Human Inheritance Patterns

Gregor Mendel's Principle of Segregation states that pairs of genes segregate during the formation of gametes (meiosis), so that each gamete has one of each gene pair but not both. The Principle of Independent Assortment states that each gene pair is distributed (assorts) independently of other gene pairs during the formation of gametes (meiosis). These principles laid the foundation for the study of inheritance that continues today.

When Sutton made the connection between Mendel's principles of inheritance and the behavior of homologous chromosomes during meiosis, the study of genetics and inheritance became the focus of much research. That research continues today, with emphasis on molecular genetics and DNA structure. With increasing information about DNA, we can better identify genetic abnormalities as well as learn more about gene interactions, gene expression and gene regulation.

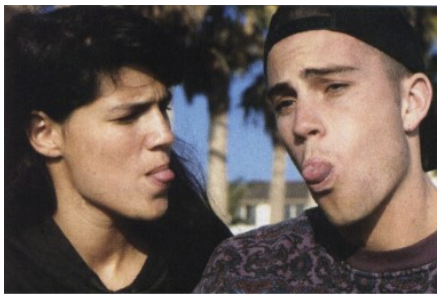
We can also study human genetic disorders by analyzing pedigrees – inheritance patterns of specific genes in families, such as the inheritance of hemophilia in the royal families of Europe in the 1800's. Pedigree analysis is useful in predicting the possibility and probability of inheriting a debilitating genetic disorder. Often, pedigree analysis uses karyotypes of individuals as a first step, along with more specific DNA analysis. A goal of the human genome project has been to facilitate the identification of gene disorders through DNA analysis. This laboratory is designed to look at some of the ways in which human inheritance can be studied.

Mendelian Inheritance in Humans

Many human traits follow Mendelian inheritance predictions. A number of human inheritance traits are listed below. Most appear to be "controlled" by a single gene that has two alternative alleles (specific forms of a gene). In each, one allele is dominant and the alternative is recessive. Your genotype is the specific combination of alleles you inherit from your biological parents. It can be homozygous dominant, heterozygous or homozygous recessive. Your phenotype is what is expressed, and for "visually observable" genes, what we see.

Tongue Curling

The ability to curl the tongue upward from the sides is a dominant trait. It is probably the result of several genes, though in genetics labs is usually treated as a one-gene trait.



Free Ear Lobe

Earlobes that hang "free" from the ear are dominant over attached earlobes that are attached directly to the side of the head.

Widow's Peak

A distinctive downward point in the hairline is known as the widow's peak. This is a dominant trait. If you have a straight hairline, you have recessive alleles for this trait.

Brown Pigmented Iris
Presence of pigment is usually dominant over absence of pigment in eye, hair and skin color. The actual pigmentation is a result of the interaction of several genes i.e., a polygenic trait. Brown eyes are the result of a brown pigment layer in the front of the iris. Blue eyes result when a blue layer in the back of the iris can show through in the absence of brown. Hazel or green eye color is the result of an unrelated gene that produces a yellow pigment. Hazel eyes have brown iris pigment while green eyes have a blue iris.

Hitchhiker's Thumb
The ability to bend the thumb backward (at least 45^{o}) is a caused by a dominant allele. The proper term for this is distal hyperextensibility. People with dominant alleles have more flexible ligaments and thus looser joints.

Shorter Big Toe
The dominant allele for toe length results in a big toe shorter than the second toe. If your big toe is longer than your second toe you have recessive alleles for this trait.

Mid-digital Hair
The presence of hair on fingers is a dominant trait. This is controlled by a number of different genes but we will treat it as a one-gene trait. Hair may not be present on all of your fingers, but if you have hair on even one finger, you show the dominant phenotype.

Interlocking Fingers
The way that you interlock your fingers is genetic. Most people will interlock their fingers so that the left thumb is on top of the right, the dominant allele. Those who are homozygous recessive place their right thumb over the left. (Try interlocking your fingers the opposite way from the way you naturally do this to compare.)

Freckles
The presence of freckles is dominant over absence of freckles.

Short Palmar Muscle
Examining the tendons that run over the inside of the wrist can tell you if you have the recessive long palmar muscle or the dominant, shorter palmar muscle. Those who are dominant have two tendons. Those recessive have three tendons. To determine this, you must clench your fist and flex your hand. Look and feel for three tendons. A middle tendon indicates the presence of the recessive long palmar muscle.

Rh Factor
The presence of the Rh coating on the surface of red blood cells is a simple Mendelian trait. Producing the Rh factor (Rh^{+}) is dominant. Rh^{-} individuals do not produce this coating and are recessive.

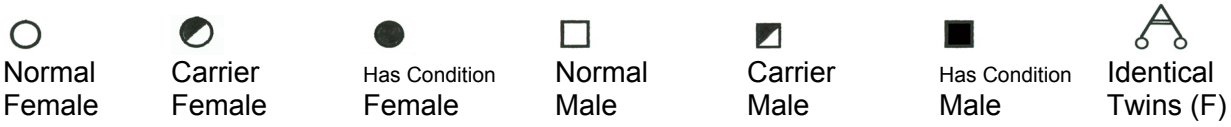
Blood Type
Although Blood Type is not a simple Mendelian trait, it is an inheritable characteristic. Within the human population there are three primary alleles for the gene that determines a surface coating for the red blood cells. There are two coating forms: "A" and "B". A recessive allele, "O" results in no coating. A and B are co-dominant since both will be expressed. An individual will have two alleles for this gene, which results in four possible phenotypes within the human population. As are many genetic traits, blood type is actually more complex than presented. There are variations in the alleles and interactions with other genes that affect this trait.

Exercise I Gene Detective

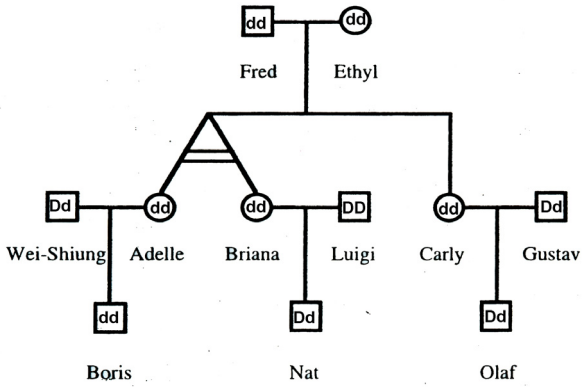
Prior to the use of DNA analysis to establish identity of individuals, pedigree analysis was used for known traits. Such tests have been used in cases of disputed paternity, and in rare cases, to determine if a child was "switched" at birth in the hospital. The case you will examine is a case of possible baby switching.

You will be examining three generations of the Menendez family. Fred and Ethyl have three daughters, including the identical twins, Adelle and Briana. Their sister is Carly. All three daughters are married and each has one son. Adelle and Brianna gave birth in the same hospital on the same day. A nurse bringing the infant sons to the mothers for feeding may have gotten confused because the mothers are identical twins, and given the "wrong" son to each mother. The nurse kept her fear that she may have switched the infants to herself until her deathbed when she "confessed" her dilemma. After hearing this, the families decided to undergo a set of six tests to see if they could find out which son was born to Brianna and which to Adelle. The pedigree results of each of the tests done are shown.

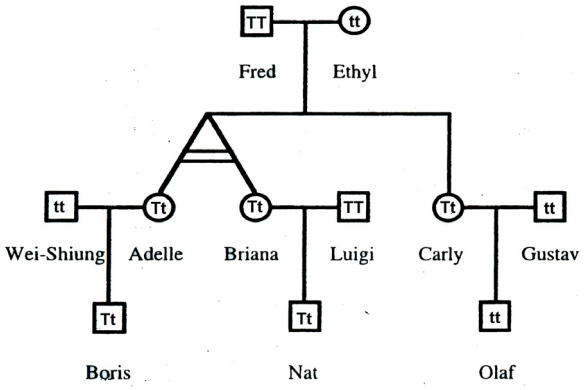
Symbols used:



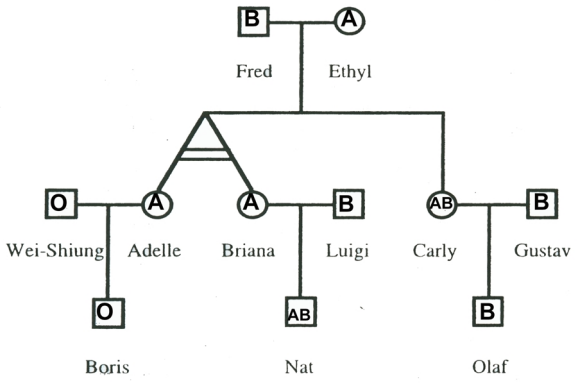
Mid-Digital Hair



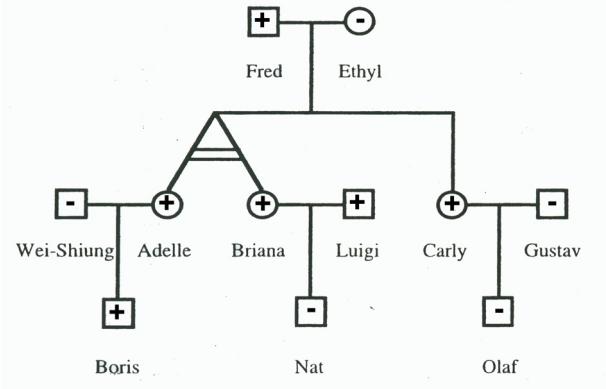
Tongue Curling



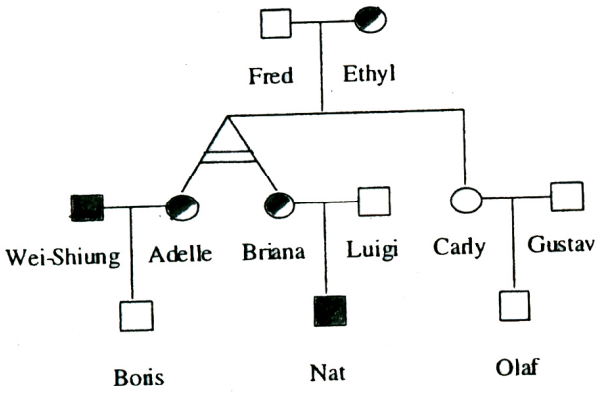
ABO Blood Type



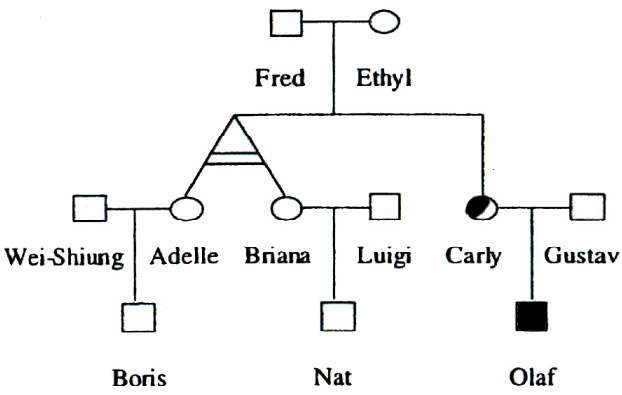
Rh Factor



Color Blindness



Hemophilia



Using the knowledge you have about human inheritance, determine from the pedigree tests who the birth parents of Boris and Nat are.

Pedigree Analysis

	Test Results (Genotypes)	Conclusion
Mid-Digital Hair		
Tongue Curling		
ABO Blood Type		
Rh Factor		
Color Blindness		
Hemophilia		

Explain which, If any, of the tests were sufficient to make a determination whether the boys were switched at birth or not?

Were the boys switched? _____

Which tests did not prove whether the boys were switched?

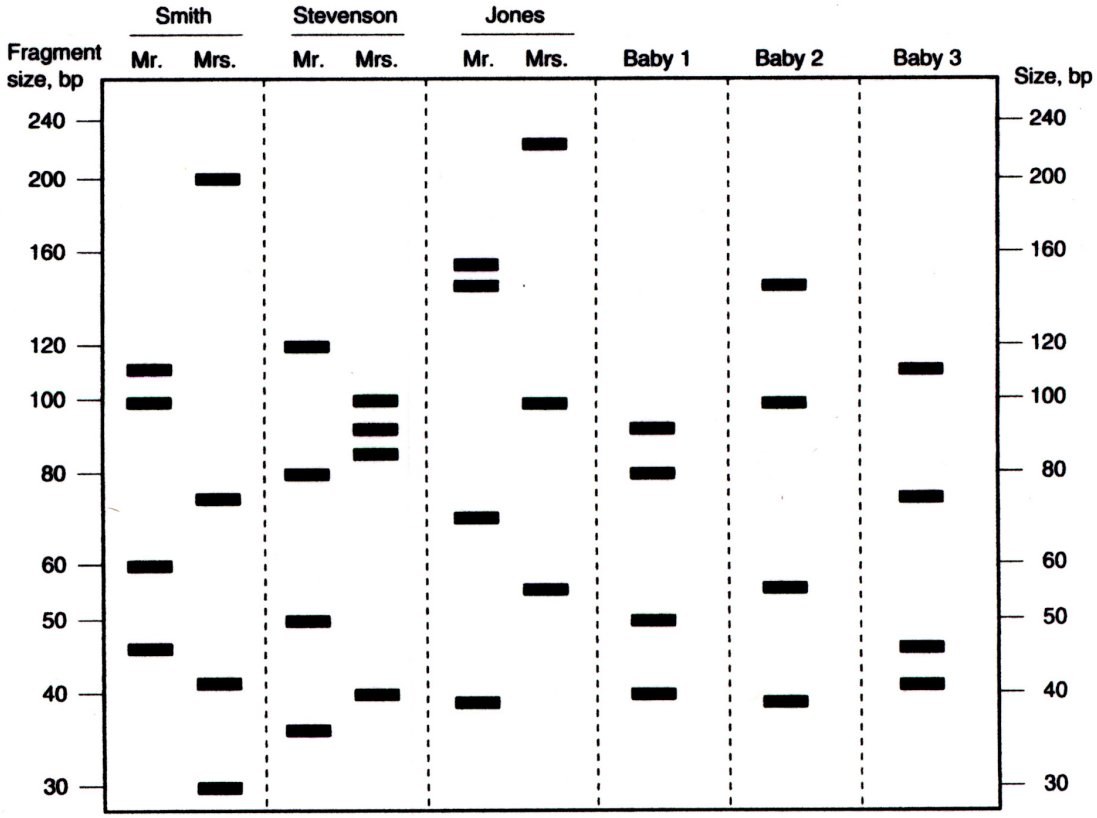
Note: This specific exercise has been adapted from the University of Washington Biology 201, Autumn 2000, Lab Manual.

Exercise II – Parental Identification Using DNA Analysis

DNA analysis is an accurate method of genetic identification. A DNA sample from a forensics scene can be compared to the DNA of a potential suspect, or used to provided positive identification in a number of situations. As discussed in the DNA technology section, restriction enzymes cut DNA molecules into unique fragments that can be separated by the technique of gel electrophoresis. The pattern of DNA fragments on the gel can be used to match samples with individuals. Just as you matched children to parents using pedigree analysis in Exercise I above, DNA analysis can match children to the correct biological parents. This exercise is designed to introduce you to DNA analysis for genetic identification.

Three baby boys were born at about 1:00 pm in the metropolitan hospital. Shortly afterward, the hospital's fire alarm sounded. The hospital staff rushed to safely evacuate all patients, including the three newborn babies. Unfortunately, the babies ID bracelets had not yet been attached, and with the confusion of the alarm, the staff was not positive which baby belonged to which parents. The hospital's head of pediatrics ordered DNA testing of the three babies and six parents.

The gel pattern result from the DNA analysis is shown below. Although not all parental bands will be found in each infant, each band of an infant's gel should match a band of either his mother's or his father's gel bands.



Identify which baby belongs to which set of parents.

For each baby, indicate which bands were inherited from the mother and which from the father by placing a F for father or M for mother above each band.

Note: This exercise has been adapted from Recombinant DNA and Biotechnology – A Guide for Teachers, by Kreuzer and Massey, 2001, Student Activity #20.

Exercise III Karyotype Analysis

A number of human genetic disorders, including non-disjunctions, can be determined by looking at a karyotype. Chromosomes at metaphase are particularly compact, and when stained with certain DNA-specific stains, such as the Giemsa stain, have distinctive bandings (G-bands) that facilitate identification of the chromosomes. White blood cells of adults or fetal cells obtained through amniocentesis or chorionic villus sampling can be used.

Procedure 1 Identifying Karyotypes

Four human karyotypes are shown below. After analyzing them, identify whether the karyotypes are male or female, and state whether they are normal, or if not nomal, what genetic abnormality is revealed in the individual karyotypes.