Hereditary Xanthinuria Test Result Interpretation

Each of the three xanthinuria tests screen for a specific DNA mutation in a specific gene. These mutations are all inherited in an autosomal recessive manner. We have designated the letter D to indicate the deleterious (xanthinuria) form of the gene and N to indicate the normal form of the gene. A dog's particular combination of N or D forms of the gene is known as its genotype.

Clear (N/N): Clear dogs have no copies of the mutation and cannot pass the mutation on to offspring. However, this test result does not rule out the possibility that a dog could be affected or a carrier for a different xanthinuria mutation.

Carrier (D/N): A carrier dog has only one copy of the mutation. Since the mutation is recessive, one copy will not cause xanthinuria and risk for xanthine stones. Carriers will, on average, pass the mutation on to half of their offspring. This does not mean that they need to be taken out of the breeding pool, but they should be bred to clear dogs to avoid producing affected puppies.

Affected (D/D): An affected dog has two copies of the mutation that causes xanthinuria (this is also referred to as being homozygous affected). They will pass on a copy of the mutation to all of their offspring, but if bred to clear dogs, the offspring will only be carriers. Many, but not all, dogs with xanthinuria will go on to form urinary stones and/or kidney disease. Signs can start at virtually any age from a few months onwards. Urinary stones cause irritation that may manifest as straining to urinate, frequent urination, urgency with urination, blood in the urine, or life-threatening urinary obstructions. If kidney damage is present, signs may manifest as excessive thirst and urination. Please see our website for basic information on management strategies.

http://z.umn.edu/xanthinuria

Breeding Outcomes

* Clear (N/N) x Clear (N/N) = 100% Clear (N/N)

* Clear (N/N) x Carrier (D/N) = 50% Clear (N/N), 50% Carrier (D/N)
(This is an average, individual litters may see anywhere from 100% Clear to 100% Carrier)

* Clear (N/N) x Affected (D/D) = 100% Carrier (D/N)

* Carrier (D/N) x Carrier (D/N) = 25% Clear (N/N), 50% Carrier (D/N), 25% Affected (D/D)
(This is an average, individual litters may see more or less of any result)

* Carrier (D/N) x Affected (D/D) = 50% Carrier (D/N), 50% Affected (D/D)
(This is an average, individual litters may see anywhere from 100% Carrier to 100% Affected)

* Affected (D/D) x Affected (D/D) = 100% Affected (D/D)