

**From:** "George Sofronidis" <[george@orivet.com.au](mailto:george@orivet.com.au)>  
**Subject: RE: DNA testing for Belgian Shepherds**  
**Date:** 12 August 2016 at 1:41:16 PM AEST  
**To:** "Jody Hafey" <[jodyhafey@gmail.com](mailto:jodyhafey@gmail.com)>

Hi Jody

DNA testing can be used and is offered by a number of genetic testing laboratories worldwide. One can determine whether a dog is

NOT SHOWING THE PHENOTYPE (NORMAL)

Or

CARRYING THE PHENOTYPE (CARRIER)

Or

POSITIVE SHOWING THE PHENOTYPE (AFFECTED).

I would simply refer to each test via the gene and cite the reference for them. Just let them know that genetic testing is available and can be used to let breeders know whether they carry the genes associated with these 3 phenotypes and therefore work towards producing affected or clear animals.

Masking is part of the E Series and E<sup>m</sup> (masked) MC1R Gene appears over the top of the A series. It is top dominant and therefore a dog only need one to have a mask. By testing and determining whether a dog carries the E then it can allow breeders to determine whether masking is present.

Reference: [J Hered.](#) 2003 Jan-Feb;94(1):69-73.

#### **MC1R studies in dogs with melanistic mask or brindle patterns.**

[Schmutz SM<sup>1</sup>](#), [Berryere TG](#), [Ellinwood NM](#), [Kerns JA](#), [Barsh GS](#).

#### **Author information**

#### **Abstract**

Black mask is a characteristic pattern in which red, yellow, tan, fawn, or brindle dogs exhibit a melanistic muzzle which may extend up onto the ears. Melanistic mask is inherited in several breeds as an autosomal dominant trait, and appears to be a fixed trait in a few breeds of dogs. A MC1R nonsense mutation, R306ter, has been shown to cause a completely red or yellow coat color in certain breeds such as Irish setters, yellow Labrador retrievers, and golden retrievers. The amino acid sequence for the melanocortin receptor 1 gene (MC1R) was examined in 17 dogs with melanistic masks from seven breeds, 19 dogs without melanistic masks, and 7 dogs in which their coat color made the mask difficult to distinguish. We also examined nine brindle dogs of four breeds, including three dogs who also had a black mask. No consistent amino acid change was observed in the brindle dogs. All dogs with a melanistic mask had at least one copy of a valine substitution for methionine at amino acid 264 (M264V) and none were homozygous for the premature stop codon (R306ter). These results suggest that black mask, but not brindle, is caused by a specific MC1R allele.

The same applies to the determination Dilute (MLPH Gene). Genetic testing allows you to determine whether a dog is carrying dilute and will be dilute and thus dilute the coat colour.

#### **Reference**

Polymorphisms within the canine *MLPH* gene are associated with dilute coat color in dogs

- Ute Philipp, et al;

*BMC Genetics* 2005;6:34

DOI: 10.1186/1471-2156-6-34

Abstract

### Background

Pinschers and other dogs with coat color dilution show a characteristic pigmentation phenotype. The fur colors are a lighter shade, e.g. silvery grey (blue) instead of black and a sandy color (Isabella fawn) instead of red or brown. In some dogs the coat color dilution is sometimes accompanied by hair loss and recurrent skin inflammation, the so called color dilution alopecia (CDA) or black hair follicular dysplasia (BHFD). In humans and mice a comparable pigmentation phenotype without any documented hair loss is caused by mutations within the melanophilin gene (*MLPH*).

Coat Length

[Science. 2009 Oct 2; 326\(5949\): 150–153.](#)

Published online 2009 Aug 27. doi: [10.1126/science.1177808](https://doi.org/10.1126/science.1177808)

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### Coat Variation in the Domestic Dog Is Governed by Variants in Three Genes

[Edouard Cadieu](#),<sup>1</sup> [Mark W. Neff](#),<sup>2</sup> [Pascale Quignon](#),<sup>1</sup> [Kari Walsh](#),<sup>2</sup> [Kevin Chase](#),<sup>3</sup> [Heidi G. Parker](#),<sup>1</sup> [Bridgett M. VonHoldt](#),<sup>4</sup> [Alison Rhue](#),<sup>2</sup> [Adam Boyko](#),<sup>5</sup> [Alexandra Byers](#),<sup>1</sup> [Aaron Wong](#),<sup>2</sup> [Dana S. Mosher](#),<sup>1</sup> [Abdel G. Elkahoul](#),<sup>1</sup> [Tyrone C. Spady](#),<sup>1</sup> [Catherine André](#),<sup>6</sup> [K. Gordon Lark](#),<sup>3</sup> [Michelle Cargill](#),<sup>7,\*</sup> [Carlos D. Bustamante](#),<sup>5</sup> [Robert K. Wayne](#),<sup>4</sup> and [Elaine A. Ostrander](#)<sup>1,†</sup>

Regards,

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