Detection of c.1623delG mutation in ATP13A2 gene causing NCL in Tibetan Terriers

Result: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation

Presence or absence of c.1623delG mutation in ATP13A2 gene causing Neuronal Ceroid Lipofuscinosis (NCL) in Tibetan Terriers was tested.

Mutation causing NCL in Tibetan Terriers is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs with N/P genotype are 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).

Method: SOP173-NCL-TT, PCR-RFLP

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