Primary Open Angle Glaucoma (POAG) / Primary Lens Luxation (PLL) in the Shar Pei

Affected breeds: Shar Pei

In the Shar Pei POAG seems to develop alongside PLL, and the two conditions are caused by a single genetic mutation. Glaucoma is a serious eye condition in which there is a build-up of pressure in the eye caused by inadequate drainage of fluid from within the eyeball. In addition, the lens of the eye is not held securely and may fall out of position. This is a very painful condition which can lead to blindness. In affected dogs the condition becomes apparent at 4-6 years of age.

The mutation is recessive, which 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 POAG/PLL (“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 POAG/PLL

This test will be reported as:
- CLEAR: no evidence of the POAG/PLL mutation
- CARRIER: carries one copy of the defect, which will be passed to 50% of offspring
- AFFECTED: carries two copies of the defect, and will have POAG/PLL

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
- CLEAR X AFFECTED = 100% CARRIER
- CARRIER X AFFECTED = 50% AFFECTED, 50% CARRIER
- AFFECTED X AFFECTED = 100% AFFECTED

References
Evaluation of ADAMTS17 in Chinese Shar-Pei with primary open-angle glaucoma, primary lens luxation, or both.
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American Journal of Veterinary Research, January 2018, Vol. 79, No. 1, Pages 98-106