

Detection of g.52737379delC mutation in NIPAL4 gene causing congenital ichthyosis in American bulldog and American Bully

**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 g.52737379delC mutation in NIPAL4 gene causing congenital ichthyosis in American bulldog and American Bully was tested. Congenital ichthyosis is an inherited skin disorder. In the affected dogs, the outermost layer of the skin is not formed correctly. The typical signs are recognizable as early as 1 to 2 weeks of age. Compared to normal littermates, the affected dogs have a dishevelled fur and the glabrous skin of the abdomen is reddened. Complications due to infection caused by fungi of Malassezia genus. Together with the growth of the fungi, the inflammation of acoustic duct and itching infection of skin folds or paws develop and even hyperkeratosis may develop in adult dogs.

Mutation that causes congenital ichthyosis is inherited as an autosomal recessive trait. That means the 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

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



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