GENOMIA
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Result certificate #012345

Detection of c.977delC mutation FAM83H gene causing CKCSID in Cavalier King Charles spaniel

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 21-12345
Date received: 01.02.2021
Sample type: blood

Information provided by the customer
Name: Lassie DEMO
Breed: Plemeno
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 1.1.2020
Sex: female
Date of sampling: 01.02.2021
The identity of the animal has been checked.

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.977delC FAM83H gene causing CKCSID (congenital keratoconjunctivitis sicca and ichthyosiform dermatosis), commonly known as dry eye curly coat syndrome, in Cavalier King Charles Spaniel breed was tested. CKCSID is an inherited disorder characterized by defects of eyes, skin and claws. Affected dogs have reduced production of tears and tear film that may result in severe disorder of cornea. Ventral abdominal skin becomes hyperpigmented and hyperkeratinised. Persistent shedding of skin leads to scratching. Footpads are hyperkeratinised from young adulthood with claws growth abnormalities causing pain and lameness. Affected dogs also tend to suffer increased dental disease.

Mutation that causes CKCSID 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, 25 % P/P and 50 % N/P.

Method: SOP171-CKCSID, fragment analysis

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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