

## How is Merle inherited?

Merle exhibits an autosomal dominant pattern of inheritance with incomplete penetrance.

## What is the Merle Mutation?

A 253bp SINE element repeat insertion that at the boundary of intron 10/exon11 in the SILV gene

## Why is there so much diversity in phenotypes?

The Merle mutation produces a multitude of alleles as a result of differing lengths attributed to the polyA tail of the SINE insertion. Moreover, the mutation exhibits varying degrees of mosaicism, this means that different cell types in the body can exhibit different Merle alleles which adds further complexity to the phenotype.

## Can results differ between different biological samples submitted?

Yes they can. Blood, Cheek Swabs and Semen may show differences in merle alleles call or possess more than 2 alleles.

## How many Merle alleles are there?

The most comprehensive description of merle alleles was published by Langevin et al. 2018. The assay design and description of the methodology is freely published and can be replicated by other genetic testing labs. The research describes 7 merle alleles designated m, Mc, Mc+, Ma, Ma+, M and Mh with associated phenotype descriptions for each. Below we listed the alleles and phenotypes described by the study.

### ALLELE

m  
Mc  
Mc+  
Ma  
Ma+  
M  
Mh

### MAJOR PHENOTYPIC FEATURES

No Merle pattern±solid coat  
No Merle pattern±solid coat  
No Merle pattern±solid coat  
No Merle pattern±diluted±brownish hue  
Muted, undefined, diluted±brownish hue  
Classic Merle  
Minimal Merle, areas deleted to white, tweed

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## What Merle assay is Orivet using?

Orivet uses the same assay and methodology described by the Langevin et al.2018 study and reports the 7 merle alleles accordingly.

## How are the Merle alleles detected?

Merle alleles are designated by the base pair size of the SINE insertion. However, depending on the instrument used to detect the mutation, the base pair number can vary slightly between labs. A lab that is accurately testing for the SINE insertion can make the required adjustments to issue results identical to the 7 merle alleles described by Langevin et al. 2018.

## Base pairs Vs Alleles

As base pair sizes will vary between laboratories it makes more sense to issue results that can be shared universally between breeders. As long as the 7 merle alleles is the standard nomenclature for issuing results, dogs tested overseas and in Australia should not show discrepancies in merle reporting.

## On contributing to Merle research

Merle is a complex phenotype and Orivet is always happy to discuss results with you and share photos of your dogs amazing coat colours. As homozygous or double merles dogs are often affected with hearing and vision disabilities we believe it is important to document these phenotypes in order to assist researchers in identifying further health risks affecting your breed. Please share with us your stories.

## Is there a patent on the merle test?

Yes there is, the patent is held by IDEXX laboratories and Orivet has acquired the necessary licence to perform the test.

## Reference

Langevin M, Synkova H, Jancuskova T, Pekova S(2018) Merle phenotypes in dogs -SILV SINE insertions from Mc to Mh. PLoS ONE 13(9):e0198536.