Basic Concepts of Classical Genetics

Deoxyribonucleic acid (DNA): the molecule that most organisms use to store genetic information, which contains the "instructions for life"
Ribonucleic acid (RNA): a molecule related to DNA that living things use for a number of purposes, including transporting and reading the DNA "instructions"

Karyotype: A Karyotype is an array of **chromosomes** created by photographing the metaphase chromosomes from one cell, cutting out the individual chromosomes from the photograph and lining them up in order from largest to smallest, pairing the appropriate homologous chromosomes. Chromosomes for karyotypes are often stained using special procedures which create banding patterns the chromosomes, thus making pairing easier.

Genome: A genome is an individual organism's total array of genetic information.

Gene: A gene is a segment of DNA which controls the production of a particular characteristic. More precisely, a gene is a recipe for the production of a specific kind of protein.

> **Allele**: Alleles are different forms of the same gene. For any gene, an individual may possess only two alleles, and a gamete may possess only one. However, the gene pool of a species may contain many alleles for any gene. Alleles are assigned symbols according to specific rules of convention. All alleles of a particular gene should be given versions of the same symbol.
> **Locus** (plural Loci): A gene's locus is its position on a chromosome. All genes have individual and unique locations characteristic of the species. What makes two organisms members of the same species is that they have the same assortment of genes, arranged according to the same gene map on their chromosomes. In other words, their various genes have the same loci.
> **Multiple alleles**: A gene has multiple alleles if there are more than two different alleles for that gene in the gene pool. For example, there are three different alleles for the A-B-O blood type gene in human populations (L^A, L^B and l).

Genotype: The genotype of an organism is the list of the symbols representing that organism's specific genetic constitution—in other words, a list of all the alleles the individual carries for its genes. In actual usage, a stated genotype typically describes only one or two genes at a time.
Phenotype: The phenotype is the actual physical expression of an organism's traits. Much of the phenotype is the product of the genotype, but environmental influence can be very important as well. Geneticists discuss the

Homozygous; A homozygous individual has two identical alleles for the gene in question. For example, BB, bb, AA, PP.
Heterozygous: A heterozygous individual has two different alleles for the gene in question. For example, Bb, Aa, Pp.

Complete dominance: If two alleles display complete dominance, it is not possible to tell the difference between the homozygous dominant individual and the heterozygous individual. The recessive allele is hidden by the presence of the dominant allele.
Dominant: The dominant allele is the one which is displayed in the phenotype of the heterozygote. In assigning allelic symbols, the convention is to assign a capital letter to the dominant allele. Dominant traits can never skip generations in a pedigree, because they can never be present but hidden—they always show.
Recessive: The recessive allele is the one which is hidden in the phenotype of the heterozygote. The recessive allele is generally assigned a lower case letter symbol. Recessive traits can skip generations in a pedigree.
Co-dominance: If two alleles show co-dominance, the phenotype of the heterozygote expresses both of the alleles completely. For example, in the A-B-O blood group, the LA and LB alleles show co-dominance. The heterozygote (Type AB) has all of the bloody type characteristics of Type A blood as well as those of Type B blood. Again, co-dominant alleles are generally assigned different versions of the same capital letter for symbols.

Wild type refers to the phenotype which is most common in nature. This generally refers to a phenotypic which is found in almost all "wild" members of a particular species. Eg, in gerbils, the brown color is the wild type. All other colors are rare in nature.
A balanced polymorphism is when more than one allele exist in the population at a stable frequency at a given time. E.g the sickle cell HBB allele versus the normal HBB alleles both exist in high and stable frequencies where malaria is present. Where malaria is not present, the normal HBB allele prevails.

A **quantitative** or **polygenic** trait is controlled by two or more different genes working in concert. Eg, human skin color is controlled by >300 loci, each with at least two different alleles. The skin phenotype of an individual is the product of all the alleles of these genes working together, plus the impact of environmental influence. The result of quantitative inheritance is generally that the trait doesn't show discretely different phenotypes, but rather demonstrates a continuous variation in the species. Human skin color demonstrates this perfectly. There are no "categories" of human skin color, just a continuous spectrum from very pale to very dark.

CRISPR:

- **CRISPR**: Clustered Regularly Interspaced Short Palindromic Repeats of genetic information that some bacterial species use as part of an antiviral system. A group of scientists, including our co-founder Dr. Emmanuelle Charpentier, discovered how to use this system as a gene-editing tool (Jinek, et al. Science 2012)

- **Cas9**: a CRISPR-associated (Cas) endonuclease, or enzyme, that can unwind DNA (helicase) and act as “molecular scissors” to cut DNA (endonuclease) at a location specified by a guide RNA
- **Guide RNA (gRNA):** a type of RNA molecule that binds to Cas9 and specifies, based on the sequence of the gRNA, the location at which Cas9 will cut DNA. The sequence that recognizes specifically the DNA is ~ 20 nucleotides long.