

➤ GENETICS: THE INHERITANCE OF TRAITS

- **genetics** = the science of heredity.
- **gene** = a sequence of DNA that codes for a protein.
- **Gregor Mendel** = an Augustinian monk who is credited with discovering the fundamental principles of genetics by breeding garden peas. Mendel lived and worked in an abbey in Brunn, Austria in the 1860's. Mendel's research was both experimentally and mathematically rigorous and his work allowed him to develop rules for how one organism passes its traits to its offspring.
- **cross-fertilization** = the fusion of sperm and egg derived from two different individuals. The mating of one individual organism to another.
- **hybrid** = the offspring of parents of two different species or of two different varieties of one species. The offspring of two parents that differ in one or more inherited traits or an individual that is heterozygous for one or more pair of genes. A mule is a hybrid animal whose parents were a male donkey and a female horse.
- **hybridization (or cross)** = the cross-fertilization of two different varieties of an organism or of two different species; also called a cross.
- **P generation** = the parent individuals from which offspring are derived in studies of inheritance; P stands for parental.
- **F1 generation** = the offspring of two parental (P generation) individuals; F1 stands for first filial. (filial means relating to offspring)
- **F2 generation** = the offspring of two first filial (F1 generation) individuals; F2 stands for second filial.
- **monohybrid cross** = an experimental mating of individuals in which the inheritance of a single characteristic (trait) is tracked.
- **dihybrid cross** = an experimental mating of individuals in which the inheritance of two characteristics (traits) is tracked.
- **allele** = an alternate form of a gene. For instance, say there is one gene that controls the coat color of an animal. One form of the gene might make the animal have a light coat color while another form of the gene might make the animal have a dark coat color.
- **homozygous** = having two identical alleles for a given gene. For instance, two copies of the dark coat color allele. This animal would be a homozygote. (the prefix homo means "the same" from Greek).
- **heterozygous** = having two different alleles for a given gene. For instance, having one copy of the light coat color allele and one copy of the dark coat color allele in an animal. This animal would be a heterozygote. (the prefix hetero means "different" from Greek).

- **phenotype** = the expressed traits of an organism.
- **genotype** = the genetic makeup of an organism that determines the phenotype of the organism. The genotype refers to which alleles an organism has for each of its genes.
- **dominant allele** = in a heterozygote (an organism that is heterozygous for a certain trait), the allele that is fully expressed in the phenotype. For instance, if an animal is heterozygous for animal coat color (it has both the light and dark alleles), if dark is the dominant allele, the animal will be dark.
- **recessive allele** = in a heterozygous individual, the allele that has no noticeable effect on the phenotype. That is, the recessive allele is usually not expressed if a dominant allele is present. For instance, if an animal is heterozygous for animal coat color (it has both the light and dark alleles), if light is the recessive allele, the animal will be darkly colored (because dark is the dominant allele).
- **Punnett square** = a diagram used in the study of inheritance to show the results of random fertilization between two parents. A Punnett square can help predict the probability of the phenotype of the offspring of two parents with known genotypes.
- **principle of segregation** = a general rule in inheritance that individuals have two alleles for each gene (one allele from each parent), and that when gametes (sperm or eggs) form by meiosis, the two alleles separate, and each resulting gamete ends up with only one allele of each gene: also known as Mendel's first law of inheritance.
- **principle of independent assortment** = a general rule in inheritance that when gametes (sperm or eggs) form during meiosis, each pair of alleles for a particular trait segregates independently of other traits; also known as Mendel's second law of inheritance.
- **incomplete dominance** = a type of inheritance in which F1 hybrids have an appearance that is intermediate between the phenotypes of the parental varieties. For instance, if one parent flower is red and the other parent is white, some of the offspring of a cross between these two parents could be pink (an intermediate color between red and white; both red and white are partially expressed).
- **codominance** = the expression of two different alleles of a gene in a heterozygous individual. Say, an animal had both the purple and orange alleles for animal color. If neither purple nor orange was dominant, the animal would be part purple and part orange because both alleles are expressed in the phenotype.
- **polygenic inheritance** = the additive effect of two or more genes on a single phenotypic characteristic. That is, the appearance of a specific trait being affected by more than one gene. In humans, most characteristics (traits) are polygenic. For instance, skin color is a polygenic inherited trait in humans. Skin color is controlled by more than one gene.
- **chromosome theory of inheritance** = a basic principle in biology stating that genes are located on chromosomes and that the behavior of chromosomes during meiosis accounts for inherited patterns. Organisms have the phenotypes and genotypes they have because of the genes (on chromosomes) that they inherited from their parents.
- **linked genes** = genes located close enough together on a chromosome to be inherited together. If two genes are close together on a chromosome, even when crossing over occurs during meiosis, these genes will probably be inherited together in the same gamete.

- **sex-linked genes** = a gene located on a sex chromosome. In humans, any gene on the X or Y chromosome is a sex-linked gene. So, all normal humans individuals receive one copy of all Xlinked genes (which is a type of sex-linked gene). Human males get one copy of X linked genes because human males have one X and one Y chromosome. Human females get two copies of Xlinked genes because human females have two X chromosomes. The Y chromosome determines maleness in humans, if an individual human has a Y chromosome they are genetically male.
- **recessive disorder** = a disorder or disease caused by a recessive gene.
- **dominant disorder** = a disorder or disease caused by a dominant gene.
- **EXAMPLES OF RECESSIVE DISORDERS** = **albinism** (lack of body pigment), **cystic fibrosis** (an inherited glandular disorder causing the production of thick mucus in the lungs and other organs), **phenylketonuria** (PKU; lack of an ability to metabolize the amino acid phenylalanine; screened for at birth in almost all states), **sickle cell anemia** (homozygous form; lack of an ability to transport oxygen in the blood due to abnormal hemoglobin), **Tay-Sachs disease** (a neurodegenerative disorder caused by the accumulation of lipid metabolites in the brain; lack an enzyme for lipid metabolism)
- **EXAMPLES OF DOMINANT DISORDERS** = **achondroplasia** (a type of dwarfism), **Alzheimer's disease** (one type, not all types), **Huntington's disease**, **hypercholesterolemia** (abnormally high blood cholesterol level)
- **Huntington's disease** = a human genetic disease caused by a dominant allele. If you have one copy of the Huntington's gene you will get Huntington's disease. Symptoms of the disease do not usually appear until after the age of 40. Huntington's is characterized by uncontrollable body movements, degeneration of the nervous system and individuals usually die within 10-20 years after the onset of symptoms. There is a test which can be taken to detect if an individual has the gene before they actually show the symptoms of the disease.
- **SEX-LINKED DISORDERS** = a disorder resulting from alleles of genes located on sex chromosomes. Most of the known sex-linked disorders involve genes on the X chromosome (the Y chromosome is small and has relatively few genes on it). More males than females display sex-linked disorders because males only have one copy of the X chromosome (men don't have a "back-up X chromosome). If a male receives the allele for a sex-linked disorder, he will get it regardless of whether the allele is dominant or recessive. Females will only get recessive sex-linked disorders if they are homozygous for the allele (if the female has two copies of the allele, one on each X chromosome). If the female is heterozygous the dominant normal gene will be expressed and her phenotype will be normal. However, heterozygous females are carriers of sex-linked disorders and thus they can pass the disease onto the children even if they are normal.

- **carrier** = an individual that is heterozygous for an inherited disorder but who does not show symptoms of that disorder. The individual has the gene for the disorder and can pass it on to his/her offspring, but the carrier does not usually have the disorder.
- **EXAMPLES OF SEX-LINKED DISORDERS** = red-green color blindness and hemophilia.
- **hemophilia** = a sex linked recessive trait. Hemophiliacs bleed excessively when injured because they have inherited an abnormal allele for a protein factor (usually factor VIII) that is required for normal blood clotting.