YOUR GUIDE TO CANINE COLOR AND TRAIT GENETIC TESTING

Coat color in canines is determined by the interaction of multiple genes, each responsible for a specific inherited trait or characteristic. Every dog has two copies of each gene; one copy inherited from each parent. Each gene may have multiple versions called **alleles,** which may result in a different physical appearance in the individual. The combination of these two copies of a gene is called the dog's **genotype** and is displayed with two letters that represent the copies of the gene received from each parent. If the two gene copies are each a different allele, the letters (or capitalization of the letters) representing each copy of the gene will differ. The relationships between the alleles that a dog inherits will determine what the dog may look like.

Inheritance Patterns: Dominant vs. Recessive

Traits are inherited in two general ways; dominant or recessive. **Dominant** alleles are those that only require one copy to be present in order to produce the associated trait and are most commonly notated by using a capital letter. For example, the merle coat pattern seen in many breeds is dominant. Traditional merle dogs have a genotype of '**m/M'** at the M locus which indicates one copy of the recessive (non-merle) gene allele (**m**) and one copy of the dominant, merle-associated allele **(M**).

Recessive alleles are those that require two copies to be present (one inherited from each parent) in order to produce the associated trait and are most commonly notated by using a lowercase letter. For example, the light colors of dogs (varying from white to red) are recessive (see below). These dogs have a genotype of '**e/e**' at the E locus indicating that two copies of the recessive allele were inherited.

Dogs that have inherited one copy of a dominant allele and one copy of a recessive allele for a trait (e.g. **E/e** at the E locus) are considered 'carriers' of the recessive trait and will not display that trait, but can produce puppies with that recessive trait if bred to another dog with at least one copy of the same recessive allele (in this example, **E/e** or **e/e** dogs).

What color tests should I order for my dog?

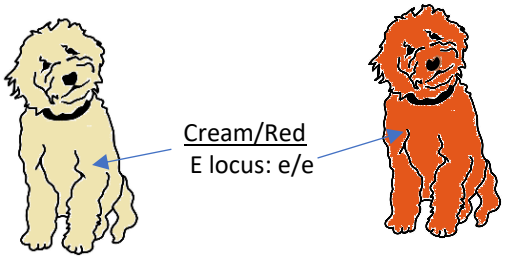
Genetic tests for canine colors and traits are given letter names followed by the word 'locus' (plural 'loci') which indicates the specific location in the genome being tested. Below is a comprehensive list of color and trait tests for which the genetic alleles are known. It is important to note that either due to historical reasons or because of a true difference in appearance, some breeds use terminology that differs from the majority of breeds. For example, dogs that are 'red' are most commonly '**e/e**'. However, in some breeds, dogs that are $\mathbf{A^y/A^y}$ or **b/b** are called red while '**e/e**' dogs in these breeds are given a different color name (e.g. lemon, cream, yellow, clear red). Refer to your breed's standard for assistance with these less commonly used terms.

Base Colors- E, B, K, and A loci

<u>These four loci determine the foundation of your dog's coat color:</u>

<u>E locus:</u> *White to Red*

Dogs that inherited two copies of the recessive allele at the E locus (**e/e**) cannot produce the pigment associated with black or brown hair and will be a color that can vary from chalk white to a deep red color. Dogs inheriting at least one copy of the dominant E allele (**E/e** or **E/E**) will still be black or brown. At this time, the genetic variants that dictate which specific color hue an '**e/e**' dog might express are mostly unknown.



<u>Cream/Red</u>
E locus: e/e

- *Note: Because white, cream, yellow, apricot, caramel, and red are recessive, dogs of these colors do not need to be tested at the E locus because they must be '**e/e**'.*

<u>B locus:</u> *Brown, Chocolate, Liver, "Red" (in some breeds)*

Dogs that inherited two copies of the recessive allele at the B locus (**b/b**) will be brown. Dogs with at least one copy of the dominant B allele (**B/b** or **B/B**) will be black. Though dogs that are '**e/e**' at the E locus cannot produce black or brown hair, the B locus still determines the color of the nose and foot pads in these dogs.

- *Note: In some breeds (e.g. Australian Shepherd and Border Collie) dogs that are '**b/b**' are called red. Because brown is recessive, dogs that are brown (or have brown/liver noses) do not need to be tested at the B locus because they must be '**b/b**'.*

<u>Brown/Chocolate</u>
E locus: E/E or E/e -AND- B locus: b/b

K Locus: *On/Off switch for the A locus*

Dogs that inherited two copies of the recessive allele at the K locus (k^y/k^y) will express (or 'turn on') the alleles that they have inherited at their A locus (see below), allowing for a variety of colors/patterns. Dogs with one recessive allele (K^B/k^y) will not express their A locus but can produce puppies that can express their A locus if bred with a dog that also carries 'k^y'. Inheriting two copies of the dominant allele at the K locus (K^B/K^B) completely blocks a dog's ability to produce puppies that express their A locus.

- *Note: Because the 'on signal' for the A locus is recessive, dogs that are sable/fawn, wild sable, tricolor, or have tan points do not need to be tested at the K locus because they must be '**k^y/k^y**'.*

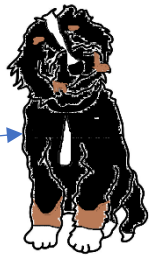
Sable
K locus: k^y/k^y
-AND-
A locus: A^Y/A^Y *or* A^Y/a^w *or* A^Y/a^t *or* A^Y/a

A Locus: *Sable/Fawn, "Red" (in some breeds), Wild Boar (Wolf) Sable, Tricolor, Phantom, Tan Points*

As stated above, expression of the A locus is dependent upon a dog also inheriting a 'k^y/k^y' genotype at the K locus. The A locus has 4 different alleles. In their order of dominance (first being most dominant) they are A^y (sable/fawn/'red' in some breeds), a^w (wild sable/wild boar), a^t (phantom/tan points/tricolor), **a** (solid/bicolor). This means that if two different A locus alleles are inherited in the same dog, the most dominant one will be expressed (e.g. Dogs that are A^y/a^t will appear sable because 'A^y' is dominant to 'a^t'. However, a dog that is a^t/a will have tan points because 'a^t' is dominant to '**a**').

- *Note: Whether a dog appears sable (black hair tips) or fawn (no black hair tips) when they have inherited an '**A^y**' varies by breed and sometimes from dog to dog. In sable, wild boar, and tan pointed dogs, the darkest coat color expressed is controlled by the B locus. **B/B** or **B/b** result in traditional sable (**A^y**) with **black** hair tips or a **black** and tan pattern (**a^t**). '**b/b**' results in chocolate sable (**A^y**) with **brown** hair tips or a **brown** and tan pattern (**a^t**). In some breeds (e.g. Australian Cattle dog, Basenji, and Dachshund), dogs that have one or two copies of **A^y**, are called "red" rather than sable. Also, in some breeds, dogs that are **A^y/a^t** may have a different appearance than dogs that are **A^y/A^y** (e.g. Shiba Inus are 'red' if **A^y/A^y** and 'sesame' if **A^y/a^t**).*

Tricolor:
K locus: k^y/k^y
-AND-
A locus: a^t/a^t *or* a^t/a



Color and Hair Trait Modifiers- D, E^m, M, S, IC, and Cu loci

These additional loci modify the base coat color and traits of your dog:

D Locus: *Coat Color Dilution*

Dogs with two copies of the recessive allele at the D locus (**d/d**) will be born with a diluted version of the coat color determined by all of the other color loci. In dilute dogs, black becomes blue/dark gray/charcoal, brown becomes silver/gray, and cream/apricot becomes champagne. Dogs with at least one copy of the dominant allele (**D/d** or **D/D**) will not display a diluted coat color at birth, but some breeds and mixes of those breeds (e.g. Poodle, Old English Sheepdog) may still undergo 'progressive graying' or 'fading' which often begins in the first two months of life. The genetic variant associated with progressive graying/fading is unknown.

- *Note: Coat color dilution associated with the D locus is not the cause of 'progressive graying' or 'fading' coat color. Although their coats may look similar, the nose and footpads of dilute dogs are also a diluted color while those of graying/fading dogs will retain the black or liver color present at birth. Since coat color dilution is recessive, dogs born with a dilute coat do not need to be tested at the D locus because they must be '**d/d**'.*

Dilute Brown- Silver/Gray at Birth
D locus: d/d (Note the dilute nose color)
-AND- B locus: b/b

E^m Locus: *Melanistic (Black or Brown) Mask*

Dogs with at least one copy of the dominant allele at the E locus (**E^m/N** or **E^m/E^m**) will display a black or brown mask (color controlled by the B locus) which can be seen on the muzzle of dogs with a sable or fawn coat. Dogs inheriting two copies of the recessive allele (**N/N**) will not develop this mask. **Note: This mask is not visible on dogs that have two copies of the recessive allele at the E locus (**e/e**) or in dogs that already have a solid black, brown, or white face.*

M Locus: *Merle, Dapple*

As stated above, merle is a dominant trait requiring dogs to inherit only one copy of the merle associated allele (**m/M**) to display this pattern. Merle is a pattern of randomly distributed, fully pigmented black or brown regions (controlled by the B locus) separated by areas of white or diluted coat color. Merle dogs have a DNA mutation with a piece of DNA known as an 'insertion' in a gene associated with pigment distribution (*PMEL*). This insertion is naturally unstable in its structure and may change size from generation to generation. Dogs who inherit the merle insertion can have wide variation in coat characteristics caused by this size variation. Dogs with the smallest size insertions often fail to show any indication of merle patterning and are known as 'cryptic merles'. Those with an intermediate size insertion tend to fall into the more classic merle appearance with diluted coat color between the fully pigmented regions. Dogs with the largest size insertions often display a 'harlequin' appearance marked by white hair between pigmented areas and/or a 'patchwork' appearance in which dogs develop multiple shades of color within their pigmented regions. Dogs that are **e/e** AND merle (**m/M**) will not appear merle since they can't produce black or brown pigment.

- *Note: M locus testing is critical prior to breeding a merle dog because dogs that inherit two copies of the merle allele (**M/M**) often display a mostly white coat and suffer from hearing and/or vision deficits. In order to avoid this situation, it is not recommended to breed two dogs together if they both have one copy of the merle allele (**m/M**) because roughly 25% of the puppies will be born as 'double merles' and potentially display disease characteristics. Instead, it would be preferable to breed any merle dog with a dog that has been shown by genetic testing to have not inherited the merle allele (**m/m**). In addition, the B locus dictates which type of merle pattern seen. **B/b** or **B/B** produces a 'blue' (black) merle while **b/b** produces a 'red' (chocolate) merle.*

S Locus: *Parti-Color, Piebald*

Dogs inheriting two copies of the recessive allele at the S locus (**s^p/s^p**) will be parti-colored (called piebald in some breeds) and identified by random distribution of white around the body. The colored portions of parti-colored dogs are determined by all of the other coat color loci. Dogs inheriting at least one copy of the dominant allele (**S/s^p** or **S/S**) will not be parti. With rare exceptions, parti-colored dogs are usually greater than 50% white.

Parti
S locus: s^p/s^p

- *Note: S Locus is not responsible for the presence of 'Irish White' spotting seen in many dogs including tricolor dogs or abstract poodles. Irish white is identified as white on the chest (sometimes around the collar), between the eyes, and on the tips of the toes and tail. The genetic variants associated with Irish white are unknown. However, dogs that have inherited one copy of the parti allele (**S/s^p**) AND are presumably also Irish white, often display expansion of the Irish white on the chest, head, toes and tail. In addition, there are other, unknown genetic factors that can produce white in dogs. Because of this, the white on dogs that are **S/Sp** can be quite variable.*

IC Locus: *Improper Coat and Furnishings*

Dogs inheriting one or two copies of the dominant 'furnishings' allele (**F**) at the IC locus (**F/IC** or **F/F**) will display long hair on the face (including the moustache and eyebrows) consistent with the generally preferred appearance of many breeds. Dogs inheriting two copies of the recessive 'improper coat' allele (**IC/IC**), will have short hair on the face like that seen on the Labrador retriever and other breeds (only referred to as improper coat in breeds in which furnishings are a standard).

- *Note: Dogs inheriting two copies of the improper coat allele (**IC/IC**) differ from others primarily by the length of hair on the face. The hair coat of these dogs is normal in all other ways. However, dogs with one or two copies of the 'improper coat' allele (**F/IC** or **IC/IC**) may tend to shed more than dogs with two copies of the 'furnishings' allele (**F/F**).*

Cu Locus: Curl

Curl is inherited in a 'semi-dominant' manner in dogs. This means that dogs inheriting one copy of the 'curl' allele (**Cu/Cu^c**) will display an intermediate, 'wavy' hair coat which falls between a straight coat (caused by two copies of the 'straight-haired' allele; **Cu/Cu**) and a coat with tight curls (caused by two copies of the 'curl' allele; **Cu^c/Cu^c**). To produce pups with wavy or curly coats, dogs that carry one copy of the straight-haired allele (wavy; **Cu/Cu^c**) should be bred to curly coated dogs (**Cu^c/Cu^c**).

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