

MENDELIAN GENETICS

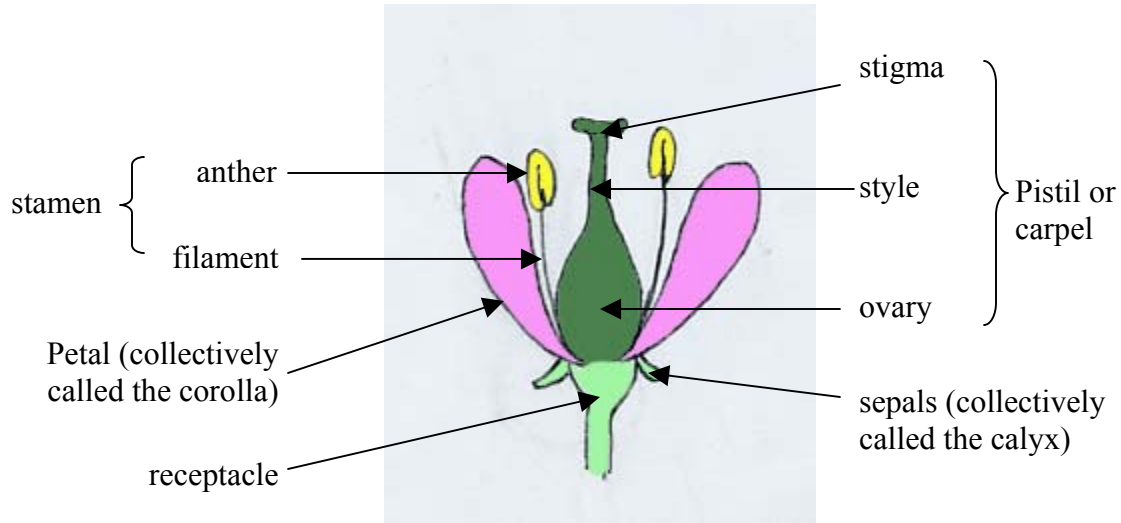
Johann Mendel (1822-1884) was born in Heinzendorf, in what is now a part of the Czech Republic. He was admitted to the Augustinian Monastery of St. Thomas in Brno in 1843 where he took the monastic name of Gregor. He began teaching in what is the equivalent of the local high school in 1849. In 1856, Mendel performed experiments with the hybridization of peas and reported his results in 1865 in an obscure Austrian scientific journal. He published in spite of the fact the famous Swiss botanist, Karl Wilhelm von Nägeli suggested Mendel do something more constructive with his time instead of performing hybridization experiments on peas. Later, three biologists, Hugo de Vries (Dutch botanist), Karal Erick Correns (German botanist) and Erick Tschermak von Seysenegg (Austrian botanist) independently came to the same conclusions of Mendel. In getting ready to publish their results, they each performed a literature search and each found Mendel's obscure work. Realizing Mendel had published the same results some 30 years earlier, they quickly gave credit to Mendel for his discoveries in hybridization of peas. Mendel's work established the foundations for the science we now know as genetics.

Some say Mendel was a brilliant scientist, others say divinely inspired by God. In any case, he was extremely fortunate. He began his studies with one of the few organisms on earth that would consistently give him repeatable results. Most organisms used in the study of genetics today, particularly fruit flies, are notorious for providing skewed data. Mendel picked the common garden pea.

Not only did he pick the one organism that gave his excellent data, he basically established the foundations of genetics without the benefit of knowledge of genes, chromosomes, and DNA. They had not been discovered at that time. Furthermore, he was, if anything, patient. He had to hand pollinate extremely small flowers, wait until they produced fruit, harvest the seeds, wait until spring to plant them, and then grow the seeds to maturity before he saw the results of his experiments – he had to wait a full year for his results!

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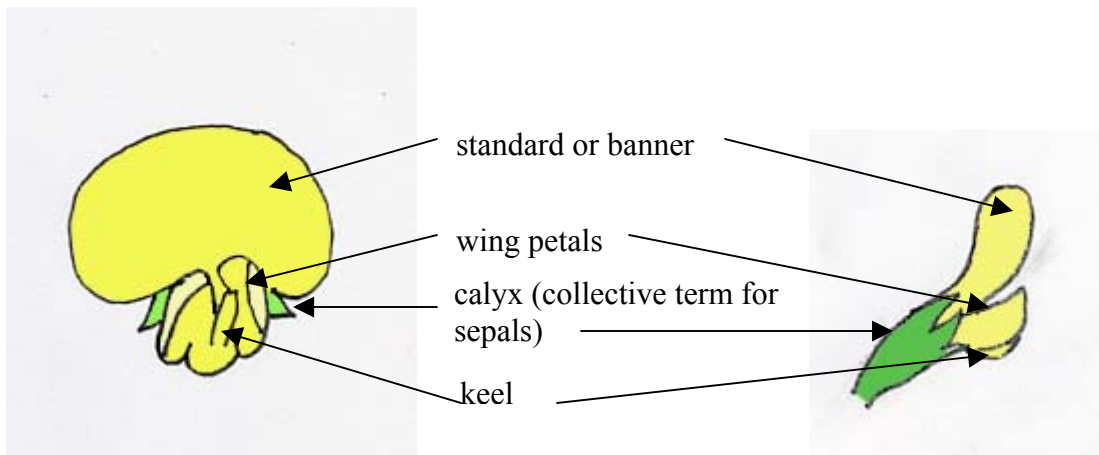
Peas belong to the genus and species *Pisum sativum*. The pea flower is especially tedious to work with and Mendel had to pollinate each flower by hand in order to study sexual reproduction in peas.



The male parts of the flower are called stamens. Stamens are composed of a filament that holds the anther – the pollen chamber. Pollen contains the sperm cells of the flower. The female part of the plant is called the pistil. The pistil has three parts: stigma, style and ovary. The stigma is the “sticky” tip of the flower to which pollen adheres. The style is a long, neck-like structure that leads away from the stigma and down to the ovary. The ovary contains the seeds or ovules. Eggs are found within the ovules and once fertilization occurs, an embryo forms in the seed.

In addition to the male and female reproductive parts, there are the more “showy” parts of the flower: sepals, petals, and receptacle. The receptacle is the swollen base of the flower to which most other parts are attached. Sepals are the leaf-like structures at the base of the flower and the petals are the structures most often recognized as the “flower”. All of the sepals are collectively called the calyx, and the petals are collectively called the corolla.

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Pea flowers are modifications of the basic structure of flowers learned in high school. The corolla (collective term for petals) is modified into a banner, a pair of “wing” petals, and a “keel” petal (similar in shape to a boat’s keel).

Pea plants often self-pollinate. The stamens are close enough to the stigma in the same flower they often reproduce with themselves. Mendel needed to cross pollinate the flowers under controlled conditions. In order to do this, he had to open each flower and remove the stamens of each flower to prevent self pollination. He then selectively tapped the stamens (and distributed the pollen) to each pistil individually. What a task! Pea flowers are very small.

The Results

What did Mendel discover? Mendel knew pea flowers came in two colors: purple and white. He crossed a pure strain of purple flower pea plants with a pure strain of white flower pea plants. By pure strain, we mean the flowers breed true. No matter how many times you plant purple flowers, you always get purple flowers.

When Mendel made the cross of the pure strain of purple with a pure strain of white, all the offspring were purple. (The pure strains are called the parents and the offspring are called the first filial generation or F1.) Where did the white flowers go? He didn’t stop there. He then crossed the F1 generation with itself. When he planted the next spring, he noticed the white flowers reappeared but in smaller numbers. Specifically, there were 3 purple flowers for every white flower.

Mendel made the observation that purple flower color must be dominant over white flower color in pea plants. (If purple is dominant, white is said to be recessive). Remember, Mendel did all of this without the benefit of knowledge of genes, chromosomes or DNA!

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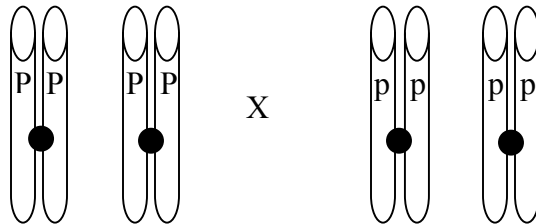
What's Going On?

We now have the benefit of knowledge of genes, chromosomes and DNA, therefore, genetics should be a snap. Let's look at the pure strain of purple and pure strain of white. Remember, on a chromosome, on a point on the chromosome (a locus) you may find a unit of heredity called a gene. Let's say that gene is for purple flowers (P). That chromosome has a homolog. At the same locus is an allele of that gene. For the pure strain of purple flowers, the allele is also "P" for purple. The gene and allele are identical – for purple. This condition where the chromosome and homolog are identical is referred to as homozygous dominant.

For the pure strain of white, the chromosome has a gene for white. White is denoted in dominant-recessive traits, not as "w" but as little "p" to show that capital P is dominant over little "p". The homolog has an allele for "p" also. This condition where the chromosome and the homolog both have identical genes and alleles for white is said to be homozygous recessive.

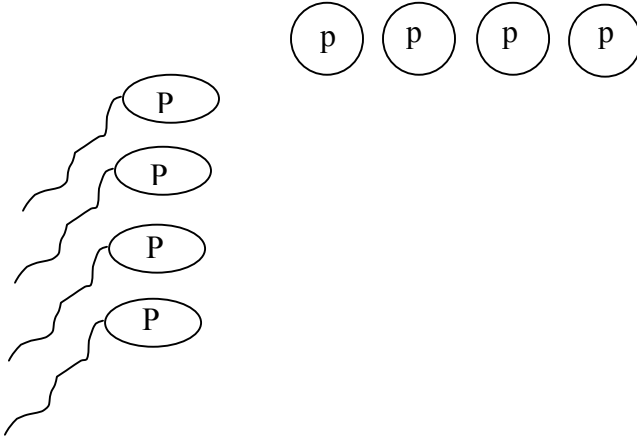
There is one other possibility. The chromosome could have a gene for "P" and the homolog could have an allele for "p". The flower color would be purple because purple is dominant over white, but the make-up of the chromosomes is not homozygous. In this case, it would be heterozygous dominant.

During meiosis of interphase, the chromosomes make exact copies of themselves. In the case of the pure strain of purple, the chromosome and its copy both have genes for "P". The homolog and its copy also have alleles for "P". For the pure strain of white, the chromosome and its copy have genes for "p" and the homolog and its copy have alleles for "p". Here's the cross:



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For the chromosomes and homologs on the left, meiosis results in the production of 4 sperm cells. Each sperm cell gets one of the chromosomes or homologs, or their copies. For the chromosomes and homologs on the right, meiosis results in the production of one egg and 3 polar bodies. However, there is no way of telling which chromosome or homolog or copies gets the egg and which gets the polar bodies, so you must assume 4 eggs are produced. The result is as shown below.



Notice that it does not matter which sperm penetrates which egg, the result will always be Pp for the F1 generation. Remember, when Mendel crossed a pure strain of purple with a pure strain of white, he observed *all* purple offspring: Pp.

The more interesting cross is the F1 cross with itself. In other words Pp x Pp. Don't worry about drawing the chromosomes and homologs anymore. Simply remember during interphase, the chromosomes make exact copies. The result will be the following: PPpp x PPpp.

These must segregate out into sex cells (gametes). For the sperm, you get 4 sperm as follows:

P
P
p
p

Remember to assume 4 eggs are produced. The eggs would be:

P
P
p
p

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It's best to eliminate any duplicate sperm and any duplicate eggs. That means the sperm with differences are P and p. The same holds true for the eggs. It's easy to predict the F₂ generation (second filial) by using a device called a Punnett square. Place the *different* sperm on the left and the *different* eggs on the top as shown below.

	P	p
P	PP	Pp
p	Pp	pp

Notice the offspring in the F₂ generation. Three are purple and 1 is white, exactly the results Mendel observed.

Phenotype

The observation of 3 purple to 1 white in the F₂ generation is called the phenotypes of the flowers: purple flowers and white flowers. The phenotypic ratio (what's observed of the trait) is 3:1, Purple: White.

Genotype

Note, however, there are two types of purple in the F₂. One purple is PP. Two of the three purples are Pp. Where phenotype is the trait observed or seen, the genotype is what is actually stipulated on individual chromosomes. In this case, the genotypic ratio is 1:2:1, PP:Pp:pp.

Monohybrid Cross

The crosses just discussed are called monohybrid crosses in the sense we looked at only one trait in pea plants – flower color.

In a monohybrid cross, heterozygous for the trait and dominant-recessive for the trait, the expected phenotypic ratio is always 3:1 and the expected genotypic ratio is always 1:2:1.

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Dihybrid Cross

Peas, obviously, have more than a single trait. When you look at two traits simultaneously, it's called a dihybrid cross. Let's keep the first trait: flower color. However, peas also grow in two heights: tall and short. Short plants are recessive to tall plants. Let's use the letter "T" for tall and "t" for short.

TT = tall
Tt = tall
tt = short

Let's cross the F1 generation (heterozygous dominant for both traits) with itself. The genotype of the parents would be: PpTt x PpTt.

The first thing you must do is to determine the number of different gametes for the sperm and eggs. *Remember! Each sperm must have one gene for flower color and one gene for height. The same is true for each egg.*

Question: What would be the result of crossing a pure strain of purple, tall plants with a pure strain of white, short plants?

Answer: All the offspring (F1) would be Purple and Tall with the genotype PpTt.

What are the possible *different* gametes from a parent with the genotype of PpTt?

PT
Pt
pT
pt

The Punnett square would look like this:

	PT	Pt	pT	pt
PT				
Pt				
pT				
pt				

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Next, fill in the Punnett Square.

	PT	Pt	pT	pt
PT	PPTT	PPTt	PpTT	PpTt
Pt	PPTt	PPtt	PpTt	Pppt
pT	PpTT	PpTt	ppTT	ppTt
pt	PpTt	Pppt	ppTt	pptt

Check the phenotypes:

Purple, Tall = 9

Purple, Short = 3

White, Tall = 3

White, Short = 1

The phenotypic ratio is 9:3:3:1.

Genotype

The genotypic ratio is determined by listing the various different combinations. You can do that, but it works out to be 1:2:1:2:4:2:1:2:1.

In a dihybrid cross, heterozygous for both traits and dominant-recessive for both traits, the expected phenotypic ratio is always 9:3:3:1 and the expected genotypic ratio is always 1:2:1:2:4:2:1:2:1.

Trihybrid Cross

When you look at three traits simultaneously, it's called a trihybrid cross. Keep the first two traits: flower color and plant height. Let's add seed texture. Pea seeds either appear smooth and round or very wrinkled. Smooth is dominant over wrinkled, therefore:

SS = smooth
Ss = smooth
ss = wrinkled.

Cross two parents heterozygous for all three traits: PpTtSs x PpTtSs. Remember, sperm and egg must each contain a gene for flower color, plant height, and seed texture.

Question: What would be the genotype of the F1 generation if you crossed a pure strain of Purple, Tall, Smooth with a pure strain of White, Short, Wrinkled?
Answer: All the offspring would be Purple, Tall, Smooth with the genotype of PpTtSs.

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For sperm, the possibilities are

PTS
 PTs
 PtS
 Pts
 pTS
 pTs
 ptS
 pts

The same is true for the eggs (note we have 8 sperm and 8 eggs. We are trying to determine the probability of combinations and have to take all into consideration, not just 4 sperm and 4 eggs.).

	PTS	PTs	PtS	Pts	pTS	pTs	ptS	pts
PTS								
PTs								
PtS								
Pts								
pTS								
pTs								
ptS								
pts								

There's no real reason to work this out unless you simply like the exercise. Perhaps you can see why after a trihybrid cross, geneticists often use mathematical models to determine the genotype and phenotype of matings.

How Many Different Gametes?

Did you notice that in a monohybrid cross, heterozygous for the trait, there were two different gametes? For a dihybrid, heterozygous for both traits, the number of different gametes was 4. For a trihybrid, heterozygous for all three traits, the number of different gametes was 8. How many do you think there would be for a tetrahybrid cross, heterozygous for all 4 traits?

Eye Color

Enough of plants. What about human traits. Eye color in humans is a simple, dominant-recessive trait. Brown eyes are dominant over blue.

BB = brown
 Bb = brown
 bb = blue

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What would be the possible results of a mating between two heterozygous brown-eyed parents? You should not have to do a Punnett square. Remember, in a monohybrid cross, heterozygous for the trait, dominant-recessive for the trait, the expected phenotypic ratio is 3:1 and the expected genotypic ratio is 1:2:1. So what would the possibilities be for the offspring?

Eye color is not as simple as it sounds. Remember, there are shades of blue and shades of brown. The degree to which a trait is shown is called expressivity. The percentage of the trait in a population is called the penetrance of the trait.

Incomplete Dominance

Incomplete dominance is where no one trait exhibits dominance over the other. A classic example is morning glories. There are three variations of color in morning glories: red, pink, and white. The genotype of red morning glories is “rr”. Note the use of all lower case letters since red is not dominant over white, nor *vice versa*. The genotype of white morning glories is “ww”. Note the use of a totally different letter for the color. Pink morning glories are represented by the genotype “rw”.

What would be the result of a mating of two pink morning glories?

	r	w
r	rr	rw
w	rw	ww

Note the phenotypic ratio. One red, two pink, one white or 1:2:1. The genotypic ratio, in this case, is the same.

Test Cross or Back Cross

Sometimes, you don't know the genotype of the individual in question, but you do know the phenotype. For example, in peas, smooth seeds could be represented by SS or Ss. If you see a smooth seeded plant, how do you know the genotype. Often, you must do a back cross. If you cross the individual whose genotype you don't know with an individual recessive for the trait, you will know for sure if you get a wrinkled seed. The offspring must have one gene from the “father” and one gene from the “mother”.

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Multiple Alleles

When more than one phenotype is expressed by a genotype, it is called multiple alleles. An example of multiple alleles is blood type.

The story of blood types has its origins in the need to stopping bleeding and the replacement of lost blood. Karl Landsteiner, an Austrian physician identified three blood types: A, B, and C (later changed to O) in 1901. Colleagues of Landsteiner identified a fourth type, AB. In 1907, Dr. Ruben Ottenburg of Mount Sinai in New York performed the first successful transfusion of blood after cross matching blood types. Later, in 1940, Landsteiner and A.S. Wiener discovered Rh factor. Today, human blood can be typed to at least 15 different types.

There are four major types of blood: A, B, AB, and O. These are phenotypes. When a person has A blood, it means the glycocalyx on their red blood cells have the “A” antigen on their surface. A person of “B” blood type has the “B” antigen, AB people have both A and B antigens on their red blood cells, and an O person has not antigens for blood type.

Who Can Give Blood To Whom?

It’s critical to know someone’s blood type and Rh factor (more about Rh factor later). If you give someone the wrong blood type, the antibodies in that person’s blood react with the antigens of the donated red blood cells and cause them to clump together (agglutinate). This clumping or agglutination causes blood clots when the mass of cells tries to get through narrow diameter arteries and veins or capillary beds. It’s quite a painful death for the individual. Who can give blood to whom can best be defined by a chart showing donors and recipients. A negative sign indicates no clumping takes place – the transfusion is safe.

	Phenotype	Antigen	Antibody	RECIPIENT			
				A	B	AB	O
D O N O R	A	A	anti B	-	+	-	+
	B	B	anti A	+	-	-	+
	AB	A & B	none	+	+	-	+
	O	none	anti A & B	-	-	-	-

Note that AB people may receive blood from anyone. O people can only receive blood from O people, but they can donate blood to anyone. That’s why O people are considered universal donors and AB people are considered universal recipients. Nature has a way of leveling things out. AB is the rarest of the blood types and O is the most common.

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The concept of multiple alleles comes into play when you look at the phenotypes and which genotypes may express those phenotypes.

Phenotype	Genotypes
A	AA, AO
B	BB, BO
AB	AB
O	OO

There are two ways to have A blood type: AA and AO. Remember the definition of multiple alleles: more than one genotype may express a phenotype.

Paternity Cases

Before the advent of DNA testing, blood types were about the only way to solve paternity law suits. One a woman would sue a man for child support, she would claim he was the father of the child. Blood typing could only decide if he was not the father. For example, if the child is A phenotype and the mother is O phenotype, and the person she claims is the father is O, he can't possibly be. Can you explain why? On the other hand, if the supposed father is O and the mother is O and the child is O, it doesn't necessarily prove he is the father.

Rh Factor

In 1940, Landsteiner and Wiener discovered that when they injected serum from a rhesus monkey into a rabbit, there was a response from the rabbit against the serum. They determined there must be another factor at work other than A, B, AB, and O (remember, we now know of at least 15 factors). They named it Rh factor to honor the rhesus monkey, a valuable research animal. People are either Rh positive or Rh negative. Seventy-five percent of the population is Rh positive. It is a simple dominant-recessive trait. There are two ways to be Rh positive and one to be Rh negative.

$RhRh = Rh$ positive

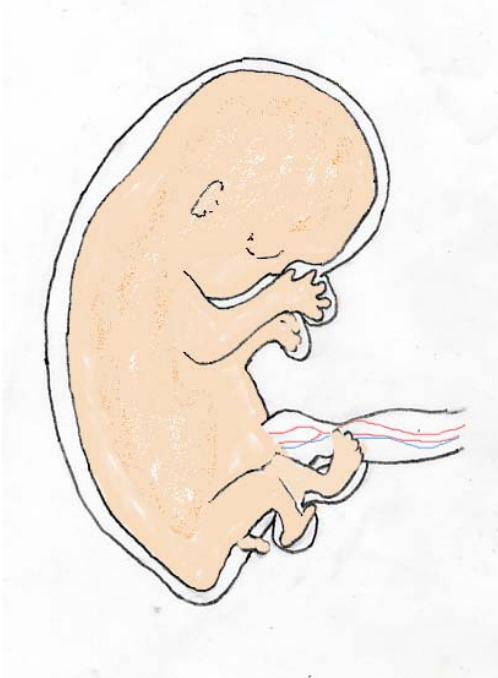
$Rhrh = Rh$ positive

$Rhrh = Rh$ negative

Don't be confused by the use of two letters to indicate Rh factor. You can just as easily use RR, Rr, and rr.

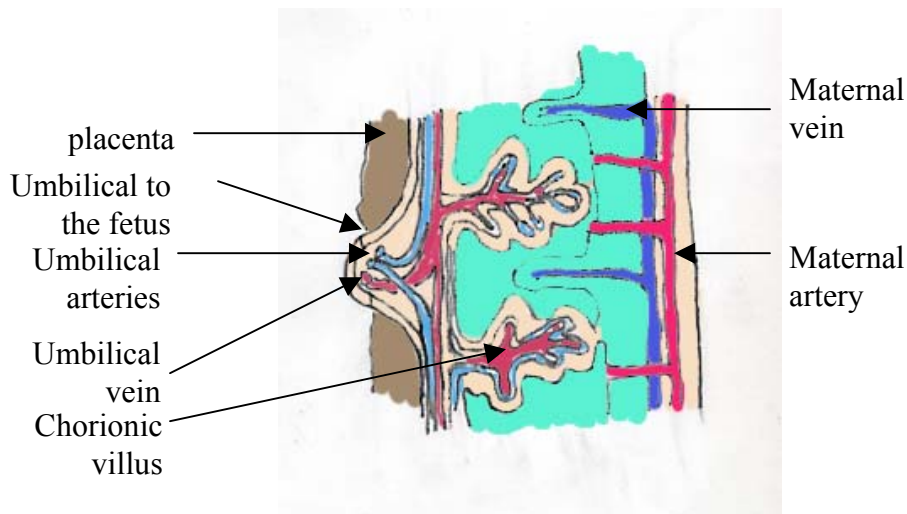
When you deal with both blood type and Rh factor, it is a dihybrid cross.

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Umbilical cord with fetal artery and fetal veins.

In the womb (uterus) of the mother, the fetus is attached to the uterine wall (mostly myometrium) by the placenta (afterbirth). The myometrium is somewhat finger-like. So is the part of the placenta that comes in contact with the myometrium. It is similar to putting the fingers of your left hand into the gaps of the fingers of your right hand. Like the above “finger arrangement” the placenta and the myometrium are separate. Blood of the fetus does not mix with the blood of the mother. Instead, nutrients and oxygen diffuse across the myometrium into the placenta and are carried to the fetus by the fetal artery to the fetus.. Likewise, the waste products of digestion –including urine – and carbon dioxide from respiration are carried by the umbilical vein to the placenta. These waste products diffuse across the placenta to the myometrium and the mother removes them through her system.



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You may have heard at one time that arteries carry oxygenated blood and veins carry deoxygenated blood. This is mostly true, however, there is one location that the reverse occurs. The pulmonary arteries in the lung carry deoxygenated blood and the pulmonary veins carry oxygenated blood. A more appropriate distinction between arteries and veins would be to say arteries carry blood away from the heart and veins carry blood to the heart. The fetus also reverses this typical process. The umbilical arteries carry nutrients and oxygen *to the fetus* and the umbilical vein carries the waste products *away from the fetus*.

Hopefully you can understand why it is dangerous for a pregnant female to smoke, drink or do drugs. In essence, not only is the female smoking, drinking and doing drugs, but so is the fetus.

It is well documented that a pregnant female who smokes will typically deliver a baby of lower birth weight. The lower the birth weight, the greater the potential for the death of the infant. A mother who drinks during pregnancy will lower the IQ of her child. This does not take into consideration fetal alcohol syndrome which often occurs when the mother is an alcoholic. Drug use in pregnant females is only now being studied over the long term and the first tests are actually rather terrifying.

Erythroblastosis fetalis

A serious condition that may arise with Rh factor is erythroblastosis fetalis (literally translated as destruction of erythrocytes of the fetus). For the condition to be possible, the mother must be Rh negative and the father of the child must be Rh positive. There is better than a 50% chance the fetus will be Rh positive. For the first pregnancy, it's not a problem. The problem develops when the mother delivers the child and the placenta rips away from the uterine wall. Some of the mother's blood and the baby's blood mixes. The mother then develops antibodies for Rh positive blood. The second pregnancy is the one in danger because if the father is Rh positive, there's a better than even chance the second child will also be Rh positive. She's already developed antibodies to Rh positive and the mother's antibodies attack the child's blood. It can be fatal. Fortunately, the mother can be desensitized between the first and second pregnancy and the second child will not be in danger.