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Merle aka dapple - colour.

Info from APBT Network

Look-a -like "pit bulls" are not "real" APBT.

Referring to the "rare" 'merle' harlequin pattern colour in "pit bulls".

The APBT standard and the emergence of the Merle pattern in the APBT breed.

Several breeds are known to carry merle as a colour pattern but the APBT is not one known to have ever carried this "infected" allele and having this colour is a definite disqualification.
By Scot E. Dowd Ph.D.

The recent appearance of merle patterned APBT's and a couple breeders specializing in the "new" and "rare" colour pattern has stirred up a controversy in the APBT community.

The general thought among those that have been around the APBT breed for the greatest number of years **is that**, these new colour patterns were brought about by **unscrupulous outcrossing to a separate breed such as Catahoula Leopard dogs.**

What is known is >> that Catahoula Leopard dogs along with pit bulls are often used in the sport of hog catching / hunting and it is known that crosses of these breeds have been made in attempts to produce more **competitive catch dogs.**

The following comments from the APBT standard committee provide prelude to a brief essay on the **merle locus in relation to the APBT**

Walt Pasko "I feel the emergence of the merle colour pattern in our breed has raised the questions of how it was introduced into our breed and what health problems the merle gene could cause.
From all information I've read, I have to recommend that the merle colour be made a **disqualification in the APBT Breed Standard.**"

Carol Gaines Stephens "I strongly oppose the colour pattern 'merle' in the APBT since it has never been there in the past and has just recently risen it's ugly head with the popularity of the **catch dogs.**
I have spoken to several people that say that they know and breeders that do so themselves, cross the APBT with the Louisiana Catahoula Leopard dog to make a better catch dog.
If the gene has never been present in all these decades/centuries then **how did it finally come about just recently?**
I am a firm believer in leaving the standard the way it was **originally**, (we talking about the APBT breed here – not just better hunting dogs) - but when something surfaces that has no rhyme nor reason, then I think we have to address the matter."

Cheryl Larum "I am in agreement with the other committee members on the merle issue"

Scot E. Dowd " First it should be noted that there are ways that the merle can remain hidden such as within a complete phaeomelanic coat where the merle would not be evident, also there are cryptic merles, however **this absolutely would fail to explain the relatively recent appearance of this colour pattern in the APBT.**

I feel that another allele with defined health problems associated with this locus, is not a positive thing for our breed – the APBT"

The following information is submitted on behalf of the NAPBTA standard committee - ***Scot E. Dowd***

Unethical outcrossing to another breed of dog.

This issue is of great concern with the merle as a colour pattern.
This practice would then have been followed **by falsely registering such an outbred animal - mutt** - either with the ADBA or UKC as a purebred APBT.
Such false registration would be **termed hanging papers.**

The health aspects of the Merle allele.

Here I will try to answer the predominant questions that arise regarding the merle allele and the APBT without making a judgment of my own other than that expressed above.

Why is a colour or colour pattern so important to the stewards of the breed?

The entire process of coloration and colour patterns in dogs starts with embryonic development.
The specific cells that become melanocytes (pigment producing cells) are derived entirely from the neuronal crest of the embryo.
This essentially means that pigment cells are directly produced along with the same cells that give rise to
the nervous system.

Though not entirely true, it can be assumed that if you have defects in genes associated with colour genetics you might also have nervous system defects because both types of cells are derived from the neuronal crest.
This provides a logical genetic indicator and explains why it is likely that certain dilute or patterned dogs, such as extreme piebald's, or other types of homozygous dilutes common in the APBT, as well as those that may be carrying the Merle pattern **are prone to psychological, neurological and/or immunological problems found in other breeds that carry these alleles.**

What is merle?

Merle like other dilution alleles acts to lighten whatever colour would otherwise have been expressed.
However, with merle the lightening effect is not spread evenly over the coat, but produces patches of undiluted colour (dappled pattern) scattered over the dog's body.

The merle gene when heterozygous Mm (only **one copy** of the gene) on an otherwise black dog produces a blue merle which is phenotypically a bluish grey dog that is dappled with full colour black spots.
A homozygous or MM dog (carrying **two copies** of the merle gene), often called a double merle or a homozygous merle, **will be a mostly white dog (similar to an extreme Piebald).**

The normal state of the merle locus is dual recessive mm and completely lacks the offending transposon resulting in normal colour.

Maybe merle has been in the breed throughout its history and only now is it being noticed?

The response to this question is also genetic in nature.

The genetic and phenotypic nature of the Merle locus and the merle allele (M) is such that **it would not remain unnoticed in a breed and suddenly appear**.

It would take **crossing to another breed** that carries the merle allele for it to be transferred into the breed.

The reason it could not remain invisible or hidden is because the Merle allele is expressed with incomplete dominance.
This means, if it is within the genome at all, even in a heterozygote (one copy of the gene) state, **it is still expressed and evident.**

The M allele **is not found in all breeds**; in fact **most breeds do not carry it**.

Finally, this specific transposon **cannot arise spontaneously or through mutation as some have claimed**.

What are the health problems associated with the merle allele?

The merle allele like a couple other dilution factors when expressed in a homozygous state is correlated to psychological, neurological, and usually immunological issues.

Here I will mention a few of the issues.

Eye development problems.

The first are eye development problems that are superficial in nature affecting appearance such as heterochromia iridis (A difference of colour between the iris of one eye and the other), thus … a dog with **one brown and one blue eye** has heterochromia iridis.

Note that this **defect** is not necessarily or always indicative of having the merle gene because it **can also be found** in dogs with extreme piebald or double blue dilution for example.

In addition to superficial indicators there are also major effects such as absence of tapetum lucidum.
Tapetum lucidum is a reflective substance that lines the back of the dog's eyes. This reflective structure acts like a mirror and reflects light back through the retina, like a satellite dish giving the retina two chances to catch the light.

Night blindness or reduced ability to see in low light.

Dogs that lack tapetum licidum have **night blindness or reduced ability to see in low light**.

Smaller than normal eye - third eyelids.

Another defect is lack of retinal pigment and microphthalmia. Microopthalmia **(smaller than normal eye)** is described as dogs having **prominent third eyelids and seemingly small eyes which appear recessed in the eye socket** (enophthalmos).

Physical cleft in a portion of the eye, particularly the iris.

Another problem known as coloboma is actually **a physical cleft in a portion of the eye, particularly the iris**.

Reduction in auditory sensitivity or complete deafness.

In addition to the eyes which are a key indicator of neurological defects, there is also evidence for effects on the ears that result in **reduction in auditory sensitivity or complete deafness** because the merle colour locus exerts epistatic effects on ear development.

Excessive white or dilution in a dog of any colour.

Excessive white or dilution in a dog of any colour can be a warning sign of potential hearing problems**.**
If there is no pigment in the inner ear the dog will be deaf;
white ears are more likely to lack inner ear pigment.

More technically, what is the genetic explanation of the merle pattern?

Transposon or transposable element**.**

The merle allele is considered to be caused by a transposon or transposable element.
A transposon is a piece of DNA that has the potential to actually jump out of, or excise from the gene it has infected (disrupted), during cellular division and genetic DNA replication.

This means that while melanocytes are migrating from the neuronal crest during embryonic development the merle transposon can remove itself from the gene in some of the melanocytes when they are derived and produce normal coloration on those parts of the coat to which they migrate.
Thus, the merle allele acts to cause eumelanic areas in the coat, to become diluted, but other areas to be fully and intensely pigmented.
Such fully coloured areas occur in scattered patches throughout the body.

The merle locus is autosomal (not carried on one of the sex chromosomes) acting as a dominant mutation (it is expressed in all dogs that carry this gene).
It should also be noted that genetically such transposons do not arise spontaneously but **must be passed from sire and/or dam to offspring.**

This means that if the APBT did not carry this allele to begin with, **then only through outcrossing to another breed**, that does carry this transposon, could it be integrated into the APBT genome.

Thus, we as members of the National American Pit Bull Association are presented with the issue of **a dilution pattern** that **may have been introduced into the breed by unethical conduct.**
As stewards of the breed we have to choose to continue to honour our current standard and allow this colour pattern or to change the standard to reflect that merle is not an acceptable colour pattern for the breed.

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Description.

Merle can affect all coat colours.
The merle forms of brown and black are usually called liver (or red) and blue, respectively.
Dogs who are recessive red can still be affected by merle, but the patches are either hardly seen or if the dog is a clear recessive red, it is not visible at all combinations such as brindle merle exist, but are not typically accepted in breed standards.

In addition to altering base coat colour, merle also modifies eye colour and colouring on the nose and paw pads.
The merle gene modifies the dark pigment in the eyes, occasionally changing dark eyes to blue, or part of the eye to be coloured blue.

Since merle causes random modifications, however, both dark-eyed, blue-eyed, and odd-coloured eyes are possible colour on paw pads and nose may be mottled pink and black.

Breed.

Merle is a distinguishing marking of several breeds, particularly the Australian Shepherd, and appears in others, including the Australian Coolie (Koolie), German Coolies in Australia, the Shetland sheepdog, various Collies, the Welsh Corgi (Cardigan), the Pyrenean Shepherd, the Bergamasco Sheepdog, the Old English Sheepdog, American Pit Bull Terrier, and Cataula Leopard Dog.
In Dachshunds the merle marking is known as "dapple".
It is also present in the Pomeranian and Chihuahua, but is a disqualification according to the FCI standards.
In the Cocker Spaniel breed, it is a recognized pattern, although incorrectly listed as a 'colour'.
(American Kennel Club, Cocker Spaniel) The merle gene also plays a part in producing harlequin Great Danes.
In several breeds, such as the Pomeranian and Chihuahua merle is an indicator of cross breeding.

Genetic basis.

A deaf 8 month old homozygous Merle Australian Shepherd.
She has blue eyes with starburst pupils and an eccentric pupil in her left eye.

Merle is actually a hetero-zygote of an incompletely dominant gene.

If two such dogs are mated, on the average one quarter of the puppies will be "double merles", which is the common term for dogs homozygous for merle, and a high percentage of these double merle puppies could have eye defects and/or be deaf.

Knowledgeable breeders who want to produce merle puppies mate a merle with a non-merle dog; roughly half the puppies will be merles without the risk of vision or hearing defects associated with double merle dogs.

In January 2006 scientists at Texas A&M University announced the discovery of a mobile genetic unit called a retrotransposon, responsible for the merle mutation in dogs.

A phantom merle or cryptic merle is one with such small patches of merle—or none at all—that it appears to be a non-merle.
This is commonly seen in dogs who are recessive red, clear recessive reds in particular, though patches can still be seen in certain red dogs. In America, a dog with the phantom merle colouring is described as being "cryptic for merle."

Health issue.

The merle gene is often associated with congenital deafness, with merle dogs being more likely than other dogs to be born deaf.

Dogs with two copies of the merle gene (homozygous merle or "double merle") have an even higher chance of being born deaf.

The UK Kennel Club has acknowledged the health risk associated with homozygous merle and will stop registering puppies produced from merle to merle mattings starting from 2013.

Merle to merle mating is currently only forbidden in three breeds.
Recent research indicates that the majority of health issues occur in dogs carrying both piebald and Merle genes.
The suppression of pigment cells (melanocytes) in the iris and in the stria vascularis of the cochlea (inner ear) leads to blue eyes and deafness.

An auditory-pigmentation disorder in humans, Waardenberg syndrome, reflects some of the problems associated with heterozygous and homozygous merle dogs and genetic research in dogs has been undertaken with the goal of better understanding the genetic basis of this human condition.

A Great Dane mix homozygous for merle displaying bilateral micropthalmia.

Ocular defects.
Dogs who are homozygous for the merle pattern gene often have visual and auditory deficits.
These dogs are sometimes referred to as 'double merle' and sometimes incorrectly referred to as 'lethal white.'

Ocular defects include micropthalmia, conditions causing increased ocular pressure, and colobomas, among others.

Double merle dogs may be deaf or blind or both, and can carry ocular **defects in blue or coloured eyes.**
Currently no studies have been done to prove whether or not the merle gene affects the eyes, causing blindness.

Auditory defects.
One study done by a German researcher showed that out of 38 dachshunds, they found partial hearing loss in 54.6% of double merles, and 36.8% of single merles.

1 out of the 11 (9.1%) double merles was fully deaf while none of the single merles were.

Another study done by Texas A&M University found that out of 22 double merles, 8 were completely deaf and two were deaf in one ear.

Out of 48 single merles, only one was deaf in one ear, none were completely deaf.

Another study done with 70 dogs, 15 of them were Catahoula Curs and 4 of them were deaf, whereas the other 86% of double merles in other breeds were deaf.

Deaf, blind, and deaf and blind dogs can have good lives when properly cared for.

There are a variety of internet groups dedicated to supporting carers of such dogs.
Deaf dogs can compete successfully in agility and there are many anecdotal reports of deaf/blind dogs earning their Canine Good Citizen certification, working as therapy dogs, and competing in dog sports like tracking or Nose work.