

COLLIE KALEIDOSCOPE

Variations in Collie Color...

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Introduction:

Armed with new research information to read suggested to me by my friend Dr. Leigh Anne Clark, we'll be doing double duty in this article. It will be both a refresher on what we published five years ago in the colliesonline.com (<http://www.colliesonline.com/may2009/coat-of-many-colors.php>) and reprinted in the Bulletin on the mapping of the merle gene and the basics of collie coat color genetics and the latest research on white coats, harlequin pattern and normal grey (Maltese). **Dr. Clark's research is funded in part by a generous grant from the Collie Health Foundation (www.chf.org).**

Let's begin with an overview of "The Coat of Many Colors" written in collaboration with Leigh Anne and another friend and CCA member Dr. Mike Vaughan published on colliesonline.com and reprinted in the CCA Bulletin. At that time, Dr. Clark et.al had mapped the merle gene in all breeds adding significantly to knowledge of merle genetics. In 2005 Dr. Clark and her colleagues successfully mapped the merle gene in all breeds and published their findings in The National Academy of Sciences PNAS journal in January 2006.

Breeders, fanciers and all collie lovers owe it to our breed to study and understand all genetic information, since even genes that seem unrelated to health and soundness, often *are* related. In some cases color and color modifier genes, as well as, color pattern genes are influential because of their proximity to other genes on strands of DNA and affect these close neighbor genes for good or ill.

Collie Colors:

The two base color genes in collies are brown (sable) and black (tri color). Most of us know that in our breed brown is dominant and black recessive. A puppy inherits one color gene from each parent. Therefore, if a collie inherits a dominant brown gene from each parent, he/she is a "pure for sable" (homozygous for brown). If on the other hand, a recessive black gene is contributed by each parent, the collie will be a tri-color (homozygous for black). Some collies inherit a brown gene from one parent and a black one from the other making them tri-factored sables

(heterozygous genetically although still brown in appearance).

Dr. Vaughan explains the process that delivers color to a collie's coat:

Specialized skin cells produce pigments called melanins that come in black or brown. These are not free dyes, dissolved in the cell water but instead are solid pigment particles built up in tiny bodies called melanosomes. The skin pigment cells pass these melanosomes into hair follicle cells which produce the hair strand.

If a hair follicle does not receive melanosomes, the hair strand it forms is un-pigmented, white, as in the majority coat of a white Collie or the neck region of colored collies (from the Irish pattern gene). If all the melanosomes that a growing hair strand receives contain black melanin, the hair strand itself will be black, as in the black areas of a tricolor collie. And if the melanosomes the follicle receives from the pigment cells are all brown, the hair strand will be brown, as in the brown coat of a pure sable or tan points on a tricolor.

This process continues throughout the life of the dog, and with each fresh coat may alter the amount of pigment. An example is the mahogany sable coat of tri-factored sables.

When puppies many look much like pure sables, and almost all the developing hair in the first coat is brown. However, as the dog ages, some black melanosomes are put into the growing hair strands, so the coat develops a deepening dark appearance.

Merle Genetics:

Unlike the collies' two base color genes, the merle gene is not a color gene; in fact, it produces no pigment at all. There is no color gene in collies for blue coat color only for black coat and brown coat. The merle gene turns some parts of a black coat to grey by not allowing the black color to come through completely. The merle gene has exactly the same effect on a collie with two brown genes, turning some parts of a brown coat lighter brown by allowing less color to come through.

Of course, when a collie has one brown color gene and one black color gene, it is still a sable, just a darker brown one. This darkening phenomenon is called epistasis (an interaction between genes). Epistasis means that the black recessive gene in a tri-factored sable is suppressed by the dominant brown gene that we call sable. The brown gene's phenotype (appearance) is expressed, so it is epistatic while the suppressed black gene is hypostatic, and simply darkens the brown. The resulting collie is still a sable. So when a merle gene with no pigment acts upon the dark brown coat, lighter shades of brown come through.

We'll need a few definitions to understand how the merle gene works. For example, SILV is a gene that produces proteins, the melanosomes that produce pigment. Another term, SINE, means short, interspersed mobile elements that insert themselves in SILV. Merles have the SINE needed to help create merling. However, SINE alone will not cause the collie in question to present visually as a merle. SINE must also have oligo(dA) also called poly(A) which creates a "tail"

within SINE. So the presence of the SINE element is necessary but not sufficient to generate the merle phenotype; a poly(A) tail in the element is also required. The length of the tail controls the degree of merling seen in a collie. Collies with longer poly(A) tails present as merles, but the tail length controls the degree of merling and is responsible for the random degree of mottling as well as the randomness of eye color.

It turns out that mutations occur in this string of “A’s” quite often as cells divide, decreasing the length of the stretch of “A’s”. The result is that as the body of a tricolor (with merle) puppy develops, (first as an embryo, and later, after birth) with clones of skin cells with mutations in the merle gene DNA, reversing the merle mutation allowing normal melanosomes to be made again. So this clone of skin cells, with normal melanosomes, loaded with black pigment, will produce a spot of hair that is black or grey producing a blue merle. When you next look at a blue merle Collie, look at the spots of black and grey on its body which give it its beautiful coat and think that you are literally looking at mutations. The merle gene is still there in those spots; it has just been turned back “on” in them, by the second mutation decreasing the string of “A’s”. One mutation is canceling the effect of another.

A collie with little or no poly(A) will present as a cryptic (a merle who is not physically distinguishable as such or who mainly looks tri-color or sable with only slight hints of merling). Imagine my surprise when Leigh Anne called one day a few years ago to say that my tri rough Wiggy has a merle gene. She has not a trace of merling anywhere. She was bred only once to another tri rough and had 6 tri color rough puppies. I asked for a recheck! No mistake, she’s a true cryptic. For Wiggy, now age 14, the findings are moot but startling.



Merle Gene Influence on Eye Color & Ear & Eye Abnormalities:

Merles, both blue (pure for black) and sable (pure for sable or mahogany sable), have random chances at pigmentation in eye color. The same SINE/poly(A) genetic information that randomly puts merling on these collies also allows for the following: both eyes brown, both eyes blue, one

eye of each color, partial blue & brown in one or both eyes and blue flecks in brown eyes. So while eye size, shape and set are genetically controllable through selective breeding, the presence of blue in the eyes of merles is not.

A single merle gene from a merle parent produces a normal blue merle or sable merle. However, when each parent contributes a merle gene to offspring, problems arise. Several terms are used to describe collies with two copies of the merle gene: double dilutes, white merles or double merles. Double merles usually have at least some vision and/or hearing impairment. However, these defects are not passed to the double merles offspring provided they are only bred to solid color collies – tri color or sable – with no merle gene. The resulting offspring will all be normal merles.

The unwanted effects of doubling up on merle in one individual need explanation. Because the merle gene is apparently in close proximity on the strand containing genes for normal eye and ear development, doubling the merle gene reduces pigmentation necessary for proper development. Melanocytes are pigment-producing cells present in many tissues, including the epidermis, hair follicle, inner ear, and choroid of the eye. This explains why collies who receive a copy of the merle gene from each parent are double or homozygous merles that may have varying degrees of vision and auditory abnormalities.

Contrary to the claims that double merles have other health problems due to having two merle genes no scientific support exists as far as I can determine. My own experience owning 4 double merle collies over the past 40 years also does not support infertility, shortened life span or more health problems than single merle or non-merle collies.



Merle Gene Modifier - Maltese:

Merle gene modifiers that breeders call Maltese and harlequin are now better understood and can be viewed separately from disease genes that may alter coat color and pattern for completely different reasons. Starting on page 103 of the Bulletin's June issue you'll find a report covering Dr. Clark and her colleagues' mapping of the collie genome and discoveries about the genetics of autoimmune diseases such as dermatomyositis (DMS). While the report's main themes are DMS genetics and genome mapping, a side product of the research, was enlightening.

Dr. Clark was able to sort out the mystery of normal grey Collie coat color (no relationship to lethal greys). In testing the DNA from several different normal greys, she discovered that these Collies are actually blue merles with an additional mutation that eliminates the visible spots and produces a nearly solid grey coat. Normal blue merle littermates to a normal grey have no more chance of producing normal greys than any other blue merle. Also, no health related problems are associated with the mutated merle gene that produces solid greys than with any other Collies. However, grey is not a color recognized in the Collie Standard; therefore, breeding collies with this mutation is undesirable.

Dr. Clark cautioned breeders against euthanizing these normal greys because they will be the same as any other Collie without the mutation, except for their coat color. However, these normal greys when bred reproduce their grey color. If breeders do not want to propagate this color, they should not breed normal greys.



Merle Gene Modifier – Harlequin:

The subject of harlequin coat pattern in blue merle collies and shelties has been an interest of Dr. Clark's for several years. Harlequin is not a color gene but rather a coat pattern. She believes harlequin in some collies results from a change in the DNA within or close to the merle gene. It is not the same gene that produces harlequin coats in Great Danes. In Danes the harlequin is predominantly a white dog with black torn patches. In collies and shelties a harlequin will usually have a white background with black patches as well, but may also have merling and tan points.

Dr. Clark and her colleagues found and mapped the harlequin gene in Danes a number of years ago. Harlequin in Danes is located on a separate chromosome than the merle gene. No other harlequin dogs outside of the Dane breed have this same mutation. Dr. Clark discovered a collie kennel in Finland with harlequin pattern collies unlike the Dane gene and unlike the harlequin seen in most American collies. The reason for this uniquely dissimilar pattern is not known at this time.

Most harlequin merles in breeds other than Danes reproduce a preponderance of harlequin blue merles, but can occasionally produce non-harlequin blue merles. Contrary to what some mistakenly believe, harlequin patterned Collies are not defective, and no additional health problems have been associated with the pattern.



New Research on White Coat Color:

White coat color in all dogs, including color headed white collies, is currently under investigation. Research published 4 months ago by Korberg, et al sheds new light on white coats in a number of breeds, including collies. White coat in collies and shelties according to this new study is probably caused by alleles for piebald. There are two (2) alleles of one white spotting gene in collies; one is “fixed” (Irish pattern) and one “segregates” (piebald). MITF is name of the gene for both. Piebald appears to be the allele responsible for color-headed whites.

Irish spotting is characterized by modest white spotting and is responsible for white collars, belly coat, legs & feet, and tail tip. All collies are “fixed” for this Irish pattern allele, meaning all collies have two copies. Piebald causes limited to extensive white spotting. This piebald allele “segregates” in collies, meaning that some collies have no copies, some have one copy and some have two copies. One copy of the allele produces white factored but otherwise solid colored collies. Two copies create a color headed white.

Not strictly on topic, but good white coat information anyway. I have sometimes heard the myth that dogs with white coats don’t make good herding dogs. I have always disagreed. Here’s a bit of herding trivia that supports the position that white dogs are not handicapped as herding dogs because of their color.

Columella, Roman agriculture authority, wrote in the first century AD that shepherds preferred a white sheep dog “because it is unlike a wild beast, and sometimes a plain means of distinction is required in the dogs when one is driving off wolves in the obscurity of early morning or even at dusk.” This 2,000 year old observation counters the belief that white dogs don’t make good herding candidates because they are the same color as most sheep, and so do not get respect from the stock.

Dr. Clark and her colleagues are studying more pieces of the collie genetic puzzle and answers and solutions will be pieced together in the future. We owe it to our collies to explore and understand what science tells us so that we can eventually view and appreciate the completed picture.

As always, many thanks to Dr. Leigh Anne Clark for her assistance with this article! Special thanks to the Collie Health Foundation for their generous, ongoing funding of Dr. Clark’s research.

To read the article “The Coat of Many Colors” see one the two links below.

<http://www.colliesonline.com/may2009/coat-of-many-colors.php>

http://www.colliehealth.org/Summer_2014_Newsletter.pdf (see page 11)

From the Breed Education Committee: The Collie Club of America's mission is safeguarding and improving the health and well-being of the breed. Double dilute merles carry a high risk of being blind and/or deaf or may not have eyes. As such, the Collie Club of America does not recommend breedings that would produce offspring having high risk of health defects.