L-2-HGA (L-2-Hydroxyglutaric Aciduria)

Affected breeds: Staffordshire Bull Terrier

L-2-HGA is a neurometabolic disease which results in the increase of L-2-hydroxyglutaric acid in spinal fluid, blood plasma and urine. The age of onset is usually from about 4 months to one year of age. The condition results in a wobbly gait, seizures, ataxia, changes in behaviour, and tremors.

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 L-2-HGA ("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 L-2-HGA

This test will be reported as:

CLEAR : no evidence of the L-2-HGA 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 L-2-HGA

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

References

Penderis, J., Calvin, J., Abramson, C., Jakobs, C., Pettitt, L., Binns, M. M., Verhoeven, N. M., O'Driscoll, E., Platt, S. R., & Mellersh, C. S. (2007). L-2-hydroxyglutaric aciduria: characterisation of the molecular defect in a spontaneous canine model. Journal of medical genetics, 44(5), 334–340. https://doi.org/10.1136/jmg.2006.042507