



Animal Disease Diagnostic Laboratory

Non-HSF4 Hereditary Cataracts (NHHC)

Disease: Non-HSF4 Hereditary Cataracts

Breed: Miniature American Shepherds

About the condition:

Cataracts develop when the typically clear lens of the eye becomes opaque; this opacity causes diminishment – or total loss, when severe – of vision. In dogs, cataracts are assumed to be inherited unless there is a known history of: ocular trauma, diabetes mellitus, advanced age, chronic uveitis, nutritional deficiencies, or exposure to specific medications or environmental toxins.

Inherited cataracts have been reported in Miniature American Shepherds, diagnosed on examination by board-certified veterinary ophthalmologists. These cataracts are not caused by the two currently-known *HSF4* variants; in other words, affected Miniature American Shepherds test clear for the *HSF4* mutations.

The cataracts in these breeds do not develop until the dog is aged 3-5 years, which is unfortunate, because some dogs may have been bred by that age, and before it was known they were affected. These cataracts are bilateral and they progress very rapidly; the dog is typically blind and requires cataract surgery within months of the cataracts first appearing.

Research carried out at Purdue University indicates that this disease is inherited in an autosomal recessive manner. We have identified a mutation within a disease-associated gene and all affected dogs have two copies of this mutation. We are now offering a genetic test which allows owners to determine their dog's status for this mutation (clear, carrier, or affected) in order to diagnose affected dogs and to guide future breeding decisions. Carriers do not need to be removed from the breeding population; however, they should only be bred to clear mates to avoid producing affected offspring. This is only the third genetic test available to aid in prevention of this condition, and the first in a gene that is not *HSF4*.