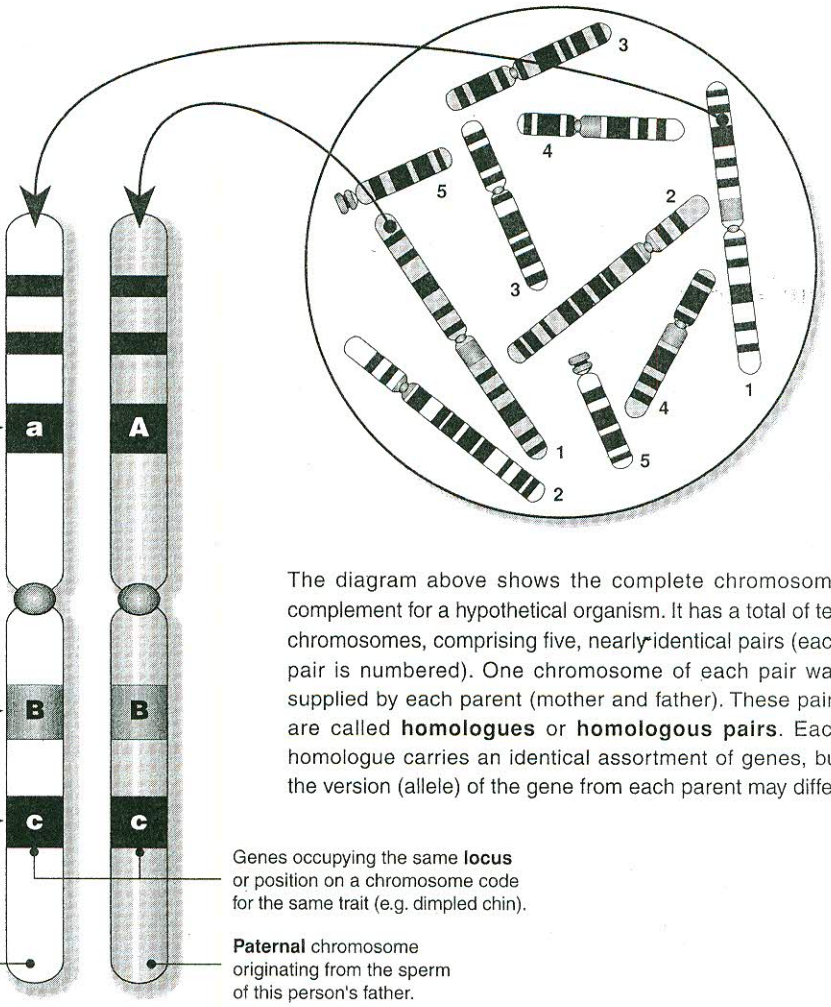
Alleles

Sexually reproducing organisms in nearly all cases have paired sets of chromosomes, one set coming from each parent. The equivalent chromosomes that form a pair are termed **homologues**. They contain equivalent sets of genes on them. But there is the potential for different versions of a gene to exist in a population and these are termed **alleles**.

Homologous Chromosomes

In sexually reproducing organisms, most cells have a homologous pair of chromosomes (one coming from each parent). This diagram shows the position of three different genes on the same chromosome that control three different traits (A, B and C).



These two different versions of gene A create a condition known as **heterozygous**. Only the dominant allele (A) will be expressed.

When both chromosomes have identical copies of the dominant allele for gene B the organism is said to be **homozygous dominant** for that gene.

When both chromosomes have identical copies of the recessive allele for gene C the organism is said to be **homozygous recessive** for that gene.

Maternal chromosome originating from the egg of this person's mother.

Genes occupying the same **locus** or position on a chromosome code for the same trait (e.g. dimpled chin).

Paternal chromosome originating from the sperm of this person's father.

The diagram above shows the complete chromosome complement for a hypothetical organism. It has a total of ten chromosomes, comprising five, nearly-identical pairs (each pair is numbered). One chromosome of each pair was supplied by each parent (mother and father). These pairs are called **homologues** or **homologous pairs**. Each homologue carries an identical assortment of genes, but the version (allele) of the gene from each parent may differ.

1. Define the following terms used to describe the allele combinations in the genotype for a given gene:

   (a) Heterozygous: _____

   (b) Homozygous dominant: _____

   (c) Homozygous recessive: _____

2. For a gene given the symbol 'A', name the alleles present in an organism that is identified as:

   (a) Heterozygous: _____ (b) Homozygous dominant: _____ (c) Homozygous recessive: _____

3. Explain what a homologous pair of chromosomes is: _____

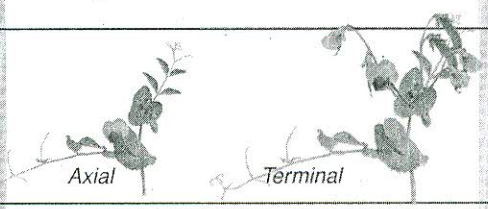
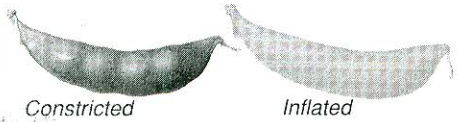
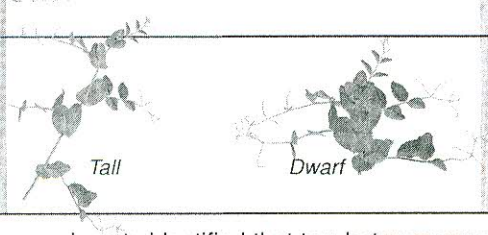
4. Discuss the significance of genes existing as **alleles**: _____

Mendel's Pea Plant Experiments

Gregor Mendel (1822-1884), pictured on the right, was an Austrian monk who is regarded as the 'father of genetics'. He carried out some pioneering work using pea plants to study the inheritance patterns of a number of **traits** (characteristics). Mendel observed that characters could be masked in one generation of peas but could reappear in later generations. He showed that inheritance involved the passing on to offspring of discrete units of inheritance; what we now call genes. Mendel examined a number of phenotypic traits and found that they were inherited in predictable ratios, depending on the phenotype of the parents. Below are some of his results from crossing heterozygous plants (e.g. tall plants that were the offspring of tall and dwarf parent plants: Tt x Tt). The numbers in the results column represent how many offspring had those phenotypic features.

1. Study the **results** for each of the six experiments below. Determine which of the two phenotypes is the dominant one, and which is the recessive. Place your answers in the spaces in the **dominance** column in the table below.

2. Calculate the ratio of dominant phenotypes to recessive phenotypes (to two decimal places). The first one (for seed shape) has been done for you (5474 ÷ 1850 = 2.96). Place your answers in the spaces provided in the table below:

Trait	Possible Phenotypes	Results	Dominance	Ratio
Seed shape	 *Wrinkled* *Round*	Wrinkled 1850 Round 5474 **TOTAL 7324**	Dominant: Round Recessive: Wrinkled	2.96 : 1
Seed color	 *Green* *Yellow*	Green 2001 Yellow 6022 **TOTAL 8023**	Dominant: Recessive:	
Pod color	 *Green* *Yellow*	Green 428 Yellow 152 **TOTAL 580**	Dominant: Recessive:	
Flower position	 *Axial* *Terminal*	Axial 651 Terminal 207 **TOTAL 858**	Dominant: Recessive:	
Pod shape	 *Constricted* *Inflated*	Constricted 299 Inflated 882 **TOTAL 1181**	Dominant: Recessive:	
Stem length	 *Tall* *Dwarf*	Tall 787 Dwarf 277 **TOTAL 1064**	Dominant: Recessive:	

3. Mendel's experiments identified that two heterozygous parents should produce offspring in the ratio of three times as many dominant offspring to those showing the recessive phenotype.

   (a) State which three of Mendel's experiments provided ratios closest to the theoretical 3:1 ratio:

   _____

   (b) Suggest a possible reason why these results deviated less from the theoretical ratio than the others:

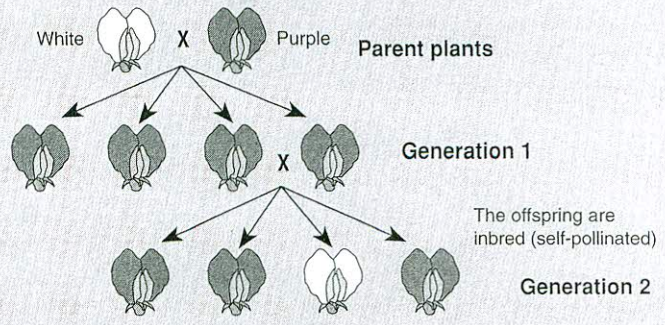
   _____

Mendel's Laws of Inheritance

From his work on the inheritance of phenotypic traits in peas, Mendel formulated a number of ideas about the inheritance of characters. These were later given formal recognition as Mendel's laws of inheritance. These are outlined below.

The Theory of Particulate Inheritance

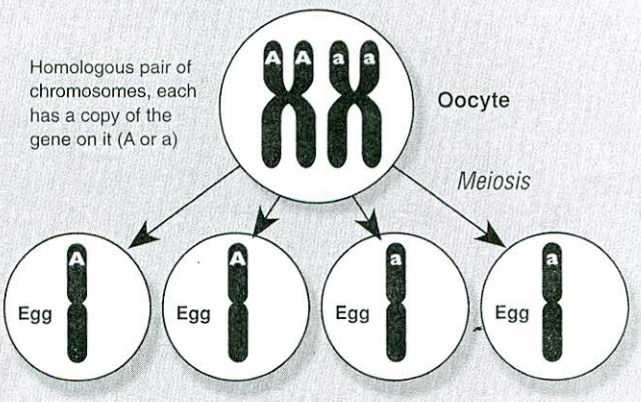
Mendel recognized that characters are determined by discrete units that are inherited intact down through the generations. This model explained many observations that could not be explained by the idea of blending inheritance that was universally accepted prior to this. The diagram on the right illustrates this principle, showing that the trait for flower color appears to take on the appearance of only one parent plant in the first generation, but reappears in later generations.



Law of Segregation

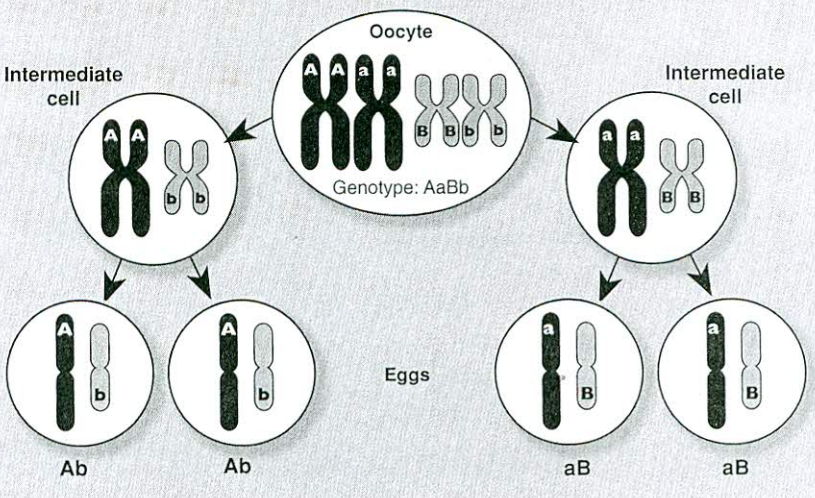
The diagram on the right illustrates how, during meiosis, the two members of any pair of alleles segregate unchanged by passing into different gametes. These gametes are eggs (ova) and sperm cells. The allele in the gamete will be passed on to the offspring.

NOTE: This diagram has been simplified, omitting the stage where the second chromatid is produced for each chromosome.



Law of Independent Assortment

The diagram on the right illustrates how genes are carried on chromosomes. There are two genes shown (A and B) that code for different traits. Each of these genes is represented twice, one copy (allele) on each of two homologous chromosomes. The genes A and B are located on different chromosomes and, because of this, they will be inherited independently of each other, i.e. the gametes may contain any combination of the parental alleles.



1. Briefly state what **property of genetic inheritance** allows parent pea plants that differ in flower color to give rise to flowers of a single color in the first generation, with both parental flower colors reappearing in the following generation:

_____

2. The oocyte is the egg producing cell in the ovary of an animal. In the diagram illustrating the **law of segregation** above:

   (a) State the genotype for the oocyte (adult organism): _____

   (b) State the genotype of each of the **four** gametes: _____

   (c) State how many different kinds of gamete can be produced by this oocyte: _____

3. The diagram illustrating the **law of independent assortment** (above) shows only one possible result of the random sorting of the chromosomes to produce: Ab and aB in the gametes.
   (a) List another possible combination of genes (on the chromosomes) ending up in gametes from the same oocyte:

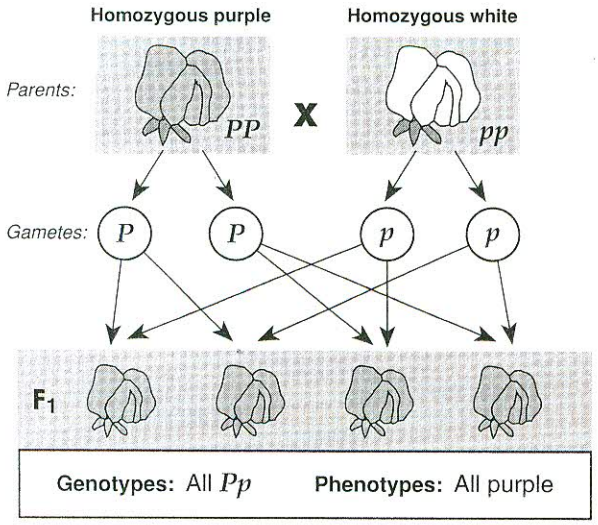
_____

(b) State how many different gene combinations are possible for the oocyte: _____

Basic Genetic Crosses

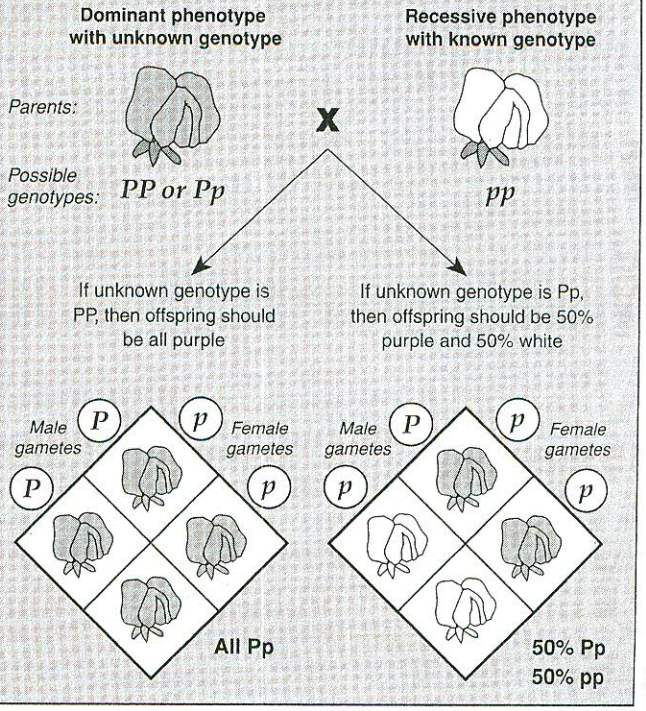
For revision purposes, examine the diagrams below on monohybrid crosses and complete the exercise for dihybrid (two gene) inheritance. A **test cross** is also provided to show how the genotype of a dominant phenotype can be determined. A test cross will yield one of two different results, depending on the genotype of the dominant individual. A **back cross** (not shown) refers to any cross between an offspring and one of its parents (or an individual genetically identical to one of its parents).

Monohybrid Cross



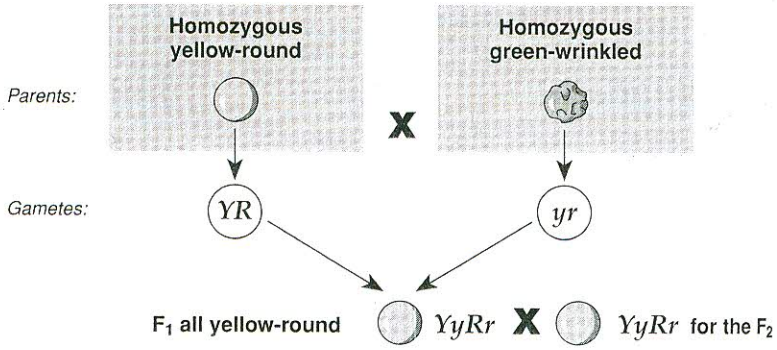
The F_1 offspring of a cross between two **true breeding** parent plants are all purple (Pp). A cross between these offspring (Pp x Pp) would yield a 3:1 ratio in the F_2 of purple (PP, Pp, Pp) to white (pp).

Test Cross



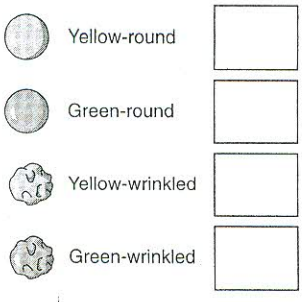
Dihybrid Cross

In pea seeds, yellow color (Y) is dominant to green (y) and round shape (R) is dominant to wrinkled (r). Each **true breeding** parental plant has matching alleles for each of these characters ($YYRR$ or $yyrr$). F_1 offspring will all have the same genotype and phenotype (yellow-round: $YyRr$).

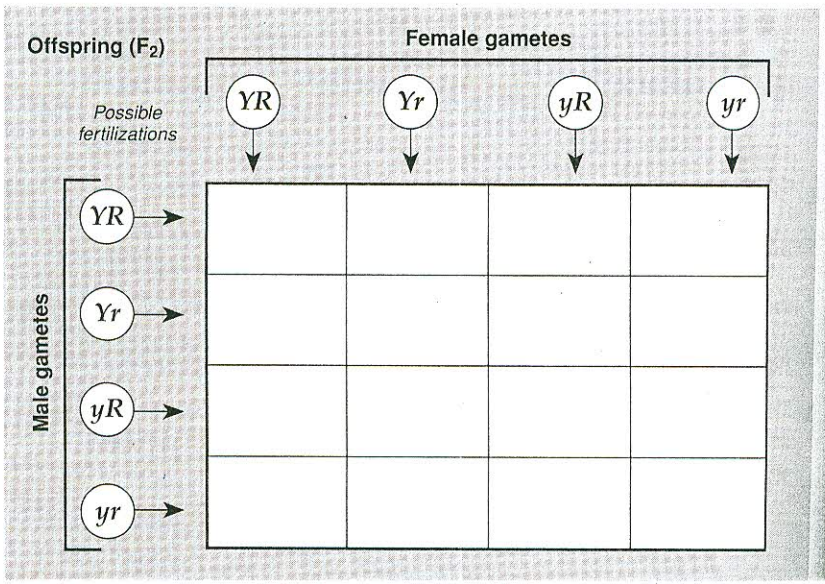


1. Fill in the Punnett square (below right) to show the genotypes of the F_2 generation.

2. In the boxes below, use fractions to indicate the numbers of each phenotype produced from this cross.



3. Express these numbers as a ratio:

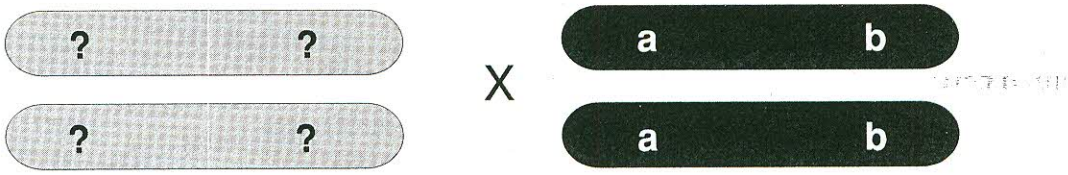


The Test Cross

It is not always possible to determine an organism's genotype by its appearance because the expression of genes is complicated by patterns of dominance and by gene interactions. The **test cross** was developed by Gregor Mendel as a way to establish the genotype of an organism with the dominant phenotype for a particular trait. The principle of the test cross is simple. The individual with the unknown genotype is bred with a homozygous recessive individual for the trait(s) of interest. The homozygous recessive can produce only one type of allele (recessive), so the phenotypes of the resulting offspring will reveal the genotype of the unknown parent. For example, if the unknown individual is homozygous for the trait, all of the offspring will display the dominant phenotype. However, if the offspring display both dominant and recessive phenotypes, then the unknown must be heterozygous for that trait. The test cross can be used to determine the genotype of single genes or multiple genes.

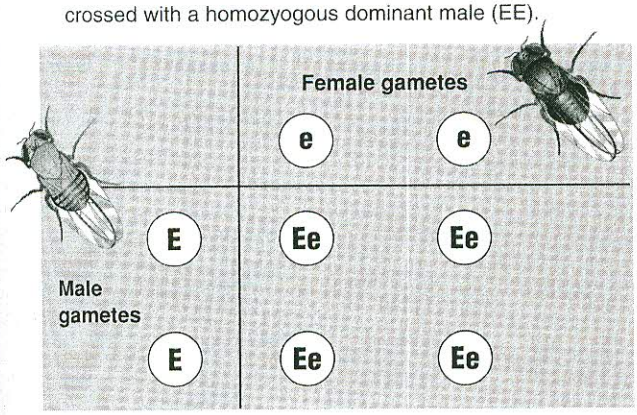
Parent 1
Unknown genotype
(but with dominant traits)

Parent 2
Homozygous recessive genotype
(no dominant traits)



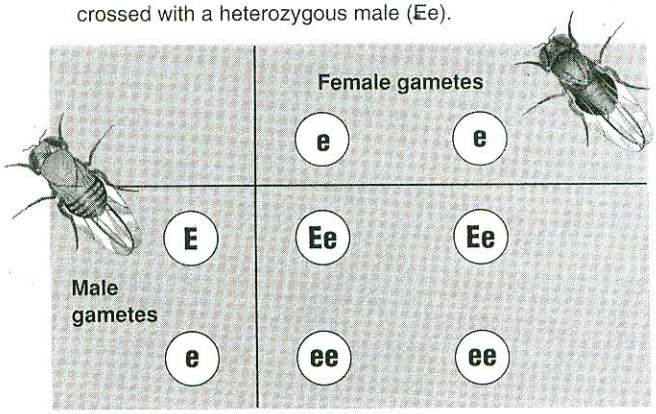
The common fruit fly (*Drosophila melanogaster*) is often used to illustrate basic principles of inheritance because it has several genetic markers whose phenotypes are easily identified . Once such phenotype is body color. Wild type (normal) *Drosophila* have yellow-brown bodies. The allele for yellow-brown body color (E) is dominant. The allele for an ebony colored body (e) is recessive. The test crosses below show the possible outcomes for an individual with homozygous and heterozygous alleles for ebony body color.

A. A homozygous recessive female (ee) with an ebony body is crossed with a homozyogous dominant male (EE).



Cross A:
(a) Genotype frequency: 100% Ee
(b) Phenotype frequency: 100% yellow-brown

B. A homozygous recessive female (ee) with an ebony body is crossed with a heterozygous male (Ee).



Cross B:
(a) Genotype frequency: 50% Ee, 50% ee
(b) Phenotype frequency: 50% yellow-brown, 50% ebony

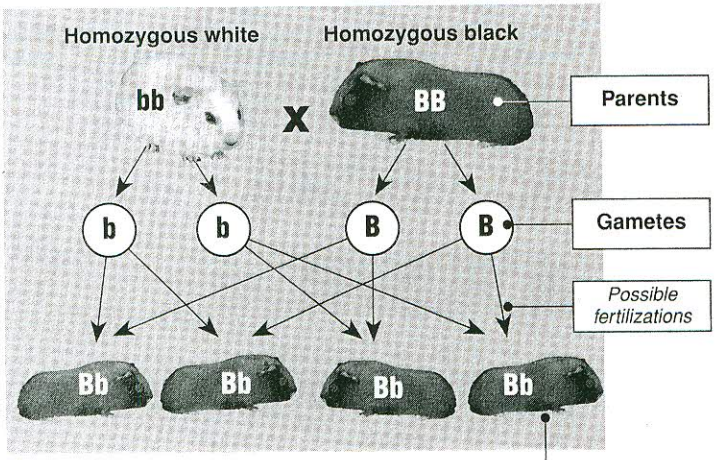
1. In *Drosophila*, the allele for brown eyes (b) is recessive, while the red eye allele (B) is dominant. Explain how you would set up a **two gene test cross** to determine the genotype of a male who has a normal body color and red eyes:

2. List all of the **possible genotypes** for the male *Drosophila*:

3. 50% of the resulting progeny are yellow-brown bodies with red eyes, and 50% have ebony bodies with red eyes.
   (a) State the genotype of the male *Drosophila*:
   (b) Explain how you came to this conclusion:

Monohybrid Cross

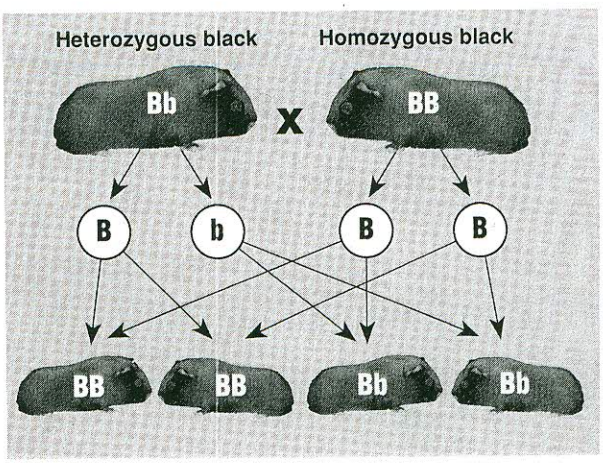
The study of **single-gene inheritance** is achieved by performing **monohybrid crosses**. The six basic types of matings possible among the three genotypes can be observed by studying a pair of alleles that govern coat color in the guinea pig. A dominant allele: given the symbol **B** produces **black** hair, and its recessive allele: **b**, produces white. Each of the parents can produce two types of gamete by the process of **meiosis** (in reality there are four, but you get identical pairs). Determine the **genotype** and **phenotype frequencies** for the crosses below (enter the frequencies in the spaces provided). For crosses 3 to 6, you must also determine gametes produced by each parent (write these in the circles), and offspring (F_1) genotypes and phenotypes (write in the genotype inside the offspring and state if black or white).



Cross 1:

(a) Genotype frequency: 100% Bb

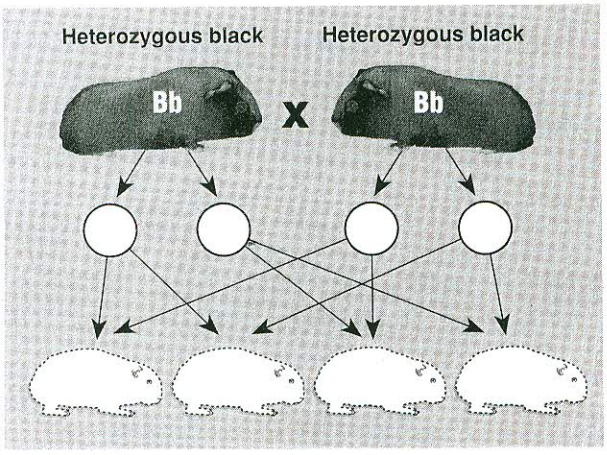
(b) Phenotype frequency: 100% black



Cross 2:

(a) Genotype frequency: _____

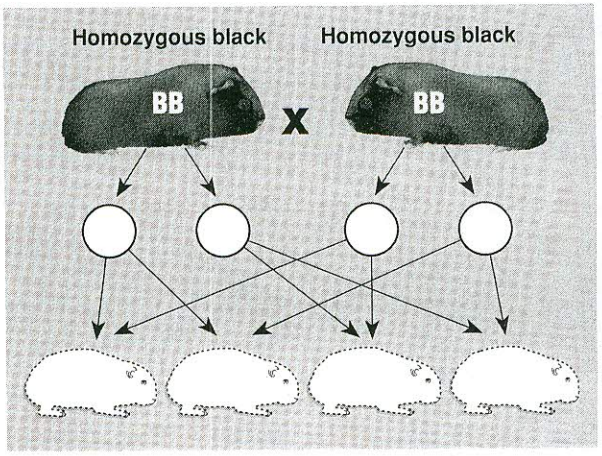
(b) Phenotype frequency: _____



Cross 3:

(a) Genotype frequency: _____

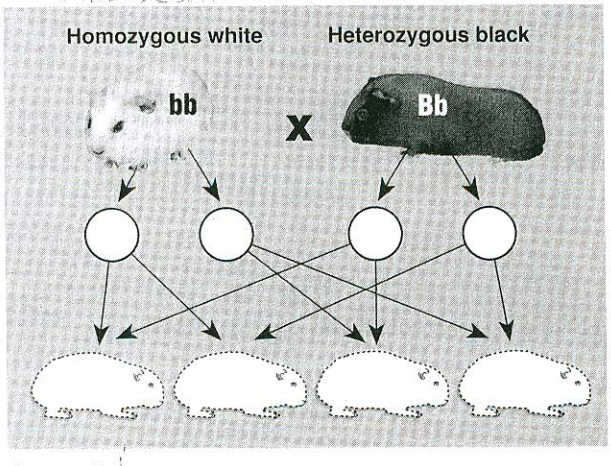
(b) Phenotype frequency: _____



Cross 4:

(a) Genotype frequency: _____

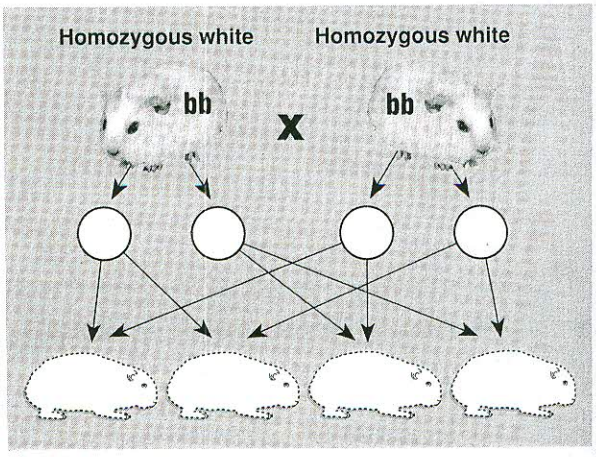
(b) Phenotype frequency: _____



Cross 5:

(a) Genotype frequency: _____

(b) Phenotype frequency: _____

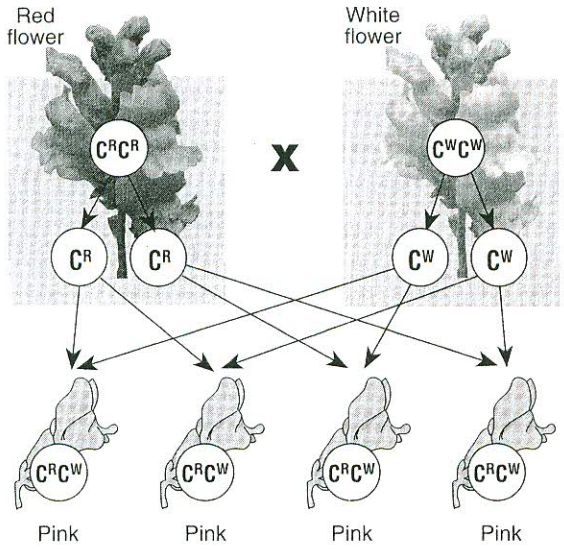


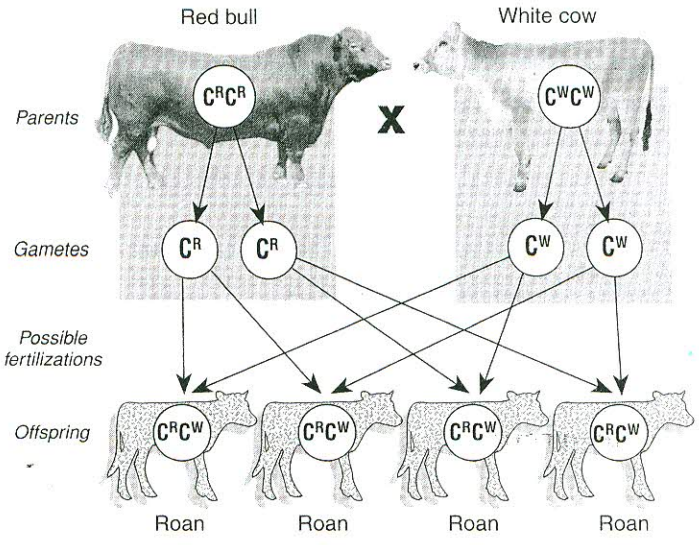
Cross 6:

(a) Genotype frequency: _____

(b) Phenotype frequency: _____

Dominance of Alleles





Incomplete Dominance

Incomplete dominance refers to the situation where the action of one allele does not completely mask the action of the other and neither allele has dominant control over the trait. The heterozygous offspring is **intermediate** in phenotype between the contrasting homozygous parental phenotypes. In crosses involving incomplete dominance the phenotype and genotype ratios are identical. Examples include snapdragons (*Antirrhinum*), where red and white-flowered parent plants are crossed to produce pink-flowered offspring. In this type of inheritance the phenotype of the offspring results from the partial influence of both alleles.

Codominance

Codominance refers to inheritance patterns when both alleles in a heterozygous organism contribute to the phenotype. Both alleles are **independently** and **equally expressed**. One example includes the human blood group AB which is the result of two alleles: A and B, both being equally expressed. Other examples include certain coat colors in horses and cattle. Reddish coat color is not completely dominant to white. Animals that have both alleles have coats that are **roan**-colored (coats with a mix of red and white hairs). The red hairs and white hairs are expressed equally and independently (not blended to produce pink).

1. In incomplete and codominance, two parents of differing phenotype produce offspring different from either parent. Explain the mechanism by which this occurs in:

   (a) Incomplete dominance: _____

   _____

   (b) Codominance: _____

   _____

2. For each situation below, explain how the heterozygous individuals differ in their phenotype from homozygous ones:

   (a) Incomplete dominance: _____

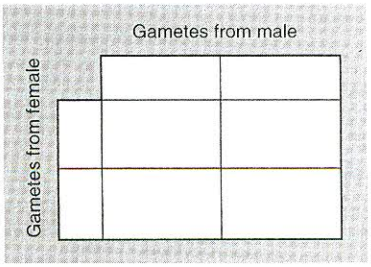
   (b) Codominance: _____

3. Describe the classical phenotypic ratio for a codominant gene resulting from the cross of two heterozygous parents (in the case of the cattle described above, this would be a cross between two roan cattle). Use the Punnett square (provided right) to help you:

   _____

   _____

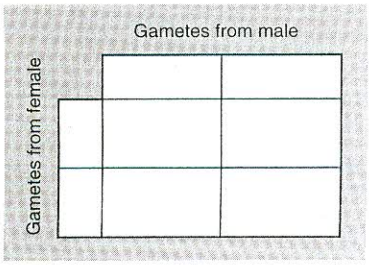
   _____



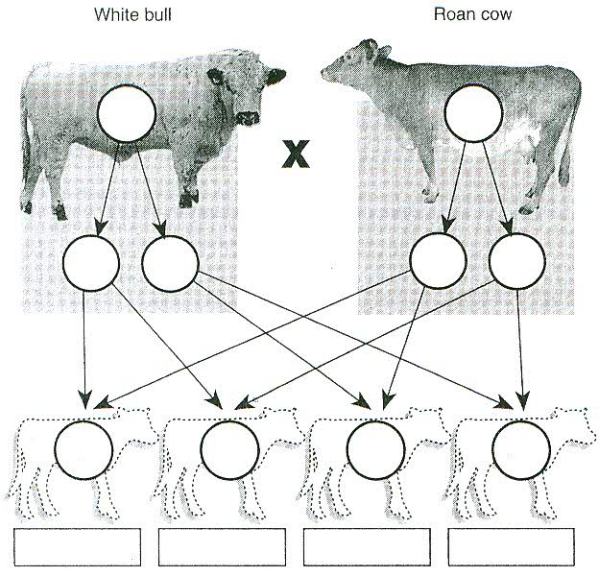
4. A plant breeder wanted to produce flowers for sale that were only pink or white (i.e. no red). Determine the phenotypes of the two parents necessary to produce these desired offspring. Use the Punnett square (provided right) to help you:

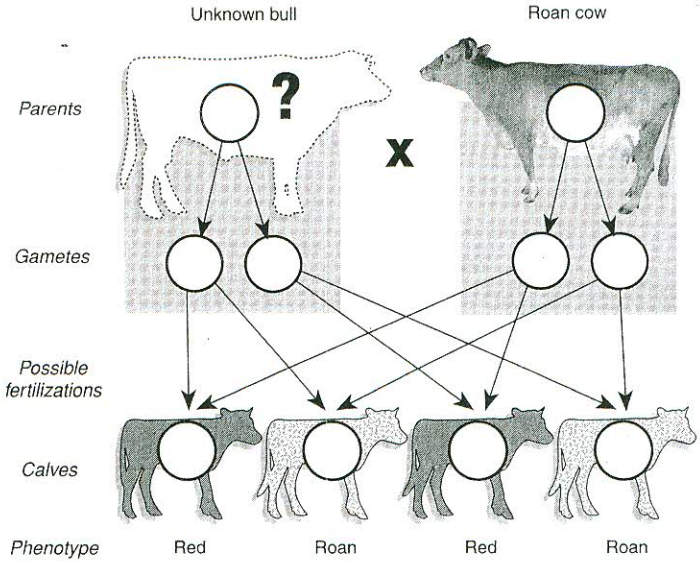
   _____

   _____



In the shorthorn cattle breed coat color is inherited. White shorthorn parents always produce calves with white coats. Red parents always produce red calves. But when a red parent mates with a white one the calves have a coat color that is different from either parent, called roan (a mixture of red hairs and white hairs). Look at the example on the previous page for guidance and determine the offspring for the following two crosses. In the cross on the left, you are given the phenotype of the parents. From this information, their genotypes can be determined, and therefore the gametes and genotypes and phenotypes of the calves. In the cross on the right, only one parent's phenotype is known. Work out the genotype of the cow and calves first, then trace back to the unknown bull via the gametes, to determine its genotype.





5. A white bull is mated with a roan cow (above, left).

   (a) Fill in the spaces on the diagram (above, left) to show the genotype and phenotype for parents and calves.

   (b) State the phenotype ratio for this cross: _____

   (c) Suggest how the farmer who owns these cattle could control the breeding so that the herd ultimately consisted of red colored cattle only:

   _____

   _____

   _____

6. A unknown bull is mated with a roan cow (above, right). A farmer has only roan shorthorn cows on his farm. He suspects that one of the bulls from his next door neighbors may have jumped the fence to mate with his cows earlier in the year. Half of the calves born were red and half were roan. One neighbor has a red bull, the other has a roan bull.

   (a) Fill in the spaces on the diagram (above, right) to show the genotype and phenotype for parents and calves.

   (b) State which of the neighbor's bulls must have mated with the cows: **red** or **roan** (*delete one*)

7. A plant breeder crossed two plants of the plant variety known as Japanese four o'clock. This plant is known to have its flower color controlled by a gene which possesses incomplete dominant alleles. Pollen from a pink flowered plant was placed on the stigma of a red flowered plant.

   (a) Fill in the spaces on the diagram on the right to show the genotype and phenotype for parents and offspring.

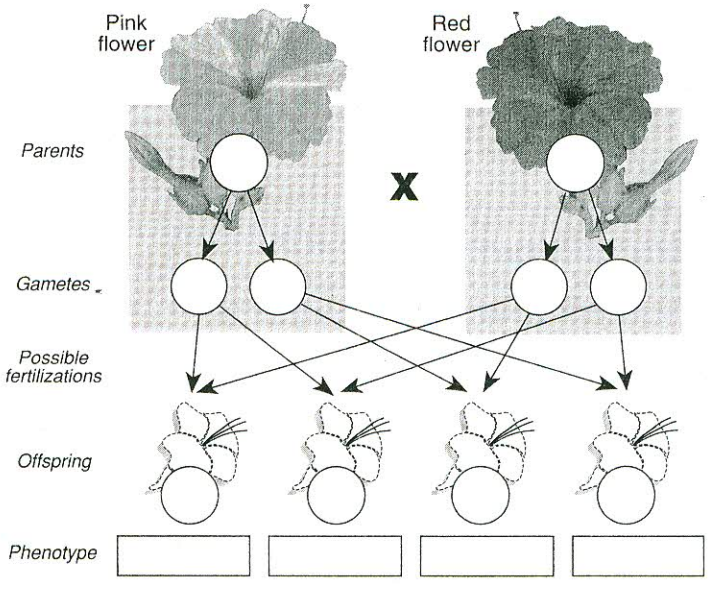
   (b) State the phenotype ratio:

   _____

   _____

   _____

   _____



Multiple Alleles in Blood Groups

The four common blood groups of the human 'ABO blood group system' are determined by three alleles: ***A***, ***B***, and ***O*** (also represented in some textbooks as: I^A, I^B, and i^O or just i). This is an example of a **multiple allele** system for a gene. The ABO antigens consist of sugars attached to the surface of red blood cells. The alleles code for enzymes (proteins) that join together these sugars. The allele ***O*** produces a non-functioning enzyme that is unable to make any changes to the basic antigen (sugar) molecule. The other two alleles ***(A, B)*** are **codominant** and are expressed equally. They each produce a different functional enzyme that adds a different, specific sugar to the basic sugar molecule. The blood group A and B antigens are able to react with antibodies present in the blood from other people and must be matched for transfusion.

Recessive allele: ***O*** produces a non-functioning protein
Dominant allele: ***A*** produces an enzyme which forms **A antigen**
Dominant allele: ***B*** produces an enzyme which forms **B antigen**

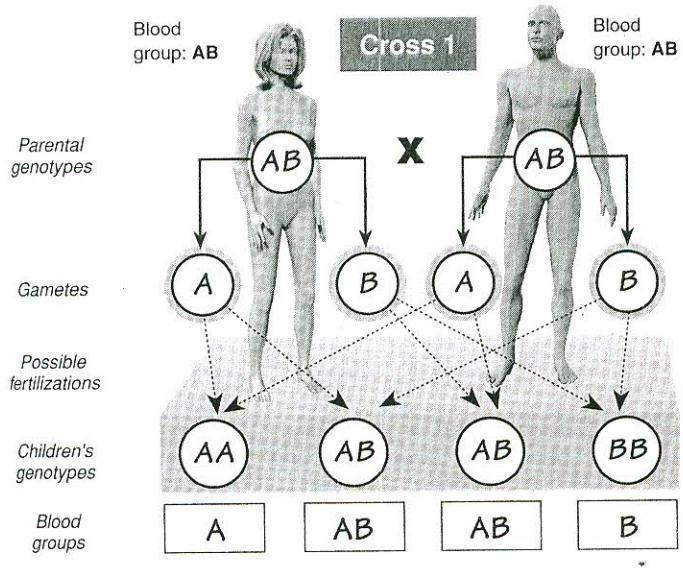
Blood group (phenotype)	Possible genotypes	Frequency*		
		White	Black	Native American
O	*OO*	45%	49%	79%
A	*AA AO*	40%	27%	16%
B		11%	20%	4%
AB		4%	4%	1%

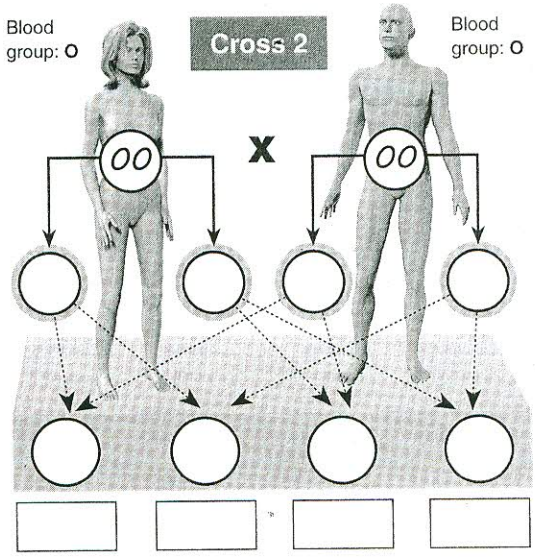
* Frequency is based on North American population
Source: www.kcom.edu/faculty/chamberlain/Website/MSTUART/Lect13.htm

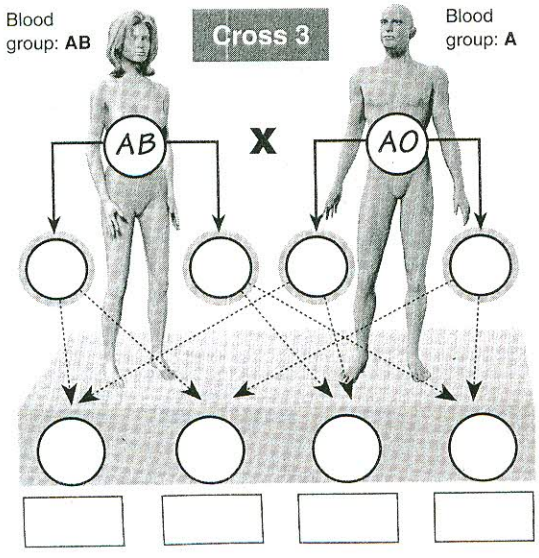
If a person has the ***AO*** allele combination then their blood group will be group **A**. The presence of the recessive allele has no effect on the blood group in the presence of a dominant allele. Another possible allele combination that can create the same blood group is ***AA***.

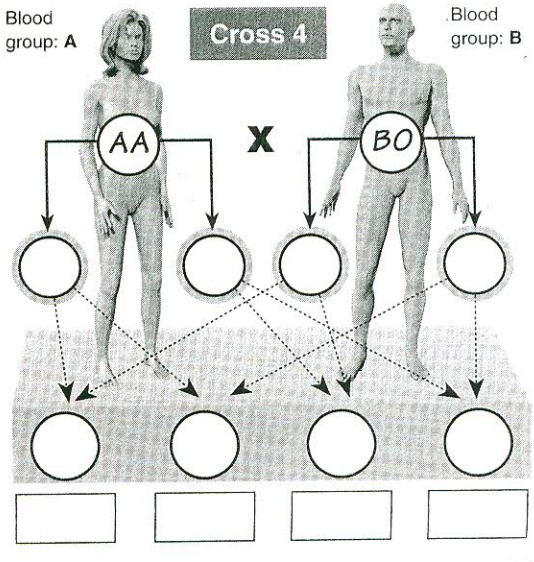
1. Use the information above to complete the table for the possible genotypes for blood group B and group AB.

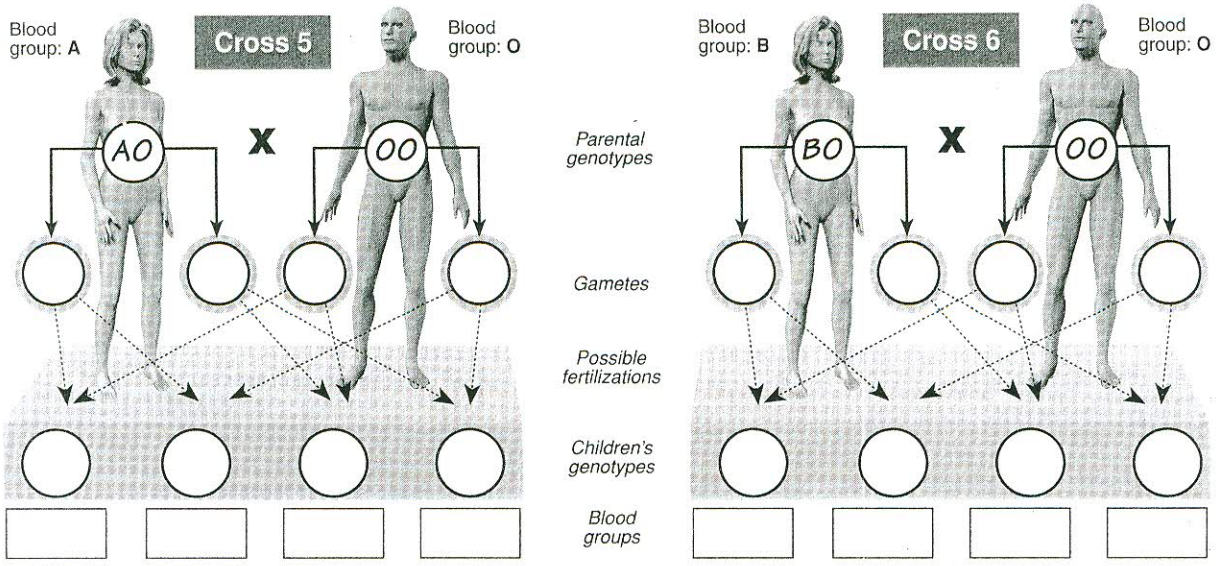
2. Below are six crosses possible between couples of various blood group types. The first example has been completed for you. Complete the genotype and phenotype for the other five crosses shown:



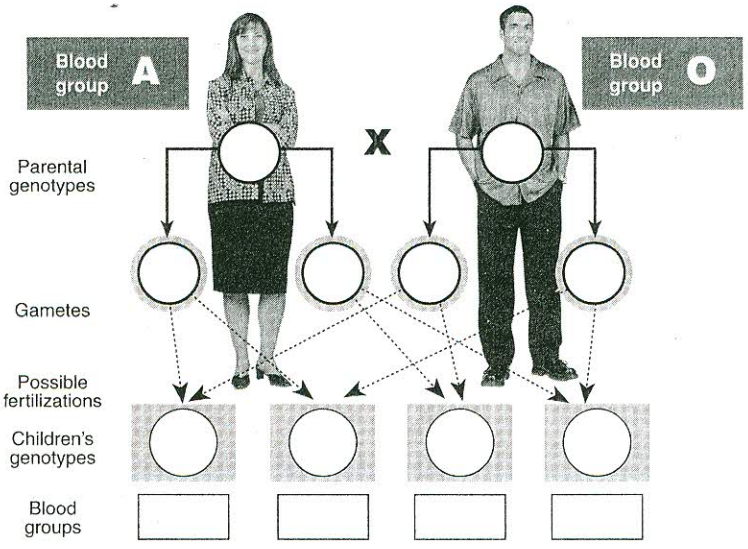








3. A wife is heterozygous for blood group **A** and the husband has blood group **O**.



(a) Give the genotypes of each parent (fill in spaces on the diagram on the right).

Determine the probability of:

(b) One child having blood group **O**:

_____

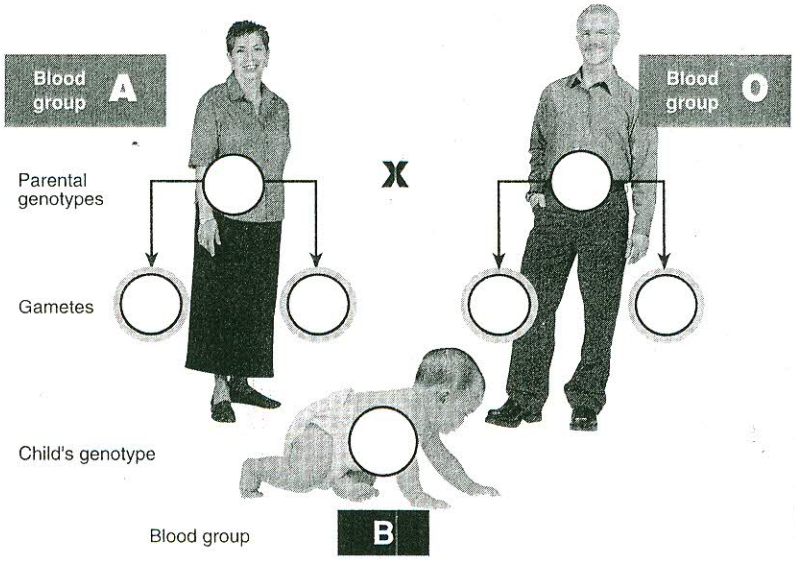
(c) One child having blood group **A**:

_____

(d) One child having blood group **AB**:

_____

4. In a court case involving a paternity dispute (i.e. who is the father of a child) a man claims that a male child (blood group **B**) born to a woman is his son and wants custody. The woman claims that he is not the father.



(a) If the man has a blood group **O** and the woman has a blood group **A**, could the child be his son? Use the diagram on the right to illustrate the genotypes of the three people involved.

(b) State with reasons whether the man can be correct in his claim:

_____

_____

_____

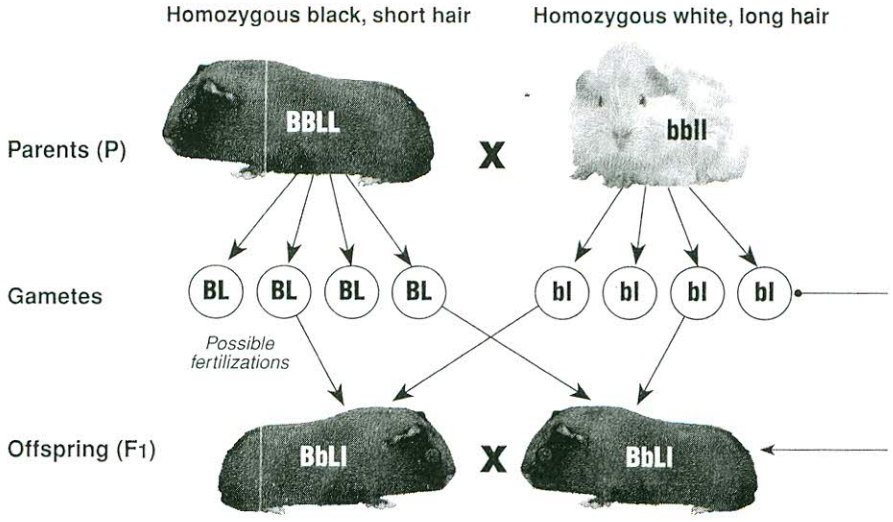
5. Give the blood groups which are possible for children of the following parents (remember that in some cases you don't know if the parent is homozygous or heterozygous).

(a) Mother is group **AB** and father is group **O**: _____

(b) Father is group **B** and mother is group **A**: _____

Dihybrid Cross

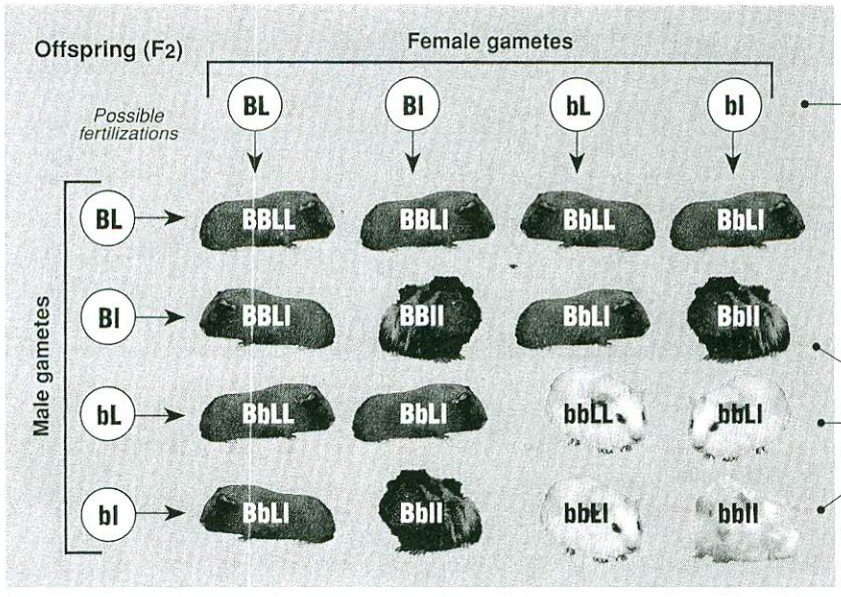
A cross (or mating) between two organisms where the inheritance patterns of **two genes** are studied is called a **dihybrid cross** (compared with the study of one gene in a monohybrid cross). There are a greater number of gamete types (four) produced when two genes are considered. Remember that the genes described are being carried by separate chromosomes and are sorted independently of each other during meiosis (that is why you get four kinds of gamete). The two genes below control two unrelated characteristics **hair color** and **coat length**. Black and short are dominant.



Parents: The notation **P**, is only used for a cross between **true breeding** (homozygous) parents.

Gametes: Only one type of gamete is produced from each parent (although they will produce four gametes from each oocyte or spermatocyte). This is because each parent is homozygous for both traits.

F1 offspring: There is only one **kind** of gamete from each parent, therefore only one kind of offspring produced in the first generation. The notation **F1** is only used to denote the heterozygous offspring of a cross between two true breeding parents.



F2 offspring: The F1 were mated with each other (**selfed**). Each individual from the F1 is able to produce four different kinds of gamete. Using a grid called a **Punnett square** (left), it is possible to determine the expected genotype and phenotype ratios in the F2 offspring. The notation **F2** is only used to denote the offspring produced by crossing F1 heterozygotes.

Each of the 16 animals shown here represents the possible zygotes formed by different combinations of gametes coming together at fertilization.

The offspring can be arranged in groups with similar phenotypes:

Genotype		Phenotype
1 BBLL 2 BbLL 2 BBLl 4 BbLl	A total of 9 offspring with one of 4 different genotypes can produce black, short hair	 9 black, short hair
1 BBll 2 Bbll	A total of 3 offspring with one of 2 different genotypes can produce black, long hair	 3 black, long hair
1 bbLL 2 bbLl	A total of 3 offspring with one of 2 different genotypes can produce white, short hair	 3 white, short hair
1 bbll	Only 1 offspring of a given genotype can produce white, long hair	 1 white, long hair

Cross N°. 1

The dihybrid cross on the right has been partly worked out for you. You must determine:

1. The genotype and phenotype for each animal (write your answers in its dotted outline).
2. Genotype **ratio** of the offspring:

_____

_____

_____

_____

_____

_____

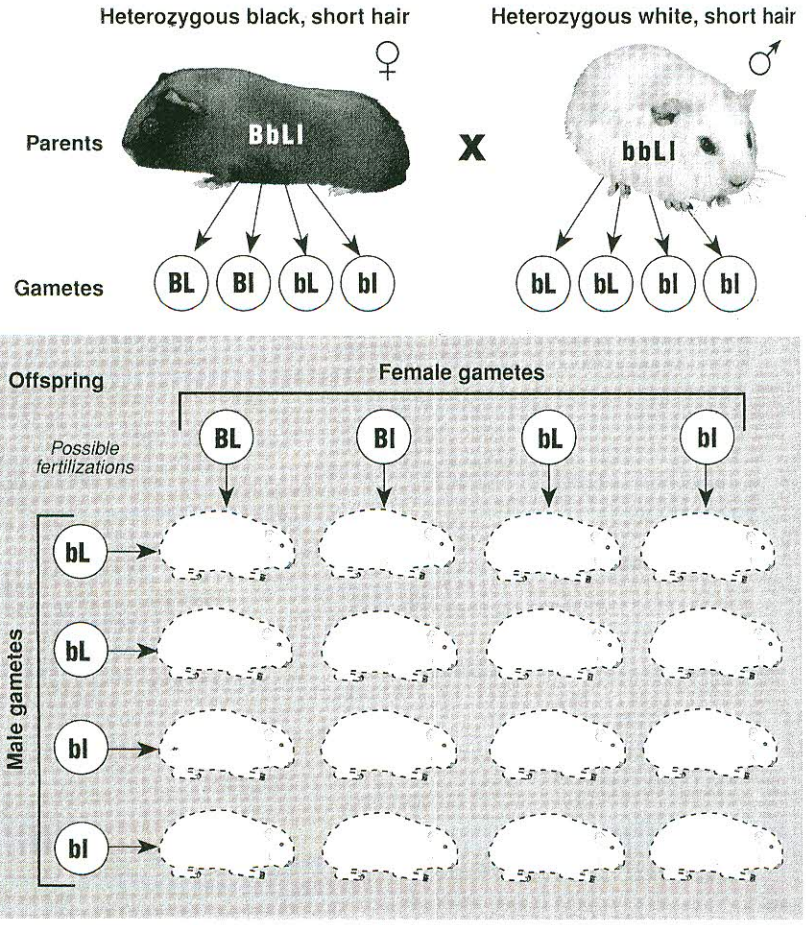
3. Phenotype **ratio** of the offspring:

_____

_____

_____

_____



Cross N°. 2

For the dihybrid cross on the right, determine:

1. Gametes produced by each parent (write these in the circles).
2. The genotype and phenotype for each animal (write your answers in its dotted outline).
3. Genotype **ratio** of the offspring:

_____

_____

_____

_____

4. Phenotype **ratio** of the offspring:

_____

_____

_____

_____