we usually make a chart called a Punnett square, in which we list all the possible gametes of the father on the side of the chart and all the possible gametes of the mother on the top.

Thus, to answer our question about the children of couple A and C we would do the following:
First, we would make a Key where we would define what traits we are considering, which letters of the alphabet we are using and which is the dominant trait. For example:

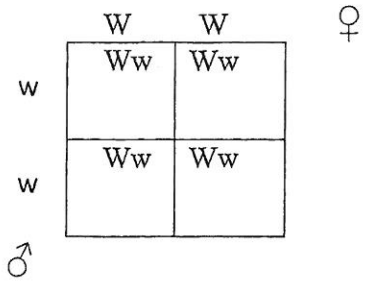
Key
W = widow's peak gene
w = straight hairline gene

Next, we would show the genetic composition of each parent (of course only with regard to the trait (s) we are considering (e.g. hairline) and the types of gametes those parents would produce.

Then we would make a Punnett Square with 2 spaces on the top and 2 spaces on the side for the parents' gametes. The Punnett Square would look like this. Finally, we would fill in each empty box by using the letter at the top of that column and the letter at the side of that row.



See the illustration below. This represents all the possible fertilization events that could occur between these two individuals.



By doing this we see that the father's gametes with the **w** gene will always combine with a mother's gamete carrying a **W** gene. Thus, all the children of this couple will have a genetic composition (genotype) of **Ww** with regard to hairline and they all will be heterozygotes. Despite the fact that they all carry a gene for straight hairline, their hairline will be a widow's peak in appearance (phenotype) because their appearance will express the dominant gene. Thus, a man with a straight hairline will only have children with a widow's peak if he mates with a woman homozygous (pure) for a widow's peak hairline.

We must review the terms genotype and phenotype. The term genotype refers to the genetic composition of an individual (s) – what traits they have and whether they are heterozygous (hybrid) or homozygous (pure) for those traits. The phenotype refers to the appearance of the individual (s) and does not concern itself with whether or not the individual is pure or hybrid for the trait.

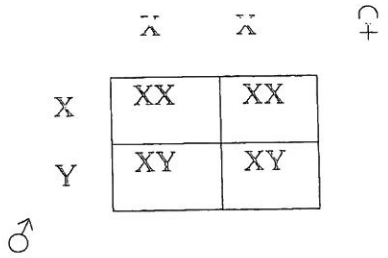
Genetic inheritance is often more complicated than simple Dominance. Some traits exhibit Incomplete Dominance; in this case both genes express themselves when in a heterozygote. This is the case with the disease Sickle Cell Anemia. In this disease the hemoglobin of the Red Blood Cell changes shape under low oxygen conditions and causes the RBC's to take on a crescent shape with pointy ends. The pointy ends of the RBC's cause the cells to get caught in narrow blood vessels and sometimes to rupture. This is a painful and possibly life threatening condition. A person homozygous for normal hemoglobin (SS) is fine. A heterozygote individual (Ss) has Sickle Cell Trait where half their hemoglobin is normal and half will be abnormal. This is not a severe condition; however, the person that is homozygous recessive (ss) will have only abnormal hemoglobin and as such, Sickle Cell Anemia. The gel electrophoresis experiment you are doing is to determine whether or not a child will have Sickle Cell Anemia by examining the migration pattern of his DNA and that of his parents.

Thus far we have examined the inheritance of traits based on the presence or absence of a specific gene. However, the presence or absence of an entire chromosome has far reaching consequences. An abnormal condition called Down's syndrome (Mongolism) occurs when a child inherits an extra 21st chromosome and so has 47 instead of 46 chromosomes. Although Down's syndrome is an abnormal condition, normal sex inheritance also depends on the presence or absence of a chromosome, a chromosome called the Y

chromosome. All normal humans have 23 pairs of chromosomes in their cells. Twenty-two we call the autosome chromosomes but the 23rd pair we call the sex chromosomes; we use the letters X and Y to symbolize these chromosomes. The Y chromosome is smaller than the X chromosome and carries very few genes. A man has an X and a Y chromosome and a woman has 2 X chromosomes. Because a woman is XX, her gametes (eggs) will all have X chromosomes but a man is XY and so half of his gametes (sperm) will carry an X chromosome and the other half will carry a Y chromosome (see below).

How is the sex of a child determined?

If we do the Punnett square we see the answer.



The sperm with the X chromosome is as likely to fertilize an egg as the sperm with the Y chromosome and so there is as good a likelihood of having a female child as a male child (or the chances are 50-50). Of course, you know this from your life experiences but now you know why. Actually, there is a slightly greater chance of having a male child. This is so because the **Y chromosome has fewer genes than an X chromosome** and so a sperm carrying a Y chromosome is likely to swim faster and reach the egg before a sperm carrying an X chromosome.

Inheritance of the Y chromosome (or some say the number of X chromosomes) determines an individual's sex.

The fact that the Y chromosome has so few genes can create a problem for a man. A man only has one X chromosome. If his X chromosome is carrying a recessive abnormal gene he will have no gene on his Y chromosome to counter-act it. On the other hand, a woman has two X chromosomes and so a recessive abnormal gene on one of her X chromosomes will be "masked" by the dominant normal gene on her other

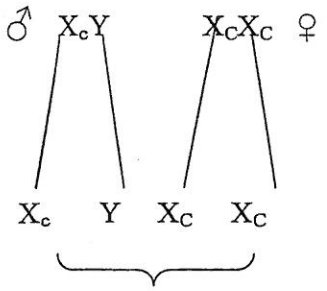
X chromosome. The traits which are carried on the X chromosome are called sex-linked traits (or X-linked) because they are inherited with the X chromosome. Abnormal recessive traits such as colorblindness, baldness and hemophilia are carried on the X chromosome and so are much more commonly found in men than women. (See the genetic cross below).

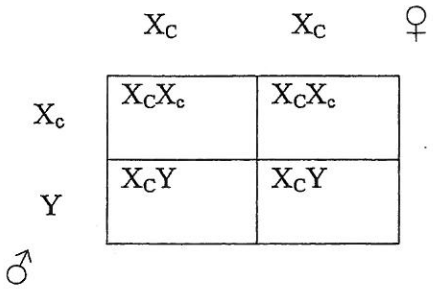
If a colorblind man and a woman who is homozygous for normal color vision mate, what will be the genotype and phenotype of their off-spring?

Key

X_c = color blind gene on x chromosome
X_C = normal color vision gene on X chromosome

Parents





Results

Genotype: Two females heterozygote for color vision. They are called carriers because even though they are not colorblind, they carry the gene for colorblindness which they can pass on to their children. Two males normal for color vision.

Phenotype: All the children will have normal color vision. (100% normal vision)

NOW IT'S YOUR TURN TO ANSWER SOME QUESTIONS.

Genetics Lab 12 SCB204 Name: _____

ANSWER ALL THESE QUESTIONS DIRECTLY ON THE QUESTION PAPER. YOU MAY USE THE REVERSE SIDE IF NECESSARY.

Define:

a. Homologous chromosome

b. Allele

c. Homozygote (pure)/Heterozygote (hybrid)

d. Dominant/Recessive gene

e. Genotype/Phenotype

f. Sex-linked trait

2. Problems – Answer each of the following problems. Be sure to show all your work for each cross. Make a key, show the genetic composition of each parent, what their gametes will look like and a completed Punnett's square.

   a. What will be the genotype and the phenotype of the children from a cross between 2 heterozygote people with widow's peak hairlines?

   b. What will be the genotype and the phenotype of the children of a woman who is a carrier of colorblindness and a man who has normal color vision?