

GENETIC ANALYSIS DNA PROFILE REPORT



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OWNER'S DETAILS

Zeljka Rakonic Goldin
10 Dvojak Purarije 26B
New South Wales HR

ANIMAL'S DETAILS

Registered Name:	DB Never Stop Dream for ingold's	Pet Name:	Kia
Registration Number:	HR1064850C	Breed:	Border Collie
Microchip Number:	380260042869811	Sex:	Intact Female
Date of Birth:	0/0/0	Colour:	

COLLECTION DETAILS

Case Number:	17109233	Date of Test:	01/01/2018
Approved Collection Method:	YES	Collected By:	Dr. Filip Tkalec

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

DNA PROFILE The DNA Profile below represents the genetic identification of DB Never Stop Dream for ingold's

P1_2 A G P3_2 A A P3_3 G G P11_3 C C P12_1 A A P24_2 A A P12_3 G G P30_3 A A
P13_1 C C P24_3 C C P31_1 A A P28_3 A A P31_3 G G P25_1 G G P32_2 C C P13_2 A A
P13_3 A C P25_2 G G P25_3 A C P32_3 A A P33_1 G G P14_1 T T P10_1 G G P26_1 G G
P33_3 G G P26_2 A A P14_2 G G P26_3 A G P14_3 C C P15_1 A A P34_1 A A P34_2 G G
P34_3 A C P10_3 C C P15_2 G G P15_3 A C P16_3 C C P35_1 G G P35_2 G G P36_1 A C
P17_1 G G P36_2 C G P37_2 G G P17_2 C C P29_1 G G P37_3 A G P38_1 C C P38_2 A G
P27_1 G G P17_3 G G P27_2 A C P4_3 A G P18_2 C C P18_3 C C P5_1 G G P11_1 A G
P19_1 T T P19_2 A G P5_2 G G P19_3 G G P2_1 G G P2_3 A C P27_3 A T P20_1 A A
P20_3 A A P5_3 G G P11_2 C C P6_2 G G P6_3 C C P21_1 A G P21_3 A G P22_2 A A
P28_1 G G P7_1 C C P7_2 A A P28_2 C G P7_3 A A P29_2 G G P8_1 A A P22_3 C C
P8_2 G G P8_3 A A P23_1 G G P9_3 T T P23_2 C C P23_3 A G P24_1 A G P3_1 G G

RESULTS REVIEWED AND CONFIRMED BY:

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GENETIC ANALYSIS SUMMARY

TESTS REPORTED

RESULT ¹

¹**Please Note:** This is a summary disease and trait report. To view more details on each test, including a DNA profile, please log in to your account and view the detailed single DNA report.

Metabolic (Associated with the Body's Enzymes and Cell Metabolism)

COBALAMIN MALABSORPTION: CUBILIN DEFICIENCY NEGATIVE / CLEAR [NO VARIANT DETECTED]

Ophthalmologic (Associated with the Eyes)

COLLIE EYE ANOMALY/CHOROIDAL HYPOPLASIA NEGATIVE / CLEAR [NO VARIANT DETECTED]

PRIMARY LENS LUXATION NEGATIVE / CLEAR [NO VARIANT DETECTED]

Neurologic (Associated with the Brain, Spinal and Nerves)

DEGENERATIVE MYELOPATHY NEGATIVE / CLEAR [NO VARIANT DETECTED]

IVERMECTIN SENSITIVITY MDR1 (MULTI DRUG RESISTANCE) NEGATIVE / CLEAR [NO VARIANT DETECTED]

NEURONAL CEROID LIPOFUSCINOSIS 5 NEGATIVE / CLEAR [NO VARIANT DETECTED]

Musculoskeletal (Associated with Bones and Muscles)

MYOTONIA CONGENITA CLCN1 NEGATIVE / CLEAR [NO VARIANT DETECTED]

Immunological (Associated with the Immune System)

TRAPPED NEUTROPHIL SYNDROME NEGATIVE / CLEAR [NO VARIANT DETECTED]

Haemolympathic (Associated with the Circulatory System)

VON WILLEBRAND'S DISEASE TYPE II NEGATIVE / CLEAR [NO VARIANT DETECTED]

Trait (Associated with Phenotype)

A LOCUS (FAWN/SABLE;TRI/TAN POINTS) a¹/a¹ - TAN POINTS - TAN POINTS or TRICOLOUR MAY BE BRINDLED [SEE K LOCUS]

BROWN (345DELPRO) DELETION BB^d - DOES NOT CARRY BROWN or CHOCOLATE (DELETION)

BROWN (GLNT331STOP) STOP CODON BB^s - DOES NOT CARRY BROWN or CHOCOLATE (STOP CODON)

BROWN (SER41CYS) INSERTION CODON BB^c - DOES NOT CARRY BROWN or CHOCOLATE (INSERTION)

D (DILUTE) LOCUS DD - NO COPY OF MLPH-D ALLELE (DILUTE) - PIGMENT IS NORMAL

E LOCUS - (CREAM/RED/YELLOW) Ee - CARRIES EXTENSION (YELLOW/WHITE/APRICOT/RUBY/RED)

K LOCUS (DOMINANT BLACK) KK - DOMINANT BLACK - SOLID [WILL NOT BE BRINDLED or EXPRESS AGOUTI]

LONG HAIR GENE (CANINE) POSITIVE - SHOWING THE PHENOTYPE

Dermatologic (Associated with Skin)

BLACK HAIR FOLLICULAR DYSPLASIA NEGATIVE - NOT SHOWING THE PHENOTYPE

GENETIC ANALYSIS

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Cobalamin Malabsorption: Cubilin Deficiency
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : CUBN on Chromosome 2
Variant Detected : Deletion of C

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Collie Eye Anomaly/Choroidal Hypoplasia
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : NHEJ1 on Chromosome 37
Variant Detected : Deletion

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Degenerative Myelopathy
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : SOD1 on Chromosome 31
Variant Detected : Base Substitution A>T and G>A

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Ivermectin Sensitivity MDR1 (Multi Drug Resistance)
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : MDR1 on Chromosome 14
Variant Detected : Deletion 4bp AGAT

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

SAMPLE COLLECTION DETAILS

Case Number: 17109233
Approved Collection Method: YES

Date of Test: 01/01/2018
Collected By: Dr. Filip Tkalec



Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Primary Lens Luxation
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : ADAMTS17 on Chromosome 3
Variant Detected : Base Substitution G>A

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Trapped Neutrophil Syndrome
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : VPS13B on Chromosome 13
Variant Detected : Deletion of GTTT

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Long Hair Gene (Canine)
Result: POSITIVE - SHOWING THE PHENOTYPE¹
Gene : FGF5
Variant Detected : Base Substitution L > I (Point Mutation)

¹ The phenotype/trait tested is present. Please Note this can vary from breed to breed and within breed.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: E Locus - (Cream/Red/Yellow)
Result: Ee - CARRIES EXTENSION (YELLOW/WHITE/APRICOT/RUBY/RED)¹
Gene : MC1R
Variant Detected : Em (point mutation) > E (wild type) > e (point mutation)

¹ One copy of black (E) and one copy of red/yellow/cream/apricot/white. These "e" colours are dependent on breed. The "e" allele is non-functional. May produce yellow/white/apricot/ruby or red offspring if mated to another carrier of "e".

SAMPLE COLLECTION DETAILS

Case Number: 17109233
Approved Collection Method: YES

Date of Test: 01/01/2018
Collected By: Dr. Filip Tkalec



Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: K Locus (Dominant Black)
Result: KK - DOMINANT BLACK - SOLID [WILL NOT BE BRINDLED or EXPRESS AGOUTI]¹
Gene : CBD103
Variants Detected : Deletion of GGG

¹ Two copies of dominant black (K) are present. No brindle/red or fawn offspring will be produced. Will not express Agouti phenotype. This can also be referred to as KB. In some breeds the K locus is fixed so all dogs will be KK. This (the K Locus) can be modified by other genes eg. liver, dilute, greying or merle. Red can only be added through the e locus.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Brown (345DELPRO) Deletion
Result: BB^d - DOES NOT CARRY BROWN or CHOCOLATE (DELETION)¹
Gene : TYRP1
Variants Detected : Base Substitution (Point Mutation)

¹ Does not carry brown – cannot have brown offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Brown (GLNT331STOP) Stop Codon
Result: BB^s - DOES NOT CARRY BROWN or CHOCOLATE (STOP CODON)¹
Gene : TYRP1
Variants Detected : Point Mutation

¹ Does not carry brown – cannot have brown offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Brown (SER41CYS) Insertion Codon
Result: BB^c - DOES NOT CARRY BROWN or CHOCOLATE (INSERTION)¹
Gene : TYRP1
Variants Detected : Base Substitution (Point Mutation)

¹ Does not carry brown – cannot have brown offspring.

SAMPLE COLLECTION DETAILS

Case Number: 17109233
Approved Collection Method: YES

Date of Test: 01/01/2018
Collected By: Dr. Filip Tkalec



Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: D (Dilute) Locus
Result: DD - NO COPY OF MLPH-D ALLELE (DILUTE) - PIGMENT IS NORMAL¹
Gene : MLPH
Variant Detected : Base Substitution

¹ Full colour, no dilute gene present. The D allele modifies the Melanophillin (MLPH) gene. This animal cannot produce "dilute" offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: A Locus (Fawn/Sable;Tri/Tan Points)
Result: a^t/a^t - TAN POINTS - TAN POINTS or TRICOLOUR MAY BE BRINDLED [SEE K LOCUS]¹
Gene : ASIP
Variant Detected : Base Substitution 246 G>T(A82S); G>A (R83H): C>T (p.R96C)

¹ Homozygous for black and tan/tricolour (no hidden colours) allele. Tri factored/white factored in dogs that have white points. No Bi Factoring (Black White & Tan). Animals are primarily black and have areas of pheomelanin (tan) which tends to be seen on the leg and stomach areas, the side of the head and spots above the eyes. Please note the colour and distribution of pheomelanin "tan" will be dependent on the breed and other colour genes.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Black Hair Follicular Dysplasia
Result: NEGATIVE - NOT SHOWING THE PHENOTYPE¹
Gene : RAB27
Variant Detected : Base Substitution G>A

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Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: Myotonia Congenita CLCN1
Result: NEGATIVE / CLEAR [NO VARIANT DETECTED]¹
Gene : CLCN1
Variant Detected : Deletion of A

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

SAMPLE COLLECTION DETAILS

Case Number: 17109233
Approved Collection Method: YES

Date of Test: 01/01/2018
Collected By: Dr. Filip Tkalec



Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: **Neuronal Ceroid Lipofuscinosis 5**
Result: **NEGATIVE / CLEAR [NO VARIANT DETECTED]¹**
Gene : **CL5**
Variant Detected : **Base Substitution G>A**

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

Sample with Lab ID Number 17109233 was received at Orivet Genetics, DNA was extracted and analysed with the following result reported:

Test Reported: **von Willebrand's Disease Type II**
Result: **NEGATIVE / CLEAR [NO VARIANT DETECTED]¹**
Gene : **VWF**
Variant Detected : **Base Substitution G>A**

¹ We have scanned the DNA and the genotype of this animal is NORMAL - no presence of the disease associated variant (mutation) has been detected. This result may also be referred to as NORMAL, "-/-" or "wild type (WT)" or "homozygous negative". The animal is clear of the disease and will not pass on the disease-causing variant. Can be mated with an untested animal and WILL NOT produce any positive/affected offspring.

SAMPLE COLLECTION DETAILS

Case Number: 17109233
Approved Collection Method: YES

Date of Test: 01/01/2018
Collected By: Dr. Filip Tkalec



EXPLANATION of RESULT TERMINOLOGY

The terms below are provided to help clarify certain results phrases on your genetic report. The phrases below are those as reported by Orivet and may vary from one laboratory to the other.

NEGATIVE / CLEAR [NO VARIANT DETECTED]

No presence of the variant (mutation) has been detected. The animal is clear of the disease and will not pass on any disease-causing mutation.

CARRIER [ONE COPY OF THE VARIANT DETECTED]

This is also referred to as HETEROZYGOUS. One copy of the normal gene and copy of the affected (mutant) gene has been detected. The animal will not exhibit disease symptoms or develop the disease. Consideration needs to be taken if breeding this animal - if breeding with another carrier or affected or unknown then it may produce an affected offspring.

POSITIVE / AT RISK [TWO COPIES OF THE VARIANT DETECTED]

Two copies of the disease gene variant (mutation) have been detected also referred to as HOMOZYGOUS for the variant. The animal may show symptoms (affected) associated with the disease. Appropriate treatment should be pursued by consulting a Veterinarian.

POSITIVE HETEROZYGOUS [ONE COPY OF THE DOMINANT VARIANT DETECTED]

Also referred to as POSITIVE ONE COPY or POSITIVE HETEROZYGOUS. This result is associated with a disease that has a dominant mode of inheritance. One copy of the normal gene (wild type) and affected (mutant) gene is present. Appropriate treatment should be pursued by consulting a Veterinarian. This result can still be used to produce a clear offspring.

POSITIVE HOMOZYGOUS [TWO COPIES OF THE DOMINANT VARIANT DETECTED]

Also referred to as POSITIVE HOMOZYGOUS. Two copies of the disease gene variant (mutant) have been detected and the animal may show symptoms associated with the disease. Please Note: This disease has dominant mode of inheritance so if mated to a clear animal ALL offspring will be AFFECTED – HETEROZYGOUS ONE COPY.

NORMAL BY PARENTAGE HISTORY

The sample submitted has had its parentage verified by DNA. By interrogating the DNA profiles of the Dam, Sire and Offspring this information together with the history submitted for the parents excludes this animal from having this disease. The controls run confirm that the dog is NORMAL for the disease requested.

NORMAL BY PEDIGREE

The sample submitted has had its parentage verified by Pedigree. The pedigree has been provided and details (genetic testing reports) of the parents have been included. Parentage could not be determined via DNA profile as no sample was submitted.

NO RESULTS AVAILABLE

Insufficient information has been provided to provide a result for this test. Sire and Dam information and/or sample may be required. This result is mostly associated with tests that have a patent/license and therefore certain restrictions apply. Please contact the laboratory to discuss.

INDETERMINABLE

The sample submitted has failed to give a conclusive result. This result is mainly due to the sample failing to "cluster" or result in the current grouping. A recollection is required at no charge.

DNA PROFILE

Also known as a DNA fingerprint. This is unique for the animal. No animal shares the same DNA profile. An individual's DNA profile is inherited from both parents and can be used for verifying parentage (pedigrees). This profile contains no disease or trait information and is simply a unique DNA signature for that animal.

PARENTAGE VERIFICATION

QUALIFIES/CONFIRMED or DOES NOT QUALIFY/EXCLUDED

Parentage is determined by examining the markers on the DNA profile. A result is generated and stated for all DNA parentage requests. Parentage confirmation reports can only be generated if a DNA profile has been carried out for Dam, Offspring and possible Sire/s.

PENDING

Results for this test are still being processed. Some tests are run independently and are reported at a later date. When completed, the result will be emailed.

APPROVED COLLECTION METHOD (YES)

The sample submitted for testing HAS met the requirements recommended by member bodies for the DNA collection process. The animal has been identified via its microchip number (Positive ID) and collected by a Veterinarian or Approved Collection Agent. APPROVED COLLECTION METHOD (NO)

The sample submitted for testing HAS NOT met the requirements recommended by member bodies for the DNA collection process.

TRAIT (PHENOTYPE)

A feature that an animal is born with (a genetically determined characteristic). Traits are a visual phenotype that range from colour to hair length, and also includes certain features such as tail length. If an individual is AFFECTED for a trait then it will show that characteristic eg. AFFECTED for the B (Brown) Locus or bb will be brown/chocolate.

POSITIVE – SHOWING THE PHENOTYPE

The animal is showing the trait or phenotype tested.

CLARIFICATION OF GENETIC TESTING

The goal of genetic testing is to provide breeders with relevant information to improve breeding practices in the interest of animal health. However, genetic inheritance is not a simple process, and may be complicated by several factors. Below is some information to help clarify these factors.

- 1) Some diseases may demonstrate signs of what Geneticists call “genetic heterogeneity”. This is a term to describe an apparently single condition that may be caused by more than one mutation and/or gene.
- 2) It is possible that there exists more than one disease that presents in a similar fashion and segregates in a single breed. These conditions - although phenotypically similar - may be caused by separate mutations and/or genes.
- 3) It is possible that the disease affecting your breed may be what Geneticists call an “oligogenic disease”. This is a term to describe the existence of additional genes that may modify the action of a dominant gene associated with a disease. These modifier genes may for example give rise to a variable age of onset for a particular condition, or affect the penetrance of a particular mutation such that some animals may never develop the condition.

The range of hereditary diseases continues to increase and we see some that are relatively benign and others that can cause severe and/or fatal disease. Diagnosis of any disease should be based on pedigree history, clinical signs, history (incidence) of the disease and the specific genetic test for the disease.

Penetrance of a disease will always vary not only from breed to breed but within a breed, and will vary with different diseases. Factors that influence penetrance are genetics, nutrition and environment. Although genetic testing should be a priority for breeders, we strongly recommend that temperament and phenotype also be considered when breeding.

Orivet Genetic Pet Care aims to frequently update breeders with the latest research from the scientific literature. If breeders have any questions regarding a particular condition, please contact us on **(03) 9534 1544** or **admin@orivet.com** and we will be happy to work with you to answer any relevant questions.