Imerslund-Gräsbeck Syndrome (IGS)

**Affected breeds:** Australian Shepherd, Border Collie

Also known as **Cobalamin Malabsorption**, affected pups typically fail to thrive and have anaemia, poor appetite, poor growth, weakness and intermittent diarrhoea. The non-specific nature of these signs probably means that the condition often goes undiagnosed. The basis for the disease is an inherent inability to absorb vitamin B12 in the digestive tract; these clinical signs can be ameliorated by regular injections of vitamin B12. Failure to treat leads to irreversible neurological damage.

IGS is caused by a recessive genetic mutation. This 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 have IGS (“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 Imerslund-Gräsbeck Syndrome

**This test will be reported as:**
- **CLEAR** : no evidence of the IGS 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 IGS

**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
- CLEAR X CLEAR = 100% CLEAR

**References**
