

# Equine Genetics

## Equine Science – Chapter 6

The field of genetics began “exploding” with new information produced by cutting-edge research during the 1990s – a trend that has continued up to the present time. Scientific technology has advanced to the point that the identities and positions of the genes that make up humans and other animal species are being determined and recorded. The new technology is called gene mapping.

Because research dollars are limited, the study of equine genetics has lagged behind work on many other species. Humans have been the first priority for genetic studies, followed by livestock species on which the food and fiber industries depend. Mice, dogs and other non-livestock species have also been studied more than equines. Still, funding has emerged to support work in equine genetics.

Genetic research on the molecular level of the gene is known as genome research. The International Society of Animal Genetics has been a leader in equine genome research projects with cooperative effort worldwide. Additionally, because there are many genetic similarities across species, especially in the field of disease, some of the information learned about humans and other animals applies to equines as well.

This document will give a brief overview of current equine genetic studies and some gene-related information that can be beneficial for all horse owners.

### Some Background

Each cell of every living organism (animal or plant) has a center, known as the nucleus. The nucleus contains chromosomes, which carry genetic information about that particular organism. Chromosomes can be seen with a high-powered microscope.

Chromosomes come in pairs, called chromosome pairs or diploid chromosomes (diploid means double or two). The genetic makeup of any individual organism contains one chromosome from each of its parents, and this makes a chromosome pair.

Every kind of organism has a certain number of chromosome pairs. For example, horses have 32 chromosome pairs. Humans have 23.

A chromosome is made up of genes. The genes are arranged like beads on a necklace along the chromosome.

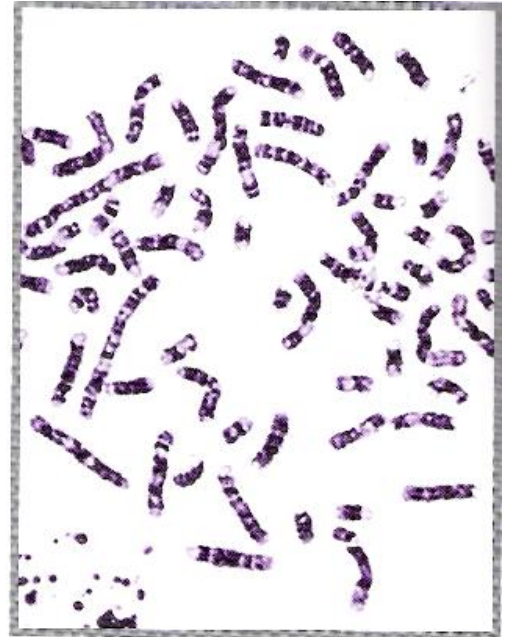
### What are genes?

Genes are so small that they cannot be seen, even with a microscope. They are tiny bits of information, contained in the cells of every living organism, that make the organism what it is. Genes are passed from one generation to the next.

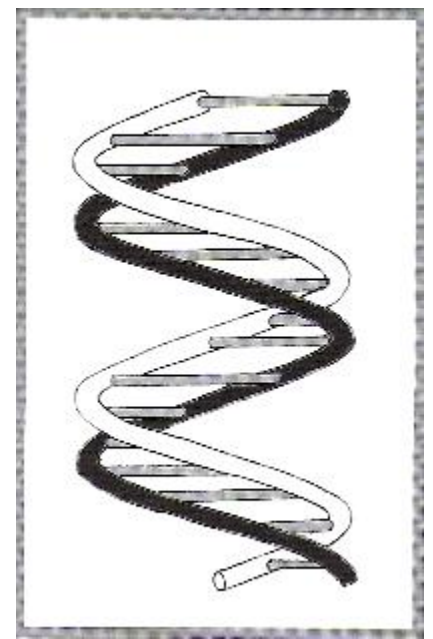
Genes are made up of complex molecules of DNA (deoxyribonucleic acid). DNA is composed of different amino acids and proteins, arranged in 2 thread-like strands that are twisted together like a coil. This coil-like structure is called a “double helix”. Between the two strands are bonds that hold the DNA together, somewhat like the rungs of a ladder.

The two pairs of a gene pair are not necessarily exact duplicates, although they are responsible for the same trait (for example, eye color or hair color).

Chromosome pairs split during one phase of the reproductive process. When this happens, half of the genetic information of each parent is passed on to the next generation. This process is called meiosis. Meiosis occurs only in reproductive cells and produces gametes, or sex cells, in preparation for the rest of the reproductive process. The chromosome now has what is called the haploid number of



Chromosomes seen under a high-powered microscope.



chromosomes – haploid means “half”. The process of meiosis occurs in the reproductive organs (the ovaries or mares and the testes of stallions).

Body cells also split in half; however, body cells are not involved in the reproductive process. When body cells split in half – this process is called mitosis – they produce identical copies of the original cell. These copies contain the diploid number of chromosomes.

As you will remember from Equine/Horse Basics, different equine species have different numbers of chromosomes:

Equine Species (Common Name)	# of Diploid Chromosomes	# of Haploid Chromosomes
<b>Equus przewalskii</b> (Przewalski's horse)	66	33
<b>Equus caballus</b> (domestic horse)	64	32
<b>Equus Asinus *</b> (domestic ass/donkey)	62	31
<b>Equus burchelli boehmi*</b> (Grant's zebra/common zebra)	44	22

\*Note – The several different ass/donkey species and zebra species each have their own number of chromosomes, which can vary greatly from one species to the next. (Ass/Donkey – from 50 to 63 chromosomes, Zebra – from 32 to 46 chromosomes)

A chromosome example:

- The horse has 64 diploid chromosomes or 32 chromosome pairs (haploid number)
- Each of the 64 chromosomes of the horse contains different genetic information
- Each body cell in the horse contains 64 chromosomes.
- A sex cell that splits in half has only 32 chromosomes.

Model genome research has developed ways to decode and take an “inside look” at genes on the molecular level. Some terms for this technology include:

- Gene mapping
- Zoo-FISH maps.

By understanding the complicated science of genetics, we can make prediction about the genetic makeup of the next generation, based on genetic information about the mare and stallion that are being mated.

The process of passing genetic material to the next generation is known as inheritance. Having a good understanding of inheritance traits is very important to the future of the horse industry, giving breeders a basis of information to consider when deciding whether to breed a particular mare to a particular stallion. Genetic selection is crucial in preserving breed purity and strongly influences the quality of the offspring, performance traits, conformation, gaits, coat color, white-spotting patterns, predisposition for disease, potential genetic problems, and more. Parentage testing is now readily available and has become a common practice in many breed associations.

## Some Genetic Language

Learning to use genetic terms accurately makes the complex science of genetics easier to understand. Below are a few more terms that will be helpful for you:

- **Locus** – the position of location of a gene on a chromosome (singular = locus; plural = loci)
- **Alleles** – genes that are in a chromosome pair and responsible for a particular genetic trait; alleles are not necessarily identical but are at the same location or locus
- **Dominant** – “simple genetic inheritance” works by allele pairs; if a gene is “dominant”, it has the ability to mask the genetic makeup of the other gene in an allele pair (Ee – “E” is dominant or “e”)
- **Recessive** – “simple genetic inheritance” works by allele pairs; if one gene cannot mask the genetic makeup of the other gene in the allele pair, it is “recessive” (ee – Neither “e” is dominant over the other “e”; both are “recessive”)
- **Homozygous** – refers to an organism whose chromosomes carry two identical members of a given allele pair (EE or ee – the two genes are the same, or are “homozygous”).
- **Heterozygous** – refers to an organism whose chromosomes do not carry identical members of a given allele pair (Ee – the two genes are not the same, or are “heterozygous”)
- **Multiple alleles** – many genetic traits do not follow the principles of “simple genetic inheritance”; some genetic traits are determined by complicated genetic inheritance involving the interaction of multiple gene pairs or “multiple alleles”
- **Polygenic traits** – genetic traits that are controlled by multiple genes, gene pairs, and/or alleles
- **Phenotype** – What an individual “looks like”, based on the genetic information of the alleles present
- **Genotype** – what the genetic makeup of an individual really is; what alleles are actually present.

## Sex Inheritance

The sex of the foal is determined by the sire, not by the dam. No other genetic trait currently known is determined solely by the sire. The pair of chromosomes carrying the genes that determine the sex of the offspring are called sex chromosomes. In the mare, these chromosomes are identical; she carries two X chromosomes. In the stallion, they are not identical; he carries an X and a Y chromosome in the chromosome pair.

**Female or Mare = XX**

**Male or Stallion = XY**

So when the chromosome pairs split during the process of meiosis (forming two gametes) in preparation for the reproductive process:

**A mare would produce:**

X            X

**A stallion would produce:**

X            Y

Because the process of sex inheritance is a “simple genetic inheritance” trait, a Punnett square can be used to determine the expected outcome of the mating. Geneticists use the Punnett square to determine expected outcomes and can determine the percentage of expected offspring that should show a particular trait. The Punnett square can only be used when simple inheritance patterns apply.

**Punnett Square for Sex Inheritance**

	Chromosome from the Stallion = X	Chromosome from the Stallion = Y
Chromosome from the Mare = X	XX Female	XY Male
Chromosome from the Mare = X	XX Female	XY Male
Expected %	50% Female	50% Male

## Color Genetics

Because of selective breeding, a wide variety of colors have become common among horses over the many centuries since domestication began. So the horse of today certainly is a “horse of a different color”! Many breed associations have been formed based on coat color, such as the American Paint Horse Association and the Appaloosa Horse Club. Other breed associations today welcome horses of colors that were extremely rare in their breeds until recently, such as palomino-colored Morgans.

As you might guess after reading the “Basic Coat Color” chapter, the genetics of coat color in horses and other equines is very complicated. Entire books have been written about it! This section will include some of the highlights of what is currently known about the coat colors of equines.

Color is one means of identification of individual horses. Determining a horse’s color is a matter of observing the animal’s phenotype – what the horse “looks like”. However, determining that genetic makeup of genotype of the same horse is much less simple. Much progress has been made in recent years to identify the many genes, gene combinations, and genetic interactions that determine coat color in horses. But as yet there is still no laboratory test to identify the complete genetic makeup of genotype for the coat color of a horse.

Color in the horse is controlled by granules of pigment in the hair, the skin, the iris of the eye, and also some internal tissues. This pigment is known as melanin. Melanin comes in two different forms:

- Eumelanin – controls black or brown
- Pheomelanin – controls red or yellow

Melanocytes are “pigment cells” made up of eumelanin and pheomelanin. The genes for color on a chromosome produce different colors by alternating between eumelanin and pheomelanin. Genes control the presence of absence of these two color pigments in melanocytes, as well as the shape, number and arrangement of the pigment granules.

### A Few Coat Color Terms and Symbols:

- Extension gene – the gene that regulates the color differences in black-pigmented and red-pigmented horses
  - Black-pigmented horses include:
    - Blacks
    - Browns
    - Bays

- Also – buckskins, duns and grullas
- Red-pigmented horses include:
  - Chestnuts/sorrels
  - Also – palominos and red duns

The “extension gene” controls the amount of black eumelanin granules in the skin and/or the coat by means of a pair of alleles. It is represented by the following two symbols:

- E – Extend or increase the amount of eumelanin; decrease the amount of pheomelanin
- e – Decrease the amount of eumelanin; increase the amount of pheomelanin

The extension gene is dominant, so:

- EE or Ee – black, brown, or bay; these horses have black pigment in both the skin and hair
- ee = chestnut or red; these horses have black pigment in the skin but not in the hair

There is a great variation in basic coat colors among black, brown and bay-colored horses. Variation is also seen among chestnut or red-colored horses. It is thought that multiple alleles are responsible for the variations in coat colors. Research about this topic is ongoing.

- Agouti gene – the gene that controls the distribution pattern of eumelanin in the hair and restrict the distribution to the “points” (the mane, tail, lower leg, and ear rims) by means of a pair of alleles.

The agouti gene is represented by the symbol A.

AA = the presence of eumelanin is restricted to the “points” pattern

a = the presence of eumelanin is not restricted to the “points” pattern

Black	EEaa or Eeaa
Bay or Brown	EEAA, EeAA, EEaA, EeAa
Chestnut/Sorrel	eeAA, eeAa, eeaa

- **Dilution gene** – there are at least 3 genes that cause the “dilution” of coat colors in horses. A full discussion of what is currently known about dilution genes is much too complicated for the purposes of this book. The following list give some of the basic coat colors that are the result of these dilution genes’ interactions:
  - Palomino
  - Buckskin
  - Cream
  - Perlino
  - Dun
  - Champagne
  - Silver dapple – NOTE: dappling is not always a direct result of a dilution gene. Some horses whose coats show a “silver dapple” effect do not have dapples. This genetic effect would have been better named “silver”.

The genetic elements that control coat color get even more complicated. Some additional colors/color patterns and the symbols of the genes that control them are listed below:

- White – Ww
- Gray – GG or Gg
- Roan – RNrn
- Tobiano – TOTO or Toto
- Overo = Oo
- Few-spot leopard = LPLP
- Leopard, blanket, varnish roan, snowflake or frosted = LPlp

More symbols to represent colors are being developed as more genes that control color are identified. Even the symbols listed here may change as the “genetic language” of coat color is developed further.

## White Markings

White markings in horse are thought to be product of both genetic and “non-genetic” factors. White markings are a result of the melanocyte cells. Melanocyte cells develop during the process of embryonic development. Interestingly enough, they develop out of the embryonic tissue from the neural crest – the area of cells from which the spinal cord and brain will eventually develop. The melanocyte cells migrate outward from this area during further cell development in the embryonic process.

White hairs occur due to a lack of pigment granules (eumelanin and pheomelanin) in the melanocyte cells. Skin that lacks pigment granules is pink (white skin that gets its pink color from small superficial blood vessels).

Most white markings in horses have pink skin beneath them. In a few instances, small, narrow white markings may have dark skin underneath. You may also see what looks like white markings as a result of scar tissue where melanocyte cells have been destroyed, or from “graying out” in older animals.

No two horses have exactly the same white-marking patterns, so white markings are an excellent tool for animal identification. For this reason, white markings are recorded by veterinarians on Coggins, rabies, and other health documents.

Genetic research on white markings has been done in identical foals. In 1984, investigators W.R. Allen and R.L. Pashen split developing foal embryos in two and implanted them in separate mares. The resultant “identical foals” were of the same sex and color but had different white markings. The “non-genetic” influences on white markings are not known or understood to this day. However, genetic research has demonstrated that white markings are relatively heritable from the parents.

## Some Genetic Disorders

A number of genetic disorders are present in the equine population and affect the horse industry. All horse owners should be aware that genetic disorders exist and be familiar with those that are most serious. This is especially true for anyone who breeds horses. The following are several of the more common genetic disorders.

- **Lethal White Foal Syndrome (LWFS), also known as “Megacolon”**
  - **Which breeds?** – Paint/pinto colored horses; most documented cases are found in American Paint Horse Association-registered animals.
  - **What is it?** – seen in blue-eyed, white-coat-colored foals; which may have very slight colored skin spots or hairs around the muzzle, ears, or tail; within a few hours after birth, the foal shows signs of intestinal discomfort, which may mimic that caused by retention of meconium (first feces); fecal material builds up in the gut and cannot be passed because lack of nerve cells in the digestive tract results in an absence of normal peristaltic movement; neither medication nor surgery can correct the problem, and affected foals usually die within a few days after birth
  - **Genetics of the condition** – “lethal white” foals are homozygous for the overo gene; foals have at least one overo-colored parent.
- Hyperkalemic periodic paralysis (HYPP)
  - **Which breeds?** – Quarter Horses of genetic lineage of the sire Impressive, as well as of breeds or cross breeds that have been bred to that genetic line (e.g., American Paint Horses and Appaloosas)
  - **What is it?** – episodes of muscle tremors, weakness, stiffness, and paralysis; during which the animal could stop breathing and die; episodes not necessarily associated with exercise or stress; visibility of the third eyelid may be a warning sign of an episode; diet that include high levels of potassium can trigger attacks; affected animals, which are usually heavily muscled, have high blood potassium levels and an increased skeletal muscle-cell membrane permeability to sodium.
  - **Genetics of the condition** – an inherited dominant gene causes a defect in the muscle physiology; testing for the presence of this gene is available.
- Hemophilia A
  - **Which breeds?** – Thoroughbreds, Standardbreds and Quarter Horses
  - **What is it?** – seen in colts only; causes inability of the blood to clot, resulting in hemotoma, internal bleeding, anemia and death.
  - **Genetics of the condition** – The defective gene is carried on the X chromosome; therefore, this is a “sex-linked” genetic trait; fifty percent of a carrier dam’s fillies would carry the gene; colts either die prior to breeding age or are not used for breeding because of the affliction; so the gene is passed on only by carrier mares with the defective X gene.
- Severe combined immunodeficiency (SCID or CID)
  - **Which breeds?** – Arabians
  - **What is it?** – a lethal disease of Arabian foals, causing improper development of the immune system; foals usually die from severe respiratory infection before 5 months of age
  - **Genetics of the condition** – a recessive genetic trait; if a foal is afflicted, both parents are carriers; in 1997, equine-genome research clearly identified the gene responsible for the disease.
- Equine night blindness (ENB)
  - **Which breeds?** – Appaloosas
  - **What is it?** – visual impairment at night; afflicted horses tend to stumble in the dark; may have high incidence of injuries occurring at night; normal horses usually have very good night vision.
  - **Genetics of the condition** – suspected to be a recessive genetic trait in horses
- Aniridia
  - **Which breeds?** – Belgians

- **What is it?** – absence of the iris of the eye, plus cataracts of the eye; most afflicted animals are blind and unfit for work; conditional has been traced back to one Belgian stallion in Sweden.
- **Genetics of the condition** – caused by a dominant gene; history of this disorder provides a good example of useful genetic selection: breeders and the breed association acted to eliminate the problem by deciding not to allow afflicted animals to be used as breeding stock.
- Rhabdomyolysis (also known as “tying up”, “Monday-morning disease” or “azoturia”)
  - **Which breeds?** – seen mostly in Draft breeds; seen occasionally in most breeds of horses
  - **What is it?** – destruction of muscle tissue, causing muscle cell necrosis; horses develop a stiff gait, especially in the hind quarters, during or just after exercise; extremely painful; in some cases the horse is unable to move; blood may be seen in the urine.
  - **Genetics of the condition** – a dominant-gene inheritance; polysaccharide-storage myopathy (PSSM) is one type of rhabdomyolysis that has been proven to be a genetic trait; research is ongoing
- Osteochondrosis (OC)
  - **Which breeds?** – breeds characterized by fast growth, such as Thoroughbreds, Standardbreds, Warmbloods and Quarter Horses
  - **What is it?** – abnormal bone growth that affects horses’ joints; may cause cyst-like lesions in the bone or flattened or depressed areas in joint surfaces; weakening of bone structure can result in bone fragments breaking away and ending up in the joint.
  - **Genetics of the condition** – OC usually occurs during periods of fastest growth in young horses; a genetic factor is thought to be the inherited ability to grow quickly; may involve a problem with bone-growth patterns or a sensitivity to nutrient deficiencies or excesses.
- Equine degenerative myeloencephalopathy (EDM)
  - **Which breeds?** – Standardbreds, Arabians, Appaloosas, Thoroughbreds, Paso Finos, Morgans, Quarter Horses, and other breeds; also Przewalski’s horses and zebras
  - **What is it?** – a degenerative condition that affects the brain and spinal cord; causes an ataxic gait in the front legs, hind legs or both.
  - **Genetics of the condition** – a genetically related sensitivity of individuals to low levels of vitamin E is suspected; thought to be a function of both genetics and environment (nutrition); ongoing research is improving the understanding of this problem.
- Neonatal isoerythrolysis (NI)
  - **Which breeds?** – Arabians, Thoroughbreds, Quarter Horses, Morgans, Warmbloods, pony breeds, and miniature horses.
  - **What is it?** – incompatibility of mare’s and foal’s blood group; causes destruction of red blood cells in newborn foal; seen 2 to 5 days after birth; foal anemia results; foal shows lethargy and elevated pulse and respiratory rate; occurs in foals from second and later pregnancies, after mare has become “sensitized” to incompatible blood types, causing her immune system to form antibodies that destroy foal’s red blood cells
  - **Genetics of the condition** – inherited “susceptibility” to sensitization by blood types; genetics not clearly understood.

## Genetic Testing

In very recent years, genetic testing has become available to the horse industry. Some breed registries now require parentage testing prior to foal registration in order to verify the sire and dam of the foal. There may be other reasons for equine genetic testing in the future, as more and more genetically related inherited traits and genetically related disorders are understood.

Some tests that are currently available for horses include:

- **Blood-group test** – blood groups of horses are similar to those of humans; parentage can be verified with equine blood group tests.
- **Lymphocyte tests** – high effective test that use the immune response and rejection of tissue from unrelated animals; can be utilized to verify parentage as well as to detect susceptibility to certain diseases in equines
- **DNA test** – can be done with blood samples or with samples of hair from the body, tail or mane; tests are accurate and inexpensive; results are available quickly.

## Summary

Great strides have been made in the field of equine genetics in a relatively short time, thanks largely to the international equine-genome project. The cooperation among scientists of many nations in this effort has been good for the horse industry worldwide.

Advances in equine genetics are changing the horse industry. An understanding in this science is important for all horse owners, but become more knowledgeable and responsible about equine genetics is imperative for breeders.

This is a field in which scientists are learning more every year. Continue to study and learn about this important and interesting part of the equine industry!