

# MERLE GENE



## Coat Color Loci in Dogs

Gene Locus	Gene Name
A	Agouti
B	Brown/Black
Br	Brindle
C	Albino
D	Dilution
E	Extension
F	Flecking
G	Graying
Grp	Gray Points
H	Harlequin
I	Intense
Ma	Mask
M	Merle
P	Pink Eye Dilution
Pp	Powder Puff
S	Piebald/Spotting
Sg	Slate grey
T	Ticking
Tw	Tweed
W	White

## Breeds with Merle Coat Pattern

Shetland Sheepdogs <sup>a</sup>  
 Collie <sup>a</sup>  
 Great Danes <sup>b</sup>  
 Cardigan Welsh Corgi <sup>a</sup>  
 Australian Shepherds <sup>a</sup>  
 Border Collie <sup>a</sup>  
 Chihuahua  
 Cocker Spaniel <sup>b</sup>  
 Dachshund <sup>a</sup>  
 Catahoula Leopard Dog  
 Norwegian Hound  
 Pyrenean Shepherd  
 Pomeranian  
 Beauceron Sheepdog  
 Pit Bull

<sup>a</sup> Acceptable coat color for show purposes

<sup>b</sup> Unacceptable coat color for show purposes

## Genetics of Canine Coat Color

Canine coat color is determined by the expression of a specific combination of genes. A gene, the basic unit of heredity, is comprised of a unique sequence of DNA and directs the production of a specific protein. Proteins are required for the structure, function and regulation of the body's cells, tissues, and organs. Genes are located within chromosomes. Dogs have two sets of 39 chromosomes in every cell, one set inherited from each parent. The location of each gene within a chromosome is referred to as its locus. While there is more than 99% DNA sequence similarity between dogs, variations in DNA sequence do occur in a small number of genes. Different forms of the same gene are called alleles. Dogs can have two identical or two different alleles for a particular gene. If both alleles are identical, then the dog is said to be homozygous at that gene; if both alleles are different, then the dog is said to be heterozygous at that gene. The genotype of an animal is its genetic identity, as identified by the alleles it carries; while the phenotype, or appearance, is the expression of those alleles. Coat color in dogs is usually controlled by a set of genes. These include the color genes, genes that affect the pigment color of hairs, and the pattern genes, those that affect the distribution of a particular color. At least 20 genes have been identified that affect coat color in dogs.

## Merle Coat Color Patterning

The merle coat color is characterized by patches of dilute pigment in combination with areas of full pigmentation. Therefore, the merle gene acts to lighten whatever coat color would otherwise be expressed. However, unlike other dilution genes, the lightening effect is not spread evenly over the coat, but is expressed as patches of diluted color scattered over the dog's body. If the basic color of the dog is black, the effect of the merle gene is a soft gray, often referred to as "blue". If the basic color of the dog is red, the effect of the merle gene is a pale red. The merle coat pattern is characteristic of a number of breeds recognized by the American Kennel Club, including the Shetland Sheepdog, Collie, Border Collie, Dachshund, Australian Shepherd, and Cardigan Welsh Corgi.

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### Genetic Inheritance of the Merle Gene

It is only recently that investigators at the Texas A&M University (reference: PNAS, 2006, 103(5):1376-81) discovered a mutation in the dog *SILV* gene and found it to be responsible for the merle coat color patterning in dogs. The merle gene (*M*) is inherited in an autosomal fashion. In other words, the trait is not linked to gender and can be passed on from either the mother or the father. The gene is incompletely dominant, or a gene that has intermediate expression. A heterozygous dog, carrying only one copy of the merle gene (*Mm*), expresses the characteristic diluted coat color pattern. A non-merle dog (*mm*) is normal in color, while a homozygous double-merle (*MM*) is predominantly white. Punnett squares can be used to determine the expected coat color of offspring when breeding dogs of known genotype (i.e. coat color genes have been identified). In the example illustrated, a non-merle dog (*mm*), indicated in the vertical column, bred to a heterozygous merle (*Mm*), indicated in the horizontal column, will give rise to offspring with an expected frequency of 50% merle (*Mm*) and 50% non-merle (*mm*). Dogs that carry the merle gene but do not show the characteristic merle phenotype, are known as cryptic merles. These dogs may give rise to merle offspring. It is suspected that the DNA sequence of the merle allele in the cryptic is shorter than the allele expressed in the typical merle dog. The harlequin coat color pattern in Great Danes is produced through the interaction of the merle locus and the harlequin (*H*) gene. In harlequin Danes, the merle background color is diluted to nearly white with fully pigmented black patches.

		Father		
		M	m	
Mother	m	Mm	mm	Offspring
	m	Mm	mm	



Tri-colored non-merle Shetland Sheepdog (*mm*)



Blue merle Shetland Sheepdog (*Mm*)



Double merle Shetland Sheepdog (*MM*)

Clarke et al., 2006, PNAS 103(5):1376-81

### Health Concerns

**Increased Intraocular pressure:** excessive pressure created in the eye.

**Ametropia:** vision impairment due to a refractive error such that images fail to focus upon the retina.

**Microphthalmia:** a smaller than normal eye due to a defect occurring early in development. Affected dogs may have prominent third eyelids. Other eye defects are common in animals with this condition, including defects of the cornea, anterior chamber, lens and retina.

**Coloboma** – a defect in ocular tissue; a cleft or missing portion of components of the eye, most commonly affecting the iris.

### Health Problems Associated with the Merle Allele

Both heterozygous merle (*Mm*) and homozygous double merle (*MM*) dogs may exhibit auditory and ophthalmic abnormalities including mild to severe deafness, increased intraocular pressure, ametropia, microphthalmia and colobomas. The double merle genotype may also be associated with abnormalities of skeletal, cardiac and reproductive systems.

### Genetic Testing for the Merle Gene

With the recent discovery of the merle gene, a genetic test is now available that allows for the identification of the merle allele. This technology is patent pending (U.S. Serial # 60/708,589) and available exclusively thru GenMARK, the DNA technology service of VITA-TECH Laboratories LLC. By testing dogs for this genetic trait, it is possible to:

- allow identification of merle dogs to prevent undesirable merle to merle breeding
- classify harlequin Danes as single or double merle
- identify cryptic merles

**For more information, please contact GenMARK.**

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