GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.583_584insC mutation in IL2RG gene causing XSCID in Cardigan and Pembroke Welsh Corgi

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: Xn/Xn

Explanation

Presence or absence of c.583_584insC mutation in IL2RG gene causing severe combined immunodeficiency disease (XSCID) in Cardigan and Pembroke Welsh Corgi was tested. This immune disorder is characterized by reduced lymphocyte counts with low (or absent) concentrations of IgG and IgA, but normal concentrations of IgM. Clinical findings include absence of palpable lymph nodes, small thymus size and chronic recurrent infections. Affected puppies do not thrive well, are plagued by diarrhoea, and occasional vomiting, and even with supportive care usually die by 4 months of age.

The mutation 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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