



Using Genomics Part I: What are DNA markers?

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Use of genomic technologies in the beef industry is ever-increasing. To understand the value and use of DNA markers, it helps to begin with basics about the genome and about DNA. DNA (deoxyribonucleic acid) provides the code needed to perform processes within an organism's body both to keep it alive and to perform in an expected way. DNA is simply many bases¹ (coded A, C, T, and G) bound together in long strings that are packaged into chromosomes within each cell. All of these chromosomes make up an organism's genome.

There are enough bases (letters) within the cattle genome to fill up approximately 2,000 New York City phone books. Given that, think of the genome² as a giant stack of large phone books. Each book is like a chromosome³ and the entire stack of books is the genome. Inside each of the books, the names and numbers represent the DNA sequence.

Cattle have 29 pairs of autosomal (non-sex determining) chromosomes and one pair of sex chromosomes (either XX for females or XY for males). Cattle inherit one chromosome from each of their parents, so for every place in the genome, there are two different alleles (alternative forms of a gene). This is why genotypes (genetic make-up of an individual) are recorded in pairs (for example, AA, BB, or AB). Some regions of the genome are the same in all animals of a species such as in cattle. Regions of the genome that vary between animals are mutations, and these mutations can be used as DNA markers. If an animal has two copies of the same mutation (AA or BB), they are homozygous⁴ (think homozygous polled, or horned). If an animal has 2 different copies of the mutation, they are heterozygous⁵ (think heterozygous polled).

There are several different types of DNA markers used in the beef industry today. The oldest type of marker currently used in the beef industry is called microsatellites⁶, and these are small pieces of DNA (a few bases) that are repeated over and over again. These markers would be scored⁷ and the genotype recorded as the number of repeats. For example, a heterozygous animal might have a genotype of 123/142 meaning that one chromosome has 123 repeats, and the other had 142 repeats. Microsatellite markers are often used in parentage testing. Because microsatellites are numbers of repeats, there are often more than two possible alleles at any given location. This makes microsatellites a very powerful tool, but also harder and more expensive to assay. A newer type of marker, single nucleotide polymorphisms (SNPs, pronounced "snips")⁸, have increased in popularity because they are simpler and less expensive to genotype than microsatellites. SNPs are a single base change in the DNA sequence and typically

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only have two possible alleles at any given location. A SNP genotype would look like A/C, or G/T if calling the bases directly, or AA, AB, or BB using generic coding.

Returning to the phone book analogy, think of microsatellites as common last names in the phone book. If you examine a phone book you will likely see the last name Smith or Jones repeated many times, just like you would see combinations of DNA bases repeated in a microsatellite. SNP markers are similar to finding small typos in the phone books. Only one letter has changed, but it can be identified and tracked within the phone book. When we perform a genetic marker test, it is akin to testing each phone book for the typos or repeated names at known positions. Unlike receiving the entire phone book, we only receive some of the "errors." Not every typo or repeat is surveyed, but given those that have been obtained, we can test for characteristics in important regions of the genome.

DNA markers⁹ are used in genomic applications just like one would use a marker on the side of a road while taking a long trip. Consider planning a trip across the United States from Boston, MA to San Diego, CA, which is about 3,000 miles. Since the bovine genome is about 3,000,000,000 base pairs, it makes an easy comparison. If there are only 300 road markers (300 SNP markers) for your road trip, you only have a marker every 10 miles (10,000,000 bases). If you have 50,000 road markers (like a 50K SNP chip¹⁰), you have a marker every 195 feet (60,000 bases). If you were given the task of finding an interesting landmark to visit on your trip, it would be much easier to find one with markers every 195 feet rather than every 10 miles. SNP markers within the genome work similarly: the more markers you have, generally



the easier it is to locate interesting regions of the genome that affect economically important traits in livestock.

Many SNP tests within the beef industry today are small (35-384 markers), but there are opportunities to genotype animals with higher density panels (7K, 50K, or 770K). As prices continue to decrease, these types of tests will become even more affordable and have greater industry acceptance in the future.

Vocabulary

- ¹ Bases- The building blocks of DNA sequence. Can be either A, T, G, or C. Because mammalian genomes are diploid (2 copies of each chromosome, one from each parent), bases are typically talked about in terms of base pairs.
- ² Genome- The sum total of all genetic material in an animal including DNA that is found in the nucleus of a cell. May also include DNA from the mitochondria (mitochondrial genome).
- ³ Chromosome- A linear (in mammals) structure comprised of packaged DNA. Bovines have 29 pairs of autosomal chromosomes plus the sex chromosomes.
- ⁴ Homozygous- The organism possesses the same allele (2 copies of the same allele) on each chromosome at a locus (For example, C/C or T/T, AA or BB).
- ⁵ Heterozygous- The organism possesses two different alleles at the same locus (For example, C/T or AB). Heterozygous animals can be referred to as carriers.
- ⁶ Microsatellite- Repeats of certain pieces in the DNA sequence that can be used as genetic markers. Genotypes are typically expressed as the number of repeats- For example, 123/126. These markers are often used in parentage panels, but their use is declining due to expense and assay difficulty.
- ⁷ Scored-Refers to the act of recording a genotype on an animal.
- ⁸ SNP- Stands for Single Nucleotide Polymorphism. Pronounced "snip". A single base mutation in the DNA sequence that can be used as a genetic marker. For example, one chromosome exhibits an A and another a G at the same position (which would be called an A/G SNP). These types of markers are gaining in popularity due to their ease and decreasing expense to genotype.
- ⁹ DNA Markers-Mutations found within a specific place in the genome
- ¹⁰ SNP chip-panel of SNP markers used to obtain genotypes on an animal. Usually, looks like a small glass microscope slide. Common SNP chips used in the beef industry include the 7K, the 50K and the 770K SNP chips.

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