

**Detection of mutation 6.47 Mb inversion in
FAM134B gene causing Sensory
Neuropathy in Border Collies****Customer:** Dr. Josette Debrincat, 20, Animal Doctors, Guze Bajada, HMR 2131 Hamrun, Malta**Sample:**

Sample: 24-26623

Date received: 01.10.2024

Sample type: blood

Information provided by the customer

Name: Scott Dell'Antica Contrada**Breed:** Border Collie

Microchip: 380 206 044 889 554

Reg. number: MKC BOR/14/2024

Date of birth: 18/12/2022

Sex: male

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 mutation 6.47 Mb inversion in FAM134B gene causing Sensory Neuropathy (SN) in Border Collies was tested. Sensory neuropathy is a severe neurologic disease caused by degeneration of sensory and, to a lesser extent, motor nerve cells. Affected dogs start to show symptoms from 2 to 7 months of age and signs include progressive loss of coordination, joint laxity and extreme stretching of limb muscles. The affected dogs are not able to feel the stretching of individual muscles and ligaments (loss of proprioception). Moreover, the affected dogs lose sensation of pain (loss of pain receptor, nociceptors) which leads to self-mutilation of paws.

Mutation that causes SN in Border Collies 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: SOP171-SN, fragment analysis

Date of issue: 15.10.2024

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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