Acral Mutilation Syndrome

Other Names:	Pain Insensitivity, Sensory Neuropathy, AMS, SN
Affected Genes:	GDNF
Inheritance:	Autosomal Recessive
Mutation:	chr4:70875561 (canFam3): C>T

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Common Symptoms

Acral mutilation syndrome is an inherited neurological disease affecting dogs. Affected dogs present around 4 months of age with an insensitivity to pain in the lower limbs demonstrated by repetitive licking and biting of their paws, eventually resulting in self-mutilation. Affected dogs are often smaller than littermates and present with severe, self-induced wounds of the feet including toe amputations, toenail loss, and bone fractures. Dogs with open wounds are subject to additional complications from secondary infections. Since affected dogs are unable to feel pain in their feet, they will continue to walk without obvious discomfort. Euthanasia is often requested by owners due to quality of life concerns and a lack of treatment options.

Breed-Specific Information for the English Cocker Spaniel

The Mutation of the *GDNF* gene associated with acral mutilation syndrome has been identified in English cocker spaniels in the Paw Print Genetics laboratory. Neither the frequency of the causal mutation nor its association with disease in this breed has been reported in the medical literature. In addition, multiple apparently healthy English cocker spaniels with two copies of the *GDNF* mutation have been identified by Paw Print Genetics. Thus, suggesting that this disease may display Incomplete Penetrance in this breed.

Testing Tips

Genetic testing of the *GDNF* gene in English cocker spaniels will reliably determine whether a dog is a genetic Carrier of acral mutilation syndrome. Acral mutilation syndrome is inherited in an Autosomal Recessive manner, meaning that dogs must receive two copies of the mutated gene (one from each parent) to be at an increased risk of developing disease. Paw Print Genetics has identified multiple apparently healthy dogs with two copies of the *GDNF* Mutation (including old English sheepdogs and English cocker spaniels). Therefore, acral mutilation syndrome may display Incomplete Penetrance, meaning that some dogs which inherit two copies of the *GDNF* mutation may not develop this disease. In general, carrier dogs do not have features of the disease but when bred with another carrier of the same Mutation, there is a risk of having affected pups. Each pup that is born to this pairing has a 25% chance of inheriting two copies of the mutation (at risk for disease) and a 50% chance of inheriting one copy and being a carrier of the *GDNF* gene mutation. Reliable genetic testing is important for determining breeding practices. Because the mutation may display incomplete Penetrance, genetic testing should be performed before breeding. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. English cocker spaniels that are not carriers of the mutation have no increased risk of having affected pups.

There may be other causes of this condition in dogs and a normal result does not exclude a different mutation in this gene or any other gene that may result in a similar genetic disease or trait.

References

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