

Customer: Natálie Jičínská, Sovolusky 3, 535 01 Sovolusky, Czech Republic**Sample:**

Sample: 25-07931

Date received: 01.04.2025

Sample type: buccal swab

Information provided by the customer

Name: Birgit Ginger Storm**Breed:** Belgian Shepherd - Malinois

Microchip: 203 098 100 455 268

Reg. number: CMKU/BOM/11340/19

Date of birth: 04.06.2019

Sex: female

Date of sampling: 29.03.2025

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.-6582_*516del mutation in SELENOP gene causing central nervous system atrophy with cerebellar ataxia (CACA) in Belgian Shepherds was tested. Clinical signs, such as uncoordinated movements, intention tremors, general elevated muscle tone and reduced swallowing reflex can be observed in puppies at 2 weeks of age. Gradual progression leads to such a degree of severity of clinical signs that the animals must be euthanized. The SELENOP gene encodes selenoprotein P, which is involved in the deposition and transport of selenium to the brain and other organs. Incorporation of selenium into selenoprotein P prevents the toxic effects of free selenium. A deletion in the SELENOP gene results in the complete absence of encoded selenoprotein P and causes a defect in selenium transport to the CNS. Blood selenium concentration is reduced by 70% in affected homozygotes and 30% in heterozygotes compared to healthy dogs.

Mutation that causes CACA 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: SOPAgriseq_canine, ngs

Date of issue: 10.04.2025

Date of testing: 01.04.2025 - 10.04.2025

Approved by: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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