

# Cystinuria

## What is it?

Cystinuria is a disorder in the urinary system occurring in Newfoundlands and other breeds, though the disease may have different origins between breeds (e.g. Mastiffs, French Bulldogs and other breeds have cystinuria with either dominant, or unknown inheritance. Most fundamentally, there is a higher concentration of the amino acid, cysteine, in the urine which can lead to cysteine crystals (stones). While both males and females are affected, males usually show the most dangerous effects due to the differences in anatomy. In fact, if unrecognized, in males it can lead to life-threatening urinary blockage.



## What causes it?

Many of the fundamental processes in the kidney work on a systematic principle. At the beginning of the process, many chemical byproducts are filtered into the developing urine. In more distal processes constituents which are conserved (e.g. sodium) are “resorbed” into the bloodstream from the filtrate. High concentrations of cysteine in the urine result from the inability to resorb cysteine. A mutation in a specific protein “carrier” which transports cysteine from the urine back into the bloodstream is the culprit. The Newfoundland community was fortunate in that a similar problem occurs in humans. This provided a “candidate” gene that proved the same in the Newfoundland and resulted in one of the early definitive genetic tests. The disease is completely genetic and is inherited as a simple recessive, i.e. it is only if both copies of the gene carry the defect that the disease results. The genetic test can distinguish clears (no mutation), carriers (one normal and one mutated, not showing disease) and affecteds (both copies carry the mutation and the dog has overt cystinuria).

## Signs & Symptoms

Clinical signs often don't appear until the dog is two years of age or even older. Males have more severe clinical signs than females because of the anatomical differences. Repeated UTI's, blood tinged urine, unsuccessful attempts to urinate are all possible symptoms. In males, the urinary blockade can be fatal.

## Prevention:

Given the genetic test, there is no reason for a reputable breeder to ever produce Newfoundlands with cystinuria. Although there is always some controversy in breeding carriers, if they are only bred to clear dogs (both copies normal) no offspring will have cystinuria. In theory puppies can be cleared by pedigree (i.e. if they are the offspring of 2 clear dogs, we know they are clear themselves) The OFA will accept one generation of clear by pedigree after which the genetic test must be performed. NCA policy allows the breeding of carriers to avoid removing superior dogs from the breeding genepool. It is expected that over a period of time carriers will be replaced by clear dogs of equal quality.

## Testing & Treatment

A test for cystine in the urine is available to test for the disorder even in the absence of overt clinical signs, although by far the most definitive test is direct detection of the mutated gene (see below).

Treatment may consist of a change in diet, urinary alkalinization/pH neutralization, and administration of cysteine-binding agents.

*This information is not meant to be a substitute for veterinary care.  
Always follow the instructions provided by your veterinarian.  
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