

Congenital Ectodermal Dysplasia/Skin Fragility Syndrome in Chesapeake Bay Retriever Dogs

Chesapeake bay retrievers (CBRs) have been recently described with a unique skin disorder comprising trauma-induced skin fragility and congenital ectodermal dysplasia, which is similar to Ectodermal Dysplasia/Skin Fragility (ED/SF) in humans. At birth, affected puppies' skin is lobster pink with blistering on the footpads. Over the first 48 hours, they develop more severe blistering and peeling of the skin on their faces, limbs, and feet. Their skin is extremely fragile and tears easily (Figure 1). There is currently no treatment for this disease.



Figure 1. CBR puppy with ED/SF syndrome. Note blistering on nose, footpads, and soles of the feet.

Affected dogs are born to clinically normal parents. Males and females can be affected, i.e. there is no sex predilection. The disease is inherited as an autosomal recessive traits meaning that both parents have to be carriers of the disease. Briefly, for every trait there are two copies of a gene, termed a gene and its allele. In this case, each parent will have a normal gene paired with the defective allele. When you breed the carrier parents, you will get four possible combinations (Figure 2) which will ultimately result in 25% affected, 50% carrier, and 25% normal (clear).

	Dam	
	A	a
A	AA	Aa
Sire a	Aa	aa

Figure 2. In this case, “A” stands for the normal gene and “a” for the defective allele (gene). “AA” is a clear (normal) dog, “Aa” is clinically normal but a carrier for the defective allele which is “masked” by the normal gene, and “aa” is a dog affected with ED/SF. This type of inheritance is called autosomal recessive.

We have recently found a mutation in a skin specific gene causing the disease in the CBRs. This test is now available for breeders to test their dogs and plan breedings (see below). Not only will the information from the CBRs help the breed but ultimately people with ED/SF.

A brief word about breeding when there is a genetic disease in the breed and a DNA test is available. Previously, if affected animals were born, we would advise not to breed the parents or litter mates. However, now that a test is available, the carriers do not need to be removed from the breeding pool. If you are dealing with a breed in which there are not many dogs, such as the CBRs, then it would be a shame to disqualify a perfectly good dog with all the desired qualities, just because he or she is a carrier. In order to preserve the good genetic “material”, this dog could be bred to one that tests clear, and then those offspring that test clear and that fit the breed standard can be retained for future breedings.

For testing: <http://research.vet.upenn.edu/Default.aspx?alias=research.vet.upenn.edu/PennGen>

The only saving factor about this disease is that you know the affected puppies well before any might be sold-meaning no one ever gets a dog affected with the disease.