GENOMIA
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Result certificate #012345

Detection of c.2839del mutation in RELN gene causing cerebellar hypoplasia in White Swiss Shepherd Dogs

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.2839del mutation in RELN gene causing cerebellar hypoplasia in White Swiss Shepherd Dogs was tested. Cerebellar hypoplasia means incomplete development of the cerebellum. In White Swiss Shepherds, it occurs together with lissencephaly, a developmental defect characterised by an abnormal structure of the brain tissue, without the typical folds, co-called gyri and sulci on the cerebral cortex of the brain. The condition also accompanies moderate hydrocephalus with dilatation of the cerebral ventricles. The disease manifests itself in puppies as early as 2 weeks of age with ataxia.

Mutation that causes cerebellar hypoplasia is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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-RELN, fragment analysis

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

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