GENOMIA
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Result certificate #171478

Detection of c. 916C>T gene variants in MC1R gene (locus E) responsible for dog coat color

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

Result: Based on gene variants examination genotype was determined E/e

Explanation

Presence of c.916C>T MC1R gene variants (melanocortin 1 receptor) was tested. The gene region is called locus E (Extension). Wild-type allele is called E; recesive gene variant c.916T is called e. Genotype e/e is manifested by red or yellow dog coat color.

Phenotype of e allele is inherited as an autosomal recessive trait and is manifested only in e/e individuals (they need to inherit gene variant c.916T from both parents). Heterozygous individuals E/e are carriers of allele e.

Method: SOP173-MC1R, PCR-RFLP

Date of issue: 10.03.2021
Date of testing: 25.02.2021 - 10.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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genetic laboratory

Result certificate #171479

Detection of d1-allele in D-locus affecting the coat colour in dogs

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

Result: D/D

Explanation

It has been examined the presence of gene variants c.-22G>A of MLPH-gene (melanophilin gene) causing coat colour dilution in dogs. The dilution is caused by d1-allele at D-locus (Dilution). The MLPH-gene is responsible for the density of pigment granules (eumelanine) in a hair. The presence of the gene variant c.-22A, d1-allele, causes the loss of pigment granules in a hair; the original black colour is diluted to blue and brown colour to lilac.

The phenotypic expression of d1-allele is inherited autosomal recessively. The colour dilution occurs only in d1/d1-dogs that inherit d1-allele from each of its parents. The dilution is not expressed in heterozygous dogs D/d1, however these dogs are carriers of this trait. Dogs with D/D result do not carry dilution.

There is other MLPH-gene variant c.705C (d2-allele) that is responsible for colour dilution in various dog breeds. The diluted dogs are also compound heterozygous d1/d2, where the d1-allele is inherited from one parent and d2-allele from the other parent.

There will be probably discovered other gene variants responsible for colour dilution. The final colour of a dog is affected by the presence of alleles at other loci (E, B, A, K and other).

Method: SOP175-MLPH, real-time PCR-ASA

Date of issue: 10.03.2021
Date of testing: 25.02.2021 - 10.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #171480

Detection of canine TYRP1 (locus B) gene variants influencing coat color

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

Result: B/B

Explanation

Presence of TYRP1 gene (locus B) variants c.991C>T (allele b^s), c.1033_1036delCCT (allele b^d) and c.121T>A (allele b^c) causing brown coat or nose color was examined. It is a set of locus B (Brown) alleles. Wild type allele is called B.

- If the result is B/B the individual does not carry brown color.
- If the result is B/b^c or B/b^d, B/b^s the individual carries brown color.
- If the result is b^c/b^c or b^d/b^d or b^s/$b^s$the individual is brown colored.
- If the result contains two or more different b-alleles the individual could be either carrier of brown color without brown color phenotype (b-alleles are inhered from one parent only) or is brown colored (b-alleles are inhered from both parents). It is not possible to summarize locus B genotype without testing the parents.

Phenotype of b allele (brown color) is inherited as a autosomal recessive trait. This examination does not exclude existence of any unknown variant of TYRP1 gene causing brown coat and nose color. Final coat color is influenced also by other loci (A, E, D, K).

Method: SOP182-TYRP1,173-TYRP1, PCR-RFLP

Date of issue: 09.03.2021
Date of testing: 25.02.2021 - 09.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #171481

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

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

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: SOP172-NCL5, direct DNA sequencing

Date of issue: 09.03.2021
Date of testing: 25.02.2021 - 09.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #171482

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

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

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: 05.03.2021
Date of testing: 25.02.2021 - 05.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #171483

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

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

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: SOP171-TNS, fragment analysis, accredited method
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: 10.03.2021
Date of testing: 25.02.2021 - 10.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #171484

Detection of mutation c.295_298delAGAT
in ABCB1 gene causing
drug sensitivity in dogs

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

Result: Mutation was not detected (N/N)

Explanation

It has been studied the presence and absence of mutation c.295_298delAGAT in ABCB1 gene leading to defect of P-glycoprotein (previous nomenclature: c.227_230delATAG MDR1). 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: SOP175-MDR1, ASA-PCR, accredited method

Date of issue: 10.03.2021
Date of testing: 25.02.2021 - 10.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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Result certificate #171485

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

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

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: SOP171-IGS-border, fragment analysis

Date of issue: 10.03.2021
Date of testing: 25.02.2021 - 10.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #171486

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

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

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: 10.03.2021
Date of testing: 25.02.2021 - 10.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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genetic laboratory

Result certificate #171487

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

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

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: SOP172-OLFML3, direct DNA sequencing, accredited method

Date of issue: 02.03.2021
Date of testing: 25.02.2021 - 02.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Result certificate #171488

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

Customer: Karolína Pincová, Koleč 180, 27329 Koleč, Czech Republic
Sample:
Sample: 21-05508
Date received: 25.02.2021
Sample type: buccal swab

Information provided by the customer
Name: Wedgewood's Warrior Within Sneaks Thru The Night
Breed: Border Collie
Microchip: 956 000 011 899 301
Reg. number: CMKU/BOC/14525/-20/19
Date of birth: 21.5.2019
Sex: male
Date of sampling: 25.02.2021
The identity of the animal has been checked by Ing. Jana Kůsová, Genomia s.r.o.

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: SOP173-raine-syndrome, PCR-RFLP

Date of issue: 26.02.2021
Date of testing: 25.02.2021 - 26.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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