Cystinuria

Affected breeds: Newfoundland

Cystinuria is an inherited metabolic disease which results in defective absorption in the kidneys. Consequently, cystine accumulates in the urine of affected individuals and crystallises to form stones in either the kidney or bladder. This leads to urinary obstruction which can be very painful and can lead to damage to the kidney or bladder. Cystinuria in the Newfoundland is particularly severe, with symptoms becoming evident at 6 – 12 months. Typically the males will suffer more severe signs than females.

Cystinuria is caused by a recessive genetic mutation. This means that 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 cystinuria ("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 cystinuria

This test will be reported as:

CLEAR : no evidence of the cystinuria 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 cystinuria

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

Henthorn PS, Liu J, Gidalevich T, Fang J, Casal ML, Patterson DF, Giger U (2000) Canine cystinuria: polymorphism in the canine SLC3A1 gene and identification of a nonsense mutation in cystinuric Newfoundland dogs. Human Genetics 107: 295 - 303