

About Color (and ASD)

Mountain Horses have some unique colors caused by the Silver Dapple gene, which occurs in several breeds. This color is associated with the gene for an eye condition known as ASD. This page discusses both of these genetic conditions and their relationship.

Base and Modified Colors

Mountain Horses come in most of the colors found in other breeds including the base colors of black, bay, and sorrel or chestnut. These three base colors in turn may be modified by other genes which are called "modifier genes" which usually dilute or add white to the base colors, so that you find roans, paints, duns, grays, palominos and buckskins, etc.

Chocolates

In the Mountain Horse breeds, and primarily the Rocky Mountain Horse registry, the color known as chocolate with a flaxen mane and tail is very popular, and has become strongly associated with this horse. At least four of the five foundation stallions of the RMHA registry were chocolate, all sons of Tobe, himself chocolate in color. The chocolate color is also found in other breeds, most commonly in Icelandics, Shetland Ponies, Miniatures and some draft breeds.

The chocolate color is caused by a color modifying gene. That is, a gene which modifies (in this case dilutes) another color. This gene has been named the Silver Dapple gene by geneticists. It is a gene that only dilutes the color black. It does not affect the color red (sorrel or chestnut). Therefore, a sorrel or chestnut horse can carry the gene and pass it on, but you cannot tell whether the horse carries the gene from looking at the horse, as there is no black on its body to modify.

The silver dapple gene is a dominant gene which produces an even more dilute effect if two of them are present in the animal (one inherited from each parent). In these homozygous horses, the manes and tails are almost white, and the black on the body is lightened up to an almost gray color. If it is a red chocolate horse (silver dapple bay), the black points on the legs are so light that horse almost looks like a solid chestnut with a white mane and tail, at least when the horse is young.

Color and the Eyes

Color genes are arranged on the chromosome strand very close to the eye genes. Often, during reproduction, groups of genes travel together in clumps and are said to be genetically linked. This can happen with eye and color genes.

Geneticists from Michigan State University have determined, in one of the largest equine studies ever done, that a gene or genes affecting the physical development of the eye is associated with the Silver Dapple gene. This syndrome is a collection of differences in the structure of the front part of the eye is called ASD, short for Anterior Segment Dysgenesis. In the vast majority of cases, ASD causes the horse no problems, it is merely a collection of harmless differences from the norm in physical structure.

This gene linkage is a color phenomenon, not breed-related. Researchers have found the ASD-silver dapple link in most breeds with this color. However, the silver dapple coloring is very popular in Mountain Horses, and Mountain Horse breeders have always bred for this color. Consequently, we have learned a lot about ASD.

It has become common for people in the breeds to refer to only the homozygous horse, who carries two of the ASD genes, one from each parent, as an "ASD horse." The horse that has inherited only one ASD gene from one parent (heterozygous), is referred to as a "cyst-only" horse. These horses usually show only one of the characteristics of ASD, small structures behind the lens called "cysts". The cysts cause no problems for the horse or its vision.

Most breeders of Mountain Horses have become convinced that the Silver Dapple and ASD genes are 100% linked that is, where the horse has inherited a single silver dapple gene, it will also have a single ASD gene. If the horse has two of one gene, it will have two of the other. If it has no silver dapple gene (i.e., true black or bay) it appears to have no ASD gene. While this assumption is not borne out by research, it is something that has been reliably experienced by breeders in over ten years of breeding since the initial MSU study. This linkage appears to occur even if the eyes of a chocolate horse examine as completely normal, without any indication of even a single ASD gene. About 13% of chocolate horses do examine this way, and they are called silent carriers. Silent carriers are usually heterozygous so they will pass on the ASD gene 50% of the time just like horses whose eyes show cysts.

The Nature of ASD

The abnormal ASD gene is a semi-dominant gene. When the horse inherits only one abnormal gene from one of its parents, there is only the very slight and harmless physical indication of cysts (or none if it is a silent carrier) behind the lens. Cysts can usually only be detected by an experienced veterinary ophthalmologist using an indirect ophthalmoscope. About 87% of horses that carry a single ASD gene will show cysts. The remaining 13% are silent carriers, and can pass on the gene to offspring to the same degree as horses whose genetic status is visible in an eye exam - that is, 50% of the time.

NOTE: Please be aware that this distinction between the results of an eye exam and the actual genetic status of a horse as a carrier of the abnormal ASD gene is important to understand. Unfortunately, many breeders or sellers of our horses either do not understand or do not properly explain that a "normal" eye exam is not in any way a guarantee that the horse does not carry the gene. There are a significant number of chocolate "silent carriers" that examine as normal but carry, and pass on, the ASD gene. Some breeders may advertise a horse that has had a "normal" result on an eye exam as being "aa" - which is the genetic symbol for a horse that does not carry the ASD gene. This is very misleading and detrimental to anyone who may want to breed the horse they are purchasing. An eye exam can only confirm the presence of the ASD gene, but it cannot rule it out. Until we have a genetic test for the presence of the abnormal ASD gene, which to date we do not, it must be assumed that all chocolate or red chocolate horses are carriers or "silent carriers" of at least one abnormal ASD gene, and will be able to pass it on in breeding.

When a horse inherits two of the dominant form ASD genes, one from each parent, the eyes of the horse will almost always show other differences in addition to the cysts. (This is the horse that in lay terms has become known as the ASD horse.) Many of these are easily seen if you know what to look for. Not all homozygous horses will have all of the various differences which are part of the ASD syndrome most will exhibit only some of them.

It has now been determined by a separate study using sophisticated equipment that only one of the various ASD syndrome characteristics adversely affects the vision of the horse**, and that is where the lens itself is out of proper position or is detached. This is also the only characteristic with a progressive component, that is, a characteristic that will get worse over time the detached lens will usually develop cloudiness that worsens with time. This can only occur with the homozygous ASD horse (symbolized as "AA").

Of the homozygous AA group of horses, only about 10% of them will have the subluxated lens which affects vision. This makes up a very small percentage of the total horse population. If you are buying a horse, you do want to make sure that your candidate does not have displaced or detached lenses. If it does not, then the horse should be perfectly fine for using purposes no matter whether it has one ASD gene, two ASD genes or none.**

The good news is that, except for the very small percentage of horses with subluxated lenses that will get more and more cloudy, the characteristics of ASD do not change over time. They are physical, developmental differences that occurred in the womb. What you (or your vet) see is what you will get, now and in the future. The changes from normal are not a disease process, cause the horse no pain, and will not come and go or appear at a later time. You can know from a competent prepurchase exam what you are getting.

Some people are concerned in looking at a horse to purchase, that the parents were both chocolate, and therefore they are worried that there is more of a possibility of problems than with a horse having one chocolate and one black parent. This should not be a concern. The offspring of two silver dapple parents that happens to inherit only one ASD gene or no ASD gene, is no different, physically or genetically, than the horse that inherits one ASD gene or no ASD gene from the silver dapple/non silver dapple parents. The only question is whether the offspring has inherited two ASD genes, and if so, whether vision is affected as a result. And in this case, a competent veterinary exam should be able to tell you whether the horse has any resulting vision difficulties.**

Color and Breeding

If you are intending to breed a horse, the ASD gene status of your horse is something you should know in order to select a mate that will minimize chances of a homozygous ASD foal, as it is only the homozygous foal that has any chance of having affected vision. Again, we believe that any "chocolate" or "red chocolate" horse will carry at least one of these genes, and until we have a DNA test for the presence of this gene, horses should be bred with this assumption, regardless of the results of a visual exam. A sorrel or chestnut horse may carry none, one, or two copies of the ASD gene. If you are unable to determine the ASD gene status of your horse from its color and/or its parentage or color of its offspring, a veterinarian trained in the techniques for detecting ASD should examine the horse. Unfortunately, even

an exam cannot eliminate the possibility of the presence of the ASD gene, but it may well confirm it.

If you desire to eliminate any chance of a homozygous ASD foal in your breeding program, the most reliable way of doing this to date has been to include not more than one "chocolate" (or sorrel with a known ASD gene) in your breeding pair. Regardless of whether the one silver dapple mate is heterozygous ("cysts only") or homozygous ("ASD"), you will have NO chance of getting a homozygous ASD foal, if one of the pair does not carry any ASD gene. Of course, the genetically normal horse in the pair (symbolized as "aa") must be truly "normal". This horse may appear as black, bay or chestnut, or any modification of these colors caused by a modifier gene that is not Silver Dapple.

How can you know if a horse is truly genetically normal, if there are silent carriers in which the presence of the gene cannot be detected? How can you tell with a sorrel or chestnut horse. The only clue breeders have had since the ASD-silver dapple relationship was discovered, is the color of the horse. Chocolates and red chocolates are assumed to be carriers; blacks and bays are assumed to be normal. The sorrel or chestnut horse could have any genetic status. Here we have a true mystery, what I call "the mystery of ASD." While geneticists have stated that they have seen "cysts" in horses that appear bay or black, and have warned breeders that they cannot rely on color to make breeding decisions that always will avoid homozygous ASD, breeders in practice have had to rely on color in the absence of a genetic test, and considering the problem that eye exams do not detect all carriers. And over the last ten+ years of countless thousands of horses bred, it has proved to be a very reliable tool. Breeders in the field who want to avoid homozygous ASD, breed chocolates to black or bay horses, and do not get ASD. It has been as simple as that. Chestnut horses are the only unknown, but chestnut horses are underrepresented in our breeding population due to the color-consciousness of our breed fanciers.

Recently, we have a new tool - a genetic test for the silver dapple gene. This may aid breeders who would like to be sure that their "normal-eyed" chestnut horse is a carrier of ASD, or perhaps a horse whose color is uncertain due to the presence of other color genes. If the horse is shown by a DNA test to carry the silver dapple gene, a breeder is well advised to assume that the ASD gene is also present.

What if you want to breed two Silver Dapple horses together? Breeders often do this, risking the small chance of getting a foal with affected vision, because the match is desirable for other reasons. This is the breeding that produces the most issues. In the chart below, the heterozygous, "cyst-only" horse is symbolized as "Aa" - one dominant ASD gene. The homozygous "ASD" horse is symbolized as "AA", and the genetically normal horse that does not carry any ASD gene is "aa".

The probabilities for eye status of the foal with each Aa- to-Aa mating are as follows:

- 25% aa (no ASD gene and no silver dapple gene - black, bay, chestnut)
- 50% Aa (a single ASD and a single silver dapple gene - chocolate, red chocolate, chestnut)
- 25% AA (two ASD and two silver dapple genes- light chocolate or red chocolate, chestnut)

It is only this last category which is at risk for affected vision 10% of this last group will have the subluxated lens. This is about a 2.5% probability altogether of eye problems as a result of this Aa to Aa breeding.

Whether or not to breed two Aa horses together must be an individual decision, based on the breeder's ability to absorb and deal with the very slight chance of producing an affected foal. In terms of the breed itself, we are advised by experts at the University of Kentucky that the Aa to Aa mating is one that should continue, so that we can continue to produce the 25% of completely normal "aa" horses the breed needs for its breeding stock. Also, as we have a large number of chocolate, "Aa" horses in our breed, we are advised that to discontinue breeding these horses to each other would eliminate sufficient horses from the gene pool that it could potentially leave us open for other, far worse, genetic problems developing within the breed.

Chocolate will always be popular. The abnormal ASD gene will never be eliminated from our horse population as long as we desire to produce this color. But we have learned to limit the production and breeding of the homozygous ASD horse. When the AA horse is bred, it should be bred only to the "aa" horse that does not carry any silver dapple or ASD gene - the true black or bay horse, or the chestnut horse that is known to not be a carrier.

Again, if you are considering purchasing an ASD "AA" foal or horse for pleasure riding, there should be no problem with its vision if a veterinary exam reveals that there is no subluxation of the lens.

Finally, an interesting footnote, some breeders and geneticists believe that the silver dapple color is genetically linked not only to the ASD gene, but also to some of the very characteristic personality traits that have made the Mountain Horses so popular! If this is true, it may be another reason for continuing to breed for the single-gene chocolate horse.

We are happy to answer questions about ASD, color genetics, the status of our stallions, and the probabilities of color and ASD status of offspring. Just e-mail us at morgan@triple-s-ranch.com, or call 541-831-1625.

Summary.

If you are purchasing a pleasure riding horse only, and will not be breeding, all you really need to know in terms of ASD eye status is that there is nothing wrong with the vision of your prospect at the time you are buying it, and it does not have detached or out-of-place lenses. None of the other characteristics of the ASD syndrome will affect vision or will change over time. If your horse is chocolate, red chocolate or chestnut, get a competent eye exam as part of your prepurchase exam.

If you will be breeding your horse, and do not want to take any chance of having an ASD foal, make sure that one parent has no silver dapple or ASD gene - the true bay or black horse, or the chestnut that does not carry a silver dapple gene. If you want to breed an Aa

horse to another Aa horse, you need to accept the risk, about a 2.5% chance, of producing a foal with affected vision.

**It was initially assumed that vision difficulties would be presented by megalocornia ("pop-eye"), a common characteristic seen in the AA horse, by making the horse near-sighted, but the tests showed that the horses' eyes had physically accommodated to this difference by the time the horse was one year old, resulting in no loss of focus of light rays on the retina.

Also, many AA horses show artifacts in the lens in the way of streaks or starburst patterns of cloudy matter, called nuclear or temporal cataracts. A vet not familiar with the ASD syndrome may be concerned about these. While these can look significant when viewed from the outside, the horse's brain appears to see past these imperfections in the same way it looks through the mesh of a fly mask, or in the way you would look through your splayed fingers put up against your face, with the result that vision is still functionally adequate.