The coat colour genes in the Great Dane panel are A, D, K, H & S

There are a number of genes which interact to dictate the coat colour of a dog – the genes tested in this panel are the main ones which dictate the coat colour in Great Dane.

Colours which dogs carry are often hidden, but become apparent in their offspring. It is in identifying these hidden genes that genetic testing has value. Genetic testing can be used to identify some of the genes that a dog carries, and to help identify the possible outcome when dogs are bred.

As with all genetic traits, every animal inherits one copy of each locus from each of its parents. So each gene test gives two results for each dog – one has come from his father and the other his mother. These are usually written one after the other e.g. at at.

Further details about each of the gene tests follows below:

The A Gene:

The Agouti Signaling Protein (*ASIP)* gene interacts with the MC1r gene to control red and black pigment switching in most mammals including dogs. Dog coat colour is further complicated by the interaction of other genes that restrict agouti expression such as the dominant black gene – Beta-Defensin 103. There are 4 known alleles (variants) of agouti listed here with corresponding colour pattern in order of dominance: **fawn/ sable (a^y)** yellow to red with some dorsal black tipped hairs, **wild sable (a^w)** banded hairs of yellow and black as in seen in wolves and coyotes, **black-and-tan (a^t)** black dorsal hairs with tan hair on cheeks, eyebrows and undersides, and **recessive black (a)** all black as seen in some herding dogs. The Eurasier dog breed has all 4 alleles while some breeds are fixed for only one variant such as the Norwegian Elkhound for wild sable and the Beagle for black-and-tan. For many breeds, there are 2 or 3 alleles possible and it may be advantageous for breeders to predict the possible colours of offspring resulting from specific matings. The agouti test is also useful to help determine the colour of dogs that have white patterns that may obscure the distribution of the coloured pigment.

This test will help determine possible coat colour outcomes from specific matings.

Results are reported as:

a^y / a^y	Homozygous for fawn/ sable.
a^y / a^w	Dog is fawn and carries wild sable.
a^y / a^t	Dog is fawn and carries black-and-tan.
a^y / a	Dog is fawn and carries recessive black.
a^w/ a^w	Homozygous for wild-sable.
a^w/ a^t	Dog is wild-sable and carries black-and-tan.
a^w/ a	Dog is wild-sable and carries recessive black.
a^t/ a^t	Homozygous for black-and-tan.

a^t/ a	Dog is black-and-tan and carries recessive black.
a/a	Homozygous for recessive black.

The D Gene

A recessive mutation in the melanophilin (MLPH) gene was identified as the cause of colour dilution phenotypes in the dog. Two alleles (variants) are described: the dominant full colour (D) and the recessive dilute (d). Two copies of dilute are needed to lighten black pigment to grey (often called blue) and red pigment to cream (also called buff). A diagnostic DNA test identifies the specific variants of the MLPH gene.

NOTE: Another as yet unidentified mutation causing colour dilution is known to occur in some breeds such as Doberman Pinscher, French Bulldog, Italian Greyhound, Chow Chow and Shar-Pei. In these breeds, and likely others as well, some dogs may carry both the known and unknown dilution mutations and present a dilute phenotype.

Results from the DILUTE test are reported as:

D/D	Full colour, no dilute gene present
D/d	Full colour, carries 1 copy of the dilute gene
d/d	Dilute, 2 copies of the dilute gene

The K Gene

The wide variety of coat colours in mammals is achieved by the production of two pigments, eumelanin (black) and pheomelanin (red or yellow). In most mammals, the switching between these 2 pigments is controlled by MC1R and Agouti genes. In dogs, original coat colour research of pedigrees suggested that a third gene, named **Dominant Black** (**K locus**), was involved. This gene produces dominant black vs. brindle vs. fawn colours in breeds such as Great Danes, Pugs and Greyhounds among others. Researchers recently have discovered that dominant black is due to a mutation in a Beta-defensin gene (*CBD103).*

This test can assist owners of black dogs to determine if their dogs are homozygous for dominant black or if they carry brindle or fawn.

Results are reported as:

K/K	2 copies of dominant black are present
K/N*	1 copy of dominant black is present
N/N	Dog does not have the dominant black mutation

* This result is sometimes associated with the brindle pattern.

The H Gene

The interaction of many genes is involved in the variety of colours and patterns of dog coat colour. Harlequin is a pattern seen in Great Danes resulting from the complex interaction of the Merle (PMEL17) and Harlequin (PSMB7) genes on black pigment. The dominant Merle gene, by itself produces dark spots on a dilute background on eumelanistic dogs. If a merle dog also inherits 1 copy of the Harlequin gene, the dark spots increase in size and the background pigment is removed altogether. Dogs that are not merle, or only have red pigment, cannot express the Harlequin gene. Two copies of Harlequin have not been observed and is presumed to be embryonic lethal, thus all Harlequin patterned dogs have only 1 copy of the mutation.

Results from the HARLEQUIN test are reported as:

N/N No copies of Harlequin mutation are present.
N/H 1 copy of the Harlequin mutation is present. If the dog has merle and is black pigmented, the Harlequin pattern is expressed. Breedings between N/H dogs are expected to result in 25% embryonic lethal offspring.

The S Gene

White spotting patterns that occur in many dog breeds do not have a uniform genetic basis. Some white patterns, such as the Irish spotting, are symmetrical with white markings on the undersides, collar and muzzle, and/or blaze such as seen in Boston Terriers and Corgis. The white pattern called mantle is phenotypically similar to Irish spotting but with more white extending onto the thigh and up the torso, as seen in some Great Danes. A pattern of less symmetrical white spotting, often called **piebald, parti or random white**, is present in many breeds. A DNA variant has been found in *Microphthalmia Associated Transcription Factor-* (MITF) gene that is associated with **piebald spotting** in many breeds.

The genetic determination of white spotting in dogs is complex. In breeds such as Collie, Great Dane, Italian Greyhound, Shetland Sheepdog, Boxer and Bull Terrier, piebald behaves as a dosage-dependent trait. A dog with one copy of the MITF variant has some white pattern expression, while a dog with 2 copies of the variant display more extreme white with colour only on the head and perhaps a body spot. In Boxers and Bull Terriers, dogs with 2 copies of the MITF variant are completely white and dogs with 1 copy display the mantle (called **flash** in these breeds) pattern. However, additional mutations in MITF or other white-spotting genes appear to be present in these breeds that affect the amount of white being expressed. In other breeds, **piebald** behaves as a recessive trait- that is 2 copies of piebald are needed to produce white spotting.

This test will assist breeders with selection of matings that can produce the desired outcome for white.

Results are reported as:

N/N	Dog has no copies of piebald
S/N	Dog has 1 copy of piebald
S/S	Dog has 2 copies of piebald

Note- expression of white patterns varies from breed to breed and among individuals within a breed. This test is specific for the mutation in MITF known to be associated with piebald/random white spotting.