

## Acral Mutilation Syndrome (AMS)

**Affected breeds:** English Cocker Spaniel, English Springer Spaniel

Acral Mutilation Syndrome (AMS) is a neurological disorder in which the nerves leading to the feet degenerate. This results in an insensitivity to pain in the lower limbs, sometimes with swelling and discomfort. Affected dogs are prone to excessive licking which can lead on to self-mutilation.

The condition is sometimes detected soon after birth, but will become obvious as the dog grows older and by 4-5 months the characteristic licking and biting begins.

The outlook for affected dogs is bleak with euthanasia being the only option as the condition progresses.



AMS is caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will develop AMS ("AFFECTED").

### **This test is particularly useful for breeders:**

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively confirm AMS in an affected dog

### **This test will be reported as:**

**CLEAR** : no evidence of the AMS mutation

**CARRIER** : carries one copy of the defect, which will be passed to 50% of offspring

**AFFECTED** : carries two copies of the defect, causing AMS

### **The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:**

CLEAR X CLEAR = 100% CLEAR

CARRIER X CLEAR = 50% CARRIER, 50% CLEAR

CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

### **References**

Plassais J, Lagoutte L, Correard S, Paradis M, Guaguère E, Hédan B, et al. (2016) A Point Mutation in a lincRNA Upstream of GDNF Is Associated to a Canine Insensitivity to Pain: A Spontaneous Model for Human Sensory Neuropathies. PLoS Genet 12(12): e1006482. <https://doi.org/10.1371/journal.pgen.1006482>