

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 Chondrodysplasia: N/CDPA**Result Chondrodystrophy and IVDD risk:** N/CDDY**Explanation**

The presence or absence of two insertion mutations in retrogene for fibroblast growth factor FGF4 in chromosomes 12 and 18 was examined. FGF4 retrogene is involved in many biological processes incl. bone development. Dogs that carry both mutations show a more drastic reduction of leg length (e.g. Basset Hounds, Dachshund, Welsh Corgi and Scottish terriers).

Chondrodysplasia (CDPA) is caused by the insertion of FGF4 retrogene into chromosome 18, is responsible for short legs in basset, welsh, dachshund, western, Scottish terrier and more. Inheritance of CDPA mutation is autosomal dominant, only one allele passed from one of the parents is sufficient to show the shortened legs.

Chondrodystrophy (CDDY) is caused by the insertion of FGF4 retrogen into chromosome 12 and explains the leg shortening of other breeds. The mutation is also connected with predisposition to abnormal growth and development of intervertebral discs. In affected dogs, premature calcification before 1 year of age results in loss of flexibility and gradual degeneration of intervertebral discs. These abnormal discs are predisposed to herniation into the spinal canal where the inflammation and haemorrhage can cause severe pain and neurological dysfunction termed Intervertebral Disc Disease of type I (IVDD).

The CDDY is inherited as a semi-dominant trait for height, meaning that dogs with 2 copies of the mutation (CDDY/CDDY) are smaller than dogs with only 1 copy (N/CDDY). As to predisposition to degeneration of intervertebral discs, the inheritance follows an autosomal dominant mode meaning that 1 allele obtained from one of its parents is sufficient to expressing the phenotype.

Chondrodysplasia result codes:

N/N	No copies of CDPA mutation.
N/CDPA	1 copy of CDPA. Mutation causes leg shortening compared to N/N dogs.
CDPA/CDPA	2 copies of CDPA. Mutation causes leg shortening compared to N/N dogs.

Chondrodystrophy (CDDY and IVDD Risk) result codes:

N/N	No copies of CDDY mutation.
N/CDDY	1 copy of CDDY mutation. Dog is at risk for IVDD. Mutation causes leg shortening compared to N/N dogs. When bred to an N/N dog will produce 50% of normal sized puppies and 50% of puppies at risk for IVDD.
CDDY/CDDY	2 copies of CDDY. Dog is at risk for IVDD. Mutation causes leg shortening compared to N/N dogs. Will produce 100% of puppies with shorter legs at risk for IVDD.

Method: SOP176-IVDD, ASA-PCR

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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