Primary Hyeroxaluria type 1 (PH1)

Description

Primary hyperoxaluria (PH) is a rare autosomal recessive disorder of glyoxylate. It is characterized by the accumulation of oxalate and subsequent precipitation of calcium oxalate crystals, primarily in the kidneys, leading to progressive kidney failure. If the storage capacity of the kidneys is exhausted, the crystals are accumulated in other tissues including bones, joints, cartilages, retina and muscles.

Symptoms

The symptoms may include intense abdominal pain radiating to the groin, blood that can be seen in the urine, and the passage of kidney stones.

Treatment

Treatment methods include medications, dietary modifications, and removal of kidney stones.

Test method

PH 1 testing is through DNA (blood and/or swab)

http://www.vetgen.com/ordertests.aspx?id=Coton+de+Tulear

Genetic/breeder information

AGXT is suspected as to be the candidate gene. Genetic testing of dogs is recommended before breeding to prevent the birth of affected offspring.

The PH I disease in Coton de Tulear is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive / positive) genotype only. The dogs with P/N (positive /negative) genotype are clinically without any symptom. They are genetically considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers). Because of high risk of producing affected offspring, mating of two N/P animals (carriers) cannot be recommended.

Stats within CdT breed

The mutation was tested in of 118 Finnish Coton de Tulear dogs, ten (8.5%) of which were revealed as carriers.

Source ofdata

http://www.genomia.cz/en/test/ph/

http://www.ncbi.nlm.nih.gov/pubmed/22486513