

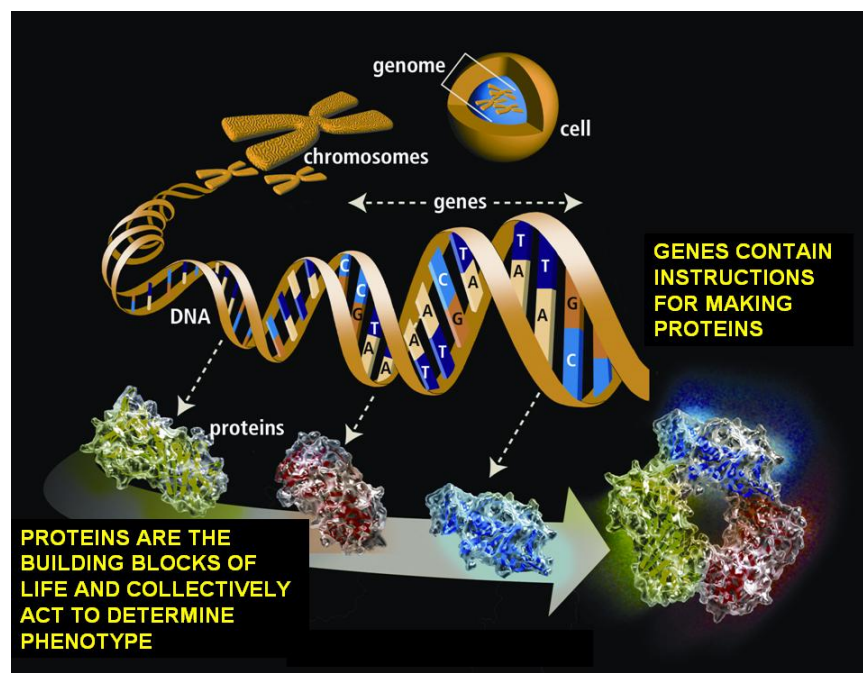
Basics of DNA markers and genotyping

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DNA (deoxyribonucleic acid) is a molecule that is shaped like a double helix and made up of pairs of **nucleotides**, abbreviated as “A”, “C”, “G” and “T” (Figure 1). DNA is packaged into chromosomes which are located within the nucleus of all cells. Every cell in the body contains all of the chromosomes that collectively make up the **genome** of that organism. DNA codes for **amino acids** which are linked together to form **proteins**. A **gene** is a stretch of DNA that specifies all of the amino acids that make up a single protein. Proteins are the building blocks of life. There are thousands of proteins in the body encoded by thousands of genes. The interaction and structure of proteins determines the visible characteristics or **phenotype** of an organism, while the **genotype** refers to the genetic makeup.

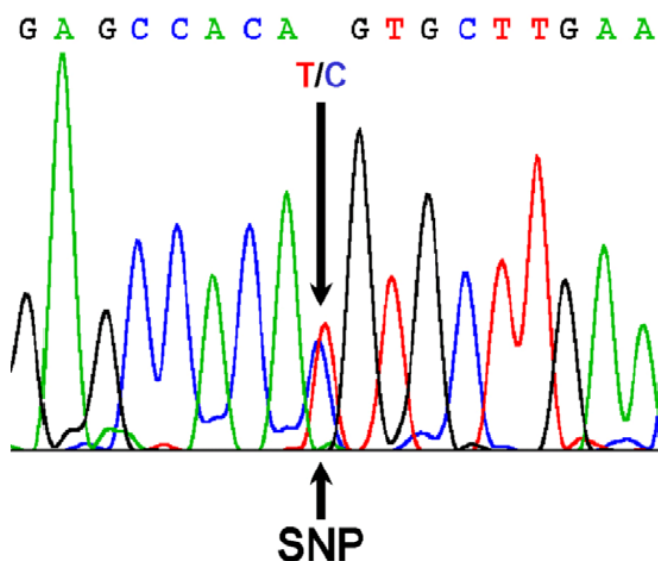


The sequence of nucleotides that encode a gene can differ between individuals. These differences are called **genetic variants**. As a result of these nucleotide differences, genetic variants or **alleles** may differ in the amino acid sequence of the protein they encode, or they may regulate the production of different quantities of the encoded protein. These differences can have an effect on phenotype.

Scientists have started to identify regions of DNA that influence production traits. They have used the techniques of molecular biology and quantitative genetics to find differences in the DNA sequence in these regions. Tests have been developed to identify these subtle sequence differences, and so identify whether an animal is carrying a segment of DNA that is positively or negatively associated with the trait of interest. There are several types of genetic markers. An animal will always carry two alleles of each marker; one from the sire and one from the dam.

Microsatellites are stretches of DNA that consist of tandem repeats of a simple sequence of nucleotides (e.g. “AC” repeated 15 times in succession). The tandem repeats tend to vary in number such that it is unlikely two individuals will have the same number of repeats. To date, the molecular markers used to determine parentage have primarily utilized microsatellite markers. Another type of genetic marker is referred to as a **single nucleotide polymorphism** or SNP (referred to as “snip”) where alleles differ from each other by the sequence of only a single nucleotide base pair. SNP genetic tests focus on detecting precise single nucleotide base pair differences among the three billion nucleotide base pairs that make up the bovine genome.

Genotyping is the term that is used to describe the process of using laboratory methods to determine which DNA-marker alleles an individual animal carries, usually at one particular



gene or location (locus) in the genome. The **genotype** identifies the marker alleles an animal carries. Because an animal gets one allele of each gene from its sire, and one allele of each gene from its dam, it can only carry two alleles of any given marker locus or gene. If an animal gets the same marker allele from each parent it is referred to as **homozygous** (e.g. “**” or “TT” or “140, 140”), or it may inherit different alleles from each parent in which case it is referred to as **heterozygous** (e.g. “*-” or “TC” or “144, 136”). DNA testing can be used to distinguish between animals carrying different marker alleles and this information can also be used for tracking parentage.

Some traits (e.g. coat color, horned/polled, Arthrogyrosis Multiplex) are controlled by one or a small number of genes and genetic tests for these **simple traits** are entirely predictive of phenotype. Most of the economically relevant traits for cattle production (birth weight, weaning weight, growth, reproduction, milk production, carcass quality, etc.) are **complex traits** controlled by the protein products of many genes, and they are additionally influenced by the production environment. The genetic component of phenotypic variation is the result of DNA sequence differences between chromosomes of individuals. When an animal has an expected progeny difference (EPD) above the base year average for a certain trait, what that means is that the animal has inherited a higher than average proportion of alleles for genes that favorably affect the trait. In other words, selection based on EPDs results in an increase in the number of favorable alleles an animal has, without knowing which specific genes or DNA sequence differences are involved. This contrasts with DNA-based selection where selection is based on the use of genotyping to identify animals carrying specific DNA variants that are known to be associated with the trait of interest. It should be noted that traditional EPD-based selection methods inherently tend to increase the frequency DNA sequences or markers associated with the alleles of genes that have beneficial effects on selected traits.