GENOMIA
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Result certificate #012345

Detection of c.90_92del mutation in RBP4 gene causing isolated microphthalmia with coloboma in Soft Coated Wheaten Terriers

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)

Explanation

Presence or absence of c.90_92del mutation in RBP4 gene causing isolated microphthalmia with coloboma in Soft Coated Wheaten Terriers was tested. Microphthalmia is an eye defect characterized by a marked under-development of the entire eye. Coloboma is a cleft defect of the iris and sometimes other parts of the eye (ciliary body, choroid or optic nerve). The manifestation is visual disturbances of varying severity, often myopia.

Pedigree analysis suggests an autosomal recessive mode of inheritance, but it was found that the disease only appears in homozygous offspring from a homozygous mother (P/P genotype). It is therefore a recessive defect with maternal transmission. The maternal effect arises from an impairment in the sequential transfer of retinol across the placenta via RBP4 encoded by maternal and fetal genomes. A dog with N/N result does not carry a mutation causing the disease, a dog with N/P result is a disease carrier.

Method: SOPAgriseq_canine, ngs

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 s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999