



Essential Genetics for the Horseman

Whether you are an owner, breeder, rider or other equine enthusiast, there are some essential genetics that can help you to make the most of your journey. This primer is meant to introduce you to the basics of inheritance.

Over the last decade, scientific research in equine genetics and the mapping of the equine genome has led to a number of interesting and valuable discoveries. Some of these are directly relevant to the Arabian breed, especially the discoveries relating to disease genes. However, a number of very interesting insights about the entire equine species have been discovered.

DNA and Genes

The basic starting material for genetics in all species is their DNA. The horse genome (the collection of all the DNA in each cell of the horse) has been completely sequenced, just like in the human. The genome of the horse consists of about 2.7 billion base pairs (the basic unit of DNA, abbreviated by G, A, T, C). This is in comparison to the human genome that has just over 3.0 billion base pairs. When these base pairs are strung together in sets of a few thousand at a time, they provide the 'blueprint' for about 20,000 different genes in the horse. About 17,000 of these genes are very similar both in sequence and in function to the corresponding human gene.

Chromosomes

These genes are physically located on 64 chromosomes in 32 pairs, located in every cell of the horse. Each foal receives a set of 32 chromosomes containing 20,000 genes from its' dam and also a set from its' sire. This means that each foal has two complete sets of genes in each of its cells. There are not 40,000 different genes, but a sire version and a dam version of each of the 20,000 genes. This is important when we discuss dominant and recessive traits.

Sperm and eggs

When dealing with the cells responsible for reproduction, the sperm and the egg only carry 32 chromosomes each (and the corresponding single set of 20,000 genes). The 20,000 genes in a sperm cell represent a random set of genes that are present in the sire. Only one of the two copies of each of the 20,000 genes is present in each sperm cell. The process is similar in the production of an egg. When the sperm and the egg are joined to produce a new foal, the complete set of 64 chromosomes and double set of genes are reconstituted.

Dominant and recessive

All versions of a gene (called an allele) are not created equal. Often one version of a gene is dominant when compared to a different version of the same gene. This is evident in the E gene, which is the basic coat color gene in horses. The version (allele) of the basic coat color gene 'E' is dominant to the other allele 'e' (sometimes dominance is expressed with a capital letter and recessive is expressed using a small letter). The 'E' allele produces a black base color and this allele is expressed whenever it is

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present, whether a foal has one 'E' or two (Ee or EE). The 'e' allele produces a chestnut (red) base color, but if an 'E' allele is present with an 'e' the base coat color is black (Ee). For a chestnut color to be produced, the foal needs to have two copies of the 'e' allele (ee). So, a foal that receives one 'E' allele from either the sire or dam and an 'e' allele from the other parent will have black as their base coat color. If the foal receives an 'e' allele from both parents the color will be chestnut. A black base coat color will be EE' or 'Ee'. A chestnut base coat color will be 'ee'. Other genes can interact with the base color genes to produce additional effects on color (see the articles [Introduction to Coat Color Genetics](#) and [Arabian Coat Color Patterns](#)).

Two words that are used to describe the arrangement of alleles in an individual are homozygous and heterozygous. Homozygous means that the two alleles of a gene are the same, such as 'EE' or 'ee'. Heterozygous means that an individual carries different alleles, such as 'Ee'. When we discuss diseases heterozygotes are sometimes referred to as 'carriers'.

Both dominant and recessive alleles are involved in expression of a variety of diseases. Sometimes the disease gene is dominant and only one copy of the disease allele is necessary for the disease to be expressed. These disease genes are fairly easy to follow because the disease gene is not masked, but easily recognizable. Juvenile Idiopathic Epilepsy is thought to be caused by a dominant allele (see [Genetic Disorders and the Arabian Horse](#)). Hyperkalemic Periodic Paralysis (HYPP) found in Quarter Horses is a known dominant disorder.

At least three of the major disease genes in Arabians are recessive. Severe Combined Immunodeficiency (SCID), Lavender Foal Syndrome (LFS) and Cerebellar Abiotrophy (CA) all require that a foal inherit a disease allele from both the sire and the dam that then results in the disease. Recessive alleles are often masked by the 'normal' allele. A sire or dam that is heterozygous does not show signs of the disease and in the past this has made it difficult to identify 'carriers' of the disease allele. Today we have genetic tests that can identify the carriers of these three important genetic disorders.

In some cases it is a little more difficult to determine whether a disease or trait is recessive or dominant. Some are incomplete dominants, meaning that they have an intermediate form of expression when heterozygous. Other traits are 'polygenic', meaning they are caused by multiple gene pairs. This is actually the rule for most traits that we see including height, weight and athletic ability.

Probability

Dominance and recessive refer only to the action that occurs when the two gene versions (alleles) are compared with each other. It does not determine whether one of the alleles will be more likely to be inherited from a sire or dam. The chance that an 'E' or an 'e' is passed to the next generation from an 'Ee' sire or dam is 50/50. Sometimes we fall into a trap of thinking that because one allele is dominant over the other allele, it is more likely to be passed to the offspring. This is not the case. The rule still holds that either allele, the dominant or recessive one, has an equal chance of being passed to the next generation.

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When the set of alleles in a foal is produced from the union of the sperm and the egg it results in two alleles of every gene; one set from the dam and one from the sire. In turn, the sets of genes available for transmission to the foal came from the two grand sires and two grand dams. The foal then receives on average 25% of its alleles from each of the four grandparents. At each generation, the sets of alleles available from either the sire or dam are rearranged, essentially randomly, and passed to the offspring. Two possible results of this process is that you can have two full brothers that may have inherited completely different sets of genes and therefore 'not related' genetically or may have inherited a set of genes that are identical with each other and therefore 'identical twins'. These cases would be extremely rare. It is more likely that two full siblings share closer to 50% of their alleles.

If a sire carries a recessive disease allele, the probability that the offspring inherits the recessive disease allele from the sire is 50%. Likewise if the dam carries a recessive disease allele the probability that it will be inherited by the foal is 50%. If two carriers are bred, the result is that a foal has a 25% chance of inheriting the disease allele from both the sire and the dam, resulting in the expression of the disease. The foal also has a 25% chance of inheriting the non-disease allele from each parent and being completely free of the disease allele, and subsequently not able to pass a disease allele to the next generation. 50% of the time a foal will inherit one disease allele and one non-disease allele from the parents. This foal will not express the disease, but is capable of transmitting the disease allele to the next generation.

Mutations

The word mutation is often thought of as something very bad. In some cases the result of a mutation does cause a significant problem. However, mutation just means that there has been a change in the DNA sequence from what is accepted as the 'wild' type. Mutations are what are responsible for all of the variety that we see in the appearance of our horses. How boring would a halter or performance class or an endurance race be if all of the horses were 'clones' of each other?

Mutations occur when mistakes are made during the replication of the DNA in the cell. Exposure to certain chemicals or radiation can sometimes increase the chance of a mutation occurring, but most mutations are normal accidents of the replication process. It is estimated that each new foal born has about 10 new mutations in its genome. Almost all of these cause no observable difference in the foal. Mutations are not caused by inbreeding. Sometimes inbreeding makes it more likely that a sire and dam may both carry a 'bad' gene that they inherited from a common ancestor in their lines. This can result in lethal cases of recessive disease more often in closely bred lines.

Other types of inheritance

There are other types of inheritance that play a role in the genetics of our horses. Mitochondrial DNA (mtDNA) is inherited exclusively from the dam. The sire has essentially no contribution of mtDNA to the foal. The mitochondria have some vital functions in the cell's respiratory cycle and the production of ATP (the molecule that moves energy around the cell). It is the powerhouse of the cell. It is likely that performance is affected in a number of ways by the mitochondria. Because mtDNA is inherited strictly from dam to foal, it is an excellent way to trace and record the maternal lineage of the foal.

The X and Y chromosomes are the 'sex chromosomes'. The genes that determine maleness are located on the Y chromosome. Males have both a Y chromosome and an X chromosome (XY), females have two

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X chromosomes (XX). Colts always inherit their Y chromosome from their sire and their X chromosome from their dam. Fillies inherit one X chromosome from their sire and one from their dam. There are some traits that are sex-linked meaning that these genes are found on the X chromosome. There are other traits that are sex-limited, such as milk production in mares. Even though both males and females have genes for milk production, they are only expressed in females.

As we continue to unravel the secrets of the horse genome, more of the genes associated with these traits are beginning to reveal themselves. As we move forward we will have many new and exciting genetic tools to help us plan our future generations of equine partners.

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