Cystinuria

Cystine is one of many amino acids that are building blocks for proteins. Amino acids are part of a normal canine diet and are absorbed through the gut. They are freely filtered in the kidney, but are normally reabsorbed (nearly 100%) by special kidney transporters and, therefore, are not lost.

In dogs or humans with Cystinuria, the kidney transporter for cystine is defective; and because cystine readily precipitates in acid urine, crystals and later calculi (stones) can form in the kidney and bladder. These calculi can result in serious illness. Cystinuria occurs in human patients and many dog breeds, but likely the most severe form has been observed in the Newfoundland Dogs.

Cystinuric dogs may show recurrent clinical signs of a urinary tract disorder from a few months of age until late in life. Dogs may experience difficulty in urination, have blood tinged urine, pass calculi or may unable to void urine despite numerous attempts. Due to their urogenital anatomy, male dogs may become completely blocked: their bladder gets extremely distended and is at risk for rupture. Furthermore, the severe urinary back-up pressure caused by the blockage may result in kidney failure. These serious complications often lead to death unless the animal receives immediate emergency and intensive care. Calculi lodged in the urethra may be flushed out or back into the bladder. In some cases, calculi in the bladder or kidney or urethra may have to be removed surgically. Special diets and medications may be helpful in preventing recurrence of calculi. Even treated dogs are likely to develop mild to life-threatening recurrences at least early in life.

Cystinuria is inherited as an autosomal recessive trait in the Newfoundland and likely other dog breeds. This means that diseased Newfoundland dogs of both genders have two mutant (diseased) genes. Their parents are either clinically healthy but carrying a normal and a mutant gene (carriers), or are also affected (cystinuric with two mutant genes). We are aware of at least one (deceased) stud Newfoundland dog that was affected and sired over 100 offspring, all of them being carriers.

Using a specific urinary screening method, the nitroprusside spot test, can make a diagnosis of Cystinuria, meaning, “affected”. This test requires only a small amount of urine (2-5 ml). You must contact one of the 3 labs for instructions for submission.

This urine test does not show whether a Newfoundland is clear or a carrier. Furthermore, stone laboratories can also analyze the voided or removed calculi (even when only a pinpoint in size) for cystine. These tests are very reliable. In contrast, examination by a veterinarian of the urine for cystine crystals is not completely reliable, since these crystals are not always present in the urine.

Finally, affected as well as carriers and clear in Newfoundland dogs can be identified by the recently developed DNA test.

In order to assist dog breeders and pet owners of Newfoundlands, there are three approved testing laboratories for testing for **DNA** (check swabs). They are:

1). The University of Pennsylvania (Dr. Urs Giger,) Section of Medical Genetics, 215-898-3375
2). VetGen Laboratories at www.VetGen.com
3). Veterinary Diagnostic Center 1-800-625-0874 who is the cheapest and seems to be the quickest.

This information has been compiled by the Southeastern Newfoundland Club, as part of their comprehensive rescue program. We are grateful for their generosity in sharing all of their hard work.- NCA 2006