

Defining - M^c - Cryptic Merle

Chapter taken from the book ~ **Merle - SINE Insertion from M^c - M^h the Incredible Story of Merle** ~

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Use of the term “Cryptic Merle”

The origins of the word cryptic come from “crypticus”, a Latin word meaning “hidden” or “to hide.” The terminology of “cryptic Merle” has been used for decades to mean exactly that - a dog who is a “hidden Merle”. You will most often find the word cryptic also used in conjunction with words such as “hidden”, “masked”, “ghost”, “phantom” and “minimal”. All the same descriptive words with the same meaning - a dog who was assumed by phenotype to be non-Merle and then bred as a Merle producing visibly Merle patterned offspring.



Indie's Merle pattern - m/M - m/266 is “masked” by e/e - Recessive Red as Merle does not express well on red pigment.

It is very important to note that the word “cryptic” in this sense has been used only as a general “descriptive” word and not in a true “genetic” manner. Used to describe something we could not understand as we did not yet have the technology available to discern the precise genetics of Merle’s poly-A tail.

In 2015 a paper was published officially naming the “Cryptic Merle” allele - M^c. However this research was done still using the old testing method and based only on phenotype, not on breeding outcomes. This resulted in an M^c allele that was much too long in length. This length encompasses the “langevin et al” three alleles of M^c, M^{c+} and M^a which are all “Non-Expressing” as heterozygous but do not breed the same.

It was unfortunate that this paper named the allele M^c - Cryptic Merle as the term “cryptic” has become so convoluted over the years; used in such general form to mean so many different things to different people and in different breeds. My choice for the allele would have been M^t - Truncated Merle. “Truncated” meaning “shortened”, “curtailed”, “cut short” which describes the M^c allele perfectly.

Setting the boundaries for each allele was an immense task that came with an immense responsibility. The base pair numbers for the M^c allele were by far the most important. We need to ensure for every breeder that when a dog testing as M^c - 200 - 230 bp is bred to M, there is no deletion of pigment to white due to the combination of both alleles and therefore no risk of vision and/or hearing impairments. Any dog tested as m/M^c or M^c/M^c can safely be bred to M with the M^c allele breeding the same as non-Merle. A dog who is M^c/M will have no pigment deleted to white due to the allele combination.

Moving forward “**Cryptic Merle**” now genetically refers to M^c, an allele on the M Locus that is so shortened/truncated that it can no longer express a Merle pattern. A dog who is m/M^c will not breed as M and will not produce offspring who are Merle patterned. Using the wording of “Hidden Merle” or “Masked Merle” is the appropriate wording for cases such as the red pigment (pheomelanin) of e/e - Recessive Red and/or A^y - Clear Sable not expressing - “hiding or masking” - a Merle pattern. The true genetic word for this is “Epistasis” but the definition also means “masked” or “hidden” - both these words have long been used in the language of Merle.

As well dogs assumed to be “Hidden Merles” due to resulting Merle patterned offspring may actually be a case of a combination of the lesser Merle alleles from each parent due to shorter “Non-Expressing” Merle alleles.

The following breeding between Kelvar and Kiva and their resulting offspring, Colt, is an excellent example of what we used to assume regarding Merle and what accurate Merle testing can now confirm.

Either Kevlar or Kiva might have been assumed to be a “hidden” or “ghost” Merle - having a full M allele and not expressing the pattern on their coat. DNA testing by a lab that can provide us with the exact number of base pairs has shown that both Kevlar and Kiva are m/M^{a+} and their offspring Colt is M^{a+}/M^{a+} .

<p>Sire - Kevlar - m/M^{a+} - m/255</p> 	<p>Dam - Kiva - m/M^{a+} - m/255</p> 
<p>Offspring - Colt - M^{a+}/M^{a+} - 255/255, D/D</p> <div>   </div>	
<p>Breed - Catahoula</p>	

<p>Sire - Rico - m/M^a - m/247</p> 	<p>Dam - Mac - m/M^{a+} - m/258</p> 
<p>Offspring</p> <div> <p>Wendy Darling - M^a/M^{a+} 247/258</p>  </div> <div> <p>Peter Pan - M^a/M^{a+} - 247/258</p>  </div> <div> <p>Tinkerbelle - M^a/M^{a+} 247/258</p>  </div>	
<p>Breed - Toy Australian Shepherd</p>	

The following is an example of a litter from an $m/M^a \times m/M^{a+}$ breeding - “Non-Expressing” Merle alleles. Without accurate Merle testing that can report the base pair numbers of each allele, it could have been assumed that a parent was a “hidden Merle” and M offspring were produced.

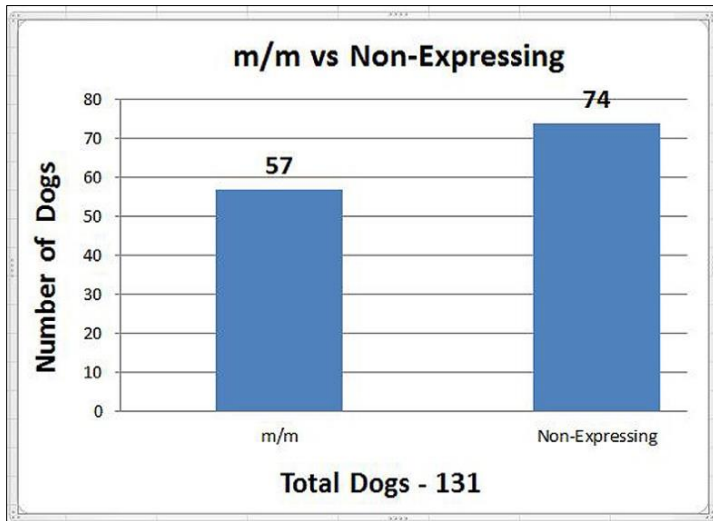
<p>Sire - Boaz m/m</p> 	<p>Dam - Selah M^a/M^a - 250/250, D/D</p> 
<p>Litter - All Pups m/M^a - m/250</p> 	
<p>Breed - Catahoula</p>	

The following is a reversed example of a dam who is homozygous for two “Non-Expressing” M^a - Atypical Merle alleles which does create a Merle pattern. All of her offspring are heterozygous for M^a - “Non-Expressing”.

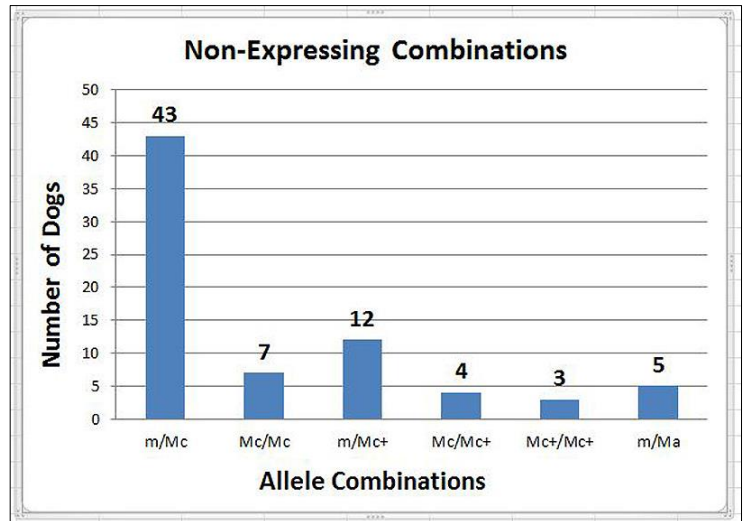
Genetic Diversity

In order to protect the genetic diversity of all Merle breeds it is imperative for breeders to have a full understanding of the M^c allele 200 - 230 bp.

In order to provide a very clear understanding of just how common these shorter alleles are, most importantly M^c , I will provide results of an online survey of Aussie breeders/owners as to the percentage of phenotypically solid dogs having a shorter non-expressing Merle allele of M^c , M^{c+} or M^a .

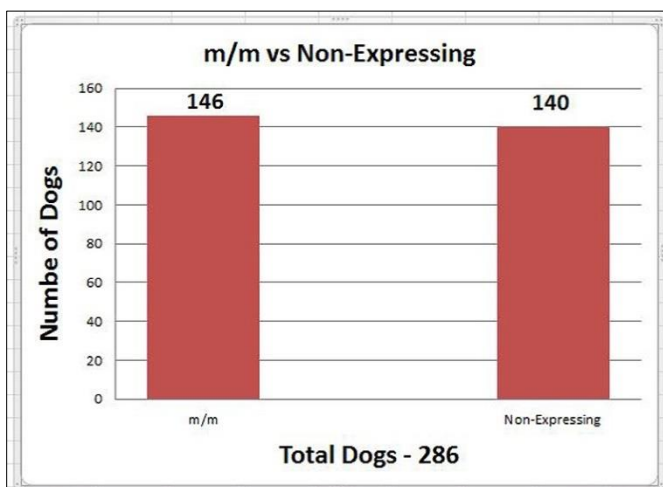


Percentage of m/m - Non-Merle - 43%
 Percentage of dogs having a shorter Non-Expressing allele of M^c , M^{c+} or M^a - 57%

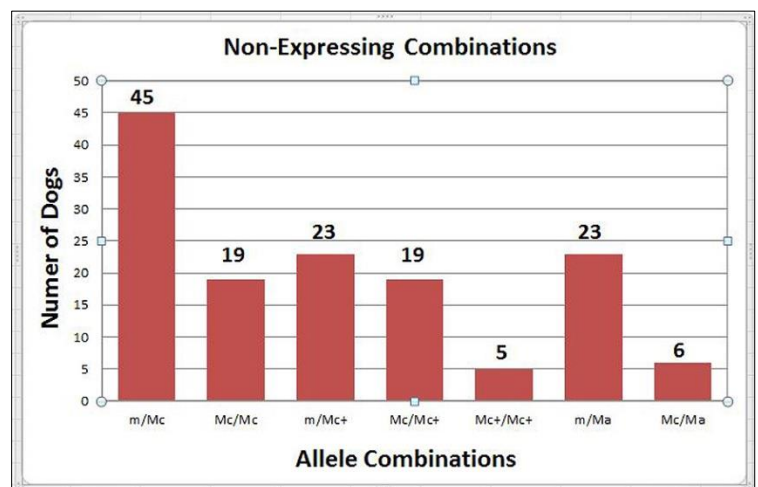


38% of phenotypically solids dogs are either m/Mc or Mc/Mc. These dogs will breed the same as non-Merle. When bred to an M - Merle mate no pigment will be deleted to white. 19% of phenotypically solids dogs have either an M^{c+} or M^a allele which can delete pigment to white when paired with M.

The following are the same results using data provided by Tilia Laboratories which includes 785 Merle tested dogs from all breeds worldwide. Out of those 785 dogs a total of 286 are non-Merle in phenotype - **146 are m/m and 140 have shorter non-expressing alleles.**



Percentage of m/m - Non-Merle - 51%
 Percentage of dogs having a shorter Non-Expressing allele of M^c , M^{c+} or M^a - 49%



23% of phenotypically solids dogs are either m/Mc or Mc/Mc. These dogs will breed the same as non-Merle. When bred to an M - Merle mate no pigment will be deleted to white. 26% of phenotypically solids dogs have either an M^{c+} or M^a allele which can delete pigment to white when paired with M.

So what do these statistics have to do with the issue of genetic diversity? There are currently 18 labs offering Merle testing. A full list is available at this link - www.merle-sine-insertion-from-mc-mh.com/labs-offering-merle-testing.

Nine of these labs are offering the new high resolution testing method but only six of these are offering base pair numbers and mosaic results. Out of these six, only five are offering up-to-date information in regard to the M^c allele.

Nine labs are still using the old method of testing and only two are offering correct genetic information in regards to M^c . That leaves us with 11 labs that have old outdated and inaccurate non genetic information on their websites. Along with literally hundreds of websites who repeat this information over and over again, as well as old information cited from old Merle studies, some that were based only on phenotype and not on genetic testing.

Any Google search for "Cryptic Merle" will result in pages and pages of websites using the old catch-all, convoluted and non-genetic meaning of the term. I have seen very limited progress over the past years from most labs and breed websites to update their Merle information. The damage being done could be disastrous to many breeds as dogs testing as M^c - 230 bp and under are neutered/spayed and removed from breeding programs due to the fear that M^c means "hidden", "ghost" and "phantom" Merle. That their M^c dog could possibly breed the same as a Merle and may produce impaired Double Merle offspring when bred to another visible Merle dog.

The following information is taken from Google search - "Many solid dogs are actually cryptic or phantom merles and can produce both Merle and double merles. A cryptic ghost or phantom Merle is a dog which phenotypically appears to be a non-merle or very faint patches of Merle that can go unnoticed. Animals that do not present the Merle phenotype may possess the Merle genotype and subsequently produce Merle offspring. These dogs are known as cryptic Merles."

Scary stuff indeed!!

Information such as this has not been overly detrimental in the past when there was really no reliable test for Merle or the M^c allele. Breeders based their information on breeding results of litters produced. Now that we have the technology available for the accurate testing of Merle's poly-A tail we are aware of just how common a result of M^c - 200 - 230 bp is at an average of approximately 38%.

What if 38% of all phenotypically solid dogs were removed from breeding programs based exclusive on a result of M^c ?!

Genetic diversity would be greatly impacted. I have already seen dogs tested as M^c spayed/neutered and removed from the genetic pool either based on information the owner found through an internet search or following the advice of labs stating not to breed M^c to M for the fear of producing Double Merle offspring and passing this "unsafe" M^c allele to the future generation. It is even more imperative in Europe that the M^c allele is understood completely as many registries and clubs do not allow the breeding or registering of litters from a Merle x Merle cross. This still includes M^c x M and even M^c x M^c . Imagine the irreversible damage that could be done to a closed gene pool by restricting the breeding of M^c dogs? A gene pool will get smaller when the number of gene variants decrease and are lost due to dogs not reproducing and passing their genetics on to future descendants.

It is a travesty to remove a dog from a breeding program based solely on a result of M^c and totally unnecessary! The base pairs for the M^c allele were set at 230 bp in order to guarantee it will breed the same as Non-Merle, that no pigment will be deleted to white when bred to M. I know that "guarantee" is a strong word here and not scientific in the least. However in this case it is a word I am confident using. The limit that was placed on the M^c allele could likely have been slightly higher, maybe even to 234 bp but I have seen examples of 235 bp x M starting to delete pigment.

NOTE: Even if every M^c dog was removed from the breeding population there would never be an end to the eradication. Mosaicism is always happening. There is no way to stop the natural shortening of any poly-A tail. With 18% of Merle dogs being mosaics and 55% of those dog having a shortened M^c allele there is just no way to "get rid of" it.