GENOMIA
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Result certificate #260967

Detection of c.118G>A mutation
in SOD1 gene causing
degenerative myelopathy in dogs

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

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.118G>A mutation in SOD1 gene causing degenerative myelopathy in many canine breeds was tested. This mutation is sometimes referred to as SOD1A. Affected dogs have progressive loss of movement and gradual worsening of the condition up to complete paralysis. The age of disease onset and symptoms severity vary among the breeds.

Mutation SOD1A 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).

The test does not exclude existence of another, nowadays unknown, mutation which can cause DM. In Bernese Mountain Dogs, there has been identified also SOD1B mutation responsible for DM - this test does not refer about SOD1B.

Analysis was performed by the partner laboratory. Genomia guarantees the quality of its partner's services.

Method: SOPAgriseq_canine, ngs

Date of issue: 24.04.2024
Date of testing: 17.04.2024 - 24.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Zpráva o výsledku zkoušky #260959

Detekce mutace c.1393C>T genu DNM2
způsobující CNM u border kolií

Zákazník: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Vyšetřovaný:
Vzorek: 22-13019
Datum přijetí vzorku: 18.05.2022
Vyšetřovaný materiál: krev

Údaje poskytnuté zákazníkem
Jméno: Hyperion z Říše fantazie
Rasa: Border kolie
Mikročip: 203 098 100 491 053
Datum narození: 23.3.2022
Pohlaví: samec
Datum odběru: 10.05.2022
Při odběru byla ověřena identita jedince.
Ověřil/a MVDr. Miroslava Sůvová, KVL 5816

Výsledek: Mutace nebyla detekována (N/N)

Komentář k výsledku

Byla vyšetřena přítomnost či absence mutace c.1393C>T genu DNM2 způsobující centronukleární myopatii (CNM) u border kolií. CNM je defekt vývoje svalových vláken. Zpočátku se projevuje nesnášenlivostí svalové zátěže, slabostí kosterního svalstva a mírně narušenou chůzí. Onemocnění je progresivní a způsobuje svalovou atrofii a strukturální anomálie svalových vláken, včetně jaderné centralizace a mitochondriálních abnormalit.

Mutace způsobující CNM je děděna autosomálně dominantně. To znamená, že k projevení příznaků onemocnění stačí jedna kopie mutovaného genu zděděná od jednoho z rodičů.

Metoda: SOPAgriseq_canine, ngs

Datum vystavení zprávy: 24.04.2024
Datum provedení zkoušky: 17.04.2024 - 24.04.2024
Schválila: Mgr. Martina Šafrová, vedoucí laboratoře

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Result certificate #260965

Detection of c.590G>A mutation in OLFML3 gene related with Goniodysgenesis and Glaucoma in Border Collies

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

Result: Mutation was not detected (N/N)

Explanation

Presence or absence of c.590G>A mutation in OLFML3 gene related with Goniodysgenesis and Glaucoma in Border Collies was tested. Goniodysgenesis is a hereditary disorder characterized by development abnormalities of anterior chamber. Due to abnormal development of intraocular fluid egress channels inside the eye the iridocorneal angle, through which the excessive chamber fluid is filtered and drained, get narrower or closed. Goniodysgenesis is significantly associated with the glaucoma and blindness.

Goniodysgenesis occurs in severe and mild forms. Severe goniodysgenesis potentially leading to glaucoma is connected with homozygosis for c.590A allele of OLFML3-gene which indicates autosomal recessive mode of inheritance. The vast majority of dogs with severe goniodysgenesis and glaucoma are homozygous for the mutation mentioned, however there are some cases of heterozygotes affected with this disease. The exact mode of inheritance has not been elucidated yet.

Result options: N/N healthy dog, N/P carrier of disposition to goniodysgenesis, P/P dog in risk of goniodysgenesis development.

Method: SOPAgriseq_canine, ngs, accredited method

Date of issue: 24.04.2024
Date of testing: 17.04.2024 - 24.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #260966

Detection of c.8392delC mutation in the CUBN gene causing IGS in border collies

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

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.8392delC mutation in the CUBN gene causing IGS (Imerslund-Gräsbeck syndrome) or intestinal cobalamin malabsorption in border collies was tested. IGS is metabolic disorder in border collies. Signs appear early in 6 to 12 week of dog´s age and include failure to thrive and chronic loss of appetite. The affected dogs can suffer from neutropia, non-regenerative anaemia, anisocytosis and poikilocytosis, megaloblastic changes in bone marrow, reduction of Cbl level, methylmalonic aciduria and homocysteinemia.

Mutation that causes IGS 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), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/P.

Method: SOPAgriseq_canine, ngs

Date of issue: 24.04.2024
Date of testing: 17.04.2024 - 24.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #260958

Detection of c.2841delT mutation in DMD gene causing DMD in Border Collies

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

Result: Xn/Y

Explanation

Presence or absence of c.2841delT mutation in DMD gene causing Duchenne muscular dystrophy (DMD) in Border Collies was tested. The disease is characterized by progressive muscle weakness that is ultimately fatal. Clinical signs begin to appear in puppies between 8 and 10 weeks of age and include a stiff gait or shortened stride, inability to fully open the jaw, difficulty swallowing, excessive salivation and marked wasting of the muscles of the body and limbs.

The mutation is X-linked. This means that it is localized on the X chromosome. Males have an X and a Y chromosome, so they can only be healthy (Xn/Y) or affected (Xm/Y). Females have two X chromosomes, so they can either be healthy (Xn/Xn), carriers (Xn/Xm) or affected (Xm/Xm). Female carriers do not show clinical signs but are able to pass the mutant allele to their offspring.

Method: SOPAgriseq_canine, ngs

Date of issue: 24.04.2024
Date of testing: 17.04.2024 - 24.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #260964

Detection of mutation c.228_231del
in ABCB1 gene causing
drug sensitivity in dogs

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

Result: Mutation was not detected (N/N)

Explanation

It has been studied the presence and absence of mutation c.228_231del in ABCB1 gene leading to defect of P-glycoprotein. P-glycoprotein is a membrane drug transporter and a very important component of the blood brain barrier that prevents entry of many potentially toxic compounds into the central nervous system. The dysfunction of P-glycoprotein in dogs can result in potentially fatal neurotoxic reaction, especially following the administration of ivermectin, acepromazin, butorphanol, doramectin, doxorubicin, loperamid, milbemycin, moxidectin, selamectin, vinblastin and vincristin.

The sensitivity to drugs develops in dogs with mutation in both copies of MDR1 gene (P/P). Some dogs that are heterozygotes (N/P) have shown adverse reaction after administration of some drugs. The specific cause of this variation is not known so far – other gene mutations, general health conditions and dosage.
It is not possible to exclude existence of other mutations of ABCB1 gene in various breeds (in Bordier collies, another two mutations have been found). Compound heterozygotes that carry two distinct mutations of ABCB1 gene may occur, where each mutation was inherited from one of the parents. The compound heterozygotes also have defective P-glycoprotein function.
The defect occurs in Collies, Longhaired Whippets, Australian Shepherds, Miniature Australian Shepherds, McNab Shepherd dogs, Silken windhounds, English sheepdogs, Shelties, German shepherd dogs, Bobtails, Border Collies and herding breed cross.

Method: SOPAgriseq_canine, ngs

Date of issue: 24.04.2024
Date of testing: 17.04.2024 - 24.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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GENOMIA
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Result certificate #260963

Detection of c.619C>T mutation in CLN5 gene causing NCL5 in border collies and australian cattle dogs

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

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 c.619C>T in CLN5 gene causing Neuronal Ceroid Lipofuscinosis type 5 (NCL5) in border collies and australian cattle dogs was tested. NCL is a neurodegenerative disorder that is characteristic by accumulation of lipopigments (coroid and lipofuscin) in the lysosomes. The beginning and clinical course of the disease are very individual. The rate of neurodegeneration increases together with the age. Mental abnormalities and ataxia usually develop in all affected dogs. Increased restlessness, aggression, hallucinations, hyperactivity and epileptic attacks can be observed as well. Accompanying symptom is damaged retina due to lipopigment storage. Affected individuals rarely survive more than 28th month of age.

Mutation that causes NCL5 is inherited autosomally recessively which means that the disease develops only in 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: 24.04.2024
Date of testing: 17.04.2024 - 24.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #260960

Detection of mutation
g.4411956_441190delGTTT in VPS13B gene
causing TNS in Border collies

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

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.4411956_4411960delGTTT in exon 19 of VPS13B gene causing Trapped Neutrophil Syndrome (TNS) in Border collie breed was tested. Due to this mutation the correct function of white corpuscles - neutrophils - is impaired. They take part in fighting bacterial infections and are important participants in acute inflammation. The failing of immune system can be seen in pups from as early as 2 weeks old and the pups die or are euthanized by approx. 4 months of age. The first symptoms may include apathy, loss of appetite, diarrhoea or poor mobility. Other symptoms depend on the type of infection the pup happens to contract.

Mutation that causes TNS 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, 25 % P/P and 50 % N/P.

Method: SOPAgriseq_canine, ngs
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

Date of issue: 24.04.2024
Date of testing: 17.04.2024 - 24.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #260962

Detection of c.899C>T mutation in FAM20C gene causing Raine-syndrome in Border Collies

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

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.899C>T mutation in FAM20C gene causing dental hypomineralization, called Raine-syndrome, in Border Collies was tested. Disease causes extensive wear of teeth, cracking of tooth enamel, brownish spots or brownish discolouration of teeth or dental pulp inflammation. Severe tooth wear leads to chronic inflammation of the pulp up to the loss of teeth.

Mutation that causes Raine-syndrome 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), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 50 % N/P (healthy carriers) and 25 % P/P (affected).

Method: SOPAgriseq_canine, ngs

Date of issue: 24.04.2024
Date of testing: 17.04.2024 - 24.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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genetic laboratory

Result certificate #260968

Detection of g.28697542-28705340del7799 mutation in NHEJ1 gene causing CEA in several dog breeds

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

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.28697542-28705340del7799 mutation in NHEJ1 gene causing Collie eye anomaly (CEA) was tested. CEA is known to affect Australian Shepherd, Border Collie, Boykin Spaniel, Lancashire heeler, Longhaired whippet, Nova Scotia Duck Tolling retriever, Rough and Smooth Collie, Shetland Sheepdog and Silken windhound.

Mutation that causes CEA 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).

Analysis was performed by the partner laboratory. Genomia guarantees the quality of its partner's services.

Method: SOP176-CEA, ASA-PCR

Date of issue: 26.04.2024
Date of testing: 17.04.2024 - 26.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #260961

Detection of mutation 6.47 Mb inversion in FAM134B gene causing Sensory Neuropathy in Border Collies

Customer: Marie Reisingerová, Hasičská 34, 463 34 Hrádek nad Nisou - Dolní Sedlo, Czech Republic
Sample:
Sample: 22-13019
Date received: 18.05.2022
Sample type: blood

Information provided by the customer
Name: Hyperion z Říše fantazie
Breed: Border Collie
Microchip: 203 098 100 491 053
Date of birth: 23.3.2022
Sex: male
Date of sampling: 10.05.2022
The identity of the animal has been checked by MVDr. Miroslava Sůvová, KVL 5816

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. Senzory 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 loose 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: 30.04.2024
Date of testing: 17.04.2024 - 30.04.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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