Cobalamin Malabsorption

Affected breeds: Australian Shepherd

This is a serious metabolic disease in which cobalamin (vitamin B_{12}) fails to be absorbed in the lower intestine. Signs of the disease usually become apparent at 6-12 weeks, but may not become obvious until adulthood. There are a range of varied clinical signs including lack of appetite, vomiting, wasting, listlessness and a general deterioration. Additional serious complications include degeneration of the immune and nervous systems. Due to the diverse clinical signs, Cobalamin Malabsorption can often be mis-diagnosed. Treatment of affected dogs involves routine injections of vitamin B_{12} for the rest of their life.

This is a serious disease caused by a recessive genetic mutation, but affected puppies can be avoided by selective breeding. Dogs which carry the mutation ("CARRIERS") are healthy but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will suffer from cobalamin malabsorption ("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 diagnose Cobalamin Malabsorption

This test will be reported as:

CLEAR : no evidence of the Cobalamin Malabsorption mutation
CARRIER : carries one copy of the mutation, which will be passed to 50% of offspring
AFFECTED : carries two copies of the mutation, and will develop Cobalamin Malabsorption

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

AFFECTED X AFFECTED	= 100% AFFECTED
AFFECTED X CARRIER	= 50% AFFECTED, 50% CARRIER
AFFECTED X CLEAR	= 100% CARRIER
CARRIER X CARRIER	= 25% AFFECTED, 50% CARRIER, 25% CLEAR
CARRIER X CLEAR	= 50% CARRIER, 50% CLEAR

References

He Q, Madsen M, Kilkenney A, Gregory B, Christensen EI, Vorum H, Hojrup P, Schaffer AA, Kirkness EF, Tanner SM, de la Chapelle A, Giger U, Moestrup SK, Fyfe JC (2005) Amnionless function is required for cubilin brush-border expression and intrinsic factor-cobalamin (vitamin B_{12}) absorption in vivo. Blood 106(4): 1447-1453