GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.910-1G>A mutation in EDA gene causing Ectodermal dysplasia in German Shepherds

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: PREVIEW RESULT LINE

Explanation

Presence or absence of c.910-1G>A mutation in EDA gene causing Ectodermal dysplasia in German Shepherds was tested. Ectodermal dysplasia is an inherited skin disease characterized by absent or abnormal teeth, impaired coat growth, missing sweat glands and reduced tear production. Puppies are born with symmetrical bald spots on the forehead and pelvic area, and often suffer from eye infections under closed eyelids. A major problem is the impaired mechanism for clearing matter from the respiratory tract, so-called mucociliary clearance (the ability to remove mucus by cilia), resulting in an increased susceptibility to pulmonary infections. Chronic nasal and ocular discharge or corneal ulceration is common.

The causal mutation of Ectodermal dysplasia is X-linked. This means that it is localized on the X chromosome. Males have an X and a Y chromosome, so they can only be healthy (Xn/Y) or affected (Xm/Y). Females have two X chromosomes, so they can either be healthy (Xn/Xn), carriers (Xn/Xm) or affected (Xm/Xm). Female carriers do not show clinical signs but are able to pass the mutant allele to their offspring.

Method: SOPAgriseq_canine, ngs, accredited method

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

verified and certified document
GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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