

PMEL Merle

PMEL is Silver and Merle

The gene PMEL (premelanosome protein) is important for storing melanin (pigment) in the cell. In horses a mutation in the PMEL gene is responsible for silver dapple expression. [1] In dogs a different mutation in the same gene causes the merle coat color. [2]. In HGG you will find both silver and merle listed as alleles of the “silver” gene.

How Merle Works

Silver is a straight forward mutation that reduces the amount of black pigment in a horse's coat. Merle is significantly more complex. Instead of a stable mutation, the merle pattern is caused by changing SINE lengths. SINE stands for “short interspersed nuclear element.” It's a short repeating bit of DNA. In the case of merle this repeating element is found in the regulatory section of the gene.

These repeating DNA elements can cause a lot of confusion during DNA replication. Sometimes they are accidentally copied many times, sometimes whole sections of them get pinched off. In this way the number of merle SINE elements an animal has can grow or shrink while it's developing as an embryo. Every time the merle SINE element is repeated it causes less pigment to be stored. Darker areas of the animal have less SINE elements, while lighter areas have more SINE elements. The random fluctuations in SINE lengths result in the varied shades found on merle animals.

Too many SINE elements is dangerous

A common myth is that homozygous merle leads to health issues. While some genes, like frame in horses, always cause health issues when homozygous merle causes issues when too many copies of the SINE element are present. It is possible for homozygous merle animals to be perfectly healthy, and heterozygous animals to be unhealthy depending on the number of SINE elements present. It is the total amount of copies of the merle SINE element that determine the health impact. Of course a very fast way to get a large number of SINE elements is to produce a homozygous merle dog.

HGG does not condone merle to merle breeding in real life.

As stated above, merle is unstable and can spontaneously become longer or shorter. This means a cross that is safe “on paper” may produce a health impacted individual anyways.

In the real world genetic testing labs test if the merle mutation is found at all, and how large the repeating section of merle DNA is. This section is often called the poly-A tail. Different labs use different numbers or codes to denote different lengths. You may also see terms like cryptic, dilute, standard, double etc. used to describe different lengths of merle. For simplicity we do not use these terms in the game. HGG uses a unique SINE length numbering system that does not correspond to any real life lab.

You may see the following terms used to describe merle dogs, but they are not used in HGG:

Cryptic - An animal that has the merle SINE mutation but at a number small enough not to show

Dilute - A merle, normally with lower SINE length, that shows silver like dilution but not classic merle patches

Standard/Classic - The typical merle expression

Double - An animal with excessive SINE length, often but not always caused by being homozygous. These individuals have large amounts of white markings, and a very high incidence of hearing and vision problems.

Harlequin gene- A homozygous lethal gene found in Great Danes that removes the diluted area of merle individuals but leaves the dark patches intact.

Harlequin pattern - A variant of merle caused by long sine lengths having strong dilution and a lot of white. Not related to the harlequin gene.

Tweed - A variant of merle with lighter patches mixed into the normal dilute and darker patch areas. Caused by the SINE length increasing in the lighter areas.

SINE Numbers in HGG

To calculate how big the total amount of SINE length your HGG horses has:

- 1) Find the silver gene in the gene test results
- 2) The merle gene is written like **Mrl(65)**. The number inside the parentheses () is the SINE length. If your horse is homozygous for merle, add both numbers together.
- 3) If your horse has a copy of the silver gene, add 15 to the total SINE length.
- 4) The - symbol means no mutation is found and does not add to the total SINE length.

In HorseGeneticsGame foals will start developing health problems at a combined SINE length of 80. Unhealthy horses in the game have a reduced training ability, though breeding ability is not impacted. The health impact is listed as a percentage. The health impact increases proportionally. A foal with a 120 combined SINE length will have much worse health than a foal with an 80 SINE length.

SINE lengths over 142 are embryonic lethal.

Health impacted foals can be corrected by the HGG university using the ability max out feature to give them normal showing ability.

Red Merles

The PMEL gene is specifically responsible for the fixing of black pigment. That is why the silver mutation only shows on black and bay horses, not on red/chestnut horses. The same is true for merle. ee individuals do not show the classic merle patterning. However they can end up with spontaneous blue eyes from the merle gene and can have health impacts at high SINE levels just like their black based counterparts. Not enough research exists yet on merle's impacts on ee individuals for us to have the complete picture. We do know, however; that there is some impact at high levels.

For simplicity, ee horses in game will have negative health impacts at the same SINE levels as E/? horses.